"IT'S NOT A SECRET BUT..." PREDICTIVE TESTING AND PATTERNS OF COMMUNICATION ABOUT GENETIC INFORMATION IN FAMILIES AT RISK FOR HUNTINGTON DISEASE

by

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ABSTRACT

The increasing transparence of the human genome has profound implications for how we understand health and illness and perceive our biological and social relatedness to others. Presymptomatic testing for adult onset conditions, in particular, creates the novel situation in which some individuals know in advance of impending illness while others learn that they have escaped such a fate. How families at risk for one adult onset condition — Huntington Disease (HD) — communicate about such information is the topic of this dissertation.

HD is often described as a ‘genetic time bomb’. It is an autosomal dominant neuropsychiatric disorder characterized by mid–life onset, involuntary movements, cognitive impairment, and depression. There is no effective prevention or cure but with the advent of predictive testing in 1987 it became possible for at risk individuals to learn if they had inherited the mutation associated with HD. Empirical studies on predictive testing for HD focus primarily on the individual psychological impacts of the test; few studies consider how families understand and attempt to manage genetic information in their everyday lives.

This dissertation begins to address these lacunae by examining the stories that test candidates and their families tell about hereditary risk and predictive testing. These stories derive from a prospectively designed study which includes 102 in–depth, at–home interviews conducted in the pre and post–results period with 16 test candidates and 33 family members. Focusing on three narrative ‘moments’, the dissertation explores how study participants storied their experiences of: 1) learning about the family history of HD, 2) deciding to request the predictive test and, 3) making sense of an informative result. Drawing upon a social constructionist approach, the analysis emphasizes the processual nature of predictive testing as well as the significance of interpersonal communication in producing and reproducing the social realities in which genetic information acquires a particular salience. Given the recent proliferation of genetic tests as well as the absence of an adequate popular discourse on embodied risk, the research underscores lay actors’ abilities to reframe existing clinical schema in order to interpret and manage hereditary risk in an intersubjectively meaningful way.
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This thesis is the product of many conversations. My interest in what it is like to live with the knowledge that one may develop an unpreventable, degenerative disease in mid-life began nearly twenty years ago when a friend explained to me that he was at risk for Huntington Disease. I had no idea, at the time, that his story would make such an indelible impression on me. Nor did I foresee the possibility that I would one day find myself struggling to articulate something about the meaning of the disease that he eventually came to understand all too well.

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The last acknowledgment is for my dear friend Margaret Harrison. She too believed in me and in this work. I miss her greatly and it is with profound respect that I dedicate this thesis to her.
DEDICATION

For my friend
Margaret Mary Harrison (1930–1999)
As she said,

“We think many of the same things, we just use different words.”
CHAPTER I
CLAIRVOYANCE

There is an allure to trying new technologies. We flex our laboratory muscles and prepare to test our latest diversions. And there is something particularly aesthetic about genetic diagnosis, with those clean, impersonal bars stretching across the lanes at their appointed positions. Without words, they can reveal past couplings or confess extramarital transgressions. And they are clairvoyant, unpeeling shrouds from the future as well as the past. (Wexler, 1990:80)

The increasing transparence of the human genome has profound implications for the ways in which we understand health and illness, for how we view ourselves and, ultimately, for how we perceive our biological and social relatedness to others. Indeed, there is no more cogent illustration of the social significance of scientific developments, than the rapidly developing field of molecular medicine.

In 1990 the Human Genome Organization launched an international effort to map and sequence the entire human genome. Mapping involves the compilation of genetic linkage and physical maps which indicate where an estimated 50,000–100,000 genes reside on each of the 23 pairs of human chromosomes.¹ Sequencing, a separate but related activity, involves working out the precise order of approximately 3.3 billion nucleotide bases.² When complete, this project will provide a computerized catalogue containing the basic biochemical “recipes” for the entire human genome.³ Proponents of the Project argue that this map will contribute to the discovery of solutions to long-standing puzzles of development and cellular function. From a public perspective, however, the primary significance of genome mapping will likely derive from its use in the prediction and diagnosis of hereditary disorders (Kevles & Hood, 1992).

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¹ The full complement of human chromosomes is comprised of twenty-three pairs or a total of 46 chromosomes: this includes twenty-two pairs of autosomal chromosomes and one pair of sex-determining or germline chromosomes (the XX pair determining a female and the XY pair determining a male).

² DNA (deoxyribonucleic acid) is comprised of two parallel strands which are linked together by four pairs of bases—adenine, thymine, cytosine and guanine. As a unit of information, each gene is made up of a specific sequence of base pairs. Variation in the sequence of base pairs gives each gene a unique structure.

³ Researchers have already compiled low resolution maps of all human chromosomes and, given adequate funding, sequencing of the entire human genome will be approaching completion by 2006. The projected cost of this project is $3 billion although this amount could easily be exceeded (Garver and Garver, 1994).
Molecular Medicine

The genetic basis for many hereditary disorders was known well before the advent of the Human Genome Project. Nonetheless, medical genetics is only now beginning to realize its potential as an overarching means of understanding and explaining the causes of human disease. Commenting on his vision of the "Holy Grail", molecular biologist Walter Gilbert (1992:94) argues that "the possession of a genetic map and the DNA sequence of a human being will transform medicine". The most immediate change will be a knowledge of genes that cause rare genetic disease. Of broader significance, however, is the suggestion that a detailed genetic map will lead to the identification of genes responsible for conferring susceptibility to a host of much more common afflictions.4

There are now over 4,000 disorders with a known genetic basis and an estimated “60 percent of the population will experience a disease with a genetic component to its cause” (Science Council of Canada, 1991:21). Further, there are over seventy genetic tests capable of detecting and/or predicting inheritable disorders. Ontario, which has the most extensive genetic testing programs in Canada, routinely offers tests for fifty-six genetic conditions.5

Genetic testing is based upon at least two common techniques of DNA analysis. If the gene has been mapped and sequenced, gene probes can be developed and used to look for the precise series of base pairs which describe a specific disease gene. This direct form of genetic testing reveals whether or not the at risk individual has inherited the specific genetic mutation associated with the disorder. Alternatively, if the precise sequence of base pairs describing the gene for a particular disorder is as of yet unknown, stretches of DNA which are known to be linked to this gene can be identified and used to estimate the probability that the disease-related gene is also present. The genetic markers used in linkage analysis exist in a number of forms

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4 A quick perusal of my clipping file reveals a cross-section of recent discoveries: “Genetics Key to Colon Cancer Cure” (Independent Senior, February 1994); “Researchers Isolate Gene in Breast Cancer Fight” (The Vancouver Sun, September 15, 1994); “Canadian Scientist Leads Discovery of Gene for Early Onset Alzheimer’s Disease” (Canadian Genetic Diseases Network, June 28, 1995); “Genetic Parkinson’s Link Probed” (The Vancouver Sun, November 15, 1996); “Scientists Closing in On Gene Believed Linked to Prostate Cancer” (The Vancouver Sun, November 22, 1996).

5 These are, however, only a fraction of the tests which may be available in research-related and private facilities elsewhere in North America (Strauss, 1995). According to the U.S. based Helix National Directory of Diagnostic Laboratories, there are at least 275 conditions for which there is now, or soon will be, an accurate genetic test (Strauss, 1995).
and hence, it is often necessary to compare the DNA of various family members in order to determine which marker is associated with the genetic disorder in each particular family.

With availability of a high resolution map of the human genome, it will become possible to link all single gene disorders with specific genetic markers — a technical accomplishment which proponents argue will permit detection of virtually any single-gene disorder. Early detection of inheritable disorders may, in some instances, lead to interventions which delay onset of disease or avert its most serious consequences. For instance, the 1–2 percent of the population with a single-gene form of hyperlipidemia may, through diet and medication, avoid early heart disease (Baird, 1990). At present, however, medicine’s ability to detect harmful genetic traits far outstrips the ability to offer treatment or cure for the resulting diseases.

In the absence of effective treatment or prevention for inheritable disorders, the provision of information about genetic risk is itself emerging as a new type of medical intervention (Cox, McKellin, & Burgess, 1995). The decision to undergo predictive genetic testing is, however, highly personal and has profound implications for at risk individuals and their families. Reflecting on some of the implications of predictive testing, psychologist and researcher Nancy Wexler (1990:80) — who is herself at risk for Huntington Disease — suggests that several aspects of predictive testing are of particular salience to understanding the increased social and biomedical significance of genetic information. First, predictive genetic testing forecasts with varying degrees of probability or certainty, the possibility of becoming affected by an inheritable disorder. Such knowledge is distinct from that offered through clinical diagnosis since most diagnostic tests are initiated after the patient exhibits signs and symptoms indicative of the actual presence of disease.

With adult onset disorders in particular, predictive genetic testing creates the novel situation in which some individuals know in advance, and with a high degree of certainty, of impending illness while others learn that they have escaped such a fate. This information may reduce uncertainty but it is as of yet unknown how it will shape the life trajectories and illness related experiences of at risk individuals and their families. As Nancy Wexler's sister Alice (Wexler, 1995:xv) asserts, the possibility of knowing, in advance, of impending illness raises stark questions about “the meanings of certainty and uncertainty and what it means to occupy a
‘third space’ outside the categories of either—or that we conventionally use to organize experience”.

Second, genetic information does not “belong” to just one person. Given that we inherit DNA from both parents and share an average of fifty percent of the same genes as our siblings, predictive testing reveals information which is both individual and familial in orientation. The modification of risk which accompanies an informative test result may, therefore, have implications not only for the individual being tested but also for his or her offspring and, possibly, siblings.6 This is in contrast to most diagnostic information which centres exclusively on the patient who has, or is suspected of having, the disease condition. Further, since it is often necessary to obtain DNA samples from multiple family members, there is always the potential for discovery of non-paternity. As Nancy Wexler (1990:80) eloquently suggests, the results of genetic testing can “without words...reveal past couplings or confess extramarital transgressions.”

Finally, genetic information has an intimate connection with our individual (as well as collective) need to define ourselves in relation to others and in so doing, recognize both our similarity to, and difference from, others. Genetic information is, therefore, intertwined with ongoing existential processes of self–identity and perception — that is, one’s sense of physical, intellectual, emotional and spiritual “beingness” in the world. Moreover, genetic information is also deeply connected to our notions of uniqueness and personhood, agency and self–determination (Brock, 1994).7

Each of us is and has a body and certain mental phenomena which are — in whatever mysterious ways — associated with that body. Further, each of us has a history and is situated within a setting of other persons, practices and ways of looking at the world. These things, most would agree, are somewhat malleable aspects of who we are. In contrast, our DNA appears to

6 With disorders which are transmitted via a pattern of dominant inheritance (such Huntington Disease), a test result confirming presence of the gene indicates that at least one parent of the person tested must be (or have been) a gene carrier, whether or not this parent has been diagnosed with the disease. In consequence, the presence of the gene in any one of the offspring alters the parent’s risk from 50% to 100% and simultaneously this result also alters the risk of all siblings of the tested offspring from 25% to 50%.

7 Scientific developments are, however, challenging our definitions of personhood. As the ominous implications of successfully cloning an adult sheep begin to worry the world, many are wondering whether human cloning might be just around the corner and, if so, whether genetic uniqueness is a necessary condition for personhood.
have a more fixed quality; it is a “blueprint” which specifies something about how we came to be and who we will become (Shapiro, 1991). Our DNA is, according to this way of thinking, a kind of “future diary” which reveals our aging selves to our more youthful ones (Elmer–Dewitt, 1994).

Metaphors used to describe the genome as a “blueprint”, “computerized catalogue” or “recipe” for who we are may render the complexities of the human genome understandable to a lay public with minimal scientific literacy, but they do not offer such enlightenment without a price. They cajole us into thinking, like molecular biologist Walter Gilbert (1992:96), that “we are determined, in a certain sense, by a finite collection of information that is knowable”, and as such they may promote a form of genetic reductionism. Lippman (1991:18–19) refers to this type of reductionism as geneticization — that is, an ongoing process “in which differences between individuals are reduced to their DNA codes”, “most disorders and behaviors, as well as physiological variations are defined as at least in part genetic in origin”, and “interventions that employ genetic technologies to manage problems of health [are] advocated.”

Geneticization shifts the focus of medical attention from the person to the genome and in so doing, it reorients the way in which problems are defined. New genetic knowledge and techniques shape not only the ability to detect, and intervene in, but also the very ways in which we think about health and illness, normality and abnormality. In consequence, geneticization has important implications for the design and delivery of health care, the priorization of health care needs, and, the structure of physician–patient relations (1991; Lippman, 1992). No longer is the individual the only patient; each “proband” has a family history which is summarized in a pedigree and investigated as a necessary and routine element of the medical genetics consultation (Nukaga & Cambrosio, 1997). The individual patient is, in this context, part of a larger (and more relevant) social as well as biological whole. For health–care providers, who must observe norms of individual patient autonomy, this creates an inevitable tension (Nelson & Nelson, 1995). As several participants in a European workshop on the ethics of genome analysis suggest, “new genetic technology not only creates difficult options for patients and medical doctors, it also transforms the social relationships of the medical world” (De Dinechin, et al., 1993:258).
**Ethical, Legal and Social Implications of Genetic Information**

As genome mapping and sequencing proceed, more and more disease genes are being identified and a wider range of diagnostic and predictive tests are becoming available. How the information derived from genetic testing is to be used is, therefore, an issue of growing social, ethical and legal importance.

In response to concerns about the potential use and management of genetic information, the U.S. based Human Genome Project established an Ethical, Legal and Social Implications (ELSI) program to prospectively evaluate the various impacts of new genetic knowledge and techniques as they are being developed. Canada followed suit in 1992 when the newly established Canadian Genome and Technology Program (CGAT) also included an ELSI program. Critics of the Human Genome Project perceive these programs as a necessary and positive step, yet many remain concerned that such initiatives do not yet take the eugenic potential of new genetic knowledge and techniques seriously enough (Garver & Garver, 1994; Hubbard & Wald, 1993; Lippman, 1991).

The movement to improve the human species either through culling out "undesirables" or by using genetic techniques to enhance the number of desirable traits has — since Sir Francis Galton coined the term "eugenics" in 1883 — enlisted its share of support from sociology as well as biology. In the late nineteenth century, sociologist Herbert Spencer popularized the particular blend of naturalistic thinking and laissez-faire economics which came to be known as Social Darwinism and, in the early twentieth century, allies of this movement seized upon the emerging science of genetics to justify eugenic notions of racial superiority, lawful and even mandatory sterilization of the poor and "feeble-minded" (McLaren, 1990), exclusionary immigration policies, and, a lack of state intervention in poverty, ill-health and discriminatory employment practices (Kevles, 1985). Reaching its most heinous apex in Nazi Germany, the

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8 Soon after it was established, the Canadian ELSI initiative was expanded to become MELSI (medical, ethical, legal and social implications). Some, such as the Social Science Federation of Canada, interpreted this as a decreased commitment to support social science research.

9 In particular, Social Darwinists emphasized that through endless competition, adaptation, and "survival of the fittest", the best and most intelligent social groups would prevail and improve the evolutionary capacity of human society as a whole.

10 In Nazi Germany, an active program of euthanasia included individuals affected with Huntington Disease (Garver and Garver, 1994:153).
eugenic ideals of racial betterment were banished in the years following World War II.

More recently, many scientists and medical geneticists as well as lay and social science critics, have voiced concern that genetic information may lend itself to new types of discrimination (Hubbard & Wald, 1993; Kevles & Hood, 1992; Knoppers, 1991; Lippman, 1991). Critics have argued that employers, insurance agencies, the government and the military will, for various reasons, want to have access to individuals' genetic information. When placed in the context of unfair employment practices, contentious claims about legal liability and an increasingly cost-efficient calculus for the provision of health and life insurance, such third-party access to genetic information becomes increasingly problematic (Billings, et al., 1992; Draper, 1991; Duster, 1990). 11

Concerned about the potential abuses of genetic information, the Privacy Commissioner of Canada (1992) cautions that existing legislation is wholly inadequate as means of ensuring that genetic technology is used for appropriate ends. Information about genetic risk is exceedingly personal and reasonable expectation dictates that it would be shared with another only under circumstances of trust and confidence. As such most clinicians as well as policy-makers maintain that it is essential for people to maintain meaningful control over the communication of genetic information (European Community Huntington’s Disease Collaborative Study Group, 1993; Huggins, et al., 1990; World Federation of Neurology & International Huntington Association, 1990). In particular, at risk individuals must be able to decide whether or not they wish to learn about their genetic status and, if so, how and when it may (or may not) be appropriate to disclose this information to others.

An adequate framework for managing genetic information will, therefore, need to have at least two components: “protection from one’s own hitherto unknown secrets” and “protection from the intrusions of others” (Privacy Commissioner of Canada, 1992:4). From a clinical standpoint, the first of these is largely satisfied through standardized protocols which emphasize the voluntary nature of predictive testing, the importance of informed consent and the need to

11 Billings et al (1992) have documented at least forty–one instances of possible genetic discrimination in the U.S. Of these, thirty–two involved insurance related issues (e.g., applications for coverage, changes in health, life and longterm disability insurance, mortgage and automobile insurance) and seven others involved various employment related situations (e.g., hiring, termination, transfer and promotion).
respect individual autonomy in decision-making. As such, most service providers maintain that just as at risk individuals retain the *right to know* their genetic status, they also retain a parallel *right not to know*._\footnote{Taking exception to this, Shaw (1987) argues rather more pointedly that individuals at risk for HD have “an ethical duty to know” whether or not they are gene carriers and to disclose such information to other family members.}

Given that test results often have significant implications for the at risk individual’s spouse/partner and family as well as a range of other third parties, the second aspect of genetic privacy is, however, somewhat more problematic. Attempting to juggle confidentiality with perceived responsibilities to the at risk individual’s family, medical geneticists express divergent views about appropriate practices of disclosure. In a survey of medical geneticists from nineteen different countries, Wertz and Fletcher (1989) found that only one-third of the 700 respondents would uphold completely the privacy of patients with Huntington Disease or parents of children with haemophilia; another third would, if asked, tell the patient’s relatives and, one-quarter would go so far as to independently seek out relatives and tell them. Genetics patients or clients also express divergent perspectives on disclosure. A survey of 283 families with a known history of chromosomal rearrangements revealed that a significant proportion of individuals with such rearrangements either do not disclose genetic risk information to relatives or, “they transmit it in such a way that relatives do not perceive the necessity to explore it” (Ayme, et al., 1993:573).

If there is a *right to know*, a *right not to know* and a *duty to know* (Shaw, 1987), how do medical geneticists and counsellors make such distinctions? Bioethics may assist in finding case or policy-oriented resolutions to such dilemmas by clarifying how important principles of clinical practice — such as individual autonomy, the avoidance of harm (non-maleficence), promotion of benefit (beneficence) and justice — come into conflict (Beauchamp & Childress, 1994). For instance, if an individual’s genetic test results reveal the presence of a gene associated with a severe but preventable heritable disorder there may be a duty to warn any relatives who may also be at risk for the disorder. If the individual being tested refuses to disclose this information, clinicians will have to choose between the competing principles of
autonomy (i.e., the individual’s right to self-determination, confidentiality and privacy) and avoidance of harm (i.e., the relative’s right to take advantage of any available preventive strategies) (Andrews, 1997).

The resolution of such conflicts may, in some instances, be achieved through an a priori ranking of the above mentioned ethical principles. Indeed, it has been proposed that the principle of autonomy should take precedence over all others (Beauchamp, 1982). Elsewhere, however, empirical studies of genetic counselling and obstetrics attest to the salience of practical concerns, emotions and cognitive strategies in understanding processes of ethical decision-making. As Wolf (1994) argues, principalism is, within the clinical context, primarily a method of providing a post-hoc rationalization for what has already been decided.

Holding that much of traditional ethics fails precisely because it attempts to sustain the objectivity of moral thinking by extracting it from all substantive context, many feminist philosophers (Sherwin, 1992) and an increasing number of ethicists (Burgess & Hayden, 1996; Hoffmaster, 1992; Hoffmaster, 1994) now advocate a more contextual approach. In this approach, moral and ethical controversies,

... are not about the major or minor premises of deductive arguments, as in philosophical ethics. Rather, they are deeply embedded in and connected to the fundamental, constitutive facts of our lives, the concepts, beliefs and values that give structure to our lives and interactions with others and determine what is important to us. (Hoffmaster, 1994:1161)

The practical and ethical dilemmas which arise in everyday social life as well as clinical practice are situational; they cannot be adequately understood without reference to the personal circumstances, cognitive strategies, socio-economic resources and/or communicative competence of those who are enmeshed in such circumstances. As such, a “new pragmatism” is slowly emerging, a hybrid of bioethics and health law which encompasses a range of more inductively oriented strategies — the revival of casuistry, a call for more empirically-based studies, a growing interest in narrative ethics and the emergence of care-based ethics are but a few of these (Wolf, 1994).

That good ethnographic studies are urgently needed to tackle the dilemmas associated with the management of genetic information now seems increasingly obvious. Despite an extensive literature which focuses on attitudes toward predictive testing, the importance of
counselling and support, the clinical disclosure of test results, and, the minimization of anxiety and other adverse events, little attention has been devoted to the everyday experiences of individuals and families as they attempt to understand and manage genetic information in their everyday lives. Moreover, as Gordon and Paci (1997) have shown in their finely nuanced ethnographic explorations of concealment and silence around the diagnosis of cancer in Tuscany, there are a host of background assumptions and routine practices which must be interpreted from the standpoint of those most directly involved. Practices of disclosure and non-disclosure are not isolated; they are enactments of our most fundamental values; and they operate within particular, historically-specific “visions of good” (Gordon & Paci, 1997).

As genetic tests become available for a growing number of heritable disorders (e.g., Huntington Disease, Alzheimer’s Disease, breast/ovarian and colon cancer), the social, ethical and clinical dilemmas associated with the delivery and management of genetic information will become increasingly prevalent. In order to provide solutions that are flexible and yet respectful of the individual and familial orientation of genetic information it is essential to know how at risk individuals and families themselves understand and communicate about hereditary risk and predictive testing. Given many service providers’ sincere desire to enhance patient autonomy and avoid medical paternalism, such knowledge could provide an important corrective to policies and procedures which do not adequately account for the perspectives of predictive testing participants and their families. Moreover, given that diminishing economic resources for health care will constrain existing levels of counselling and support available to predictive testing participants, it is vital to consider how a variety of non-clinical factors shape the short and longterm well-being of at risk individuals and their families.

From a sociological perspective, this situation presents many important and unexplored lacunae for research. In particular, there is an urgent need to know more about how growing up in a family with a history of genetic disease shapes an individual’s self-identity and relationships with others (both in and outside of the family), how and when at risk individuals and families communicate about genetic disorders, how lay beliefs about inheritance modify and incorporate new information from scientific genetics, and, how ethnicity, class and gender shape different approaches to genetic problems (Richards, 1993). Unfortunately, few of these aspects
of the new genetics have yet been explored by social scientists in any systematic way.13

“It’s Not a Secret But...”

This dissertation is a prospectively designed ethnographic inquiry into predictive testing and patterns of communication about genetic information in families at risk for Huntington Disease. In particular, I am interested in how PT candidates and their families communicate about their awareness of the family history of Huntington Disease, the implications of hereditary risk, the decision to have predictive testing, and the experience of receiving and making sense of a test result.

The title of this dissertation, “It’s Not a Secret But...”, is an amalgam of thoughts and feelings, words and silences, about the experience of living at risk for Huntington Disease as it has been conveyed to me by at risk individuals and their families. It is intended to suggest a sense of ambivalence about the experience of sharing information about oneself and related others when such information is constructed as a powerful source of, as well as an ever-present threat to, self–identity, intimacy, and social life (Goffman, 1959; 1963).

Huntington Disease and Predictive Testing

Huntington Disease (HD) is a severely debilitative, neuropsychiatric disorder for which there is, at present, no effective prevention or cure. Onset of HD typically occurs in mid–life and is characterized by loss of control over voluntary movements. As the disease progresses, it causes gradual but inexorable physical and cognitive decline (Hayden, 1981).

As an autosomal dominant disorder, HD affects both men and women. All offspring of an individual who has the gene for HD will have, at birth, a 50:50 chance of either inheriting or escaping the disease. Inheriting the HD gene is, therefore, a random event which genetic counsellors often compare to the flip of a coin. Not inheriting the gene — an equally chance event — means that the “chain is broken” and HD will not reappear in subsequent generations. Because of its typical mid–life onset and devastating progression, HD is described as a “genetic

13 Richards (1993) proposes that social scientists have been reluctant to do research on genetic issues because of a long-standing distaste for notions of human action based on biological determinism. A further deterrent may arise from the highly technical language of genetics, an aspect which may prove difficult or alienating for those without scientific training. In addition, I would add that for many social scientists there also remains the sometimes thorny issue of negotiating access to a suitable clinical or research population.
time bomb” which “remains dormant until the person reaches adulthood” (Huntington Society of Canada, 1996). The gene for HD is, however, fully penetrant which means that anyone who has inherited the gene will, if they live long enough, eventually become affected.

Until recently, the uncertainty of living at risk for HD could be only be resolved in an unfavourable direction — that is, through a clinical diagnosis of the disease. However, with the 1983 discovery of a linked marker for the HD gene a presymptomatic genetic test was developed. As the first proof of the power of new techniques in gene mapping, this discovery was of enormous significance to the human genetics and molecular biology communities (Wexler, 1995).

In 1986, genetics centres in the U.S., Canada and the U.K. began to offer the linkage test to those at 50% risk for HD. When informative, the linkage test indicated to individuals at 50% risk that they had an increased or decreased probability of developing HD. Given concerns about the lack of effective therapeutic interventions for HD and the potentially catastrophic effects of receiving an increased risk result, the test protocol included extensive psychosocial assessment, numerous counselling and follow-up sessions as well as a protracted period of time in which the test candidate could reconsider or withdraw from the program (Fox, et al., 1989). Indeed, the care devoted to devising an appropriate and ethically sound test protocol has been supported by clinical findings which suggest that most individuals who receive adequate counselling and support are able to cope well with learning their genetic status (Wiggins, et al., 1992).

With the discovery, in 1993, of the gene for HD, it became possible to provide at risk individuals with a definitive or direct type of predictive test (HD Collaborative Research Group, 1993). In contrast with linkage testing, the direct test is based upon analysis of the region of DNA believed to be directly implicated in causing HD. This region — which is located on the short arm of chromosome 4 — has been described as a kind of “genetic stutter” in which the trinucleotide sequence of CAG is repeated many more times than in the unaffected population.14

14 The degree of expansion in this region is, at its upper limits positively correlated with earlier age of onset for HD but, given the wide variance in the correlation for most repeat ranges, this data is considered useful only in a small number of cases (Duyao et al, 1993; Simpson et al, 1993). In consequence, there is at present a great deal of
Experiences in offering predictive and prenatal forms of the test to individuals at 50% risk for HD, have been central in understanding many of the clinical, ethical and psychosocial implications of predictive genetic testing. In fact, "HD represents the disorder for which predictive testing has been offered for the longest time, to the largest number of people" (Benjamin, et al., 1994:615). As Mattsson and Almqvist (1991:27) suggest, however, the accuracy and certainty of a direct test for HD imposes a heavy burden of responsibility on clinicians and researchers.

It is a serious exhortation to those who accelerate the technical development not to ignore the ethical consequences of such a test. When reliability of the test and technical perfection is 100% all hopes of not being a victim are dashed to the ground. Without such hope the human consequences of suffering, fear and irresoluteness may be high.

Such concerns have not been raised in isolation although they are perhaps indicative of a fairly profound shift — that is, from the question of whether or not to offer predictive testing to the question of how best to continue offering it. Maintaining responsibility for the immediate well-being of PT candidates, most service providers continue to monitor carefully the psychosocial effects of predictive testing (Benjamin, et al., 1994). Further, many struggle with limited resources to ensure that predictive testing continues to be offered only within the context of extensive pre and post-test counselling and support. As Hayden (1991) argues,

In these days of significant pressure to cut costs, there is the temptation and pressure for programs which are being initiated to try to limit their costs by diminishing follow-up care. In some sense, this is like performing an operation without providing post-operative care. If one is not able to provide appropriate follow-up support, then perhaps it is not advisable to do the operation in the first place.

Although it has, for obvious clinical reasons, been critical to establish the individual psychosocial impacts of predictive testing, the important task of situating these individual impacts within their familial and social contexts has, overall, received comparatively little attention. Moreover, most studies which do acknowledge the significance of the familial impacts of predictive testing have been conducted from the perspectives of various service providers. Quantitative approaches and, in particular survey methods, have been almost universally debate within the medical genetics community about whether or not repeat numbers should be provided to predictive testing participants.
favoured and the test results almost always constitute the primary independent variable. As such, there is an overall neglect of social factors which shape and differentiate the predictive testing experience.

The social and familial implications of predictive testing for HD neither begin with the decision to have the test nor end with the clinical disclosure of results. Disclosure is one pivotal point in a much larger process which must be elaborated if we are to understand the significance of this new type of medical intervention. For many families at risk for HD, the story begins with the diagnosis of a mysterious family illness. This precipitates a search for information, worry about having children who may also be at risk and, for some, the dilemma of whether or not to request predictive testing. For others, HD is an unwelcome genetic heirloom which is all too predictably passed from one generation to the next; some of its heirs may decide to confront their probable fate through genetic testing while others prefer to leave open the question of whether or not they will eventually experience onset of HD.

Patterns of Communication About Genetic Information

Broadly stated, the purpose of this dissertation is the interpretive understanding of social action (Weber, 1968). Adopting a hermeneutic approach, I am less interested in the cause and effect relationships which feature prominently in the clinical literature than I am in how PT candidates’ and their families’ arrive at an intersubjectively meaningful interpretation of the experience of predictive testing. Familial communication is central to this understanding because talking is itself a means of allocating things a definite place in the world. As Berger and Luckman (1966:174) propose, “language realizes a world, in the double sense of apprehending and producing it.”

Moving out of the clinic and into the everyday world of individuals and families at risk for HD, this dissertation therefore focuses on patterns of communication about genetic information within the sphere of familial and social rather than clinical interactions. In particular, my analysis foregrounds the stories that PT candidates and their families tell about how they communicate, in their everyday lives, about:
1) the family history of Huntington Disease and its implications for various family members,
2) the decision to request predictive testing, and

3) the process of receiving and making sense of an informative test result.

This focus on how families communicate about genetic information within everyday life gives rise to a number of questions. When and how do at risk individuals become aware of the family history of HD? With whom do these individuals discuss the implications of hereditary risk? Is there someone within the family who takes primary responsibility for initiating such discussions? How do at risk individuals decide whether or not to request the test? To whom do they disclose their test results? How do family and friends respond? What are some of the consequences of disclosure for the PT candidate and their family? Such questions point to the processual nature of predictive testing as a lived experience which unfolds in and through time. This experience becomes storied as PT candidates and their families engage in a process of “account formation” (Stimson & Webb, 1975) within their everyday lives and within the context of the interviews which form the basis of this research.

Following a prospective study design, the research which informs this dissertation includes a total of 102 in-depth interviews with sixteen participants in predictive genetic testing, thirty-three of their family members and/or close friends and one couple where the wife was at risk but did not want to proceed with predictive testing. Together these fifty-one study participants represent seventeen different families who live in both urban and rural settings in the province of British Columbia. With few exceptions, study participants were interviewed individually in their own home (or home communities). In almost all cases, participants completed at least one pre-results interview (conducted several weeks prior to results) and one post-results interview (conducted approximately six months after predictive testing results). In addition, six families participated in a third interview (conducted one to two years after results). These interviews covered a consistent set of themes but all study participants were actively encouraged to talk about what they felt was most important and to frame this in whatever ways seemed most salient to them at the time. This approach enhances the validity of participant

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15 There were only three exceptions: one PT candidate and his brother resided in the United States and the brother of another PT candidate lived in Alberta.

16 See the interview schedules contained in Appendix VI.
accounts by allowing participants themselves to pattern the timing, sequence, content, and context of topics discussed (Mishler, 1991). Interviews lasted from one to three hours and were tape recorded and transcribed for subsequent analysis. The stories and narrative accounts which derive from these interviews offer a rich source of insight into the experience of predictive testing as it is understood from the perspectives of at risk individuals, their spouse/partners, children, parents, siblings, other family members and friends.

Reflexivity and the Researcher

This dissertation is about the experience of predictive testing and the patterned ways in which PT candidates and their families communicate about genetic information. It is, however, also about the stories that people tell about their acts of communication. These two aspects are not one and the same nor are they separable. Though social scientists have methods of conceptualizing the understandings that practitioners of communication have of their own communication, most social scientists do not adequately reflect upon their own hermeneutic participation in the communication they seek to understand. Indeed, the self-referential has until recently been treated by much social theory as if it were something of an oddity: this is especially the case with theories of human communication (Krippendorf, 1994).

Before proceeding any further, I therefore need to say something about how I conceptualize my own role in this research, about how I am both observer and observed, listener and listened to. This is more than an attempt to be reflexive. As instances of the very subject I seek to study, the interviews conducted with at risk individuals and their families not only tell me about specific instances of communication, they are specific instances of communication which shape and are shaped by pre-existing familial awareness contexts. As such, the research is both enhanced and confounded by the very embeddedness of subject matter in method. This aspect of the research therefore demands more than the usual nod to interviewer effects; it demands that I locate myself, as a kind of atypical friend of the family, within the context of the communicative interactions which I describe.

17 Five of these accounts have been prepared for inclusion in a forthcoming publication from the Huntington Society of Canada. These are included in Appendix VIII.

18 I am grateful to Nancy Waxler-Morrison for her suggestion that I conceptualize my relationship to interview participants in this way.
During the interviews, I was a listener and observer but I was also engaged in an ongoing conversation which had significant implications for how at risk individuals and their families communicated with each other in their daily lives. One brief illustration will clarify. A young man whose mother was undergoing predictive testing told me, after the first interview, that he had never talked so much about HD and that he now wanted to talk to his mother and sister about some of the issues he had not previously considered. I initially thought of this as a possible therapeutic benefit of the interview but later heard from his sister that he had been quite depressed. The interview had not only given him reason to reflect on how he and his family communicated with each other; it was an intervention which shaped and thereby became a part of the patterns of familial communication that I sought to understand.

As mentioned above, the descriptive and analytic work of this dissertation is primarily based on the narrative accounts generated by interview participants. In addition, I make use of my fieldnotes as a source of data which records the metacontext of the interview and situates me (as researcher and friend of the family) within the context of what I describe and analyze. The perspective I adopt here is, however, also reflective of other research related experiences. I had the opportunity to observe a number of genetic counselling sessions and, in addition, I volunteered for several years at the annual retreat for persons with HD. This was, for me, a watershed experience through which I have come to understand quite differently my relationship to this research. In particular, the experience of being at the retreat taught me the importance of integrating (rather than merely acknowledging) the emotional as well as organizational and analytic work that is an inextricable element of doing and writing about research such as this (Kleinman & Copp, 1993; Rothman, 1986). As such, I try to retain throughout the dissertation, a sense of what the work entailed.

Like Campbell (cited in Thompson, 1996), I believe that we need to take the risk of revealing ourselves as "live sociologists at work". Further, I suggest that the judicious use of moments of introspection about personal experience can provide an alternative means for "comprehension of the other by the detour of the self" (Timmermans, 1994). Finally, the integration of reflexive commentary about the nature of the relationship between personal experience and the research also helps to establish trust: without such reflexive commentary, the
reader cannot assess the degree to which the researcher distinguishes between her own experiences and the experiences of those she studies (Reinharz, 1992).

Summary of Chapters

In what remains, I offer a brief summary of each chapter in the dissertation. This will provide the reader with a sense of how my general argument unfolds.

My purpose in Chapters II and III is to situate contemporary lay and scientific understandings of HD within their socio–historical context and locate the tensions and gaps in existing research on predictive testing for HD. Taking the naming of ‘that disorder’ as a motif, Chapter II reviews the social and biomedical history of HD — from its description, by George Huntington, as a distinctive form of hereditary chorea to its contemporary molecular characterization as a kind of “genetic stutter”. These processes of naming bring to the foreground the social history of ‘that disorder’, its hereditary nature and place in the history of eugenics, as well as the more recent progress which scientists have made in elucidating its molecular characteristics.

In the absence of effective prevention or cure for HD, predictive testing offers a new type of medical intervention which has significant implications for at risk individuals and their families. For some, the test provides resolution to the uncertainty of living at risk; for others it introduces new types of uncertainty. The predominant clinical discourse of predictive testing is, however, a “discourse of potential benefits” (Bouitte, 1988) and the results are typically constructed as a “gift of knowing” (Kenen, 1996). Chapter III considers this discourse as it shapes existing research on, and clinical approaches to, predictive testing. In this chapter, I review key aspects of the experience of diagnosis and living at risk for HD. I also evaluate existing studies of the clinical and psychosocial implications of predictive testing, pointing out the need for alternative approaches to understanding how PT candidates and their families jointly construct the meaning of hereditary risk within their everyday lives. Noting that little is known about the patterns of communication which shape, and are shaped by, at risk individuals’ and families’ experiences of predictive testing, I suggest that research on this topic has much to offer lay and social science as well as clinical understandings of the meaning of genetic testing.
In Chapters IV and V, I set out the theoretical and methodological approach taken in this dissertation. Familial communication about genetic information and the stories that PT candidates and their families tell about their communication may be seen as constituting two intersecting levels of analysis. Taken separately, each constructs a different object of analysis and invokes a somewhat different range of potential theoretical and methodological approaches. These two chapters therefore bring together several theoretical and conceptual frameworks: interactionist approaches to understanding health and illness offer an umbrella in that interpersonal communication is, of necessity, central to theorizing the social construction of reality (Berger & Luckmann, 1966). Within this rubric, my analysis of familial communication about HD and predictive testing is informed by Glaser and Strauss’s (1964; 1965) theory of awareness contexts as well as studies which have since offered some refinement of this theory. In addition, it would be difficult to examine the management of personal information without referring to the concept of stigma (Goffman, 1963) and the way that it is both felt and enacted in the lives of those with chronic illness and/or disability (Davis, 1961; Scambler & Hopkins, 1986; Schneider & Conrad, 1980; Wendell, 1996). My approach to the stories that people tell about their acts of communication is oriented by a somewhat separate literature on the role of biography and narrative in shaping and representing illness experiences (Bury, 1982; Couser, 1997; Frank, 1995; Gordon & Paci, 1997; Kleinman, 1988; Mishler, 1984; Mishler, 1991). This literature is not incompatible with the literature on awareness contexts but it suggests a different line of approach and invokes a different level of analysis.

In setting out my methodological approach, I draw upon Mishler’s (1990; 1991) dialogical approach to interviewing (1990; 1991), and Kleinman (1988) and Frank’s (1991; 1995) attention to the analysis of illness narratives. Among other things, these contributions demonstrate how the voice of medicine often interrupts the voice of lifeworld, subverting patients’ efforts to construct and tell their own story in a socially meaningful way. 

Chapter V also provides an overview to the research design, protocol for predictive testing, recruitment procedures and study sample. Moving out of the clinic and into the everyday lifeworld of study participants, I then describe the process of interviewing and, explore the dialectics of being an interviewer and an atypical kind of friend of the family. In particular, I
attend to the act of listening and how, over time, I learned to hear differently. Finally, I conclude by exploring the ontological status of the interview as a communicative event. On one level, the interviews are occasions for study participants to talk about their acts of communication. On another level, however, the interviews are instances of communicative interaction.

Having teased out these various levels of analysis, I proceed in Chapters VI, VII and VIII, to present three narrative "moments" which elucidate the substantive focus of this dissertation. This story of familial patterns of communications about genetic information has several components. Chapter VI focuses on study participants' initial awareness of Huntington Disease and the family history. Chapter VII focuses on learning about and deciding to have predictive testing, and Chapter VIII focuses on the process of making sense of the results of predictive testing. The structure of these three chapters is chronological; they tell a story of transitions in familial awareness contexts, the social factors which both enable and constrain these transitions and, some of the consequences of these transitions for families at risk for HD.

Chapter IX offers a synthesis of the substantive issues arising from the three data chapters. In this chapter, I draw upon the phrase which names this dissertation — that is, "It's Not a Secret But..." — in order to consider the intersubjective meanings of various strategies for managing genetic information. Moreover, I also consider how these strategies are embedded in and reflective of larger cultural narratives as well as gendered practices of managing genetic information.

Chapter X concludes the dissertation by summarizing and reflecting on the central findings and their clinical, ethical, sociological, and methodological implications. Also attending to what is of the greatest importance to families at risk for HD, I emphasize the practical significance of conversation and storytelling. There is, as narrative therapy suggests, a therapeutic moment which accompanies storytelling (Wiersma, 1992); this moment is sometimes cathartic but as Cruikshank (1998) demonstrates, oral storytelling also has the potential to "destabilize" and "transform official orthodoxies". This potential is, I suggest, of overarching importance if lay knowledge is to be properly valued and lay persons are to have a meaningful part in public dialogue about the new genetics (Kerr, Cunningham-Burley, & Amos, 1998).
CHAPTER II

‘THAT DISORDER’ AND ITS’ NAMING

We suddenly came upon two women, mother and daughter, both tall, thin, almost cadaverous, both bowing, twisting, grimacing. I stared in wonderment, almost fear. What could it mean? (George Huntington cited in DeJong, 1953)

At the age of eight, George Sumner Huntington (1850–1916) had his first encounter with ‘that disorder’. The young George was accompanying his father on his medical rounds in East Hampton, New York. Noting that his first encounter with ‘that disorder’ made “a most enduring impression” upon his “boyish mind”, he later recalled that it was “the very first impulse” to “choosing chorea as my virgin contribution to medical lore” (DeJong, 1953).

At age twenty-one, Dr. George Huntington presented his paper On Chorea to the Meigs and Mason Academy of Medicine at Middleport, Ohio. In this paper, Huntington described the form of chorea that now bears his name (DeJong, 1953). Elucidation of the characteristic features of the hereditary form of chorea had for some time been confounded by a confusing array of nomenclature.

Fortuitously, George Huntington belonged to a family in which there were three generations of physicians who practiced in the same community. His ability to correctly elucidate the hereditary nature of the disease thus derived, at least in part, from intergenerational observation. So succinct was Huntington’s description of the hereditary chorea, it prompted Sir William Osler to pronounce, some years later, that “In the whole range of descriptive nosology there is not, to my knowledge, an instance in which a disease has been so accurately and fully delineated in so few words” (Durbach & Hayden, 1993). This was not an honour which Huntington sought: indeed, Huntington eschewed any aspirations toward a career in research and devoted himself to the demands of rural practice and the attractions of country life.

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1 Chorea is the term which has long been used to denote the presence of involuntary movement of the limbs and torso. The word chorea derives from the Latin choreus and the Greek choros, the former alluding to dancing and the latter meaning chorus (Hayden, 1981).
Purpose and Outline of Chapter

Neurological disorders have often been named after the physicians who first described them. Taking the naming of 'that disorder' as a motif for this chapter, I review the social history of Huntington Disease (HD), including its emergence as a distinctive disease entity, its etiology, clinical manifestations, and hereditary nature. As we shall see, the stigma associated with the disease has shaped the process of naming, and naming, in turn, continues to shape the way in which stigma is both felt and enacted.

Placing Huntington’s description within its social and historical context, I then turn to a consideration of how the early twentieth century discovery of Mendelian genetics was taken up by a flourishing eugenics movement and how, in the post World War II years, scientific genetics sought to reconstruct itself. This review of the social history of eugenics and the rise of scientific genetics provides an essential backdrop for understanding the social and ethical implications of the new genetics as well as the “non-directive” stance of much of genetic counselling. Moreover, the themes of social labelling and discrimination continue to resound within contemporary debates about the probable consequences of identifying human differences with ‘spelling mistakes’ found through mapping and sequencing the human genome. Though many scientific and other published articles on genetic testing and screening rightfully acknowledge the importance of autonomy in decision-making, the lessons of eugenics are cast as a kind of “pre-history” (Zerubavel, 1996). In consequence, there seems, to me, to be a distinctly asociological perception that bioethics and an emphasis on individual choice, in particular, alleviate the need to worry about larger social consequences.

The final section of the chapter then returns to the status of HD as a problem and paradigm. From the mid-twentieth century on, molecular biologists made numerous advances in understanding the complex patterns of human heredity. Breakthroughs in unraveling the genetic

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2 For instance, Parkinson’s disease was named after James Parkinson, the British physician who, in 1817, first reported on the physical tremor. And, Alzheimer’s disease is named after the German physician Alois Alzheimer who, in 1907, presented his discovery of protein deposits in the brain of a 51 year old woman suffering from dementia (Davies, 1991).

3 With the increasing awareness that some individuals who have Huntington Disease experience rigidity rather than choreoform movements, the term “chorea” was dropped and the disease is now widely referred to as Huntington (or Huntington’s) Disease. In this chapter, however, I continue to refer to Huntington’s chorea wherever it is historically appropriate.
basis for HD have, however, only occurred within the last decade and a half. As we shall see, families affected by HD played a major role in bringing this about through organizing the Committee to Combat Huntington’s Disease as well as mobilizing resources and generating interest in the disease amongst scientific researchers.

The Origins of Huntington’s Chorea

The social history of the hereditary form of chorea described by George Huntington long predates the disease we now know as Huntington Disease. Medical historians differ on the precise origins of the hereditary form of chorea but genealogical investigations have helped to establish that it must have been present long before George Huntington’s illustrious description in 1872. In the preceding centuries, however, there was little differentiation between various causes of the dance-like movements now known as chorea.

In the first monograph ever devoted completely to the subject of Huntington’s Chorea, Hayden (1981) provides a succinct review of the history of chorea as a whole. The papyri of Ancient Egypt correctly ascribed the loss of voluntary movement to brain dysfunction, but it was not until some 4500 years later that Paracelsus (1493–1541) made a “momentous conceptual leap” and recognized that “excessive movements could also be due to underlying diseases of the central nervous system” (Hayden, 1981: 1).

Prior to Paracelsus, the earliest recorded instances of involuntary movements referred to a strange “epidemic” known as “the dancing mania”. Originating in Germany in the late fourteenth century, the “dancing mania” drew large crowds of men and women who formed circles and hand in hand “danced around in wild delirium for hours and hours, quite oblivious to the jeers and taunts of the onlookers” (Major, 1954:346). This phenomenon was, in Germany, referred to as St. Johannes (John’s) chorea in recognition of St. John the Baptist, patron saint of epilepsy and other movement disorders (Hayden, 1981). Clergymen attempted to treat the malady with exorcism and many of the afflicted were sent to the Chapels of St. Vitus’ where priests apparently had some success in curing “the strange affliction” (Major, 1954). Perhaps for this reason, the malady also became known as Chorea St. Viti (or St. Vitus’ Dance).

4 In Italy the disorder became known as tarantism since it was supposedly caused by the bite of the tarantula spider.
Paracelsus rejected the view that the dancing mania was attributable to "ghostly beings and spirits". Investigating closely the abnormal movements, he discerned three types of chorea: the first of these, as in the dancing mania, was thought to arise from the imagination; the second from "sensual desires", and; the third from an organic basis. It is possible that some of those afflicted with this latter type may have had Huntington's chorea. As Hayden (1981:3) notes, however, superstitions about the involuntary movements of the dancing mania "did not die with Paracelsus" but persisted throughout the centuries that followed: "there is, to this day, an annual procession of dancers in Meulenbeeck [Germany] on St. John's Day." Celebrating the day through various rituals such as leaping though a fire or dancing across a particular bridge, participants seek protection from serious illness in the coming year (Hayden, 1981).

The contemporary observance of such rituals is suggestive of the perseverance of a collective social memory of the dancing mania, preserved in oral tradition and handed down from one generation to the next. That this distant memory may, to this day, be kept alive in the stories of those who are at risk for Huntington Disease (HD), was a possibility I first encountered while doing an interview with a woman who described, in some detail, various attempts to locate missing pieces of her own family history.

So she [my mother] went back to England in 1947 and then again in about 1949 or 1950, desperately searching for information about her father's family as well as her biological mother's family. And at that time people said that her father had a sister that was crazy, that would dance through the fields or, you know, was described as crazy. So we've all figured out that she must have had Huntington's too. And she actually had a child but we've never been able to trace/to find that. I actually went to that area when I went to England too. But there's just nothing left. There are no records or people left because this is, of course, 40 years later. (F, 50% risk, PT candidate, 54 years, married, 2 children)

The image of a 'crazy' woman dancing through the fields has a vividness which depicts the corporeality of 'that disorder' in an almost timeless way yet, it also carries with it an air of superstition which Paracelsus and many others have since tried to dispel. Seeking a rational explanation for the dancing mania, historians are now largely agreed that the phenomenon Paracelsus referred to was "probably a form hysteria, mass hysteria in a literal sense," a kind of social reaction to the protracted misery and hopelessness of the Black Death (Major, 1954:348).

It was not until 1686 that the English physician, Thomas Sydenham described a childhood form of chorea which later became known as 'Sydenham's chorea' or 'chorea minor.'
This differed from the ‘chorea major’ of the dancing mania but Sydenham’s erroneous adoption of the term ‘St. Vitus’ dance’ perpetuated the “existing nosological confusion” (Hayden, 1981) and tolerance for the error persists to this day.5

Nosological clarity and the dispelling of fear and superstition, could not, it seems, come soon enough. In the late seventeenth century, the fear of witchcraft culminated in the Salem witch trials. Although the evidence remains in dispute (Hayden, 1983), several historians and genealogists argue that some of those who were persecuted as witches in both England and America may have been afflicted with the hereditary form of chorea which George Huntington later described. One source of support for this hypothesis derives from a letter written by the Rev. Samuel Willard, Minister of Groton, Connecticut. The letter portrays the “bodily motions, leapings, strange agitations” of one of the grand-daughters of an early settler who may have been affected with Huntington’s chorea (Hayden, 1981:18).

Popular belief held that persons so afflicted had renounced God and, as such, their facial grimacing and contorted movements were said to represent the suffering of Christ during crucifixion. Death was the recommended punishment for such renunciation and, in 1642, the first anti-witchcraft laws were passed in New England. Culminating in the Salem witch-hunt of 1692, these laws resulted in thirty or more executions in New England during the seventeenth century (Hayden, 1981). According to several accounts, there were at least seven female “witches” who were, in all likelihood, afflicted with the hereditary form of chorea. All were believed to be descendants of three male immigrants to the United States. Apparently fleeing the climate of oppression and persecution in sixteenth century England, all three men had emigrated from the village of Bures to New England. Coincidentally, a man named Simon Huntington also set sail, in 1633, from the nearby Norwich area and, as such, “both the earliest transmitters of Huntington’s chorea and the progenitor of the man who was to describe the disease left Great Britain for the United States” within the same three year period and from an area spanning only 50 miles (Hayden, 1981:17).

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5 For instance, a recent novel about Huntington Disease (Rubalcaba, 1996) is entitled Saint Vitus’ Dance. This book was written to help children understand and cope with hereditary illness.

6 Simon Huntington eventually settled in Connecticut where he helped to found the new settlement of Norwich.
Huntington’s Chorea

George Sumner Huntington’s ancestors settled in East Hampton, Long Island and both his father and his grandfather practiced medicine in the area. Each had taken an interest in the hereditary form of chorea and observed several generations of families afflicted with the disorder (Brody, 1967). Reflecting on his first exposure to the disorder, George Huntington recalled that his interest in the disease “never wholly ceased” (Huntington, 1967 [1872]).

George Huntington completed his medical training at Columbia University in 1871. Upon return to East Hampton he assisted his father with the family practice and continued his observations of the hereditary chorea. Soon completing the paper On Chorea, Huntington (1967 [1872]) prefaced his description of the hereditary form of chorea with an astute observation of its social stigma.

It is spoken of by those in whose veins the seeds of the disease are known to exist, with a kind of horror, and not at all alluded to except through dire necessity, when it is mentioned as ‘that disorder.’

Huntington’s description of the key features of hereditary chorea has a lasting relevance. Indeed, recent medical texts on Huntington Disease (Folstein, 1989; Harper, 1991; Hayden, 1981) and popular books on the genetic revolution (Bishop & Waldholz, 1990; Shapiro, 1991) continue to make extensive reference to Huntington’s original paper. Further, several of the precise phrases which Huntington employed in his description of the disorder appear regularly in lay as well as professional discussions of the disorder and its genetic transmission. The text of On Chorea is, therefore, worth examining in some detail.

Huntington described how those afflicted with ‘that disorder’ displayed all of the symptoms of common chorea but to an aggravated degree. In some, he noted, the chorea may progress to the stage where those afflicted can barely walk and “would be thought by a stranger to be intoxicated.” In contrast with other types of chorea (such as Sydenham’s chorea), however, there were three “marked peculiarities” to the type which Huntington observed: “1) Its hereditary nature. 2) A tendency to insanity and suicide. 3) Its manifesting itself as a grave disease only in adult life.”

Mendel’s laws of inheritance were not, in 1872, a part of the biomedical lexicon, yet Huntington (1967[1872]) accurately described what would soon be recognized as a classic
pattern of Mendelian autosomal dominance.

Where either or both the parents have shown manifestations of the disease...one or more of the offspring almost invariably suffer from the disease if they live to adult age. But if by any chance these children go through life without it, the thread is broken and the grandchildren and great-grandchildren of the original shakers may rest assured that they are free from the disease....Unstable and whimsical as the disease may be in other respects, in this it is firm, it never skips a generation to again manifest itself in another; once having yielded its claims, it never regains them.\(^7\)

The observable patterns of transmission in hereditary chorea revealed that it was only the offspring of an affected individual who could ultimately manifest the disorder. Further, if one or more of the children of an affected individual went through life without developing the disorder, the “thread [was] broken” and all subsequent generations would be spared from the disorder.

On the tendency to insanity and suicide, Huntington noted several instances of suicide which occurred in those afflicted with the disease or those who were from families in which the disease was known to exist. In addition, he described the considerable variation between the progress of the disease in different individuals: “as the disease progresses the mind becomes more or less impaired, in many amounting to insanity, while in others mind and body both gradually fail until death relieves them of their sufferings.” In men, the disease also appeared to manifest itself in the form of licentious behaviour. Ever the astute observer of social and as well as biomedical pathology, Huntington duly noted that there were at least two married men afflicted with HD who could not refrain from “constantly making love to some young lady”. Nor, it seems, were they “aware that there is any impropriety in it”; the effect was “ridiculous”.

According to Huntington, onset of the hereditary form of chorea occurred only in adult life. Noting that he had never seen a single case of anyone showing marked signs of chorea before age thirty, Huntington also stressed that “those who pass their fortieth year without symptoms are seldom attacked.” This remains true for the majority of people since the average age of onset is estimated to occur at thirty-six to forty-five years of age but, it has since been shown that onset may occur much earlier, as in juvenile onset HD, or much later in life (Hayden, 1981). With onset, however, the progression of the disease was, in Huntington’s observation,

\(^7\) This passage and, in particular, the phrase “it never skips a generation to again manifest itself in another” is, as I discuss in Chapter III, extremely significant in understanding how scientific facts become distorted in everyday patterns of clinical and lay communication.
gradual but unstoppable and though it "often [occupied] years in its development" the hapless sufferer eventually became a "but a quivering wreck of his former self."

No treatment was, in Huntington’s time, of any avail and indeed, the ending was, he thought, “so well–known to the sufferer and his friends that medical advice is seldom sought.” That Huntington’s chorea appeared to be “one of the incurables” may explain Huntington’s conclusion to his contribution to “medical lore” (Huntington, 1967 [1872]).

I have drawn your attention to this form of chorea gentlemen, not that I considered it of any great practical importance to you, but merely as a medical curiosity, and as such it may have some interest.

Huntington was, perhaps, too modest in assessing his contribution. The hereditary aspect of the disorder had been described earlier in the nineteenth century by other investigators but, as Hayden (1981:7) suggests, “in spite of these earlier descriptions the inherited form of chronic chorea was not generally recognized until after Huntington’s contribution.”

There are several compatible lines of explanation for the timing of George Huntington’s contribution and the subsequent recognition it received. First, up until the mid–nineteenth century few physicians would have lived long enough to observe firsthand the characteristic features of ‘that disorder’ as they occurred throughout multiple generations of the same family (Folstein, 1989). As Huntington noted in an address to the New York Neurological Society in 1909, he could not have formulated a correct understanding of the salient features of the disease had it not been for the prolonged observations of his father and grandfather before him (Brody, 1967). Second, Huntington had the good fortune to have his paper immediately abstracted in German and this resulted in his name becoming associated widely with the disease in many parts of Europe (Hayden, 1981). 8 In addition, it is interesting to note that an enhanced awareness of the disorder in the U.S. resulted, for some time, in more frequent diagnosis of affected persons and a mistaken belief that the disorder was primarily an American tragedy (Hayden, 1981). Third, the timing of George Huntington’s paper was ideal. Pre–Mendelian ideas about heredity, which predominated for most of the nineteenth century, were too poorly developed to acknowledge the significance of the pattern of inheritance which characterized this

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8 The eponym Huntington’s chorea was first used by Huber in 1887 and in the subsequent years 1885-1895, the eponym was adopted by authors in France, Italy, England and the United States (Hayden, 1981:10).
disease (Folstein, 1989). Huntington's description, however, came at a time when the medical and scientific world was poised to have a new appreciation for the concept of heredity.

**Mendelian Inheritance and the Eugenics Movement**

Turn of the century understandings of human genetics gave rise to two interwoven but increasingly oppositional developments. On one hand, basic genetic research began to identify the complexity of inheritance and the involvement of multiple genes as well as environmental factors in producing many observable traits. On the other hand, a flourishing eugenic movement seized upon Mendel's basic laws of inheritance and feverishly applied a sort of one size fits all approach to explaining and promoting the eradication of a host of what were considered socially undesirable traits.

Rescued from near obscurity at the beginning of the twentieth century, Mendel's laws of inheritance had enjoyed only nascent acceptance in the latter part of the nineteenth century. This was, in part, due to Mendel's own shyness and reluctance to tout his studies of patterns of inheritance in garden variety peas (Serafina, 1993). Seeking to explain the persistent 3:1 ratio of tall to dwarf sized and wrinkled to smooth pea plants, Mendel described what is now a famous tenet of genetics: some traits are *dominant* while others are *recessive*.

Mendel knew little of genes or chromosomes but he did advance the idea that genetic characteristics are transmitted from one generation to the next in the form of paired "particles". As such, there were three possibilities for any given offspring: two recessive characteristics (aa), one dominant and one recessive characteristic (Aa) or two dominant characteristics (AA).

Mendel presented his findings at a meeting of the Brunn Natural History Society in 1865, but it was not until 1901, when Mendel's paper appeared in the *Proceedings of the Royal Horticultural Society* that other scientists, such as William Bateson and Hugo De Vries, began to become enthralled by his ideas (Serafina, 1993).

In the early twentieth century, a growing interest in Mendelian patterns of inheritance helped to spawn a series of classic genealogical investigations. With its distinctive characteristics, long history and low mutation rate, Huntington's chorea provided an especially fertile basis for genealogical inquiry (Hayden, 1981). If, however, such genealogical
investigations ultimately contributed much to the scientific understanding of Mendelian inheritance in general (as well as to Huntington’s chorea in particular), these investigations also became entangled in the early twentieth century eugenics movement.

Despite what can only be described as a limited appreciation for the emerging complexity of human heredity, American eugenicists such as Charles Davenport and Henry Goddard, promoted “racial betterment” through negative eugenics which stressed the importance of preventing reproduction of the “unfit” and positive eugenics which encouraged the reproduction of the “fit”. The doctrine of Mendelian inheritance was an important plank in the eugenicist platform. Nowhere was this more apparent that in the eugenicists’ fervent belief in the threat of unchecked “feeble–mindedness”. Francis Galton, who initially coined the term eugenics in the late nineteenth century, had studied mental traits in Britain but had been unable to pinpoint the role of heredity in explaining his controversial findings. Mendelian analysis, however, seemed to cement the case for heredity as a plethora of family studies appeared to demonstrate a predictable ratio of 3 “normals” to 1 “defective” — exactly the ratio expected if mental defect resulted from a single recessive gene.

Chief among those who latched onto Mendelian inheritance as an explanation for ‘feeble–mindedness’, Henry Goddard argued not only that the ‘feeble–minded’ were a form of undeveloped humanity displaying “a low intellect but strong physique,” they also lacked “one or the other of the factors essential to moral life — an understanding of right and wrong, and the power of control” (Kevles, 1985:78). Davenport, on the other hand, recognized that ‘feeble–mindedness was a social rather than biological term. Goddard was, nonetheless, able to convince many geneticists that much anti–social behaviour was attributable to hereditary factors.

The dubious “results” of many family studies of feeble–mindedness and other heritable “defects” were exhibited at many U.S. state fairs and expositions and publicized in high school and college textbooks, popular books and magazines. Women constituted a large proportion of the audience for these displays. As Kevles (1985:90) notes, eugenics was “concerned ipso facto with the health and quality of offspring” and, as such, it “focused on issues that, by virtue of biology and prevailing middle–class standards, were naturally women’s own”.

Eugenics’ greatest enthusiasts were, in Britain and the U.S., primarily middle or upper
middle-class, white, Anglo-Saxon, predominantly Protestant and well-educated. Canada, which
did not escape unscathed from popular efforts to promote racial betterment, also had its
enthusiastic supporters of immigration restriction, mandatory sterilization and other eugenic
social policies (McLaren, 1990). The political persuasions of eugenicists differed widely;
nonetheless, conservatives such as Davenport mingled with radicals such as Emma Goldman or
Hermann J. Muller to espouse the view that poverty, crime, licentiousness and mental
retardation were the products of inferior breeding. The idea that it was the social system which
created these problems was, to those convinced by hereditary explanations, antithetical to
progressive social change.

For Davenport (cited in Kevles, 1985: 48–55) and many of his followers, the idea that
heritable traits were “private and personal matters” which ought not to enter into enlightened
social policy was “a narrow and false view” which must be set aside in the service of the
common “protoplasmic good”. Davenport had a long-standing interest in Mendelian patterns of
inheritance but unlike others, such as Sir Francis Galton and the population biologist Karl
Pearson, he was anxious to gather data about the individual genotype as well as the observable
phenotype. Noting the unsystematic character of data concerning the familial incidence of
various diseases in the medical journals, Davenport undertook one of the first and most
ambitious general surveys of inheritable disorders. Preparing a “Family Records” form which he
distributed to hundreds of individuals, scientists and medical, mental and educational
institutions, Davenport amassed a vast amount of information about heritable disorders as
observed in at least three generations of American families.

Funded generously, Davenport went on to form the Eugenics Record Office at Cold
Spring Harbour on Long Island and, between 1911 and 1924, he carried out an extensive project
which entailed house-to-house surveys and scrutiny of many American prisons, hospitals and
institutes for the mentally deficient, deaf, blind and insane. In order to conduct this large scale
“inventory of the blood of the community” — as Scientific American later called it (Kevles,
1985) — Davenport enlisted a contingent of more than 250 field workers, who were for the
most part women’s college graduates with some training in biology. Armed with a summer
course in human heredity and field methods as well as a “Trait Book” for guidance, this cadre of
trainees was sent off to study the incidence of juvenile delinquency, albinism and many other traits which Davenport suspected were heritable in the classic Mendelian fashion.

Published in 1916, Davenport and Muncy's study of the incidence of Huntington's chorea in the New England area described 962 persons affected with the disorder. Many of these individuals derived from four distinct families who had immigrated from Europe to the Boston and Salem areas in the early seventeenth century. Pleading for drastic measures to halt the proliferation of this "dire disease", Davenport and Muncy suggested that sterilization of "all those in which chronic chorea had already developed [was] a policy of far-seeing philanthropy". It was, in their view, the state's responsibility to "concern itself with all of the progeny of such" (Davenport & Muncey, 1916).

Davenport was, nonetheless, critical of the use of outdated scientific ideas in bolstering the argument for widespread sterilization of the feeble-minded and favoured instead the eugenic segregation of mental defectives. All the same it was, in the U.S., legal challenges rather than new scientific findings which curbed the rate of sterilization. Hence, while there were some operations performed on the insane, it was not until the famous 1927 Buck v. Bell case upheld the Virginia statute that eugenic sterilization came into wide practice in the U.S. In the notorious words of Justice Oliver Wendell Holmes (cited in Paul, 1995:83),

> It is better for all the world if, instead of waiting to execute degenerate offspring for their crime, or to let them starve for their imbecility, society can prevent those who are manifestly unfit from continuing their kind...Three generations of imbeciles are enough.

Following this case, twelve states enacted new statutes or amended existing ones to withstand constitutional challenge and thirty states eventually adopted sterilization laws. The precise number of those afflicted with HD that underwent mandatory sterilization is, in so far as I have been able to determine, unknown but, given the disorder's frequent misdiagnosis and

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9 One of the most fundamental shifts in scientific opinion emerged with the Hardy–Weinberg Law. Formulated in 1908 by the English mathematician G. H. Hardy and the German physician Wilhelm Weinberg, this mathematical statement expresses the relationship between the frequency of a particular allele and the frequency of genotypes involving that allele in a population. Assuming random mating, this law demonstrated that the frequency of a particular allele will remain constant in upcoming generations (Serafina, 1993). Mathematicians working at Cambridge used this law to demonstrate that even if all of the feeble-minded could be prevented from breeding it would take thousands of years to reduce the prevalence of "feeble-mindedness" from 3 in 1,000 to 1 in 100,000. (Paul & Spencer, 1995:302)

10 Between 1913 and 1918, seven out of twelve states which had enacted sterilization legislation were challenged constitutionally and all of these challenges succeeded (Paul, 1995).
propensity to lead to progressive "insanity" it is probable that there was a significant number amongst the estimated 60,000 individuals legally sterilized in the U.S. (Paul, 1995:83).

In Canada, mandatory sterilization laws were not enacted until 1933 but in that year British Columbia followed Alberta's lead and adopted legislation which sanctioned the sterilization of the mentally ill and retarded. This legislation was amended in Alberta in 1942 in order to permit sterilization of those with Huntington Disease and epilepsy (Cairney, 1996). Manitoba and Ontario both rejected such legislation but as McLaren (1990:91) argues, "eugenically based racial concerns were all-pervasive in interwar Canadian society and the most extreme policies tended to be advanced, not by conservatives, but by progressives and medical scientists." Indeed, it was often respected members of the medical profession who felt most compelled to respond to "a dangerous surge in the numbers of the mentally deficient", a "spectre that the professionals themselves had conjured up" (McLaren, 1990:91). Progressive women of the day did, however, also play a pivotal role in promoting legislation permitting sterilization of "degenerates" and the "congenitally diseased." As the president of the United Farm Women (the women's wing of the United Farmers of Alberta) stated in response to civil libertarian concerns raised during her 1924 address, "democracy was never intended for degenerates" (Cunn, cited in Cairney, 1996:791). Elsewhere, social activists such as Nellie McClung, Louise McKinney and Emily Murphy actively supported the eugenics campaign. Murphy, in particular, stressed that young Canadians had a moral obligation to investigate the medical history of the family they intended to marry into (Cairney, 1996).

In Germany, the eugenics movement gathered momentum with the initiation of a campaign for racial hygiene in 1933. Issued only two months after the Nazis came to power, the Law for the Prevention of Genetically Diseased Progeny extended greatly the range of what was considered a heritable condition and required doctors to register all cases of hereditary disease. The law also endorsed compulsory sterilization, whether or not the person was institutionalized.

11 Sterilization legislation was not removed from the statute books of Alberta and British Columbia until 1972 (McLaren, 1990:169).
12 It was found in 1978 that Ontario had conducted hundreds of sterilizations each year despite the absence of any legislation. The provincial government halted this practice but many doctors continued to profess a belief in the appropriateness of using medical procedures to remedy social problems (McLaren, 1990:169).
“in cases of congenital feeble-mindedness, schizophrenia, manic-depression, severe physical
deformity, hereditary epilepsy, Huntington’s chorea, hereditary blindness and deafness, and
severe alcoholism” (Paul, 1995:86, emphasis added). The Nazi regime sterilized an estimated
320,000 to 400,000 in the first four years of the law but, by 1939, the program ground to a halt.
Sterilization was, however, only the first step as Hitler’s campaign for racial hygiene reached its
most heinous apex in the extermination of millions of Jews, Gypsies, Slavs, homosexuals and
other persons of “inferior blood” or “defective character”.

Though geneticists had often found themselves at odds with the extremes of eugenic
rhetoric in the interwar years, it is important to remain cognizant of the fact that many leading
scientists from around the world were initially supportive of the Nazi sterilization law. Nazi
scientists often quoted American geneticists and, in particular, invoked a published account of
California’s large scale application of mandatory sterilization (Paul, 1995). Eugenics was also
popular in the Scandinavian countries and, although the Danes ultimately adopted a very
different approach to solving their “Gypsy problem”14, they too produced a registry which
documented the genealogy of those widely considered to be a social burden (Paul, 1995).

For reasons which are perhaps too obvious to recount, eugenics fell into disrepute as the
world awoke to the atrocities of Nazi Germany. If a nascent disfavour with eugenics in the pre­
war years had begun to erode support for restrictive policies of immigration and mandatory
sterilization, the revelations of the post-war years discredited even more completely the
foundering science of eugenics. This is not, however, to suggest that scientific research on
heritable disorders or the application of eugenic thinking came to a grinding halt. In the post­
war years, scientific genetics forged a new identity and, in conjunction with the emerging
profession of genetic counselling, developed a less polemical and more rigorously scientific
basis for new biomedical approaches to understanding heritable disease.

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13 The law was intended to be race-neutral and was applied to both women and men on a roughly equal basis.
‘Feeble-mindedness’ was the most common grounds for sterilization (up to 75% of all sterilizations) followed by
schizophrenia, epilepsy and alcoholism. A table from the 1937 edition of Erbpathologie: Ein Lehrbuch für Ärzte
und Medizinstudierende shows the breakdown for three studies of the grounds given for sterilization in the
adjudication of the 1933 Sterilization Law. In these studies Huntington’s chorea accounted for .2, 1.5 and .3% of
sterilizations amongst the respective samples of 6,052 men, 6,032 women and 325 men and women (Proctor,

14 Denmark favoured the integration of the Gypsy population into Danish society and thus, similar science was, in
two very different contexts, used to justify opposing social policies (Paul, 1995:90).
Scientific Genetics and the Rise of Genetic Counselling

As noted above, there were a number of geneticists who had publicly supported the mandatory sterilization and restrictive immigration policies of the eugenics movement. In the mid to late 1930’s and during the aftermath of World War II, many more began to denounce the flagrant misuse of genetic science. The eugenic movement had, especially at its close, become intellectually static and increasingly associated with explicitly racist policies and ideas. This reflected badly on scientific genetics and continued to overshadow emergent understandings of the complex role of human heredity in a variety of medical disorders. As such, the public association between eugenics and genetic science had to be severed.

The Evolution of Scientific Genetics

Lamenting the status of human genetic research in the war and immediate post-war years, one American geneticist recalled, in a 1982 interview with historian Daniel Kevles, that existing systems of classification for human traits were confused to say the least. Further, it would be more productive for geneticists to work with experimental organisms since “the only thing you can do with human genetics is develop prejudice. And anyone who went into human genetics was immediately classified as a person of prejudice” (Kevles, 1992:11).

In Britain, eugenic ideas were, from the mid 1930’s on, somewhat tempered by a group of human geneticists which included J. B. S. Haldane, Lancelot Hogben, and American Hermann J. Muller. Finding an ally in anti-eugenicist Lionel Penrose, these “reform eugenicists” sought to free prejudicial eugenics from its racial and class bias by developing a sound science of genetics which might be employed in preventive or therapeutic medicine (Kevles, 1992). Focusing in particular on the utility of linkage analysis15 for the identification of single gene recessive disorders in humans, a good deal of effort was expended in attempting to link blood groups with known genetic disorders. These efforts paid off for Julia Bell and J. B. S. Haldane when, in 1937, the authors (Bell & Haldane, 1937:119) reported on their pioneering work in locating a genetic link between colour blindness and haemophilia. Offering the first

15 Linkage analysis is a technique which was pioneered by fruit-fly geneticists who observed that some traits which occur in a variety of forms (e.g., eye colour or wing type) tended to be inherited together. Thus, their respective genes were likely to lie on the same chromosome and were said to be linked. If these genes were close together they would be jointly inherited with a high frequency.
concrete proof of linkage in humans, Bell and Haldane (1937: 119) speculated about the potential application of their work in developing a predictive genetic test for inherited disorders such as Huntington Disease.

The present case has no prognostic application, since haemophilia can be detected before colour blindness. If however to take a possible example, an equally close linkage were found between the genes determining blood group membership and that determining Huntington’s chorea, we should be able, in many cases to predict which children of an affected person would develop this disease, and advise on the desirability or otherwise of their marriage.

It is noteworthy, although perhaps not surprising, that the authors did not see fit to offer — and nor did the existing climate demand — any comment on the psychosocial implications of such a test. Only a scant four years earlier Lancelot Hogben (1933:21) had stated that dominant hereditary conditions such as Huntington’s chorea “could be eliminated in a generation if individuals suffering from them were not allowed to reproduce. When diseases of this class are incurable, this is the only effective method of prevention known at present.”

Somewhat more circumspect in his appraisal of discoveries in human genetics, Lionel Penrose (1959:127) prefigured much subsequent debate when he quietly concluded his Outline of Human Genetics with the words, “some people would rather not know about the unfavourable genes that they carry.” Head of the prestigious Galton Laboratory of National Eugenics at University College in London since 1945, Penrose was an anti-eugenicist and ardent critic of “wasteful” and “absurd” efforts to sterilize carriers of recessive disorders. Wary that the progress made by science “involves dangers for human happiness as well as potential benefits”, Penrose (1959:127) recognized the importance of severing human genetic research from its eugenic applications. This he attempted in several ways.16 Most centrally, Penrose was instrumental in refocusing medical genetics on the study of hereditary phenomena which could be quantitatively and objectively observed. As such, the Galton laboratory drew scientists who, like Penrose, were keen to conduct credible research on human genetics (Kevles, 1992).

In the U.S., genetics was also gaining a new momentum. Ironically, it was however, the advent of the atomic bomb and the threat of radioactivity and genetic mutation which helped to

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16 In 1954, he succeeded in changing the name of the Galton laboratory journal from the Annals of Eugenics to the Annals of Human Genetics. After a somewhat longer battle, he also succeeded, in 1961, in changing his title to the Galton Professorship of Human Genetics.
spur a renewed interest in the science of genetics. During this period, genetic science derived significant impetus from a group of disillusioned physicists who moved into biology and brought with them new and mechanistic ways of thinking about information systems, coding and control (Hubbard, 1990). Some undoubtedly felt burdened by their involvement in the creation and deployment of the atomic bomb and, sought some redemption in the more life-enhancing work of biology; others sensed that biology presented a new frontier which could yield discoveries of unparalleled importance (Kaye, 1986).

Attracted to the emerging quest for “the secret of life”, the young scientists James Watson, Francis Crick and Maurice Wilkins were drawn to molecular biology. In 1953, Watson and Crick became widely acknowledged for the discovery that genes are comprised of a double-helical strand of deoxyribonucleic acid (DNA). The honour is, however, rightfully shared by X-ray crystallographer Rosalind Franklin (Sayre, 1975). Within a decade of this breakthrough, scientists recognized that the four bases — adenine and thymine or cytosine and guanine — of DNA constituted a sort of alphabet of the genetic code. Genes were, then, a unique sequence of letters which spelled out precise units of genetic information.

By the late 1950’s, molecular biologists making extensive use of this information analogy, argued that biological systems, like other informational systems, are organized by a chain of binary reactions which act in concert to control the development and homeostasis of living organisms. Formulated by James Watson and Francis Crick, the Central Dogma (or dominant theory) of this period of molecular biology was derived from the notion that DNA (deoxyribonucleic acid) defines the structure of RNA (ribonucleic acid), and RNA, in turn, defines the composition and structure of proteins. In short, when genes are “turned on”, the

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17 Much of the early work on genetic mutation was conducted by Curt Stern, a German refugee and leading fruit-fly geneticist who emigrated to the U.S. in the mid–1930’s. Working with the Manhattan Project, Stern conducted a major investigation into the biological effects of radiation and, in the aftermath of Hiroshima and Nagasaki, one of his students, Dr. James Neel, inaugurated a study of the genetic impact of substantial radiation exposure (Kevles, 1985:223–224). Subsequently taking up the study of human blood disorders, Neel sought solid scientific evidence of harmful genetic traits and, in 1948, demonstrated that sickle cell anemia was the result of a single recessive gene (Kevles, 1992).

18 This juxtaposition of the powers of physics and the as of yet untapped potential of molecular biology inspired metaphysical reflection in some migrant thinkers such Erwin Schrödinger. In his 1943 lectures series entitled “What is Life?”, Schrödinger expressed the hope that rigorous physiochemical analysis of the gene and its processes of replication would culminate in the discovery of “other laws of physics’ hitherto unknown”; paradoxically these “laws” would not undermine human free-will but rather would reveal the mystical oneness of all living things, an “omnipresent, all-comprehending eternal self” (Schrödinger, cited in Kaye, 1986: 65–68).
synthesis of RNA and proteins ensues (DNA → RNA → proteins).

During the 1960’s and 70’s, such advances in the understanding of human genetics also fueled a resurgence of interest in the biological origins of human social behaviour. In the U.S. in particular, E. O. Wilson and other proponents of sociobiology argued that all behaviours and social relationships are biologically, genetically and evolutionarily determined (Wilson, 1975). The sociobiology of Wilson and others elicited a storm of controversy and, for a time, the nature versus nurture debate occupied centre stage in many social science publications. Furthermore, biologists who rejected the misappropriation of an outdated model of genetics were also avid contributors to this debate. Rose, Lewontin and Kamin (1984) argued that not only is sociobiology “bad science” in the sense that it misapprehends the facts, it is also a “determinist ideology” which “preserve[s] the interests of the dominant class, gender, and race”.

By the close of the 1970’s, the often vociferous nature versus nurture debate established a tentative consensus that both biology and the environment — inclusively defined as the social, political, and economic as well as material context — have a vital if often unquantifiable influence in shaping human attributes and behaviours. During the last decade there has, however, been a marked increase in publications which again claim a genetic basis for, not only human health and illness, but also a range of human social and political behaviours.

While it is difficult to establish the impact that such claims are having on public perceptions of the role of genetics, it appears that there is now a very real danger that popular representations of the workings of DNA oversimplify and distort what is known about the complex interactions of genes and the environment. In particular, the information analogy which originated with the Central Dogma, has become outdated. Nonetheless, this way of thinking about the genome has filtered into public consciousness through journalistic and other popular accounts which often represent DNA as a set of instructions or “biochemical recipes”, “a human

19 Carried to an extreme in The Selfish Gene, Richard Dawkins (1976) asserted that an organism is merely the necessary vehicle for genes to reproduce themselves. Everything organisms do, he claims, they do out of self-interest because organisms are only “living manifestations of selfish genes” engaged in the process of replicating themselves.

20 Some such claims are, indeed, incredulous. For instance, in a 1991 paper presented to the American Political Science Association, Somit and Peterson claim that there is a human genetic tendency toward “hierarchy, obedience, dominance and subordination” and, as a result, democracy is an inherently fragile form of government. “Man is not born free, but, rather in chains ... evolution has probably produced, in homo sapiens, a species with genetic tendencies that, in effect, are biased against democracy” (The Vancouver Sun, August 31, 1991).
text” or the “blueprints for who we are” (Bishop and Waldholz, 1990). These ways of thinking about DNA have, as Nelkin and Lindee (1995) point out, now assumed a stature and prominence comparable to that of early twentieth century conceptualizations of the germplasm as the essence or “very soul” of the individual.

Whimsical or not, the idea that “Genes R Us” is ubiquitous in popular culture. As the cover of a 1995 issue of Life (see Figure 1, next page) proclaims, “Knowing Your Medical Family Tree Can Save Your Life.” A baby gazes at an old family portrait and the caption poses a question which ought to concern all parents. “You may have inherited “your mother’s smile” and “your father’s eyes but have you also inherited... DIABETES? CANCER? ALZHEIMER’S?” Clearly, the moral imperative to unravel and act upon genetic information looms much larger than Life. Heredity is not destiny as long as we accept the moral responsibility to research and manage the risk of familial disease. “Ignoring family illnesses”, on the other hand, “GUARANTEES their place in future generations” (Adato, 1995:63).

The Evolution of Clinical Genetics

The hereditary basis of many common diseases now occupies a prominent place in many lay as well as scientific discussions of human health and illness. In order to situate the increasingly significant role of genetics in medicine it is, however, necessary to briefly review the origin and development of clinical genetics.

From the 1930’s on, several advances in medicine stimulated physicians to become more receptive to genetic science and knowledge of human genetics (Schild & Black, 1984). First, the emergence of bacteriology and the beginnings of chemotherapy contributed to an increase in average life expectancy; as such, physicians began to see more complex forms of chronic illness, such as diabetes and heart disease, which seemed to have a hereditary component. Second, a new emphasis on the prevention of disease was compatible with giving increased importance to hereditary factors. Third, scientific genetics was compiling a set of tools which were to have enormous clinical utility in the prediction and diagnosis of hereditary disorders.

One of the first of these tools stemmed from the identification in the 1930’s of the “inborn error” phenylketonuria (PKU). Treatment for the disorder, in the form of a modified
SUMMER TRAVEL SPECIAL:

Knowing Your MEDICAL FAMILY TREE Can SAVE YOUR LIFE

Your mother’s smile, your father’s eyes...

But have you also inherited...

DIABETES? CANCER? ALZHEIMER’S?
diet, soon followed and made possible the prevention of the mental retardation which typically follows from untreated PKU. Knowledge of human blood groups and the discovery of the Rh Factor, in 1944, also served to enhance the growing clinical significance of genetic data (Schild & Black, 1984). Then, with the 1949 discovery of techniques for chromosomal staining and the 1956 finding that humans have 46 rather than 48 chromosomes\(^{21}\) (as had been thought previously), prenatal diagnosis of chromosomal abnormalities became possible.

The first genetic counselling clinic was set up in Copenhagen and the second soon followed, in 1948, at the Hospital for Sick Children in London, England (Krush & Evans, 1984). By 1955, there were thirteen such clinics and by 1980 there were over 600 genetic service units in the United States alone (Schild & Black, 1984).

Proposed in 1947 by Dr. Sheldon Reed, the term “genetic counselling” supplanted a variety of other terms such as “genetic hygiene” or “genetic advice”. In keeping with the view that genetic services ought to provide a nondirective\(^{22}\) approach to the provision of genetic information, counselling was generally restricted to explanations of the risk of recurrence or occurrence of a genetic disorder. As Schild (1984:15) notes, however, the term genetic counselling has not been free of problems especially as it moved from the academic world of research into the clinical world of medical genetics.

The issues of terminology and definition represent much more than mere semantic issues. Any definition of genetic counseling raises substantive questions about the basic aims and purposes of genetic counseling and who should provide such a service. As a result, definitions of genetic counseling have changed often since Reed (1974) initially coined the term and defined it as meaning “a kind of genetic social work done for the benefit of the whole family entirely without eugenic connotation.”

Prior to the advent of professional programs in genetic counselling, a variety of predominantly male medical professionals and scientists provided counselling about recurrence risks and disease management to families at risk for, or affected by, genetic disorders. When, in

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\(^{21}\) First observed by scientists in the late nineteenth century, chromosomes are the tiny sub-cellular thread-like structures which contain the complex set of instructions for how each cell in an organism will develop. With the addition of a special dye, chromosomes are visible under the microscope at particular stages of cell division and growth. Each chromosome is comprised of a long strand of deoxyribonucleic acid, or DNA.

\(^{22}\) The clinical psychologist Carl Rogers originally coined the term “nondirectiveness” in 1942 in order to describe his psychotherapeutic approach to refraining from giving advice or guidance to his clients. As he came to realize that his very presence in counseling had a directive effect, Rogers altered his approach to incorporate an “unconditional positive regard for the client as a self-actualizing person; this he called a “person-centred approach” (Andrews, et al., 1994:151-2).
1969, the first genetic counselling program was developed at Sarah Lawrence College in New York, the provision of genetic counselling shifted to become a predominantly female occupation. As Rapp (1988:144) argues, it seemed fitting that with the introduction of amniocentesis women should fill the role of "gatekeeper" between science and social work...epidemiology and empathy". Translating scientific probabilities into individual estimates of risk, the graduates of Sarah Lawrence began to fill a growing niche.23

With the widespread initiation of genetic services in the 1960's, efforts to screen for chromosomal anomalies (such as Downs Syndrome) as well as various hereditary diseases expanded rapidly. Newborn screening for PKU began in 1963 and carrier detection programs for autosomal recessive disorders such as sickle cell anemia24 and Tay–Sachs disease25 soon followed. In a pertinent warning, Fraser (cited in Krusch & Evans 1984:20) cautioned in 1963, Let us not oversell ourselves and let us protect...Human Genetics from jeopardy by unqualified people speaking irresponsibly in its name. It happened before and it could happen again.

But, happen again it did. Long regarded as a “public policy disaster”, American programs for mandatory sickle–cell screening invoked all of the prejudicial sentiments and discriminatory behaviours of the earlier eugenics movement (Duster, 1990:45). Beginning in the 1960's, sickle–cell anemia became symbolic of African–American efforts to improve existing standards of living and access to adequate health care. By 1970, the disease had acquired such status that President Nixon decreed that public funds must be directed toward reversing a long

23 McGill University offered the first, and for a long time, the only degree in genetic counselling in Canada. As of 1997, the University of British Columbia began to offer a similar 2 year Master’s level degree.

24 Sickle–cell trait is prevalent in populations which trace their origins to equatorial Africa and the Mediterranean area; in part this may be due to the fact that the trait confers resistance to malaria (Hubbard & Henefin, 1985). Recognized since 1910 as an inheritable disorder, sickle–cell anemia was discovered in 1950 to be one of a class of diseases known as hemoglobinopathies: the red blood cells of an afflicted individual are impaired in their ability to carry oxygen. When these impaired cells try to move oxygen quickly, they change from their normal shape into sickle–shaped cells which block small arteries. The resulting "crisis" deprives organs, joints or muscles of oxygen and creates bouts of mild to intense pain. There is no cure for sickle–cell anemia although blood transfusions sometimes help to alleviate acute cases (Hubbard & Henefin, 1985).

25 First observed clinically in 1881 by W. Tay, a British ophthalmologist, Tay–Sachs’s disease was recognized as an inheritable disorder a few years later by B. Sachs, a New York neurologist. It was not, however, until much later that any progress was made in working out the physiology of this disease. Originally known as infantile amaurotic idiocy, Tay–Sachs’s is a degenerative neurological disorder characterized by an excess of unmetabolizable fatty substances. Molecular biologists have now been able to isolate the presence of a defective enzyme in Tay–Sachs’s children but remain unable to halt progress of the disease. Normal at birth, Tay–Sachs’s children regress quickly within the first year — development slows and progressive paralysis, blindness and other central nervous system disturbances set in. Death usually results before the child reaches age four. The incidence of Tay–Sachs is much higher amongst Ashkenazi Jews than it is in the general population (Duster, 1990).
"record of neglect" on African-American health issues (Kunitz, 1974:208). Mass carrier screening was, however, the dismal consequence. As an increasingly entrenched state and medical bureaucracy sought to manage and contain a volatile political situation, genetic screening appeared to provide a convenient ruse for addressing in earnest a litany of concerns about the poor health, inadequate nutrition and living conditions of many African-Americans.

Widespread misunderstanding of the difference between the heterozygotic status of carriers and the homozygotic status of those afflicted with sickle-cell anemia also exacerbated the situation. Surveys conducted in the Pittsburgh area revealed that 20% of obstetricians and gynecologists "did not know that the gene is the basic unit of inheritance" or that sickle-cell anemia is the result of an inherited hemoglobin defect (Kenen & Schmidt, 1978:1116). Further, many carriers of sickle-cell trait were deprived of health and life insurance or discriminated against in employment opportunities (Draper, 1991) as misinformation about the prevalence and heritable nature of the disorder stigmatized the African-American population. It comes as little surprise then that many states which once required neonatal and carrier screening for sickle-cell trait yielded, in 1976, to increasing pressure from African-American physicians and community activists to eliminate mass screening for sickle-cell trait.

In contrast with the sickle-cell experience, screening programs for Tay-Sach's were, from the start, held up as a model for the achievement of community support and involvement in lowering the incidence of a severe and fatal disorder. Beginning in 1970, it was possible to determine Tay-Sach's carrier status through a simple blood test, and diagnose affected fetuses through prenatal screening. Responding to these developments, Jewish charitable organizations altered their commitments and devoted all fund-raising to mass screening, education and prevention of Tay-Sach's disease (Goodman & Goodman, 1982). With the full support of Jewish physicians and rabbis, Tay-Sach's task forces were formed in each community and fully educated about "the characteristics of Tay-Sach's, carrier rates, the meaning of being at risk for Tay-Sach's, and the justification for the program" (McQueen, 1976:129).

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26 For instance, Kunitz (1974) describes one example in which researchers interested in the links between sickle-cell trait and intelligence gathered their data from screening programs intended to benefit the individuals being tested.
One of the first programs of mass screening for Tay-Sach's was set up in the Baltimore-Washington D.C. area. This was followed by more than sixty other programs spanning at least five countries. Couples at risk received counselling and, in some instances, the rabbinate would intervene to advise against marriage if the union was to involve two known carriers.

The prevention of genetic disease through carrier and prenatal forms of screening was, during the early 1970's, widely supported by genetic counsellors although there was, by the late 1970's, mounting evidence that many considered it important to move away from an exclusive focus on prevention and help individuals cope with genetic problems by removing or lessening guilt and anxiety (Sorenson, Swazey, & Scotch, 1981). This shift toward addressing the psychosocial needs of individuals and families was also evident in the definition of genetic counselling adopted by the American Society of Human Genetics in 1975. This definition specified that genetic counselling is a "communication process which deals with the human problems associated with the occurrence or risk of recurrence of a genetic disorder in a family."

In addition to providing information about the disorder and its management, attention was to be given to assisting individuals in choosing the "course of action which seems appropriate to them in view of their risk and their family goals" (Schild & Black, 1984:15–16).

Protracted efforts to develop sound federal guidelines for regulating and supporting genetic screening programs also resulted in the 1976 enactment of the National Sickle Cell Anemia, Cooley's Anemia, Tay Sach's, and Genetic Disease Act. This act resulted in new funds for basic and applied research and it also provided for testing, counselling, education and information programs related to genetic disease. Measures to ensure confidentiality and the quality of genetic services were also included in the act (Schild & Black, 1984). As the history of mandatory sickle–cell screening so amply demonstrated, genetic screening held out many possibilities for new types of discrimination. Further, news of being a carrier for a genetic disorder could result in prolonged anxiety and an increasingly negative view of personal health. Referring to these lessons, Kenen and Schmidt (1978:1117) pointed out in 1978 that,

Modern technology may have introduced a new biological and social label — "carrier" — with yet unknown psychological and social consequences... the newly identified carriers of mutant alleles are, in a sense, both psychological and social pioneers.
Experience in offering carrier testing and prenatal diagnosis for recessive disorders such as sickle-cell and Tay-Sachs's has provided clinicians and genetic counsellors with the opportunity to routinize many aspects of service delivery. Nonetheless, because the focus is so often on reproductive decision-making, genetic counsellors have had to confront their personal and professional values. Many recognize that these values are not always the same as those of their clients but, on balance, most continue to emphasize the importance of offering a non-directive stance and providing clients with sufficient information to come to their own decision.

As genetic counsellors and others have argued, the language of genetics plays a key role in shaping the experience of disease as a social, as well as biomedical, phenomenon. Given that genetic counselling is, fundamentally, a process of communication and further, that much of the language of genetics has its origins in the language of eugenics, genetic counsellors and other service providers must choose their terminology with extreme care and sensitivity. Used indiscriminately, terms such as "defective" or "abnormal" have significant implications for clients who must confront a diagnosis in themselves or in their fetus or child. As such, many geneticists and counsellors have become increasingly self-conscious of their own language use and many have contributed to the growing effort to "destigmatize medical language" (Andrews, et al., 1994). Calls for the "eradication" of genetic disease or the "reduction of the burden of genetic disease" to improve the overall health of society do, however, continue to slip into the utilitarian discourse of cost-benefit analyses; accordingly programs for prenatal testing which result in less than 50% of parents choosing to terminate an affected fetus are considered, by some, to be a failure (Andrews, et al., 1994:153).

Public awareness is another major factor which impinges greatly on the experience of genetic risk and/or diagnosis. As the history of eugenics demonstrates, the social construction of particular genetic disorders may owe as much to predominant moral and political concerns as to biomedical and scientific knowledge. As Kunitz (1974:211) suggests,

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27 For instance, in a 1988 statement on the social and ethical concerns raised by the Human Genome Project the U.S. Congress Office of Technology stated that: "Human mating that proceeds without the use of genetic data about the risks of transmitting diseases will produce greater mortality and medical costs than if carriers of potentially deleterious genes are alerted to their status and encouraged to mate with noncarriers or to use artificial insemination or other reproductive strategies."
Many of us assume that diseases are primarily facts that exist apart from our awareness of them. While they may indeed exist as harmful conditions independent of our awareness of them, it is our changing awareness of what is a problem that has for too long been neglected.

**Huntington Disease as Problem and Paradigm**

In the 125 years since George Huntington described ‘that disorder’, the status of Huntington Disease has shifted from “medical curiosity” to model for understanding the impact of testing for late onset genetic diseases. How has this change in awareness occurred and with what consequences?

As the first adult onset dominantly inherited disorder for which there was a predictive test, Huntington Disease now has a high profile within the scientific and medical genetics communities. And, although public awareness of the disorder is not yet high, a dedicated corps of individuals and families as well as the various Huntington Disease associations, have worked hard to enhance public recognition of the disease, establish support services, and provide scientists with the funding and infrastructure necessary to basic research. Research–funding and public awareness have not, however, come easily. In contrast with other well–known diseases, Huntington Disease has long been considered a relatively rare disorder. At present it is estimated to occur with a frequency of 1 in 10,000 for persons of European ancestry. As such, there are in Canada an estimated 2,500 individuals affected with HD and, in the U.S., about 30,000. Nonetheless, the social stigma surrounding HD may have resulted in an underestimation of the prevalence of the disorder. As Hayden (1981:42) suggests, “the completeness of collection of data and thus the estimated frequency of Huntington’s chorea in any area is inversely proportional to the social stigma attached to it.” Further, there are, for every person affected with HD, approximately five to seven times as many more persons that are at directly at risk for the disease (Huntington Society of Canada, 1996).

Many of those at risk for HD have been highly active in mobilizing support for research and improved medical and family support services. Beginning in the 1960’s, families directly affected by Huntington Disease began to organize and act on their own behalf. When, in 1967, the American folk–singer and songwriter Woody Guthrie died from the disease, his ex–wife
Marjorie Guthrie founded the Committee to Combat Huntington's Disease (CCHD). As the CCHD gathered momentum, it became the Huntington's Disease Society of America (HDSA) — a national grassroots organization which successfully lobbied the U.S. Congress, developed educational campaigns for health professionals and the public and organized services for families. Soon after the CCHD got underway, Milton Wexler organized a California-based chapter. Motivated by his ex-wife's diagnosis in 1968, Wexler was deeply committed to basic research and hoped that, with sufficient support, basic scientists could be enticed to study the disease and ultimately, to find a cure (Bishop & Waldholz, 1990).

At this time, HD was of interest to only a small number of neurologists and geneticists. Although there had, since the mid 1950's, been a considerable increase in the number of publications on HD, there was an urgent need to integrate existing approaches to scientific and clinical studies. In the half-century since George Huntington died little had changed in the ability to offer treatment and no-one yet understood the biochemical pathology which resulted in the pattern of neuronal death associated with the disorder. Some of the neurologists who had effectively used L-Dopa to treat patients with Parkinson's Disease had observed that too much of this synthetic neurotransmitter could produce chorea-like movements in their patients. Applying what was known about the action of dopamine in the brain, these researchers then began to investigate the possibility that the chorea associated with HD derived from too much dopamine or an oversensitivity to dopamine in the region of the brain known as the basal ganglia (Wexler, 1995). Originally brought together by André Barbeau, a neurologist working on both Huntington's and Parkinson's Disease at the University of Montréal, a handful of these researchers gathered in Montréal, in 1967, under the auspices of the World Federation of Neurology (WFN). At this meeting, twelve researchers — the sum total of all those who were working on Huntington's at the time — presented their work to each other and ultimately decided to form an international research group. Also present at the founding of the WFN Subgroup was Marjorie Guthrie — who had just formed the CCHD — and Alice Pratt, who had

28 Marjorie Guthrie looked after her ex-husband throughout his illness. When he became permanently hospitalized, she visited him regularly. Refusing to accept that he was mentally incompetent just because he could no longer speak, she devised ways and means of continuing to communicate with him.
recently formed the Huntington’s Chorea Foundation in Texas in order to fund basic research on
the disease. Barbeau later recalled that it was the neurologists who had inspired Marjorie
Guthrie but, as Alice Wexler (1995:100) points out, “it may have been the other way around.”

Meeting informally in 1971 for a workshop on HD research, a small but growing number
of neurologists familiar with HD began to actively exchange ideas with basic scientists and
molecular biologists. Driven in part by Milton Wexler’s determination in organizing this and a
series of other workshops, scientists began to construct a variety of new hypotheses to test in the
lab. Presented with the compelling stories of spouses and siblings as well as those affected by
HD, many scientists and other workshop participants who had little or no clinical exposure to
HD became aware of the devastating impact of the disease. Speaking with those who had
firsthand experience of HD was, as Milton Wexler’s daughter Nancy Wexler later noted, a
powerful influence which “really makes me feel like turning myself inside out to cure this

Understanding firsthand what it meant to be at 50% risk for HD, Nancy Wexler29
became a prominent spokesperson for research on HD within the expanding scientific
community. In 1972, this community gathered in Ohio to assess existing research and
commemorate George Huntington’s original paper. With 146 researchers from fourteen
different countries, this meeting celebrated “an atmosphere of optimism which [had] never
before existed in the history of Huntington’s disease” (Wexler, 1995). Although little medical
progress had been made, there were many new ideas emanating from non–medical scientists and
geneticists new to the field of research on HD. One of those in attendance was a Venezuelan
psychiatrist, Dr. Ramón Ávila–Girón, who brought with him a film of several communities on
Lake Maracaibo where there were large numbers of individuals affected with HD. Because of
the unusually high prevalence of HD in these isolated communities, Ávila–Girón’s “startling”
film was to have significant implications for subsequent research (Wexler, 1995). Recalling the
impact of seeing the film, Nancy Wexler (cited in Revkin, 1993:102) later said, “It was a total
shock.... Here were all these Huntington’s cases, practically in every household, not shut away

29 Writing about the experience of living at risk for HD, Nancy Wexler completed her doctoral dissertation in
psychology in 1974 at Columbia University.
in nursing homes like they are here, not being stared at, but accepted as part of a community.”

In the wake of the 1972 meeting, tensions had developed between those, like Milton Wexler, who fervently believed that the CCHD should fund basic research and those, like Marjorie Guthrie who, equally fervent in belief, argued that spending should be divided between science, public education and patient services. These tensions intensified and, in 1974, Wexler formed a separate organization, the Hereditary Disease Foundation (HDF). The tension between these two basic sets of priorities — scientific research leading to a treatment or cure and the provision of services which would assist families affected by the disease — would, however, remain as each group attempted to forge a different path toward a common goal (Wexler, 1995).

Eventually becoming the Huntington Disease Society of America (HDSA), the CCHD worked at a grassroots level to build a network of families, caregivers and health professionals devoted to the provision of clinical and support services. New chapters formed in many areas and the support extended to families affected by the disease served to lessen social isolation and fear. In Canada, the first meeting of what was to become the Ontario chapter of the CCHD occurred in 1973 when Ralph and Ariel Walker gathered together a handful of people at Sunnybrook Hospital in Toronto. With twenty-nine interested families, the chapter soon produced its first newsletter and, in the following year, the third newsletter proudly announced a new name and organization, the Huntington Society of Canada (HSC). There were, in 1974, 350 families on the mailing list; chapters had begun in Hamilton and Winnipeg as well as Toronto; Dr. André Barbeau promised medical backing for a Montreal chapter and, in British Columbia, Dr. Thomas Perry offered similar assistance. Noting the significant impact which local chapters can have in fostering on public awareness of HD, Ralph Walker stated in a 1974 newsletter,

It is quickly becoming clear that H.D. is not a rare disease. I am sure that as soon as we are able to receive the much needed publicity which Chapters can provide, we will discover that H.D. has touched every part of Canada.

Published epidemiological studies on HD had by this time established that although the disorder was prevalent in many counties the highest prevalence of HD occurred in populations which traced their ancestry to western Europe. Given the low mutation rate and historical patterns of migration, colonial expansion and world trade, this pattern was suggestive of a “founder effect”. As Folstein (1989:7) describes it,
When a gene is introduced by a founder (either by a new mutation or by immigration) to a particular population, and when that gene has even a very slight reproductive advantage, the frequency of the gene in that population will tend to increase.

Genealogical evidence pointed strongly to France, Germany, Holland or Norway as the site of the original mutation but since the mutation may have occurred more than once, there may have been several “founders”. Many affected individuals living in the U.S. are now believed to be distant descendants of three men who emigrated from England in the early seventeenth century but German, Scottish, Irish, Scandinavian and Italian ancestry has also been documented in some affected families (Hayden, 1981). In Canada, French and English settlers have been shown to be the primary contributors of the gene.

Assisting with the development of a register of families affected with the disease in French-speaking regions of Canada, Barbeau was able to trace all such families back to a common ancestor from mid-seventeenth century France (Wexler, 1995). Tracing the presence of the disorder in distant ancestors has, however, not always been easy; many investigations have been hampered by a lack of official documentation, especially for the years prior to 1872. As such, many family histories have been established only through painstaking attention to available details about changes in personality, anti-social or promiscuous behaviour, or diminished abilities to perform normal tasks — all of which could yield clues that might provide the determined investigator with enough information to establish a probable family history.

Such information was, however, not always easily accessible. As Hayden (1981:13) stresses,

> In view of its tragic and far-reaching social effects, denial of the existence of the disease, even when relatives have been admitted to mental hospitals, is common. Family members are often only willing to reveal their ancestry once a relationship of trust and friendship develops.

Well-documented family studies of HD with extensively detailed pedigrees have been crucial to the work of those scientists who, from the mid 1970’s on, sought to understand the genetic as well as biochemical processes of HD. Indeed, the linkage studies which were eventually to yield the first major breakthrough in localizing the gene for HD would not have been possible without the cooperation of numerous extended families affected by the disease.

Clinical studies had documented with increasing precision, the triad of cognitive, emotional and physical changes associated with the disease but by the mid 1970’s the focus of
scientific research began to shift from the more descriptive aspects of the disease to the biochemistry, cell biology and genetics of the disease. Likewise, the social consequences of HD for at risk individuals and families received additional attention when, in 1975, a group of families from New Jersey succeeded in their efforts to lobby for a Congressional Commission for the Control of HD and Its Consequences. Chaired by Marjorie Guthrie and co–anchored by Milton Wexler as Vice–Chair, the Commission held numerous meetings, workshops and hearings with government groups, medical professionals, service agencies and the public. It was, for many of the families who gave testimony to the Commission, like a “revival meeting”; many had never met another family afflicted by the disease and some had never talked to anyone about their own family history and experiences of the disease. Further, as Nancy Wexler, who served as Executive Director on the Commission, later noted, the prominence given to the project was extremely therapeutic to those within the HD community who had worked for so long to overcome ignorance and disinterest in the disease (Wexler, 1995:139).

Offering a number of proposals for the management of HD as well as a series of recommendations for further research on the genetic and biochemical basis for HD, the ten volume report of the Commission came at a time when scientists were making steady progress in developing new techniques for localizing genes to particular chromosomes. Linkage studies, which were pioneered in the 1930’s by Bell and Haldane, held out the promise that scientists might, given plenty of markers and access to enough large families, be able to isolate the gene for HD. Further, studies on other heritable disorders (such as familial hypercholesterolemia) indicated that two copies of the faulty gene could exacerbate the condition. As such, there might be some immediate utility in locating individuals who had two parents suffering from HD.

Given the extremely high incidence of HD in the communities of Lake Maracaibo, Venezuela, it seemed possible that researchers might be able to locate individuals who had a double dose of the gene for HD. Embarking on the first of many trips to Venezuela in 1979, it was Nancy Wexler who spearheaded this search and, in conjunction with a team of American and Venezuelan neurologists and geneticists, assembled a huge pedigree and collection of skin tissue and DNA samples for analysis. Families in these isolated communities were at first surprised to learn that their disease, *el mal*, existed elsewhere. Initially wary about giving blood
samples to the researchers, many were eventually won over by Nancy Wexler's disclosure that her mother had HD and that she too was at risk for the disease. As Nancy Wexler (cited in Wexler, 1995:191) later said in reference to the mark she had from a skin biopsy, "If I had not had that biopsy scar, they might never have believed me.... It really made a difference. After that, even if they thought the whole thing was crazy, at least they never doubted our sincerity."

Working with restriction enzymes, the new molecular tools which allowed scientists to snip DNA into manageable size pieces, scientists in the early 1980's were intent on devising ways to determine the order and sequence of the bases which comprise DNA. Certain stretches of the DNA were found to contain distinctive variations or polymorphisms which could be used to distinguish one person's DNA from another. These polymorphisms were transmitted from one generation to the next and, if they were located very close to a disease causing gene, it was likely that the two would travel together despite the chromosomal reshuffling which occurs during the production of eggs and sperm. Among those most intent on applying the idea that you could use a polymorphism as a marker for a disease gene was David Housman, a scientist from MIT who was involved in the ongoing workshops on HD research. Housman persuaded one his recent graduate students, James Gusella, to focus on the search for a marker for HD.

Developing a series of probes or pieces of DNA which were complementary to the sequence of bases found in a particular polymorphism, Gusella began to look for a marker which traveled with the gene for HD. Given that no one knew which chromosome the gene (or genes) might reside on, and that one could never be certain that a particular probe was only latching onto the fragment of DNA which contained the desired polymorphism, this task seemed monumental. Nonetheless, Gusella was extremely lucky. While testing his third probe on DNA samples obtained by Michael Conneally from a small HD lineage in Iowa, Gusella (1983) discovered a marker which was consistently present in members of the family affected with HD but not in those who were unaffected. Statistical checks run by Conneally confirmed the pattern in the Iowa family but a larger sample was required and thus, it was the Venezuelan kindred which ultimately confirmed, in 1983, the utility of Gusella’s marker (Revkin, 1993).

30 This family is now believed to have the largest concentration of HD in the world; there were, in 1990, 144 living affected members and over 1000 more at risk (Roberts, 1990:625).
Based on the predictable ways in which a DNA segment will break apart when digested by particular enzymes, the use of restriction fragment length polymorphisms (RFLPs) had narrowed the search for the Huntington’s gene to the tip of chromosome 4. As the first proof of the power of new techniques in gene mapping, this discovery was of enormous significance to the human genetics and molecular biology communities (Wexler, 1995). Moreover, the discovery also catapulted HD into prominence as a vital area of research in molecular biology. As Wexler (1991:80) notes, “localizing a gene whose chromosomal origin was unknown confirmed the practical value of a novel strategy applicable to almost all hereditary disorders”.

With this discovery, six teams of scientists (brought together by the Hereditary Disease Foundation) agreed to work collaboratively in the search for the precise location of the gene for HD. This search was to be longer and more arduous than expected but the collaborative venture ensured that the data, and the eventual credit, would be shared amongst scientists who would otherwise be in direct competition. Two other teams, headed by Richard Myers and David Cox (University of San Francisco) and Michael Hayden (University of British Columbia), opted to work either independently or in conjunction with other groups of geneticists.

The possibility that finding the gene would lead to an understanding of the basic biochemical pathology associated with the disease and hence, an effective treatment or cure, provided much-needed hope to families affected by the disease. If, however, therapeutic potential remained in the future, presymptomatic testing was an immediate possibility. Prior to the discovery of a linked marker for HD, there was no reliable means of determining whether presymptomatic individuals at risk for HD had inherited the gene. Early attempts to refine the diagnostic technique of L-Dopa loading had proven unsuccessful. Moreover, the experiments done by neurologists André Barbeau and Harold Klawans had elicited a storm of controversy about the ethics of using L-Dopa to provoke chorea–like movements in those at risk at for HD. As the report of the Commission for the Control of HD proposed in 1977, genetic linkage offered a more promising route toward a presymptomatic test since at risk individuals would not

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31 In the late 1980’s, the Canadian Government established a program which would offer substantial research funding to cohesive groups of scientists working together in many areas of research. As one of only 15 Centres of Excellence, the Genetic Disease Network was founded in 1990 by Michael Hayden. This network brought together many researchers and permitted the sharing of core facilities and expertise.
be subjected to the terrifying experience of chorea-like symptoms (Wexler, 1995).

By 1986, several new linked markers had been isolated and, although the precise location of the HD gene remained unknown, scientists were able to use these markers to develop a reliable predictive test for HD. In addition, the Venezuelan data had shown conclusively that having two copies of the gene for HD did not alter the clinical manifestations of the disease. Thus, individuals homozygous for the disorder were not more severely affected and HD, it seemed, was a true autosomal dominant (Wexler, 1995). In 1986, genetics centres in the U.S., Canada and the U.K. therefore began to offer the linkage form of predictive testing to those at 50% risk. Where informative, this test provided at risk individuals with up to a 95% probability that they did or did not have the gene for HD. To be fully informative, linkage testing required a documented family history and DNA samples from affected and unaffected family members. Consequently, linkage testing demanded the support and co-operation of a significant number of people. Sometimes necessary DNA samples were unavailable because significant family members were deceased or did not wish to participate. Further, the limited number of markers available for the test meant that some families’ genetic make-up was uninformative.

Given concerns about the well-being of test candidates, clinical geneticists and neurologists worked in conjunction with the International Huntington Association to develop guidelines for offering the linkage test. Initially, the test was only offered as part of a research protocol which included extensive psychosocial assessment, counselling and follow-up sessions as well as a protracted period of time in which the test candidate could reconsider or withdraw from the program (Fox, et al., 1989). As the product of extensive dialogue between scientists, clinicians and lay organizations, the protocol for offering predictive testing for HD has served as a model and a stimulus for thinking about how to offer other types of genetic testing (Quaid, et al., 1996; Wexler, 1996). As Harris (cited in McKie, 1988) confidently stated over a decade ago, 

...the complex ethical issues we face with Huntington’s chorea are exaggerated versions of many of the problems we will shortly face when introducing other forms of genetic screening. If we get it right with Huntington’s chorea we can then hope to tackle other diseases and conditions.

When, in 1993, the Huntington’s Disease Collaborative Research Group announced that
they had cloned the gene for HD, it soon became possible to provide at risk individuals with a definitive or direct test. In contrast with linkage testing, the direct test is based upon analysis of the region of DNA believed to be directly implicated in causing HD. This region — which is located on the short arm of chromosome 4 — has been described as a “genetic stutter” in which the trinucleotide sequence of CAG is repeated many more times than in the unaffected population (Andrew, et al., 1993). The degree of expansion in this region is, at its upper limits positively correlated with an earlier age of onset for HD but, given the wide variance in the correlation for most repeat ranges, this data is considered useful on an individual basis in only a very small number of cases (Andrew, et al., 1993; Craufurd & Dodge, 1993; Duyao, et al., 1993; Snell, et al., 1993). In consequence, there has been much debate within medical genetics about whether or not repeat numbers should be provided to test candidates (Burgess & Hayden, 1996).

The shift from linkage to definitive testing — which occurred in the fall of 1993 — has had other immediate implications for how predictive testing is conducted: with a reduced need to acquire DNA samples from additional family members, those at risk are able to obtain information about their HD status somewhat more quickly and discreetly than before. Nonetheless, given that there is still nothing that can be done to prevent or delay onset in those who have inherited the gene for HD, the increased accuracy and certainty of the direct test imposes an added burden of responsibility on service providers; “with 100% certainty, all hopes of not being a victim are dashed to the ground” (Mattson & Almqvist, 1991:27).

Extensive research and international cooperation among scientists has, in the last few years, yielded many advances in understanding HD. Although scientists do not yet know the root biochemical cause of the disease, progress has been made in understanding the specific DNA sequences within the gene. In particular, it has been determined that an expanded sequence of CAG repeats is associated with production of an abnormal form of the protein, huntingtin. This protein is necessary to normal development but the mutant protein may generate toxic byproducts which damage specific types of brain cells. Further, the development

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32 Hayden’s team was, at this time, also very close to finding the gene for HD. Publishing in the journal *Nature* on the day before the Collaborative Group published their findings in the journal *Cell*, Hayden’s team had discovered a candidate gene which was within 200 kilobases of the gene identified by the HD Collaborative Group (Goldberg, et al., 1993).
of a mouse model for HD permits scientists to further explore the pathogenesis of HD and test new therapeutic and preventative strategies (Nasir & Hayden, 1995). Nonetheless, an effective treatment for HD is, as of yet, elusive and ameliorative actions are still largely restricted to the administration of anti-depressant and anti-choreic drugs.

In the continued absence of effective treatment, the care provided through enhanced patient and family services is vital. Likewise, local chapters of the HDSA and the HSC ensure that families have a network of people to turn to for advice and support. Sharing a common experience with the disease, families are also able to work with various professionals to devise appropriate methods for increasing physician and public awareness of the disease.

Reflecting on the feeling of helplessness which the lack of treatment must have long ago occasioned in his grandfather, Charles Gardiner Huntington, George Huntington’s grandson, (cited in Durbach & Hayden, 1993) stated “I have known people suffering with Huntington’s disease and when I saw them I knew what it was my grandfather felt. Especially, I knew that feeling of there being so little that we could do for them.” The inability to offer effective treatment did not, however, prevent Charles Huntington (cited in Durbach & Hayden, 1993:409). from proposing that his grandfather, George Huntington, “gave people with the disease a humanity they’d never had before”.

As Frank (1991: 104) argues, “humanity” derives as much from the willingness to recognize and acknowledge suffering as it does from any medical intervention.

When we know that someone recognizes our pain, we can let go of it. The power of recognition to reduce suffering cannot be explained, but it seems fundamental to our humanity.

The recognition of suffering is essential to any definition of humanity. Yet, suffering is often threatening and what is threatening is often left unspoken or even actively denied. Medicine has, in many areas, a history of complicity in the process of denying suffering, especially where there is no available treatment or cure for the cause of suffering. It may, therefore, appear to be something of an anathema to critics of modern medicine to suggest that the naming of ‘that

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33 The article “George Huntington: The Man Behind the Eponym” provides evidence of the doctor’s commitment to patient care and talent for observing “something special about those affected by the ‘chorea’ he later described” (Durbach & Hayden, 1993).
disorder' might, among other things, bequeath to its' sufferers a sense of humanity. Steeped in
the tradition of objecting to medicine's predilection for reducing whole persons to disease
categories — the stage 4 carcinoma in room 302 or the prolapsed uterus in the O.R. — it is
sometimes tempting, for the (temporarily) healthy and able-bodied, to overlook the profound
significance of being able to name the source of one's suffering and ill health.

Attending to the possible ways in which the naming of 'that disorder' may affirm the
humanity of those who suffer from it is then, a line of inquiry which must parallel but not
eclipse other less than humanitarian consequences of naming. Scientific medicine has played an
important role in dispelling the myth and superstition which have long contributed to the social
stigma which shrouds HD. In describing the salient features of 'that disorder', biomedicine
provides those who suffer from this disease with an explanation for its occurrence. The social
utility of this diagnostic label in alleviating public misunderstanding is poignantly illustrated by
a Huntington Society of Canada pamphlet: emblazoned across a photograph of a man who looks
as if he may be staggering, the caption reads "he's not drunk, he has Huntington Disease" (see
Figure 2, next page).

The social power of diagnostic labels is, however, Janus-faced. Clumsy efforts to
catalogue families affected by the "taint" of heritable disease exist in rather close historical
proximity to the more cautious creation of scientifically informative genetic registries. And
while mandatory sterilization has, thankfully, been outlawed the rhetoric of prevention
continues to surface in contemporary social and scientific discourse on genetic disease.

Although being at risk for HD is not a disease, those at risk for the disease do share in
many aspects of what is commonly referred to as an illness experience. In Chapter III, I describe
some of the most salient aspects of this experience from the point of view of existing studies on
HD and predictive testing. Noting that much of the research tends to frame predictive testing in
terms of a predominant "discourse of potential benefits" (Boutté, 1988), I question the way in
which this discourse attaches a particular moral worth to information which facilitates rational
planning and choice while at the same time obscuring the social construction of such choice.
He's not drunk.

He has Huntington's.

Huntington's disease is a cruel hereditary brain disease. It causes relentless physical and mental deterioration, and eventual death.

Help us find a cure.

You can make a difference.
CHAPTER III
THE 'GIFT' OF KNOWING

So he [the doctor] said to me, "Well, what you receive today which ever way the results are, is a gift. You have the gift of knowledge, you know, even if you're going to get it [Huntington Disease]." And we were looking at it that way, you know, even if we found out that I inherited the gene. It was a gift. It would be a gift to find out for sure. (M, 50% risk, PT candidate, 41 years, married, 3 children)

With no effective treatment or cure for HD yet available, the insidious first symptoms and typical late onset of the disease are often a chronic source of uncertainty for many at risk individuals and their families. Until recently, this situation could only be resolved in an unfavourable direction — that is, through clinical diagnosis of the disease. With the advent of predictive genetic testing for HD, it has become possible to provide individuals at 50% risk with the "gift of knowing" (Kenen, 1996) whether or not they will become affected by the disease later in life. The individual, social and familial meanings of receiving this "gift" are, however, complex. The test results may offer a profound sense of relief or they may engender new and unexpected kinds of uncertainty. Neither experience is wedded to a particular test outcome nor are experiences of relief and/or protracted uncertainty mutually exclusive.

Clinical experiences in offering predictive and prenatal forms of linkage and direct testing to individuals at 50% risk for HD, have been central in understanding many of the ethical and psychosocial implications of predictive genetic testing. In fact, "HD represents the disorder for which predictive testing has been offered for the longest time, to the largest number of people" (Benjamin, et al., 1994:615).

Purpose and Outline of Chapter

In this chapter, I review and critically evaluate the literature on predictive testing for HD. This literature is primarily drawn from the fields of psychology, social work, genetic counselling and clinical and medical genetics. Other significant contributions to the growing literature on the social and familial implications of genetic testing (which are not specific to testing for HD) are drawn from sociology and anthropology.
The first section of the chapter focuses on the familial implications of living at risk for HD and is primarily descriptive in tenor; the second evaluates the strengths and weaknesses of existing research as well as the predominant social construction of predictive testing as a "gift of knowing".

Much of the existing research has focused on individual psychological aspects of the clinical experience — test results are constituted as the primary independent variable and the measurement of outcomes is often considered the most salient objective. The predominant research ethos is positivist rather than interpretive and, as such, much of the literature is oriented toward the description and elaboration of causal relationships. Moreover, few studies acknowledge the degree to which research and service protocols themselves shape the experience of predictive testing and fewer still consciously reflect on how knowledge about predictive testing is socially constructed, whom it is being constructed for and, with what consequences. Thus, while existing research fulfills an important mandate for clinical evaluation, it does not provide significant insight into the meaning and lived experience of predictive testing for at risk individuals and their families.

