ADDRESSING ACCESS TO HUNTINGTON DISEASE PREDICTIVE TESTING

by

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ABSTRACT

Genetic knowledge holds great promise in terms of health benefits, y*et al*so raises challenges regarding the delivery of beneficial testing and services. Addressing this challenge is especially important in rural areas where lack of access to clinical genetics is pervasive, resulting in considerable inequities in service availability. The purpose of this research study is to explore the hypothesis that a novel telehealth strategy for delivering predictive testing (PT) for Huntington disease (HD) can address the potential inequity that exists in access to PT in rural communities in British Columbia.

To address the hypothesis, the project employed a three part, highly structured, mixed method sequential exploratory approach. The first part of the research involved: a) a mapping study; b) a qualitative interview study of 33 at-risk individuals; c) a survey of 102 individuals at-risk for HD; and d) an 11-person expert workshop. The second part of the research built on results from the prior work and was to develop a telehealth PT protocol and a HD PT dedicated website for individuals considering testing. The final component of the research involved a pilot project that compared the novel telehealth protocol with the standard, Vancouver-based PT protocol. Evaluation of the pilot project was conducted by quantitative survey with 28 participants and was subject to statistical analysis.

Results revealed that PT rates are lower in rural areas and that access is a significant issue due to distance related factors and the inflexible nature of the PT process. The pilot project demonstrated that providing PT via telehealth is not only possible, but is also warranted. There were no significant differences in terms of quality of care, information, counseling and support

during the PT process between the Vancouver-tested and telehealth-tested groups. Overall, the pilot study reveals that providing PT via telehealth can improve access to PT while maintaining high quality of care and support. The work adds to a growing body of literature on the utility of telehealth services in genomic medicine in an age of increased technological innovation and comfort with such communication mechanisms.

PREFACE

This thesis is predominantly manuscript based. The first two chapters provide introductory and background information and the subsequent chapters are manuscript based papers that have either been published, or submitted for publication in the peer reviewed literature.

Research Ethics Board approval was obtained for this project. The research presented in chapters five and six, involving interviews with human subjects, received ethics approval from the University of British Columbia (UBC) Behavioural Research Ethics Board (certificate number: H10-00656). The research presented in chapter seven, involving a pilot study, received approval from the UBC Children's and Women's Research Ethics Board (certificate number H11-00033).

Chapter 3: A grand challenge: Providing benefits of clinical genetics to those in need

I developed the initial idea for this manuscript and wrote the first draft. Michael Hayden advised in the presentation and writing of subsequent versions of the manuscript. A version of this chapter has been published as: Hawkins, AK & Hayden, MR. A grand challenge: Providing benefits of clinical genetics to those in need. *Genetics in Medicine* 2011; 13: 197.

Chapter 4: Lessons from predictive testing for Huntington disease: 25 years on

I developed the initial idea for this manuscript and wrote the first draft. Michael Hayden advised on the theoretical background and literature for the manuscript. Anita Ho advised on the ethical theories and concepts discussed. A version of this chapter has been published as: Hawkins, AK,

Ho, A & Hayden, MR. Lessons from predictive testing for Huntington disease: 25 years on. *J Med Genet* 2011; 48: 649-650.

Chapter 5: When access is an issue: Exploring barriers to predictive testing for Huntington Disease in British Columbia, Canada

The initial concept and design of the study was conceived by me with the support of Michael Hayden. I developed the interview guides, obtained research ethics approval, and conducted all of the interviews for the study contained in the manuscript. Susan Creighton, Susan Tolley and Joji Decolongon assisted in the recruitment of the interview participants. I analyzed the data from each of the interview transcripts, developed the findings and results from the survey, generated the figures and wrote the manuscript. Michael Hayden and Susan Creighton advised on the data analysis and writing of this manuscript. A version of this manuscript has been accepted for publication as follows: Hawkins, AK, Creighton, S & Hayden, MR. When access is an issue: Exploring barriers to predictive testing for Huntington disease in British Columbia, Canada. *Eur J Hum Genet* 2012 Advanced Online publication: http://www.nature.com/doifinder/10.1038/ejhg.2012.147

Chapter 6: Developing a comprehensive, effective patient friendly website to enhance decision-making in predictive testing for Huntington disease

I conceived the initial design for the website and the study including the pilot test instruments and survey administration described in this chapter with support of my supervisor, Michael Hayden. I developed the interview guides, obtained research ethics approval and conducted all of the interviews for the study contained in the manuscript. Input on the website development,

content and diagrams was given by B'stro website developers[1], Susan Creighton and The Huntington Society of Canada. I analyzed the data from each of the interview transcripts, developed the website framework and diagrams with the assistance of Michael Hayden and the B'stro website development team. I generated the figures and wrote the manuscript. Michael Hayden advised on the writing of this manuscript. A version of this manuscript has been submitted for publication as follows: Hawkins, AK, Creighton, S, & Hayden, MR. Developing a comprehensive, effective patient friendly website to enhance decision-making in predictive testing for Huntington disease.

Chapter 7: Providing predictive testing for Huntington disease via telehealth: Results of a pilot study in British Columbia, Canada

The initial concept for the study described in this chapter was conceived by me and Michael Hayden and was subsequently developed in collaboration with my supervisory committee (Anita Ho, Bruce McManus, Michael Hayden) and Susan Creighton. I developed the telehealth protocol with the assistance of Michael Hayden, Susan Creighton, and Robi Blumenstein. I obtained research ethics board approval and developed, adapted, pretested and piloted each of the survey instruments. I recruited and consented individuals, scheduled all appointments, and sent out all survey tools. I performed all of the qualitative and quantitative data analysis, generated the tables and figures and wrote the manuscript. Susan Creighton and the Huntington Society of Canada assisted in the recruitment of survey participants. A version of this chapter has been submitted for publication as follows: Hawkins, AK, Creighton, S, Ho, A, McManus, B & Hayden, MR.

[1] B'stro are a professional website development firm based in Vancouver

Providing predictive testing for Huntington disease via telehealth: results of a pilot study in British Columbia, Canada.

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LIST OF ABBREVIATIONS

BC	British Columbia
BDI	Beck Depression Inventory
CAG	Cytosine-adenine-guanine nucleotide repeat
DNA	Deoxyribonucleic acid
FH	Family history
FU	Follow up
GC	Genetic counselor
HCP	Healthcare provider
HD	Huntington disease
HD+	Persons with CAG expansion
HD-	Persons without CAG expansion
NSGC	National Society of Genetic Counselors
NT	Not tested: persons that have chosen not to test for the CAG expansion
NA	No answer
OR	Odds ratio
Psych	Psychological
PT	Predictive testing
Qual	Qualitative
Quan	Quantitative
Rk	Risk
SD	Standard deviation
TT	Telehealth tested (persons who underwent PT via the telehealth protocol)

UK	United Kingdom
US	United States
VT	Vancouver tested (persons who underwent PT via the standard Vancouver-based protocol)

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Firstly I would like to thank my supervisor and supervisory committee for their unwavering support and time. As a supervisor Michael Hayden has urged me to be an innovative thinker and dynamic researcher, to push boundaries and not be afraid of taking a new or unpopular stance, as long as it is one you believe in. Anita Ho has provided mentorship and time in both my research and professional development, and has made my PhD a rich and rewarding experience. Bruce McManus has championed me and my work from the outset, and his sage advice, enthusiasm and ability to think of my work in the bigger context have been invaluable. I would also like to thank Hillel Goelman from the Interdisciplinary Studies Graduate Program for providing guidance at crucial times during my research journey.

I am grateful to Robi Blumenstein and CHDI for providing not only funding but also insights and input toward the development and translation of this research. I am also grateful for the personal financial support that has allowed me to pursue my doctoral training: Pfizer Ripples of Hope, the Child and Family Research Institute, the University of British Columbia, and the Canadian Institute of Health Research Ethics of Health Research and Policy Training Program.

This research would not have been accomplished without the tireless support of Susan Creighton who has dedicated considerable time and effort in helping me complete this project. I am also indebted to the assistance of a number of other key individuals: Susan Tolley, Joji Decolongon, Mark Guttman, Bev Heim-Myers and the Huntington Society of Canada, and members of the Hayden laboratory. Ruth Milner, Sema Aydede and Hao Luo each provided expert technical advice during different phases of the project for which I am grateful. I am

thankful for the support, time and experience of Holly Longstaff and Nina Preto who listened to my ideas, helped me work through problems and navigate the entire doctoral process.

I would also like to express my sincere appreciation to the families who generously participated in different stages of the research project.

Finally I would like to thank my incredible family. My parents, James and Flick, for supporting me through all my learning endeavors to satisfy a ‘curiouser and curiouser’ developing mind, and Molly, Sam, Anisha, Yasmin and Amir for their love, kindness and enthusiasm over the last four years. And lastly my wonderful, loving husband, Sean, without whom this would not have been possible. His constant encouragement, steadfast belief in me and patience has been my foundation throughout this entire process. Thank you all.

DEDICATION

For my parents

1. INTRODUCTION TO HUNTINGTON DISEASE AND PREDICTIVE TESTING

1.1 Access to genetic services

In very general terms, this research seeks to explore the ethical and social implications of variability in access to genetic services in rural communities. This aim is motivated by an increasing shift towards genomic based healthcare with the promise of personalized medicine, defined as a *"tailored approach to patient treatment, based on the molecular analysis of genes, proteins and metabolites"* (Davis, *et al*., 2009, p279). It is hoped that personalized medicine will lead to better disease prediction and prevention, improve treatment and decrease adverse drug reactions (Personalized Medicine Coalition, 2009). While personalized medicine holds great promise in the healthcare arena, it also raises significant challenges. One of the most prominent of these issues relates to the translation of genetic knowledge into actual delivery of genetic tests and services. This challenge is particularly acute in rural and remote areas, where lack of access to genetic services is a persistent issue. Access is of particular relevance for a devastating neurodegenerative disorder such as Huntington disease (HD), in which current testing practices often require multiple visits to a large city. Such testing regimes may be a disincentive to accessing testing, treatment and support.

1.2 Background

1.2.1 Access to genetic services in rural communities

"Most healthcare professionals have insufficient knowledge about genomic medicine and there are too few geneticists and

genetic counselors."

(National Human Genome Research Institute, 2006)

Genetics represents a specialized, novel and developing field of healthcare[2]. Due to this fact, and a limited number of geneticists and genetic counselling training programs[3], there is a limited supply of genetics professionals (geneticists, genetic counsellors and genetic nurses), even in countries with the most developed genetic services[4]. Compounding this issue is the paucity of access to genetic services in rural areas[5]. On a broad level, access to healthcare in rural areas represents a worldwide concern since half the population lives in rural areas (UN Department of Economic and Social Affairs Population Division, 2005). While these rates are much lower in developed countries, this does not translate into access to specialized healthcare services, such as genetics. Access and proximity to genetics services is usually confined to a

[2] This background discussion focuses primarily on examples from North America (US and Canada) and secondarily from other English-speaking developed nations such as the UK and Australia. The focus has been chosen for a number of reasons: 1) Genetics, genetic services provision and genetic training programs were first established in these areas and as such are more developed than in other areas of the world; 2) there is easier access and availability to data from these countries; and 3) the author's scope of work and research is focused in North America.

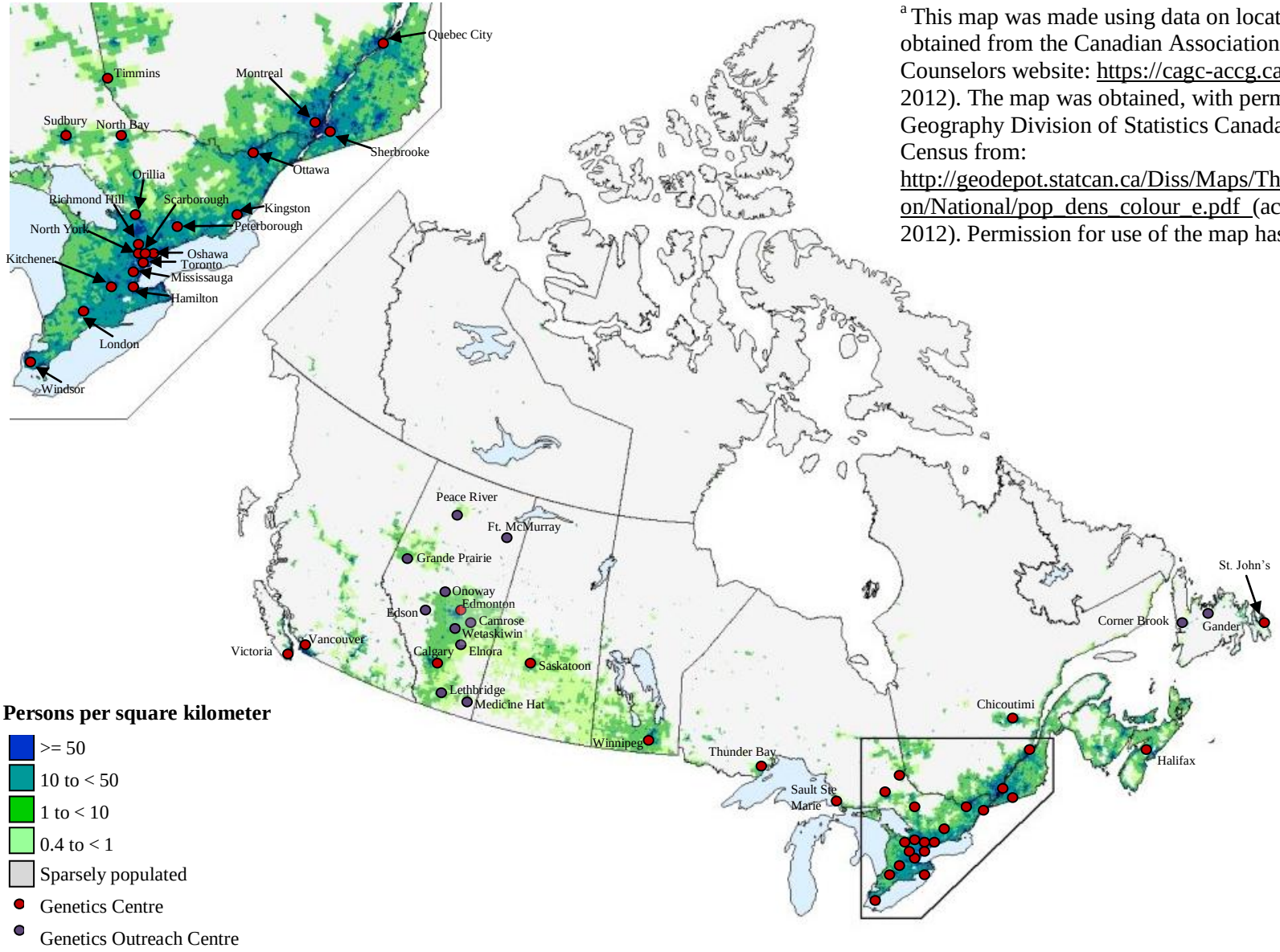
[3] For example there are currently 29 genetic counselling training programs in the US, 5 in Australia, 2 in the United Kingdom and 3 in Canada (Genetics Education Center, 2012)

[4] For example, there are only an estimated 240 registered genetic counsellors in Canada (Canadian Association of Genetic Counsellors, 2010), 2440 practicing genetic counsellors in the US (National Society of Genetic Counselors, 2008) and approximately 300 genetic counsellors in the UK (Association of Genetic Nurses and Counsellors, 2010).

[5] Due to national and regional differences in rural and urban areas, the precise definition of the term 'rural' remains problematic (UN Statistical Division, 2012). For the purposes of this discussion, the term rural refers to areas which do not have easy access to the services typically found in large urban areas. In terms of access to genetic services, this may vary between different jurisdictions. In British Columbia, where this research took place, there is one predictive testing centre for HD in the whole province. As such, populations more than two hours drive from this centre were considered rural, even if they lived in a somewhat urban setting. This is because these populations were not able to easily access the services of the predictive genetic testing clinic, the main focus of this research.

limited number of large academic medical centres (Washington State Department of Health, 2008). The geographic access issue in the United States is illustrated by National Society of Genetic Counselor (NSGC) survey data that reveals there are at least five US states, such as Wyoming and the Dakotas, in which there are no reported genetic counsellors, and at least a further ten states in which there are less than five reported genetic counsellors for the entire state (National Society of Genetic Counselors, 2008). Unfortunately data are not currently available for Canada, where there are fewer than 20 new genetic counselors graduating per year (Canadian Association of Genetic Counselors, 2012). Nonetheless, given the population distribution and sheer size of Canada, it is reasonable to assume that some sectors of the population, particularly those in the North, have difficulty in accessing genetic services which are focused in large academic medical centres that are primarily located in the more densely populated southern regions of the country.

Figure 1.1: Location of genetic centres and population distribution in Canada[a]



[a] This map was made using data on location of genetic centres obtained from the Canadian Association of Genetic Counselors website: https://cagc-accg.ca/ (accessed June 6th 2012). The map was obtained, with permission, from the Geography Division of Statistics Canada, based on the 2001 Census from: http://geodepot.statcan.ca/Diss/Maps/ThematicMaps/population/National/pop_dens_colour_e.pdf (accessed June 2nd 2012). Permission for use of the map has been obtained.

The large gaps in genetic service coverage do represent a significant issue, in terms of travel time and distance, as well as a social and financial burden, for those who currently require genetic counselling and other genetic services (d'Agincourt-Canning, 2004). This access issue is of even greater concern in terms of the future of genetic medicine: Large segments of the population have no genetic services and will therefore be ill equipped to deal with advances in technology, treatment and education for medical conditions with a genetic basis. Moreover, a lack of genetic knowledge among family physicians, who are the primary, and sometimes only, healthcare providers in these remote geographic areas, (Frueh & Gurwitz, 2004) may complicate this issue even further.

