INTERPRETING AND MAKING SENSE OF UNINFORMATIVE RESULTS OF TESTING FOR BRCA1 AND BRCA2 CANCER GENE MUTATIONS

by

Christine Maheu

R.N., John Abbott College, 232, 1987
B.S.N., L'université de Montréal, 1994
M.S.N., L'université de Montréal, 1997

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ABSTRACT

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Research suggests that a significant proportion of individuals from families at risk of hereditary breast and ovarian cancer will be found not to have a detectable mutation in their BRCA1 or BRCA2 cancer genes. Although the interpretation of genetic test results is relatively straightforward in families where a mutation has already been identified, little is known about how people who have had breast and/or ovarian cancer in the past as well as a family history of cancer considered at risk for HBOC interpret and make sense of test results concluding that no detectable mutation has been found. This problem is further compounded when they are told that such genetic test results do not completely rule out an inherited mutation because of their strong family history of the disease. While the clinical and research literature refers to these results as uninformative or inconclusive, this study shows that clients' interpretations are much more complex. To date, few studies have focused on affected individuals from families at risk of HBOC who receive uninformative genetic test results. We therefore have little knowledge of how these individuals interpret and make sense of such results and how these results affect their everyday lives, health and illness experiences.

This dissertation addresses these lacunae by using an interpretive description approach to examine clients' experiences of genetic testing. Qualitative, in-depth interviews were conducted with 21 affected individuals with a family history of cancer considered at risk for HBOC who received genetic testing and 15 family members. The interview data show that living with a personal and family history of breast and/or ovarian cancer plays an important role in interpreting and then making sense of their genetic test results and in one's perception of
probably having an inherited mutation for HBOC. Thirteen generic structures were found to organise beliefs towards the making sense process of interpreting their genetic test results while three types of interpretation of the test results were derived from the participants’ accounts. The categories of interpretation are seeing results as: a confirmation of their mutation status, ambiguity regarding their mutation status, and refutation of being a mutation carrier. On the basis of these generic structures and the three types of interpretation, it is possible to see a retrospective narrative of causal reasoning of having a probable inherited mutation that builds upon recognition of a strong family history with breast and/or ovarian cancer. This 7-stage process evolves with changes in people’s everyday lives, health and illness experiences.

The impact of receiving uninformative test results for BRCA1 and BRCA2 mutations on the lives of affected individuals and their family members requires further examination. We need to understand how such results affect cancer and genetic risk perception and potentially contribute to clients’ viewing themselves at chronic risk of cancer and of an inherited mutation. Further investigation is also needed to determine how uncertain genetic risk information is shared among and used by family members. This dissertation offers recommendations to ameliorate the experience of individuals who receive uninformative genetic test results, to improve genetic testing services, and to enhance the genetic knowledge of nurses and their clients.
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CHAPTER ONE:

Situating the Study

Prior to the advent of genetic testing for hereditary breast and ovarian cancer, the combination of breast self-examination, clinical examination and mammography was the only strategy recommended to increase early detection of breast cancer (Delisle, 1997; MSSS, 1998). For families who have a strong history of breast and/or ovarian cancer, genetic testing offers an additional way of assessing the risk of developing these diseases if family members carry inherited mutations of known breast or ovarian cancer genes. Having a personal and family history of breast and/or ovarian cancer increases the risk of carrying an inherited mutation (FitzGerald et al., 1996; Newman et al., 1998; Oddoux et al., 1996).

With the discovery of the BRCA1 and BRCA2 breast and ovarian cancer genes (Miki et al., 1994; Wooster et al., 1994), genetic testing for common adult-onset disorders such as breast and ovarian cancer emerged within clinical settings, either as part of research protocols or clinical service. Clinicians, researchers and patients all had high hopes about the efficacy of genetic testing in finding the etiology of breast and ovarian cancers and in identifying individuals at increased risk of developing these diseases as a result of having inherited mutations. Genetic testing for hereditary breast and ovarian cancer is not yet widely available nor offered in all provinces as it is not yet official, integrated clinical practice because of, among other reasons, patent issues. Nonetheless, there has still been considerable uptake of genetic testing for breast and ovarian cancer genes in Canada.

For clinicians, one of the valuable aspects of genetic testing for BRCA1 and BRCA2 is the test’s ability to identify individuals at risk of hereditary breast and/or ovarian cancer (HBOC) (Lynch et al., 1999). For individuals, genetic testing for BRCA1 and BRCA2 informs them of
their actual hereditary risk for breast and/or ovarian cancer and, in part, of their cancers' etiology for those with a past diagnosis because cancer is a multifactorial disease (Hallowell et al., 2002; Lerman, Seay, Balshem, & Audrain, 1995). This test also clarifies the risk of their children and families for hereditary breast and/or ovarian cancer (Armstrong et al., 2000; Lynch et al., 1999).

Despite the benefits (such as knowing with certain one's genetic risk status) and risks (such as increased anxiety as a result of this certainty) there are many unresolved limitations that clinicians as well as the general population need to understand. For example, how do you support clinical decision making with the uncertain information resulting from genetic testing (Frost, Venne, Cunningham, & Gerritsen-McKane, 2004)? Uncertain information resulting from genetic testing for BRCA1 and BRCA2 is mainly of two kinds. First, a positive genetic testing result is not a cancer diagnosis but a prediction of risk of developing cancer. Even if individuals are found to carry an inherited BRCA mutation, while they have a high risk of developing breast and/or ovarian cancer, there are no certainties that they will develop the disease and, if they do, no estimate can be given as to when it could appear. With a BRCA1 or BRCA2 cancer mutation, women have a 56% to 87% lifetime risk of breast cancer and a 16% to 40% lifetime risk of ovarian cancer (Di Prospero et al., 2001; Ford, Easton, Bishop, Narod, & Goldgar, 1994). Men found to carry a BRCA1 mutation have a 16% lifetime risk of developing prostate cancer while with a BRCA2 mutation carries a 6% risk of developing breast cancer (Di Prospero et al., 2001). Children of individuals carrying an inherited mutation have a 50% chance of inheriting the mutation. Second, when individuals considered at risk for HBOC are not found to have an inherited mutation, health professionals cannot confirm their risk of cancer or genetic status. The negative test results could either be a true negative, a false negative or – because researchers have not yet identified all conferring inherited mutations of breast and ovarian cancer genes – it
is possible that these individuals may carry mutations in genes other than BRCA1 or BRCA2 (Pasacreta, Jacobs, & Cataldo, 2002). Considering that only 5% to 10% of all breast cancer cases are hereditary (Claus, Risch, & Thompson, 1991; McCance & Jorde, 1998) and that there is growing interest in genetic testing for BRCA1 and BRCA2, especially among individuals with a family history of cancer considered at risk for HBOC (Peshkin, DeMarco, Brogan, Lerman, & Isaacs, 2001), we can expect to see a growing pool of individuals who are likely to receive genetic test results of uncertain value such as uninformative genetic testing results.

**Background to the Problem**

Before the discovery of the BRCA1 and BRCA2 genes, individuals with a family history indicative of HBOC seeking information about their risk of developing these diseases and that of their family members could only rely on linkage analysis and on models that determine carrier probability for the genes, BRCA1 and BRCA2 such as the BRCAPRO model (Parmigiani, Berry, & Auguilar, 1998).¹ Both linkage analysis and models yielded only a probability that one might carry an inherited mutation. Peshkin et al. (2001) note that, even back then, clinicians and scientists struggled with whether to give individuals the results of these analyses, in light of their uncertainty.

Now that genetic testing for adult-onset-hereditary-disease has become an important part of clinical genetics practice, we see an increased demand for tests, perhaps as a result of media coverage of genetic discoveries or from referrals by health professionals. In 1996, the BC Cancer Agency and the BC Provincial Medical Genetics Program formed the Hereditary Cancer Program (HCP) to provide genetic education, counselling and testing for BRCA1 and BRCA2 to

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¹ BRCAPRO is a model and software developed by G. Parmigiani, D.A. Berry and O. Aguilar at the Institute of Statistics and Decision Sciences, Duke University, U.S.A. The model uses a Mendelian approach of autosomal-dominant inheritance to assess the probability of an individual to carry a BRCA1 and BRCA2 mutation. The model is based on the individual’s family history of breast and ovarian cancer (BayesMendelLab, 2004).
high-risk individuals and families with a strong history of cancer (Williams-Jones, 2002). Although there has been an increase in the uptake of genetic testing for BRCA1 and BRCA2 and we now have an abundance of research since 1994 on the impact of the availability of this test, many unknowns and complexities still remain (Peshkin et al., 2001). The process leading up to genetic testing, receiving the results, and interpreting the results is not simple. The complexities include how to delineate eligibility criteria for testing, how to minimize negative psychosocial reactions, how to support health-care decision making in light of genetic risk values, how to interpret risk values, and how to interpret risk-descriptive statements when risk values are impossible to provide. Even with positive test results indicating the presence of a BRCA1 or BRCA2 mutation, individuals do not receive one precise quantitative cancer risk value but are given a range of the risk to develop cancer. For example, they are told that their risk of cancer will increase between percentage ranges; 56% and 87% for breast cancer (Ford et al., 1994; Struewing et al., 1997) and 22% and 44% for ovarian cancer both by age 70 (Ford et al., 1994; Whittemore, Gong, & Itnyre, 1997). Risk percentages of developing cancer also differ depending on which gene the mutation is found. In addition, these percentages are different for individuals who have already been diagnosed with breast and/or ovarian cancer where they see their risk of developing another primary cancer increased.\(^2\) Research in this area is ongoing. As for individuals within a confirmed hereditary breast and/or ovarian cancer family who receive negative test results for BRCA1 and BRCA2, the test results for these individuals are classified as true-negative. While they see their risk of cancer decrease, their risk is still not at general population's level because of their strong family history with cancer.

\(^2\) A primary cancer is the place where the cancer started growing. Recurrent cancer or metastases are escaped cancer cells from the primary site that spread to other parts of the body.
Research has found that a high percentage of high-risk families (between 16% and 65%) do not carry detectable mutations of BRCA1 and BRCA2 (Peshkin et al., 2001). When not found with a detectable mutation, these families are viewed as part of the “uninformative group”. Hence, the availability of genetic testing for these mutations has created a large pool of individuals in a quandary about their perceived and actual risk for breast and ovarian cancer (Iglehart et al., 1998). We are just beginning to see the results of research on this population and the complexities that may result from uninformative genetic test results.

Even among those who receive positive test results, many psychosocial consequences are not well understood (Pasacreta et al., 2002). Some known responses to positive BRCA mutation test results are persistent worry, depression, anxiety, confusion, and sleep disturbance (Lynch et al., 1993). While the literature documents known responses to positive and true-negative results, such as survivor’s guilt among individuals who test negative within hereditary cancer families (Lerman, Seay et al., 1995), there are still many unknowns. Providing adequate support to those who receive uninformative genetic testing results is even more challenging because of the paucity of studies. While health professionals in cancer genetics have gained knowledge in clinical practice, this knowledge is based mostly on the assumptions, beliefs, and observations of clinicians.

**Purpose of the Study**

Few studies were found that focus on individuals and families considered at risk for HBOC who receive genetic testing results other than positive or true-negative. We are just beginning to see some studies that have documented experiential knowledge of individuals’ reaction to and understanding of uninformative results of genetic testing for BRCA1 and

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3 According to K. Panabacker, genetic counsellor at the HCP, of the 255 individuals tested for BRCA1 and BRCA2 from high-risk families, less than 25% received a positive result.
BRCA2. Within the literature, where individuals with a strong personal and family history of breast and/or ovarian cancer have received a genetic testing result that indicates that no detectable mutations were found, their results are determined to be either uninformative or inconclusive (Frost et al., 2004; Hallowell et al., 2002; Hallowell, Foster, Eeles, Ardern-Jones, & Watson, 2004; Peshkin et al., 2001; Schwartz et al., 2002). At the time I wrote my dissertation proposal (the beginning of 2000), very few researchers had studied the experience of women with a past cancer diagnosis who received uninformative genetic testing results for BRCA1 and BRCA2. In light of the lack of knowledge to how individuals interpret and make sense of uninformative genetic testing results for BRCA1 and BRCA2, questions for clinicians become: How do we prepare individuals who rely on us for information when there is little experiential knowledge of how individuals with a past breast and/or ovarian cancer diagnosis interpret and make sense of uninformative genetic testing results? How can we assist individuals as they attempt to make sense of the uncertainty of their results? Inevitably, a critique has been that the arrival of genetic testing in clinics may have preceded the collection of sufficient data to support their use and the clinical knowledge required to interpret all types of results (Bell, 1998; Biesecker & Marteau, 1999; Holtzman & Watson, 1997). This study aims to address this gap by contributing to the experiential knowledge of interpreting and making sense of uninformative, uncertain genetic testing results for BRCA1 and BRCA2 among affected women considered at risk for HBOC who have gone through both genetic testing and the making-sense experience. The study overall purpose is to gain empirical knowledge about individuals’ understanding of uninformative BRCA1 and BRCA2 test results but more specifically, to explore, from these individuals’ perspectives, how they interpreted such results and how they made sense of them in light of the uncertainty and of their personal and family experiences with cancer. The study also
aimed to explore how uninformative genetic testing results affected individuals’ everyday lives, health and illness experiences. To best serve this research purpose, a qualitative interpretive description was used (Thorne, Reimer Kirkham, & O'Flynn-Magee, 2004; Thorne, Reimer Kirkham, & MacDonald-Emes, 1997).

The Research Questions

The research questions for this study are

1. How do affected women considered at risk for HBOC interpret and make sense of their uninformative genetic testing results?

2. How do these results affect their everyday lives, health and illness experiences?

The research questions are both descriptive and interpretive. That is, the first part of the first research question focuses on how affected women interpret their genetic testing results. The second part focuses on how they arrive at their interpretation based on how they made sense of them. The second research question explores how the women’s interpretation of their results and making-sense experience affected their everyday lives, health and illness experiences.

Because I assume that individuals’ perspectives differ from the views held by health professionals, I wanted to first explore the women’s perceptions. Hence, during the interview process, I did not label genetic testing results as uninformative or inconclusive, just as “your genetic testing results.” My goal in using an interpretive description was to produce a rich description of the constructed and contextual nature of individual experience while at the same time allowing for shared realities (Thorne et al., 1997).

Significance of the Research

As previously said, most studies on the experience of receiving genetic testing results for BRCA1 and BRCA2 focused on individuals who were found to carry a mutation. Furthermore,
the studies that have been done are mostly based in the United States and thus, because of the
different health care system, bring up issues that are not always relevant in the Canadian context,
such as accessibility of testing because of its cost and health and life discrimination risk. One
Canadian study, conducted by d’Agincourt-Canning (2003), produced experiential knowledge
about some of the complex social and moral issues raised by positive genetic test results for
hereditary breast and ovarian cancer.

Currently, people who receive genetic test results are assumed to interpret their cancer
and genetic risk in the same way that health professionals working in cancer genetics do
(Bottorff, Ratner, Johnson, Lovato, & Joab, 1998). However, according to Bottorff, Ratner,
Johnson, Lovato and Joab (1998), people tend to conceptualise risk using a constellation of
descriptive meanings, while health professionals tend to conceptualise risk as a statistical
measure. Further, Richards and Ponder (1996) explain that conflicts between aspects of scientific
explanations and lay knowledge of inheritance may impede assimilation of the former. Because
postcounselling after testing is not always available to people receiving uninformative genetic
test results, there is little opportunity to assess how they interpret and make sense of their results.
Yet, the need for this knowledge is urgent, given the growing number of companies, such as
Myriad Genetics, who, despite providing supportive materials, rely on family physicians rather
than certified clinical geneticist and genetic counsellors to provide counselling. The increased
availability and easier access to genetic testing will create a huge pool of individuals and families
who receive uninformative genetic test results for BRCA1 and BRCA2. Because nurses (and
more specifically genetic nurses) will most likely be the first point of contact for those seeking
further information and support before and after genetic testing, nurses need to prepare
themselves. This thesis aims to produce knowledge to facilitate this preparation.
Definitions of Terms

The following are key terms in this study.

Affected women: the word ‘affected’ refers to having previously received a breast and/or ovarian cancer diagnosis.

Primary and secondary participants: The primary participants are the affected women who have received genetic testing. Secondary participants represent family members chosen by the primary participants and are addressed as “family members” in this study.

Individuals with a family history of risk for hereditary breast and ovarian cancer (HBOC): Individuals assessed to be at increased risk of carrying an inherited mutation predisposing them to breast and/or ovarian cancer, based on having two or more of the following:

1. Cancer in several closely related people, on the same side of the family, in several generations.
2. Cancer at younger than usual ages (e.g., breast cancer in the 30s).
3. More than one diagnosis of cancer in the same person.
4. Specific types of cancer that are linked to specific genes: e.g., breast cancer and ovarian cancer (BCCA, 1999).

Having two or more of above factors in one family results in an estimated 20% probability of detecting a gene mutation (d’Agincourt-Canning, 2003).

Hereditary breast and/or ovarian cancer families: Families in which a mutation has been identified within the BRCA1 and/or BRCA2 genes.

BRCA1 and BRCA2: BRCA1 and BRCA2 are the two most common breast and ovarian cancer genes tested for in genetic testing for an inherited susceptibility to the diseases. ‘BR’
stands for ‘breast’ while ‘CA’ stands for ‘cancer’, while ‘1’ and ‘2’ represent the order in which the genes were discovered. Mutations on either BRCA1 or BRCA2 cancer genes predispose individuals to breast and ovarian cancer. In some families, there is a cluster of only breast cancers, in other families only ovarian cancer and, in still others, both. Although most participants in this study mainly have a family history of breast cancer, I chose to address them all as having a history of both cancers to reduce the risk of inadvertent identification of participants.

**Uninformative genetic test results for BRCA1 and BRCA2:** Within the current study, this term applies only to affected women considered at risk for HBOC who received genetic test results for BRCA1 and BRCA2 indicating that no mutations were identified in the two cancer genes. However, because of their personal and family history of the disease, it is impossible to confirm that they do not carry an inherited mutation or that their cancer diagnosis may have occurred by chance.

**Lived experiences:** Refers to how individuals describe and give meaning to what they have experienced as a result of having come into contact with a clinical context.

**Organisation of the Thesis**

This thesis is organised into eight chapters. Following the introduction of the study in chapter 1, I proceed to a literature review in chapter 2. The literature review focuses on the process of genetic testing and on studies of the implications of breast and ovarian cancer genetic testing. Chapter 3 details my methodology as well as decisions I made throughout the study. Chapters 4 to 6 present the research findings. In chapter 4, I begin this chapter by describing the implications of living with a family history considered at risk for HBOC presenting the women’s interest in genetic testing and their reasons for accepting it. In chapter 5, I focus on the women’s
interpretation and making-sense experiences of their test results for BRCA1 and BRCA2. Chapter 6 discusses what participants concluded from their genetic testing experience. In chapter 7, I discuss how key themes within the study findings can advance the practical science of nursing and inform other health professionals working in the area of cancer genetics. Chapter 8 begins with practical applications of key findings and their clinical applications and concludes with recommendations for clinical practice, research and policy development.
CHAPTER TWO:

Review of the Literature

According to the Canadian cancer statistics (Canadian Cancer Society, National Cancer Institute of Canada, Statistics Canada, & Health Canada, 2004), breast cancer is still the most frequent cancer diagnosis among women and the second most frequent cause of death for women, following lung cancer. However, the prevalence of breast cancer diagnoses is more than twice that of lung cancer. The Canadian Cancer Society et al. (2004) estimate 21,400 new cases of breast cancer in Canada for 2004 and 5,300 deaths from the disease.

Among all breast cancer diagnoses, an estimated 5 to 10% are said to be hereditary. Of these, the predominant cancer genes responsible are BRCA1 and BRCA2 (Lynch et al., 1999). An inherited mutation in either cancer gene increases the risk of developing both breast and ovarian cancer, but the breast cancer is more prevalent. Lynch et al. argue that initial lifetime cancer risk for carriers of BRCA1 and BRCA2 must be interpreted with caution because these estimates are based on cancer prevalence in large families and families at high risk of hereditary breast and/or ovarian cancer due to cancer diagnoses at young ages. Nonetheless, an individual found to carry an inherited mutation of either gene has a 50 to 85% lifetime risk of developing breast cancer (Lynch et al., 1999), compared with an 11% risk in the general population (Canadian Cancer Society et al., 2004). The lifetime risk of developing ovarian cancer is slightly more with a BRCA1 mutation than a BRCA2 mutation: 20 to 60% with a BRCA1 mutation, compared with 10 to 20% with a BRCA2 mutation (Lynch et al., 1999). Canadian Cancer Statistics (2004) estimates the lifetime risk of developing ovarian cancer among women in the general population to be 1.5%.
Before the discovery of these two cancer genes (BRCA1 in 1994 and BRCA2 in 1995) (Lynch et al., 1999; Wooster et al., 1994), estimating an individual and family risk for breast cancer for those with a family history was based solely on mathematical models such as the Claus model (Claus, 2001)\(^4\) and the BRCAPRO (Berry et al., 2002), although the BRCAPRO model comes closer to reflecting genetic risk.\(^5\) Individuals and families with a strong family history of breast and/or ovarian cancer demonstrated high interest in inherited susceptibility testing (Lerman, Seay et al., 1995). The breakthrough in cancer genetics with the identification of the BRCA1 and the BRCA2 genes allowed genetic testing to enter clinical settings within research protocols. Since that time, we have seen a steady buildup of studies focusing on potential and actual impacts of genetic testing on people’s lives.

This first section of this chapter aims to provide an overview of hereditary breast and ovarian cancer. The second section focuses on the process of genetic counselling and the types of genetic testing that can be addressed in counselling, as well as genetic counselling issues and ethical concerns with genetic testing. The third section presents literature on the interest in genetic testing among women with a family history to breast and/or ovarian cancer and the intended use of test information, while the last section reviews literature about currently known psychosocial issues in genetic testing for BRCA1 and BRCA2.

**Hereditary Breast and Ovarian Cancer**

Genetic susceptibility is an important factor in the development of breast and ovarian cancer, as shown with the discovery of BRCA1 and BRCA2. Other known inherited gene mutations that result in an increase risk to develop breast cancer but are not as penetrant as the

\(^4\) The Claus model was developed by Claus et al. and mainly based their estimate on family risk factors such as age at diagnosis for affected family member (Claus et al., 1991).

\(^5\) Comparisons of the sensitivity and specificity of the BRCAPRO model to genetic sequencing for BRCA1 and BRCA2 mutations demonstrated that BRCAPRO sensitivity in picking up mutations was at least 85% (Berry et al., 2002) and its specificity was 68.2% (Phillips, 2002).
BRCA1 and BRCA2 cancer genes are: the ataxia-telangiectasia mutated (ATM) gene, the 1100delC CHEK2, the phosphatase and tensin (PTEN) tumor suppressor gene, the p53 tumor suppressor gene, the androgen receptor gene (AR), the hereditary nonpolyposis colorectal cancer (HNPCC) gene and the estrogen receptor gene (Bennett, Gattas, & Teh, 1999). Although most breast cancer cases are not hereditary, 5 to 10% of all breast cancers are thought to involve one of the above genes (Claus et al., 1991), although 70% to 90% of all hereditary breast cancers harbour a BRCA1 or BRCA2 mutation (Bennett et al., 1999; Lynch et al., 1999).

A family history of breast and ovarian cancer is recognised as one of the most important risk factors for the diseases (Yang & Lippman, 1999). Other nonhereditary factors documented to increase risk for breast cancer include having a personal history of breast cancer, early menarche, late menopause, lack of breastfeeding (a protective factor), use of birth control pill, use of estrogen replacement therapy, childbirth delayed after age 30, and lack of full-term pregnancies (Kelly et al., 2004). Other clinicopathological features of individuals and families suggestive of hereditary breast and ovarian cancer include early disease onset, high incidence of bilateral breast cancer, and high incidence of multiple primary carcinomas in other organs (Anderson, 1992; Nomizu et al., 1997; Ormiston, 1996). A family history of cancer with the following characteristics indicates a probable inherited susceptibility: a woman with two or more first-degree relatives (mother, sister or daughter) with breast and/or ovarian cancer, and breast cancer occurring in a manner consistent with Mendelian transmission of autosomal dominant inheritance (Ormiston, 1996). This means that breast and/or ovarian cancer appears on the same side of the family, among same-blood family relatives.

When individuals within families are identified as having an inherited mutation such as in their BRCA1 and BRCA2 genes, their children have a 50% chance of inheriting the mutation.
However, unlike single-gene syndromes, such as Huntington's disease that follow the laws of simple Mendelian genetics, individuals with an inherited mutation of either the BRCA1 or BRCA2 gene will not necessarily develop the disease but, rather, will be at increased risk of developing it. Ormiston (1996) explains that, unlike single-gene syndromes, for hereditary breast cancer to develop, a person must have inherited one mutated gene and develop a second, somatic mutation in the same region of the gene with the inherited mutation. In sporadic cancers, both somatic mutations are acquired after fertilisation, in the same region of the gene and the mutations occur at a much later time in life. Both scenarios of mutated genes leading to cancer are known as the 'two-hit theory' of carcinogenesis (Ormiston, 1996). Cancer genes, when not mutated, actually protect against cancer by repairing DNA and suppressing tumours.

It is now possible, through genetic testing, to determine if one is carrying a mutated gene. Carrier, presymptomatic, and predisposition testing are the three general genetic tests now offered. Carrier testing indicates whether a healthy individual carries one copy of a gene mutation and involves testing for a gene protein product (Ormiston, 1996). This test has implications for the decision to procreate, in that of those identified as gene mutation carriers who have children with a partner who is also a carrier, each of their children faces a 25% chance of inheriting each of their parents mutated gene and, if they do so, becoming affected with the disease. As with recessive inherited conditions, two copies of the inherited mutation are necessary to develop the disease. If only one copy is inherited from one of the parents, the person will be known as a carrier but will not develop the disease. Examples of common recessively inherited conditions include cystic fibrosis, Tay-Sachs disease, and sickle-cell anemia.

In presymptomatic testing, healthy individuals learn whether or not they will eventually develop a genetic condition. The most common presymptomatic testing is for Huntington

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6 Three modes of Mendelian inheritance are possible: autosomal recessive, autosomal dominant and sex linked.
Disease. Only one mutated gene of a pair of genes is needed to determine whether the individual will be affected by the illness. This type of inheritance is autosomal dominant (Ormiston, 1996).

With susceptibility or predisposition testing, individuals learn if they are at increased risk of developing a condition but do not know with certainty whether they will develop the disease. The most recent common types of predisposition testing are for breast and ovarian cancer susceptibility such as genetic testing for BRCA1 and BRCA2 (Ormiston, 1996). The Hereditary Cancer Program (HCP), where the study participants were recruited, reported three categories of results for predisposing testing for breast and ovarian cancer susceptibility for individuals who met the HCP eligibility criteria for genetic risk assessment: (a) positive – mutation was found in one or both cancer genes tested; (b) negative – known familial inherited mutations were not found; or (c) uninformative – either a change in the sequence of their amino acids has been found but with no known documented clinical relevance yet reported or no mutation was found but because of their strong individual and family history of the disease, the absence of an inherited mutation cannot be confirmed.

**Challenges with Genetic Testing and Counselling**

The provision of genetic testing for BRCA1 and BRCA2 has not escaped the political turmoil. In Canada, most clinics provide genetic testing for BRCA1 and BRCA2 within research protocols only because of limitation use imposed by Myriad Genetics’ patented BRACAnalysis test. Among the Canadian provinces offering testing are British Columbia, Ontario, Alberta and

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7 Eligibility criteria used within the HCP can be viewed in table 3. These criteria tend to vary among provinces, but testing is usually limited to individuals and families with a family history of risk for HBOC. The HCP genetic testing protocol is based on a 20% priori probability of finding a mutation. According to a prior study that recruited its study participants in the same cancer agency as the current study, the 20% cut-off point represents the likelihood of finding a mutation in individuals already diagnosed with cancer compared with finding a mutation in an unaffected individual (less than 10%). However, eligibility criteria are subject to change with new findings indicating specific variations in individual and family histories of the disease that either increase or decrease the likelihood of carrying an inherited mutation, as well as newly identified mutations. In 2000, the HCP called for an ad hoc committee to review the eligibility criteria used by other centres conducting genetic testing for BRCA1 and BRCA2.
Quebec (Eggertson, 2002; Williams-Jones, 2002). The other provinces mostly provide
counselling services but send their samples for analysis to laboratories in Canada; mostly in
Ontario (Williams-Jones, 2002). While in 2000 and 2001, the U.S. Patent Office has awarded a
series of patents to Myriad Genetics that suspended testing in British Columbia in July 2001 (but
still continued to offer genetic counselling), other Canadian provinces such as Ontario, Alberta
and Quebec continued to offer testing. The Ontario government is currently challenging the
patent. Myriad is demanding that all BRCA1 and BRCA2 testing be conducted in their
laboratories for a cost of about $2500US, in comparison to a cost of approximately $1200 -
$1500 by the British Columbia Cancer Agency Hereditary Cancer Program (BCCA HCP)
laboratories. Since February 2003, the BCCA HC Program has resumed testing in its own

In 1996, the BC Cancer Agency and the BC Provincial Medical Genetics Program
established the Hereditary Cancer Program (HCP) to provide genetic education, counselling and
testing for BRCA1 and BRCA2 to individuals and families with a strong history of cancer
(Williams-Jones, 2002). Through the creation of an official Cost Centre for the HCP in 1997 by
the Ministry of Health, the HCP aimed to offer clinical service while first being established as a
research protocol.

In the United States, it is currently possible to collect your own DNA sample and to have
it sent directly to a laboratory for a substantial fee. Whereas the United Kingdom and Canada
finds direct marketing of genetic testing inappropriate without the involvement of proper
qualified personal, Myriad Genetics, the U.S. base biopharmaceutical and genomics company
that initiated an intensive five months direct-to-consumer advertising campaign for breast cancer

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8 An in-depth review of the current legal issues related to genetic services for BRCA in light of patent regulations is
beyond the scope of this study. For a thorough review, see chapter 2 of Bryn Williams-Jones’ (2002) doctoral
dissertation “Genetic testing for sale: Implications of commercial BRCA testing in Canada.”
genetic testing (Myriad Genetics, 2002) still recommends that all individuals considering genetic
testing first consult with their doctor. While laboratories conducting genetic tests are not required
to participate in quality programs for genetic testing, both the U.S. Task Force and the U.K.
Advisory committee strongly recommend against “over-the-counter” genetic testing for late
onset diseases (Jamieson, 2001).

In Europe, The Public and Professional Policy Committee (PPPC) of the European
Society of Human Genetics (ESHG) organised a workshop in November 1999 in Amsterdam,
where 51 experts from 15 European countries derived recommendations for genetic screening
programs. Recognizing the possible harms that may arise with genetic testing, the PPPC and the
ESHG recommended the establishment of proper organizational criteria similar to those found in
Canada such as: having all programs be governed by a governmental or nongovernmental body
and having in place guidelines for testing, genetic counselling and delivery of information. As
well, they recommended that all individuals who access genetic programs have the possibility to
meet face-to-face with a trained professional if needed following the receipt of test information
(Godard, ten Kate, Evers-Kiebooms, & Ayme, 2003). While maintaining provision of genetic
testing for BRCA1 and BRCA2 within local clinics may be uncertain because of Myriad
Genetics patent laws, the HCP amongst other clinics offering testing aims to continue offering
genetic services in the most holistic manner -- that is, providing pre- and post-counselling
services with genetic testing.

With genetic testing for adult-onset-hereditary-diseases becoming an important part of
clinical genetics practice, we see an increased demand for tests, perhaps as a result of media
coverage of genetic discoveries or from referrals by health professionals. While eligibility
criteria for testing may vary between testing agencies of the same country and of different
countries, the criteria usually include having a strong family history with the disease or early onset occurrence of the disease. The eligibility criteria set by the HCP at the time of the study in 2000 can be viewed in table 3. These criteria represent a 20% chance of finding a mutation in the BRCA1 or BRCA2. Because new discoveries and new developments in genetics are ongoing, regulations of eligibility criteria are likely to change as a result of these.

Within the context of the current study, participants were recruited from the BC Cancer Agency HCP. Most individuals are referred to the program by their health care practitioners. While testing agencies referral process may differ, the HCP uses a triage system set by eligibility criteria for genetic testing for risk of HBOC so that its limited genetic counselling resources can be made available to those whose family history with cancer is most suggestive of hereditary breast and ovarian cancer (HBOC). Although all individuals referred to the HCP can receive genetic assessment, only those who meet eligibility criteria are offered testing following the receipt of their genetic counselling session.

Tested individuals who are found with a mutation in their BRCA1 and/or BRCA2 meet again with a genetic counsellor and a medical geneticist to discuss further results implications for themselves and their family members. For individuals who receive uninformative results, their results are first explained through a telephone call made by a genetic counsellor and the genetic counsellor’s explanations are further followed by a written letter. The HCP does reiterate to all individuals who have received genetic testing that provision of face-to-face discussion is always possible.

From January 1997 to December 1998, the HCP reported over 250 individuals who received uninformative genetic testing results for BRCA1 and BRCA2. At the time of this study, the lapse of time between obtaining genetic testing and receiving results averaged 18 to 24
months for new cases and four to six weeks when a mutation has been identified within a family member.\(^9\) Hence, for individuals who received genetic testing between 1997 and 1998, their full results of both BRCA1 and BRCA2 were obtained in 1998 to 2000 (K. Panabacker, personal communication, January 11, 2000). These individuals constitute the population pool for this study. Recent numbers indicate that the total number of referrals to the Cancer Agency between January 1998 to June 2004 is approximately 5,900 (records were not kept previously). Of those, 80 to 90% were for hereditary breast and ovarian cancer risk assessment. Again, of the 5,900, approximately 4,000 were seen for genetic counselling and more than 508 subsequently received uninformative genetic test results, while 345 received positive results, and 192 received true-negative results. The remainder are still awaiting their results (M. McCullum, personal communication, August 12, 2004).

Although laboratories offering screening for BRCA1 and BRCA2 mutations may differ in the technology they use, three main types have been documented in studies of BRCA1 and BRCA2 mutation screening: full sequencing (Menkiszak, Brzoskoj et al., 2004), the Protein Truncation Test (PTT) requiring polymerase chain reaction (PCR) amplifications (FitzGerald et al., 1996; Iglehart et al., 1998; Malone et al., 1998), and Single-Strand Conformational Polymorphism (SSCP) (FitzGerald et al., 1996; Iglehart et al., 1998; Malone et al., 1998). A new system of testing, Denaturing High Performance Liquid chromatography (DHPLC) is now being tested.

Using DNA from the peripheral blood lymphocytes of an individual already diagnosed with breast and/or ovarian cancer, identification of a possible BRCA mutation is best achieved

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\(^9\) Reasons given to the extent of time between testing and providing results are the unexpected demand for this test and the limited availability of technicians to process all DNA samples (K. Panabacker, personal communication, January 11, 2000).
through full sequencing of the whole gene. However, full sequencing is often not possible largely for economic reasons, thus a modified screening approach is employed. Often, a combination of the Protein Truncation Test (PTT) and single-strand conformational polymorphism (SSCP) are used to detect possible mutations – which can then be characterized by DNA sequencing. Full sequencing of the BRCA 1 and BRCA 2 genes, for example, would offer 99% clinical sensitivity – meaning that the chance of failing to detect a change in the sequence of the gene is less than 1%. Changes such as large genomic deletions, however, will not be detected by full sequencing. Sequencing has the same weakness as SSCP in this regard (M. Anderson, personal communication, June 7, 2000). The PTT can only determine the length of a specific protein but cannot detect a change in the sequence of the gene. That is, the PTT can only detect mutations that create premature protein termination codons. This results in a shortened protein product that can be detected by gel electrophoresis. PTT will produce false negative results if the gene mutation results in a simple amino acid substitution (one for another) or an “in-reading-frame” addition or deletion (loss or gain of a single amino acid) (M. Anderson, personal communication, June 7, 2000).

These types of mutations may or may not have physiological significance in the function of the

\[\text{\textsuperscript{10}}\text{ However, as attested by Peshkin et al., (2001), full sequencing is still not 100% sensitive. Large deletions (loss of genetic material) can go undetected, and there are variants that are not detectable by this method. Full sequencing of a gene refers to the process of determining the order of the individual’s four nucleotide bases in a DNA molecule known as: adenine, thymine, guanine, and cytosine (University of Illinois at Chicago, 2001).}\]

\[\text{\textsuperscript{11}}\text{ The accuracy of a test is described by its clinical sensitivity and specificity. Clinical sensitivity reflects the probability that a test will be positive when a particular sequence is present (Holtzman & Watson, 1997). The failure to detect a change in a specific sequence reduces a test’s clinical sensitivity. Clinical specificity refers to the probability of the test being negative when no change in the gene sequence is found (Holtzman & Watson, 1997).}\]

\[\text{\textsuperscript{12}}\text{ M. Anderson is the laboratory technician who conducts all DNA tests at the Hereditary Cancer Program where the study participants were recruited.}\]

\[\text{\textsuperscript{13}}\text{ Here is an abbreviated glossary to genetic terms used in this paragraph: Genes are pieces of DNA; DNA is composed of four bases. Amino acids represent a combination of three of the four bases which are the building blocks of protein. Codons is a three letter word specifying either the start of a protein, end of a protein or one of the amino acids building blocks. A stretch of DNA correspond to a gene that codes for a particular protein that has a particular function. And exon is the region of a gene that contains the code to produce a specific protein (National Human Genome Research Institute, 2004).}\]
BRCA1 or BRCA2 gene. If, however, the PTT detects a change in the length of a specific protein, then sequencing will be done in that specific area of the gene to determine the specific characteristic of the mutation.

Depending on the setting, in instances where the PTT does not detect any mutation in the major exon observed likely to harbour mutations known to a specific hereditary disease, SSCP is employed on the remaining exons. SSCP is used to find gel mobility variations in the exon, in comparison with normal genes. The presence of a variation indicates a mutation. If a variation is found, sequencing is done for that area of the gene to determine the mutation. With SSCP, a risk of producing a false negative result is possible because the test will not detect large deletions on the exon.

**Genetic Counselling for Hereditary Breast and Ovarian Cancer**

Genetic counselling for hereditary breast and/or ovarian cancer is based on past practices with Huntington Disease (Wiggins et al., 1992). “Genetic counselling has evolved to mean a communication process by which individuals and families come to learn and understand relevant aspects of genetics, to make informed health decisions, and to receive support in integrating personal and family genetic information into their daily lives” (Lea, Francomano, & Jenkins, 1998, p. 111).

Genetic specialists, including nurses, participate actively in the advancement and services of genetic programs in many areas of the US and Canada. Although Canadian genetic programs and services are not as long established as those in the US, we have seen increased interest in Canada in developing both genetic programs and genetic nursing practice. The Ontario government announced in 2000 their decision to fund genetic testing for breast, ovarian and colon cancer through nine regional centres, for a total of 21 clinical genetic clinics as part of the
provincial health insurance (Williams-Jones, 2002). Since then, a group of Canadian nursing researchers began studying the expansion of genetic nursing in Canada. The research is funded by CIHR and was entitled Genetic Services and Adult Onset Hereditary Disease: Current and Future Nursing Roles (Bottorff et al., 2004).

The increased availability of genetic testing and the shortage of genetic counsellors require nurses to prepare themselves to participate in genetic counselling. Some documented roles of nurses include supporting individuals and their families during the counselling process, helping them to integrate new genetic information into their daily lives, collaborating with genetic counsellors to support individuals and families, and coordinating genetic health care (Lea et al., 1998).

Other genetic specialists involved in genetic counselling include clinical geneticists (MDs), medical geneticists (Ph.Ds), clinical molecular geneticists (MDs or Ph.Ds), genetic counsellors (health professionals with Master degrees), advanced-practice genetic nurses (nurses with Master of Science or Doctoral degrees), and genetic nurses (Lea et al., 1998). This team of genetic specialists collaborates in providing support, genetic evaluation, information, and resources to individuals and their families. The team members also obtain and interpret complex family history information, evaluate and diagnose genetic conditions, and interpret and discuss complicated genetic test results (Lea et al., 1998). The person seeking testing and their family members have an active role in decision making throughout the counselling process.

Guiding principles of genetic counselling include nondirectiveness, awareness of social and cultural differences, confidentiality, and privacy (Lea et al., 1998). Genetic counselling aims to respect individuals' right to self-determination (Michie & Marteau, 1996). The nondirective

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14 See also the following link for more information on the Ontario government announcement to funding genetic testing for breast, ovarian and colon cancer: http://www.health.gov.on.ca/english/providers/program/ohip/bulletins/4000/bul4352.html
approach also respects decisions that reflect the clients’ personal values, beliefs, and interests. It also requires genetic specialists to be aware of their personal values and how they may be reflected in communicating genetic information. Furthermore, nondirectiveness means that genetic specialists must provide all information about the risks, benefits, and limitations of a particular genetic test, and about prognosis for and management of the genetic condition (Lea et al., 1998).

Awareness of social and cultural differences calls for genetic specialists to recognise how values and beliefs can affect genetic counselling. Cultural differences are not solely associated with race or ethnicity, as Clarke (1997) points out, but also with a wide range of formative experiences. Individuals from different ethnic groups may experience problems when they are in contact with genetic services. Linguistic and cultural barriers may influence their understanding of and response to genetic counselling and screening (Clarke, 1997). Cultural differences may also affect autonomous decision making about genetic testing. Further empirical studies of beliefs about inheritance and disease are needed to improve professionals’ understanding of them.

Genetic specialists must also assess how their personal beliefs influence the way they collect and communicate genetic information (Lea et al., 1998). Personal values of clients should not be seen by health professionals as limitations to their understanding of genetic information. In fact, having a better understanding of their clients’ values and beliefs should enable genetic specialists to adapt their counselling to understand better their clients.

Confidentiality and privacy are important in genetic counselling as well. Genetic test results can affect family members, intentionally or not. Individuals do not always want their family members and friends to know of their genetic status. While not telling family members
about the test result could deprive them of information that could affect decisions about their lives, the current state of knowledge about prevention or early detection of breast or ovarian cancer does not justify such disclosure. However, legal interpretations from authors such as Burgess, Laberge, and Knoppers (1998) and Surbone (2001) suggest that this view may change if more effective treatments show in the future that disclosing genetic mutation status may prevent harm. Women with a familial history of breast cancer should usually be advised to take early detection measures whether or not they have BRCA1 or BRCA2 mutations (Burgess, 1999; Koenig et al., 1998). Moreover, the risk of cancer to other family members when an individual within a family is found with an inherited mutation is not immediate and hence, not likely to change substantially as a result of genetic information (Koenig et al., 1998). Generally, genetic specialists should honour any wish for confidentiality of the individual who seeks genetic testing. Because there is a possible risk of insurance discrimination, ensuring and accurately describing the risk related to confidentiality of genetic information becomes even more important (Burgess et al., 1998).

Privacy, confidentiality and threat of genetic discrimination are the most cited ethical issues related to genetic testing (Burgess, Knoppers, & al., 1999; Knoppers, 1998; Surbone, 2001). Describing these potential social harms and psychological distress that may arise from genetic testing is a key part of informed consent. Informed consent should always be obtained before genetic tests are conducted and opportunity for the individual to participate fully in the decision process needs to be provided (Burgess et al., 1998). Moreover, informed consent should

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15 There exist detailed theoretical literature on the ethical implications with genetic testing, however, as the current study is not focused on the moral and ethical issues with genetic testing, only an abbreviated review of the content address in documented and observed informed consent to genetic testing is presently address. For a current literature review on the contemporary bioethics literature on genetic testing for hereditary breast/ovarian cancer, see d’Agincourt-Canning (2003). Her study addressed the complex moral and social issues generated by genetic testing. Also see Burgess (2001) for a review of the ethical and social issues in genetic testing beyond informed consent.
not be limited to the technical and medical aspects of testing but should also include social and ethical implications (Surbone, 2001), as well as alternatives to proposed treatments. While full autonomy in genetic testing can never be attained fully, a standardised protocol that explicitly describes the process of genetic counselling and informed consent provides at least some guidance towards obtaining informed consent (Burgess, 1999; Burgess & d'Agincourt-Canning, 2001; Burgess et al., 1999).

Within the issue of privacy, individuals can be told that, although the clinic will keep their test results confidential, there is always a risk that genetic discrimination may occur (Koenig et al., 1998). Health professionals and others have an obligation not to disclose private medical procedures unless authorised by the patient. In addition to discrimination in life insurance, disclosure of genetic information can cause discrimination by health insurance providers and, discrimination in the work place (Burgess et al., 1998; Surbone, 2001).

Psychological distress can result when genetic testing is offered without pre- and post-test genetic counselling (Burgess et al., 1998). What genetic testing may reveal should be discussed before the test within the informed consent procedure (for example, paternity). Follow-up genetic counselling should assess how individuals have integrated the test results and their implications into their lives (Burgess et al., 1998).

Although risks to privacy and confidentiality as well as the risk of discrimination have been discussed by many in the ethics and legal communities with an interest in genetics, few of these risks have been empirically documented in Canada. However, according to Surbone (2001), cases of discrimination following genetic testing for BRCA have been documented in the United States. As Surbone (2001, p.156) warns, “we are not our genome: yet we culturally tend to see genetic data as more fundamental and more inalterable than any other equation of life.”

16 Other psychosocial issues that may arise from genetic testing are discussed in the next section of this chapter.
D’Agincourt-Canning’s (2003) doctoral study, which addresses moral agency in genetic testing for hereditary breast and ovarian cancer, proposes that, in light of the genetic information being familial as well as personal, that perhaps the concept of relational responsibility could be assessed for its potential contribution to the informed consent process. Relational responsibility complements an individual’s right to autonomy and reinforces the complex social nature of genetic testing (Burgess & d’Agincourt-Canning, 2001). D’Agincourt-Canning (2003) argues that further empirical study focusing on the experiential impact of genetic testing on people’s everyday lives is needed to understand the moral agency of individuals and families who have received results of genetic tests to hereditary breast and ovarian cancer.

What has been described and discussed so far are the technical and ethical issues related to genetic testing and counselling. What follows is an overview of the process of experiencing genetic testing and counselling.

**Experiencing the process of genetic testing and counselling**

The information presented in this section is derived from the literature, as I do not have first-hand clinical experience in genetic counselling, although I have observed three genetic counselling sessions. The process observed in these sessions is representative of the report by Lea, Francomano and Jenkins (1998) on the genetic counselling process. These authors explain that genetic counselling can occur over an extended period and usually entails more than one visit. The long process allows time for individuals and families to understand genetic information before deciding whether or not to have genetic testing done. Although the process of genetic testing may differ from one country to another, individuals and their families can be asked to first complete a survey that contains a family history questionnaire. The survey is usually provided by the family physician who, based on the family history, recommends genetic testing.
Once the completed questionnaire is received by the agency, a genetic nurse evaluates the family history of the individual to decide if the person meets criteria for genetic risk assessment. If the individual does meet the criteria, she/he is put on a waiting list to be seen by a genetic counsellor. Prior to the counselling appointment, the individual receives a telephone call from the genetic nurse explaining what may be expected from the counselling session and who can come for testing. However, the nurse will also explain that she may choose to have the family members make their own appointments and undergo the process separately. During the pre-counselling phase, the nurse will also assess the emotional and social status of the individual and her family (Lea et al., 1998). Usually all individuals seen for genetic counselling are seen again after receiving the results of their genetic test.

According to Lea et al. (1998), first phase of genetic counselling is assessment and information gathering. This includes asking individuals and family members why they were referred and what they expect from the session. According to my observations of genetic counselling and the literature, genetic counsellors then review the family history; determine the clients’ level of knowledge about hereditary breast and ovarian cancer; communicate genetic information; discuss genetic testing, its accuracy and its pros and cons; and describe existing supports such as networking groups and the follow-up genetic counselling that can be expected. Following each of the sessions, genetic counsellors, with individuals’ permission, write a report for the family physician and coordinate the care to be given by other specialists as needed. For example, when family members agree to give blood for genetic testing but live far apart, the counsellor will make the necessary arrangements to have the person tested if genetic testing is offered in their area or arrange to have their blood sample shipped to the testing agency.
While much of the information needed to make a genetic risk assessment is gathered by the genetic nurse during the precounselling phase, this information is then reviewed by the genetic counsellor with the individual during the session. Following this review, the genetic counsellor gathers and discusses with the individual her or his family history with cancer. This phase consists of filling out a family pedigree based on information shared by the individual during the precounselling phase and information shared during the session. The pedigree represents the family’s medical history and helps the genetic counsellor determine if a strong possibility exists for an inherited susceptibility to breast and/or ovarian cancer (Lea et al., 1998). The genetic counsellor collects information about the number of affected and unaffected individuals with breast cancer and other forms of cancer from the maternal and paternal sides of the family and inquires about the ages at which diagnoses were made. Based on my observation of genetic counselling sessions, the pedigree is used by the genetic counsellor to explain to individuals the differences between sporadic and hereditary breast cancer. The discussion focuses on the inheritance of cancer if the family history suggests a strong probability of this. The genes associated with breast and ovarian cancer are discussed and presented graphically and the mutation process that can lead to cancer is explained. This part of the session is usually presented by both a medical geneticist and a genetic counsellor and also includes a discussion of the risks, benefits and limitations of testing, and available treatments, if an inherited susceptibility to the disease is found (Lea et al., 1998). However, presenting and explaining notion of inheritance is not as straightforward an activity as presented in the Richards and Ponder (1996) study.

17 This part of counselling can be sensitive as, to some individuals, viewing their family history in a pedigree may bring their first awareness of a probable inherited susceptibility to breast and/or ovarian cancer.
Richards and Ponder (1996) conclude through hypothesis testing that people tend to derive their understanding of inheritance from concepts of social relationship of kinship, which Richards and Ponder (1996) explain are sustained by everyday social activities. The authors posit that closeness in genetic terms is determined by the closeness of social relationship and social obligations. For example, the authors note that breast cancer and ovarian cancer were described by their study participants as a "proneness" to cancer passed by mothers to daughters. Conversely, women from families with ovarian cancer may not see themselves at risk because they "take after their father's side of the family" (Richards, 1996). Overall, people recognise an equal contribution by both parents to inheritance but are less aware of the genetic connection between an individual and more distant relatives. Richards (1996) notes that some inheritance principles understood by lay people are that family resemblance to an affected family member makes that individual more at risk to develop the familial disease and believe that the disease will appear at the same age as the affected relative. There is also the idea of inheritance as being able to "ski" a generation. Cox and McKellin's (1999) research demonstrates that social components, rather than biological components, shape the everyday understanding and experience of hereditary risk. Their conclusion concurs with that of Biesecker (1997b), whose secondary analysis of studies and book chapters that had looked at the psychological aspects of cancer genetics and susceptibility testing shows that risk perceptions are also shaped by the complexities of experience in a cancer family and by factors influencing the understanding of risks such as personality traits, illness perception, family relations, mood, and anxiety.

Assessing how individuals interpret inheritance and how they draw upon these interpretations when understanding hereditary risks is all part of the counselling process. Whereas most sessions take up to an hour or more depending on the agency and the individual's
need, a second face-to-face counselling session is, at times, requested by individuals before making their decision for testing. Follow-up education will often be done by the genetic nurse. Concomitantly, at the end of the session, the individual and family members are informed that it takes time to understand genetic information and that a networking group is ready to help answer their questions and concerns.\textsuperscript{18}

While follow-up genetic counselling when giving out genetic testing results is recommended standard practice, it is not always feasible. When genetic testing is done through a private company such as Myriad Genetics, the onus is on the provider or on the individual to seek pre- and post-test counselling through their personal health care provider. However, in cases where genetic testing was done within a research protocol, postcounselling is usually included in the process. But, at times, because of cost and time constraints, only those who receive positive test results may be offered a one-to-one session, while individuals who receive uninformative results are given support over the telephone and sent a letter explaining the meanings and implications of their results. They are usually offered a one-to-one session only if they feel the need. Again, procedures for informing individuals and families of their test results depend on the agency (Lea et al., 1998).

Providing genetic test results is an important part of the genetic team's role. However, communicating risk information to the lay public can be challenging. One of the challenges to communicating cancer risk information is in moving beyond the individual's assessment of risk to assessing their risk within their familial context. That is, in that family history of cancer is an important risk factor in establishing personal risk, situating cancer risk information within the

\textsuperscript{18} Because most genetic testing is offered through research programs, not all settings that offer this service also coordinate the networking groups. The Hereditary Cancer Program of the BC Cancer Agency offers such a service for those who have received genetic counselling, regardless of whether or not they have cancer or received their genetic test results.
familial context becomes important. Other challenges include how risk information is communicated, as the way the message is framed can influence how the content is understood and acted upon (Sarfati, Howden-Chapman, Woodward, & Salmond, 1998). For example, following an empirical literature review of thirteen studies, Edwards and al. (2003) found that detailed personal risk communication in the form of numerical calculations of risk was associated with a smaller uptake of screening while risk communication stated as categories (high, medium and, low) or simply listed as personal risk factors presented higher uptake of screening programmes. Most screening programmes addressed in the studies reviewed were in relation to mammography screening, breast cancer risk and genetic testing. Bottorff, Ratner, Johnson, and McCullum (1999) also comment on how physicians’ struggle with conveying limitations of cancer risk knowledge for fear that this information could be misinterpreted.

The literature documents that health professionals and lay people differ in the ways they understand risk information (Peshkin et al., 2001). That is, risk figures can be given in genetic counselling as odds or as percentages (Harper, 1993). Pitfalls exist for both methods, however. With odds, some individuals may not clearly grasp that odds ratios refer to the future and not to the past. As Harper (1993) explains, just because an individual has two affected children in succession does not make it less likely that the next child will be affected since chance has no memory. Also, the possibility exists that individuals reverse or misinterpret odds ratios. Nevertheless, when risk values are framed in positive ways, for example the risk of not developing breast cancer for carriers of BRCA1 and BRCA2 gene mutations is approximately 30% as opposed to the risk of developing breast cancer for carriers of BRCA1 and BRCA2 is approximately 70%, the experience can be less threatening and may lead to different decisions
Consideration should be given to how risk estimates and figures are interpreted by individuals who seek and receive genetic testing. These interpretations need to be explored during counselling sessions, since studies have shown that lay people do not always understand principles of probability (Macdonald, Doan, Kelner, & Taylor, 1996).

Communicating cancer risk also poses challenges for people who do not always understand the meaning of lifetime risk for cancer and how such risk can be modified by individual factors (Rimer, 1999). Lipkus and Hollands (1999) suggest the use of graphic visual display to enhance the understanding of numerical risk. While diverse approaches to providing risk information may help individuals understand their theoretical risk to cancer, none can eliminate the ambiguity resulting from the inherent uncertainty associated with risk information. Future studies are still needed to assess the best approach in presenting uncertainties with risk estimates.

Studies that have looked at communication of risk information note that some individuals may feel more “prone to cancer and in having the familial known inherited mutation to cancer” when they see physical or behavioural resemblances between themselves and the individuals in the family who have developed cancer or who have been found to have an inherited mutation increasing their susceptibility to cancer (Richards, 1996; Richards & Ponder, 1996). While some individuals within one family may see themselves more prone to cancer and to an inherited mutation, others may see themselves less at risk. For example, within the context of hereditary breast cancer, men have an equal risk to women in inheriting a BRCA1 or BRCA2 mutation and passing it down to their children but men may still perceive themselves less at risk for inheriting a mutation associated with one of these cancer genes because of seeing breast cancer as less

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19 The topic of decision making with uncertain results is discussed later within this chapter under the section “interest and intended use of test information”.

relevant to them and being a women’s disease. However, even less understood than how perceptions of risk are formed and how risk is communicated are the conceptual structures individuals use to interpret and make sense of genetic test results. There is a considerable absence of studies that have empirically researched this process. As Lynch et al. (1999) attest, because identification of the BRCA1 and BRCA2 mutations is relatively recent, knowledge of how best to translate genetic information in a clinical setting is still ongoing.

Even though providing genetic test results for BRCA1 and BRCA2 may seem relatively straightforward, in many instances the process becomes complex, not only because of the cases encountered within clinical settings but because of the many complex issues in genetic counselling (Peshkin et al., 2001). Even with positive genetic test results, the process is complex, as the risk of developing cancer cannot be precisely quantified and management options are still uncertain. However, recent studies indicate that prophylactic surgeries protect against recurrence of the cancer or new primary cancer among those with an identified BRCA mutation (Narod, 2002). Affected individuals found to carry an inherited mutation have an approximately 40% risk of developing contralateral breast cancer over the next decade (Narod, 2002).

The experience of true-negative results can also be compounded by confusing interpretations because an individual may interpret the results as no longer being at risk of ever developing breast and/or ovarian cancer and experience their good fortune with survivor’s guilt over family members found with the familial mutation (Peshkin et al., 2001). Or, as is the focus of this study, individuals with a family history of risk for hereditary breast and/or ovarian cancer may receive results indicating no mutation was found without a straightforward explanation from genetic health professionals on how to interpret and make sense of their results. Thus, genetic

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20 Recall that a true negative genetic test results for BRCA1 and BRCA2 can only be received by individuals from families with an identified inherited mutation of either cancer gene.
counsellors and other health professionals providing support and education face many challenges – especially with individuals receiving uninformative genetic test results. In fact, they will likely face many such individuals, as studies have demonstrated that 16% to 66% of all high-risk families do not carry detectable mutations in their BRCA1 and/or BRCA2 cancer genes (Peshkin et al., 2001).