**Huntington Disease: A Family Matter**

The story of predictive testing and its social construction as a "gift of knowing" begins long before at risk individuals arrive at the clinic for their first counselling session. The story begins with family and the profound effects that growing up at risk for this disease may have on self-identity and social life for, if there is one thing that can be stated in absolutely unequivocal terms, it is this: Huntington Disease has significant implications for all members of the family. Siblings and spouses, children, caregivers, persons at risk for, or living with HD — all are affected by the disease. Indeed, if all close relatives of individuals affected with HD are included, "almost 10 times as many persons suffer chronic anxiety about Huntington’s chorea as actually have the disease" (Perry, 1981:1098).

In many respects, HD provides a dramatic example of the problems associated with late

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1 In fairness, these concerns illustrate more about the need for alternative approaches to research on predictive testing than they do about the limitations of existing research.
onset disorders. For families at risk for HD, the combined effects of insidious symptoms and a delayed age of onset produce a family in “chronic stress” (Wexler, 1984:131). As Hayes (1992:1449) describes it,

We had to wait, watch for minor twitching, mood swings, and forgetfulness, and hope that they did not become severe enough to confirm our worst fears. We knew that if Huntington’s disease developed there was (and still is) nothing to be done.

The effects of living with chronic stress and uncertainty have been fairly extensively documented within the literature on HD. Since the mid-1970’s, clinicians, psychologists and social workers have published case studies and reports which describe and illustrate the impact of HD on affected individuals and families (Barette & Marsden, 1979; Korer & Fitzsimmons, 1985; Phillips, 1982; Phipps & Desplat, 1984; Tyler, et al., 1983). The Congressional Commission for the Control of HD and Its Consequences published a ten volume report which contains many testimonials from families and the Huntington Society of Canada (HSC) and the Huntington Disease Society of America (HDSA) have, from the outset, featured the stories of family members and caregivers in their publications. Most of these accounts are, however, based on small and/or self-selected samples and it has only been in the last few years that more extensive empirical studies of the at risk population have been carried out.

Drawing upon her own experience as well as a series of in-depth interviews with individuals at risk for HD, Nancy Wexler (1979) was one of the first to carry out a detailed study of the “inner world” of living at risk for HD. Concerned to elaborate the social meanings of HD as well as the particular images and fears it evoked, Wexler found that HD was commonly perceived as a “time bomb.” In contrast with the pain and suffering evoked by cancer or the sense of unpredictability which denotes multiple sclerosis, HD imposes a burden of anticipation and silent apprehension. One the chief reasons for this sense of apprehension lies in the nature of HD symptomatology.

Many of those at 50% risk for HD are exposed, at a fairly early age, to the intellectual deterioration, choreic movements and personality changes of HD in an affected parent or other family member. Indeed, for many of Wexler’s study participants, the inexorable mental, physical and emotional decline associated with HD seemed to “strike at the core of their
physical and psychological self-esteem” such that many imagined “a vision of a Frankensteinian monster, one who approaches others with affection but from whom others recoil in horror” (Wexler, 1979:201).

The following discussion outlines the nature of HD symptomatology, its onset and progression, as well as the psychosocial and familial implications of diagnosis. An appreciation of these aspects of the disease is an essential but all too often overlooked prerequisite to more topical discussions of predictive testing. Indeed, in the attempt to focus on a select number of more easily operationalized variables, studies of the psychological impact of predictive testing often neglect to include such salient factors as whether or not the at risk individual has grown up knowing about HD, whether they have observed firsthand its progression, and how, in turn, this has shaped predominant images of HD.

Symptoms, Onset and Diagnosis of HD

The age of onset and progression of HD varies widely. The typical age of onset is between thirty-six and forty-five years of age but the disease may remain quiescent until the individual is well into old age. At the opposite end of the life-course, the juvenile form of HD may manifest itself during adolescence or, in rare cases, early childhood (Hayden, 1981).

Juvenile onset is associated with a more rapid and marked decline. In addition, it has been observed that early onset is often related to paternal transmission. This trend appears to defy Mendelian patterns of inheritance but, since discovery of the mutation associated with HD, the sex-dependent effects of major expansion and contraction of the CAG repeat have been linked to differential processes of gametogenesis in males and females. The offspring of affected fathers are more likely to have large expansions than the offspring of affected mothers. Conversely, the offspring of affected mothers are more likely to show no change or a reduction in CAG size (Kremer, et al., 1995). Although there have not yet been any published studies which document familial awareness of the sex-linked effects of transmission, it is likely that such awareness will have significant consequences for how women and men interpret the significance of their own family history of HD and/or engage in reproductive decision-making.

2 This phenomenon was first reported in 1968 and since then the finding has been observed in HD populations from all over the world (Harper, 1991; Hayden, 1981).
For most individuals affected with HD, the early symptoms are subtle. These symptoms include increased "nervous" activity and clumsiness as well as slight cognitive impairment and/or moodiness and depression. If HD is present, these initial symptoms will, in most cases, gradually become more pronounced. Involuntary choreic movements may, at first, be experienced as slight twitches and jerks. With progression of the disease, however, head, neck, torso and limbs may all begin to move in an uncontrollable fashion. In approximately one fifth of those affected with HD, rigidity rather than chorea may be present. In either case, the loss of control over voluntary movements often leads to difficulties with balance and walking, eye and hand coordination, speech and swallowing. Steady or sudden intellectual decline may involve diminished abilities to organize routine responsibilities, cope with unusual situations or recall recent events and, accompanying emotional distress may be evident in the form of bouts of increased irritability, depression or impulsiveness (Huntington Society of Canada, 1996).

With the progressive development of symptoms, most affected individuals become consciously aware that the disease is present. During this "incipient stage" (Bloch, et al., 1993), many will adopt strategies of denial, rationalization and/or repression. Because symptoms are initially quite mild, affected individuals may blame other people and/or stressful situations for their apparent deviations from normalcy. This response may isolate and alienate affected individuals from their own experience as well as from available support. Nonetheless, as Bloch et al (1993:369) propose, strategies of denial and repression are not necessarily maladaptive. Denial provides time to perform the important emotional work of slowly assimilating a new reality and preparing for conscious acceptance of a diagnosis of HD.

When more pronounced symptoms become a regular intrusion into daily life, awareness of HD cannot be forestalled. Anxiety about confronting the situation may, however, make it seem more acceptable to deny the origin of the problem and seek professional help under another pretext. Indeed, with the development of predictive testing, a number of symptomatic individuals have enrolled for "pre-clinical" assessment hoping that they can begin to deal with HD but at the same time forestall the day of diagnosis (Bloch, et al., 1993; Kessler & Bloch, 1989). Clinicians must exercise extreme sensitivity and neither push the person toward premature diagnosis nor hold out false hope that everything is fine (Bloch, et al., 1993).
Clinical diagnosis of HD marks an important transition for the affected individual and for family members. Even amongst those who knew prior to formal diagnosis that they were affected, there is often immense disappointment and loss of hope. Nonetheless, the resolution of uncertainty may also provide a sense of relief. Where prior knowledge about the disease and an open and inquiring attitude is supplemented by a responsive and informed network of family members and friends, positive coping on the part of the affected individual is a strong probability (Bloch, et al., 1993). On the other hand, maladaptive responses, which range from withdrawal and severe depression to suicidal ideation and behaviour are not uncommon in the weeks and months following clinical diagnosis.

Reviewing the national HD roster in the U.S., the Commission for the Control of Huntington's Disease and Its Consequences found, in 1977, that 5.7% of deaths amongst those affected with HD were attributed to suicide, a rate seven times the national average. More recently, it has been found that approximately 28% of persons affected with HD attempt suicide at least once (Kessler & Bloch, 1989:65). Stressing the importance of early recognition and intervention in depression and suicidal ideation, psychiatrist and neurologist Allen Rubin commented, in a presentation to members of the Huntington Society of Canada, that earlier reports of HD–related suicides probably reflect more about the number of individuals who were detached from a supportive and potentially therapeutic community than they do about the progression of HD itself. Suicide may be a difficult subject for professionals and caregivers to broach but “talking about troubles does not make them happen” and, as Rubin asserts, “talking about suicide can uncover solutions that can be preferred routes of response to what seem to be intolerable situations” (Huntington Society of Canada, 1996:34).

From the perspective of the affected individual, long term adjustment to living with HD requires gradual changes in many aspects of life. Self–identity and the experience of time may, in particular, seem altered and this will affect attitudes and priorities (Bloch, et al., 1993). The impact of diagnosis on family relationships, employment, financial security, insurance and health care coverage may also be extensive but as, much of the lay literature points out, a

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3 Conflicting evidence suggests that on one hand, HD related suicides occur primarily in the early stages of the illness and, on the other, they occur more often during the latter stages (Huntington Society of Canada).
diagnosis of HD does not mean that an immediate change in lifestyle is required (Huntington Society of Canada, 1996). Many persons with HD are able to continue regular employment and other productive activities for some time after diagnosis. The continuation of these activities contributes to gradual adjustment and, in addition, may alleviate, or at least postpone, the feeling of being a burden to caregivers and family.

For the affected individual’s spouse or partner and family, the actual diagnosis of HD may provide a sense of validation. Subject to protracted uncertainty and stress in the period leading up to diagnosis, many families have coped for long periods of time with HD–related problems of domestic violence, sexual aggressiveness and abuse, alcoholism and/or what appears to be a propensity to become involved in various types of criminal activity (Korer & Fitzsimmons, 1985; Phipps & Desplat, 1984; Yale & Martindale, 1984). With the news of clinical diagnosis steps can be taken to address these problems. At the same time, however, new problems arise. Financial security may be doubly threatened by a loss of income and a host of new expenditures. The family home may need to be adapted to ensure that it is safe and accessible. Assistance with caregiving and/or homemaking may be required and, particularly where it is the wife/mother who is affected with HD, concerns about domestic responsibilities and the maintenance of the family may predominate (Yale & Martindale, 1984).

As studies on the impact of mental illness indicate, gender roles play an integral part in day to day family organization and co–ordination; as such, there is often a greater impact on effective family functioning when the mother is affected than when the father is affected. This has not yet been investigated in the case of HD, but Kessler (1993a; 1993b) notes that gender differences are striking in families where children participate in caregiving for a parent affected with HD. Daughters frequently pick up their mother’s domestic duties while sons are not as often expected to contribute in such ways.

Socioeconomic status will, of course, also have an immediate bearing on whether or not the family will be able to enlist the services of a full–time nurse or companion such that the affected individual may continue to live at home for as long as possible. Being able to live at home and maintain regular ties to family life is integral to the well–being of persons with HD but it may also have significant consequences for how other members of the family cope with
HD. Stressing that perceptions and consequences of the disease differ widely amongst families according to factors which cannot be adequately conceptualized in terms of an individualistic biomedical model, Kessler and Bloch (1989:67) argue that,

...individuals whose parents dealt with the diagnosis and progression of HD with courage and dignity tend to perceive the personal threat of HD with less terror and panic than persons whose parents became dysfunctional and subjected them to abuse and/or molestation...The experience of family members in these two circumstances is so vastly different that it is as if one were dealing with a different disease entity. Economic and psychosocial factors provide the context that gives meaning to the biological aspects of HD (emphasis added).

Studies conducted by British social workers also confirm that families exhibit a wide variety of responses to the diagnosis of HD — many cope quite well with diagnosis while others have enormous difficulty in adapting to the inevitable deterioration of a spouse or family member (Yale & Martindale, 1984). Despite these differences, however, families overwhelmingly encounter frustrations related to acquiring the necessary level of professional intervention and home support. This situation has improved as awareness of the disease has increased but many problems remain. Communication between specialists may be uncoordinated or completely lacking; family physicians may be unfamiliar with the management of HD (Huntington Society of Canada, 1996), and social workers, who could assist the family are often not called in until family relations are stretched to the breaking point (Yale & Martindale, 1984).

Familial Dynamics of Diagnosis and Living at Risk

Emphasizing the importance of considering the needs of family members who are at risk for HD, Yale and Martindale (1984) argue that the time of diagnosis is particularly crucial. In many cases, diagnosis of an affected family member precipitates new discussions of genetic risk and, inevitably, there are those who discover for the first time that they are at risk. This discovery may invoke shock, disbelief and a profound sense of threat.

As Folstein (1989:172) argues, those who are well into adulthood before learning about their risk for HD are often completely preoccupied with this information. Children may be somewhat less disturbed since they may not fully understand the implications of being at risk while teenagers may in turn be somewhat shielded by a youthful sense of optimism and
invulnerability. Such age-specific responses have not, however, been the subject of detailed study and the clinical observations of Folstein (1989) and others may underestimate the adverse responses of children and adolescents to the news that they are at risk for HD.

With time and the realization that the threat is not imminent, those at risk develop various strategies for coping with knowledge of their genetic risk. In particular, there are a variety of psychological defense mechanisms which may be employed by at risk individuals who seek to manage chronic uncertainty and/or hold at bay the recognition of onset in themselves or a family member. These include denial ("it won't happen to me"), rationalization ("I could get knocked over by a truck") and attempts to banish all thoughts of being at risk (Bloch, et al., 1993:369). These strategies are not necessarily maladaptive; they provide at risk individuals with a means of slowly assimilating new information and this may eventually assist in conscious acceptance of being at risk for HD. Such attempts to manage the uncertainty of living at risk may, however, be disrupted by seemingly innocuous events such as birthdays or more ominous events such as the diagnosis of another family member.

With awareness and provisional acceptance of being at risk for HD, there is often a new impetus to obtain information and advice. Learning the facts about HD may alleviate some of the chronic anxiety of living at risk for the disorder but, as Wexler (1979:200) has argued, many individuals at risk for HD are "disease-wise" and "doctor-shy" — that is, from a fairly early age they know the odds of inheriting HD but "often regard the genetic counsellor with the same wary contempt the battlefield soldier shows for the journalist — ticking off in round numbers the daily toll of lives lost, reducing the human struggle to arid statistics."

Familial knowledge about HD often incorporates a variety of "myths and misunderstandings" and, if these are not sorted out through genetic counselling or other means, family members may draw erroneous conclusions about the implications of being at risk for HD. Ironically, one of the most common misunderstandings derives from one of George Huntington's most central and, I might add, carefully worded observations of the disease: "it never skips a generation to again manifest itself in another." In common usage this phrase is

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4 The adolescent experience of living at risk for Huntington Disease is currently being investigated by Jessica Easton and will comprise the topic of her doctoral dissertation at the University of British Columbia.
often abbreviated to “it never skips a generation.” For those who understand the genetics of HD, the phrase is a shorthand way of saying that HD cannot remain dormant for a generation and then suddenly reappear in a subsequent generation. Severed from this understanding (or the additional qualification that each child of an affected parent has a 50% chance of inheriting the disease) the abbreviated phrase appears to mean something quite different; thus, the mother who concluded that because it never skips a generation “either my son or my daughter will get it” (Korer & Fitzsimmons, 1985) can hardly be faulted for any illogic in her thinking. Other examples of family myths about HD include the notion that “every third child will get it” or “the oldest will get it and then it gets weaker down the line.”

Although such misunderstandings often do indicate a lack of awareness of the scientific facts, they may also represent an abstracted form of knowledge about how the disease tends to manifest itself within a particular family. As such, family “myths” and “misunderstandings” may sometimes signal the need for contextualization rather than straightforward correction. The possibility of recognizing family “myths” as an alternative form of knowledge is, however, not generally reflected in the clinical literature on HD and/or genetic counselling. 5

Social scientists writing about the need to engage alternative approaches to research on the social construction of genetic knowledge have found that lay beliefs about inheritance are often at variance with the Mendelian theories of geneticists (Cox & McKellin, in press; Richards, 1993). Lay beliefs about proneness to heritable and non-heritable disease provide a particularly cogent illustration of this. In families of children with cystic fibrosis, Burton (cited in Richards, 1993) found that a lack of other affected family members often accompanied confusion about heritability of the disorder:

It isn’t really an inherited condition but it does come from the parents — both parents having the same genetic fault.

In contrast, the presence of more common non-hereditary disorders throughout several generations may, despite medical evidence downplaying the influence of heredity, result in the belief that all such conditions “run in the family.” As Blaxter and Paterson’s (1982) study of

5 This is a point I shall refer to again but for now suffice it say that I am not suggesting that genetic counsellors should treat lay beliefs about HD as if they were equivalent to scientific truths; they are of a different order altogether but ought, all the same, to be acknowledged as forms of knowledge which often have a basis in the observations and lived experiences of families at risk for HD.
three generations of women in Scottish working class families demonstrates, such reasoning makes sense given that protracted exposure to a poor environment may affect several generations in the same way. Referring to these and other studies, Richards (1993:577) suggests, it “seems to be a common feature of lay beliefs — that you can only pass on what you and your close relatives have and are seen to have.” If this is indeed the case, a comparison of how families respond to the occurrence of recessive versus dominant single–gene disorders may shed further light on how individuals weigh family history and heredity in formulating understandings of individual and familial proneness to disease.

Within the literature on the psychosocial and familial dimensions of HD, symptom–seeking and patient preselection figure prominently as related phenomena which illustrate the degree to which family life may become saturated by beliefs about HD. Given that many of the early symptoms of HD are insidious, ”symptom–seeking” (Kessler & Bloch, 1989) or “symptom–searching” (Korer & Fitzsimmons, 1985) on the part of at risk family members may become a common preoccupation. In an in–depth study of families at risk for HD, Korer and Fitzsimmons (1985:589) found that parents of at risk offspring often engaged in monitoring of their children’s physical and emotional behaviour: as one anxious mother mother stated,

I worry all the time. I keep looking for signs. I worry terribly. I have dreadful nights since I found out it could be passed on. I had terrible nightmares. I keep looking for them doing things they wouldn’t normally do and then I can’t do anything about it.

Internalizing fears about the role of emotional distress as a catalyst for onset of HD, some mothers went so far as to hold themselves personally responsible for warding off the onset of HD by trying to keep their children on an unnaturally even emotional keel.

Preselection — the practice of singling out in advance which members of the family are going to develop the disease — is another phenomenon which frequently occurs in families where a parent is beginning to show early signs of HD. Where there are several children, observations of coincidental familial resemblances or mannerisms may be seen as indicators of proneness. One woman cited in Korer and Fitzsimmons’ (1985:590) study commented that:

When he’s had a drink he reminds me of his Dad. I’m a bit frightened of him when anything upsets him. I worry that he might be like his Dad. It’s always at the back of my mind. I’ve always felt that he would get it because he’s a lot like his Dad.
Analyzing the phenomenon of preselection as an aspect of the familial effects of living at risk for HD, Kessler and Bloch (1989:61) argue that because preselection delegates one family member to the “sick role” (Parsons, 1953) the behaviour of other family members becomes organized around beliefs consistent with this assignment. This creates the impression that uncertainty can be managed: “it is as if the chance nature of gene transmission is brought under control and the chronic threat of the laws of probability defused” (Kessler & Bloch, 1989:61). At the same time, however, the occurrence of symptoms in those individuals who are not preselected may create enormous difficulties — “it is not the way the script is supposed to be played out” (Kessler & Bloch, 1989:62).

The effects of living with protracted uncertainty are, for many at risk individuals, also reflected in educational, employment, marital, and reproductive decision-making. Living their lives as if they will almost certainly get HD, many at risk individuals adopt personal philosophies of “living from day to day” or “for the moment” (Korer & Fitzsimmons, 1987:533). Some forego opportunities to have a career or family while others never pursue an education or are unable to make long term financial or other commitments. These consequences of living at risk for HD ripple through the generations affecting the life-chances and decisions of those at 25% as well as 50% risk for the disease.

In their comparison of youth at 50% risk versus 25% risk, Korer and Fitzsimmons (1985) conclude that even those who are cushioned by being one generation removed from HD are still in a highly tenuous position. Given that one parent is at 50% risk and has grown up with all of the uncertainties and anxieties which the condition engenders, a strong desire to maintain family security may over-ride candid discussion of HD. In consequence, a high premium may be placed on secrecy and parents may avoid the onerous task of confronting children with their at risk status. Knowledge withheld may, however, backfire when those at risk and/or their well spouses lash out against those who have apparently failed to share information in a timely fashion (Yale & Martindale, 1984).

In summary, the literature on the familial dynamics of coping with, and living at risk for HD attests to several key aspects of this disease: 1) insidious first symptoms and a delayed age of onset create the potential for chronic stress; 2) onset may lead affected individuals and/or
family members to adopt strategies of denial and/or repression in the effort to manage a frightening reality; 3) lay beliefs about the genetics of HD are often at odds with Mendelian theories of inheritance; 4) the eventual diagnosis of an affected family member often prompts new awareness of the disease and its hereditary nature amongst other family members who are themselves at risk; and 5) factors such as socioeconomic status and the sex of the affected parent have significant implications for how families experience and cope with the disease.

One of the key ingredients in all of this appears to be chronic uncertainty. Several studies (Bloch, et al., 1993; Kessler & Bloch, 1989) have addressed the various strategies employed by at risk individuals and families who seek to manage uncertainty within their everyday lives but, with the advent of predictive testing, research has focused increasingly on the utility of testing as a means of resolving uncertainty and/or engaging in rational decision-making.

**Predictive Testing: From If to How**

In 1986, genetics centres in the U.S., Canada and the U.K. began to offer the linkage test to those at 50% risk for HD. This test provided individuals with either an increased or decreased risk outcome. Given serious concerns about the immediate and long term psychosocial well-being of predictive testing participants, linkage testing was, however, initially provided only within a research context. Required components of the research protocol included extensive psychosocial assessment, a series of counselling and follow-up sessions as well as a protracted period of time in which the predictive testing candidate could reconsider or withdraw from the predictive testing program (Benjamin, et al., 1994; Fox, et al., 1989).

To be fully informative, linkage testing requires a well documented family history and DNA samples from multiple family members — both affected and unaffected. Consequently, linkage testing demands the co-operation of a significant number of people. Not infrequently necessary DNA samples may be unavailable — either because significant family members are deceased or do not wish to participate. Additionally, there is also the possibility that the available family history and DNA samples will yield an uninformative result. With the (1993) discovery of the gene for HD, it became possible to provide at risk individuals with a direct type of presymptomatic test (Huntington's Disease Collaborative Research Group, 1993). In contrast
with the probabilistic outcomes of linkage testing, the direct test provides a definitive result.

The shift from linkage to definitive testing — which occurred in the fall of 1993 — has had immediate implications for how testing is conducted: with a reduced need to involve additional family members, those at risk are able to obtain information about their HD status somewhat more quickly and discreetly. Worried about the impact that this change in predictive testing could have for those who learn that they have inherited the gene, Wexler (1990:90) argued prior to the cloning of the gene for HD that the long and drawn-out process of linkage testing was “a brake on precipitous action and allows a client to live for some time experimenting with both positive and negative outcomes.” With no treatment for HD yet in sight, the potentially increased availability and ease of presymptomatic testing may not allow those at risk sufficient time to “delve through all of the layers of defenses that people at risk have built over time to enable them to cope and allow them to feel the emotional reality of a negative or positive test result”. In addition, numerous commentators have pointed out that the increased accuracy and certainty of the direct test imposes a new burden of responsibility on clinicians and researchers (Mattson & Almqvist, 1991). These concerns have not been raised in isolation although they are perhaps indicative of a profound shift in debates around predictive testing for HD — that is, from the question of whether or not to offer predictive testing to how best to continue offering it.

Attitudes Toward and Participation in Predictive Testing

Presymptomatic (or predictive) testing for HD has been available for more than a decade. It is now offered as a clinical service in many countries and is no longer considered to be highly controversial; indeed some argue that the clinical protocol for predictive testing ought to serve as a model for how to offer genetic testing for other late onset disorders (Quaid, et al., 1996). Nonetheless, many fewer at risk individuals than originally estimated have chosen to participate in predictive testing for HD and much of the literature now reflects the view that the rate of uptake will probably not show a demonstrable increase unless, or until, effective methods of treatment and/or prevention are available (Babul, et al., 1993).

Surveys of the at risk population conducted prior to the introduction of presymptomatic
testing indicated that anywhere from 66% to 79% of at risk individuals would request such a test when it became available (Kessler, et al., 1987; Mastromauro, Myers, & Berkman, 1987). However, after eight years of offering predictive testing in Greater Vancouver, a region where people are well-informed about the test, only an estimated 20% of at risk persons had participated in such testing (Babul, et al., 1993). In the two years following the advent of direct testing in the fall of 1993, approximately 350 at risk individuals entered the PT program in fourteen Canadian centres offering the test (Adam, 1995).

Prenatal forms of testing for HD have been even less widely utilized. In Canada, the prenatal test was requested by only seven of thirty-eight eligible candidates (or 18%) in the years prior to the introduction of direct testing. This is a much lower rate of uptake than the anticipated 32–65% derived from early survey data (Adam, et al., 1993). The discrepancy between attitudes toward the prenatal test and actual uptake is, however, not surprising given the general tendency of most pre-test surveys to forecast comparatively high levels of uptake for both linkage and direct forms of predictive testing.

Interestingly, the ratio of “increased” to “decreased” risk outcomes has not changed with the advent of the direct test — for every at risk individual who learns that they have inherited the genetic mutation associated with HD there are two who learn that they have not (Adam, et al., 1995). The primary explanation for this phenomenon derives from the fact that many PT candidates wait until they are past the typical age of onset for their family and hence, the odds of having inherited the genetic mutation for HD have declined (Bloch, et al., 1989).

Assessment of the demographic characteristics of the Canadian cohort of participants who received predictive testing results before 1990 reveals that there were twice as many female

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6 Elsewhere, participation has typically been even lower: only 5% of those at risk have been tested in the six countries with the longest established testing programs. In all, 1479 presymptomatic tests for HD were undertaken world-wide by the end of 1991. Of these, Canadian centres offering predictive testing accounted for 292 results (World Federation of Neurology & International Huntington Association, 1990).

7 There were, during this period, an average of three new predictive test participants seen at the local medical genetics clinic each month. This rate of uptake remains fairly constant although the waiting list is now shorter than it was during the period of recruitment for this research (Kaurah, 1998).

8 Where informative, linkage testing provided a result which was expressed as a probability. The assigned probability was higher or lower than the 50% apriori risk of most PT candidates; hence risk was “increased” or “decreased.” With direct testing, however, the result is no longer expressed as a probability; PT candidates are told that they have or have not inherited the genetic mutation associated with HD (or, in a small number of cases, that they have an intermediate number of repeats). For the sake of comparison, however, I continue to use the terms “increased” and “decreased” risk.
participants as male (Wiggins, et al., 1992). This differential is reported elsewhere (Meissen &
that women’s more intimate involvement with reproduction and child-rearing coupled with “the
apparently greater capacity of men to deny their feelings and avoid looking at the painful
implications of their situation” may help to explain the differential participation of women and
men in predictive testing. This clearly requires further research but it is, nonetheless, consistent
with the results of a Dutch study which assessed the pretest attitudes and expectations of
predictive test candidates and their partners. Reporting on prior feelings about test outcome,
Tibben et al (1993b:13) note that only four male (16%) as opposed to nineteen female (42%)
participants anticipated an increased risk result and that males more often than females either
expressed no opinion or thought that their risk would decrease.

Understandably, many people at risk for HD share feelings of trepidation about the
possibility of learning with near 100% certainty that they will develop an incurable degenerative
disease.9 Indeed, North American studies assessing the reasons for choosing not to be tested for
HD indicate that the most important factors include: the burden of having knowledge of the
increased risk of one’s children if one is found to carry the gene; the absence of an effective
cure; the potential loss of health insurance and other related financial implications, and; the
inability to “undo” the knowledge offered by predictive testing (Quaid & Morris, 1993).
Alternatively, the most commonly expressed reasons for testing include relief of uncertainty,
general planning for the future, specific planning in marital, reproductive,10 career or financial
areas, and the perceived responsibility to provide information to children (Bloch, et al., 1989).

Partners and companions of individuals requesting predictive testing diverge from at risk
individuals on the most important reasons for requesting predictive testing. Meissen et al (1991)

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9 In this respect, there are several important parallels between testing for HD and testing for HIV. Both deliver
information which is potentially devastating and, although not everyone testing positive for HIV will eventually
develop AIDS, the diagnosis brings fear, uncertainty and the expectation of a severely reduced lifespan. It may also
lead to social stigma and isolation. Up until fairly recently, AIDS organizations (such as the U.S. based Gay Men’s
Health Coalition) did not actively encourage HIV testing as long as people took adequate protectionary measures.
However, attitudes have now shifted as new options for treatment are becoming available.

10 Tibben et al (1993b) report that in the Dutch study mentioned above family planning was the most frequently
cited reason for testing. This may be an artifact of a somewhat younger age cohort than is present in other studies
and/or it may also be indicative of a comparative absence of worry related to obtaining adequate longterm health-
care in the Netherlands.
report that clarifying the risk of children was the most important motivation for partners while Evers-Kiebooms et al. (1989:38) found that partners tend to be concerned to the same extent about children and the consequences of HD for the relationship — at risk individuals, on the other hand, are more immediately concerned with their own future. Further, while there does not appear to be any discernable relationship between sociodemographic variables (sex, age, marital status, educational level and presence of children) and reasons for or against predictive testing, two studies report an association between age of onset in the affected parent and the strength of test participants’ stated intentions to take the test — the younger the age of onset in the affected parent, the stronger the expressed intention to proceed with the test (Evers-Kiebooms, et al., 1989; Markel, Young, & Penney, 1987). Presumably this is because potential onset of the disease is perceived to be fairly imminent in such cases.

Clinical observations and anecdotal reports suggest that test participants who have only recently learned that they are at risk for the disease often display a high degree of resolve about proceeding with the test. Many do, however, change their minds once they have adjusted to their new status and given more thought to the potential impact of the test (Tyler, Ball, & Caufurd, 1992). Although this has not, to my knowledge, been investigated with a large enough sample to draw any firm conclusions, the suggestion that people who have only recently learned of their risk for HD wish to dispense quickly with what is a new source of anxiety makes intuitive sense and fits within existing frameworks for understanding at risk individuals’ reasons for testing. It should be noted, however, that this situation may impose an additional burden on genetic counsellors; individuals who have only recently learned of their at risk status are often unfamiliar with the disease and may be resistant to learning enough about it to ensure that their consent to predictive testing is both meaningful and fully informed. The timing of testing is, therefore, significant and clinicians may find it prudent to allow considerable time to elapse between the diagnosis of a parent and the testing of any offspring (Tyler, Ball, & Caufurd, 1992). Further, proximity to the death of a relative should, according a recent position paper of the National Society of Genetic Counselors, be considered as another salient reason to defer

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11 These observations are based on personal communication with several genetic counsellors working at the UBC clinic during the period of this research.
testing. As this position paper notes, “many people need time to ‘tell their stories’ and derive therapeutic benefit from this experience” (McKinnon, et al., 1997).

Psychosocial and Familial Impacts of Predictive Testing for HD

Presymptomatic genetic testing of adults at 50% risk for HD was initially offered in several locations\(^{12}\) as part of a research program designed to determine: 1) the psychological consequences of informing people of their HD risk status; 2) whether or not it is possible to predict who will adapt well to their status, and; 3) whether educational and counselling interventions could alleviate depression and prevent suicide amongst those who learned that they were at increased risk for HD. In addition to being eighteen years of age or older, candidates for predictive testing were required to be free of major psychiatric disorders and/or serious suicidal risk at the time of testing. All candidates were required to undergo a physical examination and participate in pre and post-test counselling (Fox, et al., 1989).

Early reports describing the psychological outcomes of linkage testing offered preliminary support for the finding that people are able to cope well in a clinical environment “that includes education, pretest counselling, psychological support, and regular follow-up” (Brandt, et al., 1989:3108).\(^{13}\) Initial study cohorts were small\(^{14}\) and had not been followed long enough to ascertain anything more than the short term consequences of predictive testing but subsequent reports on the psychosocial well-being of larger cohorts followed for a longer period of time have largely substantiated the early findings described in Brandt et al (1989).

In one of the most widely cited papers on this issue, Wiggins et al (1992) reported on the first large-scale prospective evaluation of the psychological effects of predictive testing for

\(^{12}\) The first genetic centres to offer predictive testing were in Vancouver, British Columbia and Baltimore, Maryland.

\(^{13}\) Interestingly, Brandt et al (1989:3108) highlight the fact that in a pretest evaluation of subjective feelings about risk, many of those who ultimately tested negative for the Huntington’s marker had attached a lower subjective value to their risk status than those who ultimately tested positive. The authors noted that while this was not statistically significant, it did suggest that “the at risk participants and their close family members may have some covert awareness of symptoms that are not detectable by the most sensitive of clinical testing procedures.”

\(^{14}\) In 1989, Brandt et al reported on 55 people tested for the presence of the Huntington’s marker in the Johns Hopkins’ program—12 yielded positive results, 30 were negative and 13 were “uninformative” or inconclusive. And, in the same year, Fox et al (1989) cautiously reported that although 95 participants were enrolled in the pilot project at the University of British Columbia the short and long-term effects of predictive testing remained uncertain since relatively few individuals had received informative results and completed follow-up.
Reporting on behalf of the fourteen genetic centres which comprise the Canadian Collaborative Group, Wiggins et al (1992:1404–5) concluded that,

Knowing the result of the predictive test, even if it indicates an increased risk, reduces uncertainty and provides an opportunity for appropriate planning. Therefore, as our findings suggest, people who receive informative results, regardless of the content, may derive psychological benefits not experienced by those who remain uncertain. (emphasis added)

Published in the New England Journal of Medicine, this paper enjoyed a wide circulation and elicited a flood of media interest in the phenomenon of predictive testing. This had a positive impact on public awareness of HD (Tolley, 1998). Less obvious at the time, however, responses to the NEJM paper also signaled a watershed in thinking about predictive testing; clinicians and service providers would henceforth remain vigilant about monitoring the psychological effects of predictive testing but protracted concerns about the potentially catastrophic consequences of predictive testing would now have to share the limelight with evidence which suggested the potential for psychological benefits. This shift in thinking had the further effect of helping to defuse an ethical debate which was, in the wake of rapid technological advances and reassuring research findings, already changing its focus if not winding down.

Predictive testing for HD had not resulted in the catastrophic events which were widely feared when programs for offering the test were first being developed and over time, confidence in the potential benefits of predictive testing has begun to escalate. For instance, Alice Wexler (1996) recently observed that the conclusions of Wiggins et al’s 1992 paper have been considerably overstated in Lerman et al’s 1996 study of testing for the BRCA1 mutation. Lerman et al erroneously cited “the long-term psychological benefits” of testing and, in Wexler’s opinion, glossed over the adverse psychological responses to testing which the Canadian group had described in a separately published paper. Lerman (1996:1140) responded to Wexler’s concerns with a short statement acknowledging the need for continued caution, long term study, counselling and targeted support.

15 This study included 135 participants in the Canadian program: 37 received an increased risk, 58 received a decreased risk and another 40 did not receive results either because the test was uninformative or they did not want to take the test. Data from this latter group was used for comparative purposes.
As we have pointed out, examination of average responses in study groups may obscure important individual differences in functioning. Continued investigation is needed to examine the long-term outcomes of genetic testing for heritable diseases and to identify predictors of individual responses to disclosure of genetic information. Such information would be useful to identify the subset of individuals who may be more vulnerable to adverse effects and to target enhanced counselling to them.

The above statement could be the conclusion to any number of current papers on predictive testing for adult onset disorders. Lerman et al are hardly alone in advocating the need for continued careful research on the psychological effects of predictive testing for heritable diseases such as breast and/or ovarian cancer and HD. This theme is ubiquitous in the literature but it probably cannot be overstated. Indeed, after eight years of studying the effects of linkage testing and two years of offering the direct test, Hayden et al (1995:210) concluded that "little is known concerning the effect of a completely accurate test result on the psychological status of individuals at risk for genetic illness."

Alternatively, widespread recognition of the need to evaluate the social factors which shape and give meaning to the experience of predictive testing is noticeably absent from the increasingly voluminous literature on genetic testing. Moreover, few published studies consciously reflect an awareness that clinical research itself contributes to the social construction and immediate implications of predictive genetic testing. As Boutté (1988) has argued, the dominant medical discourse of genetic screening and predictive testing is the "language of potential benefits." This discourse affirms that prediction of genetic disorders resolves uncertainty and enables informed decision-making. In a social climate which values certainty and places a high premium on rational planning, it is perhaps not surprising that individuals undergoing predictive testing are able to cope well when they "have the facts" and are given extensive counselling, follow-up and support. Knowledge provides the opportunity to make choices and informed decisions: for instance, knowing that a family member carries the gene for HD means that preparations can be made to compensate for lost earnings or provide for long term care. Further, the alleviation of uncertainty which predictive testing provides may have an existential as well as rational-purposive value.

Nonetheless, there are costs as well benefits to predictive testing (Codori & Brandt, 1994a). For those who learn that they have inherited the gene, there is a new certainty that the
disease will eventually manifest itself and this may be reflected in increased practices of symptom-searching or other forms of self-surveillance. As one woman who learned several years ago that she has the gene for HD put it, "I have gone from if to when." Close to the age at which her mother experienced onset, this woman also described the anxious uncertainty of waiting for symptoms to appear or, more pointedly, the periodic crises she experiences when she suspects that her husband is aware of symptomatic behaviour of which she is unaware. ¹⁷ For those who have their own biological offspring, there may also be a heightened sense of guilt about the risk of having transmitted the gene to the next generation and, paradoxically, a worry that their children will also experience this guilt and refrain from having children of their own.

For approximately two-thirds of test participants, the news is ostensibly "good." However, contrary to what common-sense might seem to dictate, there are also costs attached to receiving news of a decreased risk for HD. What might be regarded as "good news" does not always provide a feeling of relief nor, in some cases, will it be easily incorporated into at risk individuals' and families' day to day lives (Huggins, et al., 1992; 1993a; Tibben, et al., 1993c). Some individuals who learn that they have escaped the family disease also report that they no longer feel as if they are "part of the club" (Burgess, submitted) while those who have siblings or other family members affected with HD experience a relief that is tempered by sadness and a pervasive sense of "survivor's guilt."

Of particular salience to current studies on the familial impacts of predictive testing, the finding of adverse impacts arising from a decreased risk illustrates cogently the need to situate individual psychosocial well-being within the nexus of family beliefs and dynamics (Chapman, 1993; Hayes, 1992). Patient pre-selection and survivor's guilt are but two phenomena which indicate the need to adopt a social rather than individual perspective in assessing the impact of the at risk individual's test results. Furthermore, many researchers now recognize the urgent need to assess the psychosocial impacts of predictive testing for the participant's spouse or partner and other family members (Evers-Kiebooms, et al., 1989; Kessler, 1993a; 1993b; Quaid & Wesson, 1995; Tibben, et al., 1993b; 1993c). Noting the profound importance of not

¹⁷ These observations are drawn from fieldnotes written after I took part in a workshop for individuals at increased risk for HD. This workshop was held at the 1996 Huntington Society of Canada Conference.
overlooking the impact of predictive testing for the unaffected spouse, Kessler (1993a:148) argues that,

The genetic etiology of HD tends to differentiate it from most other chronic illnesses...the way spouses of HD patients tend to experience the guilt generated by its hereditary nature influences and affects their thinking, behaviour and decision-making to an extent and in ways often not seen among spouses of individuals with other chronic illnesses and disorders.

In one of a small number of studies which addresses the familial impacts of predictive testing, Tibben et al (1993c) report that at risk individuals who were informed that they carry the HD gene indicated that “the result had affected their family more than they had previously expected”. In addition, partners of those who learned that they were at increased risk expressed greater concern about a future overshadowed by HD than did gene carriers themselves.

Noting that partners often have particular insights into the likely outcome of predictive testing, its impact on marital stability, and its potential implications for future caregiving, Quaid et al (1995) found that partners of at risk individuals entering predictive testing suffered greater depressive symptomatology than those at risk. Stressing also that those couples who completed testing (N=19) expressed greater satisfaction with their relationship than those who withdrew after the first counselling session (N=6), Quaid et al provide important corroborative evidence for other studies (Codori & Brandt, 1994b) which suggest that the people who are most likely to succumb to morbid reactions are simply not being tested. As such, favourable psychological reactions to predictive testing may be due to the self-selection of participants who believe themselves to be well-equipped to handle “bad news” (Kessler, 1994). Further, although several key studies (Quaid & Wesson, 1995; Wiggins, et al., 1992) mention significant rates of sample drop-out, there is insufficient data available on why these individuals were unavailable for follow-up. Underscoring the importance of this for the phenomenon of self-selecting samples, Quaid et al (1995:50) suggest that “those individuals who are coping the worst are also those who are unlikely to contribute follow-up data”.

Longitudinal studies of the familial as well as individual impacts of predictive testing will begin to fill in these gaps but, as recent discussions of self-selection indicate, there is also an immediate need to broaden existing research strategies in order to understand the responses and coping abilities of those who are least likely to follow through with post-results surveys or
counselling. Gaining access to what these individuals and their families have to say is, however, problematic since avoidance of HD and HD related issues often constitutes one major strategy for coping with test results (Tibben, et al., 1993c).

Disclosure of Predictive Test Results

Few studies of predictive testing for HD have yet addressed the relationship between PT candidates’ willingness to disclose test results to selected family and/or friends and their ability to adopt effective coping strategies or secure adequate levels of social support. All the same, a positive correlation between disclosure (to selected family and/or friends) and psychosocial well-being is implicit in the perspectives of many service-providers as well as the predictive testing protocol itself (Benjamin, et al., 1994). The link between these factors is also strongly supported in much of the literature on stress and coping, family therapy, and social work (Karpel, 1980; Pearlin, 1989; Schild & Black, 1984).

One of the few explicit treatments of patterns of disclosure is found in Tibben et al’s (1993b; 1993c) study of PT candidates and their spouse/partners’ overall experiences with the predictive test. In this study Tibben et al used qualitative and quantitative methods to assess pre and post test attitudes as well as impacts of the test on gene carriers versus non-carriers. Finding that 31% (N=70) of PT candidates did not discuss the decision to take the test with sibs, the authors report that pretest fears of stigmatization were a common reason. In addition, some PT candidates expressed the fear of feeling responsible should their disclosure lead to a sibling receiving an unfavourable result after being induced to take the test. Such fears were minimized by a decreased risk result and a full 92% of those with this outcome (N=39) discussed their results with sibs. In contrast, 26% of those with an increased risk (N=24) did not discuss this result or their prior decision to have the test with sibs. The primary reason given was the prevention of unwanted sympathy and the avoidance of being “in the family’s limelight”. Most were, however, “fully aware of deliberately keeping their test participation a secret” (Tibben, et al., 1993b:15).

Also of considerable interest is the finding that 20% of all PT candidates and their partners indicated a “lack of confidence or an inability to feel comfortable either with or when
discussing test results with others" (Tibben, et al., 1993c:106). This finding holds for both increased and decreased risk outcomes but is perhaps most poignant for the partners of those given an increased risk result. Nearly half of the partners reported that they missed the support of others with whom they could share their feelings. Talking about their spouse to a third party may seem like a betrayal and yet strategies of non-disclosure and avoidance do, as the authors caution, jeopardize the ability to resolve emergent problems and adequately plan for the future.

Disclosure practices have not, as mentioned above, been extensively studied. Nor is the wider literature on genetic testing and prenatal screening of much assistance when it comes to ascertaining a sense of when and how PT candidates share information about their test results with family and friends. One recent study (Eng, et al., 1997) evaluating the educational and counselling components of prenatal carrier testing for three genetic disorders (Tay-Sach's, Cystic fibrosis and Gaucher's disease) reports under the heading of “privacy issues” that test participants were more likely to share information with their family and physicians than they were with friends, although the likelihood of telling physicians and family was greater for women than men, as was the likelihood of telling friends.

The absence of studies in this area is in some respects puzzling. With respect to predictive testing for HD, clinicians and genetic counsellors routinely inquire about PT candidates’ plans to disclose their test results to other members of the family, friends and associates, the family doctor, and/or their employer; many also consider it important to know about the PT candidate’s social support network. Such knowledge has obvious clinical implications — for instance, the counsellor who sees several members of the same family is well advised to know about any family members who have not yet been informed of the test candidate’s intent to undergo predictive testing. Likewise, it may be extremely important for the counsellor to know which of the PT candidate’s family and friends can be relied on to provide support and companionship in times of crisis or depression. Test candidates who have an inordinate fear of disclosing their test results to significant others may require additional counselling and/or support and, those who appear to be somewhat cavalier in their attitude toward disclosure may need to discuss more thoroughly the implications of this for other members of the family as well as themselves.
Although these issues have for some time provided a strong clinical rationale for studying familial patterns of communication about HD, it has only been since the advent of direct testing that the area seems to have generated substantive new interest. Two particular issues support this contention; both have to do with the ways in which at risk individuals and their families talk about and attribute meaning to the genetic mutation associated with HD.

First, the advent of direct testing has raised the controversial issue of whether or not clinicians should disclose to test participants the number of CAG repeats found in the HD gene during laboratory analysis. Reluctance to disclose such information derives from the concern that test participants who learned that they had inherited the HD gene might misinterpret the significance of their CAG repeat number and erroneously conclude that it is in some way predictive of their probable age of onset. Furthermore, clinicians worried that family members who had undergone predictive testing at different genetic centres might exacerbate such misunderstandings by comparing and/or inaccurately reporting their repeat lengths to other members of the family (Burgess & Hayden, 1996).

Second, genetic testing for HD is now used in two distinct ways: in the absence of signs and symptoms, testing is predictive while in the presence of signs and symptoms, testing may be used to confirm a clinical diagnosis. Where clinical examination and/or the lack of a documented family history of HD leave room for diagnostic uncertainty, DNA analysis may now be utilized to confirm a suspected diagnosis of HD. While this practice may help to eliminate inaccurate diagnoses it also highlights the problematic relationship between phenotype and genotype.

When does an hereditary disease “begin”? At the moment of conception? With knowledge that one is at increased risk? Once symptoms are undeniable? At the stage of diagnosis? It might be argued that such questions are really a matter of semantics; if we cautiously choose the right words to differentiate carefully between predictive testing and clinical diagnosis or having the gene and actual onset of the disease, confusion and needless

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18 Policies on disclosure of repeat length vary; some centres acknowledge the PT candidate’s right to have this information and therefore have a policy of always disclosing repeat length. Others, who have a policy of never disclosing repeat length, simply inform the test participant that they have or have not inherited the genetic mutation associated with HD.
anxiety can be avoided. Alternatively, it could equally well be argued that there are, at present, a number of scientific, medical and lay discourses on genetic risk, each of which conceptualizes and expresses the relationship between genotype and phenotype rather differently.

For those who learn that they have inherited the gene for HD, the distinction between having the gene and having onset of the disease is vital. Indeed, clinicians and service providers stress that it is highly reassuring to those individuals who know that they have inherited the mutation associated with HD to hear that they are not yet demonstrating any clinical signs or symptoms of the disease (Benjamin, et al., 1994; Tyler, Ball, & Caufurd, 1992; Wexler, 1992b). Yet, despite the significant clinical implications of conflating the presence of the gene with the outward manifestation of disease, this kind of slippage occurs even within the scientific and medical discourse on genetics. For instance, medical geneticist Patricia Baird (1995:161) argues that for some single gene diseases, the relationship of a genotype to the actual disease is very clear.

...in such Mendelian disorders, having a particular gene inevitably means that the disease will be present: for practical purposes having the gene means having the disease. (emphasis added)

Baird made this statement within the context of an important argument about the dangers of using Mendelian genetics to think about much more complex polygenic and multifactorial disorders. Nonetheless, the slippage which occurs as she moves from talking about genes as signifiers of future disease to genes as signifiers of actual disease is all the more significant given her otherwise exceedingly careful analysis. If, for “practical purposes,” we neglect the temporal dimension and accept that “having the gene means having the disease”, then it follows that the disease is present from the moment of conception onward. Further, those who are at 50% risk for having the gene also seem to have some sort of illness. As Hayden (cited in Bishop & Waldholz, 1990:278) observes,

People argue that a 50 percent at risk child also has a 50 percent chance of not having the gene and living a full and normal life...But we have people here who have lived a sort of hell all their lives being at 50 percent risk. They don’t want their children to live with the same uncertainty. I think for some people the 50 percent risk is itself an illness they don’t want to pass on to their children.

The idea that risk is itself an illness may be understood and/or interpreted in several ways. Most immediately, the experience of living at risk for HD often creates a profound sense
of anxiety which may pervade and even overwhelm routine aspects of daily life. In its ubiquity, the threat of HD becomes inextricably interwoven with HD; chronic stress involuntarily animates domestic life just as chorea involuntarily animates the bodies of its sufferers. As such, no-one in an HD family really escapes the disease. Referring to the large HD kindred which she studied in Venezuela, Nancy Wexler (1984:134–5) notes that,

They say that every offspring of an Huntington’s disease parent has inherited the disorder but that only certain individuals will get sick. This distortion of genetic fact accurately expresses an emotional truth: Those at risk for Huntington’s disease have inherited some constant potential to manifest the disease and in that sense are not entirely normal, but they may or may not become ill depending on various circumstances. To inherit Huntington’s disease is for the Venezuelans a separate and unique state apart from having Huntington’s disease. At risk individuals inherit it; affected individuals have it. (emphasis added)

As these brief examples illustrate, there are at present a variety of lay and scientific ways of framing the relationship between phenotype and genotype. In some cases, shorthand ways of describing the implications of Mendelian disorders conflate the disease with its genetic marker (Baird, 1995); in others, however, it is apparent that the social experience of being at risk is itself becoming constructed as a form of illness which may have psychosocial and/or familial implications (Bishop & Waldholz, 1990; Wexler, 1984).

The emerging social construction of genetic risk as a form of illness cogently illustrates the need to move beyond the confines of the biomedical model in attempting to understand the implications of predictive genetic testing. In the effort to do so, it is perhaps inevitable that diverse research traditions and approaches will sometimes come into conflict. Reflecting on the potential of this conflict to generate new insights and understandings, Alice Wexler (1995:xxv) has proposed that Huntington Disease may “serve as a space where many discourses collide and therefore help [to] make visible the hidden stakes in [the new genetic] contest for human survival and identity in which all of us are at risk.”

Limitations of Existing Research

Thus far, I have selectively reviewed the literature on HD and predictive testing, pointing the strengths and weakness of existing studies. Although it has, for obvious clinical reasons, been critical to establish the individual psychosocial impacts of predictive testing, the important task of situating these individual impacts within their familial and social contexts has,
overall, received comparatively little attention.

Useful as existing studies are in providing basic information about individual attitudes toward and reasons for predictive testing, the relationship between test results and individual short and longterm psychosocial well-being and, the clinical protocols best suited to appropriate delivery of predictive testing, most of these studies reveal little about the social meanings and everyday lived experiences of predictive testing for HD. Moreover, the few studies that have focused on the perspectives of those at risk adopt an individualistic approach which fails to examine the reciprocal impacts of predictive and diagnostic genetic information on the immediate family and/or social support network. Quantitative approaches and, in particular survey methods, have been almost universally favoured and hence, existing studies often do little more than revisit issues which have already been recognized as having some importance. In addition, test results constitute the primary independent variable and as such, there is an overall neglect of gender, race/ethnicity, class and other factors which shape and differentiate the predictive testing experience. Finally, few studies acknowledge the degree to which research and service protocols themselves shape the experience of predictive testing and fewer still consciously reflect on how knowledge about predictive testing is socially constructed, whom it is being constructed for and, with what consequences.

I began this chapter by noting that the predominant clinical construction is that predictive testing offers “a gift of knowing.” This construction is supported by a “discourse of potential benefits” (Boutté, 1988) which emphasizes that no matter what the outcome of the test, PT candidates have the opportunity to live their lives and/or plan for the future in accordance with this knowledge. In consequence, the debate around whether or not to offer predictive testing is transformed into the more practically–oriented task of how best to offer predictive testing — that is, how to maximize the potential benefits and minimize the potential harms. This focus is oriented toward the production of desirable, measurable outcomes and away from the messy conceptual issues of how the knowledge offered through predictive testing acquires a particular historically–situated social value and moral significance.

The discourse of potential benefits and the metaphor of the gift of knowing are part of a larger metanarrative about the value of information in an increasingly risk–oriented society.
This metanarrative offers a particular schema for organizing the contemporary experience of uncertainty and attaches a particular moral worth to information which facilitates rational planning and, at least the appearance of, choice. This metanarrative is not the only available means of conceptualizing the experience of predictive testing but it does tend to elide other critical and potentially more insightful ways of framing the experience.

Even if we view human agency through the narrow lens of rational choice, the metaphor of the “gift” is inherently problematic. Gifts are given within the context of a social relationship and, particularly where there is an asymmetry of power between the gift-giver and the recipient, gift-giving is neither purely benevolent nor purely self-interested. As Mauss (1965 [1954]) pointed out in his classic essay on reciprocity,

Our terms 'present' and 'gift' do not have precise meanings...It is a complex notion that inspires the...actions we have described, a notion neither of purely free and gratuitous prestations, nor of purely interested and utilitarian production and exchange; it is a kind of hybrid.

If predictive testing is a “gift of knowing” that is given by service providers to PT candidates, it is also a gift that is given within a formalized clinical exchange. Just as PT candidates receive the “gift of knowing” whether or not they have inherited the genetic mutation associated with HD, researchers receive a reciprocal gift which is given by PT candidates and their families — that is, the DNA samples, questionnaires, counselling sessions, follow-up consultations and interviews which constitute the life-blood of social and scientific research, clinical evaluation, policy development and analysis. The collection of such data is a routine element of predictive testing; standardized clinical as well as research protocols require that PT candidates contribute DNA samples, complete a specified amount of counselling and undergo clinical assessment before they receive any test results. The value of such data is not, however, restricted to its utility in managing the individual case. Such data are essential to the social production of scientific and technical knowledge about HD and predictive testing: as such it is to be hoped that it will ultimately benefit numerous others at risk for, or affected with, HD.19

19 Banked DNA, in particular, has been essential to scientific researchers who seek to elucidate the molecular basis of HD and many remain optimistic that such research will eventually result in an effective treatment (if not cure) for the disease. Those who seek to develop sound protocols and methods of obtaining consent for the banking of human DNA do, however, acknowledge, that DNA is neither “person” nor “thing”; as such, Knoppers (1996:175) argues that “it may be more respectful of its unique status and qualities to consider its use in genetic research as a gift—a gift conditional on the individual choices made.” Under the “conditional–gift” approach, there is no
Many of the limitations of existing research on predictive testing could be rectified through more extensive multi-disciplinary approaches for no matter how well-intentioned, research which is conducted by service-providers within a clinical context must contend with a number of unavoidable constraints. First, from the service providers' point of view, it is the individual predictive testing candidate, rather than the family, which is the “patient.” As such, service providers must uphold professional responsibilities for ensuring the predictive testing candidate’s autonomy and confidentiality. This focus on individual autonomy necessarily limits the range and scope of attention which service-providers are able to devote to understanding the impact of predictive testing on other members of the family. Second, even where clinicians and service-providers are able to delve more deeply into the sphere of family relations, predictive testing candidates may, past a certain point, find this to be overly intrusive (Burgess, submitted). Third, predictive testing candidates are aware of the gate-keeping role of genetic counsellors and other service providers who retain the prerogative to delay or indefinitely postpone the disclosure of test results should there be reason to believe that the candidate is unstable, severely depressed or in the midst of too many other personal or familial troubles. Practically speaking, such delays do not occur often and if they do, it is usually with the mutual consent and agreement of the PT candidate. Nonetheless it is the test candidate’s perception that such delays or indefinite postponements could occur which is at issue here and which may, as I wish to suggest, impinge upon what Goffman (1959) refers to as “the presentation of self” within various clinical interactions.

In summary, these constraints shape the clinical interactions which constitute the experience of predictive testing and, as such, they also have an immediate bearing on the type of data which can be gathered by service-providers within a clinical context. This is not to suggest that clinical research does not fulfill an crucial evaluative role; studies of the psychosocial well-being of PT candidates who receive an informative test result have made an important

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20 This is based on my own research observations and involvement in the weekly meetings of the local predictive testing research group. Burgess (submitted) confirms that this was also the case in another major Canadian centre offering predictive testing.
contribution to the literature and, no doubt, to understanding how to better offer the clinical
service of predictive testing. The point is that existing approaches to studying the psychosocial
implications and outcomes of predictive testing tend to overlook the way that the process and
meaning of predictive testing is itself socially constructed. Moreover, research that is based on
interactions that occur in a clinical setting tends to cast the PT candidate in the leading if not
solitary role of "proband" (i.e., the patient) while other family members are the supporting caste.
This represents one perspective on the experience but it misses altogether the possibility that
there are other protagonists waiting in the wings; other sequences of interaction that compete
with and sometimes confound resolution to the main plot.

This dissertation is an attempt to address some of these shortcomings and perhaps also,
to put the process of predictive testing into another context. For, if there is anything which
might serve as a conclusion to the foregoing review of existing research, it is this: there remains
a vast and as of yet unmet need to understand the experience of predictive testing from the
perspectives of at risk individuals and their families. This means that it is necessary to turn away
from clinical interactions as the central analytic focus (Conrad, 1990) and look instead at how
people manage hereditary risk within the context of their everyday lives. As Richards
(1993:578–9) suggests, "to understand the consequences of the new knowledge and techniques
that the new genetics are creating, we must study genetic conditions, and inheritance more
generally, as part of family beliefs and dynamics." In addition to knowing how information
about genetic risk is presented within the clinical context, there is an urgent need to understand
how and when at risk individuals and families communicate about predictive testing in their
everyday lives, what the implications of such patterns of communication are for various family
members' awareness of HD, and, how such familial awareness contexts shape, and are shaped
by, the perception or fear of stigmatization and/or discrimination. These neglected aspects of
research on predictive testing for HD are beyond the purview of most clinically–based research;
they demand a different starting point and an alternative approach to eliciting participants' accounts of their experience.
CHAPTER IV
HOW THE PROBLEM (RE)PRESENTS ITSELF

Problems are not given. They are constructed by human beings in their attempts to make sense of complex and troubling situations. Ways of describing problems move into and out of good currency... New descriptions of problems tend not to spring from the solutions of the problem earlier set, but to evolve independently as new features of situations come into prominence (Schón, 1993:144).

Huntington Disease is a source of "trouble" (Zola, 1972a) and so too is the problem of how to talk about the troubles it creates. This dissertation is about these troubles: it is about the problems encountered by families at risk for HD and the ways that families communicate about these problems. It is, however, also about the stories that people tell about their communicative interactions.

I have not come to this awareness of my central research problem easily. In the initial stages of my research, I framed the problem in fairly straightforward terms: I would do an ethnographic study of "who says what to whom, when and where" and write a "realist tale" (Van Maanen, 1988) about what I "discovered" in the field. Though there would have to be a confessional element that revealed something of the subjective experience of being in the field — its dilemmas, problematics and uncertainties — I could, I believed, proceed with my inquiry by locating and describing various instances of familial communication as they were reported to me by study participants. The patterns in these communications would, I thought, emerge if I was systematic in my elicitation of participants' accounts and thorough in my treatment of the data. With this framework in place, I would then look at the how and why of these patterned interactions: I would describe how PT candidates and their families engaged in disclosure, their reasons for doing so (or not) and the consequences of these disclosures and/or nondisclosures.

I now find this framing of the problem naive and strangely lop-sided. In emphasizing the content of who says what to whom, it suggests a referential view of language and a sender—receiver model of communication. As a result, it does little to facilitate understanding of how a wide range of human capacities for communication may be employed not just to transmit information but to produce and reproduce meaning, create and sustain social relationships. Nor
does my earlier framing of the problem adequately acknowledge the depth of lay knowledge: it
neglects the understandings that communicants have of their own communication and,
moreover, it absolves the researcher (as external observer) from the need to realize her own
"hermeneutic participation" in the research (Krippendorf, 1994). For these and other reasons,
then, I have had to rethink my approach and allow the problem to re-present itself.

The stories that people tell about their acts of communication are, of course, also acts of communication. The interviews which inform my analysis are forms of communicative interaction which display many of the same linguistic and paralinguistic features as the communicative acts they are about but they do not exist in the same way as the events that they refer to. There are significant methodological differences between observing what happens when various family members talk (or avoid talk) with each other about HD, and, sitting down some time later with one or more family members to talk about the communicative interactions that I might have observed were it possible for me to do so in a non-intrusive manner. There is, as Bakhtin (1981) insists, a categorical boundary line between the actual world and the world represented in the story. This boundary line is, however, not absolute or impermeable.

However forcefully the real and represented world resist fusion, however immutable the presence of that categorical boundary line between them, they are nevertheless indissolubly tied up with each other and find themselves in continual mutual interaction; uninterrupted exchange goes on between them, similar to the uninterrupted exchange between living organisms and the environment that surrounds them (Bakhtin, 1981:254).

Thus, as I now see it, the problem lies less in determining how to pry apart differing levels of analysis than it does in figuring out how to understand each in and through its relation to the other. This relation is not static as much writing on narrative realism (versus constructivism) might suggest (Fay, 1996); it is, rather, "oscillating" (Wilden, 1987) in that it entails shifting back and forth between differing "moments" of interpretation. Each "moment" is part of "a two-way interactive mode of investigation" which involves the researcher and her study participants in discovering, and co-constructing, local worlds of meaning (Shorter, 1993).

1 Such oscillation is a trademark of the Dutch artist, M. C. Escher. Ingeniously crafted, Escher’s paintings offer a tangle of dimensions which trick the mind’s eye into shifting rapidly back and forth between competing perceptions of what is figure and what ground (Wilden, 1987).
Purpose and Outline of Chapter

In this and the next chapter I set out the theoretical and methodological context within which I have articulated my understanding of the central research problem of this dissertation. As mentioned above, I did not arrive at this understanding overnight. The unavoidably recursive nature of writing about people’s stories about their acts of communication was, for a time, a kind of vortex: it spun me around in circles until I no longer knew which reality I was attempting to study or whose story I was engaged in trying to write. On a good day, I accepted the blurriness in my thinking as an indicator that I was on the right track; on a bad day, the shifting back and forth between various levels of reality literally made me feel queasy.

At the most general level, my approach is informed by social constructionism and the critique of biomedicine’s tendency to treat diseases as objective entities. I am, however, more interested in the empirical application of this critical stance than I am in its abstract, theoretical implications. My emphasis is on the way in which social actors create and recreate meaning through ongoing social interaction thus I locate this research within the interactionist tradition. My framework for thinking about the significance of familial communication in shaping the experience of predictive testing has, from the start, been informed by Glaser and Strauss’s (1964; 1965) theory of awareness contexts as well as studies which have since offered some refinement to this theory (Armstrong, 1987b; Hutchinson, Leger-Krall, & Wilson, 1997; Seale, 1991; 1997; Timmermans, 1994). In addition, it is difficult to examine the management of personal information without referring to the concept of stigma (Goffman, 1963) and the processes in and through which stigma is both felt and enacted in the lives of those with chronic illness and/or disability (Davis, 1961; Scambler & Hopkins, 1986; Schneider & Conrad, 1980; Wendell, 1996).

My approach to the stories that people tell about their acts of communication is, however, oriented by somewhat separate literatures on the ethnography of communication (Gumperz & Hymes, 1986; Hymes, 1986; Saville-Troike, 1989) and the role of biography and narrative in shaping and representing illness experiences (Bury, 1982; Couser, 1997; Frank, 1995; Gordon & Paci, 1997; Kleinman, 1988; Mishler, 1984; Mishler, 1991). This literature is not incompatible with the literature on awareness contexts but it suggests a different line of
approach and invokes a different level of analysis. In what follows I review these somewhat disparate literatures, showing how they converge in the subject matter and approach of this dissertation.

Given that the theory of awareness contexts and much subsequent work on the experience of chronic illness originated within the social constructionist paradigm, I begin by reviewing the origins of this tradition. As we shall see, one of the hallmarks of social constructionism is its questioning of the taken for granted nature of reality.

**The Social Construction of Health and Illness**

Social constructionism is not a coherent theory or paradigm. This is not, however, to suggest that social constructionism does not have special meaning and application within medical sociology and the study of interpersonal communication about health and illness.

Social constructionist approaches emphasize the interactive rather than oppositional nature of the social and biological worlds as an unavoidable condition of human life (Wendell, 1996). Biomedicine has traditionally drawn a sharp distinction between the biological and the social. In taking for granted the independent existence of disease entities and the neutrality of biomedical knowledge, biomedicine renders the social meanings of health and illness, normality and pathology opaque (Mishler, 1981). Social constructionist approaches to health and illness offer an alternative to such taken for granted realities. Identifying as problematic the methods and content of medical practice and knowledge, social constructionism posits that “medical knowledge is a social product — not some privileged and asocial penetration of the workings of Nature” (Wright & Treacher, 1982:14–15). What counts as a biological fact is shaped by a range of socio–cultural as well as scientific and technological factors. This does not mean that such knowledge is unreal or spurious, that the activities of doctors are illegitimate or that diseases are imaginary. “Illnesses really do exist, but as sufferings which have no necessary, transhistorical, universal shape” (Wright & Treacher, 1982:14–15).

Biomedicine provides a powerful explanatory framework but it does so from the

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2 For instance, when lay beliefs about inheritance contradict scientific explanations, these alternative understandings are understood by biomedicine as myths or misunderstandings which are less than rational and therefore in need of correction (Richards, 1993). This perspective is evident in the resounding calls of some medical geneticists for an “attack on ignorance” and genetic illiteracy (Scriven, 1993).
perspective of a science which is concerned with explaining objectively the cause and effect relationships of abstract and seemingly asocial entities known as diseases. If we are concerned to understand the meaning of illness or hereditary risk in the lives of those who are affected by it, the experience of symptoms, predictive tests and/or diagnoses must be situated in their social, familial and cultural contexts. This involves more than just adding social and psychological factors to a biomedical model: it involves a fundamental challenge to the adequacy of the biomedical model as a useful depiction of what happens when a person becomes ill or a physician attempts to apply a set of diagnostic criteria for transforming symptoms into an identifiable disease (Engel, 1977).

Social constructionist approaches are, perhaps, nowhere more necessary than in the rapidly expanding field of predictive genetic testing. Even Huntington Disease, with its relatively straightforward genetics, acquires very distinct meanings for different individuals, families and cultures. Given that PT candidates are, in most cases, asymptomatic and, further, that the information derived from testing is not a diagnosis, it is important to ask how, in the absence of an appropriate popular discourse on hereditary and other forms of “embodied risk” (Kavanagh & Broom, 1998) lay actors adapt and modify culturally available frameworks in order to interpret hereditary risk in an intersubjectively meaningful way.

Disease, Sickness and Illness

Social scientists have long argued that there are important differences between biomedical understandings of disease, social perceptions of sickness and personal experiences of illness (Engel, 1977; Kleinman, 1988). Each offers an alternative mode of defining and accounting for health problems and each demands a somewhat different analytic focus (Robinson, 1990). The study of disease, sickness and illness have therefore been taken up by sociologists and anthropologists in distinctive ways. Both disciplines offer a critique of the taken for granted assumptions of the dominant biomedical model but anthropology has often done so through contrasting biomedicine’s view of disease with other cultural models of health and illness (Good, 1994; Kleinman, 1988) while sociology has tended to focus on the inherently problematic epistemological, social and political implications of viewing disease in a narrowly
reductionistic way (Engel, 1977; Waitzkin, 1991). Further, whereas medical anthropology has encompassed a focus on the cultural construction and experience of illness in an array of non-clinical as well as clinical settings (Young, 1982), medical sociology initially focused on social structural aspects of sickness and “sick role” behaviour (Parsons, 1953) while paying less attention to the lived experience of illness and non-clinical interactions (Mechanic, 1978). Indeed, it was not until a new focus on the lived experience of chronic illness began to percolate through the literature that sociologists focused on the work of managing illness in everyday life and, following anthropology, turned their attention to narrative as a powerful source of insight into how illness experiences are framed by particular world-views and social processes.

Given these differing emphases there is now an extensive literature which applies social constructionism to the study of health and illness. Reviewing a wide array of sociological contributions to this literature, Gerhardt (1989) identifies four paradigms in the development of medical sociology. Parsons (1953) established the sociological significance of health and illness through his notion of the “sick role” but Parsonian approaches did not offer a challenge to the biomedical model of disease or the power of the medical profession. With the demise of Parsonian sociology, the focus of medical sociology shifted: through an emphasis on the political economy of health and the dominance of biomedicine, conflict theory introduced a more critical dimension while interactionist and phenomenological approaches established within the field a deepening concern with micro-level politics, structures and social relations. This critical, interpretive turn introduced a renewed emphasis on fieldwork and a variety of methodological strategies: from the analysis of existing statistics and medical records to detailed descriptions of diagnostic procedures and the observation of various interactions between physicians, clinical personnel, patients, their families and friends (Gerhardt, 1989). Moreover, the anti-Parsonian impetus challenged biomedicine’s notion of disease, offering new insights on how health and illness are socially constructed phenomena.

Symbolic Interactionism and Phenomenology

This dissertation draws upon the interpretive tradition (i.e., symbolic interactionism, social phenomenology, ethnomethodology) and, in particular, those who, following Mead
Schutz (1967; 1970), Blumer (1969) and Garfinkel (1967) sought to understand the way in which meaning emerges from routine social interaction. Summarizing the basic premises of symbolic interactionism, Blumer (1969:2) stated,

The first premise is that human beings act toward things on the basis of the meanings that the things have for them. The second premise is that the meaning of such things is derived from, or arises out of, the social interaction one has with one’s fellows. The third premise is that these meanings are handled in, and modified through, an interpretive process used by the person in dealing with the things he encounters.

Here Blumer draws upon the Thomas theorem (Thomas & Thomas, 1928) — “If men define a situation as real, it is real in its consequences.” — to stipulate that phenomena have no intrinsic meaning apart from that given by actors in and through their ongoing interactions. As such, the real and/or anticipated reactions of others are always part and parcel of what is a dynamic interpretive process. Meaning is not static nor can it be determined by the structural features of a situation. It is not out there waiting to be discovered but is, rather, something that is achieved in and through social interaction.

In its emphasis on the intersubjective character of the social world, Schutzian phenomenology (Schutz, 1967) provides a complementary perspective. Encompassing several distinctive approaches (from Garfinkel’s ethnomethodology and Cicourel’s cognitive sociology to Sacks’ conversational analysis), phenomenological approaches to health and illness often centre on the way in which illness and “troubles talk” (Lynch, 1993) disrupts the normal “everyday and everynight” (Smith, 1987) routines of the social world. Moreover, both interactionism and phenomenology emphasize the importance of understanding the patient’s perspective: as such, commonsense and lay knowledge are as worthy of investigation as the abstracted knowledge of experts and medical professionals.

This emphasis on forms of knowledge brings me to one further issue that is central to defining the critical, interpretive stance I wish to adopt here — that is, the role of reflexivity in research. Constructionism, as it has developed within medical sociology, often entails a high degree of reflexivity or double reflection, both about its subject matter and its part in creating it (Bury, 1986). Rather than standing outside of social relations, sociology (like medicine) constructs as well as observes its object (1984; Armstrong, 1990); that is, sociology (like
medicine) mediates social relations just as it gives them shape and meaning. This reflexive stance is lacking in much of the existing research on predictive testing precisely because clinical research typically excludes its own role in co-creating what it purports to study. As suggested in the preceding chapter, this obscures the relationship between power and knowledge, deflecting rather than reflecting an awareness that knowledge production and expression are processes which are constructed in and through social interaction (Armstrong, 1987a).

Though it is widely accepted that pre and post-test counselling are important components of predictive testing, very few studies explicitly acknowledge that clinical outcomes are as much a measure of the interaction between PT candidates and counsellors (or other service providers) as they are of the PT candidate's individual psychosocial state. In consequence, there has been very little attention given to studying the process of predictive testing as it is constituted in and through face-to-face or other forms of communicative interaction within clinical, familial and other social contexts.

Labelling Theory and Negotiation

Within the interactionist paradigm of medical sociology, Gerhardt (1989) distinguishes between the crisis and negotiation models, each of which offer somewhat different explanations of how it is that sickness and illness are socially constructed phenomena. The crisis model, which is derived from the labelling theory of Becker (1963) and other theorists of social deviance, proposes that illness is created by societal reactions to illness stereotypes. These stereotypes confer on the putatively ill a deviant self-identity or "master status" and, as a sort of self-fulfilling prophecy, illness labels constrain people to see themselves as others do.

Whether or not people accept illness labels (and it is assumed that "good" patients will) has a contrary effect to what one would predict based upon the assumptions of the biomedical model. Biomedicine predicts that a return to normalcy (and hence, "de-labelling") will occur in direct proportion to technical advances in treatment and patients' willing compliance with prescribed treatment regimes. Nonetheless, the obverse seems to occur: as Waxler (1981) demonstrates, patients who accept and internalize institutional labels of illness often suffer a more protracted illness episode than those who do not internalize such labels. As such, the social
labelling perspective raises a difficult “chicken and egg” question about the nature of the relationship between illness labels and the experience of illness: which precedes which?

Labelling theory offers a valuable point of entry for understanding the new categories of health and illness that are being created through predictive genetic testing. Emphasizing the social power of diagnostic labels and the role of professional dominance in creating and maintaining deviant and/or stigmatized identities, this approach has often assumed an explicitly critical stance toward biomedicine. Applied to the study of the social implications of genetic testing, this critical stance highlights existing sociological concerns with medicalization (Zola, 1972b) and, more specifically, “geneticization” (Lippman, 1991), the emergence of new types of genetic discrimination (Billings, et al., 1992; Gostin, 1994) and social injustice (Duster, 1990).

For those at risk for or affected with HD, the consequences of actual or presymptomatic diagnosis (through predictive genetic testing) are complex. Indeed, the very act of speaking about a disease which was, as George Huntington noted, “not at all alluded to except through dire necessity,” remains a challenge for many individuals and families at risk for HD. On one hand, being able to name the source and nature of one’s suffering may alleviate uncertainty, social stigma and feelings of shame. On the other hand, a confirmed diagnosis or family history of HD may also become a “master status” which tends to subordinate other equal or more important sources of self-identity (Becker, 1963; Goffman, 1959; 1963).

These issues are by no means unique to hereditary disorders and/or the experience of Huntington Disease; survivors of cancer (Frank, 1991; Lorde, 1980), persons with epilepsy (Scambler & Hopkins, 1986; Schneider & Conrad, 1980) and AIDS (Powell–Cope & Brown, 1992; Weitz, 1989) share an acute awareness of the biographical and social significance of diagnostic labelling. The social power of illnesses labels — such as cancer, epilepsy and AIDS — is not, however, derived from the medical reality they represent; it is derived from the dense network of social meanings which is invoked by the label.

Some illness labels are almost unspeakable. Frank (1991:100) writes about the impact of a nurse’s refusal to say the "dreaded c-word,"
The nurse's refusal to say the word 'cancer' told me that what might be happening to my body was too awful to be called by its proper name. I was suddenly ashamed of what might be wrong with me... Now even the disease was unspeakable. In that nurse's 'c.a.' I disappeared.

Likewise, Scambler and Hopkins (1990) describe the many euphemisms employed by epileptics who reject the medical label of epilepsy but acknowledge that they suffer from "attacks," "fits," or "seizures." The acceptance of illness labels is not, therefore, a matter of fact; it is the product of a "collective definitional process in which the actor's perspective occupies a central place" (Schneider & Conrad, 1980:34).

The interactive aspect of this definitional process is sometimes lacking in labelling theory. Although Waxler (1981) stresses that it is important to consider the degree to which individual patients accept and internalize diagnostic labels, this insight has not always been reflected in empirical studies which adopt a labelling theory approach. As Scambler and Hopkins (1990) point out, many labelling theorists have taken self-labelling for granted, almost as if it were the inevitable outcome of social labelling. As such, an important general criticism of the labelling approach has been its tendency to treat patients or other "deviants" as if they were passive in the self-definitional process (Gerhardt, 1989). Moreover, labelling theory seems to endorse a strong form of linguistic determinism: it is as if saying some thing is an instance of some other thing makes it so. Both of these criticisms therefore take issue with the way that social life appears to be overdetermined; thus both also attempt to restore some degree of human agency. Agency refers,

...not to the intentions people have in doing things but to their capability of doing those things in the first place... Agency concerns events of which an individual is the perpetrator, in the sense that the individual could, at any given phase in a given sequence of conduct, have acted differently (Giddens, 1986:9).

The negotiation model, which owes its primary allegiance to symbolic interactionism and the "substantive" or "grounded theory" approaches of Glaser and Strauss (1967), emphasizes the interactional processes in and through which actors select, check, suspend, regroup and transform meaning (Blumer, 1969). Illness is, therefore, conceptualized as the outcome of a process of negotiation between health care professionals, patients and their families. As such, it is patients' as well as professionals' differential access to resources and divergent abilities to successfully "play the game" which ultimately shape emergent definitions
of the situation (Scheff, 1968). This emphasis on actors’ differential access to resources is crucial here in that it precludes an overly voluntaristic perspective on human agency. As Giddens (1986:26) argues, some versions of hermeneutics and phenomenology tend to view society as if it were “the plastic creation of human subjects.”

Sociologists interested in how illness labels are negotiated initially focused on the diagnostic encounter in order to explore how medical realities are constructed and modified through doctor–patient interaction. As Scheff (1968) points out, this process of interaction resembles bargaining. Diagnosis is, however, contingent upon patients’ validation of the illness categories proffered by doctors or other health practitioners. Without such validation, the doctor’s diagnosis can assert little influence on self–perception or behaviour. It is, after all, validation which makes “the diagnostic categorization (be)come ‘true’” (Gerhardt, 1989:137).

This emphasis on the actor’s self–perception offers a necessary corrective to the determinism of some versions of labelling theory. Moreover, in stressing the significance of the ongoing flow of interaction between doctor and patient, the notion of validation offers an explicit point of entry for studies on the role of interpersonal communication in health and illness. Conversation is not just a means of imparting information; it is an important vehicle of reality–construction and maintenance. As Berger and Luckman (1966:174) propose, “language realizes a world, in the double sense of apprehending and producing it”. Thus, the act of verbally repeating a doctor’s diagnosis to others is a means of allocating health troubles a definite place in the world.

Following Stimson and Webb’s (1975) depiction of account formation, social interactionists interested in the negotiated meanings of illness labels have also emphasized that significant occasions for validation occur during the discussions that patients have in their non–clinical interactions with family and friends. On these occasions patients describe what the

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3 Several phases can be discerned: legitimation occurs when the doctor assesses the patient’s condition and decides whether “normal” sickness status is granted; deciding and telling involve an exchange in which the doctor typically elicits information about the patient’s condition (e.g., a history–taking and/or physical examination), decides upon and tells the patient the diagnosis (Scheff, 1968).

4 One of the best examples of the moral imperative inherent to this view of the therapeutic dimensions of patient validation is found in the 12 Step Program of Alcoholics Anonymous. The first (and perhaps most significant) step comes when the alcoholic stands up and says that he or she is an alcoholic. This admission is a customary part of every meeting as each recovering alcoholic introduces him or herself by saying “My name is X, I am an alcoholic.” Without this admission, the alcoholic is not allowed to progress beyond step 1 (Steffen, 1997).
doctor has said or done and how they feel about it. Often the telling of such stories serves to substantiate or call into question existing clinical definitions of the situation (Scambler & Hopkins, 1990). Further, the responses of relatives or friends may have a major impact on coping, adaptation, and the ability to mobilize resources and social support (Pearlin, 1989). Patient validation and account formation are, therefore, integral to the work that patients and families must do in order to "manage" illness in their everyday lives (Strauss & Glaser, 1975).

The Lived Experience of Illness

Much of the early work done in medical sociology was concerned with acute rather than chronic illness. Furthermore, the emphasis on illness behaviour and the institutional context of doctor–patient interaction neglected to address the lived experience of illness. As Conrad (1990:1260) points out, a focus on the meaning and subjective experience of illness must be grounded in the social organization of the sufferer's world. Among other things, it must include...how people first notice that something is wrong and what it means to them, what kind of theories and explanations they develop to make sense of these unusual events, what they do about their problem, how they come to seek medical care...what impact diagnosis has on them, and how they cope with a medical label and managing regimes. It must examine the relationship with family members, friends and work associates...consider how people contend with formal and informal disenfranchisements...and what strategies people use simply to 'get by' in their lives.

There is now a substantial literature on the lived experience and management of chronic illness. Offering a meta-analysis of the "shifting images of chronic illness" that emerge from this literature, Thorne and Paterson (1998) note that early contributions to the field emphasized the uncertainty, loss, suffering and burden of chronic illness. More recently, the emphasis has shifted to encompass images of health within illness, empowerment, transformation and the normalization of living with chronic illness. Parallel conceptualizations of health–care relationships have also shifted from "client–as–patient" to "client–as–partner". As the authors

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5 Parsons' (1953) approach to understanding the sociological significance of being sick was modeled almost exclusively on acute illness. Indeed, the assumption that patients have an obligation to accept and act upon their physician's authority (and thereby aspire to become well again) was one of the core assumptions informing his conceptualization of the sick role. As many have since pointed out, the experience of chronic illness is markedly different (Mechanic, 1978); in particular, Parsons' assumption that sick role incumbents will eventually get well, cease to be patients, and return to their normal obligations unchanged by the experience of illness and its treatment is untenable in the case of chronic illness. These assumptions may not hold for acute illness either, especially where the illness episode is severe and/or life–threatening. Adapting Sontag's metaphor of illness as travel, Frank (1995) suggests that we are all "citizens of two kingdoms, the kingdom of the well and that of the sick"; those who have experienced illness and its remission are on permanent "visa status" and that visa requires periodic renewal.
suggest, critical interpretive analysis of the way in which chronic illness is constructed reveals many alternatives to the traditional biomedical emphasis on disease; nonetheless, it also reveals "the effects of the social and political understandings within which health researchers generate questions and approach inquiry" (Thorne & Paterson, 1998:176).

Though there is much to recommend about a reformulated image of the chronically ill person as the authority on their own illness, the danger is that we may err too far in this direction and neglect the fact that the work of managing a chronic illness can at times be overwhelming. When the body becomes unreliable, asserting itself in new and predictable ways, the structures and routines of everyday life as well as the forms of knowledge implicit in them are unsettled. Close attention to bodily states, pain, suffering, and the possibility of death force a re-examination of taken for granted assumptions and behaviours. As Kleinman (1988:45) notes, 

The fidelity of our bodies is so basic that we never think of it — it is the certain grounds of our daily experience. Chronic illness is a fundamental betrayal of that trust. We feel under siege: untrusting, resentful of uncertainty, lost.

Illness does not always lead to epiphany nor is the culturally validated narrative of triumph over adversity always available: for some, protracted bodily dysfunction and even death preclude the possibility of ever reformulating the illness experience into something empowering. As Couser (1997:5–6) notes, "what distinguishes the autobiographical subject may also extinguish it."

Recognizing the social and theoretical consequences of able-bodied persons’ abilities to disattend the embodied nature of human existence, sociologists such Irving Zola (1991), anthropologists such as Robert Murphy (1990) and feminist philosophers such as Susan Wendell (1992), draw upon their own experiences of illness and disability to argue that it is only a privileged few can afford such complacency. Moreover, sociology’s systematic neglect of the body in social theory “implies and poses major problems for the formulation of a sociological perspective on the human agent, agency and human embodiment” (Turner, 1992:35).

Sociology has traditionally focused on human agency as if it were a disembodied phenomenon — a matter of intentions, values and beliefs. When illness and/or disability intervenes, however, the body can no longer be taken for granted; illness and disability therefore call into question the Cartesian basis of most social theory. From this perspective, studies of the experience of chronic illness and disability have much to contribute to the development of social
As studies on basic strategies of symptom control and normalization indicate, the body in illness is a body that must be constantly monitored (Nettleton, 1995). Loss of mobility, strength, and control over bodily functions disrupts everyday routines, threatening the illness sufferer’s presentation of self (Goffman, 1959) and compromising access to material and social resources (Dyck, 1995; Wendell, 1992). The day-to-day management of illness thus requires constant attention to the “resources problem” and the “sociability problem” (Gerhardt, 1989).

Access to various resources may become strained long before a diagnosis is actually made and thus the work of managing illness includes processes of self-monitoring and “troubles talk” which may or may not culminate in a visit to the doctor and the attainment of legitimate patienthood status (Lynch, 1983). Given that an official medical diagnosis is an essential prerequisite to obtaining disability pensions, income assistance and/or a range of other services (Dyck, 1995; Wendell, 1996) patients and their families may experience a sense of relief or even elation when a diagnosis is finally made. Further, a definitive diagnosis may dispel the frustration and anxiety which marks the experience of having prolonged physical manifestations which are ignored, minimized or otherwise invalidated by medicine.6

This is not to suggest that the act of naming a hitherto unexplained set of bodily phenomena is always welcome or experienced by patients and their families as a moment of maximal coherence. The moment of diagnosis may mark a time of crisis in which a welter of feelings (i.e., shock, relief, fatalism, bewilderment and confusion) are aroused. Moreover, feelings of relief about finally obtaining a diagnosis may be short-lived and contingent on the longterm prognosis and/or the images that people have of their illness. As Pinder (1992) found in her study on the diagnosis of Parkinson’s Disease, the word “disease” often conjures up images of being unclean; thus feelings of relief associated with obtaining a diagnosis may be overwhelmed by a deep sense of shame.

Shame is part of the sociability problem: it has to do with the negative evaluation of self-identity or moral worth. It is a “primary social emotion” (Goffman, 1959) that Scheff

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6 Studies illuminating these aspects of the experience of diagnosis are, as Wendell (1996) points out, an important counter to a much more extensive literature that documents the dangers of over-medicalization.
(1990:18) describes as a "feeling trap" — an "intense, and automatic bodily sign" that is both recursive and contagious, "each shame state serving as stimulus for further shame". For the chronically ill and disabled, such feeling traps are especially pronounced. They magnify the suffering which accompanies loss (Charmaz, 1983) and exacerbate the feeling of being "worth less" or even "worthless" (Charmaz, 1998).

Illness experiences are, in short, "critical situations" (Giddens, 1979) in which "the structures of everyday life and the forms of knowledge which underpin them are disrupted" (Bury, 1982:169). Chronic illness, in particular, "brings individuals, their families, and wider social networks face to face with the character of their relationships in stark form, disrupting normal rules of reciprocity and mutual support."

Emphasizing the importance of understanding how people sustain a sense of self-identity and manage such disruptions within the context of their overall illness trajectories, Bury (1982), Williams (1984) and others writing in the early 1980's focused new attention on the role of biography, life history and narrative. Bury (1982), in particular, describes three interwoven aspects of illness as "biographical disruption". First, there is a disruption to taken for granted routines and behaviours; second, there are disruptions to the explanatory systems people normally rely upon such that a fundamental re-thinking of one's biography and self-concept is required; third, there is the need to mobilize resources in order to cope with the altered situation.

Focusing on the "cognitive packages" which help people to rewrite their personal biographies in order to make sense of the illness experience, Bury takes exception to the inherently anti–biomedical stance of critics such as Illich (1975). As Bury proposes, access to medical knowledge and information permits illness sufferers to conceptualize their disease as if it were separate from self. Such reification is precarious — there is an "uneasy balance...between seeing the condition as an outside force and yet feeling its invasion of all aspects of life" but it is also a "powerful cultural resource" that provides "an objective fixed point on a terrain of uncertainty" (Bury, 1982:173).

Bury's point is important because it exemplifies the need to avoid the dichotomous identification of medical thought with disease and lay thought with illness. Moreover, it also points to the duality of medicine as a cultural system and the way that researchers must, on
some level, respect "the authenticity of belief (whether doctors' or patients') whilst offering the possibility of a 'critical evaluation of the justification of belief' and the involvement of beliefs in the distribution of power" (Bury, 1982:180).

This focus on beliefs also extends to the ways in which people draw upon various cognitive schema in order to re-negotiate the meanings of illness and sustain hope in the face of uncertainty and/or a gloomy prognosis. One example comes from Wadell's (1982) study of how parents of children with cystic fibrosis "explain away" aspects of the illness that threaten faith and hope. Borrowing from Sykes and Matza's (1957) conceptualization of techniques of neutralization, Wadell describes several such strategies: for instance, parents tend to talk about the history of diagnosis rather than the uncertain aetiology of the disease or discount the lack of effectiveness of treatment regimes by insisting upon the uniqueness of each case. These techniques are not unique to cystic fibrosis; families at risk for, and affected by, HD also employ many of these strategies as a response to what remains an exceedingly gloomy prognosis.\footnote{Nor are such strategies unique to patients' management of uncertainty. As Pinder's (1992) study on the diagnosis of Parkinson's Disease demonstrates, doctors also focus on the importance of therapies and/or a comparison with worse case scenarios in order to sustain hope and faith.}

If it is possible to isolate, within such a cursory review, the most salient findings in the literature on the lived experience of chronic illness and disability, I would do so as follows:

1) there are several parallel realities that exist for patients and their families; 2) the work of managing chronic illness and disability occurs largely within a non-clinical context and it remains contingent on patients' and families' access to and ability to mobilize key resources; 3) forms of knowledge and ways of knowing are cognitive resources which play a crucial role in shaping the illness experience and; 4) illness experiences are a form of biographical disruption in that a rethinking of one's life history, self-identity and trajectory are required in order to make sense of the experience. Further, given the emphasis that I have already established on social interaction, one further theme deserves additional comment — that is, the way in which people manage (potentially) stigmatizing information.

**Stigma and the Management of Discrediting Information**

Much of the work on how people manage chronic illness and disability has been
elaborated through detailed descriptions of the dilemmas that people encounter in the attempt to normalize social relations. Some of the best work incorporates the researchers' own subjective experience of illness and disability. Drawing upon his experiences with polio, Davis (1963) was among the first to focus on how the "visibly handicapped" manage strained social interactions with so-called "normals." As he demonstrates, the socially adept handicapped person must create a feeling of safety for the "normal" before it is possible to openly discuss the "incidental capacities, limits and needs" of the handicapped person. After "breaking through" (i.e., facilitating normalized role-taking), the two can engage in something like normal sociability. The difficulty of sustaining an atmosphere of openness about the handicap is, however, a source of stress for both parties; such openness cannot ever be fully accomplished; it must be created and recreated in and through ongoing social interaction.

Following Goffman's (1963) classic essay on stigma and the management of spoiled identity, sociologists interested in the adverse social consequences of illness and/or disability have looked at the role of stigma in shaping the presentation of self and the management of (potentially) stigmatizing information. When illness visibly announces itself — be it in the form of uncontrollable choreic movements, hair loss from chemotherapy, a sudden epileptic seizure or the scars of Kaposi's sarcoma — the sufferer loses the ability to manage the presentation of self in ways that those who are healthy and able-bodied commonly take for granted. The body is highly contingent and "passing" — that is, keeping from public view that which announces a "spoiled identity" — becomes increasingly problematic.

Given that some illnesses and/or forms of disability are visible and some are not, and/or some are visible at some times and not at others or to some observers but not others, it is therefore important to distinguish between discredited and discreditable aspects of the person: in the first case, information about the identity of the person is visible and/or has been disclosed while in the latter it is not visible and/or has not been disclosed (Goffman, 1963). The problems of managing stigma differ markedly in the two situations — whereas the discredited are

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8 Persons in the early or mid-stages of HD may, for instance, show very few visible signs and symptoms of the disease until they become tired and begin to lose the ability to control balance and coordination. Likewise, it is important to note that not everyone will notice or, for that matter, know the significance of the characteristic signs and symptoms. As noted earlier on, those with HD are often mistaken for being drunk and, for many, this type of labelling is infinitely worse.
concerned with managing the tension inherent to social interactions with so-called "normals," the discreditable are concerned with strategies of concealing (i.e., "passing") and/or otherwise regulating access to information about the stigmatizing feature. Clearly the latter situation pertains to many people at risk for Huntington Disease.

Goffman's interest in the strategic role of interpersonal communication leads him to make numerous observations about the ways in which lay actors go about the work of managing stigma. For instance, he notes that "a very widely employed strategy of the discreditable person is to handle his risks by dividing the world into a large group to whom he tells nothing, and a small group to whom he tells all and upon whose help he then relies" (Goffman, 1963:95). Once aware of the stigmatizing attribute, family and others who are in the know (i.e., fellow-sufferers and/or the "wise") take on certain obligations: as such, "intimates can come to play a special role in the discreditable person's management of social situations, so that even where their acceptance of him is not influenced by his stigma, their duties will be." Such duties may involve "covering" and, where intimates share in the stigmatizing potential of the attribute — that is, where there is a "courtesy stigma" — adhering to a pre-arranged approach to determining what if anything to tell to those who remain unaware.

This is precisely the type of situation which many PT candidates and their families must contend with. Predictive genetic testing provides information which has an intimate connection with ongoing existential processes of self-identity and perception; notions of personhood and individuality; difference and similarity (Brock, 1994). In addition, it is information which is generally considered to be private (Privacy Commissioner of Canada, 1992) and relatively inaccessible to others. Information about one's genetic inheritance and more specifically, the outcome of PT results does, however, also have a special relevance for related others. It may modify risk or the perception of risk and it may also, as genetic counsellors warn, occasionally reveal unanticipated paternity issues (Benjamin, et al., 1994). Further, although it may not provide the basis for a deviant or "spoiled identity" (Goffman, 1963), living at risk or being a

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9 While I am not aware of any studies which offer empirical support for Goffman's proposition, it is certainly the case that a policy of discretion is often advised during the counselling which accompanies predictive genetic testing for HD.
known gene carrier for HD, does invoke the potential for stigmatization. Certain types of family
dynamics (e.g., pre-selection) may also invoke the same potential for non-carriers who, by
virtue of their “escapee” (Wexler, 1984) status, fracture a powerful source of solidarity and
belonging. For all of these reasons then, one might expect many FT candidates and their
families to express ambivalence about the probable consequences of sharing information about
the family history of HD.

The social threat posed by a discreditable attribute may, of course, vary throughout an
illness career. Although the diagnostic encounter has been granted special relevance in
understanding how patients and their families identify and attempt to manage the adverse social
meanings of illness labels, Goffman’s conceptualization of stigma has also spawned many
studies on the everyday management of discrediting and/or discreditable information. The key
problem is that many of these studies presume a deviant self-identity and thereby neglect the
actor’s self-perception and/or ability to intervene in and reshape the social meanings of various
illness labels. Further, there has been a protracted tendency to overemphasize the idea that the
stigmatized learn the meaning of stigma through direct exposure to discrimination. In
consequence, the perception of stigma and its meaning to the putatively stigmatized have been
overlooked (Schneider & Conrad, 1980).

Self-labelling and the disclosure of illness may offer an opportunity to achieve a kind of
metacontrol even if the circumstances which create the “opportunity” are themselves
undesirable. In The Cancer Journals, Audre Lorde (1980) writes about her resistance to wearing
a prosthesis after having had a mastectomy. Rejecting “the cosmetic reassurance” of a “little
puff of lambswool and/or silicone gel” precisely because it rendered invisible her struggle “to
come to terms with her changed landscape and changed timetable of life,” Lorde chose to affirm
her difference and share it with other women. Like the closeted epileptic (Schneider & Conrad,

10 One cogent example of this comes from Scambler and Hopkins’ (1986; 1990) finding that many epileptics who
initially resisted the diagnosis of “epilepsy” engaged in protracted attempts to persuade their doctor to revise the
diagnosis. None were successful in negotiating removal of the stigmatizing label nor were many able to obtain the
support of family and friends in disavowing epilepsy. Thus, although most persons with epilepsy eventually came
to accept the diagnosis, many preferred not to disclose their epileptic status to friends and associates unless it was
absolutely necessary; many also continued to have great difficulty in saying the word epilepsy and thus a range of
alternative descriptors (e.g., “attacks”) were employed as pseudonyms for the condition.

11 Studies on homosexuality (Humphreys, 1972) and criminal deviance (Sykes & Matza, 1957) are classic
examples.
Lorde (1980:61) recognized that persons with stigmatized illnesses are often invisible to one another. "Surrounded by other women day by day, all of whom appear to have two breasts, it is very difficult sometimes to remember that I AM NOT ALONE." Visibility is, as Frank (1995) also argues, a fundamental part of community, where community is defined in terms of a shared illness experience.

Schneider and Conrad's (1980) study on how persons with epilepsy manage the "stigma potential" of their illness, was among the first to address the relationship between disclosure of stigmatizing information and the availability of a supportive community. Demonstrating that the processes of information management used by persons with epilepsy are "much more complex than the now-familiar metaphor of being 'in or out of the closet'", Schneider and Conrad (1980:39) argue that disclosure and concealment are always contingent on "learned perceptions of the stigma...actual 'test' experiences with others before and/or after disclosure, and the nature of the particular relationship involved." As such, they find that the closet often has a "revolving door"; particularly where there is no readily available positive identity or supportive subculture to encourage the "coming out" process, the putatively stigmatized oscillate between being "in" and "out" in dealing with the practical matter of avoiding discrimination.

Also looking at the experiences of persons with epilepsy, Scambler and Hopkins (1986) extended the analytic utility of the concept of stigma by making the distinction between felt stigma (i.e., a sense of shame and fear of enacted stigma) and enacted stigma (i.e., stigma experienced as a consequence of the actions of others). Concluding that felt rather enacted stigma often predisposed persons with epilepsy toward secrecy, concealment and/or trying to pass as normal, Scambler and Hopkins (like Conrad and Schneider) underscore the importance of not viewing felt stigma as an omnipresent force in the lives of the potentially stigmatized. Shame is a highly significant aspect of stigma but it is not essential to the felt need to adopt strategies of information control. The awareness that others are likely to perceive one's attributes as negative is, in and of itself, often sufficient to ensure that the potentially stigmatized are cautious when it comes to self-disclosure.

These and other studies on how the chronically ill manage information have greatly enhanced the usefulness of Goffman's original conceptualization of stigma. Nonetheless, one
significant danger remains. Goffman has often been criticized for adopting an overly cynical view of human action and intention. In his dramaturgical model, all of life is a stage and human actors are, when all is said and done, “strategic manipulators.” As Gergen (1991:149) notes, “no actions remain sincere, simple explosions of spontaneous impulse; all are instrumental.”

Applied to the study of how people manage potentially stigmatizing information, this view of human action narrows and flattens the possibilities for understanding the meaning and significance of illness and/or disability. It assumes that interactants are always (or usually) capable and/or conscious of engineering a particular outcome or representing themselves in a predetermined way. Given the radical contingency of the body in illness, this seems to me a tenuous assumption that even the most generous reading of Goffman does not dispel.

Finally, there is the issue of the goals most salient to understanding the meaning and significance, as well as the consequences of, communicative interaction. Strategies for managing information are often goal driven but, as Daly and Wiemann (1994) point out, there are many types of goals. The pre-determined, individual goals most relevant to understanding strategic outcome–oriented communication stand in sharp contrast to the emergent, relational goals that facilitate process–oriented communicative interaction. To assume that one or the other is predominant or that both cannot be operative at the same time is to miss the complexity of communicative interaction. The aims which an actor pursues in language and interaction with others cannot, as Habermas (1984:241) insists, be treated simply as if they were conditions which can be brought about through causal intervention in the world: “for the actor, the aims of action oriented towards success and reaching understanding are situated on different levels.”

**Practices of Disclosure and the Theory of Awareness Contexts**

The sociological literature on the disclosure of “bad news” cogently illustrates the need to examine the diverse purposes and meanings of communicative interaction from the standpoint of a variety of social actors. Indeed, it is at this juncture, that interactionist approaches to understanding diagnosis, the lived experience of illness and the management of (potentially) stigmatizing information may be seen to converge with empirical studies on awareness and communication. Though I have not yet linked practices of disclosure with the
larger cultural narratives which shape as well as reflect these practices, the reflexive stance which I have thus far emphasized points toward the need to critically examine the stories we tell ourselves about our acts of communication. How do we talk about what we accomplish in and through communication and what do these ways of talking about communication imply?

Much of the literature on the delivery of bad news focuses on clinical situations where the patient has a serious, life-threatening illness (such as cancer) and/or is believed to be dying. More recent studies of risk perception, communication and awareness do, however, make substantive use of many of the key insights drawn from this literature in order to better understand the challenges of communicating about risk information (Bottorff, et al., 1996). Though the disclosure of terminal illness may be seen to represent the extreme case in communicating bad news, there is even here, much room for variation in how patients and their families as well as health care providers structure their communications. As Taylor (1988) found in her study of physicians’ practices of “breaking bad news” (Buckman, 1992), many adopt a rigid, predetermined style of communication: this routinizes a difficult task but it also neglects patients’ individual preferences about how much information they wish to receive and how they wish to receive it. As such, minimization of the patient’s discomfort and distress is only achieved in those situations where there is a fortuitous match between the physician’s adopted style of disclosure and the patient’s preferences.

Given that there is now a strong impetus toward “truth-telling” in most Western countries (Armstrong, 1987b; Buckman, 1992; Taylor, 1988), the management of uncertainty remains an important focal point in many studies of doctor-patient communication. As Davis (1960) argues, uncertainty is often feigned by doctors when they do not wish to reveal everything they suspect or know about the patient’s condition. Physicians have a great deal of latitude here since the professional dominance and authority of physicians “is based on the assumption that a professional has such special esoteric knowledge and humanitarian intent that he and he alone should be allowed to decide what is good for the layman” (Friedson, 1970:x). This is not to suggest that the practice of withholding information about the patient’s diagnosis (and/or prognosis) was (or is) done solely out of professional self-interest; many physicians who were unwilling to disclose “bad news” believed themselves to be acting in the
patient's best interests. Nonetheless, attitudes toward truth-telling and practices of disclosure have undergone substantial change.\textsuperscript{12}

In part, the shift toward increased disclosure has come about as a result of studies which demonstrate that there is little evidence to support the view that truth-telling does serious harm and/or induces adverse responses (such as suicide or severe depression) in patients newly aware of the gravity of their situation. Further, physicians who have long underestimated the number of patients who want full disclosure have had to concede that a high proportion do want to know the truth.\textsuperscript{13} Moreover, there is now an ethical and legal obligation to provide mentally competent patients with any personal medical information that they require and/or request.\textsuperscript{14} In many cases it is also essential that patients understand the nature of their disease and the likely prognosis before consenting to experimental or other therapies.

Recognizing the profound significance of disclosure practices for patients as well as health care providers, Glaser and Strauss (1964; 1965) undertook one of the first, detailed ethnographic studies of the social interaction which occurred between dying patients and medical personnel in a hospital setting.\textsuperscript{15} This study was conducted in the early 1960's and hospital staff were, at the time, concerned by feelings of ineptness in helping dying patients. Observing these situations and the tactics which hospital staff and patients employed in order to manage their communicative interactions, Glaser and Strauss (1964; 1965) pioneered the use of grounded theory methodology (Glaser & Strauss, 1967) in developing their now classic theory of awareness contexts.

An awareness context is "the total combination of what each interactant in a situation

\textsuperscript{12} Studies on American physicians' attitudes toward, and practices of, disclosing a diagnosis of cancer graphically illustrate the speed and magnitude of this change: in 1951 approximately 90\% of physicians indicated that they preferred not to disclose the truth to cancer patients while only two decades later, in 1971, 90\% of physicians indicated that they were practicing disclosure in these circumstances (Novack, et al., 1979).

\textsuperscript{13} Studies conducted in the U.S. during the 1980's demonstrated that, independent of the nature of the disease, more than 50\% and as many as 97\% of patients surveyed said they wanted to know the truth about their condition; further, these findings held when patients actually did learn the truth (Buckman, 1992).

\textsuperscript{14} In Canada, the precedent setting 1992 Supreme Court case of \textit{Mclnerney v MacDonald} established that patients are entitled to reasonable access to their medical records. Although health records are owned by the institution, clinic or doctor that created them, patients have the right to examine and, for a "legitimate" fee, copy such records. Doctors do, however, have some discretion to deny access where there is a high probability that disclosure will have adverse effects on the patient's health. In British Columbia, these aspects of patient access to information are also covered under the Freedom of Information and Protection of Privacy Act (Shaw, Westwood, & Wodell, 1994).

\textsuperscript{15} See also Sudnow (1967).
knows about the identity of the other and his own identity in the eyes of the other" (Glaser & Strauss, 1964:670). Thus, an awareness context is an analytic rather than structural unit; it represents one moment in a dynamic process of interaction between two or more interactants. Further, awareness contexts are inherently unstable; interaction changes as it continues.

Glaser and Strauss (1964; 1965) set out a number of directives which emphasize the processual nature of their paradigm for studying awareness contexts. These include a description of the social structural conditions under which each awareness context exists and the consequent changes of interaction leading to further changes of context, the tactics of various interactants as they attempt to manage changes in awareness contexts and the consequences of the initial context, interactions and transformations for the various interactants. They stress that it is important to ascertain the awareness of each interactant because each may hold different beliefs about the awareness of others.

Glaser and Strauss focused primarily on situations where two interactants (i.e., a patient and a member of the hospital staff) were engaged in the mutual problem of managing awareness about the identity of only one person (i.e., the dying patient). As such, a closed awareness context — where “one interactant does not know the other's identity or the other's view of his identity” — applied in situations where the staff person knew that the patient was dying but the patient did not. However, where one interactant “suspects the true identity of the other, the other's view of his identity or both,” a suspicion awareness context exists. This is a modification of the closed awareness context. For example, the dying patient who suspects but does not actually know for certain about the physician’s definition of him/her, may begin to ask questions. The physician may for a time continue to believe that the patient does not know s/he is dying but once “each interactant is aware of the other's true identity and his own identity in eyes of other” an open awareness context pertains. Such openness about impending death may prove difficult for the staff person and/or dying patient to acknowledge and/or sustain and thus the open awareness context may be modified such that “both interactants are fully aware but pretend not to be.” This is a pretense awareness context (Glaser & Strauss, 1964:670).

The theory of awareness contexts has been applied in examining how interactants within a wide range of settings mediate the flow of personal information through verbal and non-verbal
communication. In part, this is because Glaser and Strauss identify, in awareness contexts, an analytic framework which lends itself to understanding not only specific instances of communicative interaction and their consequences for individual interactants, but also the relationship between social-structural features of the situation, proscribed levels of awareness and the routines and practices which create and reflect such awareness. Indeed, it is these latter aspects of the theory which suggest its usefulness for the present study of interactants' stories about, as well as involvement in various instances of communicative interaction.

A range of empirical studies have now utilized Glaser and Strauss' four-fold typology of awareness contexts to examine changing patterns of awareness about death and dying (Seale, 1991; 1997), social interaction between Alzheimer's Disease clients and caregivers (Hutchinson, Leger-Krall, & Wilson, 1997), and familial processes of coping with a terminal diagnosis (Timmermans, 1994). Each study demonstrates the utility of the theory of awareness contexts for examining such issues although each also expands upon and/or modifies the conditions under which the theory is applicable. Several important substantive and methodological considerations are especially relevant within the context of this dissertation. The first has to do with the incompleteness of the four-fold typology of awareness contexts.

As Hutchinson et al (1997) note in reference to the awareness of memory loss in probable Alzheimer's Disease (AD) clients and their caregivers, there is often a period in which all parties experience diagnostic uncertainty and a lack of knowledge. Since no one has a full awareness of the facts there is no attempt to conceal information as in the closed awareness context originally described by Glaser and Strauss. This initial lack of awareness or knowledge is, therefore, a distinct type of awareness context which ought to be of particular concern in assessing the management of communicative interaction in the pre-diagnostic stages of an illness career. Moreover, the inclusion of this pre-awareness context raises the possibility that the client, caregiver and/or family may suspect the diagnostic significance of symptoms (such as memory loss in the case of AD or loss of balance and erratic movements with suspected onset of

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16 Additional studies discuss the significance of awareness but do not make explicit reference to Glaser and Strauss or the theory of awareness contexts. One such study which is of interest here examines familial awareness of the hereditary aspects of Autosomal Dominant Polycystic Kidney Disease (ADPKD) (Wilkie, et al., 1985).
HD) and therefore the suspicion awareness context is brought into play long before a diagnosis is made. Given that Glaser and Strauss were studying situations where the diagnosis had already been made it is understandable that they did not concern themselves with this type of transition into initial awareness. In thinking about how the theory might be usefully employed to understand familial awareness of hereditary disorders, it is, however, important to note that many families at risk for HD are acutely aware of the diagnostic significance of symptoms and hence, although the diagnosis may compel the explicit recognition of onset, it often does not come as a total surprise. Indeed, family members may themselves be far more aware of the “identity” of the person with suspected onset than the physician her/himself. This is, I might add, often a point of frustration for family members who have coaxed the person with suspected onset to see a doctor only to find that the doctor is unable to make a definitive diagnosis.

Incompleteness also pervades the other end of the awareness spectrum. As Seale (1997) and Timmermans (1994) point out, open awareness entails something more than just information about x (where x is the diagnosis of a terminal illness). Although more patients are now told the medical truth about their condition, “they do not always listen or make sense of what they are told” and, in consequence, “there seems to be something else necessary, apart from an increase in information, to create open awareness” (Timmermans, 1994:325). As such, Timmermans focuses on the importance of understanding how the patient and various family members comprehend and respond to the diagnosis of a terminal condition and, on this basis, Timmermans distinguishes between suspended open awareness — wherein the patient and/or family member blocks out or otherwise ignores the information that has been provided about the terminal condition — uncertain open awareness — wherein negative aspects of the information are disregarded and uncertainty is valued because it leaves a margin for hope — and active open awareness — wherein the “patient or family member accepts the full implications of the message and acts accordingly.” All three have in common the fact that the patient and/or family member has been informed of the terminal condition, hence all three are types of open awareness contexts. 17

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17 The work mentioned above on techniques of neutralization (Waddell, 1982) addresses many of the same issues raised in Timmermans’ discussion of uncertain open awareness.
Seale et al (1997:479) identify a similar need to attend to the issue of how patients and families respond to information and, in their study of the prevalence of open awareness of dying in the U.K., they operationalize a distinction between open awareness and “full open awareness”; the “latter is distinguished from the former by indicators of ideological commitment to openness and mutual talk about dying.” Hence, relatives and friends of people who died in full open awareness indicated not only that they and the deceased “certainly” knew that they were dying, but also that the deceased was “definitely” or “fairly” accepting of their death and that they and the deceased talked openly about it.

These modifications of the four-fold typology of awareness contexts broaden the scope of the theory because they demand that explicit attention be given to the role of emotions and value commitments in shaping how patients and their families receive and respond to medical information. This helps to break down the false dichotomy of knowing or not knowing; it allows people to oscillate between “half knowing” (Seale, 1997), knowing but refusing to acknowledge and/or knowing but only partially acknowledging the information in question (Timmermans, 1994). Further, Seale et al’s emphasis on the ideological commitment to openness and mutual talk about dying attaches a heightened significance to what people actually say to each other in and about their acts of communication. It becomes important to know not only that someone knew that they were dying but also that they believed it preferable to know and talk about it. From this perspective, talk about dying means something quite different to those who have a value commitment to openness than it does to those who do not and yet, according to Glaser and Strauss’ formulation, both would be grouped together as instances of open awareness.

Other pitfalls of the theory have also been pointed out: Glaser and Strauss place too much emphasis on the ways in which interactants engage in rational negotiation and as such, the cognitive abilities (Hutchinson, Leger-Krall, & Wilson, 1997), emotional (Timmermans, 1994), and behavioural responses of interactants (Seale, 1991) are overlooked. This has the effect of promoting an instrumentalist view of human action. Further, a preponderance of interest in doctor–patient interactions neglects the fact that “family members and patients are powerful actors in the construction of an awareness context” (Timmermans, 1994:335, emphasis added). Finally, Timmermans is concerned to reject the idea that awareness contexts unfold in a linear or
staged fashion and, in opposition to what appears to be a growing emphasis on the benefits of active or full open awareness, he therefore concludes that,...there is not one optimal or appropriate emotional response to a terminal diagnosis. Acceptance of the impending death may be more appropriate for some patients and family members in certain situations and it may be more functional for the organizational working of the hospital staff, but one open awareness context cannot be imposed upon all patients or family members (Timmermans, 1994:335–36).

Cultural Narratives about Practices of Disclosure

The trend toward increased openness about death and dying entails specific value commitments. These tend to be characteristic of the dominant ethos of individualism; where there is full open awareness, people are able to plan for and exercise some control over the circumstances of their death (Seale, 1997). Such openness and/or the desire for such openness is not universal even within Western Anglophone countries (Davis, 1960; Seale, 1991; Seale, 1997; Taylor, 1988). There are, however, few studies which describe the social distribution of awareness about death and dying although there is evidence supporting a direct association between high socio-economic status and the desire for openness in communication about death and dying (Seale, 1997). Practical considerations are more often referred to in order to explain variation in patterns of awareness or, more specifically, the kinds of disclosure practices which support particular awareness contexts. Where death is medically unpredictable open awareness is unlikely and where there is profound mental confusion or loss of memory, there may only be a small window of time in which open awareness is even possible (Hutchinson, Leger-Krall, & Wilson, 1997). Further, practices of disclosure vary across medical specialties; people dying from cancer are more likely to be reported as knowing what their illness was and that they would die than people who were dying from other conditions (Seale, 1991).18

The growing hospice movement has had a formative role in the shift toward greater openness since such openness is an integral aspect of hospice care (Lind, 1989). In addition, widespread recognition of the “stages of dying” (Kubler-Ross, 1969) has ensured that no matter how resistant the patient or trenchant their denial, they will be expected to progress through

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18 As one study on patients with severe stroke suggests, communication about dying is much like it was thirty years ago in oncology; family members are told rather than the patients themselves despite the fact that many such patients are able to guess that they are dying (Addington-Hall, et al., 1995).
shock, anger and grief and arrive at the realization of impending death. Reflecting on the way in which these issues have been framed in the developing biomedical and popular literature on death and dying Armstrong (1987b:654) argues that “patient denial demanded neither challenge nor complicity but investigation, a discourse on a diagnosis of not wanting to known [sic].”

Examining the salience of cultural narratives in shaping traditional as well as more modern practices of disclosure around the diagnosis of terminal cancer in Tuscany, Italy, Gordon and Paci (1997) argue that we currently understand concealment and silence — or the traditional “not-telling story” — in a very limited way. In part, this is because contemporary biomedicine (as it is practiced in the United States and other Western Anglophone countries) reinforces the predominant cultural narrative of “autonomy-control”. In its emphasis on the value of self-determination, freedom, control and pragmatism — all things which are commonly identified with contemporary projects of self-identity — the autonomy-control narrative stands in sharp contrast to the “social-embeddedness” narrative. This narrative emphasizes the relational/contextual, adaptive, and unifying features of family and social life. Seen through the lens of social-embeddedness, straightforward disclosure and the expectation of openness looks very harsh, even irresponsible.

It looks as if, at the time the patient needs to rest, be cared for and protected by others, s/he has tremendous responsibility and work to do: understand the diagnosis and prognosis, decide on what therapy to follow, work at therapy, have a positive attitude, express deep and strong emotions and thoughts, and plan “rationally” for one’s future end. Knowing looks very courageous or foolish or dangerous, and seems the cause of unnecessary suffering.

Patients and their families often know much more than is explicitly discussed: nonetheless knowing the truth about a terminal diagnosis is secondary to the issue of how one manages one’s relationships with others. Where “everybody knows the other knows, but nobody says anything”, Gordon and Paci (1997:1444) argue that non-disclosure is not experienced by the patient as a lie;

...on the contrary, the family and the patient enact it as a moral duty, a very engaged way to help each other...the responsibility of the burden is shared...and the patient feels that the family, the doctor, and friends are all cooperating to keep him/her within this little, comfortable world until the last minute of life.

19 This is an example of a “pretense awareness context” (Glaser & Strauss, 1965).
Gordon and Paci do not suggest that silence or “the not-telling story” is unproblematic for those who must work to conceal sadness and anxiety from one another. Nor do they overlook the way that many physicians now question the practice of non-disclosure yet continue with it because of family pressure and their own uncertainty. Such sources of ambivalence are, as they suggest, evidence of the growing tension between two very different sets of understandings about appropriate modes of communication and rights of access to patient information. These tensions are not exclusive to cancer nor to the medical world. Various modalities of communication (i.e., speaking or silence) and their associated practices (i.e., disclosure or non-disclosure) find their “roots and support in other contexts, social relationships, and assumptions, both medical and meta-medical”; they are connected through cultural narratives which have a broad or deep cultural influence but which are, more often than not, taken for granted and invisible, “operating in the background of attention” (Gordon & Paci, 1997:1434–35).

Cultural narratives are, as Ricouer (1978:59) suggests, deeply ideological in that they are part of “the code of interpretation of a concrete community which supports us” yet they are, at the same time, never fully transparent to us. It requires concerted effort to become aware of such codes of interpretation, to envision how they shape our daily interactions as well as our larger world views. This is especially so with respect to the stories that we now tell ourselves about the value of genetic information. All of the most bothersome moral questions seem to arise from the problem of how to manage this information, not whether or not to obtain it. As Rothman (1986b:83) suggests, there is something well beyond knowledge for its own sake that is operative here. It is,

...the idea that action is based on information, and the fullest possible information is needed to determine action responsibly....If there is information to be had, and decisions to be made, the value lies in actively seeking the information and consciously making the decision. To do otherwise is to ‘let things happen to you,’ not to ‘take control of your life.’ Such is the contemporary, secular definition of mature, responsible behaviour.

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20 As Ricoeur (1978:59) suggests, we can “nevertheless enjoy a relative autonomy” through the active taking of distance; this “distanciation is the dialectical counterpart of participation, it is the condition of the possibility of a critique of ideologies, not without, but within hermeneutics.”
Communication and Stories about Communication

This dissertation moves some distance away from the clinical interactions which constitute one important aspect of the experience of predictive testing: nonetheless, it is important to acknowledge that the forms of knowledge, value commitments and styles of communicative interaction which are characteristic features of the move toward greater openness within the clinical world may have a hegemonic influence within the lifeworld.

It is, perhaps, too soon to draw any firm conclusions about the appropriateness of various clinical approaches to the management of genetic information. Nonetheless, existing studies on familial communication about genetic information seem to presume that there is a link between access to information and rational action. Some service providers are, in particular, concerned that their patients do not “transmit” the information they receive through counselling and/or testing to their relatives in such a way as to ensure that these relatives understand the necessity of obtaining information and/or counselling (Ayme, et al., 1993). Such concerns are reflective of the “discourse of potential benefits” (Boutté, 1988; Kenen, 1996) but they are far less reflective of the standpoints of at risk individuals and/or families who may or may not experience their communicative interactions in this instrumentalist way; moreover, as the foregoing discussion suggests, there is little to recommend about models of communication which depict communicative interaction as if it were simply about the transmission of information.

With some modification, the theory of awareness contexts provides a useful conceptual basis for describing specific types of communicative interactions (e.g., concealment and/or disclosure of test results) and their implications for the social patterning of familial communication about genetic information. It does not, however, provide a theoretical and methodological basis for analyzing the social activity of making sense of such communicative interactions through the practice of account formation (Stimson & Webb, 1975) and/or conversational storytelling. What is still lacking is a description of the relationship between participants’ stories and the acts of communication that they are ostensibly ‘about’. In order to address this issue, it is necessary to consider some of the ways in which human communication has been theorized and, in particular, the way in which much research on health–related
communication neglects lay actors’ awareness of what is going on in routine instances of communicative interaction. As Giddens (1986) has long argued, the task of understanding the meaningful social world as it is constituted by lay actors involves a constant slippage between the frames of meaning generated by lay actors and the metalanguages invented by social scientists. Lay and social science understandings are not, however, merely first and second order conceptualizations of the social world: appropriation may occur in both directions when knowledgeable lay actors reflect upon sociological descriptions and mediate the frames of meaning within which they orient their conduct.