A review of available data and literature thus suggests that access to genetic services is limited, at least in terms of proximity, particularly in rural communities. However, the fundamental concern here is the actual impact of this limited access to genetic services on the affected communities. Unfortunately this remains difficult to assess due to lack of data and research in this arena, undoubtedly in part due to the novelty of the field. Nonetheless, genetic knowledge is thought to have multiple *potential* benefits for human health through the prevention, diagnosis and treatment of disease (Collins, *et al*., 2003; Hunter, 2005). Researchers hope that the elucidation of genetic information can be translated into meaningful clinical benefit via: new therapeutic targets, leading to new therapies for treatment or prevention of disease; decreasing adverse drug reactions; allowing identification of biomarkers that can improve disease prediction and monitoring of disease progression and response to therapy; elucidating environmental components of disease and allowing public health interventions; and more personalized approaches to disease management (McCarthy, *et al*., 2008).

In summary, although current research on the translation of genetics into healthcare practice is somewhat limited, genetic knowledge does, nonetheless, appear to have an important *potential* role in these areas in terms of how we diagnose, prevent, treat and practice medicine (Khoury, *et al*., 2008). The exact mechanisms by which laboratory successes will be translated into the clinical arena remain somewhat unclear, although there appears to be future utility in the areas of personalized medicine and pharmacogenomics, susceptibility and immunity, diagnosis and treatment protocols and preventative strategies (Hamburg & Collins, 2010). However, if access to genetic services is limited, one can speculate that these potential benefits may not be realised, particularly in remote areas where there is little or no availability of genetic services. Collins and colleagues highlight this issue as one of the 'grand challenges' of genomics:

> *"To ensure that genomics research benefits all, it will be critical to examine how genomics-based healthcare is accessed and used. What are the barriers to equitable access, and how can they be removed? This is relevant not only in resource-poor nations, but also in wealthier countries where segments of society, such as indigenous populations, the uninsured, or rural and inner city communities, have traditionally not received adequate healthcare."*
>
> (Collins *et al*., 2003, p843)

The problem of lack of access to genetic services is likely to increase as novel technologies and genetic therapies become available. There has also been a sharp decline in the

cost of such technologies, such as human genome sequencing[6]. Decreasing costs, together with the rapid advancements in technologies and therapies, means that going forward there is likely to be an improvement in the cost-benefit impact of genetic knowledge in the healthcare arena. However, unless there is research into this area and into the impact of access issues to genetic services, advances in genetics may lead to wider and wider discrepancies both within and between countries.

The above discussion has highlighted two important issues, 1) the realized and potential utility of genetic technologies in the healthcare system, and 2) the apparent lack of access to such services, particularly in rural areas, due to both a lack of knowledge about genetics among primary healthcare providers as well as a limited supply and distribution of genetic professionals. Recent literature has suggested that one method to address this issue would be to use telemedicine, whereby genetic services, education and counseling are provided remotely. Until this point, telemedicine techniques have primarily been used in the area of cancer genetics, and it appears that such approaches can be successful in terms of improving access and producing patient and health system cost-savings (Baumanis *et al*., 2009; Coelho *et al*., 2005).

1.3 Huntington disease predictive testing

HD is a devastating, progressive neurological condition without treatment, inherited in an autosomal dominant pattern. The disease, which on average begins to manifest in the fourth or fifth decade, is characterised by cognitive, motor and behavioural decline resulting in death usually ten to fifteen years after first onset (Hayden, 1981; Walker, 2007). The disorder is caused

[6] Human Genome Sequencing has decreased from $2.3 million USD per sequence in 2003, to $1,000 USD in 2012 (Lauerman, 2009; Markoff, 2012).

by a CAG triplet repeat expansion mutation in the huntingtin gene (HTT), located on chromosome four (MacDonald, *et al*., 1993). Due to its inherited nature, first degree relatives of those affected by the disease have a 50% chance of also developing the condition. Age of onset and progression may vary considerably among individuals family members due to length of the CAG expansion as well as other genetic and environmental factors (Andrew, *et al*., 1993; Rosenblatt, *et al*., 2006; Wexler, 2004).

Predictive testing (PT) for the disorder has been available via linkage analysis since 1986, and by direct mutation analysis since 1993 (Benjamin *et al*., 1994; Kremer, *et al*., 1994). Although there is no treatment for the disorder, PT affords individuals at risk for the disorder benefits across multiple levels including:

- Reproductive planning
- Relieving uncertainty
- Life planning (such as job choice and insurance)
- Access to services and support

(Binedell, Soldan, & Harper, 1998; Harper, Lim, & Craufurd, 2000; Kessler, *et al*., 1987; Wiggins, *et al*., 1992).

1.3.1 The predictive testing process

The decision to proceed with PT may be dynamic and unfold over time, or it may be more automatic or triggered by a particular event (such as getting married or making career choices) (Etchegary, 2006; Smith *et al*., 2002; Cox, 2003). It is shaped by clinical and socio-political contexts such as cost, availability and potential for genetic discrimination, as well as

other personal and familial factors (Bombard, *et al*., 2009; Taylor, 2004). Providing adequate support during this process entails non-directive counseling that allows individuals to consider the pros and cons of such testing, and make up their own minds as to whether testing is right for them (Elwyn, Gray, & Clarke, 2000; Harper, 1988). To aid in the provision of such non-directed supported decision-making, at risk individuals should be provided with unbiased information on the testing process and the ramifications of receiving results (Benjamin *et al*., 1994). A series of one-on-one genetic counseling and education sessions with an appropriately trained professional is a fundamental part of the informed consent process in PT, as is appropriate follow-up counseling after results are given (Green, *et al*., 2001). As such, HD PT guidelines recommend a process involving four distinct phases. The standard PT process is outlined in Figure 1.2.

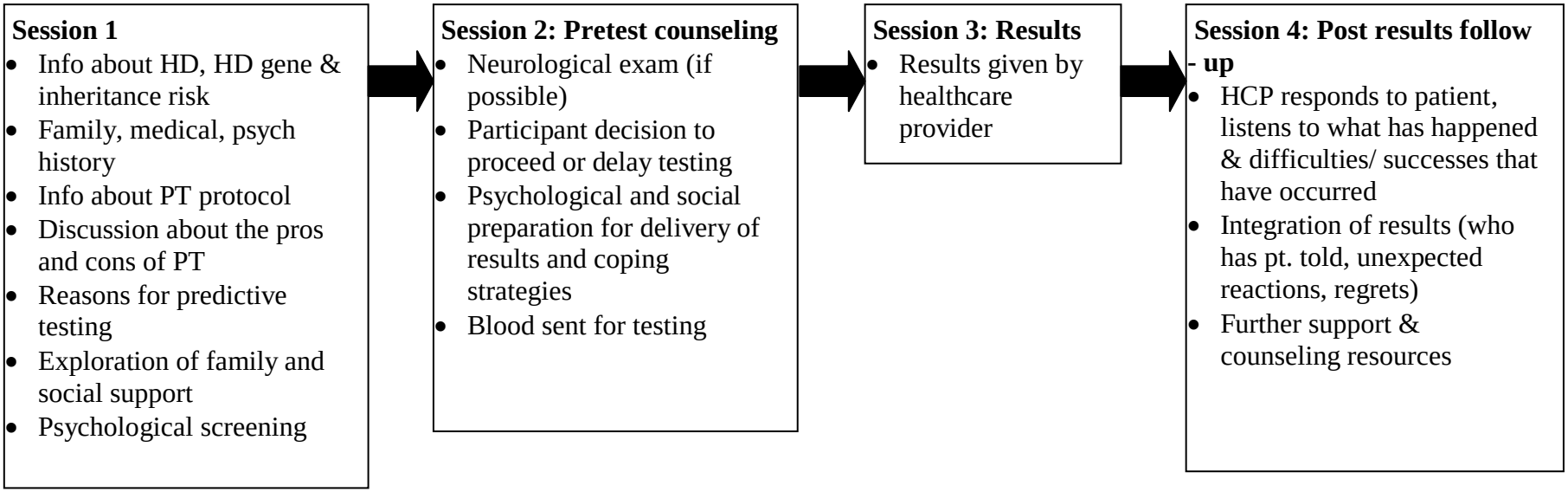


Figure 1.2: The stages of the Huntington disease predictive testing process

The Centre for HD in Vancouver serves patients from all over the province of British Columbia and Yukon Territory. In general, each of the first three sessions are conducted in-person and the final post results session is conducted via phone, or in person, depending on the patient's preference. In recognition of the difficulties of travelling to the Vancouver-based HD testing centre, the protocol has also been somewhat modified in recent years for those patients from rural areas. After attending their first visit in person at the UBC HD centre in Vancouver, these individuals are given the option of completing the PT process in their local community with the support of their primary healthcare provider (HCP). In these instances, their local HCP is provided with phone-based training and support, as well as written materials from the Vancouver-based HD PT team. The local HCP then conducts the pre-test counseling and results session in person with the patient, and acts as a local support for any further questions or follow-up needs.

1.3.2 Huntington disease predictive testing uptake

Testing uptake is defined as the number of individuals who have undergone PT as a proportion of the number of individuals at 50% risk for developing the disorder and is expressed as a percentage (Tassicker *et al*., 2008). PT uptake rates vary widely with estimates ranging from 3-25% (Bernhardt, Schwan, Kraus, Epplen, & Kunstmann, 2008; Craufurd, Kerzin-Storrar, Dodge, & Harris, 1989; Creighton, *et al*., 2003; Harper, Lim & Craufurd, 2000; Laccone, *et al*., 1999; Maat-Kievit, *et al*., 2000; Morrison, Harding-Lester, & Bradley, 2011; Tassicker, *et al*., 2008) (see Table 1.1 for more details on PT uptake rates worldwide). These rates vary considerably which may reflect the availability of testing, knowledge of testing, social, economic

and cultural factors (i.e., health insurance coverage and region-specific anti-discrimination laws, as well as the PT uptake calculation method used[7]).

Table 1.1: Predictive testing uptake rates worldwide

Country	PT uptake rate	Study year and author
Canada	21%	(Creighton, *et al*., 2003)
United Kingdom	18%	(Harper, Lim & Craufurd, 2000)
The Netherlands	24%	(Maat-Kievit, *et al*., 2000)
France	5%	(Goizet, Lesca, & Dürr, 2002)
Germany, Austria, Switzerland	<3-4%	(Laccone, *et al*., 1999)
Australia	13.0-15.4%	(Tassicker, *et al*., 2008)
South Africa	4.5%	(Futter, Heckmann, & Greenberg, 2009)
Northern Ireland	12.3–14.6%	(Morrison, *et al*., 2011)

While PT should be a carefully considered choice, these rates are lower than expected (65-80% of at risk individuals indicated they would pursue such testing prior to the availability of the test) (Hayden, 2000; Meissen & Berchek, 1987). This suggests disincentives to testing prevent at risk individuals being able to realise the potential benefits of PT. Furthermore, low uptake rates limit the number of individuals who may become aware of and possibly involved in potential research opportunities in HD research. Clinical trials for HD often require individuals who know their HD status, so low PT rates may limit the available pool of potential research participants. Such trials may ultimately lead to future treatments and improvements in care.

[7] PT uptake rates were traditionally calculated by dividing the number of people affected by the number of those at risk for the disorder (based on prevalence numbers). However, more recent calculations have also tried to take into account for timeframe, and number of minors in this pool (who are not eligible for testing) to calculate PT uptake figures.

1.3.3 Psychological consequences of predictive testing

The increased use of genetic testing, particularly in the areas of predictive genetic testing for adult onset disorders such as HD, have sparked a number of ethical debates in the literature regarding the potential harms and benefits of providing and undertaking such testing. These concerns are manifest in discussions surrounding possible psychosocial implications of testing, which may be positive or negative in nature (Codori & Brandt, 1994; Wiggins, *et al*., 1992). In terms of the advantages of PT, proponents argue that testing enables individuals to make an autonomous choice regarding knowing whether or not they will develop the condition. This choice may be motivated by a variety of desires such as relieving uncertainty and the possibility of pursuing a different life course including treatment (or screening) and reproductive options (Decruyenaere, *et al*., 1996). However the potential benefits of such testing must be weighed against potential harms that may result, including the potential for discrimination (e.g., in life/ health insurance or in employment), family and relationship difficulties, or depression relating to the knowledge that one is destined to develop a certain disease (Bombard, *et al*., 2007; Codori & Brandt, 1994; Harper, *et al*., 2004).

PT for the disorder is recognised as being an irreversible decision of great consequence (Evers-Kiebooms, Cassiman, & Van den Berghe, 1987), with substantial implications including psychological and emotional feelings of guilt (including 'survivor guilt') (Tibben, 2007; Tibben, *et al*., 1997; Williams, *et al*., 2000), shame and fear, as well as other effects such as unintentional risk alteration for biological relatives (Benjamin, *et al*., 1994; Bloch, *et al*., 1992; Wiggins, *et al*., 1992). As such, the international guidelines for HD PT recommend that individuals considering

testing undergo in-depth genetic counseling, psychosocial evaluation and work-up prior to receiving results to ensure they have adequate support and preparedness for receiving life-altering news (MacLeod, *et al*., 2012). Both internal and external factors, including effects on other family members and relationships, family planning, insurance and employment discrimination, career planning, access to testing and ability to pay for the service, come into the decision-making process regarding whether or not to proceed with PT (Binedell, Soldan & Harper, 1998; Bombard, *et al*., 2007; Harper, *et al*., 2004; Hawkins, Ho, & Hayden, 2011; Kessler, *et al*., 1987). Despite considerable concern for adverse events such as suicide, major depression or psychiatric hospitalization following predictive test results (Mastromauro, *et al*., 1987; Meissen & Berchek, 1987), these fears have not been realized. A number of studies have shown that frequencies of adverse catastrophic events after receiving test results are similar to those in the general population (0.97% or 44/4,527) (Almqvist, *et al*., 1999). Perhaps not surprisingly, individuals most at risk for difficulty in adjusting to a positive PT result are those who have just received a positive test result (Almqvist, *et al*., 1999) and those with a prior psychiatric history (Bloch, *et al*., 1992). Nonetheless, the majority of studies suggest that people are satisfied with their decision to be tested and cope well over the long term when adequately prepared to receive a HD test result, regardless of whether they received a positive or negative test result (Codori & Brandt, 1994; Decruyenaere, *et al*., 1996; Decruyenaere, *et al*., 2003; Dufrasne, *et al*., 2010; Hayden, 2000; Lawson, *et al*., 1996; Meiser & Dunn, 2000). Levels of psychological distress after testing are significantly lower once PT results are received (whether HD+ or HD-) as compared to before testing (Almqvist, *et al*., 2003; Broadstock, Michie, & Marteau, 2000).

1.3.4 Deterrents to predictive testing

To discuss the deterrents to PT we first need to understand the exact nature of these deterrents and subsequently address the issues and limitations which create such disincentives. A review of the academic literature *(*Demyttenaere, Evers-Kiebooms, & Decruyenaere, 1992; Evers-Kiebooms & Decruyenaere, 1998; Kessler, 1994), plus anecdotal reports from genetic counselors working in the field[8], suggests that one of these deterrents is limited access to genetic services. For example, Kessler reports that "*the testing protocol may act as a screen to discourage all but the most motivated and determined individuals to proceed with the test"* (Kessler 1994, p163). This issue is even more relevant in rural and remote areas where there is no access to a medical genetics department or genetic professionals.

The large gaps in genetic services coverage described above may also represent a significant hindrance for those who currently require genetic counselling and services. In regards to HD, in which there are potential therapies on the horizon (Walker, 2007), these geographic issues will be a major barrier to persons residing in under-served areas in terms of their ability to participate in PT and clinical trials.

1.4 Underlying ethical considerations

The above outlined research problem requires a consideration of the various ethical issues that surround health and healthcare delivery generally, as well as genetics more specifically. The central ethical consideration of the research involves justice and equity in relation to health and

[8] Personal communication with genetic counselors in the HD field: Nathalie Boulduc, Shaina Archer, Tina Babineau-Sturk and Susan Creighton.

healthcare services. However, while issues of justice and equity[9] are the central tenets of this research, the ethical principles relating to autonomy and beneficence/ non-maleficence[10] are also relevant (Beauchamp & Childress, 2009).

1.4.1 Justice

Justice in healthcare delivery relates to the availability and accessibility of healthcare services, treatments, facilities and other factors across the population (Kenny & Joffres, 2008; Daniels, 2001). Just delivery of healthcare resources (in a country with a universal health system such as Canada) is defined as equal distribution of healthcare resources to those in equal need of such resources. For example, an individual diagnosed with breast cancer should have the same ability and access to surgery, chemotherapy, radiation and other therapies as other individuals diagnosed with the same type of breast cancer. This is not to say that every individual should be offered the most expensive, state-of-the-art treatment available; priority setting and economic realities are necessarily important considerations involving complex ethical tradeoffs (Singer, *et al*., 2000). Instead individuals with the same condition, disorder or disease should be offered the same standard of care regardless of their ability to pay, location, gender, ethnicity and so on (Giacomini, Kenny & DeJean, 2009). Individual choice also undoubtedly plays into such

[9] Discussions of inequity and injustice in health and healthcare require a definition of certain terms: inequalities are considered unjust if they are the result of systematic differences in social structures that cause one group, or groups, to be unfairly compromised in terms of their health status in relation to another group (Starfield, 2006). What makes inequities 'wrong' is not just that they result in differences in morbidity and mortality, but because they are the expression of unfair and unjust social, political and economic circumstances (Peter, 2004). Creating equity in health involves removing barriers that lead to inequities, as well as fair distribution, access to opportunities and support (Whitehead & Dahlgren, 2007).