Within the health literature, guidance can be found for interpreting uninformative BRCA1 and BRCA2 genetic test results; however, as said earlier, most of this information is from clinical literature as opposed to empirical literature. Authors such as Lynch et al. (1999), Peshkin et al. (2001) and Iglehart et al. (1998) who have conducted studies on the complexities of interpreting uninformative genetic testing results to BRCA cancer gene mutations maintain that counselling individuals from families considered at risk for HBOC can be a huge challenge. Such as, Lynch et al. point out, “there is currently no test that is sensitive enough to pick up all possible mutations” (p. 97). Hence, individuals receiving uninformative genetic test results must be told that there is still a possibility that they carry a mutation undetected by current tests. Iglehart et al. recommend that individuals know about and understand these limitations to genetic screening before deciding to have genetic testing done. Interpretation of negative results from studies conducted on other inherited diseases such as for familial adenomatous polyposis (FAP) also show that some individuals fail to be reassured of their decreased risk to general population risk as a result of having received a negative result (Michie, Smith, Senior, & Marteau, 2003). Although representing a small scale study of nine participants, explanations given by Michie et al. provide some understanding on the reasons for the lack of reassurance experienced despite receiving negative results. Among the nine participants, four continued to perceive self at increase risk for FAP because they doubted the efficacy of a blood test to determine one’s risk
for a disease that occurs in the bowel. The participants also perceived DNA testing as too simple compared to an invasive testing of colonoscopy that can actually see inside the bowels. Other findings from their study also show that participants expressed doubts about the sophistication of DNA testing technology to detect all mutations as well as viewing their results as transitory. That is, the testing could only test for known mutations to date.

To illustrate the complexities that may arise with genetic counselling setting of individuals who receive uninformative results, I will summarise a case study presented by Peshkin et al. (2001). This case was drawn from their clinical research with high-risk individuals who received genetic counselling and testing. It is a case of an affected woman with a previous breast cancer diagnosis at the age of 41. Her family’s history of cancer comes from her paternal side and presents only with breast cancer. There are six cases of breast cancer in three generations with four considered early-onset cases (i.e., diagnosed before the age of 50), one male breast cancer and one case of bilateral breast cancer. The index case (this affected woman) underwent full genetic sequencing for BRCA1 and BRCA2 and was not found to have a mutation or any variants. The authors conclude that, in light of her strong family history that gave her an overall 90% risk of carrying an inherited mutation, hereditary breast cancer cannot be ruled out. Possible interpretations of her situation were multiple and complex. First, she was told that current tests are not 100% sensitive in detecting all mutations and, therefore, it is still possible that she may carry a mutation. As well, she was told that her suspected mutation may be in an as yet unidentified breast cancer gene. Finally, she was told that there is a possibility that her breast cancer may be a sporadic case within a hereditary breast cancer family. The authors felt, however, that this explanation is unlikely because she was a first-degree relative of a male breast cancer case and she was the youngest in the family to receive a breast cancer diagnosis.
Hence, she was advised to have other affected family members tested and that, if their tests were negative, it would be highly unlikely that all the tested women in this family would develop sporadic breast cancer considering the high occurrence of cancer in their family in comparison to what is expected within general population. Upon this, she would be recommended to enter additional research studies, such as those offering linking analysis.

The above case clearly presents some of the complexities that were faced by the genetic team at the Hereditary Cancer Program when the participants of the current study received their "uninformative" genetic test results. While Peshkin et al. (2001) recognise that the time-consuming nature of providing comprehensive services may make it difficult for one genetic professional to provide all the care, they recommend referral to an oncology nurse or a nurse specialising in cancer genetics, because they have ongoing contact with affected individuals through regularly scheduled follow-up appointments.

To conclude this section on challenges to genetic testing and genetic counselling, one area that further complicates the making-sense process of individuals receiving uninformative results is how to use such results in their medical management. The following section reviews interests in genetic testing and intended use of test information.

**Interest in Test Information**

Research involving individuals considered at risk for HBOC suggests that substantial proportions are interested in genetic testing for breast and ovarian cancer susceptibility. Comparison between two American studies on this subject showed significant differences in the level of interest between individuals recruited from the general population and individuals recruited from a breast cancer centre who had at least one first-degree relative with a past breast and/or ovarian cancer diagnosis. The latter study, by Lerman et al. (1995), showed a greater
interest among its study participants with a family history of cancer (91% or 96 out of 105 participants) compared with Tambor, Rimer and Strigo’s (1997) study of the general population in which 69% of participants showed interest in genetic testing for breast and ovarian cancer susceptibility. Other studies, such as those by Lipkus, Iden, Terrenoire, and Feaganes (1999) and a Canadian study by Bottorff et al. (2002) produced comparable results. After controlling for differences in age, education, personal history of breast cancer, and knowledge of genetics, Bottorff et al. found that individuals with at least one first-degree relative with breast cancer were two times more likely to be interested in genetic testing for breast and ovarian cancer susceptibility than those with no family history. However, their research showed lower overall rates of interest than reported in both Canadian and American studies. Bottorff et al. reported an interest rate of 28.5% among the 761 individuals surveyed who had no personal history of breast cancer and a rate of 30% among the 260 individuals surveyed who had a personal history of breast cancer. However, as other studies on interest in genetic testing have indicated, lower interest may suggest that, despite media coverage, individuals from high-risk families are not sufficiently informed about the availability of testing to decide to use this technology (Andersen, Bowen, Yasui, & McTiernan, 2003; Bottorff et al., 2002). Finally, Donovan and Tucker (2000) conclude from their study that no relationship exists between interest in genetic testing and individuals with a family history of breast cancer.

According to Tambor et al. (1997), sociodemographic factors common among members of the general population interested in genetic testing are being younger than 60 years and being Caucasian rather than African American. Tambor et al. hypothesise that younger women may be more interested in knowing their probability of carrying an inherited mutation in order to decide whether to have children, while older women may be more concerned about their risk of cancer
because of their advanced age. Bottorff et al. (2002) found that sociodemographic factors influencing interest in genetic testing were being less than 50 years old, having a positive family history of breast cancer, and having more years of education. Psychosocial factors reported to influence interest in genetic testing for breast and ovarian cancer susceptibility include having a sense of vulnerability to breast cancer (Press, Yasui, Reynolds, Durfy, & Burke, 2001), cancer worry, and beliefs that having testing will result in increased access to cancer screening (Durfy, Bowen, McTiernan, Sporleder, & Burke, 1999; Gwyn, Vernon, & Conoley, 2003).

Beliefs related to breast care influence interest in genetic testing. Those who believe that mammograms will benefit them and their family, and that they will feel more in control over their health were found to be more likely to be interested in testing (Tambor et al., 1997). As with the results of work by Lerman et al. (1995), interest in testing was associated with acquiring information useful for other family members such as daughters. The authors conclude that interest in testing differs among individuals from different age groups, races, levels of schooling and use of health care. In light of what is known about sociodemographic factors influencing interest in genetic testing for breast and ovarian cancer susceptibility, both Lerman et al. (1995) and Bottorff et al. (2002) recommend that these factors be used in targeting community-based education about genetic testing. They also recommend that research be done to test different approaches to education and counselling based on specific characteristics of target populations. Although it can be hypothesised that when planning for cancer genetic services, interest in genetic testing will always be higher than actual uptake (Cappelli et al., 1999) I found very few Canadian studies that actually compared interest and uptake in the general population. Finally, Marteau and Croyle (1998) suggest that uptake rates for genetic testing are higher when there are effective ways of treating or preventing the condition. They report that an average of 50% of
those interested in genetic testing for breast and ovarian cancer susceptibility proceed with testing compared to 10% for Huntington’s disease, for which there is no treatment and 80% for familial adenomatous polyposis, for which there is effective treatment.

**Intended Use of Test Information**

I found very few studies that looked specifically at the use of genetic testing for BRCA1 and BRCA2 with the intent to use test information to support health management decisions to reduce risk of breast and ovarian cancer among individuals considered at risk for HBOC. What is even more crucial is gaining empirical understanding of the use of genetic results to inform health-related decisions when test results are uninformative or are uncertain. Both of these contexts will be discussed below.

One study on this topic by Meiser et al. (2000) looks at intent to pursue prophylactic bilateral mastectomy among women at risk of HBOC. The study found that 19% of the 333 women surveyed said that they would consider prophylactic mastectomy if the test revealed that they carry an inherited mutation in their BRCA genes. This was in contrast with the majority (47%) who said that they would not consider a mastectomy while 34% said they were unsure and 1% had already undergone a prophylactic mastectomy. Statistical measures from the Meiser and al. study showed a correlation between breast cancer anxiety and consideration of mastectomy. As well, the highest proportion of women considering prophylactic mastectomy were within the 30 to 39 age group (Meiser et al., 2000). Perceived risk as opposed to actual risk was also found to correlate with consideration of prophylactic surgery. Although these results concern hypothetical situations whereby the individuals are only viewed as having a theoretical risk to an inherited mutation compared to an actual risk, they nonetheless provide an overview to the context of having to make health-related decisions with inconclusive or uninformative genetic
testing results. Uninformative test results are of limited use in decisions about prophylactic mastectomy because they do not provide a definitive measure of genetic mutation status and of risk of cancer. Although some recent empirical evidences suggests that prophylactic bilateral mastectomy reduces morbidity and mortality among individuals at risk of HBOC (Meijers-Heijboer et al., 2001; Narod, 2002), further studies establishing this surgery's effectiveness are still needed (Burke et al., 1997; Meiser et al., 2000). Bouchard et al. (2004) also suggest that there are cultural differences that influence recommendations of prophylactic surgery. Their study showed that cancer geneticists in Montreal (Quebec, Canada) and in Manchester (Great Britain) were more likely to recommend prophylactic mastectomies than cancer geneticists from Marseilles (France). One study that provides prospective evidence of the usefulness of prophylactic surgery for unaffected individuals identified with a BRCA1 or BRCA2 gene mutation is by Meijers-Heijboer et al. (2001). Their results show that 3 years after surgery, among the 76 out of the 139 women identified with a BRCA1 or BRCA2 mutation who chose to have bilateral prophylactic mastectomies, none developed invasive breast cancer. This compares with the 8 women out of 63 who chose regular surveillance instead of surgery that did develop breast cancer. These eight women developed invasive breast cancer. While more studies are needed to determine the long-term protection of prophylactic mastectomy for women identified with a BRCA mutation, this next study does point in favour of the surgery to confer protection against the appearance of cancer. Among the 22 carriers of BRCA mutation diagnosed with a previous breast cancer before the age of 42, at 12 years of follow up, half had developed a second primary breast cancer (Haffty et al., 2002). The majority of cancers were classified as second primary tumours.
While not all genetic scientists would agree, Narod (2002) views it prudent to recommend bilateral mastectomy for women with a past breast cancer diagnosis known to carry an inherited mutation of BRCA1 or BRCA2. He also suggests the use of tamoxifen (a cancer preventing drug) to reduce the incidence of second primary cancers. Generally, health professionals presume that those who learn of a genetic risk will be more motivated to strictly follow breast cancer surveillance measures (Bredart, Autier, Audisio, & Geragthy, 1998; Lerman, Seay et al., 1995). Studies, however, have not conclusively demonstrated this assumption to be correct in all cases. Reactions to positive genetic test results may be more complex than was initially presumed. As demonstrated in the 1992 study by Kash, Holland, Halper, and Miller, high cancer anxiety led to poor adherence to a regime of breast examinations by health professionals and monthly breast self-exams among women with a family history of breast cancer.

Although researchers hypothesize that women who carry BRCA1 and/or BRCA2 cancer gene mutations are able to reduce their risk of developing and dying from breast cancer by using preventive measures such as prophylactic mastectomy (Lerman, Hughes et al., 1998; Narod, 2002; D. L. Newman et al., 2004; Rebbeck et al., 2004), controversy exists about the efficacy of strategies for preventing cancer in carriers (including prophylactic surgery) and strategies for managing surveillance of cancer (Bredart et al., 1998). Such controversies are doubts in the efficacy of breast cancer mammography in women under the ages of 40 as well as the lack of prospective studies showing a protective use of tamoxifen, prophylactic mastectomy and prophylactic oophorectomy in reducing the risk of breast and ovarian cancer. Other associated controversies with prophylactic surgeries are complications of premature menopause, and the few studies that have addressed the quality-of-life issues in women who have opted for

Certainly more studies are needed about women’s interest in using genetic testing to support and inform health-related decisions. As put forth by the Bottorff et al. (2002) study, the most frequently cited reason for genetic testing among the 260 women with a past breast cancer diagnosis surveyed was for simple curiosity. Bottorff et al. posit that this finding might reflect individuals’ expectation that genetic testing will tell them categorically whether they will get the disease or not rather than providing a probability of cancer risk based on genetic status. Another reason for testing was to warn family members and to take preventive action, although specific actions were not mentioned by survey participants.

The utility of uninformative genetic test results in assisting individuals with risk management decisions is undocumented. However, one recent study suggests that such test results may lead to decisions based on incorrect assumptions in individuals’ personal interpretation of their results (Frost et al., 2004). This study by Frost et al. took place in the High Risk Breast Cancer Clinic at Huntsman Cancer Institute, University of Utah. The clinic is both a research facility and a clinic for individuals with a family history of breast and/or ovarian cancer. Frost et al. conducted a qualitative pilot study using focus groups and individual interviews with 15 women enrolled in their breast-cancer surveillance program. The study examined how women made health-related decisions when provided with uncertain genetic test results for BRCA1 and BRCA2. Based on the type of uncertain results, women were enrolled in three different groups:

- Group 1 (n=6)—affected women with breast and or ovarian cancer who received results indicating a variant of uncertain significance;
Group 2 (n=4)—unaffected women found to carry an inherited mutation; and
Group 3 (n=5)—affected women considered at risk for HBOC but for whom tests did not identify an inherited mutation.

The authors define uncertainty in group 3 as an absence of explanation for their personal and family history of cancer, their future risk of cancer and that of others in their family members. The authors report that women who received their results face-to-face understood the difference between uncertainty in test results and uncertainty in their risk of developing cancer. Thus, understanding such a difference in their risks as well as having the opportunity to discuss the implications of uncertain results in light of having to make health-related decisions facilitates the making sense process. While the authors did not report what were the health-related decisions to be made, they nonetheless point to the notion that when inconclusive results are unexpected and misunderstood, individuals are not able to use genetic information for health care decision making. Clearly, more empirical studies are needed to assess how individuals receiving uninformative genetic testing results are able to use such information to assist them in making risk management decisions.

**Psychosocial Issues**

**Psychological impact of having a family history of breast and/or ovarian cancer**

Learning that one has a genetic predisposition to breast and ovarian cancer is stressful and there is potential for adverse psychological outcomes (Esplén et al., 1998). However, Kelly et al. (2004) note from their study that once the initial shock of learning one’s genetic status has passed, this news may be accompanied by a decrease in an overall general stress, although most studies point to the contrary (Friedman et al., 1999; Lynch et al., 1993). One type of stress, however, that does not dissipate with time is living with a family history of cancer considered at
risk for HBOC. This stress tends to recur in each new generation with each new cancer diagnosis. Pasacreta, Jacobs and Cataldo (2002) point out that anticipation of breast cancer development among unaffected individuals from known hereditary cancer families may be more psychologically distressing than actually having or having had breast cancer. Individuals from families with histories of cancer are known to experience symptoms of general distress, have frequent intrusive thoughts and can at times deny their risk of cancer (Zakowski, Valdimarsdottir, & Bovbjerg, 2001; Zakowski et al., 1997). Bish et al. (2002a) document that distress associated with increased risk perception to cancer can have detrimental effects, such as failing to interpret appropriately genetic test results (Lerman, Lustbader et al., 1995), reduced participation in screening and surveillance programs (Kash et al., 1992) and excessive breast self-examination. Consequently, when individuals enter a clinic for genetic counselling and genetic testing, they may be already experiencing distress related to their own cancer and genetic risks, and that of others in their family. As for Lerman, Seay et al. (1995), they support that affected and unaffected individuals with a family history of cancer indicative of HBOC do live with an overestimation of their risk to develop breast cancer. Overall, women, unaffected and affected, with a HBOC experience multiple psychological stresses ranging from increased perception of risk for cancer to increased perception of carrying mutations that will increase the cancer risk of family members (Bish et al., 2002a).

**Benefits and limitations of genetic testing for breast and ovarian cancer susceptibility**

The introduction of genetic testing for breast and ovarian cancer susceptibility was accompanied by much enthusiasm among many families at risk of HBOC. They could finally learn whether they had a heightened risk of cancer and whether they would transmit mutated genes to their children (Lerman, 1997). Other perceived benefits were learning one’s genetic risk
status for breast and ovarian cancer susceptibility, planning for appropriate treatments if found with a mutation, providing information to relatives who want to know of their risk for an inherited mutation, helping to decide about life and disability insurance, and assistance in making lifestyle changes to prevent cancer (Cappelli et al., 1999).

While there are benefits to genetic testing such as assisting at risk families make medical decisions and promote cancer risk-reducing behaviours (Lerman, 1997) there are, as well, unfavourable implications to learning one's genetic status. That is, while learning more precisely about one's potential lifetime risk of developing breast cancer by knowing if one is carrying an inherited mutation has been perceived as a benefit (Biesecker, 1997a; Sagi, Kaduri, Zlotogora, & Peretz, 1998; Schwartz et al., 2002), this benefit can also be attenuated when individuals experience estrangement from family members with differing results, experience guilt over the possibility of having transmitted a mutation to their children, lose hope and, experience anxiety and depression (Biesecker, 1997b; Lynch et al., 1997).

For noncarriers within hereditary breast cancer families, a hypothesise benefit is feeling relief in discovering that their risk of developing breast cancer is the same as that of the general population (Lerman, Hughes et al., 1998). Nevertheless, because genetic testing is in its early years and most of the research in this area has focused on those who discover that they have a genetic mutation, the lack of empirical studies of individuals who tested negative to breast and ovarian cancer susceptibility does not permit us to draw conclusions of possible similarities from this group to the uninformative group about the degree to which relief from anxiety and uncertainty can be found. However, one recent study assessed the difference in cancer-specific distress and perceived risk for breast and ovarian cancer between individuals from known hereditary breast cancer families who tested positive and those who tested negative (Schwartz et
al., 2002) as well for individuals who received uninformative results. The authors had hypothesised that, since an uninformative result was determined as inconclusive to an individual’s genetic status to an inherited mutation, they did not expect to see significant reductions in distress or perceived risk. Both of these variables were assessed through a structured telephone interview which was then followed by a face-to-face counselling session. Among their participants, 78 tested positive, 58 tested negative and, 143 received uninformative results. No significant difference was found from baseline to 6 months following disclosure between the positive group and the uninformative group on the mean scores of distress outcomes. This study, however, did find that cancer-specific distress and perceived risk for breast and ovarian cancer had significantly decreased for individuals found not to carry an inherited breast and ovarian cancer gene mutation. Hence, the uninformative group followed similar path as the positive group and did not exhibit a decrease in distress as reported among relatives who received a negative test results from known HBOC families.

Another benefit foreseen by individuals going for testing is learning more precisely about the risks for their children or future children to inherit a mutation, and deciding about whether or not to have children. The desire for reassurance can be powerful but can also be counteracted if test results are not as expected (Claes et al., 2004). Hence, during genetic counselling, emphasis should be placed on the fact that, if an individual has a mutated gene associated with breast cancer, then their risk of transmitting it to their children would be 50%.

Also, the possibility exists that individuals will learn that they did, in fact, transmit a mutated gene to their children. Reactions to such situations include self-blame for passing the genetic mutation and feeling guilty for failing to protect one’s children (Biesecker, 1997b; Claes et al., 2004; Lynch et al., 1999). Although the precise effect of testing for breast cancer
susceptibility on family relationships is not yet fully understood (Macdonald et al., 1996), some effects have been documented among individuals such as daughters, siblings, parents, and first- or second-degree relatives. Individuals may unwillingly learn more about their own susceptibility following the revelation of the genetic status of a family member (Bredart et al., 1998; Croyle, Achilles, & Lerman, 1997). Family members may pressure each other to go ahead or not to go ahead with genetic testing (Lynch et al., 1999).

Adverse effects on an individual’s self-concept is another potential risk of genetic testing (Biesecker, 1997b; d’Agincourt-Canning, 2003). How mutation status disclosure may influence the quality of life of an otherwise healthy person found to be at increased risk of developing cancer in the future is largely unknown. In addition, disclosing genetic information may cause people to worry about when the disease will develop, as it has been observed that some individuals equate having an inherited mutation of their BRCA cancer genes with receiving a cancer diagnosis (Welch & Burke, 1998). While early disclosure of one’s genetic status may prompt some to be more regular with their cancer screening, early disclosure can also interfere with an individual’s present life and goals for the future (van Zuuren et al., 1997). Further, Croyle et al. (1997) comment on that offering information about the probability of developing cancer can complicate the task of maintaining a healthy concept of self. Individuals who live with the perception to being at risk of cancer because of a known mutation in their family and who learn that they do not carry this mutation may have difficulties incorporating into their self-identify that they are not at extremely high hereditary cancer risk (Lynch et al., 1999). Noncarriers may also no longer see themselves at risk or feel that their risk is below average. According to Lerman, Daly, Masny, and Balshem (1994), 72% of the women they studied who are at risk for hereditary breast cancer expected to go on worrying even if they received a result
indicating that no mutation had been found. Hypothetically, individuals from known hereditary breast cancer families who decline genetic testing may be doing so to protect their sense of wellbeing or because they feel that proven strategies for preventing cancer are still lacking. While there needs to be an understanding that not all health problems are the result of heredity, there may be a societal tendency towards genetic reductionism – providing simplistic medical explanations to multifactorial, social problems such as in the case for breast cancer when breast cancer is a multifactorial disease. Nelkin and Lindee (1995) labelled this trend genetic essentialism – reducing the self to a molecular entity and discounting the environmental context in which our genes exist. The risk is that genetic essentialism may create new forms of social eugenics and lead to new social classes based on genetic inheritance (Macdonald et al., 1996). Macdonald et al. (1996) believe that holding such a restrictive view of genetic essentialism may result in genetic testing one day becoming mandatory, so that people may lose their right to make their own decisions if they want testing or not. Genetic essentialism could also lead to discrimination, as it has in the past for certain social groups (for example, Black Americans at risk of sickle-cell anemia) (Macdonald et al., 1996).

Other limitations of genetic testing for BRCA genes include the degree to which test results are uncertain (Frost et al., 2004; van Zuuren et al., 1997). From their study evaluating uncertainty in genetic counselling process, van Zuuren et al. (1997) describe the distressing situation in which individuals and families found themselves when they were looking for reassurance from genetic testing but instead received uncertain results. van Zuuren et al. note that this inconsistency where individuals consult experts to get certainties but receive uncertainties with their genetic consultation creates a bad match. This incongruence is further exacerbated when the intent of testing is to support health decision making.
Perceived risks

There are many studies showing that despite receiving genetic counselling and testing to correct individuals’ actual risk to an inherited mutation and risk to cancer that their perceived risks remained unchanged. A study at North York General Hospital and Toronto-Sunnybrook Regional Cancer Centre in Ontario evaluating the impact of positive test results for BRCA1 and BRCA2 mutations found that perceived risk of acquiring a second cancer among 18 affected individuals increased from 46% to 57% after receiving test results. As for 6 unaffected individuals, their perceived risk for cancer increased from 27.5% to 47.5%. These findings indicate affected and unaffected people who received positive results experienced an increased perception of cancer risk while Schwartz et al. (2002) study findings indicate that people within HBOC families who received negative results experienced a reduction of perceived risk. As well, those who perceive they are at increased risk of cancer because of their strong family history may experience similar stress as those who received confirmation of their mutation (Baum, Friedman, & Zakowski, 1997; Burgess & d'Agincourt-Canning, 2001). Perceived risk can also increase not only after receiving positive results but also following genetic counselling. Bish et al. (2002a) found that after genetic counselling, perception of risk for ovarian cancer increased among affected women from high-risk families. In addition, some women’s perceived risk for cancer increased while they were waiting for their results of testing. Changes in perceived risk for breast and ovarian cancer and to risk of carrying an inherited mutation have yet to be documented for individuals who received uninformative genetic test results.

Nursing implications of genetics

Genetic discoveries made within the last two decades have slowly begun to affect Canadian genetic nursing. Although still relatively small and not yet fully recognised, genetic
nursing is a growing health care specialty within Canadian nursing (Bottorff et al., 2004; Green, 2004). Following their interviews with 22 nurses in genetic services for adult-onset hereditary diseases, Bottorff et al. (2004) report that challenges for genetic nursing in Canada include role ambiguity, lack of recognition for nursing expertise, limited availability of genetics education, isolation, and the instability of nursing positions.

Because the first point of entry into the health care system for the general population is often through nurses, there is a need to provide educational support to nurses so that they can better assist individuals as they make sense of their experiences with genetic testing. While there is currently an international effort to develop education in genetics, there are as well many issues for leadership in education in genetics. Some of these issues are the increased need to incorporate genetic knowledge by all nurses; to convince administrators and faculty of the need to add more genetics content in nursing programs; and to support nurse leaders in genetics to disseminate the importance of genetics as an essential content for health care throughout the world (Feetham, 2001). At best, what is needed from nurses working in all settings is a basic understanding of genetics to know when referral to cancer genetics agencies may profit their patients. Lashley (2000) says, “Minimally you want to develop professionals who can think genetically when approaching a clinical situation or problem that on the surface may not appear to be genetic” (p. 797-798). The International Society of Nurses in Genetics advocates that nurses recognise the integral component of genomics within their role of promoting health and wellbeing (International Society of Nurses in Genetics, 2003). Responses from 975 Canadian nurses to a survey regarding their educational needs in the areas of genetic testing and adult-onset hereditary disease indicated that they perceived themselves to be inadequately prepared to provide genetic care (Bottorff et al., 2004). Educational needs include not only the psychosocial needs and
services related to genetic testing but also how to provide support in interpreting results, as well as the ethical, legal, and social issues associated with genetic testing (Pasacreta et al., 2002).

**Summary**

This chapter reviewed various implications of genetic testing for BRCA1 and BRCA2 mutations. Empirical studies about the impacts of genetic testing on people’s everyday lives are scarce, although more are beginning to appear. A better understanding of the impact of receiving uninformative genetic test results for BRCA1 and BRCA2 mutations is needed to counsel these individuals appropriately. In the chapters that follow, I endeavour to contribute to this lack of empirical knowledge.
CHAPTER THREE:  
Research Design and Implementation

My interest in nursing practice and the type of knowledge that is useful in guiding our practice led me to examine methodologies suited to the objectives of my research. Because my intent was not to look at how individuals’ discourses can be constrained or regulated by social processes beyond individual’s consciousness, I believed that traditional qualitative methodology such as that of critical social theory and grounded theory limited the exploratory nature needed to answer the research questions. I needed an approach that acknowledged human subjectivity and interpretation in people’s health and illness experiences.

Moreover, the complexity of clinical nursing requires that knowledge generated in nursing research be not only theoretical but also inform nursing practice. I, therefore, needed an approach that provided guidance towards producing practical nursing knowledge. Because of the interdisciplinary nature of nursing, nurses are often called upon to work in innovative, evolving clinical settings where little nursing knowledge exists to guide their practice. Genetics is one such domain, where the nurse’s role is becoming more visible and expanding (Thorne, 2001). Therefore, nursing research that informs practice in this area is greatly needed, especially knowledge grounded in context and in human subjectivity that has the potential to bring understanding to lived experiences.

The challenge of describing and interpreting shared realities such as health and illness experiences from the perspective of those who live them requires a flexible research process that does not constrain participants’ liberty to express their views, without being a priori categorised. A qualitative methodology that offers such a flexible process to study shared realities without
losing sight of the individual is interpretive description – a noncategorical qualitative alternative for developing nursing knowledge (Thorne et al., 1997).

The present qualitative inquiry, then, is guided by interpretive description. The concepts most commonly associated with interpretive description are inductive research, subjectivity of experience within common experiences of health and illness, and contextualising particular cases in an everyday-life perspective (Lowenberg, 1993; Thorne et al., 2004; Thorne et al., 1997).

Interpretive description has been used as an analytic framework by researchers and scholars who find they can use it to create practical nursing knowledge out of shared human experiences and unique individual experiences (Buissink-Smith & McIntosh, 1999; Irwin, Thorne, & Varcoe, 2002; Ramfelt, Severinsson, & Lutzen, 2002; Stajduhar et al., 2002).

In this chapter, I begin by presenting the analytical framework of interpretive description, explaining how this approach is distinctively useful to nursing inquiries. I then describe the general principles guiding my study. I begin by reporting on recruitment strategies and participants selection criteria. I go onto describe the data sources, commenting on interviewing and on field notes. I then outline the data analysis procedure used to arrive at my research findings. I conclude with a discussion of the standards and credibility measures I employed to establish the scientific integrity of the research process and its ethical conduct.

**Interpretive Description of Health and Illness Experiences**

**Theoretical framework**

Thorne et al. (1997) argue that interpretive description is a valid form of qualitative inquiry for understanding how people experience their health and illness, so that nursing practice can be adapted to better meet the needs of these patients. Just as traditional empirical methods such as quantitative studies prove limited when used in inquiries of human subjectivity, so do
qualitative approaches developed in other disciplines other than nursing. A noncategorical qualitative approach called *interpretive description* as been proposed as an alternative (Stajduhar et al., 2002; Thorne et al., 2004; Thorne et al., 1997).

Like the constructivist approach, the interpretive description approach recognises the contextual and constructed nature of health and illness experiences of individuals who come into contact with clinical settings and relates how clinical context can influence an individual’s subjective interpretation to their experience. The goal of interpretive description is not to produce new truths, but to identify common clinical phenomena that, within individuals’ contexts, bring about new understandings and new meanings (Thorne et al., 1997).

The philosophy of interpretive description shares perspectives with traditions of interpretive and naturalistic inquiries (Denzin, 1989; Lowenberg, 1993; Schwandt, 1994). Epistemological foundations underpinning interpretive description include the view that, because reality is multiple and constructed, it can only be studied holistically (Thorne et al., 2004). That is, in-depth understanding comes from studying the phenomena within multiple realities, not from viewing the phenomenon extracted from the reality in which it occurred. The knower and the would-be knower are inseparable; they interact to influence one another. Because no a priori

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21 While many qualitative approaches developed from different disciplines do not seem entirely different from other qualitative research methodologies, there are main differences within the choice of phenomenon studied. Grounded theory was created by Glaser and Strauss (Glaser & Strauss, 1967) and originated in sociology for the study of social processes. Interpretive description was created by Thorne, et al. (Thorne et al., 1997) and originated in nursing for the study of clinical phenomena. Whereas grounded theory has been used to study clinical phenomena in the past, its purpose differs from interpretive description. Grounded theory aims to generate a theory by creating a schema of the phenomenon through the development of concepts. Interpretive description aims to produce a synthesis of the main themes and patterns of a phenomenon studied. While interpretive description may still contribute to generating theories whereby analysts may test concepts derived from such studies to further bring refinement of plausible relationship between them, its purpose is not focused on theory development per se (Stewart, 1998). I chose interpretive description to guide my study as I believed that too little was known of the current study population "the uninformative group" to be able to pre-determine the value of generating a theory of the underlying social processes related to the experience of making sense of interpreting genetic testing results.
theory can encompass all the possible realities of a studied phenomenon, interpretive description encourages theory grounded in the data.

Researchers who choose interpretive description believe that no research is value-free; rather research is socially constructed, historically located and shaped by both the researcher’s and the participant’s beliefs and emotions (Denzin, 1996). Emotions and beliefs permeate all aspects of interpretive research: personal lived experiences, the home and family, health and illness experiences, and in power relations. Power is viewed as the ability to define what is legitimate. The aim of interpretive description is not to determine how these emotions, beliefs and power correlate with the phenomenon studied, but to acknowledge their influences on how an individual will experience a clinical procedure. Interpretive description focuses on how the studied clinical phenomenon occurs, how individuals’ interpret their clinical experiences, and how patterns are formed and altered throughout the making-sense experience. In interpretive description, the individuals studied are not perceived as objects of research but as collaborators in the research process. Consequently, unlike traditional post-positivist approaches to the building of knowledge, researchers working within an interpretive description approach engage in the subjectivity of participants to understand and express more effectively the emergent patterns (Denzin, 1989). This reflexive, engaging, dialectic approach to research is said to reduce this objectification of study participants by paying attention to power inequalities between knower and would-be knower (Guba & Lincoln, 1994).

Engaging also refers to the reflective process that I engaged in within the study. Being aware that I could not remove all power inequities between myself and the participants, I made explicit to participants at the outset of interviews as well as in our first contact by telephone that this research was part of my doctorate. I shared with them that I do not have a family history of
breast and/or ovarian cancer, nor have I shared this experience through someone close to me. I did not pretend to comprehend fully where they are coming from in interpreting and making sense of their genetic test results. I did share, however, my many years of reading on the subject, observing genetic counselling sessions, participating in clinical reviews of individual genetic testing cases, and participating in weekly discussions with a group of researchers conducting inquiries in the ethical, social, and legal implications of genetic testing. I tried to decrease power inequities by emphasising the importance of their experiences to my learning and, more importantly, to the improvement of genetic services for those coming after them. Concomitantly, I was still aware that complete sharing of power within the research process was impossible, as I defined the orientation of the interviews by making participants aware of the focus of my study and I made the final decisions of what would be considered valuable knowledge.

Research strategies in interpretive description draw upon work in grounded theory, naturalistic inquiry and ethnography, and on phenomenological approaches for data collection (Thorne et al., 2004). Such strategies include purposive and theoretical sampling that reflects an awareness of possible structures that may bring variations within the phenomenon studied. Data analysis begins during data collection and researchers locate emerging patterns within the data and compare them to existing knowledge on an ongoing basis. The end product of interpretive description is a synthesis of the main patterns and processes in the phenomenon studied. Individual stories are used to illustrate differences and similarities, thereby rendering individuals visible within identified patterns. This form of clinical knowledge development appeals to those working directly with patients because it provides insights on how to apply aggregate knowledge to individual cases (Thorne et al., 2004; Thorne et al., 1997).
The write-up of individual cases and common patterns is thick description and interpretations derived from individuals’ lived experiences (Denzin, 2001). Thick description, as opposed to thin description that simply states facts, provides the context of the phenomenon explored; uncovers meanings that organise experiences; and traces the phenomenon’s evolution. Thick description presents meanings and feelings as a text that can be interpreted (Denzin, 1989). The interpretive description approach recognises that lived experiences reflect the social world in which the individual lives. Interpretation aims to illuminate the meanings and concepts that organise a person’s experiences of health and illness (Denzin, 1989). This aim requires that the researcher be an informed reader of the phenomenon studied. That is, unlike phenomenology, interpretive description researchers locate their findings within existing knowledge, so that the new knowledge can be linked, and contrasted, to the work of others in the field (Thorne et al., 1997) and to the researcher’s prior understanding of the phenomenon (Denzin, 1989). In addition, Thorne et al. (1997) argue that an analytic framework constructed out of a sound critical analysis of existing knowledge builds a stable platform from which to launch a qualitative inquiry.

**Research Design**

All the above features of knowledge construction created the theoretical foundation for my methodological approach to studying how people interpret and make sense of their uninformative genetic testing results and how these results are lived in their everyday lives. This approach directed me to:

1. Design a preliminary interview schedule based on existing knowledge but leaving space to reformulate the initial interview schedule.

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22 See appendix 8 for a sample of initial analysis of key statements with thick description and interpretation.

23 My personal assumptions about the phenomenon studied at the outset of the study and my evolving understanding are injected throughout the report of research results in chapters 4 to 6.
2. Recognise the importance of thematic outliers as evolving selection criteria for study participants.

3. View participants’ experiences as permutated by past and present lived experiences, including family relations and interactions, and by subjectivity reflecting complex cognitive and emotional interactions.

4. Engage in the subjectivity and interpretation of participants’ narrative accounts so as to clarify my understanding of the lay theory developed by individuals to render their experience meaningful.

5. Construct knowledge in collaboration with participants by sharing with them patterns and themes in the data.

6. Seek out ways in which aggregate processes can further enhance understanding of individual cases.

7. Consider how clinical knowledge derived from the study can be applied in the development of nursing practice.

Implementing the Study

Having set out the methodological approach that guided my research design, I now turn to the methodology itself. I will also detail the interpretive lens that guided the research and analysis process as well as document my efforts to fully engage in inductive reasoning by reflecting on the research analysis and processes. In this study, the primary participants are individuals who had genetic testing for BRCA1 and BRCA2, while the secondary participants are family members chosen by the primary participants who did not have genetic testing (except for one). In accordance with interpretive description, in-depth interviews, field notes and clinical documents were my primary data sources. Transcription of interviews and field notes was
ongoing during the research process, to facilitate location of emergent themes within the findings.

**Recruitment of participants**

Primary participants were recruited for this study with the assistance of genetic counsellors and the educational nurse at the Hereditary Cancer Program of the BC Cancer Agency. Based on initial sampling criteria presented below, the counsellors and nurse made the initial contact with potential participants, who had already undergone genetic testing for BRCA1 and BRCA2 mutations, had been informed of their results through a telephone call from one of the health professionals from the Cancer Agency informing them that no mutation had been identified in their BRCA1 and BRCA2 cancer genes, and had received a letter informing them of these results. In this initial contact, the counsellors and nurse explained my study and sought their participation. If the person was interested to hear more about the study, they were asked for permission to release to me their names and telephone numbers. If they agreed, I telephoned them to describe further the goals of my study, what their involvement would be and seek their participation. If they voiced interest, I encouraged them to take time to consider participating. In the meantime, I sent them an information letter and an informed consent form that further outline the aims of the study. I told them that I would contact them a week following the receipt of the letter to determine their interest in taking part in the study. I also explained that they could refuse to participate or withdraw from the study at any time. If they agreed to participate, verbal consent was obtained one week following the receipt of the letters and written

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24 See Appendix 3: Letter of support for recruitment from the BCCA Hereditary Cancer Program.

25 See appendix 4 for a copy of the participation information and consent form. The information letter and consent form was sent to all participants via express post to maintain respectable timeline between voicing an interest in participating and in conducting the actual interview.
consent at the day of the interview. All interviews were conducted within 2 weeks of receiving verbal consent.

Potential recruitment of family members for the purpose of answering more in depth Research Question 2 was discussed with primary participants after they had received the letter of information and informed consent form. They were also told that they were not obliged to identify potential family members in order to participate in the study. I explained that a significant family member could be an individual who they felt supported them through their genetic testing experience or someone interested in their experience. If they could identify anyone, I asked them to make the initial contact, and seek permission for me to contact the person. Only at the interview did I obtain names and telephone numbers of potential secondary participants. Among primary participants who had identified their partner as their significant family member (n=3), all partners chose to be interviewed on the same day as the primary participant. As well, one primary participant brought her sister on the day of the interview, as she also wanted to participate. Written informed consent was obtained from the sister when I conducted a separate interview with her.

Of 23 potential participants contacted, only one declined while another asked to postpone her interview because she was receiving treatment for a recurrent breast cancer at the time. She asked that we keep in contact every week or so to see if she had enough energy to set a tentative interview date as she insisted on participating. I kept in contact with her for over 6 months. The last time I called, her husband shared with me that she had passed away from recurrent breast cancer but had been very sorry she had not been able to participate in my study.

I continued recruiting participants until I heard consistent repetition of themes in the interviews. As well, as I neared 20 interviews, I heard few new insights in participants’ stories.
That is not to say that their stories were completely similar, but that I was gaining very little new knowledge; hence, I felt that I had achieved thematic comprehensiveness. In interpretive description, small samples are considered ideal for inquiries of clinical phenomena where the purpose is to capture themes and patterns within participants’ subjective perceptions (Thorne et al., 2004). However, the sample must be large enough to portray participants’ experience well enough to inform clinical practice.

As for recruitment of secondary participants, I had intended to recruit an average of five family members per primary participant. In the end, I had an average of one secondary participant per primary participant and the maximum was three. The low number of family members per primary participants reflects an analytical decision I made during the course of data collection and analysis. Early in the data collection phase I realised that the family members’ accounts of interpreting and making sense of test results were quite different from the accounts of those who had actually lived the experience as the former could only anticipate how they might have lived such an experience. Hence, I no longer emphasised the need for primary participants to identify significant others unless they really felt that someone made a difference in their experience. I had also realised that the content of the secondary participants’ interviews contributed only minimally to the research and mostly to answer Research Question 2, assuming that their everyday lives of primary participants included interaction with significant others who helped them through their experience. Hence, I subsequently focused on the primary participant data and included secondary participants’ data in the analysis only when it added depth to the primary participants’ accounts.
Participant characteristics

The final research sample comprised 21 primary participants and 15 secondary participants. Table 1 gives baseline demographics of the 21 participants and table 2 summarises the relationships between family members interviewed and the study participants. The education levels of primary participants varied: 71% had at least a college education, while 29% had completed high school only. Of the former, 57% had completed at least one year of university level or obtained a full degree. All participants were women.

26 Since the main focus of analysis is the primary participants’ accounts of their experience of interpreting and making sense of their genetic test results, participants, refers only to primary participants. When input from secondary participants input is included, I specifically say so and address them as the family members interviewed.
Table 1: Demographic Profile of Primary Participants

<table>
<thead>
<tr>
<th>Sex</th>
<th>Birth place</th>
<th>No. of offspring</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female</td>
<td>Canadian-born</td>
<td>18</td>
</tr>
<tr>
<td>Male</td>
<td>European-born</td>
<td>3</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Age</th>
<th>No. of offspring</th>
</tr>
</thead>
<tbody>
<tr>
<td>41-50</td>
<td>None</td>
</tr>
<tr>
<td>51-60</td>
<td>One</td>
</tr>
<tr>
<td>61-70</td>
<td>Two</td>
</tr>
<tr>
<td>Older than 70</td>
<td>Three</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Age at breast/ovarian cancer</th>
<th>Marital status</th>
</tr>
</thead>
<tbody>
<tr>
<td>20-30</td>
<td>Single</td>
</tr>
<tr>
<td>31-40</td>
<td>Married/common law</td>
</tr>
<tr>
<td>41-50</td>
<td>Separated/divorced</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Level of education</th>
</tr>
</thead>
<tbody>
<tr>
<td>High school diploma</td>
</tr>
<tr>
<td>College diploma</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Employment Status</th>
</tr>
</thead>
<tbody>
<tr>
<td>At least one year of university</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Residence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Greater Vancouver</td>
</tr>
<tr>
<td>Fraser Valley</td>
</tr>
</tbody>
</table>

Note. Total number of participants (N) = 21.
3 participants had more than one primary breast cancer. *Greater Vancouver areas include participants from Vancouver, North Vancouver, West Vancouver, Coquitlam, and Langley. ‘Fraser Valley area include participants from Surrey, Port Coquitlam, White Rock, Richmond, Maple Ridge, and Delta.

Table 2: Relationship of Secondary Participants (SP) to Primary Participants (PP)

<table>
<thead>
<tr>
<th>Relationship</th>
<th>Specifics</th>
<th>Total</th>
<th>Identification of PP with SP</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spouse/Partner</td>
<td>Husband</td>
<td>3</td>
<td>SP1/PP15</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP16</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP20</td>
</tr>
<tr>
<td>Sibling</td>
<td>Sister</td>
<td>7</td>
<td>SP1/PP1</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP2</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP10</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP5</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP2/PP5</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP18</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP21</td>
</tr>
<tr>
<td>Child</td>
<td>Daughter</td>
<td>1</td>
<td>SP2/PP3</td>
</tr>
<tr>
<td>Extended family member</td>
<td>Female Cousin</td>
<td>3</td>
<td>SP1/PP3</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP2/PP5</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>SP1/PP22</td>
</tr>
<tr>
<td></td>
<td>Niece</td>
<td>1</td>
<td>SP2/PP20</td>
</tr>
</tbody>
</table>

Total number of secondary participants (family members interviewed) 15
In order to provide a clearer portrayal of those who participated in the study, I built a pedigree for each of the participants from information gathered during the interviews (see Appendix 5) as well as from the demographic data collected at the end of each interview.27 I refer to these pedigrees throughout the results chapter as a visual means to orient readers to the study findings. Permission to build pedigrees was sought from the participants. I told them that I would omit familial characteristics that might disclose their identity. Participants were quite enthusiastic about the idea and even offered their medical files to help build their pedigrees. However, wanting to be clear about my position, I explained to them that my role was one of researcher, not nurse, and that this did not give me permission to access their medical files. Some participants still associated me with the health professional from the BC Cancer Agency who had first contacted them on my behalf. Some commented that “I was the first they had heard from the Cancer Agency since they had been handed their genetic test results”. Even after when I clarified my position, many participants said how good it was for them to express how they had felt about their genetic test results.

Selection criteria

In this research, the first selection criterion consisted of participants who had undergone genetic testing for BRCA1 and BRCA2 mutations and received results considered uninformative or inconclusive by clinicians. Although the clinicians and health professionals working in the BC Cancer Agency call these results uninformative, they are also called inconclusive in some of the literature on genetic testing. One of my dissertation committee members made the comment that I should not assume that participants share the view that their results are uninformative. If my aim was to seek how they interpreted their results and made sense of them, I had to avoid introducing these results as the Cancer Agency defined them. Very few participants actually

27 See Appendix 10 for the demographic data collection form used to help build the pedigree.
referred to their results as uninformative or inconclusive, although many spoke about the uncertainty of their test results. Because I viewed the results as inconclusive in terms of their outcome, I struggled to stay true to the reflective process and power issues, such as not thinking that my perception was the most accurate when participants asked for my view of their results.

The line between engaging with knower and would-be knower is vague. That is, I was always wary that participants might adopt my interpretation of their results and how I made sense of them when my intent was to learn how they interpreted and made sense of their results. I do share the view that groups from different professional and social backgrounds tend to interpret testing differently. Whenever participants asked what my position was I answered that I was most interested in hearing their view.

My aim in selecting potential participants was to obtain as heterogeneous a group as possible and to include what may seem at first outliers, as seen in earlier times with older use of qualitative methods by nurse researcher trying to adhere to the dominant rules of normal sciences approaches that aim for their sample to be homogeneous (Thorne et al., 1997). Thorne et al. explain that excluding odd cases from an inquiry results in producing identical, quantifiable data that is devoid of almost all human subjectivity. Hence, the general principles of theoretical sampling are relevant to the design of research with an interpretive description approach when the purpose is building nursing knowledge.

The variables to purposive and theoretical sampling consisted of those identified within the genetic testing literature as having potential to influence interest in uptake and interpretation of genetic testing for breast and ovarian cancer susceptibility. In the course of the interview process, characteristics other than those identified a priori came to light, such as differences among individuals who came from large families compared with small families. Hence, toward
the end of the middle of the data collection, a few additional individuals from large families were
approached to participate in the study. I used the same procedure in establishing first contact as
outlined above in the Recruitment section. Writing field notes soon after each interview
facilitated my recognition of another variable influencing the studied phenomenon: having a
mother with a past diagnosis of breast and/or ovarian cancer. Although I had already identified
this variable in the literature review and selected it as a sampling variable, I had recruited only
two such women, among the potential participants. Hence, I selected a few more participants
meeting this criterion from the available candidate pool, numbering about 255.

Further variables that potentially influence participants' interpretation and making sense
experience of their results were identified through discussion with the genetic nurse at the Cancer
Agency, as well as the genetic counsellors. They recommended certain participants because they
had observed differences among their experiences of receiving and interpreting their genetic test
results. In our discussions, I also shared with them potential influential variables I had observed
in the genetic counselling sessions at the Cancer Agency (about 24 hours' worth). I also drew
potential influencing variables from the monthly clinical review meetings I attended in the
Hereditary Cancer Program between 1999 and mid-2002, when I then moved to a different
province. These meetings provided a forum for discussion of difficult and unusual cases.
Attending these meetings were medical geneticists, oncologists, pathologists, genetic
counsellors, nurse educators, a medical anthropologist, a bioethicist, and graduate students
interested in genetic testing. These meetings were chaired either by the director or the acting
director of the Hereditary Cancer Program. All of these professionals contributed to the case
management by making recommendations about treatment and follow-up care. These meetings
also allowed staff to share and discuss new knowledge on standards of genetics, clinical practice and ethical issues. The final variables for sample selection were:

- **Age**: Below and above 50 (Reason: Interest in and use of testing can differ between younger and older women);

- **Gender**: All women (Reason: Only one man in the available pool of candidate had received uninformative results);

- **Children**: With and without children (Reason: Common incentive for interest in genetic testing);

- **Family history**: Mother diagnosed with breast cancer and died as a result of the disease vs. mother still alive or died of cause other than breast cancer (Reason: Known to influence perception of risk for an inherited mutation);

- **Size of family**: Families of two or fewer siblings in two generations vs. more than two siblings in two generations, both from same side of the family where breast and/or ovarian cancer occurred (Reason: During dialectic data collection and analysis, it became apparent that differences occurred between the two, while their similarities brought about further distinctions to the phenomenon studied);

- **Education**: University vs. college vs. high school (Reason: Education level thought to influence individuals’ interpretation of testing by assuming that college to university people might have been introduced to some more complex concepts of genetics compared to those learned in high school);

- **Country of origin**: Canadian-born vs. not Canadian-born (Reason: Differences in ethnicity might bring about different beliefs and experiences of testing);
• Residence: Greater Vancouver vs. Fraser Valley (Reason: Proximity to testing site seen as possibly influencing testing experience);

• Marital status: Live-in spouse vs. single, divorced, or widowed (Reason: Influence of support system on genetic testing experience); and

• Year genetic testing was done: 1997, 1998, 1999 or 2000 (Reason: See how events unfold over time until receipt of test results in 2000. Note: Receipt of genetic testing results for BRCA1 began in 1997 while results of BRCA2 testing began in 1998. Data collection took place in the year 2000).

My aim in using sampling variables was to produce a guide for the genetic counsellors and the genetic nurse assisting me in recruitment. I aimed to have at least two individuals identified for each variable. Because the sample was being drawn from a large population (255), this goal was possible and attained.

Data sources

The main data source for this research was in-depth, open but focused and exploratory, interviews. Following each interview, I wrote field notes.28 The field notes were a valuable source of data to begin identifying key themes that serve to guide ongoing data collection, data analysis. My secondary sources of data were, as identified above, genetic counselling session observations, monthly meetings of the Hereditary Cancer Program group, and weekly meetings with interdisciplinary scholars working in the area of genetic testing.

I conducted all of the interviews with the participants face-to-face, in participants' homes or in other places of their choosing. Nearly all interviews with family members were also conducted in their homes. The exception was telephone interviews, when either the individual lived far from Vancouver or in the United States. Most participants mostly lived in locales

28 See appendix 6 for a sample of field notes made following interviews.
around Vancouver such as Langley, North Vancouver, West Vancouver, and Coquitlam. Others lived in Lower Mainland suburbs such as Surrey, White Rock, Delta, Abbotsford and Maple Ridge. The interviews with the participants ranged from an hour and a half to two hours while those with the family members ranged from half an hour to an hour. All were audiotaped (with consent) and transcribed. All transcribed interviews were transferred into QSR N5 software for qualitative analysis for easier management and retrieval of data. As well, a database was used to keep participants' record of contact, field notes, and easy access to personal journal. All participants were asked to choose a code name to safeguard their anonymity.

Many participants told me either on the first or second telephone contact that they were not sure if they had something valuable to share. Hence, at the interviews, I began by sharing the aim of the study to establish the direction of the interview. I then invited the participants to start by telling me about their breast cancer experience. With the family members, I asked that they start with how they felt about their family member's experience of genetic testing. Beeson (1997) indicates that storytelling by individuals who have lived a meaningful experience helps to create order, make sense of their lives, and construct action. Therefore, I welcomed first-person accounts as valuable data. The knowledge gained from the participants' storytelling added richness to my understanding of their experience.

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29 I transcribed half of the interviews, while the other half were transcribed by two self-employed transcriptionists. I chose to hire professionals as I felt that they probably had an ethical protocol for working with personal information from tapes. The issue of confidentiality was discussed with both transcriptionists.

30 See Appendix 11 for a reading of interview segments from a few participants.

31 See Appendix 12 for template of database recording.

32 See Appendix 2 showing preliminary interview schedule for both primary and secondary participants. The bolded content represents ongoing additions to the interview schedule as data collection progressed. Although I took these preliminary interview schedules to all interviews, I did not always look at them during the interview because I knew the content by heart and because I did not want to interrupt the flow of the participants' narrative. The very few individuals who expected to be asked questions in a formal, directive manner felt more comfortable when I gave them possible topics that could be discussed in the interview.
A few participants expected direct questions and did not seem comfortable starting with the storytelling. Hence, with them, I would start by asking that they tell me about their genetic testing experience and then how they interpreted their results. With the exception of a few participants, most felt more comfortable talking first about their breast cancer experience, then later about their experience with genetic testing. To the interviews, I brought my list of topics to cover and the prompts to use if necessary, although the way I used prompts – if at all – varied from one interview with another. Most participants needed little prompting.

Since data collection and data analysis were done iteratively, data collection took over a year. I spent time immersed in the data interspersed with time in the field. This iteration is a principle behind many qualitative approaches. The inquiry was constantly refined through this iteration, testing and challenging emerging conceptualisations of the studied phenomenon (Thorne et al., 1997). Through writing my field notes following each interview, I reflected on key and recurring points mentioned by the participants, as well as new questions I asked that brought new perspectives. This activity led me to question some of my initial constructions and explore new constructions in subsequent interviews. I found myself asking questions such as: What led this participant to respond in this way? What am I hearing and what am I not hearing? What is different and similar within the interviews conducted thus far? I marked these insights as outstanding questions in my field notes, to be either tested or negated by the next participants I met. This reflection allowed further clarification of emerging key themes: I asked participants to reflect on what these emerging themes meant within their lived, health and illness experiences. This strategy further contributed to the validity of the findings.

See appendix 7 for the list of prompts sent to the participants as part of the letter of invitation that accompanied the information sheet and informed consent form.
Data analysis

An interpretative description approach requires an inductive analytical interpretation to data analysis. While I believe that experienced qualitative researchers would not see the need for more guidance to data analysis in interpretive description, than that put forth by Thorne et al. (2004), I felt that my limited experience in qualitative data analysis required me to find more detailed guidelines for qualitative data analysis. Reviewing methods used in other qualitative approaches that share similar philosophical principles with interpretive description (ethnography, grounded theory and phenomenology, among others), I believed that interpretive interactionism (Denzin, 1989, 2001) provided further guidance in my search to find themes and build patterns and commonalities without losing sight of individual variations within them.

I was advised by experienced qualitative researchers to be cautious of any method that claimed to offer a recipe for data analysis because there is no such thing – qualitative data analysis tends to be reshaped by ongoing inductive reasoning. I also understood that qualitative data analysis can only be learned through experience and practice (Thorne et al., 2004). As such, although Denzin’s interpretive interactionism (Denzin, 1989; 2001) provided me with some guidance to organise my initial data analysis, it was only when I began to actually analyse data that I took full notice of the intellectual juggling between data collection, data analysis, and generating valuable, plausible and credible findings. Hence, the techniques I used to arrive at an inductive, analytical interpretation, were borrowed among those recommended by Denzin for interpretive interactionism (1989)\textsuperscript{34} and from Thorne et al. for interpretive description (2004). In interpretive interactionism, as in interpretive description, the end goal is to produce a meaningful

\textsuperscript{34} Some of the techniques to data analysis borrowed from Interpretive Interactionism (Denzin, 1989) are discussed in the next pages.
account of experiential knowledge. Thus, what I present below is my reflective, learned data analysis process to knowledge development of my studied phenomenon.

The purpose of thick description and interpretation in data analysis as put forth by Denzin (1989; 2001) and by Thorne et al. (2004) is to tease out the meanings and feelings present in participants’ experience. To begin this process, I first bracketed main themes within the participants’ stories that spoke directly to how they interpreted their results and the structures that informed their belief towards their interpretation of results. Key themes were mainly recurring points within the interviews or ideas participants emphasised while telling their stories during the interviews. These themes ranged from two to as many as 10 lines or more of transcribed-verbatim accounts.\(^{35}\) I deliberately did not select one or two words to represent evolving themes, to avoid constraining the analytical process within specific categories.

Because the study had as its main focus how affected women interpreted and made sense of their genetic test results, relevant segments of the transcripts were coded under such a category. However, this was not a means to limit my view of their experiences but for initial data management. The main analysis came later when all of the segments coded under this category were analysed for possible themes and structures that could help illuminate the meanings people gave to their experience.

These emergent key statements served my initial attempt to describe and interpret the meanings found in participants’ accounts. I asked myself, what are the intentions, feelings, beliefs of the participants within the key statements? To synthesise the participants’ accounts of their making-sense process, I looked for lay theories participants might have used to understand their experiences. Denzin (1989) says that lay theories are often found in participants’ stories. By looking for and analysing some of these lay theories, I was able to identify more concepts that

\(^{35}\) See Appendix 8 for a few examples of key statement segment.
structured their interpretation and making-sense experiences. One example of such a lay theory is how one participant explained to me that, within each passing generation, breast cancer diagnoses seemed to be occurring 10 years earlier in family members with each passing generations. She felt that this pattern was too prominent to be just coincidental or the result of chance and that there were too many similarities between the ages at which individuals were diagnosed with breast cancer to also be the result of chance. This lay theory grew out of her family history with the disease and her belief that she may be carrying an inherited mutation despite being told that no mutation was found.

The dialectic approach facilitated my seeing initial differences and similarities in the participants’ experiences. For example, I noticed that some differences depended on personal familial characteristics. Participants who came from large families and/or also who had a mother who had been diagnosed with breast and/or ovarian cancer tended to attach different meanings to some of the emergent themes influencing how they interpreted and made sense of their genetic test results.

Once I located themes and structures within each participant’s story, I formulated an initial description and interpretation of their meaning based on the context of each individual.36 I then looked at the essential themes as a whole to begin understanding possible processes and patterns in participants’ experiences and what was meaningful to them. Thus, I came to see a chronology to the participants’ experience. That is, participants made it clear to me that they had not just “arrived at the decision to have genetic testing overnight,” but that there are factors that led them to become open to this new science. One of the major factors is the implications to living with a family history of breast and/or ovarian cancer. Then, before they consider genetic

36 See Appendix 8 for examples of initial analysis of key statements and the formulation of their description and interpretation.
counselling and obtaining genetic testing, there is a search for answers to the question "Why would I want such a test?" This first phase is in chapter 4. When they finish this first phase by deciding to get tested, they enter into the experience of genetic testing. This second chronological phase is represented in chapter 5.