Communication, Miscommunication and Metacommunication

Much of the initial impetus toward studying the relationship between communication and health derived from psychiatry. This is because psychopathology has long been defined in terms of disturbances of communication (e.g., “hallucinations”, “withdrawal”, “delusions”) which imply “either that perception is distorted or that expression — that is, transmission — is unintelligible” (Ruesch, 1951:79–80). Accordingly, the therapeutic task of psychiatry has been framed in terms of helping those who “failed to experience successful communication” while the scientific aim of psychiatry was to “gain information about the nature of these failures and to formulate remedial measures” (Ruesch, 1951:50–1).

Assessing the pragmatic aspects of human communication and their relevance for understanding “normal” as well as “disrupted” patterns of communication, Bateson (1951:209) was among the first to propose that there are multiple levels of communication and that these levels need to be understood and bridged within everyday interactions as well as therapeutic encounters. Introducing the concept of “metacommunication” as a way of talking about communication that is itself about communication, Bateson pointed out that all communicative

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21 The metaphor of “transmission” derives from a framework for communication proposed by mathematician Claude Shannon in 1941. This “general system of communication” involves a linear schema in which communication consists of a chain of constituent elements: a message source produces a message which is transformed into a signal that is sent as code through a channel to a decoder which reconstructs the message from the signals for the destination message receiver (Mattelart & Mattelart, 1998). As such, neither the meaning, utility or value of the messages transmitted nor the skills and/or relationships of the communicators have any relevance (Wilden, 1987:186). Interestingly, the organizing power of Shannon’s information-based theory of communication also had a formative impact on molecular biology. Borrowing the language of codes, Erwin Schrödinger used the theory to explain the workings of chromosomes in 1943 (Mattelart & Mattelart, 1998).
interaction has at least two important aspects: it conveys information (i.e., it is "about" something) but it also demands that interactants have a mutually agreed upon (if tacit) sense of what is going on (i.e., an awareness of the rules of communication).

In their formulation of the axioms of human communication, Watzlawick, Beavin and Jackson (1967:54) developed a useful terminology for applying these ideas to the analysis of dialogue. Drawing upon Bateson, they proposed that one cannot not communicate and, moreover, that "every communication has a content and a relationship aspect\(^{22}\) such that the latter classifies the former and is therefore a metacommunication." With respect to verbal communication, the relationship aspect is concerned with the understandings that interactants have about how to carry on a conversation and interpret what is going on. Thus metacommunication can be expressed verbally (as in the statement "I am only kidding" or "This is an order") or nonverbally through gestures (such a wink or nod) but each of these forms of commentary on one's own discourse requires stepping outside of it.

Interactants are seldom fully aware of the relationship aspect; nonetheless, communication would be meaningless without it. For instance, someone who is listening to someone else talk about personal appearance (or some other aspect of self) must be able to assess whether the other is making a statement about how they actually see their own appearance, how they wish others to think they see their appearance, how they see others seeing their appearance and so on. The awareness and recognition of the need to distinguish between these possibilities is the ability to engage in metacommunication. The importance of this can hardly be overstated for, as Watzlawick et al (1967:53) maintain, the "ability to metacommunicate appropriately is not only the *conditio sine qua non* of successful communication...[it] is intimately linked with the enormous problem of awareness of self and others."

Metacommunication is what makes it possible to communicate about some thing or topic. Only rarely, however, are we fully aware of and able to articulate what it is that we do in and through metacommunication. Moreover, the relationship between content and relationship

\(^{22}\) Bateson (1951) referred to these as the "report" and "command" aspects.
(metacommunication) is inherently context dependent — we tacitly understand and observe various rules or make certain kinds of inferences according to the people we are in communication with, the social setting, time of day and/or explicitly acknowledged purpose of the communication (e.g., a departmental meeting or counselling session). The phrase “how are you?” may, for instance, be taken as a greeting or acknowledgment when it is exchanged at the beginning of a meeting between two or more colleagues but, when asked by a therapist within a counselling session, the literal question may be the *raison d'être* for the entire sequence of communicative interaction. As such, each situation demands a different type of response; the former warrants a short reply such as “good, how are you?” and the latter, an extended discussion of what is going on and why it is troublesome.

Rule-breaking occurs when one or more interactants misinterpret or otherwise fail to observe the tacit understandings which are the fabric of such communication. Attempting to discover the rules which govern normal social interaction, Garfinkel (1967) devised a series of “breaching” experiments in which his experimenter-students refused to comply with the expected sequence of social interaction. When asked “how are you?” the experimenter would ask for clarification as if it were a literal question. This lead to an exasperated ‘victim’ who eventually declared, “Quite frankly, I don’t care how you are.”

The ability to metacommunicate is essential to communicative competence, but so too is shared cultural knowledge about language, its contextually appropriate use and interpretation (Gumperz & Hymes, 1986; Hymes, 1986). Especially where we are concerned with meaning as it emerges in social interaction, it is necessary to consider competence in so far as it extends to,

...both knowledge and expectation of who may or may not speak in certain settings, when to speak and when to remain silent, whom one may speak to, how one may talk to persons of different statuses and roles, what appropriate nonverbal behaviours are in various contexts, what the routines for turn-taking are in conversation, how to ask for and give information, how to request, how to offer or decline assistance or cooperation, how to give commands, how to enforce discipline, and the like — in short, everything involving the use of language and other communicative dimensions in particular social settings (Saville-Troike, 1989:21).

Problems arise when individual communicative competence is assessed in relation to a

23 As such, deliberately breaking the rules in order to discover what they are, is a strategy which is now known as “Garfinkeling.”
presumed ideal speech community since each communicative situation presents a diversity of kinds of interaction. Awareness of the complex nature of communicative competence has, however, been slow to emerge within studies on health communication and communication problems in medical settings.

**Health Communication Reconsidered**

In an extensive review of the last few decades of research on health communication, West and Frankel (1991) note that much of the interest in this area originated with the observation that problems in communication were implicated in two key issues in medical practice: that is, patient dissatisfaction and noncompliance with medical advice. Thus, it is not surprising that much early work consisted of a practical search for those factors that would contribute to better outcomes (i.e., improved patient satisfaction and patient compliance). This search began with an emphasis on the clarity and amount of information provided to patients in the context of medical dialogues, the theory being that if doctor–patient communication is clear, the information “transmitted” will be readily understood and acted upon by the patient.

Empirical support for this theory was equivocal. Longer patient visit times did not always result in greater patient satisfaction nor did the clarity of the physician’s information always improve. Moreover, longer visits were often indicative of communication “failures” that required extra time to sort out (Korsch & Negrete, 1972). Such findings therefore challenged the assumption that good communication is always clear communication. As such, attention shifted to an examination of individual characteristics and behaviours in the hope that this would explain why some physicians and patients were, respectively, good “senders” and “receivers” and some were not. This focus was equally unsatisfactory as study after study failed to explain the mechanisms involved in miscommunication in terms of the physician’s skills, training and personal attributes or the patient’s education, social class, ethnicity or other characteristics.

In short, cases of miscommunication could not be explained simply as a function of the individuals involved in them (West & Frankel, 1991). Some other explanation was needed and this came with the recognition that it was physicians’ perceptions of patients’ attributes (rather than the actual attributes) (Davis, 1963) as well as the way that patients presented themselves
within the hospital environment (Roth, 1963) that shaped the amount and type of medical information they were given. This explanation resolved many outstanding inconsistencies but as West and Frankel (1991:173) argue, it also “undermined one of the fundamental assumptions of the [sender–receiver] model [of communication]; namely that miscommunication could be traced to characteristics of senders and receivers as individuals.”

Not only was the interactional dimension missing; the sender–receiver model of communication endorsed a “telephone booth bias” (Goffman, 1967) which blinded researchers working in a clinical environment to the fact that doctor–patient interaction was not the only relevant locus for understanding the significance of communication for patient satisfaction and medical compliance. Close friends and family members are also an important source of influence in determining whether or not a patient’s symptoms warrant medical attention (Lynch, 1983); in addition such persons play a significant role in shaping the patient’s response to diagnosis and subsequent willingness to follow medical advice (Stimson & Webb, 1975). Further, as Glaser and Strauss (1965) demonstrate, patients seeking information about their condition often turn to other patients or fellow–sufferers rather than doctors and this is particularly so when relatives withhold information.

Many studies of health communication have therefore adopted a range of alternative approaches in order to address some of these deficiencies. Most notably, conversational and discourse analysis stress the importance of documenting the observable socio–linguistic behaviours that shape and reflect patients’ understandings of, and participation in, health–related communication. Through presentation of detailed excerpts from transcripts of doctor–patient communication, such studies make explicit a host of problems which were initially overlooked. Focusing on various types of interference to an effective exchange of information in the medical interview, Shuy (1993) describes how the structural aspects of discourse (such as who introduces the topic, when and how it is changed, who switches to another topic, when and how interruptions occur) contribute to patient comfort and improved accuracy of information exchange. Elsewhere, Cicourel (1993) analyzes the competing frames of reference which help to explain the disjunction between what happens in the medical encounter and the physician’s attempt to document this in medical records. And, West (1993) examines the role of physician
versus patient-initiated questions in shaping preferred and dispreferred modes of talk. Concluding that many physicians "inhabit a privileged position (if not a state of grace) with respect to being questioned by their patients", West (1993:149) asserts that empirical investigation of the structure of communication is the first step toward understanding and ultimately resolving "the alleged communication 'gap'" between doctors and patients.

In summary, conversational analysis has built a strong case for moving away from the presumption that health communication is mostly a matter of interaction between institutional message 'sources' (e.g., medical research, health care professionals, government, disease foundations) and individual 'receivers' (e.g., patients, their families, schoolchildren, employees). Nonetheless, problems remain. Reviewing a large cross section of more recent contributions to health communication research, Lievrouw (1994) argues that despite the promise of discourse-analytic and other innovative approaches, the field as a whole continues to exhibit three distinctive "pathognomic signs". First, there is an instrumental, interventionist approach to research problems and this is reflected in a preoccupation with outcomes (such as prevention, palliative care or education). Second, there is a persistent view that better communication is therapeutic in that it can repair the gap between ideal medical care and its reality and, third, the knowledge interests of the field remain subordinate to those of clinical medicine and thus there is a persistent bias toward provider interests.

Though these criticisms are directed primarily at studies of media and other forms of mass rather than interpersonal health-related communication, they also speak to much of the literature on genetic testing and counselling. As suggested in the preceding chapter, existing studies on predictive testing for HD have focused primarily on the clinical evaluation of outcomes and the importance of appropriate counselling in preventing adverse responses to the disclosure of test results. Further, most of these studies have been conducted by and for service providers and thus the patient, client or proband's perspective has been neglected.

Two studies on the use of amniocentesis for the diagnosis of genetic anomalies (i.e., Down's syndrome) do, however, illustrate the way in which qualitative studies on communicative interaction have made important contributions to the area. Rapp (1988:143) examines the language of genetic counselling "as it communicates and miscommunicates not
only medical information but also structural power arrangements, social knowledge, and popular meanings about medically defined disability” while Press and Browner (1997) show how service providers shape women’s understandings of the meaning and purpose of MSAFP screening\(^\text{24}\) by omitting reference to the possibility of terminating the pregnancy. As such, these studies focus attention on how the language of genetic testing both conditions and supports certain “choices” while obscuring the social construction of such choices.

Though many genetic counsellors are sensitized to the need to adopt a more “client-centred” approach (Kenen & Smith, 1995), the importance often assigned to viewing the client as an independent, rational being supports an epistemological stance which emphasizes the separateness of the client over and above what the client experiences (Brock, 1995) and/or their connections with others (Kenen & Smith, 1995; Rapp, 1988). Communication is, likewise, conceptualized in terms of its content and strategic, instrumental value and little attention is given to the self-referential nature (i.e., the relationship component) of communication and/or its expressive features. Accordingly, language is treated as “a system of representations or symbols according to which talk always is about something, about a world outside the speaker, as if it were not part of the same world it claims to describe” (Krippendorf, 1989:188).

Living with the Recursiveness of (Studying) Communication about Stories about Communication

This dissertation is about familial communication about genetic information and the stories that people tell about their acts of communication. The theoretical stance which informs this research is, however, one that acknowledges the impossibility of observing such phenomena without also playing a role in their construction. This is the conundrum with which I struggle.

To assert, as Wittgenstein (1953) did, that words are “deeds”, is to recognize that language is constitutive. It shapes the way in which we see and experience the world because it plays a role in constructing as well as representing that world. This does not mean that anything goes or that words can be liberated from the syntactic and semantic rules which govern language

\(^{24}\) Maternal serum alpha fetoprotein (MSAFP) testing is now widely used in prenatal testing for neural tube defects (such as spina bifida) as well as Down syndrome and other fetal chromosomal anomalies. It is only reliable after the 16th week of pregnancy and an initial positive result must be subsequently confirmed through use of ultrasound and/or amniocentesis (Press & Browner, 1997).
systems and the pragmatics of their use. Nonetheless, when we move away from the retrospective analysis of decontextualized words and turn, instead, to understanding the use of words as they are spoken within a practical communicative context, it is apparent that we must attend closely to the possibilities that are permitted or afforded by the prevailing circumstances. As Shotter (1993:43) suggests, “our talk can bear in an important sense upon what is real in those contexts, even though others may contest what we say, and claim that other (and better!) accounts capture what our circumstances afford more adequately.”

As Mishler (1991) has done such a fine job of demonstrating, interviews are practical conversational contexts in which one does not merely gather data or elicit responses. The living dialogue of an interview is the joint production of speaker and listener as each alternates between asking and answering, anticipating and responding. The flow of living conversation is therefore relational and dialogical; each conversation has a social history that emerges as speaker and listener jointly discuss and debate, question and consider, topics of mutual interest. The flow from utterance to response is the engagement we feel in conversation. Bakhtin (1981:280) amplifies this point when he writes,

> The word in living conversation is directly, blatantly, oriented toward a future answer-word: it provokes an answer, anticipates it and structures itself in the answer’s direction. Forming itself in an atmosphere of the already spoken, the word is at the same time determined by that which has not yet been said but which is needed and in fact anticipated by the answering word. Such is the situation in the living word.

Much of what occurs when people talk about their illness experiences has to do with the process of developing a story which makes sense of the experience. Such account formation is, as I have suggested above, integral to the work that must be done in order to manage illness within the context of everyday life. Because storytelling is fundamentally an act of selecting and sequencing relevant events, it is, however, also a ‘natural’ form of conversation. Stories arise at those points in conversation where talk that is “organized around consequential events” opens into an opportunity for a teller to take a listener “into a past time or ‘world’ and [recapitulate] what happened then to make a point, often a moral one” (Riessman, 1993:3).

As the illness sufferer selects relevant events and orders them into a temporal sequence, the illness experience becomes “emplotted” (Good, 1994). Much of what has been written about
illness and narrative therefore centres on how illness sufferers: 1) draw upon relevant life experiences and culturally-available schema for emplotting their accounts of the illness episode (Frank, 1995; Good, 1994; Hyden, 1997; Kleinman, 1988), 2) rework their personal biographies and reorient their life trajectories (Robinson, 1990; Williams, 1984), 3) attempt to accommodate an altered or diminished sense of self (Charmaz, 1983) and/or, 4) cope with disruptions to the taken for granted knowledge and routines of everyday life (Bury, 1982). This process of narrative construction and reconstruction is, as Williams (1984: 197) suggests, “an attempt to reconstitute and repair ruptures between body, self, and world by linking-up and interpreting different aspects of biography in order to realign present and past and self with society.”

The possibility of having near certain knowledge of one’s future illnesses presents a new twist on these themes. As studies on the experience of being HIV positive demonstrate, knowledge of impending illness imposes an intense existential as well as biographical need to reformulate one’s orientation to the future as well as the past and present (Davies, 1997). Moreover, some (Kavanagh & Broom, 1998) argue that we do not yet have an “adequate” culturally-available discourse for making sense of the “embodied risk” which shapes and is shaped by the possibility of such knowledge. It is, in consequence, especially critical at this socio-historical juncture, to be aware of the ways in which such discourses are constructed and the role that we, as researchers play in co-constructing such discourses.

Just as language may be described as “a dialect with an army,” so too is discourse sometimes equipped with a rhetorical force that upholds various regimes. Biomedicine is one such “regime” that has been extensively criticized through the lens of Foucauldian approaches to health and illness (Armstrong, 1984; 1987a; Nettleton, 1995). Further, many critics of biomedicine now argue that narrative offers a powerful medium for reclaiming personal experience from impersonal medical discourse (Couser, 1997). Many examples of published illness narratives read as testimonies to the suffering that is induced when biomedicine denies the illness sufferer the opportunity to speak for her or himself. These narratives have an

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25 I heard language described in this way at a concert given by the Chilean group Inti-Illimani. Musical director and performer Horacio Salinas, was speaking, at the time, about the experience of returning to South America after living in exile during Pinochet’s dictatorship in Chile. The concert took place not long after the furor over Dolly the sheep and Salinas remarked that he did not know what all the fuss was about since genetic cloning was just the end stage of a long process of enforced cultural cloning.
explicitly political impulse which is, in the language of post-colonialism, a “demand to speak rather than being spoken for and to represent oneself rather than being [sic] represented” (Frank, 1995:10).

This impulse toward improved listening and the empowerment of illness sufferers is to be taken seriously, nonetheless, as I have already noted, it ought to give us reason to pause and consider the possibility that when such opportunities become moral imperatives, we add to the work that illness sufferers and their families must do. As Gordon and Paci (1997) suggest, many families in Tuscany prefer not to disclose a terminal diagnosis to their ill family member precisely because such knowledge adds to the burden of being ill. In addition, I concur with Bury (1982:173) when he argues that although medical knowledge tends to separate self from disease it also permits illness sufferers “an objective fixed point on a terrain of uncertainty.”

In other words, I think it essential that researchers take very seriously the standpoint of illness sufferers and their families when considering and co-constructing the conversational and discursive possibilities for, as well as constraints on, the ability to tell some kinds of stories about health and illness and not others. This stance does not imply a naive form of realism but nor does it tolerate the endless play of words which some postmodern approaches to textual analysis seem to deify. There are always possibilities for an alternative reading as one layer of interpretation becomes heaped upon another and we all become hypervigilant about the need to demonstrate that we have gone ‘meta’, but things do not have to be so complex if we are willing to make some good (if not perfect) distinctions and walk with, rather than stand by, them.

Finally, in contending with the fractal-like character of (studying) human communication I am reminded of a set of nested Russian dolls. Communicative events which feature within the narrative are not to be confused with the narrative as communicative event: one is nested within another as the telling of one story encompasses, as its object, the telling of another.
CHAPTER V
THE SITUATION OF THE TELLING

Personal narratives are contextually contingent in a double sense: responsive to and embedded in both the current life circumstances of the teller and the situation of the “telling” (Mishler, 1992:33).

Before I began doing interviews for this research I wondered what the experience would be like, both for the people who would ultimately talk with me about sensitive and sometimes painful aspects of their lives and for me, as I listened, asked questions and probed for more information. Would I ask the “right” questions? Could I rely on myself to know when and when not to probe? Should I strive to be as neutral as possible in my responses or should I respond as if I were a “friend” of the family as well as a researcher (Oakley, 1981)?

I thought a lot about such questions; they were with me on the bus as I travelled to and from the University; they focused my observations of genetic counselling sessions; they permeated many discussions of research design and ethics. A dog-eared copy of Mishler’s Research Interviewing became a constant companion. It was, however, the people with whom I talked — on and off the record in interviews, during various Huntington Society events and at the Annual Retreat for people with HD — that taught me most about how to talk and how to listen to talk about HD. Each interview (and each conversation) was an occasion for the telling of a life story and an event in that life story. Moreover, each telling was a joint construction — responsive to, and embedded in, the current life circumstances of the teller and the situation of the telling.

Purpose and Outline of Chapter

This chapter is about situations of telling: it is about the seriousness of taking context seriously. It speaks to Mishler’s (1992:35, emphasis added) injunction that “shared cultural conventions about narratives enter into their production and are not simply brought in at the stage of analysis.”
There are several situations of telling salient to this dissertation: first and foremost, there is the interview, a somewhat structured, pre-arranged occasion for elicited telling. This occasion, like all others, was situated in time and space; it had a purpose and participants (including myself) had particular expectations about the nature of the interaction which would occur, the form it would take, what was and was not appropriate to ask or tell. These expectations varied with the life circumstances of the teller, and more specifically, their familiarity with the cultural conventions of doing an interview and/or their ease and manner of talking about themselves and others. The stories which participants offered during this context were, however, also about other stories that had been told in a second context — that is, the everyday context wherein people talk informally with each other in their daily lives.

Third, many study participants had attended one or more genetic counselling sessions prior to the first interview and, as such, many had “practiced” telling their story within a very particular institutional and clinical context. During the first genetic counselling session, predictive test candidates are encouraged to discuss issues such as their reasons for wanting to have the test and the possible implications of an increased or decreased risk outcome. These issues overlap with the topics covered in the interviews and as such it is important to distinguish between the communicative interaction of the clinic and the interview situation. How are the stories people tell at the clinic (as patients or clients) different from the stories people tell in their everyday lives (as mothers and fathers, wage-earners and students, parents and caregivers)? Moreover, how were these everyday stories shaped by the context of the research and study participants’ perceptions of themselves as research subjects?

Fourth, there is the situation of my own telling and re-telling, as it unfolds within the pages of this dissertation. Like other situations of telling, this dissertation is structured by a specific (and hopefully shared) set of expectations and these expectations shape what it is that I am able to say and how I am able to say it. Audience matters but it matters differently to me than it does to my study participants and it matters differently now than it did when I began this research. For instance, it matters for academic, ethical, legal, social and personal reasons

1 In her now classic critique of traditional criteria for interviewing, Oakley (1981) highlights the importance of such expectations in shaping the social interaction of the interview. She argues, for instance, that respondents must be allowed to ask questions of the interviewer.
that this telling is now committed to paper and circulated to a particular audience. Moreover, it matters that others, and most specifically my research subjects, may also listen and find that this re-telling is not just about their lives and their tellings but is also, in some cases, an event in those lives. In summary then, all telling is socially situated and the assumptions we bring to every telling enter into its production; as Mishler (1992:35) asserts, the effects of these assumptions are not a function of my or anyone else's "idiosyncratic interviewing [or conversational] style but are inherent to all interview situations [or conversations]."

In the first section of this chapter I discuss the methodological approach and research design — including the context of, and preparation for, the research, the process of recruiting study participants and the selected sample. In addition, I describe the protocol for predictive testing and the sequence and timing of my clinical and non-clinical interactions with study participants. Finally, I also consider some of the ethical implications of the research. Some issues were anticipated in the research design (e.g., specific provisions for informed consent and confidentiality were made prior to ethical review). Others, however, were less easily anticipated.

In the second section, I move out of the clinic and into the everyday lifeworld of study participants in order to describe the process of interviewing and, explore the dialectics of being an interviewer and atypical sort of friend of the family. In particular, I attend to two key issues: 1) the act of listening and how, over time, I learned to listen differently and, 2) the ontological status of the interview as a communicative event. How did participants experience the interview and what implications does this have for the type of analysis pursued here?

The final section of the chapter considers the iterative process of data collection, analysis and writing. It describes the form and content of the three data chapters which follow, detailing the rationale for the many difficult decisions I had to make about which stories to include in the dissertation and, moreover, which "moments" to bring to the foreground as "findings". Here I also consider the various ways in which my "findings" are inevitably partial and incomplete. This is not a shortcoming nor does it undermine the validity of the research. As Mishler (1991: 112) suggests, "it has become clear that the critical issue is not the determination of one singular and absolute 'truth' but the assessment of the relative plausibility of an interpretation when compared with other specific and potentially plausible alternative interpretations."
Methodological Approach

This dissertation is about patterns of communication in families where someone is having predictive testing for HD. An initial survey of the literature yielded scant evidence to suggest that any other empirical work had been done on this topic.² Many papers addressed, in descriptions of protocol or clinical guidelines, the nuances of disclosing results to the PT candidate and some stressed that the candidate’s willingness to communicate their results to family and friends was an important indicator of social support and psychosocial well-being (Bloch, et al., 1993; Huggins, et al., 1992). Nonetheless, despite widespread acknowledgment within the medical genetics community that predictive testing has significant familial implications (Chapman, 1990; Chapman, 1992; European Community Huntington’s Disease Collaborative Study Group, 1993; Hayes, 1992; Kessler, 1993; Mattson & Almqvist, 1991; Tibben, et al., 1993a; Tibben, et al., 1993b; Wexler, 1992), few studies were designed in a way which might elucidate this important area. None of these studies were concerned with eliciting from the PT candidate a sense of the embedded meaning and lived experience of predictive testing and, with few exceptions (Gray, 1995; Wexler, 1979) all were designed and conducted from the perspective of service providers and clinicians.

Clinical research relies heavily upon survey-based instruments chosen for their utility in diagnosing depression and/or measuring other significant psychosocial attributes of the PT candidate. I stated in Chapter III that such research serves an important purpose and I will not reiterate these contributions here. What I do wish to stress is that an alternative approach was required, an approach which would allow PT candidates and their families to express in their own words and according to their own narrative conventions, a sense of what predictive testing meant to them within the context of their own family history and awareness of HD. The methodological approach for this research was therefore shaped by the necessity of arriving at a deeply contextualized way of understanding, rich in detail and illustrative of the micro-processes which characterize the everyday social and familial interactions of study participants, yet attentive to the themes and issues which resonate across a diverse set of experiences.

² The only reference I have been able to locate to work on patterns of communication in families at risk for HD is an unpublished conference paper (Shakespeare, 1992: cited in Richards, 1993),
Short of living with and observing all of the interactions of a particular family or families, in–depth unstructured interviews, in conjunction with participant–observation, provided the most promising method of data collection. Although interviewing does not, as a general rule provide researchers with access to what are commonly regarded as “naturally occurring conversations”, there are many aspects of interviewing which allow the researcher to elicit narrative accounts which are similar in structure and content to those which might occur during more informal social interaction. (This is an issue I take up in greater detail below.) Moreover, the ethnographic component of the work allowed me to situate myself within the communicative events that I sought to understand and describe. Talking and listening, it seemed, was the best way to learn about the shared norms, cultural rules and knowledge which shaped at risk individuals’ and families’ talk about HD. Such knowledge, together with the skills that are needed to make use of it, form the basis of what is now widely recognized as communicative competence (Saville–Troike, 1989).^3

The ability to talk about HD is a resource which has immediate implications for at risk individuals and their families. It is the primary means for acquiring information, but it is also the way in which observations and feelings are shared, understandings are formed, family decisions are made. In addition, being able to “really talk” is central to the ongoing creation of identity, the sustenance of friendship, intimacy and social support. As Keen and Fox remind us, “You can’t tell who you are unless someone is listening” (cited in Randall, 1995:288–89). Conversely, the feeling and experience of not being able to talk with others about everyday hopes and dreams as well as anxieties and fears, may undermine meaningful social interaction and threaten one’s sense of self and connection with others. In this respect, the patterned absence of speech is also an important dimension of the research. As Lakoff (1995:25) suggests,

> It is easier to perceive what is there as meaningful, as opposed to discerning meaning in the absence of a phenomenon. What is explicit and apparent responds to analysis more readily than what must be inferred.

Silence in discourse can be described and interpreted in a number of ways, all of which

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3 As Saville–Troike (1989:7) notes in reference to the late 1960's convergence of sociology and linguistics, "merely accounting for what can (and cannot) be said in a language, but not what can be said when, where, by whom, in what manner, and under what particular circumstances, came to be considered inadequate as a goal for linguistics by many linguists, and by all identifying themselves as 'sociolinguists.'"
suggest that silence ought never to be regarded as an absence of content. One cannot not communicate (Watzlawick, Beavin, & Jackson, 1967). A basic distinction may, however, be made between the semantic analysis of those silences which carry meaning but not propositional content (i.e., pauses and hesitations that occur within and between the turn-taking of two or more speakers) and the pragmatic analysis of silent communicative acts (e.g., refusal to respond to a question) which derive their meaning from adjacent vocalizations but which also have their own illocutionary force (Saville-Troike, 1989:146). In addition, silence may also be examined in terms of its social and ideational significance within a particular speech community’s own conception of the symbolism of the act of speaking or not speaking. Accordingly, silence is part of a “complex set of communicative resources available to members of particular speech communities for the communication of social and expressive as well as referential meaning” (Bauman, 1983:5). Implicated here is a focus on “how communication works as a process.” Silence, like speech, is utilized in socially patterned but infinitely variable ways and these must “be discovered, not simply assumed to be akin to the investigator’s own” (Bauman, 1983:16).

My own assumptions about the place of silence in communication about personal information have been formed in opposition to what might be called a discourse of therapeutic confession. This discourse begins from the premise that life must be both examined and revealed in order to be worth living. Examination is, in this sense, much more than introspection and self-reflection; it demands an audience to whom one must tell all. There is, therefore, a compulsion to reveal oneself and, by extension, the ways in which one has chosen to live.

One confesses one’s crimes, one’s sins, one’s thoughts and desires, one’s illnesses and troubles, one goes about telling with the greatest precision, whatever is most difficult to tell. One confesses in public and private, to one’s parents, one’s educators, one’s doctor, to those one loves, one admits to one’s self in pleasure and pain, things it would be impossible to tell anyone else, the things people write books about. One confesses, or is forced to confess (Foucault, 1978:59).

In some respects the recent profusion of interest in biography and narrative (that is, ways of telling one’s story) may be seen as a more positive outgrowth of this tendency toward telling all. In its less moderate form, however, this emphasis on self-disclosure suggests that we cannot really be who we are unless we tell who we are. Secrecy and silence are cast in pathological terms not only because there may be someone who has a legitimate need to know what is going
on but because secrecy and silence are (among other things) real and symbolic attempts to
preserve that realm of self-knowledge and/or personal life which is off-limits to specific others
or even ourselves.

In approaching the issue of how at risk individuals and families communicate about
genetic information, I wanted to understand the boundary between what was and was not, off
limits. This boundary is flexible or perhaps, semi-permeable. Thus I neither hoped nor expected
to characterize particular individuals or families in terms of their “open” or “secretive”
communicative styles. People practice an array of ways of knowing and these ways of knowing
are deeply embedded in the social situation and life circumstances of the knower. Only some of
this, I realized, might actually be articulated within the interviews; the other parts of it (or at
least those that were accessible to me), I would have to learn through linking my own rational
and analytic as well as emotional awareness with what I heard and how it felt to listen. In other
words, I did not expect study participants to tell me everything that I needed to know nor did I
presume that everything I needed to know could or should be articulated in words.

Research Design

At the outset of this research, there were a number of social scientists writing about the
social implications of the new genetics. Very few, however, were conducting empirical work
from a non-clinical perspective. Richards (1993) argues that a lasting distaste for biological
reductionism combined with the specialized and highly technical language of genetics has
deterred many social scientists from engaging in empirical research on the new genetics. It is my
impression, however, that the relative absence of empirical work by social scientists is more
often related to the logistics and politics of acquiring access to a population which is at risk and/
or undergoing genetic testing for an hereditary disorder. In this, and several other respects, I was
extremely fortunate.

Context of Research

In February 1993 I met with Dr. William McKellin to discuss the possibility of working
with the Predictive Testing Research Group in Medical Genetics at UBC. Headed by Dr.
Michael Hayden, this multi-disciplinary group was formed in 1988 to study the psychological
and clinical implications of predictive testing for HD. Affiliated with fourteen other genetics centres in Canada which now offer predictive testing, this research group has followed the largest cohort of PT participants in the world for the longest period of time. The PT Research Group was, in 1993, seeking the assistance of interested social scientists in order to design and conduct a study of the familial implications of predictive testing for HD.

Participation in the weekly research meetings of this group provided a number of benefits including regular updates about new developments in predictive testing, a lively forum in which to share ideas and discuss proposed research and, the opportunity to observe and contribute to the development of clinical protocols and policies related to predictive testing for HD (Adam, Cox, & McKellin, 1996; Benjamin, et al., 1994). Working in conjunction with Dr. McKellin, I contributed to the design, development and implementation of both qualitative and quantitative strategies for studying the social and familial implications of predictive testing for HD. The objectives of the qualitative studies which received funding are shown in Appendix I.

The research which is the focus of this dissertation directly addresses objective 3 of the British Columbia Medical Services Foundation (BCMSF) study — “to investigate individuals’ decisions to share information about participation in predictive testing with family members, friends and others” — and objective 2 of the Hampton Fund study — “to understand the dynamics of communicating test results to family members and friends”. In addition, a related project funded by the Huntington Society of Canada provided the impetus to prepare five narrative accounts for a forthcoming publication on the lived experience of predictive testing.

The BCMSF study is based on pre and post–results interviews (conducted four to six months after test results) with twenty–two predictive test candidates and forty–two family members from urban and rural settings in British Columbia. The Hampton Fund study follows a sub–sample of the same families over a longer period of time and as such, a third round of interviews was conducted with selected families at eighteen to twenty–four months after the test results. The BCMSF study was designed in conjunction with a separate quantitative study which was, ultimately, funded by the Canadian Genome Analysis and Technology Program (CGAT).

Dr. McKellin was the Principal Investigator on each of these projects.

The CGAT study utilized a package of psychosocial and family assessment questionnaires which were
I was employed as a research assistant on the qualitative studies described above from May 1994 to July 1996. Prior to this, I assisted in the research design, prepared and produced study materials (including letters of introduction, pamphlets about the study, questionnaire packages for the CGAT study and interview schedules), recruited study participants and conducted the first ten interviews. During the initial phases of the BCMSF study I also recruited participants for the CGAT study. As per the pamphlet shown in Appendix II, recruitment strategies for the BCMSF and CGAT studies were integrated in British Columbia. Elsewhere in the country only the questionnaire-based family study was offered to new PT candidates.

**Predictive Testing Protocol**

The research design was, in many ways, shaped by the clinical protocol for predictive testing. This protocol has been described in detail elsewhere (Benjamin, et al., 1994; Fox, et al., 1989) but it is important for the purposes of this chapter to summarize the most salient aspects.

Participation in the predictive testing program is guided by the following criteria for eligibility: the at risk individual must “1) have a confirmed family history of HD and an apriori risk of 50% or 25%, 2) be able to provide informed consent, and 3) not have been given an established clinical diagnosis of HD (as predictive testing is for persons who consider themselves to be at risk)” (Benjamin, et al., 1994:607). The involvement of the at risk individual’s family physician is also recommended but is not a requirement of the program.

All PT candidates are encouraged to bring a support person along with them to as many counselling sessions as possible since involvement in these sessions provides the support person with a more thorough understanding of predictive testing. If the PT candidate has not had prior counselling on the genetics, natural history and clinical features of HD, this information is provided prior to actual enrollment in the predictive testing program. Further, all candidates are informed that their participation in predictive testing is completely voluntary, they may withdraw at any point, all information derived through the program is confidential and, results are only disclosed to third parties with the express written consent of the PT candidate.

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administered before, and at several intervals after, test results. The final sample for the CGAT study included forty PT candidates (and selected family members) from four Canadian genetics centres where PT for HD is offered. Sixteen of these families were from BC and, of these, thirteen also participated in the BCMSF interview study.
There are three distinct components to the clinical sessions in the predictive test protocol: 1) counselling, 2) education, and 3) clinical assessment. At the first pre-test counselling session participants discuss with a genetic counsellor their reasons for requesting the test, the possible benefits and potential harms of receiving an informative result. PT candidates are also encouraged to review all life and disability related insurance policies in light of the fact that their test result may have significant implications for health care and employment. A medical, psychiatric and family history is taken and the PT candidate’s psychological status is assessed through use of questionnaires and an interview. Finally, a neurological examination is done to assess whether or not the PT candidate is showing signs of HD. If PT candidates are showing signs but have indicated a preference not to know, they may continue with the process of predictive testing. Once the decision has been made to proceed with the test written consent is obtained and a blood sample is drawn and sent to the laboratory for DNA analysis.

The second counselling session is scheduled for one to two weeks before test results are given. The purpose of this session is to answer any outstanding questions, discuss personal feelings about the test and prepare for different test outcomes. Details about when and how the results will be delivered and who will be present are reviewed and the first follow-up session is arranged. At the next session, results are presented by the neurologist or medical geneticist in “a clear, direct, unambiguous, non-judgmental fashion” (Benjamin, et al., 1994:609). There is an extended opportunity for questions and discussion but PT candidates and their support person(s) often appreciate having some time on their own during this session since the results may, even with adequate preparation, come as a shock. A follow-up session is conducted approximately two weeks after clinical disclosure of results. This session is designed to provide continued support and contact as PT candidates adjust to knowing the results of the test. Support then continues over the next year (or for as long as necessary).

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6 If there are concerns about the candidate’s psychiatric status, a separate counselling session may be offered and/or the candidate may be referred to an appropriate mental health professional. In addition, the possible postponement of predictive testing may be discussed.

7 Prior to the neurological examination, candidates are asked if they are aware of any symptoms or whether they are concerned that they may be showing clinical signs of HD and, since some candidates may not be ready to receive a clinical diagnosis, all are asked in advance if they would want to know that signs or symptoms were present.

8 The genetics counsellor does not, at this point, know the results of the DNA analysis since this might inadvertently shape the counselling provided.
Previous research has shown that candidates who learn that they are at increased risk for HD are more likely to have an adverse response in the first few months after results (Bloch, et al., 1992) while those who learn that they are at decreased risk may have an adverse response many months after receiving results (Huggins, et al., 1992). In person counselling is therefore recommended at six months and one year after results for those who learn that they have the HD mutation while telephone contact at six months and a personal interview at one year is indicated for those who learn that they do not have the HD mutation. In addition, a twenty-four hour contact phone number is provided as part of the ongoing service program.\(^9\) The protocol as described here is flexible and, for those who have already participated in the linkage form of predictive testing, a modification in the number of sessions may be justified.

A community-based protocol has also been developed to meet the needs of individuals who live a significant distance from the genetics centre at UBC. This “rural protocol” combines the services of the genetics centre with professional support and counselling that is available to candidates in their home community. In order to participate, the PT candidate must travel to the genetics centre in order to complete the initial counselling session and clinical assessment. All other sessions, including the results session, are conducted in the candidate’s home community by a health professional (e.g., family physician, nurse, social worker or psychologist) chosen by the test candidate. The clinical team at UBC provides this professional with information about HD and predictive testing as well as the results of the candidate’s psychological assessment. At least one pre-results session is conducted in the candidate’s home community before results are given and the candidate is given the option of visiting the genetics centre again at any time.

As mentioned above, this research was designed in order to mesh with the PT candidate’s progress through the clinical protocol. Recruitment of study participants occurred during the candidate’s first visit to the genetics centre and the first round of interviews was scheduled to occur prior to the clinical disclosure of results. This allowed for a prospective design in which interviews with all PT candidates and selected members of their family were conducted before and several months after test results.

\(^9\) This service is seldom used but PT candidates believe that it is important to maintain it (Copley, et al., 1993).
Recruitment and Participation Rate

All eligible PT candidates attending their first counselling session at the local genetic clinic during the period of recruitment (November 1993 to September 1994) were invited to participate in the research on familial impacts of predictive testing. Provided with the use of an office adjacent to the Medical Genetics Clinic in the hospital at UBC, I met briefly with each PT candidate and any family members who were also in attendance at the initial genetic counselling session. During this time I explained the goals and objectives of both the BCMSF interview study and the CGAT questionnaire study. Many PT candidates received in advance a pamphlet and letter about the study (see Appendix IV) and these materials helped to facilitate the decision to participate in the research. Some PT candidates had given specific thought to which family members they would ask to participate in the study and a few had either spoken to or passed on information about the study to these family members. Where feasible, PT candidates were encouraged to invite their spouse or partner and unaffected parent to be part of the study. Siblings or other relatives and/or close friends were, however, also invited to participate in the interview study if the PT candidate so desired. In all cases, however, it was entirely up to the PT candidate to decide which family members they wished to invite to participate in the research and, in no case were family members interviewed in the absence of the PT candidate’s participation in the study.

Test candidates and their families demonstrated a remarkable willingness to participate in the research. During the ten month period of recruitment (November 1993 to September 1994), a total of thirty-four new PT candidates attended the clinic for their first counselling session. Of a possible twenty-seven eligible PT candidates who were invited to participate in the interview study, twenty-two agreed and five declined to participate. As shown in Table 1

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10 Seven test candidates were not considered eligible for the study for reasons which are described in Table 1.
11 If the PT candidate had not received materials about the study prior to their first visit to the clinic, I provided these materials at the recruitment session and encouraged the person to take some time to consider their participation in the study.
12 A separate letter about the research was prepared for family members of PT candidates and this was provided to the PT candidate along with the letter of introduction and the brochure mentioned above.
13 In the section following the question “Which family members should I invite to participate in this study?”, the study brochure explicitly excludes “anyone under 18 years of age or anyone who has been diagnosed with Huntington Disease”. These criteria for eligibility were, however, not rigidly applied in recruiting for the interview study and two persons diagnosed with HD participated in the interview study.
(see next page), this represents a participation rate of 81% for the total BCMSF research sample.

The high level of participation in this research was attributable to a number of factors. First, all predictive testing candidates received information about the study prior to their first visit to the clinic. Study materials (a letter and brochure) were included with information about predictive testing which is routinely sent by the genetics counsellor to all new PT candidates. Second, I was able to do in–person recruitment at the clinic either just before or just after the PT candidate’s genetic counselling session. I was introduced to the PT candidate by a genetic counsellor and provided with sufficient time during the clinical schedule to discuss the research and any questions or concerns the PT candidate might have. In some cases, I was also able to sit in and observe the first counselling session with the PT candidate and their support person.

Third, many PT candidates and their families had previous knowledge of, or contact with, the clinical team at UBC. These individuals were, in general, very appreciative of the support, counselling and/or other clinical service provided at UBC and this often translated into a willingness and/or perceived responsibility to do whatever they could to reciprocate.\(^\text{14}\) In some cases, family members also had a long–standing relationship with those conducting scientific research on the genetics of HD; some had banked their DNA and/or arranged for DNA samples of other family members to be sent to the DNA bank in order to assist with research. Fourth, this research — with its stated objectives of learning about the familial and social rather than individual psychological experiences of predictive testing — tapped into a pre–existing need on the part of some family members to tell their story. Although PT candidates are encouraged to bring their spouse/partner or another support person along with them to the counselling sessions, this person seldom has the opportunity to discuss their own concerns about HD and predictive testing. Likewise, those family members who do not attend the clinic seldom have the chance to discuss, at length, their thoughts and feelings about HD and predictive testing. Finally, for those who were living outside the Greater Vancouver area, travel time was a significant factor; most PT candidates and their families therefore greatly appreciated the fact that the study design eliminated any need for them to travel to the clinic in order to participate in

\(^{14}\) There were no study participants who expressed concerns about feeling as if they had to participate in the research. Many stated that they had decided to participate in the hope that it would help others in a similar situation.
**TABLE 1**

RECRUITMENT AND PARTICIPATION RATE
(TOTAL RESEARCH SAMPLE)

Period of Recruitment for Study — November 1993 to September 1994

<table>
<thead>
<tr>
<th>Description</th>
<th>Value</th>
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</thead>
<tbody>
<tr>
<td>Total intake of new predictive test candidates</td>
<td>34</td>
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<tr>
<td>Number of test candidates not eligible for study participation&lt;sup&gt;a&lt;/sup&gt;</td>
<td>7</td>
</tr>
<tr>
<td>Number of test candidates invited to participate in study</td>
<td>27</td>
</tr>
<tr>
<td>Number of test candidates that accepted invitation to participate in study&lt;sup&gt;b&lt;/sup&gt;</td>
<td>22</td>
</tr>
<tr>
<td>Number of test candidates that declined invitation to participate in study&lt;sup&gt;c&lt;/sup&gt;</td>
<td>5</td>
</tr>
<tr>
<td><strong>STUDY PARTICIPATION RATE (22/27)</strong></td>
<td>81%</td>
</tr>
</tbody>
</table>

<sup>a</sup> Reasons new predictive test candidates considered ineligible for study: results date scheduled too soon (2); service protocol atypical because candidate had been through prenatal and linkage forms of test (1); candidate at 25% vs. 50% risk and at risk parent uncertain about test (1); psychiatric referral required (1); probable diagnosis of HD (1); other (unknown) extenuating circumstances (1).

<sup>b</sup> With the exception of one brother and sister, all predictive test candidates are from different families. Of those who agreed to participate in the interview study, 20 also initially agreed to participate, with at least one family member, in the questionnaire study. (13 of these 20 families completed the baseline questionnaires, although some follow-up packages were not returned.)

<sup>c</sup> With one exception, all candidates who declined to participate were male. Their reasons for declining were not, in most cases, explicit. One candidate stated that no-one other than his wife knew that he was having the test. He, and three others who declined participation in the interview study, said they were willing to do the questionnaires only. Two declined to participate in both the interview and questionnaire studies.
the study. Further, regardless of place of residence, participants were pleased to be able to do the interviews in the comfort and privacy of their own homes.15

In summary, the willingness of the clinical team to integrate the process of study recruitment into the protocol for predictive testing ensured that I had a timely opportunity for face–to–face contact with all potential study participants. Moreover, because the study was endorsed by the clinical and research teams at UBC, PT candidates and their families appeared to place a high degree of trust and confidence in me as a researcher affiliated with the clinic. Study participants’ perceptions of my role and/or affiliation with the genetics clinic were, however, also significant in shaping subsequent interactions during the interviews. Clearly, the initial perception that I was, in some capacity, part of the clinical team had important implications for how I managed the on–going research relationship and how, in turn, study participants formed and revised their initial perceptions of me as a researcher and their expectations of the interview as a structured form of conversational interaction. In addition, my affiliation with the genetics clinic raised a significant ethical and methodological issue (which I discuss in greater detail below): many study participants assumed that I would have relatively unimpeded access to information contained in their clinical files and some assumed that I would have the ability to give medical advice and information.

Ethical Considerations

This research received ethical approval under the auspices of the protocol submitted for the BC Medical Services Foundation study; this was later modified to include the long–term follow–up interviews. Full provisions for informed consent, confidentiality and security of data were included in the application for ethical review. Copies of the letter(s) of introduction and consent form(s) are included in Appendix IV and V. Here I wish to highlight several aspects of the research design which are salient to “doing research on sensitive topics” (Lee, 1993).

First, it was important to negotiate meaningful informed consent. The chief problem I found was that study participants tended to sign the consent form quickly as if it were a

15 There were minor exceptions. Members of one large family preferred to meet in a conference room in the hotel where I was staying. On another trip (to interview a husband and wife in a different region of the province), winter driving conditions made it more practical to conduct the interviews in the hotel where I was staying.
formality to be dispensed with. In comparison with the consent required at the clinic (to proceed with predictive testing), it may have seemed a relatively minor thing to consent to be part of the interview study. Participants were, for the most part, relatively unconcerned that the interviews would stir up painful thoughts and feelings. Issues of confidentiality on the other hand, were more likely to become focal points for discussion. For instance, many study participants asked, at the beginning of an interview, about who would have access to the data. Some made jokes about obtaining access to tape recordings of other family members’ interviews but most observed stringently their family member’s need for privacy during the interview itself. As mentioned above, the PT candidate was always the index person for each family and no contact was made with any other family members without the express permission of the PT candidate. This was particularly germane to the recruitment phase of the research but some PT candidates reiterated over the course of the research that certain persons in their family were not, under any circumstances, to be contacted.

At the beginning of each interview I reminded study participants that they could, at any time, request that the tape recorder be turned off. Few actually made such requests but many participants alluded to or directly flagged certain pieces of information as “off the record.” Others were more circumspect and simply declined to follow-up on a question or probe if it seemed to be leading them into an area they would rather not discuss on tape. Many of these issues later surfaced once I had turned off the tape-recorder.

Second, the interviews provided an opportunity for PT candidates and their families to speak candidly about things which, in some cases, they had never spoken at length about before. As such, some commented that the interview had a therapeutic effect. A “good” interview did, however, also have confessional elements and I had to be extremely cautious in how I managed my increasing awareness of each family. In order to maintain confidentiality it was essential to know how and from whom I knew each piece of information. I did not want to inadvertently reveal something which was told to me in confidence but nor could I prevent study participants from making inferences and drawing sometimes erroneous conclusions based on what I did say. The actual test results were the most obvious example but many other issues were equally sensitive (e.g., the scheduled date for learning test results).
As noted above, there were ethical implications that flowed from PT candidates' assumptions that I would have access to their clinical files. In particular, it was often assumed that I would immediately be appraised of their PT results when, in fact, this was not the case. Test results were not automatically provided to me by virtue of my connection with the PT Research Group although the genetic counsellor did, with permission of the PT candidate, disclose to me the test result.

Where PT candidates and/or members of their family indicated that they were in need of more information about HD, I provided a pamphlet about the Huntington Society of Canada (HSC) and contact information for the local Huntington’s Disease Resource Centre. I also carried with me copies of the HSC newsletter Horizon and if people were unfamiliar with it I described the publication and encouraged them to subscribe. In summary, the intention was to provide basic information about HD and existing resources where appropriate but to otherwise intervene as little as possible.

Nonetheless, the interviews were not only about people’s lives, they were an event in people’s lives. Residents in rural communities planned trips into town around my arrival; family get-togethers were sometimes interrupted and, in one case, a woman on the rural protocol postponed receiving her test results until after I could travel to her home community and conduct a pre-results interview with her. I did not know that this was the case until I arrived. I was speechless. Her dedication to the research went beyond reasonable expectation and yet it was also emblematic of the profound commitment exhibited by all participants in the research.

Selection and Demographics of Study Sample

The research described in this dissertation is based on a sample of sixteen PT participants and thirty-three of their family members and/or close friends. In addition, I also interviewed one married couple where the wife was at risk for HD but did not want to proceed with predictive testing. Together these forty-nine study participants represent seventeen different families who, with few exceptions, live in urban and rural settings in the province of

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16 Material from these two interviews is integrated within some aspects of the analysis (i.e., the section on why at-risk individuals did and did not want predictive testing) but it is not otherwise singled out for special analysis.

17 One brother and sister were both predictive test candidates and family members during the course of this research and hence, are included twice.
British Columbia. This study sample was selected from the total research sample of twenty-one families recruited through the clinic for the BCMSF study and includes all of the PT candidates and family members that I have interviewed during the course of this research.

Tables 2 and 3 (see next pages) describe, respectively, the distribution of family members in the total BCMSF research sample and the selected study sample for this research. The majority of family members were, in both samples, spouse/partners or siblings of the predictive test candidate although there was a good cross-section of parents, children, other family and friends who also agreed to participate in the research. This diversity is a significant strength in that it contributes to this research an array of differing perspectives on familial experiences of predictive testing for HD. It does, however, also illustrate the problematic nature of attempting to define, for research purposes, who is and is not an eligible member of the family. Recruiting only the unaffected parent and spouse/partner (as per the guidelines for the CGAT funded quantitative study mentioned earlier) might have permitted greater comparison between families but it would also have diminished the range and representativeness of themes arising from the resulting data. As such, I find Gilgun’s (1992:24) “lean definition” of family to be a more appropriate reflection of the approach taken here: "the subjects of the research are persons who mutually define themselves as family, are in committed relationships, have a shared sense of personal history, and who usually but not always have legal and biological ties."

Within the context of this research, it was important not to impose a narrow definition of family but rather to allow families and, more specifically, the PT candidate, to say who was and was not part of their family and who it would (and would not) be important and acceptable to include in the research. Families often have spokespersons or gatekeepers who present a certain picture of the family to the outside world and these spokespersons are often women. This

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18 One predictive test candidate and his brother were from the United States and the brother of another predictive test candidate was from Alberta.
19 The woman who did not wish to proceed with predictive testing was recruited through a short advertisement about the study which was included in the newsletter of the B.C. Chapter of the Huntington Society of Canada. This ad was designed to encourage those who did not want predictive testing to be part of the study; it attracted only one response. The text of this advertisement is included in Appendix III.
20 Dr. McKellin interviewed the remaining six PT candidates and their family members as well as the brother of one PT candidate that I interviewed. The brother lived in the U.S.
21 I know of only one family member that declined to participate in the study when invited to by the PT candidate.
TABLE 2
DISTRIBUTION OF FAMILY MEMBERS
(TOTAL RESEARCH SAMPLE)

Relationship of Family Members to Predictive Test Candidate

<table>
<thead>
<tr>
<th>Relationship of Family Members</th>
<th>Total</th>
<th>Female</th>
<th>Male</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spouse/Partner</td>
<td>16</td>
<td>6</td>
<td>10</td>
</tr>
<tr>
<td>Wife/Female</td>
<td>16</td>
<td>6</td>
<td>10</td>
</tr>
<tr>
<td>Husband/Male</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Parents</td>
<td>3</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>Mother</td>
<td>2</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Father</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Siblings</td>
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<td>5</td>
<td>6</td>
</tr>
<tr>
<td>Sister</td>
<td>11</td>
<td>5</td>
<td>6</td>
</tr>
<tr>
<td>Brother</td>
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</tr>
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<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Son</td>
<td>2</td>
<td></td>
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<td>Other Family</td>
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</tr>
<tr>
<td>Male</td>
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<td></td>
<td></td>
</tr>
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<td></td>
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<td>Other*</td>
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<td>1</td>
</tr>
<tr>
<td>Female</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Male</td>
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<td></td>
</tr>
<tr>
<td>TOTAL FAMILY MEMBERS</td>
<td>45</td>
<td>24</td>
<td>21</td>
</tr>
</tbody>
</table>

*a Includes woman who did not want predictive testing and her husband.*
TABLE 3
DISTRIBUTION OF FAMILY MEMBERS
(SELECTED STUDY SAMPLE)

Relationship of Family Members to Predictive Test Candidate

<table>
<thead>
<tr>
<th>Relationship of Family Members to PT Candidate</th>
<th>Male</th>
<th>Female</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spouse/Partner</td>
<td>8</td>
<td>3</td>
</tr>
<tr>
<td>Parents</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>Siblings&lt;sup&gt;a&lt;/sup&gt;</td>
<td>5</td>
<td>4</td>
</tr>
<tr>
<td>Children&lt;sup&gt;b&lt;/sup&gt;</td>
<td>2</td>
<td>1</td>
</tr>
<tr>
<td>Other Family&lt;sup&gt;c&lt;/sup&gt;</td>
<td>1</td>
<td>4</td>
</tr>
<tr>
<td>Friends</td>
<td>0</td>
<td>2</td>
</tr>
<tr>
<td>Other&lt;sup&gt;d&lt;/sup&gt;</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>TOTAL FAMILY MEMBERS</td>
<td>18</td>
<td>17</td>
</tr>
</tbody>
</table>

<sup>a</sup> All siblings of PT candidates interviewed for this research were themselves at risk for, or already diagnosed with, HD. None of the PT candidates were only children; all had at least one sibling.

<sup>b</sup> All at 25% apriori risk.

<sup>c</sup> Other family includes: 1 grandmother, 1 honorary "aunt" and her daughter, 1 nephew and his wife.

<sup>d</sup> Includes woman who did not want predictive testing and her husband.
has significant implications for who will and will not participate in interview-based research; as Daly (1992) points out, men are typically more difficult to recruit to qualitative family studies than women. I am, however, pleased to note that half of the family members recruited for this study were male.

The decision to focus my analysis on only the families I interviewed is justified for several reasons: *first* and foremost, this research required that I have a close rapport with study participants. Face-to-face interaction was an essential component in understanding the content of, and context for, much of what was said during the interviews. Even the best transcript is a very poor substitute for actually sitting and talking with someone in person. *Second*, this research is, as I have already mentioned, also concerned with the interview as an actual instance of communication about genetic information. How I felt as an interviewer and “friend of the family” and how I responded to what was being said is, therefore, important data in and of itself: arguably, much research on self-disclosure has paid too little attention to the issue of the listener’s response to disclosure. *Third*, the selected study sample — of sixteen PT candidates, thirty-three family members and two others (i.e., woman who did not want predictive testing and her husband) — participated in a total of 102 separate interviews and these interviews resulted in approximately 5,000 pages of transcripts and fieldnotes. This data set was more than adequate in generating new insights about “what was going on” and, well before the last interview, standard indices of saturation — such as repetition and confirmation of emerging themes — became apparent (Morse, 1994).

Demographic information on the PT candidates and family members in the selected study sample is summarized in Tables 4 and 5 (see next page), and briefly reviewed here. Three-quarters of the PT candidates were female. This is slightly higher than the 2:1 female: male ratio which has been consistently observed in many genetic centres offering linkage and direct testing during the last decade but if the total BCMSF sample is considered, there is an almost exactly 2:1 ratio (fifteen female, seven male). The mean age of the sixteen PT candidates

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22 Much of this information is drawn from the demographic questionnaire which all PT candidates filled out during their first visit to the clinic. Family members who were participating in the CGAT-funded questionnaire study also provided this information and, where there were gaps, I referred to the interview transcripts.
| TABLE 4 |
| BASELINE DEMOGRAPHIC INFORMATION FOR PREDICTIVE TEST CANDIDATES |
| (SELECTED STUDY SAMPLE) |

Total Number of Predictive Test Candidates (N) = 16

<table>
<thead>
<tr>
<th>Sex</th>
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<tbody>
<tr>
<td>Female</td>
<td>12</td>
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<tr>
<td>Male</td>
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<table>
<thead>
<tr>
<th>Age</th>
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</thead>
<tbody>
<tr>
<td>Mean</td>
<td>41.8</td>
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<tr>
<td>Range</td>
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<table>
<thead>
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<th>Marital Status</th>
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<tbody>
<tr>
<td>Single</td>
<td>4</td>
</tr>
<tr>
<td>Married</td>
<td>10</td>
</tr>
<tr>
<td>Divorced &amp; now living common-law</td>
<td>2</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>No. of Offspring</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>None</td>
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</tr>
<tr>
<td>One</td>
<td>1</td>
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<tr>
<td>Two</td>
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<td>Three</td>
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<td>Four</td>
<td>1</td>
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<tr>
<td>Five</td>
<td>1</td>
</tr>
<tr>
<td>Total No. Offspring</td>
<td>27</td>
</tr>
</tbody>
</table>

<table>
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</thead>
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<tr>
<td>Other British Columbia</td>
<td>7</td>
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<tr>
<td>United States</td>
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</table>

<table>
<thead>
<tr>
<th>Education</th>
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<tbody>
<tr>
<td>&lt; High school</td>
<td>2</td>
</tr>
<tr>
<td>High school graduation</td>
<td>4</td>
</tr>
<tr>
<td>Vocational training</td>
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</tr>
<tr>
<td>College</td>
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</tr>
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<td>University</td>
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<table>
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<tr>
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<tr>
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<tr>
<td>Retired</td>
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<tr>
<td>Unemployed</td>
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</tr>
<tr>
<td>Not employed (by choice)</td>
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<table>
<thead>
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<th>Occupation</th>
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<td>Professional</td>
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</tr>
<tr>
<td>Entrepreneur/sales</td>
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</tr>
<tr>
<td>Office work/clerical</td>
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<tr>
<td>Skilled Trades</td>
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<table>
<thead>
<tr>
<th>Religious Affiliation</th>
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<tr>
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<td>Christian Alliance</td>
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<td>Not applicable</td>
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TABLE 5
BASELINE DEMOGRAPHIC INFORMATION
FOR FAMILY MEMBERS
(SELECTED STUDY SAMPLE)

Total Number of Family Members\(^a\) (N) = 35

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<tr>
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</tr>
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<td>Divorced &amp; now living</td>
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<tr>
<td>common-law</td>
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<td>Widowed</td>
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<table>
<thead>
<tr>
<th>No. of Offspring</th>
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<tr>
<td>None</td>
<td>12</td>
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<td>One</td>
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<td>Four</td>
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<tr>
<td>Five</td>
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<table>
<thead>
<tr>
<th>Residence</th>
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<td>Greater Vancouver</td>
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<thead>
<tr>
<th>Education</th>
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<td>&lt; High school</td>
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</tr>
<tr>
<td>High school graduation</td>
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<tr>
<td>Vocational training</td>
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<td>College</td>
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<td>University</td>
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<th>Employment</th>
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<td>Part-time</td>
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<td>Retired</td>
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<td>Unemployed</td>
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<td>Not employed (by choice)</td>
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<td>Seasonal employment</td>
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<table>
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<th>Occupation</th>
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<td>Professional</td>
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</tr>
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<td>Entrepreneur/sales</td>
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<td>Office work/clerical</td>
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<td>Skilled Trades</td>
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</tbody>
</table>

\(^a\) Includes brother and sister who were also predictive test candidates; woman who did not want predictive testing and her husband..
who were part of the selected study sample was 41.8 years and the range was 23 to 57 years. Most candidates were, at the time of their pre-results interview, either married (N=10) or divorced and now living common-law (N=2) and two others had a significant relationship involving the possibility of marriage. None were openly gay, lesbian or bisexual. More than half had at least one child and several had two or three children; the total number of children of all PT candidates was twenty-seven. Of the ten who had at least one child, four also had their own biological grandchildren. When all of these children and grandchildren are considered, it becomes abundantly clear that predictive testing may result in a modification of risk which has significant implications for many other members of the family.

Half of the sixteen PT candidates lived in the Greater Vancouver area and half lived in smaller communities throughout the province of British Columbia. One PT candidate was from the United States but maintained dual citizenship and had family living in BC. All attended the genetics centre located at UBC but of the eight PT candidates who lived outside the Greater Vancouver area, four chose to participate in the rural protocol outlined above and thus four received their results from a health care professional located in their home communities. Having access to PT candidates who opted for the rural protocol greatly enriched this research since little was known about how participants felt about this modified protocol. Most PT candidates were formally fairly well-educated; all but two had completed high school and ten had gone on to complete vocational training, college or university. Most candidates were employed either full-time (N=9) or part-time (N=2) while several were unemployed (N=2) or not employed by choice (N=2) and one was retired. Occupational categories represented in the sample included office/clerical workers (N=7), skilled tradespersons (N=3), entrepreneurs or professionals (N=3) and homemakers (N=2). Only one-quarter (N=4) of candidates reported engaging in regular religious practice and of these three indicated a Protestant affiliation.

For the family members participating in this study, the ratio of female to male was almost 1:1. The mean age of these thirty-five family members was, at 42.9 years, only slightly

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23 This question was not explicit on the demographics questionnaire (or within the interviews) but neither were any of the questions on current relationships/marital status worded in such a way as to presume heterosexuality.
higher than that of the PT candidates themselves but the range was greater (i.e., 22 to 72 years). Most family members were, at the time of their pre-results interview, either married or living common-law (N=21); ten were single, one was separated and two were widowed. There were no openly gay or lesbian participants although one woman described herself as bisexual. More than two-thirds had at least one child and several had three or four children.

More than two-thirds of the family members lived outside the Greater Vancouver area (N=23), one lived in Alberta and one in the United States. All but four had completed high school and seventeen had gone on to complete vocational training, college or university. Most were employed either full-time (N=18) or part-time (N=5) while several had seasonal employment (N=5) or were retired (N=2). Two were not employed by choice (N=2) and none were unemployed. Occupational categories represented in the sample included skilled tradespersons (N=6), entrepreneurs (N=4), homemakers (N=4), professionals (N=4), office/clerical workers (N=3), service workers (N=2) and students (N=5). Roughly one-quarter (N=8) reported regular religious practice and, of these, five indicated Protestant affiliation.

It is more difficult to describe the socio-economic status of PT candidates and their family members since I did not have a complete set of information about annual household and/or personal income.24 Perhaps it may suffice to say that although there were two PT candidates who were living at or near the poverty line, most individuals interviewed in this study appeared to have at least an average or better than average income and an adequate if not relatively affluent standard of living.

Finally, it is important to mention two other aspects of the study sample which are not included in Tables 4 and 5. Study participants were, in general, almost exclusively of Western European ancestry and/or origin. Many were born in Canada or the U.S. but several had emigrated as young adults (from England, Scotland, Switzerland and Denmark) and two spouse/partners of PT candidates had emigrated as adolescents (when their respective families moved to Canada from Poland and Burma). All spoke English fluently although three learned English as a second language when they arrived in Canada (from Denmark, Poland and Burma).

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24 This question was optional on the baseline demographics questionnaire filled out by PT candidates and family members who participated in the CGAT funded questionnaire study.
Timing and Structure of Interviews

One of the strengths of this research is its prospective design. In-depth semi-structured interviews with PT participants and their family members were conducted both before and after clinical disclosure of PT results. With only one exception, all study participants completed at least one pre-results interview which was, in most cases, conducted several weeks prior to results. A second interview was conducted four to six months after predictive testing and with one exception, all predictive test candidates completed this interview. In addition, six predictive test candidates and several members of each of their families participated in a third interview (conducted one to two years after results). Table 6 (next page) shows the number of individuals who participated in pre and post-results as well as long-term follow-up interviews during the period of data collection for this study (December 1993 to March 1997).

With few exceptions, all study participants were interviewed individually in their own home (or home communities). Interviews lasted anywhere from one to three hours and, with permission, were tape recorded and transcribed for subsequent analysis. The interviews covered a consistent set of themes although these themes were not set out in the form of a prescribed sequence of questions. During the pre-results interview, study participants were prompted to talk about their current family and working life, memories of growing up, relationships with siblings and others, perceptions and awareness of HD and other health-related concerns, recollections of finding out about the family history, how they heard and felt about predictive testing and, if applicable, their experiences of attending the medical genetics clinic. During the post-results interviews, study participants were invited to describe the time leading up to the clinical disclosure of results, their concerns and/or strategies for managing related stress, who they turned to for support and how people responded, what it was like to learn the results, who this information was shared with and, if applicable, any further reflections on their experiences of attending the medical genetics clinic.

Interview schedules outlining the themes covered in the pre and post-results interviews appear in Appendix VI although it is important to stress that these schedules were used only as

25 In several cases it was more convenient to meet at a family member's home and in a very small number of instances it was more convenient to utilize a hotel meeting room.
### TABLE 6
TIMING OF INTERVIEWS (102)
(SELECTED STUDY SAMPLE)

<table>
<thead>
<tr>
<th></th>
<th>Predictive Test Candidates</th>
<th>Family Members</th>
<th>Other&lt;sup&gt;a&lt;/sup&gt;</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Pre-Results</strong></td>
<td>16</td>
<td>31&lt;sup&gt;b&lt;/sup&gt;</td>
<td>2</td>
</tr>
<tr>
<td><strong>Post-Results</strong></td>
<td>15&lt;sup&gt;c&lt;/sup&gt;</td>
<td>28&lt;sup&gt;d&lt;/sup&gt;</td>
<td>n/a</td>
</tr>
<tr>
<td><strong>SUB-TOTAL</strong></td>
<td>31</td>
<td>59</td>
<td>2</td>
</tr>
<tr>
<td><strong>Minus Duplicate Counting</strong> (if applicable)</td>
<td>31</td>
<td>56&lt;sup&gt;e&lt;/sup&gt;</td>
<td>2</td>
</tr>
<tr>
<td><strong>Long Term Follow-up</strong> (Selected sub-sample only)</td>
<td>6</td>
<td>7</td>
<td></td>
</tr>
<tr>
<td><strong>TOTAL (102) INTERVIEWS</strong></td>
<td><strong>37</strong></td>
<td><strong>63</strong></td>
<td><strong>2</strong></td>
</tr>
</tbody>
</table>

<sup>a</sup> Single interviews conducted with woman who did not want to have predictive test and her husband. Not recruited through clinic.

<sup>b</sup> One pre-results interview with family member not possible due to their vacation plans. Two post-results interviews were conducted, one immediately upon their return from vacation and one several months later. Second pre-results interview missing because person not originally identified by predictive test candidate as potential study participant.

<sup>c</sup> One predictive test candidate declined to participate in post-results interview because she found it very difficult to talk about HD.

<sup>d</sup> Includes one interview conducted by Dr. McKellin. Missing post-results interviews with 6 family members due to: withdrawal from study (1); distance and inability to travel (2); inability to schedule convenient time (3).

<sup>e</sup> Actual number of interviews with family members is reduced by 3 because one was conducted with two family members at the same time and two others were dual purpose interviews (with brother and sister who were both predictive test participants and family members).
checklists for ensuring that important areas were covered. In all cases study participants were actively encouraged to talk about what they felt was most important and to frame this in whatever ways seemed most salient to them at the time. This approach enhances the validity of participant accounts by allowing participants themselves to pattern the timing, sequence, content, and context of topics discussed (Mishler, 1991).

**Fieldwork and Other Sources of Data**

The in-depth interviews I conducted with study participants constitute the primary source of data for this study. I did, however, also have access to the demographic questionnaires completed by PT candidates and their families (as part of clinical assessment and/or the CGAT funded study mentioned above). These materials provided useful background information and assisted, in some cases, in developing the demographic profile of PT candidates and family members.

Before considering in greater detail the process of interviewing and how it varied according to the life circumstances of the teller and the situation of the telling, I need to also mention briefly several other sources of data and, in particular, the significance of having the opportunity to observe a number of genetic counselling sessions and participate, as a volunteer staff person, at the annual week-long Retreat for persons with HD.

In the early stages of this research, I had the opportunity to sit in on and observe eleven genetic counselling sessions at the UBC genetics centre. These sessions were all part of the predictive testing protocol and they were conducted by two different counsellors who had or were in the process of acquiring specialized experience in working with individuals at risk for HD. Nine of these sessions were pre-results sessions, one was a results session and one was a 2 week post-results follow-up. With the exception of two pre-results sessions, all sessions were with five PT candidates who were part of this research. As such, my observations were of considerable assistance in acquiring background knowledge about the PT candidate’s family history and reasons for deciding to have the predictive test. In addition, the opportunity to observe these sessions provided an essential backdrop for understanding the dynamics of professional and lay communication about genetic information.
I did not take any notes or ask any questions during the sessions since the goal was to be as non-intrusive as possible. I did, however, write up fieldnotes after the session was over and, during the recruitment and early phases of interviewing, I relied on these notes as a means of familiarizing myself with the topics and themes which seemed to elicit the most discussion from PT candidates and their support person. I also noted how the genetics counsellors tactfully initiated topics of discussion and framed their questions or probes in an affirmative way. One counsellor, in particular, was very skilled at creating rapport and making her clients feel comfortable in talking about difficult issues. Her style contrasted with that of the geneticist who tended to ask blunt questions and probe further if he thought something was being withheld. Often he was rewarded with a crucial piece of information. These contrasting styles were instructive and I learned early on that one needs to be extremely astute when attempting to gauge when and how much to probe. These and other insights added immeasurably to the ease with which I was able to begin the process of interviewing.

During this research, I also participated, as a volunteer staff person, at five of the annual Huntington Society Retreats. The Retreat is organized by Susan Tolley (Director of the HD Resource Centre) and runs for six days in the early summer each year. The purpose of the Retreat is to provide persons with HD with the opportunity to get away from the stresses of family life, relax and engage in a range of creative, recreational and therapeutic activities which would not ordinarily be available to them. In addition, the Retreat provides some respite for family members who may often feel that their own needs are overshadowed by HD.

There are usually ten to fifteen people who are mid-stage with HD at the Retreat. Staff are there to assist with all aspects of the Retreat but we also participate in the full range of daily activities and events (e.g., water basketball, art and music therapy) along with people who have HD. The Retreat is, however, about much more than finding enjoyment and relaxation; it is a place where staff and people with HD come together as if we were an extended family. As a part of this family, I learned firsthand about many aspects of the disease which I might otherwise have been unaware of. For instance, I observed how difficult it was for some people with HD to carry on a ‘normal’ conversation and, in response, I learned how to modify and direct conversation so that it was possible to engage in a meaningful exchange. This was an important
aspect of developing communicative competence within the HD community. Being at the Retreat also had practical implications for my ability to establish rapport with interview participants; several knew about the Retreat because they had family members who had also attended and, in more than one instance, my involvement in the Retreat mattered to study participants precisely because it suggested that I already knew something about the experiences that I was so intent on inquiring about.

The experience of being at the Retreat was (and is) an honour I find both compelling and impossibly difficult to write about. I wrote extensive fieldnotes each year but I find it difficult to treat the experience as if it were a form of participant-observation. There is a circle of confidence which is respectfully drawn around specific interactions that occur at the Retreat but at the same time, it is important to acknowledge that I was there, in part, to learn and that it was people with HD who taught me about what it means to live with the disease, how it feels to inhabit an unfamiliar body, what it is like to grieve the inevitable losses and yet live each day with courage and dignity. Without having been part of the Retreat, I would not have known what it is to care deeply about people who have HD and worry about what it is like for them to live in a world which is often ignorant and unkind. Moreover, I would have been less attentive to the strong emotions which are shielded by the armor we all wear in our everyday lives. I might well have missed knowing how very unusual (and precious) it really is when mutual trust allows people to (however temporarily) shed that armor and speak openly with each other.

Communication, and even what we might on first glance regard as self-disclosure, is often about keeping one’s armor in place. As Pearce and Sharp (1973:415) point out, “some but not all divulging of personal information is an attempt to make the speaker known to his [or her] listeners.” History is a type of communicative strategy in which “past events rather than present experiences, actuarial detail rather than subjective responses, and intellectual interpretation rather than personal involvement” are presented in a detached manner which forbids the listener from becoming overly involved. Honesty, on the other hand, is characterized by the speaker’s description of experiences which invite the listener to share and respond in an empathetic manner. Honest messages may or may not be totally accurate or “true” but they are “a sincere attempt to make the speaker known to the listener.”
The distinction between history and honesty is, of course, not absolute; communication about personal information is a mixture of these two strategies. The distinction does, however, remain a useful one since it reminds us that self-disclosure is never an individual behaviour or attribute; it occurs within the context of the relationship which exists between speaker and listener. It has to do with what Mishler calls the “situation of the telling”.

The Interviews

I have already described the process by which study participants were recruited and, within this context, alluded to the significance of meeting with study participants in a clinical setting. Here I want to elaborate on how the interviews initially bridged and eventually crossed the gulf which exists between the clinic and the everyday lifeworld of PT candidates and their families. Before proceeding any further, however, it is necessary to consider how the communicative interaction of medical settings differs from that which I sought to jointly construct with the participants in this research. This aim requires a short detour.

The Voice of Medicine and the Voice of the Lifeworld

Drawing upon transcripts of doctor–patient encounters, Mishler (1984) demonstrates that the structure of medical interviews often constrains patients’ abilities to tell their stories in a coherent fashion. Within the “voice of medicine”, the structure and sequence of the doctor’s questions provide a normative order which disrupts and redirects the patient’s attempts to tell a story. Communication difficulties are then a product of speaking practices which affirm the dominance of one set of values over another. Thus, it follows that patient dissatisfaction may derive from the practices doctors and other health care professionals employ in extracting from the patient only that information considered relevant to the goals of the medical encounter.

Standard research interview practices may emulate the patterns of interviewer dominance and respondent acquiescence which Mishler (1991:54) has documented in medical interviews. “Although medical interviews differ from standard research interviews in several important respects, the two are similar in that both are methodical inquiries in which interviewers through a series of questions attempt to elicit ‘relevant’ information from respondents.” The attempt to follow closely a standardized interview schedule and ask of each
respondent the same questions in the same predetermined order does, however, jeopardize the meaningfulness of the entire enterprise since it, like the sender–receiver model of communication, is premised on the view that each question constitutes a stimulus which should, independent of context, be received and understood by the respondent in a similar if not identical fashion. Likewise, the attempt to standardize interview questions excludes from consideration the manner in which the interview itself is a "developing discourse" (Mishler, 1991) which shapes and is shaped by the unfolding sequence of exchanges between interviewer and respondent. If, however, the interview is viewed as a speech event or activity, the natural variation in how particular questions are asked and answered becomes part of the object of inquiry rather than a source of bias. In short, where the interviewer (like the doctor) adopts a narrowly prescribed interviewing structure in the attempt to elicit only 'relevant' information from the respondent, it is unlikely that the communicative exchange will result in the production of narrative accounts which are reflective of the lifeworld of the respondent.

At this juncture it is useful to define more precisely what is meant by the notion of relevance. Noting that most exchanges of talk do not consist of a series of disconnected remarks, Grice (1989) draws attention to the way in which each participant must adhere to some common purpose or sense of mutually agreed upon direction. This purpose may evolve throughout the exchange but there will always be a number of possible conversational moves that are, in some way, inappropriate to the exchange. Roughly speaking, participants are therefore expected to broadly observe what Grice (1989:26) refers to as the "Cooperative Principle."

Make your conversational contribution such as is required, at the stage at which it occurs, by the accepted purpose or direction of the talk exchange in which you are engaged.

Grice distinguishes four maxims which support the "Cooperative Principle." The first three have to do with the content of what is said: "Quantity" specifies that each contribution should be as informative as is required but not more so; "Quality" specifies that each contribution should be true (that is, do not say that which you believe to be false or that for which you lack adequate evidence); "Relation" specifies that each contribution should be relevant. The fourth maxim, "Manner," is concerned with "how what is said is to be said" and it proscribes obscurity and ambiguity of expression while prescribing brevity and orderliness.
Conceding that these maxims of “truthfulness, relevance, informativeness and manner” are now “part of the standard machinery for pragmatic analysis,” Wilson and Sperber (1987:155) propose that they may, in most instances, be reduced to a single principle — the principle of relevance. Relevance is “a relation between the proposition expressed by an utterance, on the one hand, and the set of propositions in the hearer’s accessible memory on the other” (Wilson & Sperber, 1987:169, emphasis added). Being relevant is, therefore, a matter of saying that which will encourage the hearer to expand upon or modify an initial set of beliefs or assumptions as they have been expressed in a preceding utterance (or string of utterances); if the speaker wishes to obtain the maximum possible effect on the hearer’s initial assumptions (i.e., have some impact), relevance is, however, also a matter of assessing the inferences which the hearer is likely to make at any given point within the conversational exchange.

Obtaining an understanding of the lived experiences of PT participants and their families was, from the beginning, one of the primary objectives of this research. The process of gaining access to these perspectives and experiences did, however, mean that I had to move out of the clinic and away from the expectations of relevance which tend to predominate within a clinical context. This is not intended as any form of slight toward those who provide clinical genetics services nor am I implying that the stories that people tell when they are in the clinic are in any way inaccurate or untruthful. What I am suggesting is that the stories people tell when they are “probands”, patients or clients in the medical genetics clinic differ from the stories these same people tell when they are busy living their everyday lives. As Conrad (1990:1260) argues,

Much research on illness experience has assumed that studying patienthood from the patient’s perspective — especially doctor–patient interaction — is the same as studying illness experience. It is not. Conrad’s injunction implies that researchers interested in understanding illness experience must turn away from provider–patient interaction as the central analytic focus and go beyond medical settings in order to focus on how people manage their illness when they are at home or work or doing whatever they do when they are not patients. Indeed, researchers interested in understanding the experience of living at risk for an hereditary disease might heed such advice even more rigorously since even those who learn, through predictive testing, that they are at increased risk of developing the disease are not, in most cases, symptomatic or any way unwell.
Bridging

Within the context of this research, it was significant that my first interaction with many study participants occurred in a clinical setting. As mentioned above this was an asset for the purposes of recruitment. Further, the research would not have been possible without some form of on-going affiliation with the clinic and, more specifically, the genetic counsellors and other service providers whom I turned to for advice (e.g., when an interviewee disclosed that a PT candidate had openly discussed suicide with her) and whom I depended on for information (e.g., about the scheduling of results) and assistance (e.g., in obtaining an address or telephone number for a study participant who had moved). On a practical and ethical level then, the clinical connection was essential. This said, however, my affiliation with the clinic also had a significant influence on: 1) how I conceptualized my relationship with study participants, 2) how study participants conceptualized their relationship with me, 3) the expectations study participants had about the nature of the interview as a communicative event, and 4) the assumptions that study participants had about my access to clinical files and resources as well as information about their predictive test results.

I will not dwell on the issue of how I conceptualized my relationship with study participants since I have, in Chapter 1, already described myself as an atypical friend of the family. This way of characterizing myself in relation to study participants was not something that I had in mind when I began doing the interviews but over time, it began to fit and I believe, in many cases, it also fairly represents the way that study participants eventually came to conceptualize their relationship with me.26 The stress here is on eventually although there was one instance, early on in the research, in which a respondent invited me to think about those dimensions of the research relationship which might, outside of the formality of an interview, be properly conceived of as friendship. As I later wrote in my fieldnotes under the heading “post-interview comments and observations,”

After the interview, we had tea. I talked about the research and her son talked about his studies. She seemed happy to sit and relax. She wanted to know about my research and asked about whether my parents knew someone that she knew. I needed to leave but it was difficult to do so. It

26 Oakley (1981) makes a similar point within the context of a discussion of the implications of doing repeated interviews with study participants who are at some critical stage in their lives.
seemed that there was a lot that she could still talk about. On the way out, she said that she had been apprehensive about the interview but that she was really glad I came, that I seemed like a friend who had come to visit. She wanted to know if I would be doing the other interview as well and said she hoped so, the interview was therapeutic for her. She seemed to not want me to leave. I gave her a hug and thanked her. I said I would be in touch with her again in a couple of months.

During the first round of interviews, which took place a few weeks before clinical disclosure of the test results, many study participants assumed that I was part of the staff of the genetics clinic and/or that I worked for the Huntington Society. In part, such confusion may have arisen from the fact that I often met with study participants in the HD Resource Centre since it was the only available space during a hectic clinic schedule. In addition, I have the same first name as the Director of the Resource Centre, Susan Tolley. In short, I spent a fair bit of time explaining that I was a sociologist, that I was not trained in genetics and that I was working in conjunction with researchers at the clinic on a research project which was also the topic of my dissertation. Such explanations also served as disclaimers when study participants asked me to explain the latest research developments or describe some of the intricacies of molecular biology although I now realize that my disavowal of such expertise was not, from my study participants' point of view, entirely justifiable. For those who lacked social and/or geographic proximity to information about HD and/or the opportunity to talk with a knowledgeable "professional", the expectation that I would begin to fulfill some of these needs must have been acute. For instance, when I travelled to a small town in a remote region of the province, several study participants anticipated that I would provide information and counselling. I had a difficult time retaining a sense of my role as a researcher and, after a particularly challenging interview with a young man who did not understand his a priori risk for HD, I wrote,

I was concerned at a number of points that he did not have some pretty basic information. Also concerned about finding a balance between doing an interview and providing some minimal information since I absolutely couldn't let him continue thinking things were worse than they were in terms of his own risk. I also felt sad at a number of points during the interview and had to actively resist feeling as if I was being put in a counselling role. This made me feel like I had to qualify everything but at the same time give him something. Then I also had to remind myself that just talking about some of this for the first time was a positive step and

27 As Dr. McKellin reminded me, I was a regular participant in meetings where the latest developments in scientific and clinical research were discussed; I worked with a group of experienced scientists and professionals, and I had access to an extensive collection of publications and other resources.
that I wasn’t there to do any more. I did so want for him to be able to feel better though... not through false hopes but through understanding.

It was in part the expectation that I was there as a resource person which seemed to compel additional members of the family to call me at the hotel where I was staying in order to request an appointment. The word had travelled that I was in town and the three interviews I had anticipated blossomed into seven. This family’s collective enthusiasm for the research was, fortunately, no less once I established the purpose of the interview and, upon my return some months later, I was warmed by their tendency to gather outside the small meeting room I used for the interviews, conversing with each other and occasionally — when an interview seemed to be going on for too long — dropping in to see if everything was going according to plan.

During the second round of interviews, conducted several months after clinical disclosure of test results, the issue of who I was to study participants became, paradoxically, more profound but harder to locate and trace in my growing trail of transcripts, fieldnotes and analytic memos. It surfaced as an issue when some study participants began to feel more at liberty to ask questions of me and anticipate that I would offer the same level of honesty in my responses as they had in theirs. It surfaced, in other words, as the expectation of reciprocity and resonated, within me at any rate, as a sign that our mutually developing discourse had, however partially and incompletely, tilted the asymmetry of power ever so slightly toward a more agreeable balance. Elsewhere, the issue of who I was surfaced more painfully. On two separate occasions, study participants prompted me to rethink my comfortable but minimalist approach to talking about my own life circumstances. In the absence of information to the contrary, each seemed to think that I was perhaps overly aware of the mortality of those at risk for HD but not sufficiently aware of my own mortality. As a woman who had recently lost a dear friend to HD said,

He did not live his life in vain and just wait for Huntington's to take him anymore than you're going to live Huntington's. Who knows, maybe on the way back [home] you'll just fall asleep on the bus and not wake up. It can happen to any of us Sue. (F, not at risk, family member, 55 years, married, 3 children)

This excerpt was taken from the closing portion of the first (pre-results) interview with a woman who had only a limited amount of time in which to talk. I had travelled a considerable
distance in order to meet with her and when I arrived in the small community where she lived, I
found that much of the town was on red–alert because of the brush fires which were burning out
of control in the nearby hills. She was organizing temporary shelter for those who had been
evacuated from other small communities and, somehow, managed to sandwich an interview into
the midst of this chaos. Like many other study participants, she went out of her way to
participate in the research. She knew that it mattered but she was at first uncertain about whether
or not she would be of any help to me.

Her expectations about what might or might not be relevant to the interview were, in this
sense, instructive of the process of negotiation which marked the beginning segments of many
interviews. The following excerpt from the opening segment of this interview (see next page)
begins with my reminder to the respondent that she may at some point wish to shut off the tape–
recorder (001–002). She infers that I am aware that she may wish to say something “off the
record” and this prompts her to ask about who will have access to the tapes (003–004). She is
trying to establish who her secondary audience is or may be but I reply with a somewhat vague
description of my role in the research (005–016); she affirms that she is listening (011) and
understands why it is important to talk to family members and friends as well as the PT
candidate (018), and; I check to make sure that she also knows about the questionnaire study
(016–020).28 I then offer an explicit reminder that she could also participate in this aspect of the
research (020–021) and she offers a tentative response (022–025) which suggests that she is not
yet certain about the relevance of what she has to offer. Thus, she begins her story with a
disclaimer (“I’m not really terribly well–versed with the disease”) and alludes to the fact that the
family she has known for much of her adult life told her everything she knows. I offer a neutral
affirmation that she should proceed (026) and, with only a slight hesitation, she initiates a
transition into talking about her own perspective (027–029). I offer another neutral affirmation
(030) which, in its questioning inflection, indicates my increased interest in what she is saying
and, in response, she takes up the invitation to proceed with her story (031–032), now somewhat
assured that we have arrived at a mutually relevant starting point. As the interview proceeds she

28 The questionnaires I refer to within this segment were for the CGAT–funded study mentioned above.
Transcript 1: Negotiating Relevance at the Beginning of an Interview

001 I If you'd prefer that uhm the tape recorder wasn't running, just say and we'll shut [it off].

003 R [Yup]/ like these tapes what are they/ are they used for other students or?

005 I No, this is research that I'm doing uhm in part for my thesis/ my doctoral thesis at UBC and I'm involved in this project which is looking at people who are going through predictive testing and also their family members and close friends because a lot of the research to date has really focused mostly on the person going through testing

011 R Uh huh

012 I and for good reason [you know]

013 R [Oh yeah] yup, yeah

014 I to understand what people go through/ but everyone realizes that's only part of the picture and so we're doing uhm this study which involves mostly me going around and doing interviews/ there's also a questionnaire

018 R uh huh

019 I component to it which Alice might have mentioned to you

020 R [Yes, yes she did.]

020 I and uh so I've got questionnaires as well if/ if you feel that want to do those as well that would be great.

022 R If it helps you Sue (. that's fine. I mean I don't know what I can tell you uhm (. in particular. I mean I don't/ I'm not really terribly well versed with the disease other than what the family have told me.

026 I Mm hmm.

027 R Uhm I think in that particular family there's a lot of other uhm (. circumstances uh/ I don't know what you call it, it's a very strange family.

030 I Yeah?

031 R I've known them for a long time and since dad has passed away (. things (. haven't been easy?

033 I Yeah.

034 R (...) Uhh (h) Everyone seems to be uhm (. /Well mom is a difficult person

036 I mm hmm
a very difficult person and if there isn't a problem she'll create a
problem uh/ She really likes the attention uhmm so I think that's—
where a lot of the stress comes in like there's no real like/ I, I
shouldn't say this maybe but I sometimes feel that (..) the kids don't
have a family?

Yeah.

Because uh/ and (..) families are always pulled together usually by
the surviving parent

Right.

and it doesn't matter if the kids are ninety or they're nine. The
family unit has still got to be there. And uh this ganging up on one,
there always has to be a rift in that family, Sue.

Yeah?

And then they gang up, there's 3 children and they'll gang up on
this one and then that's fine that's eventually sorted out and then/
They can't they can't be a foursome, a family of 4 with the
grandchildren. There has to be/ let's create a problem. And uhm I
finally/ Well last spring after Ben passed away it was "Are you
coming this weekend?", run to the lawyer, go to the accountant, do
this for me, do that for me, come and baby sit my dogs while I go on
holidays, shoot your cat and it just was [this]

[This] was Alice's mother?

Yes. Yeah and/ until it got to a point where I just said enough I
can't do anymore. I was having a few stomach problems myself

Ohh

and I was supposed to rest every afternoon and yet I was
also supposed to take the ferry and you know the usual thing
and it was just too difficult. And uhm/ You know you don't mind
really extending yourself to a point and I think there's a bit of a
drinking problem that the Mom has, whether the kids
recognize it or not/ alcohol(.)ism is a factor in her family

Yeah.

and it is hereditary. Uhh/ So we sort of go on with it and I just
phoned back and I said no if there's going to be any
stabilizing for these kids I guess I'm going to have to
provide it because Ben was my friend and back in the days
when we grew up and we were starting to raise families it
wasn't popular to have a female to be a friend/ friends with a male
that wasn't your husband.

Right.

so of course you had to be friends with [the mom]
I [with the whole family]
yeah, and uh I think that's where our friendship/ I just learned to
tolerate and I'm/ I'm/ I do quite well. And I do pat myself on the
back with tolerating (.) and I have a hard time saying no. So that's
how I know the kids so well, I know Alice well but not as well as I
did the older two because I used to be their baby-sitter. [And]

[Right.] You were living nearby [at the time?]

Yeah both dads] worked together. My husband and Ben were
working together and so we spent many hours but Ben/
that's where the friendship started and it well/ wasn't really until
after uh (. ) Ben passed away here last spring, did the kids sort of
realize that hey the friendship was always between Auntie
Kate as they call me and Dad it wasn't so much mom. And uhm,
but of course for Ben's sake I rushed in and tried to pick up what
pieces I could and it just became that well I was kind of the major
caregiver and with no thought for my own life or that of my
children, it was her. Not the thought of her children, the fact that
she was grieving...
starts to describe how difficult the Mom is (034–039) and then hesitates briefly (“I shouldn’t say this maybe but I sometimes feel...”) before continuing to discuss the problematic family dynamics (040–053) which ultimately lead to her intervention as a caregiver and stabilizing influence for the family. She offers an itemized list of errands she ran for the Mom (054–057) in the period immediately after her friend (the Mom’s husband) passed away, mentions the strain on her own health and well-being (059–064) and, then discloses that the Mom’s husband was a very dear friend to her and that it was not considered proper at the time for a woman to be friends with a married man unless she was also friends with his wife (072–080). I offer an affirmative “right” (076) and she begins to situate her friendship with the Mom’s husband and her long-term involvement with the family within her own life circumstances (081–083). I prompt for more information about where she was living at the time (084) and she responds with additional details about herself and her relationship to the family (085–095).

I present this particular excerpt because it illustrates fairly succinctly the jointly constructed nature of the interview as a form of developing discourse or communicative exchange. Although the respondent’s assumptions about the nature of the interview remain implicit, the tasks of establishing relevance and locating an appropriate starting point signal that important negotiations are occurring. In allowing the respondent to establish the sequence, timing and structure of her account, I signalled that her knowledge — a knowledge derived from the lifeworld rather than the world of medicine — was what I was most interested in hearing. This excerpt is not unusual: the negotiation (and re-negotiation) of relevance and perspective, turn-taking and topic introduction are features of all interviews, whether or not they are explicitly acknowledged during the process of analysis. This is not, however, to suggest that I or for that matter, other respondents, were always able to initiate such a seamless entry into the everyday lived experiences of the lifeworld. Short, closed-answer responses to my questions and prompts are scattered through-out the pages of more than a few transcripts and, although I clearly had a lot to learn about technique (i.e. not asking closed-answer questions, developing a neutral set of responses et cetera), I am hesitant to attach too much significance to any such “errors” and/or their prescribed technical solutions (e.g., rules for reducing the “biasing effects of inadequate interviewer performance”) (Mishler, 1991:52). The varied structure and format of
the interviews could, perhaps, be just as easily explained by the range of life circumstances and
narrative or conversational styles of study participants themselves — a real talker versus a
“holder-inner” (as one respondent described himself) — but this approach too must be faulted
for neglecting the dynamics of the interview as a developing discourse. The communicative
exchange of an interview demands the sustained efforts of interviewer and respondent as they
engage in the mutual process of locating shared meaning: the outcome of this process cannot be
reduced to the status of an individual attribute any more than it can be corrected through
techniques designed to eliminate interviewer bias.

Bearing in mind this preface, let me now say that, in most cases, I found it easier to elicit
from women the sort of extended narrative account that is exemplified in Transcript 1. This
may, in part, have been a product of my own expectations — a sort of self-fulfilling prophesy
— or it may have reflected some combination of gendered patterns of communication and my
level of comfort and ease in talking with women as opposed to men. When interviewing men, I
often noticed that I had to attend much more closely to the sequence of topics and the framing of
questions if I was to be successful in eliciting more than a few consecutive phrases in response. I
felt that I was doing more of the work, and in several instances, that I was not necessarily doing
it very well. On the other hand, the free-flowing narrative accounts which spilled out of some
(male and female) study participants consumed pages and pages of paper and, as I began to look
for a method of analysis which would allow me to manage this highly “attractive nuisance”
(Miles, 1979), I gained a new appreciation for the economy of some respondent’s words. There
was, within the white space of what sometimes felt like an interminable silence, a sparse but
intensely focused and chronologically ordered response. This leads me to a related issue: that is,
the ontological status of the interview as a communicative event.

**Telling and Listening**

How did participants experience the interview situation and what implications does this
have for the type of analysis pursued here? On one level, the interviews were formal pre-
arranged occasions for study participants to talk *about* patterns of communicative interaction.
On another level, however, the interviews *were* instances of communicative interaction. This
research engages with both levels of analysis and in some instances, it also must engage a third level wherein participants initiated a metaconversation about the way in which they (or we) were communicating about communication within the interview.

In some ways, this issue parallels the issue of who I was to my study participants. If I was an atypical friend of the family — and there are limits in the degree to which I am comfortable in arguing that this was the case — then the interviews were in some ways similar to naturally occurring conversations. If, however, I was first and foremost an interested but friendly outsider with a legitimate research interest in some fairly private matters, then the interviews cannot be taken as representative instances of naturally occurring conversation.

Alternatively, the better question might be, were study participants offering, in their narrative accounts and self-disclosures, a form of history (a stratagem to avoid being known) or honesty (an invitation to share their experiences)? Pearce (1973:415-16) notes that direct observation of disclosures which occur in the contrived setting of a laboratory are “perhaps best understood as the sums of confessions (to the extent that subjects acceded to the demand characteristics of the experiment), revealing behaviour, history, and — perhaps — honesty.” In agreement with Pearce, I adopt a both/and approach. I reserve the right to be both skeptical and a tad idealistic. The skeptic reminds me that power is never erased by pretending that it does not exist while the idealist knows that there are times and places and people that allow us to experience such safety that “we believe vital values will be gained if we are known in our authentic being, or lost if we are not” (Jourard, cited in Pearce & Sharp, 1973).

Knowing is, in this sense, connected to desire. It is not simply a rational/analytic process; it incorporates emotion both as an affective stance toward what is known and a condition through which knowing is accomplished. Knowing is, as Game and Metcalfe (1996:171) argue,

...in life. Thus it is an experience that is thoroughly embodied and affective. We know the world, or specific phenomena, through our affective, emotional, sensual responses: ‘this is how I feel in the face of...’

Sociologists have often shied away from rather than been informed and enriched by their affective responses to knowing the social world. Letting go of the desire to control and instead welcoming the unpredictable it is, however, possible to gain a sense of how knowledge feels
When it is in process (Game & Metcalfe, 1996).

Listening, like speaking, is an embodied way of knowing but few books on methods entertain the idea that researchers need to let go of the desire to control what is heard and gain a sense of listening in process. Perhaps poets and those who engage in narrative analysis are more attuned to this, more willing to listen to the feelings in words and less concerned with extracting meaning from the embodied and situated utterances of a speaker. In “Words on Words”, poet and playwright Vaclav Havel (1990:6) describes the chameleon-like qualities of words.

No word — at least not in the rather metaphorical sense I am employing the word ‘word’ here — comprises only the meaning assigned to it by an etymological dictionary. The meaning of every word also reflects the person who utters it, the situation in which it is uttered, and the reasons for its utterance. The selfsame word can, at one moment, radiate great hopes, at another, it can emit lethal rays. The selfsame word can be true at one moment and false the next, at one moment illuminating, at another, deceptive.... On one occasion it can open up glorious horizons, on another, it can lay down barren tracks.

And, in Holocaust Testimonies, Lawrence Langer (1991) builds a painfully convincing argument about the need to listen carefully to the “unstable resonance of particular words.” In particular, he writes with a passionate dispassion about the inability of interviewers who recorded the oral testimonies of Holocaust survivors to hear what was being said. These interviewers, and some of the historians who have written about the testimonies, could not hear the confusion, doubt and moral uncertainty of a consciousness violated by the remembered realities of disaster; could not resist the need to hear, in the testimonies, a triumph of human will, a narrative of survival and liberation. Our collective historical need for some form of reconciliation with a “milder reality” is, as Langer admits, often overwhelming. Nonetheless, Langer also confronts us with our insistence that there is always some meaning in suffering. As such, he forces us to consider the difference between what is said and what is heard.

If narrator and listener are both active participants in the creation and articulation of plot, how is good listening accomplished? The progression from learning to think about stories to learning to think with stories (Cruikshank, cited in Frank, 1995:23) is less straightforward than it might initially appear and yet we need to think with stories in order to really hear them. As Frank (1995:158) proposes,
Thinking with stories means joining with them; allowing one's own thoughts to adopt the story's immanent logic of causality, its temporality, and its narrative tensions...The goal is empathy, not as internalizing the feelings of the other, but as what Hapern calls 'resonance' with the other. The other's self-story does not become my own, but I develop sufficient resonance with that story so that I can feel its nuances, and anticipate changes in plot.

Learning to listen has been one of the most rewarding aspects of this research. Thankfully, it is not a technique to be mastered (Corradi Fiumara, 1990). It is something which unfolds through time. I was, at first, inordinately aware of what I thought the interview should be. I tried to ask good questions, give non-judgmental responses and prompt for missing pieces of information but things happened fast; I was barely able to keep up with what the story was about never mind think through it. As I began to relax and trust that I knew how to respond, I developed the ability to attend more closely to what was being said. I did, however, listen for the plot that I was most interested in and to this end, I stuck mental post-it notes on who said what to whom, when and where. I knew what the story was about and I empathized with the teller, but as a listener I had not yet heard the story through the story.

I do not know when this changed but one of the pre-results interviews I did early on in the research was pivotal in presaging the sort of awareness of story that I am talking about here. I was talking with a man whose wife was having predictive testing. He had grown up on the Prairies during the Depression and, despite my efforts to steer him onto the issue of his wife's imminent test results, his story kept gravitating back toward his observations and feelings about driving to the Prairies each summer. He worked as a "fruit peddler" and journeying back there (within his narrative) evoked a sense of nostalgia for the past — a tangible longing for simplicity, honesty and community.

See I've lived a good life even though it was always tough and that but it was okay. Whatever you had you appreciated.

...See at one time a friend was a friend, you could discuss things with him...Nowadays you're just about at the point where if you see a guy having tough luck, you're happy about it...Instead of saying "Well, the poor guy, he's got a wife and a family and that." (M, not at risk, family member, 62 years, married, 4 children)

His mother, "a little bit of a woman" had a stroke and died in her 40's. His younger sister, "raised five kids on nothing." It was "livin' too good" that lead to his plugged arteries and
eventual double bypass and it was the affluence and indifference of the "modern" world that troubled him most.

...I have a problem with people that just throw up their hands and scream cause some little thing is not right...Like I always said if we had a depression and everything went for/ went to hell, I could handle it because/ I could live. I wouldn't maybe live here, you know. I may not be watching television, but I could handle it. Somehow we would survive.

...You see, our neighbours they'd have a beautiful television and drive in a big car but they couldn't cook you up a meal. It's damn hard to make soup out of a television.

I laughed at his parody of the futility of keeping up with the "Joneses" and he became very serious.

I don't know if you understand what I mean...I mean I don't care. You have to be a survivor...if we did have a real boomer then people would wake up cause we've got things too/ really pretty easy. I'm just old enough to remember the 30's in Northern Saskatchewan.

It was nearly supper time, the family was visiting and the grandchildren were getting restless. The interview was almost over but he wanted to know that I understood that what he had been talking about was the story. He made one final pitch.

See I haven't sat down and [said] well, "these are the bad things and these are the good things." It's just everything is/ It just has to be.

If it is, it is and if it isn't, it isn't.

Thinking about his story and the relative absence of direct references to HD and/or predictive testing, I thought that I had not learned all that much about how he really felt but thinking through his story, I understood that the things he valued most — hard work, honesty, commitment and community — were enduring aspects of who he was and how he engaged in survival. His narrative was about the virtues of accepting what cannot be changed — Huntington Disease included — and finding, through such acceptance, a way to live and die honorably with "whatever happens."

Reading and Writing the Story

This dissertation is a story about stories. The task I have given myself is to re–tell the stories that I have been told, not precisely as they were told but rather as they now collectively elucidate significant aspects of communication about genetic information in families at risk for
Huntington Disease. The dissertation as story does, therefore, have its own plot and narrative style. In addition, the roles of speaker and audience have shifted. I am aware of how tenuous it feels to talk about someone and wonder if they are listening. Like Josselson (1996:69–70), I want to understand my own discomfort with this situation, "the dread, guilt, and shame" that seem to go with writing about others. She writes,

The dread is easiest to trace. There is always the dread that I will have harmed someone, that I will be confronted with, "How could you say that about me?"

The guilt is more complicated. My guilt, I think, comes from my knowing that I have taken myself out of relationship with my participants (with whom during the interview, I was in intimate relationship) to be in relationship with my readers. I have, in a sense, been talking about them behind their backs and doing so publicly. Where in the interview I had been responsive to them, now I am using their lives in the service of something else, for my own purposes, to show something to others.

Arguably, writing is the hardest part of analytic work. This is because written narrative about oral testimony inflicts difficult choices; it is the litmus test of understanding; it frustrates and forces a "concession to what words cannot do" (Langer, 1991:105); it demands the imposition of a perceived sequence of events whether or not this precise sequence existed during the period that is being preserved in language.

It is a universal feature of narrative that it has a beginning, middle and end. Nonetheless, what is beginning, middle, and end? And, moreover, what kind chronology, trajectory or teleology informs such decisions? Thus far, I have portrayed predictive testing as a process which involves, among other things, a chronological sequence of events. In chronological time one event necessarily precedes the next and, although the interval of time which intervenes between events may vary widely, there is a before, during and after. As Ricoeur (1981:278) notes, this "episodic dimension" of narrative leads the listener (or reader) to ask "and so? and then? what happened next? what was the outcome?"

The research which informs this dissertation was conducted prospectively. The clinical disclosure of predictive test results was the fulcrum around which interviews were scheduled and the storyline was anticipated to unfold. As such, it is important to maintain the sense of contingency which permeated the pre-results interviews in particular. Chronological time is, in this sense, an important organizational feature of the dissertation. It permits the story to unfold
within and through a sequence of narrated events. This is not, however, the only structure of time that will concern us here.

The art of narrating as well listening to a story requires that we are able to construct a meaningful whole out of scattered events. This "configurational operation" or "grasping together" is a reflective act. To tell and follow a story is, as Ricoeur (1980:178–9) suggests, "already to reflect upon events in order to encompass them in successive wholes." Sequence (the episodic) and pattern (the configuration) are, then, always in tension with one another but pattern can never completely overcome sequence without suppressing the narrative structure.

In a word, the correlation between thought and plot supersedes the "then" and "then" of mere succession. But it would be a complete mistake to consider thought as a–chronological.

Plot consists of a patterned configuration of events that imply an ending. Once that ending is known — as in popular narratives about founding events or particular acts of heroism — the re–telling of a story introduces an alternative to the chronological, episodic representation of time. Time no longer moves forward from past into future; it is inverted and we read the ending in the beginning and the beginning in the end. As such, plot roots human action within memory as well as chronological time. The verbal structure of autobiographical memory is, as Rubin (1996:2) asserts, "the structure of the genre of narrative that it is." Whether or not autobiographical memories are actually stored as narratives, they are usually told in a narrative form to oneself or others and thus the structure of discourse shapes the structure of recall which in turn shapes the structure of subsequent recall.

Drawing upon Heidegger's notion of repetition, Ricoeur (1980:180) explains that the role of memory in plot implies "more than a mere reversal of the basic orientation of care toward the future; it means the retrieval of our most basic potentialities inherited from our past in the form of personal fate and collective destiny." The past ("what we have already been") provides resources which allow us to anticipate "what we may have to be." For Heidegger, the future is, however, closed in the sense that being–toward–death is the untransferable individual precondition of life. Rethinking this proposition from the standpoint of narrativity, Ricouer (1980:188) objects. Narrativity opens up our mediation on time to include another horizon than that of death. It focuses our attention on "the problem of communication not just between living
beings but between contemporaries, predecessors and successors.” After all, he proposes, “is not narrative time a time that continues beyond the death of each of its protagonists? Is it not part of the plot to include the death of each hero in a story that surpasses every individual fate?”

It is perhaps Ricoeur’s interest in the role of narrativity in history that leads him to recognize the limitations of viewing narrative in an individualistic way. Narrative time precedes and exceeds the lifespan of any single person and, as such, it entails a sense of collective rather than merely individual fate. This collective orientation is helpful when considering the narratives of persons at risk for, or affected by, HD. Indeed, the very unsettled nature of where to begin the story was one of the most striking features of many participants’ accounts. As the next chapter demonstrates, family stretched back in time but it was only through the lens of the present and more specifically, the confirmation of a family history of HD, that participants could connect the individual fate of certain predecessors with their own fate.

Further, many of the storied accounts which inform this research are deeply reflective of the lived experience of temporality. For those who know that they have inherited the genetic mutation associated with HD or for those who have only recently experienced onset or diagnosis of HD, there is pronounced emphasis on living in the moment or living each day to the fullest. The tragedy of this is, however, that the future is writ large upon those who are in an advanced stage of the disease. At the Retreat for persons with HD, this mirroring of the past and future offered an infinite regress wherein those in more advanced stages of the disease saw their earlier selves mirrored in others who were more “able” and those in earlier stages saw their future selves mirrored in those who were in more advanced stages of the disease. The Retreat was, then, an opportunity for those with HD to see forward and back in a kind of kaleidoscope of lived and imagined exigencies. This kaleidoscope of past, present and future was not unique to the Retreat but that was where I became most acutely aware of it.

These various dimensions of narrative time impose certain tensions in the analysis and presentation of study participants’ accounts. Some of these tensions are described below within the context of considering the process of preparing the five narrative accounts which are included in their entirety in Appendix VIII while others have an overarching significance for the analysis as a whole.
Process of Analysis

The process of analyzing the interview data derived from the pre-results interviews started before transcription. In effect, this meant that I listened over and over to the tapes, making notes about significant events and jotting down questions that I wanted to ask at the next interview. There were, as it turned out, always aspects of each interview that puzzled me. A partial case-by-case analysis was, therefore, integrated into the preparation that I did for the second round of interviews.

With the exception of the long-term follow-up interviews, I did not do the transcription myself. The BCMSF and Hampton Fund grants mentioned above provided funds to hire a transcriptionist. Tapes were duplicated (as a precaution against loss) and I reviewed randomly selected transcripts against the tapes, checking for completeness and words or phrases which were inaudible to, or misunderstood by the transcriptionist. A list of the conventions followed in transcription is included in Appendix VII.

Documents were then formatted for entry in NUD.IST (Non-Numeric Unstructured Data Indexing, Searching and Theorizing), a software program which permits theory-building as well as descriptive-interpretive functions (Richards & Richards, 1994). Selected for its sophisticated search and retrieval capacities as well as its ability to accommodate diverse approaches to data analysis (Weitzman & Miles, 1995), NUD.IST permits the researcher to employ both inductive and deductive approaches. Through features such as on-screen coding and memoing, NUD.IST assists in working closely with the data to identify important concepts and ideas. These can be used to build or modify an index system. With a well-developed index system and coded online documents it is possible to construct and test hypotheses, generate reports on the findings, and locate evidence which contradicts as well as confirms emergent understandings. Further, self-reflexivity is enhanced through the "system closure" capabilities of NUD.IST. As such, it is possible treat the development of one's own thinking (as it is reflected in an audit trail of memos, reports etc.) as data which is available for further analysis.

29 One of the more memorable instances of a misunderstood phrase derived from an interviewee who described himself as coming from a family of "long livers". His intended meaning was that many members of his family lived well into old age. The transcriptionist, however, was concerned that she could not quite ascertain the name of what must be an unusual liver condition.
NUD.IST was invaluable in organizing, managing and searching the data. The search functions, in particular, allowed me to rapidly locate various segments of text and unearth some rather intriguing patterns in how certain words were used. This led to some wonderful “moments of eureka” in which I was startled to discover a relationship between previously unconnected ideas (Bassett, Cox, & Rauch, 1995). I did not, however, utilize NUD.IST in the way that I had originally intended. Although I developed an extensive index system, I did not engage in detailed coding of small segments of the text according to theme or category. The key problem with coding and categorizing is that it breaks apart the data and disembodies it from the person who produced it. Narrative analysis, on the other hand, stresses the importance of context and emphasizes the embeddedness of meaning in the language, structure and content of the individual’s story. As Conrad (1990:1258) asks, however, “does narrative analysis show greater respect for the integrity of the data, while reducing the possibility of analytic generalizability?”

Coding and categorizing lay the framework for a mode of analysis which, in the tradition of grounded theory, often results in the development of generalizable concepts and theories. Generalizability is, within qualitative research, measured through the applicability of such concepts to other settings, situations and/or research populations (Guba & Lincoln, 1994). Indeed, the theory of awareness contexts (Glaser & Strauss, 1964; 1965) which is so central to this dissertation was a ground-breaking example of the significant benefits which could be derived from adopting a grounded theory approach.

What to do? How to weigh the integrity of the narrative against the potential for enhanced analytic generalizability? The dichotomy is more dichotomous in its abstraction than it was in reality. In reality, confusion often reigned. I tried to work away at coding but I was constantly drawn into the story, I had to follow its twists and turns in order to know what the category I was so liberally applying actually meant. And, I became frustrated with spending so many hours in front of the computer. I printed out pages and pages of reports in NUD.IST (with line numbering down the side of each page), bought a rainbow of colored pens and sat at the kitchen table writing notes in the margin. I was happier but the text seemed flat on the page. I missed hearing the cadence and rhythm of the words, as they were spoken. I regressed even further and began listening to the tapes again. I was, for a time, utterly hopeless at what Miles
and Huberman (1994) refer to as "data reduction."

Shifting my attention to the task of locating the chronological sequence of events inherent to each study participant's story, I proceeded with the analysis of patterns of communication about genetic information by describing, for each study participant, a series of awareness contexts (Glaser & Strauss, 1965) — that is, the shifting configuration of persons who are aware or unaware of information about Huntington Disease, its particular history within the family, the implications of this history for the risk status of various family members, the decision to request predictive testing, the results of the predictive test and their implications for various members of the family. Attention to the communicative parameters of each awareness context generated a basic framework for the analysis — the who tells what to whom, when, and where part of the story — but the how and why elements were trickier; they were implicit and often ambiguous. I had to think with the story and extrapolate from its telling within the interview in order to arrive at a sense of how and why certain disclosures and/or concealments were achieved and what they meant within the larger context of each person's story. Without this understanding of what communication meant to communicants themselves, I felt that my treatment of the subject would be, at best, superficial.

The process I followed in analyzing the study participant's stories ultimately involved all of the following (not necessarily chronological or discrete) aspects.

1) Preparation of study participant summaries: Drawing upon demographic questionnaires, family trees, fieldnotes and transcripts I developed a summary of information about each study participant and a rough timeline depicting significant life events.

2) Mapping of awareness contexts: I read and reread the transcripts, noting the who, what, where, and when of significant instances of communication about the family history of HD, the decision to have predictive testing and share the results with selected others.

3) Aggregation of individual summaries and awareness contexts: I compared the timelines and awareness contexts of various family members noting similarities and differences. I also looked for exemplary instances of conversations or events which included two or more of my study participants so that I could read and reread what each had to say about the conversation or event. This was not an attempt to determine the truth or veracity of an account; it was a
search to understand from different perspectives the meaning and significance of an event or the impact of a particular disclosure.

4) Description of strategies for managing genetic information: Looking across the range of individuals and families, I noted similarities and differences in strategies of disclosure and/or concealment and their practical consequences for PT candidates.

5) Interpretation and reflection: I reconsidered the form and content of the stories that study participants told about their acts of communication, asking myself what various acts of communication meant to the interactants and, moreover, how the meaning of talking about HD and hereditary risk shaped patterns of familial communication.

This was a highly iterative process which was, in reality, less ordered and systematic than its representation now seems. I had to be grounded in the stories as they were told yet I also wanted to reflect on my own role in co-constructing each. Moreover, I often found that each reading of the transcripts turned up new questions and insights and that it was, in consequence, very difficult to decide when I understood enough about study participants’ stories to begin writing through the stories.30

Preparation of Narrative Accounts

The interview data gathered for this research offers a wealth of insight into familial constructions of hereditary risk, patterns of communication and information management, experiences of predictive testing and interpretations of clinical information amongst PT candidates and their families. This dissertation focuses on familial communication and the stories that PT candidates and their family members tell about their acts of communication.

Funding from the Huntington Society of Canada also supported the preparation of five narrative accounts. These accounts were jointly produced with each of the selected PT candidates with the intention that they would be made available for publication by the Huntington Society. The five accounts are included here in their entirety (see Appendix VIII) because they were an integral part of the process of analysis and because they provide the reader

30 The process put me in mind of something that a young man with HD said while doing art therapy at the HD Retreat. He was painting an abstract picture of a breeze in green, blue and purple, and when I asked him how he knew when to stop he said, “you have to let it speak to you. Go away for awhile and then come back and just be with it. Only you know when it is finished.”
with the opportunity to read a cross section of complete ‘uninterrupted’ stories.

The process of transforming oral accounts into a coherent written narrative presents innumerable decisions, many of which I have alluded to above. For instance, the imposition of chronological time provides a coherent structure but tends to orient the narrative around the linear development of plot. Temporal order may therefore provide a unified context but it does so at the cost of obscuring another important aspect of illness narratives in particular: that is, the way in which they are struggles to locate and define a framework within which it is possible to discuss the illness experience (Hyden, 1997). Further, illness narratives are often concerned with change and the accompanying reconstruction of identity and personal life in connection with loss (Charmaz, 1983). The sense of illness experience as epiphany is, however, not generalizable to the experience of predictive testing however much existing research may incline us toward thinking that it is. Many PT candidates actively resist this view (see Regina’s story) in seeking some kind of practical relationship to HD; to insist on framing predictive testing in this way is therefore to think against rather than with such stories.

The five narrative accounts included in Appendix VIII were derived from verbatim transcripts of the interviews and thus each presents a true story about the experience of predictive testing. These accounts were selected in order to represent an array of experiences with predictive testing and thus each tends to highlight a different set of circumstances and/or outcomes. All were read and validated by the interviewee and, where necessary factual or other corrections were made. Pseudonyms are used in all of the accounts (and throughout the entire dissertation) and in some cases identifying information has been removed (e.g., name of town where person lives).

Each account is an accurate representation of the experience as it was described to me but none are complete. This means several things: first, substantial editing and reorganization was required in order to turn a verbatim oral account into a smooth written account. “Uhms” and “aahhs” as well as repeated phrases and false starts were removed; interesting aspects of local pronunciation were lost and, in general, the feeling of orality disappeared. Furthermore, given that most interviews looped forward and back in reference to events that had occurred in chronological time, it was also necessary to rearrange the sequence of whole passages of text in
order to create and sustain the desired episodic flow.

Second, space limitations dictated that the finished account be of a publishable length. In many cases, 150 or more pages of transcript had to be whittled down to 20 pages. I had to focus on what would be most salient to the HD community and, in particular, potential PT candidates and their families. In many cases, there was no conflict between this and my own purposes in this dissertation. Issues of familial communication figured prominently in all of the interviews and the narrative accounts reflect this. In other cases, however, there were aspects of familial communication which were too sensitive to include in a publishable account. For instance, one participant commented extensively on a case of suspected non-paternity which threw into doubt the basis of the family’s risk for HD. Another said things about a family member which could, if left untempered, cause irreparable harm to the relationship. Thus a third type of incompleteness also enters the accounts.

Fourth, the finished narrative accounts render invisible the local struggles for coherence which were an important part of how participants read and validated their own stories. As one might expect, participants’ self-schema and narrative interpretation of the process of predictive testing changed subtly or even markedly over time (before and after test results). The person they were at the time of reading was different than the person they were at the time(s) of narration. In one case, the existential experience of reading the finished account prompted the participant to acknowledge her own personal growth. She said that her story made her feel proud of herself. In other cases, however, it was difficult for participants to resist the compulsion to rewrite certain portions of their story, emphasizing some themes and de-emphasizing others in the effort to make their story more consistent with their altered sense of self. All participants acknowledged the importance of allowing the finished draft to reflect their experiences as they were lived but two women, in particular, found this especially difficult. One woman initially suggested a number of revisions that would have had the effect of dampening her sense of agency but later agreed with me that some of these revisions would interfere with the readers’ insight into what the experience of predictive testing has been like for her. The other stated that she felt “exposed” but that she would not make any revisions. There is a lot more that could be said about these negotiations but, for now, I merely point out that the process of preparing the
narrative accounts is itself indicative of the open-endedness which characterizes not only narratives about illness-related experiences but all narrative.

Finally, some additional comments on incompleteness are pertinent to the dissertation as a whole. I am in a small number of instances aware of the need to disguise the identity of the speaker from her/himself such that I do not believe that it is ethically defensible to put into words that which the speaker has not been willing to articulate and may not be prepared to hear. For instance, where a study participant who appears to be showing signs and symptoms of HD is not willing to verbally acknowledge this, I also refrain from acknowledging this in a way that could precipitate subsequent harm. There are, then, things which can only be discussed through reference to a more general level of observation. Further, in some instances, I refrain from connecting various pieces of an individual account. In no case, however, have I merged different accounts and presented them as if they were derived from one person. Nor do I present otherwise “fictitious” material.