[10] In this context the principle of autonomy is concerned with respecting an individual's rights and ability to make informed choices for themselves. The principle of beneficence relates to performing and pursuing actions that will result in beneficial outcomes, and the principle of non-maleficence relates to avoiding actions that will cause harm.

considerations, as do realities such as economics, politics, scientific opinion and resource allocation (Kenny & Sherwin, 2011; Sherwin, 2000; Sherwin, 1992). Discussions of whether healthcare is justly distributed are also tied to nuanced definitions of justice, and disagreement as to whether healthcare should even be considered a justice issue (Daniels, 1985). As such, while the ideals of social justice may call for a fair and equal distribution of healthcare resources, these ideals are either *"not widely recognised or conveniently ignored"* (Beauchamp, 1976, p105). A more detailed discussion of justice and equity in access to health services (including the Canadian Health Care Act), which addresses the complexities of the issues in the healthcare and justice debate, will be explored further in chapter two.

1.4.2 Autonomy

Utilization of health services is related not just to need, but also to personal characteristics and individual autonomy (Sherwin, 2000). While a society may be exemplary in providing equal access to healthcare, this equal access may not necessarily result in equal utilization of the service. Utilization of healthcare services is determined by personal characteristics such as preferences, trust in the healthcare system, previous experiences, expectations and beliefs. Healthcare service use is therefore determined in part by an individual's ability to make an autonomous choice as to when and whether they access an available service. Individual autonomy is an important consideration when determining whether inequity in utilization is truly unfair; if an individual makes a free and informed choice not to access available healthcare, and this choice is made without influence from unfair social structures, barriers or processes, the observed inequity in utilization may not necessarily be unfair (Allin, 2008). Similarly, if differences in health status result from this autonomous choice, these

differences would not be considered to be socially unjust, as they were driven by personal choice rather than social structures.

HD PT can provide elucidation of this point: The decision whether or not to undergo HD PT is a highly personal choice that should be made in an informed manner without pressure from outside sources. If a decision regarding whether or not to undergo PT was influenced or determined by access to genetic services (not a personal choice), which is a societal/ structural issue, and this access encroached upon an individual's ability to make a free and informed choice, this would be considered unfair and unjust. However a relational autonomy approach is also relevant in understanding decisions around predictive genetic testing, as decisions are influenced by numerous factors including family structure and obligations, medical literacy, culture, gender roles and child-bearing considerations (Burgess & d'Agincourt-Canning, 2002; Ho, 2008; McLeod & Sherwin, 2010; Sherwin, 2000). On a similar vein, the ability to provide truly informed consent – a hallmark of autonomy - in predictive genetic testing is complex (Burgess, 2007) as the full ramifications for such testing for oneself, one's relationships and one's family members may be unknown and could include despair, survivor guilt, discrimination, financial pressures and so on.

1.4.3 Beneficence/ non-maleficence

Since it was first conceived, the availability of HD PT has sparked a number of debates regarding the potential harms (maleficence) and benefits (beneficence) of providing and undertaking such testing (Burgess & d'Agincourt-Canning, 2002). Concerns primarily focused on the psychosocial implications of testing, as discussed previously (Codori & Brandt, 1994;

Wiggins, *et al*., 1992; Huggins *et al*., 1992). Potential disadvantages or harms related to PT include discrimination (e.g., in life/ health insurance or in employment), relationship difficulties, and depression (Bombard, *et al*., 2007; Codori & Brandt, 1994; Harper, *et al*., 2004). Potential benefits of PT include the ability to choose to know whether one will develop HD and therefore make certain choices such as career planning, family planning and insurance planning (Benjamin *et al*, 1994; Cox, 2003; Decruyenaere, *et al*., 1996).

1.5 Gaps in existing research

In summary, the limited access to genetic services and PT for individuals at risk for HD may represent an inequity in the healthcare system which may limit such individuals from receiving the care, support and education they require. The current testing model, which requires multiple in person visits, may act as a disincentive in its requirements and could discriminate against at risk individuals in rural communities[11]. Disturbingly this may not only lead to limitations and inequities among those receiving care, it may also lead to a number of individuals undergoing improper PT which completely circumvents accepted practice. These individuals may be offered little education, support or follow-up[12]. Thus, there is a need for research to explore the potential obstacles to testing in terms of access to genetic services, and subsequently an exploration of novel and practical mechanisms by which this issue may be addressed. Such research may improve access to PT services and other potentially beneficial research

[11] In an attempt to address this issue, the BC Rural Testing Protocol has been in effect for the last several years. This protocol modifies the current accepted practice by requiring only one in-person visit, and providing all other testing services through a local healthcare practitioner and telecommunication methods.

[12] Personal communication with Susan Creighton, Jeff Carroll and Bev Heim-Myers.

advancements in the field of HD including clinical trials and new therapies. This research study proposes a proactive approach to decreasing inequities in genetic service provision for those residing in rural communities, which may, ultimately, help improve the lives of those affected by this devastating disorder.

1.6 Research objectives

The purpose of this research is to explore a novel mechanism to address the potential inequity in the healthcare system that exists in access to HD PT in rural communities. Specifically the research project aims to elucidate and address structural and societal (as opposed to personal) access issues by developing a user friendly PT protocol for HD which may be used anywhere in the world.

1.6.1 Broad goals

1) Determine whether access to genetic services is truly perceived as a barrier/ deterrent to PT for HD by at-risk individuals;
2) Assess the key components of the PT process such as information/ education provision, quality of care and support, and pre and post-test counseling, from an at risk individual and expert HCP perspective;
3) Use this information to develop a user-friendly PT protocol and website for HD which provides remote testing for HD while still maintaining quality of care, counseling, education and follow-up support;

4) Compare this novel protocol to the standard testing protocol on a variety of quality assurance measures to determine whether this novel protocol is different to the existing protocol across important outcome measures including satisfaction and support.

1.6.2 Research hypothesis

The overall hypotheses for this project are as follows:

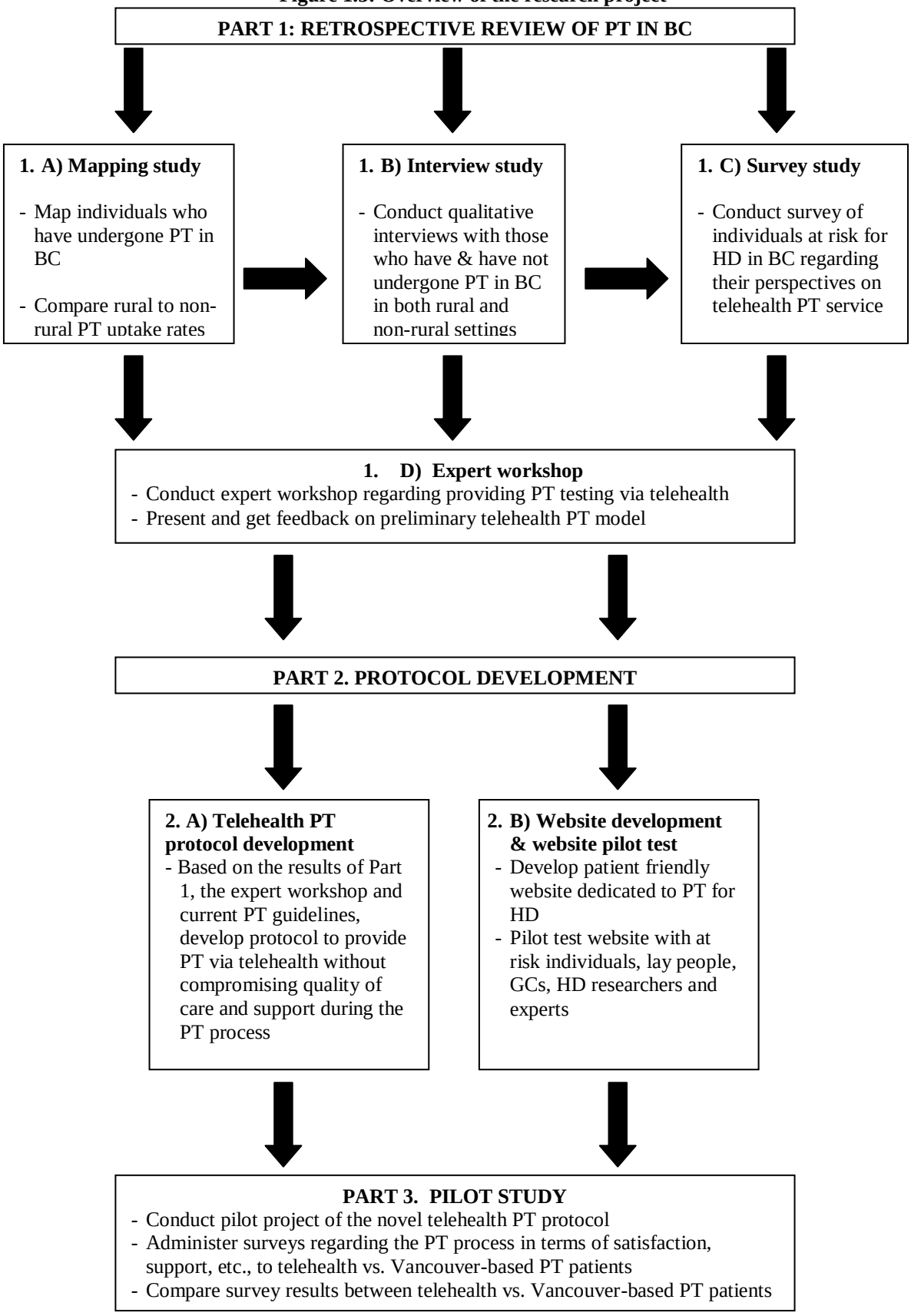
1) Access to genetic services is a barrier or deterrent to PT
2) Lack of access represents an injustice or inequity within the healthcare system
3) A novel telehealth testing protocol for delivering PT for HD can address the inequity that exists in access to PT in rural communities in British Columbia without compromising quality of care and support for those undergoing testing.

1.7 Research methodology and analytic approach

1.7.1 Overview of the research project

The proposed project will address access to PT for HD through a three part, highly structured, mixed methods research approach, as outlined in Figure 1.2. The discussion below provides a brief outline of the research methodologies used to address the aforementioned research objectives, before moving on to a discussion of the overall approach to data analysis. More specific details on recruitment procedures and analysis methods are provided in subsequent chapters.

Figure 1.3: Overview of the research project



1.7.2. Research location

British Columbia is an appropriate location to conduct this research given a number of distinct provincial characteristics:

1. BC is an established centre of excellence in the area of HD research
2. BC has considerable experience and infrastructure in providing counseling, support and services to HD families[13].
3. There is significant awareness of HD among British Columbians, and traditionally higher than average PT rates (Creighton, *et al*., 2003b).
4. 40% of the BC population lives outside the Vancouver health service area (Statistics Canada, 2010), with the rest of the population residing in more rural locations which may be up to 20 hours drive from the HD testing centre at the University of BC.

1.7.3 Part 1: Retrospective review of predictive testing in British Columbia

Part 1A: Mapping study

Overview
This component of the research project involved a retrospective review of the nearly 700 individuals who have undergone PT in BC (Centre for HD clinic records). Individuals were stratified as rural versus non-rural based on their location at the time they underwent testing.
Objectives
1. Compare rural to non-rural PT uptake rates to determine whether there is a difference in PT uptake rates between these areas

[13] The Centre for HD at UBC has been running since 1983, and offering PT since 1986. More information on the Centre for HD can be found at: http://www.cmmt.ubc.ca/outreach/hd-clinic

Part 1B: Interview study

Overview
The interview study involved 33 in-depth semi-structured interviews with those at risk for HD in BC who have and have not undergone PT in both rural and non-rural locations. The interviews sought to understand the perspectives of those at risk for HD in terms of the PT process, and access to the PT service. Interviews consisted of open-ended questions focussing on: 1) perceptions of the PT process; 2) experience of considering and/or undergoing PT; 3) barriers to testing; 4) assessment of the key components of the PT process; and 5) opinions of providing PT remotely. Interviews were tailored based on whether the respondent had undergone PT, their location and their at risk status (if known).
Objectives
1. Document the experience of patients who have undergone PT and of those at-risk who have decided not to undergo testing 2. Understand the essential components of the PT process 3. Explore how the PT process may be adapted and refined to improve access while maintaining quality of care 4. Determine the barriers/ deterrents to PT (structural and personal)

Part 1C: Survey study

Overview
This stage of the research project involved a paper-based survey of 100 HD families across the province of BC to better understand perspectives on access to PT, and the provision of PT services via telehealth.
Objectives
1. Determine the support and need for the PT telehealth service in BC

Part 1D: Expert workshop

Overview

This part of the research project involved an expert workshop, held in Vancouver, BC to foster discussion and elicit expert advice regarding the provision of PT for HD via telehealth, including remote and rural areas. Eleven experts were invited to participate in the workshop based on the following criteria:

Expert type	**Expert knowledge area**
Provides genetic services at a distance	Telemedicine and other novel communication strategies Non-traditional genetic counseling Training and education needs and mechanisms
Provides a high volume of HD testing and care	Ethical issues Psychosocial considerations Practical limitations

Objectives

1. To assess experts' perspectives on the key components of the PT process
2. To gather suggestions and recommendations on how to improve access to PT for HD
3. To propose and get feedback on a provisional telehealth PT testing protocol

1.7.4 Part 2: Protocol development

Overview
Results from the first part of the research project as well as the current PT guidelines were used to inform the development of a telehealth PT protocol, for use in rural areas, to improve access to HD PT.
Objectives
1. Develop a portable PT protocol that provides PT via telehealth without compromising quality of care and support 2. Develop a patient friendly website dedicated to PT for HD which provides the information an individual considering PT for HD would need including: a. The genetics of HD, including inheritance, the meaning of the CAG repeat size etc. b. The advantages and disadvantages of testing c. Interpreting PT results d. Deciding whether or not to pursue testing

1.7.5 Part 3: Pilot study

Overview
This part of the research project involved a year-long prospective, comparative study of PT in the BC population. The study involved a comparison of 13 individuals who underwent the standard PT protocol in Vancouver and 15 individuals who underwent PT via the telehealth protocol. The hypothesis driving this component of the research project was that the telehealth PT protocol would improve access to PT without compromising quality of care and support.
Objectives
1. Determine whether the telehealth PT protocol and standard PT protocol differ in terms of patient satisfaction, support through the process, information and education, time to attend appointments, etc.

1.7.6 Analytic approach

The research questions considered in this study were posed in a curious and open frame of mind, with the intent of understanding a novel issue, wherein paradigms are unsettled and theories are evolving. This research can therefore be deemed as primarily exploratory (i.e., exploring the barriers to PT and exploring strategies to address these barriers) rather than explanatory (i.e., explaining why these barriers exist). As the topic of access barriers to PT has not been adequately explored in the existing literature, there is a limited understanding of what these barriers might be and how they might be addressed. Thus before we can address barriers to PT, we need to first understand the exact nature of these barriers. Initial investigation by qualitative methods via a qualitative exploratory approach was deemed most appropriate to investigate and understand this issue (Chenail, 2000; Denzin & Lincoln, 2000). Once an understanding of these barriers to PT was achieved, subsequent stages of the research aimed to capture the scope of this issue, as well as exploring support of and satisfaction with a telehealth PT protocol. As such, a mixed methodological approach, including qualitative semi-structured interviews followed by quantitative based surveys, was considered most suitable to investigate the research topic at hand.

Using a mixed methods approach draws on the strengths of both qualitative and quantitative methods to gain a greater understanding of research questions by adding 'rigor, breadth, complexity, richness and depth' to inquiry (Flick, 1998). In this case, qualitative methods provide a nuanced and detailed understanding of the complexities involved in accessing HD PT and essential components of the PT process. Analysis of this qualitative data allows for the development of theories regarding access to PT, and informs subsequent stages of the

research including which questions and topics should be addressed in the quantitatively-based survey. The quantitative methods provide a mechanism of assessing and comparing perspectives on PT and the strategies developed to address these barriers.

A sequential exploratory approach (Strauss & Corbin, 1998) was determined to be the most appropriate mixed method strategy for this research project. This approach involves a primary qualitative data collection and analysis phase, followed by a secondary quantitative collection and analysis phase that is shaped by the primary qualitative phase. The primary component of this design is to explore a phenomenon via the qualitative research phase. The secondary quantitative phase is used to test any emergent theories and ideas which have been developed during the first phase of the research and determine the distribution of a phenomenon in a given population (Morse, 1991). Data are thus linked to maximize learnings that reflect both the first and second phases of work. There are numerous advantages of this approach: it is relatively easy to implement, describe and report; it enables a researcher to expand on qualitative research findings and test preliminary theories during the quantitative phase; yet it is more accessible and legitimate to those who are skeptical of qualitative research as it includes a quantitative component (Creswell, 2009). A model of this design is outlined in Figure 1.3.