Perhaps, having had the opportunity to talk about how they felt with their genetic testing experience and how they made sense of their results, all the participants reflected on the "so what?" question: What was this all worth to me? Even without prompting from me, their stories moved to this issue, which happened to be my second research question: How do their results affect their everyday lives, health and illness experiences? This third phase is represented in chapter 6.

When I began to look at the participants' experience as a whole through the lens of main themes and structures identified, I focused on attempting to classify and order them as they occurred, regardless of which participant's experience the structures belong to. This process is known in interpretive interactionism as the construction phase (Denzin, 1989). In the construction stage of data analysis, I often asked questions such as "What am I seeing here? Is there any order to all of these structures? Is there a beginning, middle, and end? What do these themes mean in participants' lives? And why am I not seeing something I expected to see? When confronted with two opposing views among participants, I asked "How does this occur? What situation brings people to have such an opposing view? How is it that I see such a pattern among these individuals but not among others?"

With the answers to these questions, I was able to derive how key themes and structures affected each other and show how they cohere to form patterns and processes.\(^{37}\) When I observed

\(^{37}\) See appendix 9 for a sample of ongoing data analysis used throughout the study to assist in constructing patterns and processes.
that there was absence of some of the key themes and structures in a personal participant’s story, I contrasted the participant’s family and personal lived experiences with other participants’ stories who presented with these key themes and structures, thereby showing how lived experiences can shape and alter patterns, processes, and structures within the aggregate experience of interpreting and making sense of uninformative genetic test results. This method also brought to light what is particular to the lived experience of an individual person (Thorne et al., 1997). This final contextualising stage puts the phenomenon studied back into the social world where it occurred and where the structures identified take full meaning (Denzin, 1989, 2001). Within interpretive description approach, the dialect activity of comparing and contrasting individual cases and patterns with the aggregate experiences of all participants forms the basis of its strength: “a respect for knowledge about aggregates in a manner that does not render the individual case invisible” (Thorne et al., 1997, p. 171).

Contrasting stories and discovering how lived experiences shape patterns and processes produces valuable clinical knowledge. Nurses can then see how aggregate knowledge can be applied to individual cases. This is a form of knowledge nurses have long demanded, one that facilitates their everyday professional practice (Thorne et al., 1997).

**Standards and credibility measures**

As understood by Crotty (1998), establishing the *credibility* of our research requires that we report our research process. This permits the reader to judge the trustworthiness of our findings and decide what she or he can retain from them. Further, Thorne et al. (2004) state that what is most important in establishing the credibility measures of our findings is not so much presenting “a litany of attributes such as trustworthiness, transferability or making claims about one’s integrity (such as reporting an ‘audit trail’)” (p.15) but rather presenting our analytic
decisions and showing how these are contextualised within the final study. I have described my
data analysis procedures above, and I have given examples of it. More examples are presented in
chapters 4 to 6.

As with other qualitative research, rigour is a critical component of interpretive
description, as research designs often evolve with ongoing data collection and analysis. Another
component critical to interpretive description is explicit accounting of researcher’s biases that
may influence our study. Although completely setting aside our biases is naïve (Thorne et al.,
1997), as we can never remove ourselves completely from the research process, I will share one
personal bias that I struggled with during the conduct of this research. One of my biases is a
belief that we, as health professionals, often interpret clinical tests and results differently than our
patients do. Consequently, although I interpret the genetic test results that participants of this
study received as inconclusive (meaning that no conclusion can be drawn from their results), in
the interviews I referred to their results just as “your genetic testing results.” I tried not to
influence their interpretations of the results by not showing approval or disapproval of their
interpretations, even if at times the participants’ interpretation seemed unrealistic.

Validity of findings in interpretive description is defined in their potential to create
mental heuristics that confirm hunches of expert clinicians from the field studied where they
would see new understandings of their reflective practice observations (Thorne et al., 2004;
Thorne et al., 1997). Hence, I sought feedback from expert clinicians within genetics throughout
my data analysis in peer-reviewed scientific presentations and nursing rounds at two different
universities. The feedback I received encouraged me to refine my data analysis, as some of my
interpreted structures were either challenged for their content or questioned for their clarity by
audiences at my presentations.
Additionally, the dialectic approach in interpretive description builds in a credibility check that is more productive in ensuring validity than taking data excerpts back to study participants. Thorne et al. (1997) recommend taking key statements in the form of interpreted data back to participants where such an approach can produce important insights when individuals reflect on why such key statements do or do not quite represent their view. To give an example within the current study, a few participants had all said something I found rather unusual: “Those who test positive for an inherited mutation are the lucky ones.” This contradicted some of the literature, in which receiving a positive test result for a mutation of BRCA1 and/or BRCA2 is perceived as stressful (Frost et al., 2004; Tercyak et al., 2001). However, this statement could also refer to viewing positive testing as eliminating uncertainty about one’s genetic status (Lerman et al., 1996). So, I brought this key statement to subsequent interviews for the participants’ critical consideration. Some said “Well it never occurred to me to think of them as the lucky ones, but I certainly do understand where this might come from, as…” and then apply their reflection to their own experience with genetic testing.

Field notes and journalling also facilitated the evolution of my reflection during data analysis and led me to question my early assumptions about participants’ experiences. In the field notes, I recorded situations, people, and places being observed, as well as my general demeanour on interview days. I also would write postinterview comments and observations in my journal, which I used to reflect on the research process. The first few journal entries focussed on my comfort with conducting open interviews. I also enjoyed having a place where I could reveal my inner thoughts on the research process. The last journal entries focussed more on struggles with my analytic framework and their resolution. Although I viewed this journal as a private, safe haven as I reflected on my integrity within the research process, I also realised that
this journal was another means by which others can judge the credibility of my findings. Hence, I reluctantly share two entries in my journal: one made following my third interview, the second following the completion of all the 36 interviews.

Today I interviewed my third participant. We were just the two of us. Again, I was surprisingly amazed at my qualities in conducting open unstructured interviews, as this is how I truly see them. However, as I was congratulating myself, I wondered what my committee would think. Would they think differently if they'd been in the interview with me? Would they think differently if they heard my responses and questions on the tape? This led me to wonder if I should do the interview having in mind my committee. In the first instance, I thought it would be good idea, as I do have to be credible to myself and to them at the same time. So, I should be comfortable with the fact that they would be listening to my every word. Well, that is just the problem. I would become so conscious that I am being evaluated and heard on every word I said that I would lose track of the interview process and probably lose the easy, comfortable feeling I have in interviewing the participants. The participants would probably feel my tension and perhaps start becoming conscious of the tape recorder and the fact that we are not really having a discussion but more, as Mishler (1986) would say, a traditional stimulus-response interview. That is totally what I am trying to avoid. Therefore, I will keep doing my interviews with the mindset that I have been doing them. That is, as if we are having a conversation, with them sharing with me a part of their lives. (Research journal entry, December 18, 2000)

The focus of the second research journal entry is on the analysis process. I am questioning the orientation analysis of my secondary participants:
I have done a fair amount of writing of my findings from key statements, although much more interpretive description needs to be brought out for result chapters. I have prepared a presentation for next Monday for the Sigma Theta Tau conference, and it has helped me to synthesise further my results and see more clearly growing patterns and processes, such as the chronology of their experience and the concept of time. Today I wanted to take a break from the participants’ analysis and go to the family members’ analysis. Somehow I suddenly felt blocked about how to code and analyse the transcripts. I have read what I wrote previously using the reflection framework I followed with the primary participants, but I am not sure how to proceed with the family members/secondary participants. Their experience seems both to differ from the participants’ and not to follow the focus of the study. With the family members and the primary participants I used the same research questions, because I assumed that this experience is lived in relation with others, not in isolation so I wanted to have the family members’ perspectives. But, in the end, I realise now that this is not the case and that the aim of this study is not to compare the two populations. Or should I be thinking about that?

(Research journal entry, October 31, 2002)

Following this entry, I went back to my dissertation committee to seek their advice. I explained that, although the family members’ stories added to the second research question (where the focus is on exploring how the participants’ experience of genetic testing affected their everyday lives, health and illness experiences), because the family members had not actually lived the experience of genetic testing, it became difficult to integrate experiential knowledge of this experience. My committee and I decided that, from this point on, the family members’ accounts would only be added when it contributed to the participants’ accounts in answering the
second research question. As theorised by Thorne et al. (2004), “If the findings look too similar to the analytic framework with which one entered the study, they may reflect the mind’s capacity to ‘fit’ data rather than to ask good questions and generate useful conceptualizations” (p.10). This experience made me realize more fully that qualitative research must remain open to change through an iterative, reflective, critical and analytical process (Morse & Field, 1995).

Throughout the data collection and analysis processes, I continually examined my preliminary beliefs and assumptions about the studied phenomenon in relation to the interpretations and decisions I made throughout the research process. This ongoing reflection forced me to reflect on my own position in relation to the research (Hall & Stevens, 1991) – an added component towards achieving scientific credibility (Lather, 1991).

I have already presented examples of how I modified the analytic process, but now I would like to reflect on my assumptions when I entered the study. Although largely influenced by my reading on genetic testing for breast and ovarian cancer susceptibility, I originally viewed genetic testing as a tool in breast and ovarian cancer screening for individuals identified at high-risk of developing the disease. However, in the course of the study, I came to realise the slippery slope in this assumption: providing individuals with their genetic mutation status, that these individuals were being offered the same treatments as high risk individuals already diagnosed with the disease (d'Agincourt-Canning, 2003). Further, I had not realised that the availability of this testing might also create a new pool of individuals categorised as at risk for cancer or the “at-risk health status” (Bottorff et al., 1999) and bring about the possible psychological harm that may result with these labels.

I also assumed that individuals from high-risk cancer families would be automatically interested in genetic testing and that they would see its availability as an opportunity to be
screened more precisely on their own cancer risk and that of others in their family. Perhaps my position as a nurse influenced the latter assumption, in that I have relied on clinical tests to assist me at arriving at clinical care decisions, either in relation to my own care, my family’s care or, most often, the care I provided to the many patients I nursed over the years. I have come to trust and rely heavily on clinical tests and their findings and just assumed that, if a new test came along that could convey additional information about one’s disease risk, that people would be interested in obtaining this test. Although this assumption was rarely challenged in the interviews, what the participants did say was, “How can you offer a test when you can’t offer a definitive result?”

In addition, I assumed that individuals who received a genetic test would automatically want to tell as many family members as possible they were having this test. It became impossible for me to maintain this assumption without sharing it with the participants, in light of what I was hearing from them. The majority had decided not to tell their family they were having the test before obtaining their results because, as they explained, “Why worry others when we don’t know the outcome of the results?” When I questioned this view, I came to learn from the participants that genetic testing can play a very small part in their everyday lives compared to more stressful events such as their breast cancer experience. I realised then that I could not discuss how they interpreted and made sense of their results without exploring what it meant to live with a personal and family history of breast and/or ovarian cancer.

Thus, the interviews helped me to redefine my initial assumptions and guided me toward new areas of inquiry that I had not previously intended to explore, such as looking at how the individual made sense of their genetic testing results in light of the implications of living with a family history of breast and/or ovarian cancer. The interviews also gave me an appreciation of
the complexities of receiving uninformative genetic testing results and the complex meanings generated by living with a family history of cancer, both of which I have not personally experienced.

Now, I would like to briefly reflect on the choices I made in the lines of inquiry I pursued with the participants which, in the end, affected the knowledge I produced (Whittaker in d'Agincourt-Canning, 2003). Seeing that there is a paucity of studies on the population studied in this research, I felt that the immediate clinical knowledge need was for understanding how these individuals interpret and make sense of their specific type of genetic test results, as opposed to focusing on the communication of risk. Although I do see the latter as important within the context of genetic testing because of its inherent complexity, I felt that such a topic would require the full focus of a study. Thus, while communicating risk and interpreting risk values are important aspects to study in genetic testing, I do not feel that I can do justice to their inherent complexities even though some issues relating to these two topics are interwoven within the findings of my research.

**Ethical considerations**

In the previous section, I discussed the standards and credibility measures applied in this study that were critical to the integrity of the research. In this section, I address responsibilities of the researcher to protect study participants – also known as responsible research conduct (Steneck, 2002). While protection of study participants cannot rest solely on the integrity of the researcher, clearance to conduct this study was obtained by the Behavioural Research Ethics Board at the University of British Columbia, and from the BC Cancer Agency.\(^{39}\)

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\(^{38}\) For references to studies looking at risk communication in the context of genetic testing for BRCA1 and BRCA2 mutations, see Bottorff et al., 1998; Bottorff, Ratner, Johnson, & McCullum, 1999; Croyle & Lerman, 1999; Edwards et al., 2003; Lipkus & Hollands, 1999; Lynch et al., 1997; and Rimer, 1999.

\(^{39}\) See appendix 3 for a copy of the letter of support received from the BC Cancer Agency.
The potential exists that people’s lives may be reshaped as a result of the researcher entering the lives of individuals by asking them to share their stories. Because researchers enter into these relationships with specific aims and goals that are independent of participant benefit, the primary responsibility of researchers should be avoiding harm to participants (Clandinin & Connelly, 1994). Responsibilities I took to heart were: ensuring informed consent throughout the research process, maintaining privacy (protecting the identities of participants), and protection from harm (physical, emotional, or any other kind) (Fontana & Frey, 1994).

Within this study, ways in which I tried to make the participants’ experience positive included respecting as much as possible the time limit for the interviews as written on their consent form. As well, I made it a point to only discuss the topics outlined in the cover letter sent with their participation information sheet and consent form. Therefore, I did not question them about other areas of their lives unrelated to the focus of the study. Finally, as a token of my appreciation for their time and their participation in the study, at the end of each interview, I offered all participants both a small scented candle and a three-piece Lindor chocolate box. As well, the day following the interview I sent all participants a thank you card. I also sent them all a Christmas card in December 2002 updating them on my progress, since I had told them that I would send them a 10-page written summary of my findings once I defended the dissertation. I explained in the card that my writing phase took longer than expected because of maternity leave and family obligations, but that I had not forgotten my commitment to them. Subsequently, I received a Christmas card from a secondary participant who wished me well and provided me with an update on her family’s newest diagnosis of cancer.

40 See appendix 7 for a copy of the accompanying cover letter. Its content represents a synthesis of the interview schedule I brought to each interview.
With the aim of minimising any possible harm of the participants having engaged in my research, before asking a question I constantly asked myself, would this question cause pain to the participant? If so, does their protection from harm preclude asking this question? I always emphasised with participants that their comfort was always more important than my need to obtain information. I wanted them to feel free not to discuss topics that they were not ready to discuss with a stranger. As an example, during one interview, discussion moved toward the diagnosis of a participant’s mother with breast cancer. Tears started rolling down her face. I immediately stopped the tape and told her that if it was too uncomfortable a topic that I did not need to hear about this experience to understand how she made sense of her results. The participant agreed to change the topic and shared how she had not yet discussed with anyone the death of her mother following her cancer diagnosis. Although I remember thinking at the time that hearing about this experience would have benefited my overall view of the emerging process within my phenomenon studied, I did not want to put the dissertation needs ahead of the participant’s comfort.

Maintaining confidentiality was another important ethical concern I tried to maintain throughout the study. Because of the nature of the research framework, whereby I interviewed different people within the same family, I found myself at times being questioned about what some family members had said, knowing that the other person’s response was not what the interviewee expected. For instance, one participant said to me, “I am sure my sister must have told you about our mom?”, yet the sister’s view of their mother’s experience with breast cancer was not the same. This event propelled me to be even more cautious about keeping all participants’ interview data anonymous. Some strategies employed were fictionalising identifiable familial characteristics (Fontana & Frey, 1994) such as asking participants to choose
a code name that only they could recognise, hence some of the code names are unusual.

Throughout the entire project, I remained highly aware of the ethical issues that may arise as a result of having entered into a researcher-participant relationship.

Consent for me to contact potential participants was given during a telephone call from either a genetic counsellor or the nurse at the Hereditary Cancer Program of the BC Cancer Agency. For those who expressed an interest in participating in the study, a letter of information and a consent form were sent prior to the interviews but following our first initial telephone contact. The information and consent form specified that the student researcher would be contacting them to find out if they were still interested in participating; it specified the estimated length of the interview, and asked that the interview take place in the participant’s home or in an alternate location if desired. The letter also asked that the participant pass a copy of the letter of information to other family members who might be interested in participating in the study. The consent form included a statement regarding risks such as becoming upset as a result of having discussed some topics. Participants were told that they were always free to contact the genetic counsellors and the genetic nurse at the Cancer Agency to discuss ongoing issues in relation to their experience with genetic testing.

Participants were also told at the beginning of the interview that they could refuse to answer any question they judged to be too upsetting and that they could ask me to erase any part of the tape recording if they felt that disclosing any information made them uncomfortable. None of the participants requested this. Participants were also told that their participation was voluntary, that they were free to stop the interview at any time, and that they could choose to withdraw from the study without jeopardising their health care or access to genetic services.

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41 See appendix 4 for a copy of the participant’s information and consent form.
While I obtained written consent at the beginning of the interviews, I also recognised that consent is ongoing throughout the interview as well as throughout the research process.

The right of participants to privacy was further protected by presenting only selected data to my dissertation committee members that would not compromise participants’ confidentiality. Interview transcripts were kept in secure computer files protected by a password and all identifying information was stored separately from the transcripts. Paper files of participants were stored without using their names and instead used numbers such as primary participant 1 (PP1) and secondary participant 3 (SP3) of primary participant 1 (PP1) that became SP3/PP1. Biographical information was modified as necessary to conceal possible identifying characteristics of the participants and will continued to be so in future published and unpublished work. The data were not destroyed once the transcripts were completed as I obtained consent from the participants to retain the data to permit future secondary analysis.

Summary

In this chapter, I presented the rationale for use of interpretive description by nursing researchers studying health and illness experiences. I also described some of the reconceptualisation that occurred in the research process as a result of its interactive nature. I outlined the research methodology and the methods employed to gather and analyse data. I addressed standards and measures used to establish credibility of research findings and described ways in which I attended to ethics. I now turn to chapters 4, 5 and 6, in which the findings of the research are presented.

42 Hence my use of breast and/or ovarian cancer when speaking of all participants’ history with cancer as opposed to saying that a particular individual only had breast cancer or ovarian cancer in their family history. Without this condition, I believe that maintaining anonymity becomes perilous.
CHAPTER FOUR:  
The Buildup to Genetic Testing

When I began my study seeking how individuals and family members interpret and make sense of uninformative genetic test results, I asked individuals to describe their personal experiences with breast and/or ovarian cancer and the experience of others in their families. Not until I did a few interviews did I take full notice of the implications of such a starting point in understanding what would lead individuals to be receptive to genetic testing. Just as Hallowell et al. (2004) found when they looked at the responses to uninformative genetic test results for BRCA1 and BRCA2 mutations among affected individuals with a family history to hereditary breast and/or ovarian cancer (HBOC), it is impossible to discuss individuals’ interpretations of such genetic test results without first providing the context of these responses.

Thus, I begin this chapter by describing how having a family history of risk for breast and/or ovarian cancer brings about certain discourses that you would not normally see in other families. In the first section, I present implications of these found in the participants’ accounts that either facilitated or brought contention to their interpretation and the process of making sense of their uninformative genetic test results. In the second section, I present how genetic testing entered participants’ lives and the reasons that led them to undergo genetic testing. In the final section, I build a foundation for exploring how participants interpreted and made sense of their genetic test results, the subject of chapter 5.

Implications of Living with a Family History of Breast and/or Ovarian Cancer

Many of the participants explained how they looked to genetic testing to explain their breast cancer diagnosis and their past family history of the disease. Before they went on to explain how they made sense of their genetic test results, all participants except one started by
sharing with me what it had been like for them to live with a diagnosis of breast and/or ovarian cancer and, more specifically, what it had been like to live with a family history of cancer. Their journey of learning to live with the disease led many to seek genetic testing as a way of finally answering why breast and/or ovarian cancer was so present in their family. Living with a family history of a disease deepened their struggles to make sense of uninformative genetic test results.

Before presenting the implications of living with a family history of breast and/or ovarian cancer as described by the participants of this study, I first present a pedigree of what constitutes having a family history considered at risk for HBOC and look at how one study participant met eligibility criteria for genetic testing. Although eligibility criteria vary among cancer programs, hereditary breast cancer susceptibility is usually characterized by early onset, cancer occurring in both breasts, multiple primary cancers; and in conjunction with other cancers (Ormiston, 1996).

First, I review the eligibility criteria for testing presented in Table 3 that study participants met, and then follow with an example of a pedigree, built from a participant’s account of her family history.

43 Although very few participants described their results as uninformative or inconclusive, mostly all participants except four viewed their test results as uninformative or inconclusive as defined in chapter 1. That is, because of their personal and family history with cancer, that it was impossible to confirm that they do not carry an inherited mutation or to conclude that their cancer may have occurred by chance.
Table 3: Eligibility Criteria for Hereditary Breast and Ovarian Cancer Risk Assessment

Genetic risk is assessed appropriate if:

A family history meets one of the following; OR A family history that includes at least two of the following on the same side of the family:

- A woman with breast cancer diagnosed at age 35 or younger OR
- A woman with ovarian cancer diagnosed at age 50 or younger OR
- An Ashkenazi Jewish woman with breast or ovarian cancer OR
- Have a blood relative with a confirmed BRCA1 or BRCA2 mutations

- Cancer in 2 or more closely related family members (i.e., parents, siblings, children, grandparents, aunts, uncles)
- Cancer diagnoses at younger ages than expected in the general population (i.e. premenopausal breast cancer) OR
- A woman with ovarian cancer diagnosed at age 50 or younger OR
- Multiple primary cancers in one individual OR
- A male with a breast cancer diagnosis

Note. The eligibility criteria presented here are from the Hereditary Cancer Program, BC Cancer Agency (BCCA) and represents those used within the year the study was conducted. Reprinted with permission from the BCCA.
Starting from Table 3 and looking at Sherry’s \textsuperscript{44} family pedigree, Sherry had to meet at least one criterion from the left column or at least two criteria from the right column. Sherry met the first criterion: in the left column. Sherry was diagnosed with breast cancer at the age of 35. Her family history also met other criteria from the left column. Her niece was diagnosed with ovarian cancer at the age of 34. As for the last two criteria, Sherry was not an Ashkenazi Jew nor does she have in her family an individual with a confirmed BRCA1 or BRCA2 gene mutations. Nonetheless, she meets the criteria for testing under this category by having received a breast cancer diagnosis at the age of 35. Looking at the right column in Table 3, Sherry’s family history met three of the four criteria – some of them more than once – while only two are necessary for eligibility. For the criterion breast and/or ovarian cancer in at least two closely related women on one side of the family, Sherry’s family history meets this criterion in many ways. That is, both

\textsuperscript{44} All first names presented in the dissertation represent code name chosen by participants.
she and her sister had a diagnosis of breast cancer, as did her mother. Sherry's two nieces who are siblings were each diagnosed with cancer, one with breast and the other with ovarian.

As for the second family history criterion, cancer diagnoses at younger ages than expected in the general population (i.e. premenopausal breast cancer), Sherry's family history again meets this criterion more than once. Sherry was diagnosed with breast cancer when she was premenopausal, as were her two nieces. While Sherry's sister was diagnosed at the age of 50, Sherry said during the interview that her sister was premenopausal at the time she was diagnosed. The third criterion is having at least one individual with multiple primary cancers. Sherry's sister received two primary breast cancers, one cancer occurring at the age of 50 and the other at the age of 63. As well, Sherry's other sister, a family member interviewed in this study identified as SP on the pedigree, sent me a letter in January 2003 saying that Sherry had been diagnosed with lung cancer, believed to be another primary cancer. I remembered Sherry saying in her interview that she had always been a healthy person, jogged regularly and had never smoked a cigarette in her life. The fourth criterion is having a case of male breast cancer in the family. Sherry's family has none.

Sherry was told that no mutation had been found in the two breast and ovarian cancer genes tested for but because of her personal and strong family history alone, the clinic could not conclude with certainty that there was no inherited mutation and that the family should continue screening as previously recommended. These conflicting results have been a challenge to make sense of for Sherry, in light of what she has come to live with in her everyday life.

In the next section, having illustrated what a family history considered at risk for HBOC can look like, I now discuss the themes derived from participants' interviews about the implications of living with a family history of breast and/or ovarian cancer.
**Expecting breast cancer to appear at some point.**

Individuals with a family history of breast and/or ovarian cancer and who have been in close contact with family members who have developed the disease often speak of constant thoughts that the disease must be inherited. A study by Iglehart et al. (1998) showed that affected women with a family history tend to overestimate their risk of carrying hereditary factors for breast and/or ovarian cancer. However, the authors explain that such feelings may come from prolonged care for someone significant who had breast cancer, such as a mother. As a result, individuals tend to internalise their risk to cancer from embodied experiences of others to cancer. For many of the participants, this has been their reality. One participant, whose mother developed breast cancer while still a preteen, described the experience as follows: “I basically prepared myself to have cancer all those years. It was like I knew that probably at some point I would have cancer.” As described by Chalmers, Thomson and Degner (1996), perception of risk for cancer can be intensified by the breast cancer experience of others within the family. This experience has been termed *empathetic* (d'Agincourt-Canning, 2001; 2003), referring to the subjective knowledge derived from personal, close experience with others in the family who experience cancer.

**Increased awareness of risk for breast and/or ovarian cancer in self and others.**

This increased awareness of risk to breast cancer in self and others brings about different ways of being. For example, one participant spoke of her need to always be on the “lookout” for breast cancer, while another participant spoke of being “attuned” to verbal and written information about breast cancer. She explained that she did this for her two sisters and herself. As a result of her breast cancer diagnosis, she found fear of a second primary cancer or a recurrence difficult to let go of.
For many of the participants with multiple cases of breast and/or ovarian cancer in their family, dealing with some family members being diagnosed with more than one cancer, or a recurrence, was not unusual. This brought about additional concerns: Perhaps they as well would have to live through a recurrence or another cancer. Just as Vickberg (2003) concluded, it was the younger participants in this study who often feared a recurrence. Many of the participants looked to their family history to assess their possible risk for cancer and that of others in the family. This has been suggested by Richards (1996) as a way lay people explain inheritance. Other lay theories include physical characteristics and personality traits. In other words, if you resemble a family member with cancer, you may inherit the disease.

I found in this study that participants not only use notions of physical characteristics and personality traits to establish their risk for another cancer or a recurrence but also use logical concepts of pathophysiology to explain their perceived risk for cancer. Participants whose mothers' breast cancer recurred definitely thought that “perhaps cancer was still lurking inside of them.” However, according to the Jobsen, Meerwaldt and van der Palen (2000) prospective cohort study, a family history of cancer is not predictive in cancer recurrence among women older than 40.

Nonetheless, participants have learned to live with a perceived risk of recurrence. With the disease being part of their family history, participants with children (especially daughters) commented that they also had to live with the thought that perhaps one day their daughters too would be diagnosed with breast cancer, just like them. One participant, Donna, even said that her daughter does not fear cancer because she has come to accept that it will happen to her. Here is Donna sharing a conversation she had with her daughter about the daughter’s view of her risk of developing breast cancer:

45 Recall that all 21 participants have been affected by breast or ovarian cancer.
My daughter said, ‘I know what happened to my grandmother. I know what happened to you. I expect it to happen to me but I don’t really want to think about that now.’ My daughter has kind of accepted that this is something that’s going to happen. She’s just waiting for the other shoe to drop. She’s a very strong young woman, but she’s also a very pragmatic young woman. And so I think, for her, and I think I can speak for her cousins too, that...I mean they’ve seen this in three very close family members and they just expect it. It’s going to happen. (Donna, PP14, field conversation, September 7, 2001 speaking of her 20 year old daughter)

A second participant also contemplated what it must be like for her daughter to live with the notion that her mother, her grandmother and her great grandmother were all diagnosed with breast cancer:

My daughter is 20 years old now. I just can’t imagine what it must be like for her now to be fourth generation, and knowing that all women in the past three generation have had it. I grew up with that so I can be kind of cavalier about it, of course I am going get cancer, right. But in my heart I could hope that I wasn’t. But by now not having found the mutation, I hope that the cycle will stop with me. (Stephanie, PP12, field conversation, July 18, 2001)

Both of the above participants held fatalistic views that, because their mothers had breast cancer and they had breast cancer, it is almost inevitable that their daughters too will one day be diagnosed with it. With many of the individuals within the study there exists a burden of anticipation that seems internalised by the family’s history of cancer, which then influences the perceived risk of cancer for themselves and others. Burden of anticipation based on the family legacy of breast cancer was also documented in d’Agincourt-Canning’s study (2003), where she also found an anticipation of cancer among individuals at high-risk for breast and/or ovarian cancer who underwent genetic testing. However, fear of recurrence has also been documented among individuals with or without a strong family history of breast and/or ovarian cancer as a result of a cancer diagnosis (Gil et al., 2004; LaTour, 2002; Vickberg, 2003). These studies found that illness uncertainty continued long after cancer diagnosis and treatment. The most frequent trigger of this fear was hearing about someone’s else cancer (Gil et al., 2004). As explained by some participants in this study, “Hearing of someone else’s recent cancer diagnosis
within the family is not an unusual event” and therefore, participants have come to internalise that no one is immune to cancer, even if they have already been diagnosed with one. Whether or not cancer appearing in the participants’ families is a true reflection of them having an inherited mutation or the result of sporadic cancers, this is the reality that participants are living with on a daily basis. In his book on illness narratives, Frank (1995) named the “chaos narrative” to reflect this continued perception of vulnerability that some individuals experienced following an illness or life threatening health condition.

**Becoming comfortable with the word cancer.**

Some participants explained that, as a result of having come to terms with their increased risk for breast and/or ovarian cancer because of their family history, they have also learned to be comfortable with the word “cancer,” as it became an expected part of their future. As Stephanie said, “What else was I to assume?” Before she finally received her breast cancer diagnosis at the age of 50, she had already gone through two previous cancer diagnoses, the first at 20 years old when she was diagnosed with precancerous cells in her cervix and the second at 35 when she was diagnosed with uterine cancer. Just like her mother, who had received two cancer diagnoses, one at the age of 32 of breast cancer and the other at 66 of lung cancer, Stephanie was not surprised to have more than one cancer. D’Agincourt-Canning (2003) documented similar findings in her study. She found that unaffected individuals drew on other family members’ cancer experiences to construct how cancer could eventually happen to them. Family patterns of breast cancer have certainly been used to inform the participants’ perception of their risk of future cancers.

Within the literature, becoming comfortable with the word cancer has also been seen as a coping strategy among individuals from high-risk families. Appleton, Fry, Rees, Rush and Cull
(2001), who studied the psychosocial effects of living with an increased risk of breast cancer among 25 unaffected women, reported that women use a variety of cognitive strategies to cope with this increased risk. They focus on the present, avoid potentially worrying breast cancer cues, and think positively. Thus, they adopt an optimistic attitude about the future.

The strategies I found are similar to those within Appleton et al.’s study (2001); however, they also found that the participants in their study had to learn to live with this threat, whereas the participants in my study said they had become comfortable with the word cancer as a result of seeing breast cancer as part of their families – as a family artifact. Breast cancer was omnipresent within their lives, they explained. Within their homes were pictures that remind themselves and others of those who had received a breast and/or ovarian cancer diagnosis. The scars were not only psychological but physical: the pictures showed that some of the women in the family are breastless. Although all participants experienced stress upon their cancer diagnosis, they also celebrated their cancers’ departure. One participant described how, when it came time for her to have her breast removed as part of her breast cancer treatment, she and her sister had a huge celebration the night before. They took pictures of her naked breasts and ones with lingerie to celebrate the departure of one breast, scheduled for a mastectomy the next day.

Becoming comfortable with the word cancer also seemed to come as a result of witnessing a close family member’s experience with cancer. The idea of having cancer was not foreign, even before they receive their own diagnosis. One participant, Juniper, explained how she felt she had already gone through the cancer experience even before receiving her own cancer diagnosis because of the way her mother involved Juniper in her emotional and physical experience with cancer. Here is my reinterpretation of Juniper’s story of what it was like for her to grow up with a family history of breast cancer:
When Juniper’s mother was diagnosed with breast cancer at the age of 45, she pulled Juniper and her sister, both still in their preteens, out of school so that they could look after their younger brother. The mother felt she was unable to do so anymore as a result of her breast cancer. Juniper said that her mother went into a depression and showed the many facets of her cancer experience to her children. The family’s support system was limited to the immediate members because they had migrated from another country and had no relatives in Canada. With the diagnosis, her mother, having no outside support system, further isolated herself. Her young children became her only support. Her mother showed her two daughters all her scars – physical ones and the mental ones. Juniper mentioned how her mother’s mental pain was more difficult to deal with than her physical pain, even when her mother showed them the gruesome scars from her radical mastectomy. And they did look gruesome back then, Juniper added.

There began Juniper’s story of feeling as if she had already been through the experience of breast cancer, even before receiving her own diagnosis. Juniper also shared how her mother later had her other breast removed out of fear. However, 5 years after her mother’s breast cancer diagnosis, Juniper explained that her mother viewed her experience with cancer as an epiphany. Her mother realised she wanted to live life to the fullest and thereafter embarked on many projects that were meaningful to her and, at times, to her family as well.

Her mother’s epiphany was a learning experience for Juniper. She knew she could now live through cancer, as her mother did. However, she promised herself that, contrary to her mother, she would not fear the cancer experience and that she would not share all of her intimate emotional experiences with her children. Like her mother, Juniper was diagnosed with breast cancer at the age of 45. When Juniper made the decision to have genetic testing, she did not share this news with her mother but did tell her children that no mutations had been found.
Becoming comfortable with the word cancer applies mostly to participants in whose families cancer occurred in the previous generation. Those whose families’ cancer experiences appeared only within their generation did not talk about becoming comfortable with talking about cancer – illustrating an evolving level of comfort with the word cancer.

While some participants became comfortable with the word cancer and comfortable with their increase risk for cancer, a few were not comfortable at the thought of losing their breast to cancer. Although images of breastless women were familiar to some of the participants, this did not mean that they were at ease with the notion. The following quote speaks to this issue:

Having most of my adult life seen my mother with no breasts, I knew what it was going to look like. It wasn’t as if I didn’t know what it was going to look like. You get used to the word cancer and are almost comfortable with it even when one gets a diagnosis. But what has been harder is losing the breast. It’s like, there’s two different issues happening. I had a harder time with losing my breasts as opposed to having cancer. (Juniper, PP2, field conversation, December 13, 2000)

In light of what is known about individuals living with a high-risk of disease, we can hypothesize that time allows acceptance of one’s risk of cancer, while having to accept a mastectomy as a treatment for breast cancer is an immediate decision that does not allow much time for reflection. However, according to two studies that explored how well women accepted loss of a breast following a mastectomy, only 7% in Kiernan et al. (1981) study reported negative problems resulting from their surgery – work, social or sexual problems – whereas among this 7% (20 women), sexual problems were cited most often. From their study, Metcalfe et al. (2004) also concluded that 97% of the women surveyed were satisfied with their decision but that younger women below the age of 50 were less likely to report satisfaction than older women. The women in the Kiernan et al. study who expressed greater concern with losing their breast were also younger than most of the other women. Just as in the Metcalfe et al. study, for women in my study under the age of 50 who underwent a mastectomy to treat their breast cancer or
prophylactic mastectomy, having reconstructive surgery immediately after facilitated their acceptance of losing their breasts. A study by Maguire (1989) demonstrated that psychiatric morbidity was lower among women who underwent reconstruction immediately following mastectomy while Rowland et al. (2000) study shows that women from the lumpectomy group (n=1119) compared to women from the mastectomy with reconstruction (n=327) or the mastectomy-alone group (n=511) reported fewer problems with their body image and feelings of sexual attractiveness.

Cancer in our family is part of our upbringing – it is the norm.

Individuals with a strong family history of breast cancer in my study explained how the topic of breast cancer came to be a common subject of conversation within the family and at family gatherings. Reminders of breast cancer around the house became keepsakes of the family’s history. Although the following quote is from only one participant, many others expressed in similar terms this view that cancer was part of their upbringing:

My children are aware of everybody’s cancer in the family and have always been aware that I have had breast cancer. They see the old pictures of me and they are like reminders of my past history with breast cancer. I had a mastectomy and although I am wearing a prosthetic bra in these pictures, they all feel they could tell the difference. (Sherry, PP21, field conversation, September 18, 2001)

Sherry later described how she came to view breast and/or ovarian cancer as an expected event by making a parallel with how society has come to view car accidents as an expected part of having so many cars on the road. She has come to view cancer as the norm in her family. Before her own breast cancer diagnosis, there were three confirmed breast cancer diagnoses in Sherry’s family. After Sherry’s breast cancer diagnosis, there were two more relatives who received cancer diagnoses -- her two nieces who are siblings. One developed breast cancer while the other developed ovarian cancer.
It used to be that it became common to hear about car crashes and people being killed. There just seemed to be so many people, it just became normal that some individual would be killed in a car crash. Or to hear every couple of months of somebody you knew being dead. That was sort of the norm. And so this is what it is like for me with breast cancer. Breast cancer in my family is the norm. (Sherry, PP21, field conversation, September 18, 2001)

As a result of their perceived increased risk for cancer from seeing so many cancers in their past and present family, some participants shared that “There is always the thought that, someone else in the family will be diagnosed.”

The participants of my study have all had breast cancer and have a strong family history of the disease. By virtue of their family’s history, they feel at risk to develop another cancer. For those who share these latter thoughts, the idea that their family members are likely to develop cancer is often omnipresent. Hearing of another breast cancer within the family is an almost inescapable reality that they learned to live with.

Role of family guardian.

Many study participants adopted the role of family guardian. They make it their responsibility to look out for their children, siblings and sometimes extended family members. As family guardian, they tried to reduce the family’s risk of cancer, and to provide them with information to increase their screening behaviour and to make them more aware of their increased risk for cancer. The concept of family guardian as related to the ill relative is not new within the literature. Past studies have described how the ill relative within high-risk families becomes the manager of cancer-related information for others in the family unaffected by breast cancer (Chalmers et al., 1996). This guardian position is also seen as providing a role model that one can survive their cancer and for the need to be proactive in decreasing their risk of cancer.

Seeing herself somewhat as the family guardian – being the eldest sibling, Ginger explained that she felt she needed to gather as much information as possible about her disease on
behalf of her siblings. She felt that, the more information one has, the more power one has over the disease. She did, however, specify that, although she was going through genetic testing as a means of obtaining more information about her cancer risk, she had not decided what she would do with the information.

While the participants may have enacted the family guardian role unconsciously, some of the family members interviewed also spoke of viewing their family member who underwent genetic testing as role model -- a role model of living through a cancer experience.

Like I mean, the whole idea of saying you know, the survivor of breast cancer, I mean it seems like a term that’s being used so much and it really kind of bugs me but I mean, you have somebody who’s lived through it. And so I have sort of a living role model and the fact that if you developed breast cancer, they are means of ways of getting over it. Not to say that it is not a killer, I mean it is. People do die. It’s just depends on how it gets into the system right. (Jill, SP2/PP3, Joyce’s daughter, field conversation, December 18, 2000)

Although Jill has not lived through a cancer diagnosis, she described her experience of living with a family history of cancer using terms similar to those used by affected individuals. Having become aware of her increased risk for cancer because of her family history with the disease, Jill has learned to live with this risk. She does not think that one survives breast cancer but that one can learn to live with having a diagnosis. Other individuals in her family who have lived through their breast cancer diagnosis are role models for her. They show that a cancer diagnosis is not a death sentence.

Need to be proactive.

Because of the cancer risk in their family, the participants spoke of their need to proactively prevent another cancer in themselves and to prevent cancer in their family. As said previously, seeing oneself and others in the family at higher risk of developing cancer when one has experienced the disease is not unique to high-risk families, but is often seen among cancer
survivors (Macleod, 2000; Mast, 1998; Mishel, 1988, 1990). Being proactive may also be an adaptive mechanism to help them deal with concerns they frequently mentioned such as, “Why did I get breast cancer?” and “Was it something I did?”

According to Macleod’s (2000) study, illness uncertainty is positively linked to fear of recurrence and symptom distress. Hence, although unaffected individuals such as the family members interviewed who do not live with cancer and are therefore most likely not experiencing the same probability of illness uncertainty, they could nonetheless be experiencing what is known as living with chronic risk (Kenen, Ardern-Jones, & Eeles, 2003b).

Just as most of the participants viewed their genetic test results as uncertain, they also viewed the origin of their cancer as uncertain. In attempting to understand why they got breast and/or ovarian cancer, they compared themselves with others in the family, in terms of who they believed was more at risk of developing the disease. Even participants who strongly believed they had an inherited mutation and that this mutation was the cause of their cancer still struggled with the idea that current genetic testing technology could not confirm their genetic mutation status. Hence, the participants also learned to live with uncertainties about the origin of their cancer and with uncertainties about their genetic mutation status. This double uncertainty lends weight to the need to explore further the Kenen et al.’s (2003b) concept of living with chronic risk. A couple of participants explained that if they could not figure out why they developed breast cancer in the first place, how could they know they are being diligent enough now to prevent another cancer? Not having an answer as to why they developed breast and/or ovarian cancer left them constantly in search of, “What did I do wrong?”

Bottorff, Ratner et al. (1998) explain that uncertainty exists when the probability of outcome is unknown, while risk is associated with the probability that each outcome is known. Yet, the authors acknowledge that even known probabilities do carry some form of uncertainty.
When I asked participants how they felt about individuals who were told that a mutation had been discovered in their breast and ovarian cancer genes, three participants answered in the following ways:

Barney: They are the lucky ones, for at least now they know why they have developed breast cancer. (PP7, field conversation, December 4, 2001)

Stephanie: I would not say they are lucky but I would say that I would envy them. At least they have something to hang their hats on. (PP12, field conversation, June 27, 2001)

Erika: I was looking for an explanation for my breast cancer, but I would not have liked to be told that I had the gene. (PP10, field conversation, September 7, 2001)

The above examples expose the complexity of the participants' capacity to tolerate uncertainty. The participants were forced to struggle with the uncertainty of the etiology of their cancers and the uncertain outcome of their genetic test results without finding relief. Messecar and Kendall (1998), who studied the meanings of uncertainty and the adjustment of wives whose husbands were called to active duty during the Persian Gulf War, formulated the concept of struggling without relief as one of the three main ways wives came to terms with the separation from their husbands when their return was uncertain.

Continuous uncertainty about the origin of the participants’ cancers may explain their need to seek ways to be more diligent. One participant, Erika, reported becoming more aware of everything that could be considered carcinogenic. She mentioned how she had, at one point, almost stopped eating for fear that all food contained carcinogenic agents. Another participant,
Victoria, decided to grow an organic garden in the summer and, the rest of the year, refused to eat any vegetable or fruit that was not organic.

Participants explained that, when you have been diagnosed with breast cancer and have had many experiences with breast cancer among your family and friends, these proximity experiences remind you of your risk of developing another cancer and the need to be constantly vigilant. As Erika said, “Yours ears just perk up as soon as you hear the words breast cancer, and respite is very seldom.” However, Erika explained that this overabundant information caused her to “switch off at one point,” because she was unable to process the information anymore. Her metaphor of switching off parallels turning a light on and off. A study by Appleton et al. (2001) found similar results; some women’s heightened sensitivity to breast cancer cues prompted them either to increase their vigilance or consciously avoid the cues.

The findings of my study suggest that the role of family guardian and being a living role model to others in the family can be overwhelming. The responsibility of keeping abreast of all “anti-cancer behaviours,” as a participant described it, may fulfill their need to stay diligent. This responsibility, however, should perhaps not come without support. That is, Stephanie mentioned how it is “just too difficult to leave it up to us to find our cancers and to help others around us.” Participants felt they needed specialised support to enact their roles effectively. Hence, Stephanie requested on going assessment by a high-risk breast cancer clinic because she believes such a clinic has the latest evidence-based knowledge of breast and/or ovarian cancer.

Three participants held the opposite view. They viewed the suggestion from their high-risk clinic that they could now be monitored by their general practitioner as “being released” from the high-cancer-risk population. They perceived their cancer risk to be lowered. However,

47 Proximity experiences of family’s and friends’ breast cancer have also been defined as embodied experience – as opposed to bodied experiences. I find the distinction interesting, as both terms convey anxiety-provoking feelings. For a fuller description of the meanings attached to embodied experiences, see d’Agincourt-Canning (2003).
most participants did not share this view. As some said, “A genetic test, no matter what the results are, does not erase our family history of the disease.” Here is Stephanie expressing her need to be screened in a high-risk clinic. I must add that Stephanie is convinced that cancer runs in her family with a “100% certainty” and, therefore, firmly believes that she carries an inherited mutation: “You can’t find this disease by yourself, ‘breast or ovarian cancer’ and so that is why I want to stay in this specialised clinic.” (Stephanie, PP12, field conversation, June 18, 2001)

Reassurance from being assessed in a high-risk breast cancer clinic has been documented in another study in which the psychosocial effects of living with an increased risk of breast cancer were examined (Appleton et al., 2001). The authors described how many women not only felt privileged to be assessed in specialised clinics but also felt confident about both the expertise of the staff and the equipment.

**Increased exposure to breast and ovarian screening and testing.**

It can only be speculated that, perhaps as a result of their family history with breast and/or ovarian cancer, the participants have gone through much more than the usual amount of screening and testing. Other studies indicate that women with a family history of breast cancer used screening procedures such as mammography more often than those without such a family history (Costanza, Stoddard, Gaw, & Zapka, 1992; McCaul, Branstetter, Schroeder, & Glasgow, 1996). Participants in the Mahon and Casperson (1995) study commented that they participated in more screening activities as a result of the reminders sent by their cancer screening centres.

Costanza et al. (1992) specify that it is only when women are aware that having a family history of cancer is a risk factor for breast cancer that they increase their use of mammography. However, other studies focusing on high-risk women have documented that rates of mammography do not differ between high-risk women and women of average risk (Andersen et
al., 2003; Drossaert, Boer, & Seydel, 1996). Constanza et al. (1992) comment that, based on their study findings, further education is needed among family physicians to increase mammography utilisation among high-risk women who do not know that having a family history of cancer increases your risk of breast and/or ovarian cancer. The increased use of follow-up screening within my study could reflect participants already being clients of a cancer agency, identifying themselves as high-risk, and being identified as such by their oncologists.

Some participants remarked that they understood that having a family history of breast cancer increased their risk for the disease. For example, participants were very aware that, when they found a lump, it needed to be taken seriously. As Juniper explained to me during her interview, “I understand that my lumpy breasts are not the same as your lumpy breasts” – knowing that I do not have a family history of breast cancer. Participants commented on how, for example, a suspicious lumpy breast or a fibrous cyst was investigated more thoroughly. Hence, before some of the participants received their first diagnosis of breast cancer, they had already experienced many screening and testing procedures for cancer. The following account looks at one participant’s experience.

By the time I was barely over 30 years old, I had already experienced my first mammogram. By the time I was 40, I had about five or six done. And by the time I was 41, I had one breast aspirated from a fibrous cyst. Yes, I had been thinking about cancer for a long time. Then, you also have those worrisome mammograms that tend to lead to other mammograms soon after. (Juniper, PP2, field conversation, December 13, 2000)

A new breast cancer diagnosis in the family is often a pivotal moment reminding others that perhaps they should have follow-up screening and mammography done, regardless of whether or not they are due for their regular checkup. This again increases their exposure to screening procedures. One participant commented on how, when she was found to have breast cancer, her three sisters had their mammograms redone. As a result of this retesting, two sisters
were told their mammograms were normal while one was found to have a suspicious lump, which led to more tests revealing a cancerous tumour.

The increase in screening and testing procedures results not only from participants’ requests following a family member’s recent breast and/or ovarian cancer diagnosis, but also from recommendations by their doctors. Stephanie describes how her doctor recommended that she consider prophylactic surgery in light of her family history of breast cancer:

When the Cancer Agency found out I still had my ovaries, they panicked and said you shouldn’t have those because of your breast cancer history and your family history with the disease... He said, ‘You still got your ovaries?’ and I said yes. And he said, ‘You have breast cancer?’ I said yes. He said, ‘There is a huge connection with family history. Whether or not it’s a gene it doesn’t matter, families that have a history with breast cancer often have ovarian cancer.’ So he said, ‘Well it’s got to come out. (Stephanie, PP12, field conversation, June 27, 2001)

When family physicians are aware of their patients’ strong family history of breast and ovarian cancer, these individuals typically receive more intense screening. And, as described in past studies, physician recommendation for screening is a strong predictor of acceptance and adherence to regular screening (Lerman, Rimer, Trock, Balshem, & Engstrom, 1990; Stefanek & Wilcox, 1991).

Having to go through more than their usual share of screening and testing is not without consequences. Not all participants found a sense of security in being screened so intensely. Gilligan, echoing others, expressed how she came to dread her yearly mammograms. Gilligan was diagnosed with breast cancer at the age of 26 and had bilateral prophylactic mastectomy in her 40s:

Prior to my decision to have prophylactic surgery, going through my yearly mammograms was just a big fear. Usually what would happen is that I would go in for my mammogram, and it would come back questionable. So I would have to go in for a second mammogram and again they were having questions. And so you know I’m in there for a couple of hours, which feels to me like a very long time. I then would have to go through many visits, repeated mammograms, and then to ultrasound – to finally be
told that the mass turned out to be just a cyst. Just going through that every year, I was beside myself and so it confirmed in my mind that a mastectomy was the way to go. There was a huge fear there that you keep under control a lot of the time but yeah, at one point, you have enough. (Gilligan, PP16, field conversation, August 14, 2001)

“Having enough” meant to Gilligan that she did not want to live with this constant fear that perhaps one day she might be found to have another breast cancer, or a recurrence like her mother. Following this response, Gilligan described how she felt a huge sense of relief from her decision to have a bilateral mastectomy followed by reconstructive surgery. She expounded on how she had come to view her breasts as a major source of stress and anxiety and, by having had her breasts removed, she felt she had removed a major risk in her personal life and was now more ready to take on new risks in other parts of her life, such as in her professional life. Her husband, who was interviewed as part of this study, spoke of the impact of her surgery on their lives. Here are the field notes taken from his interview:

His wife’s surgery was lived as a major turning point in his life. He describe that he felt his wife became more relax and hence, he became relaxed. He also emphasised many times how this change had loosened the tension in their relationship and had made it so much better because she no longer feared for cancer. (Pierre, SP1/PP16, Gilligan’s husband, field notes, August 16, 2001)

Such an experience from affected women with a family history of risk for hereditary breast and ovarian cancer (HBOC) who received uninformative results to their BRCA1 and BRCA2 genetic testing has also been documented in Hallowell et al.’s (2004) study, in which a participant described that having her breasts removed followed by breast reconstruction was like being born again, in the sense that she could now move on with her life.

Within participants’ lives, there was a sense that having so many screening and testing procedures was the norm. Just as some participants came to view breast and/or ovarian cancer as the norm in their family, so were multiple screening and testing procedures. Gladys, along with
two sisters, developed breast cancer, spoke casually about the number of mammograms one of her sisters had:

My sister chose not to have a double mastectomy. She chose to have a mastectomy – only on one side. And she has mammogram four times a year, I think. So they watch her very closely and nothing else has occurred. I mean her surgeon indicated that there's a very good chance that she will get cancer again, but nothing so far has happened and she is being carefully followed. (Gladys, PP17, field conversation, August 30, 2001)

Familiarity with clinical trials.

Having a strong family history of breast and/or ovarian cancer meant that many of the participants and their family members met many of the criteria required to enter clinical trials related to breast and/or ovarian cancer. A few participants shared how they were at times solicited and encouraged to participate in clinical trials because of having such a strong family history. These participants made the analogy that, just as they easily met many criteria to enter different clinical trials, they also easily met the eligibility criteria for genetic testing for BRCA1 and BRCA2 mutation analysis. Meeting these criteria for different trials was interpreted by some as a confirmation of their high-risk status and their high likelihood of having an inherited mutation.

Ginger said, “Genetic testing is just one more tool to manage my life, to manage my chronic illness.” In this statement, Ginger acknowledged that she viewed her past breast cancer experience as a chronic illness that she needs to manage. As expressed earlier in this section, many participants spoke of their concern that breast cancer may reoccur or that they were more at risk of a recurrence or just generally more at risk as a result of having a diagnosis of cancer. In the quote above, Ginger also makes reference to how breast cancer is now part of her reality, part of her life. Breast cancer was not a state outside of her in the sense that once she received treatment, the cancer was no longer with her. On the contrary, like other participants, Ginger
emphasised that breast cancer was her “chronic illness to manage” and that it was now part of her life. Ginger’s view that breast cancer is a lifelong disease has been reinforced by outside experiences, such as reports of recurrences in her support group:

More and more, it seems women are getting breast cancer like 7 years later or 9 years later or 12 years later…In this support group where I used to go, I met many wonderful older women who are all survivors. But many of them have had, like my mother, two cancers. (PP11, field conversation, May 7, 2001. Ginger’s mother died following her second breast cancer diagnosis)

The chronic illness perspective has also been documented by Kenen et al. (2003b). In their study, women with a family history of breast and/or ovarian cancer used various coping strategies to “get on with their lives” and not be dominated by thoughts of long-term risk for cancer. These included adopting a health lifestyle, reducing stress and participating in annual screening. The authors found that women in their study faced issues concerning adaptation and an uncertain future similar to those faced by individuals living with chronic illness. Hence, they describe this process for individuals with a family history of breast/ovarian cancer indicative of HBOC as living with chronic risk.

Having to deal with broken plans.

Having to deal with broken plans reflect some of the participants’ reality whereby as a result of many diagnosis of cancer in the family or having to deal with mortality as a result of some of these cancer, some participants were not be able to follow through with plans they had made. When making short – and long – term plans, most individuals do not have to consider, what if someone gets a diagnosis of cancer? How will this affect our plan? The following family in this study had to address these issues after the fact. Gladys, a participant, relates how her sister’s plans and hers sadly changed as a result of her sister’s second breast cancer diagnosis.
Their other sister had been diagnosed with breast cancer while still dealing with her rectal cancer.

But they said, 'Well, you know it looks all right now. We don't see any gaps in the bowel cavity and, you know, maybe try a little bit of liquids and so on. And she started to eat and started to get better. And so after 2 weeks my sister G and I said, 'Well, you know we can't just stay on here. In a month or more, we have to go home to our lives.' So we left and G said to me, 'Look, I have been over three times and you also have been over three times, but I'm retired and my husband's retired so I am more flexible.' So she said I didn't have to go again, if... when N dies - because we knew it would be within the next month or two - that she would handle it. She was going to go to the funeral. So I said, 'that's fine.' So, she was all pumped up to organize it and, when N died, G was diagnosed with a second lump and was told she had to have a mastectomy right away. So, she couldn't go to the funeral. So I had to go to the funeral. And that was so upsetting to G that I think that's probably why she's had all this trouble and all this pain because she was... she was absolutely destroyed at not being at the funeral. She and N had been very close in age; they were about 13 months apart. (Gladys, PP17, field conversation, August 14, 2001)

Here is a reinterpreted biographical account of Emma's experience of broken plans resulting from her two sisters dying from breast cancer as shared during her interview:

It only had been recently that Emma could speak about the death of her sisters. Emma had made plans to grow old with her two sisters. Instead, she saw them die of breast cancer as she nursed them both to their deaths. One of her sisters was like a mother to her. Upon this sister's diagnosis, Emma and her husband made the decision to sell their home and move closer to her dying sister. Soon after their move, Emma took her sister into their home and cared for her till the end. Following the death of both of her sisters from breast cancer, Emma was then diagnosed with breast cancer as well, at the age of 43. Emma said that, for a long time and still now, she suffers from survivor guilt. (Emma, PP20, field notes, October 27, 2001)

Perhaps this section could also be called adaptation to living with a family history of breast cancer. However, adaptation differs between participants who had children and those who do not. Those who had young children often said that, "Dying is not an option." Evelyn, who did not have children, spoke of her risk of dying from her disease as something to expect. Evelyn was first diagnosed with breast cancer at the age of 46 and again at the age of 50. She said she was still premenopausal when she received her two breast cancer diagnoses. Evelyn explained
what she meant with seeing death as something to expect because of her lived experience with cancer:

Like living an uncertainty is what one of my psychologists said, you know, and you do but you don’t dwell on it, you know. And I’m learning to be more comfortable with the thought of dying and watching, you know, I’ve watched a lot of friends die and see how gracefully and graciously they do it but it does, I mean it’s, it’s why I’m retiring in two years. I mean my odds of living to 65 are not great, so why spend all that time working? (Evelyn, PP5, field conversation, March 23, 2001)

Evelyn goes on to explain:
The head librarian, I saw him just after Christmas and he asked how my holidays were and I told him that my dad had just died and it was unexpected. And he said, um, oh, that must be really hard, it’s a long time ago that my parents died but it really causes you to face your own mortality. And I looked at him and I said, ‘David, been there, done that twice already.’ (PP5, field conversation, March 23, 2001)

In the above quote, Evelyn refers to her poor prognosis which had been given to her on both of her cancer diagnosis. In conclusion, after hearing how participants coped with their strong family history of cancer, I now understand more fully their description of genetic testing as “nothing in comparison to their experience with cancer and their family’s history of it.” Certainly, having understood these individuals’ experiences with cancer through the nine themes presented above has given me a better sense of why they came to be interested in having genetic testing. Through these themes I have also come to understand how living with such a history would make individuals more receptive to genetic testing because they have had much time to adapt to living with their perception to an increased risk of a subsequent cancer and risk of cancer for their family members, and, to their belief that perhaps all the cancer cases in their family results from an inherited mutation.

In the next section, I look at how genetic testing entered the participants’ lives and the reasons that led them to accept genetic testing. Many of the reasons operate within the
participants' experience of living with a family history of cancer and, more specifically, within the themes just discussed.