Deciding on a Structure: Three Narrative Moments

In Chapters VI, VII and VIII, I present three ‘moments’ in the story of predictive testing. Chapter VI focuses on learning about Huntington Disease and the family history as well as understanding the implications of the family history for the risk status of various family members. Chapter VII focuses on deciding to have predictive testing, and Chapter VIII focuses on receiving and incorporating the results of predictive testing. The structure of these three chapters is chronological; they tell a story of transitions in familial awareness contexts, the social factors which both enable and constrain these transitions and, ultimately, the consequences of these transitions for families at risk for HD. As such, each chapter is concerned with the meaning and significance of communicative interaction in shaping and representing the experience of predictive testing.
CHAPTER VI
WHEN ‘IT’ STARTED

Narrative is language used to connect events in time. The connection, whether conceived as a closed pattern, beginning-middle-end, or an open one, past-present-future, whether seen as lineal or spiral or recursive, involves movement “through” time for which spatial metaphor is adequate. Narrative makes a journey. It goes from A to Z, from then to then-prime (Le Guin, 1989:38).

Meaning derives from the ability to formulate events into a story with a beginning, middle and end. Indeed, the sequential “arrangement of the incidents” in a directional, temporal order is, as Aristotle maintained, the basis of plot (Le Guin, 1989). In narrative time, beginnings, middles and ends are, nonetheless, relative. Beginnings mark a point of entry into the story but there is no imperative to begin at the beginning of a linear sequence of events. Though chronological time presents a structure which many storytellers adopt in relating a sequence of events, this not essential, even in history.

While history claims to represent reality in a way that literary texts do not, historians like novelists offer “explanation by emplotment” — that is, historical events make sense when they are transformed into a story (White, 1973).¹ History is therefore both scientific and artful: it is how we systematically recollect and document past events, illuminate their origins and explore their implications but it is also an aesthetic endeavour which implicates the attentive reader or listener in considering not just what is remembered but ways of remembering.

The act of recalling past events and reflecting upon the meanings these renderings of the past hold within the present is not an asocial process. The people and events we recall as well as the particular tone in which we recall them are shaped by the social context of remembering. The distinction between a memorable “history” and a forgettable “pre-history” is, therefore, neither logical nor natural; it is “an unmistakably social, normative convention” (Zerubavel, 1996:288). As the “very first thought community in which we learn to interpret our own

¹ This correlation between works of history and works of fiction does not imply that (written) history may be treated as a literary artefact: “history is both a literary artefact (and in this sense a fiction) and a representation of reality” (Ricoeur, 1981:290).
experience,” the family plays a particularly critical role in “mnemonic socialization” (Zerubavel, 1996:286). In this and other thought communities we learn “social rules of remembrance that tell us quite specifically what we should remember and what we can or must forget.” Moreover, some people (such as our parents or older siblings) have better access to parts of our past than we do and such “mnemonic others” may act as witnesses who prop up and affirm or contradict our own recollections.

**Purpose and Outline of Chapter**

This chapter is about beginnings and, more specifically, how study participants recalled and narrated their initial awareness of the family history of HD. It is the first of a sequence of three chapters which explore three ‘moments’ in the story of predictive testing. The next two chapters focus, respectively, on the decision to have predictive testing and the role that communication plays in shaping the meaning and significance of the test results for PT candidates and their families.

Many participants in this research expressed a sense of ambivalence about where and when their story ought to begin. The boundary between history and pre-history — that is, the beginning of the story — was therefore problematic. Almost all study participants who were themselves at risk for, or diagnosed with, HD had known about the family history of HD for more than five years and slightly more than half had known for at least fifteen years. Nonetheless, many commented on the fact that they were uncertain about exactly when such awareness had crystallized into knowing that they were at 50% or 25% risk for the disease. Moreover, many were also careful to point out that they could not always distinguish between what they could themselves remember and what they knew (or thought they knew) by virtue of stories they had heard from other significant members of the family.

In what follows, I begin by summarizing key aspects of the family history of HD for all study participants at risk for, or diagnosed with, HD. This summary represents what was known by study participants at the time of the first interview. I then explore how PT candidates recalled and narrated their initial awareness of the family history of HD.
**The Family History**

Existing literature stresses the clinical importance of ensuring that PT candidates are properly informed about the hereditary nature of HD, its symptoms, typical age of onset, progression, and diagnosis. In addition, all PT candidates are informed that although there is at present no effective prevention or cure for the disease, there are some interventions which are of assistance in controlling movements and depression. Without such knowledge, PT candidates cannot give meaningful informed consent to predictive testing. Further, it has always been considered important to ensure that PT candidates have a judicious amount of time in which to consider the meaning and significance of information about HD within the context of their own family history. Predictive testing is, for this reason, contra-indicated in situations where the PT candidate has only recently acquired the knowledge that they are at risk for HD (Benjamin, et al., 1994; Fox, et al., 1989; Quaid, 1992; Quaid & Wesson, 1995).

In reconstructing how the at risk individuals interviewed in this study first became aware of the family history of HD, it is important to recall that the first (pre-results) interview was conducted with PT candidates and each of their family members at approximately one to three weeks before the scheduled date for test results. All PT candidates (and family members who were support persons) had attended at least one counselling session and some had been to two or more sessions. By virtue of their decision to proceed with predictive testing, PT candidates had, therefore, been prompted to reflect on their personal history and awareness of HD in way that other family members might not have. Their recollections of the family history and the process through which they first became aware of HD must, for this reason, be situated within the context of discussions which had only recently occurred within a clinical setting. Moreover, I too had observed a number of the pre-test counselling sessions with study participants and was, in these cases, already somewhat familiar with the family history.

**Profile of Family History at Time of First Interview**

In this section, I provide a profile of some key aspects of the family history of HD for all individuals at risk for, or diagnosed with, HD at the time of the first interview. These key aspects are summarized in Table 7 (see next page). The right hand column of this table provides
TABLE 7
FAMILY HISTORY OF HUNTINGTON DISEASE
(AT TIME OF FIRST INTERVIEW)

<table>
<thead>
<tr>
<th>Selected Study Sample</th>
<th>49</th>
<th>PT Candidates Only</th>
<th>16</th>
</tr>
</thead>
<tbody>
<tr>
<td>Not at risk&lt;sup&gt;a&lt;/sup&gt;</td>
<td>20</td>
<td>Not at risk</td>
<td>n/a</td>
</tr>
<tr>
<td>At risk or diagnosed&lt;sup&gt;b&lt;/sup&gt;</td>
<td>29</td>
<td>At risk</td>
<td>16</td>
</tr>
</tbody>
</table>

<table>
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<tr>
<th>A Priori Risk for HD</th>
<th>7</th>
<th>2</th>
</tr>
</thead>
<tbody>
<tr>
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<td>7</td>
<td>2</td>
</tr>
<tr>
<td>50% risk</td>
<td>20</td>
<td>14</td>
</tr>
<tr>
<td>Diagnosed&lt;sup&gt;c&lt;/sup&gt;</td>
<td>2</td>
<td>n/a</td>
</tr>
<tr>
<td>Showing signs of HD&lt;sup&gt;d&lt;/sup&gt;</td>
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<table>
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<tr>
<th>HD Family History</th>
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</thead>
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<tr>
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<td>17</td>
<td>9</td>
</tr>
<tr>
<td>Maternal</td>
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<td>1</td>
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<th>5</th>
</tr>
</thead>
<tbody>
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<td>Living</td>
<td>6</td>
<td>5</td>
</tr>
<tr>
<td>Deceased</td>
<td>16</td>
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</table>

<table>
<thead>
<tr>
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<th>16</th>
</tr>
</thead>
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<td>29</td>
<td>16</td>
</tr>
<tr>
<td>No</td>
<td>0</td>
<td>0</td>
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<table>
<thead>
<tr>
<th>Has Sibling(s) Diagnosed</th>
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<th>7</th>
</tr>
</thead>
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<tr>
<td>Yes</td>
<td>11</td>
<td>7</td>
</tr>
<tr>
<td>No</td>
<td>18</td>
<td>9</td>
</tr>
</tbody>
</table>

<sup>a</sup> Persons not at risk (N=20) include spouse/partners (12), parents (2), other family (4) and friends (2).

<sup>b</sup> Persons at risk for or diagnosed with HD (N=29) include predictive test candidates (16), parents (1), siblings (7), children (3), other family (1) and woman who did not wish to have predictive testing (1).

<sup>c</sup> Persons diagnosed with HD (N=2) include 1 parent and 1 sibling.

<sup>d</sup> Based on personal observations at time of first interview. (Both at 50% risk.)

<sup>e</sup> No documented family history of HD in either parent (mother deceased and father living).

<sup>f</sup> Excludes persons at 25% apriori risk.
the same information for PT candidates only. Much of this information about the family history and is required in order to ascertain eligibility for predictive testing and as such, it is routinely gathered during an initial counselling session. Often the genetic counsellor also draws a family pedigree at this time.

As indicated in Table 7, more than half of the individuals interviewed for this study were themselves at risk for HD: twenty had a 50% a priori risk, seven had a 25% a priori risk and two had been diagnosed with HD. These individuals represent sixteen different families (thus some are PT candidates while others are parents, siblings or offspring). As indicated, fourteen PT candidates had a 50% a priori risk while two had a 25% a priori risk. Further, there were two PT candidates who may have been showing signs of HD at the time of the first interview.

For nine of the sixteen PT candidates, the family history of HD originated with the paternal side; five traced the family history of HD through the maternal side and, in one case, there was no documented family history in either parent. Five PT candidates had a living parent diagnosed with the disease and nine had a parent who had died from HD (or related complications). It is also of interest to note that none of the PT candidates interviewed for this research were the only biological offspring of their affected parent; all PT candidates (and indeed all at risk individuals) interviewed for this study had at least one sibling at risk for HD and/or diagnosed with HD.

Fourteen of the sixteen PT candidates interviewed in this study were, to the best of their knowledge, the first in their generation to request predictive testing. Of the two PT candidates who were not the first in their generation to request predictive testing, one had a brother who had participated in linkage testing and the other was one of two siblings who both proceeded with predictive testing during the course of this study. Four PT candidates had, prior to the development of a direct test, requested the linkage form of testing: three had received an informative result; two had a decreased risk and one had an increased risk. The fourth PT candidate had a brother who had participated in linkage testing and the other was one of two siblings who both proceeded with predictive testing during the course of this study. Four PT candidates had, prior to the development of a direct test, requested the linkage form of testing: three had received an informative result; two had a decreased risk and one had an increased risk. The fourth PT candidate had a brother who had participated in linkage testing and the other was one of two siblings who both proceeded with predictive testing during the course of this study.
candidate withdrew from the program after a sample mix-up made it impossible to proceed without gathering new DNA samples from a number of distant and somewhat reluctant relatives.

In summary then, the sixteen PT candidates interviewed for this study were strikingly similar in several important respects: almost all had observed firsthand the way in which HD affected a parent; all had siblings at risk for, or diagnosed with, HD and, with only two exceptions, each PT candidate was the first in their immediate family to request predictive testing for HD. While there were, in all cases, one or more family members who had in common the experience of living at risk for HD, there were no immediate family with whom PT candidates shared the experience of predictive testing. As these aspects of family history suggest, there was a strong “pioneering” (Kenen & Schmidt, 1978) spirit amongst PT candidates; each was embarking on a quest which no one else in the family had yet undertaken. As additional family members begin to request predictive testing, this will no doubt change: It is to be expected that new PT candidates will, in the future, be more likely to have a sibling, parent or other family member who has already been through predictive testing and who is, hopefully, also available as a source of information and support. Knowing the results of a sibling’s predictive testing may, however, have significant implications for at risk individuals’ intersubjective experience of risk (Cox & McKellin, in press) and this may, in turn, be a salient factor in whether or not such individuals accept or reject predictive testing as a viable option for themselves and/or other untested siblings.5

What it Means to “Know” About the Family History

While I learned many details about PT candidates’ family history through clinical observation of pre-test counselling sessions, I nonetheless always began each pre-results interview by asking the respondent to tell me about themselves. Most study participants began

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5 One example illustrates how what is perhaps the most common scenario—that is, “the gambler’s fallacy” (Tversky & Kahneman, 1982)—may come into play in such situations. In one family interviewed, the eldest of six siblings was the first to request predictive testing. The linkage form of testing revealed that she had a decreased risk of having inherited the genetic mutation associated with HD. Since then, all but one of her siblings has had predictive testing. Two were found to have an increased risk and two were found to have a decreased risk. At the time that I interviewed the eldest sibling, the only untested sibling was considering having the direct test. The eldest sibling knew and understood that her younger sibling had a 50:50 chance of having inherited the genetic mutation associated with HD but, nonetheless, could not escape the feeling that this 50:50 chance would also be mirrored in the final ratio of siblings with the gene to siblings without the gene. Given that three siblings had not inherited the gene and two had, she felt that it was somehow inevitable that the final sibling would even up the score (i.e., have the gene for HD).
in the past and worked forward in time, sketching in relevant biographical details about childhood and adolescence, education, employment and current family life. Most also introduced, or alluded to, the presence of HD in their lives without any direct prompting from me but only a handful independently mentioned the point at which they first became aware of the family history of the disease. As such, the one question which appears, without exception, in all transcripts from pre–results interviews is, "So when you think back, where in your history and growing up do you remember first learning about Huntington’s being in the family?"

Not surprisingly, answers to this question were seldom crisp and precise. Many study participants struggled to locate and define their initial awareness of HD by counting forward or back from other more resilient memories of related life circumstances and/or significant political or other world events. This is consistent with research which shows that the dating of events within autobiographical memory is often achieved through reference to related events rather than a chronological time line (Brown, Shevell, & Rips, 1986). Explicit recall of absolute dates is relatively unusual in long–term memory and thus people arrive at a plausible date through a series of temporal inferences which relate the events in question to each other or to a smaller number of somewhat privileged events that are linked to a specific temporal locus. These “marker events” (Linton, 1986) may include the dates of extraordinary occurrences such as the assassination of John F. Kennedy (as in one participant’s account cited below) or they may be drawn from the autobiographical domain in so far as major periods in a personal chronology are often bounded by events that significantly alter the social, educational, occupational or geographic situation of one’s life.

The ability to recall specific event memories is closely tied to the perceived salience of the events. Certain memories “survive, thrive and populate the domain” (Linton, 1986:51) of autobiographical memory because they are perceived as important, experienced with a high degree of emotionality and/or are recalled and rehearsed on a regular basis. The perceived salience of events is, however, always contingent and in relation to other events which may, with the passage of time, assume a similar or greater prominence. Some events may, therefore, lose their distinctive character, become blended together with other events or become altogether lost as identifiable memories. This inevitable process of forgetting some things while recalling
others takes several logical forms; some things one knows must have happened even though there is no memory of their happening; other things are logically implied by virtue of things that are available to recall; and still other things are both plausible and compelling but cannot be recalled or deduced from other memorable events (Linton, 1986). These multiple aspects of autobiographical memory are nicely illustrated by one young man, Jason, who attempted to pin down his first awareness of the family history of HD in the following way.

Somewhere about grade 7 or grade 8 we used to have [Huntington Society] meetings at our house every now and again so it was just obvious to me that it was just obvious exposure. There were a few people with Huntington’s there and you know, I was around/ I didn’t shy away from it but I wasn’t really a participant. So it’s really hard to say when I knew about it. I think I knew about it quite young. I don’t think we ever avoided the issue. I don’t remember a day in my life when they sat me down and said, “look son, there’s this thing in our family” so I don’t know when it started. (M, 25% risk, family member, 29 years, single, 0 children)

Jason was unsure about exactly “when it started.” He began by defining the time period in question through reference to his educational situation (grade 7 or 8) and through reference to events that he was able to recall (meetings of the Huntington Society). He inferred from his memories of the “obvious exposure” to people with HD that he must have known about HD even though he was not “really a participant” in the meetings which the Huntington Society held at his home. He could not recall his parents ever sitting him down and telling him that “there’s this thing in our family”. As such, there was no definitive event to anchor his awareness in chronological time.

Jason rationalized this gap in his childhood memories by stressing that, as a child, he had a “lack of strong feelings” about HD. He had never known his maternal grandmother (who died from HD) but he had known “for years” that he took his first plane ride when he was twelve days old — “I had to go to my grandmother’s funeral.” Jason therefore grew up knowing something about the family history of HD but, as he repeatedly suggested, it was the stories told to him by his mother and older sister which actually allowed him to “remember” the events that he could not specifically recall.

For other at risk individuals, the diffuse sense that something was not quite right with a parent or other family member long preceded the sort of focused awareness which follows from having a specific diagnosis and/or a documented family history of HD. As such, several study
participants commented that they had, in anticipation of the interview, talked to their unaffected parent, siblings or other relatives in the effort to determine when an affected family member had begun to show signs or symptoms of the disease. At risk individuals were, in particular, anxious to ascertain the correct dates and establish when, in turn, their own awareness of the disease would have begun to emerge. In the following excerpt, Maggie recalls a recent conversation with her sister. She recounts some of the circumstances which anchor her memories (e.g., where she was living) but — in contrast with Jason — it was ultimately her felt and enacted memories of her father's unusual behaviour which allowed her to recall the process through which she became aware of Huntington Disease.

My sister and I were talking about that [first awareness of HD] and we figured we might have been 16 (...) 17, maybe somewhere in there that we thought/THOUGHT OF IT and saw that it was in the family. When I was, I'd say/I'D SAY closer to really knowing that it was there I was 27. Let me see, 26. 27. My dad came to visit me in {the valley} when I was living in {the Fraser Valley} and he would sit there and (unpleasant sound of fingers scratching repeatedly on the surface of the table) you could see the scratch, scratch/ and his feet were shuffling under the table (muffled sound of shuffling) all the time and it just sent me up the wall. And he never came home without a new dent in the car. E-v-e-r-y day. And how he managed to get through all this insurance and everything I don't know. But we/1 knew something was wrong and then/1 guess it had been mentioned somewhere in the family that Huntington's was there but it didn't penetrate to us at the time cause we didn't know what it really was. You know, "this old person got Huntington's." We didn't realize it was a hereditary disease or anything. So somewhere between 17 and 27 was before it really sunk in, I think, for me. (F, 50% risk, PT Candidate, 57 years, married, 5 children)

Maggie peels away several layers of awareness and, slightly rearranged, her accounting of the process yields this chronology. An elderly relative had an illness and someone somewhere may have mentioned the word “Huntington’s”; if so, it did not make a lasting impression; it didn’t “penetrate” because she didn’t know what it was. Then, somewhat later, something was wrong with her father; he was difficult and somewhat annoying to be with. She thought about “it” and somewhere between age seventeen and twenty-seven, she found out that her father had HD. She was twenty-seven when she was “closer to really knowing that it was there” but only when she learned that HD was an hereditary disease, did it really “sink in.”

Maggie’s account is instructive because it demolishes the apparent simplicity of my question (i.e., when did you first learn about Huntington’s being in the family?). The “when” part of the question provided an opening for Maggie to tell a story about how she learned that
Huntington Disease was in her family. This story revealed several discrete but interrelated aspects of her awareness: 1) recognizing and attaching significance to the words “Huntington Disease,” 2) observing or otherwise becoming aware that something was wrong with a family member (or was wrong with an ancestor), and 3) acquiring information about the symptoms, onset, severity and/or hereditary nature of HD.

Like Maggie, most at risk individuals had difficulty in distinguishing clearly between the age at which they first learned about HD being in the family and the age at which they understood that they (and other members of the family) were also at risk for HD. Most had been aware of each of these aspects for some time and it was, in consequence, very difficult to recall the lived experience of a more partial awareness. It was hard to know exactly when ‘it’ started. Some participants indicated that they were in the past (or are, even to this day) confused by certain aspects of the genetics of HD, but very few noted any significant temporal disjunction between initial awareness of the family history and comprehension of the fact that the hereditary nature of HD conferred some level of a priori risk for themselves and/or other family members. The two aspects were, more often than not, intertwined aspects of the way in which at risk individuals reconstructed their initial awareness.

The lack of separation between these two aspects of awareness may be an artifact of the process of memory in that it is difficult to recall when one did not know something that one has now known for quite some time. Alternatively, it may testify to the fact that it was the comprehension of actual or potential personal risk which ultimately compelled at risk individuals to take more serious note of that which might otherwise not have “sunk in.” In either case, it is noteworthy that very few at risk individuals recalled hearing of HD in some other context (e.g., through references to the famous folk singer Woody Guthrie) prior to the time at which they first became aware of the family history. And, none of the at risk or diagnosed individuals interviewed for this study professed to have known anything about the hereditary nature of HD prior to the time at which they first became aware of the family history.

This will no doubt change for subsequent generations but given that HD is not a common disorder and that it does, even now, have only a modest public profile, it is not surprising that few individuals interviewed in this study could recall ever having heard of the
disease prior to learning about the family history. As such, the relatively unknown character of HD was one of the most salient structural features shaping the experience of initial awareness for many at risk participants interviewed in this study. In contrast with HIV/AIDS, cancer or heart disease, HD did not seem to offer a semiotic handle; it did not announce itself widely or portend an inherent meaningfulness until something personal happened to make it relevant. Finally, as Maggie’s recollections of initial awareness of the family history of HD so clearly illustrate, the relevance of HD sometimes takes awhile to “sink in.” Even where the words “Huntington Disease” have been uttered and even where there is some level of awareness that HD is “in the family” it does not always follow that the disease acquires a high degree of topical relevance (Schutz, 1970) or a lasting autobiographical salience.

Age of Initial Awareness and Duration of Awareness

Before looking in detail at how various experiences of initial awareness were recalled and narrated, it is useful to briefly consider what PT candidates and other at risk individuals had to say about when they first became aware of the family history of HD. ‘When’ has, in this respect, two important components: age of initial awareness and duration of awareness.

Existing research (Quaid & Morris, 1993; Steenstraten, et al., 1994) suggests that the age at which at risk individuals first find out about being at risk for HD may have significant implications for how such awareness is constructed and for whether or not the at risk individual is likely to want to participate in predictive testing. This makes intuitive sense given that childhood, adolescence and early adulthood each impose a unique set of developmental challenges (Klein & White, 1996) which must, on some level, intersect with and shape the experience of learning about being at risk for HD. The relationship between age of initial awareness and subsequent desire for predictive testing is, however, not yet well–understood and it may be that the duration of awareness is at least as significant a factor as age of initial awareness or that the two are inextricably interwoven aspects of the experience (Cox & McKellin, in press). Service providers have always stressed that at risk individuals need time to

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6 Studies conducted in the Netherlands (Steenstraten, et al., 1994) suggest that at risk individuals who found out about being at risk for HD during adolescence were less likely to request predictive testing than those who found out about being at risk as adults.
assimilate new information about being at risk before proceeding with predictive testing. Especially where an adult has only recently learned about being at risk, they may not be in a good position to evaluate the potentially adverse consequences of undergoing predictive testing. Indeed, it may seem that the test offers the opportunity to simply get HD out of one’s life.

For the purposes of this research, I consider age of initial awareness in the following way. It is a conservative estimate which best represents my understanding of the point at which at risk individuals knew that a) someone in their family had HD and, b) HD is a hereditary disease. Knowing that HD is hereditary means that the individual understood that the presence of a family history confers some a priori risk for the disease; it does not necessarily mean that the individual possessed a scientifically accurate and/or explicit understanding of what the a priori risk of each potentially affected family member (including themselves) was. This is an important distinction to maintain for two reasons. First, there were some study participants who were aware of the hereditary nature of HD but confused about the characteristic pattern of inheritance. Second, there were others who clearly understood the scientific calculation of risk but preferred to adopt an alternative schema for thinking about the probability that they or another family member would eventually become affected with HD. The phenomenon of pre-selection (Kessler & Bloch, 1989) discussed in Chapter III offers one example of this.

In looking at the age at which at risk individuals participating in this study acquired initial awareness of the family history of HD, I found a wide variation in age: the earliest reported age was five while the latest was fifty. Nonetheless, almost all study participants who were themselves at risk for, or diagnosed with, HD (N=29) had known about the family history of HD for more than five years and approximately half had known for at least fifteen years. The individual who had known for the longest period of time (thirty–three years) was nine years old at initial awareness. Only two at risk individuals had known about the family history of HD for less than one year. Looking at PT candidates only (N=16), all but two had known about the family history for more than five years and six had known for twenty years or more.

7 Two items on the demographics questionnaires filled out by all PT candidates were helpful (i.e., estimates for age first learned about HD in family and age first learned at risk). Two of sixteen PT candidates recorded the same age for both items and this corresponded fairly closely with what was said in the interviews. In the few cases where the demographics questionnaires reflected a discrepancy with what was said in the interviews, I pooled all available information and derived my own best estimate.
Although the sample described here is too small to offer any generalizable conclusions about the relationship between age of initial awareness and duration of awareness, the data are suggestive of a pattern which makes intuitive sense given that predictive testing has now been offered for a little over a decade — that is, those who found out about the family history of HD early on in life had a longer duration of awareness prior to predictive testing than those who found out later in life. Age and duration of awareness are, however, only two of a number of important aspects of at risk individuals’ initial awareness of the family history of HD. They provide a useful composite picture but taken on their own, they tell us very little about the social interactions which shape, and are shaped by, individual and familial awareness of HD.

I have described above how difficult it was for study participants to distinguish between various components of their initial awareness of HD (i.e., hearing about HD, knowing that there is a family history, understanding the hereditary nature of the disease) and stated that it is, however, the narrative representation of these events which is ultimately the object of this analysis. What then are the central themes which organize and animate study participants’ narrative accounts of where and when ‘it’ started? And, how do specific instances of familial communication and/or miscommunication figure in the sequence of narrated events?

Transitions into Initial Awareness of HD

Alice Wexler’s book, *Mapping Fate*, offers a compelling depiction of how the saga of HD unfolds in one generation after another.

First there is the grandfather who has died of “nervous trouble” on the back ward of a state hospital, the uncle who attracts whispers and stares from the neighbors as he stagers down the street, the doctor who says, “Women do not get it.” Rumors of hereditary insanity linger about the family in question, along with a certain atmosphere of secrecy and suspicion. Divorce, arrests, abandonment, suicide punctuate the action. There is always a moment of discovery, when the protagonists finally learn the truth, usually after having several children. In the end, the characters all come to resemble one another, and the action winds down to a predictably gruesome close, with no resolution or release and always the promise of more performances to come. This is the drama of families with Huntington’s disease (formerly called Huntington’s chorea), played out with minor variations on stages around the world. (Wexler, 1995:xi, emphasis added)

The themes which Wexler sketches in this passage — the family history of “nervous trouble,” the lingering rumours of insanity, the moment of discovery and the depressing knowledge that the saga will be repeated — resonate strongly within many families’ experiences
with HD. Nonetheless, I wish to suggest here that while such summative depictions are powerful tools in eliciting insight and solidarity, they also evoke the desire to organize experience around a master narrative — "the drama of families with Huntington's disease."

"Minor variations" notwithstanding, this drama presents and represents itself as the archetype for understanding the common experiences of families with HD.

The power of an archetype is, however, also its downfall. In focusing on the common patterns, we lose sight of the individual life circumstances which shape, and are shaped by a changing biographical as well as social and historical awareness of HD. Given the shifting and uncertain burden of responsibility which new genetic knowledge and techniques confer on families at risk for HD, there is a need to move away from the tendency to depict familial experiences of HD in such homogenizing terms. Things are changing and intergenerational variations in the experience of living at risk for HD demand that we attend more closely to the significant variation in how at risk individuals and their families construct and represent their experiences of HD. The relatively recent advent of predictive testing for HD, in particular, demands that we now assess more closely the changing social and historical circumstances which structure experience, and shape the narrative frameworks of diverse biographical accounts. There is, as Frank (1995:158) argues, a need to attend to the nuances of the other's self-story and, in so doing, "anticipate changes in plot."

Moments of discovery are not all cut from the same cloth. Indeed, analysis of the way in which at risk individuals recalled and described the process of acquiring initial awareness of the family history of HD yielded at least four different types of transitions. In keeping with the themes and sequence of events emphasized most strongly in at risk individual's accounts, I refer to these four types of transitions as 1) solving the mystery, 2) discovering the secret, 3) learning by osmosis and, 4) out of the blue. These four types of transitions into initial awareness are not necessarily mutually exclusive; nor do I wish to suggest that they are exhaustive. They do, however, collectively illuminate the diversity in how at risk individuals recall the experience of becoming aware of the family history of HD. Further, only one of these types of transitions — discovering the secret — revolves around a moment of such singular importance that it might be summarized by Wexler's phrase "the protagonists finally learn the truth."
The five narrative accounts which are included in Appendix VIII situate each of the four types of transitions into initial awareness within the larger context of each narrator’s self-story. In what follows, I draw upon the stories which inform these accounts, as well as excerpts from the verbatim transcripts of these and other study participants, in order to describe and illustrate the diversity of responses to my overarching question about when participants became aware of the family history of HD. The primary focus is on how PT candidates narrate their recollections of initial awareness but, where appropriate, I also incorporate the perspectives of family members knowledgeable about the circumstances shaping the PT candidate’s initial awareness.

Solving the Mystery

For six of the sixteen PT candidates interviewed for this research, initial awareness of HD was preceded by a lengthy period in which a family member exhibited odd behaviour and/or experienced some form of mysterious illness. In the absence of a documented family history of HD, the cause of the ‘odd’ behaviour or illness was worrisome to the family (and no doubt the affected individual) but, more often than not, it was initially attributed to some relatively benign origin. Once the illness and/or ‘odd’ behaviour progressed beyond a certain point, however, its impact could no longer be minimized. Medical investigations were initiated and the diagnosis of HD was eventually ascertained and communicated by doctors to the affected individual and selected members of their family.

Colin’s story describes and illustrates this type of transition into initial awareness of HD. Colin was forty-one years old and at 50% a priori risk for HD at the time of our first interview. He had been married for seventeen years and had three daughters. Colin had a “normal childhood and family life” until his father “took sick” in 1966. For several years the family attributed his father’s illness to the trauma of a serious car accident and it was not until his father’s employer became concerned about safety in the workplace that a medical investigation was forcibly initiated by the company management. As Colin noted,

And the reason we, uh/how we ended up with him being diagnosed with Huntington’s disease was that his employer was quite concerned about his safety at work because they had noticed the difference in him, and certainly we did and we were still, you know, thinking it was something to do with the car accident or something was going on (…) But (…) again, looking back, it must have been very scary for him because he never, that I know of, never talked about the
changes that he must have noticed in himself. And then it must have been hard for him when the employers said, “Look, either go to the doctor and figure out what’s going on or you don’t have a job.” I think that’s basically what it came to. So the company actually put him in the hospital, I think, for I think he was there for about two weeks because it took them a long time to figure out what the heck was going on because it wasn’t such a... Huntington’s didn’t have the profile in the medical community that it does now. And it took a long time before one of the doctors kind of recognized what it might be and a number of them got together and pushed... (tape turned off briefly while Colin answered the telephone, then turned on again)...so, uh, anyways. That how we found out about Huntington’s Disease.

Colin was about seventeen when his father was diagnosed. As he recalled, HD was not well known within the medical community at this time (i.e., 1970). In addition, there were few medical records which might have documented Colin’s family history of HD and thus it took some time to diagnose his father’s illness. Prior to 1970, Colin and his family had no knowledge of HD but, with a diagnosis of HD, the pieces began to fall into place. Colin’s paternal grandmother had, before her death in 1934, been institutionalized because she “was acting different.” Her death certificate recorded the cause of death as “homesickness.” Colin knew very little about what her symptoms were but, in light of Colin’s father’s diagnosis, the family consensus seemed to be that she had probably had HD. During the course of the interview, Colin repeatedly expressed the wish that he could talk, as an adult, to his now deceased father about the family history because, as he put it, “you don’t ask questions, you don’t think about those things when you’re a kid.”

Like Colin, many of those at risk for HD acquired their initial awareness of the disease through a gradual process that was, more often than not, precipitated by the growing suspicion that something was not right with a parent or other member of the family. In the absence of a documented family history of HD, odd mannerisms or other suspicious behaviour were, however, often accommodated and normalized by a variety of lay and medical explanations for the causation of such troubles. More often than not these explanations were bound up with aspects of personal biography. For instance, Colin’s grandmother had immigrated to Canada from England and her odd behaviour was interpreted as a manifestation of a restless longing for home — a “homesickness.”

Looking across the narrative accounts of study participants, the prevalence and diversity of such explanations is striking. Familial explanations for the undiagnosed and mysterious
illness of an ancestor often revolved around some type of traumatic event; car accidents were, for both male and female ancestors, one of the most prominent of these traumatic events, the date of the accident invariably becoming a watershed in explaining and recounting the onset of illness. Elsewhere the trauma of war was seen to have lasting effects. A woman who had recently lost her husband to HD recounted how her father-in-law’s odd behaviour had long been attributed to a rare tropical disease which he contracted while stationed in India during WW I. And, in another family, the sister of a PT candidate described her family’s persistent belief that an aunt’s mysterious illness was related to “war nerves” — the aunt had been in England during the air-raids of WW II.

In the absence of a documented family history and/or diagnosis of HD, alternative explanations for the protracted odd behaviour of a family member are schema which organize and to some extent normalize phenomena that are otherwise incoherent and disturbing. They do, however, achieve this normalizing effect only in so far as they are capable of accommodating existing lay beliefs about disease causation and/or perceived vulnerability to illness. In this respect, it is interesting to note that men seemed to suffer from a difficult adjustment in the period leading up to and immediately following retirement. This difficulty in adjustment helped to explain the repeated occurrence of otherwise unexplainable odd behaviour. Reflecting on the behavioural changes he had observed in his aging father-in-law, one study participant stated that he had, prior to the diagnosis of HD, viewed these problems as a manifestation of “age, atrophy and lack of attitude.”

In contrast, the mid-life onset of ‘odd’ behaviour in women was often attributed to a range of female problems associated with menopause. Referring to her mother, who at the age of thirty-two abandoned her role as wife and mother in order to pursue charity work with everything from St. Vincent de Paul to “the blind dog’s society!”, one woman talked about how she and her siblings assumed that her mother was going through early menopause and, more generally, “just going daft like my grandmother was going daft.” Another woman talked about how her mother “went religious” and the doctors said, at first, that “she was just going through her change of life.” A third woman, recalled that in the absence of any other explanation she always thought that her mother, who was beginning menopause, had experienced “a nervous
breakdown.” Elsewhere women who were perceived as being excessively flamboyant, inappropriately subject to wanderlust and/or promiscuous behaviour were labelled as “eccentric” and “crazy” or misdiagnosed as schizophrenic or manic depressive.

For Colin’s father, the diagnosis of HD came at the end of a two-week hospital stay initiated by his employer. Colin recalled that although there were a number of tensions — or, as he put it, a “lot of physics” — in the family at the time of his father’s diagnosis, he didn’t really remember “too many incidents where it came to a head.” He and his family were, in many ways, fortunate because the Huntington Society of Canada was formed in the same year (1970) that his father was diagnosed. As Colin put it, “we went from not knowing anything about it [HD] to fairly quickly knowing a lot more about it than doctors or anybody else.” Further, Colin “always knew” that he, and his older brother, had a 50% chance of having inherited the disease but he didn’t worry about it at the time because “younger people tend to be invincible.”

Colin had, at the time of our first interview, been aware of the family history of HD for approximately twenty-four years whereas for Adam, initial awareness was a much more recent phenomenon. Adam’s family, like Colin’s, had no documented family history of HD. At the time of our first interview, Adam was twenty-three years old and he had learned of his father’s diagnosis of HD when he was sixteen. He was not married but did have a serious relationship with a woman who had a five year old son. Like Colin, Adam recalled that his initial awareness of HD was preceded by a protracted period of time in which he observed and gradually came to the realization that there was something not quite right about his father’s behaviour.

Adam’s first observations were of his paternal grandfather, Ward — “Granddad sits on the balcony and kind of shakes a little bit. Old people do that.” Some time afterwards, Adam and his parents were watching a television program (Nova) about the high concentration of Huntington Disease in Venezuela.

Looking at it, my Mum and my Dad said, “that’s Granddad. Granddad’s got that.” Or “Granddad acts similar to that.” And then I think that’s when it sort of clicked.

That was when I sort of went “hmm, you know, maybe it is something more.” Adam knew that his grandfather had, some years earlier, been through extensive medical testing but it was not until I spoke with Adam’s grandmother, Ethel, that I began to understand how
long and drawn out the process of diagnosis had been. For Ethel, it all began sometime in the mid-1970's when her sister noticed that something was not right about her husband Ward. Ethel could see that Ward had "started to move a lot" and she recalled in particular that his eyebrows would wiggle up and down. For a time, the family kidded that "it was the Danforth wiggle" but the movements became increasingly suspicious. Ethel began to piece together her memories of other men in Ward's family.

And I remember/ Now I think back, I remember his grandfather... he was quite bad but he lived to be in his, oh gosh I don't know, 80's or 90's. Then his [Ward's] dad/ I noticed he was shuffling a bit but/ his dad died of lung cancer the year that President Kennedy was shot [1963]. (F, not at risk, family member, 72 years, widowed, 2 children)

Ethel had become increasingly concerned by her husband's behaviour but it was not until her son Mitchell began to have the same type of movements that doctors began to talk about chorea.

...my son had started [the movements] in his early 30's/ and they brought him in and they [Ward and Mitchell] went before a panel of doctors and they ruled out Huntington's and for want of a better name it went out to UBC as the Danforth's/ unique to the Danforth family as Danforth's Chorea. This has upset me for a/ I'm still a little bit put out about it. I never did go back to tell him [the first doctor] that he had misdiagnosed. And the only way that I knew he had was that Ward was getting worse. I don't know how many years had gone by...I think it was 1986 and... we had another doctor... and I went to [this doctor] and I said that Ward was getting worse, could he go back to see [the doctor] to see if he could do anything for him. So she said..."I know a nice young neurologist... I'd like him to see." So we went to [see him] and he [the neurologist] put Ward in [hospital] for... a day... gave him a brain scan and...predicted it's Huntington's. So I made an appointment to go see the doctor at UBC/ cause there's two different opinions/ and the doctor [at UBC] looked at Ward and he said..."It definitely is Huntington's disease."

Prior to the diagnosis, Ethel had looked up the words "Huntington Disease" in the dictionary and, like Adam, she recalled seeing a program about it on television. She knew that HD was a "devastating" disease and remained annoyed about the misdiagnosis since there were, as she put it, "other implications." Had they known sooner that it was HD, and not a somewhat more benign form of chorea, she might have been more understanding of husband's moodiness and odd behaviour. Likewise, an earlier diagnosis would have meant that her husband could have been eligible for certain disability benefits. Ethel's only comfort was that her son Mitchell and his wife Joyce (that is, Adam's parents) had started a family well before the misdiagnosis and as such, any false reassurance that might have arisen from the misdiagnosis could not have been a factor in their reproductive decision-making.
Ethel was at a loss to explain why the misdiagnosis of Danforth's Chorea persisted for as long as it did. She could not recall the doctors ever explaining to her why they had initially ruled out HD. Her son, Mitchell (Adam's father), did, however, have a theory that it had to do with the way in which the disease had manifested itself within his family. As Mitchell said,

There was no diagnosis of the disease in our family when I was his [Adam's] age. So until I was diagnosed with the disease it was called Danforth's chorea... Some neurosurgeon in Vancouver made a big mistake. And my dad was in the hospital, the General, and went through all kinds of batteries of different tests and spinal taps and all kinds of things. They had both of us down at the same time touching our tongues to our noses and "watch our fingers." They got 25 or 30 doctors in the room all watching us. And I guess in our family it manifests itself in the male members only. And I guess that kind of gave them a loop or whatever. They made a mistake... In retrospect, maybe it was a healthy mistake, maybe... our family didn't have to know this then. (M, Diagnosed with HD, 47 years, married, 2 children)

Adam and his younger brother were both living at home when their father Mitchell's diagnosis of HD was confirmed. Remembering the years leading up to the diagnosis, Adam recalled that he had always thought of his father's movements and hyperactivity as part of a particular constellation of behaviours that were simply an aspect who his father was.

My dad has never been able to sit still, ever. Hyperactivity describes my dad. He's/ get up in the morning, go cut the lawn/ get up in the morning, work on the boat. You know he's always busy, you know. Or/ I remember talking about it at a restaurant or something/ "Dad behave yourself. I'm going to nail your feet to the floor for kicking me under the table." You know, stuff like that. I never really, you know, just/ Dad's just wired. Just the way Dad is, right?

Even when he learned that his father had HD, a part of Adam maintained that, it's just Dad you know. It's just the way Dad is right. And it's not like Dad has Huntington's, and this is why Dad is this way, it's just Dad acts this way cause Dad is Dad, you know. It's just I've always/ It's always been that way.

Adam's parents also recalled that he and his younger brother were quite accepting and nonchalant about the news that Mitchell had been diagnosed with HD. As Mitchell described it,

...when we told them [Adam and his brother] what the scoop was, in my mind I had all this horrible things, and then in reality, you know, they said, "well you're old and I'm young and it's never going to happen to me." Like that was their attitude. That I'm a dad and they're a kid. There's going to be a lot of years before they're my age and so don't worry about it. (M, Dx, family member, 47 years, 8

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8 Mitchell is referring to various aspects of a standard neurological test utilized in the clinical diagnosis of HD. This test is known as the QNE (Quantified Neurological Examination) and it is considered a highly sensitive indicator of whether or not a patient is showing signs and symptoms of HD. Patients are asked to perform a number of routines which test cognitive abilities and memory, posture and coordination, conversational speech and eye movement.
married, 2 children)

Adam’s mother, Joyce, also attributed the boys’ nonchalant response to their youthfulness but she was careful to add that she and Mitchell had gradually laid the basis for “breaking” the news to them.

By the time he [Mitchell’s father, Ward] was diagnosed with Huntington’s he had chorea movement. He was, I think, starting to slur his words and so uh I think/ [one of the doctors at the clinic] had suggested that you know, children/children adapt...if you just bring it into the conversation instead of making a big deal out of it and so we would talk about granddad, you know, that granddad was having trouble pronouncing his words and so forth and that it was a genetic disease and that we hadn’t known anything about it before they were born... And we discussed it not at any great length. Kids just/ It just seems to be like water off a duck’s back...

We tried to talk about it with them and I guess we just kind of made it so that we could talk about it, you know what I mean. It wasn’t something that was hidden from the boys. They had to know that it was genetic. But they don’t bring it up.

(F, not at risk, family member, 47 years, married, 2 children)

When I asked Adam if his father’s diagnosis had come as a surprise to him, he replied,

For me I don’t think it did cause I remember Dad coming home. I was in the kitchen cooking and... they came back from Vancouver...and I just looked and I was like, “oh yea, I know what’s going on here.” And Dad came up and told me and said “Give me a hug kid” and he just broke down and started to cry. It was like “whoa, Dad doesn’t do this. This is something new.” Like Dad cries when the Canucks win a hockey game. Dad cries when things like that happen but Dad doesn’t you know/it was just like “whoa.”...I don’t know if it was a case of it being a surprise or whatever, but I don’t think it was...I think I knew about it or decided that this is the way that things are going beforehand. Like with me I don’t look at/ I don’t tend to focus on the bright side of things. It’s like, “okay, you’ve got it.” If you don’t, it’s an added bonus. I do it that way because if you get your hopes really wired and then everything comes falling down around your ears I hate that. I really hate that. I used to do that a lot. I don’t know. It makes me feel like I’m a real nihilist or something like that. (emphasis added)

Adam attempted to answer what seemed to be a fairly straightforward question (e.g., whether or not he anticipated his father’s diagnosis) by first recounting his memories of his father coming home and telling him the news and then reflecting upon why he thought he remembered the event in this way. The emotional intensity of his memory — the experience of his father crying and the recognition of this being “something new” — was, however, shaped by his awareness of how he now prefers not to get his hopes up. Thus, he became uncertain about how or if he “knew” or decided beforehand that “this is the way things are going.”

For Adam, who was at the time of our first interview only three weeks away from learning whether or not he too would eventually have HD, the tension between hope and
nihilism was profound. His telling of the story of his father's diagnosis was not simply an effort to recall and describe what happened seven years ago; it was also an effort to say something about how the diagnosis of HD forces a re-evaluation of all that has gone before it ("Dad acts this way cause Dad is Dad, you know. It's just I've always/ It's always been that way"). What was HD and what was "just Dad"?

The transitions into initial awareness described by both Colin and Adam involved a protracted period in which there was a growing awareness that something was not right about their respective fathers' (and grandfather's) behaviour. In the absence of a documented family history of HD, Colin's family attributed this odd behaviour to the trauma of a car accident while Adam thought of his Dad's behaviour as a normal manifestation of personality and his grandfather's shaking as an aspect of aging ("That's what old people do."). With their respective fathers' diagnoses of HD, however, Colin and Adam each acquired new information. Colin emphasized that he and his family were fortunate in that the newly formed HSC provided them with a wealth of information about HD. In consequence, he "always knew" that he had a 50% chance of having inherited the gene for HD. Adam, on the other hand, gained his initial understanding of the hereditary nature of HD from several sources: there was the television program he recalled watching with his parents and, as his mother recalled, there were casual conversations about HD as it related to Adam's granddad's condition.

Neither Colin nor Adam were able to pinpoint a particular communicative event as pivotal in changing their awareness of HD. The process of acquiring initial awareness was gradual; it involved firsthand observation of an affected family member, a period in which the affected family member was undergoing medical investigation and finally, the news that the family member had been diagnosed with HD. Colin and Adam were, however, living in close proximity to these events; they were both in their teens and still living with their respective families. Not all PT candidates who talked about their experience of initial awareness as an instance of solving the mystery were as youthful and/or unfettered in terms of family responsibilities. Others (such as Maggie) had left home and begun a family of their own before becoming aware of the family history of HD and, as such, the impact of learning about the family history and hereditary nature of HD came to acquire an additional layer of significance;
personal risk of having inherited the disease was compounded by the concern that the next
generation was also at risk. Carla and Gabriella’s stories, discussed below, illustrate this and
other concerns arising from a quite different type of transition into initial awareness.

Discovering the Secret

The narrative accounts of Carla and Gabriella contain elements of solving the mystery;
there is a protracted period in which a family member suffers from an illness which is
mysterious to one or more other family members. Carla and Gabriella’s accounts are, however,
distinctive in that the diagnosis of HD in a family member was concealed by one or more family
members for a prolonged period of time. In this sense, HD was more than a mysterious illness.
It was a secret which was, sometimes guarded and sometimes simply not talked about. The
family history of HD had to be actively discovered by the protagonist; circumstances alone did
not simply reveal the truth. As such, both narratives contain what I shall refer to as pivotal
disclosures — that is, disclosures which confirm and/or compel a revised or even fundamentally
different definition of the situation on the part of one or more interactants.

There were four PT candidates interviewed in this study who described their initial
awareness of HD in terms of discovering the secret; the parallels between Carla and Gabriella’s
accounts will be used here to illuminate the social dynamics of this type of transition into initial
awareness. In contrast with Colin and Adam, both Carla and Gabriella were young adults at the
time of initial awareness. Further, both Carla and Gabriella knew, at a very young age, that their
respective mothers were unwell and both struggled to understand the impact that this had on
their childhood and early family life. Both laboured for a long time with the suspicion that they
were not being told the (whole) truth about the mysterious illness which seemed to lurk in their
respective families but neither were they able to obtain from other family members and/or
doctors the information which would confirm or deny their suspicions.

At the time of our first interview, Carla was forty-two years old and at 50% a priori risk.
She had known about the family history of HD for twelve years. She was divorced but living
with her partner Tom; neither had any children. When Carla was born, her mother was incapable
of looking after her; thus, Carla spent the first twelve years of her life with an aunt and uncle
Carla did not say very much about her mother’s illness during the interview but the combined memories of losing her aunt and her mother prompted Carla to reflect on the feelings that she had about the times she visited her mother in the hospital.

She [my aunt] passed away when I was 13, so that was a real loss for me. And then my mom passed away when I was 15. She was in the mental hospital mostly those years and I mean, my dad used to take us up there to see her and it was whacko. I mean it was. It was like a/ I couldn’t believe/ I mean, now, that I think about it, I mean it’s hard to believe that you can allow somebody you love to be in one of those places. I mean especially in the 50’s...

At the time she visited her mother in the mental hospital, Carla did not know that her mother had HD. Carla had nonetheless mentioned, during her first pre-results counselling session, that her mother had the “typical choreic movements” and moreover, that her mother did not seem to recognize her; she was “in another world.” During the interview, however, Carla’s recent memories of visiting her younger sister Nicki (who had, at the time of our interview, been diagnosed with HD for nearly ten years), prompted her to offer a less-medicalized perspective on her mother’s illness.

...I remember going to see her [my mother] and it was really funny because all she wanted to do was eat sweets, eat sweets, and smoke cigarettes and drink strong coffee. (laughs) And it’s funny because when I see Nicki now, my sister is the same way. It’s like, “Did you bring me chocolates?” (laughs) You know, “did you bring me any chocolates or any sweets?” And they always like to drink lots of really strong coffee. I don’t know if that’s a symptom. (emphasis added)

Carla’s mother and sister shared, in this instance, a very specific set of desires. Carla was struck by the intensity and similarity of this and wondered if the craving for chocolate, strong coffee and cigarettes is a symptom of HD.

Carla returned to talking about her mother’s illness and went on to say that her father had told her (and her siblings) that her mother had some type of “nerve damage.” Doctors had been treating her mother for TB with a very intense antibiotic and Carla said that she recalled hearing the doctors talking about a “scar on her lungs.” After her mother died, Carla persisted in asking her father about her mother’s illness. He wouldn’t “really answer” her and every time Carla talked to him she would get a different answer — “it just didn’t make sense.” But her father was
a “great storyteller” and, as Carla admitted, “when it’s your father speaking to you, well, for me anyway, whatever he said, I believed him.”

Carla had grown up in California but she left home soon after finishing high school and moved to B.C. to train and subsequently work as a dental receptionist. She travelled extensively but always maintained fairly close ties with her father and his second wife as well her two sisters and brother. She visited at least once a year but it was not until she was thirty years of age that she “found out about Huntington’s.” On one of her visits home, Carla and her two sisters were over at an aunt’s (her mother’s sister) house.

...we were sitting around the table this one night and my aunt was telling us about how her father had passed away. And he was found hanging in the barn of his farm... and my mother was the one that found [him]. Now they don’t know whether or not he had Huntington’s but he probably did.

Carla also found out (from her maternal aunt) that her mother’s illness may have begun at an early age. Before Carla’s parents were married, the aunts and uncles tried to tell Carla’s father that “maybe it wasn’t a good marriage” and that “she [Carla’s mother] just didn’t seem to be all there.” Her father wouldn’t listen and, in hindsight, Carla admitted that because “he’d tend to overlook things and pretend that things were okay” she had no way of knowing if he really had “any inclination as to what was happening.” Perplexed as to why no one had told her about any of this sooner, Carla learned that the aunts and uncles had collectively assumed that she (as well as her brother and two sisters) knew that Huntington’s was in the family. They observed that “none of us married and had children” and “they figured that this was a choice we had made so nobody said anything to us.”

It was possible that Carla’s eldest sister had known some of this before the aunt’s disclosure “because she was pretty inquisitive” but if so, she had not communicated with Carla about it. Recalling that she had felt relieved when her aunt told her about the family history, Carla stressed that it cleared up all of her questions, “it was good because we could carry on...I could make some choices about my own life from that point on.”

Why Carla’s aunt decided to talk about her father’s (i.e., Carla’s grandfather’s) suicide and the family history of HD at the time she did remains uncertain. Carla did not speculate about this or mention that she and her sisters had been prompting her aunt for such a disclosure
but it is possible that Carla’s younger sister Nicki (who was also visiting with the aunt) was beginning to show signs and symptoms of HD. Nicki was diagnosed with HD only two years after the aunt’s disclosure and thus it may have been that the aunt wanted to know with some certainty that her nieces also knew about the family history of HD. Perhaps she also hoped that Carla or one of her sisters would ensure that Carla’s brother, who was not present at the disclosure, knew about the family history. Carla could not recall when she spoke to her brother about this but she thought she had written him a letter and/or spoken with him about six months later on her next visit home.

Like Carla, Gabriella also experienced the profound impact of HD at a young age. Gabriella did not actually learn that there was a family history of HD until she was twenty-five years old, married with one child and another on the way. When I first spoke with Gabriella she was fifty-four years of age and married to her second husband. Gabriella’s mother was about forty when her “symptoms appeared” and Gabriella was about seven.

Recalling that she grew up in Alberta in the 1940’s, Gabriella described how she and her two older sisters “lived in fear a lot of the time” because “my mother really beat us.” The beatings were worse for her sister Annette than they were for her, or her other sister Karen, because Annette was “a lot like my Dad” and “my mother was rather paranoid about my Dad.” Having recently attended a funeral, Gabriella was surprised at how being there took her “right back” to when she was young and she and her sisters would go to church. “Church was the place that saved me from the bogeyman, the dark at the bottom of the stairs.”

When Gabriella was fourteen, she and her sisters were told that her mother had been diagnosed with schizophrenia.

They thought she had schizophrenia. When I was young/ 13 or 14/ we went to a psychiatrist who said that “weren’t we lucky because the first few years of our lives were fine” and that my mother had schizophrenia. Then she was going to go and have shock treatments and all sorts of things. She was trying to find something. And I really feel sorry for her because she didn't have a clue, of course, what was wrong with her.

When Gabriella was sixteen, her mother left home and went to live in Vancouver. Her mother’s odd and sometimes dangerous behaviour (e.g., setting fires in her apartment) resulted in frequent evictions from where she was living and, by the time Gabriella was twenty her
mother had been admitted to a mental hospital near Vancouver. She was later transferred to a facility in Alberta. Gabriella wrote to the psychiatrists at the Alberta facility to ask if they had been able to diagnose her mother's illness. Gabriella was, by this time, married with one child and living in Ontario. She was concerned because her aunt Sylvia (i.e., her mother's sister) appeared to have "an illness which was quite similar to my mother's, although perhaps not as bad." She mentioned this in her letter but the psychiatrist did not write back. "So that got me nowhere."

In the summer of the following year (1965), Gabriella's mother "took a terrible down turn." Now pregnant with her second child, Gabriella went back to Alberta to see her mother. Doctors then told the family that they thought Gabriella's mother had HD but that they would not know for certain until after she died and they were able to do an autopsy. The timing of this news could not have been worse. Gabriella had a "really difficult time" in the months leading up to and immediately following her pregnancy. She walked the streets wondering "What am I doing having had this baby?" Her doctor (in Ontario) received a letter from the Alberta mental hospital stating that "HD was only inherited through the female" and that it "skipped generations." Gabriella said that she and her doctor "knew this wasn't true."

When the autopsy confirmed that her mother had indeed had HD, Gabriella then asked her uncle if her Auntie Sylvia (his wife) had been diagnosed with HD. Gabriella was still hopeful that there had been some mistake, that her aunt and her mother had "different diseases." Her uncle said he "didn't know." Several years later, however, Gabriella learned that her uncle had not told her the whole truth about what he knew.

Shortly after she and her family returned from a two year period working with CUSO in Africa, Gabriella received a letter from her uncle. In the following exchange she describes the letter and her feelings about the pivotal disclosure that it contained.

G: Anyhow, then I went and taught with CUSO and in 1969 I came back/ I came to Vancouver. My aunt who had Huntington's died/ of cancer and my UNCLE (.) wrote me a letter saying, "I think it's/ I don't know WHY I didn't tell you before but your aunt was actually diagnosed as/ that they were suspicious that she had Huntington's in 1962."

S: Oh really?

G: YES.
S: So even when you'd asked him [about her/]

G: [He KNEW.] He KNEW. He KNEW before my children were even born.

S: [Oh.]

G: [I KNOW.] And he wrote this letter saying, "I don't/I don't understand my reasons now but I'm writing to tell you that I'm really sorry and here's the drugs she was on and here's all the situation, and"/I, I really love/I really liked that uncle a lot. And I liked that aunt too. And I could never figure it out and unfortunately he died about 3 or 4 months later, so I/ Cause I really was going to go back to Ontario to visit him. [See] and then he died.

S: [You didn't] have a chance to see him again?

G: So I never got a chance to talk to him or see him. But I still have the letter upstairs. So I don't understand his reasoning. But you know, people are so funny about this kind of thing. (...) I mean even my own sister, I don't know whether she would, you know, she wouldn't disclose that kind of thing necessarily.

The uncle's disclosure was pivotal in that it fundamentally changed Gabriella's awareness of what had occurred. Her repeated emphasis on the words "he knew" underscores the profound significance that this moment of discovery held for her. Had she known in 1962 that doctors suspected her aunt had HD, Gabriella would have had the opportunity to consider the implications of this for reproductive decision-making. In describing these events, however, Gabriella did not explore the possibility that she might not have had her two children if she had known in 1962 that there was a family history of HD. My flat sounding “oh” and her emphatic “I KNOW” seemed, within the context of the interview, to be sufficient signifiers of the profound implications this information might have had for Gabriella in reproductive decision-making. Thus Gabriella immediately went on to finish summarizing the letter and recount how much she liked her aunt and uncle. Any regrets she might have had about the timing of her uncle’s disclosure therefore seemed to centre on the lost opportunity to talk to her uncle and understand his reasoning. As she concluded, “people are funny about this kind of thing.”

The lost opportunity is a theme that we have already encountered in Colin’s story. The issues are slightly different but Colin, like Gabriella, expressed regret about not being able to talk with a family member (his father) about the implications of the family history of HD. Further, Colin and Gabriella both highlighted the desire to talk specifically about issues of family communication. Colin noted that his father never talked about his experience of onset but
it must have been "very scary." Would it have been any less scary for Colin's father or Colin for that matter, if they had talked about it? And Gabriella pondered whether or not her sister would disclose "that kind of thing." Would talking to her uncle have helped Gabriella to understand why people are "funny" about talking about HD? And, more specifically, how to respond when other people are "funny"?

Carla and Gabriella experienced a long period in which their suspicions about their respective mothers' illness went unconfirmed. Both had made protracted attempts to discover what was going on but both were ultimately stymied in their attempts by one or more family members who were unable and/or unwilling to talk about the family history of HD. As Carla understood it, her awareness of HD was limited by father's "denial" and her aunts and uncles' collective assumption that she and her siblings must already know about the family history. For Gabriella, awareness of HD was first thwarted by her mother's misdiagnosis (as schizophrenic) and then by her uncle's protracted unwillingness to tell her that her aunt had HD. The respective communicative interactions of Carla's father and Gabriella's uncle were, in this sense, key to understanding how the process of initial awareness unfolded. Both men could have provided information which might have eased our protagonists' transition into initial awareness but neither chose to do so. Further, neither Gabriella nor Carla were able to elicit such information — in both cases, the disclosures which ultimately resulted in their initial awareness of HD came from other sources (i.e., Carla's aunt and Gabriella's mother's physicians).

Carla's moment of discovery — which came long after her mother's death — occurred when her aunt told her and her sisters about the family history of HD. At this point, Carla also learned why her aunt had never talked about this with her before. Gabriella's moment(s) of discovery were tied to several events: a few months before her mother's death she learned from doctors that her mother probably had HD. Then, after her mother's death, an autopsy report confirmed the diagnosis. It was not, however, until Gabriella received her uncle's letter that she knew for certain that her suspicions about her aunt's illness had been correct and further, that her uncle had known since 1962 that his wife probably had HD. Gabriella does not, even now, fully understand why he did not tell her sooner and why, despite her questions, he denied having any knowledge of HD.
Non-disclosure of information about a hereditary disease such as HD may be motivated by a number of concerns. It is important, however, to recall that in this instance we are talking about non-disclosures as they occur within the context of a relationship between someone who is in the know but is not at risk (i.e., Carla's father or Gabriella's uncle) and someone who is not in the know but is at risk for HD (i.e., Carla and Gabriella). From the standpoint of Carla's father or Gabriella's uncle, non-disclosure might have been in accordance with their respective wife's desires; it might have been a strategy for protecting others from worry and anxiety; or it might have been a method of avoiding being the bearer of bad news.

At the time of the first interview, Carla had not observed any positive changes in her father's ability to talk about the family history of HD. He's "more mature" and "he has more stories" — "I gave you this information way back when, don't you remember?" — but he has, ever since her sister Nicki's diagnosis, alternately maintained that Nicki does not have HD — "she's got something else" — and/or that it was "that damn car accident" that brought it on. In contrast, Gabriella's uncle wrote a letter which was, among other things, an apology. He said he was "very sorry" and that he did not understand his own reasoning. He provided information about the various drugs his wife had been taking. He initiated a conversation that Gabriella now wishes she had been able to continue.

**Learning by Osmosis**

Learning by osmosis is, in many ways, the anti-thesis of discovering the secret. As the metaphor of osmosis suggests, this type of transition occurs gradually. It is the product of living in close geographic and/or social proximity to a family member diagnosed with and/or aware of, the hereditary implications of HD. Unlike the previous two types of transitions — solving the mystery and discovering the secret — there is someone in the family who is both aware of the family history of HD and (somewhat) willing to talk about it. The tell-tale signs and symptoms of HD do not appear as manifestations of a mysterious illness and, though it may not have been a regular topic of conversation, the family history of HD was not hidden. As such,

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9 Osmosis is a process of diffusion wherein fluids pass through a semi-permeable membrane so as to equalize the concentration of certain elements on either side of the membrane. Diffusion occurs from a solution of low concentration to a solution of higher concentration.
HD did not have to be discovered by the protagonist; nor was there a protracted period of suspicion. There were five PT candidates interviewed for this research who talked about their initial awareness is this way.

At the time of our first interview, Regina was twenty-eight years old; she was married and had an eight year old son. She was at 50% a priori risk and, less than two years earlier, had lost her father to HD. Retracing her awareness of the family history HD, she explained that she and her older brother and sister had grown up knowing about it. Her paternal grandfather had died of HD on the day after her parents were married and her grandmother, in turn, became quite consumed by the topic of HD. In answer to my question “where in all of this did you first learn about the family history of Huntington’s?” Regina replied,

Uhm (...) Probably as soon as I was old enough to understand because grandma was the type of person that ate slept and breathed it. It was like almost every second sentence/this was what was out of her mouth the whole time I was growing up so/when we were very young it was always in the foreground when grandma was around our house...

Her grandmother’s persistence with the topic of HD did, however, become tiresome. From Regina’s point of view, it was as if her grandmother thought that nobody else ever talked about HD; that they had never heard of it; or that they were trying to hide from it. To Regina, HD “was just there” but to her grandma “it was everything.”

…it’s just always been/even before Dad got sick it was always there. Always. She wasn’t a bad person, she just drove me nuts when I came to it and you finally say “enough’s enough, we have a life, we don’t want to hear about it anymore,” cause we all knew about it/there was nothing that we didn’t know about it that she was going to tell us because if we ever had any questions Mom and Dad always answered them and they were always very honest with us...

As Regina stressed, her parents were always open to talking about HD or any other issues. The family history of HD,

...was never kept a secret/it was/one thing about Mom and Dad/they were always/ if we had a question about anything we could always ask it. Sometimes we’d get shit afterwards but we could always ask. They were like that when it came to anything/drinking, drugs/and they were/they used to tell us the pros and cons to everything. It wasn’t “don’t do this because it’s bad” or because/ if there was any good to be found they would try to do it, right.

Regina’s father was in the military and the family moved a number of times, eventually settling near Vancouver. The grandmother who had been so persistent in talking about HD was many miles away in Ontario but the “always thereness” of HD persisted; it just wasn’t “in the
forefront." Regina was about sixteen when her father was diagnosed with HD. She was living at home at the time but her two older siblings had left home. Her mother was the first to notice early symptoms but Regina also recalled starting to see “little signs” — the “handwriting goes” and there’s a “lack of coordination in some things.” Her parents did not have a family meeting and there wasn’t “a big to do made about it” so Regina and her brother and sister “noticed” at different times.

I asked Regina if she remembered a time when she and her family discussed their awareness that her father was having signs of HD or whether it was something that didn’t really need to be talked about because everyone knew that everyone else knew. Regina explained,

...we just never talked about it [HD], not cause we were hiding from it, just because, it wasn’t part of our life yet/ like it was there but not in the forefront of our lives, and even when Dad did get sick, it was still, we went around doing a lot of the same things/ take him to football games and shopping and whatever else.

In talking with Regina, I began to understand how it was that too much “talk” about HD could signify the inability to get on with living and enjoying life. As Regina said in reference to her grandmother’s obsession with the topic, “you finally say ‘enough’s enough, we have a life, we don’t want to hear about it anymore.” Hearing too much about HD was akin to not “having a life,” not having other more important, interesting and enjoyable things to do and talk about. Moreover, too much talk about HD, as it was personified in her grandmother, stood for everything that was wrong with the way that people with HD were and, still are, treated.

Regina’s grandfather had been put in a psychiatric facility before he died “because that’s what they did with these people” but, for Regina, there was no explanation for the fact that her Uncle Conrad, who also had HD, was (at the time of our interview) living in a facility in Ontario that was “almost the exact same type of place they put grandpa in before he died.” It was “a barrack”; the walls were “filthy dirty”; “everybody was barred into their bed.” Her uncle had a feeding tube and was not allowed to have anything orally. He had few if any joys left in life. This was profoundly disturbing to Regina and she recalled that she had, for some years, fought with her grandmother about the quality of care that her uncle was receiving. Her grandmother would, however, always say “Well, I know what’s happening...I’ve dealt with Huntington’s before.” This was “maddening” to Regina — “they haven’t done anything differently than what
they did when my grandfather had it. Which is stupid. Stupid, stupid, stupid.” In contrast, Regina’s father received the best care possible. “Nobody had the attitude ‘no Dad, you can’t do that cause you got Huntington’s.’ It was ‘well, we’ll find a way to do it and if that means, whatever/ we’ll do it’...like it was a totally different attitude from what I was raised with around grandma.”

Toward the end of his illness, Regina’s father lived in a nearby facility. Significant family discussions about HD occurred when there were decisions which had to be made about his care. These discussions were, however, never simply about her father; they were discussions which included her father.

Dad was included in things/ even when it came to financial things/ there was certain things/ it was like when/ I don’t know what Mom was doing/ she was changing the address on his pension or something and the woman on the other end of the phone said “you’ll have to get power of attorney over him to do that.” Mom said “I’m just changing his address, she said “I don’t want to get power of attorney over him”/ it was really funny cause the point came where there was some stuff/ legal stuff that Mom had to take care of/ she just couldn’t get Dad’s signature anymore/ so she asked him for power of attorney. Dad said “yes, I will give you power of attorney, if you will give me power of attorney.” So that’s what happened. They gave each other power of attorney. Mom said “that’s fine”/ that’s kind of the way it was done...that’s the way we did it...because it’s his life...he’s the one who’s sick, he’s got the options to make, and/like some people where/ like in Uncle Conrad’s case/ the decisions and all the questions were always answered by somebody else, he’s never been asked. It’s just not right/ there’s no reason for it.

Regina was very close to her father and, when he died (about a year before the interview cited above) the loss was, in many ways, profound. He had, as the above passage suggests, never been allowed to die a “social death” (Sweeting & Gilhooly, 1997) before it was time to die a biological death and one of the most salient implications of this was that he somehow held family communication together. Regina and her two siblings had, at one point or another, been on the “outs” with their mother — “she’s always got one of us that she’s not talking to” — but as far as she could remember the only time that they had all been on speaking terms was just before her father died. “When my Dad was alive he would calm her down. He would say ‘Okay Donna, you’ve overstepped your bounds, that’s enough’.”

Like Regina, Nigel and his younger sister Landis also became aware of their family history of HD through observation and a gradual process of assimilating information. Nigel and Landis were, however, unique in this study because it was their father’s predictive test results
(rather than diagnosis) which transformed their 25% a priori risk into a 50% a priori risk. The fact that their father had *not* been diagnosed with HD was, as we shall see, a significant influence on how each experienced initial awareness of the family history.

At the time of the first interview, Nigel was twenty-nine years old and single with no children. He had known about his father’s test results for about nine months. Recalling with some difficulty the point at which he first knew that his paternal grandmother had been diagnosed with HD, Nigel stated that “it wasn’t a significant emotional event that stuck in my mind. I think that’s what they call it when you remember something like that from the past.” Because Nigel recalled visiting his grandmother before leaving on a trip to Europe, he was able to determine that he must have been about fifteen. He knew that HD was hereditary but he did not, at that time, understand that he was also at risk for the disease. Nigel had heard that “the women in the family are the ones that generally carry Huntington’s” and thus he believed that his “aunt would be at more risk” than his father and that there was only “a marginal chance” that his father would ever have it. When I asked Nigel where he had heard this idea he said,

> I think my father. And uh (..) back then he really had a hard time about it (clears throat). It was definitely a serious strain on him and, uh, you just couldn’t say Huntington’s disease around him. And uh/ and uh/ it was really tough for him to (..) even visit my grandmother. You know, it’s kind of a family curse.

Nigel’s recollections of his father’s anxiety about the family history of HD prompted me to ask him if it had been his father who had told him about his grandmother’s diagnosis. Nigel said that “it was either him or my mother, I can’t remember” but then added, “my mother seemed to deal with those kinds of things uh/ with us/ he was so freaked out about it.” Somewhat later in the interview, however, Nigel explained that he had never really been concerned about being at risk for HD because “we were all convinced that my Dad didn’t have it cause he was so convinced and said that and let everybody know it.”

At the time of the first interview, Nigel’s younger sister Landis was twenty-seven and thinking about having the predictive test. She too was single with no children and, like Nigel, she had been convinced that her father did not have HD. In contrast with Nigel, however, she had only just learned about her father’s predictive test results about two weeks before the interview.
Landis recalled that her initial awareness of the family history began with her grandmother’s eccentric behaviour and subsequent diagnosis of HD. Nonetheless, Landis emphasized that it was in and through discussions with her friends, and then her father, that she initially came to perceive HD in a fairly non-threatening way. As she explained,

"I don’t have any recollection of anybody even really explaining it to me. It was just like, “your grandmother has Huntington’s. It’s like Alzheimer’s or Parkinson’s.” Seemed to be happening to a lot of people. I rem-/ and then later on/ maybe a couple of years later sort of having Dad say “Well, okay, you can’t get it unless one of your parents has it. So if one of your parents has it you have a 50% chance of getting it.” And I still don’t recall it having any great impact on me whatsoever."

Landis noted that Huntington’s initially seemed to her to be like Alzheimer’s. Many of her friends talked about the crazy things their grandparents would do — “you know, watering the driveway and whatever” or “my grandfather’s got it [Alzheimer’s] and you wouldn’t believe what he did last week” — and these stories reminded her of her grandmother’s “eccentric” behaviour. At parties these stories provided “a little bit of a source of amusement” but, as Landis cautioned, “we were never exposed to the horror that say, like a spouse would be exposed to...we didn’t really understand it.” For Landis, there was nothing “earth-shattering at all” about the fact that her grandmother had HD. It was perceived as “something that affects you in your old age” and “it was really a bit of a non-issue.”

Nigel and Landis acquired their initial awareness of the family history in their mid to late teens. With news of their grandmother’s diagnosis both also learned that HD was hereditary but neither perceived HD in an overly threatening way. Even when their aunt began to exhibit the characteristic jerky movements and slurred speech neither recalled that they had been overtly concerned about their own risk for HD. Nigel initially believed that it is only women that carry HD (hence, his aunt was at greater risk than his father) while Landis thought of HD as a disease which only affects old people. Although these ideas were scientifically untrue, they were accurate representations of what Nigel and Landis knew and observed about the way that HD had manifested within their own family. When their father received his predictive test results the situation changed and personal risk took on a new significance.

Nigel was fairly certain that he had known for about “7 or 8 or 9 years” that there was a 50:50 risk for each offspring of an affected parent but he recalled checking with the genetic
counsellor about this and the “male/female [issue]” when he went to the clinic with his father. Recalling that he had been more upset with his father’s predictive test results than his father, Nigel said,

He [my father] just went “ah, nuts. That’s/ you know/ That’s really too bad.” I felt like punching the wall. I just couldn’t believe it. I was just in shock. But then I pulled it together and (...) just looked at everybody in the room and just said, “Well, nothing’s changed here. You had it all your life and only difference is now you know about it and/ and uh (...) you know, it’s not lymphatic cancer or/ It’s not/ You don’t have 30 days to live kind of thing. So.

In telling this part of his story, Nigel seemed to be very focused on the impact of the news for his father. He laughed when I concluded that perhaps he had not been thinking about the implications of the news for himself. “No. But I knew that day I wanted to get tested.” Nigel’s story was not simply about his concern for his father but nor was it simply about Nigel’s concerns about his own risk — each was in tension with the other. As Nigel moved from talking about the remembered emotion of the moment (“I felt like punching the wall”) to talking about a more rational response (“I pulled it together...nothing’s changed here”) he shifted his framing of the event from a recounting of the past to an imagined present. Nigel was recalling the moment of discovering his father’s test results but in his story this moment also became an opportunity to articulate and perhaps rehearse a potential response to the clinical disclosure of his own test results.

When Landis talked about the impact of learning about her father’s test results, she too described a predominantly rational rather than emotional response but, in contrast with Nigel, Landis emphasized the importance of past events in structuring her current matter-of-factness. Her father had not told her about his test results when he first received them because she had, at about the same time, lost her fiancé in a tragic accident. When her father decided that he would tell her about his test results the news came as a surprise (since she had not known that he was having predictive testing) and she was “very bummed out for him” but, given the magnitude of her recent loss, the implications of this news for her own risk status were “not a major concern.” Furthermore, although she had, since her aunt’s diagnosis, thought about her cousins’ risk of inheriting HD, her own risk was not something that she had ever dwelt on.

...it might be something that [my aunt’s] kids have to worry about but it’s certainly never occurred to me up until last week when Dad said “well, you
know, I think you’re going to want to know this/ I got the answer I wasn’t really interested in getting or wasn’t too pleased about getting, but I really thought I was home free.” And I wasn’t/ I mean it was just the two of us having dinner and I still was like “well, wow...” and then out came his binder and all the latest information and the fact that they’d identified the gene and blah blah blah blah blah. And it’s still/ You know/ and I’m a very emotional person. I’m not what you would call a matter of fact person, but I’ve remained completely matter of fact. And I’ve thought, you know/ spent a couple of days thinking about it. I can’t really come up with any reason to um/ I mean I can’t control any of it. So there’s not really a heck of a lot I can do except continue and I still am not/ It’s not a major concern. (emphasis added)

Landis was able to “continue” by focusing on the present and “living each day” to the fullest.

Making plans for the future doesn’t really work too well. My life changed in one second [with the accident] (...) so to worry about something down the road is just nonsense. It really is.

Worrying about her own chances of having inherited the gene for HD was not something that Landis could “generate any energy toward” and although she was “fascinated by the science side of it and very encouraged by it” she stressed that it was only important to her because it was important to her father. Paradoxically, Landis stated that she felt no real need to talk about HD.

That’s the thing, I’m not like, “No I don’t want to talk about/ I don’t want to deal with it." It’s like I’ve already dealt with it.

but then in the next breath she went on to say that,

...I mean if I was to be told well, you know, there’s a pretty good chance after being/ if I was diagnosed with it, there’s a pretty good chance it’s something that’s going to affect you in your 30’s and your 40’s, yea, then I’ll get interested in it. But I haven’t sort of been given any information along those lines at all/ and again I still, you know, I feel like I have a lot of time and I feel like things [scientific research on HD] are happening really quickly.

On one hand, Landis had “already dealt with it” yet on the other, she noted that she had not yet been really “interested in it.” Her story thus mirrored her practice of living in the moment. HD and other incontrovertible evidence of mortality was both behind and ahead of her in time.

Out of the Blue

Many of the stories of initial awareness described thus far illustrate how at risk individuals’ perceptions of HD are shaped by a combination of firsthand observation and/or knowledge of the way that Huntington Disease manifested in one or more family members. In contrast, those who had never knowingly been “exposed” to someone with HD, struggled to

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10 In this instance, Landis uses the word “diagnosed” to mean having the gene for HD rather than a clinical diagnosis of the actual onset of the disease. This usage is quite common and often the source of confusion.
make sense of their sudden awareness of the family history in a relative information vacuum. HD seemed to come *out of the blue* and though this type of transition may also have elements of solving the mystery and discovering the secret, it is unique in that it is marked by geographic and/or social distance from a family member diagnosed with and/or aware of, the hereditary implications of HD. There were two PT candidates (Helen and Rosalind) who described their initial awareness in this way.

At the time of our first interview, Helen was forty-seven, divorced and living in a small coastal community with her common-law partner. She had three adult sons from her first marriage. Neither of her parents had been diagnosed with HD and, given this family history, Helen was at 25% a priori risk. Helen had only known about the family history of HD for about eight months and thus her story centres on events which occurred comparatively recently.

Helen knew that her deceased Aunt Beatrice had an undiagnosed neurological illness but it was not until she received a late Christmas card and letter from her cousin Malcolm in England that she knew anything about the family history of HD. In the letter, Malcolm mentioned that his sister Katherine had been diagnosed with HD and that their mother’s (i.e., Beatrice) death certificate had been changed to reflect a posthumous diagnosis of HD. Helen did not know anything about HD at this point but she did recall that her now deceased mother had suspected that “something’s not right” with her sister Beatrice.

She had seen her sister once when she was ill. She was bedridden. I don’t know how far advanced you would have to be to be bedridden but I would consider that it had been a long time. My mother was going to England and she wrote to say “I’m coming to visit” and she got a letter back saying that her sister was ill. Until that point my mother had never been told that her sister was ill. And I believe that Beatrice was pretty well bedridden when my mother found out. So my mother’s association with Huntington’s is that 2 or 3 week period/ you know/ there was never any information coming from England. I know at a later date my mother had questioned my uncle about what it was but he didn’t know. I think my mother suspected that what Katherine had was hereditary from her mother [Beatrice] but there was no way of confirming it. Because my mother made such an effort to find out what it was I think she suspected a lot more than she told us. We didn’t pay much attention to it. I mean we were all, “well, what the hell?” I mean/...“these are all strangers.”

Never having known anyone with HD, Helen was unfamiliar with the characteristic symptoms and progression of the disease. When she received her cousin’s letter Helen therefore called her family doctor looking for information about HD. The receptionist was only able to tell
her that “if you are suspect [for HD] there should be genetic counselling for the kids.”

Concerned about the implications of this for herself and her adult sons, Helen delved into the outdated medical books in her local library, photocopied everything available (which was not much), and emerged with the overriding sense that most people with HD end up committing suicide. As Helen recalled, she was by this time “bouncing off the wall.” Refusing to see her own doctor until he had more information, Helen then contacted one of the genetic counsellors who worked with the predictive testing program at UBC. Helen spoke with the counsellor on the telephone and soon arranged an appointment. This was, she said, “probably the first sanity” she had since receiving the letter from her cousin.

Based on what she knew about the family history, Helen accepted that scientifically she had a one in four chance of developing HD. Remaining fairly certain that her mother had not shown any signs of HD before dying from cancer at age sixty–seven, Helen did, however, think of her risk as quite a bit less (about one in twenty was the figure she put on it). Describing why, Helen reiterated that it was because of her mother’s advancement beyond the typical age of onset and,

...because I’ve never been exposed to it and it’s still something that is really foreign to me as much as I know that it shouldn’t be. It’s something that they have over there that doesn’t come over here.

...I don’t know that side of the family. And my cousin who has it is probably my age, I think a year younger and she’s had it for several years. I can’t remember when. I’d say at least 10 years, maybe 15...I figure the fact that I am that much older is a good sign. We’ll give that another 2 or 3 percentage points, you know, all these things I add up in my head.

Helen’s method of accounting for her risk of having inherited HD reflects the fact that she had no established social relationship with her overseas cousins. She had never “been exposed” to HD and it was, therefore, “foreign” to her. She had lived her life with some awareness of the fact that her mother had suspected a family illness but it had never really occurred to her that this illness was something that should or would concern her in a much more immediate sense.

Like Helen, Rosalind also received “a real surprise” when she first learned that HD was in her family. There had been some dispute about the cause of her younger sister Patty’s protracted illness and, for a time, Rosalind believed that Patty had multiple sclerosis (MS). Patty was later diagnosed with HD, and, as Rosalind put it, “that was the first we ever knew.”
"Nobody in our family had had anything like that so it was a big surprise to us."

At the time of our first interview, Rosalind was fifty-seven; she was married and had two adult children. Patty had been diagnosed with HD for about seven years and was beginning to encounter pronounced speech and movement-related difficulties. Neither of Rosalind’s parents had ever been diagnosed with HD and the hereditary origin of the disease was unclear. Her mother was long since deceased and her father was in his late seventies and showing no signs of HD. Because of her sister’s diagnosis, Rosalind had originally been given a 50% a priori risk for the disease. Rosalind did, however, request linkage testing — about a year after her sister’s diagnosis — and the test results indicated that there was an 85% probability that she too had inherited the genetic mutation associated with HD.

When I asked Rosalind about her first awareness of HD, she recalled that she had actually known something about “Huntington’s Chorea” before she learned of her sister’s diagnosis.

I knew/ I knew of Huntington’s Chorea. I knew of chorea but I didn’t know very much about it other than/ chorea to me was you know, was spastic, uncontrollable action but I knew very little of it.

...I didn’t know a whole lot about it other than it was neurological. And uh/ I felt, figured that it would be something rather debilitating if it was neurological. I just figured that/ you know/ it’s not good but I didn’t know at the time when she was diagnosed that it was genetic.

Rosalind’s sister Patty lived in Alberta and thus Rosalind did not have easy access to a doctor who was knowledgeable about HD. Nor did she have a family doctor of her own at the time. She went to see her husband’s doctor and asked him what he could tell her about HD and he said “nothing...I really don’t know anything about it...give me a couple of days to read up.” She went back and he sent her to see a geneticist at the hospital who explained the characteristic pattern of inheritance. This made a big impact on Rosalind.

As soon as I knew that it was a genetic problem I figured well then we’d all better sit up straight and pay attention because uh/ we all could be in for some unpleasant news.

Rosalind was the eldest of five siblings and Patty was the youngest; in between were Ben, Alice (who had not been seen or heard from for very long time) and Dougie (who was adopted). All five siblings had health problems as children and, as Rosalind explained, her
father “never knew what it was to be without medical bills.” It was her mother’s illness, however, that Rosalind’s family seized upon in trying to make sense of the sudden appearance of HD.

Rosalind’s mother was diagnosed with Hodgkin’s Disease at age twenty-eight and, while pregnant with Rosalind’s sister Alice, she underwent radiation therapy. After Alice was born Rosalind’s mother also had surgery. Rosalind remembered that there was “a lot of sadness in the house” and that she had overheard someone say that her sister Alice had been burned by the radiation.

So uh/ this is why I wonder what ever became of my sister Alice. I really would be very surprised if she were still living today because she was the one that you know was really damaged I think by my mother’s illness at the time.

Rosalind’s mother “outlived” all of the doctor’s predictions and nearly ten years after her initial diagnosis of Hodgkin’s disease she became pregnant with Rosalind’s sister Patty. The pregnancy was totally unexpected because doctors had assumed that the massive radiation therapy would have caused permanent sterility. When Rosalind’s mother became pregnant with Patty, the doctor tried to obtain permission to do an abortion since Rosalind’s mother was “terminally ill” but when the hospital committee finally agreed to the abortion the pregnancy was too far advanced.

So as a result, Patty was born and Patty shouldn’t have happened. Just in that/ Not/ I mean/ My mother was on, you know, she was dying/ that wasn’t going to change. You can’t say that because Patty was born it shortened what time she had/ maybe it did/ maybe it didn’t/ but just the family didn’t need this, my father didn’t need this, Patty most of all didn’t need this. She used to fret and I can remember at 6 years old she said to the neighbour she was scared to come home from school in case mummy was dead when she got home.

When Patty was diagnosed with HD, Rosalind’s family re–evaluated the family history of various illnesses hoping to find some sort of explanation for the apparently unexplainable appearance of HD. It was Ben’s wife Sandra, however, who proposed that the de novo appearance of HD in the family was related to the radiation therapy that her mother–in–law had received while undergoing treatment for cancer. As Rosalind explained of her sister–in–law Sandra,

...she’s got it all figured out that the radiation scrambled something and Patty got/ it was passed on from my mum to Patty and that’s the end of the issue. Genetic damage’s been done over the years and then when my mother finally got
pregnant she passed on this damage to Patty. And as far as Sandra’s concerned that’s it.

Doctors had ruled out any possibility that the radiation could have caused a new mutation in germ-line cells and, thus it seemed to Rosalind that Sandra was simply looking for a means to deny that her husband Ben, and their two children, could also be at risk for HD. Moreover, Rosalind was troubled by the fact that Sandra seemed to blame Patty for somehow bringing HD into the family. This became an increasing source of tension between Rosalind and Sandra and ultimately undermined the closeness of the relationship.

For Helen and Rosalind both, the news that a family member had been diagnosed with HD came out of the blue. Neither had any cause to be suspicious that there was a family history of HD although both had known that a family member was unwell. Helen’s mother and father were both deceased and geographic and social distance intervened to preclude Helen from knowing much about her overseas cousin’s illness. Rosalind knew that her sister was unwell but a misdiagnosis of MS coupled with the absence of a similar illness in any other family members precluded Rosalind from having any suspicions that the disease was hereditary.

Helen and Rosalind were, at age forty-seven and fifty-seven, the two oldest PT candidates interviewed for this research and their stories of initial awareness demonstrate a number of significant parallels. Both Helen and Rosalind perceived the news of their family member’s diagnosis as troubling and both responded by actively seeking out information about HD from a family doctor. Neither doctor was knowledgeable about HD and thus both women initially experienced frustration in trying to obtain basic information about HD. Neither Helen nor Rosalind had known anyone with the disease but their first impressions were somewhat alarming: Helen read that most people with HD commit suicide while Rosalind noted that she assumed that because HD was neurological it must be bad news. Moreover, there were no straightforward explanations for the origin of the family history of HD for either woman. Helen and Rosalind had both lost their mothers to cancer. Moreover, neither could trace the family history through their father. Helen’s father was long since deceased from causes unrelated to HD and Rosalind’s father was well into his seventies and not showing any signs of HD.

With no confirmed diagnosis of HD in either parent, both Helen and Rosalind were
uncertain about the hereditary origins of HD. Helen thought of her own risk as less than 25% in part because HD was "foreign" to her and, while Rosalind did not accept her sister-in-law's theory about the effects of radiation, she did explain that in her family too, there was a tendency to view HD as if it were something external to the family. Finally, in reference to an earlier discussion of the relationship between duration of awareness and the desire for predictive testing, it is interesting to note that upon learning about the family history of HD both Helen and Rosalind immediately opted to become involved in predictive testing. At the time of the first interview, Rosalind had already had linkage testing and was awaiting the results of the direct test. Helen stated that the knowledge that there was such a test offered her the first "real sanity" she could remember since receiving her cousin's letter.

Discussion

The preceding stories illustrate four different types of transitions into initial awareness of HD. Colin's and Adam's stories exemplify the type of transition I refer to as solving the mystery — the mystery in this case being the undiagnosed illness and/or protracted odd behaviour of a family member who eventually turns out to have HD. Given the documented lack of medical records and low profile of HD within the medical community, this scenario has, until recently, been fairly common.

Carla's and Gabriella's stories are variations of discovering the secret but this sub-plot is nested within a larger narrative of solving the mystery. Like Colin and Adam, Carla and Gabriella knew for a protracted period of time that something was not right with a (respective) family member. Despite their efforts to find out what was wrong, both Carla and Gabriella were, however, unaware of their respective family histories of HD as family members concealed or withheld significant information.

Regina, Nigel and his sister Landis, told stories of learning by osmosis. This type of transition has little to do with mystery and, with respect to initial awareness, it has nothing to do with secrecy. As the name might suggest, this type of transition is the product of living in close geographic and/or social proximity to a family member diagnosed with and/or aware of, the hereditary implications of HD.
In contrast, Helen’s and Rosalind’s stories depict the upheaval created by a sudden, unanticipated transition into initial awareness. As such, HD seemed to come out of the blue. This type of transition may also have elements of solving the mystery and discovering the secret, but it is unique in that it is marked by geographic and/or social distance from a family member diagnosed with and/or aware of, the hereditary implications of HD.

Each of these four types of transitions into initial awareness may be summarized according to several dimensions. These include the timing of initial awareness (i.e., in childhood, adolescence or adulthood), the process through which initial awareness emerged (i.e., gradual or sudden), the nature of familial communication in the period leading up to awareness (i.e., relatively open discussion or active concealment of information), social and geographic proximity to a family member affected by or openly aware of HD, and access to various sources of information about HD (i.e., family, doctor or genetic counsellor, the Huntington Society of Canada or media). These dimensions of each type of transition into initial awareness are summarized in Table 8 (see next page).

Further, as suggested in Table 8, each type of transition has a particular social and historical significance. The most common type of transition described here is solving the mystery. Where there is no documented family history of HD and/or physicians have little or no awareness of HD, families (such as Colin’s or Adam’s) experience a protracted period in which diagnosis is, at best, uncertain. It is, however, probable that with improved diagnostic capabilities and increased physician awareness, this will become a less common experience. Families coping with a new diagnosis will, therefore, be more likely to have a documented family history of the disease and less likely to encounter the confusion and frustration occasioned by a misdiagnosis.

Many aspects of PT candidate’s stories about their initial awareness of the family history of HD are not, however, so easily abstracted and compared. In this respect, three specific themes have an overarching significance for several or, in some instances, all of these types of transitions into initial awareness. First, there is the tenuousness of beginnings — that is, there is a sense of ambiguity that characterizes many PT candidates’ attempts to define when “it”
<table>
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<tr>
<th><strong>type of transition</strong></th>
<th><strong>solving the mystery</strong></th>
<th><strong>discovering the secret</strong></th>
<th><strong>learning by osmosis</strong></th>
<th><strong>out of the blue</strong></th>
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<tr>
<td><strong>description</strong></td>
<td>no documented family history and protracted period where family member experiences mysterious illness and possible misdiagnosis</td>
<td>no documented family history and protracted period where family member experiences mysterious illness and possible misdiagnosis</td>
<td>documented family history of HD and observation of family member who has HD or interaction with family member highly aware of family history</td>
<td>may or may not be documented family history of HD but if so, interactant is initially completely unaware of it</td>
</tr>
<tr>
<td><strong>timing of awareness</strong></td>
<td>adolescence to adulthood</td>
<td>adulthood</td>
<td>childhood and adolescence</td>
<td>adolescence to adulthood</td>
</tr>
<tr>
<td><strong>process of awareness</strong></td>
<td>gradual</td>
<td>gradual or sudden</td>
<td>very gradual</td>
<td>sudden</td>
</tr>
<tr>
<td><strong>nature of family communication</strong></td>
<td>may be open (where there is genuine uncertainty about the diagnosis) or may involve staging of information or suspicion on part of interactant(s)</td>
<td>closed, involves active concealment of information and/or paternalism</td>
<td>open, may involve exposure to person with HD, staged, tactful or even excessive discussion of information about HD and family history</td>
<td>non-existent or closed then revelatory</td>
</tr>
<tr>
<td><strong>social and geographic proximity</strong></td>
<td>some proximity to HD but no one in family openly aware of family history</td>
<td>some proximity to HD but only select individual(s) aware of family history</td>
<td>close proximity to HD and/or family member aware of HD</td>
<td>distance from HD and/or family aware of HD</td>
</tr>
<tr>
<td><strong>sources of information about HD</strong></td>
<td>doctors as primary source but HSC, genetic counsellor and/or media may also be important</td>
<td>family member(s) (or doctor, as in case not discussed here)</td>
<td>family member with HD or aware of HD, media, doctors, HSC</td>
<td>family member or doctor</td>
</tr>
<tr>
<td><strong>historical and social significance</strong></td>
<td>may be decreasing because of documented family history, improved physician awareness and diagnostic abilities</td>
<td>non-disclosure continues to be a means of protecting self/others and social stigma (or perception of stigma) still prevalent</td>
<td>may be increasing because people with HD are diagnosed earlier and more now have a documented family history</td>
<td>affected by social and geographic proximity of family, variable need to provide medical history for adopted children, and unknown incidence of new mutations</td>
</tr>
<tr>
<td><strong>examples</strong></td>
<td>Colin, Adam</td>
<td>Carla, Gabriella</td>
<td>Regina, Nigel and Landis</td>
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started. "It" stands in for many things, among them the inkling that something is wrong with a family member, the distant recollection of someone uttering the words Huntington Disease, the explicit knowledge that a family member has been diagnosed and the self-conscious realization that HD is an hereditary disease which has a range of significant social and biological implications for self and related others. When "it" started is, therefore a complex question which may be interpreted in multiple ways. Maggie’s description of her emerging awareness was, in this respect, exemplary; her self-reflexive process of accounting for how and when she became aware of the family history highlighted the importance of avoiding the tendency to view awareness as if it were a binary, off and then on, phenomenon.

As I began this chapter by saying, memory is bound up with the salience of a particular event and salience, in turn, varies according to the perceived importance and emotionality of the event as well as the degree to which the event is recalled and repeated to oneself or others within the intervening stretch of time. Maggie, like many other study participants, had difficulty in defining what should count as a meaningful level of awareness of HD. She provided multiple answers to my question and though she knew for certain that she must have been about twenty-six when “it really sunk in” she, like Jason, was ultimately unsure about exactly “when it started.”

On one level, then, a concern with “when” is reflected in participants’ uncertainty about when their *individual* awareness of the family history of HD actually began. The title of this chapter — when ‘it’ started — does, however, allude to much more. As many of the stories presented above illustrate, the diagnosis of HD in one family member often compels a re-evaluation of all that has gone before. This re-evaluation is particularly profound for those families who do not have a documented history of HD or who have only recently acquired the knowledge that HD is in the family (that is, those whose experience fits the solving the mystery or *out of the blue* type of transitions).

For instance, when Adam learned that his father had been diagnosed with HD he began to question how much of his father’s behaviour had been due to HD and how much was “just Dad.” He could not quite fathom that the Dad he had always known might have had HD for quite some time and, in consequence, that life as he had known it was not life as he now knew it.
Adam's grandmother Ethel extended this re-evaluation back still further; she had observed several generations of the Danforth family and when her husband Ward began to demonstrate the same shuffling gait as his brother and father before him, they too became implicated in what she was beginning to understand as an hereditary illness. Finally, in Colin's and Helen's families, the diagnosis of HD also lead to the reinterpretation of illness in other, now deceased, family members. Colin no longer believed that his grandmother had died of "homesickness" and Helen learned that her aunt's mysterious illness must have been HD. As such, beginnings were inherently problematic for families who collectively sought to locate and trace the origins of the disease within their own family history.

A second overarching theme is the significance of familial communication. This theme emerges most clearly in the contrast between stories which exemplify the two types of transitions that I have called discovering the secret and learning by osmosis. In these two types of transitions, specific patterns and instances of communicative interaction are of paramount importance to understanding the sense of plot which structures the narrative. Tension builds around the issue of who talks to whom and under what circumstances and this is reflected in the episodic sequence of events within the narrative.

Where there is a secret, information is withheld from someone who has a specific and, from their point of view, legitimate interest in knowing about that information (Karpel, 1980). Tensions arise when the protagonist becomes suspicious but unable to find out exactly what is going on. These tensions culminate in what I have referred to as a pivotal disclosure. For instance, Carla did not explicitly state that she had long held suspicions that there was more to her mother's illness than her father would admit but, in concluding her story about her aunt's disclosure of the family history of HD, she stated that the disclosure had "cleared up" all of her questions and allowed her to get on with making some choices about her life. Likewise, Gabriella recalled that she had, for a number of years, wondered about whether or not her aunt had been diagnosed with HD. When her uncle first denied that this was the case and then, some years later, affirmed that he had withheld information about his wife's diagnosis, Gabriella's suspicions were confirmed.

In contrast, Regina's story exemplifies the opposite pole of what we might envision as a
continuum between secrecy and openness. There was no moment of revelation, no pivotal disclosure. In Regina’s words, there was “nothing we didn’t know about HD.” Talk about HD was, however, over–emphasized by Regina’s grandmother and, in consequence, Regina felt that talk sometimes substituted for meaningful action.

A third theme which runs throughout these stories links together the preceding themes of the tenuousness of beginnings and the significance of familial communication. This third theme is the “re–membering” of family. By this I mean the way in which families engage in an ongoing process of defining and redefining their sense of connectedness across time and space. As I wish to suggest, this process is intimately tied to existing patterns of awareness about the family history of HD — which side of the family it came from, who was diagnosed and/or likely to have been affected by HD and how HD tended to manifest itself within other family members. Re–membering therefore invokes memory but memory is, in this instance, collective rather than individualistic; it is the product of communicative and miscommunicative interaction, family myths as well as misunderstandings, secrecy as well as openness and, perhaps most significantly, the intersubjective experience of social and biological connectedness.

Helen’s and Rosalind’s stories provide explicit instances of the way in which re–membering occurs and, in turn, how the news of a diagnosis of HD may precipitate a strengthening or weakening of feelings of connectedness within families. Helen had no meaningful social ties with her aunt and cousins. They lived in Britain and she did not really know them. The news that her cousin had been diagnosed with HD did, however, precipitate a new sense of connectedness for Helen. It caused her to reconsider the meaning of her mother’s journey to visit her sister in Britain; it prompted her to think about and even quantify her own risk for HD in social as well as biological terms.

In contrast, Rosalind talked about how familial awareness of HD diminished the social connectedness within her family. When Rosalind’s sister Patty was diagnosed with HD, Rosalind’s sister–in–law Sandra insisted that the de novo appearance of HD must have been due to genetic damage sustained during Patty’s mother’s radiation therapy. Biological ties were, in Sandra’s view, irrelevant to understanding the family history of HD thus, it followed, her
husband and their two children were not really at risk for the disease. HD was external to Sandra’s definition of family and this, in turn, eroded Rosalind’s ability to engage in meaningful communicative interaction with her sister-in-law and, to some extent, her own brother.

There are numerous other examples which might be drawn upon to further develop these themes as they pertain to initial awareness of, and familial communication about, the family history of HD but, I wish to conclude by briefly considering the relevance of the issue of “when it started” for the argument of this thesis.

First, the issue of “when it started” constitutes an important but often overlooked point of departure for studies on the experience of predictive testing. Most studies of predictive testing impose a baseline against which to gauge the impact of the test on individual psychosocial well-being. Little attention has been devoted to understanding the perceived significance of events that long precede such baseline measures and, in consequence, those studies that do inquire about age of initial awareness or duration of awareness do so in a rather cursory fashion. Furthermore, there has been a tendency to homogenize the experiences of families at risk for HD — as if there were such a thing as “the HD family”. Within the current context, this tendency to homogenize obscures the way that new genetic knowledge and techniques are perhaps changing the experience of being at risk for HD. And, it does little to illuminate our understanding of when and how at risk individuals first learn about the family history of HD or, ultimately, whether the transition into initial awareness has any real bearing on at risk individuals’ desire to have predictive testing.¹¹

Second, the issue of “when it started” is interwoven with the issue of how at risk individuals and their families recall and narrate the events which are an integral part of their personal biographies. This process is never a solitary enterprise; remembering is, as Zerubavel (1996) argues, a social and intersubjective phenomenon that involves much more than the individual recall of events (although this is important). Remembering invokes a sense of the individual as s/he is situated within the context of a particular type of mnemonic community — in this case, the family. As I have suggested above, family is not, however, strictly bounded by

¹¹ The links between PT candidates’ patterns of initial awareness and processes of storying the decision to request the predictive test are discussed in Chapter VII.
chronological time and/or mortality. Stories told about long-deceased ancestors — their particular mannerisms, odd behaviour and/or other attributes — contribute to the sense that the family history of HD extends infinitely backward in time.

Third, the issue of "when it started" amply demonstrates that memory has a strong emotional as well as cognitive dimension. It has to do with the particular tone in which stories are told and the way in which these stories are nested in a web of felt memories and connections to other people and events. Maggie was able to recall her experiences of her father's illness precisely because his behaviour irritated and perplexed her and, in her recounting of these recollections, she invoked the same feelings that she described. In telling her story, she scratched the surface of the table and shuffled her feet beneath the table. Elsewhere, Gabriella talked about how her recent experience of going to church helped her to recall the feeling of safety that she used to experience when she and her sisters attended church; it was her refuge from "the bogeyman," and "the dark at the bottom of the stairs"; it allowed her to escape, however temporarily, the relentless impact of her mother's illness.

This chapter has focused primarily on the stories of individual PT candidates and other at risk family members but, even here, it is apparent that these stories are shaped and informed by the storied recollections of other family members. Some (such as Jason) were explicit in acknowledging that their recollections derived from stories that they had heard about events that they could not themselves recall while others (such as Regina) only tacitly acknowledged that this was the case. As such, there is considerable variation in the degree to which participants distinguish between memories of the events which comprise their own personal biography and the memories of events which are perhaps more properly understood as part of a collective, familial biography. The existential fusion of the two is significant here not because it in any way undermines the validity of participants' narrative accounts but because it exemplifies so completely the need to situate individuals' stories — as well as genetic information — within the context of an expanded appreciation for the importance of history and family.

12 The importance of emotion to memory emerges again in Chapter VIII when I consider how study participants recalled and narrated their experiences of learning and making intersubjective sense of the test results.
CHAPTER VII
STORIES IN DECISIONS

At certain moments the need to decide upon the story of our own lives becomes particularly pressing — when we choose a mate, for example, or embark upon a career. Decisions like that make sense, retrospectively, of the past and project a meaning onto the future, knit past and future together, and create, suspended between the two, the present. Questions we have all asked of ourselves such as, Why am I doing this? or even the more basic What am I doing? suggest the way in which living forces us to look for and forces us to find a design within the primal stew of data which is our daily experience. There is a kind of arranging and telling and choosing of detail — of narration, in short — which we must do so that one day will prepare for the next, one week prepare for the next week. (Rose, 1983:5-6)

There is a narrative structure to life as it is lived (Cruikshank, 1990). The stories we tell ourselves and others about what is happening in our lives are an essential part of our ability to understand what we are doing and, in turn, pursue what we perceive to be a meaningful course of action. In this sense, narrative is a fundamental aspect of agency (Fay, 1996).

As intentional agents, we all “live within stories that we must constantly tell ourselves as a condition for being able to perform...intentional acts ” (Fay, 1996:191). This fundamental aspect of narrative is, however, overlooked when narrative is understood as if it were simply the retrospective imposition of structure on a formless flow of events. Beginnings, middles and ends — or as Fay (1996) puts it “starting points, endings and climaxes” — may not be absolute but they do exist as “natural joints” between events and it is, in part, our ability to recognize and order such events in terms of their temporal orientation that enables us to “go on in life” (Giddens, 1986). For instance, when we decide to pursue a particular course of action we look toward the future in anticipation of the possible outcome(s) we believe it will bring about — that is, we have intentions. When we consider our reasons for making a particular decision, we look back in time to reflect on how we came to our present situation. The moment of acting is “precisely the coming together of the agent’s sense of his or her past history, present situation, and future possibilities” (Fay, 1996:192). The intentional agent is, therefore, constantly engaged in a process of knitting together past and future, beginning and end.
Purpose and Outline of Chapter

This chapter is about the stories that PT candidates and their families tell themselves and others about the experience of learning about and deciding to request predictive testing. It is about the narrative process of making decisions that make sense of what has already happened and what appears to lay ahead. It is about the confluence of reasons and intentions, the knitting together of past and future — that is, the stories that are in decisions.

This chapter is the second of three chapters which, taken together tell the story of predictive testing. In the previous chapter, I looked at how PT candidates recalled the process of becoming aware of the family history of HD. In this analysis, I emphasized the inchoate sense of beginning which characterized many study participants' responses to the question of "when it started." Further, I demonstrated how an awareness of the familial implications of HD is seldom (if ever) a simple matter of knowing or not knowing the scientific facts about HD. It is a social process which shapes, and is shaped by, a diverse array of familial interactions; some of these patterned interactions facilitate "active open awareness" (Timmermans, 1994) while others tend to deter it.

This chapter is the middle of the story as it was told to me by study participants at the time of the first (i.e., pre-results) interview. It focuses on how PT candidates and their families arrive at the decision to request the test and, moreover, what it means to be in the midst of predictive testing. This part of the story is, then, a sub-narrative; it is a story unto itself but it is also part of the larger story of predictive testing. The narrators do not, in the narrative now, know what the outcome of the predictive test will be nor, at the time of the pre-results interview, did I (or anyone at the clinic) have access to this information. Thus, while this sub-narrative has a beginning (e.g., learning about the test), middle (e.g., considering the test) and ending (e.g., the decision to proceed with testing), it also loops forward in time in order to incorporate one or more imagined outcomes and it loops backward in time to reflect upon and make sense of past events.

and a handful that examine at risk individuals' reluctance to undergo predictive testing (Bloch, et al., 1989; Evers–Kiebooms, et al., 1989; Quaid & Morris, 1993). Very few, however, mention, much less explore in any depth, the process of how PT candidates and their families arrive at the decision to request the test. Given that published clinical guidelines for offering the test (Benjamin, et al., 1994; Craufurd & Tyler, 1992; Fox, et al., 1989; World Federation of Neurology & International Huntington Association, 1990) consistently emphasize individual autonomy in decision–making and, further, that service providers stress the importance of ensuring that PT candidates have adequate social support during the process of predictive testing, this is a curious gap.

In what follows I am, therefore, especially concerned to understand how PT candidates, sometimes independently and sometimes in and through consultation with others, arrive at the decision to request predictive testing. The events and circumstances that figure most prominently in this process follow from having an awareness of the family history of HD. For PT candidates in particular, the story of learning about and deciding to request the test includes the following (not necessarily chronological) events: 1) learning about the existence of the predictive test, 2) discussing (or not discussing) the test with others and deciding (privately or with the input and involvement of others) to request the test, 3) visiting the clinic for pre–test counselling, 4) anticipating the outcome of the results session and its implications for self and others and, 5) telling (or not telling) other family members, friends and associates about the decision to have predictive testing. These events and circumstances may recur in a somewhat different configuration where PT candidates who have already participated in the linkage form of predictive testing return to the clinic in order to have the direct test.

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1 See the European Community Huntington's Disease Collaborative Study Group (1993) and Huggins et al (1990) for a series of case studies that examine, from a service provider’s perspective, the ethical dilemmas in clinical decision–making (e.g., coercion, third party requests for testing and/or the clinical disclosure or nondisclosure of information about test candidates’ results).

2 This is not an exhaustive list of all of the topics that were discussed in the pre–results interviews. The preceding chapter presents those parts of the pre–results interviews most salient to understanding the beginning of the story—that is, when the PT candidate first became aware of the family history of HD.

3 As noted in Chapter II, the two forms of predictive testing differ significantly in terms of the nature of the information that is provided and the level of familial participation that is required. Linkage testing provides probabilistic information whereas direct testing provides a definitive result. Further, linkage testing requires DNA samples from multiple family members whereas direct testing does not. In this research, I interviewed three PT candidates who had participated in linkage testing and received an informative result; two others initiated the process but subsequently withdrew due to difficulties with acquiring DNA samples from other family members.
In order to provide a sufficiently detailed and contextualized understanding of study participants’ experiences of learning about and deciding to request predictive testing, I present three pairs of imbricating stories. In the first section, I present the stories of the PT candidate Colin and his wife Emily; in the second, I present the stories of the PT candidate Helen and her common-law husband Duane and in the third, I present the stories of the PT candidate Regina and her close friend Denise. As these stories demonstrate, there are at least three ways that PT candidates story their experiences of deciding to request predictive testing.

Each type of story sheds a different light on the process and meaning of making this decision. In addition, each reveals that PT candidates and their families make many related decisions about if and when to tell other family, friends and associates about the decision to request the test. These instances of telling (or not telling) selected family, friends and others about the decision to have the test are both a part of the story and a formative influence on the story as it unfolds. Some PT candidates suggest that it is therapeutic to tell their story while others are reluctant to do. Further, many PT candidates and their family members are acutely aware that in telling other family members and friends about the decision, they are creating an additional burden of emotion work for themselves and others. Especially where others, such as young children, do not need to know, there is for most PT candidates a reluctance to share this information with them until the test results are known.

Though I am focusing almost exclusively on the stories of PT candidates who have decided to request the test — since this dissertation is, after all, about the experience of predictive testing — the choices and, indeed, the social construction of the decision to request the test are not as simple as they might initially seem. As Rothman (1986:51) suggests with respect to women’s decisions about the use of amniocentesis, what I am calling a “decision” (i.e., to request or reject the predictive test) “may be more or less consciously experienced as a decision, an opportunity for choice.”

Choices are always hedged in by constraints; we are not free to decide upon just any course of action nor are we ever positioned in such a way that we can see what the full range of choices might consist of. As mothers and daughters, fathers and sons, sisters, brothers, aunts, uncles, cousins, spouses, life partners and friends, we exist in and through our social and
familial ties with others. For those at risk individuals who are in the process of deciding whether or not to request predictive testing for HD, such social and familial ties loom large (McKellin, 1997). They limit just as they give meaning to the range of acceptable choices; they are constraints on action that are often experienced as moral phenomena. The unspoken commitment to provide care for an affected sibling, the heartfelt desire to relieve a father’s guilt about the possibility that he has passed on the gene, the strong conviction that it is not acceptable to have children without first ensuring that they will not be at risk for HD\(^4\) — all are forms of constraint that shape and narrow the perception of what constitutes an acceptable choice yet all are examples of the social relationships and moral commitments that bind family and friends together within the context of everyday life. How these and other aspects of choice are incorporated into the stories that PT candidates and their families tell about the experience of deciding to request the predictive test is the subject of this chapter.

Learning About and Deciding to Request Predictive Testing

The stories that are presented here are drawn from a number of different families: some have been introduced in the preceding chapter while others have not. Further, although I have foregrounded the stories of the PT candidates, the stories of various family members are not peripheral; they present a range of perspectives that contribute substantively to understanding the jointly constructed nature of PT candidates’ stories about the experience of learning about and deciding to request predictive testing. In many cases, the decision is constructed by PT candidates and their family members as if it were the PT candidate’s alone. As we shall see, however, there are few stories that uphold the ideal of autonomous decision–making if it is understood to mean rational, self–interested and self–directed behaviour. The decision to request the test occurs in a familial context; it is shaped by supportive and occasionally nonsupportive others and it is informed by the felt as well as explicitly articulated commitments and responsibilities that PT candidates and family members enact in and through their ongoing relationships with one another.

\(^4\) Downing (In progress) examines two distinct aspects of reproductive decision–making in families at risk for HD: 1) what it means to be a parent at risk for, or affected with, HD and, 2) what it means to pass on the risk for HD to offspring. Though it is no less significant in understanding reproductive decision–making, the former issue is often overlooked in studies which focus primarily on the risk of recurrence in the next generation.
In reading these stories it is important to recall that, at the time of the pre-results interview, most PT candidates (and their support persons) had attended at least two pre-test counselling sessions. In these sessions, the counsellor has a responsibility to provide full information about the implications of testing and “assess whether a decision is being unduly influenced by other persons or institutions” (Huggins, et al., 1990:5). During the last pre-test session, PT candidates also consider the possible outcomes of the test, imagining and perhaps talking about how each outcome might feel and what might happen next. As such, these sessions provide an opportunity for PT candidates to articulate and rehearse their stories. This has two significant implications for understanding the stories that PT candidates tell about the decision-making process.

First, the decision to request the test is, by all clinical standards, supposed to be an autonomous decision — that is, PT candidates ought never to feel coerced by family members, their physician, life insurance agency or any other person or agency. This is without doubt an important criterion for predictive testing but where it becomes translated into an apparent prescription for independent decision-making, it creates a source of tension; PT candidates who are acutely aware of the implications of the test for others as well as themselves may feel that they must articulate their “own” reasons for wanting the test and, further, that they must minimize or even disavow the involvement of other family members in the making of the decision. Likewise, family members may feel that they have no business “interfering” in what is supposed to be the PT candidate’s decision.

Second, the process of imagining and talking about the possible outcomes of the test is, within the clinical context, a key part of the process of preparing for the clinical disclosure of the test results; it is, however, also a significant aspect of the narrative process as it was invoked during the interviews; it is a time of deciding how to live the story as it is being told and how to tell the story as it is being lived. Though the interview context provides a very different opportunity to story the experience of deciding to request the test, the opportunity to rehearse this story in the clinical setting may shape PT candidates’ and their support persons’ understandings of what is “properly tellable” (Gubrium & Holstein, 1998).
Hearing the Difference: Three Stories in Decisions

In writing about the stories that study participants told, I (like my study participants) must perform a "kind of arranging and telling and choosing of details" that is, as Rose (1983) points out, an act of narration. The choosing of detail and, indeed, the selection and ordering of the stories that are presented in this chapter reflects the manner in which I arrived at a particular understanding of the stories that PT candidates and their families tell themselves and others about the process of learning about and deciding to request predictive testing.

Early on in the cycle of pre-results interviews, I realized that it was sometimes difficult to elicit from PT candidates the story of how they had arrived at the decision to request predictive testing. Moreover, very few PT candidates talked about actively involving other family members in the making of this decision; it was almost invariably constructed by PT candidates and family members alike as the PT candidate's decision. This was intriguing given that most PT candidates were acutely aware of the implications of the test for other members of the family as well as themselves. I wondered if I was I doing something wrong in terms of my interviewing technique and whether or not I was missing an important part of the story. There was, for a significant number of PT candidates, no single question or prompt that served to evoke a story about the weighing up of options, the to-ing and fro-ing, and wondering 'if' that I so expected to be the visible trace of making of such a significant decision. I initially thought that this silence and/or gap in the story might be due to the fact that most PT candidates had already worked through their options and made a commitment to proceed with predictive testing. The interview was, in consequence, an occasion for affirming and justifying rather than reliving the process of arriving at the decision; it was a time to express certainty and resolve rather than doubt or indecision.

Though there is no doubt in my mind that some PT candidates were particularly concerned to manage their "presentation of self" (Goffman, 1959) at the pre-test counselling sessions, I now understand that such concerns offered, at best, a partial explanation for what

5 As is the case throughout this dissertation, there are aspects of these stories that cannot be presented here without violating the limits of confidentiality and/or inadvertently revealing something that one family member said to me, in confidence, about another.

6 As mentioned in Chapter III, PT candidates and their support persons are aware that they must "negotiate"
was occurring during the interviews. It was not simply a matter of establishing a relationship of trust with study participants and/or getting PT candidates to talk about something that they would prefer not to. It was a matter of acknowledging that, for some PT candidates, there was no question about whether or not they wanted to proceed with predictive testing; it was a matter of when and/or how quickly it could be arranged. Their silence on the process of decision-making was, therefore, telling. The making of the decision was a non sequitur for some PT candidates because it presumed that there was a process that PT candidates had to consciously enter into, that they did not already know, with some certainty, that they wanted or needed to know whether or not they had inherited the genetic mutation associated with HD. The idea of not having to decide — of just having to know — was, however, foreign to me as I have never, in doing this research, been able to decide whether or not I would request predictive testing if I were myself at risk for HD. My understanding of what it means to come to such a decision thus predisposed me to look for some kinds of stories and not others.

I came to this recursive understanding of my own expectations, and hence my own inability to hear certain kinds of stories, through talking with Colin. Indeed, it was Colin’s framing of the contrast between his experience of “evolving toward” the decision to request the test and the experiences of those who knew right away that they wanted to request the test that allowed me to see there are many pathways to the predictive test; some entail the conscious experience of the decision as an opportunity to make a choice while others do not. In this sense, some stories of deciding to request the test were explicitly emplotted while others were, in a sense, “unplotted” (Good, 1994:147). These stories focused on the immediacy of having the test in the life of the PT candidate but the making of the decision was not featured as having a beginning or ending; it just was. These stories were, therefore, organized less as stories of making decisions than were those in which discussions of emergent outcomes were paralleled by a sense of beginning and some sort of progression toward the decision.

This basic difference in the selection and temporal unfolding of events is instructive; it

(Scheff, 1968) their participation in predictive testing with the genetic counsellor and other service providers. Most centrally, each PT candidate must be seen to be certain about the desire to know the test results, psychologically prepared to learn the test results and aware of the implications of the test for self and others before clinical disclosure of the results.
helps to account for several very different kinds of stories about deciding to request the test. First, there are stories, like Colin’s, which entail a gradual and/or incremental process of decision-making. In these stories of *evolving toward it* the narrator moves from being opposed to and/or somewhat ambivalent about the test (and/or making a decision) toward a sense of resolve; a feeling of readiness thus culminates in the decision to request the test. Second, there are stories like Helen’s in which the decision is an almost self-evident act. In these stories of *having to know*, the narrator is certain about the decision to request the test; there is no trace of ambivalence and little or no conscious reflection of the decision as a decision *per se*. Finally, there are stories, such as Regina’s, that reflect aspects of each of these patterns but do not fit easily into one or the other pattern. In these stories of *taking the decision*, the narrator does not initially perceive that there is a decision to be made and it is not until something changes that the narrator acknowledges that the test presents an opportunity for choice. Once the opportunity is apprehended as such, there is little or no difficulty in deciding to request the test.

These pathways to the decision are not necessarily mutually exclusive; nor do I wish to suggest that they exhaust all of the possible story lines for emplotting the decision to request the test. They do, however, begin to map out the diverse ways in which PT candidates and their families recall and narrate their experiences of arriving at the decision to request the predictive test. Although other plot structures could no doubt be formulated, I propose three basic types which are differentiated by the temporal unfolding of relevant events and the degree to which the decision is constructed as an opportunity for choice.

Further, each of the three plot structures is linked, albeit more explicitly in some cases than others, to the narrator’s orientation toward talking (or not talking) about the experience of being at risk for HD within the context of the interview and within the context of everyday communicative interactions with selected family and others. As we shall see in Colin’s story, the plot structure of *evolving toward it* is shaped by Colin’s view that you can’t talk about being at risk for HD with most people; it is too “scary”. Helen’s story of *having to know* is shaped by quite a different metacommunicative imperative. Helen says you “have to talk about it” in order to cope with it thus she feels compelled to share her story with others and put into words that which is often “hidden” in everyday social interactions. Regina’s story of *taking the*
decision is shaped by her communicative pragmatism: she does not want to dwell on being at risk for HD but if someone asks her a question about it, she feels an obligation to provide an honest answer. The three plot structures are, then, reflective of what was said (or not said) and how it was said (or not said).

Evolving Toward It

I begin by looking at Colin's story of evolving toward it, not because I view this as the prototype for a “good story” about making the decision but rather because it exemplifies the kind of story that I initially expected to hear. As mentioned above, this type of story about deciding to request the test entails the conscious recognition that there is a decision to be made; requesting the test is therefore not an immediate response nor is it any way self–evident. There is a period of weighing up the implications of the test for self and others and there is a tentativeness that is only gradually replaced by a feeling of being ready to proceed with the test.

Five of the sixteen PT candidates interviewed for this research narrated the experience of arriving at the decision to request predictive testing in this way. All shared a number of common experiences. Each had a long awareness of the family history of HD that began before moving away from home; all had subsequently lost a parent to the disease or had a parent in the mid to late–stages of HD and, all but one, had a sibling that had been diagnosed with HD within the preceding ten years. Four had some involvement with the activities of the Huntington Society of Canada, received regular newsletters and kept up to date with the latest research on HD. All had given some thought to the linkage form of testing and two (Gabriella and Marie) had initiated the process only to find that it would cause too much in the way of family upheaval to acquire the necessary DNA samples.7 Perhaps most significantly, however, four experienced sporadic or chronic feelings of anxiety about the possibility of already having symptoms of HD and a fifth shared a profound sense of anxiety about imminent onset; thus none was able to disavow the feeling that they had the gene for HD.