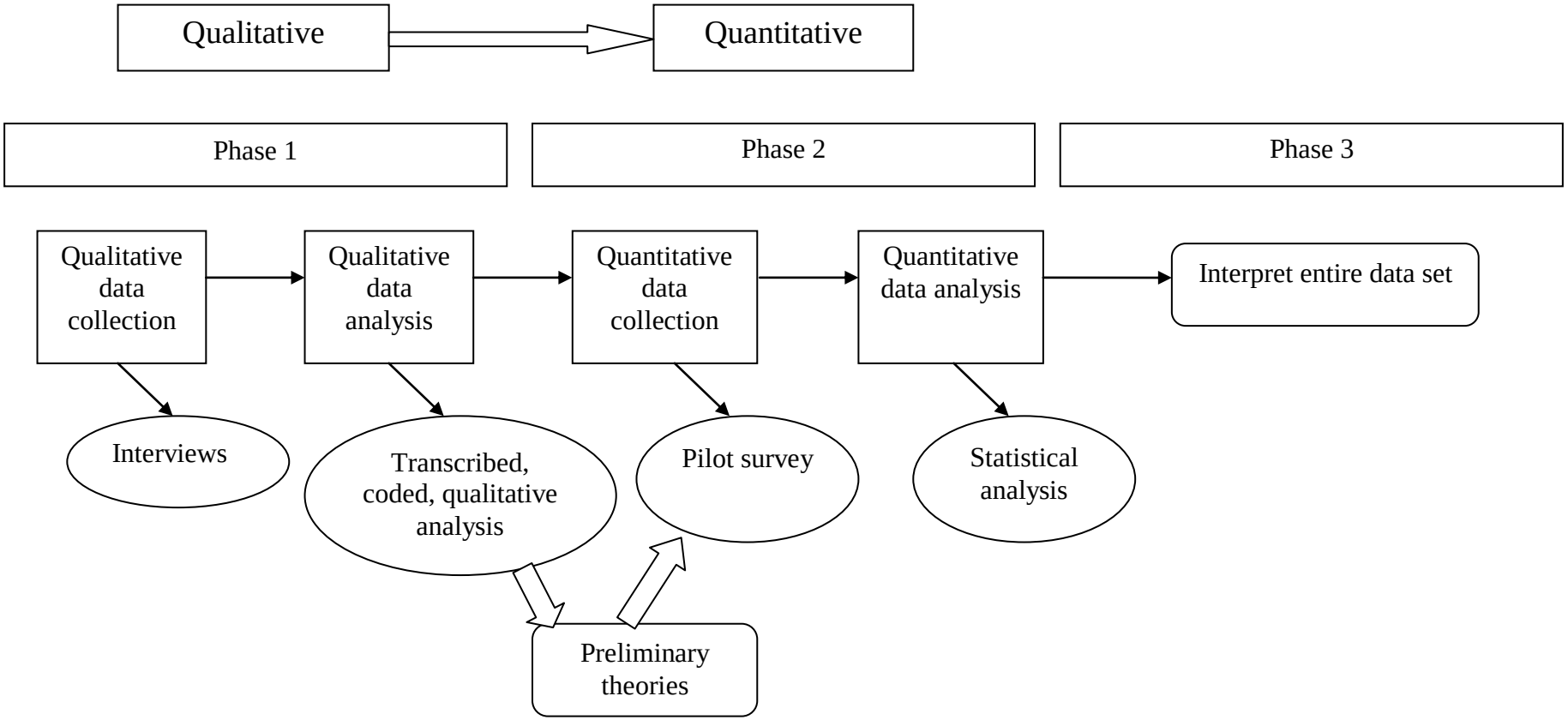


Figure 1.4: Research design and analysis flowchart

2. ACCESS TO HEALTHCARE IN RURAL COMMUNITIES: EXPLORING THE ETHICAL ISSUES OF INEQUITY AND JUSTICE IN THE CONTEXT OF GENETICS

2.1 Introduction

"The growing field of genomics raises questions of just and equitable access to health services for all."

(WHO, 2010)

Genetic research, techniques and knowledge have greatly expanded in the last two decades with the completion of the Human Genome Project and other major advances in research and diagnostic technologies. With this expansion, there is growing interest in the clinical application and utility of such technologies on population health, including the promise of personalized medicine. It is hoped that genome based healthcare will lead to better disease prediction and prevention, improved treatment, and decreased adverse drug reactions (Personalized Medicine Coalition, 2009). While the application of genetic research in the clinical realm holds great promise, it also raises significant challenges. One of the most prominent of these issues relates to the translation of genetic knowledge into actual delivery of genetic testing and services. This challenge is particularly acute in rural and remote areas, where lack of access to genetic services is a significant issue.

This chapter will explore the aforementioned assumption and examine the hypothesis that lack of access represents an injustice or inequity within the healthcare system. The chapter will begin with a definition of equity and justice and outline the distinction between equity in health and equity in healthcare. The chapter will then provide an overview of the major ethical theories of equity and justice in the context of health, including a discussion of the capability approach, libertarian theories, egalitarian perspectives and critical and feminist approaches. Examples of

inequities in health and healthcare will be provided along with a more detailed description of access issues (geographic, cultural and economic) as they relate to equity and health.

The chapter will then discuss how the aforementioned theories might be applied in the context of genetics, focusing specifically on access to genetic services in rural areas to establish: first, that access does appear to be limited in rural areas, and second, that genetic services are likely to be of importance to human health and well-being. Throughout this section an important distinction will be made between the current state and potential future state of genetic service delivery and its impact on healthcare. The chapter will consider whether this lack of access actually represents an injustice in the healthcare system, either at the current time or potentially in the future.

2.2 Equity and justice

According to Whitehead and Dahlgren, in their 2007 report for the WHO, inequities in health are defined by three distinguishing factors: they are systematic, socially produced and unfair. A systematic difference occurs when there are differences in health status between different social groups, for example, an increase in morbidity or mortality from a certain condition with declining social position. Socially produced differences are differences in health status that are socially, not biologically determined; such social processes are potentially modifiable. Inequities in health are deemed unfair because they are both generated and maintained by ‘unjust social arrangements’ that do not allow individuals to achieve their full potential due to socially determined circumstances (Whitehead & Dahlgren, 2007).

Thus, health inequalities are considered unjust if they are the result of systematic differences in social structures that cause one group, or groups, to be unfairly compromised in terms of their health status in relation to another group (Starfield, 2006). What makes inequities 'wrong' is not just that they result in differences in morbidity and mortality, it is because they are the expression of unfair and unjust social, political and economic circumstances (Pauly, Mackinnon & Varcoe, 2009; Peter, 2004; Sherwin, 1992;). If inequities in health are characterised by systematic and potentially remediable differences, the converse is the absence of these health differences in geographic, social, economic or demographic sectors of the population (International Society for Equity in Health, 2005). Creating equity in health necessitates removing barriers that lead to inequities, fair distribution, access to opportunities and support (Anderson *et al*. 2009; Whitehead & Dahlgren, 2007).

In consideration of inequity, it is important to make a distinction between equity in healthcare and equity in health; equity can be understood in terms of equity in access to healthcare as well as in equity in health outcomes (Pauly, MacKinnon, & Varcoe, 2009). Equity in the former does not necessarily result in equity in health status as access to care is only one of many factors that determine the ultimate health status of an individual (Peter, *et al*., 2001). The importance of social determinants of health, including social position, education, financial standing, cultural considerations and other factors must not be underestimated (Baylis, Kenny & Sherwin, 2008; Denton, Prus & Walters, 2004; Judge, Mulligan & Benzeva, 1998). These factors are sometimes referred to as the 'social' gradient in health whereby health status is relative to social standing (Marmot, 2003; Marmot, 2005; Powers & Faden, 2000). There is also an important distinction between the goals for equity in health and equity in healthcare; the ultimate

goal for equity in health is the removal of all differences in health status between different social groups, whilst the ultimate goal for equity in healthcare is to match health services to health need, so that all those with the same health needs receive the same access to healthcare services (whether or not they chose to use them) (Whitehead & Dahlgren, 2007).

The distinction between equal health and equal healthcare is important to make in understanding the relationship between access to health services and health outcomes. Utilization of health services is related not just to need, but also to personal characteristics and relational autonomy (Ho, 2008; McLeod & Sherwin, 2000). While a society may be exemplary in providing equal access to healthcare, this equal access may not necessarily result in equal utilization of the service. Utilization of healthcare services is determined by personal characteristics such as preferences, expectations and beliefs, as well as individuals' relationships, family structure, culture and other influencing factors (Sherwin, 1988). For example, just because a healthcare service, such as mammogram screening, is available, not all women will access the service due to cultural barriers and considerations (Bell *et al*., 1999; Sassi, Luft & Guadagnoli, 2006; Sheinfeld Gorin & Heck, 2005). Lack of trust in the healthcare system, or healthcare providers, may also deter certain groups from accessing services. Healthcare service use is therefore determined in part by an individual's ability to make an autonomous choice as to when and whether they access an available service (Pauly, 2008).

Autonomy is an important consideration when determining whether inequity in utilization is truly unfair; if an individual makes a free and informed choice not to access available healthcare, and this choice is made without influence from unfair social structures, barriers or

processes, observed inequity in utilization may not necessarily be unfair (Allin, 2008). Similarly, if differences in health status result from this autonomous choice, these differences would not be considered to be socially unjust, as they were driven by personal choice rather than social structures. However, these arguments are complicated by the fact that healthcare decisions are not made in a vacuum and as such may never be truly 'free'. Instead such decisions are influenced by many competing factors such as family, relationships, gender, social status, culture and so on. While it may be argued that decisions are never truly 'free' from outside influences and contextual factors, some decisions may be more constrained than others, particularly given one's social circumstances. As such, a relational model of autonomy is a more useful model to apply in health and healthcare decision-making (Ho 2008; Sherwin 1992). Consider, for example, a preventative national screening mammography program for breast cancer. Some members of the population may not wish to undergo mammography due to personal reasons, including culture, concepts of womanhood, relationship status, religion, age or culture (Bell *et al*., 1999; Sassi, Luft & Guadagnoli, 2006; Sheinfeld Gorin & Heck, 2005). Forcing them to undergo the screening would encroach on their autonomy and be unfair. However for the program to be deemed equitable on a national level it would need to ensure equal access to screening with no geographic, cultural, social, economic or other barriers (or at least minimizing these barriers as much as possible). Trust considerations would also need to be addressed, so that people who wished to use the service could not only do so, but they could also trust that their privacy, information and personal beliefs were respected. If barriers in access and trust existed, and prevented access to the screening, this program would be considered unjust and unfair.

2.3 Major ethical theories of justice and health (care)

The forthcoming sections will move on to a discussion of the major ethical theories of justice and how they may be applied to health and healthcare. Doing so will establish that some theories of justice have been extended to health and healthcare, while others have not. This discussion will enable the reader, particularly during the latter part of this chapter, to reflect on whether and how these theories may be used in the context of access to genetic services. In the following sections, relevant ethical theories will be outlined before a final summary section is presented that provides a brief comparison of these viewpoints. Finally, a suggestion for which of these theories may be most applicable and useful in the context of access to genetic services is presented.

2.3.1 Libertarian theories

Libertarian viewpoints on justice can be traced back to seventeenth and eighteenth century philosophers such as John Locke and David Hume. Their theories are primarily concerned with creating a society in which individuals may be 'free and equal'. Such a society would enable individuals to develop and express themselves as they please, and protects them from arbitrary interference and restrictions by the government or other members of society (Held, 2006; Nozick, 1974). Libertarian theories underpin the dominant North American conception of economics and capitalism, with perspectives of justice underlying market justice (Pauly, MacKinnon & Varcoe, 2009).

With respect to healthcare, libertarian theories of justice consider healthcare to be a commodity that should be determined by market forces. Additionally, there is a strong emphasis

on an individual's responsibility for his or her own health and well-being, with a limited role of government in people's lives being relatively hands off to ensure people are free to pursue their own opportunities. Under this viewpoint, lack of available healthcare, and other socially desirable commodities such as public services, education, housing and employment, are unfortunate, but not unfair as these resources are determined by the marketplace and individual choice and responsibility (Alkire & Chen, 2004). Thus while libertarian theories acknowledge the importance of health and healthcare to society, they do not consider them to be an issue of justice. Health differences are thus undesirable, but do not represent a social justice issue (Powers & Faden, 2000). As such, libertarian theories would not consider variation in access to testing or screening to represent an injustice or inequity in society.

This viewpoint is congruent with the increasing availability of 'for-profit' healthcare services, most apparent in the United States, where traditionally there has been no universal healthcare system, although the new Affordable Care Act (commonly known as 'Obama-care') and the recent Supreme Court Ruling on this divisive issue may change this (Krugman, 2012)[14]. Insurance-based systems are predicated on the assumption that individuals should take personal responsibility for their health and access to healthcare, and make related decisions in the context of their circumstances and desires. Inadequacies in health should be dealt with through philanthropy and promoting a humanitarian response. Such a safety net response however, has limitations as it creates a power imbalance that relies on the generosity of the benefactor and 'does not address the structural or root causes of inequities' (Pauly, MacKinnon &Varcoe, p121).

[14] In recent years Canada has also seen an emergence of private or supplemental healthcare that must be paid for out of pocket such as cosmetic surgery, dental services and private health clinics.

2.3.2 The capability approach

In contrast to libertarian theories which consider healthcare to be a commodity and an individual's own responsibility, the capability approach views healthcare as essential in assisting individuals to be functioning and contributing members of society. Sen outlines the capability view of health in which health is not just an end, in and of itself, but is viewed as an important mechanism to achieving other objectives in life such as education and employment. Achieving and maintaining health is a way of promoting functioning and the freedom of individuals and societies (Sen, 1993). Sen argues that health equity should be judged in terms of capabilities and achievements rather than just healthcare activities and outcomes. Ultimately, people value the capability to achieve good health (Alkire & Chen, 2004). This viewpoint is further developed by Peter (2001):

> *"What matters is what people can do with the resources they have access to – a notion of freedom. A person's capability tries to capture that. Capability is the set of 'functionings' – the various 'doings and beings' – that a person can achieve (but may decide not to). Examples of functionings are being adequately nourished, being able to read, or, more complex, achieving self-respect. Sen often cites good health as an example of functioning. It may, however, be more promising to see health as a capability of its own, or, more precisely, as a sub-set of a person's capability. The idea is that health itself is*

> *composed of several functionings – such as, for example, being able to move around, not being tired, etc."*
>
> (Peter, 2001, p162)

Thus the capability viewpoint gives precedence to human flourishing and the ability to function in terms of what people are able to do and their freedom to pursue the lives they value. Health capability is given a special moral importance so that society is morally obliged to prevent or resolve any loss of an individual's physical functioning. This also encompasses human agency; the ability to live a life that has value for the person that is living it. Human agency subsequently impacts mental health and well-being (Ruger, 2004). As such, social policy should aim to maintain and improve capabilities by providing health services and by meeting an individual's health needs.

The capability view does not specify which services or basic benefits should be supplied in a society. However Sen does present a concept of the basic capabilities: the capability to avoid morbidity and premature and preventable mortality (Sen, 2002). Policies and resources should therefore be allocated in efforts to avoid preventable deaths and illnesses that do not allow individuals to live a life of normal length. Further, healthcare resources, in order to be just and to avoid inequity, should be distributed in terms of need, not in terms of one's ability to pay. As such, a just genetic screening and testing program should not be determined by ability to pay for and access such services.

2.3.3 Critical and feminist perspectives

Many critical and feminist approaches to health inequity are commonly associated with the ideas of Iris Marion Young. In her book, 'Justice and the Politics of Difference', Young argues that social differences can be deemed inequitable if they represent systematic structural differences in a society (Young, 1990). This is similar to the definition of inequity/ injustice outlined earlier in the chapter. Part of establishing an injustice in society has to do with determining an individual's status relative to that of other members of society. Unequal distribution of a certain good or service may lead to inequalities that are unjust if the unequal distribution represents or even perpetuates a systematic structural difference in society, and these differences cannot be attributed to individual choices or preferences, luck or accident (Caufield Burgess & Williams-Jones, 2001; Sherwin, 1996; Young, 2002).

However judgements of injustice are not simply concerned with the unequal distribution of a good or service. Distribution represents only part of what needs to be considered and may be a clue as to the social processes or structures that are in place that restrict opportunities (McLeod & Sherwin, 2000). These processes are unjust if they represent generalised social structures that exist and enhance the capabilities and opportunities of one group, while ignoring or restricting access and capabilities of another group. Differences in distribution are therefore just the starting point for a theory of justice – these differences, which may reveal themselves in many places, such as ethnicity, gender and class, limit individual freedom and well-being and impact the development of inequities (Pauly, 2008; Young, 2002).

Social structures and processes that result in unjust situations may result from a number of different factors such as institutional practices, political viewpoints, rules, laws and regulations that govern society (Sherwin, 1988; Kenny, Sherwin & Baylis, 2010). These structures refer to "*the relation of basic social positions that fundamentally condition the opportunities and life prospects of persons located in those positions […] The unintended consequences of the confluence of many actions often produce and reinforce such opportunities and constraints, and these often make their mark on the physical conditions of future actions, as well as the habits and expectations of actors"* (Young, 2002, p14-15).

These structural processes may result, for example, in social oppression, limitations in individual options and actions, stereotypes, gender differences through socialisation and economic dependence (Arber & Ginn, 1993; Pauly, MacKinnon & Varcoe, 2009). In turn these may result in inequalities between groups that can be seen as unjust, not simply because they are the result of social processes but also because they are modifiable. Structural inequities represent constraints that people encounter in their freedom and wellbeing in comparison to others who may be less constrained and have greater access to benefits (Wallace & Villa, 2003). Young recognises that these inequities do not necessarily lead to worse outcomes for those who are constrained due to personal attributes and differences such as luck, diligence and intelligence. However, those who overcome significant obstacles to reach a certain endpoint cannot be seen as equal to those who have had to overcome fewer obstacles to reach the same endpoint. Young offers this definition of injustice:

> *"If observation discovers that a category of individuals is unequal to others on certain important measures of well-being, and a plausible story can be told about how the relations, rules, expectations, and cumulative consequences of collective action specifically condition the lives of members of that group, then there are grounds for saying that members of the group suffer some injustice."*
>
> (Young, 2002, p15)

The critical/ feminist perspective is relevant for consideration of justice in both health status/outcome and distribution/access to healthcare, including access to HD testing. This is due to the notion that social processes and structures result in different social, historical, economic and political conditions that result in both inequities in health and in healthcare (Pauly, MacKinnon & Varcoe, 2009). In the context of HD, although there is no cure for the disorder PT may afford individuals multiple benefits including relieving stress, enabling reproductive planning, and financial and career decision-making (Binedell, *et al*., 1998; Harper, Lim & Craufurd, 2000; Kessler, *et al*., 1987; Wiggins, *et al*., 1992). Thus inequity in access to such testing and support services due to economic, social, political or other reasons, may result in psychological and emotional consequences (including stress, depression, etc.), as well as economic and other detrimental consequences due to an inability to plan (e.g. purchase appropriate life and disability insurance).