**Encountering Genetic Testing**

As I began to analyse the reasons that brought the participants to pursue genetic testing, it became apparent that a process took place before the participants arrived at in genetic counselling. In this section, I begin by describing how participants became aware of the availability of genetic testing for breast and ovarian cancer susceptibility. Then, I present the different reasons for having genetic testing described by the participants in accepting to have genetic testing.

**Hearing of the availability of BRCA1 and BRCA2 mutation testing**

Very few participants had inquired about the availability of a test that could assess whether their cancers were inherited. Of all the 21 participants, only 5 self-referred for genetic testing for breast and ovarian cancer susceptibility after becoming aware of its existence either through the media or through breast cancer support groups. Five other participants had been contacted by a family member to see if they would have testing done on their behalf and 11 other participants had been made aware of the availability of this test by one of their health care providers.

**Self-referral.**

As discussed in the previous section, Implications of Living with a Family History, a newly diagnosed case of cancer in the family often acted as a catalyst for change, by reminding family members of their increased risk to cancer and to perhaps leading them to have a follow-up screening test. This applies to four of the five participants who self-referred. As also discussed in the previous section, each new case of cancer further influences their perception of risk and a
need to proactively reduce the risk. These two events led many participants to become attuned once more to all that is said about breast and/or ovarian cancer. Hence, three of the participants described how during this period they came to hear of the availability of genetic testing either through the media or through a breast cancer support group. One participant, Marcy, asked about genetic testing after the death of a cousin from kidney cancer. As Marcy explains,

Bernadette – that’s my mother’s sister’s daughter – she had breast cancer when she was 60. That’s my first cousin. Her brother had just died of kidney cancer, so we’ve got quite a few in the family. When you start thinking about it and writing them all down, you start to see how they add up. So that’s the reason we wanted to be – asked to be – genetically tested. (PP18, Marcy, field conversation, September 13, 2001. Marcy’s two sisters, two female cousins and herself were diagnosed with breast cancer. One female cousin was diagnosed with colon cancer. All are on the maternal side of the family)

The fact that they all lived in a province where genetic testing for BRCA1 and BRCA2 mutations became available led them to be more aware that such a test was actually possible for them.

Upon a family member’s request.

Similar to those who had self-referred following a reminder of their increased cancer risk, family members (5) who approached primary participants about having genetic testing on their behalf had the same catalysts. Joyce, who had been approached by her cousin Stacey to have genetic testing, explained how her cousin had recently lost her sister to breast cancer. I had the opportunity to interview Stacey. The following quote tells how Stacey came to hear of genetic testing:

Every few years I’d hear of another cousin. Right now, out of the three generations, there are 13 women that have had breast cancer in our family. My sister was the last one. So, once my sister developed breast cancer at the age of 46, it got quite personal then. And, um, just watching her go through what she was going through, um, and thinking of her children, (she has three daughters) it just made me want to do as much as I can to find out what’s going on with our family. Then several years ago I saw an ad in the paper about this breast cancer research thing. So I asked my doctor if he would refer me, which he did. And that is how the whole thing started for our family. (Stacey, SP1/PP3, Joyce’s cousin, field conversation, June 4, 2001)
Joyce offers the following interpretation of her cousin’s interest in genetic testing:

My cousin had asked me to go for genetic testing. She has since lost her sister to breast cancer. Last April she lost her. So she’s the one that asked me. She phoned and asked if I would mind and I didn’t. It didn’t bother me to go down and to see. (Joyce, PP3, field conversation, December 12, 2000)

Joyce explained that, before her cousin called, she did not know of the availability of this test but, after discussing it with her cousin, felt that it would be a good idea. Joyce then made the first call to the Hereditary Cancer Program, the clinic her cousin told her offered the test. Joyce understood that she was being asked to have genetic testing, because she was the only living individual with a breast cancer diagnosis who was locally available. Four other participants where also contacted by a relative to have genetic testing on their behalf following a recent diagnosis of breast cancer in one of their close relatives. All of the five family members who contacted a primary participant in this study to seek their willingness to have genetic testing on their behalf all lived close to the Hereditary Cancer Program and were aware of its existence. Hence, it was after these relatives had consulted a health professional from the Cancer Agency that they contacted the primary participants.

Of the five participants who were approached by an extended family member, four said that they did not object to having been contacted. They said that they understood their relatives’ suddenly increased perception of risk for breast cancer because of having a close family member diagnosed with the disease. Besides, they themselves could now see value to this test for themselves and their own immediate family members. The one participant who had objected to being contacted by an extended family member to have genetic testing on her behalf, spoke to the issue of timing in terms of when she felt it was appropriate to approach a family member about genetic testing.

48 Recall that, in order to be eligible for genetic testing for BRCA1 and BRCA2 mutations, a blood sample from an affected individual with breast and/or ovarian cancer is needed.
It was a cousin that phoned me, and she’d heard about it, and she asked me if I’d go in and give some of my blood. I was just going through chemo and radiation at the time. And I, well I hadn’t talked to her in years. Well, phoning me up to ask me to do that after not talking to me in years! Well, that kind of threw me for a loop. But I said, sure, no problem on my part. And that’s where she phoned the genetic counsellor, and then the genetic counsellor phoned me. And I said, yeah, I will come in and let them take my blood and stuff like that. (Barney, PP7, field conversation, December 4, 2001)

Barney later explained that she was upset at her cousin “calling her out of the blue like that” after not having talked to her for years. Perhaps Barney felt, although she was receptive to genetic testing, it was not the best time to have it done because she was in the midst of her own breast cancer treatment at the time. Certainly, the timing of the approach of when to contact a third party to have genetic testing for someone else’s behalf needs further inquiry.

Although it was not the purpose of my study to assess and analyse family dynamics and family interactions as a result of genetic testing, this is an area that would certainly be interesting to pursue in future studies, in light of Barney’s response. Research questions could include: What are the best clinical guidelines for deciding who should inform third parties of another individual’s interest in genetic testing and when is the best time to approach them? As Barney later specified in her interview, she would have preferred to be approached by a genetic counsellor or a genetic nurse rather than being asked by a cousin whom she had not spoken to in years. There is still little empirical knowledge about the best way to approach family members about genetic testing while creating as little harm as possible, when the request is made on behalf of a third party.

Another participant, Victoria, explained that her cousin had not sought her own sister’s interest in being tested, despite the sister being recently diagnosed with breast cancer but, instead, called upon her. Victoria’s cousin felt that her sister was having a hard time adjusting to her breast cancer diagnosis while concurrently receiving chemotherapy and would find genetic
testing a burden. Victoria’s diagnosis of breast cancer dated back 11 years. Victoria explained that she had heard of genetic testing and had always been interested in it but had never pursued this interest. Through this call she received from her cousin’s and her mother’s genetic counsellor, she found out that both of them had independently pursued genetic testing and that her mother had actually had gone ahead with it. She passed away from breast cancer before obtaining her results. Victoria’s mother had not shared with her that she had pursued genetic testing.

Victoria’s cousin, who wanted to have genetic testing done following her sister’s diagnosis, inquired about the test but found out that one of the eligibility criteria for testing was a past diagnosis of breast or ovarian cancer in their own body. As noted in the clinical trial study by Loader, Shields, and Rowley (2004), the individual who initially requests testing appears to be the one who undergoes the most stress when asking for genetic testing. This stress associated comprises distress from cancer risk, genetic counselling, distress about general health and emotional health, and breast cancer worry.

**Recommendation of a health care provider.**

Eleven of the 21 participants had heard about genetic testing from one of their health professionals while receiving follow-up care or follow-up screening at the Cancer Agency. Contrary to Barney, who felt upset being called “out of the blue” by an extended family member, none of these 11 participants mentioned feeling any anxiety over their health professionals’ recommendation that they have genetic testing done.

**Recommendation from a health care provider has been reported in other studies as the most frequent method of referral for individuals with hereditary cancer syndrome (Campbell et

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49 I did not have the opportunity to interview Victoria’s cousin. Hence, I do not know how she came to hear about the availability of genetic testing and why she felt that it was improper to ask her sister.
al., 2003; Mahon & Casperson, 1995; van Zuuren et al., 1997), especially when physicians know that genetic testing can be obtained locally (Wideroff et al., 2003). While these studies indicate that health professionals do know how to identify and know when to refer women with a family history of breast cancer for risk evaluation, not all studies point in that area. A recent review by Hutson (2003) showed that most women who attended these programs tended to be self-referred and had underlying psychological distress. Based on this finding, Hutson recommended that more education be given to health care providers to help them identify individuals at increased risk of hereditary cancer who could benefit from genetic counselling and screening. This recommendation is also supported by recent studies indicating that lack of genetic knowledge and lack of local availability for genetic testing is a barrier to physician referral (Wideroff et al., 2003). When I asked all the participants how genetic testing entered their lives, many first said they were uncertain but that it most likely would have been through one of their health care providers. Given that all of these individuals were being followed in a high-risk-breast-cancer clinic, I thought that most would have been referred by health professionals.

Of the 11 participants referred by a health care provider, only 3 actually said that their health professionals had requested a referral for genetic testing to help them make immediate decisions about their breast cancer treatment. Obtaining their first genetic test result took (for some) almost a year; they did not in the end use the test results to decide whether to have a bilateral mastectomy and prophylactic oophorectomy. Instead, following their family physicians' and oncologists' recommendations, they opted to follow through with a bilateral mastectomy.

In summary, the process of genetic testing did not always begin with a personal interest. Rather, most individuals were either advised to have such a test by one of their health care providers or received unexpected requests by a relative to get tested, because the relative was
ineligible. Among the 21 participants who had genetic testing done, in 16 cases, the genetic testing process was initiated by others. Nonetheless, all 21 could find good reasons for getting tested. These reasons represent the last phase of the process to the built up of genetic testing.

**Motivations for having genetic testing**

Just as Hutson’s (2003) found in her study, in my analysis of the motivations given by participants for having genetic testing, the main motivators given were not polar opposites but more scaled from having genetic testing for themselves to having it for others as well. However, within these scales, priority distinctions were found. The distinctions reflected aspects of self in relation to others. The same view has also been documented in recent research by d’Agincourt-Canning (2003) who also studied the experience of high-risk women and families with genetic testing for inherited susceptibility to breast and ovarian cancer.

Four categories of motivations for having genetic testing were constructed within my study:

1. For others only.
2. For self and others without priority.
3. For others first, and then for self.
4. For self first, and then for others.

Within these categories, some priorities were assigned by the primary participants. I derived these four categories from my analysis of the motivations that led the participants to decide to have the genetic test done. The conditions for each one of the categories will be discussed below, but, to begin, here is a brief description of the categories.

The first category of motivators given for having genetic testing was *for others only*. The second and third, although containing the same words, *for self and for others*, differed in the
priority to each word given by the participants. That is, having genetic testing was either done for oneself first, then for others; or for others first, then for oneself as well. This third category is represented as for others first, and then for self. The fourth category is for self first, and then for others. The difference between the fourth and the second and third categories is that, initially, when participants decided to get tested, their motivations for having the test done was for themselves only – until they received genetic counselling and realised that their test results could be important to other family members. Then their motivations shifted categories: from for self only to for self first, and then for others. The latter (fourth) category shows the aspect of self in relation to others evident when individuals make decisions about medical testing in the context of genetics. The four categories, although I present frequency rates for them among the 21 participants, are not mutually exclusive. Rather, I use these categories to illustrate the complexity of decisions involved in genetic testing that the participants underwent. When participants said that they were having the test done for others, they meant others to include immediate family members as well as extended family members. The one participant for who other represented only her children, expressed how she did not feel that it was her responsibility to let her extended family know that she had gone for genetic testing.50

For others only.

Of the eight participants whose reasons for having genetic testing were for others only, four had been asked by a third party to have genetic testing done: two by a niece and the other two by a female cousin. Participants explained that they felt genetic testing was too late to tell

50 The issue of responsibility for communicating genetic information is a delicate one, because of the ethical implications involved. These include, whose duty is it to inform? What are the harms and benefits of communicating, or not, that one has received genetic information? Although this study’s focus was not to assess the ethical issues of genetic testing for individuals and their families, certainly some of the participants brought up some valuable ethical issues. These issues are discussed later in this dissertation. For reviews of some of the ethical implications of genetic testing, see d’Agincourt-Canning's (2003) doctoral dissertation and Burgess (2001).
them if they were at increased risk of developing breast and/or ovarian cancer, since they had already developed the disease. Hence, they viewed the test as only useful for family members still unaffected by the disease. Just like in d’Agincourt-Canning (2003) study, very few participants had considered genetic testing just for themselves only. However, as other studies have reported, many individuals have genetic testing out of a sense of responsibility to others, such as for their children (d’Agincourt-Canning, 2003; Lerman & al., 1996; Lerman & Croyle, 1995; Lerman, Daly et al., 1994; Lerman et al., 1996; Lynch et al., 1999).

Participants believed that, by having the test done, at least family members would have the option of being genetically tested to see whether they themselves carry an inherited mutation. The participants also felt that, if they were found to carry a mutation, they could at least use this information to convince their family members to be more proactive about their health and about cancer screening. This links with the view of some participants that they are family guardians and are always looking for ways to make their family members aware of their risk for cancer. Having genetic testing for others only has also been documented by Bottorff (2002) and Hallowell (2002), who describe how their participants wanted to have genetic testing in order to warn family members of their possible increased risk of an inherited mutation.

The following situation illustrates the category for others only as reflected by one participant within this study:

I just thought it was something I could do that, if I tested positive, then they would be more aware and they would have more information. But I really think that it’s more important for the people that have not tested positive or have not had cancer. You see, genetic testing was nothing to me. It’s more for other people, members of the family. I know I am more at risk to cancer. I have already been diagnosed with breast cancer and was told that I was terminal 5 years ago. I mean, what if I had the gene? It made no difference. (Macy, PP1, field conversation, December 11, 2000)

51 In the Hereditary Cancer Program, one of the criteria for unaffected individuals to have genetic testing is that an individual within the family must have an identified BRCA1 and/or BRCA2 cancer gene mutation. Not all cancer genetics programs have this criterion.
Like Macy, the other seven participants who gave reasons in this category also felt that genetic testing was “too late for them.”

**For self and others without priority.**

Having genetic testing done *for self first, and then for others* is the second category of motivations. Here eight participants could see a benefit for themselves, as well as valuable reasons to have the test on behalf of their family members.\(^{52}\) They wanted to find out why there were so many cases of breast and/or ovarian cancer in their families and if all of these cancers resulted from an inherited mutation. Among those eight, three had the test done to assess their risk for ovarian cancer and to help them make informed decisions about having prophylactic mastectomy and/or oophorectomy. These three participants felt that their risk to breast and ovarian cancer was “high enough,” with or without the mutation, and went ahead with their surgeries without waiting for their test results. One participant had a prophylactic oophorectomy, while the other two had bilateral prophylactic mastectomy. One of these two had received a lumpectomy as part of her breast cancer treatment but still felt at risk for a subsequent breast cancer. As described in the first section of this chapter, this participant, like others, constantly felt at risk for a subsequent breast cancer and for a recurrence especially since her mother had died from recurrent breast cancer.

The participants hoped that genetic testing would provide them with information that could at least tell them if their cancer was hereditary. Juniper explained:

> I wanted to be more informed. I wanted to know that if I had a genetic type of cancer or a hereditary type, or whatever you call it. I felt that not only did I owe it to myself to know

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\(^{52}\) When the participants spoke of having the testing done on “behalf” of their family members and extended family, I do not know if the participants had understood before having genetic counselling that their genetic information would also have implications for others. The genetic counselling process was not studied within this research, but the evolution of one’s understanding of the implications of genetic information for others would be an interesting area to study.
that so that I could make decisions regarding my other breast and decisions regarding my whole body – my health, you know health wise, that it was also imperative having a sister. I owed it to my sister to avail myself to anything that is being offered to me that could help her out and ease her anxiety about not only having a mother at 45 with breast cancer but having now a sister with breast cancer. I owed it to what I originally thought were only my daughter and my two nieces. But I didn’t realize that I also owed it to my nephews, because I didn’t realize that it can carry along that way. I felt that I owed it to my whole family; that, if this information was there and I could avail myself to that information, that it was really important for me to get it because it would help us be able to make decisions no matter which way it went. It would help us make the right decisions for our bodies. (Juniper, PP2, field conversation, December 13, 2000)

Juniper added further that, while she already had one breast removed as part of her breast cancer treatment, she had seriously considered having her other breast removed if genetic testing showed that she carried an inherited mutation. Although many other studies have documented similar reasons for having genetic testing that go beyond the individual’s need (Bottorff et al., 2002; Burgess & d’Agincourt-Canning, 2001; Hallowell et al., 2002; Lynch et al., 1999), Juniper’s response shows the aspect of the self in relation to others and how it is difficult to dissociate the self from the rest of the family members in the context of genetic testing. The reasons reported by the above studies are similar to those expressed by the current study participants: to use genetic information to increase utilisation of cancer screening among family members, to adopt more proactive behaviours aimed at reducing general risk to cancer, and to know the etiology of their cancer.

Ginger, whose reasons fall under this category, explained that, although uncertain about what she would do with the genetic information, she believed that information was power and that, if the information was available, she should try to obtain it. Although Ginger had no intention of telling her daughter about her results, since her daughter was only 9 years old at the time of her genetic testing, “At least, if one day she wanted to know, then the information would be available for her.” Just as the participants of d’Agincourt-Canning’s (2003) study that viewed
knowledge as power, participants in my study also viewed gaining information as a form of control over their disease and felt that it is better to know if one has an inherited mutation than not to know.

For others first, and then for self.

Having genetic testing done for others first, and then for self is the third category of reasons. Here, participants’ reasons to have genetic testing were for others first then for themselves. Three participants fell under this category.

Here are two quotes that distinguishing between the second and third category of reasons for having genetic testing:

For self and others
I was having it for my children and also for myself. (Gilligan, PP16, field conversation, August 14, 2001)

For others to self
I was first doing it (genetic testing) for the kids then for my self. (Mimi, PP15, field conversation, August 21, 2001)

Gilligan’s quote does not give priority to one reason, whereas Mimi’s quote does. Evelyn, one of the three participants who said that they were first having the test done for others, then for themselves, explained that she was first having the test done for her siblings and extended family members but that she was also having it done for herself. She said that she did not need this test to tell if she was at increased risk for breast cancer as she already had two breast cancers but did want to know if she was at increased risk to ovarian cancer.

For self first, and then for others.

The final and fourth category of reasons is for self first, and then for others. Although, as in the third category, only two participants are within this category, its particularity is worth
discussing. The *then for others* reflects how, at first, these two individuals’ acceptance of genetic testing was for self only and then became directed toward usefulness to others following the genetic counselling session. Both had entered genetic testing by a recommendation from their physician.

The two participants, Becky and Louise, explained that it was during their genetic counselling session that they first became aware of the value of this test for other family members. Louise explained that her initial reason for testing was to confirm that her ovarian cancer diagnosis was “not her fault” – that it was not the result of anything she did in the past. As for Becky, she explained that she “had just gone along for the ride, like one would when recommended to be followed up by a nutritionist.” The following quote describes the feelings Becky experienced when she realised the information her test could provide to her family:

> So I go down there and I was nervous, I was surprised. I didn’t know why I was so nervous, but then Dr. X did a very good job at putting me at ease. Then Dr. X starts explaining me that my family history with breast cancer is quite interesting cause – you know – no ovarian cancer but so many late onset breast cancers. And Dr. X said, ‘For example, your grandmother and her three sisters were in their 80s, 90s when diagnosed with breast cancer. Now, we’d be interested in knowing what happened like what protected them, you know – if there was a gene that protected them. But, basically, she said there are too many breast cancer cases in my family to be due to chance and she said, you know, we can maybe help you and you can help us.’ So I’m starting to think maybe I can contribute something to science. But somehow I was still thinking this is just me. This is just my decision, okay, and I decided now why am I doing this anyway? I mean what good is it going to do and then I decided well, no, I think I want to do it because, what if something changes in the future and there is something they can do about it? Then they will already know that, yes, I am a candidate or no, I’m not a candidate. I mean you never know. And I just feel that information is a good thing…Then Dr. X said, ‘You know, we’d like to test you.’ I said yeah. She said, ‘We like to test the people who had cancer. So we’d like to test you. We’d like to test your aunt Katherine and we’d like to get tissues that maybe still exist from your two aunts that died and your grandmother.’ All of a sudden I’m going, ‘you mean I have to tell all those people that?’ And it didn’t sort of hit me, uh, till then. I thought they were just going to test me…. (Becky, PP8, field conversation, April 17, 2001)
No participants in this study were simply self-interested in testing. This fact may speak to the notion that genetic information has implications not only for the self but for other family members as well and to what Hallowell (2002), a sociologist, attributes as a sense of ‘genetic responsibility” towards others. In an empirical study, Hallowell (2002) noted that women who participated in genetic counselling and testing for breast and ovarian cancer did so out of sense of responsibility to their family (past, present as well as future) to determine their risk to an inherited mutation to the disease. Hence, the current finding further supports the need to assess individual’s decision to having genetic testing as to determine the potential impact and lack of if they receive uninformative genetic testing results.

In this chapter, I described the process that led individuals to be interested in genetic testing and the reasons that led them to have it. I suggested that there is a buildup before individuals become receptive to genetic technology, which includes living with a family history of breast and/or ovarian cancer and the implications of living with such a history. I offered a list of four categories of motivations for having genetic testing with their respected reasons as offered by the participants. What these categories provide to clinics is a condensed list that encompasses the many reasons already documented in other studies by distinguishes how the self is situated in the reasons people bring to having genetic testing. Further, the four categories also represent incentives for having genetic testing done. These incentives also serve to locate the participants’ satisfaction or dissatisfaction with their genetic testing experience, after receiving their results. This is further discussed in chapter 6.
CHAPTER FIVE:
Interpreting and Making Sense of Genetic Test Results

This chapter seeks to extend the understanding of participants' interpretation of their genetic test results by exploring the meanings attached to their interpretations. Although the participants were told that their results from the test for BRCA1 and BRCA2 mutations were negative (meaning that no mutations were found), the participants had different views on how such results ought to be interpreted. My goal here is to demonstrate how patterns of interpretations are shaped and altered throughout the making-sense experience that is grounded in individuals' belief systems. These belief systems influence how they interpret their test results.

First, I present the types of interpretation participants gave to their test results. Second, I lay the groundwork for the structures informing their beliefs to how they interpreted their test results. Then, I move to contrasting individuals' stories to identify differences and similarities that render individual cases visible within the common patterns identified (Thorne et al., 1997).

Types of Interpretation Given to Their Test Results

Table 4 presents the three types of interpretation that I derived from the participants' accounts. When I asked the participants how they interpreted their test results, they would start off by saying, "It is negative for BRCA1 and 2," then go on to say "but," "or," or "and." They would follow this with a sentence that resembled the ones in the meaning row of Table 4.
Table 4: Types of Interpretation Derived from the Participants' Accounts to Uninformative Results

<table>
<thead>
<tr>
<th>Interpretation</th>
<th>Confirmation of mutation carrier status</th>
<th>Ambiguity regarding mutation carrier status</th>
<th>Refutation of mutation carrier status</th>
</tr>
</thead>
<tbody>
<tr>
<td>But we (our family) do have a mutation; either</td>
<td>Maybe we have or maybe we don’t have a mutation and that the results are incomplete or inconclusive</td>
<td>And we do not have the genetic type of cancer.</td>
<td></td>
</tr>
</tbody>
</table>

The largest number of participants (11 out of 21) interpreted their results as ambiguity regarding mutation carrier status. The other two types are opposite, but not extreme opposites. That is, participants who interpreted their results as a confirmation to their mutation carrier status (six participants) understood that no mutation was found in either gene, but they still believed with certainty that they carry an inherited mutation. There was no doubt in their minds that they are carrying an inherited mutation while the participants who interpreted their results as a refutation to their mutation carrier status (four participants) did not doubt the outcome of their results and believed that they were not carrying an inherited mutation. As for the participants who interpreted their results as ambiguity regarding their mutation carrier status (11 participants), this group leaves room for uncertainty.

Although three types of interpretation were found in the participants’ account, some of the participants’ interpretations do not fit exclusively into one category. That is, in some parts of their interviews, participants would at times say, “But I know I have an inherited mutation, they just have not found it.” Later in the interview, they would add, “My daughters were glad to hear
that I do not have an inherited mutation.” As well, four participants’ interpretations moved at times from saying they were certain they had an inherited mutation to saying that “Maybe we do or perhaps we do not have an inherited mutation and vice versa.” As for the four participants who believed they did not have an inherited type of cancer, one would at times comment on how she understood that there is always a possibility she might carry some other yet-unidentified gene mutation, or perhaps the current technology missed her mutation but that for the time being, she is interpreting her results as a refutation to her mutation carrier status. Nonetheless, when I asked her specifically how she viewed her test results, she answered that she was found “negative – meaning that no mutations had been found in her two breast and ovarian cancer genes analysed.

How participants arrived at their interpretations of genetic testing results is discussed by presenting generic structures found to inform their beliefs about whether or not they have an inherited mutation. The generic structures represent key statements located in the participants’ experiences. The term “generic” in this context portrays how there is not one participant experience that captures all the structures described nor does one structure alone represent an individual’s experience. In general, a participant experience will present with some of the 13 structures derived from the synthesis of all the participants’ experiences. Hence, the generic structures represent a beginning point which individuals might have used to interpret and make sense of their result. They are generic as well in the sense that they do not imply any linear or causal model but provide rich, detailed description and interpretation to the phenomenon studied. Some people's interpretations are informed by generic structures usually found within one type of interpretation such as to explain why they believed they did not have an inherited mutation, while others used the same structures to explain why they believed they had an inherited mutation.
The participants’ interpretations and making-sense experiences were extensively coloured by the four possible interpretations in the letter they received from the Cancer Agency (provided below). Recall that the purpose of this study was neither to assess the content of the genetic counselling process nor to evaluate how test results were communicated. Hence, although I highlight part of the letter participants received, I only do so in the spirit of providing the context in which the participants’ interpretations developed when they tried to make sense of the test results contained in the letter.

The four interpretations offered to the participants were:

1. A mutation may exist in the region of the BRCA1 or BRCA2 gene that our lab looked at, but it has not been detected by our current testing method OR
2. A mutation may be present in the as-yet-untested portion of the BRCA2 gene OR
3. The responsible mutation may be in another, as yet unidentified, hereditary cancer gene OR
4. You do not have an inherited breast/ovarian cancer gene mutation, which means that your cancer diagnosis may have occurred by chance.

The Cancer Agency’s letter continues with, “We must highlight that this result does NOT mean that we have completely ruled out an inherited breast/ovarian cancer gene mutation in your family. Based on your family history of cancer, we recommend that cancer screening continue as previously recommended.” It ends by stating that, at this time, they do not have the necessary resources to undertake additional testing such as full sequencing of both genes.

The clinical interpretations above have also been reported in the literature (Bish et al., 2002a; Claes et al., 2004; Frost et al., 2004; Hallowell et al., 2002; Hallowell et al., 2004; Iglehart et al., 1998; Lynch et al., 1999; Peshkin et al., 2001; Wong et al., 2001). Peshkin et al.

53 Only 65% of the BRCA2 cancer gene was screened. The Agency explains that this region represents the surface area of the BRCA2 gene where most clinically significant mutations have been found to date.
54 For a complete copy of this letter, see appendix 1.
(2001) conclude that, when no mutation is found in an affected individual with a family history of risk for hereditary breast and ovarian cancer (HBOC), that several explanations are possible: (a) a mutation might still exist in the BRCA cancer genes tested but was not detected by the methods used, or (b) the individual might have a rare or as-yet-undiscovered mutation, or (c) that the individual and the clinic should consider that perhaps the person tested represents a sporadic case within a possible hereditary cancer family. Their clinical conclusions are quite similar to those proposed to the study participants by the Cancer Agency.

After receiving their letters from the Cancer Agency, the study participants were faced with making sense of their results. As Joyce put it, “I mean there isn’t much in the letter that they haven’t said to cover their butt, like my doctor said.”

This negative reaction is not surprising. When I asked others what their reactions were on receiving their results and reading the ways in which they could be interpreted, they used the following terms: dumfounded, completely let down, no relief, like receiving a nonanswer, knew their results would be negative (that no mutation would be found) because they had received them first over the telephone, felt the whole process was a waste of their time, left in limbo, and back to square one. The following quote describes many of the meanings attached to these reactions:

I wasn’t at the time thinking there might be another mutation that they haven’t identified. Like I was thinking it was like black or white, you know. And I’m thinking, okay if we don’t have the mutation then I’m home free. This was just an aberration – what’s happened in our family. It’s just the luck of the draw sort of thing. But, um, now, you see finding out has sort of tempered the happiness, I think, because I’m not black and white about it. Well, I’m thinking there is a mutation there, you know. I didn’t realise that there might be another gene there. I think ... I thought they had identified all of the genes. Like I thought the BRCA1 and BRCA2 were the only genes that there was. And then how it changed from black and white is when Dr. X said there is likely another mutation. And I thought, oh okay. (Joyce, PP3, field conversation, December 12, 2000)
Many participants said they had not realised that they might receive any results other than “You do have a mutation” or “You don’t have a mutation.” Similar findings have been reported in the literature. Some studies indicate that individuals expected that they would receive either positive or negative genetic test results and that some people were surprised when they did not get either (Frost et al., 2004; Hallowell et al., 2002; Peshkin et al., 2001). These authors, and some others, comment on how some people wrongly thought that receiving an uninformative or inconclusive genetic test result equalled receiving a confirmation that no mutation had been found (Bish et al., 2002a; Claes et al., 2004; Frost et al., 2004; Hallowell et al., 2002). This interpretation is correct only when an inherited mutation has already been identified in an affected individual within the family, and then another family member undergoes genetic testing for that specific mutation and is found not to carry it. This individual’s test result represents a true negative result.

The Making-Sense Experience

Having established that the participants interpreted their genetic test results in three main ways, I looked further to how they sustain such beliefs towards how they interpreted their results. Within the data, I searched for key statements that related to their beliefs. To do so, I asked participants to describe how they made sense of their results in light of their interpretation of them. Specifically, I asked “When you say you interpret your results as still having an inherited mutation – one that they have not found,” how do you make sense of such an interpretation and how do you make sense of the clinics’ conclusion that no mutation was found?"\textsuperscript{55} In this section, I describe the generic structures found to inform the type of interpretation given to their genetic

\textsuperscript{55} Depending on how individuals interpreted their results, the question would be formulated to fit their interpretation.
test results. The structures are group under seven general headings. The 7 general headings with all the 13 generic structures are:

1. **Doubt and uncertainty about the outcome of their results because of**
   1) Untested portion of BRCA2 gene.
   2) Being offered a more complete testing in the U.S.

2. **Too many to be coincidental**
   3) Too many cancers in the family to be coincidental.
   4) Too many deaths from cancer in the family to be all coincidental.

3. **Age at cancer diagnosis**
   5) Cancer diagnosed at the same age and at a young age.

4. **Attributing unique features to their mutation**
   6) Having a mutation that can only be transmitted to females or males only.
   7) Having a mutation that increases the risk of either breast or ovarian cancer but not both.
   8) Having a mutation that is specific to their ancestral lineage.
   9) Having a mutation that is weaker and that needs an internal or external trigger to activate cancer.

5. **Influence of time**
   10) Between testing and receiving results.
   11) Between individuals with cancer across generations and within the same generations.

6. **Resemblances among individuals with cancer**
   12) Similarities and differences among individuals with cancer.

7. **Presence of children**
13) Having children or not, whether children are young, and children’s gender.

When attempting to make sense of their experience with genetic testing, participants did so in the context of their previous experience with cancer and their family history with the disease (Hallowell et al., 2004) as well as within the context of their genetic testing experience. The complexity inherent in the four possible interpretations given to them in the letter from the Cancer Agency led many participants to draw on their lived experience to interpret and make sense of their test results.

The 13 structures found within the participants’ belief systems, although having no order of their own, led me to develop the following process that may be lived and relived differently by each participant. The process is viewed as a retrospective narrative of causal reasoning of self with a probable inherited mutation once one perceives they have a strong family history with breast and/or ovarian cancer. The process is influenced by the thirteen structures identified above. The process brings these structures together. It has seven stages:

1. Beginning to question the origin of cancers in the family.

2. Reflecting on the number of cancer cases in the family and the number of deaths from cancer.

3. Recognising the nonconformity of the number of cancers with what is observed in the general population.

4. Reflecting on one’s age at cancer diagnosis and those of others in the family.

5. Finding physical and behavioural similarities among individuals in the family with cancer.

6. Realising that the cancers in the family could result from an inherited mutation.
7. Becoming convinced that the family cancers must result from an inherited mutation when a new cancer diagnosis occurs in a young individual, or when an individual with a previous cancer experiences a second primary cancer or a recurrence.

The presence or absence of the thirteen structures influences how the process runs its course. The process of viewing self with a probable inherited mutation is never static because the people's lives are never static. Individuals' beliefs about their risk of carrying an inherited mutation change over time; they oscillate with each new case of cancer in the family and with absence of cases (for example no ovarian cancer). A suspicion of an inherited mutation increases with each new case of cancer in the family – even more when participants identified characteristics usually attributed to HBOC families, such as having both breast and ovarian cancer cases. Other characteristics that participants associated with a possible inherited mutation are having a family member diagnosed with breast cancer at a young age; seeing a pattern within the age at which individuals within the family develop cancer; noticing similar physical characteristics among those who develop cancer; and doubting and viewing uncertainty about the outcome of their genetic test results.

The findings that follow describe the 13 structures relating to how they interpreted and made sense of their results. My analysis focused only on the individuals who experienced genetic testing, not on the family member (the secondary participants) because they did not go through the experience. However, the content of their interviews were analysed and added when it contributed to the participants interpretation implications in their every day lives. As I describe each of the structures, I contrast some of the participants' stories to illuminate how the structures shaped the making-sense process. I do this by locating the participants’ making-sense process in their personal biographies and everyday lives, health and illness experiences – incorporating
their family members’ stories, for those participants whose family members participated in the study.

**Doubt and uncertainty about the outcome of results**

**Untested portion of BRCA2 gene.**

Much of the doubt and uncertainty about the interpretation of results came from the realisation that not their entire BRCA2 gene had been tested and, as well, from being offered the opportunity of further testing in the United States. Both of these statements were in their letter from the Cancer Agency (appendix 1). Recall that, depending on which year they were tested, their letter stated that either 65% or 72% of their BRCA2 gene had been tested, and that their entire BRCA1 gene had been tested. In discussing their making-sense experience in relation to their uncertainty in interpreting their results, participants often said that only a portion of their BRCA2 cancer gene had been tested and that perhaps the specific mutation in their family might lie in the untested portion of that gene. This is how Evelyn saw it:

> You know what is interesting, one thing that I’m thinking about is, as soon as I heard that it didn’t show up on 60 and 90 percent, my immediate reaction was, well, I’ll call those people from another genetic lab that tested the whole gene, my gene. I got the paperwork out next to my bed where all my piles of paper are and I still haven’t done it – which is interesting, because I suspect it is also going to be negative. So it is sort of like, why bother, but it is not like I’m afraid to do it. It is just I haven’t gotten around to do it. It is not because I am an obsessive-compulsive person in having my blood tested at two different labs, it is just that, you would like to know what that the whole gene says and not just part of it. (Evelyn, PP5, field conversation April 2001)

Evelyn’s reinterpretation of the percentage tested in each gene is common in other participants’ accounts. Evelyn’s response shows that she has reinterpreted the lack of testing of a

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56 One of the health professionals that work in the Hereditary Cancer Program explained to me that only certain sections of the BRCA2 gene, known as axons, were tested, as that was where most mutations of clinical relevance had been found thus far. Clinical relevance means that, if a mutation is found in any of these areas, it results in an increased risk of cancer. Mutations in the untested portions were highly unlikely and if some were found, no clinical relevance had yet documented for them. This explanation certainly made sense to me, but the participants did not receive this explanation in their letter.
portion of her BRCA2 gene (when she mentions 60%) to mean that her other BRCA1 gene had not been fully tested (when she mentions 90%), even though the Cancer Agency's letter indicated that all of her BRCA1 had been sequenced. Not only that, but she also reinterpreted the percentages. In the year that Evelyn had genetic testing, the Cancer Agency was screening 65% of the BRCA2 gene. While Evelyn recognises that no mutation was found in the areas tested, she nonetheless believes that the untested portion of her BRCA2 gene may say something different. She leaves room to doubt her results.

This doubt about results because one portion of the BRCA2 gene was not tested differs from the perception of women in Bish et al.'s (2002a) study. These authors found that, although they had screened only two thirds of the BRCA1 gene and none of the BRCA2 gene, when they told the 46 women that their results were inconclusive, the perception of these women that they were mutation carriers decreased 6 months after counselling. These women interpreted their results as indicating that no mutation was present. The authors do not say that their participants interpreted their test results in other ways. Contrary to Bish et al.'s findings, the untested portion of the BRCA2 cancer gene in this study acted as a catalyst for individuals to continue perceiving themselves with the likelihood of being mutation carriers.

Many other participants besides Evelyn believed that their genetic testing for BRCA1 and BRCA2 gene mutations was incomplete. They expressed a need for certainty and finality in their interpretation of their test results. They often expressed their view that the testing was incomplete by recalling what percentage of their genes had been tested, although very few restated the correct percentages as given in their letters. As van Zuuren et al. (1997) note, the reason people consult genetic services is to obtain certainty about their perceived likelihood of carrying an inherited mutation predisposing them and that of their family to cancer. Further
uncertainty was also lived by the study participants with the true outcome of their genetic tests by being told that a more complete screening of their BRCA1 and BRCA2 genes was possible through a private laboratory in the U.S.

**Being offered a more complete testing in the U.S.**

Many of the participants’ making-sense experiences evoked topics addressed in the letter they all received from the Cancer Agency. These were (a) the inability of current technology to detect all mutations, (b) the four possible ways to interpret their genetic test results, (c) the limited financial resources of the laboratory to undertake additional testing, and (d) the availability of complete assessment by an American laboratory.

Participants who interpreted their results as a confirmation of their mutation carrier status were more likely to interpret the opportunity of further testing in the U.S. to mean that the Cancer Agency acknowledged the possibility that its technology could not detect their inherited mutation, or that their mutation might lie in the untested portion of their BRCA2 gene. The Cancer Agency’s statement in the letter indicating that they could have a more complete assessment done in the United States should they wish to at a cost of approximately $3,850 CDN created much anger and confusion. Some of the participants felt betrayed for having invested their time and trust in the Hereditary Cancer Program, only to be advised that a more complete assessment could be obtained in the United States. The huge fee was interpreted by these participants to mean that the Cancer Agency must believe that they likely have an inherited mutation, otherwise why would they suggest investing such a huge sum of money in another test?
While Marcy believed both of her breast and ovarian cancer genes were tested as extensively as current technology could offer, she nonetheless told me that only 72% of her BRCA2 gene had been tested and that she therefore interpreted this situation to mean that the Cancer Agency did not have the technological sophistication to assess all of her gene and that was why they were offering her to have a more complete testing in the United States. To her, this left the possibility that a mutation could still exist in the untested portion. Here is Marcy’s explanation:

They said they did the test on both cancer genes as far as they can go and 72% they’ve got in brackets. But the other percentage, it could mean that there is a mutation, in that they just don’t have the sophistication yet in the test that they’ve done. (PP18, Marcy, field conversation, September 13, 2001)

To Marcy, until all portions of her breast and ovarian cancer genes are tested, there was still a possibility that a mutation would be present. Her letter from the Cancer Agency clearly stated that not all regions of the BRCA2 gene had been tested and that no laboratory could detect all mutations in the two genes tested; a view shared by Lynch et al. (1999). Lynch et al. believe that no current test is sensitive for all possible mutations.

To almost all of the participants, the untested portion of the BRCA2 cancer gene left room to doubt their genetic testing results. As Donna said, “One way they say no and the other they say 65%. There is still room for doubt.” This doubt was also fuelled by participants’ understanding that the Cancer Agency also suggested that, for a huge fee, they could obtain more complete testing in the United States.

Further, some participants questioned whether the percentage tested on the BRCA2 gene was arbitrarily chosen for each participant. Participants who interpreted their results as either

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57 In the year that Marcy had her test done, the HCP had increased the portion of the BRCA2 gene tested from 65% to 72%. Hence, the difference in percentage with earliest participant genetic testing and comments related to this issue.
confirming their mutation carrier or ambiguity regarding mutation carrier focused on the untested portion to explain their beliefs that they still might had an unidentified mutation. The following is an example of how a participant, Beatrice, combined the untested portion of her BRCA2 gene and the offer of further testing in the United States to form a new meaning.

I have thought about going to the States to have further testing, because apparently they could only test a certain fraction of my blood. It doesn’t test hundred percent. (Beatrice, PP6, field conversation, April 19, 2001)

Beatrice sees her genetic test results as incomplete rather than inconclusive. Although the explanation of the untested portion and the offer of further testing in the U.S. were in different parts of the letter, many participants who commented on both issues brought them together. They interpreted this combination of statements to mean that the Cancer Agency believed they carried a mutation. Some participants have also interpreted the offer of further testing in the U.S. to mean that the Agency did not have the technology to detect all mutations. Donna stated:

I don’t make sense of that letter... I laughed when I read that letter quite honestly... It does not make sense to me that a test would be offered if you cannot completely do the whole testing. I can understand them not having the money and all, but why offer the test? And I have just heard that this whole test has been patented by the United States. (Donna, PP14, field conversation, August 22, 2001)

Both Donna and her sister were diagnosed with breast cancer at the age of 49. Her mother passed away from breast cancer in her late 50s. Donna cannot put to rest the thought that she most likely has a mutation. She later specified how she would be willing to be tested in the U.S. if her daughter really wanted to know with certainty that the family carries a mutation. Hence,

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58 Donna is referring to the patent of Myriad Genetics, a Utah-based biopharmaceutical company that has patent and testing rights to BRCA1 and BRCA2 genetic testing. Through an agreement with a Canadian laboratory, MDS, Myriad has given MDS exclusive rights to provide this testing. This news came about at the end of the period in which I conducted the interviews. The resulting moratorium on testing did not affect the women I interviewed but it limited access to testing for those coming after them. The moratorium on BRCA testing began in July, 2001. The Hereditary Cancer Program was forced to stop testing by the B.C. Ministry of Health Services. Other Canadian provinces immediately opposed the cessation, such as Ontario and Quebec. Although Ontario and Quebec never ceased to offer genetic testing, British Columbia resumed testing in January, 2003 by sending blood samples to Ontario (Kent, 2003). For a more complete history of the patent debate surrounding BRCA testing, see Williams-Jones’ dissertation “Genetic testing for sale: Implications of commercial BRCA testing in Canada” (2002).
the test results did not give her an answer nor were they final to her in determining her mutation carrier status. As Donna concluded, if she saw her results as affirmative to either confirming or refuting her mutation carrier status, she would not contemplate further testing in the U.S.

Some participants took different parts of the letter out of context and put them together to form a new whole. They then reinterpreted this new version in ways to show how uncertain their results could actually be. In reviewing Donna’s comments in her interview transcript on the letter received by the Cancer Agency (refer to appendix 1), we can see that she has mixed some content of the letter taken from the top of page 2 with other content taken from the bottom of that same page. She refers to the first two statements made at the beginning of page two and the last statement made on that same page. The two first statements discuss the availability of “a more complete assessment of the BRCA1 and BRCA2 genes,” then refers to another statement within the same paragraph that explains, “We estimate that there is a less than 10% chance that additional testing will identify a mutation that is known to be clinically significant.” Finally, she puts together a comment made in a footnote at the bottom of the second page that says, “This DNA sample was screened for mutations in 100% of the coding sequence of BRCA1 and 65% of BRCA2…” with the last sentence of that paragraph, which reads, “We do not have the resources to undertake additional testing at this time.” While Donna questioned why the Cancer Agency would offer genetic testing if they could not complete it, what the letter implied was that their method used of testing for mutations differed from what is offered in a private laboratory in the U.S.

Although I chose to present only Donna’s comment, her view of the untested portion reflects many similar comments by other participants. First, much confusion arose from the note explaining that only a certain percentage of the BRCA2 gene had been tested. Many of the
participants understood this to mean that the Agency could not complete their testing because of a lack of funding or because it did not have the technology to conduct full tests on both cancer genes. Perhaps the participants would have been better served had the letter described why only a portion of their BRCA2 gene had been tested. As said earlier, only one study documented individual’s perceptions of having had only a portion of their breast and ovarian cancer genes tested. This study indicates that people did not see this untested portion as creating doubt about the certainty of test outcomes (Bish et al., 2002a).

Here is another participant’s interpretation of the untested portion of her BRCA2 gene to mean that the Cancer Agency seemed to have run out of funding to complete her testing:

I don’t think the results have actually made any difference because the first one, it took about 2 years and then they told...I can’t remember what they actually told me but it was just to say that they were in...They had to do another set of tests and I got this health professional saying, ‘Well, sorry about the test. We had to do it up through funding and if you want to know any results, just go to the American Cancer Society.’ So it was really weird, you know, so like ‘Okay, take your blood, give enough funding and we would have to pay for it down in the States.’ It’s like...I don’t know...I’ve got cancer so...I really don’t know the results. I don’t really see any results. (Red, PP4, field conversation, June 28, 2001)

Red feels that her test results are incomplete. When she was told over the telephone that the HCP would have to do the same test on her BRCA2 gene as on her BRCA1 gene, she interpreted this to mean that a new set of tests had to be done on both genes. This created even more confusion about actual results on the BRCA1 gene. However, based on my field observations and exchanges with health professionals from the Cancer Agency, when Red received a call to inform her of the results for her BRCA1 and BRCA2 gene tests, she also would have been informed that she could obtain a full screening of both genes at Myriad Genetics in the U.S. Being unaware of the relationship of Myriad Genetics to her genetic testing at the HCP, it was not this name that Red remembered but a cancer agency in the States, the American Cancer Society. Consequently, Red interpreted the offer to mean that, if she wanted any results, she
should contact the American Cancer Society and pay for them herself. She later argued, “What is the point of getting people started in testing or getting them to participate in research if you do not have enough funding to complete the study?” In the end, Red interpreted her results as ambiguity regarding her mutation carrier status and made sense of her interpretation by saying that she does not know the true outcome of her results as she does not see any.

As discussed by Frost, Venne, Cunningham, and Gerritsen-Mckane (2004), uncertainty is not an uncommon element with genetic testing, as it is with some other forms of medical testing. While some genetic tests can provide certainty about some disorders (such as testing for carrier status of cystic fibrosis or Huntington disease), with testing for BRCA gene mutations, even those who receive positive results only know with certainty that they carry an inherited mutation but not if they will eventually develop breast and/or ovarian cancer. Their risk of developing breast and/or ovarian cancer still remains uncertain; it is only a probability value. Conversely, individuals within HBOC families who receive negative results actually do obtain certainty, as their results represents a true negative –the individual does not have the identified familial inherited mutation and their risk of breast and ovarian cancer is less than family members found with the mutation (Lynch et al., 1999).

Perceiving a lack of quality and finality in the outcome of their results, some participants began to wonder if their whole experience was part of a research project, in which, in their view, some tests would intentionally be left uncompleted.59 This perception seems to not only stem from the untested portion of their BRCA2 genes but also from the multiple interpretations options offered to them in the letter. Recall that some participants described that they view

59 Although the availability of genetic testing in Canada mostly occurs through research funding, the HCP considers this test part of their clinical services and presents it as such to the population.
agreeing to be tested as entering one more clinical trial for which they had met eligibility criteria, just like other trials they had participated in.

Louise, diagnosed with ovarian cancer at the age of 23 described her experience as being part of a study.

I think at the end with the results, I was a little confused, not too sure if the study was even conclusive. But then I found out that I am kind of like in the middle. (PP13, field conversation, July 18, 2001)

This statement contains malaise. Louise struggled with the notion that, if this was a research study, then it was possible that the results might be inconclusive. Had it been a real test, she would have received a real result.

Louise’s interpretation of her test results above falls into the ambiguity category regarding mutation carrier status. However, she did express later in the interview that she felt relieved finally knowing where her ovarian cancer came from: an inherited mutation. Hence, Louise oscillated between interpreting her results as confirmation of her mutation carrier status to ambiguity regarding mutation carrier status. When asked how she came to this conclusion, she said the test results confirmed there was a possible inherited mutation within her family. Although her interpretation is accurate, it represents only one of the four possible ways in which her results could be interpreted.

The reason participants felt that their genetic testing was done as part of a research study could flow from an assumption that, if a medical test was offered to the general population it would be capable of providing definitive answers (Burke, Pinsky, & Press, 2001), rather than four different ways to interpret the results. Hence, Donna concluded that, because her testing was done as part of a research study, perhaps the Cancer Agency did not have the best technology available. Had it been real clinical testing, then perhaps the Agency would have used the same
equipment as in the United States. Thus, Donna believes that she carries a mutation but that technology used by the Agency could not find it. While this participant is not totally wrong, all participants had been informed by the Cancer Agency that no current testing can detect all mutations and that the Agency believes their false negative rate to be less than 10% for this type of test.

Barney, a participant, expressed how upset she was that the Cancer Agency offered a test that could not provide a definitive answer. Barney strongly believed that her family had an inherited mutation predisposing them to breast cancer and therefore felt that the Cancer Agency should have been able to find this mutation if they offered her testing. She did not undergo genetic testing to find out if she carried an inherited mutation but to confirm her perceived genetic risk. Hence, she concluded that the Cancer Agency did not have the proper technology to conduct her gene analysis properly. Her belief that the Cancer Agency did not have the proper equipment to be able to identify her suspected mutation was further reinforced by her interpretation of the Cancer Agency's proviso that she could send her DNA to the U.S. for a more complete assessment. She interpreted this to mean that genetic testing agencies in the U.S. must know "how to find mutations there whereas Canada does not."

Well I think they should have found something. If they can find the genes in the States, why can't they find it here in Canada? I call it genes. With the States, you will get results and they are finding it down there, so why can't they find it here? Why can't the two get together and tell Canada how they are finding the genes down there? I mean cancer is cancer no matter where you go in the world. I read in the paper that you can get the testing done here and they will send it to the States. It will cost between $400 to $6000. Well, why don't they get together and find out instead of having to send down and pay for it? I'm not going to send that money down to pay for it; my kids can't afford to do that. It's not right if you can't find out if they're going to get it. It should be done down here. I think they should find out how to do it here. You get enough funding, well don't they get enough funding? I think they are wrong here. There's got to be something there, it's hereditary and they said, okay, well send it down, you can send it to the States. Are the States going to find it where we can't? Why? (Barney, PP7, field conversation, April 12, 2001)
Only a few participants understood that no medical test could provide complete certainty. In her attempt to make sense of her results in light of the uncertain outcomes with every medical test, Ginger gave the following interpretation:

That doesn’t say that I am totally free of it, just that they couldn’t find anything. BRCA1 and 2 are just one type of genetic mutation. And maybe they weren’t able to find it because of their limited ability. But if I really don’t have those two, it doesn’t mean that I don’t have something else. (Ginger, PP11, field conversation, July 5, 2001)

Ginger took a pragmatic approach to the interpretation of her results. In her statement, she reflected the uncertainty established by the Cancer Agency in their letter by providing four possible interpretations and stating that no laboratory can detect all mutations. Being a health professional, she has most likely experienced that very few medical tests can provide absolute certainty.

The uncertainty of their results may explain why the majority of participants still believed that they may carry an inherited mutation. Even when it was not the untested portion of their BRCA2 that they doubted, they also believed that perhaps the laboratory did not find their mutation because it lies within aspects of a cancer gene not tested. Bubaloo interpreted her results to mean that perhaps she had an inherited mutation that the current technology could not find. In effect, in the following quote, Bubaloo spoke about the accuracy of the test, also known as its specificity and sensitivity.

If and when I say my results are negative, I mean to say that they did not find anything. It is not to mean that the mutation is not there. They just have not found a way to locate the mutation or to an absolute 100%. From what I understand, there are so many facets to our genes that all they have done is got a miniscule test of what they can actually test for. Hence, my results do not mean a mutation is not there but only that the technology is not to the point of being capable to find the mutation. However, the gene might show up soon or whenever they come up with more sophisticated equipment to test for the gene. (Bubaloo, PP9, field conversation, July 24, 2001)

Thus far, I presented two of the thirteen generic structures constructed from the participants’ making-sense experience that inform their interpretations of their test results. These
two generic structures – were compiled under the general heading of “doubt and uncertainty about the outcome of results.

**Too many to be coincidental**

Too many cancers and deaths from cancer in the family to all be coincidental.

The two structures, the number of breast and/or ovarian cancer cases in the participants’ family history and the number of deaths from cancer, were often intertwined when participants used them to make sense of the way they interpreted their results. Naturally, participants who have large families are more likely to have more cases of cancer and more deaths from cancer than participants from small families (no siblings or cousins). Nevertheless, participants from small families still noted how unusual it is that “all the women in each of the last three generations” were diagnosed with breast and/or ovarian cancer, when there is only one woman in each generation. They began to speculate that this pattern is not due to chance alone.\(^\text{60}\) When participants considered that there are just too many cancers in their family to be all coincidence, they contrasted their perception to the number of deaths as a result of cancer, slowly building their case for a possible inherited mutation in the family.

I think they’ve missed it. I think it probably is there. As the letter says, it may not be BRCA1 or 2, but one is there. How else can you explain that I was only 23 when my mother died of breast cancer and I had just watched my father die of lung cancer? My mother developed cancer in the same year my father died. She was the first one of her sisters that came down with it (breast cancer), and then it was one sister after another. And the only sister that missed, her two girls got it at the same time I had; two out of three girls. The oldest girl ended up having a double mastectomy, because she wasn’t going to go through what her sisters did...cause you see, one of the girls died from her breast cancer. (Joyce, PP3, field conversation, December 19, 2000)

Joyce explained that she is convinced that her family must have an inherited mutation. In her view, what else could explain the 12 cases of cancer on one side of her family – 10 of whom had breast cancer and only three of whom survived? The only reason she believed she survived

\(^{60}\)See Stephanie’s pedigree (PP12) in appendix 5 for a visual understanding of this interpretation.
was because she had a mastectomy after her second diagnosis of breast cancer. Joyce further explained that the other members of her family probably believed as well that they carried an inherited mutation, whereas one of her cousin decided to have prophylactic mastectomy while her maternal uncle, who following the discovery of cysts in his breast had a bilateral prophylactic mastectomy.

In contrast to Joyce’s belief that her family must have an inherited mutation because of the number of deaths from breast cancer, a study by Verhoog et al. (1999) suggest that no correlation exists between the survival rate in sporadic cases of breast cancer compared with carriers of BRCA1 and BRCA2 mutations. However, what Joyce implied in her lay theory was that, while she recognised that perhaps her family may not carry an inherited mutation on their BRCA1 and BRCA2 genes, they may still carry some other yet-unidentified hereditary cancer gene mutations that would explained the multiple cases of breast cancer found in her family and the deaths from this disease.

Inevitably, the number of women in any given family increases the potential number of breast and/or ovarian cancers cases. After all, being female is a major risk factor in developing breast and/or ovarian cancer. Therefore, a large number of breast and/or ovarian cancer cases in a large, predominantly female family could be expected in Joyce’s family. Still, the number of breast and/or ovarian cancers within a large family can still seem overwhelming. In defining small and large families, I did not necessarily count the specific number of family members, but tried to go with the participants’ definition of their family’s size. Joyce’s family has many females, and she defined her family as large. As Joyce added, “I mean, I’m sure some families may have maybe, you know, one or two aunts and a mother with breast cancer maybe, but not five in the generation before you.” Individuals from families at risk of HBOC in other studies

61 See Joyce’s pedigree (PP3) in appendix 5.
have expressed the same view – that having so much cancer in one’s family leads one to think that their family might carry a mutation (Frost et al., 2004; Verhoog et al., 1999). There are also studies that conclude that having a strong family history of breast and/or ovarian cancer is an important risk factor in having an inherited mutation (American Society of Clinical Oncology, 2003; 1996; Newman et al., 1998; Oddoux et al., 1996). While having multiple cases of breast cancer in a family may lead individuals to think that perhaps the cancer occurrences may be the result of an inherited mutation, studies such as Ford et al. (1998) and Frank et al. (1998) indicate that 16% to 66% of high-risk families are found not to carry a detectable mutation in either of the two genes. Further, Hopper (2001, p. 367) notes that the “‘typical BRCA1/2 family’ consists of a ‘sporadic case’, not multiple-case family.”

Even for those whose family history of cancer is not from past generations but more so within their own generation, such as Emma’s case, when the number of cancers seems too high to be considered bad luck, this convinces the participants that an inherited mutation might be responsible.

So what they are saying is that, since my two sisters and I all had breast cancer, there is likely a reason. (Emma, PP20, field conversation, October 27, 2001. Emma was diagnosed with breast cancer at the age of 43. Both her sisters were diagnosed with breast cancer at 42 and 53 respectively, and both her sisters died as a result of their cancer. All three cases of cancer appeared within 8 years of each other.)

When Emma said, “they are saying,” she was referring to health professionals in the Cancer Agency who informed her of the test results. Hence, her interpretation of the results was based on, among others, her understanding of what she has been told, and also through her own experience with breast cancer and her two sisters’ breast cancers and deaths. Emma believed that, although the Cancer Agency staff said her test results were “negative to BRCA1 and

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62 See Emma’s pedigree (PP20) in appendix 5.
BRCA2 gene mutations”, the results did not show that there is no genetic component to the three cancers in her immediate family.

Both Joyce and Emma came to their belief that they had an inherited mutation not just on their own experiences with cancer but in relation to others in their families who developed cancer as well. Hence, it is no longer the self alone that was assessed for possible inheritance of the disease but the self in relation to others. The self comes to represent the whole (for example, the three sisters in Emma’s case), and it is this new self that is compared against the possibility that their family may carry an inherited mutation. D’Agincourt-Canning (2003) also found that participants of her study compared the self with others in the family to assess their genetic risk of cancer.