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7 Gabriella did not pursue the linkage test any further when she encountered difficulties with obtaining samples from some members of her family. After some initial resistance from various family members who lived elsewhere in the country, Marie arranged for the required samples but a mix–up occurred somewhere along the line and thus the wrong samples arrived at the lab. Not wanting to put her family through the whole process again, she decided to with draw from linkage testing.
In these narratives, the unsettling experience of having possible signs of onset is a recurrent theme that introduces a source of tension that competes with what may be a more rational idiom for expressing the desire to plan and, therefore, exert some control over the future. Colin and Gabriella explicitly acknowledged this anxiety and integrated it into the story of deciding to request the test; Carla and Rose were ambivalent about acknowledging that they might already be showing symptoms and, while Marie did not actually believe that she was showing any symptoms, she did find it overwhelming to talk about her lived experience of risk.

Colin was able to integrate these feelings of anxiety into his story of deciding to request the test by creating a narrative in which there were two parallel story lines. One was shaped by what he called his "innermost thoughts" and fears while the other was more public in tenor. As we shall see, the presence of this private story provides a coherent explanation for why Colin goes about making the decision to request predictive testing in the way that he does. This explanation fits with the story that Colin's wife Emily tells about her experience of learning about "his decision" and thus while the two stories are told from very different points of view, they add up to a "workable whole" (Rosenwald & Ochberg, 1992).

Colin and his wife Emily were living in a quiet farming community and on the morning that I met with them to do the interviews, their three daughters were at school. It was the day before Colin and Emily's seventeenth wedding anniversary and only six days before Colin was to receive his test results. Colin had postponed the results session (and interviews) because he wanted to make some adjustments to his life insurance policy. He was extremely busy with the management of a successful retail business and Emily was feeling the strain of balancing the demands of family with the change in schedule occasioned by her recent return to paid employment. They were also in the midst of doing major renovations on their

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8 Colin was forty-one years old at the time of this interview and, as the reader may recall, he was one of six PT candidates who talked about his initial awareness of HD in terms of solving the mystery. He first learned about being at risk for HD when his father was diagnosed with the disease in 1970. Colin was seventeen at the time and, although he understood that he was at 50% risk for the disease, it was not something that really worried him; he had a youthful sense of being "invincible".

9 Emily was thirty-nine years old at the time of this interview. She had recently returned to paid employment after spending a number of years being a full-time mother and homemaker. Her parents had separated the year before and her relationship with her mother was, in particular, strained.

10 Their daughters were eight, eleven and thirteen years of age.
Colin heard about the predictive test through the newsletters of the Huntington Society of Canada. This was shortly after the linkage form of testing became available in 1986 and, although Colin did not connect the two events in telling his story, it was also around the same time that his older brother Brad was diagnosed with HD. Soon after the diagnosis, Brad’s wife left him. This made an indelible impact on both Colin and Emily and, as such, the meaning of love and fidelity in the face of chronic illness and uncertainty was a recurrent theme in both interviews. Colin told me that he had “the best possible mate” and Emily stressed that she knew Colin would care for her until her “last days” if she ever had “a disabling illness...come upon” her. Both also talked about the positive impact of seeing a television documentary that featured the story of a young woman who married a man who had been diagnosed with HD. Colin thought this was both unusual and “admirable”. It stood in contrast to this brother’s experience and served as a reminder that his brother was struggling to cope with the disease on his own. As we shall see, this concern for his brother figured prominently in Colin’s account of how he arrived at the decision to request the predictive test. It was one of many affirmations that such decisions are seldom ever made purely in light of rational self-interest.

Early on in the interview with Colin I asked him to tell me about when he first heard about the test and, without pause, he told me the story of how his thinking had shifted, from his initial view that the test “was stupid” to his current view that it was an “instrument” that he could use in planning the future. In the following passage we see the movement in Colin’s thinking as he acknowledges that his ‘risk situation’ is both problematic and amenable to strategic intervention.

I heard about it [the linkage form of predictive testing] through the newsletters of the Huntington's Society and thought it was stupid. Like why in the world would you want to know? And I saw a show on CBC probably 5 or 6 years ago and they interviewed a few people that were at risk for Huntington's that were going for the disease/er the uh predictive testing as it was then and that sort of thing and I thought these people are nuts. Like why would you want to know? And there's just no way that I would entertain something like that. I figured you're not supposed to know what's going on in the future. You live by faith for one thing and why dwell on it?

But I was in my early 30's then so life has a different perspective. And then you get to 40 and you know, I think as part of being around 40 you start to look at life/anyways whether you got this hanging over your head or not. I still think
you start to look at where you've been and where you want to go and make any course adjustments that you have to do.

But all through this last five years anyways I thought that I should at least, for my children's sake get my blood banked in case I get bumped off by a bus or something, that they can/ if they ever in the future want to know what they're/ whether they're going to get it or not, there's some blood on hand and you know, they can get the predictive test done that way.

And that started me thinking around well uh/ At the same time the Huntington's cloud was gathering/ gathering on the horizon and it was getting closer and closer and started hovering over me and (telephone rings) you know, it was quite tough to, there's/ I didn't think about it heavily every day (telephone rings again) but more and more days you're thinking about your (telephone rings again) risk situation and how it may affect your future and having your own business you have to plan ahead and that sort of thing. And then your children. And you're thinking all these things and my thinking went from not wanting the test at all to at least getting my blood banked to thinking that maybe I would get the test so that I can plan my future/ use it as an instrument for planning my future.

The previous year had been especially difficult with the business and Colin had thought about selling. Emily was having a "hard time with her family" since her parents were splitting up and there were "too many things working on the family". Moving back to Ontario was appealing; it would allow them to "start fresh" and spend time with Colin's mother who was not in good health and his brother who was getting to the point where he needed some assistance with managing his affairs and perhaps moving into a "group home situation."

But in my mind I thought I can't do that right now. I need to know whether I'm going to get the disease or not because I wouldn't want to take my family, my wife especially from her family such as it was and her friends and support system that she may need should I get the disease. So I think that's sort of what brought me around to wanting to/ to know for sure. So I/ actually before that point I thought well maybe I would get the test but I wouldn't get the results, I would just have the test so if somebody else in the future needs to know they can find out for sure.

Colin was intrigued to analyze how his thinking had changed.

Over two years a complete 180 degree turn, and, uh/ in my thinking, which was really interesting to me. And thinking about it it's pretty interesting that a person can change/ can see all the events and you can see your thinking evolve through that relatively short period of time. And basically that's where we're at now. I mean if/ if we were/ if I was diagnosed that I have inherited the gene or whatever, for sure I wouldn't think of moving/ because I think it would be too disruptive.

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11 Colin is referring to the linkage test here and acknowledging that it is necessary to have DNA samples from both parents in order to proceed with the test. For Colin's children to be tested it would, however, also be necessary to have samples from an affected family member.
Here Colin shifts from talking about the implications of a diagnosis for himself and his wife — that is the “if we were” part of what he saying — to the realization that it is only he that can actually be diagnosed. With this realization, he reframes the imagined event and its consequences. This shift in framing is subtle but significant in light of what Colin has to say about Emily’s involvement in making the decision to request predictive testing. When I asked “was this a decision that you came to pretty much on your own or did you feel like this was a process that you and Emily went through, talking about this together?” Colin was emphatic.

No. It was something I came to on my own. I didn't talk to her about it. Big problem with this disease is you can't talk to anybody about it uh/ including your wife! I mean we talk about my Dad or my brother or whatever, but/ And she'd often ask me “do you think you've got it or you're going to get it?” or something. And I honestly never felt that/ I somehow felt that I wasn't going to get it. There's no scientific reason to feel that way but that's the way I felt. Until the last two years/ then it's sort of a grey area but if she'd ask I'd still say no.

And yea, she would see me do kind of/ do something (...) or, not think properly or whatever and every once in a while I think she would ask one of my friends if/ if he's seen any change in me, just double checking, you know. Which is fair enough. And no one admitted to seeing any difference in/ if I'm acting a little strange or not thinking right it's cause I'm stressed out. But it's easy to blame it on the disease cause then if I trip over something I'm sure I've got it. But you know it’s just not paying attention because you have too much on your mind.

Colin referred to the stress of balancing work and family life, especially as it provided a means of explaining his occasional clumsiness or bouts of “not thinking properly” and when I asked if he felt as if these manifestations of stress might be signs of HD he said that had been his “first thought” for years. It was, however, a “mind trap” to think that way and he did not want to attribute too much to it; most people get that way when they are over extended.

I think that’s the point where we're at. I wouldn't attribute it to the disease. I guess we're about to find out who is right. But even then/ if I had the gene and maybe I'm not even in the/ showing any signs of onset yet either so I'm still maybe fooling myself.

To us, you know, when we have discussed it/ as I say I haven't discussed my feelings with Emily/ getting back to what we were talking about/ it's because/ or too much/ because I don't want her to be concerned. And, you know, it's sort of my way of protecting her. I mean if she/ if I really shared my innermost thoughts over the last few years, I would tell her at times that I'm scared, you know? And I am scared when I trip once in a while. Sometimes it does concern me, but I'd never say that to her, because then/ Or to anybody else cause then they'd just start looking for it. And so you just dummy up.

For Colin, the decision to request the test was the product of a long process of introspection. He did not share his “innermost thoughts” with Emily because he did not want
her to be concerned. In addition, he did not want Emily to “start looking” for signs of onset when there might in fact be none. I said that he seemed to be very able to analyze his own behaviour, almost as if he was watching himself from a distance and he explained that it was because the HD “cloud” that was “hovering over him” had “consumed so much” of his thinking in the last few years. The idea that he could “lift the cloud one way or the other” was then, what eventually brought him to “getting the test”. Whether it was “good news or bad news”, Colin thought that it would be “nice to know”. As he asked somewhat rhetorically, “uncertainty is probably the worst thing in the world, wouldn’t you say?”

To briefly summarize, Colin described the following sequence of events in his story of arriving at the decision to request predictive testing. He went from 1) hearing about the test, living “by faith” and actively not wanting to know to, 2) considering why he ought not to rule out the idea of banking his DNA to, 3) deciding to bank his blood just in case something happened to him, to 4) weighing up the usefulness of actually knowing the results to, 5) deciding that he wanted to know and was ready to know whether or not he had inherited the genetic mutation associated with HD. The storied nature of his decision was explicit; each step in his thinking built on what had gone before and on what it was that he intended for the future. His reasons for requesting the test were fluid and changing rather than static. Before the direct test became available, he felt an obligation to provide information about his genetic status in case his daughters should need it. With the change from linkage testing to direct testing, this became less important and he shifted toward thinking that the information provided through predictive testing might be of assistance to him in making decisions that would have a major impact on his family. As a brother and son, he was concerned about the need to take a more active role in caregiving but as a husband and father, he wanted to ensure that the move he contemplated would not be to the detriment of his family’s well-being should it turn out that he had HD. Both experiences dovetail in Colin’s story and thus he speaks both as a potential caregiver (for his brother and mother) and as a potential recipient of his own family’s care.

Emily supported Colin’s decision to request the test even though she had not been directly involved in making the decision. As she said, “he’s made up his mind that he needs to do this and I back him up 100%”. She did, however find the counselling sessions “really
stressful.” It was “mentally exhausting” to go through all of the preparation for learning the results and she wondered if it was really necessary. She tried to take things “one day at a time” since this approach had helped about a year earlier when Colin was “just acting really weird”.

Recalling this period of time, Emily said

I was really, really scared cause I thought “what is going on with him?” Like he’d forget things and he just wasn’t himself, and I was really concerned. And so I spoke to my Dad and asked him/ and you know, “do you think/ like have you noticed anything? Have you noticed that Colin’s different?” No, he didn’t notice anything. He said it’s just your imagination cause you know that it’s time, like you’re just kinda looking for things. And I think maybe I was, you know.

Emily said that it was “upsetting” when Colin told her he wanted to bank his DNA: he had always insisted there was “no way” he would have the test.

Then all of a sudden last fall/ he turned forty last year, so he’s, you know, at the turning point. In the fall he said to me/ and it was upsetting for me but I didn’t admit it to him/ “I’ve decided to go for it.” And at that point he said “I’ve decided to go for it but I don’t want to know the results.” The reason for that was he wanted the girls to be able to/ if he was ever killed or died or whatever, they could go and know. He wouldn’t be leaving them with uncertain terms, like “Do I have it or not? Did Dad have it or not?” So he didn’t want to do that to them so he decided he would just bank it and leave it. But then when we got talking to {the genetic counsellor} he said, “I changed my mind, I want to know.”

I asked if this shift had come as a bit of a surprise and Emily said “yes.”

He decided what’s the difference if we know. Like he took it very well, very excited/ just hit me like a ton of bricks. Like, whoa. I was glad he postponed the date because I don’t think I could have handled it. I wasn’t ready. And my stomach’s in knots still and it’s because of Tuesday {results day}.

Emily spoke quickly. She did not elaborate on the “difference” that knowing might make to her but nor did her anxiety about the upcoming results session preclude her thinking that the test was, on balance, a good thing. It would enable Colin to make some long term plans with the business and it would allow them both to make decisions that would ensure that they lived life to the fullest. Emily would, however, have preferred to be “the one that’s being tested” because she did not want her husband “to have to look forward to something like that”.

Emily did not see herself as having a role in making the decision to request the test; she was Colin’s “support system” and she did not want to “interfere in his decision.” At the same, time, however, she found that not being able to talk about the decision was a source of anxiety.

He made the decision on his own. And I told him/ I said I’d support it 100%, you know, it’s available to you. I questioned him years before that because he was so against it and I said, “why?” And I often questioned him on that cause I/ But I/
Personally I don't feel that I have any right, even if I am his wife/ I have no right to make that decision for him or interfere in his decision because it's his life and you know, whatever he decided I just went 100% with it. If he decided he didn't want to know that was fine too because he's/ he's the one that has to live with it and he's the one that it's going to affect, you know? And we'll always be his support system. So, um, that/ that's always been his decision. We didn't really discuss it a whole lot.

And it's interesting to me and it sort of scares me a little bit, um, the waiting period that we've had, I've asked him periodically, So what are you thinking? Like "have you thought about your decision?" And he says, "What decision?" Like/ like he doesn't want to talk about it. I think partially because he doesn't want me to worry and be upset and all that, you know, because he knows what I'm like. But I would prefer that he did. For me it's more peace of mind if he talks than it is not to talk. But everybody's different.

Colin had not told anyone about his decision. He mentioned that Emily had told her father, her sister and a couple of friends and that he had wanted to tell his closest male friend but it was "too scary". He didn't like the idea of alarming his friend with his news but nor did he like to contemplate other people learning something about the future that they might not be ready to deal with. He thought that genetic testing was moving too fast.

You know you're going to be predisposed to cancer or predisposed to heart disease or whatever, do you really want to know that? The technology is coming faster than/ than the psychological side I'm sure, and that's the scary thing cause I/ you know there'd be a lot of people jumping off bridges when they find out these things. It sounds like a good idea maybe, right? and then But if you don't think it through (...) you don't know how you're going to handle it.

I asked Colin if his daughters understood that they were at risk for HD and whether he and Emily had told them about predictive testing. He paused for a few moments before saying, I don't know whether (...) No I don't think so. I mean it's/... I don't think they're old enough to. I mean they know Granddad had it, that Uncle Brad has it/ they know I can get it. I'm sure they know that, if they thought about it, they've been told that it can keep going on. But it's not something that we've sat there and tried to give them a big education on/ however depending on the results of the tests would be how we handled that. You know, handle it with them as they get older. But if we're/ we have/ They don't know that we're taking the test. They know that we're in some sort of research program, thinking it will help Uncle Brad and people like him. And that's the way we'll leave it, because if it comes back negative then there's nothing to talk about. And if it comes back that I have got it then, you know, they don't need to know right now anyways cause it doesn't mean I'm going to go out and start acting weird tomorrow.

Colin saw the test as a "really personal thing" and emphasized that he couldn't really talk to anyone about it. I asked him to say a bit more and he explained that his hesitancy to talk about being at risk for HD derived from his concern that people might see him differently.

I can't talk to/ certainly can't let anybody know at work. I can't even let them
know what my brother has by name, and things like that because all it takes is somebody to know, "okay it's a genetic thing, you know, the odds are 50/50 chance" and they put two and two together and then they start looking at you, or whatever.

Colin was "glad" that he had not talked about his thinking about the test since, as he put it, he had completely reversed himself.

Where was I two years ago? I thought the whole thing was stupid. Not really stupid but for me it's not something that I would ever want to participate in and two years later or three years later, whatever, probably a little longer than that, but/but you know, I'm 180 degrees so. But that happens a lot in life in lots of things. You reverse yourself and things look different to you. All of a sudden things that you've been (hammering sound starts) believing all your life go out the window. It just is quite amazing to watch. You always have to be careful and, you know, I'm glad I didn't tell the world that the whole thing was stupid and now I've reversed myself.

Colin did, however, stress that there was one place that he felt very comfortable in talking about being at risk and deciding to request predictive testing.

The Huntington's Society has an at risk group that meets once in awhile. It's really good to go there. I've been a couple of times to that particular group and it/there you can share your feelings because people can relate to it...And they can empathize with you because they've had the same feelings. And you can always talk about how you've come through different periods of time too.

You can't share that totally if you have a best friend that's not at risk, they could never totally understand what you're facing. And you may get a listening ear but you can't get the same sort of thoughts and advice back. So I guess there's some real camaraderie in/in talking to people that are ((in the same place as you)). And I think in any disease there's these sort of groups where you draw a lot of strength and the same with/for my wife there's groups/support groups for people that/family members of people that are affected, not so much at risk. It's important for them because they have particular set of frustrations or problems that other people wouldn't understand.

I affirmed that it sounded as if he had derived great benefit from talking with others who were at risk for HD and he added that he also thought that he had, perhaps, helped several people who were at risk for HD or newly "confronted with the disease".

And I think from what the facilitator said that my comments were/and how my thinking had evolved as I explained it to you. It really helped with that. And that feels good. It always feels good to help somebody else. And I felt good just being able to/being able to share my thoughts with people that have been there or are there, whatever, themselves. Cause, you know, even my wife can't totally understand the thoughts that I have. Nor do I particularly share them with her because it's uh/sometimes they get a little scary and why alarm somebody?

Colin had not told his mother or brother about having the test but it was his closet friend that seemed to trouble him most. He thought that he perhaps owed it to his friend to tell him —
"as a courtesy" — since this friend knew that Colin was at risk for HD. Moreover, Colin did
not want the fact that he had not told his friend to have other undesirable consequences.

Say I get it, I'd hate to be showing signs and have Emily phone him and say,
"Have you noticed anything wrong with/" and not have shared it with him. You
know, I would like to be able to/ And I would like to tell him about this process
with the testing and all that stuff but it just hasn't worked out. The time/ Yea. As.
I said earlier in the conversation I can't just/ It's not just something you phone up
and say, you have/ “Did you know that I was getting my future told/ foretold?”

Colin had just spent a whole day with his friend so this not telling was very much on his mind.

By the time they “had caught up on all the normal things” there was no time left to have “that
kind” of conversation. Moreover, Colin worried about the impact that his news might have.

You have to do it in the right setting because you can blow somebody away
with/..if they're not ready, and really put a burden on them, you know, if you
don't treat it right. And who wants to do that to somebody?

Emily took a different stance toward telling others about the test. She told her Dad and
he was “really concerned”; he phoned them almost every night, just to check in that things were
okay. Emily had not, however, told her mother; she did not want her mother's support “because
it wouldn't be positive”. She was also disappointed in her sister and her husband because they
knew about the original date for the results session and there was “no follow up call” to see
how things went. This was, she put it, “an indication that I can't really rely on them either.”

There was “a sort of a testing period” where it became clear to Emily that some people
could be relied upon to be supportive while others could not. Aside from her father, Emily
mentioned two people who were supportive.

I have a girlfriend that I've had for years. She/ I work with her. She's been
actually in really bad shape over this. And in fact she wants me to phone her
today after you leave. Like she's just really/ I'm really worried about her because
she's been/ Well she's so concerned about Colin. And she's/ she's somebody that
will be there in a minute for me, you know. And my neighbor across the street
has phoned/ every time we've gone to UBC, has phoned when we got in the door
to make sure everything's okay and stuff.

So we have a specific few... I sort of thinned it out, you know/ there'll be my
neighbor across the street and my girlfriend and my Dad. Those will be the
three that we will sort of lean on. I think part of it is a lot of people don't know
what to say. Like they just don't know what to say. And so I/ I don't want to put
them in the position where I'm making them feel uncomfortable either so I'm
just thinning those people out. And if they ask we'll share it with them and if
they don't we'll just leave it alone.

I asked Emily about their plans for telling their daughters about Colin's test results and she
outlined two scenarios.

If he doesn't carry the gene we're telling them for sure. On our family night we'll tell them for sure it's over, there's nothing to worry about. Cause they/ they know that he's at risk, we've told them that. And they know that he's at risk. They know that they're at risk if he's at risk. They understand all that and they're/ they're concerned about their cousins because they know that Uncle Brad/ Like we've been with him and they've helped him and cared for him and/ Like they're very very good that way. It doesn't scare them and uh/ Their concern is {his two sons} which is interesting to me. Like they realize that those kids are at risk. And our oldest one actually asked Colin to/ “Have you talked to Jamie and Chris cause maybe somebody should talk to them?” Like they understand all that. I'm not quite sure that we would tell them if Colin has the gene. I think we'd probably wait. Till a few years down the road. But if he doesn't have it we'll definitely tell them.

When I asked Emily if she had a sense of what the results would be, she said that she was feeling as if “he’s got it” but hoping that he did not.

You know why I think that is? I think it's because, um (..) the meetings {counselling sessions} are all negative. The meetings are all as if he has it and I think that that's why I'm saying I'm not sure these meetings are good for the spouse, you know/ for the other person, cause the meetings are all as if he has it. And the last meeting we had I actually was kind of/ I was not myself because I was watching her {the genetic counsellor} (...) to see her emotions, how she was reacting/ to get some signals. And I got the sense that she wasn't positive that day and so I outright asked her, “Do you know?” Like, “do you know if he has it or not?” And she said, “No. I don't know until/” And it was just a total relief to me. But it's all this negative/ Like to me it's negative anyway. Maybe it isn't but it just seems negative to me.

You know what it is? And there's a lot of psychological stuff happening with me right now. You know what I think it is? Cause I thought to myself too, why am I thinking this? Like why am I/ Cause my girlfriend’s really bugged by it. She said, “1/ What am I going to do with you? Think positive.” And I said, “Well what do I do then if he has it?” You know, like/ And she/ and she said, “I really don't think Colin has it.” And just to hear that, like to me when she says that it's like, “thank you!” you know, like it's something positive. Somebody said something positive cause there hasn't been any of that. And so/ I don't even recall them asking us if he doesn't have it, how would you feel? I don't even recall them/ I'm sure they must have asked us that question but I don't recall it.

But anyway I asked myself “why am I thinking that way?” And I think it's because of all the stuff in my past, my own personal past. I think I'm almost feeling like I deserve this. I deserve this burden that I'm going to have ahead of me. Like my Mum's done such a number on me that I think that's part of it. I'm positive that's part of it... Always putting me down. And always telling me what to do...And even/ until she left here she had her thumb on me. And she still does. And, um/ it's been a real burden. And I almost feel like/ if Colin has the gene, all of the grief that we're going to go through twenty years from now is going to be because I deserve it. You know? Like, it's/ that's the attitude I have. And it's starting to affect me. And I think what happens is as you get older, things come to the surface cause I/ I'm thinking why is this all bothering me now? Like it's this roller coaster thing going on.

Emily mentioned that she had thought about going for counselling but that she had
decided to wait until after they had the test results. She did not really like to share her private feelings and she was only telling me so that people would understand what is like for the spouse. She also reiterated that she and Colin were “blessed” with some good friends and that they both felt that there was enormous hope given all of the recent developments in research.

Colin and Emily were both looking forward to having the results session over with. Colin was “quite up to know” but he reserved the right to change his mind. His most salient piece of advice to others who might be considering the test was, therefore, simple.

I think you really have to do it when you’re ready. If you don’t, I think you’ll have probably a resentment that you had/went through the process if you were pushed or resentment that you know, you’re not quite prepared for it.

Colin was not sure what the test would ultimately mean but concluded that it was not possible to know in advance: as he said, “we’ve done all the thinking we need to do and we’re committed so let’s just do it and then, you know, figure out afterwards/what it means to us.”

Emily was more pragmatic; the waiting had “taken a lot of the oomph out of everything”; “irregardless of what the decision is/it just won’t matter/like it will just be good to have Tuesday (results day) finished.” Both told stories of how they felt that they had been prepared by the genetic counselling sessions to hear “the worst”, but as the results session approached it was the waiting rather than the anticipated outcome that generated the most anxiety. There was ultimately no way of knowing, in advance, what results day or, for the matter, the day after results day, would bring. As Colin said,

So (sigh) (...) I don’t know. Wednesday morning will be more interesting than Tuesday morning. I haven’t really thought of that, what Wednesday morning will be like. Every time I think about it, I don’t want to think about it (...) Which probably is fair enough.

Cause on a good day I figure knowing for sure is better than not knowing. But uh/you know, that’s easy to say when you don’t know for sure...when you know for sure well, as I say, then you start looking at the positive aspects. Only thing that I really (...) find interesting in this whole experience for me is that I seem to/in my thinking, seemed to think in more/uh/like I have it than I don’t.

I asked if he thought that he was thinking this way to protect himself from disappointment.

I’m hoping that’s what it is. Or just trying to be realistic because I certainly can adjust real easy if I don’t have it. I mean I can adjust to that news so I want to make sure/I think it’s the way I am. I’m just trying to adjust for that reality if that’s what it’s going to be so that I don’t jump off a bridge and all that kind of stuff. I don’t want/So hopefully that’s all it is. Not sort of intuition or something.
Both Colin’s and Emily’s stories were open-ended in that neither was committed to incorporating a particular outcome of the test into their story. Colin explained why he felt that he probably “had it” and Emily felt that they had been prepared to hear the “worst” but each sustained the possibility that it might not be so. Moreover, the open-endedness of Colin’s story in particular alluded to the limitations of rational thought as a means for deciding what the test means. As Colin said, they had done “all the thinking” they needed to do, “let’s just do it and then...figure it out afterwards, what it means to us.” There were, then, obvious constraints on the kind of closure that any story could bring at this point in time. Emily hoped that they would have a double celebration of their wedding anniversary and the results and yet she was also thinking ahead to a time when Colin might have HD and envisaging how, in contrast with his brother, Colin’s “calm and passive” nature might mean that the disease would take effect more slowly. Moreover, as both Colin and Emily noted, it was not like getting cancer or being hit by a bus. They had a time frame and, whatever the outcome, they could live “one day at a time”.

As we shall see in Helen’s story, this juxtaposition of cancer with HD has a very different resonance and meaning when HD is a relatively unknown disease. In so far as cancer represents the worst case scenario for Colin and Emily, it is in Helen’s story a part of her life experience that has equipped her with the strength and insight that she needs in order to cope with being at risk for HD.

**Having to Know**

Of all of the PT candidates that I interviewed, Helen was one of, if not the most, certain about wanting to proceed with predictive testing. In contrast with Colin, Helen wanted the test before she even knew for certain that it existed. My challenge in hearing Helen’s story was, then, to understand the biographical sources of her certainty about knowing and, moreover, to locate how it was that her story of *having to know* made sense within the context of an *out of the blue* transition into initial awareness of the family history of HD.12

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12 As the reader may recall, Helen was one of two PT candidates that experienced an out of the blue transition into initial awareness. Both of Helen’s parents were deceased and she had only recently learned of the family history of HD. This revelation came when her overseas cousin Malcolm sent her a late Christmas card and letter which contained the news that his sister Katherine had been diagnosed with HD and that their mother’s death certificate had been changed to reflect a posthumous diagnosis of HD. At age forty-seven, Helen was therefore slightly past the typical age of onset for HD before learning that she was at 25% apriori risk for HD.
As mentioned above, the plot structure that I refer to here as *having to know* does not entail the conscious recognition that there is a decision to be made; requesting the test is an almost immediate response to learning about being at risk for HD. It is the only thing to do and, in this sense, the decision is not a decision per se; it is a self-evident act. There is no period of weighing up the implications of predictive testing for self and others and there is little in the way of ambivalence about being ready to proceed with the test.

Five of the sixteen PT candidates interviewed for this research described the experience of arriving at the decision to request predictive testing in this way. All had in common the experience of learning relatively late in life that there was a family history of HD and/or realizing fairly late in life that they were at greater risk for HD than initially thought. Albert, Rosalind and Heather all requested the linkage form of testing very soon after learning that a family member with a mysterious illness had been diagnosed with HD\(^\text{13}\) while Helen and Nigel did not, respectively, know or fully understand about being at risk for HD at the time that the linkage test was being offered. Heather was the only one that had ever lived with a family member who had HD. Perhaps most significantly, however, none experienced periods of protracted anxiety about showing signs of onset and all five were highly motivated to speak with other family members about the decision to request predictive testing; each wanted in their own way to set an example for others in the family to follow.

In these narratives of *having to know*, there is a strong sense of agency that is attached to the act of having the test. The test is an accomplishment which the narrator brings about; it is not a matter of passive acquiescence to a doctor’s referral. Although the decision to request the test appears to be self-evident, the narrator is empowered by the ability to acquire information.

It is also salient to note that, with one exception, each of the PT candidates who told this type of story about the decision was overwhelmingly concerned with the implications of the test for other family members. For Helen and Rosalind, this concern had to do with a perceived responsibility to provide the next generation with information about their risk status. In

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\(^{13}\) Albert and Heather learned through the linkage test that they had a decreased risk for HD while Rosalind learned that she was at increased risk. As such, Albert and Heather were able to approach the direct test with somewhat less anxiety than they might otherwise have experienced. The direct test was, then, a matter of tidying up loose ends. This was not the case for Rosalind.
Heather’s case, the test was significant for reproductive and career-related decision-making. Nigel, on the other hand, had no children but wanted to have the test in the hope that he would “break the chain” and relieve his father of any guilt he might feel about the possibility that he had passed on the gene to his children. Albert was the one exception to this pattern in that he was more concerned with being able to clarify his own risk status than he was with its implications for other family members. He, like Nigel, did not have children.

Helen was at 25% rather than 50% a priori risk for HD but she was one of the two PT candidates who felt there was a moral imperative to advise her children of their risk status. She had three adult sons and, although none was married, they were at an age where they might soon contemplate having a family. Moreover, her older sister Norma also had a son who was recently married and Helen believed that she should also look into having the predictive test.

As I noted in my fieldnotes, I had the sense during the interview that I was helping Helen “to find words”. The interview was, therefore, about her struggle to articulate her thoughts. Helen’s husband Duane, on the other hand, was “not a big talker”; as he said, he was prone to “running out of narrative.” He was a “holder-inner” when it came to talking about his thoughts and feelings and thus I felt, at times, as if I were extracting his story from him. What Duane did not really want to talk about was what Helen most wanted to talk about and their stories about their acts of communication with each other mirrored this particular dynamic.

I met with Helen and her common-law husband Duane14 in their home in a small and somewhat remote coastal community. It was a hot summer evening only two days before Helen was scheduled to receive her test results. Helen prefaced her story of requesting the predictive test with a lengthy story about how it was that she had become more outspoken about her thoughts and feelings than she used to be. Much of this story had to do with the experience of being a caregiver for her mother when she was very ill and dying from cancer.15

I had no idea what to do for her. I knew that she had a suitcase full of medication that she brought home from every doctor she saw, and every time she saw her doctors she would tell them that she didn't have any and they would

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14 Duane was in his mid forties at the time of this interview. He worked seasonally in the fishing industry. He and Helen had been together for nearly ten years and though he did not have any of his own biological children, he had a close relationship with Helen’s three sons.

15 At the time of this interview, Helen’s mother had been deceased for five years.
give her more. And I knew I had to do something but nobody would talk to me. And the doctor at the Cancer Clinic? I sat on his doorstep for two days from the minute they opened to the minute they closed and he wouldn’t give me the time of day.

I was devastated. And when I came back here I phoned her doctor and her doctor said, “Well you and your Mum can come in. I won’t see you but you come in with your Mum.” I would not say anything in front of my Mum because my Mum would never have spoken to me again. I did not cross my mother. That was the way things were. So after that appointment I phoned her doctor and I said “I want to talk to you, can I get an appointment?” And she said, “you’ll have to make one at the desk like everybody else.” So that was fine (extreme sarcasm). This was exactly ten days after my mother died.

Helen had “an awful lot of anger” associated with this experience and she could not deal with her mother’s death for some time “because the anger had to be released first.” She went to see her own doctor and he encouraged her to write a letter to all the people that were hurtful to her. Once she had done so, she was able to deal with her mother’s death. Moreover, she decided that it was time she started telling people what she really thought. Duane said she was “getting bitchy” and friends told her that she had changed but as she put it,

I just decided I didn’t ever want to go through what I had been through before and I was old enough to speak my mind now that my mother couldn’t slap my fingers for it. I have never really talked about how I felt until the last maybe three or four years. But I’m really trying to do it now. Not so well sometimes but I am trying.

I agreed with Helen that it is not an easy process.

No, it’s not. I find that my tactic more is to say it and run. “God you’re horrible, I can’t stand you when you’re like this. I’m going to go home” you know (laughs).

Given that she lived quite a distance from Vancouver, Helen elected to receive her test results through her local doctor. She did, however, feel this task would be difficult for him.

I found myself feeling very sorry for him with my coming appointment this week. As I left my last appointment he asked me how the results will be coming from Vancouver and whatever and I told him basically how I thought it was and I talked to [the genetic counsellor] later and she has told me that what I told him was correct and he says, “You know, this is the first time I’ve ever had to do anything like this.” He says, “I don’t know how it’s going to be,” he says, “but you’re going to walk in and take one look at me, if I’m grinning from ear to ear you know it’s good. If I won’t look at you you know it’s bad.” He said, “You won’t have to wait, you’ll know.” And I actually found myself feeling very very badly, you know, “why did I do this to him? This is stupid.”

__16__ Under the rural protocol, the genetics counsellor at the clinic in Vancouver works with a health care professional in the test candidate’s home community. At least one pre-test counselling session is done at the clinic in Vancouver before blood is drawn and sent to the lab. Subsequent counselling and delivery of test results is provided by the local health care professional.
But it's just how I feel about him/ I really like him and I care that I'm giving him a really dirty job because it isn't pleasant. I'm sure that it's not going to be pleasant if the results aren't good (.) which I'm sure they will be.

Helen felt that she could “say anything to him” and that even if she was “really nasty” he would not take offense. He would know that it was just “nerves”.

I was curious about the speed with which a whole sequence of events had unfolded for Helen. Only eight months beforehand, she had known nothing about the family history of HD. She had never met anyone with HD and, at the time of the interview, she was only two days way from learning her predictive test results. Recalling how this whole period of time felt like a “blur”, Helen told me the story of how she took charge of her situation and managed to find out about the predictive test. Her local doctor did not know anything about HD and Helen “kind of bounced off the wall” for a few days when she first learned from her cousin that she could be at risk. She was adamant that she did not want to see her own doctor until she knew what she wanted to ask him. As she put it, “if I go in not knowing anything then I’m dead.” The receptionist tried to insist that Helen come in for an appointment but Helen reiterated, “Until you get information, don’t call me, just get what you can for me.” Helen spoke to the receptionist again a day later and learned that there was research on HD being done at UBC.

As soon as she started calling down there I started calling her. “What did you find out today?” “Why not?” So she said, “Listen, we keep leaving messages and we don’t get an answer.” I said, “To hell with this give me the number, I’ll find out.” And having worked in an office long enough I immediately asked for his secretary. I don’t know if I got his secretary, I have no idea who it was that I got and she said, “Well he’s out of town.” And I said, “Well what do I do? Do something. Give me somebody.” And she gave me two names, one of which was [a counsellor] and I phoned her and she made an appointment for me right away and that was probably the first sanity I had. I was going pretty crazy by this time.

Having booked an appointment with the genetic counsellor, Helen considered what she ought to do about telling her sister Norma about the family history of HD. Norma was coming out of a period of depression and was planning to remarry within a few weeks. Helen “knew” that if she told Norma about being at risk for HD, Norma would cancel the wedding. Helen decided to talk to Norma’s fiancé and tell him “there was some genetic thing wrong with the family.” She “made him promise” he would not tell Norma because she didn’t want to “go behind her back” but nor did she want Norma to know at this time.
And he said that he didn’t want to know anymore, it didn’t make any difference what was in the family, he wanted to go through with the wedding regardless. Don’t know what I would have done if he’d backed out.

I agreed that this would have been very difficult and Helen went on to explain that she had always been protective of her sister; she felt it was her responsibility to look out for Norma. She looked after her mother when she was ill and she looked after Norma when her former husband died quite suddenly a number of years before. Indeed, this sense of responsibility seemed to be what entitled Helen to decide whether or not she would tell Norma about the family history.

So I’ve always kind of felt that it’s up to me to look after everybody and make sure that they’re fine which is what gave me the right to make up my mind whether or not I was going to tell her...I couldn’t see any sense in telling her but I felt very strongly that Ken had to know. Why I made the decision I don’t know. I don’t really remember it much except that I know that I did it. Just had to.

Once Helen had booked the counselling session, she felt she “had to tell” Norma. Helen wanted Norma to attend the session with her since she too had an adult son that ought to know about his risk for HD. Norma was “not particularly impressed” that Helen hadn’t told her to begin with but she did attend the counselling session with Helen. This session had a big impact on Helen. It wasn’t until then that she felt that she “really knew anything” about HD. Finding out that there was an accurate predictive test was, however, the single most important thing and “I have to know” was her immediate response.

The idea that there was actually, what is it 97% of the time a 100% positive or negative, you know, that I could get an answer was probably my salvation. That was the big thing for me, just to find out.

Helen did not think of the test as if it were for just for herself. “I mean I’d want it for myself too but I have to have it for my kids.” I wondered about why she was so certain and asked “when you first learned about the fact that there was a test, did you know right away you wanted to take the test?” She said “I wanted to take the test long before I even knew there was one.” There was “no decision”. It was something that she just knew. As she put it,

I have twice before dealt with the fact that I could die/when I had cancer at thirty and I was in a quite severe car accident that everybody kept saying to me “you should have died, you should have died.” What a hell of a thing to say. You know, “I can’t believe that you’ve come out of this and you’re only smashed up.” So I have dealt with the idea that life doesn’t go on forever. So this isn’t really making a huge psychological difference other than it’s renewing things that I already knew.
Once the genetic counsellor confirmed the fact that there was a test, Helen knew exactly what she wanted.

She said think about if you want it done. I said, “Don't have to, this is it, I'm having it. How soon can I be there?” And she said, “well it's several months away.” I said, “Several months! I can't wait that long.”

I asked if she was surprised that the test involved so much counselling and that it had to take place over an extended period of time and she said,

Probably, but I had no preconceived notions. I was completely a blank slate. I had no idea what to expect. I was floored as I said that everybody didn't automatically get tested. That to me was unthinkable. So because in my head everybody automatically got tested as soon as the tests were available. It just never occurred to me that there would be anything else there. It didn't really matter what there was, I would do whatever you told me to because I was going to get this. If I had to tell you I had green hair and purple eyes I would tell you. I mean anything you wanted to know I would tell you just to get it done. I guess maybe I was a little surprised but/ Once I found out there was testing and that was the big thing I was so happy that there was testing that nothing else really mattered.

Helen did not mention anything about her husband Duane's involvement in this decision and when I asked whether she had talked with him she said, “no” but that she would have liked to. He wasn't “as supportive as he should have been” and so she didn’t tell him much until he inquired about her trip to Vancouver for genetic counselling.

We talked about it somewhat that night. He said that he feels that he had known my Mum/ he never met my Dad but he knew my Mum and we were together when my Mum had cancer/ and he said “I've known your Mum and I know that she didn't have any symptoms,” so he said, “as far as I'm concerned there's nothing to worry about. Why get upset with it if there's nothing to worry about?” Which may have seemed logical to him but it wasn't to me. And we haven't talked about it a lot. I think basically for the reason he is absolutely 100% certain that things will be fine and there's not much sense discussing it. Which is the way he is with all things, it's not just this. I mean that is typical Duane.

For Helen, the decision to request the test was a self-evident act; she had to have the test for the sake of her sons and, moreover, she assumed that everyone got tested anyhow.

There was no real choice but nor was there any hint of ambivalence, on Helen's part, about knowing the results of her test.

To briefly summarize, Helen described her process of arriving at the decision to request predictive testing in the following way. She went from 1) learning that she was at risk for HD and assuming that there must be a test to, 2) making contact with the clinic and confirming that there was a test to, 3) learning about HD and seeing that there was a parallel in her prior life
experiences with cancer and a serious car accident, to 4) formally requesting the test and
getting herself on the waiting list. The storied nature of Helen’s decision was, therefore, quite
different than Colin’s. Helen began with a certainty that she had to know if she had the gene
and the assumption that there must be a way of finding out. Her reasons for requesting the test
were unchanging. She felt a strong moral commitment to ensuring that her sons were informed
about their risk before they decided to marry and have children. She also felt responsible for
her sister Norma and her son; she hoped that Norma would also consider having the test and
that she would provide her son with information about his risk. As a mother and a sister, Helen
was therefore concerned to set an example for others. This was, however, no less important
than her desire to relieve the anxiety she felt about her own chances of developing HD.

Helen’s husband Duane supported her decision to proceed with predictive testing
although he was somewhat peripheral to the process. He had been “very” reassured to learn that
it was unlikely, given that Helen’s mother’s had died at age sixty–seven without showing any
signs of HD, that Helen would ever have HD.

...there’s a niggling doubt. I mean, you know... but trying to be scientific about
it, strictly scientific and not emotional kind of thing I think it’s almost as close to
guaranteed negative as you can get.

I asked Duane what he thought about the whole idea of Helen having predictive testing and he
said “oh absolutely. Absolutely. I can not fathom anybody not.”

I just cannot see/ when a disease could be eradicated you know, by genetics and
decisions about having children and not having children/ and some people are
going to be screaming “Hitler!” and you know this and that, but/ there’s a
common theme you know about what the eco {ecology} thing/ about what the
kids are going to inherit and this and that. And people seem to be pretty
concerned about the pollution and blah blah blah and the debt and I think that
this probably goes along the same track only in spades.

The concern about reproduction was especially important, he thought, for Helen’s sons since
they did not yet have children. He had not spoken with them about the test as it was Helen’s
“show” but he thought they might be a little “miffed” about not knowing what was going on.

Duane was looking forward to having the whole thing over with; Helen had “been
grouchier and miserable”. He tried to be supportive but was “not really good at taking shit from
anybody”. He saw himself as employing two strategies when things were really getting to him.
“I can either just walk away or say what’s on my mind and I usually don’t mince words.” I
asked if there was anything in between and he sighed and said,

I would imagine she probably thinks I'm pretty non-supportive. I don't know, but I would think she probably does because I found her pretty hard to get along with lately. I guess I'm more recoil than try to reach out because I could just, you know, figure I'm going to keep getting shot down all the time. I guess I'm not real forgiving either. Probably not good.

I asked if there was anyone that he had talked with about how he was feeling about Helen having the test and he said, "actually I haven't mentioned it to anybody. I'm pretty much a holder in-er." "How about your family?" I asked.

Well they know. Like my parents know that she's going through a test and uhm/ We're just not that kind of a talking family, you know. My Mum's English, stiff upper lip, and my Dad's a stubborn Kraut.

And because I really don't think there's anything to worry about/ I don't get into really any/ maybe that's why I have a hard time dealing with her grouchiness but I really don't think there's anything to worry about. And sometimes I'm kind of wondering is she's just being grouchy and this is an excuse.

Duane thought that Helen did not react well to stress but he had an idea from his own experience with a cancer scare what it must be like.

I can only speak for myself and I'm not going to go into great detail but I had a pretty good worry about the big C a few years ago and I had to wait for a specialist. I had to wait and wait, and the day I was supposed to see him it was foggy and the plane couldn't come out so I had to wait another week. So I try to equate that and I don't think it really affected/ I don't think I was as miserable to be around as she is. That might be prejudicial too but I don't think I was.

Duane wondered how other men coped with the same situation. "I think that they would be wanting to/ really wondering if they were doing the right thing or thinking the right thing or whatever." I asked if he thought that men and women would cope quite differently with being married to someone going through predictive testing. He said he was sure there was a big difference; he didn't know what it was but it was there. I said that perhaps it had to do with how much men and women felt they needed or wanted talk about it.

Probably. I would think that women would be probably be more open and men more holding within, I would think. I mean cause that's just the way we are.

I asked if he was suggesting that this might make things harder for men. He did not think so and I said "just different then?"

Just different. That's exactly the word. You beat me to it, I was going to say "different". Not necessarily harder, just different. I would think at a real serious/ Like a 50:50 risk or something I would think it would be pretty unpleasant. I wouldn't even want to think how unpleasant that would be on a real 50:50.
I asked if he meant that the waiting would be harder and he said yes, “for either person”. He did not see Helen’s test as a “real 50:50” and he wondered why she was so grouchy, whether the test was “an excuse.” Likewise, he could not understand why Norma did not want the test. He did not see it as a “big deal”.

...it's just leak some blood, isn't it? The actual testing. It's a blood sample. It's not like they have to conk you out and operate on you or anything, it's not a biggie.

Duane wanted me to clarify what the outcome of Helen’s test would be for Norma since as he put it, it was sort of like Norma was taking the test too. I said yes, to a point, he was right since if Helen turned out to have the gene for HD, it would mean that Norma’s a priori risk would go from 25% to 50% and he continued,

Absolutely, yea. Yea. If {Helen's} not {positive} it doesn't really prove that she's {Norma's} not either but significantly it would make it point zero zero or whatever of a percent I suppose.

Helen was less inclined to minimize Norma’s risk but, like Duane, she thought that Norma should at least consider having the test. According to Helen, Norma’s husband Ken did not, however, seem to think there was “any sense in it.”

He {Ken} knew my mother, he said there's no way she could have been ill and we didn't know. He thinks it/ I think that he thinks/ let's make that clear, because/ I think that he thinks it's a waste of time. And I think she's being influenced somewhat by him. I think she's probably feeling really pulled both ways because I feel so strongly that you have to be tested. Her son is twenty-five. I feel either she or he has to be tested now.

So Ken's on one side and I'm on the other and we're both pulling. So we really haven't discussed it.... I haven't said anything because she hasn't other than very casually. She knew I was coming down to be tested but that's about it.... I haven't wanted to push it. It's not hidden. It's just not something that has come up at a time that we could discuss it openly.

Helen thought that Norma had her own way of dealing with things — “her way of dealing with things is not to deal with them.”

That's her way/ and Duane’s way is not to deal with it until he has to/ my way is to sulk. (chuckle) “If you're not going to talk to me I'm not going to force you. You can drop dead and I won't talk to you.” It's not that bad but that is/ Oh god, am I really saying this? But that's true. I withdraw if I put out feelers and I don't get a positive response. I withdraw and I probably have been quite withdrawn.

I said it sounded as if she might be testing the water to see how people would respond and she replied that she had two girlfriends that were very supportive of her decision to have the test.

One I'm very close to and I feel I can say anything and it wouldn't be held
against me...I have a sounding board there. I have another girlfriend too who I don’t see a lot of. She had cancer about two years ago, maybe three. She had a breast removed and I talk to her when I get the what—if’s because she I know gets the what—if’s too because with her it’s “what if the cancer comes back? What if?” you know. So I’m very lucky in having a sounding board there when I get these really stupid feelings because some of them are really stupid. I call her and you know, “Got to talk to you, I’m having a hell of a day. What if I rot to death and nobody finds me?” And she says, “yea, I feel that way too. I used to think I’d wake up and the other breast would be gone and I wouldn’t know what happened.” So it’s kind of morbid but it’s a nice match.

Helen thought that it was very important to be honest about her feelings. It was, however, much easier to talk to a girlfriend than it was to talk to her husband. She wondered if this was “sulky” behaviour and I said it sounded to me as if she was really working at sorting out how she felt.

Well I decided right from the start that having/ or the possibility of having Huntington’s is basically the same as having cancer. There’s no reason it should be any harder to discuss than it would be if it was cancer or anything else.

Helen told most of her friends that she was “being tested.” They all said the same thing, “there’s absolutely no way it could be positive, we know that you’re okay”. Helen thought that this was “kind of nice” but “what else could they say?” Duane’s family also knew about her decision to have the test. And, when Helen talked with Duane’s mother about it she found that his mother knew someone in Vancouver with HD so “she had some idea about the symptoms.”

So I do have a lot of people I do talk to. I haven’t made a secret of it. Possibly telling my friends was therapeutic for myself. I have to tell people therefore I am not ashamed of it, therefore I can deal with it better. It’s probably just a coping thing. I don’t think it’s a feel sorry for me because I really don’t want anyone to/ other than when I’m feeling depressed then the whole world should feel sorry for me. (laughter) So I do have people I talk to. And if I said to Duane, “sit down and talk to me,” I know that he would but I just find it difficult to feel that I’m forcing him to talk to me.

I was interested to know how Helen felt about living in a small community where it was hard to keep things private and so I asked if there were people that she was concerned should not know about any of this. She replied that she did not want her sons to know until she knew “one way or the other to tell them.” Otherwise, she did not really think it made much difference to her. She then mentioned that she would be looking for work in the fall and this caused her to question whether or not a “positive” test result might make a difference in getting a job.

I don’t know if the tests are positive, it may make a difference whether or not I’m going to get a job. This is a small town. That’s a possibility. But I really feel that if somebody wouldn’t hire me because I’m going to get sick in ten years, I don’t want to work for them. Which is fine to say until I really need the job. That would bother me. But that would be the only way. I mean I don’t have anybody I
feel I have to impress. I don't feel in need anybody's approval. I am what I am and if you don't like it just get the hell out of my hair, you know. Just go away.

I asked if there was anyone who had responded in a way that made her uncomfortable and she said no, that there were even a few people who knew something about HD.

One of my girlfriends knew somebody who/ this weird old man who lived around them when they were a kid and their mother said, “he's not drunk, he’s got Huntington's, just go around him.”

For the most part, however, Helen found that those who did not know anything about HD would not ask for more information. Helen thought that this was because people that knew her also knew that she would talk about it if she wanted to but that she might “get nasty” if they asked too many questions. I said that perhaps some people were reluctant to ask because they were afraid of sounding ignorant or as if they were feeling sorry for her. Helen replied,

I think you've got it right on. I have discovered that the quickest way to end a conversation is to use the word dead. People don't talk about death. When my Mum died, people of my age group did not talk about it. It was as if she didn't exist. I had had a little bit more experience. My father died first and my Mum was very hurt by the fact that people just never discussed it. He just didn't exist. And then I had read/ when Norma’s husband died, her common-law husband/ I had read into it as much as I could because it was my job to protect my sister. And I realized that you have to talk about the dead.

But the reaction that I get when I mention that I'm being tested for this genetic thing that may be in my family/ I get the same reaction as I got when I would mention something to do with my mother after she died. Anything to do with something that was/ belonged to my mother/ or my mother used to think/ I get the same very friendly response but reserved and/ “let’s talk about the dogs or the trees or hunting or fishing.”

I agreed that people often have great difficulty talking about death and Helen said that it seemed to her that “if it's not talked about it's got to be hidden or bad or the old term dirty.”

It was only two days until Helen’s results. She said she felt “good” but that she had cried all morning on the day before her last appointment with her doctor. She had been scared but now felt that it was part of her “ups and downs”. She had been putting a lot of energy into a computer course and, upon reflection, she decided that she probably had not let herself really think about the test until the last few weeks.

So /last month/ was really bad because I was really recognizing it emotionally for the first time. I didn’t really deal with it head on for the first six months. And I told myself from the start that crying didn’t do any good for anything and I

17 Helen’s father died about ten years before her mother. He had a history of heart trouble.
would not allow myself to be overcome by something that was/ unless it was real, you know, because to me at this point it’s not real, it’s a maybe.

I feel that mentally I’m strong. I’m very strong. Stronger than most of my friends. And what I need is the tools to work with and the information. Without the information I can’t do anything. And I feel that I can handle most anything as long as I know what I’m dealing with and that is very important to me.

Duane said he had not contemplated the possibility that the results might not be negative. Thus he anticipated that things would be “like normal” once Helen had her results. He envisioned saying “phew, I’m glad that’s over”. Helen’s story was, in this respect, somewhat more circumspect. She allowed herself to believe that she would not have to deal with learning that she had the gene but she was far more cautious than Duane in saying so; it was a “maybe”. She said that, regardless of the outcome, she would need to come home, shut the bedroom door and deal with it. In framing her experience with the predictive test as an opportunity to “renew things” that she already knew, Helen perhaps saw no reason to expect that this confrontation with mortality would be any different than the others that she had already experienced. HD was, as she put it, “like any other disease” and that to her, was “the turning point.”

As we shall see in Regina’s story, this theme of renewing things that are already known has a different resonance. For those who are all too aware of HD and what it means to lose a family member to the disease, familiarity brings a certain trepidation about how the past may repeat itself; this is no less pronounced for the spouse than it is for the person at risk for HD.

Taking the Decision

Regina’s story of deciding to request the test has some obvious parallels with, as well as differences from, Colin’s and Helen’s stories. Like Colin, Regina talked about how her views on testing had shifted over a period of time but, unlike Colin, Regina “all of a sudden” experienced the feeling that something had changed. Like Helen, Regina was very decisive about what she wanted once she knew what it was but, unlike Helen, Regina was familiar with HD. There was no question for Regina that there was a 50% chance that she might have inherited the mutation associated with HD and there was no doubt that she knew exactly what the disease entailed. The pivotal point of Regina’s story was, then, the realization that there was an opportunity for choice; that she could “take a very hard decision” and move on with her life.
As mentioned in the introduction to this chapter, the plot structure that I refer to here as *taking the decision* involves a fairly sudden shift from not knowing, fully understanding and/or caring about the test to wanting it right away. As such there is an abrupt change from not being in a position to decide and/or not wanting to decide to feeling that it is time to decide; this change is marked by the realization that there is a decision where none seemed to exist before. A diverse array of events and circumstances are pivotal in bringing about this change although the basic plot structure remains in evidence in all of the stories of *taking the decision*.

Six of the sixteen PT candidates interviewed for this research told stories of *taking the decision*. These stories are differentiated by the events and circumstances which prompt the narrator to realize that the test presents an opportunity for choice. Two PT candidates (Regina and Adam) openly acknowledged that they were at 50% a priori risk but endeavoured to get on with life and not dwell on it. Both were in their twenties at the time of the interview and had a parent who had onset of HD in their thirties. Both had been aware of, and potentially eligible for, the linkage test but neither felt compelled to consider having the test. This changed for Regina when, some months after her father’s death, she began to acknowledge that it was time to make a decision. Adam had thought about it somewhat but put off deciding until his fiancee suggested, with increasing regularity, that it was important to know sooner rather than later.

In the other four stories of *taking the decision*, a combination of factors obstructed the narrator’s ability to recognize and/or proceed with making a decision. This was most pronounced in Maggie’s story. Maggie considered having the linkage test when her brother was diagnosed with HD some five years before the direct test became available. Her local doctor, however, advised that the test was expensive\(^\text{18}\) and said that she didn’t need to worry about it. Living in a remote region of the province, she did not pursue it any further until she heard through her local doctor that a direct test was available. Merle was also at 50% a priori risk for HD and like Maggie, lived in a remote region of the province; she did not recall knowing about the linkage test at the time it was being offered thus did not inquire about her eligibility. She

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\(^{18}\) To be informative, linkage testing requires DNA samples from affected and unaffected family members. In addition, the lab work is more complex and time-consuming. The cost of testing would, however, have been covered by the provincial Medical Services Plan and possibly the grants which supported offering the test in the context of a research (rather than service) protocol (see Chapter III).
only happened to hear about the direct test during the course of a routine check-up with her family doctor. Neither Maggie nor Merle received the newsletters from the Huntington Society.

The remaining two stories of *taking the decision* were told by Brenda and Landis. These two stories were marked by the narrator’s lack of anxiety about being risk for HD. Both had grown up knowing about the family history of HD but neither had given it much thought and each was, in consequence, only prompted to think about having the test when, among other things, a health care professional familiar with HD suggested that they might want to look into it. Each had, up until this time, seen no reason to pursue having the test. Brenda was at 25% a priori risk but had for a long time been under the impression that she was not at risk since her father had died (of colon cancer) before showing any signs of onset. Moreover she also assumed that he had been tested and that she had nothing to worry about. Landis had long been aware that HD only affected older people in her family and, as such, she had never worried. She had, at the time of the interview, only just learned from her father that he had the gene, that she was at 50% risk and that her brother Nigel was also having the test.

In these narratives of *taking the decision*, the common plot structure derives from the absence and then sudden presence of the narrator’s realization that there is a decision to be made. Once this realization occurs the narrator is decisive. The decision is "taken" as it is presented and there is little question about whether or not to proceed. Regina’s story offers a clear illustration of this pattern and, moreover, it is the only story that I encountered in which a PT candidate decided to proceed with test despite their spouse/partner’s objections.

I met with Regina¹⁹ at her home about two weeks before she was scheduled to receive her predictive test results. She had been at work all day and her eight year old son Geoffrey was unwinding after a day at summer camp. Her husband Mark was out of town and would not return until after she received her test results. He had attended one of the counselling sessions with her but, as she explained, she had not been able to schedule her results session until after his work commitments were already set. "It just kind of happened, it wasn’t done on purpose."

¹⁹ Regina was twenty-eight years old and, as the reader may recall, she was one of five PT candidates (discussed in the previous chapter) who had grown up knowing about HD. Her father, who had HD, had died only about a year and a half before. Regina had been married for nine years and she and her husband had an eight year old son.
When I talked with Regina that evening about who else she might consider asking to be part of the research, she mentioned her close friend Denise and an old family friend, “Auntie” Ruth. She did not want to include her mother or her sister in the research.

Regina spent a long time telling me about her family and, in particular, her father. He had HD and his death hit her “very hard.” She had been very close to him, “probably Daddy’s little girl.” She did not have a close relationship with her mother and she and her two sibs had all left home “on really bad terms”. As far as Regina could recall each of them had been “on the outs with her” so it was not “one-sided”. She said this was “definitely normal” in her family.

Going to church was not a part of Regina’s life although she was living in a strongly religious community. Her mother was Catholic and her father Protestant but neither converted when they married. Regina did not believe that it was necessary to go to church “to believe” and noted that wherever there is a lot of religion there are also “a lot of hypocrites.” I asked about her own beliefs and she said that for awhile she really didn’t care, “cause we had lost Dad and then three days later my grandmother died in (Ontario) and I thought yeah right, there is a God? Yeah? There is a God?”

I asked about Mark’s family. She said that he had five sisters and that although she got along well with most of them, it was “too many women to get along with all at once.” We laughed and I asked about her friend Denise. They were close friends and, because Denise had two boys about the same age as Geoffrey, it worked out well for Regina and Denise to do things together. As it turned out, their husbands also worked together.

Regina and Mark had been married for almost nine years. They worked hard to pay down the mortgage on their home and give Geoffrey the opportunity to take part in a variety of activities. They probably would have had two children “if it wasn't for the Huntington's” but they decided after Geoffrey that “one was enough to take the risk with at the time” and Regina had a tubal ligation when Geoffrey was about six months old.

...that would have made me twenty/ which is very young. They won't usually do it for you when you're that young. And of course, that was the reasoning behind it and the doctors okayed it because of that reason. So of course we discussed

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20 Denise was in her early forties. She and her family lived near to Regina and their husbands worked together as firefighters. Denise had known Regina for about six years.
that. But it's just not something we talk about a lot. It's there but it's not there.

I asked Regina about how she had felt about the pregnancy at the time and she explained that she "was too far along to do anything about it" even if she had wanted to. She was "one of the lucky ones" who had three pregnancy tests come back negative so that she was nineteen and a half weeks pregnant by the time that it was confirmed. As she said, "I'm not saying I ever would've changed it but I never had a choice." Regina thought there was, however, one thing that she had done that was "probably very strange."

I don't know how long ago/ Dad was quite ill at this point and I sat down and told Mark that if he had some/ if he had second thoughts about being able to handle it that the opportunity for him to get out of this relationship was now because once I got sick it was too late And he said no, no he was in for the duration and then people would say "you're so mean for putting him in that position" and I said "it's cause I know in my heart if I get sick he's there because he wants to be not because he feels obligated or trapped to do it", and I said "that means a lot to me, it was never said to be mean, it was said so if he doesn't feel he was trapped into doing something."

I said that it sounded to me as if she was saying that this was something that she needed to know and she replied quite emphatically that she "didn't want anyone's pity".

I asked how she and Mark felt about talking with their son Geoffrey about HD. Regina said that he had grown up knowing about it because of her Dad and because,

We've always been a firm believers that anytime he asks a question it's answered honestly, but again we don't beat him down the throat with it. So if he's asked the question we answer the question, if he hasn't asked we haven't volunteered the information, cause we figure when he gets older he can ask then.

Now if my test comes back negative we don't ever have to worry, the only thing we have to tell him is "It's done with you don't have to worry about it anymore." If it comes back positive well then he is going to have questions and again they'll be answered at the time whenever he asks them. I don't think it's really a topic that you need to sit down and say "okay Geoffrey’s getting older now we'll sit down and have the talk with him." No.

I started to ask Regina if she had talked with friends about the family history of HD but I trailed off without finishing my question, wondering if I was somehow over dramatizing the issue. From Regina's perspective, it was not all that complicated.

If the conversation happens to be running that way (toward the topic of HD), then of course it will come out and of course there are people that know. It's not like I've ever tried to hide it, but I also don't volunteer the information cause I don't think it's something people want to hear about, "Oh by the way did you know?" Like just imagine that would scare off a whole pile of friends real fast, they'll think you're a nut case I'm sure.
Regina figured that she had known about the test for a couple of years and that up until recently she really “didn’t care” about it. I asked if she meant that she had not been interested in having the test and she said, “I never made the decision one way or the other/ didn’t care whether I did or didn’t know, wasn’t willing to make that decision in my life.” I waited and then asked why things had changed for her.

I don't know why it's changed/ it's ahh/ I don't know whether it's cause my life's been so busy for such long time that I never had time to think about it/ cause Dad was so sick and everything else/ ...that my life has finally come down enough/ has slowed me down enough that I've finally had time to think about it.

I don't know/ because I know my Mom was upset when my brother had his linkage testing done. I told her he deserved a lot of credit because he had taken the decision/ a very hard decision/ and made the decision one way or the other, whereas I was riding the fence. I never decided whether I wanted it done or didn't decide that I didn't want it done/ like I was sittin’ on the fence. My sister she's totally different. She said if she ever gets it {HD} she'll just commit suicide, so that's kind of her philosophy on life right/

And then all of a sudden I just, it was like it was bugging me all the time, and I wasn't sleeping and I thought/ not being a real religious person/ I thought “somebody somewhere is telling me that it's time I made the decision in my life” so I figured like/ because/ if not/ it wouldn't be in the forefront so much/ like I don't mean forefront in the sense of always talking about it but (...) instead of being in the subconscious it was kind of consciously there all the time. I thought for something that's never bugged me before there's got to be a reason for it, so I just made the phone call and did it.

Like other PT candidates who requested the direct test at about the same time, Regina was on a waiting list for about six months. She thought that this was “pretty long” to wait.

I'm very impatient, like if I go shopping and I want to buy something and they don't have it, God help you cause I'll go somewhere else and buy it. I'm very much impulse on some things. I want it now. Don't wait.

Wondering whether or not her brother Giles’ experience with predictive testing had played a role in Regina’s decision, I asked if she had known ahead of time that he was having the test. Regina explained that she had not known at the time because he had “kept it quiet” and that it was not until her mother “got her hands on the information” from one of Giles’ old girlfriends that he had “confided in” that she heard about it.

And all of a sudden Mom was sitting in her house one day and there was a whole pile of people around and she just decided to announce the information. That's one of the reasons why she does not know I'm having the test done and

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21 Regina’s brother received his linkage test results some months before their father passed away. He did not, however, disclose the results to his family at the time.
she will never know the results. My brother knows but he won't tell her cause he
even said I don't want her to do to you what she did to me because it was/ if
Giles wanted us to know he would've told us and he confided in somebody and
somebody killed that trust and it's not a nice thing to do.

She's very selfish in a lot of ways, she's just “me me me me oh look at what this
is doing to me.” She doesn't think about what it's done or what it could do to us
kids. Those are kinda little examples of stuff that she's done and it's just/ sorry,
to me you just don't do stuff like that.

I asked Regina if she thought that her brother's test results had an impact on her own decision.

I don't know maybe indirectly it did, but it was quite a while after that that I
finally called to get it done, maybe (yawning) indirectly it did but, I don't think it
made a big difference.

Giles doesn't say much, he's the type of person/ he doesn't butt into your life,
unless there's a real big reason for it, he encourages me in what ever I want to
do, and then I'm sure deep down he's probably got mixed emotions because of
the way his test came about, and I'm still, I'm 99.9% sure that his test has come
back positive, but I haven't physically heard that out of his mouth/ I've kind of
heard it from everybody else/ but he doesn't say one way or the other/ but the
difference is when Giles had his done it was not a 100% sure/ I think there was
only 85% accuracy or something at that time. Cause he had his done before
{they found the gene}/ whereas now it's 100% accurate so. I don't know if he'll
get it done again, I don't know what he'll do.

Regina was, as she put it, a “very spontaneous person.” She called the clinic as soon as
she decided to request the test. I asked if she talked with anybody about her decision before
calling the clinic and she said she talked to Mark and that “he was totally against it.”

We were on totally different sides about this, and it finally came down to “well
it's my body my life.” It's not that I didn't listen to what he had to say, I did.
(phone rings, tape turned off briefly and then turned on again)... I weighed the
pros and cons and when it came down to it/ when push came to shove he did
dvoice his opinion quite vocally, and I just said “well”. He said he didn't want to
know and basically I've come down and told him “fine I won't tell you. If and
when you're ready to know you can ask me and I'll tell you, but I'm not such a
mean person that I'm going to come home and say oh by the way it was this and
was that way. Not only will you not find out it's good news, you won't find out if
it's bad news either.” So I told him if and when he's ready then I'll tell him and if
not, I won't tell him.

Regina acknowledged that he would probably have some “mixed emotions” but that was just
the way things were. She had listened to his concerns but she had made her decision and

...that's where it stands you know, nothing anybody says or does is going to
make me change my mind, so he can squat till the cows come home and that's—
fine, he's just letting off steam.

I asked what Mark's main concerns were and she explained,

He said he's afraid, because you see Mark was a very big part of my Dad's life
and he was very close to my Dad. He had a very hard time when Dad passed
away and he's even said that he's afraid more than anything else, and he figures it's like finding out you're gonna die because it is terminal/ I do know it is terminal/ we just have very different feelings about it that's all.

I was still unclear about what had prompted Regina to shift her thinking from not caring about the test to deciding that it was time so I reiterated what she had said about how HD seemed to be in her consciousness in a way that it had not been before. She said,

No it's not just that/ one thing/ is Geoffrey because either way it's going to answer a lot of questions about Geoffrey and for Geoffrey. But also there's little things that we've always decided one of the things. Like I always wanted to do is go on a cruise/ always wanted to go on a cruise. Well I'd like to go when I'm in my thirties rather than when I'm fifty, because I plan on having a hell of a good time.

Regina also talked about financial concerns. She wanted to be sure that they were able to assist Geoffrey in going to university if he wanted to and that it was important to meet their goals for paying off the house so that there would never be a big debt to deal with. They had always done things “a certain way” but their plans were based on having two incomes. Regina did not want their goals to change but thought this might require some additional planning.

To briefly summarize, Regina described her process of arriving at the decision to request predictive testing in the following way. She went from 1) knowing about the test and not caring about pursuing it any further to, 2) either feeling ambivalent or not wanting to make a decision, to 3) losing her father to HD, hearing about her brother’s test results and feeling that “something had changed” to, 4) acknowledging that “it” was bugging her all the time to, 5) deciding that it was time that she made the decision to, 6) wanting to have the test right away despite her husband’s objections to knowing the results.

In Regina’s story her reasons for testing remain quite distinct from the phenomenological experience of making a decision. Her story was, in this respect, far more focused on the experiential than the rational dimensions of the decision and it was a perspective that I found intriguing since most PT candidates had integrated their reasons for testing so closely within their story of deciding that it was very difficult to extricate a sense of what, in the narrative now, people recalled about how it felt at the time to perceive that there was a decision to be made. This lack of separation between reasons for testing and the process of deciding was especially evident in the stories of having to know.
In Regina’s story we also see that there may be an important distinction to be made between not caring about the test and “sitting on the fence.” Not caring about the test was one thing but once Regina framed what she was doing as “sitting on the fence” it was no longer acceptable to her to *not* decide one way or the other. Deciding to not decide was not a decision and she was, ultimately, a decisive person. She was “very much impulse on some things” and once she had made up her mind that she wanted the test, there wasn’t anything that anyone, including her husband, could say that would change her mind. She listened to his concerns and “weighed the pros and cons” but as she put it, it finally came down to “it’s my body, my life.”

Mark was not due to arrive home for at least a day or two after Regina’s results session so I asked if anyone else would be with her when she went to the clinic. As it turned out, her friend Denise had arranged to “be there for her.” When I spoke with Denise she immediately established that she was not a fair-weather friend and that no matter how the test results turned out, it would not affect their friendship.

Like to me that’s not the biggest part of Regina. I mean Regina/ It would be the same thing as her/ Like I have sort of a high tolerance I guess or I can sort of empathize with people that have something wrong with them. For me I was burned really bad when I was about five years old so I’ve always had kind of a/ like I’ve always/ through growing up and what not I’ve had a lot of trouble with people that are sort of/ that shun you because you’re different. So I try not/ If anything I go the other way by being overly understanding.

Like I said, either way, which ever way the test results/ For Regina I’m glad because like I said, now while she’s still got the health, while she can still travel and move on it would be a good way for her to sort of plot out the rest of her life because there’s a lot of things that she can do now that she probably won’t be able to do later and that’s what she wants to find out more than anything. So I’m actually going in with her on {results day}. I’ve got the afternoon off with her when she goes in to get the results.

Denise and Regina had been friends for about six years and, as Denise explained, she had known Regina’s father and been to the hospital many times with Regina.

So I know what to expect and I know if it comes out positive that the same thing is going to happen to Regina so I’m/ Like she said, there’s things that she wants to do, cruises and places to go and things to see and do that she’s not going to be able to do once the symptoms really start taking hold. But like I said, either way, it’s not going to affect our relationship. We’re too bloody much alike. (laughter)

Denise and Regina enjoyed doing things together, especially shopping. Denise was in awe of the way that Regina was able to drive a bargain with the sales clerks. The two women also got together regularly on Saturday nights to watch “the kind of movies that the guys don’t
want to watch." It was an affectionate and caring friendship and both women supported each other through the tough times each had in coping with family and health related concerns.

Like I said since Regina and I have known each other since {my husband's} Dad passed away and that was a difficult time. Then I had an operation as well. I had a hernia operation. She came out and really helped me out with that because for two or three days I could barely walk, let alone carry on and look after kids and do what not. So she helped me out with that. And then when her Dad died/ and then the odd time where you're not getting along with spouses or work or whatever, you know, things like that that bother you, we can talk about it.

Denise mentioned that Regina had a very difficult time with her mother and that her mother did not seem to approve of anything that Regina did.

I mean my mother and I we don't always see eye to eye but I always knew that my Mum loved me and she never ever put me down or anything like that. But I can't understand Regina's mother. She's not happy in life. She's playing the kids against each other. She's only got one favorite at a time and she pushes a wedge between the other ones.

What was most amazing to Denise was Regina's strength of character and ability to thrive.

...this is why I'm not too worried about how she's going to handle the {test} results {next week}. Either way/ because she is a strong person. I think she's had to be, being raised with a mother like that. Like I think that her Dad must have been one hell of a person too because you can see a lot of him in her.

HD was not a regular topic of conversation. As Denise explained,

It was just accepted. We never really sat down and like/ she's volunteered/ like if she wants to talk about it I talk about it with her but I don't poke or prod or whatever/ if she wants to talk then she'll talk and that's how the base of most of my information has come is information that she has volunteered.

Y'know like/ since her Dad died we've spent more time talking about it than what we did before and that's when she said that/ I asked her why she hadn't gone for the testing before, she said "well I just never really wanted to know before, but now I'm comfortable with it and I would like to know cause there's plans I would like to make should I get it in later years."

And I've asked her the odd question like with her Dad/ like when did the symptoms start with her Dad/ and I think that has something to do with it too because she said that he started his symptoms in his thirties and she's starting to come up to thirty/ and I think that has a lot to do with it too as she's starting to get to the age where if she's got it symptoms are going to start to show/ probably within the next ten years or so, it would be a good time to find out now so that she can do the things that she wants to do. I would feel the same.

Denise also recalled asking Regina about why her brother wasn't married. She had thought that there was something amiss since both Regina and her sister were married.

But I don't know, that's the only other time we've really discussed it, the only time that we ever really talked about it has been when she has volunteered information or we've talked about something else that has a relation to it, but we
never really sat down and seriously discussed/ like you're going to die kind of thing, I look at it the same way as/ I could go out tomorrow and get hit by a bus you know. Things happen all the time. You take that chance whenever.

As Denise explained, there was always an element of uncertainty. “You take your chances... life is life and you got to take it one day at a time.” I asked if she recalled talking with Regina before she decided to have the test and she said, “nope, she told me after the fact.”

It's the same thing, we don't talk about the death of her Dad. Like we did a lot around the time and after for a while/ while she was grieving and then again you go on from there. Like I said she's a lot happier now, she's a lot more in control of her life, a lot more comfortable about who she is and where she's going and what she's doing. She's more independent now I think uh/ (..) she's healing and she's getting better... she was a strong person before but now she's got more inner strength than what she had before her Dad died.

I asked Denise about going with Regina to the clinic on results day and she said, “I want to be with her.” I asked how she thought the day would go and she replied,

Either way, we'll be there for each other, cause I know for myself if it was me, I know I'd be nervous, kind of anxious. I'll imagine she'll be like that, she says she's not but, it's there. She's just not letting on.

I'm hoping that it comes back saying negative but/ but in the event it comes back positive I'll be there to help her, give her moral support if nothing else, as I said it's not going to affect our relationship or anything we'll just have to sit down and have more shopping trips, plan more, get more things done. But, I think I'm probably feeling somewhat the same as her in that she's a lot like me in the fact that she doesn't like to be on that fence, she doesn't like things up in the air, she likes things resolved and I think she'll feel much better when it's resolved/ it's not something that's sort of hanging there/ that's an unknown.

Regina valued Denise’s support but results day was “no big thing”. As Regina said,

It's not in the forefront of my life, it's not like I'm trying to hide from it, but I don't want to dwell on it either. I just go on as if life's quite normal/ it's not like anything's going to change/ and say I knew was going to be the lucky one/ once I know either/ so, I'm either going to let out a huge sigh of relief or (..)/ I just keep telling myself that it hasn't missed a generation in a really long time in our family, as far back as I know it hasn't missed a generation and I figure I'm due.

I asked Regina if she had a hunch about how her test results would turn out and she said, “way back in my mind yeah.” She could not see the point in dwelling on things that could not be changed. Thus she, like many other PT candidates, would only allow herself to go so far in articulating what it might mean to know that she was “going to be the lucky one”. The focus was firmly on the value of knowing and then moving on rather than worrying and anticipating the day. It wasn’t “like anything’s going to change.”
Discussion

As proposed in the introduction to this chapter, narratives are in life as well as about life. When we make a decision or ask ourselves a basic question such as “why am I doing this?” we must find amidst the infinite details of our daily experience, some sort of pattern that makes sense of the past and projects a meaning onto the future. This is a narrative process and though the stories that we subsequently tell about such decisions may reframe the stories that are in decisions, this is a part of their “truth”; it is what they have come to mean (Frank, 1995).

The preceding stories give rise to three different plot structures for narrating the experience of arriving at the decision to request predictive testing. These plot structures are not necessarily exhaustive nor are they unique to Colin’s, Helen’s and Regina’s stories. They do, however, provide a useful comparative framework for understanding how PT candidates (and to some degree, their family members) story their experiences of the decision.

Colin’s story exemplifies the gradual process of evolving toward it. In this type of story, the decision is consciously apprehended as a decision and it is, in consequence, the explicit subject of effortful critical reflection. There are, however, limits to this process of critical reflection. As Colin concluded, “we have done all the thinking we need to do...let’s just do it and then figure it out afterwards, what it means to us.”

Helen’s story of having to know illustrates how the ostensibly self-evident act of deciding is anchored in other relevant life circumstances. In this type of story, the decision is not apprehended as a decision per se and it is, in consequence, a way of coping that cannot be fully articulated. The presuppositions, values and beliefs that inform Helen’s certainty about requesting the test are part of the routine knowledge that allows her to go in life without subjecting everything that she does to critical reflection. She is, as she puts it, “renewing” what she already knows.

Regina’s story of taking the decision does not fit either of these patterns; it begins with an absence that she refers to as “not dwelling on it” and then something changes, “somebody somewhere” tells her that “it is time.” This raises intriguing questions about the process of perceiving that there is a decision to be made. How is it that the background practices which have allowed Regina to go on in life have shifted such that she consciously apprehends the
need to make the decision to request the test?

The three plot structures underlying the story of deciding to request predictive testing may be compared across several dimensions. These are summarized in Table 9 (see next page). Most centrally, the plot structures are differentiated by the temporal unfolding of events and the degree to which the narrator reflects upon the decision as an opportunity for choice. The stories that revolve around these plot structures are also differentiated by the phenomenological aspects of deciding and most salient meanings of the test (i.e., as instrument for planning the future, an end in and of itself or an opportunity to live more fully). Further, each plot structure points to a number of questions that have a larger social, as well as clinical, significance.

Finally, there are also distinctive patterns that emerge from looking at the common life experiences and family history of those who tell each type of story. This is perhaps obvious given that the defining features of each plot type are closely linked to the narrator's family history and awareness of HD (i.e., long or short duration, based on abstract knowledge or firsthand experience of HD); it is nonetheless important not to overlook the fact that the narrator must select and order a sequence of relevant events when storying the experience of deciding to request the test; the plot structure is, then, not a foregone conclusion nor does it mirror precisely the experience of deciding. There is an inevitable gap between the lived and told story such that persons confronting similar events often configure them into different phenomenological events (Riessman, 1989). The process of configuring events into a story is therefore both active and agentic just as it is patterned and coherent.

As I began this chapter by saying, it was apparent to me during some interviews that I was not hearing a part of the story that I expected to hear. When I stopped listening for the anticipated story and started hearing its collective absence, I realized there were a group of PT candidates for whom the decision was a self-evident act. Though there were, on the face of it, many differences in these stories of having to know, there was an unmistakable certainty about proceeding with the test which distinguished these stories from all others. Moreover, the narrator had a strong sense of agency. These were stories that said: I am going to do something about being at risk for HD; I am decisive and there is no question that I have to know.
<table>
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<th>evolving toward it</th>
<th>having to know</th>
<th>taking the decision</th>
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<tr>
<td>temporal unfolding of events</td>
<td>gradual or incremental process that occurs over a period of time; involves continual or staged changes in thinking; weighing up of the implications of predictive testing for self and others; arriving at a feeling of readiness and preparedness</td>
<td>immediate response that does not require consideration; involves certainty about need to know for self and others; may involve moral imperative and/or time constraints (e.g., reproductive or other decision-making that rests on outcome of test)</td>
<td>abrupt change from not caring about test and/or not wanting to decide to feeling that it is time; involves seeing a decision where none existed before either through new information, example of others or changed life circumstances</td>
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<tr>
<td>perception of decision as an opportunity for choice</td>
<td>conscious perception of entering into process of decision-making hence intense awareness of need to choose between knowing and not knowing</td>
<td>decision is a self-evident act; may be assumed to be an automatic response to knowing about being at risk for HD</td>
<td>circumstances vary but perception of choice is ambiguous; decision not to decide or to put off deciding presents one kind of choice</td>
</tr>
<tr>
<td>phenomenological aspects of deciding</td>
<td>period of ambivalence precedes deeply examined sense of why it may be the right thing to do accompanied by weighing up of responsibilities to self and others</td>
<td>inexplicable feeling of being driven to know accompanied by certainty about responsibilities to others</td>
<td>experience of not caring or thinking about it suddenly replaced by new consciousness that it is time to decide; certainty about proceeding as inchoate experiential phenomena</td>
</tr>
<tr>
<td>salient meanings of the test</td>
<td>an instrument for planning the future and means of making course adjustments and/or relief from the uncertainty of experiencing what may be signs and symptoms of HD</td>
<td>an end in and of itself, the fulfillment of obligations to others, an opportunity to rule out HD, an achievement, a contribution to the research</td>
<td>varies but may be an opportunity to live life to the fullest by acknowledging HD but not dwelling on it; may also be an opportunity to rule out HD</td>
</tr>
<tr>
<td>patterns in narrator's family history and awareness of HD</td>
<td>long awareness of HD; immediate, significant, and/or lasting experiences with HD</td>
<td>limited awareness of HD derived fairly quickly; lack of immediate experience with HD but other life experiences may be relevant</td>
<td>long awareness of HD may be accompanied by desire not to dwell on it and/or confusion about genetics of HD and relevance of test</td>
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<td>historical and social significance</td>
<td>confirms expected pattern for those with long awareness of HD and underscores need for adequate safeguards to prevent premature decision to request test</td>
<td>raises questions about culturally expected alternatives, routinization of testing and meaning of fully informed consent especially where there is little awareness of HD</td>
<td>demonstrates complexity of how decisions emerge as decisions; raises questions about role of cognition and emotion in deciding</td>
</tr>
<tr>
<td>examples</td>
<td>Colin (and Emily)</td>
<td>Helen (and Duane)</td>
<td>Regina (and Denise)</td>
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We have already seen in Helen’s and Duane’s stories that this certainty may derive from the conviction that there is a strong moral responsibility to provide information to the next generation. The decision to have the test is, in this instance, also a decision that is made for others in the hope that they will, in the future, make their own fully informed decisions about reproduction. This was not, however, Helen’s only source of certainty about requesting the test; her experiences as a caregiver for her mother convinced her of the need to be assertive in her communication with others especially where such communication centres on the need for information. Moreover, she had twice confronted her own mortality and thus she was able to draw on her prior experiences to cope with what seemed be an analogous situation. In short, Helen found interpretive relevance in her personal biographical knowledge and this, coupled with a high degree of certitude, allowed her to proceed with predictive testing as if it were a routine “recipe” for action (Schutz, 1946). Others who told stories of having to know also found that ostensibly unrelated illness experiences contributed to the ability to cope with being at risk for HD. As Rosalind’s story demonstrates, however, the realization of one’s own mortality does not alleviate the burden of knowing that the next generation(s) may be affected by HD. Unlike Helen, Rosalind was, at the time of the interview, already a grandmother.

Rosalind grew up in a family where chronic illness was a part of life and she was, at the time I spoke with her, coping with a potentially operable brain tumor, persistent “facial numbness” and intermittent bouts of double vision. She also anticipated having a hysterectomy because of some “masses” that her doctor was concerned about. These health problems were, however, an “aggravation” that paled in comparison to the familial risk for HD. Like Helen, Rosalind had a very strong moral commitment to informing her two adult children of their risk for HD. She was “really driven”, even “obsessed” to be a part of the linkage testing when she first learned that her sister had been diagnosed with HD. As she said, I was a real “thorn in my family’s side”; “all I could think about was the kids and my duty to the kids”. She was, in consequence, the one who got the family “organized” to send in all of the requisite blood samples. She thought that her kids would “either love or hate” her for doing so but she believed that to be “forewarned is to be forarmed.” Soon after she found out through the linkage test that she was at increased risk for HD, her son also learned that he was at increased risk. As
Rosalind recalled, her biggest “sadness” was that he and his wife went ahead and had children. Though she adored her two grandchildren, she also felt great ambivalence about their existence. As she said, “my first instinct would have been to terminate the pregnancy.”

Rosalind’s certainty about proceeding with the direct test was not lessened by the fact that she already had grandchildren. She knew the direct test was more accurate and she “desperately needed to know the numbers” but she also said she derived a feeling of belonging from her continued involvement in the research. It was important to her “just be part of the program”. It wasn’t just a matter of learning the results or providing information that “somebody in your family is going to be grateful for”, however: her continued involvement made her feel that her life was worthwhile.

I’m not/ I sort of/ in this life I haven’t contributed a great deal to this life, to the world. I sort of more or less take up space and breath the air that some more worthwhile person could use so uh/ you know this is probably the only worthwhile thing that I’m really/ maybe some people don’t consider it worthwhile/ maybe they consider that I’ve just been an aggravation about it but to me its probably the only worthwhile thing I’ve done really.

There was only one other PT candidate that articulated similar feelings; his story was not etched with the same profound sadness as Rosalind’s but it did echo this relationship between self-worth and the act of participating in predictive testing.

Like Rosalind, Albert had been through the linkage test soon after he learned that there was a family history of HD. He was in his early forties and unmarried. He had several degrees but was working in a poorly paid night clerk’s job. He felt that he had, in contrast with his older brother, not done “brilliantly career-wise.” When Albert first heard that his father had been diagnosed with HD he figured that he might also get the disease. He thought of HD as a “granddaddy of a mental illness” and this became something of a “crutch” for him. As he said, “okay no problem what to do with my brains. No brains, oh well.” Getting the “good news” about his linkage test results did, however “put a responsibility” on him; it caused him rethink his “rationale” and see that, even though he had not yet established himself in a successful career he had, unlike his brother, done something about being at risk for HD.

Albert framed his certainty about proceeding with both forms of testing as if it were the logical reflection of his personality. His initial response to learning about the linkage test (from
his mother) was simply to “go for it”, “pole-vault the Berlin Wall.” When I asked why he had wanted the test he said, “Oh it’s just my personality, it’s do or die. I would rather have bad news than uncertainty.” And when I later asked about his decision to request the direct test he said quite simply, “I wanted to go ahead. It was my personality.”

In the stories of evolving toward it these sources of certainty are either ambivalent or missing; the story line weaves in and out of control; it is hedged in by the chronic or sporadic experience of anxiety and this may, as in Colin’s story, comprise a kind of parallel story line. Though there were many differences in these stories, there was in each a definite movement toward the decision that culminated in a feeling of readiness. This sense of movement from a place of ambivalence or even outright rejection of the test distinguished these stories from all others. These stories said: I want to live my life to the fullest but my risk situation is getting the better of me; besides, I can use the information that predictive testing provides to enhance the quality of life for myself and my family; I have come to a decision and I think that it is the right one although I am not certain about how I will feel once I know the results of the test.

It is, in these stories, as Heidegger (cited in Dreyfus, 1991:4) suggests: “every decision...bases itself on something not mastered, something concealed, confusing; else it would never be a decision.” Colin’s story is the most explicit in setting out the language of rational choice as the primary idiom for the expression of agency and control yet even he is ultimately unable to map out exactly what the test means. His decision to request the test initially emerges from his felt obligation to provide his three daughters with information should he get “bumped off by a bus”. He then begins to feel that the information might be useful in planning the future; he wants to move back east to care for his brother who has HD yet he does not want to move his family away from their support system in case he also develops HD; he wonders about making decisions in his business and decides that the test is an opportunity to make some “course adjustments”. At the same time, he cannot unthink the “mind trap” of wondering if he is showing symptoms nor can he talk about this with his wife Emily. This introduces a feeling of ambivalence that is not easily resolved. The test is ostensibly about the potential onset of illness and yet the presence of this other ‘internal’ story line suggests that the future may have already arrived.
Others who told stories of *evolving toward it* shared the unsettling experience of wondering if they were showing signs of onset. Some also mentioned the awareness of onset in a sibling as an equally unsettling experience. Such awareness brings risk closer and though it does not alter the abstract calculation of a priori risk, it changes everything to do with the lived experience of risk. Not all were able to articulate these experiences as an explicit part of the story. Marie, in particular, taught me about how and why it is sometimes so difficult to talk about the awareness of risk.

Unlike Colin, Marie expressed some confidence that she was *not* showing signs of onset. She asked to know the results of her neurological examination and the doctor told her they were within the normal range. Marie’s younger brother had, however, begun to show definite signs of onset. This brother lived nearby and as such, HD was an undeniably tangible presence in Marie’s life. It was, as her husband Peter said, “coming closer than we thought.”

As Marie said, “nothing was spoken about too openly” in her family and this had been the case as long as she could remember. There “wasn’t a lot of information” when the family first learned about HD and no-one was certain about which family members might have been affected. Referring to her father and his brothers, Marie said,

> They feel it was from their mother/ their mother/ but none of them would accept the fact and they wouldn’t want to talk about it. It was something like you know/ she had done something wrong/ If you/ If you knew/ if/ if you said she had it which is kind of/ you know/ that’s what you’re dealing with here.

Marie knew that her uncle had HD and that her father had been “quite scared” when he went to visit and found his brother “strapped down to the table because he was moving so much.” She was unsure about when she realized that HD is hereditary but knew for certain that she began “thinking about it more seriously” when she was in her early thirties. This was about the same time that her father was diagnosed. Marie said that for a long time there wasn’t “a strong concern for me having it.” I pressed a little and asked if she remembered talking with her siblings about their risk for HD and she replied, “No I don’t/ W–We didn’t talk much about it.” I then asked “how about your mother, did she talk with you about it?” Marie answered,

> Well, it’s not the kind of thing that/ I mean we saw it whenever we went home/ what was happening/ but as far as discussing it too much it wasn’t really talked about too much.
It wasn’t really talked about too much and we were, as I later realized, talking about it too much. “Did you find yourself thinking about it very much at the time, perhaps when you would go to visit or...” Marie was hardly breathing as she said

Uhm (...) Well IT was/ IT was THERE (..) I mean IT was uhm (...) I think I just want to get some fresh air right now.22

Upon repeated replaying of the tape, the “it” signified to me everything that could not be put into words — the fear, the endless introspection and the unavoidable always thereness of HD. “It” loomed too large and demanded too much. “It” ruptured the conversation because “it” made me and Marie acutely aware that I was asking her to put into words that which she had told me “wasn’t really talked about too much.” I had pressed Marie to the edge of what she was willing to say and the moment in which Marie refused and/or was unable to say anything more about what her experience of “it” was like was the moment in which I understood, with humility, the space between what is and is not said, what is and is not there, what I could and could not ask. This space is the always thereness of HD that resonated within the ‘narrative now’ in each of the stories of evolving toward it.

In the stories of taking the decision, the experience of being at risk for HD has a somewhat less immediate or ominous presence. Though these stories are more diverse in terms of the circumstances and events that compel the narrator to consider having the test, they all begin with the narrator reflecting on a period of acceptance and/or minimization of risk. This acceptance and/or minimization of risk may be framed in terms of a strategy for living (as in Regina’s emphasis on “not dwelling on it”); it may be the product of misinformation or confusion about the genetics of HD and the availability or relevance of the predictive test or it may, as in Merle’s story, be shaped by a quiet faith that “everything does happen for a reason.”

Merle’s mother had died some years earlier from HD–related pneumonia but it was not until after her mother’s death that she learned that her mother had HD. Merle had some “crazy” years in trying to sort out what this meant to her but, at age forty–eight, she had come to accept that her risk for HD was simply a part of life.

22 Marie went outside for a few minutes at this point. When she returned she did not seem overly distraught but did request that I not turn the tape recorder on again. We talked for awhile about safe topics such as the log house she and her husband built.
I think your life is planned and I think you just go along with it, you know? You don’t have a lot of choice in a lot of things...I think all these things, the good things and the bad things that happen to you, happen to you because you have to learn something, and if you don’t learn something from them they’re going to happen again until you do learn from them.

Merle “always figured” that she would get HD but she did not see that this presented her with any real choices; thus, although she had visited the clinic in Vancouver to have a PET scan she “never went looking for too much” in the way of information about genetic testing. Not long after she married the man who was, as she put it, “the best thing that ever happened to me”, she learned from her family doctor that scientists had discovered the gene for HD and that there was a test available. She said “yes right away” but noted that she “wouldn’t have done it before” because she “didn’t have someone to hang onto.”

Like others who told stories of taking the decision, Merle encountered a turning point in life, a juncture that enabled her to see that there was an opportunity for choice where none had previously existed. She felt secure in the knowledge that she would not be trying to cope with the test on her own and thus she was able to decide right away that she wanted the test. She thought it would allow her, unlike her mother, to have a chance to make some plans. Her risk for HD was, as she put it, “a bit like gambling” but her dream had always been to go to Reno.

Stories of taking the decision are, then, stories which feature a point of change that enables the narrator to perceive that there is a decision to be made. This change is, in most stories, immediately followed by the decision to request the test. These were stories that said: I have not up until now perceived the need to make this decision but now that X has happened I see that I am able and/or do want to know whether or not I have the gene. I have no doubt about my decision to request the test; it feels right but it was not something that I originally thought about and/or expected that I would do.

Regina talked about how HD had not been in the forefront of her life; she didn’t hide from it but nor did she want to dwell on it. She did not “know” exactly why things changed but, some time after her father died, she felt as if “somebody somewhere” was telling her that “it’s time” to decide. She thought that there “had to be a reason for it” and so she “just made the phone call and did it.” This was a sort of impulse toward requesting the test and she equated it to the way that she felt about shopping, “I want it now, don’t wait.”
In Brenda’s story, the turning point came with the realization at age thirty-five that she was, after all, at risk for HD. Brenda’s father had died from colon cancer and she had long assumed that the “chain was broken” because he had never shown any signs of onset. Not long after the gene was discovered, however, Brenda found out from her psychologist that you “could be tested for it.” Brenda immediately phoned the clinic for more information. There was no doubt in her mind that she wanted the test; she was, as she said, “like that”; she liked “to know things” and felt that she would be better able to deal with it if she “had the facts.”

So I thought, “oh well, I want to put this thing to rest for once and for all.” Now that I knew there was a possibility that Dad could have had it, you see/ that opened the book again because I always thought he didn’t get it so we wouldn’t have it. Well now I realize that that was a very ignorant way of thinking, that wasn’t necessarily true/ that now for myself and my kids I could find out for sure. So I thought it was great. I would have been tested that day if I could have done it that quick.

As Brenda’s, Merle’s and Regina’s stories demonstrate, the plot structure of taking the decision is similar to evolving toward it in that there is a series of events that folds into and shapes the narrator’s realization that there is a decision to be made. Though these events may vary, they are constructed as turning points and, as such, they serve to distinguish stories of taking the decision from all others. Brenda apprehends a turning point when she realizes that her father could have carried the gene; furthermore she frames her response to knowing about the test in terms that preserve an element of choice; her statement that “I could find out” thus stands in contrast to the imperative that emerges in stories of having to know.

In summary, the three basic plots were prominent features of the sub-narratives that I heard from PT candidates and their families about the experience of learning about and deciding to request the test. Each plot ordered the selection and sequence of events and provided the stories with a source of coherence. These plot structures were, then, both about decisions and embedded in decisions.

Each of the plot structures makes a distinctive contribution to understanding how PT candidates and their families experience the process of deciding to request the test. Stories of evolving toward it confirm what the literature has long suggested; that is, that many at risk individuals who wish to have predictive testing are experiencing a period of anxiety about the possible onset of HD. Colin adopts a popular metaphor in framing this anxiety as the HD cloud
that is "hovering over" him while Marie finds her propinquity to risk too overwhelming to put into words. These stories also reveal that the knowledge that there is a test has a bearing on the awareness and experience of risk. There is a sense in which the at risk individual is drawn into the logic of the test and this emerges in the teleological character of evolving toward it.

The apparent inevitability of deciding to request the test is, of course, a retrospective phenomenon; in stories of evolving toward it the narrators have already decided to request the test. The events and circumstances leading up to the decision are, therefore, selected and ordered with this particular outcome in mind; the story of deciding is revised in light of having decided and some events and circumstances do, in consequence, take on a greater significance than others. But what of those who have decided not to have the test? or those who have not decided? There is, as I want to suggest, a teleology here as well that has significant implications for understanding how we talk about what it means to decide whether or not to request the test.

Naomi was the only person to respond to a notice about the research placed in the British Columbia insert of the Huntington Society newsletter; this notice was placed with the intent of recruiting those who were undecided about and/or did not want to have the predictive test. Naomi was at 50% risk for HD and also had a history of serious heart trouble. She recalled that she got quite "worked up emotionally" when she heard about the breakthrough that led to the development of the linkage test in 1986: "Like I didn’t know what I was going to do. I had to basically shut down." Not long after hearing about the test, Naomi spoke with a doctor who was very involved in setting up the predictive testing program in British Columbia. She thought about whether or not she should "follow through" but decided not to because she was also having to deal with the question of whether or not she needed heart surgery.

The most salient feature of Naomi’s story, however, is the ambiguity inherent to her construction of what it means to make a decision. She was, at the time of the interview, "not sure" about whether she wanted to have the testing done or whether she wanted to "just let things be" and thus she described herself as a "fence-sitter." She was, at age forty-two, beginning to feel that she was in a "safe zone" since her mother and her two brothers had onset of HD in their late twenties. She saw the test as providing an opportunity for choice but ultimately any decision that she might make was only a decision if it was a decision to request
the test; any other decision that she made (such as “setting it aside” and deciding not to decide or deciding to delay deciding) was infinitely open-ended. Thus she said, “I’ll probably just make a decision one day and do it” and more tentatively, “I’ve decided, sort of, not to have it but I might change my mind/ I don’t know but I mean it’s a dilemma” and still more tentatively, “maybe tomorrow I’ll have a different feeling. I haven’t reached a decision.”

Naomi thought that she had “gotten more into control” of her “emotional thoughts about the test” although she said that “anytime I’ve tried to talk to myself about whether or not to go ahead and have counselling done and then a test done, I would get quite overwrought, I’d get jittery.” She was “jittery” as she spoke but when I asked if she ever wished that she did not know that there was a test, she immediately said, “no, I think it’s a great breakthrough because of what it will lead to.” Thus for Naomi (and perhaps others who have not decided to request the test), knowledge of the existence of an accurate predictive test is both a chronic source of biographical uncertainty (Bury, 1982) and a promising source of hope for the future. On one level, the knowledge that there is a test means that there is a perpetual question mark beside the decision and on another level, it is reassuring to know of the test because it represents the possibility that scientists are closing in on some form of treatment if not cure.

Stories of having to know shed a different light on what it means to decide to request the test. Though the decision itself is, on the face of it, unplotted because there is no sequential development in having to know — it just is — these stories are, on a deeper level, anchored in relevant life experiences that enable the narrator to know, without a period of critical reflection, what it is they want to do. Helen talks about how her experience with being a caregiver contributes to her take charge attitude toward finding out about the test. As she says, “if I go in not knowing anything then I’m dead.” Many of these stories do, therefore, defy any narrowly psychologistic interpretation of personality as a causal influence. Personality is rooted in a life history of social relationships and experiences; it is not an immutable set of inner characteristics; it is a learned and practiced way of responding, an aspect of “habitus” (Bourdieu, 1990) that informs the feeling of decisiveness that is the defining feature of having to know. There is in this sense a story behind the decision rather than in the decision.

Stories of taking the decision pose some of the most profound questions about what it is
that allows at risk individuals to see that there is a decision to be made; they single out a moment in time as a significant turning point that has a before and an after. What happens in between may remain inchoate as in Regina’s story or it may be very specific and therefore open to critical reflection as in Brenda’s story. Hence the diversity that exists within these stories suggests that there is only so much of human experience that can be subjected to critical reflection; there is, in consequence, a limit to the degree to which PT participants are able to consciously articulate what happened and how it happened.

This leads me to a final point that has to do with the relationship between cultural expectations, life experience and the story lines that people adopt in telling others about their decision to request predictive testing. Earlier on I described some of the patterns in the narrators’ family history and awareness of HD that seemed to play an important role in shaping the selection and sequence of events that were featured in the each of the three types of stories that I heard. Without exception, those who told stories of having to know had a short duration of awareness and little or no firsthand experience of HD. There was, in these stories, very little in the way of an explanation for how the decision was arrived at. These stories were, on the face of it, unplotted. This may, as Helen’s story suggests, be indicative of a more generalized expectation that everyone will automatically get tested. Helen’s story was the strongest example of this but there were others that endorsed similar views. This pattern is in keeping with studies on women’s decision–making in prenatal screening; those who choose what they perceive to be the culturally expected alternative do not experience as great a need to justify their choice as those who believe themselves to be choosing the dispreferred option (Santalahti, et al., 1998). In any case, where this construction of the decision as a self–evident act is coupled with a short awareness of being at risk for HD, it ought to sound a cautionary note about the pace at which this relatively novel form of testing is becoming routinized. The belief that the test is the culturally expected alternative is prevalent amongst the HD community that resides in British Columbia and there is, in consequence, little awareness that the vast majority of individuals at risk for HD have not requested the test (Tolley, 1997).23 Though such cultural

23 Though I can hardly claim to make any generalizations about this, a quick polling of students in several of the classrooms where I have been invited to give a lecture on predictive testing for HD has shown that the vast majority tend to assume that most people at risk for HD would request the test and, moreover, that if they were at
expectations did not comprise the only source of certainty in stories of having to know, it is possible that such expectations will, in the future, comprise a much more significant aspect of the experience of those who have only recently learned that they are at risk for HD.

Where there is a long period of awareness of HD and firsthand experience of the devastating consequences of HD precedes knowledge of and/or access to the test, there is a degree of ambivalence about the decision to request the test. This ambivalence appears in stories of evolving toward it and, to some degree, stories of taking the decision and it may, as some service providers suggest, be important to cultivate. As Meissen et al (1991:409) wrote in one of the few published articles on understanding the decision to take the predictive test for HD, “research has repeatedly found that, once a psychological commitment is made, especially if it is shared with others, individuals tend to rationalize and justify their decision, which in turn strengthens their commitment.” Fearing that PT candidates will feel locked into their decision once a verbal commitment is made, Meissen et al stress that there should be adequate safeguards in place to ensure that PT candidates do not come to a premature decision to request the test and, moreover, that there is ample opportunity to reconsider and withdraw at any time.

The problem with this is, however, that many PT candidates arrive at the first counselling session with a firm sense of resolve about their decision to request the test (Meissen, et al., 1991). Many have been on a waiting list for several months before beginning the pre–test counselling and thus the wait itself may serve to strengthen such resolve. This was certainly the case in most of the stories that I heard and, as a result, there were very few PT candidates who situated their process of decision–making within the clinical context. The counselling sessions were generally considered helpful in that they provided PT candidates and their support persons with the opportunity to reflect on the decision and its implications (as well as prepare for the results session), but they were not seen as significant in shaping the decision itself. Especially where the PT candidate is well–informed about the test and its implications it may be very difficult for the genetic counsellor to revisit the making of the decision and hence, hold out the possibility of deferring the decision and/or making an

risk themselves, they too would want the test.
altogether different decision. What happens before PT candidates arrive at the clinic and after
they arrive home again is, therefore, especially critical to understanding how the decision is
made and, moreover, who has a say in its making.

The decision to request predictive testing is, by all clinical standards, supposed to be an
autonomous decision — that is, PT candidates ought never to feel coerced by family members,
their physician, life insurance company or any other person or agency. This is without question
a necessary and laudable objective of predictive testing. Nonetheless, the emphasis on
autonomy presupposes that which most needs to be examined — that is, how do PT candidates,
who are often extremely aware of and concerned about the implications of the test for others,
decide how to decide? Is there a schema for decision-making that provides a means of
weighing the personal desire to know (or not know) against the social responsibilities and
moral commitments that PT candidates feel toward others? Or, does this formulation of the
problem lean too much toward rational choice? emphasize too strongly the intentional character
of human agency? underplay the spontaneous unfolding of events in everyday life?

If there is one central lesson to be learned from the diverse ways that PT candidates and
their families story the experience of deciding to request the test it is this: the process of
deciding whether or not to request the test is seldom a completely conscious, rational or self-
interested process. Sometimes one’s life experiences and/or the exigencies of the situation seem
to point, without hesitation, toward one and only one course of action. As Helen suggests, these
moments re(k)new what we already know. Further, though decision-making is, as Regina
suggests, sometimes a matter of “weighing the pros and cons”, it is also a matter of heeding the
ephemeral voice of somebody somewhere saying that “it is time”. And though the decision to
have the test may make sense in light of one’s perceived responsibilities to others, it is as Colin
suggest, also a matter of arriving at a feeling of personal readiness. This is an embodied as well
as cognitive phenomenon. When you feel your heart sink — as Colin did when he was
unexpectedly asked if he would like to know the results of his neurological examination — you
are aware of things that cannot be fitted into the idiom of rational thought. It may well be then,
that we must cast the net very broadly indeed if we are to understand what it means to make a
decision about whether or not to request predictive testing.
CHAPTER VIII
TIL I GOT THE PIECES RIGHT

L: The thing I remember is cleaning the stove that I have downstairs in the TV room and taking the chimney out/ the connection to the flue/ and not being able to put it back properly. And that took my mind off it for probably half an hour/ til I got the pieces right again and the right ends.

S: And this is while you were still waiting to hear {Albert’s test results}?  

L: This is while I was waiting, yes... It was the only thing that I did in my busy, busy jobs that really engaged me for a little while and that/ I suppose that’s why it sticks in my mind. (F, not at risk, family member, 72 years, widowed, 2 children)

I observed only one results session.¹ My notes are sparse, detached, staccato. Albert was “exactly on time”. He was alone and when I glanced into the waiting area in the medical genetics clinic he was perched on the edge of his chair, half–standing, half–sitting. I looked at my watch. The doctor was running a few minutes late. The genetic counsellor said that Albert seemed “very nervous”. She asked if I knew whether it was possible to make a long distance call from the room where he would receive his results. I said I didn’t know.

I was thinking of Albert’s mother Lillian. She was at home waiting to hear from her son. During the pre–results interview she told me about how she and her husband (i.e., Albert’s father) went to the clinic with Albert when he received his linkage test results.² Her memories of the event were vivid.

L: We went on a Monday to get the results and he {Albert} had come home on Saturday afternoon. And the whole week before that I was playing sad music to make me cry: Operas, La Bohème³ mostly. It always makes me cry. So I wouldn’t have any tears left. You know how you cry and cry and cry? You run out of tears if you do it all the time.

S: And then you’re quiet and dry?

¹ There were limits on attending these sessions. It was necessary to have attended the pre–test counselling sessions and have the consent of the PT candidate. Some (understandably) did not want anyone other than the genetic counsellor and the doctor to be present. Further, genetic counselling students often had priority on clinic days.

² Albert requested the linkage test soon after it became available in 1987. When he received his results, Lillian’s husband (i.e., Albert’s father) had been diagnosed with Huntington Disease for about one year.

³ La Bohème is one of Giacomo Puccini’s (1858–1924) most heartrending operas. Set in Paris in the mid–nineteenth century, it is about the tragic love of the poet Rodolfo and the seamstress Mimi, both of whom live in poverty. They become separated because of Rodolfo’s jealousy only to be reunited when Mimi is on her death bed.
(...) Yes, you're dry, you're sort of numb inside. Well that's what I wanted to achieve and I did achieve it. So I didn't... I was just sort of stupid. In fact I said [to the doctor] "Oh, you mean he's 98% certain that he's going to get it?" [The doctor said] "NO, I didn't say that!" (laughing) I almost argued with him. But then it sort of came upon me, you know. I didn't connect with the ground anymore. It was sheer bliss. It was probably the happiest day of my life, even happier than my wedding day.

As I waited for the doctor to arrive for Albert's results session, I wondered what Lillian was doing, whether she listened again to La Bohème, if she felt the same need for numbness. I knew that she was somewhat skeptical about the accuracy of Albert's linkage test results and that she would not allow herself to anticipate "good news". "Misplaced confidence", she said, "always precedes a fall."

The doctor arrived, said hello to Albert and motioned to the genetic counsellor and me to follow him into the room where Albert would learn his direct test results. As soon as everyone was seated, the doctor said there was "good news". Albert would never get HD; he did not inherit the gene; the result was 100% certain. Save the possibility of a sample mix-up, the margin for error was "very, very, very tiny." Albert wrote all of this down. He said he wanted to remember exactly what the doctor said. He then asked to call his mother.

Lillian was busy cleaning her woodstove but when the phone rang she was overjoyed to hear the news. As she recalled during the post-results interview several months later,

I put the 1812 on (laughing) to celebrate. You know, the last/ when the Russians are winning and the bells are going and the cannons are going off. That's sort of triumphal music to me. That and Chopin's Polonaise (humming) "dum da dum, dah da da dah dum da dum." That's sort of happy music. "Gee, I'm great (. ) I've got a son who's not got Huntington's you know!"

The 1812 is a dramatic score and it was, I thought, an appropriate musical idiom for contemplating the implications of receiving a 'negative' test result. Learning that one of her two sons had not inherited the genetic mutation associated with HD was, for Lillian, a kind of liberation, a moment in which she celebrated having "a son who's not got Huntington's".

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4 There were four of us present at the session — Albert, the genetic counsellor, the doctor and myself.
5 The Russian composer, Peter Tchaikovsky (1840–1893), wrote the 1812 Overture to commemorate the seventieth anniversary of the burning of Moscow. After a devastating battle with Napoleon's Grand Army, Russian forces withdrew beyond Moscow allowing the French to enter the city. Napoleon had barely occupied the city when it went up in flames. Built almost entirely of wood, the city was destroyed, leaving only the great stone churches and palaces. There is much speculation about whether Russian patriots set fire to the city in order to drive Napoleon out. In any case, Napoleon's army was forced to begin a ruinous retreat in October of 1812 (Burr, 1965).
6 Lillian's husband (i.e., Albert's father) died from Huntington Disease one year before this interview.
For Albert, the news was somewhat anti-climactic. He was relieved but accepted the results quickly and, at the time of the post-results interview (four months after receiving his results), he explained to me that the test results seemed to have had very little impact on him. He had not made a move to improve his work situation nor had he established a “love life”. Things were just the same as before. Musing about the lack of change in his life, he said somewhat circumspectly, “I didn't justify the good news.”

**Purpose and Outline of Chapter**

Storytelling is an ongoing process of composition: like music, it transforms experiential chaos into coherent and decipherable forms (Gubrium & Holstein, 1998). This chapter is about how study participants storied their experiences of hearing and making sense of the results of predictive testing. It is, in short, about the intersubjective processes of interpreting and integrating the results within a temporal and evaluative framework. As the preceding story suggests, these processes may be as profound for family members as they are for PT candidates.

The material which informs this chapter is drawn from the post-results interviews conducted with PT candidates and their family members at four to eight months after clinical disclosure of results. There are several components which appear with differing levels of emphasis in most (if not all) study participants’ accounts. These include: 1) a description of what occurred at the clinic during the results session, 2) an evaluation of how it felt to hear the test result and how others present at the results session (i.e., the doctor, genetic counsellor and support persons) behaved and/or responded, 3) reflections on the process of making sense of the test results, their implications for self and others, 4) a chronology of telling (or not telling) family members and friends about the test results, and 5) a synthesis which places the results into a larger context and offers some sense of closure, however tentative or ambiguous.

The title of this chapter, “til I got the pieces right”, derives from the task that Lillian sets herself as she awaits news of Albert’s direct test results — that is, cleaning her woodstove. The task of reassembling the stove presents a puzzle; Lillian is uncertain about whether she has

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7 There were also six families who participated in a second follow-up interview at eighteen to twenty-four months after results. Where I refer to material from these interviews, it is specifically noted.

8 The latter two components are also taken up in the next chapter.
properly reconnected the chimney and flue, whether she has figured out "the right ends". The task is mundane but its gestalt\(^9\) offers a useful "generative metaphor" (Schön, 1993). I relate to it because I am, in this chapter, engaged in understanding how PT candidates and their families make sense of the test results within the context of their everyday lives. According to Gestalt theory, the patterned structures of the whole cannot be apprehended by examining each element in isolation nor can the specific properties of an integrated whole be understood without reference to the way in which its elements are interconnected (Frankl, 1984).

This somewhat abstract point and its implications for my approach to the subject matter of this chapter require some brief explanation. In writing this chapter, I am aware of an array of possibilities for configuring and reconfiguring the predominant themes which emerge from study participants' stories. The test result, the piece of information that is the crux of predictive testing is, paradoxically, the least helpful in this regard. It comes with too much baggage; it is granted altogether too much explanatory, even causal, power.

As mentioned in Chapter III, the test results are, within much of the published literature, treated as if they were an independent variable or stimulus that creates a range of measurable responses, some beneficial (e.g., enhanced ability to plan for the future), others adverse (e.g., increased anxiety or depression). Though this reductionist approach has a certain utility in survey–based studies which seek to describe and predict generalizable patterns of response to clinical interventions (such as predictive testing), it is inappropriate to an interpretive, meaning–centred approach. It neglects to consider how study participants’ familial relationships, communicative interactions, biography, life trajectory, and access to material, social and cultural resources shape the possible meanings an informative test result may have. In addition, this approach obscures the way in which the expectations and interventions of clinicians and genetic counsellors inevitably render some story schema more available to PT candidates and their families than others. The task of understanding the interpretive processes through which PT candidates and their families make sense of the results is, therefore, narrowed and flattened by the predominant focus on the observable outcomes and/or measurable consequences of the test.

\(^9\) The Gestalt experience is the "Aha!" moment wherein "order is reordered" and "knowledge becomes recognition" (Wilden, 1987:249, emphasis added).
These consequences are not insignificant but I want to plot another course here, work out how to surface and tell other kinds of stories, stories that are glossed over when the test result is privileged above all else. I want to resist, as many of my study participants themselves do, the idea that it is possible to isolate and catalogue the specific 'effects' of the information provided through predictive testing and subsequently assess the impact 'on' PT candidates and their families. It is the way in which this information is integrated within familial relationships and the array of events that comprise a life story that matters here.

The test results and the process of predictive testing — like the chimney and flue — have no inherent meaning or utility when they are disconnected from the context of everyday life: they must be positioned in relation to a flow of events and situated within a purposive, evaluative framework (Gergen & Gergen, 1983). This positioning occurs in and through the act of storying the experience of predictive testing; it is, therefore, not a solitary enterprise. It is socially negotiated and contingent upon the formative relationships which exist between clinicians, PT candidates and their families and, between researchers and research participants. Such relationships enter the narrative because the actions of others are events within the narrative and because emergent narratives undergo modification as they are told to others and thereby subjected to social evaluation. Indeed, whether a given narrative can be told at all depends at least in part upon the narrator's ability to negotiate, with a specific audience, the meaning of events in relationship with each other (McKellin, Cox, & Smith, 1995). Such processes of negotiation also point to the issue of narrative ownership and whose voice is heard when the story is told to others within a particular setting: as Gubrium and Holstein (1998) suggest, occasions may "own" stories as much as people do. The central point is, then, that narratives should not be treated as if they have an existence quite independent of the context in which they emerge and/or the audience to which they are directed.

This chapter therefore attempts to do (and undo) several things. In the first section, I summarize the test results for all PT candidates involved in this research showing how the predominant focus on outcomes must be reoriented toward process if we are to understand the various ways study participants make sense of, and story, the experience of predictive testing. This is followed by a discussion of the language that is employed during the clinical disclosure
of results. An appreciation of the problems inherent to this language is, as we shall see, critical in grasping how PT candidates and their families interpret and talk about the test results.

In the second section of the chapter, I consider how study participants’ perceptions of the normative expectations of the clinic shape the experience of disclosure. Here, I draw upon a series of exemplars which show how PT candidates and their families adopt and modify available schema for making sense of the test results. The overarching dynamic that emerges here has to do with the narrative significance of ‘not.’ As we shall see, the test results do not determine a life story or trajectory; moreover, PT candidates do not bluntly accept the clinically available story line in making sense of their results. As in the preceding chapters, I emphasize here the need to attend to both the content and form of the stories. This applies not only to the stories that study participants tell but also, at a metalevel, to the story that I, as narrator, now tell about the stories that I have heard.

The Test Results

My point of departure is to acknowledge an obvious constraint on the story that I am able to tell — that is, I cannot think through my study participants’ stories without also thinking through the categories which now inscribe them. This person is a gene carrier and this person is not; this spouse/partner will likely become a caregiver for someone with HD while that one will probably not. Everything seems to coalesce around this knowledge. It is not possible for me to unknow the test results and attend to study participants’ stories as if I were naive, though I now wonder what I might find if this was possible. This abiding loss of ignorance is, then, a source of constraint as well as insight. It muffles my ability to hear some aspects of what I was told and, no doubt, amplifies my attentiveness to others.

From Outcome to Process

During the results session, PT candidates learn that they have either inherited the mutation associated with HD or they have not inherited the mutation associated with HD. In addition, a very small number (2 to 3%) learn that they have an intermediate allele (with a CAG repeat size of 30 to 36) (Benjamin, et al., 1994). To the at risk individual who, by virtue of their participation in predictive testing, asks “will I eventually develop onset of HD?” the clinician
might therefore reply: 1) “yes, provided you live long enough” or 2) “no, definitely not” or, less commonly, 3) “maybe you will and maybe you won’t, we cannot be absolutely certain”.

When participants in this research received their test results (i.e., 1994) it was not routine practice to disclose the CAG repeat size to the PT candidate. The primary reason for non-disclosure was the avoidance of potential harm: specifically, clinicians and genetic counsellors were concerned that PT candidates might misinterpret the personal significance of the statistical correlation between repeat size and age of onset (Benjamin, et al., 1994; Burgess & Hayden, 1996). Exceptions were, however, made for PT candidates who learned that they had an intermediate allele and/or a CAG repeat length that was just within the affected range (i.e., 36 – 38) as clinicians felt that these individuals needed to understand more fully the ambiguities and potential implications of such a result for themselves and their offspring and/or siblings.

Table 10 (see next page) shows the direct test results, by gender, for each of the PT candidates participating in this research. Six female and four male test candidates learned that they did not inherit the mutation, while four female (and no male) candidates learned that they had inherited the mutation. In addition, there were two female test candidates who had an intermediate allele: one had a CAG repeat number of 35 and was counselled that it was very unlikely that she would ever develop HD. The other had a CAG repeat of 36: this result was just inside the range where it was possible that if she lived well into old age she might develop HD.

In brief, there were in the sample of PT candidates described here, approximately twice as many PT candidates who learned they did not inherit the genetic mutation associated with HD as learned that they did inherit the mutation. This 2:1 ratio is consistent with trends reported in many countries where linkage and direct forms of predictive testing have been or are currently offered (Adam, et al., 1995; Bloch, et al., 1989; Brandt, et al., 1989; Codori & Brandt, 1994; Evers–Kiebooms, 1989; Fox, et al., 1989; Holloway, et al., 1994; Meissen, et al., 1991; Quaid & Wesson, 1995; Tibben, et al., 1997; Wiggins, et al., 1992).11

10 For the majority of individuals with repeat sizes of more than 36, the 95% confidence intervals for age of onset are very broad: hence the information is not useful in predicting individual age of onset (Brinkman, et al., 1997).