2.3.4 Egalitarian theories

Egalitarian approaches suggest that individuals should be provided with an equal distribution of goods, such as healthcare, to reduce inequality (Beauchamp & Childress, 2009). Such theories have their foundation in liberal theories of justice stemming from John Rawls' book 'A Theory of Justice'. In his book, Rawls outlines two principles of justice, the first of which focuses on basic liberties and argues that:

> *"each person is to have an equal right to the most extensive scheme of equal basic liberties compatible with a similar scheme of liberties for others"*
>
> (Rawls, 1971, p60)

These basic liberties include political liberty, freedom from arbitrary unrest, freedom of speech and gathering, freedom of personal property and liberty of conscience.

The second principle of justice proposed by Rawls relates to the difference principle; social and economic inequalities should be arranged so that they are of the greatest benefit to the least-advantaged members of society. Social goods (i.e., liberty, opportunity, income, wealth and the bases of self-respect) need not necessarily be distributed equally; unequal distribution is acceptable if such distribution is to everyone's advantage. According to Rawls, health is not a social good but a natural good. While Rawls acknowledges that natural goods, including health, are influenced by social goods, he believes that natural goods are not so directly under society's control. Natural goods do not have to be distributed according to the second principle of justice (Rawls, 1971).

Daniels (2001) builds on Rawls' framework and maintains that healthcare, as opposed to health alone, should be a primary good in the Rawlsian framework. He argues that Rawls' conception of justice as fairness supports the claim that we have an obligation to promote and protect everyone's normal functioning. Daniels maintains that healthcare is of special moral importance, as it allows individuals to maintain their ability to be fully functional citizens. According to this argument, in a just society, the healthcare system should be designed so that everyone is as close as possible to an adequate minimum level of health - which he calls "normal species functioning". Disease and disability impair normal species function, and in doing so, restrict an individual's range of opportunities. In the context of HD, lack of access to PT services and support may limit an individual's reproductive choices, ability to receive insurance, career planning and other economic opportunities (Binedell, *et al*., 1998; Harper, Lim & Craufurd, 2000; Kessler, *et al*., 1987; Wiggins, *et al*., 1992). In addition, the psychological and emotional burden of not knowing one's status may have considerable consequences on one's ability to function, make decisions in others areas of life, interact with siblings, spouses and friends, and so on (Higgins *et al*., 1992; Wiggins *et al*., 1992; Almqvist *et al*., 2003).

In considering equitable access to healthcare, Daniels draws upon Rawls' principle of justice as fairness, and develops the concept of fair equality of opportunity. According to Daniels, healthcare makes an important contribution to protecting an individual's equality of opportunity; the opportunity to be fully participating citizens in the social, economic and political aspects of their society. Healthcare systems should thus aim to develop a fair or level playing field, to reduce barriers that limit opportunity (Williams-Jones & Burgess, 2006).

Daniels defines unequal care as being a situation in which two individuals of comparable health status requiring appropriate care do not have an equal chance of actually receiving such care. Access can be deemed equitable if there are no informational or financial barriers, or supply issues that prevent access to a 'reasonable' or decent basic minimum of health-care services (Daniels, 2001). Throughout his discussion, Daniels acknowledges that there are many determinants of health in addition to healthcare. As Ruger notes: *"healthcare is not the only health determinant…and one must not assume that more and better healthcare is all that is needed to improve health"* (Ruger, 2004, p1076).

Daniels does acknowledge the difficulty in deciding what exactly defines a reasonable or basic minimum of healthcare services. Rationing is inherent to healthcare allocation, given that society does not have enough resources to provide everything that may help promote health and functioning. This rationing process is intimately linked to priority setting and distribution, and there are difficulties in determining on what basis we prioritize some services over others, or some people or conditions over others. Daniels believes that the problems of rationing creates a 'moral controversy' which is pervasive in every healthcare system due to resource constraints. This problem is eloquently outlined by Williams-Jones and Burgess:

> *"Just access to healthcare might be thought to entail equal access to every needed medical service, but taken literally and especially if "need" is construed broadly, this approach is unworkable. The current political reality is characterized by limited financial resources available for healthcare and the*

need to fund other important goods such as social services, education, and public works"

(Williams-Jones & Burgess, 2004, p118).

In recognition of such rationing and other limitations in healthcare, Daniels considers how society can make 'fair' decisions about such limitations. He proposes, together with Sabin, a decision-making approach that provides 'accountability for reasonableness'. Such an approach appeals to rationales that are both publically available, and that reasonable people would agree are relevant to meeting healthcare needs in the context of limited resources (Daniels & Sabin, 1997). This approach also requires transparency in the decision making process as well as an appeals mechanism for revising decisions in the light of sound objections (Daniels, 2000). By following such an approach, decisions regarding the rationing and distribution of healthcare can be judged as legitimate (Lauridsen & Lippert-Rasmussen, 2009).

Daniels can be considered to have a moderate egalitarian viewpoint; one that is committed to equality of opportunity and that recognises the importance of healthcare services as strategically important in improving health status and therefore providing a fair chance in life (Cookson & Dolan, 2000). Stronger egalitarian viewpoints can be found in the work of Veatch (Veatch, 1981) who views justice in terms of equal welfare, where every member of society should be provided with health that is as close as possible to the health of other members of society (Powers & Faden, 2000).

2.3.5 Summary of ethical theories of justice and health (care)

Each of the theories discussed above, excluding the libertarian perspective, do consider health or healthcare to be a justice issue if certain criteria are met. While the details of the feminist, capability and egalitarian arguments differ slightly in their reasoning and rationale, each of them, at a basic level, relies on the notion that health, and secondarily healthcare, impact an individuals' ability to function in society, and live both a productive and unencumbered life. Egalitarian and capability approaches are primarily concerned with the curtailing of individual ability and functioning, whereas feminist and critical perspectives focus on societal processes, and whether these processes are potentially modifiable to determine societal injustice.

In contrast, the libertarian viewpoint on justice is not particularly applicable in consideration of whether a recognised healthcare or health service difference represents an injustice since libertarian perspectives consider health and healthcare to be a commodity, and therefore not a justice issue. However the premise of this chapter is not to prove, show or demonstrate *whether* health inequities and justice issues exist, but rather *how* one may determine if difference in health status or heath care actually represent an inequity. Although the libertarian concept of justice is useful in some areas, it considers health a commodity, and as such its justice arguments cannot be applied to health and healthcare. For this reason, the libertarian perspective does not provide fruitful insight for the subject matter at hand and as such libertarian theories will not be considered for the remainder of this discussion.

2.4 Examples of inequity in health and healthcare

Multiple studies have established that disadvantaged groups have worse health and higher mortality and morbidity (Coburn, 2004; Kawachi & Kennedy, 1997; Lundberg, 1991; Whitehead & Dahlgren, 2007; Wilkinson, 1997; Williams & Collins, 1995) consistent with the 'social gradient' in health status. There are multiple documented examples of this disparity including: 1) a thirteen year gap in life expectancy between men with the highest and lowest levels of education in Estonia (Leinsalu, Vagero, & Kunst, 2003); 2) a strong association between socioeconomic status and health in Canada with those in a lower socioeconomic standing needing more health services [15] (Asada & Kephart, 2007); and 3) a ten year life expectancy gap between individuals born in the poorest areas of Glasgow and those born in the wealthiest neighbourhoods (Acheson, Baker, & Illsley, 1998). These differences in health are partially due to income, however, there other important socioeconomic factors that relate to the resulting inequity including education, complementary insurance for prescription drugs and dental care and, in some cases, region of residence (Allin, 2008). The interplay of these forces is an important consideration in healthcare utilisation. In addition, other factors, such as proximity to services, economic and informational resources that allow one to make use of available services, and the appropriateness of services or cultural considerations also determine utilization (Sutherns & Bourgeault, 2008).

Let us now move on to a discussion of access to healthcare services; an issue that has been frequently discussed in terms of inequity in the health system. Access includes economic (i.e., the ability to pay for a service) cultural (i.e., the social and cultural acceptability of a

[15] In Canada there is a recognized gap between the health of Aboriginal and non-Aboriginal Canadians, with Aboriginal populations suffering from a disproportion burden of ill health and suffering (Adelson, 2005; Garner, Carrière & Sanmartin, 2010)

service) as well as physical issues (the primary interest of this chapter). Physical access to services is increasingly recognised as important in debates about inequality (Iredale, *et al*., 2005). People living in rural areas may be significantly disadvantaged in terms of access to secondary and tertiary services as these services are not as readily available in rural areas. Furthermore, there is an inverse relationship between distance and utilisation of healthcare services. For example, a number of Canadian studies have revealed that rural residents have fewer available resources, greater financial obstacles, and limited insurance resulting in less physician visits, limited access to specialists and ultimately a lesser life expectancy compared to rural residents (Pong *et al*., 2011). Individuals living in rural communities have to travel both further and longer to receive care and have more limited transportation options (i.e., less access to public transportation) (Zhang, Tao, & Anderson, 2008).

Recognition of the importance of region of residence in health and healthcare was famously noted by Tudor Hart in the inverse care law whereby the availability of good medical care varies inversely with the need for it in the population served (Hart, 1971). Hart studied coal-mining valleys of Wales and observed that location and quality of healthcare services were much worse in poorer, rural towns. Examples of such inequities in service availability by location can be found in other areas. For example, a Swedish study revealed that there are more private specialists in affluent neighbourhoods in Stockholm compared to less advantaged neighbourhoods (Dahlgren, 2008) and a Manitoba study revealed that physician visits are lower in rural populations than urban populations (Martens *et al*., 2003). Furthermore, Watt and colleagues, who investigated health and healthcare of rural populations in the UK, found that while health outcomes for rural areas were worse than in urban areas, contradictions exist, and

access to health service was the primary problem in rural areas. They also noted ‘distance decay’, or the notion that the further away a service is, the less likely patients will use it (shown in visits to general practitioners and hospitals) (Watt, Franks, & Sheldon, 1994).

Interestingly, there are a few examples of mechanisms to address inequity in access to healthcare services. For example, Starfield (2006) notes that some countries do try to regulate the distribution of healthcare practitioners so resources are distributed more equitably; in some countries in Northern Europe physicians are not reimbursed if they settle in areas with a surplus of physicians, and in some Canadian Provinces, they may be incentivized to practice in rural communities (Rourke, 2008). Moreover, a number of national policies, such as the national health services in the UK, recognise equality as being a primary goal of the national health system. The Canadian Health Act specifically names accessibility as one of the five objectives of the healthcare service: *"to protect, promote and restore the physical and mental well-being of residents of Canada and to facilitate reasonable access to health services without financial or other barriers"* (Canada Health Act, 2005: Section 3).

This discussion will now move to a consideration of how access to healthcare may be considered in the context of genetics, specifically focussing on access issues in rural areas.

2.5 The current situation of genetic services in the healthcare arena

> *"Most healthcare professionals have insufficient knowledge about genomic medicine and there are too few geneticists and genetic counselors."*
>
> (National Human Genome Research Institute, 2006)

Genetics represents a specialised, novel and developing field of healthcare. Due to this fact, and the limited number of geneticists and genetic counselling training programs (Genetics Education Centre, 2012), there is also a limited supply of genetics professionals (geneticists, genetic counsellors and genetic nurses), even in countries with the most developed genetic services. For example, there are only an estimated 240 registered genetic counsellors in Canada (Canadian Association of Genetic Counsellors, 2010), 2440 practicing genetic counsellors in the United States (National Society of Genetic Counselors, 2008) and approximately 300 genetic counsellors in the United Kingdom (Association of Genetic Nurses and Counsellors, 2010). These numbers result in a low number of genetic counsellors per capita: in Canada there is approximately one genetic counsellor per 140,000 people; in the US there is approximately one genetic counsellor per 127,000 people; and in the UK there is approximately one genetic counsellor per 207,000 people[16].

This is not to say that every member of the population requires genetic services. This data merely presents crude figures to serve as a starting point for discussion. Moreover, it should not be claimed that the situation for genetics is any better or worse than for other medical

[16] This number is based on calculating the number of genetic counsellors per population size, as reported by the UN Department of Economic and Social Affairs, Population Division (2008).

disciplines; it is difficult to compare these numbers to per capita estimates in other healthcare service areas. No attempt is being made here to argue that the situation for genetics is substantially worse, or even comparable to other areas of healthcare. Nonetheless, one can imagine that such large per capita ratios may result in limitations in terms of the number of people who are able to receive genetic services. In addition, it may be argued that access to genetic services, and HD PT, is not as important or pressing an issue as, for example, providing access for primary care or public health. This discussion does not seek to determine and argue whether one aspect of healthcare is more or less important (in terms of healthcare impact) than other areas of healthcare. Instead it seeks to point out that access to genetic services and PT may be limited based on geographical location, and thus unfairly favour one sector of society over another.

Compounding this issue is the paucity of access to genetic services in rural areas. On a broad level, access to healthcare in rural areas represents a worldwide concern since half the world's population primarily lives in rural areas (UN Department of Economic and Social Affairs Population Division, 2005). While these rates are much lower in developed countries, at 20% and 18% in the United States and Canada respectively (Colin 2007; Central Intelligence Organization 2009), this does not translate into access to specialised healthcare services, such as genetics. Genetic services are usually confined to a limited number of large academic medical centres (Washington State Department of Health, 2008). Unfortunately, detailed genetic service provider per capita data is not available in BC or other areas of Canada, so it is not possible to state whether per capita levels are significantly lower in rural areas. However, Canadian data does suggest that there is maldistribution of physicians, particularly specialists, in rural areas

(Rourke, 2008). For example, a recent Canadian study found the number of specialists per 10,000 people was just 1.6 in rural areas as compared to 9.6 per 10,000 in urban areas (Pong *et al*. 2011). As such it is reasonable to expect that there is also a smaller distribution of genetic specialists in rural versus urban areas. Furthermore, as in the US, geneticists and genetic counselors in Canada are more likely to practice in large academic medical centres located in major urban centres, representing a significant geographic coverage issue, in terms of travel time and distance, for those who currently require genetic counselling and other genetic services (see Figure 1.1, chapter one).

This access issue is of even greater concern with respect to the future of genetic medicine as large segments of the population have no genetic services and are thus ill equipped to deal with advances in technology, treatment and education for genetic conditions. Moreover, a lack of genetic knowledge among family physicians, who are the primary, and sometimes only, healthcare providers in these areas, (Frueh & Gurwitz, 2004) may compound this issue even further.

The above discussion illustrates that access to genetic services appears to be limited, at least in terms of proximity, particularly in rural communities. The underlying concern with lack of access to genetic services relates the actual impact of this limited access on those in rural communities. This impact is unfortunately difficult to assess due to lack of data and research in this arena (Braithwaite *et al*. 2004; Hogarth, Javitt, & Melzer, 2008), partly due to the relative novelty of the field. Nonetheless, genetic knowledge is believed to have multiple *potential* benefits for human health through the prevention, diagnosis and treatment of disease (Collins, *et*

al, 2003; Hunter, 2005). For example, there is thought to be a genetic component to most major common diseases (Provincial Advisory Committee on New Predictive Genetic Technologies, 2001). In cancer, one of the greatest causes of morbidity and mortality in the developed world, (WHO, 2007) the genetic contribution of disease is estimated at 5-10% (Garber & Offit, 2005). While extensive research efforts have not yet revealed the exact genetic basis of other common human diseases, genome wide association studies (GWAS) and other novel genetic technologies are thought to hold great promise (Wellcome Trust Case Control Consortium, 2007; McCarthy & Hirschhorn, 2008; Norrgard, 2008).

For example, recent GWAS studies have been successful in identifying susceptibility variants underlying many diseases thereby increasing the likelihood that this information, in the future, can be translated into meaningful clinical benefit by or through: new therapeutic targets, leading to new therapies for treatment or prevention; allowing identification of biomarkers which can improve disease prediction and monitoring of disease progression and response to therapy; elucidating environmental components of disease and allowing public health interventions; and more personalized approaches to disease management (McCarthy, *et al*., 2008).

Genomic medicine is also something envisioned to play an important role in decreasing the number of adverse drug reactions (ADRs), one of the leading causes of morbidity and mortality in the developed world. In the U.S. alone, ADRs are thought to be responsible for 100,000–218,000 deaths at a cost of $137–177 billion (USD) each year (Ernst & Grizzle, 2001). Although multiple factors, including patient age, drug interactions, environment and organ function are important in drug reactions, genetic factors are thought to account for 20–95% of

variation in drug response variability and as such make a significant contribution to the incidence and severity of ADRs (Ross, *et al*., 2007). Research into the area of pharmacogenetics – the study of gene-drug interactions – is thought to hold great promise for improving health outcomes, particularly in the area of complex disease (Craig, 2008).

Genetics have also been shown to play an important role in susceptibility to certain infectious diseases and reaction to antiviral treatments. For example, individuals with a mutation in their CCR5 gene are immune to infection by human immunodeficiency virus (HIV). This knowledge may lead to new strategies to prevent HIV infection (Craig, 2008; Hunter, 2005).