Sherry, who was from a large family, explained that they must have a defective gene, “I just don’t believe there could be this many coincidences.” When Sherry said that there cannot be this many coincidences, she was referring to the number of breast and ovarian cancer cases in her family. During the interview, Sherry shared that there were eight cases of breast cancer in three generations to date, with one person having two primary cancers. Among these cancers, six were breast cancers, although one case was currently undiagnosed but presumed to be breast cancer and the most recent was an ovarian cancer diagnosed in her niece at the age of 34.63

Considering her family’s history of cancer, Sherry agreed with what she heard in her genetic counselling session: that there must be a “defective gene in her family.” Further, relying on what she understood about the possible interpretations in the letter from the Cancer Agency, Sherry concluded that there must be a mutation but the Agency could not locate it or that perhaps the mutation lay in the untested portion of her BRCA2 gene. Within the making-sense process of

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63 See Sherry’s pedigree (PP21) in appendix 5 for a visual account of her family history with breast and ovarian cancer.
self with a probable inherited mutation – which represents the retrospective narrative of causal reasoning presented on page 136, Sherry was past stage 6 of coming to the realisation that the cancers in her family could be the result of an inherited mutation to being convinced that there is an inherited mutation in her family. Sherry’s interpretation illustrates the temporal aspect of the process. She explained, as many other participants did, that, for the time being, the family’s mutation could not be located.

Having multiple cases of breast and/or ovarian cancer in the family also made it more likely that individuals noticed a pattern of cancer and thus suspect an inherited mutation. Seeing possible inheritance of cancer, participants tended to interpret their results as a confirmation of mutation carrier status. Even among the participants coming from small families, if they observed a cancer pattern, they tended to think that a mutation was responsible. Also, if the current family pattern of cancer seemed to replicate past ones, participants were even more inclined to believe that the cancers resulted from an inherited mutation. Sherry spoke to this issue.

In my family, it is two girls out of four who got breast cancer, which is 50% who have had it, 50% who have not. To me this indicates it may be a gene, you know, two have and two have not. Um, yeah, I do think the numbers are there and there’s going to be more. Yeah, I do expect that. And in my niece’s family, she had breast cancer, her sister has ovarian cancer, she’s got two other sisters who are fine so far. (Sherry, PP21, field conversation, September 2001)

Sherry’s lay theory of inheritance within her family is the following: 50% of women within the same generation can expect to develop breast and/or ovarian cancer because her family history carries both types of cancer.

Stephanie, who comes from a much smaller family compared to Sherry, still saw a pattern of breast cancer within her family that led her to believe that “it runs in her family at

64 Like Sherry, many participants used having a gene to mean having an inherited mutation.
According to her lay theory, one woman in each generation will get breast cancer, regardless of whether she has an identified inherited mutation. As Stephanie said, she “never did think it was not hereditary” until she had genetic testing done. After, she received her results, she added the possibility that their cancers may have resulted from “bad luck,” given that the Cancer Agency offered that as one possible interpretation. Stephanie’s interpretation of her results oscillated between viewing them as confirmation to her mutation carrier status to ambiguity regarding her mutation carrier status, although she still could observe a cancer pattern within her family and therefore now worried about her only daughter. Here is how she viewed her family cancer pattern:

One hundred percent in our family in the last three generations – every woman. Now there has only been four in three generations. My grandmother was the only girl, my mother and her sister were the only girl, and I am the only girl... Now my daughter she is 20 and she is the only girl. I just can’t imagine what it must be like for her now to be fourth generation, and every woman has had it. I grew up with that knowledge. (Stephanie, PP12, field conversation, July 18, 2001)

Cognitively, Stephanie acknowledged that, at this time, there is no genetic explanation for her cancer, but emotionally, she never questioned that her cancer and others in the family were not hereditary. The making sense of cancer-occurrence patterns within one’s family has also been documented by Richards (1996).

The next excerpt is from Becky, who comes from a rather large family. The breast cancer history was on her father’s side. All of her father’s three sisters developed breast cancer in their 60s, while all of his four aunts developed breast cancer in their 80s. Becky’s diagnosis came at 52. Although Becky sees a pattern in the age at which the women developed breast cancer (10 years earlier with each generation), she did not attribute this pattern to a possible inherited BRCA 1 or 2 gene mutation. Becky’s interpretation of her results oscillated between seeing

65 For a visual display of the age at which breast cancer appeared in each generation of Becky’s family, see her pedigree (PP8) in appendix 5.
results as refutation of her mutation carrier status to ambiguity regarding her mutation carrier status despite having a strong family history of breast cancer. However, when I asked her how she interpreted her results, she said that she was not found with a mutation. Although there have been many breast cancers in her family and six out of the eight caused death, Becky drew her interpretation from her understanding that families with an inherited mutation of either the BRCA1 or 2 genes will also have a history of both breast and ovarian cancer. Not seeing any ovarian cancer in her family history, she concluded that she does not carry an inherited mutation while not completely relinquishing the possibility that perhaps her family has some other yet unidentified mutation that increases their breast cancer risk.

Just looking at the overall, the results of the testing combined with my history, I just have a belief that we probably don’t have those genes because of the late onset, no ovarian. We don’t fit the pattern. But likely it could be another gene. But if it is, well, it sounds to me like it is not as awful as the early onset breast cancer and the ovarian cancer link. (Becky, PP8, field conversation, April 17, 2001)

Following her genetic counselling session in which she was told of the increased risk of developing ovarian cancer when one is found to have an inherited mutation of either the BRCA1 or 2 gene, Becky suspected that no mutation would be found in her BRCA1 and BRCA2 cancer genes because she did not see her family history fitting the pattern of hereditary breast and/or ovarian cancer. Becky also spoke about her understanding that, if a family is suspected of carrying a BRCA1 or 2 gene mutation, then it would be more likely to have early-onset breast cancer in the family history. She asserted that all the breast cancer cases in her family were late onset, although Becky mentioned that she was pre-menopausal when she was diagnosed with breast cancer. Becky recalled from her counselling session that the health professional had said her family did not really fit the pattern of inherited breast cancer but because breast cancer was on her father’s side, the clinic was still interested in having her do the test. Although Becky
tended to believe that a gene mutation did not cause her family’s cancers, she struggled with what seems to be her family’s reality. In her view, breast cancer was occurring 10 years earlier with each generation. This would mean that her daughter may get breast cancer in her 40s.

Mimi – who comes from a small family with no history of breast or ovarian cancer before her three primary breast cancers and her sister’s breast cancer – also saw her situation as too unusual to be just a coincidence. However, she interpreted her results as indicating that she may not have an inherited mutation. Like Becky, Mimi struggled to make sense of her three breast cancer diagnoses. She explained how unusual it was to her to see cancer occurring in a family as health conscious and physically active as hers. She, her mother, father, husband, children, and nieces all run marathons.

Mimi was aware of the value of keeping healthy and fit in preventing cancer. At times Mimi had to admit that her cancers could be the result of a mutation and/or an interaction of different reasons. Mimi felt that cancer was caused not just by one factor but by a multitude of factors. However, being so health conscious, she could not think of any environmental reasons that could have caused her to have cancer three times. Contrary to other participants, she could not look to her family history to explain the appearance of cancer in herself and her sister. “Exactly what it is, nobody knows,” she said. Nobody could tell her if it was “because she drank too much coffee in the morning, or because she did not have that extra glass of milk.” It would seem reasonable for Mimi to conclude that, being so health conscious, her three breast cancer diagnoses were the result of an inherited mutation. However, lacking a family history of the disease, she doubted this possibility.

All the participants understood that an assessment of genetic risk for cancer was done by looking at only one side of the family. There was a clear understanding of the importance of
lineage when establishing a link between the cancers in a family. Barney specifically stated that, although her mother’s brother is her half-brother, his cancer diagnosis still counts because he is from the lineage in which breast cancer was found – that he has her “grandmother’s blood.”

The establishment of a possible pattern of inherited cancer by the participants reinforced their view towards having an inherited mutation or to being ambiguous of their mutation carrier status. Despite participants’ certainty about patterns of inherited cancer in their families, I still pursued with participants whether it was possible for them to consider the fourth possible interpretation presented in the letter from the Cancer Agency: your cancer diagnosis may have occurred by chance. Evelyn confirmed my suspicion that it was difficult for participants to consider this possibility. Evelyn had nine cancer diagnoses in her family in the past three generations but on both side of the family. During the interview, I asked: “Evelyn, do you at least have some peace of mind that the Agency says that you may not have an inherited mutation in your family?” She replied:

No, because I probably fit in the familial somewhere in the middle, maybe a recessive gene that has not been located yet. That would make sense, considering cancer is on both sides of the family. But then, that would put my sisters at 25% risk instead of 50% risk, so yes, a recessive gene. And I’ve read somewhere about genetic breast cancer, familial breast cancer and nonspecific or whatever they call it, no history. Because the BRCA 1 and BCRA 2 are autosomal dominant, it’s unlikely that we have that because my mother is 70, almost 79 and hasn’t had breast cancer. Okay, so it’s unlikely if it is, I’ll carry it in an autosomal-dominant manner. It’s clearly familial, because clearly there’s a fairly strong, you know, association of first, and second degree-relatives so it’s sort of in that middle-type thing and particularly, I mean I look at their list, bilateral breast cancer before the age of 50, um, you know; two primaries in each of two seconds degree relatives, you know. I mean it doesn’t take a rocket scientist but, but there are people that had even more profound family histories than I did that tested negative also. So I think there’s, well, early breast cancer is, is genetic, you know. I mean there was a genetic predisposition that was triggered by something, but it’s not one of those two genes probably. Not sporadic, I don’t think it’s sporadic. So I think it’s familial and, and whether or not it’s hereditary, it doesn’t, it clearly doesn’t seem to be autosomal dominant. Although my mother could have the gene and just be one of the 20% that
hasn’t showed it, you know, shown it. So that’s a possibility. (Evelyn, PP5, field conversation April 2001)

This statement contains Evelyn’s explanation of why she does not consider the breast cancer cases in her family to have occurred by chance. She invokes the different categories of breast cancer (hereditary, familial, spurious) and the theory of inheritance (autosomal dominant inheritance versus recessive genes). The theory of inheritance says that if a cancer is spurious, it is unlikely that one individual will have more than one primary breast cancer. If cancer is not inherited, there is less risk of seeing breast cancer in a male in the family. Evelyn has both characteristics in family history. The theory of inheritance says, as well, that BRCA1 and BRCA2 mutations are carried in an autosomal-dominant fashion; hence, cases of cancer are usually seen on the same side of the family – unless both sides carry an inherited mutation.

Evelyn’s family history shows cases of breast cancer in both sides of her family. She pragmatically used the theory of autosomal-dominant inheritance to explain why she believes that the cancers in her family may not be from these two autosomal-dominant gene mutations but, more likely, from a recessive gene. Evelyn has three different types of cancer in her family history on both her maternal and paternal sides. There are three cases of breast cancer on her maternal side, including herself (with two bilateral primary cancers) and one aunt (with both breast cancer and melanoma). On her father’s side, Evelyn’s grandmother had breast cancer, while her father and uncle died of esophageal cancer and the uncle’s daughter developed fibrosarcoma at the age of 20. The cancers on both sides of the family explain Evelyn’s view that she may carry a recessive cancer gene not yet identified. The lack of a pattern on one side of the family reinforces her view that she and her family do not carry an inherited mutation of either BRCA1 or 2 but does not dismiss the possibility that her family may carry some other inherited mutation predisposing them to cancer.
A definitive family history of breast and/or ovarian cancer does seem to reinforce participants’ beliefs that there must be an inherited mutation to explain the occurrence of all the cancers. A common denominator among those who think that there are just too many cancer cases in the family to all be the result of coincidence and who doubt the outcome of their test results is the appearance of breast and/or ovarian cancer, and sometimes other cancers, in a young family member.

**Age at cancer diagnosis**

*Cancer diagnosed at the same age and at a young age.*

Having many family members and relatives diagnosed at a similar age was a defining factor in their belief to having a possible inherited mutation. When many members of the same family were diagnosed at a similar age, participants explained how they feared for family members when they approached what they considered to be the critical age – especially those with features similar to affected family members. Evelyn spoke about this critical age.

Because my aunt was 46 and I was 46, I got really nervous when they (my two sisters and favourite cousin) turned 46. They made it through fine, they’re 49 now, you know. So, it just seemed kind of ironic. (Evelyn, PP5, field conversation, July, 2001)

Barney, with four women in her family history diagnosed breast cancer at similar ages stated:

Well, you see all the four women in my family diagnosed with breast cancer were all in their 40s and were also all diagnosed in the same breast. (Barney, PP7, field conversation, April 12, 2001)

For Barney, turning 40 is a critical age to find out if one has inherited the disease because her lay theory is so strong. Barney explained that she entered genetic testing not to be told if she had an inherited mutation or not but to confirm what she already knew. She, like other participants who believed their family carried a mutation, was convinced that she carries it too because of the similarities she saw among those in her family who had been diagnosed with cancer. This has
been described as *perception of proneness to cancer* by Richards and Ponder (1996) who, through hypothesis testing, concluded that lay theories of inheritance are derived from social relationships within kinship.

Several other studies also found that individuals' perceived susceptibility to familial cancer increased when they approached the age at which others in the family were diagnosed with the disease (Chalmers & Thomson, 1996; Frost et al., 2004). This finding was also observed among individuals with a family history of Huntington Disease (Cox & McKellin, 1999; Richards & Ponder, 1996). However, contrary to multi-factoral diseases such as breast and ovarian cancer where the basis for similar age of onset with disease occurrence is less, with Huntington Disease, there is a correlation of age of onset with the repeat length of the CAG gene associated with disease risk (Chen, Ferrone, & Wetzel, 2002).

While participants' age at cancer diagnosis had an impact on how they made sense of their experiences, not all diagnosed at a young age believed that their cancer must be the result of an inherited mutation. What created the difference was the presence of many breast and ovarian cancers within the family, with some individuals being diagnosed at a young age and some having multiple primary cancers. When this pattern was present within the family, participants were more inclined to firmly believe they had an inherited mutation.

Many of the participants defined early breast cancer as that occurring before menopause. Individuals with early-onset breast cancer are the most likely to carry an inherited mutation (Ormiston, 1996). During their interviews, I asked participants if their breast and/or ovarian cancer had occurred before menopause. Out of the 21 participants, 19 said their cancer(s) occurred before menopause. Among the two who said their breast cancer occurred after menopause, Gladys firmly believed that neither she nor her family carried an inherited mutation
of either breast/ovarian cancer gene. Gladys has two sisters and a mother diagnosed with breast cancer; all three cases occurred after menopause. Although Marcy shares a similar breast-cancer family pattern (two of her sisters were diagnosed with breast cancer after menopause) her interpretation of her results was as viewing them as ambiguity regarding her mutation carrier status. The difference between Gladys' and Marcy's interpretation is that Marcy mentioned four more cases of cancer among her cousins, two of which are breast cancer. Gladys did not identify any cousins with cancer. This suggests again the influence of having many cancer cases in the family on one's making sense experience to interpreting genetic test results.

Among the other 19 participants who said their breast and/or ovarian cancer occurred before menopause, all but Juniper and Erika continued to believe that they may have an inherited mutation. Juniper was diagnosed with breast cancer at 45 and Erika at 35. While they each view their cancer diagnosis as unusual because of the age of occurrence, they nonetheless did not believe they carried an inherited mutation, because they felt they lacked a family history of breast cancer. Although this may be true for Juniper, Erika actually had four cases of breast cancer, including her own, in her family history. She said, however, that she felt little physical or emotional connection with her extended family members because they live in another country and did not entertain any relationships with them.

Comparing age of diagnosis with type of interpretation, out of the nine women diagnosed at or before 40, all but one either interpreted their results as confirmation of mutation carrier or ambiguity regarding mutation carrier. Finally, as introduced at the beginning of this section, the age at diagnosis structure, by itself, did not seem to influence whether or not participants believed they have or have not inherited mutation. However, it did influence beliefs when put in combination with other structures, such as past family history with breast and/or ovarian cancer.
While participants diagnosed with breast cancer at a young age and who had multiple cases of cancer in their family were more inclined to interpret their results as most likely having an inherited mutation, interestingly, a case-control study by Newman et al. (1998) found that diagnosis at young age alone did not predict BRCA1 carrier status but that in contrast, BRCA1 mutations were sufficiently frequent in families with both breast and ovarian cancer, or with at least four cases of breast cancer, at any age (Newman et al., 1998). Hence, individuals need to be supported in their increase perception of risk to an inherited mutation as a result of having received a breast cancer diagnosis at a young age and need to be informed that not all breast cancer diagnosis at a young age are the result of an inherited mutation.

**Attributing unique features to their mutation**

Four structures lie under this general heading. In general, these structures speak to the participants' understanding of genetic risk and how they construct their own theories about it. A study by Cox (1999) found that individuals with a family history of an inherited disease seldom make sense of their hereditary risk using Mendelian theories but do so by constructing theories within the context of familial beliefs that highlight social, biographical and temporal factors. The four structures, 6 to 9, are presented on page 135. These include having a mutation that can be transmitted to males or females only, that increases the risk of either breast or ovarian cancer but not both, that is specific to their ancestral lineage or that is weaker and needs an internal or external trigger to activate cancer. Although I try to separate these structures, participants often combined them to explain how they made sense of their genetic test results.
Having a mutation that can be transmitted to females or males only.

Many participants found it difficult to comprehend how men in their families could inherit a mutation known to be associated with a female disorder and pass it to their children yet never develop the disease themselves. Joyce expressed this as follows:

The thing is there are two girls out of that family where one died of breast cancer. And my uncle has been fine. That is his two girls. But he was the only boy in that family, so why did he pass it on?” (Joyce, PP3, field conversation, December 19, 2000)

The lay theory formed by some participants that a breast and ovarian cancer gene mutation can be passed down through males as well as females prompted some participants to reinterpret their results. Becky, whose family history includes only breast cancer, concluded that, because the breast cancers were on her father’s side of the family, she must have a weak gene but not a BRCA1 or BRCA2 mutation. Her conclusion was reinforced mainly by two beliefs. First, if her family had a BRCA1 or 2 mutation, ovarian cancer would have appeared (it did not). Second, she believed that her breast cancer most likely resulted from a “fluke” as she put it, because her family history of cancer comes through her father. On the other hand, she sees her paternal cousins as being much more at risk of carrying an inherited mutation, even one associated with BRCA1 or 2 because their family history of cancer comes through their mother (Becky father’s sisters). Becky’s family history of cancer consists of the following:  

Within her father’s generation, all three of her father’s sisters developed breast cancer in their 60s, while his one brother developed bone cancer. Two of his sisters died as a result of their disease. Her father died at the age of 76 from causes other than cancer. She added that her father’s mother also died from breast cancer at the age of 90. Two of her grandmother’s sisters have suspected cases of breast cancer, while a third passed away from breast cancer. In total, her paternal grandmother had three sisters, and all four developed breast cancer during their lives. In her own generation,

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66 For a visual description of Becky’s paternal history with cancer, see Becky’s pedigree (PP8) in appendix 5.
Becky has no sisters, only one brother. Becky was the only women among her female cousins known to have developed breast cancer. However, Becky did not know for sure because she has lost touch with her cousins from overseas and most in Canada as well, after moving back and forth between both countries at age when family links are usually made.

Many participants held the belief that breast cancer mutations were usually passed down through mothers and daughters. Just as some lay people can be unaware that males can inherit a mutation that causes “female cancers”, many health care providers are unaware as well (Lynch et al., 1999; Yong, Zhou, & Lee, 2003). Participants whose family cancer history comes from their paternal side often used this mistaken notion that only women can pass on BRCA1 and BRCA2 mutations to explain why they did not perceive their family cancer history as fitting the pattern of HBOC. Most participants viewed the risk of inheriting and then developing breast cancer as lower if the family cancer history came from the paternal side than coming from the maternal side of the family. Conversely, in Joyce’s view point, had her breast cancer come through her father’s side of the family instead of her mother’s side, she would have seen her risk of developing cancer as much higher. Joyce seemed to believe that physical characteristics usually attributed to men, such as being physical strong, make their genotypes stronger. This belief could have been reinforced by the experience of her cousin, who was diagnosed with breast cancer at 46, had died of her disease at 52, and whose breast cancer history came from her father’s side. Joyce commented that her cousin’s father had been diagnosed with suspicious cysts in his breast and decided to have a bilateral prophylactic mastectomy.

Thinking that an inherited breast cancer link between father and daughter is weaker than one between mother and daughter could come from the common perception that breast and
ovarian cancers are women' diseases. Bubaloo, who along with her father, had been diagnosed with breast cancer, describes her view of the breast cancer link between father and daughter.

It seems to me that there is more mother/daughter combination happening and it’s a bit of a link that would be great that it could be addressed, you know, in that aspect. And yeah, okay, dad and I, yeah, we met the criteria even though we were kind of the oddball situation. And, okay, that’s fine, you know, because I still firmly believe there’s a genetic link. It’s just that it’s not as common as a mother/daughter situation, but I know inside of me that there is some type of link. (Bubaloo, PP9, field conversation, July 24, 2001)

In effect, Bubaloo’s risk of inheriting a mutation from her father is the same 50% risk as it would be if she had inherited a mutation from her mother. The difference between the two situations is that, when a man from a HBOC family carries the mutation, his risk of developing breast cancer is less. It also depends on whether the mutation is in his BRCA2 gene or his BRCA1 gene. A mutation in a man’s BRCA2 gene confers a lifetime breast cancer risk of 6% (Easton et al., 1997; Struwing et al., 1997), while the association between BRCA1 and male breast cancer is less clear (Basham et al., 2002).

When participants perceived that their family history of breast cancer comes down through both men and women, they constructed a theory of heredity whereby the possible inherited mutation could be transmitted as much through males as through females. Seven participants had such a family history.

Sherry explained why she truly believed something “defective” was playing out in her family. She told me that her brother’s two daughters – both under the age of 40 have developed breast and ovarian cancer. In addition, her female cousin, the daughter of her maternal uncle, developed and died from her breast cancer. These two facts confirmed her that there is something defective about her family’s genes. Sherry had her own explanation as to why, perhaps, their specific family mutation has not been found. She said that she was told that breast and ovarian cancer mutations can only be passed down by females. Yet, within her family, a cancer mutation
seems to have been passed down by a male as well. She interpreted this to mean that she does not think the men in her family are necessarily more at risk of cancer – as none have developed cancer – but that their daughters are more at risk, because the men are passing down the defective gene.

Holding the view that they may have a mutation and that it can be transmitted as much through the males as the females in their families and that both males and females would be at risk of developing breast cancer led participants to believe that perhaps the mutation the Cancer Agency tested for is one that increases the risk of breast cancer for both males and females. This would explain why they say they believe the Agency was unable to locate their specific mutation.

For all of the participants who believed they have an inherited mutation that has not yet been identified, this belief is strengthened not only by the factor described directly above but with some of the other structures discussed, such as the untested portion of their BRCA2 gene. For example, participants stated that perhaps their specific mutation, one that can be transmitted as much through males as through females could lie in the untested portion of their BRCA2 gene, and that is why the Cancer Agency was not able to locate it. For example, Lisa, an unaffected secondary participant, whose family history of cancer was on her fathers’ side, was surprised to hear from her sister who had genetic testing done that not all of their BRCA2 genes had been screened, considering the high likelihood that their suspected mutation most likely lied in the BRCA2 gene. Lisa explained how she had come to learn of the increased risk of having a mutation in one’s BRCA2 gene if one has a family history of male breast cancer. Her grandfather had been diagnosed with breast cancer.
As said at the beginning of this chapter, although I try to present the structures in separate sections, many of them were used simultaneously by the participants in describing their experience of interpreting and making sense of their genetic test results. To illustrate, Becky believed that, if she is carried a mutation, it is not a BRCA1 or BRCA2 mutation, because she sees having a mutation in these two cancer genes as being associated with ovarian cancer as well as with breast cancer, and her family’s history only included breast cancer. Hence, she concluded that her mutation was one that only “attacks” one organ thus, ruling out the possibility that her mutation could lie in her BRCA1 or BRCA2 gene.

**Having a mutation that increases the risk of either breast or ovarian cancer but not both.**

The participants who learned of the possible link between a BRCA1 or a BRCA2 cancer gene mutation and risk of developing not only breast but ovarian cancer theorised that their specific mutation may be one that is linked only either with the breasts or the ovaries, depending on their family history of the disease.\(^{67}\) Of course, this belief only occurred when there was only one type of cancer in the family history. While a recent study points to the possibility that there may be an unidentified ovarian-cancer-susceptibility gene (Menkiszak, Gronwald et al., 2004), we do know that within HBOC families, there is a much higher risk of developing breast cancer than ovarian cancer; thus, there is an increased likelihood of seeing more breast than ovarian cancers. Another recent study indicates that the cumulative lifetime risk (by age 70) of someone with a BRCA1 mutation is 65% for breast cancer and 39% for ovarian cancers. With a BRCA2 mutation, the corresponding risks are 45% and 11% (Antoniou et al., 2003).

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\(^{67}\) Although only one participant had a family history only of ovarian cancer and comparing many family histories of breast cancer with only one family history of ovarian cancer can be seen as methodologically flawed, I still believe that it is more interesting to observe how having a family history of only one type of cancer influences how participants make sense of their test results, than to observe the influence of having both types of cancer in the family. Hence, I combine these two types of family history.
Through their genetic counselling sessions, many participants of the current study learned of the link between a family history of breast and ovarian cancer and an increased likelihood of carrying an inherited mutation in either the BRCA1 or BRCA2 gene. With this new understanding, some participants formulated new heredity theories to explain their genetic test results. For example, Becky explained that, while she understood having a family history of breast and ovarian cancer was associated with an increased risk of carrying an inherited mutation, because her family history did not contain any ovarian cancer and the breast cancer cases are all late onset, she was more inclined to interpret her results as not having an inherited mutation in the BRCA1 or BRCA2 gene but perhaps in some other yet-unidentified cancer gene that only causes late-stage breast cancer.

The absence of ovarian cancer in their family informed some participants’ interpretations of their test results. They used this structure to eliminate the first two possible interpretations in the letter received from the Cancer Agency. They focussed on the third option; that is, the mutation responsible for their cancer may be in another, as-yet-unidentified, hereditary cancer gene. They concluded that their mutation was not in their BRCA1 or 2 cancer gene, because they lacked a family history of ovarian cancer. Individuals whose family history was of ovarian cancer alone believed the opposite: They saw themselves having a mutation that specifically develops into ovarian cancer. Participants who believed that they have a mutation that affects only one organ (breasts or ovaries) were more inclined to interpret their results as ambiguity regarding mutation carrier status. Victoria talked about being certain she has an inherited mutation but not in her BRCA1 or 2 genes because she has no ovarian cancer in her family:

Doctor A (the oncologist there) she said that there is some mutation playing out in my family. We obviously have some mutation they have not identified. I see it that way too. I’m sure that there is a mutation. Um, because how would you explain all the breast cancer in my family? But I think it’s a mutation that, I don’t know if it’s BRCA 1 or
BRCA 2 that has a high incidence of ovarian cancer but we don’t seem to have any ovarian cancer in our family. So, I think there is a mutation there. But it’s just one they haven’t identified. Maybe it is a BRCA 3 or whatever. (Victoria, PP22\textsuperscript{68}, field conversation, September 26, 2001. Victoria was diagnosed with breast cancer at the age of thirty-seven.)

Only Becky, whose family history only included breast cancers could comfortably say that she was somewhat more inclined to believe that her breast cancer might be a “fluke,” accepted concurrently the third and fourth interpretations options in the letter from the Cancer Agency. However, she arrived at this conclusion not as a result of the last structure alone (have a mutation that affects only one organ) but by combining a number of structures: the breast cancer cases in her family history were all late onset, there were no cases of ovarian cancer, and her family’s cancer was transmitted through her father’s side. These three structures led her to believe that, if her family did have an inherited mutation, it was most likely of lower penetrance.\textsuperscript{69} Hence, with these beliefs, she explained that she saw her own breast cancer as fluke and the other breast cancers in her family as resulting from a weaker inherited mutation, one that targets specifically the breast and that makes cancer appear later in life.

Louise, diagnosed at the age of 23 with ovarian cancer, described how she understood that her inherited mutation specifically targets the ovaries:

The other day I heard a statistic about one in three people who have had cancer, it reoccurs – one in three. So I’m going, oh, you know, 20 years later it could come back, right? So I’m a little afraid, but, on the other hand, I’m thinking, I don’t care, you know. I’m just not worried about breast cancer, because I just feel that, you know, the cancer chose a place. It was either going to be the breast or the ovary and it went to the ovary so. I don’t know with the testing; it kind of indicated that, you know, you have the ovarian

\textsuperscript{68} Although I have 21 participants, Victoria’s code number is PP22, because one participant (coded PP19) was never interviewed. She passed away while we repeatedly tried to set a date for the interview. During those 6 months, I added 3 new participants to my database, PP20 to PP22. I had already begun data analysis while waiting to conduct PP19’s interview, so I did not want to change the code numbers. As well, I kept PP19 in my database out of respect for her ongoing interest in participating in my study. She kept in contact with me for many months, always trying to see if by next week she would feel strong enough to do the interview.

\textsuperscript{69} BRCA1 and BRCA2 gene mutations are highly, but not fully penetrant – not everyone who carries a mutation will develop cancer (Ponder, 1997). In comparison, the genetic mutation for Huntington Disease is fully penetrant – everyone who inherits this mutation will develop the disease.
cancer, it is not likely I’m going to get the breast cancer. (Louise, PP13, field conversation, July 18, 2001)

Louise’s understanding is that, had she been found to have an inherited mutation in either her BRCA1 or 2 genes, she would have developed breast cancer as well as ovarian by now.

However, when she said that she is not afraid of a recurrence, this is because she had a total oophorectomy (removal of both ovaries), and there was no chance that cancer could recur in her ovaries. She believed that, since her mutation chose the ovaries instead of the breasts, she need not fear a recurrence. Louise felt that her genetic test confirmed that her ovarian cancer was hereditary. Because she met the eligibility criteria for genetic testing, she believed that the clinic suspects she carries a mutation that predisposes her to ovarian cancer. She mentioned that the letter she received from the Cancer Agency said that there was a possibility that she might carry some yet-unidentified cancer gene mutation (the third possible interpretation of four).

Having a mutation that is specific to their ancestral lineage.

The following statement by Joyce describes how she used her ancestral lineage to formulate a theory of genetic risk inheritance.

When I read about this more, I found that the two genes that they were looking for was actually found in 11 or 12 families of Hebrew, Jewish extraction. I mean that’s how they found the genes. So who’s to say that the gene they found are not strictly Jewish extraction and that they may not be Anglo-Saxon, they may not be Afro-American, you know? (Joyce, PP3, field conversation, December 19, 2000)

This interpretation echoes a few participants’ belief that the mutations localised and identified in the BRCA1 and BRCA2 cancer genes are predominantly, if not only, found within the Jewish population. Holding this belief, some participants suspected that the Cancer Agency missed their specific mutation by only screening for the mutations found in the Jewish population, instead of looking for mutations known to be associated with other ancestries. Although this belief does not

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70 In Louise’s family, there is a history of ovarian cancer: three known cases in two generations. See Louise’s pedigree (PP13) in appendix 5.
totally contradict empirical findings, whereby three mutations – 185delAG and 5382insC in BRCA1 and 6174delT in BRCA2 – are associated with an Ashkenazi Jewish ethnicity and have been shown to account for a high rate of, and early onset of, breast and ovarian cancer in this group (Friedman et al., 1999; Olopade, 1997; Struwing et al., 1997). Founder mutations have also been associated with populations in Quebec, Poland, Scotland, Iceland, Singapore, and Finland (Lee et al., 2003; Lubinski et al., 2004; Mikaelsdottir, Valgeirsottir, Eyfjord, & Rafnar, 2004).  

The view that genetic testing missed their specific ancestral lineage mutation was further reinforced when there were high numbers of breast and/or ovarian cancer cases in the family. Participants with this view interpreted their results as confirmation of their mutation carrier status and ambiguity regarding their mutation carrier status. As Joyce expressed, “Who’s to say that the Agency’s screening was only done for the Jewish decent mutations and that these mutations may not be the same as those from other ancestral lineage.”  

**Being weaker – needing an internal or external trigger to activate cancer.**  

When participants said that their specific mutation requires “trigger” in order to produce cancer, they were attempting to explain their continued belief that they have an inherited mutation despite being told that no mutation had been found. They held the view that their specific mutation required a trigger to becoming active – to produce cancer. They also held the belief that the mutation must be from a weak strand or one of a lower penetrance.  

Some participants inferred that a weak mutation produces a type of breast cancer with a better prognosis, based on the fact that all or most of their family members had lived more than 5 years following their breast and/or ovarian cancer diagnoses. These participants reasoned that, a

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71 Founder mutations refer to mutations that occur among people with the same ancestral lineage. They occur very frequently within certain populations because of the group’s low genetic heterogeneity and homogenous environmental exposure (D. L. Newman et al., 2004).
weak mutation can stay inactive until triggered by an internal factor, such as stress. Here is how Victoria saw her situation:

    Like I always think, if I have these certain foods and try to keep my stress level down and exercise, you know, go for walks and those kinds of stuff that it won’t be triggered, to go to cancer. I think that genetic testing is the underlying sort of base but, you know, if you don’t have that genetic mutation, then there is nothing to trigger. So, you know, but I have that underlying mutation – that if I get all run down and everything, I can trigger the mutation to cancer. (Victoria, PP22, field conversation, September 26, 2001)

Participants who believed that their mutation is not only weaker but that it needs to be triggered by internal factors such as stress or by external factors such as environmental pollution interpreted their results as ambiguity regarding mutation carrier status. Victoria referred to an internal factor that she said is associated with BRCA1 and BRCA2 mutations.

    They said that most mutations in the BRCA1 and BRCA 2 turned up in people that were estrogen negative, which I was. So, I think that, um, that ... I don’t know I was just thinking that, if anyone was going to turn up with the mutation it would be me, because my cancer was estrogen negative, while my cousin’s cancer was estrogen positive. (Victoria, PP22, field conversation, September 2001)

In essence, Victoria linked having a breast cancer that is estrogen negative with risk of carrying an inherited mutation for BRCA1 and BRCA 2 cancer genes. After no mutation was found in either of these genes even though her breast cancer was identified as estrogen negative, Victoria concluded than that she carried a mutation not in the BRCA1 or BRCA2 genes (as she was told that none were found) but that she must carry a mutation that needs a trigger by factors such as stress or by not proactively decreasing one’s risk of breast cancer.\footnote{Proactively reducing one’s risk was one of the categories discussed in the section Implications of Living with a Family History of Breast and/or Ovarian Cancer.} Victoria also specified that she no longer felt at risk of ovarian cancer, because she does not have a BRCA1 or 2 mutations. While she was uncertain whether the unidentified weak mutation she believes she has is also linked with ovarian cancer, she said that she suspected it is not, because her family history contains no ovarian cancer cases.
Participants who believed that they may have a weak mutation predisposing them to breast and/or ovarian cancer were not only those who expected their test results to show a mutation. Participants who had less cases of cancer in their family but were diagnosed with breast cancer at a young age or had more than one primary breast cancers looked in their immediate and past internal and external environment for explanations. Concluding that their breast cancer could not result from chance alone, they considered the possibility of carrying a weak mutation that needed an environmental trigger to activate the cancer. Participants gave examples such as living in the city compared to the suburbs, having sedentary lives and eating too few fruits and vegetables, and drinking too much coffee. The common factor among these participants was the lack of a strong family history of breast and/or ovarian cancer. This demonstrates how the generic structures discussed in this chapter interacted to influence the retrospective narrative of causal reasoning of self with a probable inherited mutation presented in this study.

The above participants’ beliefs are not unsubstantiated. While carrying an inherited mutation in one’s BRCA1 or BRCA2 genes increases one’s risk of cancer, it does not automatically result in cancer. Researchers caution that all types of cancer – sporadic or inherited – remain multistep diseases with multifactorial etiologies (Wild & Kleihues, 1996). These authors conclude that individuals’ responses to environmental agents may depend, to a significant extent, on their genetic makeup. While the role of stress in producing cancer is still unclear (Baltrusch, Stangel, & Titze, 1991; Ben-Eliyahu, Page, Yirmiya, & Shakhar, 1999), other studies evinced a link between diet and increased risk of breast and ovarian cancer (Holmes & Willett, 2004; Webb, Byrne et al., 2004; Webb, Purdie, Bain, & Green, 2004).
Some participants relayed that between receiving a telephone call informing them that no mutation had been found and reading the four possible interpretations in the letter from the Cancer Agency, they had not fully realised that there might be other gene mutations responsible for breast and/or ovarian cancer. Coming to this realisation, as well as talking to their family physicians, led some participants question whether they carry an as-yet-unidentified hereditary cancer mutation. Most concluded that it is likely a weaker strand, depending of which other generic structures they used to formulate their lay theory. For example, those in whose families' breast cancers suddenly started appearing close together and saw a possibility that their cancer could be the result of internal and/or external factors postulated that it could have also resulted from a weaker inherited mutation that was triggered by one of these factors.

**Influence of time**

Time is another structure found to inform participants’ interpretation of their test results and how they made sense of them. There are two ways in which time influenced participants’ interpretations: waiting time between testing and obtaining results and time lapse between cancer diagnoses. These two ways represent structures ten and eleven out of the thirteen found to inform the participants’ interpretation. I first address the structure of time between testing and obtaining results.

**Influence of time between testing and receiving results.**

To begin, the participants had to wait almost 2 years before obtaining all of their results. At the end of the first year, they learned through a telephone call from the Cancer Agency that no mutation had been found in their BRCA1 gene and that now, the Agency was going to test their BRCA2 gene. It took almost another year to obtain the result of the second test. Some participants interpreted this long wait to mean that their results must be negative and/or that the
Agency lacked funding to conduct tests in a timely manner, and/or that the testing technology was unable to detect mutations rapidly.

Victoria, whose blood sample was taken in 1999 and who received her results in 2001, explained how she interpreted this waiting time:

I was going every 6 months to see my oncologist. She actually said to me, ‘because the results take so long to come back, I suspect that they didn’t find any mutation.’ So, I thought, well I didn’t know this. I didn’t realise that taking a long time would mean that so, then I thought, sort of thought, oh, okay. And, I guess, sort of thought in my mind, well, there is no genetic mutation. So I mean that, no BRCA1 and BRCA2 mutation has been identified. And then I, I guess I sort of thought, after she said that well, you know, I sort of made peace with it. So, when I got the actual word, that’s what I was expecting. So when the genetic counsellor phoned, I was not surprised. (Victoria, PP22, field conversation, August 27, 2001)

Victoria’s oncologist interpreted the long wait to mean that she was probably not carrying a mutation. This view certainly reflects common medical practices in which the person about to be tested is told beforehand that they will only be contacted if an abnormality is found. The comments of Victoria’s oncologist influenced her interpretation of her test results; she viewed her results the same way he did: negative for BRCA1 and BRCA2 mutations. Although she viewed her results as negative, as explained in the previous section, she also believed that she has a weaker inherited mutation that was triggered by stress.

A study by Bish et al. (2002a) reports that while affected women from families at risk of HBOC experienced a decrease in perceived likelihood of carrying a mutation 6 months after genetic counselling with inconclusive genetic test results, at 12 months postcounselling, they believed again they had a mutation. The authors hypothesise that, during the time that had passed since they received their inconclusive results, their anxiety returned to pretest levels.

Hallowell et al. (2004) found that women’s perception of risk did not seem to be affected by the length of time that passed after receiving test results. Many participants in my study who
discussed the length of time between having the test done and receiving their results mentioned how, at times, they had even forgotten about the test and figured that the results must be negative (otherwise they would have heard from the clinic). Of course, this was mostly the case when they did not have a family member regularly calling them to see if they had obtained their results. Barney mentioned how frustrated she became with this one cousin who called her up every couple of months. She told her cousin to call the Cancer Agency directly to find out when she would receive the results. Bound by confidentiality rights, however, health professionals cannot disclose to a third party that a family member has received medical assistance.

Time between individuals with cancer across generations and within the same generation.

The amount of time between participants' cancer diagnoses and those of others in their families also influenced how they made sense of test results. I observed that participants commented on their overall family history of both breast and ovarian cancer within past and present generations. Those who observed a similarity in the age at which cancer appeared within each generation were more inclined to believe that their family's cancers were the result of an inherited mutation. Following is a description of two participants' views on the subject of time.

Donna and her only sister, who both had genetic testing, were certain that their results would show they carried a gene mutation. They were both diagnosed with breast cancer at 49, and their mother was also diagnosed with breast cancer around the same age. To Donna, this indicated that a mutation was playing out in their family. In contrast, Gilligan and her mother, both diagnosed with breast cancer within two years of each other's cancer diagnosis, thought there was a pretty good chance that no mutation would be found.

73 Although family dynamics and communicating genetic testing results within families were not the focus of my study, some discussion around these topics did take place during the interviews. I plan in the near future to conduct a descriptive interpretation of these discussions.

74 See Donna's pedigree (PP14) in appendix 5 for the complete family history she shared during the interview.
Time lapse between diagnoses across generations seemed more influential on participants’ belief in having or not having an inherited mutation than time lapse between diagnoses of members of the same generation. One of the differences between Donna and Gilligan (and among other participants) was the time lapse between breasts and/or ovarian cancer diagnoses of other family members and their own. This difference in time lapse produced differing interpretations. That is, the longer the time between, for example, a mother’s diagnosis or an aunt’s and theirs, the more they were inclined to believe that the cancers in their family were inherited – especially if cancer appeared at a similar age in each generation. Participants explained that they saw this as proof that the past was repeating itself in coming generations.

Conversely, when the time lapse between cancer diagnoses was short, for example, their mother’s diagnosis and their own, they wondered if an explanation could be found from similar environmental exposure in their immediate environment such as exposure to a carcinogen, or they looked for a shared personal risk factor, such as being overweight. However, participants only held this view if their family history with cancer was not strong. Although the literature indicates that individuals from families at risk of HBOC tend to think their genetic risk is high (Lerman, Kash, & Stefanek, 1994), my findings indicate that there are other possible influences such as the family history’s distribution of cancer that can influence individuals’ perception of genetic risk. The influence of time between individuals with cancer across generations and within the same generations needs further inquiry as the actual influence of time in this study cannot be calculated independent of other influences. The study design does not permit such assertion.

**Resemblances among individuals with cancer**

Similarities and differences among individuals with cancer.
In order to make sense of their genetic testing results, participants at times commented on how they shared similar physical or psychological characteristics with others in the family who had cancer and how they saw these similarities as some sort of proof that, if they could inherit these features then they could inherit a cancer gene mutation. Bubaloo’s account reveals this proposed link between inherited features and the possibility of inheriting “our parents’ diseases.”

In my mind I knew that there was some form of, um, genetics kicking up. I did, I really did. And the thing is, when I look at my parents, like my mother and her side of the family, they have thyroid conditions. Well, of course, I have an inherited thyroid condition. From my father’s side, you know with my breast cancer at 40, well, ok, I have inherited my father’s cancer. But you know it’s not just that it’s the physical traits, it’s the mental traits. It’s everything like that. And I could always see it, you know, and I am a brand of both but it seems like I carry a strand of each, you know. I just knew in my heart that there was something. Both my dad and I have been diagnosed with breast cancer in the same breast. I mean it is too close to say this is just a coincidence, even though when they first said, ‘well we tested for the BRCA1 and it’s a negative’ and they said ‘well we are going to try for the second gene you know’ and that was like a year and a bit later. Well, that was anticlimactic. It was well after the fact, and they said, well it was negative you know. But I am going, well, we’ve just touched the tip of it. There is just so much there to know and it could be the third gene. It could be more, whatever. Like, I knew in my heart that there is, there is something out. It’s just that we haven’t got the sophistication or the knowledge to find it yet, and we may never have in my lifetime. (Bubaloo, PP9, field conversation, July 2001)

Similar features among those who develop a familial disease can create an increase perception of risk among family members who resemble each other. Increase perception of risk for an inherited disease was expressed by many participants who resembled affected family members. They used this resemblance to explain why they felt that, although no mutation was found by their test, they still feared that either they, their children, or their siblings may have an inherited mutation.

Well you know, you get blue eyes and brown eyes but, when they get the mutation, they may have the Barnes genes and not have to worry about my genes at all. That’s the way I feel; I don’t know if that’s the way it works or not. I hope so, because I see my husband’s genes as stronger than mine. And my children, my daughters have inherited more of my husband’s genes. They have: they’re larger people and I’m tall but they’re....my son is small like me. Well, I’m not small but he’s more my stature than the girls are. Sue is 5
foot 10 or 11 and Bobbie is 5 foot 8 or 9. And they are both stocky. My sons are smaller. (PP18, Marcy, field conversation, September 13, 2001)

Marcy’s lay theory (also shared by other participants) of who in the family could be at risk for an inherited mutation could be reinforced if one of those family members would actually be diagnosed with breast and/or ovarian cancer. If they did not develop cancer before passing what the participants term a “critical age” at which most of the cancers in their family occurred, the participants would conclude that this particular individual had not inherited the mutation, because “they take more from the other side of the family.” Sherry’s account exemplifies this lay theory.

My sister’s body style is more like my father’s side than our mom, which might explain her lesser risk of having inherited that family mutation. Carl, our brother, who has two daughters both of whom have developed breast and ovarian cancer, he looks more like me. (Sherry, PP21, field conversation, September 18, 2001)

Sherry, who interpreted her results as ambiguity regarding her mutation carrier status theorized why one of her sisters had not developed breast cancer, while both she and another sister did. She believed her unaffected sister inherited more genes from her father’s side, where there is no history of breast cancer.

To date, both Sherry’s lay theory of inheritance (the risk of the women in her family to develop breast and/or ovarian cancer is two women out of four within each generation) and her belief that she carries an inherited mutation and has developed cancer are consistent with the pattern of diagnosis. In this sense, Sherry understands the scientific notion that one has a 50% risk of inheriting a disease. Her lay theory has not been undermined by the “facts”. However, when a new cancer diagnosis within the family disproves the lay theory of inheritance, participants review their lay theory and may modify their conclusions about whether or not their family does or does not carry an inherited mutation of cancer.
My study findings concur with previous research, which suggests that individuals' lay theories of inheritance often incorporate resemblance in physical characteristics and personality traits (d'Agincourt-Canning, 2003; Richards, 1996). Studies conducted with people who have Huntington's disease show that individuals refer to family resemblances to gauge their risk of developing the disorder (Cox, 2003; Richards, 1996).

Participants also interpreted a similar interval between cancers' appearances with each generation as indicating that an inherited mutation was responsible for this similarity. That is, Becky described how, with each generation, breast and/or ovarian cancer was appearing 10 years earlier. She explained that four women in her family two generations before hers developed breast cancer in their 80s, three women in the generation before hers developed breast cancer in their 60s; the women in her own generation were getting breast cancer in their 50s (Becky was diagnosed with breast cancer at 52); and now her son had just been diagnosed with leukemia at 37. Thus, the participant saw a pattern of cancer diagnosis based on age and had difficulty letting go of the possibility that her family carries an inherited cancer gene mutation. Although Becky said that, for the time being, she interpreted her genetic test result as indicating no inherited mutation to at least her BRCA1 and BRCA2 genes, she is still unsure of the risk to herself and other family members for cancer.

Many of the participants used lay theories, such as those described above, as a way of understanding why, despite certain family members being more at risk of developing breast and/or ovarian cancer than them (in the participants' view), they did not develop the family disease. As reported by d'Agincourt-Canning (2003) in her study with HBOC families, when faced with a number of disturbing events, individuals insist on looking for patterns in their attempts to understand, to cope with and to control what is happening.

75 See Becky's pedigree (PP8) in appendix.
Presence of children

Having children or not, whether children are young, and children’s gender.

Previous work suggests that individuals have difficulty assessing inheritance risk of a disease usually associated with one gender (Richards, 1996). The perceived risk of passing on a genetic mutation for breast cancer among participants who only have sons was obviously discussed less often by the participants in my study. When they did acknowledge that they could transmit a breast cancer gene mutation, participants mostly did so in the context of their current or future daughters’ risk.

Participants with young children and adult-age children said that at least knowing that no mutations were found in either their BRCA1 or BRCA genes provided them with some relief. They now felt secure that they had not passed a breast and ovarian cancer mutation to their children – at least not a mutation in the two known cancer genes. They felt that they could at least “back off a bit” in pressuring their daughters to have early screening for breast cancer.

While age, gender and number of children were mentioned by few participants, those who did spoke of these factors with much emotion. Participants with children under 20 expressed how they preferred to think of their children as not likely to have an inherited mutation. When participants believed that their children did have an inherited mutation, they usually thought it was a mutation that required a trigger – hence being a weaker gene mutation. Participants would search for reasons why they felt that their children had a better chance than they did of never developing cancer. They explained that they perceived their children’s living conditions and lifestyle to be healthier and therefore their risk of developing cancer to be lower.

Among the four participants who, following their test results concluded that they do not have an inherited mutation and were “happy” to say that they had not passed an inherited
mutation to their children, two had both a boy and a girl below the age of 10 while a third has two sons over 18. These mothers believed that maintaining their children's health was part of their responsibility to nurture their children physically, emotionally and intellectually. d'Agincourt-Canning (2003) study that looked at individuals moral experience with genetic testing for BRCA1 and BRCA2 also arrived at similar results.

**CONCLUSIONS:**

**Types of Interpretation, Integration of Generic Structures, and the Retrospective Narrative of Causal Reasoning**

Although empirical studies have identified factors influencing perception of genetic risk, none have described and interpreted these factors utilising experience near-concepts such as the 13 generic structures described in this chapter. The generic structures discussed in this chapter have brought to the forefront what underlies these factors. The study's findings, further, show the importance of recognising the interplay among structures in informing individuals' interpretation of their genetic test results and their making-sense experience. While participants link some of the generic structures presented in this chapter, these connections are never static because they evolve with changes in participants' personal and family histories of breast and/or ovarian cancer.

Participants attempted to make sense of their experience in light of their past, current and future lived experience. Within the presented retrospective narrative of causal reasoning about a probable inherited mutation, participants may move from one stage to another within the proposed seven-stage process, and some stages may be more prominent in certain times in their lives. However, not all participants experience the process in similar ways. For example, I observed that, when participants and their relatives approach what is perceived as the age at
which most breast cancer diagnoses occur in their family, participants may have used this observation to explain why they believed that her family was more at risk of cancer at that time. This situation is an example of the fourth stage in the retrospective narrative of causal reasoning of self with a probable inherited mutation proposed in this study. For other participants who interpreted their results as refutation of their mutation carrier status, their retrospective narrative of causal reasoning to a probable inherited mutation tended to circle around stage one to two where they recognise that some of the cancer cases in their family history may be unusual such as early onset diagnosis. These participants did not attain all stages where the last stage represents the conviction that one has an inherited mutation increasing their risk for cancer and that of others in their family.

New diagnosis or recurrence in the family also influenced the process of perceiving self with a probable inherited mutation. Participants reassessed their breast cancer experience and arrived at new conclusions about their genetic risk status (or confirmed the existing conclusion). Similarities presumed to be genetic that are noted among those with the same cancer diagnosis seem to influence one’s view about the family’s proneness to an inherited mutation.

The belief by many of the participants that their family history with cancer shared characteristics associated with hereditary breast and ovarian cancer seemed to underlie the transition through the retrospective narrative of causal reasoning of self with a probable inherited mutation. Participants who did not think that they had a strong family history of breast and/or ovarian cancer did not seem to enter into this process. Rather, they underwent genetic testing to gain more insight into their disease, not to find an inherited mutation. In contrast, participants who felt that they had a strong family history of the disease underwent genetic testing convinced that they had an inherited mutation and expecting the results to confirm their conviction. These
participants often, in the end, interpreted their results as confirmation of their mutation carrier status.

The participants whose interpretations fall within viewing results as ambiguity regarding their mutation carrier status (maybe we have or maybe we do not have an inherited mutation, either in our BRCA1 or BRCA2 genes or in some other yet-unidentified cancer gene) entered into genetic testing uncertain of what the results would show, although slightly suspicious that they might have an inherited mutation in viewing of their unusual family history. When they reviewed the four ways in which their results could be interpreted (in the letter from the Cancer Agency), most of these participants felt that it was difficult, if not impossible, to conclude whether they did or did not have an inherited mutation, based on the fact that the Agency wrote in their letter that there is still a possibility that they may carry a mutation in light of their personal and family history.

Inasmuch as the possible interpretations offered within the letter influenced the participants’ interpretations, they were not the most influential factors; their lived experiences with cancer and those of others around them were. Each new case of cancer transformed their perception of genetic risk. Each new case concurrently transformed their view of their family’s history of cancer as strong or not. Thus, biographical context became the force moving participants through the process of seeing themselves and others in their family at risk of inherited susceptibility to cancer.

This chapter illustrated that women from families at risk of HBOC interpret uninformative genetic test results in three ways: Confirmation of mutation carrier status, ambiguity regarding mutation carrier status and, refutation of mutation carrier status. These three types of interpretation are connected to the generic structures that informed how they made sense
of their test results to derive their genetic risk. Most of the generic structures speak to the
enormous role that personal and family experience with cancer plays in the construction of
perceived genetic risk for oneself and others. The exceptions addressed doubts and uncertainties
about the outcome of their genetic test results and the influence of time between testing and
obtaining results.

Having looked at how the participants’ interpretations and making-sense experiences has
been shaped by their family’s history, their own lived experience with cancer and genetic testing,
I now turn to participants’ concluding reactions to their experience of genetic testing.
CHAPTER SIX:

What Participants Concluded from their Genetic Testing Experience

In the preceding chapter, I described and interpreted the primary participants’ experience of genetic testing. I now turn to participants’ concluding reactions to their experience of genetic testing. I will look at such issues as, what did participants gain from their genetic testing experience?” As discussed in previous chapters, there are considerable buildups (such as living with a family history of breast and/or ovarian cancer) that led participants to accept or become interested in genetic testing.

Some participants hoped genetic testing would provide them with information to better their own lives and those of their family. As when I asked participants what they gained or lost with genetic testing, they answered my question using concepts of relief, benefits, limitations, and harms. I therefore begin by describing these concepts, and then follow with a discussion of specific views held by study participants on their test results.

My goal is to demonstrate how the experiences of women who receive genetic test results with multiple possible interpretations differ from those of individuals who either receive a positive or a true-negative test result.76 My overall intent in this chapter is to show how the participants’ genetic testing experience impacted their overall lives.

Emotional Reaction to Genetic Testing Results

Contrary to other study findings (Bish et al., 2002b; Friedman et al., 1999), very few of the study participants said they found relief through their test results. Researchers have hypothesised that individuals found not to carry an inherited mutation would feel less anxious as a result of knowing that they are not genetically predisposed to cancer and will therefore not

76 By true-negative I mean results that confirm that the person tested does not have a BRCA1 or BRCA2 mutation already identified within a blood relative.
transmit a genetic mutation to their children (Bish et al., 2002a; Cappelli et al., 1999; Claes et al., 2004; Lynch et al., 1997). A Canadian based prospective study by Wiggins (1992) that followed three groups (increase-risk group, decreased-risk group, and the group with no change in risk) at risk for psychological consequences as a result of genetic testing for Huntington’s disease found that individuals who received results that indicated either an increase or a decrease in their risk of inheriting the gene for the disease did provide potential benefits for their psychological health. Hence, those whose tests confirmed risk are expected to feel relief from reduced uncertainty. What was problematic for the participants of this study is that their results were uninformative – the results did not conclude with certainty if they did or did not carry an inherited mutation. They are not true negative, in the sense that the recipient knows with certainty that they do not carry an inherited mutation. This lack of certainty may explain why very few participants found the relief they expected to accompany a negative result.

The question thus remains, were the participants better off having this test done? I addressed this question by analysing the participants’ answer to the question: do you find any sort of relief for yourself and others around you regarding the risk of having an inherited mutation and an increase risk for breast and/or ovarian cancer? Hence, in this section I first describe how relief or lack of it was experienced when they received their results. While relief is just one type of subjective benefit that can be assessed to the outcome of experiencing genetic testing, other measures such as objective assessment have also been instrumental to the psychological understanding of experiencing genetic testing. Some of these studies are discussed in comparisons to the current study findings.
Those who found relief

The four participants who interpreted their results as refutation of their mutation carrier status experienced relief. The types of relief they described are similar to others documented in the literature for individuals found not to carry a mutation: emotional relief, relief for children, relief in informing siblings and partners that they do not carry an inherited mutation, and a general feeling of happiness (Claes et al., 2004; d'Agincourt-Canning, 2003; Dorval et al., 2000; Lynch et al., 1997).

Only Juniper, Erika, and Gladys were completely convinced that they did not have an inherited mutation, while Becky wondered whether her family might carry an inherited mutation. As for Erika, she described her relief as only partial. That is, she was relieved to know that she did not carry an inherited mutation and therefore has not transmitted one to her daughter, but she had hoped to learn why she had developed breast cancer in the first place. As Erika so clearly expressed it, “I wanted to know why I had breast cancer, but at the same time I didn’t want to hear that I had the gene.” She considered herself to have a healthy lifestyle and therefore could not comprehend how she could have developed breast cancer at 35. Like many other studies report, one of the main reasons for people to have genetic testing is to find out about the etiology of their cancer (Hallowell et al., 2002; Lerman, Seay et al., 1995) as well as to learn about their children’s genetic risk (Armstrong et al., 2000; Lynch et al., 1999).

For many participants who viewed their results as ambiguity regarding their mutation carrier status, there was some temporary feeling of relief expressed as, “At least for the time being I can let go a bit.” They could let go of the idea that they and their family members may carry a genetic risk until proven otherwise. As some participants said, it was easier to have some control over risk factors in your environment than it was to control a genetic risk. As for the
participants who interpreted their results as refutation of their mutation carrier status, while they expressed experiencing relief to their perceived genetic risk for cancer, they still continued to experience uncertainty about the etiology of their cancer and lack of control because of this. These findings echo those found by Claes et al. (2004): women from families at high risk of HBOC where no BRCA1 or BRCA2 mutation were found expressed relief but were still aware of their increased risk for cancer.