11 The 2:1 ratio is counter-intuitive in that one might expect that with an autosomal dominant disorder there would be a 1:1 ratio of each test outcome. Recall, however, that many PT candidates have surpassed the average age of onset at the time of testing and thus their risk has begun to dip below 50%.
TABLE 10
DIRECT TEST RESULTS BY GENDER
(SELECTED STUDY SAMPLE)

Total Number of Predictive Test Candidates = 16

<table>
<thead>
<tr>
<th></th>
<th>Does Not Have Gene (CAG &lt; 30)</th>
<th>Intermediate Allele (CAG 30–36)</th>
<th>Has Gene (CAG &gt;36)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female</td>
<td>6</td>
<td>2</td>
<td>4</td>
</tr>
<tr>
<td>Male</td>
<td>4</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td></td>
<td>10</td>
<td>2</td>
<td>4</td>
</tr>
</tbody>
</table>

TABLE 11
MODIFICATION OF RISK THROUGH DIRECT TEST
(SELECTED STUDY SAMPLE)

Total Number of Predictive Test Candidates = 16

<table>
<thead>
<tr>
<th>DIRECT TEST RESULT</th>
<th>Does Not Have Gene (CAG &lt; 30)</th>
<th>Intermediate Allele&lt;sup&gt;a&lt;/sup&gt; (CAG 30–36)</th>
<th>Has Gene (CAG &gt;36)</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Apriori risk</td>
<td>Linkage test result</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>25%</td>
<td>-</td>
<td>2</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>50%</td>
<td>-</td>
<td>6</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>50%</td>
<td>Decreased Risk</td>
<td>2</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>50%&lt;sup&gt;b&lt;/sup&gt;</td>
<td>Increased Risk</td>
<td>-</td>
<td>1</td>
<td>-</td>
</tr>
<tr>
<td>50%&lt;sup&gt;b&lt;/sup&gt;</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>2</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>10</td>
<td>2</td>
<td>4</td>
</tr>
</tbody>
</table>

<sup>a</sup> Intermediate range was 30 to 36 repeats when research participants received results (1994).

<sup>b</sup> Appeared to be symptomatic, based on my observations at the time of the pre–results interview.
What is striking and, from a researcher’s point of view highly serendipitous, is the presence of two PT candidates who learned that they had an intermediate allele. As mentioned above, intermediate alleles are relatively unusual: in 1994, the Canadian Collaborative Study of Predictive Testing for Huntington Disease reported that only 2 of 209 direct test results provided to at risk, asymptomatic individuals showed a CAG repeat length of 30 to 36 (Benjamin, et al., 1994). Since then, it has been suggested that the CAG size of 30 to 35 need no longer be considered part of the indeterminate range. There is no evidence of anyone manifesting with HD who has a CAG repeat of less than 36. There is, however, an increased chance that offspring may inherit an expanded allele particularly where the transmitting parent is male (Brinkman, et al., 1997). These uncertainties make it more difficult to interpret the significance of the information derived through predictive testing. They add a complexity that is often overlooked when the test results are treated as dichotomous. As such, this research is greatly enriched by the experiences of the two PT candidates and their respective family members who shared with me their struggle to interpret the meaning and significance of having an intermediate allele.

Bracketing, for a moment, the definitiveness of the test results, it is important to note that focusing exclusively on the test outcome glosses much of the complexity inherent to predictive testing as a process of risk modification. Even on an objective level, it lumps together too many different kinds of experiences, too many different journeys. It is equivalent to saying that the only thing that matters is your destination: where you started, how far you travelled and, indeed, whether you have made such a journey before are of little or no consequence.

Albert was one of several PT candidates who stressed the importance of understanding predictive testing as a process. Albert obtained an informative result from linkage testing soon after it was first offered and was told that there was a 98% certainty that he would never develop

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12 This figure does not include the two PT candidates who were participants in this research.

13 Recent data on the relationship between repeat size and age of onset, along with earlier findings concerning the phenomenon of anticipation and the effect of sex of origin on severity of illness (Hayden, 1993), suggest that the genetics of HD are more complex than originally thought. Juvenile onset occurs more often with paternal transmission. This phenomenon was first reported in 1968 and the finding has since been observed in HD populations from all over the world (Harper, 1991; Hayden, 1981). The trend appears to defy Mendelian patterns of inheritance but the sex-dependent effects of major expansion and contraction of the CAG repeat in the HD gene have been linked to differential processes of gametogenesis in males and females. As such, the offspring of affected fathers are more likely to have large expansions than the offspring of affected mothers. Conversely, the offspring of affected mothers are more likely to show no change or a reduction in CAG repeat size (Kremer, et al., 1995).
HD. Unlike his mother, Albert sustained a high degree of confidence in these results. Though he did not want to be complacent, he more or less expected to hear "good news" when he received his direct test results. When I asked what he recalled about his results session, he said:

Only just waiting for the doctor to come... And then wham! good news, but (sigh) (...) expected. It makes a difference going from a 50:50 [chance of having inherited the mutation] to a 100% {certainty}, as opposed to going from a 98% to a 100% {certainty}. (M, 50% risk, PT candidate, 46 years, single, 0 children)

Albert understood that direct testing offers a greater degree of certainty than linkage testing, but he did not distinguish between the type of information each form of testing provides. He also had an affinity for numbers and often framed things in quantifiable terms. As such, he conceptualized the modification of risk that occurs with an informative test result as if it were a distance that must be traversed: the greater the potential modification of risk (i.e. from 50 to 100% rather than 98 to 100%), the longer the journey and the higher the emotional impact.

Albert's schema draws upon a coherent system of orientational metaphors. These "metaphors we live by" (Lakoff & Johnson, 1980) provide an interpretive resource which effectively conveys a sense of how the modification of risk is experienced as a spatialized and embodied phenomenon. In this system of orientational metaphors, there are two sources of movement through a time–space continuum. First, the test result may be conceptualized as an object which gathers momentum as it travels through space; the further it travels, the greater the momentum and hence, the greater the potential impact. Second, the PT candidate may, like Albert, think of themselves as moving toward or away from a particular result. Once again, however, the same system of orientational metaphors applies: the further the PT candidate must travel, the greater the potential impact of the journey.

Albert said that he didn't do anything special to celebrate his direct test results "because it was a 98% to a 100% there was nothing in particular, just the phone call to my mother and that was good enough." But, when I asked how he might have responded if he had received "other news", he leaned back in his chair, looked at the ceiling and said,

Ooooh, yes. That would have been worse. That would be going from 98% good news to 100% bad. A long way. Oh yes. Oh right. It doesn't bear thinking about.

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14 See Cox and McKellin (in press) for a discussion of how at risk individuals draw upon this system of orientational metaphors to describe the fluctuating relevance of hereditary risk over the life course.
Later in the interview I asked Albert to compare his experiences of the linkage and direct test.

I was going from 50% to 98% [with linkage testing]. There was a real issue then. And I had palpitations going in for the news. But this time it was a very small gap... It was a very intellectual thought. It didn't affect me emotionally that it could go the other way.

If, as Albert suggests, we look at the transitions between one level of risk and another, it becomes important to know each PT candidate’s a priori risk and whether they have already had linkage testing. This permits a more nuanced understanding of the objective change and/or transition that occurs through direct testing. For some PT candidates (such as Albert), the magnitude of the shift in objectively measured risk also offers considerable insight into the subjective dimensions of the experience.\(^{15}\)

Each of the transitions or risk modifications pertinent to the PT candidates who participated in this research is shown (above) in Table 11. As the table demonstrates, the emphasis on predictive testing as a process of risk modification yields an array of seven different transitions between various risk categories and test outcomes — for instance, reading from left to right, the first row describes the shift from being at 25% a priori risk to knowing with near 100% certainty that there is virtually no possibility of developing HD.

Of the thirteen PT candidates who had not had linkage testing, two (i.e., Helen and Brenda) began the process of direct testing with an a priori risk of 25%. Both defined themselves as having only a minimal risk (in comparison to a “real 50:50”) and both learned that they did not inherit the mutation associated with HD. The remaining eleven PT candidates began with an a priori risk of 50%: of these, six (i.e., Merle, Colin, Nigel, Maggie, Adam, and Landis) learned that they did not inherit the mutation, four (i.e., Carla, Rose, Marie, and Regina) learned that they did inherit the mutation and one (i.e., Gabriella) learned that she had an intermediate allele with a CAG repeat size of 36.

There were three PT candidates who began the direct test after having already received

\(^{15}\) This is not to suggest that Albert’s schema has subjective relevance for all PT candidates. There was one woman I interviewed who learned from her relatives of an instance of non-paternity in her family. If her facts were correct (and she believed they were), there was no chance that she had ever been at risk for HD. She did not, however, obtain this information until after she received her test results and, when I asked how she felt about having been through what was perhaps an unnecessary ordeal, she stressed that when it comes to being at risk for HD “there’s not much difference between never was and am not.” She did not need to worry in either case and hence, the outcome was all that mattered to her. She had no inclination to resolve or even delve into the paternity issues as she felt they were a family secret better left undisturbed.
an informative result from prior participation in linkage testing: the two who obtained a
decreased risk (i.e., Albert and Heather) confirmed, through the direct test, that they did not
inherit the mutation while the one who obtained an increased risk (i.e., Rosalind) found, through
the direct test, that she had an intermediate allele with a CAG repeat size of 35. Given what is
now known about this repeat size, her direct test result was, in effect, a risk reversal — that is,
she went from having a high probability of eventually developing HD to a very small, even
negligible risk of ever developing HD.

As Rosalind’s story demonstrates, however, it is not always possible to predict from
such objective measures how the results will be intersubjectively evaluated. Rosalind viewed
her CAG repeat of 35 as a number “to stay away from”. Despite the fact that this news was a
risk reversal for her (and hence ‘good’ news) Rosalind said, “it was not the number we wanted
to hear.” Though Rosalind knew that her son had not inherited an expanded allele (because he
also had predictive testing), she worried that her son’s children had inherited an unstable allele
and that they might eventually develop HD and/or pass it on to their children. To Rosalind, the
number 35 was, therefore, “like a death in the family, the death of a close person.”

Thus, while I wish to stress that it is helpful to be aware of the objective change in risk
that occurs with an informative test result, the abstract scientific language of risk modification
cannot be mapped directly onto the complex intersubjective realities which characterize lay
assessments of risk. As Gifford (1986:230) proposes more generally, lay risk is “not objective,
cannot be quantified or measured, and is not static,” it “must be understood as a dynamic
experience of personal uncertainty.”

The Language of Disclosure

All but one of the PT candidates participating in this research agreed that I should learn
their results through the genetic counsellor.¹⁶ This arrangement simplified things for all
concerned: it alleviated any responsibility that PT candidates might have felt for contacting me

¹⁶ I asked each PT candidate what they would prefer at the close of the pre-results interview. One man said that he
would prefer to tell me his test result himself. As he later explained, this was because he wanted to be certain that
no-one learned of his test result before he did. When he learned that he had not inherited the mutation he told the
counsellor that she could inform me of his test result.
to let me know their results; it also meant that I was aware of the results when calling to schedule the post–results interview and that I would not ask. As such, the post–results interviews were conducted within an open awareness context (Glaser & Strauss, 1965).  

An appreciation for the language of disclosure is, nonetheless, integral to my analysis of how PT candidates and their families talk about the meaning and significance of the test results. As speech–act theorists such as John Austin (1962) assert, utterances may be treated as expressions of a speaker’s communicative intention but they may also be seen as acts which do more than merely inform. Depending upon the speaker’s and the listener’s shared conventions for communicating meaning, utterances may promise, warn, request, or perform a ritual function as in the act of diagnostic labelling. Further, the language of disclosure shapes and constrains the way in which PT candidates and their families interpret the information acquired through predictive testing. This is not to suggest that the language of disclosure is deterministic; it is rather to reiterate the importance of viewing language as both a symbolic resource and a “medium of practical social activity” (Giddens, 1993:25).

The production of sense in communicative acts (such as the clinical disclosure of test results) is often taken for granted but it is, as Giddens (1993:25) suggests, the “skilled accomplishment of actors”. This skill does not reside in the ability to put one’s thoughts and feeling ‘into’ words in order to effectively ‘convey’ them to others. As Reddy (1993:186) argues, this way of talking about human communication is both misleading and dehumanizing: it “encourages us to talk and think about thoughts as if they had the same kind of external, intersubjective reality as lamps and tables.” Further, the expenditure of effort that is involved is, in this “conduit” model of communication, localized almost entirely in the speaker (or writer).

17 I spoke with one PT candidate at her two–week follow–up session. I knew that she had inherited the mutation but she was unsure whether I knew; she immediately disclosed her result to me when she saw me. I also received two telephone calls. The first was from a PT candidate who phoned soon after her results session to let me know that she had not inherited the mutation. The second was from the father of a PT candidate: he was overjoyed to hear that his son had not inherited the mutation and called me right away to share this news. I was, unfortunately, not at home for either call, although both left messages on my answering machine.

18 The only exceptions to this occurred with two post–results interviews with family members. Both are unidentified here for reasons that will soon be apparent. The first misheard a family member’s test results and believed this person to have inherited the mutation when, in fact, they had not. The second was unaware of a family member’s results at the time of the post–results interview. In the first case, I wanted to reveal the actual test result since it was, after all, ‘good’ news. In the second, I was surprised to find the study participant totally unconcerned with finding out the test results. In neither case did I disclose the respective PT candidate’s results nor did I knowingly alter their respective family members’ awareness.
The active engagement of the listener (or reader) is therefore trivialized.

The radical subjectivist paradigm, on the other hand, makes it clear that readers and listeners face a difficult and highly creative task of reconstruction and hypothesis testing. Doing this work well probably requires considerably more energy than the conduit metaphor would lead us to expect (Reddy, 1993:168).

Following Reddy, my point is that PT candidates and their families are not passive recipients of information that is ‘disclosed’, or even more misleadingly, ‘given’ to them during the results session. As listeners, they must engage in the demanding task of reconstructing and testing an array of possible meanings. They must make sense of what they have heard. From this perspective, it is intriguing to examine how the language of clinical disclosure creates confusion (about the intended meaning) and resistance (to its implied significance).

Many PT candidates and their families found it difficult to frame their experiences in terms of a bifurcated set of test outcomes. The social meanings of ‘negative’ and ‘positive’ results or ‘good’ and ‘bad’ news, are neither simple nor absolute yet the language of disclosure gives the impression that such categories are both mutually exclusive and exhaustive.

Given the finding that some PT candidates have difficulty adapting to the news that they have not inherited the mutation associated with HD (Huggins, et al., 1992), service providers have become more cautious about framing the test results in terms of ‘good’ versus ‘bad’ news. The social meanings of the knowledge that is derived through predictive testing are never quite this bald. The terms ‘negative’ and ‘positive’ are therefore attractive because they hold out the possibility of a value-neutral alternative. Further, within the lexicon of medical genetics at least, these terms have very specific meanings: a ‘negative’ test result indicates the absence of a genetic anomaly while a ‘positive’ result indicates the presence of such an anomaly. This dichotomy is, however, inherently problematic for several reasons. First, it sets up an opposition between two statements that are not of the same logical type. Second, it oversimplifies current scientific knowledge and confounds lay actors in their efforts to understand the genetics of HD. And, third, it inverts commonsense ways of thinking about ‘good’ and ‘bad’ news.

19 The possibility is, at best, theoretical. Clinicians, like their patients, communicate through a variety of verbal and nonverbal channels and, as several PT candidates noted in the post-results interviews, it was not difficult to tell from the facial expression, body language, and intonation of the doctor and/or genetic counsellor that there was ‘good’ or ‘bad’ news.
Let us first consider how the logic of this dichotomy sets up a false opposition. A 'positive' result denotes the presence of something unusual while a 'negative' result denotes an absence. The two test results are, therefore, of a different logical type: the former asserts while the other negates. To see the significance of this, we must briefly consider the syntactic significance of the word 'not'. 'Not' is a uniquely linguistic device which achieves its effect solely by virtue of its placement within an utterance or statement. As a “communication about an affirmation”, 'not' is, therefore, “not of the same logical type, or level of communication, as the other words in the sentence in which it appears” (Wilden, 1987:245). The syntactic ‘not’ as it appears in the statement ‘you have not inherited the mutation associated with HD’ is a metastatement about a statement at another level. In making sense of the metastatement it is essential to refer to the possible meanings of the statement.20

What is at issue here is the process of making a symbolic distinction between the ordinary or normal (the “unmarked”) and the special or abnormal (the “marked”).21 Each category informs the other and draws its distinctive meaning from its positioning within a particular context. The meaning of symbols thus derives not from their own inherent properties but from the way in which they are contrasted in our minds with other symbols. As Zerubavel (1997:73) suggests,

In order to fully understand the meaning of a symbol we must transcend the narrow confines of a strictly semantic analysis and consider also the syntactic context within which it is structurally embedded (that is, the way it is semiotically contrasted in our minds with other symbols).

The inseparability of syntax and semantics is particularly evident with the language of disclosure when the contrasting relations between each of the terms thus far discussed are depicted in a “semiotic square” (Zerubavel, 1997:73–4) (see Figure 3, next page). For example,

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20 The Huntington Society of Canada pamphlet (see Chapter II) offers a cogent illustration of the logic of this sense-making operation: the slogan ‘He’s Not Drunk’ rules out a plausible explanation for stumbling and slurred speech and then inserts an alternative, ‘He has Huntington Disease.’

21 Within linguistics, the study of “markedness” calls attention to certain types of asymmetries within categories. As Lakoff (1987:60) suggests, such asymmetries designate “one member or sub-category as somehow more basic than the other (or others).” Such “prototype effects” challenge classical theories of categorization because they reject the idea that there is a direct symbol-to-object correspondence that defines meaning in general and, instead, suggest that “human categorization is essentially a matter of both human experience and imagination — of perception, motor activity, and culture on the one hand, and of metaphor, metonymy, and mental imagery on the other. As a consequence, human reason crucially depends on the same factors, and therefore cannot be characterized merely in terms of the manipulation of abstract symbols” (Lakoff, 1987:8, emphasis added).
**FIGURE 3 — SEMIOTIC SQUARE: THE MARKED AND THE UNMARKED**

+ semantic association
– syntactic contrast

<table>
<thead>
<tr>
<th>Marked</th>
<th>Unmarked</th>
</tr>
</thead>
<tbody>
<tr>
<td>a. positive</td>
<td>a. negative</td>
</tr>
<tr>
<td>b. abnormal</td>
<td>b. normal</td>
</tr>
<tr>
<td>c. presence</td>
<td>c. absence</td>
</tr>
</tbody>
</table>

(-) (+)

<table>
<thead>
<tr>
<th>Marked</th>
<th>Unmarked</th>
</tr>
</thead>
<tbody>
<tr>
<td>a. bad</td>
<td>a. good</td>
</tr>
<tr>
<td>b. sickness</td>
<td>b. health</td>
</tr>
<tr>
<td>c. something</td>
<td>c. nothing</td>
</tr>
</tbody>
</table>
within the discourse of medical genetics, a ‘positive’ result is semantically associated with ‘bad’ news and syntactically contrasted with a ‘negative’ result. Further, this association can only be appreciated within the context of the homologous association between a ‘negative’ result and ‘good’ news. Here it is important to note that I am not pointing to any inherent properties of language so much I am to “the unmistakably sociomental connections between symbols and the particular thought community that uses them”; “the meaning of symbols is a property of the way they are socially used” (Zerubavel, 1997:78).

Temporarily bracketing these sociomental associations, let us now consider how the ‘positive’ versus ‘negative’ dichotomy oversimplifies current scientific knowledge of the genetics of HD. This is one of several problems that ultimately contradicts the tidy relations of presence and absence which are depicted in Figure 3. Here a ‘negative’ test result means that the PT candidate has inherited two normal alleles while a ‘positive’ result means that there is at least one ‘abnormal’ (i.e. expanded) allele. Common ways of speaking about the test results obscure this understanding: saying that someone ‘has the gene for HD’ or, more correctly, that someone has inherited the genetic mutation associated with HD, neglects the fact that everyone has, at the same locus on the tip of both copies of chromosome 4, a stretch of DNA that is comprised of a series of CAG repeats. The absence of the mutation is therefore not (as in Figure 3) ‘nothing’; It is ‘something’ but that something (i.e., a normal allele) may be taken for granted by service providers and therefore not included as a routine part of disclosure.

The association between ‘normal’ and ‘nothing’ creates the potential for confusion. In this research, such confusion was especially pronounced for those who struggled to understand the implications of having an intermediate allele. For instance, Rosalind’s husband Erik assumed that not having the mutation associated with HD meant that there were not any CAG repeats on the tip of chromosome 4. In consequence, Erik thought that a “good” result would have to be “a zero”. This misunderstanding was exacerbated by the fact that he and Rosalind were provided with only one CAG repeat number, a 35, during the clinical disclosure of her test.

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22 I am especially grateful to Rosalind, Gabriella and their respective family members who taught me much about how difficult it is to make sense of an ambiguous result (i.e., an intermediate allele) and how important it is to attend closely to the language of disclosure.
results: the number of CAG repeats in the other (i.e., her ‘normal’) allele was unreported and thus it seemed to Erik that he was correct in assuming that it must be zero.

Jason also had difficulty with the way the results were framed, though for different reasons. His mother, Gabriella, learned that she had a repeat size of 36 and, when Jason tried to explain the concept of an intermediate allele to a former girlfriend, she objected to the ambiguity. Noting that we “like things that are final, like you have it or you don’t”, Jason asked,

Would it be appropriate to suggest they change the wording? Because we do all have the Huntington's gene, we just have it in different forms. (M, 25% risk, family member, 27 years, single, 0 children)

Jason started “talking about it differently” when he learned that his mother had an intermediate allele. It became important to clarify to others that they also had a series of CAG repeats on chromosome 4 and that the distinction between having and not having the gene was quite subtle.

...you've got pretty well all the same bits of DNA, you're just missing a CAG repeat and that changes the complete nature of that gene? Well (.) I mean I think people would agree that it doesn't. You still have basically the same thing it's just slightly different.

The social consequences of labeling, as absolute, a difference that is, perhaps, better understood as a continuum, can be profound. As Jason suggests, our ways of speaking about the gene as an isolated entity that one either does or does not have are a means of constructing difference rather than similarity; they emphasize a dichotomous distinction between those who have and those who have not inherited an expanded allele.

If such categorical distinctions based on genotype seem arbitrary to Jason, it is in part because he cannot ‘see’ where the difference lies. Distinctions based on phenotype are possibly less problematic though here too caution is advised since lay knowledge does not always distinguish between the two (Richards & Ponder, 1996). When Colin learned that he had not inherited the mutation associated with HD, he considered sharing this news with his older brother (who had been diagnosed with HD) but then decided against it. As Colin said,

...I don't tell him about vacations or things that I've done that much cause I just don't want/ I'm afraid that's a reminder to him that you know/ of what he's missing. And so I guess for the same reason I don't really want to tell him, you know, that he received and I didn't. (M, 50% risk, PT candidate, 41 years, married, 3 children)

Colin's framing of the difference between himself and his brother — that is, “he
received and I didn't" — points to yet another problem with the language of disclosure, that is the contradiction which emerges from the competing clinical and everyday frameworks for representing relations of presence and absence. In Figure 3, the abnormal allele is semantically associated with sickness while the normal allele is associated with health [b]. For Colin, however, the presence of sickness denotes the absence of enjoyment and fulfillment in life. The two frameworks are therefore contradictory when situated within the context of everyday life.

The clinical–scientific and everyday evaluative uses of the terms ‘negative’ and ‘positive’ offer a more familiar instance of the same type of contradiction. As Rapp (1994:7) suggests, genetic counsellors lay special claim to words by inverting common–sense understandings: “a ‘positive family history’, for example, is anything but, as it refers to the presence of a serious, genetically transmissible condition.” This particular inversion of everyday language poses a quandary. As Rothman (1986) found in her study on women’s experiences of amniocentesis, many women said they worried about getting a ‘negative’ result when, in fact, they were worried about what their doctors would refer to as a ‘positive’ result.23

I encountered the same difficulty in this research. Study participants rarely succeeded in using the terms ‘positive’ and ‘negative’ to refer to the test result without struggling to clarify their intended meaning and frame of reference (i.e., clinical or everyday).24 As Lillian said toward the end of the post–results interview,

> It must be very difficult to deal with people who get a negative result/ who get a/ I mean who get a bad result/ uh?/ I don’t know, it must be one of the most difficult things in the world to tell them.

The inevitable slippage that occurs between clinical and everyday uses of the terms ‘positive’ and ‘negative’ also had a marked effect on how PT candidates heard and tried to comprehend the initial disclosure of their test result. As Merle recalled,

> I was sort of in shock cause I was/ I had to really think about it. Negative means

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23 Rothman avoided such confusion in her study by adopting her research participants’ point of view. Instead of referring to the results of amniocentesis as “negative” or “positive”, she asked the women she interviewed about the implications of “good” and/or “bad news”. This, however, had the effect of both clarifying and perpetuating the problem of how people interpret their results. Rothman’s study participants knew what she meant but as Rothman (1986: 160) herself suggests, there is ultimately no simple or unambiguous distinction between good and bad.

24 Widespread public awareness of HIV/AIDS has, perhaps, had an impact here: as a recent campaign promoting AIDS awareness illustrates, polysemic phrases such as ‘HIV positive’ are effective because the play between different meanings deconstructs taken for granted assumptions. The intended message was, of course, that it is possible to adopt a positive (i.e., accepting and constructive) stance toward being HIV–positive.
no. Like I couldn’t think of “I don’t have it.” I was thinking negative means no. No means I don’t (...) you know? Like I had to go through the whole process of “I don’t have it.” (F, 50% risk, PT candidate, 48 years, married, 2 children)

Listening to study participants talk about the test results, I also felt challenged to hear the intended meaning. Not only was the language riddled with inversions, it was overlaid with another type of complexity that had to do with what was being positively or negatively evaluated. As Denise, a close friend of the PT candidate Regina, said in reference to her expectations about how the results session would go,

...either way/ whichever way it turns out/ it’ll be a relief and that’s the way I’m looking at it. Like it’s positive even if it’s negative (laughing) (...) y’ know what I mean? (F, not at risk, family member, 44 years, married, 2 children)

Denise is saying that it will be good to know the results even if the results themselves are not good (i.e., her friend has inherited the genetic mutation associated with HD). Her meaning is not immediately apparent, however, for two reasons. First, we as listeners (or readers) do not immediately know whether Denise is invoking the clinical or everyday understandings of the word ‘negative’. Second, Denise uses the word ‘it’ to refer to and evaluate several things: a) the actual test result, b) the state of being informed (i.e. having the test result) and, c) completion of the process of predictive testing. As such, the listener must perform a kind of “double double unthink” (Bush, 1983) in order to grasp the sense of what she is saying. This requires running through a mental checklist of possible meanings, some nonsensical or even paradoxical, others possible and perhaps directly indicated to the competent listener by virtue of the context of the conversation (Grice, 1989). It is, then, no wonder that Denise asks if I know what she means!

Such moments are more than routine instances of the way in which speakers must cooperate in order to accomplish meaningful conversation (Grice, 1989): as I wish to suggest here, these moments present researchers interested in understanding the social meanings of genetic information with a useful heuristic. They are “rich points” or problematic bits of language that are, as Agar (1994:100) says, “putted thickly into far-reaching networks of association and many situations of use”. They create confusion, contradiction and ambiguity and, hence, like Garfinkel’s (1967) breaching experiments, they make tangible the taken for granted knowledge and skill that are required to produce communicative sense.
(Not) What They’re Looking For

Thus far, I have shown how viewing the test result as an outcome neglects the processual nature of predictive testing. I have also described some of the problems inherent to the language of disclosure: even the ostensibly value-neutral terms ‘positive’ and ‘negative’ are, when situated within the context of everyday life, freighted with ambivalent meanings. Finally, as Denise’s comments so amply demonstrate, talk about the results must be parsed into several distinct objects (i.e., the actual test result, the state of being informed and, completion of the process of predictive testing) each of which may be evaluated somewhat differently. I now expand this discussion to incorporate one further set of issues central to understanding how study participants narrated their experiences of receiving and interpreting the test results — that is, the ways in which PT candidates, in particular, engaged in making sense of the process and outcome of predictive testing by drawing upon various interpretive resources and relevant life experiences. Herein we shall meet the syntactic ‘not’ on another, more explicitly “dramatistic” level (Burke, 1989).

One of the most striking themes to emerge from analysis of the post-results interviews concerned the way in which PT candidates, in particular, engaged in making biographical sense of the process of predictive testing in light of what this experience was not. ‘Not’ is, as we have already seen, a peculiarly linguistic resource which achieves its effect by making a metastatement about an assertion. Syntax is crucial in this as the semantic significance of the metastatement varies according to the positioning of ‘not’ in a sentence or utterance. Saying what something is not, is for instance, different than not saying what something is. Though I now wish to consider the significance of ‘not’ as a narrative stratagem which elucidates much about how PT candidates and their families make sense of the process and outcome of predictive testing, the same general rule applies. The syntax of a life story matters because it is the context within which the narrative ‘not’ takes on a particular meaning and significance.

To see how this dramatistic emphasis on the act of negation differs from the propositional negative that we have thus far been engaged in examining, let us first briefly consider the range of assertions that the narrative ‘not’ may be uttered (or not) in response to. Equipped with this awareness, we will be better-positioned to hear the presence and narrative
significance of 'not' within study participants' stories. The first sort of assertion is one that we have already encountered within the preceding discussion on the language of disclosure — that is, the statement 'you have inherited the mutation associated with HD.' None of the four PT candidates who received this news responded by formulating an account that explicitly denied such a reality though Carla, for instance, indicated that it was painful to accept this truth. Likewise, none of the ten PT candidates who learned that they had not inherited the mutation explicitly negated this assertion though several, such as Colin, initially found it very difficult to believe. Not hearing either of these definitive assertions, Rosalind and Gabriella had more latitude to decide for themselves the meaning and significance of an intermediate allele.

The second sort of assertion that is relevant here shifts the framing from the level of specific utterances to the story schema and other interpretive resources which PT candidates and their families draw upon in order to make sense of the experience of predictive testing. These story schema and other interpretive resources derive from many sources, including the clinic. In *Chapter III*, I considered how the clinical discourse on predictive testing promotes the idea that having knowledge of one's genetic status is a 'gift'. Though I argued against the suitability of this metaphor, there were several PT candidates who drew upon the metaphor of the gift, as it was presented to them in the clinical setting, in order to elaborate the meaning and significance of predictive testing. Moreover, the interpretive utility of the 'gift' metaphor was not tied to the experience of receiving a particular test result. Colin referred to the 'gift' of knowing in order to underscore the utility of genetic information\(^{25}\) in making important life decisions while Carla said that even though it was painful to learn the truth, knowing that she had inherited the mutation associated with HD was a 'gift' because it enabled her to make some positive changes and live every day to the fullest.

Though the 'gift' was the most commonly invoked metaphor, study participants also drew upon a wide range of other images, metaphors, and analogies to convey a sense of how they experienced the act of coming to know the test results. Many referred to other illnesses as a means of anchoring their awareness of how HD was both similar and different. It was, in this

\(^{25}\) Recall that Colin learned that he had *not* inherited the mutation associated with HD.
respect, striking to note how frequently HIV testing emerged as a point of reference. This was, I believe, attributable to the fact that HIV testing provides a rich source of potentially useful schema for conceptualizing the social meanings of genetic testing.

Studies on the long term experience of being HIV positive have shown that there are many ways of conceptualizing such knowledge: it may be seen as a gift, burden, reward, punishment, relief, threat, opportunity for growth and/or irreparable loss (Adam & Sears, 1996; Couser, 1997; Davies, 1997). Though information about one’s HIV status differs in socially and morally significant ways from information about one’s heredity, these ways of seeing also shape perceptions of what it means to know whether or not one has inherited the mutation associated with HD. They are cultural resources which “provide material for the narrator to construct his or her own story as distinguishable from others who may be similarly placed in life” (Gubrium & Holstein, 1998:167).

Though such resources are sometimes taken for granted and difficult to articulate, they are not bluntly accepted or rejected nor do they determine how they are used. They are context-sensitive and may shift in content as they emerge in conversation. Further, they are entangled in an array of preferred and dispreferred story lines, rooted in strong emotions and likely to evoke a reaction from others. As Kleinman (1988:122) says, such resources are “our representations of the cultural flow of life experience; consequently... they congeal and unravel as that flow and our understanding of it firms up in one situation only to dissolve in another.”

The clinical interactions that occur during counselling and disclosure of the test result also contribute a preferred story line and thus they too are a formative influence on, as well as event in, study participants’ stories. The test protocol (Benjamin, et al., 1994) sets out the timing and sequence of various events (e.g., requesting the test, having blood drawn, learning the results, etc.) and, the pre-test counselling sessions, in particular, contribute to the development of a script which provides the PT candidate and their support person(s) with a sense of how the results session will unfold, what should happen when and how to envision the probable impact.

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26 Here I wish to draw attention to the fact that there is a growing need for comparative studies which elucidate the social and moral significance as well biomedical implications of information about different types of “embodied risk” (Kavanagh & Broom, 1998).
of each possible test result. This script also shapes the expectations and available repertoire of responses which service providers bring to clinical disclosure and post-result follow-up.

The script is, in ideal-typical terms, the anticipated unfolding of events. It contributes to the sense of drama which is a predominant feature of predictive testing; it constructs the disclosure session as the crux of the action; and it points, among other things, to the reduction of anxiety and uncertainty as a desirable resolution. Though this script for predictive testing implies that an informative test result will have significant implications for PT candidates (and, perhaps, their families), it also emphasizes that, regardless of the test result, there are benefits to knowing one’s genetic status (Wiggins, et al., 1992). Though it may take time to resolve emotional turmoil, depression or other psychosocial issues (e.g., survivor’s guilt), most PT candidates will, with appropriate support and guidance, eventually accept their results and utilize the information to more accurately envision, and plan for, the future. The ability to engage in rational planning and informed decision-making is, along with relief from anxiety and reduced uncertainty, a part of “the gift of knowing” (Kenen, 1996) one’s genetic status.

Though qualitative studies on the interactions which occur during the clinical disclosure of results might well demonstrate that service providers do not communicate any particular expectations for how PT candidates ought to understand and/or respond to a particular test result, almost all PT candidates and some family members interviewed in this research emphasized to me that they both evaluated themselves and/or felt as if they were being evaluated by service providers in terms of some such idealized expectations. This is perhaps to be expected given that it is a routine aspect of social interaction to monitor oneself from the standpoint of others (Mead, 1934). Nonetheless, with the exception of the four women who received their results through the rural protocol, this theme appeared in virtually all PT candidates’ stories regardless of the test result and regardless of how satisfied (or not) the PT candidate and/or their support person were with the way in which service providers conducted the disclosure session.

Of the twelve PT candidates who received their results in the clinic in Vancouver, most stated that they were somewhat or very pleased with the information, counselling and support they received. None said they left the clinic with unanswered questions and, though some
remained uncertain about the implications of the test results, most noted that they knew they could phone the genetic counsellor to clarify information and/or obtain further counselling. This was especially important to Rosalind and Gabriella (and their respective families) since there were many questions about the meaning and significance of having an intermediate allele.

The test protocol recommends that PT candidates bring a support person with them to the counselling sessions. Not all PT candidates choose to do so but of the twelve PT candidates who received their results in Vancouver, all but one (Albert) had a family member(s) or friend accompany them to the results session. Several of these support persons said that they felt the results session was over-dramatized. As Gabriella’s husband Brian said,

I do recall that they/ the way they do it you know/ they sort of say “and now (...) TA DAH! the test result is (...)” and they sort of/ they don’t give you any hint ahead of time/ they just sort of do the test result all of a sudden. And I remember feeling a bit irritated at that/ almost a little ceremony you know like opening the prize envelope and you are now Miss America/ that kind of stuff. But anyway may be that’s the way it has to be. But that kinda thing as I say tends to sort of irritate me. (M, family member, not at risk, 60 years, married, 4 children).

For others, it was the clinical setting which warranted comment. The room used for disclosure was “too clinical”, “too sterile”, and “too white”. There were “too many walls” and “no windows”; indeed, there were no signs of anything that offered any link with the lifeworld. Some thought this was in keeping with the way that “everything is handled so precisely and so carefully” while others found it incongruous that the genetic counsellor had a few tears and that the doctor could be empathetic in such surroundings. As Colin’s wife Emily said in reference to the way in which the doctor handled their disclosure session,

We were very surprised on the decision day, we totally expected the doctor to come in in his white coat and tell us yes or no and leave. Like that's what we expected and then we thought (the genetic counsellor) would just be with us and visit and whatever. And he (the doctor) came in/ no white coat/ came in, sat down, uh, totally relaxed. Talked with us, wasn't in a hurry. We were shocked. Like we thought personally “wow!” because we felt really special because we thought this guy's busy. I mean he doesn't have a lot of time. You know, and he's sitting here not rushed, totally engrossed in our personal decision here that we've just received, it/ and it was wonderful. (F, not at risk, family member, 39 years, married, 3 children)

As mentioned above, however, virtually all of the PT candidates who received their

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27 Of the four women who received their results through the rural protocol, two (Rose and Merle) were accompanied by one or more family members, one (Helen) went alone, and one (Maggie) received her results over the telephone.
results in the genetics clinic in Vancouver mentioned having an amplified awareness of the normal practices of monitoring self from the point of view of others. For instance, many PT candidates talked about how they felt as if they failed to demonstrate the appropriate type and/or intensity of emotional response at the moment of disclosure: some said they thought they were too emotional, others not emotional enough. The prevalence of references to such feelings was intriguing: it suggested that PT candidates felt constrained by the script for predictive testing and, in some sense, obligated to respond by disconfirming or otherwise expressing their unease with the clinical definition of the situation.

The experiences of the four women who received their results through the rural protocol warrant special comment here. All four women lived at least a day’s travel from Vancouver. Merle, Helen and Maggie elected to have their family doctor provide them with the test results and follow-up counselling while Rose opted to see a local genetic counsellor. All four women were pleased with these arrangements and stressed the benefits of being able to receive their results in their home communities. It was, however, only Helen and Merle that described the disclosure session in any detail.\(^{28}\) Though both women learned that they had not inherited the mutation, both expressed great empathy for their doctor in having to cope with giving an informative result for the first time. In addition, neither Merle nor Helen recalled feeling as if their respective family doctor had any particular expectations for how they ought to respond to the disclosure: it was a new and difficult situation for both doctor and patient. Hence, it was perhaps somewhat less fully scripted than it was for those who received their results in the predictive testing clinic in Vancouver. For instance, Merle stressed that her doctor found the experience of preparing to give the result unlike anything she had ever done before.

She hadn’t read much about that disease (HD) before because she never had to deal with anybody with it... "Well," I said, "You said you’d do it." She said, "yea, I know." She just didn’t realize how much a person doesn’t know about that disease. You know? Like she didn’t know hardly anything about it....And she’s a doctor! (laughs). (F, 50% risk, PT candidate, 48 years, married, 2 children)

This lack of familiarity with how to ‘do’ predictive testing was a source of symmetry in the

\(^{28}\) Maggie received her results over the telephone because winter driving conditions precluded travelling several hundred miles to see her family doctor. This was a substantial deviation from the test protocol but, given that she did not inherit the mutation, her physician felt that it was appropriate. Rose did not describe in any detail her recollections of the clinical disclosure session in which she learned that she had inherited the mutation. I do not know whether she was unable to recall or unwilling to discuss these details.
doctor–patient relationship and it shaped the way in which the disclosure session unfolded.

Merle’s doctor did not know Merle’s results until the moment of disclosure. As Merle recalled,

I was really nervous when I went in there but the doctor didn’t hum and haw and build it up/ she just said/ she went and got the fax and she opened it up ER/ it was folded and she opened it up and she read it and right away said “it’s negative.” And we were sitting there and she was/ she was crying and I was crying and Charlie {my husband} was crying (laughing)/ we were all crying and it was really emotional.

Merle’s recollections of the powerful emotion evoked by the disclosure point to another salient issue in understanding how study participants storied their experiences of making sense of the results. Affect is often what first comes to mind when recalling an event. It is “rather like a general thumbprint of the schema to be reconstructed” (Bruner, 1990:58); as such, the construction made on the basis of this attitude tends to justify and expand upon the affect. Moreover, my experience of how it felt to listen to study participants talk about how they felt at the moment of disclosure suggested that such emotion talk was also a potent stimulus to memory. It recreated the feeling of the event even though many of the details of who said what were not easily recalled. It also affirmed that the “tone” (Zerubavel, 1996) in which we remember events is a key feature of the schema that help to organize memory.

The afore mentioned example of the husband who commented on the overdramatization of the results session provides a good case in point. His affective stance toward the clinic was one of irritation and much of his story amplified this affect. It was part of a larger frame which derived from the adversarial nature of his work life and it came into play precisely because his feeling of irritation was a “hook” (Agar, 1994) that connected with the attitudes, conversations and events central to his working life. He was not unaware of this; indeed, he later talked about how his “bellyaching” about the clinic might have been “doing them a real disservice”.

Other examples point more directly to the raw emotion of results day as it was later recalled by PT candidates. Some study participants mentioned feeling anxious or extremely nervous although others said that there had been such a build–up that they were, by the day of the results, feeling “numb” or relieved just to be getting it over with. The most resounding theme that emerged from such talk about the disclosure session was, however, to do with self–consciousness and the monitoring of self from the point of view of others. As Colin
emphasized, he did not know what the doctor and counsellor thought of him when he did not have any visible reaction to hearing that he had not inherited the mutation.

I think they did such a good job of preparing you for the results to go either way that we had prepared ourselves for the worst...we expected the worst so when the doctor gave me the results it just went right over my head. I mean I heard him but it was/ he started off by saying/ by coming into the room and saying that he had some news that he thought I would be pleased with today so/ and my mind said/ I didn’t say it out loud/ but my mind said “and then he’ll tell me the bad news.” So this went on in my mind and uh anyways he told me and I really/ it didn’t fizzle me at all... I looked at them you know and I’m wondering what they’re thinking/ that I should be going up and down the hall doing cartwheels and shrieking and hollering/ but I mean it was nothing. (M, 50% risk, PT candidate, 41 years, married, 3 children)

Rosalind, on the other hand, was embarrassed about her lack of composure during the results session. She said that she had not expressed sufficient gratitude for the “good news” she received. She had obtained an increased risk from her linkage test but, with the direct test, she learned that she had an intermediate allele (of 35 CAG repeats). She understood that it was very unlikely that she would ever develop HD; moreover, she knew that her son had not inherited an expanded allele since he had also had predictive testing. When I asked Rosalind why she characterized herself as “ungrateful”, she said

Well it/ it appeared that/ you know it had to appear that way. You know I should have been (.) just you know/ and I was grateful but I should have/ I should have/ it came across/ I mean I was really, and to this day, I am really ticked off with myself. And I think it was inexcus—/ inexcusable to be like that you know. I burst into tears and (the counsellor and the doctor)/ I mean what did they think of me? They probably thought “oh my God, if she does this when we give her good news, what would she do if we gave her bad news? She probably would have slashed her wrists!” (laugh) I mean, you don’t/ this was just awful/ I mean really, just disgusting. (F, 50% risk, PT candidate, 57 years, married, 2 children)

Regina had a very different experience. She recalled being annoyed (and seemed to be annoyed as she spoke) because she felt that clinicians were looking for a “textbook” reaction that, in her case, was not forthcoming. She recalled that she had, during the results session, responded very calmly to the news that she had inherited the mutation.

I don’t feel that {the doctor} and any of the people in that place {the clinic} understand me or understand what I’m about and I just know that they seem to be looking for this textbook reaction that they’re not getting.... I don’t know what it is ‘cause it seemed when I first got my test results back it was like/ my friend was with me and we were talking on the way home and it seemed like they just dragged out and dragged out and dragged out the discussion. And I said to
Denise like it seems like they were waiting for something that they never got, cause I didn't get emotional, none of that stuff right. And she goes, "Yea, I thought they were waiting for something." And then a couple of weeks later I got a phone call, you know, "Just phoned to see how you're doing and if you're okay." And I'm going, "Like what the hell's going here? Like what's/ Is there something wrong me? Am I going to be walking down the Mall and all of a sudden like I'm an emotional basket case?" Like what are they looking for? (F, 50% risk, PT candidate, 28 years, divorced, 1 child)

Colin, Rosalind and Regina each recalled the clinical disclosure of results in terms of their affective stance. Each was, however, also attempting to make sense of their response in light of what they understood to be the normative expectations of the clinic. These expectations were a stimulus to articulate what the experience of learning the results did not seem to entail. Indeed, the post-results interviews as a whole, differed from the pre-results interviews in that PT candidates, especially, seemed to be engaged in making sense of their experience in light of what this experience was not.

As I discovered, 'not' is a powerful interpretive resource when it comes to the task of making sense of what is, at this socio-historical juncture, a fairly novel type of health-related information. Regardless of the test outcome, most PT candidates and their family members seemed, at the time of the post-results interview, able to offer a coherent account of the value of predictive testing for themselves and/or their respective family member(s) — that is, the process of predictive testing, if not the actual test result, had come to make sense, however tentatively, within the context of study participants’ lives. ‘Not’, I realized, played a big part in this sense-making process though ‘not’ was not an interpretive resource reserved for the narrative modification or rejection of the clinically-available story schema. It has, as we shall see, a much broader resonance in study participants’ stories.

Living Dialogue

It took a lot of interpretive effort to see what is now apparent — that is, that the constraints which exist on how we go about making sense of unfamiliar experiences are also resources which enable us to narrow down, without foreclosing, the range of meanings some

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29 Regina’s friend Denise accompanied her to the results session as her support person.
30 I wish to underscore that this portion of my analysis derives from the stories that PT candidates and their families told me about their experiences of learning the results. Though I did observe a number of counselling sessions and was present at one results session, I am not at this point reporting on my firsthand observations and fieldnotes about these sessions.
thing or event may have for us. Part of the difficulty in seeing the significance of ‘not’ had to do with the rich array of similarities and differences which emerged from participants’ stories. I could not seem to locate a framework that adequately encompassed both and yet also attended to the interpretive act of making sense of each type of test result. Like a kaleidoscope, the pattern kept shifting as my emergent understanding jiggled the pieces into a new juxtaposition.

The question of how the test results are intersubjectively interpreted and evaluated is a complex and extraordinarily context-sensitive phenomenon. Earlier, I stated that such evaluations must be parsed into their constituent components such that the value that is attached to having information is not conflated with the actual test result and/or the relief that almost inevitably flows from having the results session over and done with. The problem is, however, that the “trustworthiness” (Guba & Lincoln, 1994) of applying such analytic distinctions is very difficult to sustain when working with words on a page.

In living dialogue, “every concrete act of understanding is active: it assimilates the word to be understood into its own conceptual system filled with specific objects and emotional expressions, and is indissolubly merged with the response, with a motivated agreement or disagreement” (Bakhtin, 1981:282). In the highlighted neon text of my transcripts, the basis of such “motivated agreement or disagreement” was sometimes difficult to reconstruct. Propositions metamorphosed into questions which sent me forward (and back), searching the interview tapes, my fieldnotes and memory for enough detail to sufficiently recontextualize and explore the possible readings I was making. Like many study participants, I found it easier to rule out possible interpretations than pencil (or if I was confident, ink) in specific ones.

I thought a lot about how I was piecing together my understanding of study participant’s stories. I itemized the constraints on my ability to hear some types of stories but not others. I worried about what I was writing in a way that I had not before. I could not, I realized, reach the same level of data saturation as I had with the pre-results interviews. Each process of risk modification was different and I had in, some cases, only one or two PT candidates’ stories to work with in understanding each experience. Further, the prospective study design adopted in this research precluded the possibility of pre-selecting a sample stratified by gender and test
outcome: thus I did not interview any men who learned that they had inherited the mutation.31

At the same time, many participants' stories became woven into my own life such that story and life seemed to (in)form each other. The network of people, associations and connections between the life worlds in which I lived and worked seemed all of a sudden to have become exceedingly dense. Life was, as one of my study participants said, "really awkwardly complex." I had been doing research on HD and predictive testing for nearly six years and it was as if these worlds had merged. People that I had come to know through the HD community were now friends and friends were, in turn, drawn into what had been the quasi-separate world of my work. I felt saturated with life and work, acutely aware of time, and immeasurably saddened by the illness and subsequent loss of a wise and wonderful friend. What I was writing was painfully close to what I was living and it was, for a time, very difficult to write at all.

I blathered on, deleting whole passages almost as quickly as they appeared on the screen. I listened to music and tried to stop monitoring so closely the inadequacy of my words as they refused to mean what I intended. Then, somewhere in conversation with my (significant) "hermeneutic other,"32 I began to understand that it was the spaces between the words — the silences and omissions, pauses and retractions — as well as the explicit statements of what the test results did or did not mean that animated many of my study participants' stories. Moreover, I realized that what I had been doing in ruling out possible interpretations of ambiguous bits of text was not all that different from what many PT candidates and their families did in making sense of the test results: saying what something was not, had not however, occurred to me as way of proceeding. In my desire to studiously avoid reading my study participants' stories as if they were simply justifications of the test outcome, I forgot that the act of defining what is, often proceeds through the dialectical process of defining what is not. One interpretive act works in conjunction with the other; both "the authenticated and the dismissed, the nominated and the exnominated" are required (Anderson, 1996:10).

Applying this dialectical stance to my analysis of study participants' stories, I found that

31 Chapter X discusses this and other limitations as well as strengths of the prospective study design adopted here.
32 I thank Laurine Harrison for listening so attentively and for her uncanny ability to ask the questions that I needed to answer. This is, to me, what hermeneutics is about.
the interpretive processes most pivotal to understanding the sense-making activities of PT candidates and their families cut across the range of test results. Though individual stories differed according to the degree of emphasis placed on the cognitive, emotional and biographical dimensions of the interpretive work entailed in making sense of the test results, the stories as a whole reflected a collective emphasis on the strategic significance of 'not' as an interpretive resource which preserves the narrator's agency and sustains a sense of contingency. This is not to suggest that there is nothing distinctive about the task of making sense of each type of test outcome or objective process of risk modification; it is rather to acknowledge (in paraphrasing Marx) that *do* write our own life stories even though we seldom have the luxury of doing so under conditions of our own choosing.

In what follows, I present a series of exemplars which illustrate how selected PT candidates storied their experiences of making sense of an informative test result. These exemplars were selected in order to elucidate the cognitive, emotional and biographical dimensions of the interpretive process as they emerged in study participants' stories. Bracketing for the time being, the degree of intersubjective agreement entailed in PT candidates' and their respective family members' stories, I go about this task by juxtaposing selected stories related to each particular test outcome.

**Not Bad News**

There were three predominant themes which featured prominently in the stories of the ten PT candidates who learned that they had *not* inherited the mutation. These themes had to with: 1) acts of cognition, that is how the narrator came to believe that the test result was true, 2) the presence and absence of emotion, that is how the narrator felt about and responded to the test result, and 3) processes of narrative (re)construction, that is how the narrator went about (re)formulating a life story in order to make sense of and/or justify the absence of bad news.

Here I focus on three stories which were selected for their clarity in elucidating the above themes. These themes were not equally represented in all stories; indeed PT candidates tended to emphasize only one or two themes as a means of framing and representing the most prominent aspects of their experience. The narrative significance of 'not', however, cut across
all of the stories as did the salience of the narrator's stance toward the significance of talking (or not) with others about the test result.

(Not) Believing There's Nothing There

Sometimes it is very difficult to believe that news of something significant, whether good or bad, is true. We say that we cannot believe our ears; we hear but do not comprehend; we say that it will take time to sink in and become real. With respect to receiving 'good' news with predictive testing, part of the difficulty has to do with the language of disclosure. As Merle’s comments so amply demonstrated in the preceding discussion, the word 'negative' invokes two distinct frames of meaning and it is not always immediately apparent that 'negative' means 'no, you have not inherited the mutation.' Further, the counselling sessions prepare PT candidates and their families to hear 'bad' as well as 'good' news. Thus 'good' is, more often than not, first experienced as the absence of bad news.

As the reader may recall, Colin anticipated learning that he had inherited the mutation associated with HD. When he heard the opposite result, he had difficulty in believing that his test result was true; it was not what he and his wife Emily expected to hear. Moreover, he stressed that he could not get excited about the news since there were very few people that he could tell. In Colin’s story, there is therefore an emphasis on the interpretive work that is entailed in coming to believe that the test result is true — that is, that there is “nothing there.”

During the post-results interviews, Colin and his wife Emily both recalled having a very difficult time during the week before results day. Both said they experienced a feeling of numbness coupled with a profound desire to have the whole process of predictive testing over and done with. As Emily said,

We really both did think he {Colin} had it {the mutation} for sure. And we both/ we both talked about the kids because we both/ it hit us the same week that you know/ why did we ever have these kids? What have we done? Like we’ve been totally irresponsible. Like/ it hit us both that week.

When, as Emily put it, they received their “decision”, neither could believe it was true. Colin said it went right over his head; “it didn’t fizzle” him because he was so prepared to hear that he had inherited the mutation associated with HD.

Your mind is a really strange thing. I guess we wanted to make sure that we
wouldn’t have this big let down/ that we prepared ourselves for it. Unfortunately I think I did have a big let down. The let down was that, you know/ this thing that I’ve been carrying on my back for twenty–four years of knowing/ of my risk for twenty–four years/ has been such a big part of me that it/ you know I can’t just throw it off my shoulder and walk out and do a few cartwheels.

Emily said that while she was relieved, especially for their three daughters, Colin’s result was not something she could absorb quickly; HD had been a presence in their lives for so long that “you couldn’t just erase it in three seconds.” It was not easy to know how to think about the test result, much less how to make it seem real.

The doctor offered two things which were of assistance: one was a letter and the other was the idea that knowing one’s genetic status is a ‘gift’. As Emily noted, both were significant in helping Colin to realize that he had not inherited the mutation and moreover, that the knowledge was something that he could share with others.

You know what he {Colin} clings to? He has the letter from the doctor in a/ the doctor underlined that, uh, sentence, it says/ I forget how it’s worded. But it says that he has no trace of the gene or something like that. It’s very definite anyways. And he underlined it for Colin. And he {Colin} keeps that and he keeps looking at it and “see, I have no trace of it.” You know, just cause I guess you don’t totally believe that it’s all/ that there’s nothing there. You know, and he keeps looking at this piece of paper and “see, I have no trace of it.”

Here Emily refers to a very specific understanding of what it means not to have inherited the mutation associated with HD. The language of disclosure, the underscored sentence, emphasizes the absence of an expanded allele rather than the presence of a ‘normal’ allele. Thus an important aspect of the process of making sense of a ‘negative’ result involves coming to believe, that there is “nothing [significant] there.” An equally important and related aspect involves finding a framework for thinking about what it means to have such knowledge. This is where the metaphor of the ‘gift’ comes in.

Colin was reticent about sharing the news of his test results with his brother Brad. Like many other PT candidates who learned they did not inherit the mutation, Colin felt a sense of “survivor’s guilt,” a concept that he had read about in the literature on predictive testing. Moreover, Colin did not want to share the news with his mother since he believed that she would be too excited to refrain from telling Brad. This issue came up during the results session when the doctor asked Colin about sharing his results with others. As Emily recalled,

{The doctor} was very very good. You know what he said... it sticks with Colin
probably till the day he dies... He said to Colin/ cause Colin said “I’m not going to tell my Mum and I’m not going to tell my brother.” He {the doctor] said “why?...don’t you think that this would be a nice gift for them to receive?” And Colin said “never looked at it that way.” He {the doctor] said “don’t you think they’ve been worrying about this for a long time just like you? They’d be totally relieved if they knew either way/ so they could put it to rest. Ever thought of it that way?” He said, “Colin you’ve been given a gift. You know? This is special.” And he just, whoa, you know, it just hit him {Colin}. And he keeps saying to me/ he says “I keep thinking that the doctor said, I’ve been given a gift” and that’s how he’s treated it, like a gift, that he does not have it.

In the week after he received his results, Colin changed his mind and decided that he would share the news with his mother. As it turned out, he was very glad that he had done so.

I phoned my Mum within a week of the results and told her. She was so thrilled. Then I thought how nice that I told her because I had planned to tell her the next time I saw her which was... the week after she actually died. So if I had waited till that trip it would have been too late. So I thought/ you know/ I was able to pass the gift along... But she did share it with others. I met some family friends and they had heard the good news and so (...) my brother may know but he hasn’t said anything. I don’t think he knows but he may know. But anyways my worst fear was realized/ that she wouldn’t be able to/ cause she was just so excited herself. I mean you can’t blame her.

Reflecting on the fact that he had not had time to properly grieve his mother’s death, Colin said that he had heard that “one day it hits you” and you realize that your parent is gone. He wondered if the same thing would happen with his test results; the knowledge was, he said, “just below the surface” and one day it “won’t be pushed back anymore.”

Colin thought it unwise to talk about HD outside the family thus he did not share the news of his test result with many others. Colin thought that his inability to tell others did, however, help to explain why it was so difficult for him to accept his test result.

We had very few people that we could tell. Because of the nature of this disease you don’t tell anybody, you know, who has it in your family because if they understand that it’s hereditary and all that sort of thing then they put two and two together and you’re really jeopardizing your position in work and in insurance matters or any of that sort of thing. So you don’t tell too many people. And of course we didn’t tell very many people at all about the test. And so when you can’t/ when no one knew, there was nobody to tell the good news to.

Colin and Emily had a few friends that knew he was having the test, however, and some went out of the way to offer support. One of Emily’s friends left a card and a bouquet of roses for her on results day while a neighbour phoned as soon as they returned home and said that she had to know how things turned out. Such inquiries were important to Colin and Emily. As Colin said, “one of the only times I sensed any excitement was when I got to tell somebody.”
Colin was not sure when the “threat” of HD seemed to lighten but several months after his results he was preparing to visit his brother and he “started to feel fortunate” knowing that Emily would not have to cope with the same situation his mother had coped with for years. He also said that he found it easier to spend time with his brother Brad. Explaining why, Colin said that the “one big change” that had come about for him since the results had to do with the realization that he knew something about what his probable future would not be.

I can look at him {Brad} or go to the Huntington’s convention and see people {with HD} and realize that’s not me in five years or two years or whatever. That I don’t have that fear anymore. And that’s when you feel your load’s lightened.

*(Not) Feeling in My Right Mind*

Helen and her husband Duane focused on very different aspects of the experience of making sense of the knowledge acquired through predictive testing. Helen had no difficulty in accepting her test result; it was what she and Duane anticipated hearing. Nonetheless, Helen, more than any other PT candidate interviewed for this research, emphasized the unexpected cycle of emotions which followed closely on clinical disclosure. The intensity of these emotions took Duane by surprise and, as such, he had great difficulty in making sense of what he called Helen’s “negative, negative.” In Helen’s story, we therefore see how and why many service providers continue to emphasize the sometimes unpredictable responses that PT candidates may experience in response to hearing what is ostensibly ‘good’ news.

In Helen’s story, it is the intense and unpredictable nature of her response to obtaining an informative result which requires interpretation. It is contrary to everything she and her husband Duane expect and, though her story ultimately offers a high degree of closure, the process by which she arrives at this point is, in contrast with Colin’s story, highly dramatic. For both Helen and her husband Duane, predictive testing was the only responsible thing to do. Helen had to know her genetic status because she had three adult sons and it was important to rule out HD before they decided to marry and have children. Further, the idea of any parent not knowing their genetic status was, to Duane, like “leaving a can of gas and matches on a school ground.”

When Helen learned that she had *not* inherited the mutation, she was at 25% apriori risk. Helen’s mother died of cancer without ever showing signs of onset of HD. Helen had also had
cancer and felt that this had prepared her to deal with HD and predictive testing as if it were a matter of renewing what she already knew. Further, at the time Helen received her result, her only affected relatives lived overseas. She had never had much contact with this side of the family and therefore viewed predictive testing as a means of ruling out the threat of an hereditary disease that, in her experience, came out of the blue.

As the reader may recall, Helen lived in a remote community and decided to receive her test results through the rural protocol. She had great confidence in her local doctor and did not foresee any problems. Duane didn’t see that it was “a biggie” either because Helen was not, as he said, a “real 50:50.” As Duane began the post-results interview by saying,

I knew damn well she {Helen} was going to be negative. I mean there was no doubt in my mind... I mean you like to know for sure... you’re not unconcerned but not too worried about it... as I said before there was nothing with her mother to indicate {that she had HD}. (M, not at risk, family member, 42 years, common-law, 0 children)

Helen did, however, have an intense emotional response to learning that she had not inherited the mutation. When I asked her about what happened she (like Colin) described feeling quite numb beforehand. Duane had wanted to go with her to get her results but Helen insisted that she was strong enough to handle it by herself. She later regretted this but, as she explained,

I went in. I got the results, I felt I should be ecstatic and I didn't feel anything. I was no different than I had been walking in. Then I took a taxi home because I hadn't wanted to drive, I thought I wouldn't be capable and I was fine. (F, 25% risk, PT candidate, 47 years, divorced, 3 children)

As Helen recalled, her doctor

...was a little nervous about it. He had told me that I won’t have to ask him. “When you walk in you can read my face. I can’t hide things. If you come in and I’m looking in a corner and won’t face you, you’ll know it’s bad. But if I’m smiling and happy you’ll know it’s good.” And he was a blank.

Helen said that her doctor had a “short piece of paper” with about four lines of text on it, but she wasn’t curious enough to even have a look at. She was, as she put it, “insane calm.” Not long after she arrived home, Duane came in from work and, not seeing Helen in the back yard, went out again. Helen didn’t realize what had happened and waited to tell him her “wonderful news.” When she saw that he’d gone out again, she was furious.

As Helen recalled and narrated the intense cycle of emotions she experienced, her story became a long monologue quite unpunctuated by questions or prompts from me.
I came in the house and I doubled up and I cried. My world fell apart. I just/ Everything hit me at that time...I was absolutely devastated. And when he did come in I was probably the meanest I've ever been in my entire life. I was really really really nasty... I accused him of not caring enough to be there to hear the answers and he said "well then tell me what happened." Because I am just beside myself crying. And I said, "If you don't care enough to be here you don't deserve to know." And I wouldn't tell him, and I went in the bedroom and I cried and I asked him to pack his bags and leave. I can't believe I actually did that. I really was absolutely vile. And he said no he was going to stay twenty-four hours, if I wanted him to leave in twenty-four hours he would go. And it was probably an hour or so later when I finally got my head together enough to come out and tell him the results. Still not feeling guilty, still thinking he's deserving everything I'm doing to him. I can't explain it other than the fact that I just completely snapped. And went through basically the rest of that day with me crying and telling him how horrible he was and I didn't want him around. And then by the next day I was beginning to see the light a little bit and realize what I had done and he was very calm through the whole thing and he's never said anything other than he understands that I was under a lot of pressure and that I had really hurt him but he knows that I wasn't in my right mind.

Helen was grateful for her doctor's intervention. Reflecting on how he had called her that night and again on the day after her results, Helen said that she was astounded by the way he seemed to have anticipated her reaction much more than she did. Helen also said that she continued to feel "terribly guilty" about the way that she treated Duane but that these ongoing feelings were partly due to the fact that she had not been able to talk with Duane about what happened. She said that she had, for a time, been quite worried that Duane might want to leave her but when she broached the subject with him some weeks after her results he said it was "so ridiculous" that it wasn't even worth discussing. As Helen said,

I look back and think how silly it was but at the time I was so busy putting down everything I was feeling and putting everything into neat little compartments that I put him into this compartment where he really didn't belong. I feel pretty sheepish about it. It happened and there's nothing I can do to change that.

Duane referred to Helen's response to the news that she had not inherited the mutation as a "negative, negative". It rattled him deeply and he was, during the post-results interview, often inclined to scrutinize his own previously taken for granted assumptions and look for reassurance from me that he was making sense. He was, in a word, uncertain about what to make of the whole episode. As Duane said,

I don't know. I cannot figure that kind of thing out. Makes no sense to me. But I don't know/ hysterical would be the understatement of the year. ...I could see hysterical in bad news but not in good news so the whole thing is sort of unbelievable. Not a very good answer but just not what I expected at all. Probably/ I hate to use the word again/ but a female thing. I'm a male, is that a normal reaction?
Duane concluded that it would have been nice to know that “you can go freaky on a negative result” and suggested that it would be good to “sort of brace the partner that something like that might happen.” He had no problem in accepting Helen’s result — in fact he assumed that she would be “negative” — it was her response that made no sense to him. Upholding the value of being informed, Duane nonetheless found that his experience with Helen had sensitized him to the difficulty of making categorical distinctions between ‘good’ and ‘bad’ news.

For Helen, the experience faded quickly. She was pleased that she had faced up to “something unpleasant” and was thankful for her doctor’s concern but all that really mattered to her was that she did not have the mutation.

As soon as I was over the initial week or two shock, it’s almost like I dreamed it, like it didn’t happen, it’s in the past, I guess it has to be dealt with somewhat, but it feels more like a bad dream, I have to remind myself it actually did happen... I’ve got too many other things to worry about... I don’t have Huntington’s but I may have cancer. I don’t think I have AIDS. Heaven forbid but I may have all sorts of things and/ and maybe I won’t have forever...

Unlike Colin, Helen had few concerns about who knew her test results. So long as she had the opportunity to tell her sons first, she didn’t care if the whole town knew. Recalling her own experience in trying to obtain information about HD from the local library, she thought she might be able to help somebody else in the same situation. Moreover, Helen found it helpful to talk about her experience. Helen’s only regret was that her sister Norma did not provide her son (i.e., Helen’s nephew) with an adequate amount of information about HD. In Helen’s view, Norma had given him a “sugar coated white-washed version” of HD. This troubled Helen in that her nephew and his girlfriend were expecting a child. Helen’s only ongoing quandary thus revolved around whether or not she ought to go against her sister’s wishes and speak with her nephew about the family history of HD. With a close friend’s input she decided that the “least worst thing” to do was to keep her mouth shut.

(Not) Justifying the Good News

When Albert learned that he had not inherited the mutation associated with HD, the news quickly became routine. As he said in response to my question about how he felt on the long drive home from the clinic, “I was just glad to have something squared away, like tidying up my room.”
Though Albert’s mother Lillian was ecstatic with the news and later commented extensively on how she thought Albert did not seem pleased enough, Albert was more concerned with explicating how it was that he felt he had “not justified the good news.” The confirmed absence of the mutation associated with HD meant to Albert that he no longer had a convenient “rationalization for not getting on with things.” In consequence, Albert’s story focused on how he had come to re-evaluate his prior expectations about the significance of having an informative test result. Albert’s mother Lillian and brother Reg, played an important role in this process.

During the post-results interview, Albert was uncharacteristically talkative (or so his mother Lillian later said when she mentioned that she had overheard parts of the interview and could not believe that Albert had so much to say). Albert was, in particular, intent on describing how, in retrospect, he had come to understand that his experience of both the linkage and direct test was shaped and informed by clinical research on predictive testing. After requesting the linkage test, he read several published articles on the psychosocial implications of predictive testing and concluded that no matter what the test result, it would probably have a profound impact on his life. This expectation was, however, largely unfulfilled. Albert felt that he “didn’t justify the good news” because the knowledge that he had not inherited the mutation associated with HD did not “move” him to make the changes that he once believed he would if he knew for certain that he would not develop onset of HD. Further, he began to question whether the expectation of such change was an artefact of clinical research on the impact of predictive testing. In answer to my question about why he had expected his life would change, he said

A: I was influenced by Vancouver’s {the predictive test clinic’s} expectations that it {the experience of having the test} would have a change in your love life, a change in your work life. And it was simply not so. So it’s sort of like the case of uh/too many anthropologists studying a native American village and they change the culture.

S: Because they’re studying it?

A: Right. I think that was the case here. Except it didn’t change me. I was just concerned that I didn’t live up to what seemed like expectations. I didn’t justify the good news.

Albert’s mother Lillian thought that Albert was “not as pleased as he ought to be” with his test result. Albert did not disagree. In explaining why, however, he stressed that it had to do
with the way in which he had come to rely upon his risk status as a “crutch” which helped him
to make sense of his life situation. He was unmarried and, at the time of the post-results
interview, working in a night clerk’s job. Despite the fact that he had an advanced degree, he
seemed unable to better his employment situation or move ahead in a career. Given that he was
in his mid-forties and that his older brother Reg was married and successfully established in his
chosen career, this lack of advancement perturbed Albert; he felt troubled about not using “his
talents”. Though he was half joking, he admitted that part of him always looked for the easy
way out and that if he had “bad” news he could “live a life of crime.”

If I had bad news I could say “Oh I can’t do anything anyway. Relax.” It works
that way with me. There’s a down side to the good news too.

As we talked, however, it became clear that Albert took pride in the fact that he had been willing
to take “the risk of having the test,” not just once but twice. As he said,

I was willing to take the risk, the chance, just to get it squared away. That’s me...
I’m willing to pole-vault the Berlin Wall and nobody else in my family is.

I was intrigued by the contradictions in Albert’s construction of his own sense of agency.

He had accomplished something in going through with the test twice and yet he presented
himself as unable to fulfill other important life goals. When I asked him about this, he sighed
and then explained that nothing seemed to “move” him, that he needed what he called “a new
rhinoceros”. HD had been a “rationalization” for not advancing at work and not seeking a life
partner but, with good news rapidly becoming “old news”, he had to find a new way of
accounting for his stasis in life. As he said in response to my question about how he thought
about his direct test results,

In general terms I feel I’ve failed in getting a girlfriend, failed in getting a job.
There’s one big success and then a string of blah in my life. This {predictive
testing} is the one big climactic thing where I’ve had success. And everything
else, it’s just middle age and (...) my career petered out/ felt a little depressed
about that/ so it’s a high point/ a gift/ it is something that I’ve accomplished.

Albert’s sense of accomplishment spilled over into his desire to share the news of his test
results with others. When I asked him who he had told, he said that he “got out the Alpine horn”
and told his friends as well as all of his colleagues at work. This was, he said, in keeping with
his decision to “make everything public to anyone” soon after he learned the results of his
linkage test. The one problem was that Albert’s brother Reg refused to discuss the subject of
predictive testing. This was a sticking point with Albert and he admitted that he often had to
hold himself in check in order to avoid pressing Reg to have the test.

I have to put a damper on my own feelings of wanting to take the risk/ bear in
mind that for some people taking the gamble is not right. They could not live
with the results. So I caution myself not to advise them. Like my brother...I mean
emotionally I am all geared up to say “take the test, just go for it.”

Though Albert understood that his own test result did not modify his brother Reg’s a
priori risk, he sometimes spoke as if it did. Assuming that Reg had inherited the mutation,
Albert contemplated what this might mean for Reg’s two children (i.e., his niece and nephew).

I’ve got the good gene but I really haven’t got a better job or anything whereas
they have thriving careers/ they could really use it.

I asked if he meant that they were more likely to justify having good news and he said,

If I could give it {the good gene} to them... I would consider it. But would I in
actual practice if I did have the ability? I’m only feeling generous because there’s
no possibility of generosity.

Lillian was also concerned about Reg and remarked that she always kept “one eye open
for Huntington symptoms” in him and his two children. Reg was “this little niggle” that
reminded her of her husband and it was hard not to worry that he would eventually develop HD.
Lillian (like Albert), however, respected Reg’s wishes and did not press him to consider having
the test even though she too found the uncertainty hard to live with. During the post–results
interview, she said that knowing that one of her two sons would not develop HD gave her a
great deal of relief. It was only just over a year since her husband died from HD and she vividly
recalled how difficult the last months were. Knowing for certain that she had at least one son
who’s “not got Huntington’s” was as she said,

... a nice sort of comforting thought that came to me every now and then. “Oh!
Oh! Albert’s clear of it.” And I’d smile to myself idiotically and/ That’s how it/
Just from time to time. I’d be doing something and suddenly I’d think/ Maybe,
maybe reading about somebody else’s misfortune or something. I suppose it’s a
sort of what the Germans call chardenfreud. You know, uh, I don’t know the
English translation, it means joy at somebody else’s misfortune. Hey I'm out of
that. Albert’s been cleared, you know. It's a human feeling. It's not a very... noble
feeling, anything but. But that's what happened, you know. I just... I just sort of
hugged the thought to myself. It's nice. I think I'm doing it right now.

Having the certain knowledge that he would not develop HD was, to Albert, an
accomplishment which distinguished him from his older brother; it was something that he
counted as his “one big success”. Paradoxically, he could not, however, be as pleased as Lillian
thought he “ought to be” because this one success also illuminated his lack of success in other important areas. Near the end of the interview, he said

I’ve got good news as opposed to bad news so I can do something with my life. Though/ so if I don’t do something I feel restless and that’s been a part of it too/ a certain restlessness/ because I have the power to do it.

Not Good News

The four PT candidates who learned that they had inherited the mutation storied their experiences of making sense of the test result around three themes which paralleled those which emerged in the stories of those who learned that they had not inherited the mutation. These themes had to do with: 1) acts of cognition and embodied awareness, that is how the narrator came to accept that the test result was true, 2) the presence, absence and intensity of emotion, that is how the narrator felt about and experienced the test result, and 3) processes of narrative reconstruction and/or containment, that is how the narrator went about (re)formulating a life story in order to make sense of and/or minimize the impact of not hearing good news. These three themes were, however, more closely interwoven with the narrator’s sense of temporality than they were in the stories of those who learned that they had not inherited the mutation. The narrative significance of ‘not’ was, therefore, often positioned within the syntax of ‘when’. Further, the narrator’s stance toward talking (or not) with others about the test result was directly related to the (real or imagined) immanent, actual or potential onset of HD.

As in the preceding section, these three themes were not equally represented in all stories; each of the four PT candidates who learned that they had inherited the mutation tended to emphasize one or two of these themes within their stories. The narrative significance of ‘not’, however, cut across each of the stories as did the salience of the narrator’s stance toward the significance of talking (or not) with others about the test result.

(Not) Deserving HD

Carla found it very painful to accept that she had inherited the mutation and, moreover, that she was showing signs of onset of HD. In rejecting her partner’s view that she somehow deserved HD, Carla was however able to make a number of positive changes which enabled her to live every day to the fullest. Being able to talk with others about these changes was, as Carla
insisted, integral to her ability to start the "healing process." As such, Carla’s close friend Eva said that Carla taught her something important about living in the moment.

Carla, like Colin, adopted the metaphor of the ‘gift’ in order to make sense of her experience of predictive testing. I begin with a statement which offers a sense of how the metaphor of the gift informs Carla’s story.

For me it {knowing my test results} has been kind of like a gift. I feel like I’ve taken every day and made that day special for me. (F, 50% risk, PT candidate, 42 years, divorced, 0 child)

Carla made this statement during the second of two follow-up interviews. The first was conducted six months after Carla learned that she had inherited the mutation and the second, nearly two years later. Though it was a painful process, Carla gradually accepted her test results and, with the support of friends such as Eva, returned to the clinic to obtain a diagnosis of HD. This was one of many changes which allowed Carla not just to survive, but to thrive.

During the pre–results interview, Carla acknowledged that she felt “a bit unclear” in her thinking. Nonetheless, she felt confident that her results would be ‘negative’. She imagined that she would get the results, phone all her friends and “have a party.” This was not to be. During the post–results interview, six months after her results session, Carla recalled that it had been “devastating” to learn that she had inherited the mutation associated with HD.

It was like hitting rock bottom. I don’t know. I guess I had/ it didn’t surprise me in one respect cause I did feel like there were certain things happening in my life that hadn’t ever really happened before for/ to me. Yet I felt that I wasn’t sure where it was coming from so/ I mean getting the results... it was a real blow. (F, 50% risk, PT candidate, 42 years, divorced, 0 child)

Carla’s partner Tom attended the results session with her and then drove home. As Carla said, We stopped at the liquor store on the way home and bought/ I think it was Valentine’s Day or the day after... and we bought a big box of chocolate and came home and ate and drank a bottle of brandy. I was crying. I was pretty upset for a couple of days.

Carla’s close friend Eva was not surprised to hear Carla’s test result. During the post–results interview, Eva remembered how she had helped Carla to sustain a certain pretense about the possibility that she might be showing signs of onset.

I think we were both denying the situation in a way. I would say “oh, don’t

33 I am grateful to Jessica Easton for her clarity on this often overlooked distinction.
I was trying to comfort her because she would point at things like "oh look at me, I'm tripping over stuff... I can't even ride a bicycle anymore." So she actually verbalized what was happening... but I would do stuff like say "Oh, you have always been clumsy."... so there was definitely a prelude to what uh/ followed. (F, not at risk, family member, 48 years, single, 0 children)

The experience of receiving 'bad' news was, at the time of the first follow-up interview (four months after disclosure), still somewhat raw for Carla. Parts of her story tumbled out unordered and chaotic, as a stream of "and then and then and then." As Carla explained, it was sometimes very difficult to talk about knowing that she had inherited the mutation. In talking about talking about it, however, Carla recalled that it was through her connections with family and friends that she was able to start "the healing process." With each phone call, she relayed the news of her test results and gradually found that she was getting stronger.

For me it's really important to talk to people. Tom {my partner} couldn't believe that I was phoning all these people and he said, "you shouldn't be doing that," because I was all upset and crying. And I said, "Oh, it's okay. I feel better. At least I've talked to somebody." It was therapeutic for me and I needed that. It's good for me to be having that kind of contact with people.

As Carla reiterated several times, the "work of telling" others made the experience of knowing that she had inherited the mutation seem "more familiar." She even managed to find the occasional opportunity for laughter in the process. When she called her father in California and he refused to talk about her test results, she spoke with her step-mother. As Carla recalled, her step-mother suffered chronic pain from fibromyalgia and her response to the news was,

Ah, don't worry Carla. It's not any big deal. I mean we all have weird genes. It's not to worry. I've been in bed for nearly twenty years, so what? It's not a big deal. I'll send you a bunch of books.

Carla also found that some people gave her what she called "valuable feedback." When she spoke with her cousins, two said that they were not surprised to learn her test results because they had noticed "some changes" in her the last time she visited. Though it wasn't always easy to hear the truth, Carla said that she appreciated it when people "didn't mince their words".

Obtaining confirmation from others that she might have been showing signs of onset before she had the test helped Carla to understand the troubles that she had in holding down a regular job and performing routine activities such as balancing a cheque book. As such, Carla's experience began to make sense to her.

Carla's friend Eva noted that Carla was exceptional in her ability to care for others who
did not know how to respond when she told them about her test results. As Eva recalled, she and another friend visited Carla the day after her results.

And she supported us, because we didn’t know what to do, what to say, how to comfort her, and she comforted us.

Eva found it difficult in the weeks and months that followed, however, since Carla had such an intense need to talk about her test results.

She is full of words, she’s just full of/ She wants to unload herself so badly that she needs an outlet for it. And friends are there for that but speaking with my other friend about/ she said she had the same experience/ spent the weekend with Carla and she was exhausted at the end of... those two days because Carla was unloading so much and we are basically just a listener and it’s not even a dialogue anymore. It’s just a monologue and we don’t really have (dog barking)/ I don’t have any means of knowing how to react to this situation because I’m still a friend and I want to dialogue.

Carla’s partner Tom did not share Carla’s understanding of HD and what it meant to know that she had inherited the mutation. As such, it became increasingly difficult for Carla to sustain the relationship. As she explained during the second follow-up interview, she decided to leave Tom several months after learning her test results because he acted as if she was giving in to HD, almost letting it happen.

I was getting quite pulled down. He {Tom} was very negative and very upsetting to me, telling me things like “you deserve to have Huntington’s” and you deserve to be in the position that you’re in because you’re not really doing anything to help yourself.

Carla said that Tom believed there was something that she could do to prevent or delay onset of HD. Eva stressed that Tom had not been to most of the counselling sessions and that,

He seemed to be under the impression that it is something that you can overcome by positive thinking or diet or meditating or spiritual/ other spiritual practices. And perhaps, I don’t know/ I asked Carla about it and there apparently hasn’t been a case throughout the history of this disease where the symptoms would be reversed.

Nonetheless, Carla (like almost all PT candidates involved in this research) expressed no regret about proceeding with the test and/or receiving an informative result. Even at the first follow-up interview, Carla was adamant in stating that,

I’m feeling stronger. I feel like can kind of feel again/ just from finding out the information. But I’m definitely feeling some changes. And I think as I kind of make some moves into getting/ whether I’ll get welfare or some sort of income assistance/ that will make a big difference for me because at least I’ll feel like I’m doing something for myself. You know, something that’s going to give me a sense of independence.
Carla’s material circumstances were a significant part of the equation. Before deciding to request the test, Carla felt “trapped” by her situation. She had not had regular employment for months and was financially dependent upon her partner. They lived in a tiny apartment and Carla spent a lot of time on her own. When, several months after her test results, Carla was ready to acknowledge that she was showing signs of onset, she returned to the clinic to obtain a diagnosis (of HD). With the diagnosis, she became eligible for disability benefits. These benefits provided her with a small income of her own and a stimulus to think about making other changes. At the time of the two-year follow-up interview, Carla had, with the support of friends, left her partner, bought a dog (that became a constant companion), started seeing a Tibetan healer and travelled to Africa. Reflecting on these changes, Carla said, “I have placed myself at the top of the ladder.”

Carla’s view of herself and her ability to live each day to the fullest stood in stark contrast to the message that she was given by a neurologist. While visiting her family in California, Carla accompanied her sister (who also had HD) to the clinic and as Carla recalled,

He [the neurologist] kept looking at me and said “oh, so you’ve been diagnosed. Well, I don’t see any symptoms there and he said “I guess you’d better go home and get ready for the almighty hell to hit!” I just looked at him and said, “no, I am going to get ready for my life/ ready to really start living.”

As Eva maintained, Carla was able to talk openly about her test result and diagnosis, and ask for support where she needed it. This Eva saw as a real strength.

She is looking at life a little differently now. Really enjoying what she has... By doing that she’s really supporting all of us because we sometimes tend to rush through life without really taking a moment to enjoy and this is something that I have learned from her in the last month or two, that we are not/ that we are all very vulnerable. ...And Carla is doing very well on that level... she’s been able to really come through for people... and open her heart....So consequently people are reaching for her which gives her strength.

Out of that sadness, for all of us, something else grew and it is so absolutely wonderful.

(Not) Dealing with Reality

In so far as Carla’s story was about the struggle to sort out a way of living with HD, Regina’s story was about finding ways to just carry on and live life without HD. Both responses are sometimes considered to be forms of denial.
Regina had no difficulty in accepting her test result. Indeed, it was the absence of emotion that was most pronounced in her story. As Regina and her friend Denise emphasized, clinicians seemed to be looking for something that was not forthcoming. This puzzled Regina as she was not denying HD, it was just that HD was in the future and did not need to be dealt with until it actually occurred. Her stance toward talking about her results with others reflected this pragmatism: if someone asked a question, she would give an honest answer.

I had some difficulty arranging a post–results interview with Regina because she was extremely busy with an active work, family and social calendar. When I finally met with her, it was fifteen months after her results session. We began by talking about how she felt about hearing her test result and how her approach to making sense of her result differed from what is often referred to as denial. Regina had responded very calmly to the news that she had inherited the mutation associated with HD. There was no reason to have a “big” emotional response; it was just “one little bit of information.”

It (my test result) is something that's been dealt with but it needn't be dealt with again until the time that it (HD) occurs in my life but I don't feel that (the doctor) and any of the people in that place (the clinic) understand me or understand what I'm about and I just know that they seem to be looking for this textbook reaction that they're not getting. (F, 50% risk, PT candidate, 28 years, divorced, 1 child)

Elaborating on how this differed from denial, Regina said,

Nobody says what I feel is correct. Some people agree with me, some people don't. My mother doesn't agree with me just because that's her. I don't know. I just/ I don't think it's denial... it's called going on with your life and dealing with reality and you running your life not allowing Huntington's to run your life.

Regina’s story was, in one respect, very simple and straightforward. Regina had no signs of onset and repeatedly stated that the test result had no immediate relevance for her.

I'm still the same person, I just have this one bit of information that other people don't have. It doesn't make me any different. And I guess that's what I've told myself is that knowing what I know/ just sit back and think about it realistically, is it altering my life now? I say no. Is there anything I can't do? No. Well then why walk on egg shells? Why not live your life the same way you were living it before? Nothing's changed. Nothing physically has changed at all, you have one little bit of information that not everybody has the opportunity to get and that's it. ...I don't know if that's right but it makes sense to me.

Much of Regina’s story centred on her efforts to make sense of how clinicians seemed to be looking for a response from her that was not forthcoming. As Regina recalled,
Once they told me [my test result] I just kind of said “okay”. Denise and I started joking back and forth and talking about booking our cruise and that was basically it. They kind of kept asking questions and drawing the session out and we were sitting there and sitting there and sitting there and then when we left Denise and I basically almost said at the same time that it felt like there was something that was supposed to happen that didn’t happen.

On subsequent visits to the clinic, Regina felt as if the doctor and counsellor assumed that she must be holding something back. Recalling the last session she attended, Regina said,

{The doctor} didn’t like my attitude... something along the lines of he was not impressed with my attitude and he felt there was a lot of anger and hostility and frustration there. I said “well I don’t want to be here. Cut and dry, that’s it.”

I guess I deal with things very differently than most people. Like he says “what’s happening in your life? Oh I forgot, right. Oh you got a really busy life and lots of things are happening to you.” (sarcasm) ... And I said “you deal with it, you go on and I’m pretty happy with my life right now. I’m involved with somebody else and I have Geoffrey [my son] and it doesn’t look too shabby anymore.” I said “so you just play the cards that you’re dealt and go on.” I’m not one to talk about the problems that are going on in my life. There’s always somebody else out there that’s having worse ones.

Regina stated that no matter what she did, it was contrary to what people expected of her. Regina was, more than any other study participant, quite adamant that she defied all the norms. This was especially evident in what Regina had to say about the expectations of clinicians. As she said,

I guess I’m not a good subject, I have a very good/ I guess maybe I have a very callous attitude. I don’t know what it’s called. I think it’s a realistic attitude. I guess I was never going to be the textbook patient.

Regina’s insistence on writing her own life story also emerged within the conversational space of the interview when she asked about the relevance of her story to my thesis. As she said,

I’ve always had this feeling that I’m a square peg in a round hole. I haven’t really fit into/ or felt like I have fit into any other part of this (clinical research on predictive testing) so how am I going to fit into yours? You know/ usually when you do a thesis they want you to come to a conclusion at the end right? Well from what I’ve seen through the rest, I’m probably going to throw your theory all to hell.

I tried to clarify that I didn’t know what conclusion I would come to; I just wanted to listen and Regina replied with a long description of how she felt about filling out the battery of questionnaires required for clinical assessment. The process of checking boxes and circling

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34 Denise attended the results session with Regina since Regina’s husband was out of town and generally not supportive of Regina’s decision to proceed with predictive testing.
numbers on a Likert scale made her skeptical about the value of clinical research. It was “too impersonal”; it made her “feel like a number”.

I don’t think everything is as cut and dry as people are making it out to be. They’re forgetting that there are emotions and there are lifestyles and there are other people involved here and it’s not just a questionnaire on a piece of paper with a bunch of numbers and I guess that’s why I feel so strongly... I know I’m not a number. I’m me, this is who I am. I’m pretty proud of who I am.

Regina teased me about whether I would give her a separate chapter in my thesis and pushed me to explain how her experience compared with other stories that I had heard. As I came to realize, it was this insistence on writing her own story that infused Regina’s ability to get on with life. Talking about how she resented the way that everything she did seemed to be cast as if it were a response to knowing that she had inherited the gene, Regina said,

HD did not make me. It may kill me but it did not make me. People around me and my life experiences make me.

As a sociologist, I had a great deal of empathy for what Regina was saying though I am not certain that I managed to convey this to Regina since she was quite relentless in trying to ensure that I understood her point of view. She had, in the year following her results, separated from her husband, met someone new, returned to college and found a new job. She excelled in all of her courses and seemed pleased with her new employment.

Bad has turned into good, you know. Like I got laid off and I got separated but because I got laid off and I got separated my situation made it so that I knew had to go back to school, right. So whereas if those things hadn’t happened I never would have been in that situation where I could do it. So kind of I’m starting to believe more and more what they say that everything happens for a reason, right. And that’s it. You know, like you got to be optimistic somewhere in there.

Part of Regina’s sense of accomplishment also derived from the fact that she defied her mother’s expectations. She had done well for herself despite difficult circumstances and was, understandably, proud of herself. Regina’s friend Denise also commented on Regina’s resilience in the months after learning her test result.

Like it wasn’t the result that either one of us wanted but I think an awful lot of good has come from there. A lot of it. But the big plus is the fact that Regina is such a strong person. That’s why she’s been able to go on from that point... when you’re at the lowest point there’s only one way. You got to go up and she has.

When I asked Regina about how her husband Mark responded to her test results she stressed that he had not initially wanted to know the results. He had been very close to Regina’s
father when he had HD and Regina said he was afraid of going through the whole process again.

But Mark did come to me and say that he was ready to know my test results. When he asked me I said “what do you think my results were?” And he said “well I thought it was good news. You’re fine and nothing’s been bothering you.” And I said “well, actually you’re wrong.” He didn’t quite know what to say and basically from that point on he just totally withdrew.

Recalling that her test result was “the icing on the cake more than the cause of the problems,” Regina stressed that there were other tensions in the marriage and that her separation was for the best. Her “biggest hurdle,” as she put it, came in telling David (the fellow she had been seeing since her separation) that she had inherited the mutation. He had been very sick but was starting to feel better. They were sitting at the kitchen table at his place and Regina said,

“Well now that I’ve nursed you back to health... is there anymore medical conditions that I should know about you?” He’s got a degenerative tissue disease which I don’t know a lot about. ... He’s got arthritis, got rheumatoid arthritis as well, so we’re not talking about a really healthy person on the other side of the fence either, right. So maybe that’s why he can relate so well. And it was just the mode of the conversation was when the opportunity presented itself I said, Well while we’re on the topic,' so I told him. He said “okay.” And I said “well do you know what it is? And he goes “yea.” Basically end of conversation. “So it doesn’t bother you?” No. I thought that was really weird and a couple of days later, “Are you sure you understood what I told you the other day?” “Yea.” He didn’t care.

When I asked Regina about telling friends about her test results, she said that if someone asked her a question or the topic came up in conversation, she would talk about it, otherwise, it was on a “how I feel comfortable basis.” Friends would listen but never “pump for more information” and Regina could not see any point in dwelling on it.

See I don’t want to reach anything deeper. I’m quite happy with where I am right now in my life. And you know that’s what I said to my Mum. My Mum said “you know maybe he [the doctor] was right. Maybe you are in denial.” ...and I said “well let’s put it this way, if I am I’m having a hell of a good life right now.”

Regina had spent a lot of time considering why she did not feel more emotional about her test result and, in the end, her theory was as pragmatic as she was.

I thought well usually when you have an emotional reaction there’s something that’s caused the emotion. Like you stub your toe, you cry cause it hurts. Or somebody says something to you that hurts your feelings, it upsets you, but it’s usually relevant at the time. Sure the news wasn’t great ... but it’s really something that’s not relevant right now... maybe the time will come that you know I’ll start showing signs and symptoms of it. That’s when I’ll feel that it’s an appropriate time to have a reaction whereas right now? Nothing’s really happened to me... to make me react that way. I guess that’s really the way I think about it. ...They were telling me the future, that’s all they were doing. It’s not now, it’s not that I’ve gone in there and they’ve said you’ve got cancer, we have to take out half your lung, it was none of that. ...I don’t know. Delivering Geoffrey, that was
emotional! It hurt but it was emotional. But there was a reason for the emotions, right? There hasn't been a reason for the emotions yet. Maybe when there is, that's when it'll hit me.

(Not) Talking About It

Marie did not wish to talk about her test result or participate in a follow-up interview and thus her story is told in and through her husband Peter's story. Not talking about HD or sharing news of her test results with friends was important to Marie because it enabled her to concentrate on creating a “positive” mindset. Thus Peter, perhaps more than any other study participant, was constrained by being unable to talk with friends about how he and Marie were coping with what he referred to as the “immanence” of HD.

When Marie decided that she would prefer not to do a post-results interview she agreed to have her husband Peter continue his involvement with the research. The post-results interview with Peter occurred about one year after Marie’s test results. Peter was a bit anxious and noted that Marie had been quite apprehensive about the scheduled interview. He had encouraged her to meet with me and she had planned to do so up until the day of the interview when she changed her mind and decided to go out for the day.

Marie was forty-four years old and, like Regina and Carla, she was at 50% apriori risk before predictive testing. As Peter began the interview by saying, Marie was, after one year, “still acclimatizing” to her test results and trying to sustain a positive point of view. Recalling the build-up with results day as if it were only the week before, Peter said they were both “pretty shocked” to learn that she had inherited the mutation. They had built themselves up for “good news” so it was “devastating.” Marie was “hit so hard.”

Peter thought that Marie’s recollections of seeing her Uncle in the advanced stages of HD had a lot to do with how she responded to the news.

She remembers her Uncle more vividly than I do/ I never met the man/ but what she describes to me/ it wasn’t pleasant and it posed a certain amount of terror to her I’m sure/ ugh/ because you know it’s a vivid description that she gives to

35 I was scheduled to meet with Marie and Peter several months earlier but winter driving conditions delayed the interview and when I tried to reschedule, Marie sounded reluctant. I therefore delayed rescheduling until several months later in the hope that Marie might then be willing to meet with me.

36 As the reader may recall from the preceding chapter, Marie found it very difficult to talk about what it meant to her be at risk for HD. Part way through the pre-results interview, she asked me to stop the tape recorder while she went outside for some air. We later resumed by talking about safe topics.
me...it’s certainly been an imprint on her mind. (M, not at risk, family member, 46 years, married, 0 children)

Peter was familiar with HD because he knew Marie’s father and because he occasionally accompanied Marie to the Huntington Society meetings. His sense of HD was that people gradually became “locked in their bodies”. Because of this, it worried him when he thought that Marie was becoming more and more “rigid” in her outlook. It seemed to him that,

It’s all or nothing {with Marie}... no alternative. Black and white. I would say this started when she was about thirty–four years old/ she seemed much more flexible and easy going but then she got a bit more rigid. It’s a word I don’t like to use but...

Peter also noted that Marie seemed to be less and less communicative. She wouldn’t talk about how she was feeling nor did she want him to discuss her test results with any of their friends. Peter nonetheless felt that Marie’s worries about HD were not going to go away “by not talking about it.” The problem was that he didn’t want to push too hard.

I was kind of hoping that, in time, Marie and I could come to terms with this and then I could open up to my friends when she uh feels we should/I mean they’re such close friends (...) I don’t know/ it’s a containment for me. It’s a containment. Oh yeah. Yeah.

Peter found it hard not to talk about Marie’s test results with friends. He especially missed being able to have a good heart to heart talk with a couple of his closest friends. Peter knew that they were concerned because they had not heard from him.

I have to deal with this with Marie first and then I have to clear this matter up a bit because you know (...) it’s a block/ it’s a blockage. I’m not feeling good about it that’s for sure... I was thinking of talking to {Jerry} actually and haaa (long exhalation)/ but there’s no way around it unless I talk to Marie first.

Peter felt divided. His respect for Marie’s wishes were competing with his own need to talk about what was happening. As Peter said, this was “the crux of the matter.”

I’ve got to think of myself as well. I’m not a person to bottle things up... I like things on the table... I like talking about things so that I come to understand them.

I need to talk to my friends more about (. ) what’s on my mind and... we have to talk more about it you know/ about her {Marie’s} feelings on this... we have to open up more about how she’s feeling you know/ from her subconscious more it seems like/ cause she doesn’t say very much... we have to work on... find out exactly why she feels the need not to tell anybody right now and why we should carry on that road because I don’t think it’s the right route myself but this is where we’re differing right now.

Peter and Marie believed in doing everything possible to stay healthy and live a
“positive lifestyle.” They enjoyed the outdoors, took plenty of holidays, and maintained an avid interest in a range of alternative health practices. Peter was hopeful that a traditional Chinese Medicine practitioner would be of help to Marie. He had also seen a recent television documentary about electronic implants used to control the tremors associated with Parkinson’s Disease. He often repeated his belief that there would soon be an effective treatment for HD but noted that it was very hard to know what Marie was thinking. As he said, “Huntington’s is its own inside thing.”

Peter was concerned that Marie thought she might already be showing signs of onset and that HD was not just “immanent” but actually beginning. The more that Marie tried to avoid HD, the more it seemed to be on her mind and the more it was on her mind, the more her strategy of avoidance seemed to Peter to be something of a paradox.

There’s more concern and more worry that this may be it...it isn’t today and it isn’t tomorrow hopefully but the word immanent is/ is true you see... As I say we’re doing as much as we can to avoid/ for Marie avoidance is in everything it seems like/ in her mind/ with her friends/ and avoidance is imperative right now it seems to pursue good health.

But it’s not truly avoidance is it because it’s on her mind. It’s a paradox there. She doesn’t want to talk to her friends about it. She’s not denying it’s not going to happen. She thinks I’m denying it’s not going to happen cause I don’t accept immanent you see/ to some extent. She thinks it’s going to happen. She just wonders when and she’s worried about when it seems like.

Peter often managed to introduce a high level of ambiguity in responding to what seemed to be straightforward questions. There were, he stressed, no easy answers. The idea of predictive testing was, he said, very strange when he stopped to think about it. “It’s an odd thing isn’t it to tell somebody immanently you’re going to get this {HD}?” Peter nonetheless had a lot of faith in the scientific research and mentioned a particular interest he had in knowing more about what happened with other PT candidates who learned they had inherited the mutation.

I’d like to be in touch with a group of people who have the marker but don’t have the illness. “What are you doing? Where are you? What do you think?” I think that would be good for me.

Peter’s need to connect with others was pronounced. As he explained, he wanted to understand what the test result meant to Marie and, moreover, he wanted to do whatever he could to assist her in maintaining a positive outlook and delaying as much as possible the onset of HD. The frustration came with knowing that Marie needed to, as he put it,
...progress at her rate... Every time I try to accelerate that rate it just frustrates her and gets her mad at me.

Peter worried that predictive testing was a bit “like playing God” and wondered if there were many people who ended up committing suicide. Thus Peter questioned whether or not it was always a good thing to have more information. He thought that it might sometimes be dangerous to know too much although he quickly added that “being prepared is still the best answer for us.”

Toward the end of the interview, Peter said that he felt a “release” from having had the opportunity to talk. He also reiterated that it was especially important to hear from people who did not want to talk about their test results and, then, realizing the paradox embedded in what he was saying, he smiled and said that it was my job to figure out how.

Discussion

As the preceding exemplars suggest, the process of account formation is integral to the “work” (Strauss & Glaser, 1975) that PT candidates and their families must do in order to make sense of genetic information. Though the experience of obtaining an informative result may either confirm or disrupt existing expectations, it must somehow be accommodated within the flux of everyday life. No matter what the outcome, it is necessary for PT candidates and their families to re-examine taken for granted assumptions, reconstruct (or, reconsider) a sense of personal biography (Bury, 1982; Williams, 1984) and perhaps, renegotiate significant relationships. Even the ability to just “carry on” as before (Charmaz, 1991) is a way of living which must be “reconstituted amid new meanings and negotiated around new constraints or uncertainties” (Adam & Sears, 1996:xiii).

How lives are renegotiated — or in some cases “pieced back together” (Adam & Sears, 1996) — in the weeks and months after clinical disclosure is contingent upon the resources available to PT candidates and their families. The process is shaped by material factors (such as job security, health status and access to necessary social services), social and biographical factors (such as having a spouse/partner, a supportive network of friends and/or living in a disability-friendly community). Further, family and friends also have a profound impact on processes of validation and account formation when they uphold and affirm, challenge, or deny
the schema and explanatory models (Kleinman, 1988) adopted by PT candidates.

As PT candidates and their families talk about the meaning and significance of the test results, they jointly produce an account which says something about what the results do and do not mean. This emergent account then shapes, and is shaped by, ongoing communicative interaction. Account formation is therefore an effortful process which must be accomplished rather than assumed: the complexity inherent to the language of disclosure is but one indicator of how and why this is so.

The dichotomies which structure the language of disclosure are riddled with syntactic and semantic contradictions. These contradictions shape the process of interpreting ‘positive’ and ‘negative’, ‘good’ and ‘bad’ news and, as such, the stories that PT candidates and their families tell about the meaning and significance of having an informative test result are seldom straightforward or unambiguous. Further, though there are perhaps different cultural resources available to those who must make sense of knowing that they have inherited the mutation than there are available to those who learn that they have not inherited the mutation, the two interpretive challenges inform and, therefore, have much to say to each other.

There are, as the preceding stories suggest, many similarities, as well as differences, in how PT candidates and their families story their experiences of hearing and making sense of very different test results. Some of these similarities cut across the stories of those who received different test results while many differences exist within the stories of those who received the same test result. Even when viewed as a process\(^{37}\) (as opposed to an outcome), the modification of risk that occurs with predictive testing cannot be seen to determine a life course, biography or trajectory; nor does it impose a particular story line, plot, genre or style of narrative. As Riessman (1989) argues, individuals confronting what are ostensibly the same “objective” events seldom configure these events as if they were the same “phenomenological” events.

For reasons that I have already recounted, it is difficult to offer the same level of resolution in this chapter as I have in the two preceding chapters. The stories I heard from PT candidates, in particular, did not cluster around one central interpretive theme nor did they sort

\(^{37}\) Recall that when considering the test results as a process as opposed to an outcome, there are seven different transitions or modifications of objective risk (see Table 11, above).
according to the test result. The overarching dynamic and, hence the one striking source of coherence that emerges from the stories as a whole, has to do with the narrative significance of ‘not.’ Each of the stories I heard was (in)formed by an assertion and a negation; the former encompassing the array of expectations, cultural resources and story schema available to PT candidates and their families, and the latter being the various ways in which the narrator modified, revised or rejected such resources in the effort to make sense of the meaning and significance of the test result within the context of everyday life. Saying (or, as in Marie’s case, not saying) what the experience did not entail and, moreover, how it deviated from the expectations of others, was a means of sustaining a sense of contingency and preserving the possibility that things can always turn out otherwise.

The first and perhaps most important metastatement that emerges from the analytic task of undoing such narrative ‘nots’ is one which service providers have acknowledged but perhaps not yet fully appreciated — that is, there are few stories in which ‘good’ and ‘bad’ news is unqualified. The way in which good news is experienced is more accurately interpreted as the absence of bad news and, likewise, the experience of bad news is more accurately interpreted as the absence of good news. Let us reconsider, briefly, how and why this is the case. Albert and his mother Lillian both acknowledged the way in which all such news must be interpreted within the context of familial relations and the ongoing struggle to live out a full and meaningful life. For Albert, the absence of bad news imposed a new responsibility: he felt that he had to do something more with his life in order to justify the good news. Though he might well have come to this conclusion quite independently of predictive testing, this was nonetheless the context within which he made sense of the test result. Moreover Albert, like Lillian, also compared his test result with the ongoing uncertainty associated with his brother Reg’s status. As Lillian acknowledged in talking about how it felt to her to know that Albert would not get HD, joy is seldom self-contained; it exists in relation to an awareness of sadness and misfortune.

There does not seem to be a word, in English, that describes the experience of feeling simultaneously two equally intense but profoundly contradictory feelings: if for instance, joy is present, then sadness is supposed to be absent when in actual fact, one enfolds the other making knowledge of, or perhaps the existence of, the other possible. If we are constrained linguistically
to articulate the experience of sadness and joy as they co-mingle and arise in one and the same moment, how much more bereft are we in understanding the entanglement of good and bad?

There is as Rothman (1986:160) says,

"... more or less good, and more or less bad news one can get: a diagnosis can be made that is not good, but not...that bad. Thinking about these ambiguous bad-news results lays bare the moral system, or the values, underlying definitions of "good" and "bad" news. Positive and negative are presented as opposites. Good and bad, as we use them in our everyday life, are shaded. What is the criterion by which we rank and order good and bad news? How much less-than-good must results be before we consider them bad news?"

Carla's story was, perhaps, the one that ought to suggest some response to this question of how ostensibly 'bad' news is evaluated. Prior to learning her test result, Carla said that she felt confident that her result would be 'negative'. Though a part of her recognized that she could be showing signs of onset, she was not ready to acknowledge this and thus the test result came as a real blow. Listening to her story, however, it is apparent that there is 'good' news embedded in the 'bad'. Carla is clear on what her needs are and, despite her partner's obstructions, she reaches out to others in order to make sense of the experience. In the process it becomes more familiar and she finds that she is able, with the help of friends such as Eva, to make a number of positive changes which allow her to live her life more fully. Hers is a story of triumph over adversity and, in this, it speaks directly to clinicians and others who presume that sickness is associated with being miserable and health with happiness.

A second and no less significant metastatement that emerges from study participants' stories, has to do with how PT candidates and their families come to accept that the information acquired through predictive testing is true. Test candidates and their families are not passive recipients of the information derived from predictive testing. They must, as I have argued, actively make sense of their results. This is a fluid process which demands that PT candidates and their families find an intersubjectively meaningful way of situating the results within a temporal and evaluative framework. Moreover, PT candidates must also renegotiate a sense of self and identity in relation to the information acquired through predictive testing. Such processes present many challenges but the way in which they are resolved is not predetermined by the test result: it is the narrator's sense of agency and ability to choose amongst competing interpretations which shapes the meaning and significance of the result.
Colin initially has a hard time believing that he has not inherited the mutation. His ability to make sense of this result is, for a time, confounded by the difficulty he has in accepting that there is "nothing there." Being at risk for HD has been such a big part of him for so long that he is unable to think of himself without the HD cloud hanging over him. His sense of past and present are interwoven and it is only when visiting his brother that he realizes that HD is not his future; his brother is not him five years down the road. Further, Colin finds the metaphor of the ‘gift’ useful as an interpretive resource: sharing his results with others is what makes the knowledge become intersubjectively real.

Regina, on the other hand, accepts the news that she has inherited the mutation very quickly. She says that it is one little bit of information that is not relevant to her in the present. She conceptualizes her sense of self as distinct from the influence of HD, separates the present from the future and, most importantly, distinguishes genotype from phenotype. As such, Regina adapts the logic of biomedical knowledge to suit her own life circumstances; she creates for herself “an objective fixed point on a terrain of uncertainty” (Bury, 1982:173). She is in command of her life and, in defiance of the clinical expectations that she finds both puzzling and constraining, she constructs her own life story in a way that preserves contingency and sustains a strong sense of her own authorship.

The third narrative ‘not’ that makes an important metastatement about the experience of making sense of the test results has to do with the reciprocal implications of story schema and familial patterns of communication. This ‘not’ has two related aspects. First, where the schema that provide the story with its coherence are not shared by narrator and audience, the story may be unintelligible, misunderstood, and/or a source of conflict or tension. Second, some stories cannot be told because the schema which provide a sense of coherence does not promote dialogue with others. There is a metacommunicative stance that works against, though it never absolutely precludes, talking about some events and circumstances.

I shall deal with the second aspect first. In the preceding chapter, I showed how the narrator’s orientation toward talking (or not talking) with others about being at risk for HD had a significant bearing on how the decision to request the test was storied, both within the context of the interview and within the context of everyday life. Colin felt that he could not talk about
being at risk for HD with most people as it was too “scary”. Helen said that she had to talk about it in order to come to some resolution. Regina explained that she did not want to dwell on being at risk for HD but if someone asked her a question about it, she felt obliged to provide an honest answer. These storied patterns of communication also emerge within the context of this chapter as each schema for making sense of the test results suggests, but does not impose, a stance toward a greater or lesser degree of openness about talking with others about the test results.

Though I do not want to overplay the significance of this, it is apparent within several stories that the metaphors which structure our ways of thinking and speaking about communication are embedded in, as well as a formative influence on, study participants’ stories about their interpretive acts. Peter’s story about Marie’s desire to avoid talking about HD offers the most cogent illustration. Marie finds that her test result is unspeakable. She is, therefore, closed and Peter wants her to open up; he likes to have things out on the table and yet she is bottled up. He misses the connections he once enjoyed with friends and describes the experience of knowing Marie’s test result, but being unable to talk about it, as a “containment.” Helen, on the other hand, has an intense need to talk about her experience of learning her test result. When she cannot immediately share the news with Duane, she has an unexpected cathartic reaction. Once she has been able to talk it out, the experience of predictive testing quickly fades, much like a bad dream.

Returning to the first aspect of the schema–communication connection, let us now consider how the coherence of stories is always contingent on the relationship between narrator and audience. As Gergen and Gergen (1983:268) suggest, “whether a given narrative can be maintained depends importantly on the individual’s ability to negotiate successfully with others concerning the meaning of events in relationship with each other”. Further, one can only maintain one’s narrative constructions so long as others play their supporting roles. This mutual dependence or reciprocity leads to a “delicate interdependence of constructed narratives” which requires ongoing negotiation if periodic challenges to the validity of one’s narrative are to be avoided (Gergen & Gergen, 1983:270). One way in which people avoid such challenges is to incorporate others’ narratives into their own. For instance, Albert and Lillian had differing interpretations of the accuracy of linkage testing. Lillian was concerned that Albert was
overconfident about his direct test results. Incorporating her concern into his story, Albert talked
about how he developed a strategy to prevent himself from taking his linkage test result too much for granted.

I figured well, gee whiz, if I had a 2% probability for HIV how would I feel? (...) Devastated! ... It (2%) was still a significant amount.

One further example makes the point still more poignantly. As the reader may recall, Rosalind did not find her risk reversal to be cause for celebration. Indeed, she worried that her grandchildren might have inherited an unstable allele and that they would one day develop HD. This was an ongoing source of tension as her husband Erik insisted that she was making too much of a fuss about HD. The family was getting tired of hearing about it and, moreover, Rosalind had other serious health concerns that were, to Erik, of much greater concern.

Near the end of the post-results interview with Rosalind, she alluded to the way in which she had integrated HD within the framework of her other health concerns. Though I am here referring to my intuitive sense of how she intended this rather than anything explicit she said, I believe the point has a general relevance which ought not to be overlooked and which therefore justifies my somewhat more tentative interpretation. HD was, to Rosalind, a metaphor which allowed her to speak of her own mortality and perhaps, the way in which she grieved its potentially immanent realization. Speaking as if she were advising someone else who was about to go through the process of learning their results Rosalind said,

When you get your results then you have to/ you know it’s like someone dying and then you have to sit down and say/ you either mourn or you know/ there’s a bit of mourning and a bit of rejoicing involved. I mean if someone has been ill you’re sad they’re gone but you’re happy for them. You know uhm/ it’s not all bad. ... In a way it’s a little bit/ yeah, it’s a little bit like death.

The experience of knowing that she could not rule out HD in her family, was to Rosalind, also like a death because we do not generally like to hear about or imagine death. Indeed, we often do not allow people to speak of death; we try to stay away from it by changing the topic, glossing the subject with convenient platitudes and/or simply not making ourselves available to those whom we suspect may wish to speak of death.38

38 See also the preceding chapter in which Helen describes how difficult it is for others to allow her to speak of her mother’s death.
This brings me to a related issue which warrants some comment — that is the fairly uniform level of agreement which PT candidates and their families expressed with respect to the value of predictive testing. Though Peter was unusual in expressing some reservations about the potential dangers of knowing that one has inherited the mutation associated with HD, it was striking to note that there were very few study participants who expressed any regrets about their (or their family member’s) decision to proceed with predictive testing. This held, regardless of the test result. Several possible avenues of explanation suggest themselves. First, the research design was itself a factor which contributed to the finding and, perhaps creation, of such intersubjective agreement. Though I know of several instances where the siblings and spouse/partners of PT candidates were opposed to predictive testing, these family members were not invited by the PT candidate to participate in the research. Second, it is difficult to articulate regrets about a ‘choice’ that is constructed as freely taken; indeed, the old adage “you made your bed, now lie in it” is an example of the sort of response one might wish to avoid. Third, there is a growing impetus toward viewing illness and other evidence of one’s mortality as an opportunity for growth and empowerment — a learning experience. Predictive testing provides the opportunity to tell this type of story, however, where there seems to be a moral imperative to tell (or listen for) this story it may be very difficult to articulate (and indeed, hear) stories in which such epiphanous experience is neither sought nor found. The social price of not viewing illness and other threats to one’s mortality as a learning opportunity may therefore be high; if one is going to talk about one’s illness experiences and not risk one’s moral standing one had better tell a story about small victories in the face of adversity or the pleasures of living in the moment. Suffering and pain, like death and dying, are not topics that readily find a willing audience or, as one of my study participants said, “a listening ear.”

Could I hear the absence of meaning as well as its presence? Did study participants feel as if they could express anguish and suffering as well as relief from uncertainty in knowing the test results? Many study participants offered, in their stories, a metacommentary which supports the more general thesis that there are, at any given time, preferred cultural narratives about illness experiences and, moreover, about communication about illness experiences. Whether it was a subtle resistance to the expectations that PT candidates perceived in how they were
monitored during the results session or an outright rejection of the clinical schema, study participants expressed in varying degrees that they felt pressed to respond in prescribed ways to the experience of learning their test results.

Many PT candidates thus offered a metacommentary on how medical genetics seems to "hail" its subjects to be specific sorts of people, calling upon them not only to fill certain functional roles, but to tell certain types of stories about their experiences (Frank, 1995). Regina was adamant about this but the fact that participants in this research experienced such hailing is, in and of itself, not surprising: there are now many studies demonstrating biomedicine's sizable ideologic imprint, whether it is manifested in and through routine doctor-patient interactions (Waitzkin, 1991), the mechanistic metaphors that structure everyday understandings of the body (Martin, 1987) or normative expectations for participation in self-help groups such as Alcoholics Anonymous (Steffen, 1997). What is compelling, however, is the degree to which many PT candidates framed the experience of being hailed as an opportunity to define for themselves which story (not) to tell and how (not) to tell it.

In agreement with McKellin (1998) as well as others (Kavanagh & Broom, 1998; Sandelowski & Corson Jones, 1996) who have arrived at a similar position when studying other forms of embodied risk and diagnostic testing, I suggest that the test results seem to say more about what the story will not be than what it will. They are a kind of biographical negation; they bound and thereby describe what will not happen, yet leave open a field of other possibilities.

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39 The notion of "hailing" originates with Althusser's discussion of ideological hailing. It has been adapted by Waitzkin (1991) to elucidate how medical diagnoses are a form of hailing in which a medical definition is imposed upon the patient's situation.
A path, a riddle, a jewel, an oath — anything can be kept secret so long as it is hidden, set apart in the mind of its keeper as requiring concealment. It may be shared with no one, or confided on condition that it go no farther; at times it may be known to all but one or two from whom it is kept. To keep a secret from someone, then, is to block information about it or evidence of it from reaching that person, and to do so intentionally. (Bok, 1982:5–6)

Like a sieve, a secret presume the ability to set apart what is secret from what is not. Moreover, a secret presupposes a separation between insiders who are in the know and outsiders who are not. As Bok (1982) suggests, however, secrets also invoke “a family of related meanings”: the defining trait of intentional concealment is, in every practice of secrecy, intertwined with some admixture of mystery, sacredness, intimacy, privacy, silence, evasiveness, and/or deception. These meanings co-mingle and thus we often experience secrecy in highly ambivalent ways.

Where a secret concerns something mysterious, it may be both daunting and fascinating. Such a secret may evoke powerful feelings of dread and/or allure. The effort to unravel the mysteries of the human genome is, perhaps, the ultimate secret of this sort. In Watson and Crick’s words, the drama of molecular biology is a “calculated assault on the secret of life” (cited in Keller, 1992:42).

Intimacy and privacy represent another important aspect of secrecy. This aspect is reflected in the etiology of the German word heimlich — that is, that which pertains to home and hearth, the intimate, private realm in which some things are kept from the view of strangers (Bok, 1982:7). This sense of the private (as opposed to public) realm is often seen as indispensable to notions of Western individualism; it is that realm within which we feel we ought to be able to share our innermost thoughts and feelings; it is our as “haven in a heartless world” (Lasch, 1975). The private realm is, nonetheless, also that domain within which women’s and other forms of oppression have been most effectively concealed (Rich, 1979:189). Thus, the double life of privacy functions both to obscure issues of power and protect central
and defining aspects of human life (Boling, 1996:159). Privacy may protect our ability to safeguard personal information, choose our defining relationships and form our own identities but it may also conceal connections between "the personal troubles of milieu" and the "public issues of social structure" (Mills, 1959:8).

Silence conveys yet another aspect of secrecy — that is, that which is unspoken and/or unspeakable. Silence may, in its poignant and aesthetic shadings, convey a sense of profound appreciation or respect for the ineffable when, for instance, we engage in quiet reflection on something that moves us beyond words. This is not, however, the same thing as intentional concealment. The silence which is the "first defense of secrets" (Bok, 1982:7) evokes a sense of the prohibited, shameful or abominable. Here silence may signal tact and the ability to disattend something embarrassing or excessively revealing (Karpel, 1980). In its most potent forms, however, silence bespeaks repression and avoidance, especially where the emotions associated with felt and enacted stigma render some aspect of self-knowledge too painful and/or humiliating to admit (Scheff, 1990).

Finally, secrecy also suggests evasiveness and various forms of deception such as lying. This link is, nonetheless, itself deceptive. While deception requires some level of secrecy, not all secrecy is intended to mislead (Bok, 1982). The secret ballot, for instance, is a practice of concealment which is intended to ensure uncoerced participation in elections. There is no intent to deceive because it is citizens alone who have a legitimate interest in the placement of their X.

This link between intentional concealment and legitimate interest raises an important issue which has thus far eluded attention. Secrecy is sociologically meaningful when it refers to those practices which involve the intentional concealment of information from someone who has a legitimate interest in the secret (Karpel, 1980) — that is, a secret concerns something that another party needs to know, ought to know, and/or has a responsibility to know. From the standpoint of the apparent secret–holder, a secret is therefore not a secret when it concerns information that another party has no legitimate interest in; such information may be something that is private and, as such, worthy of being treated with respect and discretion but it is not a secret.

Further, a secret is not a secret once the secret–holder discloses the information to the
previously unaware. Though the practice of secrecy may have lasting consequences when something important has been withheld from someone who has a legitimate interest in the information, a secret ceases to be a secret when all concerned parties both possess the information and the knowledge that is has been revealed to other concerned parties. In such cases, an open awareness context (Glaser & Strauss, 1964) prevails and, unless a new secret is formed or it becomes apparent that one party is unaware that what now stands revealed was, for a time secret, the social dynamics of concealment have run their course.

Or so, in an ideal speech community, the story might go. In everyday life as well as in the world of research on human communication, things are infinitely more complex. The task of understanding human communication — whether secretive or not — requires that lay actors as well as researchers understand the understandings that communicants have of their own communication. A key part of such understanding, as it emerges at both levels of the “double hermeneutic” (Giddens, 1986), derives from close attention to context and the situation in which communicative interaction occurs.

The norms and practices of communicative competence vary widely in different settings (Saville-Troike, 1989). As the preceding chapter demonstrates, confusion often arises when PT candidates and their families talk about the meaning and significance of a ‘positive’ or ‘negative’ test result. Even within the context of face-to-face communication, the language of disclosure makes it difficult to communicate clearly and unambiguously about the results of predictive testing. There is a large amount of tacit knowledge that is required. Further, both listener and speaker must devote concerted attention to monitoring the flow of conversation in order to draw the correct inferences and assess whether or not others are also drawing the same inferences. Practically speaking, it is therefore very difficult to ascertain with any certainty, what one speaker’s understanding of another speaker’s understanding is (Krippendorf, 1994).