The above discussion has focussed on the many *potential* future applications of genetics in the clinical realm. Undoubtedly, much of the current research and excitement regarding genetic research has focussed on future possible uses and, at the present time, may be more 'hype' than actual realised application (Caulfield, 2000; Epstein, 2004; Ioannidis, 2009; Smith, *et al*., 2005). However there are some limited examples of the current clinical utility of genetic knowledge in both the diagnosis of Mendelian genetic disorders (i.e., single gene defects usually associated with rare genetic disorders) and in pharmacogenomics and therapeutics.

In terms of the latter, Knight believes that *"pharmacogenomics is likely to have the most immediate impact on the clinical practice of the general physician"* (Knight, 2009, p767). Perhaps the most successful example of pharmacogenetic research in terms of clinical utility is in warfarin treatment. Warfarin, also known by trade name as coumadin, is an anticoagulant with over 21 million perscriptions per year in the U.S. (Ginsburg & Willard, 2009). Research has

revealed that two genes, CYP2C9 and a VKORC, account for 57-63% of the variability in warfarin dose response (Takahashi, *et al*., 2006). Incorporating such knowledge into clinical practice by pre-prescription genotyping and individualized warfarin dosing has the potential to significantly reduce the risk of haemorrhage while maintaining effectiveness (Wadelius & Pirmohamed, 2006). In 2007 the U.S. Food and Drug Administration (FDA) updated warfarin labeling to include dosage guidance based on genotyping for CYP2C9 and VKORC and suggested physicians consider genotyping prior to prescribing the drug (Food and Drug Administration, 2007).

The warfarin case provides an example of how genetic research may be translated to actual *clinical* benefit. However, a study conducted by Eckman and colleagues into the actual cost-effectiveness of genotype guided warfarin dosing concluded that *"at its current cost, routine genotyping before warfarin dosing is not economically attractive"* (Eckman *et al*. 2009 p74). The study found that while genotyping did lead to better outcomes, the cost was $170,000 (USD) per quality-adjusted life-year, based on the current cost of genotyping of approximately $400 (USD). The authors concluded that the cost of genotyping would need to drop substantially, to approximately $100 (USD), for the test to be deemed cost-effective (Eckman, *et al*., 2009). This has led Iodannis to warn that such tests may not yet be ready for routine clinical practice and implementation (Iodannis, 2009). Unfortunately, analyses such as those conducted by Eckman and colleagues on the cost-effectiveness of warfarin are limited in number. This represents an opportunity for future research as more and more genetic services become ready for 'prime time' application, as discussed in more detail below.

In summary, while genetics has not yet demonstrated major benefit in terms of decreasing costs and improving clinical outcomes across the entire health spectrum, it does, nonetheless appear to have an important *potential* role in these areas in terms of how we diagnose, prevent, treat and practice medicine (Khoury, *et al*., 2008). The exact mechanisms by which laboratory successes will be translated into the clinical arena remain somewhat unclear, although there appears to be future utility in the areas of personalized medicine and pharmacogenomics, susceptibility and immunity, diagnosis and treatment protocols and preventative strategies. However, if access to genetic services is limited, one can hypothesize that these potential benefits may not be realised, particularly in remote areas where there is little or no availability of genetic services.

The problem of lack of access to genetic services is likely to increase as novel technologies and genetic therapies become available. There has also been a rapid decrease in the cost of such technologies, such as human exome sequencing, which has decreased from $2.3 million USD per sequence in 2003, to $5,000 USD in 2009 to less than $1,000 in 2012 (Lauerman, 2009; Markoff, 2012). This decrease in cost, together with rapid advancements in technologies and therapies, means that there is likely to be an improvement in the cost-benefit impact of genetic knowledge in the healthcare arena. However, unless there is research into this area and into the impact of access issues to genetic services, advances in genetics may lead to wider and wider discrepancies both within and between countries.

The above discussion has highlighted the fact that 1) the field of genetics has potential future clinical utility in the healthcare system, and 2) there appears to be a lack of access to such

services, particularly in rural areas, due to both a lack of knowledge about genetics among primary healthcare providers as well as a limited supply and distribution of genetic professionals. Unfortunately it is difficult to assess the impact of this lack of access due to the paucity of evidence and research exploring this issue and the fact that genetics is a relatively new and rapidly evolving area of healthcare. Recognizing these limitations, the discussion will now consider whether theories of justice in healthcare can be applied to this issue and specifically whether limited access to genetic services represents an injustice from an ethical perspective.

2.6 Theories of justice applied to genetic services

The introduction to justice and access to healthcare services has provided a context for further consideration of the main point of interest for this chapter, namely access to genetic services and justice. Egalitarian, capability and critical/ feminist theories have each provided an argument as to why, in a just society, access to healthcare ought to be considered a fundamental component of an individual's equality of opportunity and ability to be a fully functioning member of society. Thus, a lack of equal access to healthcare, and subsequent health, resulting from modifiable social processes, may be deemed a societal injustice.

Let us now consider how access to genetic services may be viewed in this context. The discussion at the beginning of this chapter focused on providing an argument for the increasing utility of genetic services in the healthcare setting. The discussion acknowledged that at the current time, genetic services are limited in their impact on healthcare (Grosse & Khoury, 2006; Scheuner & Rotter, 2006). Nonetheless, possible future directions for genetics in the healthcare arena were provided to show that genetics seems to have great *potential* to improve healthcare

outcomes through improvements in diagnosis, through a tailored approach to therapy and through decreasing adverse drug reactions (Gupta & Lee, 2007; Pagon, 2006). If the potential benefits to genetic services are realized, then it appears that such services *may* fall under the reasonable, basic minimum level of care needed to allow for normal species functioning and capability. To extend Daniels' egalitarian and Sen's capability argument further, unequal access to genetic services would result in an unlevel or unfair playing field, such that individuals were not given 'equality of opportunity', deemed to be a requirement of a just society. By the same token, it can then be argued that inequity in access to genetic services leads to unequal opportunity, and therefore represents an injustice in society, if due to modifiable social structures and processes.

This conclusion and the application of theories of justice to health and healthcare in the realm of genetics is, however, dependent on a number of factors, which also represent key areas for future research:

1) First and foremost, the above argument depends on genetic services proving to be of significant and tangible utility in the healthcare system (Khoury, Jones, & Grosse, 2006). For such a new field, this is difficult to assess, and there is a definite need for evidence based research (Khoury, *et al*., 2007) to explore the impact of genetic testing and services in the healthcare arena, both from a clinical outcome and economic perspective. Doing so will facilitate access to services in a just and efficient manner (Nunes, 2003). For example, in the case of warfarin, while there does appear to be clinical utility of genotype driven dosing, cost-effective analyses do not seem to support broad application at the current time. A larger number of such

studies will need to be completed in the future to determine the economic and clinical utility of new developments in genetics; without such research, genetics may see limited uptake in the healthcare system, or be prematurely or inappropriately applied resulting in a wasteful use of public resources. Further, for genetic services to be appropriately used, there must be a coordinated approach to allocation decisions; without such an approach, there may be *ad-hoc* decision making and implementation resulting in unfair distribution of resources (Adair, *et al*., 2009).

2) Second and somewhat related, there is insufficient research to determine the effect of unequal access to genetic services. While one might reasonably suppose that lack of access to a seemingly useful service would result in worse outcomes for those not receiving the service, there is little empirical evidence to validate this claim at the current time. However unless and until we have access to data determining the impact of unequal access, it is difficult to imply an inequity exists from the aforementioned justice perspectives. This is in line with the distinction made between healthcare and health status outlined at the beginning of this chapter. Lack of such evidence makes it difficult to make a case for the increased provision of genetic services, making the justice question difficult to resolve. Such research requires coordinated data collection and reporting both within and between nations. Without such research and understanding of the impact of access issues to genetic services, advances in genetics may lead to wider and wider disparities both within and between countries.

3) Third, it is unclear how genetic services compare to other healthcare services and whether, in rationing and distribution decisions, genetic services have more or less utility in

terms of actual clinical outcome than other medical specialties or more traditional public health measures. To aid rationing and distribution decisions, genetics needs to be contextualized in terms of established specialties such as cardiology and oncology. For example, consideration will need to be given to the utility of genetics in comparison to provision of cancer treatment and cholesterol lowering drugs. Similarly, if one is to increase access to genetic services, one must also consider the impact on macro allocation considerations such as basic public healthcare measures in the area of infectious disease. Of course, genetics is not the only field in which there are considerable medical advancements and future potential applications of research; thus genetics needs to be compared not only to current standards and provisions, but also account for future changes and improvements in other aspects of health. A further important consideration in this area is the actual type of healthcare service and delivery mechanism; for example, genetic services may not necessarily involve complex medical equipment and technologies such as MRIs and may be provided by using blood draws and informational sessions. Thus consideration needs to be given to needs and ease of service delivery, particularly in rural areas, where access to expensive technologies may represent more of a distribution issue than access to services, such as genetic counseling, which can potentially be provided relatively inexpensively and effectively via novel telecommunication methods (Baumanis, *et al*., 2009; Coelho, *et al*., 2005; Gattas, *et al*., 2001).

4) Fourth, the importance of the demographics of the society under consideration must be understood. At the current time it is unclear whether genetics may be more or less useful in certain populations. However, given that most developed nations have an aging population (WHO, 2012) it is plausible that the utility of genetics, in comparison to other areas of healthcare

may change in the future. In the case of less developed nations, there needs to be a consideration of available healthcare services in general, especially where there are rationing and priority based decisions to be made between the delivery of genetic versus other healthcare services.

5) Finally, genetics is a rapidly evolving technology, with ever-changing and broadening implications and applications in the healthcare arena. Given this fact, whether genetic services represent a 'reasonable or basic minimum' according to Daniels' or capability theories, must be determined with substantial evidence-based knowledge of the clinical, economic and ethical aspects of genetic service delivery. This also highlights the difficulty in determining what is 'medically necessary'; for example, the Canada Health Act does not consider alternative medicines, such as naturopathy and homeopathy to be 'medically necessary' so they are not usually covered by the Canadian healthcare system (Canada Health Act, 2005). This will need to be constantly re-evaluated over time, to account for the rapid evolution of genetics, not forgetting the context of genetics in other areas of healthcare provision, as discussed under point three above.

2.7 Conclusion

In conclusion, the limited access to genetic services appears to represent an injustice in the healthcare system in certain sectors. However, due to the rapid evolution of genetic services and potential improvements in clinical utility and cost-savings this may, in the future, represent a wider injustice in the healthcare system which may prohibit individuals from receiving the care, support and education they require. Given the existing access issues in rural areas, it is plausible to imagine that inequities in realizing the benefits of genetic knowledge may be particularly

acute in rural communities. However, for this claim to be strengthened, there is a mounting need for research into the area of genetic service delivery including: an exploration of rationing and distribution of genetic services, further research regarding the impact of services on human capability (morbidity and mortality), studies to determine which aspects of genetics are most salient in terms of improving outcomes in a cost-effective manner and finally, possible strategies to remove access barriers.

While limited access to genetic services does not, at the current time, represent a significant social injustice, and the above suggestions for research may help determine whether such an injustice exists in the future, we should not wait until such an injustice emerges before we consider strategies to ameliorate the problem. In actuality, research should focus not only on the utility and impact of genetic services, but should also simultaneously explore effective strategies to deliver and improve access to genetic resources not just for the rural, but for the general population as well. This strategy is a proactive rather than a reactive approach to health service delivery, and will potentially enable genetic services with proven utility to be implemented in a timely and efficient manner. Such an approach should, hopefully, result in access to genetic services being distributed in a way that can be deemed fair and equal from a social and ethical perspective.

Finally, while this chapter focuses on the ethical issue of justice, a discussion on the ethical issues involved in rural access to genetic services should acknowledge other potential ethical issues that might arise in implementing genetic services in rural areas. For example, people from rural areas receive medical care in a different social context than urban dwellers in

that there are close social networks and familiarities within a small community. Such familiarity raises questions of informed consent, confidentiality and privacy of information and threat of discrimination. In this context, principles such as respecting privacy and confidentiality to an individual, may actually conflict with a healthcare provider's obligation to families and the community (d'Agincourt-Canning, 2004). Thus, research aimed at preventing potential injustice in the translation of genetics into the clinical realm should also consider important psychosocial, familial and other ethical implications, as expanding access to services may not be the only ethical issue at stake. A further discussion of these ethical issues in the context of PT for HD, including autonomy, justice, beneficence and non-maleficence, will be discussed in the following chapter.

3. A GRAND CHALLENGE: PROVIDING BENEFITS OF CLINICAL GENETICS TO THOSE IN NEED

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3.1 Introduction

Genetics represents a specialized, novel and rapidly developing field of healthcare. A major issue is the limited number of training programs for geneticists and genetic counselors (Burke, *et al*., 2009) resulting in an inadequate supply of genetics professionals. For example, it is estimated that there are less than 3,000 genetic counselors in total worldwide (Edwards, Greenberg, & Sahhar, 2008). As was discussed in the previous chapters, compounding this issue is the paucity of access to genetic services in rural areas, as such services are usually confined to a limited number of large academic medical centers (Washington Department of Health, 2008). In the United States, there are over fifteen states with less than five reported genetic counselors, and several states which no counselors are reported (National Society of Genetic Counselors, 2008). For most rural residents accessing genetic centers means travelling to an urban center, often a considerable distance away.

The large gaps in service coverage for those outside urban centers represents a significant issue and deterrent for those who currently require genetic counseling and genetic services due to travel time, cost and distance. As with other areas of healthcare, access to services is a major barrier for rural populations creating not just a financial burden (e.g., travel, accommodation, time off work) but also a psychological barrier in leaving family and social supports to obtain necessary care (d'Agincourt-Canning, 2004). This issue is of even greater concern in terms of the future of genetic medicine as large segments of the population will be ill equipped to deal with advances in technology, treatment and education for inherited conditions (Khoury, 2003). Moreover, a lack of genetic knowledge among family physicians, who are the primary, and

sometimes the only, healthcare providers in these areas, will further compound this issue (Frueh & Gurwitz, 2004).

3.2 Predictive testing for Huntington disease: highlighting the issues

This issue is of particular relevance for a neurodegenerative disorder such as HD. Predictive genetic testing for HD has been available since 1986 and represented the first genetic test available for individuals at risk for an autosomal dominant illness (Brandt, *et al*., 1989; Fox, *et al*., 1989). Currently, the standard approach to such testing includes a protocol whereby at-risk individuals are required to make several visits to a genetics department to undergo genetic counseling, physical and psychosocial evaluation (Benjamin, *et al*., 1994; Went, 1994).

Uptake of PT rates for at-risk individuals ranges from 5 to 25% (Creighton, *et al*., 2003b). While such testing should be a thoughtful and individual choice, these uptake rates are low given that 65-80% of at-risk individuals suggested they would pursue such testing prior to the availability of the test (Hayden, 2000). Certainly the absence of therapy to alter the course of illness is one component of this limited uptake (Codori & Brandt, 1994). However other disincentives or barriers to testing need to be considered as they result in at-risk individuals being unable to realize the potential benefits of testing including: reproductive planning, relieving uncertainty, making insurance arrangements and getting access to services and support (Codori & Brandt, 1994; Decruyenaere, *et al*., 1996; Wiggins, *et al*., 1992). Furthermore, low uptake rates limit the number of individuals who may become aware of research opportunities which may lead to future treatments.

Notwithstanding personal reasons for choosing not to pursue testing, physical access to genetic services has been suggested to be a structural deterrent which acts as a disincentive to undergoing testing (Demyttenaere, *et al*., 1992; Evers-Kiebooms & Decruyenaere, 1998; Kessler, 1994). To provide granularity to this issue, the estimated prevalence of HD is 4-8 per 100,000 (Harper, 1992). In the US this translates into a minimum estimate of 30,000 affected individuals with a further 150,000 individuals at risk for the disease (National Center for Biotechnology Information, 2009). According to the NSGC, only 2% of genetic counselors practice in the area of neurogenetics (National Society of Genetic Counselors, 2008). Therefore even if *every* genetic counselor working in neurogenetics *only* saw only those at risk for HD, this would result in 3061 at-risk individuals for every one genetic counselor. This number, coupled with the above geographical constraints, means that under the traditional mode of genetic service delivery, individuals at risk or affected with HD may be unable to realize, or even have knowledge of, the potential benefits of future treatment, research and psychological support.

As novel therapies for HD become a tangible reality (Bonelli, Wenning, & Kapfhammer, 2004; Milnerwood, *et al*., 2010; Okamoto, *et al*., 2009; Walker, 2007), it is increasingly imperative to address and resolve this issue. Mutation positive individuals undergo neurological, cognitive and structural changes in discrete regions of the brain long before the physical manifestation of the disease (Tabrizi, *et al*., 2009). Thus to be most effective in protecting the vulnerable neurons affected by HD, it is likely that therapies will need to be in place prior to the onset of symptoms. This will provide a strong impetus for knowing genetic risk status early, so that preventative measures may be initiated before irreversible neuronal damage occurs.

3.3 Inequalities in access to genetic services

The fundamental concern illustrated by PT for HD is the impact of limited access to genetic services on people at risk for HD. This issue has much broader implications for clinical genetics in general as such knowledge - when accessible - may confer multiple benefits for human health through the prevention, diagnosis and treatment of disease (Collins, *et al*., 2003; Hunter, 2005). The elucidation of genetic information will be translated into meaningful clinical benefit by influencing modes of therapy; monitoring for adverse drug reactions for those at high risk; and facilitating a profoundly different approach to disease management, especially in cancer treatment (Knight, 2009; McCarthy, *et al*., 2008). In addition, genetic testing will likely become routine and require frequent use by practitioners not currently using genetic medicine routinely such as family physicians and oncologists. Genetic tests will aid these physicians in their practice with regard to medication prescription (to avoid adverse drug reactions), screening and treatment. Access to genetic testing and services is one of the 'grand challenges' of genomics that must be addressed to ensure genetics and genomics research is beneficial for everyone (Collins, *et al*., 2003).