As for the participants who are convinced that they had a mutation predisposing them and their family to cancer (the confirmation of mutation carrier status group), they felt a lack of control over their disease. This lack of control over their disease was often expressed within many of the structures presented in the section, Implications of Living with a Family History of Breast and/or Ovarian Cancer. That is, participants reported recurrent thoughts that they may develop cancer again. However, those who interpreted their results as ambiguity regarding mutation carrier status but seeing a possibility of having a weaker mutation needing an internal or external trigger to activate cancer experienced some relief in believing that they had some control over their disease by adopting healthy lifestyle behaviours. They further experienced some relief in thinking that their cancers were not their fault – a similar finding as found within Hallowell et al. (2004) study. Consequently, participants with younger children, especially daughters, expressed relief in knowing that most likely they have not passed an inherited mutation to their daughters, so they did not need to feel responsible for something that may happen to their daughters and over which they had no control. Those who expressed relief on reading their test results preferred to think that internal or external factors may have caused their cancer, such as poor eating habits, lack of exercise, or stress, since controlling the outcome of an inherited mutation was harder in their view than controlling one’s environment.
Just like Erika and Juniper, Becky (who interpreted her results as refutation of mutation carrier status) said that she found relief through her results and, with this relief, came a greater sense of control over her familial disease. This is how Becky expressed her relief:

I still think that my family is at risk to some other inherited mutations to breast cancer, but I definitely think the saving grace is that I find so horrifying, is that with the screening, it does indicate that we are not at risk to ovarian cancer as we don’t have a mutation in either BRCA1 or 2. So I felt relief that we came up negative for either genes, and I guess the reason I feel relief for that, you know. And I have well understood all the caveats that they gave me: this does not mean it’s not there, it does not mean that there is not some other gene at work, blah, blah. But just looking at the overall, the results of the testing combined with my history... I just have an instinctive belief that we don’t have it. (Becky, PP8, field conversation, April 17, 2002)

Becky later added that she was relieved to know that she does not have the mutation and that she and her family had been spared “the horrifying aspects with BRCA mutations”\textsuperscript{77}. In essence, Becky’s relief is linked to having more control over her familial disease. Even though she recognises that she may have some other mutation increasing her risk of breast cancer, she believes it would be less aggressive because her family history of breast cancer is of late-onset and would not increase her risk of ovarian cancer, which she greatly fears. A family member whose sister had genetic testing also spoke about this renewed sense of control over her future risk of breast cancer:

I think it made me feel more in control of what might happen. I think it made me feel less at risk, less vulnerable because it wasn’t BRCA1 or BRCA2. (Janet, SP1/PP5, Evelyn’s sister, field conversation, April 5, 2001)

Many of the participants mentioned how relieved they were to find out they did not have a mutation in their BRCA1 or BRCA2 genes because of the high risk of developing breast and ovarian cancer associated with a mutation in either of the BRCA1 or 2 genes. The relief was often unaffected by their knowledge that they may have another yet-unidentified cancer gene

\textsuperscript{77}You may recall that Becky believes that she most likely does not have a BRCA1 or BRCA2 cancer gene mutation because, in her view, families that carry such mutations would also have a history of ovarian cancer and early breast cancer – which her family history does not.
mutation. Such findings are also noted in Claes et al. (2004) study where the 36 individuals who received inconclusive genetic test results reported a renewed sense of control over ovarian cancer. Victoria, who had become aware of the link between having a mutation in either BRCA1 or BRCA2 genes and an increased risk for ovarian cancer as well as to breast cancer, described her new-found relief:

When the genetic counsellor said, “You don’t have the genetic mutation,” I said that’s great. I thought, I don’t have to worry about ovarian cancer now. That was my big worry. (Victoria, PP22, field conversation, September 26, 2001)

This is certainly the same belief that Becky held when she stated that she was relieved to see that her family has been spared the “horrifying aspects with BRCA mutations”. However, for others, this newly understood link between breast and ovarian cancer following their genetic counselling session stopped them from feeling complete relief, even if they interpreted their results as ambiguity regarding their mutation carrier status, because they believed that they still might have a mutation in their BRCA genes. Ginger, who interpreted her results this way, said that she now feared developing ovarian cancer. While Ginger had learned to live with her risk of breast cancer, she, like many other participants, was shocked to hear of their risk for ovarian cancer.

My genetic testing did not show that I have this hereditary type of breast cancer that also might include a risk for ovarian, but I still believe in some ways that there was a genetic component – at least a very clear predisposition. Before I had this genetic testing, I had no fear of having ovarian cancer but once I was part of this test, I thought, Holy Shit, I am going to die like Gilda Radner. But I felt that it was better to know than not know. And then, when the final results were in, they said they could not identify a specific mutation in either BRCA genes. Well, I guess that doesn’t say that I am totally free of it; just that they couldn’t find anything. So this is a new fear I have to worry about. (Ginger, PP11, field conversation, July 5th, 2001)
Ginger’s account shows how her experience with genetic testing and counselling influenced her perceived risk for ovarian cancer. The next account shows how Victoria viewed her risk of ovarian cancer to be lower after receiving her test results.

Knowing that at least nothing was found in those two genes was a relief to me. It did give me relief, because I have a real thing about ovarian cancer. You see, from the hospital where I do staffing, one of our favourite nurses, healthy as anything, nicest person you could meet, got ovarian cancer. She wasn’t even 35 years old. And, um, she died. It was horrible. She didn’t know anything was wrong with her. She just had a horrible pain all of a sudden and next thing we knew she was on the gynecology ward. She was dying. We were all, like, just so horrified that it would happen to her with no warning. I don’t like ovarian cancer, because it doesn’t give you any warnings. So, to me, it was a big relief to know that I wasn’t high risk for ovarian cancer. But I’m at the general population risk for ovarian cancer. (PP22, Victoria, filed conversation, September 26, 2001)

Both of the above examples show how valuable participants’ accounts are because they show how participants derived different interpretations from the same type of genetic test results. For participants who still see a possibility of carrying an inherited mutation (the majority), their newfound risk of ovarian cancer provoked anxiety. In Hallowell et al. (2004) study, similar findings were reported were women expressed experiencing increase anxiety from their newfound risk of ovarian cancer. Hallowell et al. described this increase anxiety about ovarian cancer as “biographically disruptive” to self-identity and that the anxiety experienced was similar to that of receiving a cancer diagnosis. However, the few participants in this study who interpreted their results as refutation of their mutation carrier status felt relief from no longer perceiving themselves at risk of hereditary ovarian cancer and saw no need to worry about reducing their risk.

Conversely, Louise, whose family history was only with ovarian cancer, felt relief in understanding that she was no longer at risk of hereditary breast cancer. Her relief, however, reflected how she interpreted and made sense of her test results. That is, Louise interpreted her results as having an inherited mutation predisposing her to ovarian cancer only. In her view, the
yet-unidentified cancer gene would be one that targets only one organ because her family history is only with ovarian cancer. This is how Louise illustrated her point:

I would be really surprised if I ever developed breast cancer. As the testing kind of indicated, because I had ovarian cancer, it is unlikely that I am going to get the breast cancer. I also find relief that I have survived my ovarian cancer and that I can impart this onto others. (Louise, PP13, field conversation, July 18, 2001)

Louise and Ginger's accounts above show how individual’s interpretation of their test results can have implications for their future use of screening procedures. These accounts also show how individuals who view their results as negative may be falsely reassured about their risk of breast and/or ovarian cancer. However, of the 21 participants, only one viewed her risk of breast cancer as being the same as the general population. Research still shows that having a family history of breast and/or ovarian cancer, with or without an identified inherited mutation, increases one’s risk of cancer above the general population (American Society of Clinical Oncology, 2003; Anderson, 1992; Goodwin, 2000; Jobsen et al., 2000; Menkiszak, Gronwald et al., 2004; Petrisek, Campbell, & Laliberte, 2000; Unic, Stalmeier, Peer, & van Daal, 1997).

To Bubaloo, who viewed her results as ambiguity regarding her mutation carrier status said that, for her, “No news is good news at this point.” Had she been found with a definitive mutation, “It would have been like feeling that there is a weight on your shoulders.” Although Bubaloo stated that she understood there were no guarantees with her results, she still saw a positive outcome. Like Erika, Bubaloo first struggled when asked what her immediate reaction to her results was. At first, she started to say “Uh no, I mean I was, it would have been kind of nice...Well no, I guess it wouldn’t have been nice.” Bubaloo was about to say that it would have been nice to know where her cancer came – it being so unusual for a 40-year-old woman and her father to both develop breast cancer at the same time. She quickly changed her response to, “No, it would not have been nice to know that the cancers’ cause was a mutation.”
Most participants, including Bubaloo, who looked to genetic testing to explain why they developed breast cancer were not ready to hear that it was because they had an inherited mutation. Participants in this situation were confronted with two challenging needs: the need to know where their cancer originated and the need to know that it was not the result of an inherited mutation. The underlying struggle could also be about gaining control over one’s health and how control could be diminished by an inherited mutation. Bubaloo gave an example of seeing the negative result as a positive outcome:

Ok I didn’t get any definitive information but I didn’t get any negative information, saying that yah we found it so I take that my negative reporting as being a positive outlook. (Bubaloo, PP9, field conversation, July 23, 2001)

Those who have interpreted their test results as a positive outcome referred to this interpretation as temporary to mean that as far as current testing technology could tell them, they do not have an inherited mutation. About half of the study participants shared this temporary positive outlook, corroborating Bish’s (2002a) findings (34 women in his study interpreted their inconclusive results in a positive light).

Like Bubaloo, Ginger found “a little bit of relief” with her results.

If we did find that mutation, then I would have been a little bit more concerned about my sister, her daughter and my daughter. So I guess I am relieved a little bit that they didn’t find anything for sure. And I know that doesn’t really put me out in the clear, you know, that it still could be there. But I have a little bit of relief that they didn’t find anything on the first go. (Ginger, PP11, field conversation, July 5, 2001)

Hence, for some participants, living with the certainty that they are at greater risk for breast cancer was preferable to living with the certainty that they carry an inherited mutation. As one unaffected, young family member said:

I was happy when we could not be tested. Well, this was one less thing to have to worry about. If it happens (developing breast cancer), then it happens, and then I will deal with it then. (Linda, SP2/PP20, Emma’s niece, field conversation, November 13, 2001.)
Linda’s mother and two aunts were diagnosed with breast cancer. Linda was physically and emotionally present for her mother’s breast cancer experience until her death.)

According to a study by Lim et al. (2004), unaffected individuals who had received a positive result in genetic testing for a mutation in either their BRCA1 or BRCA2 genes said that, once the initial shock of knowing they carried an inherited mutation had passed, they felt relief from this certainty. Lim et al.’s results echoes the views of a few participants in my study: Relief would have been greater had they received certainty with their results – certainty that they carried or not an inherited mutation predisposing them and their family at increased risk for breast and/or ovarian cancer.

Looking at the possible links between relief and how participants interpreted their results, those who interpreted their results as refutation of their mutation carrier status expressed greater relief than those who interpreted their results as confirmation of their mutation carrier status. Those who interpreted their results as ambiguity regarding their mutation carrier status gained some relief.

Stephanie, who interpreted her test results as confirmation of her mutation carrier status said that she immediately felt relief concerning her daughter. For the time being, she did not have to worry that she may have inherited a breast/ovarian cancer gene mutation until one was identified. Concurrently, however, she reported feeling sad at not getting an explanation for her breast cancer. At first, I thought this was distinct from the others’ experience but later on, two more participants expressed feeling no relief from their results. In fact, they saw individuals who receive positive results as the “lucky ones” for they knew why they had breast and/or ovarian cancer and why these cancers were happening in their families. Stephanie acknowledged during her interview that women in her support group had thought she was “weird” for feeling sad that she would have preferred a positive result for herself but not for her daughter.
Well actually it’s sad, because I thought that I wanted to have the gene so that I could understand what was going on. So I was sad, I was very sad, but then, in a day or so, I was able to step back and read the letter again… Uh, you know, I was looking for a reason and I still don’t have it, but now 2 years have passed and I still don’t have it, no big deal. I am sad, I wish I did have, but you got to deal with what was given to you. But then this heavy family history, but not one of these so-called mutated genes that we have.
(Stephanie, PP12, field conversation, July 18, 2001)

Stephanie gained partial relief from her test results only for her daughter, not for herself. She went into genetic testing hoping to find an answer about her cancer origin for her daughter’s sake. Still, she felt that, for the time being, she “can back off a bit” at pushing her daughter to get early breast cancer screening. With the passage of time, Stephanie found more relief. She continued to struggle, however, with her past family’s history with the disease and the risk of those who come after her. As Stephanie said, “All this history but not one of these so-called mutated genes.”

In summary, participants who felt relieved on receiving their genetic test results expressed relief in the following ways:

1. Knowing that they do not have an inherited mutation even if they do not know why they developed breast and/or ovarian cancer, nor what they can do to prevent cancer from reoccurring or appearing in their family members.

2. Knowing that they did not increase their children’s risk of developing breast and/or ovarian cancer by passing an inherited mutation.

3. Gaining control over external risk factors that are perceived as easier to control than controlling an inherited mutation.

4. Believing that they were no longer at increased risk for ovarian or breast cancer, depending on their family history of cancer.
5. Viewing their risk and their family members' risk of breast and/or ovarian cancer the same as the general population's risk.

**Disappointment and frustration with the results**

The participants who expressed disappointment and frustration with their results generally felt that, "These results are not telling us anything." Participants who interpreted their results as either confirmation of their mutation carrier status or ambiguity regarding their mutation carrier status felt that the test did not tell them if they were at higher risk of breast and/or ovarian cancer. Those who interpreted their results as refutation to their mutation carrier status were disappointed that test results did not explain all the cancers in their family.

As discussed in chapter 4, many participants had decided to have genetic testing for others first, then for themselves. Participants who were hoping to provide their family members with an assessment of their genetic risk felt great disappointment of being unable to reassure their family.\(^{78}\) Conversely, Frost et al. (2004) hypothesised that greater frustration would result among those who wanted genetic testing for personal decision making than those who wished to provide information to family members. Their hypothesis has limited applicability to my participants' sources of frustration since many of them explained that genetic testing had little relevance for personal decision-making because they already had cancer. What they really wanted to know was whether their family members were at increased risk as a result of an identified inherited mutation. Most of these participants felt frustrated at not getting an answer.

Participants told me they were unaware that they could receive any other result than a definitive positive or negative. Although this awareness may not reflect the protocol used by the Hereditary Cancer Program, it does reflect the recollections of participants. When I asked Joyce

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\(^{78}\) You may recall as well from chapter 4, that many of the participants came to view themselves as the family guardian and one of their roles is to provide them with concrete answers about their risk of cancer.
if she had found some relief in that perhaps she had not passed a genetic mutation to her children, she responded, “No, who’s to say that I did or I did not since the results are inconclusive?”

The following comments are typical of those who said they found no relief because of the lack of answer genetic testing provided them:

In a way, one would almost like to know...to have the other answer: Yes, you do carry this gene. I mean, I think that you would...one would get more of a sense of relief hearing that than this: “One they say ‘no’ and the other is 65%. There is still room for doubt.” I think we’re all looking for that answer. And I think these results do not give you something to sort of hang your hat on to. And I don’t feel this is an answer. The answer isn’t there. It’s not negative results. Not negative in the sense that we don’t have a mutation. (Donna, PP14, field conversation, August 22, 2001)

Contrary to some other participants, Donna’s need to know why breast cancer clusters in her family was greater than her fear of hearing that it was due to an inherited mutation. As Donna pointed out early in her interview, the first she knew of breast cancer in her family came from her mother who was diagnosed in her late 50s. Apart from her mother, she does know of anyone else in her family who has had breast cancer. Now, both she and her sister developed breast cancer at the age 49. She truly fears for her daughter and nieces, and she would have liked to provide them with a definitive answer to their risk of hereditary breast cancer. Contrary to Donna, Erika shared how she was hoping to find an answer with her genetic testing, yet she did not want to hear that she had the gene.

In line with Donna and others who shared her view, a participant from the Hallowell et al. (2004) study also described how, although she hoped her genetic test results would be negative, she would rather receive a positive result than live with the uncertainty of not knowing whether she carries an inherited mutation. Had her test results provided Donna with certainty of her genetic risk status for an inherited mutation for breast cancer, she would have viewed this result
as a positive outcome because “Now I would finally know.” However, the results she received just delivered more uncertainties.

Barney also would have felt more relief had she obtained a positive result (been found with a mutation). Here is how Barney expressed her lack of relief:

I didn’t really find relief. I think it would have been better to know, because then you’ve got, you can fight that more. But not knowing – now it’s a waiting game. I think it would have been better to find out you’re positive. (Barney, PP7, field conversation, April 12, 2001)

Barney interpreted her results as confirmation of her mutation carrier status. She still felt that her daughters were at “100% risk” of developing breast cancer one day. She was disappointed with her results because she hoped her daughters could have used the results to convince their doctors to consider them at high risk of developing breast cancer. She had, further, hoped that her daughters could then become eligible for chemo-preventive therapy, such as starting to take tamoxifen. Not knowing with certainty if they were at risk of carrying an inherited mutation, Barney feared that her inconclusive results would prevent her daughters from having this option or others, such as prophylactic surgery. She said, “Now, it’s a waiting game”: Her daughters must wait for cancer to appear before taking action. Had she received a positive result, her daughters could have been tested themselves.

Most of the family members interviewed were also disappointed about the lack of options available to them, since these options depended on their family member receiving conclusive results. Even male partners spoke about the same concern for their daughters and nieces.

I felt disappointed in the way that my understanding was that if, Emma tested positive – well, knowing that she has cancer anyway, and knowing that there is a family history and that is probably the reason why she got it – then I understood her brother could have gone in for testing and her nieces to see if they carried the gene. And it would give them a piece of information to go on. What they would do with the information was up to them but, at least, if she tested positive, it didn’t have to end there. Now, with these results, there is nothing next for them to do. I believe there would have to be a link anyway. And
when she tested negative, I was a little disappointed, because I was so sure she would test positive because of the strong family history there. (William, SP2/PP20, Emma’s husband, field conversation, November 27, 2001)

The next participant, Gilligan, said she felt no relief when she got her results, because they were not definitive, compared with what her oncologist had told her.

I was probably 40 or 41 at that point. And so when my oncologist said to me about a 50% chance of getting another cancer regardless of the results of the genetic testing, I decided that I would go for a mastectomy and reconstructive surgery, which I have done. And so, when I got the results of the genetic testing, I was, it’s a let down. I mean like a nonanswer more than, um, it saying that you are clear. I never felt any relief or anything. I just felt dumbfounded. (Gilligan, PP16, field conversation, August 14, 2001)

Gilligan first developed breast cancer at 26. As with her mother’s first diagnosis, life remained relatively unchanged until her friend was diagnosed with breast cancer, almost 15 years after Gilligan’s diagnosis. It was then that Gilligan decided to investigate further her risk of another breast cancer. Having gone through genetic testing, Gilligan did not find her results helpful in any way. She rather saw her results as a letdown. Perhaps Gilligan was looking to testing for corroboration of her surgical decision. Instead of corroboration, she received ambiguity. However, she did specify that she did not need the test results to make her decision to have her bilateral prophylactic mastectomy because she felt that her risk of developing a subsequent breast cancer was high enough based on her oncologist’s assessment. To Gilligan, the information she received from her oncologist was more helpful to her in deciding to have the surgery.

In contrast to participants who experienced relief in comparison to those who said they did not, those who perceived their genetic testing experience as providing “no results” often defined the whole genetic testing experience as useless. Those who contemplated the possibility that they may not have an inherited mutation obtained some relief, being able to see that their family members may not have an inherited mutation as well.
In summary, participants who felt little to no relief on receiving their genetic test results have expressed lack of relief in the following ways:

1. Not having been given an answer on whether or not they carry an inherited mutation.

2. Testing having created more uncertainty about their possible risk of carrying an inherited mutation.\textsuperscript{79}

3. Living with uncertainty to cancer risk was more stressful than living with the certainty that you carry an inherited mutation.

4. Results providing no confirmation of their genetic risk status, therefore reducing opportunities to take part in preventive measures offered to known carriers of BRCA gene mutations.

5. Results providing no support for health-related decisions.

\textbf{Informative Value of Testing Results for Health-Related Decisions}

There were only two participants who entered genetic testing to help them make health related decisions such as to have prophylactic surgery to decrease their risk of breast and ovarian cancer. Juniper described her view of the value of her genetic testing in deciding on prophylactic surgery:\textsuperscript{80}

> Should we be having things cut out of our body simply not to get cancer? Uh, ethically, I don’t know if we should be doing that unless, unless they get proof that it’s very important that you do this because, you know, of genetic proof. (Juniper, PP2, field conversation, December 13, 2000)

\textsuperscript{79} This view especially held true for participants who had came in to genetic testing not really considering that they may carry an inherited mutation. After testing, they had to consider this possibility, because it was one of the possible interpretations offered in the letter from the Cancer Agency.

\textsuperscript{80} Although this topic was not a main focus of the study, I discussed it within the context of reasons for having genetic testing done. Research on this topic would be tremendously useful. We could look at how individuals who asked to have genetic testing done in order to help them make health related-related decisions dealt with receiving what clinicians view as uninformative/inconclusive results.
Juniper explained that she had felt pressured by her oncologist to have prophylactic mastectomy of her remaining breast and oophorectomy because of her personal and family history of breast cancer. Because she had long ago decided she would not fear the disease as her mother had, she had decided not to have this surgery and instead to “wait it out” to see what her genetic test results would say. Having interpreted her results as refutation of her mutation carrier status, Juniper was happy with her decision not to have the surgery. Juniper questioned whether it was ethical for health professionals to recommend prophylactic surgery without “genetic proof” that an individual had an inherited mutation. This comment certainly speaks about the difference between potential risk (such as being considered at risk for HBOC) and actual risk of HBOC (being identified as having an inherited mutation in the BRCA1 or BRCA2 cancer genes) and the value of this information in making health-related decisions. Certainly, the literature agrees with Juniper that, when there is a known mutation in the family, identifying individuals with or without the mutation is important – so that they may consider prophylactic surgery (Meijers-Heijboer et al., 2000; Narod, 2002).

In essence, Juniper viewed genetic testing as a tool that could provide individuals with information to help in decisions about the removal of otherwise-healthy body parts. Had her test results been positive, Juniper said she would have agreed to a full hysterectomy with oophorectomy and a prophylactic mastectomy of her other breast.

Stacey, a family member interviewed, also decided to wait for her cousin’s test results before making any decision on prophylactic surgery. She had asked her cousin, Joyce, to have genetic testing for the entire family, since Stacey was not a candidate. She, nonetheless, felt quite at risk of breast and/or ovarian cancer after having cared for her sister, who died after being diagnosed with breast cancer at the age of 46. After Joyce’s test results, although Stacey still
struggled with the possibility of having an inherited mutation, she decided not to have prophylactic surgery.

It's got to be genetic with all of those people. But once the first negative result came in, I was warned that quite a few people are finding the second test is also negative and there is a group of people that they just haven't found what's going on with them yet. It's kind of a mixed reaction. Like on one hand, I was thinking, okay, so, um, so I was prepared at one point if they did find that we do have the gene that I want to be tested and know for sure if I was. Because I am proactive, I was going to do something about it. Um, that option was kind of put on the back burner waiting for the second test and then, when I got a negative result on her second test, um, kind of talking with the doctor and we decided, well, at this point, we're really doing as much as we can. Like, I don't want to go in and have radical surgery, because, I mean, I still might not be a carrier or not have that, you know, genetic problem. (Stacey, SP1/PP3, Joyce's cousin, field conversation, April 6, 2001)

Juniper and other participants seem to believe in the power of genetic information – what is known as genetic determinism, the view that we are the product of our genes and any negative outcome in our lives can be blamed on defective gene (Lippman, 1998). However, as with other multifactorial diseases with genetic components, even if an individual inherits a BRCA1 or BRCA2 mutation, her lifetime risk of developing breast cancer is not 100% but 50 to 85% (Lynch et al., 1999). According to the above account, Juniper certainly believes that genetic testing can provide “black or white answers.” That is how she has interpreted her genetic testing results as well: that she does not have an inherited mutation in either BRCA cancer genes. Most of the other participants do not share Juniper’s view; they see their results as ambiguous – they cannot use such information in deciding about risk-reducing medical procedures.

As I briefly alluded in the section Implications of Living with a Family History of Breast and/or Ovarian Cancer, some participants spoke of genetic testing as being one more tool they could use to gain control over their “chronic disease.” They planned to use the test results to consider how often they should have clinical breast exams and mammography and to motivate themselves to more regularly examine their breasts. They also planned to use the information to
convince family members to modify their lifestyle behaviours and diet. However, because of the uncertainty of their test results, they could not be used for the above purposes.

Also, participants like Bubaloo wanted reassurance that they had previously made a correct decision to have prophylactic surgeries. Following a recommendation from her surgeon, Bubaloo had requested genetic testing before deciding to have a prophylactic bilateral mastectomy. However, seeing how long it was taking to get her results, she decided to go ahead with the surgery. The waiting period did become an issue – and a source of dissatisfaction for her. Very few participants had entered genetic testing to help them make a decision about prophylactic surgery. Certainly, some did say that if they had been found to carry a mutation, they would have considered having their remaining breast removed (most participants who received genetic testing already had one breast removed as part of their treatment) and prophylactic oophorectomy (if not already done).

Although Bubaloo and the other participants did not get the reassurance they sought from testing, they still feel happy with their decisions to have had prophylactic surgery. As most participants said, “It doesn’t matter what the results say. I am still at high risk, and we still have all this family history of cancer.”

Bubaloo commented that, had she waited to receive her test results before having prophylactic surgery, getting uninformative results would have confused her. As other participants complained, the lack of information from uninformative results put them in a difficult position in terms of what to recommend to family members even without assuming the role of family guardian. The participants who did think that their children needed to consider prophylactic mastectomy or oophorectomy still felt the same way after obtaining their test
results. It is not so much genetic composition that they felt determined someone’s risk of a
disease but the family’s history with it (Frost et al., 2004).

Contrary to affected participants whose risk of cancer may feel more real, not all
unaffected family members interviewed opined that having a strong family history of breast
and/or ovarian cancer was reason enough to go ahead with prophylactic surgery, unless one is
found with an inherited susceptibility to the disease. Of all the 15 family members interviewed, 3
of which were husbands, none had decided to have prophylactic mastectomies. Meijers-Heijboer
et al. (2000) described that 35 out of 68 unaffected women with an identified mutation eligible
for prophylactic surgery chose to have bilateral mastectomy, while out of the 36 of those 60
eligible for oophorectomy chose to have the surgery. This shows the high demand for
prophylactic surgery among unaffected women. Their findings support the findings of Meiser et
al. (2000), which indicate that a significant proportion of unaffected women at high risk of
developing breast cancer may consider prophylactic surgery should they be found with a
mutation.

At this point, I would like to relate Bubaloo’s experience of how she arrived at her
decision to have a prophylactic mastectomy, because her account speaks to the value of certainty
– either in medical results or in medical recommendations – when making health-related
decisions. Bubaloo’s first encounter with genetic testing was through her surgeon. Following two
unsuccessful lumpectomies in which no clear margins were reached, the surgeon suggested that
perhaps a unilateral mastectomy, or even a bilateral mastectomy, should be considered instead of
a third lumpectomy. Up to this point in her life, Bubaloo said that she had been a vibrant woman
engaged in sports, work and hobbies. It was actually following a fall while skiing that Bubaloo
decided to consult her doctor about a bruise on her upper arm that would not heal.
During the examination, her doctor suggested that she have a clinical breast exam. Bubaloo had just turned 40 and at the time did not think there was any reason to worry. Upon examination, the doctor felt a lump. He suggested she go for a mammogram. Bubaloo said that her mammography result was not conclusive but highly suspicious. Hence, she was sent for a fine-needle aspiration and was told the cells extracted were cancerous. However, given the density of her breast tissue and the depth at which the lump was situated in her breast, the clinicians could not specify the size of the lump. Bubaloo was also told that both breasts had many cysts and calcium deposits and that these should be assessed as well. She was then sent for a core-needle biopsy.

Following this test, Bubaloo had a lumpectomy. Tissue analysis of the lumpectomy revealed a fast-growing cancer, “an aggressive type of cancer,” she said. Thereafter, Bubaloo’s surgeon recommended a second lumpectomy, since they had not achieved clear margins with the first. She also had chemotherapy following this second lumpectomy as recommended by the Cancer Agency to which her tissue had been sent and reanalyzed. The Cancer Agency also recommended that she either consider a third lumpectomy or a mastectomy considering her family history of breast and ovarian cancer.\(^8\)

This is when her surgeon approached Bubaloo about going for genetic testing, in the hope that it would help her make a decision about having a bilateral mastectomy. Bubaloo said that he had the impression that obtaining genetic testing results would be a quick process; they know now that it is not. Tired of waiting, Bubaloo decided to have the bilateral mastectomy followed by reconstructive surgery after her oncologist said, “Despite the outcome of your results, because of your family history, your risk of having another breast cancer and/or a recurrence is 50%.”

\(^8\) In Bubaloo’s family history, there are both breast and ovarian cancers. Her father and paternal aunt were diagnosed with breast cancer, while another paternal aunt was diagnosed with ovarian cancer.
Bubaloo shared how she felt she could no longer live in fear of a recurrence or a second primary cancer, and that it became easier for her to think of living without her breasts instead. Her first priority, as she said, was survival. Although Bubaloo went ahead with the surgery before obtaining her results, she does not regret her decision. What Bubaloo found useful in helping her make a decision about the bilateral prophylactic mastectomy was the information she obtained from her oncologist. Bubaloo felt it was definitive enough information on which to base her decision.

Conversely, none of the participants said they had actually undergone prophylactic surgery following the receipt of their results (or for the secondary participants, their family members’ results) perhaps because they all felt the test results were uninformative. However, I must add that, out of 21 participants, 15 had already had, as part of their cancer treatment, mastectomies and/or oophorectomies before undergoing genetic testing and only two participants had their surgery while waiting for their test results. The remaining 4 had lumpectomies as part of their breast cancer treatments. At the time of the interview, none of the remaining 4 participants said they were considering prophylactic surgery following the receipt of their genetic test results. Thus, the result of their genetic testing was not key in assessing their risk of future cancers.

Some participants said that they had hoped that genetic testing could help them convince their family members to consider prophylactic surgery, but the uncertainty of their results prevented them from doing so. For example, Barney was convinced that her daughters will one day develop breast cancer. Hence, she really had hoped that her test result would motivate her daughters to have prophylactic mastectomy and prophylactic oophorectomy or take tamoxifen, once their families were complete. In the end, Barney saw very little, or no, value in her genetic...
testing results toward this purpose. She said that she felt cheated because health professionals may now use her test results to deny support to her daughters should they choose one day to have prophylactic surgery. Study participants in Frost et al.’s (2004) research who received uninformative genetic test results commented that their siblings would have probably changed their coping strategies, had their test results shown they carry a mutation.

In reality, only two of the study participants said they underwent genetic testing to help them make health-related decisions. Although Macy had a second, prophylactic mastectomy while waiting for her test results, she wanted genetic testing not for herself (as it was too late) but for her family. Macy explained that, at the time she decided to have her second breast removed, she had already been told that no mutation had been found in her BRCA1 cancer gene and that her second BRCA cancer gene was being screened. She had decided to have a bilateral mastectomy when she was offered it as part of the treatment to her right-side breast cancer. She also asked to have both of her ovaries removed because of her perceived increase risk for ovarian cancer and because she “would already be under general anesthesia.” Macy said the last decision was more emotional than rational. She just “wanted them gone,” speaking of her ovaries and breasts. After Macy received the result of her second test (on BRCA2 gene), she said that she was still happy with all of her decisions, considering that the Cancer Agency could not confirm that she does not carry an inherited mutation.

Not all the participants would so willingly have mastectomies as part of their breast cancer treatment or prophylactic mastectomies and oophorectomy further down the road. If they did, they reached such a decision not just based on the potential outcome of genetic testing but on their need to survive and their need to reduce their cancer risk. Emma was diagnosed with breast cancer at 43 and lost her two sisters to the disease specified:
I was offered a mastectomy as part of my treatment but asked to have both removed. It was a hard decision, but it wasn’t. It was like a decision that was already made up for me. There was no choice. I didn’t have a choice. ‘Cause, you know, it’s either die or live without breasts. Well, I guess, it’s live without breasts that are going to win out. They are not that important over dying. (Emma, PP20, field conversation, October 27, 2001)

Another participant spoke to the same issue, the need to survive “at all costs”. Unlike Emma, who has no children, Sherry had two young children at the time of her breast cancer diagnosis (at 35).

I was determined that I was going to see them graduate. Death wasn’t an option, so it didn’t matter what choices they gave me. Whatever it was that was going to give me the best chance for survival was the choice I made. (PP21, Sherry, field conversation, September, 18, 2001)

In all of the preceding cases, lived experience (seeing death from cancer in others in the family) was a major factor in deciding to have mastectomy and/or oophorectomy and not the potential outcome of genetic testing. To this end, other studies also show that there is a tendency towards use of mastectomy at younger ages among individuals with a family history of breast cancer considered at risk for HBOC (Gershenwald et al., 1998; Meijers-Heijboer et al., 2000; Meiser et al., 2000).

After discussing with participants whether they felt relief on obtaining their test results and whether these results had value in helping them make health-related decisions, I then asked: In light of the above, what are some benefits and/or potential harms you perceived or experienced as a result of having gone through genetic testing? Potential harms refer to negative consequences foreseen or lived as a result of genetic testing and ethical issues identified by the participants.

Benefits and Lack of Them

Participants who had genetic testing for others only described the benefits they foresaw as follows: “The results make no difference to me, only to my family.” For the others who had
genetic testing for self and others viewed the benefits either as few or many, depending on the type of interpretation they gave their results. For example, those who interpreted their results as refutation of their mutation carrier status identified as a benefit knowing that their children are no longer at risk of hereditary breast and/or ovarian cancer.

Participants who, before entering genetic testing, felt they most likely carry an inherited mutation said that they gained very little benefit from their genetic testing experience. However, given their prior expectation, they did not put much value on their test results, since they had already been diagnosed with breast and/or ovarian cancer and therefore knew they were at increased risk for it. As many of these participants expressed, “I don’t need to know that I am at increased risk of developing breast and/or ovarian cancer as I am living proof that I am by the mere fact that I have developed breast cancer.” (Field notes, August 2003)

But specifically, Becky said,

I have already been diagnosed with cancer, so having the gene isn’t going to matter. (Becky, PP8, field conversation, April 17, 2001)

Participants who entered genetic testing hoping to find their cancer’s origin did not share Becky’s view. What would have benefited them would have been finding out with certainty whether or not they carry an inherited mutation, so that other family members could be tested if they wished too. Hence, these participants may have felt greater dissatisfaction with their uninformative genetic test results. The four participants who interpreted their results as refutation of mutation carrier status were satisfied with their test results and derived benefit from them: knowing with certainty that they do not carry an inherited mutation and consequently have not passed one to their children. While they listed another benefit expressed, knowing that their siblings are no longer at risk of an inherited mutation, in reality the Agency would judge other
family members’ genetic status to be undefined. Becky, although still seeing some possibility of having a mutation but not holding strongly to this belief, felt testing benefited her and her children in that she now knows she has not passed a mutation to her children.

For most of the participants who felt that their test results did not provide them with certainty, they still felt that they gained some insight into possible causes of their breast and/or ovarian cancer. They could either conclude that the possible inherited component to their breast and/or ovarian cancer is not one of the tested mutations or most likely not in the tested mutations. This is more information than they had before testing and gives them a deeper understanding of their own cancer and their family’s cancer history. However, when some of these participants thought further about the utility of this information, they either realised its limited value in supporting health-related decisions or were uncertain what they could do with it.

Evelyn, who interpreted her results as ambiguity regarding her mutation carrier status said that she gained very few benefits from her genetic testing experience, because very few decisions can be made with “inconclusive results,” as she called them. She had hoped that her test results would urge one of her sisters to start tamoxifen because nothing prevented her from taking the drug, unlike her other sister who had a history of phlebitis. Evelyn said that although her results gave her more information about the origin of her own breast cancers and those of her family, the results did not provide her with more power nor with knowledge to encourage others to make risk-reducing decisions. As a few participants who undertook genetic testing for others first, and then for self put it, “Receiving uncertain results took away my ability to probe my family in order to protect them.” Hence, they saw their results as having no benefit for themselves or for others. Here is Evelyn’s opinion:

82 The family members, genetic risk status would be judged as undefined because they still could have inherited a mutation (if one is being transmitted within the family). The risk of inheriting a mutation is 50%. That one member did not inherit the mutation does not decrease a sibling’s risk.
There are no benefits with these results. If there was a gene in the family and they (her sisters and cousin) didn’t have it, then I could stop worrying. But if they did have the gene, I don’t think they would have a problem getting their ovaries out, although breasts probably would be a different story. But maybe the medical field would follow them more closely, like they do here. At least it would give them peace of mind. (Evelyn, PP5, field conversation, April 9, 2001)

The expectation that tested individuals and their family members receive better health surveillance after a positive test result has been documented by other studies (d'Agincourt-Canning, 2003; Di Prospero et al., 2001). As Di Prospero (2001) asks, are genetic testing agencies creating a false sense of security by promoting the belief that surveillance may reduce the risk of cancer occurrence or recurrence? The current study participants saw as one benefit of genetic testing that their names would be kept on a contact list for when new breast and/or ovarian cancer genes are discovered. They expected to be among the first people tested for any newly discovered genetic mutations and to be told about any new treatment or identified risk factors. Parsons, Beale, Bennett, Jones, and Lycett (2000) also found that participants in their study held such beliefs. The group of 43 women at high risk of breast cancer talked about the reassurance they had found from their results, because they had met with an expert and become members of a surveillance group. These authors also question the potential tension between individuals’ expectations about ongoing surveillance and the realities of health care provision.

The few participants (4) who felt that genetic testing provided them with definitive answers about their genetic risk status were more inclined to view their results as informative.

It makes me feel better, it does, it definitely does, you know. It feels good to be able to say, oh, I don’t have that gene, it feels good to be able to tell my husband, oh, you know, I got the results, and. And it feels great to be able to tell my sisters that too. (Erika, PP10, field conversation, July 9, 2001)

Contrary to many other participants, Erika felt she received a definitive answer about her genetic risk status. Contrary to others as well, she did not expect her results to be anything but
negative – refutation of mutation carrier status. Seeing how these 4 participants’ interpretations different from those of the other 17 participants who interpreted their results as confirmation of their mutation carrier status and ambiguity regarding mutation carrier status, I looked for some characteristics that these 4 participants might have in common that the 17 others did not. I found one major difference: Three of the four individuals did not seem to have been exposed to as much breast cancer physically or emotionally, in that they lived far from affected relatives or were not emotionally close to them. Hence, the amount and the quality of secondary experiences of cancer in family members influenced the participants’ perception to a possible inherited mutation. Connectedness and knowledge of other family members experience with cancer in influencing personal perception of risk for diseases reflects what Abel and Browner (1998) termed “empathetic knowing.”

As Erika explained, neither of her two sisters had experienced breast cancer, she had no contact with her cousins because they all live overseas and, although her aunt and grandmother died of breast cancer, she had had very little contact with them because telephone service in her country of origin was poor. Consequently, as hypothesised by Cox and McKellin (1999), one’s perception of risk for an inherited mutation may be influenced not by the amount of exposure one has to the disease but by the quality of this exposure. Hence, this low-level of exposure to her relatives’ cancer may explain Erika’s expectation that her test results would show she does not carry an inherited mutation.

In contrast, another of the four participants who interpreted her results as refutation of her mutation carrier status had lived through her mother’s cancer diagnosis, physically and emotionally, yet put her faith in the testing technology and believed that as far as what the
technology could test for, she did not have an inherited mutation. However, she and her mother were the only breast cancer cases in the family history.

One final difference I found with the four participants who interpreted their results as refutation of their mutation carrier status is that they entered genetic testing expecting to be told that no mutation would be found while 12 out of the 17 participants who either interpreted their results as confirmation of their mutation carrier status or ambiguity regarding their mutation carrier status either entered genetic testing expecting to be told they had an inherited mutation or knew of the possibility of being told that they could have an inherited mutation. The other five of the 17 participants entered genetic testing not expecting to be found with an inherited mutation. Once they received their letter explaining how the results could be interpreted, four of these five individuals interpreted their results as ambiguity regarding their mutation carrier status while one now perceives herself to have an inherited mutation because of her letter establishing this possibility. She now interpreted her results as confirmation of her mutation carrier status. In all, five participants changed their personal risk assessment to a probable inherited mutation from pre-test to post-test. However, these findings have to be interpreted with caution as the link between pre-test and post-test to their perceived likelihood of carrying an inherited mutation to breast and/or ovarian cancer cannot be directly ascertained with the current study design. The study is retrospective and, therefore, the participants accounts may not be totally dependable.

Conversely, those who felt genetic testing had not provided them with definitive answers were more inclined to see limited informational value in their test results. As one participant
said, "I just don’t think they are letting people know enough that they may not get any results."

As recommended by Frost et al. (2004), affected individuals considered at risk for HBOC who undergo genetic testing should be told that three categories of results are possible: Individuals can either (a) be found with a known BRCA1 or BRCA2 gene mutation, (b) be found with a BRCA1 or BRCA2 gene variant of uncertain significance (meaning that the clinical relevance is unknown), or (c) be told that the test did not demonstrate an identifiable mutation but, because of their family history, the result is considered inconclusive in establishing their genetic risk status. While the categories of results recommended by Frost et al. are reflective of the categories given to individuals who have genetic testing at the BCCA Hereditary Cancer Program (HCP) where the study participants had their testing, the HCP goes further in their explanation about the third category recommended by Frost et al. That is, when there are no specific mutations found in the BRCA1 or BRCA2 in individuals considered at risk for HBOC, the HCP further acknowledges the possibility that a mutation may still exist in the regions of the BRCA1 or BRCA2 that their lab looked at, but that it has not been detected by the current testing method. Individuals tested at the HCP are also told that a mutation may still be present in the as yet untested portion of the BRCA2 gene.

While for some participants, their initial perception to a possible inherited mutation had changed from pre-test to post-test, all participants except Erika felt that their test results had not, however, significantly changed their perceived increase risk for cancer. Gladys’s comment illustrates this group’s continuing fear of cancer.

Every since I had breast cancer, I’ve never felt cured and having come across so many people that have been ‘cancer-free’ from 1 year to 20 years and then it comes back. I just don’t feel that you can ever say that you’re cured. (PP17, Gladys, field conversation, August 30, 2001)

Marcy also spoke about this continuing fear.
I was doing it for the rest of the family, to put them at ease, but it hasn’t put anybody at ease. Because it just leaves you up in the air. It just makes me realise I just have to, I have to do the same thing and so do they. And I’ve told them that, you do exactly the same thing. (Marcy, PP18, field conversation, September 13, 2001)

After Marcy said this, I asked her if, because she felt “left up in the air with her results,” if she felt disappointed in having invested time in going through genetic testing. Marcy replied,

No, I don’t, because eventually, if they find that they (a relative) have got a lump or something like that, they might want to then go farther and find out about the other test that they can take. And because we’ve already got our names down in the thing, I think the procedure is they don’t have to wait as long. They can just go in and get tested. (Marcy, PP18, field conversation, September 13, 2001)

Other participants also mentioned this benefit – that, by having had genetic testing themselves, their family member would be able to get genetic testing sooner and obtain results faster by “already being on the list.” Perhaps this assumption comes from what they partially recall from their genetic counselling sessions. They were told that, if a mutation is identified, it becomes faster to test other family members’ genes, because the laboratory technician knows exactly where to look on their gene. This was not the case for any of the participants who received uninformative results because none of them were found to have a mutation.

The participants waited an average of 2 years before obtaining their results. This combination of a long wait for results and then not seeing any benefit from them led many participants to conclude that their entire genetic testing experience was a waste of time. Barney was straightforward about this.

We were hoping for benefits but we didn’t get anything out of it. I think it was a waste of time. And I’m glad my kids didn’t go for counselling and the rest didn’t go for counselling. It would have been a waste of time. (Barney, PP7, field conversation, April 12, 2001)

After getting their results, some participants saw themselves at less risk of ovarian cancer or breast cancer (depending on their family history). This they listed as a benefit of testing.
Those who felt relief from seeing themselves less at risk of ovarian cancer were those who interpreted their results either as not having an inherited type of cancer (and therefore their breast cancer does not increase their ovarian cancer risk or if they do have an inherited type of cancer, it is one unrelated to BRCA1/2) or having an inherited mutation that specifically targets one organ, the breasts or the ovaries. In the following quotation, Victoria discussed how she saw benefit in her test results because of the above reasons. Following that, Red speaks to the lack of any benefit from her genetic testing experience.

Victoria has no family history with ovarian cancer but was diagnosed with breast cancer at the age of 37 and was 48 at the time of the interview.

To me it was a big relief and a benefit to know that I wasn’t high risk for ovarian cancer, but I’m at the general population risk for ovarian cancer. (Victoria, PP22, field conversation, September 26, 2001)

In Victoria’s mind, she has an inherited mutation but not a mutation in the BRCA cancer genes. Otherwise the Agency would have found it with currently available technology. So, after receiving her test results, she felt that she is no more at risk of ovarian cancer than the general population. To her, this is a benefit of testing.

Red, in turn, saw no benefits from genetic testing at all.

There is no benefit to me. Absolutely not! Because I think a lot of it is dreadful. And why would I want someone telling me I am a candidate for breast cancer when I’ve already had breast cancer? Do you know what I say? The benefits and risks for having done the testing are nil at present and I think it is a waste of funds and tax dollars. (Red, PP4, field conversation, June 28, 2001)

Red’s view echoes those of most of the participants who had already internalised their increase risk for cancer and did not need a genetic test to confirm this perception but, instead, hoped the test could bring some certainty to others in the family about their risk. They identified further limitations of their genetic testing experience.
I don’t think the results have any information. It’s not black and white; I mean people without the gene get breast cancer obviously. It leaves me in limbo really. I think we said the results were inconclusive – that’s limbo right! (Laughing) Well, and that’s like saying you’re back to square one, you really don’t know...Me having a positive result would not necessarily mean that my cousin and sisters would have the gene, but it would mean that they could at least get tested and then they would know more information. (Evelyn, PP5, field conversation, September 2001)

Family members interviewed who were direct blood relatives listed some benefits resulting from their family member’s test results: (a) having a more positive outlook on their perceived likelihood of carrying an inherited mutation predisposing them to an increase risk for breast and ovarian cancer and (b) recognising that they need to adopt more risk-reducing behaviours and have regular screening. Note that the first listed benefit only held true for family members who interpreted their relative’s test result as either ambiguity regarding their mutation carrier status or refutation of mutation carrier status.

It made me feel more in control of what might happen. I think it made me feel less at risk, less vulnerable because it wasn’t BRCA1 or BRCA2. (Janet, SP1/PP5, Evelyn’s sister, field conversation, April 5, 2004)

As a result of having had her sister go through genetic testing, Susan says that her two daughters have become more aware of their risk to cancer and have become more regular with their screening. (Susan, SP1/PP21, Sherry’s sister, field conversation, October 9, 2001)

It has helped me to think more clearly, to make sure I keep up with breast self-exam – especially since the results were inconclusive and I don’t see them as a negative. (Diane, SP3/PP5, Evelyn’s cousin, field conversation, June 19, 2001)

I found that family members interpreted their relative’s test results the same way that the relative did. Thus, when their relatives interpreted their results as ambiguity regarding mutation carrier status or refutation of mutation carrier, family members felt the results gave them a better perspective on their increase risk for breast and ovarian cancer. However, family members whose relatives interpreted their results as confirmation of mutation carrier status saw few benefits in genetic testing. They saw few benefits because while they believed their family
carries an inherited mutation predisposing them to an increased risk for breast and/or ovarian cancer, the testing did not confirm nor contradict their suspicion.

**Potential Harms of Genetic Testing Foreseen by Participants**

In chapter 2, I reviewed risks of genetic testing as reported by authors who have studied the impact of such testing on people's lives. Much has been written on the potential and actual risk of genetic testing, but most of this knowledge comes from individuals who tested positive for an inherited mutation in their breast and/or ovarian cancer genes, or from knowledge derived from asking individuals what possible risks they could foresee if they accepted genetic testing for BRCA1 and BRCA2. In this section, I discuss potential harms that participants saw from their experience of genetic testing and as result of obtaining uncertain results. At times, they framed these potential harms as ethical issues. Potential harms identified by the participants fall within two main groups: risk of discrimination and risk-related to communication issues.

**Risk of discrimination**

Embedded within the risk of discrimination as a result of having had genetic testing are two different forms of fear. The first is fear of losing extended health and life insurance coverage. The second is fear of losing privileges accorded to people identified as high-risk for developing cancer, and their family members.

All the participants said that they had purchased extended health and life insurance before their cancer diagnosis and they therefore foresaw very little risk of losing it had testing found an inherited cancer gene mutation. They doubted that they would be denied coverage because they had already developed cancer. They did, however, foresee a risk for their family members. One family member, Kimberly, said that, because she felt protective legislation was not clear about
discrimination on the basis of genetic risk status; she declined to have genetic testing done. This family member was not actually eligible to be tested because she was unaffected.  

My insurance company or my work, whatever – they can’t say I won’t hire you. You can’t have insurance if the information doesn’t exist. Whereas, if you are already diagnosed with it like my sister, you got it. You are not in jeopardy. (Kimberly, SP1/PP1, Macy’s sister, field conversation, December 11, 2000)

Thus, the risk of discrimination as a result of genetic testing may differ according to whether or not individuals have already had cancer, even if they share the same family risk. However, Susan, a family member who had already had breast cancer, feared that her daughters could be discriminated against for life insurance if she chose to have genetic testing. Hence, she told her daughters to purchase life insurance before she signed her consent form. Both of her daughters did so.

Stephanie discussed potential life and extended health insurance discrimination and its meaning for her daughter.

All my life insurance policies and everything are all in place. They can’t take that away from me, they can’t say well you have cancer, you can’t be insured. Which is, I know, the big thing for a lot of people is can they get medical coverage? They may be turned down job or whatever. But you see for myself that, it had no bearing, OK. Those kinds of consequences that have bearing for other people probably would have bearing on my daughter, but we didn’t get there. The instinct of a mother for her child, there’s nothing like it. I had to know for myself and I had to know for my daughter. But when it came to the testing, it is a fact that I do not have a sister, I do not have cousins who my results were going to affect. So it didn’t even enter my mind, my results are going to only affect my daughter. (Stephanie, PP12, field conversation, July 18, 2001)

As for other participants, they feared that, as a result of their genetic testing, family members could be discriminated against in insurance coverage. However, they understood as

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83 Knoppers and Joly (2004) note that, in contrast to a large number of European countries, Canada still has not clarified its position regarding genetics and life insurance, prompting the creation of the Canadian Genetics and Life Insurance Task force. Knoppers and Joly (2004) report a Quebec case in 1990 where a family was denied benefits because an individual had not shared a family condition, even though he was unaffected by it at the time he bought life insurance. In Australian, while the Australian Law Reform Commission recommends avoiding unique laws for genetic testing compared to other medical information, they nonetheless recommend amends to the Disability Discrimination Act 1992 to clarify that the Act applies to past, present, possible future or imputed disability, including discrimination on the ground of genetic status (Australian Law Reform Commission, 2003).
well that they could use the uncertain outcomes of their test results to say that they do not have an inherited mutation. Conversely, among those who saw their tests results as incomplete and noted that further testing could be done in the United States, many said that, because of possible insurance discrimination, they would not pursue genetic testing any further unless their children asked them.

When they consented to genetic testing, they assumed that their results would be confidential and that, if the Cancer Agency was asked if a certain individual had genetic testing, the Agency would not answer without their approval. It is true that, in their genetic counselling sessions, participants were told that, unless they authorized it, their genetic testing results would not be disclosed to a third party. Nonetheless, professional confidentiality does not protect against possible extended health or life insurance coverage because insurance contracts are often based on disclosure of health records with the consent of the patient.

Sherry, who had started worrying about being denied life insurance benefits, said that, during her genetic counselling session, she specified to the Cancer Agency not to send her results to her doctor because she feared that, if more people knew of her results, the more risk there would be of discrimination. She also told her genetic counsellor that she did not want these results to be in her medical file and that, if her doctor wanted to know about her results, she would tell her verbally. Sherry interpreted her results as ambiguity regarding her mutation carrier status. Many participants said that, if their results had shown with certainty that they carry an inherited mutation, they would have asked family members who were interested in knowing their results to consider buying insurance before being told of the results.

A second form of fear of discrimination addressed by a few participants was losing their privileged access to screening as a result of not having been found with a mutation and therefore
being seen at less of risk of cancer by the clinic. This fear of discrimination could affect their
family members as well.

Well, I'm hoping that the doctors are bright enough to realise that the result of the testing
does not mean there's nothing hereditary and does not mean even that it's not the BRCA
1 or 2. And that therefore they wouldn't, the fact, the very fact that their family history
might have been brought to their attention, you know, focus on the fact that we were as a
family accepted into this program (genetic testing). And so, I am hoping that the doctors
and the members of my family would be even more alert and vigilant and that their
doctors wouldn't do that. (Becky, PP8, field conversation, April 17, 2001)

However, an equal number of participants, upon being told that in light of their results
they could now be monitored by their regular physicians, feared being seen as less at risk for an
inherited mutation. For Victoria, however, being told that she could now be monitored by her
regular physician felt like she was being “released” from the high-risk clinic. But, at the same
time, she felt she could lose her privileged access to cancer screening if she stopped going to the
Cancer Agency. She felt that this was better than being monitored by her family doctor.84 One
family member interviewed, Lily, said that, if her doctor saw her as less at risk as a result of her
sister’s genetic test results and no longer referred her for yearly mammograms (which she had
been getting since the age of 35), she would change her family physician.

I would say that to him, I would simply say that’s not fair. I shared some confidential
information. I didn’t need to give you the results, I didn’t need to even tell you that my
sister went for genetic testing. (Lily, SP1/PP2, Juniper’s sister, field conversation, April
4, 2001)

Erika’s account displayed her conflicted feelings after being told she no longer needs to
attend a high-risk clinic:

They told me at the cancer clinic that I didn’t have to go there for my follow ups
anymore. Even though I was happy about that, because it’s, you know, it’s not the
happiest place in the world, I still felt mixed about leaving the cancer clinic, because, in a
way, you feel comforted in the fact that you are in this fancy institution that people seem

84 In 2002 one of the health professionals at the Hereditary Cancer Program told me that their long-term aim was to
follow only families with a known inherited mutation. Hence, the participants’ fear of losing access to care by
cancer specialists may not be unfounded.
to know what they’re doing and you’re being looked after. But at the same time you’ve got that association of drugs going into your body and your veins feeling like they’re freezing. And, you know, and burnt-out veins and things like that. And all that association is with it. So I was happy when I didn’t have to go to 6th floor anymore. I was only going to the second or third floor to get my genetic test done and that was okay. To me, it meant a good sign that I didn’t have to go back up there as I didn’t want to go back up there. So, when they told me, it was like one step closer to being better. At the same time it’s like cutting that umbilical cord, like. And I sort of thought, oh, but my doctor will she know enough? I sort of wanted that security of going to the clinic where they specialise, and this is their thing, you know. They know everything about it. And so I felt a little, you know, apprehensive about not going there anymore. (Erika, PP10, field conversation, July 9, 2001)

Other participants feared that they and their family members might be less diligent with breast cancer screening, a fear that some family members interviewed also shared. Emma explained that, if she had been told that she did not carry an inherited mutation before being diagnosed with breast cancer, she likely would not have been as diligent at screening her breasts, despite having a strong family history of the disease.\(^8^5\) Whereas, by not having this information beforehand, she always felt and knew that one day she would develop breast cancer just like her two sisters did. Hence, although she was diligent with her breast screening, when her lump was found, she was told that it had been there for over a year. She imagined that her story would have been different had she had genetic testing prior to her diagnosis. Having survived her breast cancer and seeing her two sisters die from it, Emma added that, had she been told she had a mutation in her breast cancer genes, she would have decided not to have children. She felt strongly about this, even though she does not have children and is no longer able to. Emma interpreted her results as ambiguity regarding her mutation carrier status. She underwent genetic testing mostly for her niece, whom she views as a daughter. However, Emma said that she was surprised to find out that her niece was not interested in hearing of her genetic testing experience.

\(^8^5\) In contrast, participants from known HBOC families in d’Agincourt-Canning’s (2003) study who decline genetic testing said that they did so because they did not think that the results would change their medical management.
Emma thought her niece’s lack of interest was because she had shared with her niece that, if she were to be found with an inherited mutation, she would have decided not to have children.

During my interview with Emma’s niece, Linda, she did mentioned that she was engaged to be married and that she and her fiancé had discussed starting a family soon. She did not tell her fiancé that her family might be at risk of an inherited mutation. She said she did not want to discuss this topic with her fiancé without knowing whether her family has an inherited mutation for breast and/or ovarian cancer. Linda added that: “The breast cancer had more of an impact for me than the genetic testing, even with deciding to have children or not. (Linda, SP2/PP20, Emma’s niece, field conversation, November 13, 2001) However, later in the interview, she shared the following: Like her aunt, Linda said that she will have to think about whether she is going to have children or not, for fear of passing the mutation to them and seeing them die before her. Linda said that she does not see her risk of developing cancer as less because of her aunt’s genetic testing results. (Linda, SP1/PP20, Emma’s niece, field notes, November 14, 2001) Linda’s plan is that, if and when she gets breast cancer, she will deal with the situation at that time. In the end, it was not so much that Linda was not interested in her aunt having genetic testing, but more that Linda felt no matter what the results were, she knew that she is at high risk of developing breast cancer as a result of having three close family members diagnosed with the disease. Now, Emma’s and Linda’s situations take us right to the heart of the next area of potential harm identified by participants: risk communication issues.
Risk Communication Issues

Although the foci of this study do not include how individuals and families communicate genetic testing results, what I bring forward in this section are issues that participants themselves identified as potentially creating harm.86

Like in Di Prospero et al. (2001) study, more than a few of my study participants discussed how, after having genetic testing done, they realised how few of their immediate and extended family members were actually interested in hearing the results. Disappointment was mostly felt by those whose motivation for genetic testing was for others only. Because these participants wanted to give family members an opportunity to know their risk for an inherited mutation and their risk for breast and/or ovarian cancer, they still shared their test results with some family members who had said that they did not want to know. By doing so, these participants actually took away family members’ freedom as to when they would be ready to find out their risk for an inherited mutation.