Purpose and Outline of Chapter

The substantive focus in this chapter is on what study participants had to say about disclosure and/or non-disclosure of the results of predictive testing. As the reader has, perhaps, anticipated ‘not’ continues to have some purchase here. The phrase which names this chapter —
"It's Not a Secret But..." — is an amalgam of thoughts and feelings, words and silences, about the lived experience of hereditary risk for Huntington Disease. This phrase is intended to suggest a sense of ambivalence about the experience of sharing information about oneself and related others when such information is constructed as a powerful source of, as well as an ever-present threat to, self-identity, intimacy, and social life (Goffman, 1959; 1963). The phrase is, however, also intended to evoke a series of questions which have to do with the complexities and ambiguities as well as alembicating features of interpersonal communication about predictive testing and Huntington Disease. “What is the ‘it’?” “Why is ‘it’ not a secret?” “From whom is it not a secret?” and “If ‘it’ is not a secret, what then is it?”

I proceed to address these questions by first briefly reconsidering the significant familial, temporal, existential and social implications of the information derived from predictive testing. I then present a synopsis of the patterns that emerged from the stories study participants told about their strategies for managing disclosure and non-disclosure of the test results. These strategies or “attitudes” (Burke, 1973) are, broadly, ways of naming situations that occur sufficiently often that we need a name for them. Such strategies are, however, also reflected in the micro level interactions which comprise the who tells what to whom, when, and where part of the story. As we shall see, gender is especially significant here in that it is women who take on the primary responsibility for communicating within the family about genetic information and, moreover, it is within the home that much of this “genetic housekeeping” (Shakespeare, 1992) occurs. Indeed, it is the centrality of home as a locus for family which most readily defines the boundaries between those who have a legitimate interest in knowing the test results and those who do not.

In summary, this chapter offers a somewhat different perspective on the substantive issues arising from the three preceding chapters: it is one step removed from the detailed presentation of study participants’ narratives and thus I feel somewhat unburdened of worries about revealing something that I should not or psychologizing too extensively any one study participant’s account (Johnson, 1982). The patterns in communication which are of concern here are, nonetheless, evident in the narratives presented in the three preceding chapters. Thus, it is hoped that this chapter will weave together the multiple threads I have thus far unloosed.
Genetic Information Reconsidered

One of the questions which arises from the phrase “it’s not a secret but” is what is the ‘it’ that is being referred to here? This question is best addressed through a brief reconsideration of some of the most salient dimensions of genetic information and their implications for familial communication.

The word ‘it’ stands in for many things and thus it has been problematized throughout this dissertation. Within the context of Chapter VI and the question of ‘when it started’, ‘it’ referred to the inkling that something is wrong with a family member, the distant recollection of someone uttering the words ‘Huntington Disease’, the explicit knowledge that a family member has been diagnosed and the realization that HD is an hereditary disease which has a range of significant implications for self and related others. Likewise, I suggested in Chapter VIII that, when considering the way in which PT candidates and their families evaluate the test result, ‘it’ may refer to the test result, the state of being informed or the process of predictive testing. Within the context of this chapter, ‘it’ refers to the test result and all that this information implies for self and related others. As such, a similar parsing exercise is required here in order to tease out the multiple implications of the test result for various practices of disclosure and non-disclosure.

There are several dimensions of the information acquired through predictive testing salient to understanding the meaning and significance of study participants’ practices of disclosure and non-disclosure. First, the information does not belong to just one person: it is both individual and familial in orientation. Second, the information has a temporal dimension: it forecasts the probable onset or absence of hereditary illness. Third, the information is intertwined with existential aspects of self and identity, agency and personhood. Given that the self is a fundamentally social construct, however, these aspects are also rooted in the ongoing flow of social interaction (Bakhurst & Sypnowich, 1995).

An example will illustrate the significance of these issues. Jason was an especially astute and self-reflexive informant when it came to understanding how each of these dimensions of genetic information shaped his experiences of talking with others about the information acquired through predictive testing. As the reader may recall, Jason’s mother Gabriella inherited an
intermediate allele (with a repeat number of 36). When I asked Jason what this result meant to him, he began by saying that it confirmed that he and his sister were “really at risk for HD.” He also said that although this knowledge didn’t change him, it did change the way he viewed himself. Moreover, he also felt that it changed the way others perceived his family.

It {HD} becomes one of the things that is in my family and one of the effects is that you have this news that you can tell people... I don't like the term but our family has this affliction. Whereas before it was just a sort of nebulous concept, maybe now it's a bit more concrete.

It {my mother’s test result} doesn't change the way I am. It doesn't change whether I have Huntington's or not. I either do or I don't....It changes the way I perhaps view myself a little bit, 50% versus 25% {risk}. But since her results are marginal it really softens that blow.

As Jason suggests, predictive testing provides information that ‘belongs’ to more than one person. Given that we inherit DNA from both parents and share an average of 50% of the same genes as our siblings, predictive testing reveals information which is both individual and familial in orientation. The modification of risk which accompanies an informative test result may, therefore, have significant implications not only for the PT candidate but also for offspring, siblings and/or parents. This is in contrast to most diagnostic information which centres almost exclusively on the patient who has, or is suspected of having, the disease condition.

The implications of this for familial communication about genetic information and, in particular, the disclosure of the test result to others, are profound. Framed in terms of the legitimate interest that family members have in knowing a PT candidates’ test result, many PT candidates feel an obligation to disclose their test result to those family members who, like Jason, receive a modification of their own apriori risk along with the news of the PT candidate’s result. This type of dual disclosure is, as one might well imagine, often very difficult for both (or all) concerned parties.

Further, predictive testing forecasts, with varying degrees of certainty, the probable onset (or absence) of hereditary illness. Though a test result indicating the presence of the mutation associated with HD does not specify age of onset or predict potential severity, it does create the novel situation in which some individuals know in advance of impending illness while others learn that they (and hence their offspring) have ‘escaped’ such a fate. Jason knows neither for
certain, but his mother’s test result does not rule out the possibility that she will develop HD and/or that he or his offspring (if he becomes a father) may eventually experience onset. Jason is, in consequence, “really at risk” for HD. Having this knowledge does not alter what will happen (i.e., whether or not Jason will develop HD) since this possibility is contingent on what has already happened (i.e., whether or not Jason has inherited an expanded allele) but it does make hereditary risk “more concrete.” It is “news” that Jason says he “can tell people.”

Finally, genetic information shapes the way in which we see ourselves as well as the way in which we see others seeing us. It is connected to experiences of uniqueness and personhood, agency and self-determination (Brock, 1994). Jason says that his mother’s result changes the way he sees himself since he is now at 50% rather than 25% risk, but he adds that the ambiguity of her “marginal” result softens what might otherwise be a real blow. His use of the word “affliction”, however, also suggests that he has some trepidation about disclosing his mother’s test result and/or his own risk status to others. He doesn’t like the term but employs it to underscore the social stigma that is often attached to hereditary diseases such as HD.

In this respect, Jason is somewhat unusual: he speaks from the direct experience of “enacted stigma” (Scambler & Hopkins, 1986). Though most study participants acknowledged that the possibility of discrimination and/or stigmatization impinged upon their practices of disclosure and non-disclosure, few could recall an instance in which they had directly experienced stigmatization when disclosing to family or friends. When I asked Jason how his friends responded to the news of his mother’s test result, he told me that he felt quite perturbed when a former girlfriend responded inappropriately.

...in talking with Monica about it she really/ I think to some degree got the view that/ yeah, she knew my Mum had the gene and we have other health problems/ you know, Brian [Mum’s husband] has hemochromatosis and needs a hip replacement and she [Monica] just got this feeling that we were a bunch of people that should have fallen under natural selection or something.

Based upon this experience, Jason revised his thinking about how to present such information to those outside the family.

So I think in presenting this information to other people outside my family that yeah/ I might think about this and try to present it in a different way...not my Mum or us having the gene/ just having a propensity to develop Huntington’s and that’s all that’s been tested for/ not whether we have it or we don’t but the likelihood we will get it at various ages. Maybe that will lose people in the
clouds but maybe that is better than them thinking you’re defective... I mean I say those sort of loaded words and they don’t/ I don’t think that’s the sentiment that people really have on the word defective/ they don’t see me or my Mum as defective... so I don’t know. It’s something that’s come to me in talking... but I think it’s important in presenting it to other people. Monica was instructive in this.

Though Jason was unusual in having directly experienced the social stigmatization attached to hereditary diseases such as HD, the issues that he raises have an overarching significance for understanding PT candidates and their family members’ strategies for managing genetic information. As Jason’s story demonstrates, there are even in the most straightforward disclosures a host of fairly subtle distinctions that must be made in deciding how to present the information to others.

This points requires a brief aside. During conversation participants generally adhere to some common purpose — that is; they have an agreed upon direction which shapes the quantity, quality and relevance of each contribution to the conversation. As Grice (1989:26) suggests, however, it is often the clarity or manner in which information in presented which allows participants to draw the correct inferences from what is being said. More specifically, Grice emphasizes that participants must attend to a common concern with how things are said as well as what is said — that is, we must attend to both the content and relationship aspects of communication.

Herein lies an intriguing tension: a high degree of clarity in communication is integral to the effective transmission of information. Nonetheless, clarity also prescribes obscurity and ambiguity of expression while prescribing brevity and orderliness. This may not, as Jason suggests, always be desirable when disclosing potentially stigmatizing information. As such, it is necessary to weigh the clarity of what is said against the anticipated response of one’s audience; one must evaluate the consequences of the disclosure for self and other family members and be prepared to change one’s strategies if and when they do not have the desired effect. Ambiguity is, in this regard, sometimes an ally for, like the narrative ‘not’ which preserves a sense of contingency and open-endedness, a cultivated ambiguity about the meaning and significance of the test result sustains the possibility that some degree of confusion on the part of one’s audience may be preferable to clarity. As Jason says, it is better to “lose people in
the clouds’ than have “them thinking that you are defective.”

The tension strained in both directions, however, as the permissibility of ambiguity varied according to perceptions of the legitimate interests of the intended audience. From the standpoint of those who were concerned that a family member did not understand the seriousness of being at risk for HD, the amount of information provided, as well as its truthfulness and clarity was paramount. As Helen noted with dismay, her sister Norma provided her son (i.e., Helen’s nephew) with “a sugar-coated white-washed version of the truth” about HD. Clearly, this was not enough, in Helen’s view, to fulfill the requirements of a proper disclosure to someone with a legitimate interest in the information.

In summary, the familial, temporal, existential and social dimensions of the information acquired through predictive testing have significant implications for how PT candidates and their families size up the social situations in which they decide to withhold and/or disclose genetic information to specific others. As we have seen, even in this abbreviated discussion, practices of disclosure and non-disclosure cannot be easily prised apart. Each act of disclosure may, as Jason suggests, withhold something in order to achieve the speaker’s desired objectives. Thus, in considering the question of why genetic information is ambiguously cast as ‘not a secret but’, we have already gleaned one response: it is not a secret because some portion is revealed but, at the same time, some other portion is also withheld.

Outside of a court of law which requires that one testify to the truth, the whole truth and nothing but the truth, the partial nature (and hence partiality) of all communication is inevitable. Such partiality is a necessary feature of the ways in which we frame our understandings of the world and separate what matters to us from what does not. Within the context of this research, however, the partial nature of communication was seldom taken for granted. In consequence, study participants frequently remarked not on whether someone knew about the family history of HD or the test result — indeed several male family members, in particular, exclaimed that they could no longer recall who did and did not know — but rather on how the information was presented and what, in turn, the recipient of the disclosure seemed to understand and infer from what they heard. As such, the issue of how much to say to others, and how to say it, was central to understanding PT candidates’ and their families’ strategies for managing genetic information.
Strategies for Managing Genetic Information

The understanding of what it means to communicate about hereditary and other types of "embodied risk" (Kavanagh & Broom, 1998) must emerge from an awareness of what it makes sense to say, as well as what is said, at this particular socio-historical juncture (Armstrong, 1984). Such understanding requires a "rhetorical–responsive" stance toward language and its role in constituting and reconstituting the social world. This stance emphasizes that all human activity involves "the contingent flow of continuous communicative interaction"; as such, it underscores the way we make and remake our social worlds just as they make and remake us (Shotter, 1993:13–14).

Talk about ‘strategies’ and the ‘management’ of genetic information may, within the context of these stipulations, sound as if it errs in suggesting an overly conscious, deliberate and/or predetermined approach to understanding how PT candidates and their families communicate about genetic information. If this is the case, then ‘method’ might perhaps suffer a similar fate in sounding “too methodical.” Perplexed by trying to find a word which effectively conveys the fluid way in which we approach and deal with social situations, Burke (1973:297–8) takes courage from a more militaristic idiom: “one seeks to ‘direct the larger movements and operations’ in one’s campaign of living. One ‘maneuvers’ and the maneuvering is an ‘art.’” Burke’s (1973:300) main point, however, is this: the way in which we size things up and determine what is promising and what menacing, is

...the strategic naming of a situation. It singles out a pattern of experience that is sufficiently representative of our social structure, that recurs sufficiently often mutandis mutatis, for people to ‘need a word for it’ and to adopt an attitude towards it.

As Burke suggests, another name for strategies is “attitudes.” Following the same logic, another name for managing might be coping. Nonetheless, where managing suggests too much control over the process and outcome of communication, coping (with the emphasis on just barely) suggests too little. We had best, perhaps, just accept that the polysemic ‘managing’ suggests a both/and rather than either/or approach.

Strategies for managing information are often goal driven but the intentional goals most relevant to understanding strategic outcome–oriented communication need not compete with the
emergent, relational goals that facilitate process-oriented communicative interaction (Daly & Wiemann, 1994). To assume that both cannot be operative at the same time is to miss the complexity of communicative interaction. As Habermas (1984:241) insists, “for the actor, the aims of action oriented towards success and reaching understanding are situated on different levels.”

**Naming the Situation**

In the preceding chapters, I presented three moments in study participants’ stories about their acts of communication; these moments were defined by the transition into initial awareness of the family history of Huntington Disease, the decision to request predictive testing and, the experience of receiving and making sense of the test result. Following Burke (1973), I now wish to focus briefly on those patterns of communication which are representative of the stories as a whole, that recur sufficiently often that we need words to describe them and ways of sizing them up.

The first pattern of experience and cluster of related attitudes toward disclosure, I call *having to talk about it*. We have already met many of the most salient features of this attitude in preceding chapters. Briefly, however, it entails an impetus toward the active initiation of open discussion. It emphasizes the therapeutic dimensions of disclosure and the possibility that HD is no different than most other diseases. Talking about HD is, accordingly, evidence that it is not something that must be hidden, it is, in short, nothing to be ashamed of.

The second pattern of experience and cluster of attitudes toward disclosure, I refer to as *can’t talk about it*. In preceding chapters, we have seen how this attitude contains the impetus toward open communication. In emphasizing the significance of language in making things become more real, this attitude upholds the value of silence. To talk too much about HD is to run the risk of making what I have referred to as ‘the always thereness of HD’ still more real. It is to rupture the ability to contain that which is most deeply feared and so go on in life as best one can. This attitude thus contributes to a more traditional stance on disclosure; it values not telling (all) because it is through the ability to disattend some things that anxiety is reduced, and hope sustained.
A third pattern or cluster of attitudes names itself according to the spontaneous if it comes up. This pattern of experience and cluster of attitudes toward disclosure is defined by the shifting relationships and ongoing flow of interaction which constitutes social life. The impetus toward or away from open communication is more variable; it does not reside within the narrative or the narrator to the same degree as it appears to in having to talk about it and can't talk about it. The emphasis on spontaneity is, nonetheless, also a way of pre-figuring recurrent situations: to talk (or not) about HD is to assess one's need or felt obligation to engage with one's intended audience but it is also to listen for cues and respond accordingly. In this way, the responsibility for communication is perhaps more equitably distributed between narrator (or speaker) and audience.

To talk about HD is, accordingly, to respond to the particular situation in a dialogical fashion. It is to evaluate on an ongoing basis the relationships one hopes to sustain as well as the goals and objectives one seeks to fulfill. This narrative therefore sustains a "vision of good" (Gordon & Paci, 1997) which upholds some admixture of the preceding two strategies and a sensibility which is both reflexive and postmodern. It values neither telling all nor not telling: each is implicated in the other and present as a potential within each communicative event.

These three ways of typifying strategies for managing genetic information are summarized in Table 12 (see next page). This table also indicates some of the tactics study participants described in enacting particular strategies for managing their communications about genetic information. In keeping with Burke’s emphasis on the need to avoid “terminating” categories, I do however wish to underscore the importance of not viewing these strategies as if they were a typology of individual attributes or fixed and therefore immutable patterns of behaviour. They are “definitions of the situation” (Thomas & Thomas, 1928); contingent inclinations which, in C.W. Mill’s (1940) terms, describe a “vocabulary of motives”; or as Burke (1973) would suggest, cultural frameworks which contribute a set of devices which are symbolic depictions of possible interpretive realities.

How we think about various situations in life and use language to represent them to ourselves and others is an integral aspect of studying human communication. Words and concepts are not neutral; they have consequences which infuse the social world with possible
### TABLE 12: STRATEGIES FOR MANAGING GENETIC INFORMATION

<table>
<thead>
<tr>
<th>strategy</th>
<th>have to talk about it</th>
<th>can’t talk about it</th>
<th>if it comes up</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>description</strong></td>
<td>active desire to initiate discussion because of therapeutic dimensions of talk and emphasis on truth telling</td>
<td>containment and shrinking from discussion because of acute awareness of potentially adverse consequences of talking about being at risk for HD</td>
<td>contextually defined, fluid endorsement of responsibility to answer questions and respond to situation as it unfolds</td>
</tr>
<tr>
<td><strong>implications for managing information</strong></td>
<td>the possibility of having HD is no different than the possibility of having cancer or most other diseases and it should not be any harder to discuss talking about it proves that it is no different because what is talked about is not hidden and if it is not hidden there is nothing to be ashamed of</td>
<td>possible symptoms of HD are scary but talking about this may encourage others to look for these behaviours knowing that others are monitoring these behaviours exacerbates the experience of possible symptoms leading to increased self-surveillance and a need to monitor others’ behaviour in order to detect whether or not they have detected something</td>
<td>there is no need to initiate a big discussion on the topic since talking about it all the time keeps it in the forefront but, where a question or request for information arises during conversation, it ought to be respected and answered honestly</td>
</tr>
<tr>
<td><strong>other considerations</strong></td>
<td>can’t force others to talk but frustration may lead to saying things that will later on be a source of regret</td>
<td>talking about it may alarm people and/or lead them to worry about their own future</td>
<td>respect for what people do not need to know and concern that talking about it may scare friends away</td>
</tr>
<tr>
<td><strong>tactics</strong></td>
<td>find a good sounding board; say it and run or sulk</td>
<td>dummy up, keep it to yourself or talk with those in the same position</td>
<td>don’t volunteer information, let someone else do the initiating</td>
</tr>
<tr>
<td><strong>implications for others</strong></td>
<td>may entail high demand for supportive behaviour and good listening skills</td>
<td>for the unaware, this may lead to surprises and, for the aware, it may be unsettling to be unable to elicit talk</td>
<td>requires sensitivity and good judgment about when to ask and when not in order to avoid appearance of poking and prodding</td>
</tr>
<tr>
<td><strong>limitations</strong></td>
<td>there is a proper time and place for talking openly and those who have common experience of “what–ifs” are best audience</td>
<td>talking with others who are at risk and who have same feelings leads to sense of camaraderie and it feels good to help others by sharing own story</td>
<td>talking with others is important when there is some practical decision to be made and/or specific action that could be taken</td>
</tr>
</tbody>
</table>
experiences. They typify and prefigure social situations. Language is, in this sense, indispensable to human experience and as such, it must be understood according to what it accomplishes, how it affects the situation and audience to which it is addressed.

Sociologists, like lay persons, use typological concepts to achieve a sense of order and coherence which enables understanding. This typifying activity can, however, be carried too far when terms and concepts are reified and become “terministic” abstractions that reduce the complexities of human acts and launder away all trace of ambiguity (Burke, 1973). Rather than pointing to phenomena of interest, they become concretized descriptions of realities which are endings unto themselves. Such may be the case with typologies that distinguish normal from pathological forms of familial communication. Further, much of what is promulgated in the popular media about family secrets might also be criticized for this tendency (Imber-Black, 1998). In this sense, it is refreshing to find that lay actors have much to say about how their strategies for, and attitudes toward, practices of concealment and revelation often do not fit the neat typologies that philosophers, psychotherapists, sociologists and others have constructed for them.

Secrets and Shame

The qualified disavowal of secrecy as a strategy for managing genetic information was, in this respect, one of the most intriguing themes which emerged from the post-results interviews with PT candidates’ and their families. When I asked (in roughly paraphrased form) “so, in thinking about your (or your family member’s) test results, are there people that you are concerned to (or have been asked to) withhold this information from?”, I encountered a range of responses which pointed to the ambivalence associated with secrecy and its family of related meanings.

Within this research, study participants identified an array of possible meanings associated with secrets, some positive, some negative, but all of which prefigured social situations in terms of a particular “attitude” (or “strategy”) in Burke’s sense of the word. Secrets were occasionally associated with finding the key to happiness and well-being — that is, they were good things to know about because they described an attitude which enhanced the quality
of one’s life, as in the “secret to eternal health”, the “secret to married life” (or even, in the more Northern reaches of the province, the secret places one goes to harvest the lucrative pine mushrooms!). Elsewhere study participants also talked about practical matters such as the impossibility of keeping secrets. Intriguingly, many such references pointed to the natural curiosity and astute observational powers of children who discovered whatever it was that was presumed secret. Colin and Emily offered one such story. They had decided not to tell their three daughters that Colin was having predictive testing but, much to their surprise, their daughters somehow knew. As Colin explained,

"...these little darlings figured it out. Somehow they figured it out. Because the morning that Emily and I were going in for the test/ I wasn’t here but the girls were at breakfast and the little one was saying grace and she was asking God “please don’t let Dad have Huntington’s disease.” So how they figured this out you know/ I mean we had tried to be so careful because we just felt that if I did have the disease and I wasn’t showing any signs we wouldn’t tell them for a while."

Secrets were, however, far more frequently cast in ominous terms and, as such, the majority of those who spoke of secrecy or other practices of concealment sought to dissociate themselves from any possible inference that they might have something shameful to hide. For instance, one PT candidate (who had not inherited the mutation) responded angrily to my question about who did and did not know her test results by emphatically asserting,

"I told you before! I don’t care who knows... I’m not hiding anything!"

Hiding things was, in this and many other similar instances, equated with the presence of “dark secrets” such that those who were most adamant in disavowing secrecy also talked about something within their own personal or family history which was bad, dishonorable or shameful. Mental illness, alcoholism, drug addiction, sexual abuse, extra-marital affairs, questionable paternity, dishonesty, lying and/or other forms of actual or contemplated deceitful behaviour were among the chief examples. These were family or personal secrets that were not generally (or in some cases had never been) talked about, things that were “buried” or otherwise “hidden”, and that were, in consequence, “bad”, “taboo”, or “dirty”. They presented secrecy in its most negative terms and pointed to that which is shameful.
Cultural Narratives About Secrecy and Openness in Communication

The gross lack of understanding, social discrimination and stigmatization which infuses much of the social history of HD was a recurrent element in many study participants’ stories. Moreover, this history has an overarching importance in understanding how everyday patterns of communication both shape and reflect broader cultural narratives about the desirability of various degrees of openness and/or secrecy in communication.

Though it is no doubt obvious, it bears repeating that commonsense and Burkean attitudes alike, are not isolated. Such attitudes, the various modalities of communication (i.e., speaking or silence) and associated practices (i.e., disclosure or non-disclosure) they endorse find their “roots and support in other contexts, social relationships, and assumptions, both medical and meta-medical”; they are connected through cultural narratives which have a broad or deep cultural influence but which are, more often than not, taken for granted and invisible, “operating in the background of attention” (Gordon & Paci, 1997:1434–35). In this sense, they are, as Ricouer (1978:59) suggests, also deeply ideological; they are part of “the code of interpretation of a concrete community which supports us” yet they are, at the same time, never fully transparent to us. It requires concerted effort to become aware of such codes of interpretation, to envisage how they shape our daily interactions as well as our larger world views.

Many study participants who disavowed secrecy as a strategy for managing genetic information sought to break with a past in which HD was misunderstood and highly stigmatized. Moreover, many spoke from personal experience in recalling instances in the not too distant past wherein a family member with HD was hidden away, strapped down, out of sight, in a back room, basement or institution. As the sister of one PT candidate recalled, We weren't {there} when my aunt had it {HD} or we weren't around it, we don't know if it was hushed up and years ago it would have been. You would have been locked in a bedroom and hidden and nobody would have ever seen you. Come to think of it... I can remember going...to visit relatives... and there was an old lady locked up/ well she wasn't locked up but she was upstairs. She was loony. I think. I could hear her hollering and moaning. “I want to get up.” And everybody just carried on like she wasn't there and I thought that was really weird but that's just the way they did it. Where nowadays people know enough/...people are in homes when they are like that/ which is maybe not good but.. those people were hidden away.
Furthermore, the lack of understanding which led to the practice of hiding people away was itself shameful and hence, the secrecy which surrounded HD bespoke the recursiveness of all such "shame states," as Scheff (1990), in particular, has done so much to illuminate. The recursiveness of such "shame states" became painfully clear to me when, in several interviews, I felt extreme discomfort in hearing a study participant describe the shame associated with the social ostracization of persons affected with HD.

In describing how her father had become deeply ashamed of her mother when it became apparent that she had onset of HD, one woman looked away from me as she haltingly recalled her own complicity in the process,

I don't think we realized/ (...) we didn't understand the (...) mentally ill back then so you hid them (...) sort of thing. Even my generation/ it/ I was kind of bad (...) but it/ actually my Dad was the one that really didn't want to talk about it/ you know it was all really really degrading to him.

And in a second instance, a much younger woman reconstructed what it must have been like for her grandfather to be institutionalized because no one really understood how to care for or communicate with persons affected with HD.

Dad's dad was institutionalized long before I think he needed to be because it was such a hush-hush thing you know/ you didn't talk about your sicknesses and it was also considered much more of a mental thing rather than a physical thing. ...

Writ large in one reading of such stories is the view that things are different now. Huntington Disease is no longer so grossly misunderstood. Though public awareness is not yet high, there has been real progress in understanding the disease and assisting families to cope with its most devastating consequences. Further, though there is not yet an effective prevention or cure for the disease, scientists are rapidly closing in on potential ways and means of forestalling onset and improving the ability to manage distressing and disabling symptomatology. There is no reason not to have hope and no reason to treat those who suffer from the disease as less than fully human. Moreover, we need not regard having a family history of the disease as if it were some kind of taint.

Not having anything to hide was, in this reading of such stories, a resounding
endorsement of progress and the importance of acting in ways which promote the laudable goals of demystifying and destigmatizing the disease. It was, in Gordon and Paci’s (1997) terms, that historically-specific “vision of good”, or means of enacting our most fundamental values, which sanctions openness in communication. It is through such openness that truth is discovered, human integrity sustained and individual autonomy upheld. The value of scientific knowledge and access to the latest information is, accordingly, held in high regard because knowing what to anticipate is the precursor to responsible action and informed decision-making. Seen through this lens, predictive testing provides those at risk for HD with the opportunity to “own up to the truth”, “see the light” and stop “running away from the solution.”

Another reading of how study participants’ stories reflect larger cultural narratives and their associated “visions of good” (Gordon & Paci, 1997) reveals a far less optimistic view. Scientific understandings of HD are advancing but not quickly enough to have an immediate or imminently foreseeable impact. Huntington Disease is still widely misunderstood and though there has been some progress in unraveling the molecular mysteries of the disease, it remains a dreaded family heirloom or curse which is best referred to as little as possible. Predictive testing, it follows, is something of an anathema. It is both alluring and fearsome in its capacity to confront those at risk for HD with the opportunity of knowing and articulating what might be better left unknown and unsaid.

A third reading of study participants’ stories yields a more contextually situated attitude which is, perhaps, characterized less by its optimism or pessimism than it is by its inherent pragmatism. Science will, in this view, proceed and there will eventually be an effective form of therapeutic intervention for HD. This may not happen overnight but there is cause for hope. Further, Huntington Disease is not the worst thing that can happen. There are other illnesses and human tragedies which exact a similar or greater toll and, given the imposed choice between proceeding with life and waiting for HD to happen, there is but one way to go on. Predictive testing is neither good nor bad; nor is the information acquired through predictive testing inherently promising or menacing; its meaning and significance is contingent on what PT candidates and their families jointly make of it.
Genetic Housekeeping

Strategies for managing genetic information are created and recreated in and through the micro level interactions which occur in the “everyday and everynight” worlds (Smith, 1987) of PT candidates and their families. Within the context of this chapter, the patterns inherent to such micro level interactions comprise the who tells what to whom, when, and where part of the story.

As I have stressed throughout this dissertation, it makes little sense to focus on such aspects of communication without first (or also) engaging in the struggle to understand what such communication means to communicants in their everyday lives. In emphasizing the content and outcome of who says what to whom over and above the meaning and significance of such communication, there is a real and omnipresent danger of slipping into a referential view of language and a sender–receiver model of communication. Neither facilitate understanding of how a wide range of human capacities for communication may be employed not just to transmit information but to produce and reproduce meaning, create and sustain social relationships. Moreover, such approaches often tend to absolve the researcher (as observer) from the need to realize her own “hermeneutic participation” in the research (Krippendorf, 1994). For these and other reasons, then, I have endeavoured to remain firmly within the interpretive realm. The time is now right, however, to refer to other outstanding aspects of the patterning of familial communication about genetic information.

The central theme that I present here has to do with what Shakespeare (1992) refers to as “genetic housekeeping” — that is the sometimes tidy and, at other times disordered, practices of communication which are, like housekeeping, a necessary but not always attractive aspect of everyday life. As we shall see, gender is especially significant here in that it is women who take on the primary responsibility for communicating within the family about genetic information and, moreover, it is within the home that much of this “genetic housekeeping” occurs.

As I began this chapter by saying, secrecy is often associated with privacy and privacy, in turn, with the intimate world of home and hearth. The overlap of secrecy and privacy is, indeed, often so complete as to preclude any meaningful distinction between the two (Imber–Black, 1998). Notions of privacy — that is, what is “nobody’s business outside the family” —
are, however, central in understanding how study participants distinguished between those who have a legitimate interest in the information acquired through predictive testing and those who do not. To make such a distinction is, after all, to set apart those who are the subject(s) of a secret from those who are merely unaware. The dynamics of disclosure are, in these two situations, fundamentally different.

Here it is helpful to outline briefly some of the most basic features of the gendered dimensions of genetics activity. As medical geneticist Peter Harper (1996) maintains, “for the most part it is women who form the central focus of the family–based problems that genetic diseases create.” The reasons offered for this are numerous but a short list would have to include mention of the way that women are typically at the centre of the web of family communications (The Women’s Project in Family Therapy, 1988); as “kin–keepers” who keep track of and exchange knowledge of kin, women also bind family together. Further, women are unpaid health workers for their families and often have primary responsibility for communication about, and translation of, clinical information within the family (Stacey, 1996).

With respect to Huntington Disease, in particular, women often take on the primary role in exchanging information and communicating about the family history of HD. Further, it is women who marry into the HD family who often play the most central role in instigating such discussions (Shakespeare, 1992). Further, though it has been observed for some time that twice as many women as men request the predictive test, there are few studies which acknowledge gender as significant in understanding how at risk individuals interpret hereditary risk and its modification through predictive testing. Bloch et al (1989:222) suggest that women’s more intimate involvement with reproduction and child–rearing coupled with “the apparently greater capacity of men to deny their feelings and avoid looking at the painful implications of their situation” may help to explain these gendered aspects of genetics activity. None of these aspects have, however, yet been adequately examined.

Not having had the opportunity to interview any men who learned that they had inherited the mutation associated with HD, my ability to respond to such assertions is somewhat constrained. I am, nonetheless, confident in stating that there were, in this research, many male study participants who spoke quite openly about painful and difficult subjects. Maggie’s older
brother Jerry was one. Contemplating how the women in his family had always taken responsibility for family health concerns, Jerry offered a thumbnail ethnography of the gendered dynamics of genetics activity within his own family. Much of what he had to say supported the idea that there are some things that men just do not tend to talk about.

As Jerry began by saying, it was his sister Maggie who had first noticed the signs of onset of HD in their brother Louis. As such, she prompted Louis to go to the doctor and request a neurological examination and, when he subsequently received a diagnosis of HD, Maggie also took on the responsibility of communicating this news to several members of the family. As Jerry said,

{My sister} told me first. I had all the low-down before Louis {my brother} even mentioned anything... She pressured him to see the doctor and go for these tests... perhaps she's more observant than I am on these sort of things. Most women might be...I guess because they're always bringing up the family at home. Kids getting sick and that. Mother's got to deal with it before father finds out about it.

And also the women seem to be more mentally able to handle the situation than men. They seem to stay calm under the duress of a sickness, where a man tends to go to pieces...and they don't have to deal with it as much either so they don't understand it as much which would make them kind of afraid of it... Cause I know most of my uncles and they wouldn't say anything when we were sick. A man just didn't admit to those sort of things.

Jerry was unusual in articulating such a fine-grained accounting of the reasons for women's disproportionate involvement in, and responsibility for, matters of family health. Most male study participants were not inclined to comment on the issue at all; thus it seemed as if interest in the subject was itself gendered.

Women, on the other hand, spoke frequently about the gendered nature of familial communication. Indeed, most of the female PT candidates involved in this research were themselves the most active communicators in their family when it came to discussing the family history of HD and its implications for various family members. Heather lived too far from most of her family to take on this role but, as she explained, it would be no problem to get the news out to family when her predictive test results came in.

My Gran is the communicator in our family. She just passes it [any news] on the minute you hang up it's like the tom tom drums, jungle drums, she's on the phone and she'll tell all my brothers and sisters... then she'll tell my godfather who is also her son and my uncle who is her other son then she'll tell Karl and Elaine who just had their second baby and then Auntie Charlotte ... then of course there's the ladies in the tea room so she's like the little matriarch of the family she
just passes it all around...so that's how it [my test results] will be distributed. She'll expect a call Friday, Thursday cause she knows I'm going Wednesday so she'll expect a phone call first thing Thursday morning which will be afternoon for her.

Heather's family was not unusual in delegating such responsibilities. Indeed, there were very few families involved in this research in which one, or sometimes two, women (who were not necessarily at risk for HD) did not take on the role of key communicator. The duties and responsibilities of such a role were many but one important function was to serve as a sort of repository of the family history of HD. Women who took on this aspect could generally be relied upon to know who in the family had HD, when they first began to show symptoms and, significantly, who else in the family knew about and/or was concerned about issues of familial risk.

Study participants also described numerous instances in which some especially sensitive disclosure or other communicative interaction was reserved for this person. The disclosure of the family history of HD to a potential son or daughter-in-law was one such task. As Elizabeth recalled, she and her sister Brenda first learned about the family history of HD when they overheard their mother explaining to their brother's fiancee that there was "a thing in the family". After that, it was

off and on with Mum... it came up again when the next brother was getting married and then I would have wanted to ask some questions too.

As Elizabeth explained, her mother's role was to ensure that potential spouses knew what they were "getting into." Part of what her mother did, however, also concerned something a bit more subtle. She taught her daughters to observe the bounded nature of what was "family business."

It was important (to Mum) that the family know (about the family history of HD) but it wasn't important that other people know. ...Like it was just/ like a little bit different. Like private things were private things and it wasn't that you couldn't talk about it in our house. It wasn't that way. It's just, if it's your family business, you didn't necessarily talk to somebody else about it.

The family history of HD was a private thing but this did not mean that it wasn't talked about; there were appropriate limits and they were, in many instances, proscribed by notions of home and the more intimate relations which pertain within the private sphere. Home was, therefore, the one place that study participants almost universally identified with the
conversational spaces in which it was permissible to talk about HD.

This literal association between home and talk about HD does not, however, exhaust the relevance of housekeeping as a metaphor which provides an overarching sense of coherence in locating the understandings that study participants had of the boundedness of their communicative interactions. The talk which occurred within the container of home or family was itself structured by the metaphors of housekeeping. As such, study participants talked about how at risk individuals are often either “in or out of the closet” about the family history of HD. Some families try to “sweep it [the family history] under the carpet”. Finding out about the family history therefore requires “digging up a lot of dirt” and misunderstandings often need to be “ironed out”. Predictive testing may be a process of “tidying up” or “putting it all {HD} to bed”. And, depending upon the outcome, the test may mean that it is important to put Huntington Disease “on the back burner” or that “everything gets washed away” so you can start with a “clean slate”.

Genetic housekeeping is, then, both a type of domestic work that is entailed by genetics activity and a metaphor which helps to elucidate the central aspects of study participants’ conceptual schema for describing and carrying out this work. The question that remains is ‘is housekeeping a good metaphor’? Does it generate an appropriate array of possibilities for communicants as well researchers who seek to understand familial communication about genetic information?

Let us briefly consider some features of housekeeping as housework and, more generally, as the mundane domestic issues that one must take care of on an ongoing basis. It is now widely recognized that housekeeping is work and that this work is essential to the maintenance of everyday life (Cowan, 1989). It is most visible when it needs doing and yet its overriding feature is that it is never finished. It requires knowledge, skill and expertise yet is often taken for granted and thus, unacknowledged. There are many responsibilities that are seldom equitably shared amongst all parties. It is gendered and it often reflects the sexual division of labour in its historically and culturally specific forms. It is not something that many people admit to enjoying and thus it may be treated as an activity that is best dispensed with as quickly and easily as possible.
Feminist philosopher Virginia Warren (1992) tackles a different issue from a similar vantage point. Warren argues that there are, within contemporary bioethics, two categories of moral issues — crisis and housekeeping issues. These categories contrast in several ways that are of immediate relevance here. First, with crisis issues, moral decisions are (more or less) final. A problem arises, a decision is made and there is a sense of accomplishment. With housekeeping issues, however, “the problematic situation is ongoing, rather than resolved once and for all” (Warren, 1992:36–7).

Second, the significance of crisis issues is immediately apparent. They are testaments to the difficulty of facing a challenge and housekeeping issues, by contrast, appear trivial. Even when housekeeping issues are handled well, there is little sense of accomplishment to mark their collective significance. Third, crisis issues typically present a narrow range of alternative actions. By contrast, “housekeeping issues commonly require us to reassess large parts of our lives: our character traits, how we think about ourselves, and how we relate to others. Their impact is thus felt long after a particular crisis is past” (Warren, 1992:37).

Crisis issues may also require that we reassess our lives and how we think about ourselves but Warren’s (1992:38) point, and the point I wish to conclude with here, is that even though crisis issues may have many far-reaching consequences, housekeeping issues require that we examine things in a wider context and on an ongoing basis. They call into question our taken for granted assumptions and ask us to reconsider our how we ought to act, within the constraints of our everyday lives, in order uphold our most basic moral values and commitments.
A PROMISING CONVERSATION

The prototype of a “good” conversation offers a refinement: Many dialogues are judged worthwhile because the speakers wish to continue them and not because a truth has been attained, a problem solved, or a bargain closed. Often the speakers feel rewarded because the dialogue has turned into an as yet unplumbed opportunity for understanding themselves more fully. We say it is a promising conversation. (Rosenwald, 1992:273–274)

Rosenwald expresses precisely the feeling that I most enjoy when engaged in stimulating conversation. Everything else recedes in importance as the dialogue carries speaker and listener along in a unending stream of questions and assertions, anticipated and unanticipated responses. There are, as Bahktin (1986) suggests, no last words in such conversations because “the first and foremost criterion for the finalization of an utterance is the possibility of responding to it, or more precisely and broadly, of assuming a responsive attitude to it.”

In finalizing a very long utterance, this chapter also now invites response. The outline that I pursue here is as follows. I first summarize the findings of this dissertation, emphasizing the most central substantive as well as some of the more easily overlooked methodological aspects. Following from this, I also: 1) set out specific recommendations for clinical practice and research; 2) consider the practical relevance of the research findings for families at risk for HD as well as the HD community at large, and 3) describe some of the significant ethical and methodological aspects of doing qualitative research on hereditary risk and predictive testing. I then discuss some of the limitations of the research design, study sample and method of analysis. This provides the necessary backdrop for consideration of other significant practical and theoretical implications of the work.

Finally, I conclude by offering some recommendations for further research on the social meanings as well as clinical experiences of hereditary risk and predictive testing. Attending to what is of the greatest importance to families at risk for HD, I emphasize the practical significance of conversation and storytelling. As Cruikshank (1998) demonstrates, oral storytelling also has the potential to “destabilize” and “transform official orthodoxies”. This
potential is, I suggest, of overarching importance if lay knowledge is to be properly valued and
lay persons are to have a meaningful part in public dialogue about the new genetics (Kerr,
Cunningham-Burley, & Amos, 1998).

**Risk and Uncertainty in the Age of Molecular Medicine**

We understand now that uncertainty is not a temporary nuisance, which can be
chased away through learning the rules, or surrendering to expert advice, or just
doing what others do — [it is] a permanent condition of life. (Bauman, 1995:287)

In the absence of effective therapeutic intervention, predictable genetic testing is about
risk and the management of uncertainty — two key aspects of modernity which have recently
garnered considerable attention (Beck, 1992; Giddens, 1990). Moreover, predictable genetic
testing creates a “third space” which exists outside of the categories of either–or that we
conventionally use to organize health and illness related experiences (Wexler, 1995). Learning
what it means to inhabit such a space and what in turn, it may mean for an increasing proportion
of the population to inhabit such a space is surely one of the most intriguing and omnipresent
questions of the late 20th century.

As Rothman (1996:32) suggests, it is “in the nature of this growing world of genetic
prediction that we offer information ‘out of time’, abruptly and without context of lived
experience.” Illness no longer comes “in a context, in the course of a life, unfolding itself over
time”. It comes before its time. Genetic information is, for this and other reasons, full of
paradoxes. It is highly personal and yet it is also a fundamentally social artifact which tells us
not only about our own past and probable future but also about that of those who have come
before and will come after us. Further, because genetic information or DNA has become widely
perceived as the molecular text which defines each of us it holds out a special symbolic
relevance as well as a certain mystique. As Nelkin and Lindee (1995) argue, the gene has
become a “cultural icon” with enormous potential to shape public reactions to the institutional
and political uses of genetic information.

Given that sociology has had a long and tumultuous history of confrontation with
sociobiology and other forms of genetic determinism (Lewontin, 1991), and moreover, that the
latest salvos in this battle cast doubt upon well-established notions of social equality and human
agency (Brock, 1994), it seems only a small stretch to suggest that all sociologists ought to concern themselves with the new genetics. To what ends genetic information will be used, who will have access to it and how it will shape our understandings of health and illness, uncertainty and risk are questions that remain, even at this socio–historical juncture, open. As Kenen and Schmidt (1978:1117) noted over twenty years ago with respect to the African American experience with mandatory sickle cell testing, “modern technology may have introduced a new biological and social label — ‘carrier’ — with yet unknown psychological and social consequences ... the newly identified carriers of mutant alleles are, in a sense, both psychological and social pioneers”. It is within the context of efforts to chart this new terrain that I situate the findings of this research.

Summary of Findings and Their Implications

Despite the rapid proliferation of new discoveries in the field of molecular medicine, there are, at present, few empirical studies on the social shaping and implications of the new genetics (Richards, 1993). Moreover, although there is now a large literature on Huntington Disease and predictive testing for a variety of adult onset disorders, most of this literature describes the findings of quantitative studies conducted by service providers within a clinical setting (Lerman, 1997). Such research serves an important evaluative purpose but it seldom considers the familial and social dimensions of predictive testing (Hayes, 1992). Further, such research almost invariably treats the test outcome as the primary independent variable and the PT candidate as an isolated, autonomous individual. There is, in short, an overall neglect of the array of factors which both shape and differentiate the predictive testing experience.

The research presented here breaks with these traditions in order to investigate the meaning and significance of hereditary risk and predictive testing for HD within the context of PT candidates’ and their respective family members’ everyday and “everynight” lives (Smith, 1987). Following a prospective study design, the research includes a total of 102 in–depth interviews with sixteen participants in predictive genetic testing, thirty–three of their family members and/or close friends and one couple where the wife was at risk but did not want to proceed with predictive testing.
Emphasizing the interpretive understanding of social action (Weber, 1968), the hermeneutic approach of the dissertation underscores the processual nature of predictive testing as well as the overarching significance of interpersonal communication in producing and reproducing the social realities in which genetic information acquires a particular salience. The focus on communication is integral because talking is itself a means of allocating things a definite place in the world. As Berger and Luckman (1966:174) propose, "language realizes a world, in the double sense of apprehending and producing it." Likewise, the emphasis on narrative reflects the way in which account formation is an integral aspect of how people go about making sense of their health and illness related experiences (Stimson & Webb, 1975).

Focus on Everyday Life

One of the strengths of this research is its focus on the everyday lifeworld of PT candidates and their families. As Conrad (1990) suggests, the task of understanding the intersubjective meaning and experience of illness must be grounded in the social organization of the sufferer's world. Much research on illness experience has tended to presume that studying the experience of patienthood is the same as studying illness experience and it is not. This has significant research implications: it suggests that researchers must turn away from provider–patient interaction as the central analytic focus and go beyond clinical settings in order to explore how at risk individuals and their families understand hereditary risk when they are at home, work or doing whatever they do when they are not patients. The research locale is extremely important in this: the stories that people tell when they are 'probands', patients or clients in the clinic differ from the stories that people tell when they are busy living their lives (Burgess, submitted). In particular, the range of elements which have topical and interpretive relevance (Schutz, 1970) are different.

The medical genetics clinic offers an institutional space for the telling of stories about the decision to request predictive testing; informal discussions with family and friends, organized Huntington Society support group meetings and/or the at–home interviews conducted for this research offer other such spaces for the telling of stories. Each such space offers the opportunity to tell a slightly different story; that is to say the narrator's selection and ordering of
relevant events is always conditioned by the local expectations of both the narrator and listener for what is "properly tellable" and moreover, how properly accountable stories ought to be told (Gubrium & Holstein, 1998). Although there are many different kinds of stories, the coherence of all stories is at least partially "preordained" by the communicative genre through which it is conveyed. Coherence is, however, also "occasioned" in that the coherence of a story produced under one set of narrative conditions is not necessarily the same coherence that is produced in another (Gubrium & Holstein, 1998). Different details may be selected and different explanatory frameworks (Linde, 1987) deployed.

Within the context of this research, study participants were encouraged to talk about their experiences in whatever ways seemed salient to them at the time. This approach therefore generated a very different set of stories than are typically heard in the clinical setting. In particular, study participants drew upon a wider array of life experiences and events than they might otherwise. This enhanced both the scope and depth of the stories (Mishler, 1991); moreover it brought new stories to the fore which sometimes challenged and, at other times, upheld existing clinical assumptions. The variation in the stories was, however, a testament to the fact that there is, at this socio–historical juncture, no prototypical HD family or experience of hereditary risk. Indeed, if there is one overarching message that I would like to convey to researchers and clinicians, it is this: we must attend more closely to the way in which we listen for some stories and not others.

Foregrounding study participants’ storied accounts of their experiences, the dissertation focuses, in particular, on three narrative ‘moments’: as such, the central data chapters explore how PT candidates and their respective family members described and narrated their experiences of: 1) learning about the family history of HD, 2) deciding to request the predictive test and, 3) hearing and making sense of an informative result. These three narrative moments correspond roughly to the beginning, middle and ending of the story of predictive testing as it was related to me by study participants during the course of this research.

Together these three narrative moments situate study participants’ stories about familial communication within the context of the unfolding sequence of events which comprise the story of predictive testing. Moreover, each illuminates the diversity of experiences described by PT
candidates and their families and thereby broadens existing understandings of what it means to undergo predictive testing. Finally, each addresses an aspect of predictive testing that has thus far been relatively neglected within the existing literature.

**Becoming Aware of the Family History**

The story of predictive testing begins long before at risk individuals arrive at the clinic for their first counselling session: it is, however, not always easy to specify exactly when the story begins. As I discovered during the pre-results interviews with PT candidates in particular, the transition into initial awareness of the family history of HD is sometimes gradual and at other times marked; it may occur when a mysterious family illness is finally diagnosed (as in stories of *solving the mystery*) or it may emerge when information that has long been withheld is finally revealed (as in stories of *discovering the secret*). Alternatively, awareness of the family history may become a taken-for-granted aspect of the experience of growing up in a family where someone has been diagnosed with HD (as in stories of *learning by osmosis*) or it may come *out of the blue* when a socially or geographically distant relative is suddenly found to have the disease or when there is a new mutation in a family with no previous history of HD.

An appreciation for the diversity in how PT candidates experience initial awareness of the family history of HD is significant for several reasons. *First*, it underscores the importance of considering whether recent advances in biomedical knowledge have indeed translated into a corresponding increase in physicians' as well as families' awareness of the disease. Misdiagnoses and/or the lack of a documented family history of the disease have, as several study participant's stories demonstrate, been significant factors in shaping patterns of familial awareness. *Second*, an appreciation for the diversity in how PT candidates became aware of the family history focuses attention on patterns of doctor-patient as well as familial communication. Information may, at times be withheld from those who have a legitimate interest in it, and though the shift toward full disclosure (Nelson & Nelson, 1995; Seale, 1997) and increased emphasis on patients' rights of access to medical information (Shaw, Westwood, & Wodell, 1994; Shaw, 1987) now implies that this is less likely to occur within the clinical context, the concealment of information about the family history of HD remains as a salient factor within the
sphere of familial and social interactions. Third, there has, within the existing literature on predictive testing, been much speculation about the combined effects of age and duration of awareness in shaping PT candidates’ desire to request predictive testing (Quaid & Morris, 1993; Steenstraten, et al., 1994). Little attention is, however, given to unpacking the complex meanings of such awareness. As this research suggests, knowing or not knowing that there is a family history of HD cannot be treated as if it were a dichotomous variable nor can it be assumed that such awareness unfolds in a linear fashion.

Awareness of the family history of HD involves several discrete but interwoven aspects. These include: 1) recognizing and attaching significance to the words ‘Huntington Disease’; 2) becoming aware that something was (or is) wrong with a family member, and; 3) acquiring information about the symptoms, onset and hereditary nature of HD. Only when each of these aspects comes together is it meaningful to talk about the sort of “active open awareness” (Timmermans, 1994) which is often presumed (rather than explicitly investigated) in much of the literature on awareness contexts. Further, it is important to note here that the fluid nature of such awareness means that it ebbs and flows over time; awareness is, in this respect, a social phenomenon which is intricately bound up with the social and emotional as well as cognitive process of remembering (Zerubavel, 1996). In short, memory involves much more than the mere recall of events; the social process of remembering creates the opportunity to tell a story and, in so doing, re-evaluate the meaning and significance of past events (Riessman, 1993).

For many of the families who participated in this research, the storied act of remembering was often tied to a reassessment of a deceased family member’s odd behaviour. Equipped with the knowledge that such family members probably suffered from HD rather than insanity, homesickness, war nerves or some form of illness, some study participants expressed profound regrets about not understanding what was wrong and hence not responding with an appropriate level of empathy and support. Others commented on the degree to which the discovery of a common bond (i.e., hereditary risk) invoked powerful new feelings of connectedness.

The awakening of such feelings of connectedness and belonging is part of what I refer to as the process of “re-membering.” Re-membering is, in turn, central to understanding how an
awareness of hereditary risk shapes at risk individuals’ sense of self-identity within the relational context that is family. Here it is, however, also important to note that the transition into initial awareness of HD may disrupt feelings of connectedness and belonging. This is especially the case where family members adopt competing explanatory models (Kleinman, 1988) for making sense of the emergence of HD and/or hold divergent views about the desirability of communicating openly within the family about HD.

**Understanding What it Means to Make a Decision**

In framing the experience of deciding to request the test as a “decision” which presents an opportunity for choice, it is often assumed that PT candidates and their families consciously perceive that there is a decision to be made. A close reading of study participants’ stories about deciding to request the test revealed that this construction of what it means to make a decision does not necessarily apply.

The stories that PT candidates tell about deciding to request predictive testing reveal that there are many pathways to the clinic. The plot structure of these stories is differentiated by: 1) the temporal unfolding of events; 2) degree to which decision is constructed as an opportunity for choice, and 3) narrator’s orientation toward talking or not talking with others about the meaning and implications of being at risk for HD. As stories of having to know suggest, some at risk individuals do not enter into a conscious process of decision-making; the decision is, for all intents and purposes, a self-evident act that does not, on the face of it, require consideration or much in the way of discussion with others. Such stories stand in sharp contrast to stories of evolving toward it in that the latter feature a gradual or incremental process of weighing up the implications of testing before arriving at a feeling of readiness and/or willingness to reveal the decision to others. Finally, there are also stories of taking the decision which do not fit either of the preceding patterns. In such stories, the PT candidate experiences an abrupt change from not caring about the test or wanting to decide to feeling that it is time; thus these stories feature a pivotal point which involves the narrator in seeing that there is an opportunity or new impetus to make a choice where there did not initially seem to be one. Such pivotal points may arise with the acquisition of new information about the availability of the test (e.g., from the doctor) or
they may seem, in hindsight, to be quite inexplicable.

Many studies have investigated PT candidates' reasons for testing (Binedell & Soldan, 1997; Bloch, et al., 1989; Decruyenaere, et al., 1995; Evers–Kiebooms, et al., 1989) but very few consider how PT candidates (and their families) arrive at the decision to request the test (Meissen, et al., 1991). From a clinical perspective, this is an important question because the decision to request the test is supposed to be voluntary and autonomous; it should not be unduly influenced by any other person or agency. The stories PT candidates and their respective family members told about the decision were, in this sense, intriguing. On one hand, almost all family members upheld the clinical stance that it is the PT candidate alone who must ultimately decide whether or not to request the test. On the other hand, however, there were very few PT candidates who did not mention a felt obligation to others as an integral part of their story of deciding to request the test. Such tensions stretch the meaning of autonomy and point to the inadequacy of viewing decision–making purely through the lens of rational choice.

The diversity in how PT candidates and their families experience the process of deciding to request the test suggests that we must cast the net very broadly indeed if we are to understand what it means to make a decision about whether or not to request the predictive test. Stories of *evolving toward it* confirm what the existing literature suggests — that is, that many at risk individuals experience a period of protracted anxiety and uncertainty that is an integral aspect of their decision to request the test. Nonetheless, these stories also demonstrate that the knowledge that there is a test has a significant bearing on the awareness and experience of risk. In particular, such knowledge contributes to the teleological character of deciding. Imagining the decision to proceed with the test as the only definitive outcome alters the construction of what it means to make a decision.

Stories of *having to know* may feature what seems, on the face of it, to be a snap decision. Nonetheless, PT candidates who tell this type of story may well know exactly what they want and need to do because they have already experienced other similar life events. In particular, past experience with serious illness and/or other life–threatening events may contribute a feeling of familiarity, and perhaps greater ease with, issues of human mortality. Alternatively, stories of *having to know* may reflect the taken–for–granted expectation that all at
risk individuals who are eligible will automatically get tested. Where this is the case, such stories ought to sound a cautionary note about the pace at which predictive testing is becoming routinized. Moreover, such stories also raise the possibility that misinformation about the prevalence of predictive testing is, in and of itself, a factor which shapes the normative expectations of at risk individuals and their families.

Stories of taking the decision pose significant questions about how it is that a decision is perceived as a decision, and why some moment or event is singled out as a significant turning point. As such, these stories would seem to recommend measures which preserve rather than resolve the sense of ambivalence which precedes the decision to request the test. Especially where a potential PT candidate has not yet informed others of their intention to request the test, it may be important, as Meissen et al (1987) suggest, to preclude verbalization of a premature sense of commitment to the test. Nonetheless, given that many PT candidates have already articulated such a commitment to others long before they arrive at the clinic, it may be that there is very little that service providers can do to cultivate such ambivalence and preserve a sense of contingency in deciding whether or not to proceed with the test.

One significant implication of this focus on the diversity in PT candidates' experiences of deciding to have the test is that it would be beneficial to at risk individuals who are considering having the test to have access to the stories of those who are undecided and/or who have decided against having the test. Such stories could provide a valuable counter to the predominant clinical "discourse of potential benefits" (Boutté, 1988) by highlighting how decisions become constructed as decisions. As one woman who decided against having the test suggested, it is difficult to view the decision not to have the test as a decision when there is so much emphasis placed on the value of being informed. Having access to all of the available information is, at this socio–historical juncture, generally seen as an integral aspect of responsible action (Rothman, 1986).

**Hearing and Making Sense of an Informative Test Result**

Though many at risk individuals and their families deviate from the framework of scientific genetics when articulating their understandings of hereditary risk (Cox & McKellin, in
press; Richards & Ponder, 1996), there is a sense and logic to lay understanding which ought to be viewed as a form of expertise rather than a deficit (Kerr, Cunningham-Burley, & Amos, 1998a, 1998b). People are not tabula rasa and it is important to acknowledge the significance of such lay expertise in understanding how PT candidates and their families arrive at an intersubjectively meaningful interpretation of the process and outcome of predictive testing.

The language that is used during clinical disclosure of the test results both constructs and perpetuates problems which contribute to the deficit model of lay knowledge about genetics. In trying to distinguish between everyday and clinical frames of reference, study participants often expressed their frustration; the terms used to designate the presence or absence of an abnormal allele—that is, a ‘positive’ or ‘negative’ result — were, in this sense, especially problematic. Further, such problems in communicating about the outcome of the test spilled over into the process of interpreting the meaning and significance of an informative result.

Making sense of the test results is an active process: information is not merely disclosed or, even more misleadingly, ‘given’ to PT candidates during disclosure. Nonetheless, much of the existing literature on predictive testing for HD treats the test outcome as if it were an independent variable or stimulus that creates measurable effects. In contrast, the findings of this dissertation support the view that the test results have no inherent meaning when disconnected from the context of PT candidates’ everyday lives, familial relationships, communicative interactions, and access to material, social and cultural resources. As such, the test results do not determine a life course, biography or trajectory, nor do they impose a particular story line, plot, genre or style of narrative. Test candidates and their respective family members make sense of hereditary risk and the process of predictive testing within the context of their ongoing life history and biography (Frankenberg, 1993; McKellin, Cox, & Burgess, 1995). The meaning and significance of the test outcome is therefore seldom unambiguous when situated amidst the messiness and contingency of human life.

Just as it was important to parse study participants’ awareness of the family history of HD into several discrete but interwoven aspects, so too was it important to parse study participants’ evaluations of the test results into their various components. Many PT candidates rejected the bifurcated set of possibilities which the clinical schema for interpreting the test
results seemed to impose: good and bad news was, in consequence, seldom straightforward. Exactly what was being referred to when PT candidates and their families talked about their experiences of making sense of an informative test result was, however, sometimes difficult to discern. At times, the context of the emergent conversation suggested that study participants were referring to the actual test result, whether or not it was expected and how it generated feelings of ambivalence. At other times, however, study participants shifted to talk about the importance of having the information and being prepared. Finally, study participants also incorporated, as an integral part of their stories, some expression of relief that the process of predictive testing was complete. Each of these components was evaluated somewhat differently and, I suspect, contributed to the way in which an informative result was seldom unambiguously presented as ‘good’ or ‘bad’.

If the language of disclosure and these interwoven components of what it means to have an informative result are not already a complex amalgam, there is an additional problem for the researcher or clinician intent on discovering how PT candidates and their families make sense of the information provided during clinical disclosure. Much research on health–related communication neglects to consider lay actors’ self–reflexive awareness of what is going on during communicative interaction. Nonetheless, as Giddens (1986) has long argued, the task of understanding the meaningful social world as it is constituted by lay actors involves a constant slippage between the frames of meaning generated by lay actors and the metalanguages invented by social scientists and, I might add, clinicians. Lay and social science or clinical understandings are not, however, merely first and second order conceptualizations of the social world: appropriation may occur in both directions when knowledgeable lay actors reflect upon and monitor the frames of meaning within which they orient their conduct and within which they observe others observing their conduct. Such monitoring was an integral aspect of how PT candidates, in particular, experienced clinical disclosure. Indeed, an overarching theme that emerged from the post–results interviews concerned the way in which PT candidates felt as if they were not responding to the disclosure session in the clinically–expected way.

Resistance to clinical and other expectations was reflected in the way in which many PT candidates drew upon the narrative ‘not’ as a stratagem for interpreting the meaning and
significance of an informative result. This stratagem appeared in various forms in the stories of those who learned that they had not inherited the mutation as well as those who learned that they had. Moreover, it was salient to the ways that PT candidates and their respective family members described the cognitive, emotional, biographical and communicative dimensions of their sense-making activities. Saying what their experience did not seem to entail and, moreover, how it deviated from the expectations of others, was a means of preserving a sense of contingency. Further, it allowed PT candidates and their respective family members to arrive at an intersubjectively meaningful interpretation of the situation by selectively adopting and modifying rather than bluntly accepting clinical and other culturally available schema.

In summary, many study participants offered, in their stories, a metacommentary which supports the view that there are, at any given time, preferred cultural narratives about illness experiences and, moreover, about communication about illness experiences. Whether it was a subtle resistance to the expectations that PT candidates perceived in how they were monitored during the results session or an outright rejection of the clinical schema, study participants expressed in varying degrees that they felt pressed to respond in prescribed ways to the experience of learning their test results. In this respect, it is essential that researchers take very seriously the standpoint of illness sufferers and their families when considering and co-constructing the conversational and discursive possibilities for, as well as constraints on, the ability to tell some kinds of stories about health and illness but not others. The voice of medicine often interrupts the voice of lifeworld, subverting the patient/layperson’s efforts to construct and tell their own story in a socially meaningful way (Mishler, 1984; 1991). The contemporary impulse toward the empowerment of illness sufferers is a positive development which counters the medical world’s tendency to interrupt. Nonetheless, when such opportunities for personal empowerment become moral imperatives, we add to the work that illness sufferers and their families must do. As several PT candidates emphasized it is sometimes important to put HD “on the back burner” and just get on with life: the endless introspection and teasing apart of one’s experience that is expected in counselling and, perhaps in research such as this, may therefore sometimes become a burden rather than a much vaunted opportunity to tell one’s story.
Strategies for Managing Genetic Information

The phrase which names this dissertation — “It’s Not a Secret But” — is intended to suggest a sense of ambivalence about the experience of sharing information about oneself and related others when such information is constructed as a powerful source of, as well as an ever-present threat to, self-identity, intimacy, and social life (Goffman, 1959; 1963). The phrase is, however, also intended to evoke a series of questions which have to do with the complexities and ambiguities as well as alembicating features of interpersonal communication about genetic information and predictive testing.

Within this context, genetic information is ambivalently cast as ‘not a secret but’ for several distinct reasons. First, as many study participants emphasized in talking about specific instances of disclosure of the family history of HD and/or the test results to family and friends, genetic information is not a secret because some portion is revealed but, at the same time, some other portion is withheld. Though partiality is a feature of all human communication, it is especially salient to understanding the way in which genetic information is presented to others. Each disclosure varies in terms of the quantity, quality and clarity of information presented (Grice, 1989). Clarity is especially significant to understanding such disclosures, however, since it highlights the tensions inherent to many study participants’ stories about their acts of communication.

Clarity is, on one hand, important, if one wishes to effectively convey information to others. The purpose of interpersonal communication is, however, not always so narrowly circumscribed. As Daly and Wiemann (1994) point out, the pre-determined, individual goals most relevant to understanding strategic outcome-oriented communication stand in sharp contrast to the emergent, relational goals that facilitate process-oriented communicative interaction. To assume that one or the other is predominant or that both cannot be operative at the same time is to miss the complexity of communicative interaction. As such, clarity in conveying information to others may be less desirable when a certain level of obscurity and ambiguity actually work to sustain important social and familial relationships. Further, ambiguity may be positively valued because it retains a degree of uncertainty and therefore the possibility of hope; it is, in Waddell’s (1982) terms, a strategy of neutralization.
A second and closely related issue concerns the meaning and significance of language and verbal communication to speakers and their audiences. Study participants varied widely in their willingness to articulate some aspects of their experience. Those who believed that they were experiencing signs of onset and/or those who learned that they had inherited the mutation were, in this respect, especially attuned to the way in which language makes things real by allocating them a definite place in the world. As such, the phrase “it’s not a secret but...” points to the possible response “I can’t talk about it.” This type of response warrants much greater attention since those who are least able to talk about their experiences are often left out of, or unable to participate, in interview-based research.¹

A third set of more pragmatic issues lends other insights into why genetic information is “not a secret but.” The idea of secrecy was overwhelmingly constructed in negative terms. Indeed, most study participants actively disavowed secrecy as a strategy for managing genetic information because: a) secrecy implied that information was being withheld from someone with a legitimate interest in the information, and b) secrecy seemed to suggest that there is something shameful that must be hidden. Given the need to justify the with-holding of information from someone with a legitimate interest, it is not surprising that few study participants talked about engaging in deliberate acts of concealment. Moreover, given the long history of social stigma and misunderstanding surrounding HD, many study participants felt that it was especially important to move away from viewing hereditary risk and illness as a source of shame. Ironically, however, study participants sometimes shifted the burden of shame onto other actual or potential illness sufferers in order to make this point: the comparison between going public with one’s genetic versus HIV/AIDS status was, in particular, invoked on occasions when study participants sought to distinguish between the moral dimensions of hereditary as opposed to lifestyle risks. Being at risk for HD was nothing to be ashamed of because it was something that happened to you, rather than a reflection of something you did.

Finally, study participants also drew an important distinction between secrecy and

¹ Survey-based research may suffer from similar deficits in understanding the diversity of PT candidates' experiences. Response rates typically drop in the post-results phase and it is likely that such attrition is patterned by many of the same factors.
privacy. This distinction underscored the links between several salient themes. In contrast with secrecy, privacy is a means of establishing boundaries and preserving some aspects of self from public view. In keeping with the way that complex objects are often represented (in English) by their containers (Reddy, 1993), the home was, in this sense, one means of signifying privacy and representing the containment of genetic information within the family. Home was an “information preserve” (Zerubavel, 1982) and the work of managing genetic information was conceptualized as a kind of “housekeeping.” The gendered dimensions of this work were reflected in several ways: women took on the chief responsibility for communicating about HD within the family and, moreover, the work of managing such communication was itself constructed in and through a series of metaphors oriented around the theme of housekeeping.

**Lessons From the Field**

Sometimes our most spectacular failures are what offer up the greatest lessons in life. This is no less true when it comes to the doing of research. Though I hope that the findings of this research, as I have summarized them above, will facilitate new understanding and prompt ongoing conversation, I am at this juncture, humbled by what I have not accomplished in the doing and writing up of this research.

I was blessed with an extremely large, rich and diverse set of stories about the experience of hereditary risk and its modification through predictive testing. The process of gathering these stories took me inside many worlds. I spoke with PT candidates, their mothers, fathers, spouse/partners, daughters, sons, sisters, brothers, aunts, and friends. There was also one grandmother. Moreover, the prospective design of this research meant that I had the opportunity to speak with study participants several times and to learn, firsthand, about how their stories unfolded in and through time.

Each of these aspects of the research was, however, also a source of complexity. The size of the data set meant that I often felt overwhelmed. There were approximately 6,000 pages of transcripts and fieldnotes. Further, it was difficult to envisage how I could effectively encompass the multiple perspectives of PT candidates and their respective family members. Finally, the prospective design meant that each study participant’s story shifted as it unfolded.
All of this required that I had make some difficult choices about what to foreground and how.

Such choices are inevitable but it is important to acknowledge their implications for the findings of this research. This dissertation focuses primarily on PT candidates’ stories. Though family members are certainly given a presence, the dissertation as story would be very different were it to have adopted the perspectives of spouse/partners or siblings as central. Further, although the sixteen PT candidates’ stories offered much more than I could adequately analyze here, it was sometimes difficult to achieve a sense of data saturation. This was especially the case in the post–results interviews since there were, amidst the sixteen PT candidates, a total of seven different objective modifications of risk. Moreover, four received their results through the rural protocol while twelve attended the local medical genetics clinic. Finally, there were no men who learned that they had inherited the mutation so it was difficult to gain a sense of how gender factored into the process of interpreting this test outcome.

Recruitment of study participants was conducted in the clinical setting and this contributed greatly to the high level of participation in the study. Nonetheless, I have in hindsight, some reservations about the degree to which this form of recruitment shapes the stories that it is possible for study participants to tell. To begin with, I was initially perceived by many study participants as a member of the clinical team. With time and repeated interviewing, this initial confusion dissipated. The degree to which study participants felt constrained to tell a particular story is, however, something that is very difficult to assess. Adopting an alternative form of recruitment was not, however, feasible given the prospective study design and the need to collaborate closely with the clinical team in coordinating the timing of pre and post–results interviews. Were I to undertake a retrospective study, I would consider utilizing snowball sampling through the community and Huntington Society contacts that I have now established. As Downing (In progress) found when recruiting participants for her study on reproductive decision–making within families at risk for HD, word of mouth is often a very effective means of locating persons who do not attend the medical genetics clinic or belong to the Huntington Society.

Research on sensitive topics is an intervention in study participants’ lives. This is especially the case when the topic of the research requires a high degree of introspection and
willingness to share very personal aspects of one’s life. Within the context of this research, the embeddedness of the research activity in the subject matter of this research posed many practical as well as analytic challenges. Study participants often went out of their way to accommodate my schedule and ensure that multiple members of the family were available to do an interview. Moreover, I was sometimes caught unaware in trying to anticipate the kinds of problems that could arise when study participants began to incorporate my research–related communications with their family members into their strategies for managing genetic information. To give but one example, I found on one occasion that I had been given permission to contact a family member about their participation in the study and when I called to arrange an interview, it became clear that the family member did not yet know that the PT candidate was going ahead with the test.

This brings me to one final issue of overarching significance in shaping this research — that is, the limits that one must observe in writing up sensitive material. I have, for instance, found it necessary to think through each verbatim quote from the vantage point of various family members. Informed consent notwithstanding, this remains one of the most challenging aspects of conducting in–depth qualitative research within a collectivity: there are never any guarantees that one has not overlooked something or, for that matter, that a study participant will not draw an incorrect inference from something that one says or does.²

Where possible, I have therefore tried to adopt several strategies to remain accountable to both study participants and the reader. Throughout this dissertation I have made a point of observing myself in the act of observing others. Such moments are, as Timmermans (1994) argues, a means of comprehending the other “by the detour of the self”. Further, I have also presented myself as “a live sociologist at work” in order to demonstrate how I arrived at the conclusions I did. Following the reflexive turn in sociology and much of contemporary ethnography (Hertz, 1996), such writing strategies are hardly unorthodox. Nonetheless it bears repeating that the integration of reflexive commentary about the nature of the relationship

² This, I found especially difficult with respect to the timing of the pre–results interviews since some family members involved in the study did not know for certain when the PT candidate was to learn their test result: those who understood the research design could, however, make a fairly accurate guess and thus, in at least one instance, it appeared as if I had disclosed the results date when I should not have.
between personal experience and the research helps to establish trust: without such reflexive commentary, the reader is no position to assess the degree to which the researcher distinguishes between her own experiences and the experiences of those she studies (Reinharz, 1992).

Though I am now moving beyond a discussion of the limitations of this research, it is important to note that there were many significant methodological and ethical issues that emerged from the doing of this research. Working with the study participants who are featured in the narrative accounts was, in particular, an experience through which I learned much about the difference between orality and textuality, narrative and chronological time. Moreover, I also found that participants' self-schema and narrative interpretation of the process of predictive testing changed subtly or even markedly over time (before and after test results). The person they were at the time of reading was different than the person they were at the time(s) of narration. Thus great sensitivity was sometimes required in negotiating with study participants who wished to revise their finished narrative accounts. The text of these accounts, however, renders invisible the local struggles for coherence which were an important part of how participants read and validated their stories.

Reflections and Recommendations

Here I wish to conclude by offering some reflections on the findings of the research and their specific implications for clinicians, researchers, at risk individuals and their families.

Though there is much evidence to support the principles and practices of the original test protocol (i.e., allowing sufficient time to reconsider and withdraw from predictive testing), the findings of this research suggest that it is important that service providers reevaluate the degree to which PT candidates are expected to engage in introspective analysis of their experience of predictive testing. In practice, this might mean allowing PT candidates and their families a greater degree of autonomy in deciding the content of various counselling sessions. Further, there are a number of very specific issues arising from the language of disclosure. Predictive testing is about the provision of information: the quantity, quality and clarity of information presented to PT candidates and their support persons during counselling and disclosure of the test results is therefore crucial. As such, it may be important for service providers to re-evaluate
the use of phrases which assume a high degree of tacit knowledge (e.g., 'it never skips a generation') and avoid the use of terms such as 'positive' and 'negative'. Further, where CAG repeat numbers are provided, consideration ought to be given to providing the PT candidate with the CAG repeat number for both alleles.

Within the research context, it is important to avoid placing too much emphasis on the test outcome. What comes after initial awareness of the family history of HD and before clinical disclosure of results is an extremely important, though often neglected, part of the story of predictive testing. This part of the story has been neglected in the published literature for at least two related reasons. First, existing research and clinical evaluation have tended to focus on the psychosocial and other effects of predictive testing for the at risk individual (and, in a small number of studies, their spouse/partner). This focus on the effects of predictive testing mirrors what has been a long-standing problem in many studies of technological change; that is, an overemphasis on the social implications of technological change and an underemphasis on the social shaping of technology (Cox, 1991; Elliott, 1988).

This critique may be applied on a number of levels but with respect to research on the psychosocial effects of predictive testing, it implies that it is at least as important to understand the ways that PT candidates arrive at the decision to have the test as it is to look at the post–test effects. Moreover, this critique also suggests that it is important to investigate the nature of the relationship between awareness of the test and the experience of being at risk. In order to proceed with predictive testing, hereditary risk must come to the fore as an issue that requires some resolution for the PT candidate and/or various members of their family. This heightened awareness of risk and its implications for self and others need not precede the knowledge that predictive testing is available; indeed, knowledge of the test may in and of itself precipitate the very tensions which then seem to demand resolution through the decision to request predictive testing.

3 Stated more broadly, social values shape the design and use of technology just as technology, in turn, shapes social values.
4 Some clinicians and service providers have commented on the dangers associated with viewing predictive testing as a technological imperative—in particular, Adam et al (1993) stress that it is sometimes important to remind those who are considering the prenatal test for HD that the existence of the technology should not in and of itself mandate its use. Nonetheless, there are as of yet no studies which attempt to assess the difficult question of how knowing that there is a test factors into the perception that being at risk is something that ought to be resolved. The metaphor
Second, in order to measure the psychosocial effects of predictive testing much of the existing research is structured around the imposition of a pre versus post-results dichotomy. In this dichotomy, the middle is the fleeting moment that fractures the experience of predictive testing into two disparate pieces; everything that comes before this decisive moment is “pre-results” and everything that comes after it is “post-results.” The middle is, in this sense, missing; it is positioned between the before and the after but has, itself, no significant presence. Studies of predictive testing that adopt this dichotomous view therefore reveal little about how being at risk is understood and experienced in relation to the knowledge that there is an accurate presymptomatic test for HD; indeed, the complex sequence of events and experiences which culminate in the decision to request predictive testing are, more often than not, collapsed into a static list of “reasons for testing”.

Many of the items that are included in such lists are future-oriented and instrumental in character; the emphasis is on what predictive testing will accomplish and the experience is constructed as the means to a particular end or ends. Little attention is given to how PT candidates and their families became aware of the test much less to understanding the diverse pathways though which the PT candidate arrives at the decision to request predictive testing. This is an odd omission given that there is, within the clinical context, a great deal of emphasis placed on ascertaining whether or not the PT candidate is making an autonomous and fully informed decision.

The findings of this research also have significant implications for PT candidates, at risk individuals and their families, and the Huntington Society. First and foremost, I believe that it is especially important to ensure that there are formal and informal opportunities to hear about the experiences of other persons who share a common experience of HD. There was, amongst my

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5 Studies of predictive testing that adopt this dichotomous perspective take as their focal point the change that occurs in response to a particular stimulus—that is, knowledge of the outcome of predictive testing. As such, changes in the self-identity, psychosocial well-being and/or life-plans of the PT candidate are understood as effects or consequences that follow from such knowledge.

6 The “reasons for testing questionnaire” is a standard part of many PT protocols. Typically, PT candidates are asked to use a numerical scale to rank the importance of an itemized list of reasons for requesting predictive testing for HD. For instance, the “reasons for testing questionnaire” that is routinely administered as a part of the clinical protocol at the local clinic asks PT candidates to rank, on a scale of 1 to 4, each of eighteen reasons for testing, thirteen of which are prefaced by the phrase “it will” or “it could”. (e.g., “it will help me (and my partner) to make decisions about having children” or “it will help me (and my partner) to make long-term plans such as retirement.”)
study participants, an overwhelming interest in what others had to say. Such a need can in part be met through the chapter meetings and publications of the Huntington Society; nonetheless, there is also a need to encourage informal discussion.

Second, and related to this, there is an urgent need to dispel the prevalent idea that most at risk individuals undergo predictive testing. As genetic testing becomes routinized, it becomes increasingly difficult for at risk individuals to decide not to request the test without also feeling the burden of having to justify this decision. Family members and friends can assist by encouraging at risk individuals to explore ambivalent feelings and avoid premature disclosure to others about the (potential) decision to request the test.

Third, communication is not always about imparting information; it is also about sustaining relationships. Hence communication may not always be evaluated in terms of its clarity and/or completeness but may rather be evaluated in terms of how it expresses care, demonstrates trust and/or sustains hope. Ambiguity may in this respect have a moderating influence which is in tension with the desire for full and open disclosure. The most important implication of this for PT candidates and their families is that there is not one optimal or appropriate level of awareness about hereditary risk and/or the results of predictive testing. Awareness may wax and wane as may the desire to talk about it.

If anything, this supports the stance taken by some service providers (see Bloch) who argue that those who are not ready to acknowledge that they are showing signs of onset should be allowed to proceed through predictive testing as if they were presymptomatic. This understanding does, however, need to be extended to those who wish to put HD on the ‘back burner’ and not talk about it or live their lives in anticipation of it. The lack of willingness to delve deeper into analyzing the meaning and significance of knowing that one will develop HD is not necessarily ‘denial’ — it may be, as one study participant said, “simply a matter of getting on with life”.

Finally, this research emphasizes the meaning as well as the strategic significance of talking about HD and hereditary risk. Story telling (or account formation) is an important means of rendering the experience of predictive testing coherent. It is especially helpful because it emphasizes the processual nature of the experience and invites reflection on change and the
subsequent restorying of experience. Further, there is, as narrative therapy suggests, a therapeutic moment which accompanies storytelling (Wiersma, 1992); this moment is sometimes cathartic but as Cruikshank (1998) demonstrates, oral storytelling also has the potential to "destabilize" and "transform official orthodoxies". This potential is, I suggest, of overarching importance if lay knowledge is to be properly valued and lay persons are to have a meaningful part in public dialogue about the new genetics (Kerr, Cunningham-Burley, & Amos, 1998).

Researchers must, however, also take some responsibility here. It is customary for researchers to acknowledge responsibility for disseminating the results of research in the form of scholarly publications and papers. Further, clinicians and service-providers are aware of the need to modify existing clinical protocols according to the outcome of specific evaluative studies. Less often, however, do researchers recognize and respond to their research subjects' expectations of what constitutes meaningful output. Does the researcher return her findings to the community from which they derived? Are laypersons asked for their feedback and/or involvement in the design of community-based and/or participatory action research? What practical implications follow from research? As one study participant reminded me in a long overdue post-results interview,

...if it's going to help my son or somebody in my family or even some stranger then no problem, my life's an open book. If it's so that somebody can write a piece of paper, no, my life's not an open book. And you know I'm a pretty caring person but if there's no need to express myself I won't. ...Why should I throw my life out on the table for you just so that you can get an A on your paper?

The five narrative accounts prepared for the Huntington Society of Canada represent one means of returning the research to the HD community. In addition, I also served on a Huntington Society Task Force which was given a mandate to discuss and make recommendations to the Society on the needs of persons who were "gene-positive." The research which informs this dissertation was of immediate value in contributing to the Task Force report.

**Recommendations for Future Research**

There are many existing lacunae for further research on, and comparative studies of, the
social meanings and experiences of hereditary risk. A short list of topics related to HD and the
test would include: 1) qualitative studies on the experiences of those
who do not want to have predictive testing; 2) comparative studies focusing on gender and
'genetics activity', with particular attention to the issues of a) why more women than men
request predictive testing and, b) why women assume primary responsibility for communicating
about genetic information; 3) focused interviews on specific patterns of disclosure and
nondisclosure of the test results to family and friends (such data might also inform the
development of an appropriate survey which could be used to obtain a broader sense of the
desirability of openness in communication about genetic information). Within a clinical setting,
it would also be especially valuable to undertake: 4) qualitative studies on clinical
communication (and miscommunication) about genetics and hereditary risk, especially within
the context of the disclosure of predictive test results; and 5) analysis of the differences in
clinical and lay expectations, how information is presented, what types of questions are asked
and by whom.

Within the larger community and sociocultural context, it would be valuable to
investigate: 6) the impact of going public and sharing stories about experiences of hereditary
risk and predictive testing; and 7) lay and professional groups views on, and representations of,
hereditary illnesses in campaigns designed to raise public awareness. Such research could no
doubt encompass a community-based and/or participatory action design that would both involve
families at risk for and affected by hereditary disorders such as HD in the design of research
and, more generally, enhance lay involvement in public dialogue about the new genetics.

Finally, comparative studies on: 8) participants' experiences of predictive testing for HD
and other adult onset hereditary conditions (e.g., Polycystic Kidney Disease, breast/ovarian or
colon cancer), would no doubt shed new light on the salient moral and ethical as well as social
and familial issues related to hereditary and other forms of embodied risk as would 9)
comparative studies on experiences of genetic and other forms of presymptomatic testing (e.g.,
HIV testing, cholesterol testing).

In summary, we have much to learn from the novel social situation of those who elect to
know, through genetic testing, something about their probable fate. Predictive testing for
Huntington Disease has, in particular, raised many significant clinical, social and ethical issues. Recently, however, it has been suggested that HD is an inappropriate model for thinking about how at risk individuals and their families will respond to the information derived from predictive testing for other more complex polygenic, and multi-factorial adult onset disorders. HD is, in short, too simple; the issues are too black and white. This is a claim which this and, I hope, other research will do much to alleviate. Even on the molecular level, HD is proving to be more complex than originally thought. Furthermore, as tests become available for a variety of other late onset inheritable disorders (e.g., Alzheimer’s Disease, breast, ovarian and colon cancer), it will become increasingly important to know how those most closely affected by inheritable disorders understand, adapt to and manage genetic information.

After Words

In closing, I refer the reader to the following two poems: they are, among other things, about understanding the importance of communication and the limits of what it is possible to say in words. The first poem appeared not long after I joined an HD related email group. It was written by Jean Miller, for her daughter Kelly, and it is included here with their permission. The second poem is a response to Jean and Kelly’s poem that I wrote when a friend who has HD began to experience great difficulty with verbal communication.
IF I CAN'T TALK, AM I STILL HERE

I was a magical weaver of dreams, a solid and steady friend
For hours and hours at a time,
we talked as if time would never end.

You always valued my opinions,
often we'd open up and pour out our hearts,
time was an endless millennium,
always difficult when we had to part.

Then, slowly, this disease robbed me of my ability to communicate well.
Does that mean that deep within me, there are no dreams left to tell?
Why is it God, I ask in my heart, as someone so very ill
that most people so deeply fear this?
Have they forgotten the magic I instilled?

Although Huntington's has taken my health,
and maybe it's stolen some dreams
it is, oh, so very much worse.
Dear Lord, it's taking my self esteem.
Because of this, I must ask "If I can't talk, am I really here?"

Please, come take my hand, there really isn't anything to fear.
And now, yes now more than ever, does my very soul cry out
for you to come sit by my side and speak of times,
when we were the best of friends.

by Jean E. Miller for Kelly E. Miller
(May 30, 1996)
“IF I CAN’T TALK, AM I STILL HERE”: A REPLY

Even if you can’t talk, you speak
and even if you are not here
i want you to know i am listening

i will never say i know because i don’t and can’t
i am caught up in thinking
about the meaning of contemplating not being able to speak
not being able (as you say) “to communicate well”

what does it mean?
to come to know this outside in and bit by bit
a syllable at a time
as words slip and slur
and only those who know the you you were
(and still are but are unable to say)
must try ever harder
to
hear
you
never pretend to understand your meaning
only repeat the fragments
in exchange for your look
your look that says “try again”
“try some more”
“try until I think you understand something”
“some part of what I am trying to say”

(you watch as i think i hear you and nod “okay you may go on”)

there is much more in this awkward loss than i know how to say
how can you say what it means to be unable to say?

and coupled with this
coupled with this is one way of knowing that says
you cannot survive
you cannot survive this disease
but you are still here
and the want for words wrestles sleepless in these pages
choking innocent sheets

breathless
    indelible
this loss

i can’t say what it is
but to be with it and in it
cloaked heavy with trying
knowing it like the skin on the back of your hand
in grace and time
and precious time

by Sue Cox
(written October 1, 1996, revised May 11, 1999)
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APPENDIX I

RESEARCH OBJECTIVES
**OBJECTIVES OF FUNDED QUALITATIVE RESEARCH ON FAMILIAL IMPACTS OF PREDICTIVE TESTING**

(N.B. Related objectives are shown across each row of the table.)

<table>
<thead>
<tr>
<th>BC Medical Services Foundation</th>
<th>Hampton Fund</th>
<th>Huntington Society of Canada</th>
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<tbody>
<tr>
<td><strong>Objective 1</strong> — explore factors &amp; decision-making processes shaping individual decisions about whether or not to undergo predictive testing</td>
<td></td>
<td><strong>Objective 1</strong> — work with people at risk for HD to prepare accounts about experience of living at risk for HD &amp; where applicable, meaning of predictive test</td>
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<tr>
<td><strong>Objective 2</strong> — elicit &amp; describe individuals' understandings of impact of the predictive testing program &amp; test results on their lives &amp; relations with others</td>
<td><strong>Objective 1</strong> — assess longer term impact of predictive testing on test candidates, family members &amp; others</td>
<td><strong>Objective 2</strong> — work with people who learn that they have the gene for HD to prepare accounts about meaning of living with this knowledge</td>
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<td><strong>Objective 3</strong> — investigate individuals' decisions to share information about participation in predictive testing with family members, friends &amp; others</td>
<td><strong>Objective 2</strong> — understand dynamics of communicating test results to family members &amp; friends</td>
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<td><strong>Objective 4</strong> — assess &amp; make recommendations on impact &amp; availability of counselling &amp; support for individuals tested &amp; family members</td>
<td><strong>Objective 5</strong> — provide test candidates’ &amp; family members’ assessments of counselling &amp; test protocol as input for policy makers</td>
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<td><strong>Objective 5</strong> — evaluate existing clinical protocols &amp; recommend modifications to ensure candidates are adequately informed to give consent to predictive testing</td>
<td><strong>Objective 3</strong> — investigate individuals’ understandings of relationship between kinship relations &amp; genetics</td>
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<td><strong>Objective 4</strong> — examine individuals’ understandings of test results &amp; relationship between genetic status &amp; symptoms of HD</td>
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8. Will my predictive testing results be kept confidential?
Yes, absolutely. You may decide to tell family members about your test results but researchers will not disclose any information concerning your predictive testing results.

9. What happens if I (or any of my family members) start the research study and then decide to withdraw?
Participation in all components of the research is entirely voluntary and you or your family members may withdraw at any point. This decision will not adversely affect your or their participation in the predictive testing program or any other medical care you or they receive.

10. Once I have made up my mind that I am interested in participating in the research, what happens next?
If you are interested in participating in the family study, a member of the research team will be available at your first counselling session to provide you with further information and to answer your questions. If you and your family would like to participate in the questionnaire component of the study, you will be provided with the appropriate instructions and questionnaires to take home with you. If you and your family would like to participate in the interview component of the study, a member of our research team will contact you and your designated family members by telephone to schedule an interview.

Additional Questions??
Additional information is contained in the letters which you received with this pamphlet. If you have concerns about any aspect of the research that you would like to discuss before your first counselling session, please contact: Caroline Benjamin (Genetic Counsellor/Research Coordinator) 822-7195.
1. What is the purpose of the research?
This research study aims to help us better understand the social and psychological impact of predictive testing for Huntington Disease on at risk individuals and their families.

2. Who is conducting the research?
The research is a Canadian collaborative project involving researchers from medical genetics and the social sciences. The research team is based at the University of British Columbia and is headed by Dr. Michael Hayden.

3. If I do not wish to involve my family will it still be possible for me to participate in the research?
Yes. Although the family study provides the opportunity for us to gather some very important information, we do realize that you may prefer not to have any family members involved at this time. If you are in this situation you could still participate in a portion of the study by completing 3 research questionnaires (1 hour total) at 3 of your clinic appointments.

4. If I do wish to participate in the family study, what will be required and how much time will this take?
The family study consists of a questionnaire and an interview component. You and your family members may choose to take part in either or both components. The questionnaire component will require that you complete 5 questionnaires (approx. 1 hour) after your first counselling session and then again at 2 weeks, 6 months and 12 months after your test results. The interview component of the family study will involve two separate interviews—one held before your predictive testing results and the other 3 to 4 months after results. Each interview will require about 1 hour and may (at your convenience) be held in your own home or at UBC.

5. What will be expected of my family members if they agree to participate?
Your family members will be asked to complete the same questionnaires and/or interview schedule as you. The questionnaires can be completed at home, so there is no need for family members to travel to UBC. You may return completed questionnaires when you visit the clinic, or family members may return them by mail in the envelopes provided. Interviews will (at your family member's convenience) be held in their own home or at UBC.

6. Which family members should I invite to participate in this study?
When possible, we ask that you first consider your spouse or partner (if applicable) and a parent. If it is not possible to obtain the participation of either or both of these family members, then another close family member or companion may also be eligible. At this point we cannot, however, include anyone under 18 years of age or anyone who has been diagnosed with Huntington Disease.

7. How do I ask family members to participate?
Since the purpose of this research is to obtain a clearer understanding of how predictive testing affects you and your family members, we will need to know very early on whether or not you and your family members are willing to participate in the study. We ask that you talk to the appropriate members of your family and share the information contained in this pamphlet. If they are willing to consider participating in the research we also ask that you pass on the enclosed letter and complete the “interested participants” section (in this pamphlet) before attending your first counselling session.

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Family Study: Interested Participants

This section provides a space for you to list the names of one, two or more family members who are willing to participate (or consider participating) in the questionnaire and/or interview components of the family study. We ask that you complete this section before your first counselling session and bring it along with you.

---

Family Member #1

Name: ____________________________
Relationship to you: ____________________________
Address: ____________________________
Telephone: ____________________________
Best times to call (to schedule interview) ____________________________

Interested in participating in:
☐ Questionnaire & interview
☐ Questionnaire component only
☐ Interview component only

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Family Member #2

Name: ____________________________
Relationship to you: ____________________________
Address: ____________________________
Telephone: ____________________________
Best times to call (to schedule interview) ____________________________

Interested in participating in:
☐ Questionnaire & interview
☐ Questionnaire component only
☐ Interview component only

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See reverse side
APPENDIX III

RECRUITMENT ADVERTISEMENT
WHAT DO YOU THINK ABOUT PREDICTIVE TESTING?

We are conducting a study to better understand how at risk families think about predictive testing for Huntington Disease. Part of this study involves looking at the experiences of those families in which an at risk member is uncertain about or has decided not to have predictive testing. A second part of the study looks at the experiences of those families in which an at risk member is currently undergoing predictive testing.

If you (or someone close to you) are considering predictive testing or have decided against it, we would like to hear about your experience. Participation in the research involves one informal interview conducted at your convenience in your own home or at UBC. Confidentiality is assured. For further information please contact: Professor Bill McKellin or Ms Sue Cox by telephone at: 822-2756 (Department of Anthropology and Sociology, University of British Columbia, 6303 N.W. Marine Drive, Vancouver, B.C. V6T 2B2).
Thank you for your inquiry regarding the predictive test for Huntington Disease (HD). As you may be aware, the recent discovery of the gene for HD has allowed us to offer a new, highly accurate predictive test for individuals at risk for HD.

Counselling and predictive testing is offered by genetic counselling centres across Canada. A standard clinical protocol which includes pre and post test counselling and the completion of clinical questionnaires also forms part of the Canadian Collaborative Research Study on Predictive Testing for Huntington Disease. The current research study has two parts:

The first part is designed to help us to understand why individuals request testing and to study the impact of predictive testing on at risk individuals. This section of the research includes the completion of additional questionnaires (25 minutes) at each of your clinic visits to help understand the psychological and social effects of testing.

The second part of the study is designed to explore the effects of testing on the family, and requires identification of at least two family members who would be willing to participate. Previous research has indicated that the effects of testing are not limited to the individual but extend to their families as well.

We would like to invite you to participate in either or both parts of this research. Details of the study are enclosed. This research is very important to enable health care professionals to give appropriate support to families at risk for Huntington Disease and will provide information of relevance for other genetic conditions.

The opportunity to participate in this research project is offered to all individuals requesting testing for predictive testing for Huntington Disease. Participation in either section of the research program is voluntary and individuals are free to withdraw from the study at any time. If you decide not to participate, this decision will not influence the availability of testing to you or your family. If you have any questions concerning participation in the study, please contact Caroline Benjamin at the number below.
Dear Family Member:

CANADIAN COLLABORATIVE STUDY ON PREDICTIVE TESTING FOR HUNTINGTON DISEASE

The Impact of Predictive Testing for Huntington Disease on You and Your Family

We are writing in response to your family member's request for the predictive test for Huntington Disease. The new form of predictive testing for Huntington Disease tells individuals whether or not they have inherited the gene for Huntington Disease. The results of the test may have an important impact on the individual receiving the test. Participants in an earlier Huntington Disease testing program also suggested that the results of the test can have significant effects on their families.

Because we are introducing a new form of testing, and hope to provide appropriate counselling, we believe that it is important to understand the impact of the test on the individuals tested and their families. We have asked individuals receiving predictive testing for Huntington Disease to invite two or more family members to participate in a study of the psychological and social effects of predictive testing for Huntington Disease. We are inviting you to participate in this study.

The family study has two parts. In one you will complete a set of questionnaires about your psychological well being, family relations, and social support. (Most of the questionnaires will be the same as those completed by your relative during counselling sessions.) The questionnaires will be sent to you four times: one set before your family member receives the test, and three sets during the year following the test. If, after reading the questionnaires you decide not to participate, simply return the uncompleted questionnaires.

The other portion of the study consists of two interviews, one before your family member is tested and one four months to a year after the test.

The information you provide in the questionnaires will be kept confidential. No information will be provided to anyone outside the research/clinical care team without your specific written consent.
APPENDIX V

CONSENT FORMS
PART I - QUESTIONNAIRE BASED STUDY

As a participant in the clinical testing program, you will complete a set of several psychological and social questionnaires before you receive the test results. You will also complete the set two weeks, six months, and again one year after you learn the test results.

In this study we request that you complete three additional questionnaires concerning family relations and the social support you receive from members of your family. You will receive these additional questionnaires from your genetic counsellor or by mail at the same intervals as the clinical questionnaires. You may fill them in at your convenience and return them by mail. The additional questionnaires should take approximately one hour to complete each time (total - three hours for the study).

Information provided in the questionnaires will be coded to maintain confidentiality. No information will be provided to anyone outside the research/clinical care team without your specific written consent. You may obtain the results of your answers to the questionnaires though your genetic counsellor.

You are free to ask questions about the study or withdraw from the study at any time. Your decision will not adversely affect your or your family member's participation in the Huntington Testing Program or any other clinical care you or they receive.

If you wish to participate complete the bottom of this page and return it to your genetic counsellor. Keep a second copy of the complete letter for your own reference.

CONSENT

I have read pages 1 and 2 of this letter which explains the questionnaire based part of the study of the Impact of Predictive Testing for Huntington Disease on You and Your Family, Dr. Michael R. Hayden (Principal Investigator). If I decide to withdraw I will notify the clinic of my decision. I have had all questions answered to my satisfaction and understand the conditions of my participation described in the letter. I give my consent to participate and have retained a copy of this three page letter for my own reference.

NAME ____________________________________________

SIGNATURE ________________________________________ DATE __________
PART II - INTERVIEW STUDY

The goal of the interview study is to provide a different kind of information than gathered by questionnaires and to allow you the opportunity to express yourself more completely about the impact of predictive testing for Huntington Disease on you and your family.

We invite you to participate in two informal interviews concerning predictive testing. You will be interviewed at your convenience in your home or at the University of British Columbia. Each interview will last approximately one hour (total two hours for the study). In the first interview, which will be conducted before you receive your test results, you will be asked to describe your previous experience with Huntington Disease, your attitude towards testing, how you decided to take the test, and the kinds of impact that the results of the test may have on you and your family. In the second interview (four months to one year after you learn the test results) we will ask you to describe the impact of the results of the test on you and your plans, and your perception of the test's impact on your relations with family members and friends.

The interviews will be tape recorded. You may ask to have the names of any individual deleted from the tape during or after the recording is made. We also ask your permission to tape-record your sessions with the genetic counsellor as part of this study. Information provided during the interviews and counselling sessions will be kept confidential. Tapes and transcriptions will be identified by number, not by name. No information will be provided to anyone outside the research/clinical care team without your specific written consent.

You may ask questions or withdraw from the interview study at any time. If you decide not to participate or to withdraw, your decision will not adversely affect your or your family member's participation in the Huntington Testing Program or any clinical care you or they receive. If you have any questions relating to the interview study please feel free to contact us.

If you are willing to participate, please sign the consent for the interview study at the bottom of this page and return a copy of the complete letter indicating that your questions about the study have been answered to your satisfaction and that you understand the conditions of the study.

CONSENT

I hereby consent to participate in the interview based study of the Impact of Predictive Testing for Huntington Disease on You and Your Family, Dr. Michael R. Hayden (Principal Investigator), as described on pages 1, and 3 of this letter.

I have retained a copy of the three page letter describing the conditions of the study for my own reference.

NAME ____________________________________________

SIGNATURE ________________________________________ DATE ________

FAMCLICO.BC Revised February 15, 1994
PART I - QUESTIONNAIRE BASED STUDY

We have enclosed a form requesting basic personal information about you and psychological and family relations questionnaires which will also be completed by the person who has received the Huntington test. If you decide to participate in this study, we would like you to complete this set of questionnaires and return them to us before your family member receives the results of his or her test. You will also receive three more sets of questionnaires: one small set two weeks after your family member receives his or her test results and two complete sets like this one six months and twelve months after the testing results are received. The complete set of forms and questionnaires should take about two hours to complete each time. The shorter set should take about one hour (total of seven hours for the study).

The complete questionnaire set consists of seven questionnaires concerning your personal background, psychological well being, recent life events, quality of life, and family and social support. The shorter set contains only the psychological questionnaires which your family member will also be completing at that time.

The information you provide on each questionnaire will help us understand the short and long term effects of testing and will also help to assess and improve the counselling available to individuals who receive predictive testing and their families. Information provided in the forms and questionnaires will be coded with family and individual identification labels and kept confidential. No information will be provided to anyone outside the research/clinical care team without your specific written consent.

Returning the completed questionnaires indicates your consent to participate in this study. You may withdraw from the study at any time. If you decide not to participate, or to withdraw, return the uncompleted material. Your decision will not adversely affect your family member’s participation in the Huntington Testing Program or any other clinical care he or she receives.

Please feel free to contact us if you have any questions regarding any part of the study.

CONSENT

I have read pages 1 and 2 of this letter which explains the questionnaire based part of the study of the Impact of Predictive Testing for Huntington Disease on You and Your Family, Dr. Michael R. Hayden (Principal Investigator). If I decide to withdraw I will notify the clinic of my decision. I have had all questions answered to my satisfaction and understand the conditions of my participation described in the letter. I give my consent to participate and have retained a copy of this three page letter for my own reference.

NAME

SIGNATURE _____________________________ DATE __________
The goal of the interview study is to provide a different kind of information than gathered by questionnaires and to allow you the opportunity to express yourself more completely about the impact of predictive testing for Huntington Disease on you and your family.

We invite you to participate in two informal interviews concerning predictive testing. You will be interviewed at your convenience in your home or at the University of British Columbia. Each interview will last approximately one hour (total two hours for the study). In the first interview, which will be conducted before your family member receives his or her test results, you will be asked to describe your previous experience with Huntington Disease, your attitude towards testing, and the kinds of impact that the results of the test may have on you and your family. In the second interview (four months to one year after your family member learns the test results) we will ask you to describe the impact of the results of the test on you and your plans, and your perception of the test's impact on your relations with the person and other family members and friends.

The interviews will be tape-recorded. You may ask to have the names of any individual deleted from the tape during or after the recording is made. Information provided during the interviews and counselling sessions will be kept confidential. Tapes and transcriptions will be identified by number, not by name. No information will be provided to anyone outside the research/clinical care team without your specific written consent.

You may ask questions or withdraw from the interview study at any time. If you decide not to participate or to withdraw, your decision will not adversely affect your or your family member's participation in the Huntington Testing Program or any clinical care you or they receive. If you have any questions relating to the interview study please feel free to contact us.

If you are willing to participate, please sign the consent for the interview study at the bottom of this page and return a copy of the complete letter indicating that your questions about the study have been answered to your satisfaction and that you understand the conditions of the study.

CONSENT

I hereby consent to participate in the interview based study of the Impact of Predictive Testing for Huntington Disease on You and Your Family, Dr. Michael R. Hayden (Principal Investigator) as described on pages 1 and 3 of this letter.

I have retained a copy of the three page letter describing the conditions of the study for my own reference.

NAME

__________________________________________

SIGNATURE

__________________________________________ DATE

Name of family member tested and your relation to him/her

__________________________________________

FAMEMBCO.B.C.

Revised February 15, 1994
Thank you for your continued interest in our study. The goal of this study is to understand the experience of predictive testing for Huntington Disease from the perspective of at risk individuals and their families. Your continued participation in this study will help us to better understand the long as well as short term impact of predictive testing.

Here we invite you to participate in one follow-up interview concerning Huntington Disease and predictive testing. As in previous interviews you may, at your convenience, be interviewed in either your own home or at the Department of Anthropology and Sociology, University of British Columbia. This interview will last approximately one hour. During this interview you will be asked to describe changes in your personal and family life which may be related to Huntington Disease and to discuss any further thoughts about Huntington Disease and/or predictive testing which now seem relevant.

The interviews will be tape recorded. You may ask to have the names of any individual deleted from the tape during or after the recording is made. Information provided during the interviews and counselling sessions will be kept confidential. Tapes and transcripts will be identified by number, not by name. No information which would identify you will be provided to your other family members, or to anyone outside the research team without your specific written consent.

You may ask questions or withdraw from the study at any time. If you decide not to participate or to withdraw, your decision will not adversely affect you or your family member's participation in any clinical care you or they receive. There is no monetary compensation. The interviews are an opportunity for you to express yourself more completely about the impact of predictive testing for Huntington Disease on you, your family, and friends. We hope that the results of this study will assist genetic counsellors to provide appropriate information and guidance.
APPENDIX VI

INTERVIEW SCHEDULES
PRE-RESULTS INTERVIEW SCHEDULE

1. Life history/background
   Please tell me about yourself
   family background & memories of growing up
   current marital/relationship status, children
   regular contact with family
   education, occupation, employment
   social activities, sports, religious affiliations, hobbies
   contact with close friends
   a typical day (or week)
   general health, previous or current health problems

2. Family history and knowledge of HD
   When did you first learn about the family history of HD? (probe for circumstances, people
   involved, their responses)
   What did you understand about your (or your family member's) at risk status?
   Did you have any prior knowledge of HD? Was this connected to your family history?
   Have you known anyone diagnosed/affected with HD? (probe for experiences of HD
   related to affected family members or friends, caregiving experiences)
   Were there particular sources of information that you turned to? (probe for people, books,
   HD Society newsletter, resources or meetings,)
   How often was HD in your thoughts? a topic of conversation? (probe for circumstances—
   what, when, where, how often, with who)
   Could you describe how knowing about HD affected your relationships with family
   members, friends, or other significant people?

3. Managing information about HD and risk status
   Who knows about your (or your family member's) at risk status? who doesn't? why?
   (probe for family, friends, associates, family physician)
   What can you tell me about what it was like to tell other family members, friends, or
   associates about your (or your family member's) at risk status? (probe for circumstances—
   who, when, where, why?)
   What were their responses/your responses?
   Were there particular people or circumstances which seemed comfortable? difficult?
   Do you ever think or imagine that you are experiencing symptoms of HD? (probe for what
   symptoms, when)
   Are there particular strategies you have to help you (and/or your family member) cope
   with being at risk for HD?

4. Predictive testing for HD
   When did you first learn about the predictive test? (probe for circumstances—where, who
   was involved, what was learned, your responses)
   Were you (or other members of your family) eligible to have the old type of linkage test?
   (if yes, probe for whether this was considered, decision, reasons)
   Did learning about the test have any effect on your understanding of HD or what it means
   to be at risk for HD?
   Are there other family members or people you know that have been through predictive
   testing? What was this experience like? (probe for possibilities & pressures)
   What impact did this have on you?
How were other family members involved in your (or your family member’s) decision to have predictive testing? (probe for circumstances, when, where, who, major issues) What was this experience like? (probe for possibilities & pressures)
What were the main things that you felt were important in deciding about predictive testing?
What sort of impact do you expect that predictive testing will have on you? on other members of your family?
What do you think are the advantages (or disadvantages) of knowing your (or your family member’s) at risk status? (probe for which of greatest concern & why)
Do you have any unresolved feelings about predictive testing?
Are you anticipating the results? Do you have a hunch about what they will be? (probe for stability or change in outlook, mood, relationships, activities, plans)
How do you expect to feel the day before your (or your family member’s) results? the day of your (or your family member’s) results?

5. Predictive testing and counselling
How would you describe your visit(s) to the clinic? Did anyone go with you? Did you learn anything new or come away thinking about anything differently?
Are there any particular questions that should be asked when people are thinking about predictive testing? Any advice that you would offer to other families?
Is there any particular information, support or counselling that would be helpful at this stage? or that you anticipate needing?
Are there any things you would have preferred to do differently?
Are there any other important areas or experiences that we should discuss?

6. Participation in research
Is there anything you want to say about this interview and what we hope to learn through this research? What do you think is most important about your experience?
How would you prefer that I learn your predictive testing results? (i.e., through the clinic or would you prefer to tell me yourself?)
Is there anything in particular that we have talked about that you would not want other family members to know about?
POST-RESULTS INTERVIEW SCHEDULE

1. Background

   How have you been since I met with you last time?
   Is there anything that you would like to clarify from the previous interview? (e.g., changes in information, attitudes, what was said about other family members etc.
   Also review any missing or confusing information (e.g., family names, dates etc.)

2. How are you feeling today? this week?

   What is a typical day/week like for you right now?
   Have there been any changes since we last talked (at first interview)?
   Probe for: general mood, daily patterns, recent events related or unrelated to predictive testing

3. Results day

   What did you do before going to the clinic? How did you feel?
   Who was present in your (or your family member’s) results session? What happened?
   How were your (or your family member’s) results reported? What was actually said?
   What did you say? What did others who were present say?
   What do you remember about your reactions to learning your (or your family member’s) test results?
   What did you do when you left the clinic?
   How was the next day for you? (Probe for: general mood, health, sleeping and eating patterns, changes in activities, work schedule, relations with others, what was expected and unexpected)

4. Days immediately following results day

   What did you do? How did you spend your time?
   Who did you tell your (or your family member’s) results to? When? What did you say?
   What did they say? How did they react? Who did you not tell your results to? Why?
   (Probe for: general mood, health, sleeping and eating patterns, changes in activities, work schedule, relations with family members, friends, others)

5. Last few months

   How have you been spending your time?
   Who did you tell your (or your family member’s) results to? When? What did you say?
   What did they say? How did they react? Who did you not tell your results to? Why?
   Also probe for: general mood, health, sleeping and eating patterns, changes in activities, work schedule, relations with family members, friends, others

6. Implications of the results

   Do you think that knowing your (or your family member’s) predictive testing results will change how you live your life? the decisions you will make? immediate or future plans?
   (Probe for: marital or reproductive decisions, employment or educational decisions, financial planning etc.)
   How does knowing your (or your family member’s) predictive testing results affect the way that you feel about yourself? Your perception of your (or your family member’s) day to day health? Do you think that this will change over time? (Probe for: self-identity, experiences of early signs and symptoms, bodily awareness)
What are the advantages of knowing your (or your family member's) test results? What are the disadvantages? (Probe for: which of greatest concern & why? and, if applicable, what are the advantages or disadvantages of this type of testing over linkage testing?)
Do you have any regrets about going through predictive testing? Are there any things you would have preferred to do differently? Is there any additional information, support or counselling that would have been helpful? (Probe for: expected or unexpected outcome and impact, level of information and support provided by clinic, family or friends, anything that would have been useful to know about (especially from others who have been through predictive testing).

7. Advice and/or reflections on experience
   for those who are currently considering predictive testing?
   those who are not considering predictive testing?
   families at risk for HD?
   clinical and other support persons?

8. Participation in research
   Why did you decide to participate in this research study?
   What do you hope that we will learn?
   Do you have any advice about what is most important?
   Is there anything you want to say about I have conducted the interviews? or any other part of the research process?
   Is there anything else that we should discuss?
   Review: open-endedness of study; possibility of future contact and possible publications
1. Background

How have you been since I met with you last time? Is there anything that you would like to clarify from the previous interview? (e.g., changes in information, attitudes, what was said about other family members etc. Also review any missing or confusing information.)

2. How are you feeling today? this week?

What is a typical day/week like for you right now?
Have there been any changes since we last talked (at second interview)? (Probe for: general mood, daily patterns, recent events related or unrelated to predictive testing, other illness of self or family members. Offer reminders about last interview if needed).

3. Predictive testing, onset and/or diagnosis of HD

How often do you see/ hear from your family member(s)? (Probe for: frequency of contact with family member(s) (more or less than around time of PT), observations of family member, own or family member’s decisions related to outcome of PT, indications of increased or decreased closeness because of HD and PT

How are you now feeling about your (or your family member’s) predictive testing results? Have your feelings changed at all since the last interview? If yes, how? If not, why not? Have the results of predictive testing changed your perception of your day to day health? The significance of other family members’ health?

Do you think knowing your (or your family member’s) predictive testing results has changed how you live your life? the decisions you make? immediate or future plans? (Probe for: marital, reproductive, employment or educational decisions, financial planning etc.)

Was the one year anniversary of learning your (or your family member’s) results significant to you in any way? your birthday? How so? (Probe for: relationship between current age of family member and average age of onset in family. Link with outcome of PT.)

Have you been to the clinic or had any sessions with a genetics counsellor since we last talked? If so, for what reason? Any new information or understandings about HD and/or predictive testing?

Have you or has anyone in your family been diagnosed with HD since we last talked? begun to show signs or symptoms of HD ? (If PT candidate has HD gene probe for: attentiveness to possible symptoms and self-consciousness about monitoring of behaviour and if PT candidate does not have HD gene probe for: attentiveness to possible symptoms in other family members’ behaviour and/or self-consciousness about not monitoring family member’s behaviour.)

Has anyone in your family requested predictive testing for HD since we last talked? is anyone else thinking about requesting PT?

4. Talking about HD and predictive testing

Do you and your family member(s) talk about HD? the predictive test results? How often? In what context and how does the subject arise? Are you comfortable with this? Do they seem to be comfortable with it?

Do you think that you have been told everything that your family member(s) has? knows? (Probe for: inferences about what others know e.g., “the impression I get is...’ or “wives seem more likely to share information...”)

Have you discussed your predictive testing results with anyone other than your family member(s)? If so, with whom? Why? When? What did you say? If not, why not?
Are there things that you would like to be able to talk about that for whatever reason you feel you can’t? Are there people that you would like to be able to talk to about this with but for whatever reason you feel you can’t?

Thinking more generally, are there situations that you can imagine in which people have some kind of obligation or responsibility to tell other family members about HD and/or predictive test results? (Probe for feelings about telling or not telling others because of worry about how they will react, decisions they will make e.g., in 3 generational families consider grandparents worried about children not having children)

Are these responsibilities specific to HD or do they extend to families at risk for other inheritable disorders? How does being at risk for HD differ from being at risk for other inheritable disorders? Are there patterns of behaviour which seem to you to be typical of a family at risk for HD? (Probe for: perceptions of importance of dominant versus recessive disorder, ways in which HD is more or less severe, significance of late versus early onset, lack of therapeutic interventions etc.)

5. Advantages and disadvantages of predictive testing

What are the advantages of knowing your (or your family member’s) test results? What are the disadvantages? (Probe for: which of greatest concern & why? and, if applicable, what are the advantages or disadvantages of this type of testing over linkage testing?)

How do these relate to your understandings of your (or your family member’s) reasons for wanting the PT?

Do you now have any regrets about going through predictive testing? Are there any things you would have preferred to do differently?

Is there any additional information, support or counselling that would have been helpful? (Probe for: expected or unexpected outcome and impact, level of information and support provided by clinic, family or friends, anything that would have been useful to know about especially from others who have been through predictive testing).

Do you have any advice or comments for those who are currently considering predictive testing? those who are not considering predictive testing? families at risk for HD? clinical and other support persons?

6. Clinical experiences, genetic testing for HD and other conditions

What are your feelings about the nature of your experience with the genetics clinic? the value of counselling sessions? the kinds of questions you were asked?

What things seemed to be most important to the genetic counsellors and other clinical personnel? Were these the same things that you thought were important? Were there particular words or phrases or ways of understanding things which seemed at odds with your ways? (Probe for: constructedness of experience, differences between what is perceived to be expected and what is really significant, i.e., pressure to present a particular face to the clinic)

Now that you (or your family member) have had predictive testing, will it make it easier or less problematic for others in your family to have the predictive test?

Do you think that in general predictive genetic testing for HD is becoming more routine? less exceptional? How about in comparison with other genetic disorders? or in comparison with other kinds of medical tests?

7. Participation in research

What do you hope that we will learn? Do you have any new advice about what is most important?

Has being involved in the research had any impact on you? on your family member(s)?

Is there anything you want to say about I have conducted the interviews? or any other part of the research process? Is there anything else that we should discuss?

Review: open-endedness of study; possibility of future contact and possible publications
TRANSCRIPTION SYMBOLS AND NOTATION

Capital letters: Start of utterance
/
      Minor, non-final phrase boundary marker
.
      Down intonation at end of utterance
?
      Up intonation at end of utterance
ALL CAPS: Loudness and/or strong emphasis
()
      Description of audible sounds
(...) Speech pause, one period for each second
(()) Doubtful hearings or unintelligible word
[] Overlapping speech
{} Clarification of thing or person refereed to in speech and/or substitution of generic information in order to preserve anonymity

Adapted from:
CARLA

I spoke with Carla several weeks before she was scheduled to receive her predictive test results. At the time, she was living with her partner Tom in an apartment on the upper floor of an East Vancouver home. She had been working for a number of years as a dental assistant but was currently unemployed. She and her partner were considering moving away from the city and Carla felt that the timing was right for her to have the predictive test. Carla began her story by talking about her mother’s unnamed illness and the events which led to her finding out, at age 30, that she and her three siblings were at 50% risk for Huntington Disease.

I was born in 1952. At the age of 6 months I went to live with my aunt and uncle because my mother was not capable of looking after all of us. They raised me and then, when I turned 12, I went to live with my father, my sisters and brother.

When I was 15 my mother passed away. She had Huntington's but nobody told us that she had it. My father hadn't explained to us that she had Huntington's. I don't think he really knew she did at that time. She was in a mental hospital for about 12 years and then she passed away. They didn't say at that time that it was Huntington's. They didn't really know what it was.

When my Mum married my Dad some of my aunts and uncles tried to tell him that maybe it wasn't a good marriage, that maybe my Mum wasn't a good candidate for marriage because she just didn't seem to be all there. But my Dad didn't really listen to them. I don't know whether or not he had any inclination as to what was happening because he'd tend to overlook things and pretend that things were okay.

My father had told us that she had another disease and that they were treating her for TB and giving her some really intense antibiotics that they were trying on people in the 50's. She did have a scar on her lungs that I remember one of the doctors talking about. My Dad told us that this was a result of nerve damage from taking this medication.

I didn't find out about Huntington's until I was 30 years old. My eldest sister might have known because she was pretty inquisitive and she would ask a lot of questions. My aunts and uncles had assumed that we knew that Huntington's was in the family because none of us married or had children. They figured that this was a choice we had made and so nobody said anything to us. I couldn't ever quite figure out why things were happening the way they were because it just didn't make sense. My Dad wouldn't ever really answer my questions and I found it really hard to talk to him because every time I would ask him a question I'd always get a different answer. And when it's your father speaking to you, well, for me anyway, whatever he said, I believed him.

When I found out about Huntington’s being in the family I was visiting my mother's sister. We were sitting around the table and she was telling us about how her father had passed away. He was found hanging in the barn at his farm and my mother was the one that found him. They don't know whether or not he had Huntington's but he probably did.

When my aunt told us about the family history I felt kind of relieved. I felt that it was good because we could carry on and have some answers. I thought I could make some choices about my own life from that point on. For me it just cleared up all the questions.

Talking It Over

After my aunt told us about HD I seriously thought it over for a few years and decided to have a tubal ligation. I really didn't want to see children suffering anymore than they had to. There wasn't a lot of testing being done at that time so it was clear to me that I didn't want to pass this horrible thing on. I knew that clearly. So it was either I make a choice about my body and what I was going to do with my future in terms of having children or pass it on to my offspring. So I was 34 when I had a tubal ligation.

I've always had a woman friend that I'm really close to. Eva and I have been very close friends for quite a few years. She and I would sit and talk about Huntington's. But before her, I had
another really close woman friend and we'd just sit and talk about what was going on and she'd say, "Well it's okay. I think it's great that you're having this tubal ligation. I mean who wants to see other people suffering? I mean that's great." She was always really supportive. So I've always had that network and I know it's there and I know I need it. I was always really uncomfortable with talking to men about those sort of things.

I think it comes from having to kind of deal with my Dad. If I could deal with that I think my life would probably be a lot easier. But it's really hard for me. He doesn't really change. He's more mature and he's got more stories. "I told you you should do this." "I told you you should do that." "I told you. I gave you this information way back when, don't you remember?"

One of my sisters has Huntington's and she lives about 10 miles, from my brother. She was diagnosed 10 years ago. It's really interesting because if you sit him down and you look him straight in the eye and say, 'Dad, Nicki has Huntington's.' He won't look at you. It's like, "No, she's got something else." Or, "Well, if she wouldn't have got in this damn car accident, it wouldn't have brought it on and she'd probably be still working today." It's that kind of denial. So I never even bother to bring it up with him. I talk to him about things that he enjoys like his gardening. I'm sure he's done as best he can for us.

I got a lot of information from my sister's medical doctors because they would always send me updates and information about the disease. I told friends about my family history and that my sister has Huntington's and that it's something that I'm being tested for at this point in time. And everybody says, "Yes! Go ahead and do it."

I found out that there was a test three years ago. I came home with some pamphlets and I had read some information about the updates that are being done and Eva had an article that she found in the Vancouver Sun when she was at school one day. So the two of us were like chatting this over and saying "Isn't it great what's going on now. How things have really changed," and "Isn't it wonderful to be able to find out that you can have a concrete answer."

I knew I wanted the test. And I also knew that I could see myself moving away from here, so I'd like to know. I'd like to clear that off my conscience before I leave. So the timing is good for me. When I found out that you could have a definitive answer I knew that was the choice I wanted to make. My family physician was the person that recommended me to the clinic here so she knows I'm having it done. She thinks it's a great idea.