The problem of lack of access to genetic services is likely to increase as novel technologies and genetic therapies become available. Decreasing costs, such as the promise of the $1,000 human genome (Lauerman, 2009), together with rapid advancements in technologies and therapies, suggest that there is going to be an improvement in the cost-benefit impact of genetic knowledge in the healthcare arena (Eckman, *et al*., 2009). However, unless there is further research to establish cost-effectiveness, as well as developing approaches to improve access, advances in genetics may lead to wider discrepancies both within and between countries.

This crisis raises significant ethical challenges. The intimate relationship between healthcare and health status means that barriers of access will likely result in worse health outcomes among rural populations (Peter, *et al*., 2001). This presents a potentially burgeoning inequity impacting an individuals' ability to function in society and live both a productive and unencumbered life (Alkire & Chen, 2004; Daniels, 2001; Ruger, 2004; Sen,1993; Williams-Jones & Burgess, 2006). As a result, some sectors of society will be unfairly compromised in terms of their health status in relation to others (Starfield, 2006). Given that rural populations are already more likely to be uninsured, have lower incomes and less education (Casey, Thiede, & Klingner, 2001; Zhang, Too, & Irwin, 2000), and may also contain more individuals from a specific minority groups (such as Native Americans and First Nations groups), already marginalized or disadvantaged groups may suffer from further disparity. While individual autonomy (e.g., personal characteristics, preferences and beliefs) remains a crucial component of realizing access to genetic services (Allin, 2008; Beauchamp & Childress, 2009), these personal characteristics are not socially determined (i.e., formed/ impacted by social policy). Improving access to healthcare involves removing structural barriers including access to services (Whitehead & Dahlgren, 2007).

This issue must be considered in relation to a country's stated policy toward, and degree of commitment to, universal access to healthcare. For example, national healthcare systems such as those found in Canada and most European jurisdictions are committed to providing reasonable access to medically necessary healthcare services across their population "*without financial or other barriers"* (Canada Health Act, 2005). In the US the position is more complex, with varying political and public opinions regarding universal healthcare and what constitutes a societal versus

a personal responsibility. Nonetheless, at a minimum there does appear to be a commitment and appeal to provide access to healthcare to those who desire, plan for and request such services. President Obama's campaign is committed to "*provide comprehensive and portable coverage for every American"* (Obama, 2008)[17]. In addition, the US Department of Health and Human Services vision includes consistently leveraging genomic medicine in clinical practices across the nation and suggests information technology is key to realizing this goal (Ginsburg & Willard, 2009).

3.4 Potential solutions

The first step in providing solutions to inequities in access to genetic services involves recognition of the problem and the realization that scientific discovery, novel treatments and advances in genetic knowledge are the beginning, not the end of the research endeavor. To maintain momentum and funding in the genetics arena it is imperative to realign research and financial resources to focus not just on discovery, but commitment to novel mechanisms and approaches to translation. Translation in this discussion relates not just to the production of new drugs and treatment options, but to actually translating research results into healthcare decision making and routine clinical practice. This requires a different set of research skills from creating new tests and therapies. To fully translate research from 'bench to bedside' requires developing and evaluating field studies in the clinical setting by drawing on a variety of disciplines in implemental science including policy, qualitative and mixed methods studies, behavioral science and epidemiology (Woolf, 2008).

[17] The Affordable Care Act (popularly known as 'Obamacare'), that was recently upheld by the Supreme Court, seeks to improve access to health insurance and healthcare, particularly for those from low income groups (Krugman, 2012).

Another essential component of resolving genetic access issues involves an in-depth, step-by-step, exploration of genetic counseling, testing and service modalities to highlight limitations and barriers. Important questions arise: What steps are needed to provide this service across geographical barriers? How can models of provision of this service (such as PT) be simplified without undermining their effectiveness? What partners need to be identified to implement more widespread testing? What are innovative solutions that may be adopted by other service providers? How do we determine the need for increased capacity? And, finally, how do we mobilize appropriate resources to implement structural changes that would fulfill our ethical and justice demands? Specifically in the case of HD PT these issues include: knowledge of genetics and HD and availability of testing by rural healthcare practitioners; understanding of the benefits and limitations of testing; and the ability to assess and provide psychological and technical support. Other areas of genetic testing, such as genetic screening prior to medication prescription (pharmacogenomics), require a completely different approach, as these tests may not necessarily require a specialized genetics practitioner. Issues to consider in this area will include: appropriate education of family physicians and rural nurse practitioners; the ability to provide testing and results quickly and clearly; and ensuring widespread uptake of testing with proven benefit. Recognizing that removing access barriers to genetic services will require multiple and different strategies, depending on the specific service to be provided.

Other areas of success in the healthcare delivery sector broadly provide insights that can be applied to this situation. For example, successes in immunization and awareness campaigns in the public health arena provide some insights relevant to application and expansion of clinical genetic services. These include educating the public and healthcare providers through novel

education programs that are easily implemented, efficient, effective and scalable. In addition, the success of vaccination campaigns involved exploring and addressing local barriers to services, such as cultural perspectives. This may be of particular relevance for communities less trusting of genetics, such as the African American or Aboriginal populations who are less likely to use genetic tests due to past discrimination (Arbour & Cook, 2006; Corbie-Smith, Thomas, & St George, 2002). Such considerations are key to developing culturally sensitive counseling and genetic testing protocols, contributing to long-term acceptability and uptake of genetic tests (Lerman, *et al*., 1999).

Furthermore, coordinating efforts, maintaining flexibility and pursuing innovative methods is crucial to developing effective, efficient and expandable strategies for equitable uptake of genetic services. Such mechanisms include the use of novel telemedicine strategies that may also allow assessment of a person's physical status remotely. This will also entail awareness campaigns to increase education among primary healthcare providers and the public. With telemedicine, genetic services, education and counseling are provided remotely via telephone, videoconferencing or using internet based services such as Skype. Until this point telemedicine techniques have been used in a limited capacity for clinical genetics services, primarily in the area of cancer genetics, to provide genetic counseling, test results and follow-up care via videoconference and telephone to individuals in rural communities. Small preliminary studies have suggested that such approaches are successful, culturally acceptable and welcome (Baumanis, *et al*., 2009; Coelho, *et al*., 2005). There is a need for large scale research into outcomes, together with coordinated capacity building in this area (particularly in other areas of

genetics), as well as follow-up and assessment of patient experience and satisfaction with such services.

Another critical component to assure the successful uptake of strategies aimed at improving access to genetic services involves ensuring that novel approaches have the support of health insurance companies (public and private) and disease advocacy groups. To realize this, data on efficacy and cost-effectiveness – taking into account both the cost of the genetic test and service provision - will be helpful. Lack of proof of cost-effectiveness has ethical implications on allocation scarce resources and represents a key factor in the poor uptake of genotyping prior to warfarin prescription in the US (Eckman, *et al*., 2009). Moreover research results and findings must be conveyed to policy-makers, healthcare professionals and the public in an accessible and direct manner via face-to-face meetings, as well as marketing and awareness campaigns. Developing strategies for translating research discoveries and pilot project findings will be essential to improving healthcare for the whole community. Building capacity in each of these areas will enable genetic technologies with proven utility to be implemented in a timely and effective manner and result in improved fairness and more equal distribution of genetic services for society as a whole, not just those fortunate enough to live close to a major genetic center.

4. LESSONS FROM PREDICTIVE TESTING FOR HUNTINGTON DISEASE – 25 YEARS ON

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4.1 The increased availability of predictive genetic tests

The availability of predictive genetic tests has rapidly expanded in the last two decades. We can now provide testing for a range of adult-onset conditions including certain cancers, metabolic diseases, cardiac diseases and neurological disorders. These developments have recognized benefit including determining the necessity of additional screening or preventative options, relieving uncertainty and informing reproductive planning However, despite these benefits, predictive tests raise questions about the ethical delivery of genetic testing, results and services. To respond to these challenges, PT protocols, such as those for HD, have required several in-person appointments, spread over several weeks or months, in order to undergo counseling, testing and receive test results (Went, 1994). Originally, these multi-step, multi-visit protocols were developed to both protect individuals from the potential for serious psychological damage from receiving increased risk results, as well as to ensure that individuals undergoing testing made a fully considered decision. In addition, incorporating post-test result appointments into the testing protocol aimed to ensure adequate follow-up care and support.

4.2 Bioethical principles and predictive genetic testing

By providing opportunities for informed decision making and reduced potential harms, such guidelines respect some of the ethical principles outlined in chapter two and three which form the foundation our healthcare system, namely: 1) Respect for Autonomy; 2) Nonmaleficence; 3) Beneficence and, 4) Justice (Beauchamp & Childress, 2009). While some suggest that the first of these - autonomy - holds a place of primacy in the North American medical system, all are considered important.

4.3 Challenging the current predictive testing models

However, for HD, after 25 years of predictive test experience, a fresh look at PT raises the question as to whether all of these ethical principles have been equally upheld by the current testing protocols. For example, while testing protocols were designed to 'do no harm' by reducing or avoiding psychological impact, studies tracking individuals for several years following test results have revealed that initial concerns of frequent immediate and long term adverse events (e.g., suicide and depression) have not been substantiated (Broadstock, *et al*., 2000). Moreover, an increasing recognition of the importance of patient-centered care has drawn attention to suggestions that instead of respecting autonomy, lengthy testing protocols could, in fact, be viewed as paternalistic, overly time consuming and infringing upon a person's 'right to know' (Dufrasne, *et al*., 2010; Kessler, 1994). Finally, requiring multiple, in-person appointments may contravene the principle of justice and fair resource allocation, as those who do not reside in a major urban centre face barriers to access (in terms of costs, time away from family and stress) (d'Agincourt-Canning, 2004; Hawkins & Hayden, 2011). The latter issue may even be harmful in nature; if those who may benefit from PT have restricted access to genetic services, they may be unable to realize the benefits of testing (Hawkins & Hayden, 2011; Williams-Jones & Burgess, 2006). These benefits include the possibility of screening and intervention either currently available or foreseen in the future (Milnerwood, *et al*., 2010; Walker, 2007). The issues of access also infringes upon personal autonomy in that it limits choice and informed decision making.

4.4 Implications for other areas of predictive genetic testing

While the HD PT paradigm provides an elegant illustration of the problem of providing PT in an ethical manner, the issue is not unique to HD. It is directly relevant to other areas of clinical genetics such as susceptibility testing for Alzheimer's disease and across multiple hereditary cancer predispositions. Calling attention to the inadequacy of current rigid testing practices underscores the need for pragmatism and flexibility in exploring novel mechanisms to provide predictive genetic testing and services in a patient-centered, respectful, just and beneficial manner. In some areas of genetics this evolution and exploration has begun. This is most notable in the area of cancer genetic counseling where novel telemedicine approaches have proved successful, not just in terms of patient satisfaction and well-being (Baumanis, *et al*., 2009; Coelho, *et al*., 2005), but also in terms of cost-effectiveness (Buchanan, *et al*., 2011). Such forward-thinking approaches promote the accessibility and portability of beneficial clinical services.

4.5 Potential solutions

Nevertheless, more consideration and research is needed to determine flexible patient centred approaches to PT. The issues, benefits and risks need to be carefully assessed and discussed. In agreement with Knoppers and Chadwick, we think that bioethics principles can and should evolve in their application as new health technologies, such as predictive genetic testing, become available (Knoppers & Chadwick, 2006). As such we have to move away from rigid testing practices and adapt our policies and protocols so that they are tailored to individuals' needs, circumstances and constraints. For HD, this may exceptionally mean testing persons

below the age of eighteen, testing persons at 25% risk and also devising different strategies to deliver PT to those living distant from a genetic centre. Doing so requires prospective, controlled studies to design and evaluate novel customized approaches to testing that respect the diversity of the populations we serve. In the case of HD, such a collaborative tailored approach could involve providing pre-test counseling via web-based education and telemedicine methods, and incorporating novel communication tools including local delivery of results with the aid of trained and trusted community healthcare provider. The objective is to provide testing in an ethical and, patient-centred manner that improves the provider-patient relationship while maintaining support and the importance of informed consent.

The need to challenge and rethink current practices in the delivery of genetic testing, and to respond to patient concerns will facilitate novel approaches and adaptive solutions in the best interest of serving our patients. In the next chapter I will illustrate this challenge, and potential solutions, with the example of HD testing in BC.

5. WHEN ACCESS IS AN ISSUE: EXPLORING BARRIERS TO PREDICTIVE TESTING FOR HUNTINGTON DISEASE IN BRITISH COLUMBIA, CANADA

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5.1 Introduction

As noted in chapter one, HD, is a neurodegenerative disorder characterized by chorea, dementia, personality and mood disturbances (Walker, 2007). It is inherited in an autosomal dominant fashion and is caused by a CAG expansion mutation in the HD gene (MacDonald, *et al*., 1993). Although age of onset varies, the mean age of first symptoms is 35 - 50 years. There is a progression of the cognitive, movement and psychiatric symptoms with death usually occurring 15 to 20 years after first symptoms (Hayden, 1981; Walker, 2007). There is no treatment to alter disease course however standards of care for managing HD do exist (Rae & Simpson, 2012).

PT for the disorder has been available since 1986 (via linkage) (Brandt, *et al*., 1989) and via direct mutation testing since 1993 (Kremer, *et al*., 1994). PT to determine risk status enables people to access benefits including: relieving uncertainty, reproductive planning, improved ability to plan for the future, access to support, and research opportunities (Codori & Brandt, 1994; Creighton, *et al*., 2003b; Wiggins, *et al*., 1992). In keeping with the International HD PT guidelines (Went, 1994), the PT process in BC, Canada usually involves three to four in-person appointments at the Centre for HD in Vancouver to undergo clinical evaluation, genetic counseling and education, psychosocial evaluation, results and follow-up (Benjamin, *et al*., 1994; Went, 1994) (table 5.1). This process usually takes 4 weeks from initial appointment to the results session, and individuals are encouraged to bring a support person (e.g., spouse), particularly to the results session (although this is not required). Depending on specific circumstances (including distance to Vancouver) this protocol may be adapted so that only one appointment occurs in Vancouver, with the remainder of the sessions and results being provided

by a local GP with the guidance and support of the Centre for HD genetic counselor. The protocol was designed to promote informed decision-making and to help protect individuals from the potential for serious psychological injury of receiving HD test results.

Table 5.1: The predictive testing process in British Columbia

Appointment	Timeline	Topics covered/ session content
1: General counseling session	Week 1	1. Psychological Screening Questionnaires 2. Neurological Exam (if possible) 3. Genetic counseling & info about PT and PT protocol 4. Reasons for PT 5. Exploration of family and social support
2: Pre-results session	Week 3- 4	1. Participant's decision 2. Psychological & social preparation for results
3: Results session	Week 4 - 5	Results of PT
4: Post-results sessions	Week 6-7 & 6 months post results	Integration of results, coping & further support

Despite the availability of comprehensive PT programs, PT rates among individuals at risk for HD are lower than the 65-80% initially expected (Hayden, 2000; Meissen & Berchek, 1987) with reported uptake rates ranging from 5 to 25% (Creighton, *et al*., 2003b; Hayden, 2000; Morrison, *et al*., 2011; Tassicker, *et al*., 2008). While PT for HD should be a carefully considered individual choice, there may be barriers to testing so that at risk individuals who would like to pursue testing cannot realise the potential benefits of PT (Hawkins & Hayden, 2011; Hawkins, Ho & Hayden, 2011). Although the cost of PT is likely to be a significant barrier to testing in countries where PT is not covered by the healthcare system (PT is covered in Canada), other barriers to PT are likely to exist. One such barrier, particularly relevant in BC, may be the accessibility of genetic services (Demyttenaere, *et al*., 1992; Evers-Kiebooms &

Decruyenaere, 1998; Kessler, 1994). The PT uptake rate in BC is 25%. To understand predictive test uptake variation across the Province further, we conducted a systematic chart review of the location of each individual who had undergone PT at the time they underwent testing (Figure 5.1). Data points were then applied to a map of BC, and organized by region according to census classifications. Individual test locations were then stratified as rural (areas more than two hours drive from the PT clinic) versus non-rural areas. Comparison of PT figures in the rural versus non-rural groups reveals that testing rates are lower in the non- rural areas with PT uptake rates of those at 50% risk for HD at 30% for non-rural populations and 16% for rural populations. These numbers are based on the actual number of tests in the study period / actual number of individuals at 50% risk and eligible for testing to avoid the recognised difficulties in accurate uptake calculations (Tassicker, *et al*., 2008). These data suggest that access may be a barrier to testing, particularly for those living in rural areas.