Participants shared why they decided to share their results even when they knew the other person did not want to hear them. The first speaker is Stephanie, who shared her results with her 18-year-old daughter.

You see, I am kind of a forceful person and I don’t think she was given that option of wanting to know or not. If we are going to find out if I have the gene, then we will decide what we are going to do with it. I really don’t think I pushed her on the testing until I knew whether or not she was a candidate. And, as it turned out, she wasn’t. So, she said to me “Forget it.” I said, ‘No, we can’t forget it.’ You see, my primordial instinct is to protect my children, and that is why I still told her. (Stephanie, PP12, field conversation, July 18, 2001)

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86 Some discussion on the topic of communicating results did take place with some of the participants when they did bring it up. Topics addressed were how they communicated results to others, what information they communicated, and when they decided to communicate. These will be analysed at a future date.
Next, Emma relates how surprised and disappointed she was to realise how others in her family were not that interested in her experience of genetic testing, when her main reason for getting tested was to help her family know their risk of breast and/or ovarian cancer.

Nobody seemed to call me and ask me about it, you know. I told all of them what I was doing, having genetic testing and all and would they be interested in hearing the results. And they were all like, oh yeah, oh yeah, but not one of them called me back after that day to ask what’s happened. But, when I received the results, I told my niece that I am very close to. I didn’t give her a choice. I just kind of told her. It just kind of came blurring out. That I got the letter and that there would not be any further testing. And she was like, oh. But she didn’t really say anything else. And, I think it was that day that I found out that she didn’t really want to know the results anyways. She wasn’t really interested. (Emma, PP20, field conversation, October 27, 2001)

Marcy and Becky explained that, although they knew their family members did not want to know their test results, they still told them. However, had their results showed with certainty that they carry an inherited mutation, they would not have shared those results with family members.

Before I went for the genetic testing, I asked them if I should go and if they wanted to know. And they said yes. They said you don’t have to tell us anything, we consider that we are high risk and that’s the way it’s turned out. But when the genetic counsellor told me it was negative, that there was no genetic mutation as far as what they could test for, that is what I told them. (Marcy, PP18, field conversation, September 13, 2001)

Becky confessed the following action at the end of her interview:

I did do something that was probably naughty, but I did it anyway. Like I told you, I was surprised actually at the number of my cousins who said, ‘We don’t want to know the results. And my, I was surprised my daughter said, ‘I don’t want to know the results.’ I would have thought she would have wanted to know. But she explained ‘If I don’t know if I have the gene, then I can’t disclose it to the life insurance company. But the benefit is that I am getting the best screening I can. That’s good enough for me.’ But when the results came back negative, I told her. I said, ‘I know you told me you didn’t want to know. And if it had been positive, I wouldn’t, you know. But it’s not, we don’t have, you know. Apparently it’s not a 100%, but it appears we do not have those genes and I just want you to know that.’ She just started rolling her eyes and said OK. (Becky, PP8, field conversation, April 17, 2001)
Many participants who had requested genetic testing (the proband) or had genetic testing done (the index), told me it had not occurred to them before their genetic counselling session that they would have to contact family members either to seek their interest in being tested if they had a past diagnosis with breast or ovarian cancer or consider sharing their test results. A few said they were uncomfortable at the thought of having to communicate with family members with whom they had had little contact for many years.

Stacey, one of the family members interviewed, contacted affected family members to seek their interest in being tested on her behalf. (She was not a candidate, because she had never been diagnosed with breast and/or ovarian cancer.)

There was one cousin that, when I first approached her on the phone...I normally, she’s closer to my mother’s age, or she associates more with my mother. She’s like 70 years old. Um, it was a little awkward talking to her because we never talk. Christmas cards maybe; that’s about it. So, she was surprised that I was phoning and asked did something happen to your mother (laughing). You know, why are you calling? So I just sort of briefly told her what it was all about and she didn’t really answer. She said she’d think about it and she sent a message via my mother that she decided that she didn’t want to do it. And she didn’t really, wasn’t really interested in it. (Stacey, SP1/PP3, Joyce’s cousin, field conversation, April 6, 2001)

Stacey said that only one family member was receptive to her request: Joyce did not mind being called out of the blue by her cousin. Out of the five primary participants who had been contacted by a family member to have genetic testing, only one reported a negative experience. Her response was previously discussed in chapter 4 in the section of Hearing of the Availability of BRCA1 and BRCA2 mutation Testing.

As for communicating their genetic testing results, a few participants brought up the question of whose responsibility it is to communicate their results to family members who might benefit from the information. None of them thought there was an obligation to tell others of their

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87 The proband represents the first individual in a family to seek genetic counselling from the HCP while the index case represents the family member who consented to genetic testing.
results unless they chose to do so. In fact, these individuals decided not to tell any extended family members about their genetic testing unless the results showed that they carry an inherited mutation. Thus, as Lerman, Peshkin, Hughes, and Isaacs (1998) comment, most participants who had genetic testing desired to maintain control over the diffusion of genetic information to relatives.

Below are the accounts of two participants whose views echo a few others who chose not to share their results with family members.

Mimi said,

Like, if it came back positive and it was a mutation they discovered, well, then would we tell the other female relatives? Or, at that point, is it something we’ve done for...and I’ve done just for my girls. (Mimi, PP15, field conversation, August 21, 2001)

Mimi did not tell any of her relatives that she had gone for testing because until the results came in, she felt there was nothing to tell. But she did not think it was her responsibility to tell others. (Field notes, September 15, 2001)

Ginger said,

When I found out what the results might be, then I would decide whether or not to share it with other people...but as it turned out, there wasn’t much to share. (Ginger, PP11, field conversation, July 5, 2001)

Louise interpreted her results as confirmation of her mutation carrier status and chose to share her results with her extended family.

It was a very awkward moment when I went back east for a funeral. My cousin, who had the ovarian cancer, I told her about the study and she said, ‘I’m not surprised, you know. I am not surprised with your results and the fact that it brought up that, you know, ovarian might be a genetic thing.’ (Louise, PP13, field conversation, July 18, 2001)

Louise said she told this cousin about her results, because one of the health professionals at the Cancer Agency had advised her to tell her family members who were more at risk of cancer. Louise interpreted this advice to mean those in the family who had also developed ovarian cancer.
Among the participants who interpret their results as confirmation of their mutation carrier status and ambiguity regarding their mutation carrier status, very few shared with me during the interview that they communicated their results to anyone outside their immediate family. Most felt that their responsibility was to their children and siblings only and not to their extended family such as aunts, uncles, and cousins. Victoria is one of the participants who shared her results with a cousin from the side of her family where there was no history of cancer. I interviewed this cousin and this is what she had to say about her relative’s test results: “Victoria told me that she was told that she has some mutation but that they are unable to find it at this time.” (Duchess, SP1/PP22, Victoria’s cousin, field conversation, October 16, 2001)

Duchess’ interpretation of the test results echoes Victoria’s. This shows the clinical importance of following up with all individuals who have genetic testing to find out how they interpreted their results, and how and what they will communicate to their family members. Although Victoria’s interpretation of her results was correct, she failed to share with Duchess the other three possible interpretations in the letter she received from the Cancer Agency.

Only two participants mentioned having made copies of the letter from the Cancer Agency to share with family members, while another decided to give copies to all her extended family members following her interview with me. She said that being interviewed made her realise the importance of her genetic information for her family even if they had, at one point, told her that they were not interested in hearing of her test results.

A few participants mentioned how they chose not to involve their mothers in the genetic process, fearing that they would feel even more responsible for them having been diagnosed with breast cancer, if found to carry an inherited mutation. Erika and Juniper worried that sharing
their experience of genetic testing might create more anxiety to their mothers. Erika’s mother did not have breast cancer, but her aunt did and died as a result of her disease.

I did not discuss with my mum that I was having genetic testing done. Well, I think for my mum it was very scary – the idea of what was happening to me. I didn’t tell her all those kinds of things, no, I’d just told her that I was going to be fine and everything was okay. And I guess I’ve had to just be very strong, because it was just too difficult for her. She just, she, she wanted it to be her instead of me. (Erika, PP10, field conversation July 9, 2001)

Junipers’ mother had been diagnosed with breast cancer at the age of 45. Although Juniper had been told that her mother would also be a good candidate for genetic testing, she chose not to involve her mother in it.

Well, I discussed it with my husband first, and I also discussed it with my sister. I didn’t discuss it with my mother, because we don’t talk a lot about cancer because of her fear (of a recurrence). For me having cancer, I’m sure she was very worried about me. But her way of expressing was that it just made her worried about herself again, because it just brought back all of that for her. So I chose not to involve my mother in a lot, even if I was told that she would be a good candidate to test as well. (Juniper, PP2, field conversation, June 28, 2001)

For many of the participants, going through genetic testing was like reliving the fear and worries of their cancer experience. In reliving this experience, they reviewed the support system they felt they had (or did not have) during their cancer experience. Some said, “My mother was not present for me during my cancer experience as much as I was during hers.” Another participant shared how her husband was upset that she was getting back talking about breast cancer by having genetic testing. As she said, “He wanted me to forget about it, and I told him I would not.” Hence, prior to having genetic testing, participants had to decide who was physically and/or psychologically strong enough to handle reliving their cancer experience and receiving genetic information.
Views Unique to the “Uninformative Group”

How different is this “uninformative group” from people who test positive or negative to an inherited mutation? According to my study participants’ accounts, the main difference is the uncertainty of their test results and the multiple ways in which their results can be interpreted. The multiple ways in which their results could be interpreted left most participants “in a quandary” about how to assess their risk of carrying an inherited mutation. As Joyce said like her doctor had told her, the Cancer Agency was not “sticking their neck out to say one way or another”. Participant were faced with either choosing between one of the four interpretation options in the letter or forging a new interpretation; such as believing that they have a weaker mutation that needs a specific interaction with the environment to produce cancer, having a mutation that is unique to their ancestry, or having a mutation that affects only one organ.88

The paradox of their experience is that these individuals came out of their genetic testing experience with fewer answers than before they started. As a result of the genetic testing experience, some participants perceived themselves chronically at risk; at term described by Kenen et al. as (2003b), living continually with a heightened awareness of risk for cancer and for an inherited mutation in themselves and in others: a very uncertain future. While the Kenen et al. study represents unaffected women from suspected HBOC families, the participants in this study discussed how their experience with genetic testing had not altered their increased risk perception for cancer and for future cancers for themselves and family members but had changed their perception as to a possible inherited mutation.

88 Recent discoveries indicate that a low-penetrance susceptibility gene mutation, CHEK 2, confers a modest risk of breast cancer in families without BRCA1 or BRCA2 mutations (Meijers-Heijboer et al., 2002; Wooster & Weber, 2003). The CHEK 2 variant was found in 5.1% of individuals with a family history of breast cancer without a BRCA1 or BRCA2 mutations compared to a frequency of 1.1% in the general population. The difference in frequency is believed to be attributed to a possible interaction effect between CHEK2 and other as-yet unidentified breast cancer predisposition genes in these families. Hence, the participants’ belief that they carry a weaker mutation needing a certain interaction with the environment to produce cancer may not be so implausible.
Some participants used the word *loss* to describe their experience. While for most participants, their experience may not have changed their perception about an increase risk for cancer and a possible inherited mutation, the different forms of loss represent perceived possible consequences as a result of not having been found with an inherited mutation.

- loss of “at risk” status to HBOC;
- loss of the option to participate in clinical trials reserved for untested individuals at risk of HBOC and individuals from confirmed HBOC families;
- loss of opportunity for family members to be tested;
- loss of opportunity for others in the family to recognise themselves at increased risk of HBOC;
- loss of screening privileges reserved for HBOC families;
- loss of risk-reducing surgical options, such as prophylactic mastectomy and oophorectomy usually recommended for HBOC families; and finally
- loss of access to the most up-to-date preventative procedures to high-risk breast and ovarian cancer clinic.

In the end, many participants were left struggling with figuring out their true risk of cancer and their genetic status and that of their families. According to the 2003 recommendations of the Canadian Coordinating Office for Health Technology Assessment (Ho, Banerjee, & Mensinkai, 2003) on the clinical impact of genetic testing, individuals with a strong family history of cancer in whom no mutation is found following genetic testing of the BRCA1 and 2 genes remain at increased risk of cancer and should continue cancer screening as before testing. Hence, the Cancer Agency, which only follows individuals from confirmed HBOC families, could be underestimating their cancer risk. The multiple possible interpretations about
uninformative genetic test results in the letter from the Cancer Agency left many participants with a difficult dilemma: No matter which interpretation they chose, they and their family were still at risk of carrying an inherited mutation of some kind. This dilemma reflects the majority’s position (17 participants out of 21) of interpreting their results as either confirming their mutation carrier status or ambiguity regarding their mutation carrier status. As for the other four participants, because they interpreted their results as refutation of their mutation carrier status and chose to accept the beginning explanation that no mutation had been identified in their BRCA1 or BRCA2 cancer genes, they did not go on to consider any of the four interpretations possible to this negative results offered by their Testing Cancer Agency.

**Conclusion**

In this chapter, I looked at how participants’ interpretations of their genetic testing results and the way they made sense of them affected their everyday lives, health and illness experiences, thereby addressing my second research question: How did the participants’ results affect their everyday lives, health and illness experiences? This inquiry revealed that very few study participants found relief with their results. They entered genetic testing hoping to find some certainty about their genetic risk and the etiology of their cancer but, in the end, got no answers from their results. However, participants who interpreted their results as refutation of their mutation carrier status were able to find some temporary relief as to their genetic risk but not in relation to their increase cancer risk. That is, some felt their children were now at a lower risk of an inherited mutation, since, for the time being, current technology could not identify one. Participants who interpreted their results as ambiguity regarding their mutation carrier status also felt a temporary relief associated with not being found with an inherited mutation for the time
being. However, for all, relief felt was only partial because they still do not know why they
developed breast and/or ovarian cancer in the first place.

Further, those who interpreted their results as ambiguity regarding mutation carrier status
and confirmation of mutation carrier status found some relief in the thought that perhaps their
specific mutation is not one linked to the BRCA1 or BRCA2 genes but to some other as-yet-
unidentified cancer gene of lesser consequences.

Although a few participants did openly share how they would have preferred positive,
certain, test results, almost all definitely felt that continued uncertainty is better than knowing
with certainty that a family carries an inherited mutation. The irony in this view is that many
participants said they needed to gain control over their disease. Now, they had to learn to live
with uncertainty. Along with “the wait and see” who in the family will next develop cancer, their
situation was now also “wait and see what the next set of tests will tell us,” as one participant so
clearly said, representing the temporal relief experience with receiving uninformative results.

While their genetic test results provided information of little value, very few participants
had expected genetic testing to help them make health-related decisions, such as whether or not
to have prophylactic surgery. Many of the participants had already made that decision either as
part of their cancer treatment by virtue of their own cancer diagnosis and their family history
with the disease. Instead, many participants said they had genetic testing in order to provide
information to family members, to help them make cancer-risk-management decisions.

Seeing few benefits for themselves from their genetic testing experiences, some
participants felt disappointment. Not only did they not know anymore about their genetic risk
status, they and their family felt more at risk for discrimination related to life and extended
health insurance and losing privileged access to cancer screening. Further, a few of the women
felt more at risk of developing another type of cancer (either breast or ovarian) because of learning that a mutation in the BRCA1 and BRCA2 genes can cause both. An additional disappointment was realising that some family members lacked interest in their genetic testing results, after most of the participants had undergone genetic testing for others first, and then for themselves. A potential family disruption was identified; even when family members told them they were not interested in learning of the participants’ results, some participants went ahead and told them anyway. These participants justified their actions by saying that, had the test results been positive, they would not have told family members. Others, who viewed their results as uncertain, either chose to share or not to share their information with family members. While communicating results was not a main focus of this study, participants who chose to share results with extended family members (aunts, uncles, and cousins) in comparison to those who did not, had said in the course of their interviews that they had close emotional and/or physical ties with them. Further, they had identified extended family members as further motivation to have genetic testing so that they could learn of their possible increase risk for an inherited mutation. Participants who chose not to share their results said they had decided to have genetic testing to inform themselves first and their immediate family member such as children and siblings.

The results presented in this chapter, along with the findings in the previous chapters, illustrate that the greatest impact of the participants’ experience of their genetic testing and their results interpretation takes place within the context of their everyday health and illness experiences. In chapter 7, I locate the study findings within a broader nursing philosophy: the individual, who in interaction with her/his environment, lives health and illness experiences (Fortin, 1996). This philosophy can guide nurses’ understanding of how individuals, in the face of health and illness uncertainties, turn to their everyday experiences to make sense of, and deal
with their reality. The analysis in chapter 7 paves the way for recommendations to assist the clinic in providing more comprehensive services to those who receive genetic testing.
CHAPTER SEVEN:
Situating the Study Findings in the Broader Clinical Context

Much past research about the impact of genetic testing focussed on at-risk women with no personal cancer history and was done using structured questionnaires that allow little room for fully comprehending individual’s results interpretation and making-sense experiences. The interpretive description approach I used, however, permitted this. Using a dialectical approach created space to clarify the meanings people brought to their results and allowed in-depth understanding of how affected women with a family history of risk for hereditary breast and/or ovarian cancer (HBOC) interpreted their genetic test results. My research design also facilitated the analysis of how participants made sense of their genetic testing results, illuminating the generic structures that organised their results interpretation experience.

The overall goal of this study was to understand how affected women considered at risk for HBOC interpret and make sense of uninformative genetic test results for BRCA1 and BRCA2 mutations. I began my study by examining the context in which the participants’ responses to testing were embedded. Thus, I focussed on the participants’ perception of receiving a cancer diagnosis and living with a family history of breast and/or ovarian cancer. From there, I turned my attention to how genetic testing entered their lives and the reasons why they underwent testing. Then I explored how they interpreted their test results and made sense of the results. I looked at how individuals integrated their interpretations and making-sense experiences into their everyday lives, and health and illness experiences. This chapter is divided according to the themes outlined above – living with uncertainty as to their increased risk for developing further cancers and to their genetic risks, the generic structures derived from their making sense experience of interpreting uninformative genetic testing results to BRCA1 and BRCA2 followed
by a discussion of what comes next for participants who have received such results. The chapter ends with a presentation of the study’s findings limitations.

**Living with Uncertainty**

Becoming comfortable with the word *cancer* when living with a family history of it has been theorised to mimic coping strategies seen in individuals suffering from chronic illnesses (Kenen et al., 2003b). Kenen, Ardern-Jones, and Eeles (2003b) describe this concept as “living with a chronic risk perspective.” The participants of this study are not only learning to become comfortable with the word *cancer* as a result of living with a family history of it, they also face living with the uncertainty about whether they carry an inherited mutation. Although theories of uncertainty in illness have been created (Mishel, 1988, 1990), authors have only created them within the context of chronic illness, not chronic risk. Therefore, much remains to be discovered about the impact of living with chronic, unconfirmed inherited risk for a disease.

Most genetic test results are essentially uncertain as, they convey probabilities that estimate level of risk (van Zuuren et al., 1997). Many of the affected women who participated in this study live with chronic risk. The question is, “How does anxiety from knowing one’s genetic risk compare with having a family history of cancer but not knowing one’s genetic risk? While being given a percentage for one’s increased risk for cancer and for a genetic risk can be both unsettling and reassuring, so can living with uncertain risk. However, among Hallowell et al. (2004) study participants, although there was no difference between the anxiety levels of individuals who tested positive and those whose results were inconclusive for BRCA1 and BRCA2 mutations, their rates were significantly higher than the individuals who received negative results. Concurrently, as empirically shown by Mishel (1984), uncertainty increases
stress. Thus, individuals who interpret their genetic test results as uncertain experience the same amount of increased stress related to genetic testing results as those who receive positive results.

Further, my findings suggest that, not only must most participants learn to live with multiple forms of uncertainty regarding different risks; they must also come to accept that their test results brought them little relief. Instead of giving them the certainty they longed for, genetic testing gave them more uncertainty, creating further stress. As van Zuuren et al. (1997) found within their study, unpredictability and ambiguity in genetic testing results adds to stress and further creates a negative experience overall. Just as the authors van Zuuren et al. concluded, the current study findings show that there is a poor match between clients’ needs and purposes in genetic testing and the often-uncertain test outcomes that individuals receive.

As participants in my study explained, hearing of another cancer diagnosis in the family was almost expected, but never welcome. They lived each new case of cancer with much emotional struggle because it forced them to review their family’s increase risk for cancer and for a probable inherited mutation predisposing them to cancer. Participants with a family history of cancer that spanned many generations were less upset by each new cancer diagnosis than participants whose family history of cancer had occurred close together. Study findings indicate that participants’ adaptation builds over time, over generations. That is, the emotions expressed by participants whose family history of cancer stretches across many generations suggest greater acceptance of the increase risk for cancer and of the possible inherited risk for cancer, while participants whose family history of cancer is more recent have stronger reaction to each new diagnosis. Knowing the family history of cancer may have the benefit of emotional readiness for subsequent diagnosis. Additional benefits also arise from knowledge of family history. For example, telling one’s family physician that one has such a history may lead to more intense
follow-up when a suspicious mass is detected by clinical exam or mammography, opportunities to participate in early cancer detection programs, and psychological support when needed. While there may also be some drawbacks from more intense screening, data still show that breast and ovarian cancer morbidity and mortality declines the earlier cancer is found (Thulesius, Lindgren, Olsson, & Hakansson, 2004).

Research indicates that families with hereditary cancer and at risk of HBOC participate more often in screening activities (Mahon & Casperson, 1995). Findings from this study point in the same direction – that the participants’ use of screening and testing procedures was above that recommended for the general population. Of the 14 participants who still had their breasts, 12 said they have mammography every year, while 2 said they have it every 6 months. Of the 21 participants, 14 said they have clinical breast examination every 6 months, while 7 have it done every year. As for the general population, the breast screening guidelines of the Canadian Cancer Society recommend that women between the ages of 50 and 69 receive mammography every 2 years and clinical breast examination, at least every 2 years (Canadian Cancer Society, 2003). While this may be true for the general populations, for women at increased risk, Smith et al. (2003) of the American Cancer Society recommend initiating mammography screening as early as at the age of 30 with shorter mammography screening intervals such as every six months. Despite being told that no mutations had been found in their BRCA1 and BRCA2 cancer genes, the studied population continued to receive the same cancer screening as before testing. Their screening practices reflect the recommendations made by their Cancer Genetic Testing Agency (to continue screening as previously recommended) and by the American Cancer Society for women at increased risk for breast cancer such as those with a strong family history of cancer. As such, participants of this study understood that even if no mutation to an inherited
susceptibility to breast and ovarian cancer had been found, their negative results did not erase their family history with breast and/or ovarian cancer.

While the participants' cancer screening may reflect current recommendations for women at increased risk, there is a need for clinics to document uptake of cancer screening and testing in individuals still suspected of carrying an inherited mutation. Reasons for documenting cancer screening and testing relates to the probabilistic, yet still undefined, findings that indicate an increase sensitivity to radiation in individuals carrying an inherited susceptibility to cancer (Fasouliotis & Schenker, 2000). Such a conclusion, however, may be premature as this topic is found to be under-researched (Pierce et al., 2000).

The high frequency use of cancer screening reported by the participants of this study contrasts with other studies documenting that increased awareness of risk for cancer usually reduces adherence to regular screening (Kash et al., 1992; Lerman et al., 1990). Such difference in findings might reflect that, in both the Kash et al. study and the Lerman et al. study, the women surveyed, while at high risk of developing breast cancer, had not been affected by the disease. As the participants of my study had said, they had become comfortable with the word cancer and had come to expect cancer from having lived with a strong family history to cancer. They also explained that, as a result of having experience cancer, they had learned to be comfortable with their continuous risk for cancer. Hence, anxiety about screening for cancer would seem to apply more strongly to unaffected women at increased risk for developing breast cancer and would therefore explain the difference in the findings of this study where participants were actively engaged in cancer screening. Concurrently, a recent study by Hallowell et al. (2004) suggests that accommodation to genetic risk is also influenced by the degree to which individuals have internalised their increased risk for cancer in their biography following their
diagnosis and treatment of cancer. Thus, the Hallowell et al. study and the current findings may suggest that acceptance of increased risk for cancer reduces anxiety which positively affects adherence to cancer screening and uptake of genetic testing to breast and ovarian cancer susceptibility.

One more concern raised by some authors is that individuals from families at risk of HBOC who now believe they do not have an inherited mutation following the receipt of negative genetic test results may be falsely reassured and may relax their screening practices (Carter & Hailey, 1999). There is a need for high-risk individuals to understand the difference between “no mutations detected” and negative genetic test results within families with an identified mutation (Claes et al., 2004; Iglehart et al., 1998; Wong et al., 2001). Although the Cancer Agency told the study participants to continue with screening as previously recommended for high risk individuals, one participant actually concluded that her breast cancer risk was the same as that of the general population. This participant is one of the four who interpreted her results as refutation of her mutation carrier status. While the other three participants also concluded that they were not carrying an inherited mutation predisposing to an increased risk for hereditary cancers, they still perceived their risk for cancer and that of their family to be above the general population’s risk because of their family history with cancer.

Use of the Generic Structures as Guides to Clinical Genetic Testing Practice

Although the unique group of individuals I studied has been recently documented within empirical studies (Bish et al., 2002a; Frost et al., 2004; Hallowell et al., 2002; Hallowell et al., 2004; Kelly et al., 2004; Lynch et al., 1999; van Zuuren et al., 1997; Wong et al., 2001) and within clinical reviews (Peshkin et al., 2001), there are still no studies that have documented the underlying structures organising individuals’ interpretation of uninformative genetic test results.
Additionally, although I found a study that assessed the individuals’ perceived level of risk of breast and/or ovarian cancer gene mutation (Struewing, Lerman, Kase, Giambalresi, & Tucker, 1995), still absent from the scientific literature is a study of what brings individuals to perceive themselves at high risk, or not, of carrying an inherited mutation when interpreting their genetic test results. For individuals who receive uninformative genetic testing results, we know even less about how they make sense of such a result and how they arrive at their conclusion.

In order to provide effective, evidence-based support to cancer genetics clients, health professionals must comprehend how individuals make sense of uninformative results. When I asked about their results in the interviews, participants would first repeat what their letter had said that no mutations had been found in their BRCA1 and BRCA2 genes but would then add their own reinterpretation. That is, they would add that they themselves interpreted their results as either: But we have an inherited mutation; or Maybe we do or maybe we do not have an inherited mutation; or And we do not have the inherited type of cancer. Some participants stated that, although they understood that no mutation had been found in their BRCA1 and BRCA2 genes, they still believed that they carried an inherited mutation. Participants described how simply adopting the view that they do not carry an inherited mutation was impossible for them in light of the number of breast and/or ovarian cancer cases in their family history and the number of deaths from these cancers. At times, some commented as well that they could not believe their testing results that showed that no mutations had been found in their BRCA1 and BRCA2 cancer genes because the facts showed that, in their families, cancer seems to be appearing sooner with each generation.

From their reinterpretation of their results, thirteen generic structures were derived from their making-sense experience that sheds light on their belief of still carrying or not an inherited
mutation predisposing them and their family to an increased risk of breast and/or ovarian cancer. These generic structures are not seen as encompassing all possible structures likely to inform all individuals who interpret and make sense of uninformative genetic testing results, but are seen as baseline data which clinicians can use to assess how their clients arrive at making-sense of their results. For example, clinicians could elicit client’s view on the meaning of their family’s cancer history using the generic structures as prompts. That is, clinicians could ask their clients’ perception in relation to the meaning that all cancers in their family have only occurred through females or males only. Such engagement with clients prior to genetic testing and to receiving their results might facilitate their making-sense process especially in the context of receiving uninformative results. As well, these structures can also be used following the receipt of uninformative results where individuals seek further professional support to help them understand the meaning of their results. Use of the generic structures to assist clients in making sense of their results could either be done by telephone or face-to-face depending on the protocol used by the Testing Agency. At the time of the study, participants were first informed of their uninformative results by a telephone call made by a genetic counsellor. This call was later followed by a letter further explaining the meaning of their results. While some other Testing Agencies’ protocols for delivering genetic testing results may differ in that all types of results (positive, true-negative and, uninformative) are provided through face-to-face counselling, all clients who receive uninformative results could be assisted in making sense of them through the use of the generic structures derived in this study.

This study’s purpose was not to attribute outcomes to the nature of the presentation of results, as the design was retrospective. Rather, its larger intent was to acknowledge the full context in which the participants’ experience of genetic testing took place and the way in which
this entire context has influenced their interpretation and making sense process related to their uninformative results. Toward this end, one such context within their testing experience, apart from the manner of presentation of results, includes the time lapse between testing and receiving results. While the current study design was not conceived to explore the impact of time on participants’ genetic risk perception, my data suggest that the length of time between giving a blood sample and receiving test results affected participants’ interpretation of their results. The average wait of 18 months was interpreted by many participants to mean that their results must be negative; however, upon receiving the possible meaning of their results given to them in the letter written by their testing Cancer Agency, many participants then entertained the possibility that they could or could not have an inherited mutation, resulting in the majority interpreting their results as ambiguity regarding their mutation carrier status. Further, once the participants were advised that no mutation had been found in their BRCA1 gene a year after having their blood drawn and told that it would most likely take another year to analyse their BRCA2 gene, some participants said that they did not hold much hope that a mutation would be found in the second cancer gene. The long wait was also interpreted as a lack of available funding to conduct their analysis in a timely manner. These interpretations could be avoided by addressing these issues in genetic counselling and follow-up sessions. In particular, as genetic testing moves from the research setting to becoming a publicly-funded clinical service, waiting time will need to be addressed (Di Prospero et al., 2001). While reasons for the waiting time to receive genetic testing results were addressed with participants during their counselling, few attributed this wait to under-staffing of professionals to analyse their DNA. As participants of this study rather attributed the long wait between testing and obtaining results as a reflection of their likelihood of being found with an inherited mutation, genetic professionals within their counselling sessions
might consider explaining that waiting time has no bearing to their likelihood of being found with an inherited mutation.

Comparison was analysed between participants who interpreted their results as confirmation of their mutation carrier status versus participants who interpreted their results as ambiguity and refutation of mutation carrier status. The comparison led to another observation where time was observed as influencing participants’ belief about their genetic status. That is, when cancer diagnoses from two generations occurred within a short time of each other (fewer than 10 years difference), participants seemed less likely to believe that the cancers could be the result of an inherited mutation and more so from a recent common exposure to a carcinogenic compound. Conversely, the longer the time between cancer diagnoses from two generations, the more likely participants seemed to suspect an inherited mutation – especially when cancer appeared at similar ages within each generation. As for cancer diagnoses that appeared closely together within the same generation, participants were more likely to interpret this occurring as probably being the result of an inherited mutation.

Another particularity noted between the interpreted structures and the genetic literature occurred where participants believed that resemblance to an affected family member put them at greater risk for having inherited the same cancer susceptibility. While the literature suggest that women from families with ovarian cancer may not see themselves at risk if they perceive that they “take after their father’s side of the family” (Richards & Ponder, 1996), this possibility could not be properly assessed with this study’s data because of the limited number of individuals who had a family history of only ovarian cancer. However, what we have learned further within this study is that, for individuals whose family history with cancer was with either breast or ovarian cancer only, participants interpreted this family history as a fact that perhaps
their specific mutation increasing their susceptibility to this one disease may be a mutation unrelated to those associated with BRCA1 and BRCA2 cancer genes. Participants believed that individuals carrying mutations in either gene would be equally at risk of developing both breast and ovarian cancers.

While it could be possible that some or all of the individuals in this study who received uninformative results may actually not carry an inherited mutation predisposing them and their family to cancer, the possibility that their cancer diagnosis might be a sporadic case within a hereditary cancer family should be considered (Peshkin et al., 2001). Current genetic testing practices corroborates the recommendation by Peshkin et al. (2001) that testing another affected member is warranted in instances where the family history indicates a probable inherited mutation (Peshkin et al., 2001). These authors recommend that clinics carefully choose the affected individual. It should be the person who is suspected to have the highest likelihood of an inherited mutation. Although how best to identify such an individual is still a subject of debate within the scientific community, some helpful models are available and have provided statistically significant results. One example is the BRCAPRO model, based on the work of Parmigiani et al. (1998). However, Peshkin et al. recommend that such models be used in combination with traditional pedigree-based, qualitative assessment by trained genetic health professionals. Using both approaches would facilitate identifying possible individual suspected of carrying an inherited mutation within families considered at risk for HBOC. Some participants in this study had commented themselves on how they had felt that perhaps they were not the best candidate to be tested to assess for the likelihood of carrying an inherited mutation since their cancer diagnosis had occurred after menopause.
The Now What Experience of Genetic Testing?

Participants who viewed the final outcome of their results as uncertain felt little relief, but experienced much frustration with their test results. Among the few who expressed some relief, one wonders if they could have gained it not only from their genetic test results but from genetic counselling. While Hutson (2003) reports that women with a family history of breast and/or ovarian cancer have underlying psychological distress that is not usually relieved by genetic counselling, other studies show that genetic counselling and testing can produce psychological benefits and improve the accuracy of risk perception (Butow, Lobb, Meiser, Barratt, & Tucker, 2003; Esplen et al., 2000; Frost et al., 2004).

Participants in this study who interpreted their test results as ambiguity regarding mutation carrier status and refutation of mutation carrier status nonetheless felt that “for the time being, we are off the hook.” Contrary to what clinicians might assume: that those who receive uninformative/inconclusive genetic test results experience relief similar to those who test negative (Bish et al., 2002a), my data revealed that, even among those who felt relief, the relief is only temporary. Participants felt their relief was temporary because they understood that the field of genetics is evolving and that the likelihood of finding further genetic mutations conferring a risk for breast and ovarian cancer is high and that they could, therefore, later be found to carry an inherited susceptibility to these diseases.

Many of the emotional reactions to the test results documented in this study relate to uncertainty and lack of control over one’s fate. While uncertainty has been hypothesised to lead to increased stress (Mishel, 1988), a prospective cohort study by Lerman and al. (1998) also put forth the finding that uncertainty about genetic risk among individuals who decline testing leads to increased depression rates in comparison to those who underwent testing. Lerman and al.
study showed that, among the 327 individuals from HBOC surveyed for depressive symptoms at baseline and at 1- and 6- months, depression rates in decliners increased from 26% to 47% at 1-month follow-up while depression rates in non-carriers decreased and in carriers showed no change. Thus, while these authors concluded that individuals who decline testing should benefit from education and counselling and be monitored for potential adverse effects, the current study findings also points to the need for continued monitoring among individuals who receive uninformative results. Both groups (from Lerman’s study and the current study) are left with uncertainty to the true nature of their genetic status. As attested by Lerman’ study findings, among the individuals from hereditary breast and ovarian cancer families who were found not to carry the known familial mutation, their depression rates decreased following receipt of results. However, a small scale study by Lodder et al. (2003) who surveyed 13 women at risk for HBOC who had decline testing, showed no significant differences between this group and the tested group on the scale assessing depression levels. Thus, many questions remain unanswered as to the possible link between living with the uncertainty about one’s genetic risk status and depression risk. Further studies are needed that can assess this link and document the similarities and differences among those who decline testing and those who receive uninformative results. Such findings would further support the need for follow-up counselling for these two different groups of individuals.

Findings from this study indicate three ways in which genetic testing entered people’s lives: (a) by self-referral to a cancer agency, (b) through a family member’s request, and (c) following a recommendation by a health care provider. For those who self-referred, these data indicate that this referral was preceded by a new case of cancer – either in themselves, from a close relative or from a friend. Knowing that increased awareness of cancer risk contributes to
distress (Baum et al., 1997; Gil et al., 2004), and that those individuals who seek genetic testing are usually experiencing a significant amount of stress at the time of the request (Loader et al., 2004), one might hypothesise that individuals seeking genetic testing may be under stress and consider whether the clinic should assess the saliency of their cancer and genetic risks at that time. From their research on the impact of genetic counselling and testing with 87 families at risk for HBOC, Loader et al. concluded that the greatest stress was experienced by those who requested genetic testing – the “probands” – but they were told that they had to ask an affected relative to have the test on their behalf, since they themselves were ineligible. Ineligibility for testing was experienced by one family member, Stacey, whom I had the opportunity to interview and whose story corroborates the above conclusions. Stacey’s sister had recently been diagnosed with breast cancer and was given a poor prognosis, while her father had been diagnosed with suspicious cysts in the breast and was advised to have a bilateral prophylactic mastectomy.

Out of the three generations, there are 13 women that have had breast cancer in our family. My sister was the last one. So, once my sister developed breast cancer at the age of 46, it got quite personal then. Just watching her go through what she was going through and thinking of her children – she has three daughters – it just made me want to do as much as I can to find out what’s going on with our family, right? (Stacey, SP1/PP3, Joyce’s cousin, field conversation, April 6, 2001)

It could also be argued that probands could experience further distress if they are never informed of the results of their relatives’ tests. For example, Stacey said that, although her cousin told her that no mutation had been found, she had not given Stacey a copy of the letter from the Cancer Agency that presented the four different ways in which the genetic test results can be interpreted for individuals considered at risk for HBOC who are not found with a mutation. Although all participants had been encouraged to share a copy of their letter with interested family members, very few did. How participants mostly shared their results was either in person or by telephone. Guidelines to sharing genetic information with family members may require
further assessment, as family dynamics can be disrupted by such news. For example, it is
difficult to know how to respect some family members’ preference for not knowing the genetic
testing results when informing others will automatically reveal information about them. As well,
how do we assist the individual who had genetic testing to make the decision on who should be
informed of these results? A qualitative study by Kenen et al. (2003c) looking at communication
patterns in families with a history of breast and/or ovarian cancer in interviews of twenty-one
unaffected women who attended a cancer genetic risk clinic reports that perceptions of family
norms can dictate communication patterns. Some of the women in the Kenen et al. study used
self-censorship when sensing that someone did not want to talk about the cancer in the family or
when they feared that talking about cancer to a specific person may cause anxiety. Through their
study, Kenen et al. recognised that family dynamics can be destabilised and disrupted when the
family context meets with the genetic counselling context.

Patterns of communication of genetic information can also lead to ethical complications
such as: (a) does someone have the right to ask someone else to undergo BRCA testing
(Surbone, 2001), (b) is the proband entitled to receive the test result of the individual in the
family who had testing on their request, and (c) who is better placed to make the request, the
relative or the health care professional? These complications require further discussion as well as
assessment of the potential harm they might create. Perhaps, as suggested by Burgess and
d’Agincourt-Canning (2001), today’s debate about rights in the context of genetics should focus
not only on individuals’ rights but on relational responsibility.

As Surbone (2001) reminds us, with BRCA testing, all health professionals working in
the area of genetics are confronted with situations in which the balance among autonomy,
beneficence, and justice is not easily reached. To this end, participants in my study have
confirmed important issues that are continuously being raised in the genetics literature: (a) communicating genetic testing results may create more fear of cancer in the family, (b) uncertainty about responsibility to communicate result (c) potential for harm when family members’ choice of not wanting to hear results is not respected, (d) risk of discrimination in life and extended health insurance (mostly for unaffected family members), and (e) fear of losing privileged access to screening, new procedures, and clinical trials if identified at lesser risk for cancer than individuals from known HBOC families.

Previous studies suggest that worry about discrimination can be a barrier to genetic testing (Ball, Ondrusek, & al., 1998; Knoppers et al., 2004; Lerman & al., 1996; Lerman et al., 1996; Lynch et al., 1999). Although the current participants were informed of the potential risk of health and life discrimination as part of their genetic counselling, only one participant expressed fear of discrimination in this area and all proceeded to their testing. As they have explained many times, because they were already living with a past diagnosis of cancer, whatever they could not get from extended health insurance or life insurance as a result of having a past diagnosis of cancer would not change anything because of having in their file a suspected genetic risk for cancer. While in the United States, much of the debate with insurance discrimination has centered on health insurance, in Canada because of our universal health care system, the focus of discussion has been on access to life insurance as a basic socioeconomic good (Knoppers & Joly, 2004). The concern of risk for life insurance discrimination prompted patient advocates, geneticists and researchers involved in the Genetics and Society Project at the Université de Montréal to create the Canadian Genetics and Life Insurance Task Force to debate the subject of life insurance and genetics in Canada (Knoppers & Joly, 2004). While this task force is still just proposing recommendations, there is currently no specific legislation relating to
the use of genetic information in Canada (Burgess et al., 1998). While critics warn that fear of discrimination may hinder people from seeking genetic testing (Ball et al., 1998; Knoppers et al., 2004; Lerman & al., 1996; Lerman et al., 1996; Lynch et al., 1999), Knoppers et al. (2004) hope that eventually legislation will be in place that will facilitate Canadians to avail themselves of the health benefits of the genetic revolution without fear of discrimination.

Many participants said that their genetic testing experience was nothing in comparison to their cancer experience; many unaffected family members interviewed shared the same view explaining that, in comparison to witnessing their family members’ experience with cancer, knowing that a family member had gone for genetic testing was not majorly stressful. Taylor, a family member interviewed, explains the difference:

Well, the cancer had a huge impact on me; the testing not as much. I mean the cancer when she was sick, yeah, that had a very huge impact. It was very difficult. It was difficult to be here, to be so far away, not to be with her and wondering if she needed me there. I was there at one point when she was going for chemotherapy and going to the hospital with her – which I felt good about, because just to show support and when you’re so far away you feel kind of removed a little bit. But as far as the testing, it didn’t affect me as much. I think because I didn’t really understand exactly maybe what was really involved. So it didn’t have as much of an impact on me. (Taylor, SP1/PP10, Erika’s sister, field conversation, February 2, 2001)

Many participants said they were surprised that their family members were not interested in their genetic test results. One possible reason for the lack of interest by family members could result from not having attended the genetic counselling sessions and perhaps not foreseeing that a family member had undergone genetic testing might have implications for them. The testing itself could be, in the family member’s view, a simple blood test that involves little physical, or emotional pain, in comparison with witnessing family members’ cancers (Hallowell et al., 2004).

The current findings suggest that individuals who interpreted their results as being uncertain experienced frustration and stress. Others experienced stress following their genetic
counselling session. They told me that it was only during their counselling session that they became aware that having an inherited mutation in the BRCA1 or BRCA2 genes would put them at risk of both breast and ovarian cancer. This new-found awareness made them anxious about ovarian cancer. However, I do not mean to imply that there is a direct link between receiving genetic counselling for BRCA1 and BRCA2 and an increased perception of risk for ovarian cancer. Bish et al. (2002a) also found that individuals with past breast cancer diagnoses showed a higher level of concern about ovarian cancer, compared with unaffected individuals, prior to and after genetic counselling. Bish et al.'s findings (and my findings) contrast with the Kelly et al. (2004) and Hallowell et al. (2004) studies. Hallowell et al. found that former cancer patients did not necessarily experience more anxiety about their genetic risk of developing ovarian cancers following genetic testing – both for people who tested positive and inconclusive, while Kelly et al. found that perceived risk of ovarian cancer actually dropped between pre- and postcounselling.

I hypothesised that, if the participants expected that testing would provide them with a result that either confirmed or refuted their suspicion that they carry an inherited mutation, they might gain a benefit from the testing. Many participants did benefit from testing, in that they felt more control over their “chronic disease” by the information obtained from their testing. However, not all participants felt the results brought them greater control over their disease. Although very few participants spoke of having genetic testing in order to control their future risk for cancer, some did use their interpretation of their results to justify past medical decisions made to reduce their risk of breast and ovarian cancer. Others could find no comfort in their results because the results did not justify past decisions relating to risk-reducing surgical procedures based on an actual genetic risk. What this seems to imply is that when individuals
request genetic testing, their reasons for having testing should be explored, as they may incorporate unrealistic expectations of genetic testing especially if test results turn out to be uninformative. In this case, individuals and their family members will have to make medical management decision within a context of uncertainty (Frost et al., 2004).

I would like to compare similarities of trajectories between chronic risk perspective and chronic illness described by Kenen et al. (2003b) with others found with the retrospective narrative of causal reasoning to a probable inherited mutation proposed in this study. Kenen et al. describe how both the chronic risk and chronic illness trajectories have three phases: stable, downward, and comeback. These phases temporarily raise or lower perception of risk. Within my study, I found the retrospective narrative process to be similarly fluid. The narratives vary with how the individual derives meanings from their family history of cancer, their lived experiences with cancer, and their experience with genetic testing. Accordingly, affected individuals considered at risk for HBOC who have gone through the experience of genetic testing and were not found with a mutation could be seen as living in a state of chronic risk – chronic risk in relation to recurrence of cancer and for a probable inherited mutation to breast and/or ovarian cancer. When they are diagnosed with cancer, they leave the chronic risk trajectory and enter the chronic illness trajectory (Kenen et al., 2003b). If they are also found to carry an inherited mutation, they leave the retrospective narrative related to a probable inherited mutation to breast and ovarian cancer and enter into a state of certainty about their genetic risk for cancer that increases their susceptibility to breast and ovarian cancer. Conversely, unaffected family members of this affected individual who has undergone genetic testing and was not found with a mutation could be seen as living in a state of double chronic risk – chronic risk associated
with an inherited mutation and an increased risk for breast and/or ovarian cancer as a result of having a strong family history with these diseases.

**Limitations of Study Findings**

One major limitation of the current study is that data collection was done retrospectively of the participants’ experience with genetic testing. A further limitation associated with this being a retrospective study is that while the participants’ experience with their genetic counselling process had many implications to how they interpreted and made sense of their results, it is not possible to attribute outcomes to the manner of how the possibility of uninformative genetic testing results was presented to them. Recognising these limitations, although this study was not meant to assess change in stress levels specific to gene status between pre- and postcounselling, the findings point out that participants’ belief about carrying an inherited mutation in their BRCA1 or BRCA2 genes did not diminish. In some cases, participants believed that they now carry an increased risk for an inherited mutation as a result of receiving uninformative/inconclusive genetic test results. While the literature indicates that stress levels increase in individuals found to carry an inherited mutation, I cannot draw conclusions about any such change in stress level before and after their pre- and postcounselling with receiving uninformative test results. Nor do my findings compare to the Kelly et al. (2004) empirical study, which concluded that stress specific to gene status declined following genetic testing counselling.

Some of my findings have to be interpreted with caution, because there are very few other studies with which to compare them. There is still relatively little other research that focuses on exploring the experience of affected individuals considered at risk for HBOC that have received uninformative genetic testing results (Bish et al., 2002a; Claes et al., 2004; Frost et
al., 2004; Hallowell et al., 2004; Iglehart et al., 1998). Studies of people with the same characteristics as my sample employed research questions different from this study. Recognising that most individuals with a strong family history of breast and/or ovarian cancer considered at risk for HBOC who, in the future, decide to have genetic testing for BRCA1 and BRCA2 will receive uninformative results, I do hope that future studies of this population will bring further understanding.

The data for this study were gathered from individuals who were all clients of a single testing agency. While this allowed for consistency in the way test results were given to clients and interpreted by the clinic, it also limited the ways in which clients could interpret uninformative test results. Clients of other testing agencies might be given different possible interpretations and might come up with more of their own.

All the participants were residents of one province and all resided near the BC Cancer Agency in Vancouver. Individual differences might be more pronounced if the study had included rural individuals and those of different ethnic backgrounds, socioeconomic status and education level. Although the educational level in this study varied between the studied participants (see table 1), there were still only 21 individuals and thus must be considered a small sample.

While 15 family members were included in the study, the data gathered from them did not yield answers to the research questions. During data analysis it became apparent that, not having gone through the experience of cancer themselves, not having struggled to make sense of genetic test results, and not having received a copy of the letter from the Cancer Agency, family

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89 Recall that from January 1998 to June 2004, approximately 4,000 individuals were seen at the HCP for genetic counselling for HBOC. As similar to other genetic testing agencies, their detection rate for high risk individuals stand at 21% for new cases. The remaining 79% are classified as uninformative; hence representing a significant size of individuals with uninformative results.
members' accounts did not yield much knowledge about the research foci. The differences I found between affected primary participants and unaffected family members interviewed (secondary participants) suggest that these two populations are quite different. Further exploration of these differences is needed to establish clinical guidelines within genetic testing clinics. Although I integrated some of the family members accounts into this study whenever they provided insight into the participants' experiences, the foci of this study did not address interactions between tested individuals and their significant others,\(^9\) nor the impact of uninformative testing results on unaffected family members. Given this limitation, it is important to read what I say about family members with caution.

Finally, I would like to share a methodological coincidence that occurred during data collection that allowed me to better understand how participants interpreted and made sense of their genetic test results. When I began contacting individuals by telephone to schedule interviews, the first few told me they were not sure if they knew enough about genetics to be considered for an interview. Hence, in the hope that it would be easier for them to start talking about a topic they felt they knew more about, I began each interview by asking participants to start with their personal and familial experience with cancer. Had I not done this, I would not have been able to interpret the many structures found to inform their making sense experience for the interpretations of their test results. That is, all the participants had experienced cancer. When confronted with test results that said no gene mutation had been found, they did not accept these results at face value, but instead drew on their personal and familial experience with

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\(^9\) A recent qualitative study as been conducted by Kenen, Arden-Jones, and Eeles (2004) investigating the communication and interactions between twenty-one healthy women from families at risk of HBOC with their significant others. While their findings provide a more in-depth analysis of the communication and interactions issues between these two populations than the current study, they also underscore the importance of the need for further research in this area that specifically focuses on communication and interaction aspects. Such a focus would also be greatly needed for individuals who receive uninformative genetic testing results in light of the amount of inherent uncertainty in this type of result.
cancer, as well as social discourse about cancer and genetics, in order to make sense of their results.

A final caution when interpreting results is that, while this study provides new understanding of how individuals considered at risk for HBOC interpret and make sense of genetic testing results that no gene mutation had been found, it nonetheless represents people’s experiences at one point in time. As explained in chapters 4 to 6, their interpretation and making-sense experience occurred within a temporal process that continuously evolved in parallel with their ongoing life experiences. Further, individual and family members’ responses to genetic testing may change as a result of new genetic knowledge and its medical implications. The retrospective narrative of causal reasoning of self with a probable inherited mutation may also be affected by political decisions that make genetic testing more accessible, or not. Thus, clinicians need to keep in mind that my findings represent data from lived experiences at one point in time.

Summary

In this chapter, I attempted to situate the findings of this study in the clinical context, expanding on the reporting and discussion of my study findings in chapter 4 to 6. I also explained the limitations of my findings. In the next chapter, I construct a path connecting the participants’ experiences considered at risk for HBOC to nursing practice that can help people deal with uninformative genetic test results more effectively. I present practical applications of key findings and offer recommendations for clinical practice, research and policy development based on the study findings.
CHAPTER EIGHT:

Summary of Findings and Recommendations for Clinical Practice, Future Research and Policy Development

The overall goal of this study was to understand the experience of affected individuals with a family history of risk for hereditary breast and ovarian cancer who, despite their strong family history, were found not to have an inherited mutation. I entered this research believing that being in such a situation would lead all of the participants to interpret their genetic test results as inconclusive. I had little understanding of how they would reach this conclusion. As I shared with the study participants, I do not have a family history of cancer and did not fully comprehend how such a history would impact someone’s life. However, in the course of doing this research, I learned a great deal. I hope that the knowledge derived in this study will appeal to nurses and other health professionals working directly with individuals who inquire about, and undergo, genetic testing for breast and ovarian cancer gene mutations.

Summary of Findings

This study’s findings suggest that interpreting genetic test results for affected individuals considered at risk for HBOC who are found not to carry an inherited mutation is not straightforward (i.e., one has or does not have an inherited mutation). Rather, participants appended to the clinic’s interpretation – that they had not identified a specific mutation in their BRCA1 or BRCA2 genes – one of three additional meanings to their specific interpretation:

1. The results may be negative but we do have a mutation: either one that they have not yet identified or one that they have missed.
2. Maybe we have or maybe we don’t have a mutation.
3. The results were negative and we don’t have the genetic type of cancer.
These three identified meanings given by the participants to their uninformative results were reformulated as:

1. Confirmation of mutation carrier status.
2. Ambiguity regarding mutation carrier status
3. Refutation of mutation carrier status.

After the participants described how they interpreted their results, I sought a more in-depth understanding of how they made sense of them. It was in this context that the impact of their personal and family histories with breast and/or ovarian cancer became evident. I found 13 generic structures that represent beliefs that helped organised how participants made sense of their test results. The thirteen generic structures are:

Doubting their results because of

- the untested portion of their BRCA2 cancer gene, and
- being offered a more complete testing in the U.S.;

Seeing

- too many cancers in the family to be coincidental,
- too many deaths from cancer in the family to be all coincidental,
- resemblances among individuals with cancer,
- cancer diagnosed at the same age and at a young age, and
- having children or not, whether children are young, and children’s gender,

Attributing unique features to their mutation such as

- being transmitted through females or males only, or
- choosing only one organ (either the breasts or the ovaries), or
being specific to their ancestral lineage, or

being weaker – needing an internal or external trigger to activate,

Time

between testing and receiving results, and

between individuals with cancer across generations and within the same generation.

While these structures were found to organise most of the participants’ beliefs about their results, not all these structures were present in each individual’s making-sense experience. Out of these 13 generic structures, I derived a retrospective narrative of causal reasoning of self with a probable inherited mutation, which has seven stages:

1. Beginning to question the origin of cancers in the family.
2. Reflecting on the number of cancer cases in the family and the number of deaths from cancer.
3. Recognising the nonconformity of the number of cancers with what is observed in the general population.
4. Reflecting on one’s age at cancer diagnosis and those of others in the family.
5. Finding physical and behavioural similarities among individuals in the family with cancer.
6. Realising that the cancers in the family could result from an inherited mutation.
7. Becoming convinced that the family cancers must result from an inherited mutation when a new cancer diagnosis occurs in a young individual, or when an individual with a previous cancer experiences a second primary cancer or a recurrence.

This retrospective narrative is viewed as temporal and fluid and represents a speculation of how participants might work through the generic structures. Further, the narrative is seen as fluid as individual’s making sense process in relation to a probable inherited mutation may not
necessarily start at stage one but in further stages to then move back to previous stages, reflecting the ongoing nature of lived experiences bound to influence their saliency of genetic risk for breast and/or ovarian cancer. Nonetheless, the creation of this retrospective narrative of causal reasoning may be shown to be a useful tool when assessing how individuals perceive themselves to have a probable inherited mutation associated with breast and/or ovarian cancer susceptibility. As the findings indicate, affected women’s responses to and interpretation of their genetic test results are not straightforward and need to be understood within the context of their previous cancer experiences (Hallowell et al., 2004), and those of others in their families, as well as their evolving understanding of personal, familial and genetic risk.

Contrary to other study findings (e.g., Friedman et al., 1999), few women in my study expressed relief from their genetic test results. This difference could be explained by the fact that in my study, participants understood that their genetic test results did not eliminate their risk of HBOC, as explained to them in the letter from the Cancer Agency. The women in the Friedman et al. study may not have received such information. Nonetheless, although a few participants said that they were quite upset with their genetic testing experience, only one explicitly said regretting having gone through genetic testing. For the others, while they did not receive specific answers about the etiology of their cancers, their future risk of cancer and that of an inherited mutation (for themselves and that of their families), participants still felt that they had gained some information towards their “chronic disease” that is, living with a past cancer diagnosis.

Frost et al. (2004) recommend that new cases of affected individuals considered at risk for HBOC who come in for genetic counselling and testing need to be told that there are three categories of possible test results: (a) being found with a BRCA1 or BRCA2 mutation (a positive result), (b) being found with BRCA1 or BRCA2 mutation whose clinical relevance is unknown
or (c) a result that does not indicate a mutation but, because of their strong family history with cancer is considered inconclusive/uninformative. While these possible result outcomes reflects actual genetic testing practices, the findings of the current study indicate that few participants fully appreciated or understood their likelihood of receiving uninformative genetic testing results. Perhaps individuals should also be told what a negative and uninformative result might mean for their families; that is, they need to understand the difference between an uninformative result and a true negative result. When an individual receives a negative genetic test result after an individual in the family has already been identified with an inherited mutation, then the person’s result is a true negative. More needs to be learned about how genetic information is understood and remembered by individuals who receive uninformative test results. While all participants were told that there was a possibility of obtaining uninformative results, very few had expected to receive this type of results. According to their accounts, it would seem that they were expecting to receive either positive or negative results, with negative meaning that they and their family are not at risk of hereditary breast and ovarian cancer. In light of the participants’ experience, it might be useful to review the way risk information is provided during counselling and in providing their results. Further, the time between testing and obtaining results (which for the current study population took between 18 to 24 months) needs to be considered. Certainly, the long wait could have contributed to participants’ forgetfulness of the possibility of receiving results other than positive or negative as well as their dissatisfaction with their testing experience. While their dissatisfaction revolved mostly around the long wait for receiving results as opposed to the counselling process and subsequently receiving uncertain results, all but one said that they did not regret having gone through the experience nonetheless.
Reactions of the primary participants to the current testing ranged from anger, dissatisfaction, and bewilderment; although many were dumbfounded, some were satisfied with their experience. It can be speculated that having a shorter time around between testing and receiving results might have helped the individuals remember the possibility of obtaining uninformative results. A shorter time might also contribute to greater satisfaction with their decision to have genetic testing. Finding ways to reduce the waiting time between testing and providing results might be critical to enhancing individuals’ satisfaction and understanding about receiving uninformative genetic testing results.