I feel that there's a really strong network of women working at the clinic and I really feel very comfortable about phoning and asking questions. Even phoning when I'm distressed and saying "Help". It's that sort of network of women that I find really very comfortable for me. But maybe that's because I'm a woman. Men might have a different response.

When I Get the Results

The results will definitely effect my future. I feel they're going to effect my life in terms of my plans for what I'll be doing in the next 5 years of my life, in terms of my career, in terms of my relationship with my boyfriend. If I do have Huntington's, my lifestyle will definitely change in terms of just physically getting around.

When I get the results, I'll be excited and I'll phone all my friends and have a party. I am quite confident that they're going to be negative.

I don't find myself thinking about having the disease because I really don't feel I have it, but I do think about how I'm dealing with it. I feel like I'm a little bit unclear at times and I haven't really talked a lot about it... I feel like those things, those thoughts will just be verified and cleared up and I can get on with my life. I feel good about how I'm dealing with it. Very good. Getting outdoors helps a lot. Exercising. Being with Tom. I've done a lot of great things in my life so it's like now I really want to just stop and smell the roses.

Several weeks later Carla went to the predictive testing clinic and received her result. Her partner Tom went with her although he had not been present at her other pre-test counselling sessions. Carla was, in her own words, "devastated" to learn that she had the gene for Huntington's.
Huntington Disease. She told me this news herself when I saw her at the clinic for a two-week follow-up appointment with her counsellor. Several months later she described the emotional impact of her predictive test results and her concern that she might already be showing signs and symptoms of Huntington Disease.

Results Day

I was devastated. It was like hitting rock bottom. I don't know. I guess it didn't surprise me in one respect. I did feel like there were certain things happening in my life that hadn't ever really happened before to me yet I felt that I wasn't quite sure where it was coming from. So getting the results was a real blow. I was pretty upset emotionally and I do have days like that now. Whenever I can't cope with something or I have to tell people and be really clear about something or make a decision I don't have it, I just kind of fall apart. I get worried because it must be happening.

Tom went to the clinic with me. He drove and we stopped at the liquor store on the way home. I think it was Valentine's day, or the day after Valentine's day, so we bought a big box of chocolates and came home and ate it and drank a bottle of brandy. I was crying. I was pretty upset for a couple of days.

Feeling Stronger

I was supposed to phone people after I received my results and I didn't. I just found I could only phone about one person a day after that, and talk to them and I would get pretty upset. But then I got stronger. As the healing process took place I got stronger.

The first person that I called was Eva. She is kind of like my sister, my long lost sister that I never really had. She and I, we talked a lot about our upbringing and shared a lot of good times together. She's a very up person, very positive. I've had a lot of laughs with her. I think I probably had the most fun in my whole life with Eva. She and I spent a lot of time together. We lived together for about 5 years. She's having a hard time dealing with this, with me, because she notices some differences. She was pretty upset too. But she was really positive and she said, "Well don't worry about it. Be optimistic and a lot of things can change. There's a lot of new changes and new developments so you never know." And she's been really supportive of me and booked me in to see a Tibetan healer. She said, "Don't worry, Mamma Eva will take care of you. I'll keep you happy." She said, "If you're going to get it well you'll be the most traveled, most happy person in the world." So she's very up, very cheerful. But it was hard. It was really hard.

The next person I phoned was my girlfriend Pat. She and I have been friends for about 15 years. She came over that day and we went for a walk.

Then I phoned my brother and I spoke with his wife. She was very upset. Very very upset. She's having a real problem living with Huntington's in the family. It's just been really bleak for her. She made a lot of choices in her life because of this monster as she calls it.

Then I spoke with my brother. My brother is wearing a lot of hats. Plus he's living with two kids and a wife who are all very worried about this whole monster of Huntington's being in the family. So he's trying and he wants to have the test done but there's no way that he would because he just feels it would change his life too much. He's seeing all of us go through changes. So for him it's like he doesn't even want to find out because he spent years labouring over his kids and giving them lots of confidence and a lot of support so that they wouldn't be treated the way that we were treated as kids because we were raised in a very male chauvinist environment.

I spoke with my step-Mum too and she said, "Aw, hell, Carla it's not any big deal, I mean we all have problems. We all have weird genes." And I wrote my Dad this long letter and I didn't hear from him for about 6 months. I ended up phoning him and talking to him myself. He never did really address it. That was before I went down there to visit. It's typical. It's nothing unusual. So I wasn't really overly surprised but I was hurt. I mean at a time when you need to have support from somebody. Just for somebody to say they care, just "How are you doing? Sorry to hear this or that with the results."

Narrative Accounts/Carla
Tom couldn't believe that I was phoning all these people and he said, "you shouldn't be doing that," because I was all upset and crying. And I said, "Oh, it's okay. I feel better. At least I've talked to somebody." It felt good. It felt better. I felt like I was getting stronger after each one because for me it's really important to talk to people. It was therapeutic for me and I needed that. It's good for me to be having that kind of contact with people. That's what my needs are at this time. A lot of friends and family.

But it was interesting. I did phone a couple of my cousins and they weren't surprised to hear what the results were. They felt that they had seen changes in me the last couple of times I've been down there. Some people are a little a bit more critical. The people who said "I've noticed a big change in you," were people that I expected that from because I know they're like that. My cousin was like that. But then I thought that's Joanne, she shoots from the hip. I know that that's the way she is and that's her way of being protective. That's her way of being kind. I knew to just take her with a grain of salt and not let it bother me. My brother's kind of like that too. He's very analytical. He doesn't mince words. He's being honest. So you just kind of know what to anticipate with people. That's what people are like.

Feeling Some Changes

I'm definitely feeling some changes. I always used to just get on with life and have a good time and put a smile on my face. That's always been my direction in life and my family see that's changed. But the physical part of it they don't really see very much whereas I'm feeling some changes happening in my body, myself. I find my balance isn't as good as it used to be and I have this really kind of twitchy feeling inside, like there's something in me that just wants to keep moving around. It's a really bizarre feeling inside.

It could be that I'm not as strong as I used to be but I've been walking an hour to three hours a day, sometimes. So I don't know whether I'm going through changes that way or whether it's because this gene is showing or the Huntington's is showing it's effect on me.

I've also noticed a lot of changes on my own management kind of level. I find I'm having problems with doing things like paperwork or balancing my checkbook, and I get really freaked out. I'm quite emotional about it because it's not ever been something I've enjoyed doing but it never used to bother me. I used to just do it and get on with something else but now it bothers me.

Those are things my family noticed. And that I would get upset by things. I notice a big change in my memory because I used to be able to remember people's names or dog's names or horse's names and now it's like I'm stuck. It's like I can't come up with it and then I get frustrated because I know that I used to know that person's name or that person's kids. It's very frustrating. That part is something I noticed in a big way.

My partner Tom is really great. He's so supportive. He's just, "Oh, don't let it worry you. It's no big deal. I'll take care of it for you." When I do things that require dexterity I don't feel very safe. I'm always cutting myself. So Tom does a lot of cooking.

I think it took a lot of courage for me to go ahead and get the test results. It was devastating but at the same time it wasn't. I definitely think more now about how I am coping on an everyday level. I am more dependent on other people to make choices for me and I want somebody to just take control. That's nothing unusual though. I've always been that way but it's definitely very comfortable letting somebody else run my own ship. That's no change for me.

It's been quite painful at times. The truth has been painful for me. And it's had a lot of disadvantages for a lot of my friends. I think that's what kind of hurts me the worst—having to tell my friends that I have it. It's the pain it causes them. Eva was great. She went around and she was telling all my friends so I wouldn't have to go through that. That part's been hard for me.

But I'm happy that I've had the test done. I don't have any regrets. It's been painful but it gives reason to what's been occurring in my life over the last few years. I've had a lot of problems keeping jobs and when I phoned up some of my ex-employers and they said "that doesn't surprise me. It seems to me you were having some problems when you were here and we were
worried about your health.” It was feedback that I’d never sort of had before. Up until then it had always been positive feedback or “everybody here sure misses you,” or “such and such a patient was in yesterday and was asking about you.” That still happens with people that I’ve worked with 5 or 10 years ago but in the last few years that’s changed. I haven’t had a very good time. It’s been a hard year for me working. And that’s why I went to have the test done.

The Emotional Impact

People don’t usually get to know what’s going on inside the heads of people that have it. The doctors are very clinical and they’re into that kind of clinical arena. They never really touch the emotional impact or what’s happening inside a person’s head or heart.

I feel like there’s a very clinical way that it was treated in terms of receiving the results. I just felt that I was in this very clinical environment and I just wanted to go and run into the skirts of the nearest woman and cry my eyes out or something. I felt like it was everywhere I looked. It was those white walls. But I think the doctor has done a great job. He’s got a support staff that’s really very empathetic...but there could be more support. Maybe that’s what my needs are though. Maybe that’s not what everyone else’s needs are.

I was in dentistry for 25 years and in one of the offices my job involved being the support person for all the staff and patients, so a lot of people used to phone me up and talk. Whenever there was some concern or somebody had not had a good experience I’d always phone them up at the end of the day or the next morning just to check and make sure that they were okay, just so that they knew that there was someone there that they could talk to. A lot of people change when they are in the home environment. They totally change and they end up telling you things that you might never have known before. So I thought it was great when the genetics counsellor phoned me back two days after my results session, but I could have used that everyday. “How are you doing?” “It’s okay, we’re thinking about you.” That kind of feedback to me is probably the most valid that you can have. It’s kind of like somebody who you feel very special about. You just want to tell them that you care. I found that really helped people plus it helps you feel better about yourself because you feel like you’re helping someone.

Boy, I’ve done a lot of talking to people. It helps it feel more familiar.

The Next Part of It

Right now I’m not working at all so I’m thinking about going and getting some income assistance or something like that. And that’s hard for me. I’ve always worked. It’s very humiliating for me. But I need something to do so I’ve been thinking about going to the Huntington’s Society and doing some volunteer work. I have also made an appointment to go back out to the clinic and have the neurological tests done.

I’ve seen changes and that’s why I decided to have the genetic test done originally and I want to have the follow-up because I just want to get on with the next part of it. So I’m ready to have the next set of results done.

A lot is going to depend on what happens when I go in to the clinic in August but we are probably going to be taking a long trip to Mexico or something like that. We’re thinking about buying an RV and staying in some of the state campgrounds for the winter or for a couple of months or so. I don’t know. I’m not sure. I do know that I really like being, or need to be around my friends so they can stop by and give me a hug every once and awhile. That’s important to me. So we might be moving away from here but maybe not too far.

Later that summer Carla went back to the clinic for neurological tests and was diagnosed with Huntington Disease. Nearly two years later I spoke with her about the impact of her diagnosis and her extraordinary ability to begin “healing on another level”. She looked relaxed and happy as she talked about the many positive changes that she had made in her life. Admitting that her partner had not been supportive of these changes, Carla had left him and embarked on a journey of her own.

Narrative Accounts/Carla
Healing on Another Level

For me the whole experience has been kind of like a gift. I’ve taken everyday and made that day special for myself.

One of the things that I did was I left the man I was with living with. I wasn’t really getting a lot of support from that relationship. I just felt like I was needing to be in an environment where I’m supported and healthy and happy. I was getting quite pulled down. He was very negative and very upsetting to me, telling me things like ‘you deserve to have Huntington’s and you deserve to be in the position that you’re in because you’re not really doing anything to help yourself.’ So that’s when I started to go and have some counselling. The counsellor kept saying ‘Carla, you don’t have a problem. It’s not your problem. You don’t have to keep all the fish that are swimming in the sea, you know, you can throw the odd one back’. So I said, ‘okay fine’.

I also saw a Tibetan healer and he said ‘well, you know, it’s one thing to have whatever it is you have. That’s okay but you shouldn’t be afraid. You should be accepting it. You should be enjoying your life not running away from people. If you’re not getting the support, then change it.’

So I gave up the man and I bought a dog. She and I walk almost everyday. We’ll walk 8 or 9 miles a day sometimes. We go hiking in the woods or around the seawall a few times. I talk to her and tell her everything that’s going on and she’s pretty perceptive about what’s going on in my mind. So I’ve moved on and moved through that relationship and I feel a lot better for it. I have enjoyed the changes that I’ve arranged for myself. I just feel like I’ve done a wonderful job of making changes that are good for me. I have placed myself at the top of the ladder.

I had always wanted to go to Africa so I ended up buying a ticket as a treat for myself. Many people in my support group were very supportive of me and said ‘yeah, you can do it, you’ll have so much fun.’ So I ended up buying a 5 month journey to Africa and I was down on an overlander trip that went from Cairo to Cape Town. And that was fun. I really, really enjoyed it. The thing I found so nice though was that I was around a lot of people. There were 20 or 30 people on the trip with me and that was nice because there were always people to go out with. I think that’s what I find the hardest with the Huntington’s: it’s not having a network of people and not having people to work with. When you’re working you have that whole circle of people. I’ve been in touch with a lot of my friends but everyone is so busy with their lives.

But I’m getting better and healing on another level and I’m also having cranial acupuncture. That is another reason why I came back home. I really missed having the treatments. I started having them 2 years ago. It’s a Tibetan style of acupuncture. They put needles in your cranium and then they put moxa on your spine and they do body work for a couple of hours. I have that done once a week. And that, the cranial acupuncture, has really been a very healing mode for me.

When I returned from Africa, I also met Nancy. I met her at one of the support groups. She was diagnosed last summer. And both of us have been sharing a lot of concerns that we have in dealing with Huntington’s and the world. A lot of times there’s a lot of shock out there. I find that once people find out that you have some sort of medical concern, they treat you totally differently. They back off. They really do. That happened with one of the girls that was on my trip with me. It was funny because she and I were traveling together for 2 or 3 months and then all of a sudden we got to Nairobi, the capital of Kenya and we were staying there for a week and she was leaving at that point and that’s when I told her. She said ‘oh, it can’t be, you’re just like me, you’re just like me’. Then in the middle of the night we were having some fun and partying and this girl was quite drunk I think, and she was saying ‘yeah well you’re the one that has a health problem, you’re the one who has a health problem.’ It was just like everything changed and I’m going, ‘no, no you do, you’re the one who is weird and drunk and out of it. But I found once people know, it’s different.

It’s kind of like you know a secret and you can’t keep it from them. Like with some of my friends. They’d say ‘Is that the same old Carla I know? And I go ‘yeah, you can hug me. I won’t pass out in front of you.’

Narrative Accounts/Carla
The big part is that people just need to be treated with some normal respect. I remember going a couple of times with my sister in California when she went to see her neurologist. And he was very cold. He said, 'Oh, Nicki, say your name.' ‘Nnniicckkii’ (she’d say her name slowly). ‘What’s that?’ he’d say. He just made her feel awful, so edgy. The next time they wanted her to go in to see him, she didn’t go, she wouldn’t go. I don’t blame her. When I went with her to see the neurologist he kept looking at me and said ‘oh, so you’ve been diagnosed. Oh, well I don’t see any symptoms there.’ And then he said ‘I guess you’d better go home and get ready for the almighty hell to hit.’ I just looked at him and said, ‘no, I’m going to get ready for my life, I’m going to start living’.

When I had the test done, I knew I’d be taking a chance but I really didn’t feel like I had the symptoms yet and I still feel the same. But now I’m not working and then I was so that’s been the big impact for me really, the lack of work. But I’ve found other ways to fulfill my life and I have a new direction. You’ve got to do what you do in your life if you’re going to do it. There’s no point in doing it half heartedly. You make the best out of what you’ve got. That’s what the experience has been for me. But it’s a really hard one to decide on. It has helped me to make changes in my life that are good for me. I had been hanging on a lot to my relationship with Tom (the man I was living with) because I felt like nobody else would really want me. But when I was traveling in Africa everyone kept saying to me ‘there’s nothing wrong with you, I don’t see anything wrong’. And some of these women I traveled with for 2 or 3 months, day and night, they all said ‘just forget about it, just carry on, if you want to share your life with somebody, you should. Go home and find yourself somebody who is a kind loving individual.’

On another level, I have met some of the most wonderful people I’ve ever met in my life in the support group. And my friends always make sure I’m invited out for Christmas dinner or they’ll write me a poem or something. So I get a lot of feedback in that way and it’s good. I feel very well loved. I was thinking about moving away but I enjoy being around people, being around the city.

Like my sister, she’s doing really well right now. She’s had Huntington’s now for 12 years. She’s basically living at home but probably should be in the hospital. But what my sister— in—law has done is gone out and found an angel of a caregiver to look after her. She interviewed probably 40 people and she ended up with Susan. She takes Nicki everywhere, she has so much fun. And when I was down there I was over there everyday. She and I, we’d go out to the bar and play darts, have all boys come over to see her. She just laughs and that’s the way I think people should be. I mean we should enjoy the rest of our lives. Not feel sick. We don’t want to feel sick, you know we want to enjoy life.

That’s where you need caregivers who are very loving and caring, who don’t take you and stick you in a room. They’ll take you and show you to the rest of the world because you’re like everyone else. And I think that’s been the ongoing thing with people that have any kind of disease. We all need each other. We’re all part of same family. That’s what I say. I don’t really feel lonely because we’re all part of the same family, we’re all part of the same people. It’s the way that we put ourselves in our society that makes us feel separate. And I think that’s what we need to change. That’s where people need to be changing the way that they’re caring about other people.
I spoke with Colin about one week before he was scheduled to receive his test results. He and his wife Emily were living with their three daughters in a quiet farming community about an hour and a half’s drive from Vancouver. Colin was 41 years old at the time. He began his story by talking about his early memories of his father’s illness.

I grew up in Hamilton, Ontario. I was a city guy but now I’m more of a country person. It’s taken me almost 40 years to get that way. I have one brother, five years older than me. We had a normal childhood and family life until my Dad took sick.

We had a serious car accident when I was thirteen years old. That was in 1966 and for a few years we thought my Dad’s illness was something to do with the car accident. In hindsight we can see that it was the onset of Huntington’s. It wasn’t until 1970 that we actually found out it was Huntington’s. My Dad would have been in his mid 50’s. His employer was quite concerned about his safety at work because they had noticed a difference in him and certainly we also did. Looking back, it must have been very scary for him because he never talked about the changes that he must have noticed in himself. It must have been hard for him when the employers said, “Look, either go to the doctor and figure out what’s going on or you don’t have a job.” The company actually put him in the hospital and it took them a long time to figure out what was going on because Huntington’s didn’t have the profile in the medical community that it does now.

Prior to that we had no knowledge of it. He must have inherited it from his mother. She died in 1934 and her death certificate recorded the cause of death as homesickness. I don’t know what her symptoms were because I wasn’t able to talk to my Dad about it. I have two uncles and one of them told me a lot about my Dad. But I wish I could talk to my Dad because you don’t ask questions or think about those things when you’re a kid. Both of my uncles married and it is kind of interesting to us that they didn’t acknowledge the disease, especially the one who has children. They know nothing about the disease but maybe they don’t have to worry.

Our Philosophy

I moved out to BC in 1972 because I had a job opportunity and wanted to go into hotel management. I really enjoyed that business but it was pretty hard to have a life. When I was dating I could only go out on Tuesday mornings. It limits your choices. And I came to the realization that if I got more successful in that business I’d always be working weekends and evenings and holidays. It wasn’t the lifestyle that I wanted. I wanted to get married and have a more traditional lifestyle.

I met my wife Emily in 1974. My new career was selling light fixtures and her Mum came in to buy light fixtures where I was working. As a good salesman I always got involved in people’s lives a little bit. Then when I found out that she had two daughters I got real interested, and it sort of went from there.

I told Emily that I was at risk for Huntington’s before we were engaged. I told her what I knew about the disease and what my chances were. And before we got married we had genetic counselling so that she could get a professional point of view. I don’t think there’s anything that the doctor could have said at that time that would have scared her off. We talked about it. I tried to get her eyes as wide open as possible but when you’re in love it doesn’t matter. It is not until later in life that you find out what your love is made of.

Seventeen years whizzed by for us. We have 3 children aged 13, 11 and 8. Three girls, three weddings. We’re pretty busy but I’ve always tried to find a balance. I’m sure I could be more successful business-wise if I didn’t worry about the family but I’m not going to have the business rob me of all my time with the family. Huntington’s aside, you never know how many years you have together. Our philosophy has always been that we try to have fun along the way because you just never know. We’ve always tried to have time together and, whether it’s weekends away or vacations or whatever, we have something to show for each year.
There Were Fewer Options When My Dad was Sick

The year that my Dad was diagnosed was about the same time as the Huntington's Society was being formed. I was there at the initial meeting in Toronto for the Huntington's Society of Canada. Marjorie Guthrie was there too. In those days there were quite a few myths about Huntington's but one thing I always knew was that there was a 50/50 chance of inheriting the disease.

My brother Brad was away at college in those years so he escaped a lot of the family turmoil. Then after college he got married and he and his wife denied the whole thing. They never wanted the Huntington Society newsletter, never wanted to go to any meetings, they just didn't think about it. My sister-in-law is the type that never had any hardship in her life so once she saw that my brother was getting Huntington's she wanted out. This happened about 8 or 9 years ago. When they first split up I flew my brother out here to stay with us for a couple of weeks, just to talk because it was quite devastating.

Brad has Huntington's and he's not doing very well right now. When I phone him it's hard to get him to converse. He's isolating himself a little more. I haven't seen him since I went down there to see his doctor and arrange for Meals on Wheels and home care. He knows what's in store for him after seeing my Dad. He set up different friends as sort of a committee to watch over him as he starts to go down hill. He appointed one friend the power of attorney years ago because that was a big struggle with my Dad. We could never get him to sign that over to my Mum and it was really tough because cheques would come in and he wouldn't sign them. So Brad was able to think about those things and make sure that we're not going to have the same problems. But I spend a lot of my time during the year feeling guilty about being three thousand miles away with my Mum in bad health and my brother in failing health and not being able to improve their quality of life.

I know what has to be done. It's time for him to go into a group home situation. That's a hard thing to do. There were fewer options when my Dad was sick. No one understood. There was no Huntington's Society going out to the care homes and giving them specific training on how to understand and deal with someone with the disease. There was none of that and my Dad spent some time in the Ontario hospital. It was sad because we'd go to see him and he had no business being there. Mentally he knew what was going on but he wasn't able to communicate.

A 180 Degree Turn in My Thinking

I first heard about the predictive test through the newsletters of the Huntington's Society and I thought it was stupid. Why in the world would you want to know? There was just no way that I would entertain something like that. I figured that you're not supposed to know what's going on in the future. You live by faith so why dwell on it? But I was in my early 30's then so I had a different perspective on life. You get to 40 and you start to look at life differently whether you have Huntington's hanging over your head or not. You start to look at where you've been and where you want to go.

All through this last five years I thought that I should at least, for my children's sake, get my blood banked in case I get bumped off by a bus or something. That way, if they ever wanted to know whether they're going to get it or not, there's some blood on hand and they can get the predictive test done. That started me thinking. At the same time the Huntington's cloud was gathering on the horizon and getting closer and closer. Then it started hovering over me. I didn't think about it heavily every day but there were more and more days where I was thinking about my risk situation. Having your own business you have to plan ahead. And then there are the children. I was thinking about all this and my thinking went from not wanting the test at all to at least getting my blood banked to thinking that maybe I would get the test so that I could use it as an instrument for planning my future.

I had a time last year where things were happening in the business and I thought maybe I might want to pack it in. It was not a great time. At the same time my wife was having a hard time with her family so there were too many things working on the family. It looked a lot better to go back to Ontario, start fresh and try to enhance my brother and my mother's life. But I thought I

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can't do that right now. I need to know whether I'm going to get the disease because I wouldn't want to take my family and my wife away from family and friends especially. She would need her support system if I should I get the disease. That's what brought me around to wanting to know for sure. Over two years I did a complete 180 degree turn in my thinking and that's where we're at now.

**The Big Problem with This Disease**

My decision to have the test was something I came to on my own. I didn't talk to Emily about it. I haven't discussed my feelings because I don't want her to be concerned. It's sort of my way of protecting her. I mean if I really shared my innermost thoughts over the last few years, I would tell her at times that I'm scared. And I am scared when I trip. Sometimes it does concern me, but I'd never say that to her or to anybody else because then they'd just start looking for it.

The big problem with this disease is you can't talk to anybody about it, including your wife. We talk about my Dad or my brother or whatever and she'd often ask me "do you think you've got it or you're going to get it?" I somehow felt that I wasn't going to get it. There's no scientific reason to feel that way but that's the way I felt at least until the last two years. Then it became sort of a grey area. But if she'd ask I'd still say no.

She would see me do something and every once in awhile I think she would ask one of my friends if he'd seen any change in me. Just double checking, which is fair enough. No one admitted to seeing any difference. I was just stressed out but it's easy to blame it on the disease. If I trip over something I'm sure I've got it. It's a mind trap to think that way but during the last couple of years this big cloud has been hanging over everything.

**You Don't Just Spring it on Somebody**

I came to conclusion that I should take the test a long time before I told Emily. And she was glad because she had thought about it too. And the friends that she's told are quite happy that I'm doing it for our daughters' sake. I could live without the knowledge but you get to the point where you feel you owe it to your kids.

I don't think my kids know that they are at risk. I don't think they're old enough. They know Grandad had it, that uncle Brad has it, they know I can get it. I'm sure they know that and they've been told that it can keep going on. But it's not something that we've sat there and tried to give them a big education on. Depending on the results of the tests we'll handle it with them as they get older. They don't know that we're taking the test. They know that we're in some sort of research program, thinking it will help uncle Brad and people like him. And that's the way we'll leave it, because if it comes back negative then there's nothing to talk about. And if it comes back that I have got it then they don't need to know right now. I remember it was pretty hard when I found out about my Dad. You are confronted with mortality in a new and different way. I remember going through that process myself.

My father-in-law, sister-in-law and a couple of my wife's friends know I'm having the test. I haven't told one of my good friends yet. It's not just something where you phone up and say, "Did you know that I was getting my future told or foretold?" I saw my friend a few days ago but by the time we caught up on all the normal things we talk about there was no time left and you have to do it in the right setting because you can blow somebody away if they're not ready, and really put a burden on them if you don't treat it right. You don't just spring it on somebody.

**An Opportunity**

We will make whatever lifestyle adjustments we feel we need to make sure we have a relatively good quality of life and enjoy each other. So in that way this test is a real opportunity. We view it as an opportunity even if the results aren't the way we want them. We know we have a time frame. It's not like somebody with colon cancer, that's got two months to live and has to go into chemotherapy the day after they are diagnosed. But I only look at it that way because I've gone through the process of thinking about what the test really means to me.
I feel good now but I think it's probably because I'm so busy that I'm not even thinking about it. When I went to the clinic the first time they told me that when they did the neurological exam, they would know right away if I'm showing signs. They said they could tell me that day if I wanted to know. Well my heart sank. My wife's heart sank. I said, "no, I don't want to know anything like that yet." I'm not ready for that. I'm ready for the test but not that. Then we canceled an appointment in April because we couldn't get our insurance reviewed and in line quickly enough.

I wanted to increase my insurance. I told the company I was dealing with that I had been rated as an insurance risk by another company. They asked why and I told them it was something my Dad had. They already knew about it since they have a data bank for all that stuff but they still came back without any additional rating. That was the best news I've had for a long time. It's not a lot of insurance but I can't afford to buy a whole lot. They did ask me if I'd taken any test for the disease. I said no, which was a half truth I guess, but I don't know any results. And they didn't come back and tell me that I should have the test as a condition of insurance. Another insurance company that was on Market Place on CBC said, "there's a test, if you want insurance you get the test." And it's pretty scary that the insurance companies are taking that line because then I feel discriminated against. Just because there happens to be a test for Huntington's I have to take it to get insurance. If I wanted a cause I would take this on and try to get some dialogue going at the political level. The government has to talk about this because the science is going faster than politics or psychology. These things have to be talked about before the insurance companies have it all their way.

When you are young you have your health and everything looks great. But you really have to come to terms with mortality. And when you have the possibility of something like this you need to look into it and know as much as you can about the disease. I think you have to come to terms with the reality in your life. All of a sudden things that you've been believing all your life go out the window. It is quite amazing. The technology is coming faster than the psychological understanding. Testing sounds like a good idea but you've got to think it through.

I'm glad I didn't tell the world that the whole thing was stupid since now I've reversed myself. I think the only thing that I would hope for other people is that they're able to go through the process with somebody else and if they have access to it, the Huntington's Society has an at-risk support group. It's really good to go there. You can share your feelings. People can relate to it, they can empathize with you because they've had the same feelings. There's a real camaraderie in talking to people in the same situation. You draw a lot of strength and I think from what the facilitator said my comments about how my thinking had evolved really helped others at the group. And that feels good. It always feels good to help somebody else.

I Can Lift the Cloud

I had an idea that the pre-counselling would have more to it than it does. I don't feel like I've garnered anything from it other than I know exactly what to expect with the process of testing. It hasn't helped me in my thinking but from what they tell me Emily and I are more prepared for this than almost anybody else they've seen.

I really appreciated the fact that my wife was able to go with me. She was there, she could see what was going on, she could hear for herself, she could ask her own questions. Emily is a bit paranoid that everyone there is walking around knowing what my future is, which isn't the case but it's an odd feeling. It is kind of unsettling. We've had assurances from the genetic counsellor that she won't know until we know but somebody knows that whatever number I am is one way or the other.

I'm ready now but on Tuesday morning it might be a whole different story. Emily's working in the morning but she's arranged to have the afternoon off so she can come in with me. We had it planned more or less because of the kids. They're going to be away over night.

Wednesday morning will be even more interesting than Tuesday morning. I haven't really thought of what it will be like. Every time I think about it, I don't want to think about it. On a good day I figure knowing for sure is better than not knowing. But that's easy to say when you
don't know for sure. I seem to think more as if I have it than I don't. I may be trying to protect myself or I may just be trying to be realistic. I certainly could adjust real easy if I don't have it. I can adjust to that news so I want to make sure that I can adjust for the other reality if that's what it's going to be. It's consumed so much of my thinking in the last few years. I have thought about it more than almost anything else. It took me a long time to decide I wanted to do it. I had to evolve towards it but I can lift the cloud.

I spoke with Colin again about six months later. He and his wife Emily were relieved to know that the results were negative and that their children were not at risk for HD. But as Colin explained, it took a long time for it to sink in.

The last few days before we went to hear the news was a busy time. We were busy at the store and the house was a mess because we were renovating. The neighbours all thought we were nuts because on top of everything else we had a big truckload of top soil delivered to the backyard. And then I just got bark mulch for Emily's garden and the neighbours thought, "how many projects do you want at once?"

It wasn't a good time to be going through predictive testing but when is a good time? I wasn't going to forego the chance because at that point I was committed to wanting to know the answer. I wanted to go through the process and I wanted to know as quickly as I could.

I know I was getting pretty testy before results day. One of my friends got off the phone with me and said to his wife "I don't know what's the matter with him, I've known him for 20 years and never heard him talk like that or be that way. Is something going on over there that we don't know about or what's happening?" I think I was quieter at work. I don't remember having any problems with increased temper or anything like that but my office manager asked me a couple of times if everything was all right with me.

I know it was really hard for Emily because we both came to the conclusion during the last few days that I had the gene. We could deal with it for us, it was the children. We were told before we got married that we shouldn't have kids but you live and hope that first of all you're not going to get it, and secondly if you do get it there will be treatment and by the time you have kids there will be a cure. And on top of that you still have a life until it happens, even with kids. But when you're confronted with it and you realize that you've done something that you could have prevented, you've had three children and you could have prevented passing it on, it wasn't a nice feeling for us as parents. And that sort of came to a head just a few days before.

Please Don't Let Dad Have Huntington's Disease

We didn't tell the girls what we were doing. We told them we were helping with research for Huntington's disease but not about the test. But these little darlings figured it out. Somehow, they figured it out. I wasn't there but on the morning that Emily and I were going in for the results, the girls were at breakfast and the little one was saying grace and asking God "please don't let Dad have Huntington's disease." So how they figured this out I don't know, we had tried to be so careful. Maybe the walls have ears.

It was a long drive in to the clinic, literally and figuratively. We still had a certain peace because we had prepared ourselves for the worst and honestly felt that we were ready for it. And we had looked at the positive side of the worst and felt that we could deal with it. So when I got the results I was not ready, it just went right over my head. The doctor started off by coming into the room and saying that he had some news that he thought I would be pleased with. I didn't say it out loud, but my mind said, "then he'll tell me the bad news." So he told me that I did not have the gene and it didn't fizzle on me at all. I looked at him and the genetics counsellor and wondered what they were thinking. Should I be going up and down the hall doing cartwheels and shrieking and hollering? I had no reaction, it was nothing.

For people that know me, my reaction was probably predictable. I just sat there and listened. I'm not an excitable type of person as a rule. I was relieved but we honestly weren't expecting that result. We had convinced ourselves that I had the gene and probably already had the disease. I guess we wanted to make sure that we wouldn't have this big let down but unfortunately we did
I have a let down. I've been carrying this monkey on my back for about 24 years and it has been such a big part of me that I can't just throw it off my shoulder, walk out and do a few cartwheels.

When we left the clinic we phoned the girls at their grandfather's house. The middle daughter answered the phone and the first thing she said was "do you have Huntington's disease?" So it just reminds us that there aren't any secrets with kids. You think there are and maybe they're just being polite, but they don't talk to you about some of the things they hear. Of course they were really thrilled but I don't know that they really realized one way or the other how it would effect their lives. When we saw them, we talked about it and it was like "oh yea". They changed the subject. They're still young, and they're not that interested. Kids are wonderful, they just shrug it off and go onto the next thing.

I Should Be the Life of the Party

For me, the results certainly didn't sink in that night. It sunk in a little bit each time we told somebody. I guess we were just sort of shocked, we weren't prepared for that answer. We walked out of there kind of numb, not knowing really how we should feel. But we went out and had a nice dinner. I don't even know what we talked about but it was just nice to go and enjoy ourselves and unwind. Then we came home and stopped at a friend's house and a neighbour's place to tell them because they were standing by waiting for the results too. We had planned not to tell anybody for a couple of days but I guess by the time we got home we had to tell somebody and that felt pretty good. They were probably wondering why I wasn't giddy and ecstatic. It would be easy to say, I should be the life of the party but it didn't go that way for me. But it was nice to have shared it with them. Then we went home and I don't know if we opened a bottle of wine or not, we were pretty tired and emotionally exhausted.

After that, it was business as usual. I got up the next morning and went to work and I don't think I was any cheerier than usual. I didn't feel any different because there's always the work at hand and that's all you deal with. There's days where I'd just as soon go find a 9 to 5 job somewhere and stop and smell the roses. Then maybe some of this stuff would sink in. It took a long time for it to sink in, about 3 months. We were immediately relieved for the girls' sake but I still don't think it's fazed me properly. Part of it is that I've just been too busy to really think about it.

The other reason is that we have very few people we can share the news with. Because of the nature of this disease you don't tell anybody that it is in your family. If they understand that it's hereditary they put two and two together and you're really jeopardizing your position in work or insurance matters. So we didn't tell very many people at all about the test. It was only when I was able to tell somebody that it worked out well, that my enthusiasm for the results really came to light.

I Have to Deal with 'Survivor's Guilt'

My mother passed away quite suddenly in September so we all went back to Ontario to deal with that. My brother was there as well. He's still independent but probably not for more than another year. I found that with the disease not being a threat to me now it's easier for me to deal with it with him. Before when we'd spend time together he used tell me what he was experiencing at each stage. I could only take a certain amount of that because I was still at risk myself. I didn't want any other reminders of what my future could be. But going back in September to see my brother it was a lot easier to deal with. I think it will help me to be more understanding because I don't have that emotion or threat. The only thing I have to deal with is survivor's guilt and I have a great deal of that.

I figured the less people know the better. I never told Brad about the predictive testing. I did get a blood sample from him. I told him and my Mum that it was just in case our girls ever wanted to be tested. I didn't tell them that I was doing the test. But when I got the results, the doctor asked, "well, who are you going to tell?" I said, "I'm not telling my brother or my mother." He asked why and I said "because I feel guilty, especially in my brother's case I feel guilty." The only reason for not telling my Mum was that I was afraid that she'd unintentionally tell my brother.

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But the doctor said to me, “what you received today is a gift. You have the gift of knowledge, you know.” We were looking at it that way but the doctor said “would you not want to give that same gift to your brother and mother?” I said I’d love to tell Mum but I’m not sure I want to tell my brother. If I tell my brother it wouldn’t be over the phone, I would want to spend some time with him and see how receptive he might be. His hunch was that my brother would be really thrilled and I suspect that maybe he would be too.

But I still can’t bring myself to tell him. I’d go back there, fly in, rent a car, go see him, stay in a hotel and go for dinner and it all must have looked really glamorous compared to where he is in life. I don’t tell him about vacations or things that I’ve done because I’m afraid that’s a reminder to him of what he’s missing. So I guess for the same reason I don’t really want to tell him that he received the gene and I didn’t. Maybe I will some day but I’m not there yet.

A Real Gift

I did tell my Mum and I’m glad. I phoned her within a week and she was thrilled. I had planned to tell her the next time I saw her in person but it would have been too late. As it turned out, I had tickets to go down and see her for the week after she died. So I was able to pass the gift along but she did share it with others. I met some family friends and they had heard my news. My brother may know too but he hasn’t said anything. So my worst fear was realized, my mother was just so excited you can’t blame her. And it was probably good for her because she had one less kid and three grandchildren not to worry about. In that way it was a very real gift to her.

There are certain people you can’t tell afterwards even though you have good results. They will feel bad that you never thought enough of them to share it with them before. Or, in my case, with a business and with people that work closely with me, they would probably feel pretty upset retroactively to know that I got into a business relationship with them knowing that I was at risk. You can’t just come back one day and say “Hi,” you know, "this is what I did," and no worries. You always have to be thinking who you can tell and who you can't, even afterwards. Now if they happen to find out then I’ll just tell them, “No problem, I had a test.” I don’t have to tell them it was this year or last year. It could have been 20 years ago. I just had a test, it’s not happening.

Emily had friends that she could share it with and I know she got back to them. I have one friend and I really wanted to share it him but we just couldn’t get together to do it. And I still feel bad about that. I wanted to tell him before the test but we didn’t have a chance until afterwards. We went on a fishing trip and I told him the whole story from start to finish without telling him the conclusion until the conclusion. And then I apologized that I hadn’t shared that with him before. It was good to tell him. And then he shared something with me, a concern about his wife’s health which had him facing similar emotions. Not the same devastating sort of thing but there were some problems that a buddy could empathize with.

I also told my cousin and her husband that I was having the test. She is a nurse and they’re both in the medical field so they could relate to it and they’re always interested in my brother. But I feel really bad because I had forgotten that we had told them so of course I didn’t phone them with the results. And then I phoned them on something else quite a few weeks later and they asked, “Oh, by the way, how did you make out?” And I felt like two cents. People that knew of course didn’t want to ask. And it must have been hard for them to wait to find out because they didn’t know one way or the other.

One Day I Will Turn a Cartwheel

The other thing that I’m living with right now is that I haven’t grieved my mother’s death. I had to go back east, I had to do the funeral because my brother wasn’t capable. I wanted to take an hour go down to the cemetery and just sit there, say good–bye in my own way, all by myself. It’s sort of just below the surface but you are so busy that you keep pushing it back, pushing it back and then one day it won’t be pushed back anymore, it’ll come. So I guess that’s the way it’ll happen. And probably the results of the test will happen the same way. One day I will turn a cartwheel.

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I do feel that I would like to move back to Ontario and really try to give my brother a better life in his remaining years, let our family be his family. Emily is quite prepared to do that. We had a wonderful extended family experience while we were back there and realized really how many friends and acquaintances we have. We realized that we could pick up and move into a community and have an immediate circle of friends.

Since I learned my results it would be easy for me to turn my back and say, “I’m out of here. Twenty-four years I’ve lived with this, it’s history. I don’t have to pay my dues to the Huntington’s society anymore. I don’t have to go to the meetings anymore.” But I don’t feel that way at all. I feel a sense of duty. I don’t have a lot of time but if I can be available for somebody that wants to have a chin-wag, especially at risk people, I think the story that I have to tell could really help a lot. I’m not trying to brag but I’ve never been so aware of how a person can go 180 degrees in their thinking. I’ve never done that on any other issue on my life and I think it is important to understand how each person and family affected gets through these things. When you have a monkey on your back you’ve got to have it on your back to know what it’s like. You can imagine but it’s not quite the same.

I wish I could have taken two or three days after the results to think about what happened. I would have liked to have had some quiet time to process everything and put it to rest. I also wish I could talk about it to more people. But again, I think most of my pleasure from the whole thing is for my daughters. It’s nice that they can look at their uncle and realize that they don’t feel threatened just as I don’t feel threatened. I can look at him or I can go to the Huntington’s convention and see people and realize that that’s not me in five years or two years. I don’t have that fear anymore. That’s when you feel like your load’s lightened.

Postscript: I spoke to Colin and his wife about two and a half years later. They had moved back to Ontario and both found work. They were able to locate a good facility for Colin’s brother but they continue to face many challenges in caring for him.
I talked with Gabriella about three weeks before she was scheduled to receive her predictive test results. At this time, Gabriella was 54 years old. She had finished teaching for another year and was trying to enjoy her summer vacation. Huntington's was, however, very much on her mind. Gabriella began the interview by telling me about her recollections of her mother's illness and its impact on her and her sisters.

I grew up in Alberta in the 1940's and 50's. I have two older sisters, Karen and Annette. I'm sure that if it had been 1980 or 90 my sisters and I would have been apprehended by Social Services. My mother really beat us. We lived in fear a lot of the time. I think it was worse for Annette than it was for Karen and me because my mother was rather paranoid about my Dad and Annette was a lot like my Dad.

When I was young I always went to church. We all did; the three of us sisters used to sit there in our parkas. For me, church was a major crutch until I was about 19 or 20, and then I didn't go very often. Now I go to church with my sister Karen when I visit as she really is a strong religious person. I'm not an atheist. I do have some kind of spiritual belief but I'm not attached to any particular church. I went to a United Church funeral awhile ago and it just took me right back to when I was young and we would go to church. As a little girl, I always felt safe; church was the place that saved me from the Boogie Man, the dark at the bottom of the stairs.

People Said... She Would Dance Through the Fields

My grandfather probably had Huntington's but he was never diagnosed because he died of cancer when he was 54 years old. After he died in 1944, my mother went on an amazing search for her roots.

My grandfather and grandmother came to Canada together when they were very young but my grandmother died when my mother was only 11 days old. My grandfather would never tell my mother anything about my grandmother so she went back to England in 1947 and then again in 1953. She was desperately searching for information about her father's family as well as her mother's family. People said her father had a sister that was crazy and that she would dance through the fields. We figured that she must have had Huntington's too. My grandfather's sister also had a child but we've never been able to trace this. When I went to England I checked but there's nothing left, no records and no one to talk to that would know.

Trying to Find Something

I didn't know about my grandfather's symptoms of HD until much later. My mother would have been about 40 years old when her symptoms appeared so I was 7. When I was a teenager, we were told she had schizophrenia. Later, when she lived in Vancouver, she was often evicted from her apartment. She would talk to the heat registers and she often had fires. When I was 20, she was admitted to a mental hospital and she was never released on a permanent basis after that. In the mid 1950's she was going to have shock treatments. She was trying to find something and I feel very sorry because she didn't have a clue what was wrong with her.

After my daughter Suzanne was born, I wrote a letter to the psychiatrist at the mental hospital to ask if they knew what my mother had. I told the psychiatrist that I thought my aunt in Ontario had an illness which was quite similar to my mother's, although perhaps not as bad. The psychiatrist did not write back to me so that got me no where. That was about 1964. Then, in the summer of 1965, my mother took a terrible down turn and my sister phoned to say I had better come. I was living in Ontario at the time and was pregnant with my son Jason. I went out to Alberta and that's when the doctor told us that they thought my mother had an inherited disease but they wouldn't know for sure until they did an autopsy.

My son was born on December 2nd and my mother died on December 14th of that year. Then my Dad got married on December the 30th. He had wanted to wait until my mother died because he didn't want to even get divorced from her. He used to go and visit her all the time.
Then she hung on and hung on so he made plans to get married. She died two weeks before his wedding date. They didn't postpone the wedding which was fine with me, but it wasn't fine with everybody.

I don't remember worrying about Huntington's but I know I did. I had a really difficult time with Jason as a baby because I kept thinking that I shouldn't have had him. I can remember walking the streets thinking what am I doing, having had this baby? At that time we were told that Huntington's was inherited through females only. My doctor in Ontario actually got a letter from someone at the Alberta mental hospital saying that it was inherited through the female and that it skipped generations. I knew that this wasn't true and so did my doctor so he wrote back and said “aren't you incorrect about this?”

In the spring the autopsy results confirmed that my mother had had Huntington's. I went and asked my uncle in Ontario, if my Auntie Sylvia had been diagnosed with Huntington's because I was still hoping that my aunt and mother had different diseases. My uncle said he didn't know. Then, several years later when my Auntie Sylvia died of cancer my uncle wrote me a letter saying, “I don't know why I didn't tell you before but your aunt was actually diagnosed with Huntington's in 1962.” He knew before my children were even born. In his letter he said “I don't understand my reasons now but I'm writing to tell you that I'm really sorry.” I really liked that uncle and I liked my aunt too. I could never figure it out. Unfortunately he died about 3 or 4 months later, so I never had the chance to go back to Ontario to talk to him. I still have his letter but I don't understand his reasoning. People are funny about this kind of thing.

Some Advice

Huntington's was one of the reasons why I went with CUSO to teach school for two years in West Africa. I said to myself, you better get on and do something with your life. So, along with my first husband, I taught with CUSO in West Africa. We moved back to Canada in 1969 because he was going to get his Ph.D. at the University of British Columbia.

In 1970 I had my tubes tied and I really regret it now. I had gone to see a geneticist because I wanted some counselling about being at risk for Huntington’s. It was a different type of counselling in those days. All I wanted to do was get some advice but when I went into her office and said I wanted to talk about the idea of having my tubes tied she just said “there's absolutely no question about it, of course you should get your tubes tied.”

I am a strong feeling type of person and I'm ruled by my heart much more than my head. I was much more vulnerable to people in authority than I am now. So I went ahead and did it, and that was a very hard thing for me to do. I always wanted to have six children. It was a total nightmare, that's how I remember it, just saying to myself “you have to do it, you have to do it, you have to do it.” I was depressed and really upset but determined to do what seemed to be right.

About a year later my husband and I separated and I went back to university to get my teaching certificate. I met my current husband Brian and we started living together in 1973. Somehow or other we managed to have all of our children at home every weekend. I had my son and daughter and Brian had four children as well from his previous marriage. We used to have lots of family dinners that were lively and fun. We were and still are a really close family. Our kids have always been very important to us.

I remember exactly when I told Suzanne and Jason about Huntington's. They were 10 and 12 I think, old enough to have some comprehension. We were driving along in the car and I remember saying to them, "You know the blood test that we just had? It was part of a research study that a person in Saskatchewan is doing on a disease that grandma died of and it's a disease that's inherited". I just explained it. I don't remember a horrendous discussion or crying or lying awake worrying. I don't remember any of that. Of course they had never lived with anybody with Huntington's so I don't think it was real to them until they got older and realized what it was.

In those days I used to have Huntington's meetings at our house. This was before the Resource Centre was set up. There was some young guys with Huntington's that came to the meetings but

Narrative Accounts/Gabriella
not when Suzanne and Jason were around. I was quite frightened of exposing them to people with Huntington's. I was reluctant to do that.

Times Change

After Brian and I got married, I tried to get my tubes untied. I was going to do this before we married and then I decided I couldn't do it unless we were married. So after we got married I went back to the gynecologist all prepared. This area of medicine was his specialty at the time. Brian and I had gone through all of the preliminaries and it was all set in my mind. But then when I walked into his office he said, "I can't do it, you're 36 years old. I just came back from a conference in New York where I said the cut off date for this kind of thing was 35." He said, "Go home and count your blessings, I have only one daughter." I was crestfallen. That was in 1976 and given that a lot of women are now having their first child at age 40, 36 seems like an unreasonable cut off date. So times change.

I'm not sure when I started going to the Huntington's Society meetings. I was very involved in the 1970's. My sister Karen and her husband have always been involved too. They were major fund raisers in Canada for years. So I got a lot of information about Huntington's from Karen but I also went to the medical library to read about the latest research. I was a pretty independent person. That's been one of the things I've had to learn to moderate. I never relied on my first husband for HD related support and I certainly didn't after I got my tubes tied. My sister Karen has been the person that I talk to about it the most. She is basically the matriarch, the stable person in our family. She's lived in the same house, she never got divorced. My children all just love her and she has been my support in a lot of things.

The Most Amazing Relief

My life was dominated by Huntington's for a long time but the most amazing relief for me was when I had a PET scan. This was when PET scans first came out as a diagnostic tool. I can remember exactly what I felt like just before I got the result and how exhilarated I was afterwards. It was my first 24 hour period of freedom from Huntington's since I'd found out that my mother died of it.

Karen and I decided to be part of the PET scan research so we had been going for a PET scan every two years. It wasn't until they started offering predictive testing that I could get the results though. This was when the study was no longer just research. In those days that PET scan was a god-send. Before I had the PET scans I could lose my temper three times in a row and people, including me and Brian would say "Oh oh, she's probably got Huntington's." Even normal behaviour didn't get treated as normal, and that retarded some of my own personal growth. I could never accept the behaviours as my own because I just couldn't deal with them. So the PET scan freed me in other ways and allowed me to accept myself. I would have to be responsible for my behaviour not Huntington's.

When researchers first started offering the predictive testing (using linked markers) our family had trouble obtaining enough blood samples. Also, I didn't really want to go ahead with it then because of the uncertainty. I felt that even if I got news that I was 95% safe, that wouldn't be good enough for me. I'd still be nagging over the 5%. So I wasn't interested in pursuing it. Then, in 1989, my sister Karen was diagnosed with Huntington's which would have made predictive testing easy to pursue, but for some reason I didn't feel the need to.

It Wasn't a Very Nice Situation

Karen was about 54 when she was diagnosed with Huntington's. That was six years ago. It wasn't a very nice situation. She came out here to have a neurological test and a PET scan. When the neurologist walked out of the room, he left the results of her test sitting on the table and she looked at them. A couple of hours later she came back to our house and said "I've got something to say to you." She was really distressed but she was determined to tell everybody herself. I admire her for that.

Narrative Accounts/Gabriella
Her husband and her daughter had suspected that she might have Huntington's. And I think I knew it too. When I came home from picking her up at the airport I had said to Brian "Karen's got Huntington's. I can tell from the way she picks up her suitcase. It's that extra movement.” I just went to bed and cried.

**Tired of the Worry**

Once they discovered the gene and the 100% results were available, the road seemed clear. A few years ago I dithered around about getting the results but now, I feel that if I have the gene I want to be part of any research that's going on. It also seems different now since I need to think about planning for retirement. If I have the gene, I don't think I'll teach more than two more years. I'm very dedicated to my job but I recognize that it has taken up more of my energy than I want it to. I have little energy left over for anything else so I'm thinking seriously about what I'm going to do in the next few years.

I talked about it with Brian and I certainly talked about it with Suzanne and Jason. I needed their okay. I don't know what I would have done if Suzanne said, “Don't do it because I can't handle it.” I might not have gone ahead. But they both said go ahead. Jason doesn't seem to make a major thing out of it and Suzanne said “I can see why you want to do that and I think it's important for us to know.” As Suzanne said “Mother, I think it's really important because if you've got the gene we need to plan our lives with the possibility of helping you.” That's a pretty warm thing to say, that was her attitude.

Brian’s responses vary with his moods. He's in favour of it right now, I know that. In fact many years ago he said to me, “You're worrying about it too much, go to a psychiatrist.” This was before the PET scans were available and no diagnostic neurological tests were available either. He said “Go to a psychiatrist and ask his opinion and then take his opinion.” I did follow Brian's advice and that probably lasted me about 6 months. I even went back one other time. Brian encouraged me to do that and he’s always encouraged me to be part of the PET scan. I think he's really tired of the worry of it all and the unknown as well, and how much it's dominated our life. I'm sure he's fed up to the teeth with it and I am a bit myself.

**I've Got Myself in This Huge State**

I've been hanging around the house for 2 days working and doing some of the things I like but I've got myself in this huge state. I'm busy making jam. I need to clean out the basement. I'm going walking with two neighbours and I'll also get my bike out. I've also been thinking about taking an extra course but what I really need to do is unwind.

It's a little bit more serious than I had thought. It seems to be hitting me in a heavier way than I had anticipated and I'm a bit worried that I'm not understanding myself well enough, that I might react to the negative news more strongly than I'm anticipating. Maybe it's just a calm me down period that I'm going through right now and it'll all come out fine.

I think the results are going to be okay. But how do I know? It's not just wishful thinking. I've done some scientific thinking too. Knowing that I have 4 years following a clear neurological exam, I think that at age 58 my risk of having Huntington's is getting lower. But then again my aunt who has Huntington's was 60 when she was diagnosed. My sister Annette is now 58 and she doesn't have it. So I say, “well two out of three sisters or one out of three sisters?” I don't know.

All of our kids know I’m having the predictive test and probably three or four of my friends. And my Dad and his wife Sharon and my sisters of course, they all know. I almost mentioned it to a student. He had leukemia when he was young and was really worried about having a recurrence. I've had a lot of personal conversations with him and I was almost tempted to say something but I thought no, not school, not kids, not parents.

I did come out of the closet at school about ten years ago because I decided to sell the Huntington Society Christmas cards. Then to my surprise I found out that one of the other teachers I know well was also at risk. So there are people at school that know and they know I'm having the predictive test too. On the last day of school one of the other teachers asked me when
I would get my results. So I do have people that I’ll have to phone. But I feel almost embarrassed about it. I don’t like being the centre of attention and to some extent phoning people makes me feel that way. It makes me uncomfortable, as if people will feel sorry for me or something.

If I don’t have the gene I’m going to wish I was in Alberta to give my sister Karen a hug. I feel to some extent that I’m cheating her if I don’t have it because then she’s the only one. On the other hand, if I do have the gene that’s going to be hard for my Dad. It’ll make him, as well as a lot of other people, sad.

What we’ll do with the news if it’s not good I’m not sure. I honestly cannot predict how I’m going to react. If the news is bad it isn’t different than it was yesterday because I’ve always had it. I’ll do my best to forget it until I have to not forget it, but I certainly won’t like it for my kids, I know that.

I was thinking of getting my results and then going away but I can’t do that if the news is bad. I can’t walk away from Brian, Suzanne, and Jason. I need to be with them. But I also might have to go away by myself for a couple of days. I don’t know. I’m aware that I’ve missed out on a lot of support because I have this independence thing. I don’t seek help unless I’m desperate. I always find things out myself unless I get to the point where I have to ask someone. I don’t find it easy to ask. “Gee what have other people done in this situation?” But I am learning that maybe that’s a jolly good idea. In this certain circumstance I might really benefit from doing that.

Gabriella did not want to lose her entire summer holiday to worrying about HD so when the opportunity arose to obtain her results a little sooner she adjusted her plans. Her husband, daughter (now age 31) and son (age 29) went with her to the clinic and this was, in her view, a good thing since the results were a bit complicated and everyone had a lot of questions. Several months after receiving this news, I talked to Gabriella again and she began by explaining how she dealt with the anxiety of awaiting results day.

Moving Rocks

During the week before my results we went to an island where friends of ours own property. It’s very barren but it’s an absolutely beautiful place. Our friends were using dynamite to clear the area where they’re going to build their house and so I ended up moving a lot of rocks. I worked for the whole week. The weather was fantastic and I spent a lot of time by myself because Brian was working with two other men who were building a wharf down the island a little ways. I’d make lunch and they came back for lunch and then we’d all go back to work. I loved it. I just spent hours and hours moving rocks. It was so therapeutic.

We came home on Sunday night and I got the results on Tuesday. I was very anxious on the morning of my results so it was really nice for me that Suzanne was here. I just wanted to get it over with. But I always do all the things that are going to make me feel good, like I wash my hair and wear something I like. I always do that for these things.

This is Complicated

Suzanne and Jason and Brian went with me to get my test results. I can see everybody and where they were sitting but I’m quite blank about the visit. Getting the news was a shock, I don’t know how else to describe it. Getting the news was a shock, I don’t know how else to describe it. I expected to hear that I didn’t have the gene.

The geneticist told me how many repeats I had. Then he said “we don’t usually tell people how many repeats they have but we’re telling you that you have 36 repeats because you’re in this odd category of people.” He said it was complicated. The clinical team didn’t think it was going to be like this. He said, “we’ve looked at all of the data and there are only about 15 people who have between 36 and 38 repeats. Some of them don’t have Huntington’s and they’re in their 80’s, there are some who are in their 70’s, some in their 60’s and some in their 50’s.” We had a long discussion about this. We also discussed the inheritance pattern as it differs between males and females, what that meant for Jason and Suzanne, and, how odd it is that my sister and I have
quite a difference between our number of repeats. We also wondered how many repeats my mother had since she got Huntington's when she was 40. It seemed that we had a bit of an odd pattern in my family. It was interesting too in that if I'd had the predictive testing several years ago (with the linkage test) I probably would have been told that I had very high expectations of having Huntington's.

The geneticist said he felt like he was giving a university lecture, we had so many technical questions. He gave us a few articles to read because we like knowing what there is to know. He also asked us not to tell anybody about how many repeats I had. He said they didn't generally talk about it as they were re-thinking the process of how to provide people with their test results.

Jason was really upset. I was amazed and worried at how upset he was. He started crying not very long after I got my test results. I had worried a little ahead of time because he had never acknowledged worrying about it. He said he always felt like I never had it and so it was a major shock for him. Suzanne was fine. In fact she was stronger than I thought she would be.

**Bad News with a Good Flag**

After I got the test result, it was almost like a high. It was so odd. I guess it was just the fact that the anxiety was over. It was also so peculiar because I had these odd results. It's like you have it but you don't have it. So I had to focus on the positive, the fact that it was kind of bad news with a good flag.

We ended up having dinner together, Brian and Suzanne and Jason and I, and I think one of Brian's children. I remember not being very upset for some reason. I phoned all of our kids that night. I spent half that night on the phone, I remember that. My step-son Nathan said to me, "Gabriella this is like not having any news. It's almost worse." And I said, "You know, Nathan, for some reason it isn't worse. It's bad news with a good flag whereas before I was neither here nor there." And he said, "Well, you're kind of like that now." And I said, "No, for some reason I don't feel like that." The news is better than it could have been and that's what I'm interested in hanging on to.

**You Sure Know You've Got Some Good Friends**

On the day after I got my results I went in to the clinic to have another blood test. Brian had suggested that it would be good to make sure that it really was my blood that had been tested since we had this odd combination of repeats. Whoa! It just hit me at UBC. They were taking the blood and I thought, "oh oh, they're analyzing it for the bad gene not for the good ones and that's what they're looking for and that's what I've got—these bad genes." It got to me and I had to cry for about 2 hours.

I phoned my friend Deidre that evening because we often go for walks together, and I said to her, "Deidre do you want to go for a walk?" And she said, "Oh Gabriella, I just came in from a walk with the dog and I can't go." I said, "Okay, well I've got some news to tell you, I'll tell you over the phone, it's not great." And she said, "Oh my god, don't tell me. I'll be at your door in a minute, I'm just going to put my shoes on." So I said she didn't have to and she said, "Yes I do. Just wait, I'll be at your door in half a minute." And so we went for a good long walk.

What I remember about the week after is that you sure know that you've got some good friends. I don't have a lot of energy or lot of time to spend with friends, although I have made an effort, in the last year because I've decided that's important to me. What I remember thinking, though, is you might as well focus on the positive cause there sure is a lot out there. One friend gave me a gift certificate for a manicure and some other friends came over with a big bouquet of flowers. And on the same day that I got the results I went to London Drugs and bumped into my allergist. He just happened to be there and I saw him and told him. He's a very comforting person. And that's what I have to say, people are really nice. I know a lot of really genuinely caring people and it wasn't hard to tell them.

Kyle, one of my colleagues at school, knew I was getting the test and I phoned him the day before school started and told him my results. I said, I just want to tell you because I don't want
to talk about it at school.' So he said, "I've been thinking of you." He said he knew exactly where he was on the day of my results and that he was thinking about me. And then I told Trudy, who I work with and one other person on the staff at school because she and I have taught there for a long time and I knew she was concerned. I haven't told anybody else and I don't intend to tell anybody else.

**God Bless This Family**

A few days after I got my results, Suzanne, Jason and I went to Alberta for three days. I felt badly because Brian didn't go but it was Suzanne who said "Mum have you actually thought that somebody should be staying with Brian? You've got this news and you left Brian at home." I was really impressed with Suzanne's concern.

I told my sister Karen my results before I went to Alberta and everybody there knew. One night while we were visiting our whole family went out for dinner. Sharon, my Dad's wife was sick and couldn't be there but she had arranged a cake that said "God Bless You" on it. I would have liked a cake that said "God Bless this Family" because I felt like I was having undue attention. I didn't feel comfortable about it. My sister's daughter Marlene was great though. She got up and said "Well there's been a lot of tension in this family, there's been a lot of phone calls back and forth," and she said, "when I get tense, I shop." Well she bought a goofy present for everybody there. It was such a great way to make everything lighter. I really appreciated it because I thought, it's not me, it's our whole family.

I had many good conversations with my middle sister Annette, more open conversations about Huntington's than I've ever had with her. I never knew that Annette worried about it because she never wanted to talk about it. My Dad hasn't dealt with it. He doesn't understand it and I don't think he wants to, that's the odd thing. He told my aunt in England that the news was good and so I had to write and tell her that it wasn't quite that way. He's having a hard time dealing with health problems so I decided I wasn't going to have a big confrontation with him about it. He's got enough with Sharon's health and his own. He's really said nothing to me about it. We just enjoyed visiting with him. We had some good laughs and I didn't want to fill in all the details.

**A Year Away From It**

The second time that the lab ran my test, the result was the same. Maybe I had a little hope that it would be different but I was really expecting it to be the same. You don't have any control over what the results are obviously so you've got to take what you get. I think what I heard was it's bad news with a good flag. I just had to say to myself "Don't think about the bad part of it. Concentrate on the good side. Make some plans if they're necessary but don't focus on the negative part of it."

The other thing is that everything is not known. I know that it's 36 repeats and so I know there isn't some sort of fine line between 36 and a half and 35 and a half, that it is actually 36 repeats. But I also know that I don't believe every bit of research that I hear. So I just say to myself I'm not even ready to believe that it's 36, it could be 35, it could be 37. I'll just think in the most positive way I can.

I made up my mind that I didn't want to have anything to do with Huntington's for a year so I said to Suzanne and Jason, "Let's not talk about it every time we see each other. Every three months we'll just say how are you doing, anything new?" We've got this year thing that we're all doing. I told them that I really appreciated them staying in this neck of the woods for the next year but that they shouldn't put any more of life on hold. If it comes to some other crisis we'll make the changes then.

I also decided to let go of my involvement with the Huntington's Society. This was after I finished helping with preparations for the local HD conference. At this time I felt like an outsider. But this is not the reason I decided to become less involved with the Huntington's Society. I just needed a break from Huntington's.

In a year from now I may go out to the medical library to read the latest research but not right now. I have a lot of other things on the go. We're going to renovate our basement so that we can
have a student live with us. Then we'll have some income from this house. I'm very conscious
of the fact that I like gardening and living in this neighbourhood. I don't want to live in an
apartment or condominium so we have to do some things now so that we can maintain our
house.

We see our kids a lot. Kathy, one of Brian's kids, is expecting twins so we're excited about that.
Years ago I used to think I'll never get to be a grandmother. I can remember crying when I was
about forty walking around the block with Brian when a grandmother was saying good-bye to
her grandchildren, I can remember feeling sorry for myself and saying I'm never going to get to
be a grandmother.

I have this need for perpetuity or something. I know I want to be important in my
grandchildren's lives but I also feel like I need to be important now just in case I'm not around
or I'm not able to in the future. I want to have the fun now. And there are niggling little worries
too. I remember when Jason was a baby and we lived in Ontario we used to go and visit my
Auntie Sylvia. She had Huntington's and I always had to be really careful because I would only
let her hold him when she was sitting down. So with Kathy having these babies I thought to
myself that it's going to be awful if I end up having to be the one that everybody has to be
careful about with their children, if I have to go through all this carefulness again. But that's just
unnecessary worrying at the moment. I always have these fleeting visions that go through my
mind but I don't dwell on them.

All the Layers
I just need a break from it all. My Dad and Sharon are not well. And the time will come when
Karen will need a lot more support from me than she does right now and I'll be willing to do
that. When I went to visit her in the summer, I'd made up my mind that we were going to have a
good heart to heart talk because we hadn't had one when she was here. But, I'm worried about
whether Karen is able to do that anymore. I can't seem to have in-depth conversations with her
in the same way I used to because she's too twitchy or she interrupts all the time or she can't
keep her train of thought. She can't stay involved. She can't go through all the layers.

Karen seems to want me to have Huntington's because then she won't be so lonely. That was
one of the things that I got from her. A couple of times she said things like, "Well, you know
you better plan for it," things that were outright declarations that I will have Huntington's,
almost as if we're in the same shoes. I thought to myself "no way, I'm not going to start thinking
that it's a fait accompli." It's partly because she's lonely, it's a lonely road for her. But she is an
amazingly brave woman. She's still good. She never misses birthday cards and continues to
write newsy letters.

I do think about it, on days when I'm down, days that my allergies bother me. Then I start
thinking "gee I'm right back into this, what am I going to do about it?" I've made up my mind
that I might have to work longer because I don't want to give up the chance for disability
insurance. Brian has to have a hip replacement operation and if I get Huntington's we will need
care. It's a natural process that you might have to put some effort into caring for your parents but
I don't want our kids to have more than their share of the load.

My other fear is that I won't be able to handle living with Brian because he will over-function.
We will need counselling and there would have to be guidelines because I'll have trouble losing
my independence. I will also have a hard time with Brian, who I think will get immensely
frustrated about what the right thing for him to do is. He'll have a hard time, and so will I,
deciding what's normal and what's Huntington's.

One of the good things about Huntington's is that it encourages you not to waste a day. Your life
has to be meaningful. At school, I think it's a benefit to the kids I work with because I have an
intense need to have an honest relationship with them. I have an intense need to have positive
energy in my classroom and I really encourage them. This need to have what I call meaningful
rather than superficial relationships with my students is good and I would say that part of it is
due to my confrontation with Huntington's.
I think Brian's like me. He just made up his mind to look at the positive aspects of it. I need to ask him though if it is something that he's worrying about. To some extent I'm too scared to. I don't want to find out that he's worrying about it since we have enough to worry about in the here and now with his health. Needing to have a hip operation is a real disappointment for him. He's already had three operations. But even at that, you say to yourself thank goodness there is an operation and let's get on with it and get the darn thing over.

The PET scans were all good news and now I've got to deal with something closer to the real thing. I had made up my mind that I wanted the predictive test so it doesn't make me wish that I had gone for another PET scan. I can remember going for a walk before the first PET result and saying to myself, “You might never look at these blossoms the same” and all sorts of nonsense. But after the first result, I remember the feeling of being absolutely free of something that I had had as a burden for a long time. That was amazing for me. And then the second result was good too. I think I had three PET scans in a row. The PET results were great. The genetic result was not so great. It is a kind of task to deal with. It is work, this one.

My gut reaction to the idea of Jason or Suzanne having the predictive test is “don't bother, it's too yucky. You're too young.” Obviously they're going to do what they want to do and maybe it would be good for them to know one way or the other. But I don't hear them talking or worrying about it. If the thing's nagging at you, that's different, then you've got to do something about it but if it's not nagging at you, why bother?

I got the unexpected bad news and it's not that bad but the thought has crossed my mind that Suzanne is 31 now and if my mother got Huntington's when she was 40, Suzanne could too. But I haven't thought about that very much. When I used to be really upset about Huntington's a lot, I would look at Suzanne and Jason at the dinner table and I couldn't bear the thought of them having it. But they probably won't and if they do there'll probably be some kind of treatment or cure. And they certainly don't have to have children that have it.

\textit{Nearly two years later, I spoke with Gabriella again. She was very pleased about seeing her twin grandsons on a regular basis but had found, over the last year, that her life seemed to be dominated by health concerns. She began the interview by talking about the recent loss of her father.}

After I visited my family in Alberta last summer my Dad's health deteriorated a lot, and from last August until now, the situation has dominated my life. I had not anticipated any of the difficulties and it was very hard for me because he chose to die. He was not well physically but he was recovering. There was a discussion of living wills that came up and he just decided he wasn't going to take his medication. I have tried hard to understand but I will never really know why. This was very distressing for me because I've always been brought up to "never say die, only try". It was startling for me to see this man make a decision that was so counter to the way we were brought up. I found myself going back to being a child and I never could have guessed I would do this.

\textbf{The Real Rock in My Life}

My sister was amazing. On the day my Dad made the decision he was going to die, I had a terrible time dealing with it and I went over to Karen's place. She just said come on Gabriella we're going to have a big hug. And, then, we just sat and read a passage in the bible about time and acceptance of when certain things occur. It was a most perfect passage. That's my sister—my sister who's always been the real rock in my life and that rock is still there. I thought Karen you are an amazing person. You have had to live with this darn Huntington's by watching my mother and now you've got to live it over again yourself and you're doing it in the best possible way you ever could. I'll be no good if I do get it. I don't think I'll be able to handle it like Karen although I guess I'll have to.

\textbf{A Teeter-Totter}

To me, my test result put me on a teeter-totter. It's like I'm in the middle and what it means is that there's still a possibility of getting Huntington's if the teeter-totter goes down, but there's
also quite a good possibility of not getting it. I used to say to myself “you’ve always been lucky so you’re going to be lucky here” but then I wasn’t particularly lucky or I was sort of half-lucky. I haven’t worried enough to think about retiring early which is one of the things I would do if I knew I had it.

I have no desire to have another PET scan either because the neurological exam is as good as the PET scan. I can’t be bothered. I didn’t go for the neurological examination last year, I forgot all about it. That is how much it’s slipped to the back of my mind. I even let the annual date go by without thinking about it. I do wonder though if the neurological tests could detect early signs of Huntington’s before I could or before Brian or my kids could. If so, I suppose I should be going regularly so that I could begin taking any experimental medications right away.

I haven’t volunteered at bingo which makes me feel guilty but I know I’ll find it hard to be around people with Huntington’s. I want to be with my sister as much as I can and talk to her every week but I can do other volunteer things. I just know that if I’m around people with Huntington’s I’ll think about it too much. So I’m still at that place.

Brian’s health is so bad that I don’t think we think about me very much and we don’t have any reason to either. We got to the point of saying we’ll talk about it when we have to. We talk about changing our wills because we have to look at whether my pension covers Brian.

The subject of living wills is also on my mind. My aunt who died of HD lived for years being fed through a tube in her stomach and there is no way my sister wants that. How does that ever get dealt with? How can you make a living will very specific on these issues? I have found that the term “no heroic measures” is very loose. One of my good friends lost her father not long ago too so when we go for a walk we often talk families or how to change our wills. Wills are such a weird convention. I have decided that I am going to go through all of my belongings or at least the ones that anyone could possibly want and put a tag on them. This convention of not even telling anyone about who your beneficiaries are before you die is bizarre.

It’s funny but I now understand why people say that you get wiser as you get older. I really didn’t know what it felt like to lose a father until I was there. It’s so odd, so unpredictable. As I said to Jason when I called him from Alberta, “I’m always worried about you and Suzanne and I worry about something happening to Brian but I never ever worried about something happening to my Dad.” And Jason said “that’s true Mum, after all you’re immortal too you know!” It’s odd isn’t it, maybe very naive but I guess you just get so accustomed to always having your parent there for you.
I spoke with Helen two days before she was scheduled to receive her predictive test results. At the time, Helen was 47 years old and living with her common-law husband in the small coastal community where she had grown up. She had just finished a course at the nearby college and was taking the summer off before beginning to look for work. Because she lived at some distance from Vancouver, Helen elected to receive her test results through the rural protocol. Under this protocol, the genetics counsellor at the clinic in Vancouver works with a health care professional in the test candidate’s home community. At least one pre-test counselling session is done at the clinic in Vancouver before blood is drawn and sent to the lab. Subsequent counselling and delivery of test results is provided by the local health care professional. Helen chose her family doctor since, as she explains, she had great confidence in him.

I grew up here on the coast, went through school, married, had kids and divorced. I've been living common-law with Duane for about 12 years. I have three sons, all in their twenties. My eldest is back east at university and the other two are here. After I divorced my first husband I was raising the boys on my own. Duane never tried to be a father to them but he taught them to fish and hunt and he was always a good friend. I was authority, they did what I said.

I have one sister, Norma, who is eighteen months older than me. We were always close. She has one son who is about 4 months younger than my oldest. All four boys did sports together and when she and her husband split up she moved in with us for awhile.

I have worked as a book-keeper for all of my adult life but recently I went back to school. I've been training to do troubleshooting in computers. I love it but there's not much call for it here and for this last while predictive testing has been an excuse not to go back to work.

The Letter

I didn't know there was Huntington's in my family until January the 6th of this year. I got a letter from my cousin Malcolm, in England, telling me that his sister Katherine had been diagnosed with Huntington's. That was the first either my sister or I ever heard about it. I don't think my cousin felt that my sister and I could be at risk because as far as we knew my mother had never had any symptoms and she was sixty-seven when she died.

In the letter my cousin talked about his memories of his mother and her illness and how hard it is now for him to watch Katherine's children going through what he went through. We never knew anything about his mother's illness but he said that Katherine is going through the same stages. I think he assumes that I know what the stages are but I don't.

I knew my aunt Beatrice died of some type of neurological disease but I had no idea it was Huntington's. My mother went to England about 10 years ago and tried to find out about what her sister died from. It was very much a secret that my aunt had even been ill. My mother didn't come back with any information although she made such an effort to find out that I think she must have suspected a lot more than she told us. It's strange to us but I don't think the doctors in England tell people as much as our doctors do. My cousins were shocked when they found out how much I knew about the cancer my mother had.

Bouncing off the Walls

When I got my cousin's letter, I didn’t know anything about Huntington's. I naturally assumed that the doctor's office would be able to tell me something but they couldn't. Nothing. I talked to the receptionist who has been there longer than the doctor has. I've known her since we were kids and she said “All I can give you is this little blurb” and it said that if you are suspect there should be genetic counselling for the kids. I said, “Fine, where do I get more information?” And she said, “the library?”

I went to the library and looked at the medical books and it was not pleasant. When I came out of there the one thing that stuck in my head was the fact that they really didn't know what the normal cause of death was because most people with HD committed suicide. That may or may
not be true but that was the one thing I learned from my little foray into the library. I kind of bounced off the wall for a few days.

I talked to Ellen at the doctor's office again. I said how do I get tested? Can I get tested? And she said we don't know but I'll make an appointment for you right away. Normally it takes three weeks to get in to see my doctor, he's very booked. And I said I don't want to see him, I'm not coming until you get more information. The next day she told me that they knew that there was research being done in Vancouver at UBC. They called and I called and finally I got through to one of the genetics counsellors. She made an appointment for me right away and that was probably the first sanity I had. I was going pretty crazy by this time.

I remember bouncing off the walls. I was absolutely up and down. "I want to know," and "I don't want to know," and "It's happening and it's not happening." Very little was getting in.

I Couldn't Tell Her

When I got my cousin’s letter it was the 6th of January. My sister Norma was to be married at the end of January and I didn't know what to do. I've always been considered the strong one in the family. I've always felt that it's up to me to look after everybody and make sure that they're fine. Knowing that Norma has been through a major depression I thought she would call off her wedding if she knew and I didn't see what good that would do for anyone. I had very little information and I couldn't see any sense in telling her but I felt very strongly that her fiancé Ken had to know. So I went and told Ken that I had this letter and that there was a genetic disease that may or may not be in our branch of the family. I made him promise that he wouldn't tell Norma that I talked to him. I didn't want to go behind her back but I couldn't tell her. He said that he didn't want to know anymore, it didn't make any difference what was in the family, he wanted to go through with the wedding regardless.

I don't know what I would have done if he'd backed out.

About two days after the wedding, the genetics counsellor called me back with an appointment date. Then I had to tell Norma. I thought she should know. She was not particularly impressed that I hadn't told her to begin with and that I'd only given her something like two days notice before the appointment but she went to Vancouver with me. It really wasn't until I went to Vancouver that I really knew anything. A little bit of knowledge made things easier to deal with.

I would have liked to talk to Duane when I first got the letter but I felt that he wasn't as supportive as he should have been. But then we talked about it and he said that he knew my Mum and that she didn't have any symptoms so there was nothing to worry about. "Why get upset with it if there's nothing to worry about?" This may have seemed logical to him but it wasn't to me. He is absolutely 100% certain that things will be fine and there's not much sense discussing it, which is the way he is with all things, it's not just this.

Little Things I Add Up in My Head

The idea that I could have predictive testing was probably my salvation. That was the big thing for me, just to find out. I wanted to take the test long before I even knew there was one. That was my first response, I have to know. I wanted to know for the sake of my sons.

I've had two visits to the clinic in Vancouver. Norma went with me to the first session. It was the first information we had. The genetics counsellor also sent information to my doctor so he'd have an idea what's going on and she confirmed the fact that there was testing. She said think about if you want it done. I said, "Don't have to, this is it, I'm having it. How soon can I be there?" She said that it was several months away. I said, "Several months! I can't wait that long." I would have had the testing done immediately if I could. The biggest shock to me was the fact that everybody didn't automatically get tested, that people could live without knowing.

I'm starting predictive testing at less than 50% risk. The doctor at the clinic in Vancouver made a point of saying that if your parent has not been confirmed as not having HD then your chances are one in four of having the gene. I actually consider it to be much less. My mother was 67 when she died. My cousin is a year younger than me and she's had HD for at least 10 years and

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her mother developed it at the same age. So I figure the fact that I am that much older is a good sign. And because I've never seen or been exposed to it, it's still something that is really foreign to me. I don't know that side of the family and it's like it is something that they have over there that doesn't come over here. These are all the little things that I add up in my head.

The “What-ifs”

I don't usually share what I'm thinking with anybody. I'm very closed. Even five years ago just before my Mum died, I never really shared with her how I felt. It's just something that I've never been able to do. I'm practicing it a little bit more now. I don't know if it's good or not because I feel that I'm stepping on a lot of people's toes. I've had friends tell me that I have changed. I just decided I was old enough to speak my mind now that my mother couldn't slap my fingers for it.

Telling my friends was therapeutic. I have to tell people therefore I am not ashamed of it, therefore I can deal with it better. Being alone fighting something is really tough. It is important to have somebody you can talk to. My immediate reaction was I don't want anybody to know but as soon as I came to terms with the fact that it's no different than any other disease I felt much better about it.

Mentally I'm very strong. Stronger than most of my friends. What I need is the tools to work with and the information. Without the information I can't do anything. I can handle almost anything as long as I know what I'm dealing with.

Most of my friends know that I'm being tested and they all say the same thing, “there's absolutely no way it could be positive, we know that you're okay.” This is nice but what else could they say? Very few people know anything about HD. Nobody really seems to know what it is. I get questions on what it is and why I'm at risk but nothing very detailed. I have 2 girlfriends who I have really discussed it with. One of them had breast cancer a few years ago and I talk to her when I get the “what-ifs”. I know that she gets the what--if's too. I'm very lucky in having a sounding board there when I get these feelings. I call her and she says, "I feel that way too." So I do have people I talk to.

I Have Great Confidence in Him

I am really pleased with my family doctor. I've been with him for fifteen years or more. He was the one who cornered me when I was having such trouble dealing with my mother's death. And he always either started off or finished my appointment with a hug. That made me feel very good. With all the talk now about sexual harassment, it must be very hard for a man, especially a doctor to give a patient a hug. But this is just the way he is. You're family, we care, come in. I really appreciate it and I have great confidence in him.

I found myself feeling very sorry for him with my coming appointment to get my test results. He said, “You know, this is the first time I've ever had to do anything like this. I don't know how it's going to be. You're going to walk in and take one look at me, and if I'm grinning from ear to ear you'll know it's good. If I won't look at you, you'll know it's bad. You won't have to wait, you'll know.” I actually found myself feeling very badly, thinking "why did I do this to him? This is stupid." I really like him and I care that I'm giving him a dirty job. I feel I can say anything to him which makes it easier. I know if I tell him he's a quack and he can't read the results, he will know that it's just nerves. I feel very comfortable with him. For me that is very important.

Renewing Things I Already Know

My results are two days away. I'm not sleeping as well as I would like to. I worry about this testing on and off. I'd like to just say that it doesn't bother me but it does.

The day before my last appointment with my doctor, I cried all morning because I was scared. I have these ups and downs but at the moment I'm feeling really good. I told myself from the start that crying didn't do any good and that I would not allow myself to be overcome by something that was only a maybe. I wouldn't allow myself to cry until the day before my test. And the day before my test I could sit down and have a damn good cry and that way it would be over and

Narrative Accounts/Helen
done with no matter what happened. Whether or not I actually will I don't know. At this point I feel that it's too late. My target date for breaking down is tomorrow but because I've allowed myself to do it, I probably won't.

I don't want anybody with me when I get my results. Before I start my appointment I will have my doctor's receptionist call a cab so that it will be there for me when I come out. I want to get home right away. The house will probably be empty which is what I'd prefer. There's an outside possibility that Duane will be home but even if he is I will probably shut the bedroom door. I will deal with it myself before I talk to anyone. After that I have no idea. I'm not going to go out on a shopping spree to celebrate or anything strange like that. I'll tell Duane. He and my few closest friends will know first. My kids won't know for two weeks because Norma's away and I don't want my kids to tell her son.

If the results are positive I will probably look for work that is similar to what I've always done and I know that I'm comfortable with. The test result might make a difference in whether or not I'm able to get a job. This is a small town but I really feel that if somebody wouldn't hire me because I'm going to get sick in 10 years, I don't want to work for them anyway. My only concern is that my sons might find out before I've had a chance to sit down and tell them. They would not forgive me and I don't blame them.

If the results are positive I will definitely want to know more. I would expect my doctor to get more information because at this point I know more than he does. I know there is a Huntington's book for doctors, I've seen it advertised.

There are a lot of things I want to do so I probably would make other changes in my life too. Nothing radical. I have twice before dealt with the fact that I could die. I had cervical cancer at age 30 and I was in a quite severe car accident. I have dealt with the idea that life doesn't go on forever so this doesn't really make a huge psychological difference. It's renewing things that I already know.

I met with Helen about six months later to talk about the impact of her predictive test results. Under the rural protocol, Helen was able to receive her results from her family doctor. Being in familiar surroundings was very important to her since, as her story demonstrates, even "good news" can unleash a flood of unexpected emotions. Helen began by her story by talking about how calm she felt before she heard the news.

Numb

Before I got the test results, I was quite numb. I was scared but I was in a lot better shape than I thought I would be. Duane wanted to come with me and I told him I was planning to go by myself. I didn't realize at the time that he wanted to be there with me. I felt that I was strong enough to handle it by myself and that he should be working. I think I really hurt him by that, by not bringing him into it. But I went in and got the results by myself.

My doctor was a little nervous about giving me my test results. He had told me that I would be able to read his face, but he was a blank. Other than the initial, "sit down", he told me right off the top, "I got your results. They're good. You're fine." There was no beating around the bush and I'm glad. It would have been really quite painful if he hadn't blurted it out. Then after he told me he went over the fax that he received from the clinic in Vancouver and asked me how I felt. There was a short piece of paper with about four lines on it. I wasn't even curious enough to have a look. I really was insanely calm.

My doctor wanted to know if Duane was waiting for me and I said I sent Duane off to work and that I was okay by myself. I really didn't have any questions. I think he wanted to talk but I had nothing to say. I felt I should be ecstatic but I didn't feel anything. I was no different than I had been walking in.

Furious

I took a taxi home and I lay outside in the hammock for about 2 hours. Then Duane came home with my son Robert. Robert talked to me for a minute but Duane hadn't seen me out there. He
thought I wasn't home so he went out again. I was furious with him, just furious. I went into the house and doubled up and cried. Everything hit me at that time. I had told myself that I wasn't going to get upset but I guess I don't have as much control over my mind as I thought.

When Duane came home I was probably the meanest I've ever been in my entire life. I was really nasty. I accused him of not caring enough to be there to hear the answers. He asked what happened and I said, "If you don't care enough to be here then you don't deserve to know." I wouldn't tell him. I went in the bedroom and I cried and I asked him to pack his bags and leave. I can't believe I actually did that. I was absolutely vile. And he said that he was going to stay for 24 hours and if still I wanted him to leave he would go. It was probably an hour or so later when I finally got my head together enough to come out and tell him the results. Still not feeling guilty, still thinking he's deserving everything I'm doing to him. I can't explain it. I just completely snapped and I went through the rest of that day crying and telling him how horrible he was and that I didn't want him around.

Guilty

By the next day I was beginning to see the light a little bit and realize what I had done. He was very calm through the whole thing and he's never said anything other than that he understands I was under a lot of pressure. I really hurt him but he knew that I wasn't in my right mind. I told my doctor this and he told me that I had to talk it out with Duane. So a month later when we were on holidays I tried to talk to him about it and he said, "I don't want to talk about it." And I said, "I have to. I have to tell you that I really think you were special and that, if you had turned on me I would have understood it." And he said, "I really don't want to talk about it." I said, "okay, as long as you realize how I feel and that I really am sorry and I really figure I'm very lucky to have you here." He said that was fine. Other than that we've never talked about it. I don't think that is particularly good. I've got this terribly guilty feeling but I guess I deserve that.

I knew he hadn't seen me outside. We talked about that and I told him that several months before I had said to him that I was afraid that he would leave me if my results were bad. He had just shrugged his shoulders and turned away without saying anything. In my head he was telling me that he wasn't going to stay. I was afraid he was going to leave me. And when I asked him about it he said that was so ridiculous it wasn't even worth discussing. It was in my head and I know that and I look back and think how silly it was but at the time I was so busy putting everything I was feeling into neat little compartments that I put him into this compartment where he really didn't belong. I feel pretty sheepish about it.

I think my doctor realized I wasn't as together as I thought I was because he called me that night and the next day. He stayed very close making sure that I was going to be all right. I think he expected my reaction more than I did. I was astounded by it. I have great faith in him and I think he handled it very well. It mustn't have been a pleasant experience for him but then doctors have lots of unpleasant experiences they have to deal with.

An Even Keel

Things started to shift after about the third day but it was another two weeks before I was on an even keel. I told each of my sons separately. I waited till they dropped by the house so I could talk to them. They both said thank you for not telling us until you had the results. They took it a lot better than I thought they would. They don't plan to have kids anyway but they were both adamant that unless there was some cure they wouldn't have kids if it had turned out that I had the gene.

When I phoned my eldest son who was back east at university, he blew me right out of the water. He knew what Huntington's was. He knew all about it and I'd never heard of it before my cousin's letter. I figured that my kid has no business knowing things that I don't. But he was the only one that wasn't sure about the predictive testing. His attitude was more that everybody has something wrong with them and whatever happens happens type thing. Talking to my other two sons in person was much easier. I could see that were not having a bad reaction but with Russell being so far away I was afraid that I would upset him and that he would have nobody to talk to. It was much harder. The telephone masks a lot of things.

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I also talked to the girlfriend that I stayed with in Vancouver when I was down there. We go back right to childhood. I found myself resenting what she was saying to me. I don't know why but I felt myself putting up a guard. I felt that she was saying “I know what your feeling and what you’re going through” and my reaction was “you have no idea what I'm feeling, don't tell me you know how I feel.” It’s a childish way of reacting but I found it better to let the subject be forgotten and not brought up rather than cause resentments.

Rift

I told my sister Norma my results within a week. She had decided not to have the test but I told her that she had to let her son Larry know right away. I have a pamphlet that describes Huntington's and has the address and phone number on the back. I told her that I wanted to give it to Larry. I feel that he, like my kids, had the right to know where to go for information. Family is not the best place to go. Norma told me that I had no business talking to him and we haven't spoken properly since. It has caused a real rift. It's not the fact that it could be in the family, its the fact that I feel I have an obligation to her son and she feels I have no business talking to him. Larry and his partner now have a baby and I do think the baby's mother should know there is a possibility. If the kid should ever get sick and they don't know what's wrong, having it hidden from them is not right.

Norma and I have both told each other how we feel and why we feel that way. And other than that there's not much that can be said. I haven't talked to Larry. I want to but I haven't. I keep hoping that one day one of my kids will say something to him and he will have a question and he will come to me and ask. I do understand her not wanting to tell him, and I think that is her decision and she has every right to make it. I don't feel that she is wrong in not telling him, it's in not letting anybody else do it.

I basically say what I feel, probably I've gone a little bit overboard. It's hard to know where to draw the line between being honest and being too honest. I am a lot more open now than I was. I'm not so afraid of being hurt by having somebody come back on me with something I've said. I'm beginning to think that old age has it's definite advantages.

I Have to Remind Myself

As soon as I was over the initial week or two shock, it's almost like I dreamed it, like it didn't happen, it's in the past. It feels more like a bad dream now and I have to remind myself it actually did happen. It's not really something I think about too much anymore. I've got too many other things to worry about.

I have spoken to people that I wasn't close to about the fact that I had waited for test results and people want to know what it is, and why I thought I might be at risk. Maybe it's because my results are negative that I haven't had anybody give me any feeling that I'm different or strange. If it is people's business and somebody else comes up with the same thing I did and somebody says hey I know somebody who might know something, that would be great.

If the results had been positive I might not feel as open about it. I would probably feel that people were looking at me wondering if I'm just having a bad day or if I'm beginning to show symptoms. I think I would be more self conscious but as it is I don't care if the whole town knows. It doesn't bother me and it may help somebody at some time.

I approve of the fact that I had predictive testing. That's a strange way of putting it but I feel that it has given me a bit of confidence that I can face up to things even though they're not pleasant. I can do what I know is right. I think I've always faced up to what had to be done, but this one was particularly hard. Even though I knew I had to do it, it was hard for me to deal with. I realize that now.

Allow Your Family to Help

I can't say I have any complaints about the way things were handled. I wish I could have had my results faster. It was almost 9 months and that was a long time to wait. The initial lack of information was the main frustration though. I also think I would have been in serious trouble if
I hadn’t been able to get my results in my home town. I was feeling so confident that I would have gone to Vancouver by myself. And I would have left the clinic saying I’m just fine, I don’t feel anything. And then when I got somewhere where there was no help, nobody to standby me, I think I would have been in serious trouble. I mean if I had been really vile to a stranger they couldn't have put up with me.

My advice is never to be over confident that you're handling it well. You really don't know. I was sure that I was fine and it was a shock to me to find out that I wasn’t. There’s really no advice I could give to anybody other than to allow your family to help you. I felt that I could handle it and I didn't want to burden Duane but looking back on it I was shutting him out. If I had it to do over again I would let him in on how I was feeling. But it’s too late now, I had tunnel vision. I was looking at how it was going to affect me and not considering that it was affecting everybody around me as well.

Nearly two years later, Helen and Duane had made some big changes in their lives. Duane had always enjoyed hunting trips in the interior of the province and on one such trip he and Helen spotted an appealing log house and acreage. They purchased the property. Duane was fishing for the summer while Helen was living there on her own, gardening, taking a course in first aid and enjoying a very low stress existence far from most amenities.

We were here for 6 weeks before we even bought a clock. It is really good for me. I’m more relaxed. I’m not asocial but I’m quite comfortable out here by myself. If I want to see somebody then I’ll go and see them. I don’t like lots of people around, just family.

For the first time in my life I’ve relaxed enough to let Duane support me. We’ve been together for 15 years and I’ve always worked and paid my way. It was very difficult for me to have him support me. It was a big thing for me to allow that level of trust, to know that he’s not going to turn around and walk out and leave me stranded. After my predictive test results, I realized how little trust I actually put out. I have a lot more trust now.

My sister Norma and her husband Ken came up here for a day about a month ago. We don’t have any arguments but I felt that there was a bit of a barrier there. Huntington’s was the catalyst but it could have been any number of things. I care very deeply about her son and I still find it really difficult that he didn’t get all of the information he should have.

It’s funny, I never thought about how my values related to other people’s values because my friends all thought the same way I did. But with moving up here, I’ve given a lot more thought to the way I feel and why I feel the way I do. I don’t think I would be looking into myself as much if I hadn’t moved up here.

**It Fades Quickly**

I seldom think about Huntington’s other than when it was on the TV the other week. It’s like it never happened. It was the focus of my life. It was the thing my life revolved around and now I don’t think about it. My life is so different, it’s like it happened to a different person. I’m just so satisfied up here. I love my home. I love the air, I love the peace, it’s very easy to just get in my own little cocoon. It would be different if I knew anybody who had Huntington’s.

Duane doesn’t talk about it either but I don’t push him the way I used to because I figure that pushing him probably doesn’t do any good, if he’s not going to talk about it, he’s not going to talk about it. I think it’s easier for women to talk. Duane has difficulty talking about his feelings and I think I’m very open. I will say anything to anyone anytime. Looking back to my parents it was always my mother who handled things like this with the Huntington’s. I think it is considered women’s work. Women do the caregiving. Men are a little bit like ostriches, they don’t like to admit there’s something wrong with them. I think men can ignore things that a woman can’t. She ultimately will be the one who deals with it in the home.

I don’t know if I’m any different than I was other than I’m more relaxed. I don’t think my views on predictive testing have really changed. At the time it was so important and now it’s very far back in my mind. It fades quickly.

**Narrative Accounts/Helen**
REGINA

I talked with Regina about two weeks before she was scheduled to receive her predictive test results. At the time of this interview Regina was 28 years old. Married with an eight year old son, Regina was also working full-time at a nearby building supply store. Regina began her story by talking about some of her memories of growing up and what it meant to lose her father to HD.

I was born in Ontario. I have one brother and one sister who are both older than me. My grandfather died of Huntington's the day after Mom and Dad got married. Things were done very differently back then. They put my grandfather in a psych ward before he died because that's what they did then.

I knew about the family history of Huntington's as soon as I was old enough to understand because my grandma was the type of person that ate slept and breathed HD. Even when we were very young HD was always in the forefront when grandma was around our house. It was everything to that woman. She wasn't a bad person, she just drove me nuts when I came to it. We all knew about HD and there was nothing that we didn't know about it that she was going to tell us.

If we ever had any questions Mom and Dad always answered them. They were always very honest with us. They were like that when it came to anything—drinking, drugs—and they used to tell us the pros and cons of everything. It wasn't a matter of "don't do this or that because it's bad".

He's Still My Father

My Dad was in the military so we moved around a lot. We lived in Germany for four years and England for a year. I've been all across Canada a couple of times. We moved back here to B.C. because it was going to be Dad's retirement posting.

My Dad was put out of the military about 8 years ago and he probably started showing signs of HD about 4 to 5 years before that. At first it was nothing really major. You start seeing little signs—the handwriting goes, you lack coordination in some things. Mom was the first one to notice it but there wasn't a big to do made about it. It's not like we had a big family meeting about it. Whenever us kids noticed, we noticed.

We had a totally different attitude from what I was raised with around my grandma. Things were always done as much as possible in the way Dad wanted. That's the way it worked, within reason. Nobody had the attitude "no Dad you can't do that cause you got Huntington's". It was a matter of finding a way to do it.

At the point where Dad started getting really bad, my sister lived in Saskatchewan and my brother lived in Ontario. I was here in BC. Mom had some legal business that she had to take care of and she just couldn't get Dad's signature anymore so she asked him for power of attorney. Dad said "yes, I will give you power of attorney if you will give me power of attorney." So that's what happened. That's the way a lot of things were done, and Mom never used it other than when she absolutely had to.

Mom was a registered nursing assistant and is now a prison guard. Dad had the best care you could ever imagine. She just would not allow anything other than that. My uncle Conrad (Dad's brother) is in the hospital in Ontario with the same disease. He's been there about 10 years, and he's 10 years younger than my Dad was. My Dad only spent about a year and a half in the hospital though. The hospital Dad was in was nearby and he'd come home on weekends. It wasn't like he was in there full time. We spent a lot of time there and my husband would take Dad to football games and hockey games. He would also do a lot with Geoffrey; Geoffrey was very close to my Dad. They'd go touring around in his motorized wheelchair.

We fought with the nurses at the hospital all of the time. Some of the staff there just didn't care. They had become numb. I had more than one fight with those people. One time when all the
nurses were on strike there was a notice up for the family members asking if we would help with our family member's personal care during the strike. One of the nurses came up and asked "are you going to help us?" "No." She says "Why not? That's your father." I said "that's exactly why. He's 51 years old and you want me helping with his personal care?" I said "what a way to demean a person, let alone embarrassing him. What's it going to do to me? If it's an emergency I'll do it." I said "do your job. It's not a medical requirement, it's not an emergency that I need to do it. I'm not willing to put my Dad in that position." They didn't understand. They thought it was terrible I would say no. They're not thinking. He may be sick but he's still my father and how many 51 year old men do you know of having their kids do their personal care for them? Forget it, won't do it.

I was very close to my Dad, I was probably Daddy's little girl. We were it, him and me. So losing Dad hit me very hard, very hard. Dad was a very special man. I never heard anyone say a negative thing about him. He would do anything for anybody. He would give the shirt off his back.

My Mother

I love my mother because she's my mother but I don't have a close relationship with her at all. She worked shift work, full time the whole time us kids were in our younger years so my Auntie Ruth looked after us for quite a while. She has two boys and a girl and we're all very close in age. We played together when we were growing up. Then my family started moving around a lot more but we kept in touch with her. She's basically like a mother to me, always there. I go and see her as often as I can.

All of us kids moved out of home very early. We were 16, 17, 18, in there. All of us left on bad terms. Since then each of us has, at one point or another, has been on the outs with my mother. She's always got one of us that she's not talking to. As far back as I can remember, the only time we've all been speaking was just before Dad died, during that process. When my Dad was alive he would calm her down. He would say, "Okay, Donna, you've overstepped your bounds, that's enough."

The Reasoning Behind It

My husband Mark is a full time fire fighter but he also works part time as an ambulance attendant. We're all very active. I play ball and Mark usually plays baseball and hockey and he curls and scuba dives. Geoffrey is also into baseball and hockey. I couldn't imagine doing that with 2 children. I don't have any time now, never mind with another one kicking around.

I probably would've had 2 children if it wasn't for the Huntington's. My ultimate was always to have 2 children. I always wanted a little girl. I'm glad that I have a son though because Mark and Geoffrey are very close.

I had my tubes tied when Geoffrey was six months old. That would have made me 20, which is very young. They won't usually do it for you when you're that young. We had decided that we were willing to take the risk with one child and we felt that risk was okay to take but we weren't willing to take it with more than one. That was the reasoning behind it and the doctors okayed it because of that.

Huntington's is not something we talk about a lot. It's there but it's not there. When Dad was quite ill, I did however do one thing that was probably very strange. I told Mark that if he had any second thoughts the opportunity for him to get out of his relationship with me was now. Once I got sick it would be too late. He said no, he was in for the duration. People would say to me "you're so mean for putting him in that position" and I said I have to know in my heart that if I get sick he's there because he wants to be, not because he feels obligated or trapped. That means a lot to me. It was never said to be mean. It was said to make sure that he doesn't feel he was trapped. The last thing I want from anybody is pity.
It’s Time I Made the Decision

I found out that there was a predictive test a couple of years ago. At first I really didn’t care. I wasn’t willing to make that decision in my life. I don’t know why it’s changed, whether it’s because my life was so busy for such a long time that I never had time to think about it.

I know my Mom was upset when my brother Giles had his test done. I told her he deserved a lot of credit because he had made a very hard decision, whereas I was riding the fence. I never decided whether I wanted it done or didn’t. Then all of a sudden it was bugging me all the time and I wasn’t sleeping. Not being a very religious person I thought, somebody somewhere is telling me that it’s time I made the decision in my life. Instead of being in my subconscious it was kind of consciously there all the time. I thought there’s got to be a reason for it.

I’m a very spontaneous person so as soon as I decided I called. Then I was on a waiting list for about 6 months. That seemed pretty long, because I’m very impatient. If I go shopping and I want to buy something and they don’t have it, God help you cause I’ll go somewhere else and buy it. I’m very much impulse on some things. I want it now, don’t wait.

He Said He Was Afraid

I talked to Mark and he was totally against me having the test. We were on different sides about it and it finally came down to “well, it’s my body, my life”. It’s not that I didn’t listen to what he had to say. I did. I weighed the pros and cons.

He said he was afraid. You see Mark was a very big part of my Dad’s life. He was very close to my Dad and he had a very hard time when Dad passed away. He figures the test is like finding out you’re going to die, because Huntington’s is terminal. I do know it is terminal. We just have very different feelings about it that’s all. That’s where it stands.

He said he didn’t want to know. Basically I told him “fine, I won’t tell you. If and when you’re ready to know you can ask me and I’ll tell you.” I’m not such a mean person that I’m going to come home and say “oh by the way it was this or that way.” So I told him if and when he’s ready then I’ll tell him and if not, I won’t tell him. So, that’s how it is.

He’s not going to be here when I get my results since I didn’t get my appointment until after his work commitments as a firefighter were set. It just kind of happened, it wasn’t done on purpose. He did go with me to my counselling appointment and he did voice his concerns. That’s fine, I listened to them but I’ve made my decision and that’s where it stands. Nothing anybody says or does is going to make me change my mind.

How Else Are They Going to Learn?

There wasn’t a lot that people at the clinic told me that I didn’t know already. I knew about all the research that had developed because they found the gene. That was about a month before Dad died. When Dad passed away, the Huntington’s research received whatever tissues they wanted to use for their studies. That seems to help me a lot because I know that Dad was the type of person who always wanted to help if he could. Knowing that he would feel that he went through this and he lost his life for a reason has helped me get through a lot of this.

It’s like when I go to the clinic and they ask if a student in genetic counselling can sit in on my appointment. I don’t mind because it just brings more people in that know about it. How else are they going to learn? If there’s anything I can do that’s going to help somebody I’ll do it.

A lot of people don’t know what Huntington’s is. I don’t think the public is educated. People ask is it like Parkinson’s or Lou Gherig’s? Some people have never heard of it and then other people have heard of it but they still don’t know what it is. Even in the professional field they’ve got to be educated more. The family doctor that I have is the same one my Dad had. This doctor said he’d never dealt with a patient with HD before and that we’d learn together. I would go in there for an appointment and he’d come in and ask “How’s your Dad doing today?” or “do have you anything new? Oh I read this article.” He was very willing to learn. He was fantastic, but even he didn’t know. My Dad was the first person he had ever dealt with that had Huntington’s disease.
Somebody Killed That Trust

My brother Giles doesn't say much. He is the type of person who doesn't butt into your life, unless there's a good reason for it. He encourages me in what ever I want to do but I'm sure deep down he's probably got mixed emotions because of the way his test came about. I'm 99.9% sure that his test has come back positive, but I haven't physically heard that out of his mouth. I've kind of heard it from everybody else, but he doesn't say one way or the other. When Giles had his done it was not a 100% sure thing, there was only 85% accuracy at that time because he had his done before they found the gene, whereas as now it's 100% accurate. I don't know if he'll get it done again.

My brother had kept his test quiet and nobody knew about it. Then all of a sudden Mom got her hands on the information from an old girlfriend of his that he had confided in. She wound up telling Mom and then Mom just decided to announce the information to a whole group of people who were over at her house. That's one of the reasons why she does not know I'm having the test done and she will never know the results. My brother knows but he won't tell her. He said I don't want her to do to you what she did to me. Giles would have told who ever he wanted to tell and he just never had the opportunity to do that. It was done for him which isn't fair. He confided in somebody and somebody killed that trust and it's not a nice thing to do.

Anytime He Asks

Geoffrey has grown up knowing about HD. We've always been firm believers that anytime he asks a question it's answered honestly. If he doesn't ask we don't volunteer the information because we figure when he gets older he can ask then.

Geoffrey doesn't know anything about my test. What am I going to tell him, to make him understand? Right now it hasn't come up so there's no sense bringing it up. He's only 8 years old. And you're not talking about cancer where sometimes you find out and they give you a month to live if you're lucky. We're talking however many years before it starts showing and then it could be 10, 15 or 20 years after that before I lose my life because of it. We're talking about quite a long stretch of time. Anything can happen. I don't want to jump the gun too much.

If my test comes back negative we don't ever have to worry. The only thing we have to tell him is it's done with and you don't have to worry about it. If it comes back positive, then he is going to have questions and they'll be answered at the time he asks. Any of the questions he's asked up to now have been answered honestly. We've never lied to him about anything but we wait and let him do the initiating.

Either Way

Either way the test is going to answer a lot of questions about Geoffrey and for Geoffrey, but also there's little things that Mark and I always decided. One of the things I always wanted to do is go on a cruise. I'd like to go when I'm in my 30's rather than when I'm 50 because I plan on having a hell of a good time. Also, we have always agreed on the fact we can comfortably afford Geoffrey no matter what he wants to do. We can help him through university if he decided he wants to be a doctor or lawyer or whatever it may be. We can do it without going into a great deal of debt because we set our goals for paying our house off.

If I got sick, I would still want Mark to be able to lead the type of life we had decided we were going to lead before I got sick. I don't think that should have to change. I know certain things would change but the long term goals don't have to. So, if before I start getting sick, we can have the house paid off we can have a little bit more money put away for Geoffrey's education then spend a little bit more money on my cruise. Those are the little things, they're not massive things.

Everybody's got their own circumstances, their own reason as to why they want to know what they want to know. I think it would make a very big difference if somebody was making this decision and they didn't have some kind of support around them. Then I don't think it would be a smart move. If I didn't have the type of people around me that I have for support I probably wouldn't have made the decision to have the test. But I do have a lot of support, and that makes

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a big difference. I know that even if Mark ever does decide he wants to know he will still support me.

**Say I Knew I Was Going to Be the Lucky One**

There are other people that know I'm having the test. It's not like I've ever tried to hide it, but I also don't volunteer the information because I don't think it's something people want to hear about. I told my friend Denise when I first called to get the test done. I don't think it ever came up after that, but she knew roughly when my appointments were. I just go on as if life's quite normal.

Results day is no big thing. I have to work till I go for my appointment, then I'll drive into Vancouver and if somebody wants to come with me that's fine. It's kind of hard to get somebody to go with me though as most people work Monday to Friday, and it's a 2 hour drive. I'm fine with it though. I had originally intended on doing it on my own anyway so nothing has changed. It's not like anything's going to change.

Say I knew I was going to be the lucky one. Once I know, I'm either going to let out a huge sigh of relief or I'll just keep telling myself that it hasn't missed a generation in a really long time in our family. As far back as I know it hasn't missed a generation. So I figure I'm due.

Two weeks later Regina went to the predictive testing clinic and received her test result. Her friend Denise went with her although she had not been present at Regina's other pre-test counselling sessions. Regina was, in her own words, never going to be a textbook patient. When Regina learned that she had the gene for Huntington Disease she did not have an emotional reaction. When we talked again, about a year after her test results, Regina explained why this was the case and why she has still not felt any need to dwell on her test results.

Just before going to the clinic I felt good about my test results, believe it or not. Dreams and any thoughts I had associated with getting my test results were all positive. I have no idea why. I'm a fairly positive person but if there's something negative to consider I'll usually find it. Or at least, I used to be like that. So it was really strange that with something like this there were no negative thoughts. I was kind of convinced that maybe it would be good news.

Once they told me my test results I just said okay. Denise and I started joking back and forth and talking about booking our cruise and that was basically it. They kept asking questions and drawing the session out and we were sitting there and sitting there. When we left Denise and I almost said at the same time that it felt like there was something that was supposed to happen that didn't happen. I didn't have an emotional reaction, I didn't break down. And I still haven't.

I thought about that and I thought why? Usually when you have an emotional reaction there's something that's caused the emotion. Like when you stub your toe, you cry because it hurts. Or somebody says something to you that hurts your feelings. It upsets you, but it's usually relevant at the time. Well the news wasn't great but I'm wondering if maybe the reason I didn't have this big emotional reaction I was supposed to have was because it's really something that's not relevant right now. Maybe the time will come when I'll start showing signs and symptoms and that's when I'll feel that it's an appropriate time to have a reaction. Right now I guess I could break down and cry but what am I going to be crying for? Nothing's really happened to me. I'm not sick. I haven't stubbed my toe. Nobody's hurt my feelings, so nothing really happened to make me react that way. I control my life not HD.

They were telling me the future, that's all they were doing. It's not now. Giving birth to Geoffrey, that was emotional! But there was a reason for the emotions. With this, there hasn't been a reason for the emotions yet. Maybe when there is that's when it'll hit me. Crying over something that hasn't happened yet is kind of like putting the cart before the horse.

**I Was Never Going To Be the Textbook Patient**

A couple of weeks after I got my test results I got a phone call from the genetics counsellor. They wanted to know how I was doing and if I was okay. And I'm thinking “what the hell's
going here? Is there something wrong with me? Am I going to be in the shopping mall and all of a sudden be an emotional basket case? What are they looking for?"

Are they really looking realistically at what people are doing and dealing with and how it's affecting their lives? Before your test results you have to fill out a package of questionnaires. I might understand the relevance of the questionnaires if they were personalized but I don't like being forced to choose one or the other answer if it doesn't relate to me. It's like speeding down the road, going through a red light and being pulled over by a police officer and the police officer saying, "Did you do it?" "Yes I did do it but." "No, no there's no but." "Well sorry but my child in the back seat just cut his thumb off and I'm rushing to the hospital." "No, no buts allowed."

I guess I deal with things very differently than most people. You just play the cards that you're dealt and go on. I'm not one to talk about the problems that are going on in my life. There's always somebody else out there that's having worse ones. I guess maybe I have a very callous attitude. I don't know what it's called. I guess I was never going to be the textbook patient. It's not denial, it's called going on with your life and dealing with reality and not allowing Huntington's to run your life. I have my life to think about and I have Geoffrey's life to think about. Nobody says what I feel is correct. Some people agree with me, some people don't.

He Went Through it Once

Mark did come to me and say that he was ready to know my test results. When he asked me I said "what do you think my results were?" He said "well, I just thought it was good news. You're fine and nothing's been bothering you." And I said "well, actually you're wrong." He didn't quite know what to say and basically from that point on he just totally withdrew. He was a totally different person than what I expected.

Mark was very supportive of my Dad when my Dad was sick. He did everything with Dad. And he was right there by his side no matter what he needed, any time of day, moral support, emotional support, everything. He was there. I could never down him for that. You couldn't have asked for a better person. And he just said that he went through it once and he didn't think he could go through it again. That was kind of like the icing on the cake more than the cause of the problems. There was all kinds of stuff thrown in there but that didn't help at the time.

My Life Right Now

I'm pretty happy with my life right now. I'm involved with somebody else and I have Geoffrey. It doesn't look too shabby. My relationship with Mark, who is now my ex–husband, has it's ups and downs. I've made a lot of sacrifices for Geoffrey and put up with a lot for him. But I figure I have broader shoulders than he does and I'll bounce back.

Geoffrey is coping pretty well and that's one thing that has been pretty consistent. No matter how Mark and I feel about each other Geoffrey is still number one. He spends a lot of time with his Dad and that will only change if his Dad wants to change it. I would never do it because they're too close. It's joint custody, joint guardianship with one main residence and undefined access, everything's very open.

I have a new job working in reception for a law firm. It's okay but I will never be a lifelong receptionist. To change careers you have to start somewhere. So it's definitely just a starting point. I also just finished six months of school. I dedicated probably 99% of my time to it and I came out with a 95.8% over all average. I took computer courses and accounting courses. You have to branch off and start somewhere.

Bad has turned into good. I got laid off and I got separated but because I got laid off and I got separated I knew had to go back to school. If those things had not happened I wouldn't have been in a situation where I could do it. So I'm starting to believe more and more that everything happens for a reason. You have to be optimistic somewhere in there.

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If the Opportunity Presents Itself

David is an accountant, a tax manager. The girl that I commute with every day is his receptionist at work and she set us up. She just did her little ole matchmaking. I felt like Cinderella. He would buy me flowers. I had never been wined and dined like that before, ever. We used to fight over the bill all the time when we'd go out for supper. He's old-fashioned and I'm new fashioned. I said either we split the bill every once and a while or I'm not going out with you, take your pick. I don't take things well. I'm used to doing it all on my own.

He's very quiet, the exact opposite of any man I've ever been with. Very quiet, not shy but he's soft-spoken. He's not a real talker. If he's got nothing to say, he's not going to say it. He's not into idle conversation. It's his biggest adjustment to living with Geoffrey. "Does he always talk so much in the morning?"

The biggest hurdle I have had to cope with was telling David about HD and my test results. I guess it was a big hurdle because of the reaction I got from Mark when I told him. David had been really sick and I knew that I was going to have to tell him sooner or later. He was almost better and we were sitting at the kitchen table at his place and we were talking. We hadn't been dating very long, maybe two or three months. I said, "Well now that I've nursed you back to health, are there any other medical conditions that I should know about you?"

David has a degenerative tissue disease and he's got rheumatoid arthritis as well so we're not talking about a really healthy person on the other side of the fence either. Maybe that's why he can relate so well. So when the opportunity presented I told him. He said "okay." And I said "well do you know what it is?" And he said "yes but you can get killed walking out in the street tomorrow by a bus. Cancer can hit you, Alzheimer's can hit you, a gazillion other things can hit you." He said, "this just tells us that if nothing else does, this will." So that's kind of his attitude. Very pragmatic. That's David. "So it doesn't bother you?" "No." I thought that was really weird and a couple of days later I asked, "Are you sure you understood what I told you the other day?" "Yea." He didn't care. And that was basically the end of the conversation.

I don't really remember telling anyone else about my test results. I didn't do the old sit everybody around the kitchen table and go "hey now everybody listen up. I'm going to tell you once, I'm doing the talking, you're doing the listening." It doesn't work like that. If the opportunity presents itself then you do it. If you don't have to or you don't want to, you don't.

The friends that knew I was having predictive testing listened to see if I had anything to say and if I didn't have anything to say they wouldn't pump me for more information. So it's on a how I feel comfortable basis. That's how it works and that's fine with me. People that have to know, know.

I've had to tell my lawyer, out of fairness to her, so that she knew everything that was going on. And also because I have to work there and files were accessible. She could make files inaccessible.

I don't go around advertising it to the world. Some people knew about Dad and they know the disease. They'll ask about my risk and I have no problem telling. I'm still the same person, I just have this bit of information that other people don't have. It doesn't make me any different. That's what I've told myself.

On My Own

I wouldn't tell my mother about my test results until I was ready. She didn't know until at least a couple of months after. I think I was mad at her when I told her. She was shooting my brother down, his decision to get the test and I just said that it was his choice. "You think you know everything. I've lived through this with your father." And I said, "we're all dealing with it from a different point of view. Giles and I are staring it right down the face." She said, "What do you mean by that?" And I said, "What do you think I mean? I got my test results and they're exactly the same as Giles."

My mother hasn't been there for me. I've been through everything on my own with my friends. I am lucky in that I have some really good friends. Not that I needed it but never once have I
gotten a phone call from my mother or my sister asking “How are you doing? We’re there if you need us, if you need anything.” Not once. So I’ve done it all on my own. That’s just fine with me.

My mother thinks I should not be having a relationship right now because I haven’t had time to find out who Regina is. I’m jumping from one relationship to another one. But as I told her, I know who I am more than I ever have before. I’m quite happy with my life and the way it’s going. I’m not out there for everybody’s approval. What I’m doing makes me happy and it makes Geoffrey happy, so that’s what counts. I don’t care what anybody says. I’ve spent too much of my life trying to please my mother and trying to live up to her and my sister’s expectations. It’s never going to happen and I’m just not doing it anymore. This is who I am. I like myself. If that’s a bad attitude that’s fine but it’s me.

I guess you learn to deal with things differently when you have to go to the outside for support. Nobody else is going to tell you you’ve done a good job and pat you on the back. The only person that would have is Dad but he’s not around any more. It still bothers you though because there are people that you want to be proud of you and that will never change no matter what the relationship with my Mom and I is. I’ll never change me wanting her just once in my life to say that she’s proud of me and I’ve done a good job. I know it won’t happen but I’m not going to dwell on it and wait for it. So maybe that’s why my attitude is different when it comes to HD too. It’s part of life, deal with it and go on. I could be a basket case if I wanted to be but if I let all that stuff run my life I wouldn’t be anywhere or anybody. I would be no good to anybody and I’d be a useless mother. I think I’m stubborn now because I’m at the point where I’m so used to doing for myself all the time.

I’ve Gone On With My Life

Going to the clinic causes a lot of uneasiness for me, in my life and in my sleep, and none of that is there other than around the time I go in and just after I’ve been. It brings so much back again that I’ve already dealt with and put behind me, stuff that I don’t think needs to be brought up again right now. I can’t really see the relevance of me going back to the clinic again unless it’s going to help me or it’s going to help Geoffrey.

Maybe I don’t utilize what’s there in the counselling sessions so maybe that’s why I don’t feel like I come out of there with a whole heck of a lot. I don’t want to reach anything deeper. I’m quite happy with where I am right now in my life and that’s what I said to my Mom. She said “maybe they were right. Maybe you are in denial.” I said “well let’s put it this way, if I am I’m having a hell of a good life right now.” On the other hand, I also know that with the support network that I have I’ve got a lot of people around me that I can go to. The doctor and everyone else at the clinic are all very wonderful people if you need them. But I don’t need them. If I did, I’d be the first one to go there. But I have so many other people around me right now that will be there for me.

I guess I look at it from a very different standpoint than a lot of people do. I racked my brain more than once trying to figure out why there’s this difference and I don’t know what it is. Either I’m a little wimp and I’m running away from the world or everybody else is. You know, there’s always a right person and a wrong person and I haven’t quite determined which one I am but I feel the way I’m doing things is right. I’ve gone on with my life, I’ve done all these things since the test results so obviously what I’m doing is not totally wrong.

For some reason I seem to be the abnormal one in this picture and I don’t quite understand why. I go in to the clinic and they want to know what’s happened in my life in the last year and a half. I say “Oh not much.” “Why do you say it’s not much? You got laid off and you got separated and you have a new relationship.” Yea I do, but it’s my life, it’s no big thing. Do they want me to say all of this is caused by HD? It’s not. It could have happened if there wasn’t HD in the family. I’m just too damn stubborn to allow HD to alter my life and change who I am. Maybe that will be to my disadvantage down the road but a round peg just doesn’t fit into a square hole.

I still don’t really know why I wanted the test. I haven’t altered the things that I do in my life. The only thing I might change is I don’t worry about putting money into an RRSP anymore. It’s
kind of useless because I sit back and I see the amount of money my Dad had put away and he never got to spend a penny of it.

I'm optimistic though that they are going to find a treatment. And then the information from my test might come in handy. They might find a drug they think can prevent onset of HD. If they didn't know who was and wasn't going to get it, they wouldn't know who to give it to. So there's an advantage. If that test wasn't there they would only have a 50:50 chance of finding people that fit into the right categories.

Knowing what I know, I just sit back and think about it realistically. Is it altering my life now? I say no. Is there anything I can't do? No. Well then why walk on egg shells? Why not live your life the same way you were living it before? Nothing's changed. Nothing physically has changed at all. You have one little bit of information that not everybody has the opportunity to get and that's it. That's the only thing you've got that's different. So why should it change your life? I don't think it should. I don't know if that's right but it makes sense to me.