With receiving uninformative genetic testing results, there is the possibility that individuals may be falsely reassured about their breast and ovarian cancer risk (Bish et al., 2002a; Claes et al., 2004; Frost et al., 2004; Hallowell et al., 2002). However, in this study, out of the 4 individuals who view their results as a refutation that they were mutation carriers, only 1 of them interpreted their test results to mean that their cancer risk is the same as the general population. The others understood that their test results were to be interpreted with caution, because of the uncertainty as a result of having a strong family history with cancer but with no identified mutation. However, many participants who still believe they may have an inherited mutation but not one associated with a BRCA1 and BRCA2 cancer genes that would also confer them an increased risk for ovarian cancer said that they now perceived themselves to be at lesser risk for ovarian cancer. As was observed within the findings, with the receipt of uninformative results to BRCA1 and BRCA2, some participants believed that their risk for breast cancer or for ovarian cancer was lowered – depending on their family history of cancer and hence, waived one risk with another. For example, these participants explained that if they were no longer at increased risk for breast cancer for lack of not having been found with a mutation in their
BRCA1 or BRCA2, then they felt that their risk for ovarian cancer was also lowered. While the participants were informed that no mutations were found in both of these cancer genes tested, the clinic did not firmly conclude to them that they did not have an inherited mutation in either of these genes or some other as yet unidentified breast and/or ovarian cancer gene. Hence, there will be an ongoing need for the clinics to refine their procedures for explaining to future clients the implications of receiving uninformative results for BRCA1 and BRCA2 among individuals considered at risk for HBOC especially those who judge these results to represent a lowered risk for other cancers as well.

As more is learned from the genetic components of common adult-onset disease, it can be expected that genetic testing for susceptibility to these conditions will gradually gain entrance into mainstream clinics. To ensure that susceptibility testing does not create harm to people's lives, there is a need for research that will lead to evidence-based protocols for genetic testing and follow-up care that will include communicating and providing support to those who receive uninformative genetic testing results to BRCA1 and BRCA2.

While some general recommendations have been offered above, what follows are recommendations based upon concerns raised by the study participants. While giving voice to individuals in qualitative research is valuable in itself, as nursing researchers we also have a social mandate to produce knowledge that can potentially improve nursing practice and the care of those who come into contact with health agencies. Thus, I offer recommendations I believe will improve clinical practice by helping to address the needs of individuals who come into contact with cancer genetics clinics. These recommendations are then followed by directions for future research and policy development.
**Recommendations for Clinical Practice, Education, Research and Policy Development**

**Communicating uninformative genetic testing results**

Prior to receiving genetic testing, participants’ perception of risk for an inherited mutation was grounded in their personal and familial context to cancer. Participants came to assess their risk of having an inherited mutation and their future risk of cancer for self and their family members in relation to others. In this context, it is no longer just the self that was assessed for risk of cancer and for an inherited mutation but the self in relation to others. Hence, although it has been stated that providing genetic risk information beyond an individual’s assessment of a family context can be challenging for the clinicians (Bottorff et al., 1999), findings of this study show that participants did assess both future risk within the larger familial context. Clearly, finding ways to facilitate the provision of genetic risk information to individuals from a familial context requires further consideration and inquiries. Within the genetic counselling session, exploring further how individuals could make sense of uninformative results in light of their family history of cancer would facilitate both the individual and the counsellor understanding about how one comes to understand and integrate such results.

Along with assessing their likelihood of carrying an inherited mutation within their personal and family experience to cancer following their experience with genetic testing and upon receiving uninformative results to BRCA1 and BRCA2 cancer genes testing, other participants now recognised, within these results, their possibility of carrying an inherited mutation increasing their susceptibility to future breast and ovarian cancer. In light of their family history, some participants interpreted their results as confirmation of having a probable inherited mutation. Considering that these individuals entered genetic testing with the hope that the results could confirm their perception to having an inherited mutation, the genetic
counselling process should perhaps include not only informing individuals of the likelihood of not finding any mutations but also asking individuals to consider the scenario that, if the testing does not find any mutation, what would such results mean to them in light of their family history with cancer? The structures described in this study could prospectively serve as a guide to health professionals in assisting their clients to make sense uninformative results. Having a chance beforehand of making sense of possible uninformative results before the actual receipt of them might facilitate understanding of their results on the day of receipt added to the potential to increase satisfaction with their genetic testing experience. Further, such an exploration might enhance the health professionals’ understanding of how family histories with cancer may interact to influence the perception to carrying an inherited mutation at the individual level using aggregate knowledge derived from this study. For example, findings from this study show that individuals on the one hand, whose family history with cancer followed some form of pattern such as breast cancer appearing within similar ages across two or more generations, were more likely to interpret their results as confirming their perceived likelihood of carrying an inherited mutation. On the other hand, individuals who did not necessarily see a form of pattern occurring within their family history of cancer were more likely to interpret their results as a refutation of not carrying an inherited mutation or to having a weaker type of mutation, not one likely associated with a BRCA1 or BRCA2 mutation.

Considering the growing number of individuals who are likely to receive uninformative genetic testing results, communication of genetic information should stress the likelihood of receiving this type of result in comparison to being found with a mutation. While current practice in genetic counselling does present the possibility of not finding mutations, the findings of this study raise the question of how much more content should be added and emphasised related to
the possibility of receiving uninformative genetic testing results and the implications of such results in establishing one’s genetic risk status and future cancer risk. I would recommend that, ideally, individuals should be assessed prior to consenting to testing of their understanding of the possibility of receiving uninformative results. Further means of ensuring individuals understanding to the possibility of receiving such type of results needs further assessment in light of the dissatisfaction reported in this study.

In response to the study participants’ multiple and complex interpretations of their test results, development of educational material for clients seems warranted as well as further support mechanisms. In order to provide long-term support, I would recommend that, ideally, regardless of test results, all of those undergoing testing be contacted again after disclosure to discuss issues of concern such as test interpretation, screening options and to evaluate the need for further referral. This support could be given in parallel to educational materials explaining how test results can be interpreted in light of a strong family history with cancer, what medical management options they can consider, and ways they can live with a chronic risk for an inherited mutation such as discussing life style choices to reduce their risk for cancer in light of their possible inherited mutation. As recommended by Frost et al. (2004), individuals should be given educational material before consenting to genetic testing so that they know that genetic test results are not straightforward (i.e. you do have a mutation or you do not). By understanding this in advance, individuals can make better informed decisions about whether to pursue genetic testing or not. Further, providing some time between receiving educational materials and obtaining genetic testing might prove more beneficial to client’s understanding to the full implications of receiving uninformative genetic testing results for themselves and for their family.
Providing support with uninformative genetic testing results

As detailed in Chapter 2, individuals who received genetic testing from the HCP and who are found to have an inherited mutation are asked to return to the clinic for receipt of official results while those whose results are uninformative are informed of their results first by telephone followed by a letter discussing the possible ways their results can be interpreted. Despite being invited to contact the Cancer Agency to discuss further any inquiries they may have concerning their test results, very few participants said they did. While the Cancer Agency also offered three in-house information sessions, only two of the study participants mentioned having participated despite many expressing a need to further discuss their interpretation of their results. Perhaps they felt that, since they only had a few questions, coming to a session was not worth their time. It could also be hypothesised that the timing of support was too early – that individuals had not yet had a chance to make sense of their results and the impact of these on their everyday lives. That is, all participants expressed how helpful it had been for them to discuss how they interpreted and made sense of their results at the time of the interview – which in most instances was a year to two years following receipt. Hence, the findings suggest that it will remain important for clinics to explore further ways to provide for long-term support for making sense of uninformative results. Frost et al. (2004) showed that women who received their genetic testing results (either positive or uninformative) face-to-face from a genetic counsellor dealt better with the results. As Iglehart comments, “It is a job to counsel these people because you have to tell them that they do not have any mutations that we can find, but we still think something is wrong” (Iglehart et al., 1998, p.383). In light of the study findings and those from Frost et al. study, the benefits of receiving face-to-face counselling might further be emphasised in the letter sent to individuals receiving uninformative results and the encouragement to seek an
appointment with the HCP should they feel the need to discuss further the meaning of their results might be strengthened. Recognising that most genetic testing agencies are already working beyond their physical resource capacity, providing support in interpreting uninformative genetic testing results to breast and ovarian cancer susceptibility might also be shared by general health care practitioners so that they, in turn, could provide assistance to their clients in making sense of such results. This context would help to attain the goal of providing face-to-face counselling for all those who receive uninformative results.

While participants were provided with resources to help them interpret genetic testing results and to interpret uninformative results, most of them still struggled on how to make sense of them. While the letter received by the participants informing them of their results helped them to interpret and make sense of the results from a clinical perspective, some content of the letter created further confusion as to how their results could be interpreted. While this study is not a prospective study but a retrospective and did not include evaluating the format by which support was given to participants on how to make sense of their results, it is problematic to comment on how the content of their letter might have been modified to facilitate making sense of uninformative results. Nonetheless, what can be brought forward, in light of the study findings, is the idea that content areas within the letter may lead to contentious issues for the participants’ making sense process. For example, individuals described how uncertain they were to interpret their results as either still having or not having an inherited mutation when their own testing Agency recommended that they seek further complete testing in the U.S. for a significant fee. They reinterpreted this statement from their letter to mean that perhaps their own testing

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91 Recall from Chapter 2 that as part of the genetic counselling process that individual who received uninformative results were first informed by telephone then followed by a two page letter informing them on how their results could be interpreted in light of their strong family history of breast and/or ovarian cancer. Further, all individuals who receive genetic counselling from the HCP also receive an informational booklet on hereditary breast and ovarian cancer.
Agency believed that they are carrying an inherited mutation when in reality; the testing Agency’s intent was only to provide them with further options where full sequencing of both genes could be obtained. Being told that only a certain percentage of their BRCA2 gene had been tested represented the second contentious issue that caused dissatisfaction to some participants. Very few participants understood that the percentage tested of their BRCA2 represented areas where most mutations to date of clinical relevance have been identified. In reality, learning of the untested portion of their BRCA2 gene created doubt and uncertainty regarding their true genetic status for a possible inherited mutation. Modifying these two parts of the letter could likely enhance individuals’ making sense process for interpreting uninformative genetic testing results.

Support in providing uninformative genetic testing results for BRCA1 and BRCA2 is also needed for individuals who choose to share their results with family members. Among the study participants who chose to share their results and among the few who decided not to share as they felt there was little information to tell, both struggled with what they should have told – should they be telling family members that they do have an inherited mutation or be telling them that they do not or most likely do not. In this effect, interpreting uninformative genetic testing results in light of their strong family history with breast and/or ovarian cancer is complex and further means of support should be developed for individuals who chose to share their results with family members. As explained by Kenen, Ardern-Jones and Eeles (2003a), when faced with complex and upsetting knowledge, individuals may turn to the use of heuristics (inferential cognitive shortcuts) to simplify complicated situations. Although all individuals who received uninformative results were told that they could choose to share with family members their letter received by the Cancer Agency informing them of their results, very few did send a copy of their
letter but chose to give results by telephone or in person but nonetheless, expressed struggling with what to tell family members.

Just as some of the study participants’ perception of having an inherited mutation seemed to have remained unchanged with having received uninformative results, Watson’s et al. (1999) concluded from their study that 61% of the women with a family history of breast cancer continued to overestimate their cancer risk following genetic counselling that provided them with corrective information related to their lifetime risk. Croyle and Lerman (1999) have explained that perceptions of personal risk for cancer can be resistant to standard education and counselling approaches and that other factors such as stress can interfere with one’s ability to see an alternative view of self. Considering that living in constant threat of cancer and of a possible inherited mutation may affect one’s quality of life (Esplen et al., 1998; Pasacreta et al., 2002), assessing the level of stress of individuals seeking genetic testing may seem warranted.\(^\text{92}\) Ritvo et al. (2000) did find that a significant proportion of the women studied experienced substantial distress during initial assessment of genetic risk and at follow-up.

**Directions for education**

Nursing in genetics is not currently a recognised speciality in Canada nor can nurses obtain credentials in genetics here. Yet, as people become more aware of the impact of genetics on health, nurses will be called upon by clients and their families to answer questions about genetics, genetic testing, and/or risk-management options. There is an urgency to create more sophisticated and specific nursing responses with the recent advances in basic and translational

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\(^{92}\) While there exist multiple valid, reliable tools to measure cancer distress (such as intrusion subscale of the Impact of Events Scale [IES] and the Centre for Epidemiological studies Depression [CESD] scale), there are almost none that can accurately assess distress related to perceived genetic risk. Although instruments such as the Distress Specific to Gene Status have been used in research, their clinic validity and reliability have not been reported (Kelly et al., 2004). Research assessing and developing tools to assess perceived risk of an inherited mutation is needed to increase resources available to health professionals.
Nurses have been instrumental in strategising the delivery of genetic testing in relation to hereditary risk for breast cancer and the psychosocial support needed with genetic testing (Thorne, 2001). While there are nurses who directly provide genetic services in the area of adult-onset hereditary diseases, for others, genetics is a small but significant part of their work (Bottorff et al., 2004). Bottorff et al. (2004) report that the Canadian nurses they surveyed who worked in clinical areas that require some genetic nursing knowledge felt ill-prepared to address patient questions about genetics. Their research further exposed challenges in genetic nursing, such as role ambiguity, lack of recognition for nursing expertise, limited availability of genetic education, isolation, and the instability of nursing positions.

As Bottorff et al. (2004) conclude, there is an urgent need to develop genetics education in Canada to support nurses both new and already practicing as they integrate genetics into their roles. However, the current teaching content in undergraduate, graduate and continuing education programs still limits the potential roles that nurses could play in genetic services. “Because genetics is not identified as a basic competency for nursing licensure exams, there is little incentive for schools of nursing to include genetics in their programs” (Bottorff et al., 2004, p. 6). At the very least, nurses with an undergraduate degree working in family medicine should receive training on the role of genetics in health and disease process, in assessing who could benefit from screening for cancer risk, and when to make referrals and where.

The study findings offer directions for an initial role for nurses working in genetics. Some of the findings also indicate areas where nurses need further education to prevent harm through misuse and misunderstanding of genetic information and technology (Kirk, 2004). Knowledge derived from this study that could be integrated into undergraduate and graduate nursing programs and could apply to nursing practice includes: (a) the implications of living at
risk of breast and/or ovarian cancer and its relation with perception of risk for an inherited mutation, (b) the types of test results interpretations, and (c) the generic structures that organise how individuals make sense of their results. The above topics could provide guidance to nurses in supporting individuals who receive uninformative genetic test results as they try to interpret and make sense of them.

**Directions for further research**

In chapter 4, I described how participants have come to learn to live with their increase cancer risk and now must learn to live with their possible increase genetic risk for breast and ovarian cancer susceptibility. A salient finding of this study is that uninformative genetic test results entail many uncertainties. Such as, the results did not eliminate conclusively their risk for carrying an inherited mutation in their BRCA1 and BRCA2 cancer genes or the possibility that they may still carry some other as yet unidentified other hereditary breast and/or ovarian cancer susceptibility. Hence, affected women and their family members from families with a history of breast and/or ovarian cancer face a great deal of uncertainty and are faced with learning to live with many chronic risk perspective (Kené et al., 2003b). Innovative research looking at the implications of living with multiple sources of uncertainty in a chronic risk situation – chronic cancer risk and genetic risk perspective – seems warranted so that the complexities of such situations on individuals’ health and illness experiences can be better understood. Findings of such studies could provide guidance towards effectively supporting individuals in making health-related decisions with uncertain genetic information. As Mishel (1984) has shown, there is a link between uncertainty and increased stress while stress is hypothesised to hinder one’s ability to see self in other embodied perspectives (Croyle & Lerman, 1999). For example, increase stress created from fear of carrying a possible inherited mutation could limit individuals from
interpreting uninformative genetic testing results as other than confirmation of mutation carrier status or ambiguity regarding mutation carrier status. As Renn (2004) noted, subjective risk perception will also be influenced not only by how risk is communicated but also based on an individual’s psychological mechanisms for processing uncertainty and earlier personal experience of danger. Risk perception can also be affected by the extent of fear associated with a possible outcome (Slovic, Fischhoff, & Lichtenstein, 1980). To maximize quality of life, further research is warranted to find ways to reduce uncertainty associated with a possible inherited mutation and to help individuals learn to live with uncertainty regarding their mutation carrier status and uncertainty associated with their increased risk for cancer. As well, longitudinal studies are needed to fully understand the long-term consequences of living with uncertainty and being an “at risk individual” to cancer, and more specifically, living with a chronic risk for HBOC in the event where their true genetic status cannot be determined.

While this study did not address the influence of test results on medical management, very few participants intended to use their genetic test results to decide how to reduce their cancer risk (although all women in this study had already had cancer). As few researchers have focussed on individuals who receive uninformative test results for breast and ovarian cancer susceptibility, there are many possible areas of inquiry. Research could focus on: (a) how affected individuals considered at risk for HBOC have used uncertain genetic test results to assist in medical management, (b) how they communicated their results to family members, (c) how such results influenced their perceived risk of cancer and their perceived inherited risk for cancer and, (d) how results affected their overall quality of life.

There is also a need for longitudinal studies addressing how unaffected individuals from families at risk of HBOC respond to and cope with uninformative results from the testing of a
family member and the impact of these results on their cancer screening and risk management behaviours. While past research exists on the psychological consequences of living with an increase risk perception for breast cancer as a result of having a family history with the disease, future research will need to address the psychological consequences of living with a heightened perception of risk for an inherited mutation following the receipt of uninformative results from a family member who has undergone genetic testing.

Another potential area of research is investigating the impact of the family guardian role that many participants in this study spoke of. To be adequate family guardians, participants said that they needed support to keep apprised of the latest cancer and genetic discoveries. While having greater depth of understanding to the impact of enacting the family guardian role among families at high risk for cancer and for an inherited mutation may prove worthwhile, Appleton et al. (2001) also cautioned that family members who adopt this role may be chronically distressed and may be poor role models for the next generation who are likely to face the same constant threat to cancer.

Participants in my study spoke of seeing genetic testing as a way of gaining control over their chronic disease – that is, participants spoke of their past cancer diagnosis as a chronic disease that they must come to learn to live with. As most expressed, genetic testing produced no where near as much anxiety for them as dealing with cancer. However, their genetic testing result was a disappointment for them as it created uncertainty about their actual genetic risk for an inherited mutation to breast and/or ovarian cancer. As pointed out by Baum, Friedman, and Zakowski (1997), uncertainty can lead to increased stress. Future research is needed to compare the presence and level of stress before and after receiving uninformative genetic testing results. Findings from this potential research could lead to a study comparing the effect of receiving
uninformative test results versus positive test results on recipients’ stress levels. The findings of such a study could be the basis for policy guidelines towards providing long-term support to individuals who receive uninformative test results such as those who receive positive results to BRCA1 and BRCA2.

Based on my study results, it could be argued that receiving uninformative genetic test results may have a greater impact on extended family members (Frost et al., 2004) than on the women tested who have already been diagnosed with cancer. Future research could examine the expectations of family members who ask an affected family member to undergo genetic testing on their behalf, and how these family members understand the test results.

In the previous section, I recommended improving how uninformative results are communicated during counselling as well as finding ways to provide long-term support for those receiving uninformative results. Within this context, future research is needed towards examining the best means of providing support and comparing the effectiveness of these. Such studies could compare the effectiveness of face-to-face support versus telephone support as well as the effectiveness of the follow-up letter in enhancing understanding of cancer and genetic risk, ensuring continuance of cancer screening and in decreasing stress associated with the uncertainty to receiving uninformative results. Such findings could delineate the negative and positive outcomes of different approaches and determine which causes the least harm, is financially viable, and timely.

**Directions for policy development**

Clearly, policy decisions must be based on evidence far beyond the scope of a single qualitative study. However, findings from a study such as this one, considered in the context of the larger body of emerging knowledge, can help illuminate some of the policy questions that we
might properly be asking as we continue to make decisions related to genetic testing for at-risk populations. As new genetic testing technology is invented, policy makers and researchers need to continue delving into the potential impact of new tests: their cost versus their benefits, their quality, and equity of access to them (Morgan, Hurley, Miller, & Giacomini, 2003). In the context of cost, continuous evaluation is needed to assess how the availability of genetic testing for breast and ovarian cancer susceptibility changes risk management behaviours. While the basic cost covers the screening itself, what are the long-term costs and benefits to individuals, families and society if the state chooses to fund such services? For example, many participants entered genetic testing not knowing what to expect with the results. After receiving the results, almost all participants now perceived themselves to a greater likelihood of carrying an inherited mutation – either in their BRCA cancer genes or in some other as yet unidentified cancer gene. Considering that increased uncertainty to one’s health status causes stress (Mishel, 1984), such a transition in viewing self needs further inquiry to determine if the availability of genetic testing creates more stress and uncertainty without sufficient benefit.

The above issues and others related to the benefits and harms of knowing or not knowing one’s genetic risk are areas of research valuable to policy makers in determining the financial, psychological and medical benefits of funding genetic testing. While this study has broadened the understanding to how uninformative genetic testing results are interpreted and made sense of in light of a strong family history with breast and/or ovarian cancer, like other studies, however, I am left with numerous questions. Questions to consider for future genetic testing guidelines are in what context should communicating uninformative genetic testing results take place and how should we support individuals who receive such result. For the former, there might be a need for Canadian genetic testing guidelines to reflect the UK guidelines to offer genetic testing only after
individuals from high risk families who have a 20% chance or greater of having a BRCA1 or BRCA2 mutation in the family receive two sessions of pre-test counselling (Public Health Genetics Unit, 1998). Having two sessions prior to deciding on going forward with genetic testing might facilitate integration of the possibility to receive uninformative results and the implications to receiving such results for self and family members. As for the latter recommendation to how individuals who receive uninformative results be supported, considering their struggle at making sense of uninformative results, testing Agencies might need to consider delivering such results in the same format as those receiving positive results. Recall that individuals who received genetic testing at the HCP obtained positive results face-to-face while individuals who received uninformative results were informed by a telephone call from a genetic counsellor followed by an informational letter.

Inquiry is also needed to determine who should get genetic testing because clinicians and task forces are clear that screening of the entire population is not warranted (Burke et al., 1997; Koenig et al., 1998). Further research is needed to deduce eligibility criteria that will maximise the likelihood of finding mutations within BRCA1 and BRCA2 cancer genes, therefore limiting the population to be tested, keeping the overall cost of screening down, and minimising distress as a result of receiving uninformative test results. As emphasised by Morgan et al. (2003), targeting screening to populations at risk reduces the incidence of false positives; thus improving the test’s predictive value.

If as a society we choose, however, to make genetic testing available to all, then we need to ask whether this universality diverts health care resources away from more cost-effective means of promoting health (Morgan et al., 2003). We still do not know whether testing for BRCA1 and BRCA2 mutations improves screening adherence, morbidity, or mortality. Although
empirical studies have shown that when a mutation is discovered, an individual’s lifetime risk of developing breast and ovarian cancer rises, their risk for cancer is not 100%. Indeed, they may never get these diseases.

In conclusion, this study sought to examine the complex experience of interpreting uninformative genetic test results, and how such interpretations are altered through the making sense of results. By focusing on individuals’ retrospective interpretation of receiving uninformative results within a clinical setting and their effects on everyday issues, I hope that I will have contributed knowledge that will have relevance for individuals who in future will encounter such settings and clinical utility for those who provide practical care. I hope that I have planted the seeds for improvement in the way genetic services are provided to individuals and families by showing clinicians a more complete picture of their clients’ experiences over the whole trajectory of genetic evaluation.
REFERENCES


Appendix 1

Letter received by the study participants informing them of their genetic test results
Dear «Title» «LastName»:

You were seen in the Hereditary Cancer Program to review your family history of breast cancer and/or ovarian cancer. Following your genetic counselling session you provided a blood sample and agreed to participate in testing of the BRCA1 and BRCA2 genes. The goal of this testing is to identify a mistake, or mutation, in one of these genes that would explain to a great degree your personal and family history of breast/ovarian cancer. Once the family mutation is known, any blood relative could choose to have genetic testing to determine if they have or have not inherited an increased risk for breast/ovarian cancer.

This letter provides you now with a summary of your genetic testing results. Our laboratory uses a combination of two methods to look for mutations in the complete coding sequence of BRCA1, and the majority of BRCA2. (See note below for more technical details.)

To this date, we have NOT identified a specific mutation in the BRCA1 or BRCA2 genes. The result can be interpreted in the following ways:

- A mutation may exist in the regions of BRCA1 or BRCA2 that our lab looked at, but it has not been detected by our current testing method OR
- A mutation may be present in the as yet untested portion of the BRCA2 gene OR
- The responsible mutation may be in another, as yet unidentified, hereditary cancer gene OR
- You do not have an inherited breast/ovarian cancer gene mutation, which means that your cancer diagnosis may have occurred by chance.

We must highlight that this result does NOT mean that we have completely ruled out an inherited breast/ovarian cancer gene mutation in your family. Based on your family history of cancer, we recommend that cancer screening continue as previously recommended.

We hope to be able to offer further testing in the future and will keep you informed if anything is identified. We would like to advise you that a more complete assessment of the BRCA1 and BRCA2 genes (DNA sequencing) is immediately available through a private laboratory in the U.S. at a cost of
approximately $3,850.00 CDN. No laboratory can currently detect all mutations in these genes. We estimate that there is less than a 10% chance that this additional testing will identify a mutation that is known to be clinically significant. If you are interested in this option, we can provide information about how to proceed.

We encourage you to share this letter with your family doctor or other health care provider. You may also wish to share this letter with your relatives, especially those who know you have been waiting for genetic test results. You may recall that we are unable to provide this information to anyone else without your specific consent. As further testing may be available in the future, we ask that you inform us of any change in your address/telephone number. We will not conduct further testing without first contacting you to see if you want further testing at that time.

Thank you for your patience and understanding. If you have any questions about this letter or its implications for your ongoing health care please do not hesitate to call your genetic counsellor in the Hereditary Cancer Program in Vancouver at (604) 877-6000, local 2198, or in Victoria at 250-727-4176.

Sincerely,

David Huntsman
Acting Lab Director

Barbara McGillivray
Medical Geneticist

Karen Panabaker
Genetic Counsellor

cc: «DRNAME»

**Technical details:**
This DNA sample was screened for mutations in 100% of the coding sequence of BRCA1 and 65% of BRCA2 (including exons 9, 10, 11, 20, 23, 25) using a combination of two methods – the Protein Truncation Test and fluorescent Single Strand Conformational Analysis. No test method will detect 100% of existing mutations. Neither our approach nor DNA sequencing will detect deletion of complete exons or genes, or errors unrelated to the DNA exon sequence. We believe that our false negative rate is less than 10% for sequence-based mutations. Advances in genetic testing technology may allow for more complete analysis of this DNA sample in the future. We do not have the resources to undertake additional testing at this time.

H:HCP2/gcdoc/brc1a2neg
Appendix 2

Preliminary interview schedule
SAMPLE PRELIMINARY INTERVIEW SCHEDULE FOR
PRIMARY PARTICIPANTS

Begin interview by restating the purpose of the study.

1. Life history (biographical data)
   - Please tell me about yourself.
   Prompts:
   - Can you tell me about your breast cancer experience?
   - Can you tell me about your family’s history of breast cancer?
   - Have you experienced a close family member’s cancer experience?

2. Genetic testing for breast and ovarian cancer susceptibility
   - Please tell me about your experience with genetic testing.
   Prompts:
   - How did you come to know of the existence of genetic testing?
   - How long have you known about the availability of genetic testing?
   - Did you make that decision alone? Who first requested testing?
   - How would you describe your experience with genetic testing?
   - What did you expect your results to be?
   - How do you describe your results? Positive, negative or something else?
   - How would you describe your experience of receiving your genetic test results?
   - How did you feel before obtaining your results?
   - What were the risks and benefits of testing?
   - How do you see your risk of developing breast cancer or not, now that you have received your results?
   - Do you feel that you have done things that decrease your risk?
   - What do you know about heredity and how it plays out?
   - How do you explain the appearance of your breast cancer?
   - Do you feel that there is a mutation in your family?
   - Do you feel that you or another member of your family inherited a breast cancer mutation? If so, how do you explain this?
   - What was waiting for test results like for you?
3. Genetic testing and family
- Please tell me about your experience of undergoing genetic testing.

Prompts:
☐ Did you attend genetic counselling by yourself?
☐ Did you share your experience of genetic testing with other family members and, if so, with whom and for what purpose?
☐ Did you share your genetic test results with family members?
☐ How did your family respond/ react to your genetic testing experience?
☐ What do you see your family’s risk to be, considering your genetic test results?

4. Wrapping up
- Is there anything you would like to say about this interview? What do you hope I learn from your experience?
- What is most important about your experience?
- Is there anything else you would like to share with me about your experience?

At the end
➤ Ask participant to choose a code name.
➤ Are you interested in receiving a written report?
➤ If a follow-up interview is needed to clarify some issues, may I call you?
Begin interview by restating the purpose of the study.

1. Interaction with relative’s genetic testing
   - What do you know about your family history of breast cancer?
   - How has breast cancer affected your life?
   - When your (relationship to primary participant) was diagnosed with breast cancer, did this influence what you thought about your own risk, or the risk of others in your family members?
   - How do you explain the appearance of breast cancer in your (relationship to primary participant)?
   - Please tell me about some of the times you and your (relationship to primary participant) discussed genetic testing and what you talked about.
   - In what ways did you support your (relationship to primary participant) as she was going through her experience with genetic testing?

2. Knowledge and use of test results
   - Has (relationship to primary participant) shared her genetic test results with you and, if so, what did she say her results were?
   - Did you want to know of your (relationship to primary participant) results? Can you explain why you wanted to know or not?
   - Were you ready to receive genetic information about you and your family when (name of primary participant) decided to go for genetic testing?
   - Do you feel that knowing your (relationship to primary participant) results offered you any clinical advantages? If yes, what advantages?

3. Perception of cancer risk
   - How much do you know about heredity and how it plays out in your family?
   - Do you feel that there is a breast cancer mutation in your family?
   - Do you feel that you or another member of your family inherited a breast cancer mutation? If so, how do you explain this?
   - What did you expect your (relationship to primary participant) results to be?
What do you see your family’s risk of breast cancer to be, considering your (relationship to primary participant) genetic test results?

4. Reaction to results
- Did you get any relief from genetic testing in terms of your risk and your family’s risk of breast cancer? If yes, what relief?
- What are the benefits and risks of genetic testing?
- Your (relationship to primary participant) had genetic testing in (year) and received her results in (month/year). What was the waiting like for you?

5. Wrapping up
- Is there anything you would like to say about this interview? What do you hope I learn from your experience?
- What is most important about your experience?
- Is there anything else you would like to share with me about your experience?

At the end
➢ Ask participant to choose a code name.
➢ Are you interested in receiving a written report?
➢ If a follow-up interview is needed to clarify some issues, may I call you?
Appendix 3

Letter of support from the BCCA Hereditary Cancer Program
Appendix 4

Participant information and consent form
PARTICIPANT INFORMATION AND CONSENT FORM

RESEARCH FOR A GRADUATE THESIS
INTERPRETATIONS AND MEANINGS OF RECEIVING NEGATIVE GENETIC TESTING RESULTS
TO BREAST CANCER SUSCEPTIBILITY
FOR MEMBERS OF FAMILIES WITH A STRONG HISTORY OF BREAST CANCER

INVESTIGATORS:
Christine Maheu
Ph.D. Student
School of Nursing and Centre for Applied Ethics
Dr. Sally Thorne,
Professor, Dissertation Supervisor
School of Nursing
For more information, or to arrange participation, please contact Christine Maheu at: (604) xxx-xxxx

PURPOSE
The purpose of this project is to understand and interpret the experience of individuals and their family members of receiving genetic testing results and to describe how the availability and use of genetic testing affects their every day life, health and illness experiences. We are particularly interested in understanding the experience of individuals who have a family history with breast cancer, but whose genetic testing results have found no genetic mutation. In addition, we are
interested in learning about the experience of family members who have not participated in the genetic testing.

PROCEDURES
In this study we want to understand the specific meanings that characterize the experience of going through genetic testing for members of families who have a family history with breast cancer, including family members who have not had genetic testing. We are inviting you to consider this study because either you or the family member who gave you this form has participated in genetic counselling and received genetic testing results.

You have received this information and consent form because you have voiced an interest in participating in this study to the genetic counsellor of the Hereditary Cancer Program at the BCCA. Within the following weeks the investigator, Christine Maheu, will contact you to see if you still agree to participate in this research. If you still agree, Christine will discuss with you the study and arrange for a mutually convenient time and place to do the interview. The interview will require about 90 minutes.

In order to clarify her understanding of ideas discussed during the initial interview, Christine may contact you to discuss these ideas or to request a second interview within a few weeks of the initial interview. If you would prefer not to be contacted further, your wish will be respected. Further, if you think of issues arising from the interviews that you would like to discuss or wish to clarify any of your comments following the initial interview, you will be free to contact Christine for that purpose. If you are interested in receiving a written report of the research, this will also be provided to you.

We may also ask you to pass on a copy of this “Participant information and consent form” to other family members who might be interested in participating in the study themselves. Christine will discuss with you who in your family it would be most helpful for us to interview. People who you identify as possible candidates for such interview must feel entirely free to participate or not according to their own decision, and if they decide to participate, it will be left up to them to contact Christine to make arrangements.

CONFIDENTIALITY
The discussions will be audiotape and transcribed. The typed transcript will not have your name on it; the interviewer will assign code names for all persons mentioned in the interview. Only the investigators will know your identity. The consent forms, the list assigning pseudonyms and the transcripts will be kept in a locked cabinet in the project office. The audiotapes will be erased once they are transcribed. Written transcripts, with no names or identifying information on them, will be retained by the researcher for a period of five years to permit future secondary analyses.

RISKS
There are no physical risks to this research but, because some of the topics are sensitive ones, information will be available regarding appropriate referrals should you require further support or discussion about this or any other topic. In this study, we will not provide information about familial and genetic risk for breast cancer. Such information is available through the BC Cancer
Agency’s Hereditary Cancer Program (877-6000; extension 2325).

CONTACT
Your participation is entirely voluntary and you should feel free to ask questions at any time. If you have any questions, please call Christine Maheu at (604) xxx-xxxx, or Dr. Sally Thorne at the UBC School of Nursing (822-7482). If you have any concerns about your rights or treatment as a research subject, you may contact Dr. Richard Spratley, Director of the UBC Office of Research Services and Administration at 822-8598.

CONSENT
If you decide to participate in this study, you may withdraw your consent to participate at any time and for any reason. If you agree to participate, you will be free to refuse to answer any question, or to have all or portions of the audiotaped interview erased at your request.

Whether or not you participate in this project cannot affect your health care in any way. You will be given a copy of this “Participant Information and Consent Form” to keep for your records when you sign the consent form at the beginning of the interview.

Your signature here indicates that you have had the study explained, read this consent form, had any questions answered to your satisfaction, and agree to participate in the interviews described above.

________________________________________
Name (printed)

________________________________________
Signature                                      Date signed
Appendix 5

Family pedigrees of the 21 primary participants
PP1: Macy

SP1: Kimberly - Sister

INDEX: PP

PROBAND: PP

Female
Male
Deceased
Prostate cancer
Esophageal cancer
Osteo-sarcoma
Breast cancer
Skin cancer
Do not know gender
Attended genetic counselling session

Dx [#] = Diagnosed at age #
Dx [#] = Deceased at age #

Note:
INDEX had genetic testing; PROBAND initiated genetic testing process.

PP2: Juniper

SP1: Lily - Sister

INDEX: PP

PROBAND: PP

Female
Male
Deceased
Do not know gender
Breast cancer

Dx [#] = Diagnosed at age #
Dx [#] = Deceased at age #

Note:
INDEX had genetic testing; PROBAND initiated genetic testing process.
INDEX had genetic testing. PROBAND initiated genetic testing process.
Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.

PP5: Eschen
SP1: Janet - Sister
SP2: Lisa - Sister
SP3: Diane - Cousin

INDEX: PP
PROBAND: PP

Female
Male
Deceased
Esophageal cancer
Breast cancer
Skin cancer
Fibrous sarcoma

Dx[45] = Diagnosed at age #
Dx[40] = Deceased at age #

Dx [35] benign fibro adenoma
Dx [60]

PP6: Beatrice
INDEX: PP
PROBAND: PP

Female
Male
Deceased
Prostate cancer
Breast cancer
Colon cancer
Lung cancer
Ovarian cancer
Lymphoma
Thyroid cancer
Uterine cervical cancer

Dx [45] = Diagnosed at age #
Dx [40] = Deceased at age #
INDEX had genetic testing; PROBAND initiated genetic testing process.
Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.

Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.
Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.
INDEX: Louise

PROBAND: Louise

Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.

PP14: Donna

INDEX: PP and sister

PROBAND: PP and sister

Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.
Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.
Note:
INDEX had genetic testing. PROBAND initiated genetic testing process.
PP20: Emma
SP1: William - Husband
SP2: Linda - Niece

Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.

PP21: Sherry
SP1: Susan - Sister

Note.
INDEX had genetic testing. PROBAND initiated genetic testing process.
INDEX: PP and Alex
PROBAND: Alex and cousin

PP22: Victoria
SP1: Duchess - cousin

Female
Male
Deceased
Do not know what type of cancer
Breast cancer
* Attended genetic counseling session
Dx [#] = Diagnosed at age #
Dc [#] = Decayed at age #

Note:
INDEX had genetic testing, PROBAND initiated genetic testing process.
Appendix 6

Sample: field notes
Sample: field notes

Date/time/place of interview
Thursday, April 12, 2001 from 10:30 a.m. to 12:00 p.m.

Field notes written on:
Thursday, April 12, 2001, 4:00 p.m.

Notes on initial contact
She greeted at the door. I saw her husband in the window, looking out maybe to where I was parking. He went upstairs as soon as I came in. We had talked on the phone twice and both times the conversation was jovial. I asked her if this was still a good time to do the interview and she replied “of course”.

Informant description
Women in perhaps late 50s, wearing eye blue makeup. We did the interview sitting at the dining table next to a window. She was comfortable with me and helped me find a plug for the tape recorder, although she said, “I hope I’ll forget about that thing,” pointing to the microphone. Told her she would, but I also moved the tape recorder and microphone away from her.

Surroundings
We sat across from each other at the dining table next to a window. On our left was the living room with another window. The place was small but cozy. The room was rather cold, and I did get cold during the interview. By chance that morning, I had worn two pairs of socks but forgot to bring a vest. The place was private and the husband stayed upstairs throughout the interview.

Preinterview comments/observations
I really looked forward to this interview, as she had said on the phone, “Quite frankly my experience with genetic testing was not good.” I was intrigued with what she had to say.

Informant’s mood
She was awake, alert, ready to talk and share. The whole house seemed ready to receive me as there were fresh flowers everywhere and she had tea ready on my arrival.

Researcher’s mood
At first I was afraid that I might be too tired that morning to conduct a good interview. I had not slept much, following the news that my father had had a massive stroke the day before. However, as soon as I entered the house, I became extremely focussed and alert. Establishing rapport was easy because she was so welcoming.

General observations during the interview
At first it felt that she didn’t know where to start the interview, so I told her to tell me a little of her family’s history with breast cancer and some of her own personal experience. After she started talking about her experience with breast cancer, it became easier for her to continue talking and fall into her experience with genetic testing.

Interesting points/recurrent themes
- She explained how her experience with genetic testing had been negative. She saw nothing good in her experience for herself and her family, especially for her daughters.
- She was angry about the results. “They couldn’t be right. They don’t know what they’re doing. Otherwise they would have found it, because it’s there, the hereditary. Just look at the family history.”
- She said something new that set her apart from other interviewees: “The lucky ones are those who test positive, because at least now they know. They know what they are fighting against and
know for certain what needs to be done.” We went back on that concept a few times and she did not find any relief with her results.
- She also said that she was upset to have been called “out of the blue” by a cousin she had not spoken to in years, to see if she would accept genetic testing on the family’s behalf.

**Outstanding questions**
- Tell me more of what you mean by “those who tested positive are the lucky ones?”
- Do you at least see a possibility that you may not have a gene mutation? Her answer to that was, “No, because there is a mutation. They just didn’t find it. At least the other people now know.”

**Difficulty encountered**
None. The start of the interview was easy. She knew where we were going to sit and directed me to the table. Upon beginning, I saw her looking at her husband, and he went upstairs. When alone, she began the conversation to start the interview.

**Validity of the interview**
She never contradicted herself whenever we returned to previously discussed topics. She was often spontaneous and, when she realized she was being interviewed, would lower her voice when she swore or used coarse language to describe situations.

**Interruptions during the interview**
Her dog would at times make noise with his toys and would bring the toys over to her. As well, because I am afraid of dogs, I had to ask her to make sure that the dog stayed on her side of the table. Although the dog made me nervous, his presence did not distract me during the interview.

**Other postinterview comments/observations**
Again, the time flew right by. I had not realised we had talked for over an hour and half before I first looked at my watch. Our scheduled time was up, but no more time was necessary. She had said a lot, and I don’t think I would have gotten more. She said that she quite enjoyed her participation in this research, and said that I was easy to talk to. She walked me to the door, made sure I knew my way back, and wished me a safe ride back home. She also said that she would call me later in the month to give me the names of her nieces, and the cousin who had asked her to go for genetic testing, and her daughters. Three weeks after the interview, I called her to see if she had contacted her relatives. She said she did call her cousin but had not heard from her. She said would call me if she heard anything. I did not want to push her too much, so I never called her after not receiving another call from either her or her relatives.

N.B. I would like to thank Sue Cox for sharing her field note template that had been previously prepared by Rauch, Bassett and Cox. Under each heading, Sue had also shared prompts to what could be included. For example, under the heading Interesting points/recurrent themes, prompts would include: key topics discussed, new insights or recurrent themes, reflections on whether interview conformed to expectations and if not, why not and new questions arising.
Appendix 7

Letter of invitation to participate
Dear Ms xxxxx,

First, I would like to thank you again for giving me the opportunity to explain to you my doctoral study. As discussed in our telephone conversation on August 28, I am sending you a copy of the information and consent form. I have also included a list of questions that I would like to discuss with you, should we meet for an interview. I will contact you again on Monday, September 12 to see whether or not you are interested in participating in my research.

You will see that the consent form mentions that, as part of this study, I am also interested in interviewing family members or significant others. Although having a family member participate in this study is not absolutely necessary in order for you to participate, I would appreciate if you could start thinking about who was most concerned with your results and would be valuable for me to interview as well. We will certainly discuss this subject further in our next telephone conversation. Please feel free to contact me at any time. I will be happy to answer any questions you may have concerning my study.

Here is a list of possible questions I would like to discuss, should we meet for an interview. Please note that this is a suggested list and can be expanded to include other questions or concerns of your choice. Not all of these areas need to be covered, should they make you feel uncomfortable. Please also note that there is no right or wrong answer to any of these questions, and all of your thoughts are important.

➤ What is your family history of (type of) cancer?
➤ How has (type of) cancer affected your life?
➤ What circumstances led to your decision to have genetic testing for breast and ovarian cancer susceptibility?
➤ What benefits and risks did you perceive prior to and after having had genetic testing?
➤ How did you feel when you received your results?
➤ Do you find that your genetic testing results have or have not given you new answers to explain the appearance of (type of) cancer within your family?
➤ Concerning the letter you received from the BCCA, what does their explanation of your results mean to you? For example, I am no more at risk OR still at risk; I still need to be vigilant.

I understand the sensitivity of these issues and I thank you sincerely for your time and consideration.

Christine Maheu

(yyy-yyyy)
Appendix 8

Sample initial analysis of key statements
First example
PP1 - Macy

Key statement 4:
(360-367) (smack of the lips) Hmm, grandmother, and mother. Grandmother was, I think in her 70s, my mom was 50, first cousin was 50, I have another first cousin who’s dealing with it at the moment and me. That’s five in what, one, two generations? Is it environmental? I don’t know? Is it in our genes? Is it environmental, something kicks in? I don’t know but I feel really sure. My 19-year-old nephew, he doesn’t have breast cancer, but it is cancer. Uh, 19-year-old nephew and he just passed away. Oh, ya, I’m sure there’s something. I have no doubt about it.

Description:
She feels strongly that there is something more, because when she looks at her family history, there are just too many cancers there. As she puts it, apart from having a genetic predisposition, there is nothing else that could explain all the cancers in her family. There is her grandmother and her mother: her grandmother was in her 70s, her mom in her 50, her first cousin was 50, and now she as another first cousin who is dealing with breast cancer at the moment, and her. As she says, there are five people in two generations. She wonders if the reason lies in her genes or an interaction between her genes and the environment. And then there is her 19-year-old nephew. He has just passed away from osteo-sarcoma. Yes, she has no doubt there is something there.

Interpretation:
The conceptual structure informing Macy’s belief that there is most likely a genetic predisposition in her family is the number of cases of cancer in her family. There are just too many people with cancers to not think that there might be a possibility that they have an inherited mutation. Certainly Macy is aware that a family would not have such a strong history of cancer if all the cancers occurred by chance.

Second example
PP5 - Evelyn

Key statement 10:
I probably fit in the familial, somewhere in the middle – maybe a recessive gene that has not been located yet. That would make sense, considering cancer is on both sides of the family. But then, that would put my sisters at 25% risk, instead of 50%. But then, I was diagnosed with two primary breast cancer and all before menopause.

Description:
Evelyn has three different types of cancer in her family history, on the maternal and paternal sides. There are three cases of breast cancer on her maternal side, including herself, with two bilateral primaries and one aunt with both breast cancer and melanoma. On her father’s side, Evelyn’s grandmother had breast cancer, while her father and uncle died of oesophageal cancer. The uncle’s daughter developed fibrosarcoma at the age of 20. Having cancer from both sides of the family explains Evelyn’s view that she may carry a recessive gene not yet identified.

Trying to see if Evelyn had considered all the possible options given to her in the letter received from the Agency explaining how her results could be interpreted, I wanted to see if she had at least considered the last option: “You do not have an inherited breast/ovarian cancer mutation, which means that your cancer diagnosis may have occurred by chance.” And so I asked her, “Evelyn, do you at least have some peace of mind that the Agency is saying you may not have a mutation in your family?” No, because I probably fit in the familial somewhere in the middle, maybe a recessive gene that has not been located yet. That would make sense.
considering cancer is on both side of the family. But then, that would put my sisters at twenty-five percent risk instead of fifty percent risk, so yes.

**Interpretation:**
This statement contains an interpretation as to why Evelyn does not consider the breast cancer in her family to have occurred by chance. She invokes the different etiology of cancer (hereditary and spurious) and the theory of inheritance (autosomal dominant versus recessive gene).

Etiology theory says that if the cancer is spurious, it is unlikely that an individual will have more than one primary breast cancer. If cancer is not inherited, there is less likelihood of seeing breast cancer in male in the family history. Evelyn has both in her personal and family history. As well, the theory of inheritance says that BRCA1 and BRCA2 mutations are transmitted in an autosomal-dominant fashion and hence, cases of cancers usually occur on the same side of the family, unless both sides carry an inherited mutation. Evelyn’s family history shows cases of breast cancer from both sides of her family. She is pragmatically using the theory of autosomal-dominant inheritance to explain why she may believe that the cancers in her family do not come from these two autosomal-dominant gene mutations, but most likely from a recessive gene.
Appendix 9

Sample ongoing data analysis
The centrality of temporal view of self with a probable inherited mutation: Structures observed to organise the three types of interpretation

OR  Maybe we do have OR maybe we do not have an inherited mutation

• Cases of cancer at a young age and physical/emotional/geographical proximity of each case
• Premenopausal bca = likely mutation
• PP looking for an explanation of early diagnosis of breast cancer
• Number of cases of ca (esp. when all siblings diagnosed; too many to be coincidental)
• Presence of second primary cancer or recurrence
• No ovarian cancer
• Length of time between cancer cases
• Time between genetic testing and receiving results
• Cancer comes down through males
• Similar physical and behavioural characteristics among individuals with breast or ovarian cancer
• Very few survived breast/ovarian cancer
• Seeing somewhat of a pattern of inheritance
• Assess self rather than family history
• Current testing could not test for their specific mutation characteristics: mutations passed down from paternal side
• Sees possibility that mutation could be in the untested portion of BRCA2
• Met eligibility criteria demonstrating increased likelihood of an inherited mutation
• Letter received from Cancer Agency raises possibility that they could have a mutation
• Doubts ability of current technology to detect all mutations, especially when strongly believes they have a mutation, and therefore the technology should have been able to detect it
• Have a mutation that need specific interaction with the environment

AND  We do NOT have the genetic type of cancer

• Little to no contact with second degree relatives (lives out of Country)
• Not seeing a pattern of inheritance
• No ovarian cancer
• Late-onset breast cancer
• Assess self rather than family history
• Viewing breast cancer from paternal side as lower probability of brca1/2 mutations
• Have younger children; Have no daughter

BUT  We do have a mutation, even if not in BRCA1/2

• Too many breast/ovarian cancers to be coincidental, too many deaths from cancer, too many young people with cancer
• Seeing a pattern of inheritance across generations as within own generation
• Current technology could not test for their specific mutation characteristics: mutations passed down from paternal side, specific to another ancestral lineage, their targets only one organ
• Met eligibility criteria demonstrating increased likelihood of an inherited mutation
• Letter received from Cancer Agency raises possibility that they could have a mutation
• Doubts accuracy of results: sees results as incomplete - not all of BRCA2 was tested; were told more complete testing is unavailable in the U.S.
• Doubts ability of current technology to detect all mutations
• Have a mutation that need specific interaction with the environment
• Could be a mutation of a third breast cancer gene
Appendix 10

Demographic questionnaire
1. How often do you have mammograms? once/year__, twice/year__, other__
2. How often do you have clinical breast exams? every #months__, once/year
3. How often do you do a breast self-exam? q month__, every #months__, once/year
4. What year was your genetic test?
5. At what age were you diagnosed with breast cancer? _____ Any recurrence or a second primary ____________
6. How old are you now? ______
7. Number of daughters____ Age(s)____________________
8. Number of sons __________ Age(s)__________________
9. How many sisters? ____ Any diagnose with breast or ovarian cancer? ____________
10. How many brothers? ______ Any diagnose with cancer? ______ Type ______
11. Do they have any children? Ages ________________
12. Are you of Ashkenazi Jewish decent? Yes__ No__
13. What is your ethnicity? __________________________
14. On your mother's side, who in the family has or has had cancer?
   - Your mother, Yes____ No ____ Type _____________
   - Deceased? Yes____ No __
   - Did she die of breast cancer? ______
   - Your mother’s father: Cancer? Yes____ No ____ Type __________
   - Your mother’s sisters: How many?____ BCA Yes____ No __, Other___________
   - Your mother’s brothers: How many?____ BCA Yes____ No __, Other___________
   - Your mother’s cousins: How many?____ BCA Yes____ No __, Other___________
   - Your mother’s aunt: How many?____ BCA Yes____ No __, Other___________
   - Your mother’s uncle: How many BCA Yes____ No __, Other___________
15. On your father’s side, who in his family has or has had cancer?
   Your father’s Mother: BCA Yes____ No __, Other________________
   Your father: Was he diagnosed with cancer? Yes____ No __
   Your father’s sisters: How many____ BCA Yes____ No __, Other________________
   Your father’s brothers: How many____ BCA Yes____ No __, Other________________
   Your father’s cousins: How many____ BCA Yes____ No __, Other________________
16. Did any of your family members die of cancer? Yes ___ No ___.
   Who? __________________ Type _______

17. At the time of your diagnosis, where did your family members live?
   ________________________________

18. Marital status:
   single ___, married ___, divorced ___, common-law ___, widowed ___

19. Do you plan to have (or have more) children? Yes ___ No ___

20. Level of education:
   Less than high school ___, high school diploma ___, college ___, university ___

21. Employment status:
   Full time ___, part time ___, retired ___, unemployed ___, unemployed by choice ___

22. Residence: Greater Vancouver _______, Fraser Valley _______

**DEMOGRAPHIC QUESTIONNAIRE – SECONDARY PARTICIPANTS**

1. Relationship to primary participant: _______________________

2. Current age: _______

3. Number of siblings and their ages: _______________________

4. Who in your family has had breast cancer? Sit on your mother’s or father’s side? ____
Appendix 11

Transcript segments from a few interviews
First example
Yeah, so it was a 100%; 100% in our family in the last three generations, every woman. Now there have only been four women in three generations. My grandmother was the only girl. My mother and her sister were the only girls and I am the only girl. We have all had it, every girl. So okay, yeah, every girl in the last three generations has had breast cancer. (Stephanie, PP12, field conversation, July 18, 2001. Interprets her genetic test results as confirmation of mutation carrier status for self and her family but that, for the time being, this mutation has not been detected)

Second example
Genetic testing to me is just extra ammunition, just as having your yearly mammograms. I was not too worried about having the gene. I feel our kids are going to be so much better informed to help deal with the eventuality that there might be a gene... The testing was still worthwhile, even if no answers were obtained from the results, because at least the results gave me more information, more insights into the possible causes of the breast cancer... I have thought about going to the States to have further testing, because apparently the test they did on my blood only represents a certain fraction; it doesn’t test 100%. I probably would do it, just to have more information for the kids. It wouldn’t be worthwhile just for me, as I no longer have breast tissue or my ovaries. (Beatrice, PP6, field conversation, April 19, 2001. Interprets her genetic test results as ambiguity regarding mutation carrier status.)

Third example
When I was at the clinic, um, receiving – I think it was during one of my follow-ups or maybe while I was having my treatments – the oncologist asked me if I would ever be interested in genetics, and if I’d be willing to let them do some blood testing. And I said sure. Like I said, I wanted to know as much as I possibly could and I was interested in finding, you know, the answers. And so I said I would be interested, and it was that... I never thought about the negative results or, I mean the positive results I guess you’d want to say, but I, I guess I was just hopeful that I, that everything would be great. But at the same time like you said, if I don’t have those genes, why did I get it? In some ways it would have almost have been better to say, okay, you’ve got the gene, that’s why you got it. You know, because that’s a big question, why did I get it and if it is environmental or diet, am I still doing something wrong? And like I think about that a lot... But when I received the results, I felt relieved when she told me that I didn’t have it. I felt good about that, I felt positive. I didn’t feel, uh, and I don’t know if I questioned it any more than that. I think I have more or less arrived at a place where I feel like, okay, I’m going to be okay. (PP10, Erica, field conversation, July 9, 2001. Interprets her genetic testing results as refutation of mutation carrier status to mean “I don’t have the genetic type of cancer” and sees her breast cancer as being the consequence of environmental exposure.)

Fourth example
That doesn’t say that I am totally free of it; just that they couldn’t find anything. BRCA1 and 2 are just one type of genetic mutation. And maybe they weren’t able to find it because of their limited ability. But, if I really don’t have those two, it doesn’t mean I don’t have something else. (Ginger, PP11, field conversation, July 5, 2001. Interprets her genetic testing results as ambiguity regarding her mutation carrier status. She sees her results as inconclusive.)
Appendix 12

Database record with dummy data
### Primary Participant Record

<table>
<thead>
<tr>
<th>Part ID</th>
<th>Name</th>
<th>Last Name</th>
<th>Code Name</th>
<th>Secondary Name</th>
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</thead>
<tbody>
<tr>
<td>104</td>
<td>Lady Di</td>
<td>Spencer</td>
<td>Diana</td>
<td>Prince Charles</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th></th>
<th></th>
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<th></th>
<th></th>
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<th></th>
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</tr>
</thead>
<tbody>
<tr>
<td>Time</td>
<td>09:00</td>
<td></td>
<td></td>
<td>Time</td>
<td>05/21/04</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Place</td>
<td>At the Palace</td>
<td></td>
<td></td>
<td>Place</td>
<td>At the Palace</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

#### Phone Calls List

<table>
<thead>
<tr>
<th>Date</th>
<th>Time</th>
<th>Call Summary</th>
</tr>
</thead>
<tbody>
<tr>
<td>5/21/2004</td>
<td>08:45</td>
<td>Called Lady Di from my car with my cell phone to tell her that the guards at the gate did not want to let me in. She gave me a password to tell them which was “Firestone” and they let me in immediately. I thank her kindly and told her that I was looking forward in meeting her in a few minutes.</td>
</tr>
<tr>
<td>5/15/2004</td>
<td>14:00</td>
<td>Called Lady Di back. She said that she has read the consent form and is still quite interested to participate. She even asked for her husband to participate and he has agreed. We have converge that I would do both interviews on the same day but at different times. The interview is set for May 21, 2004. Lady Di has kindly given me detailed information on how to get to the Palace.</td>
</tr>
<tr>
<td>5/5/2004</td>
<td>13:00</td>
<td>Called Lady Di. Explained that I had received her name from her Cancer Agency where she had her genetic testing and that she had informed the personnel that she would be interested to participate in my research. She said she was still interested and was much</td>
</tr>
</tbody>
</table>

#### To Do List

<table>
<thead>
<tr>
<th>Date</th>
<th>Time</th>
<th>To Do</th>
</tr>
</thead>
<tbody>
<tr>
<td>5/21/2004</td>
<td></td>
<td>Send thank you card to both Lady Di and Prince Charles for their participation</td>
</tr>
<tr>
<td>5/5/2004</td>
<td></td>
<td>Send letter of information and consent form along with invitation letter. Send by express mail.</td>
</tr>
</tbody>
</table>

#### Field Notes

- **DATE/TIME/PLACE OF INTERVIEW**
  - Friday, May 21, 2004 from 09:00 AM to 10:30 AM
- **FIELD NOTES WRITTEN**
  - Friday, May 21, 2004 10:00 PM
- **NOTES ON INITIAL CONTACT**
  - I was first greeted by the Palace guards who required that I show identification paper. Afterwards, I was directed down a hall to a big blue powered door. Another guard opened the door and inside I saw Lady Di sitting at a sofa drinking tea. She came to greet me at the door with a smile. She said she was very interested in my work and was much looking forward to this interview.
- **INFORMATION DESCRIPTION**
  - Young beautiful woman with blond hair and light blue eyes in early thirties. She was