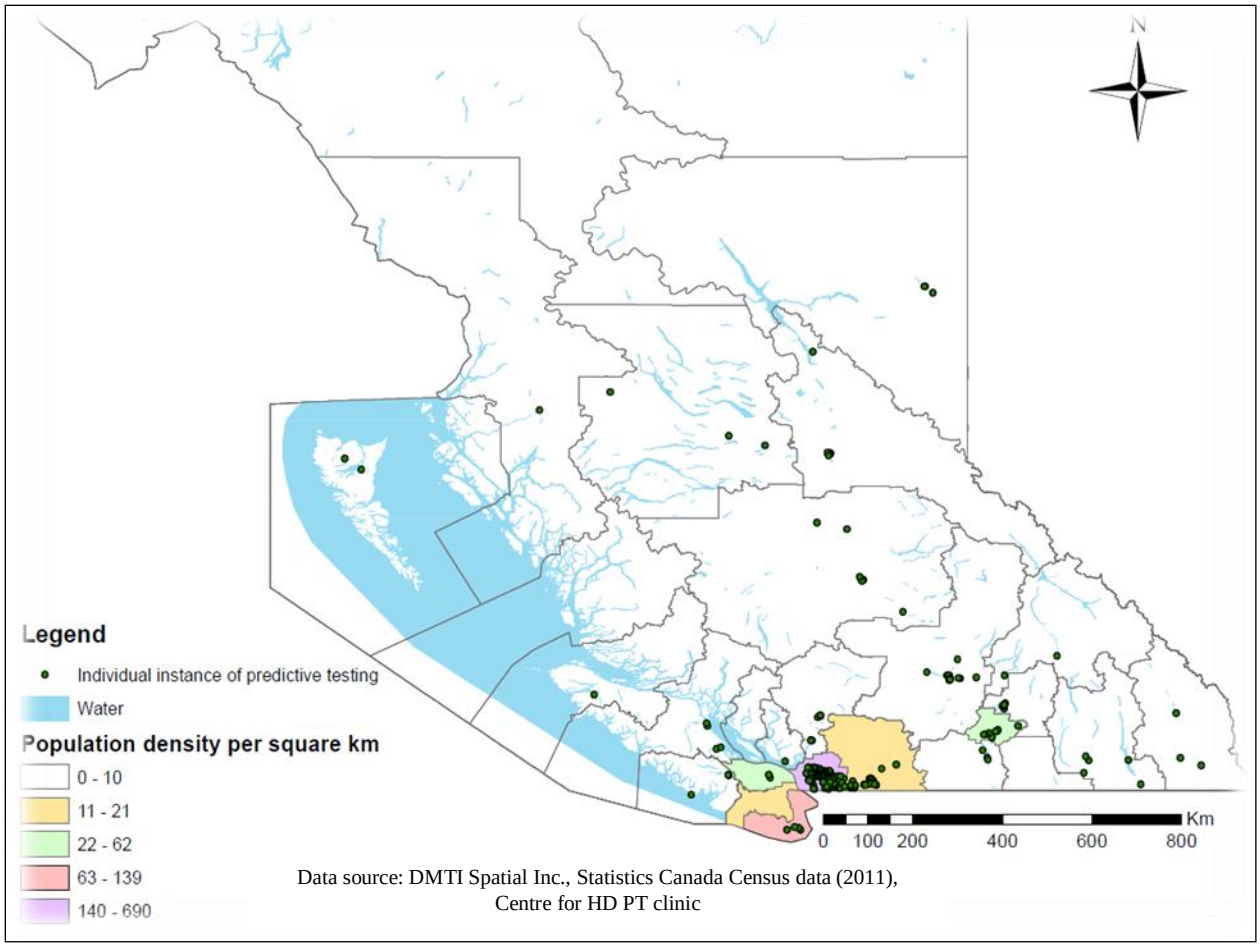


Figure 5.1: Map of British Columbia illustrating the locations of individuals who have undergone predictive testing (1993-2010)

In order to understand and explore whether accessibility of PT services is a barrier to testing, we conducted an interview study of individuals at risk for HD throughout BC. BC has a population of approximately 4.5 million people, 2.5 million of whom reside within a 2 hour drive of downtown Vancouver (Canada, 2010). The remainder of the population is widely dispersed and may have to travel for up to 24 hours to reach the PT centre in Vancouver. This research sought to understand the obstacles to testing in terms of the accessibility of services in

Vancouver, as well as exploring the mechanisms by which this issue may be addressed. Research Ethics approval was obtained for this study.

5.2 Study sample and recruitment

Interview recruitment was based on a purposive non-probability sample[18] with respondents chosen based on availability and convenience. Recruitment aimed for a diverse sample stratified for location (rural versus non rural), testing status (tested versus not tested) and test result (CAG in the disease range versus normal CAG size). These categories were chosen to ensure a range of viewpoints and to minimize bias toward a particular sub-group (e.g., individuals with a high-risk result might perceive the process differently than someone with a normal result). While every effort was made to obtain a similar number of participants in each stratification, this was not always possible due to: 1) the demographic distribution of BC; 2) people who had not undergone PT were more difficult to recruit as they were not as likely to be known (e.g., not on relevant mailing lists); and 3) of those tested, individuals who had a CAG expansion were more likely to be engaged and willing to be involved in research (those who received a normal result were less likely to stay active in HD research).

Recruitment involved a multi-pronged approach. First, patients who had gone through PT within the last five years through the Centre for HD were contacted via letter and returned a form if willing to participate. Second, to access those at risk but not tested, we sent notice of the study via the Huntington Society of Canada, BC Chapter newsletter, and also to those on the Centre for

[18] A non-probability sample does not involve random sampling. This method was chosen in order to achieve a diverse sample to recruit participants from a number of groups (rural, non-rural, tested, non-tested, HD+, HD-).

HD study database. Finally, we used snowball-sampling to recruit family members of those who had already been interviewed. After initial interest was confirmed, all participants were asked to sign a written consent form agreeing to participate.

5.3 Data collection

Interviews were conducted in person or via phone and usually lasted 45-60 minutes. Interviews consisted of open-ended questions focussing on: 1) perceptions of the PT process; 2) experience of considering and/or undergoing PT; 3) barriers to testing; 4) assessment of the key components of the PT process; and 5) opinions of providing PT remotely. Interviews were tailored to the participant based on whether they had undergone PT, their location and their at risk status (if known). Interviews also included a number of questions regarding HD PT resource needs and decision-making aids, the results of which will be discussed in chapter six. Interviews were conducted until theoretical saturation was reached, which occurs when additional interviews provide repeated themes but no further insight (Creswell, 2009; Strauss & Corbin, 1998).

5.4 Data analysis

All interviews were recorded, transcribed and entered into analysis software (NVIVO 9, QSR INTERNATIONAL). Transcripts were read several times to generate categories of information and reveal prominent themes. Data were then coded for themes and subjected to thematic analysis, adjusting coding methods until saturation was reached (Creswell, 2009). The interview,

transcription and coding process occurred concurrently so that emerging concepts or questions informed subsequent interviews to improve focus, clarity and granularity on specific issues (Charmaz, 2006; Starks & Brown Trinidad, 2007). This analysis method, known as constant comparison (Corbin & Strauss, 2008), allows a conceptual framework to be developed, and subsequently refined and verified, to ensure the final categories form a comprehensive model of the data.

5.5 Results

5.5.1 Sample characteristics

A total of 33 interviews were conducted for this study, with participants stratified by location, testing and result status, as outlined in Figure 5.2.

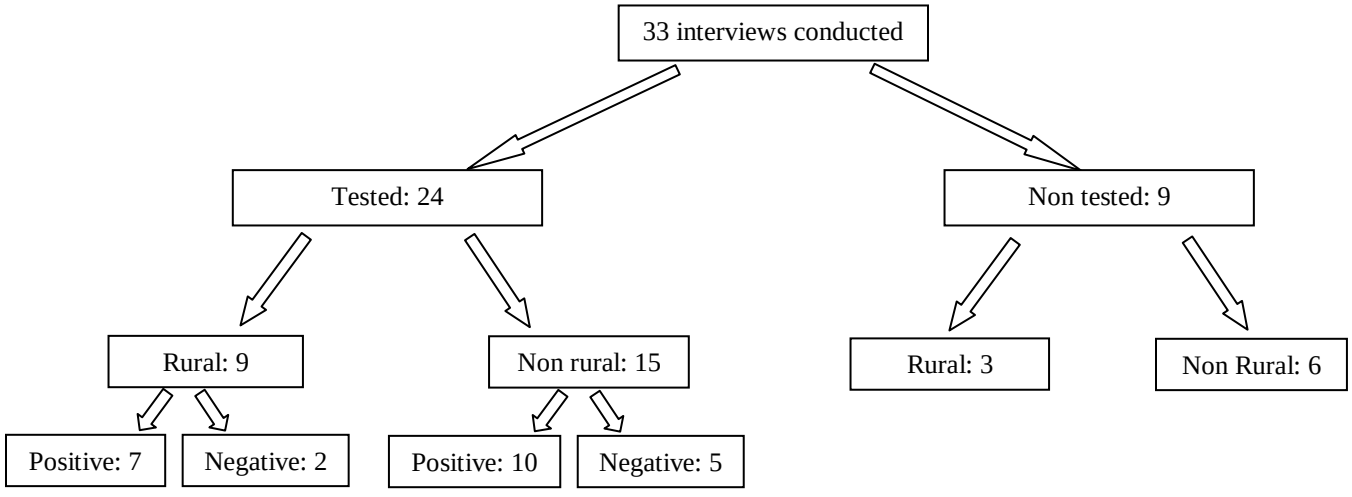


Figure 5.2: Interview participant characteristics

5.5.2 Accessibility of predictive testing services as a barrier to testing

Results of the interview study reveal that the accessibility of PT is a significant barrier for those considering PT for HD in BC. Detailed analysis of the interview transcripts reveals that accessibility is a complex barrier, and can be broken down into two major sub-barriers or themes. The first of these, distance, is structural in nature and relates to difficulties such as travel, financial and opportunity cost of attending sessions, stress related to travel and distance from support. The second major sub-barrier that emerged relates to accessibility of the actual testing process including counseling and support person requirements, the structure of the results session and the process length. These barriers and sub-barriers are outlined in Figure 5.3, and described in more detail below. Direct interview participant quotations have been used where relevant to highlight specific issues. While this discussion focuses on access barriers to PT, factors that improved access (such as travel reimbursement and use of local GPs to deliver results) have also been highlighted.

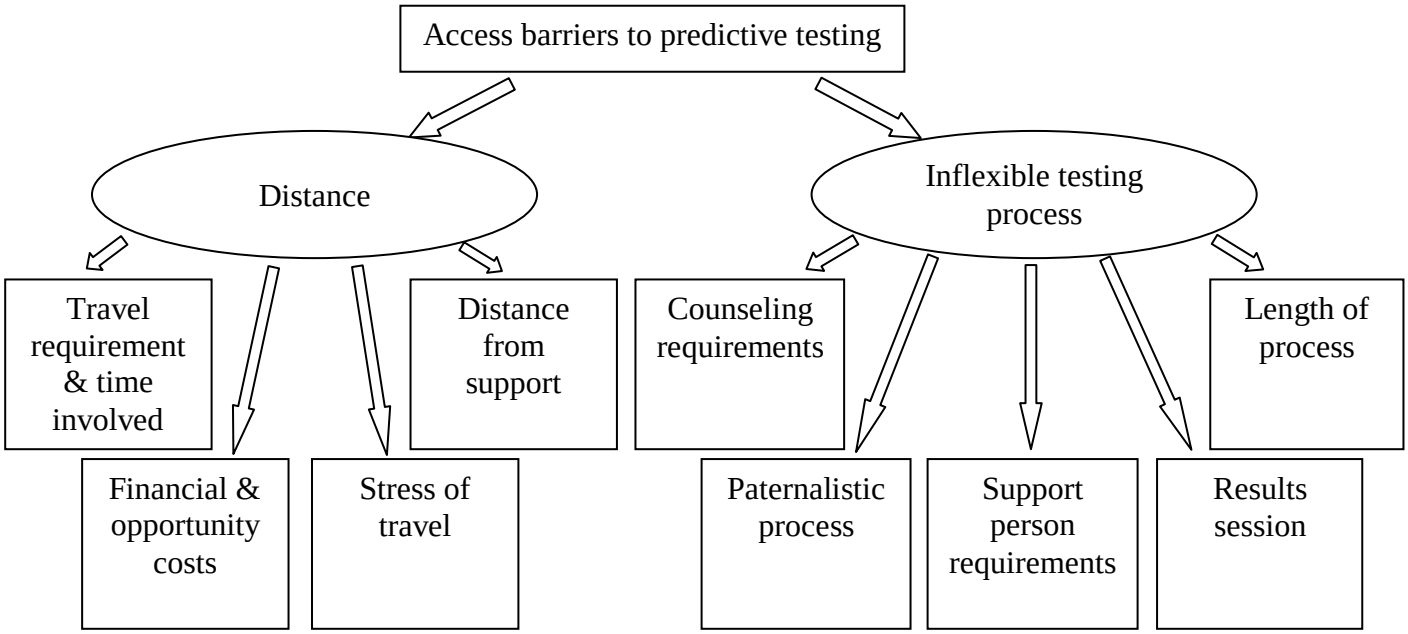


Figure 5.3: Access barriers to predictive testing: themes revealed in the interview analysis

5.5.3 Distance

The first major barrier in accessing PT that emerged through the interview analysis related to distance to the clinic in Vancouver.

Travel requirement and time involved

Perhaps the most obvious access barrier is the necessity of travelling to undergo the appointments typically involved in the PT process. This was of particular concern for those from rural communities who need to take a variety of means of transportation (e.g., airplane, ferry, bus) for long periods. As one rural respondent (not-tested) explains, this can be a big commitment: *"We'd get up at 4 o'clock, we wouldn't get back till 10 o'clock that night....that was a lot, that was a big commitment."* Others noted the financial and time considerations involved

with such travel: *"not everybody is going to be willing to travel like that. Some people can't afford it, some people just don't have the time"*. Another participant reported delaying results for two years due to distance and travel time: *"I would have had to take so much time off work that it would have cost me a lot of money to go to Vancouver. The whole reason we ended up going in March was because we were going on holiday and we decided to try to fly through Vancouver…so the process actually made me wait longer to go ahead with my results*".

Several interview respondents noted that siblings and other family members living in more rural communities had not undergone testing and that the *"main drawback for them was distance"*. As one individual living close to Vancouver explained, *"my dad was...living in a rural area, very rural. He had to come [to the clinic] and it was just terrible. He had to keep coming back and forth and it was really hard on him... He left...being told that he had Huntington's...I made him stay with us for four days...it's an eight hour drive on a long road."*

Financial and opportunity costs

While those from rural areas of BC may be eligible for provincial financial support and assistance in travelling to medical centres, this only goes some way to improving access. Shortcomings of this system were noted including length of the reimbursement process and difficulties in obtaining/ and completing appropriate forms. Moreover, this assistance does not address lost opportunity costs including the need to take time away from work and family. This is particularly relevant for those with young children or those who may be caring for affected relatives. As one participant describes, these issues played a role in her decision not to undergo

testing: "*I was a working mom. I couldn't take the time off to go down and do that…Distance definitely played a part [in not having testing].*"

Stress of travel

Somewhat surprisingly, access was an issue even for those relatively close to Vancouver due to the stress of driving to the city and rush hour traffic that could result in a three hour journey home. These individuals felt that they needed to take a whole day off for an appointment. As one respondent voiced *"it would make it easier if we didn't have to go to the clinic. [It] is forever...away.*"

Distance from support

Others noted that when receiving results they had had to travel to the HD clinic very early (or the day before) and then had to return home at the end of the day. The individual receiving results usually had to attend the appointment alone (as the support person could not afford to take time off). He/she then faced a long journey home before he/she reached his/her support system to begin to deal with and process his/her results. One individual, who received results from his family doctor, described how important it had been for him to receive the results in his local community, on his terms (including bringing a beer to the results session): *"[My doctor] said he'll make it the last appointment so there's no distractions and lots of time... we were there for a couple of hours and I drank my beer and he shook my hand and gave me a hug and apologized...But all the supports were in place. I knew exactly what the plan was."* This individual also noted that the possibility of having results delivered by his local GP (rather than the clinic in Vancouver) had facilitated his access to PT.

5.5.4 Inflexible testing process

The second major barrier that emerged from the interviews related to the accessibility of the actual testing process. These concerns related to the inflexible nature of the process, which did not take into account an individual's specific circumstances and needs.

Length of the testing process

Interview respondents felt that the time it took to go through testing was lengthy and frustrating. The initial waiting period was a source of frustration for those who had "*made up their minds*" and were "*on a mission*" to receive results. As one interview respondent explained: "*Just tell me what I'm dealing with so I can move on and make plans"*. Some respondents voiced this frustration more strongly: "*I hate waiting. I wanted to do it"* and *"[the waiting process is] just gut wrenching".* Many participants, including tested and non-tested, also perceived the number of appointments to be somewhat unnecessary. This may have detrimental effects on other family members and relationships: *"I think the time frame was just too long. It doesn't need to be....I just found it very frustrating to have to wait…And I came to a point—well my dad, he didn't even know. He died. I didn't even get to tell him actually—tell him that I tested positive. So it was really heartbreaking, because he died the same year....I wanted him to know."*

Paternalistic process

While the testing process was seen as too lengthy by some, several interview participants mentioned that in retrospect they understood why the process was so involved. Others felt the process could have been "*somehow optional or individually tailored"* to the needs of the specific patient being seen. They felt that the process was somewhat paternalistic in that they already

understood and knew the disease and the ramifications of PT. Some felt that the counseling prior to testing and psychological workup involved *"too much handholding"* and that they were *"over babied."* Others felt there were too many questions regarding depression and suicide and *"a lot of things that I don't have... They dwelled on it a lot....I didn't think was necessary for me."*

Counseling requirements

Similarly, for both those who had and had not had testing, the length of the PT process, and counseling involved (whether real or perceived) was a barrier to testing. For example, one respondent recalled his reaction to hearing what was required: *"When we first got the protocol related to the testing and the three or four visits that were required and all of that, talking amongst my siblings, it was kind of like, why? Why do we have so many?"* One individual who had not undergone testing explains this barrier further: "*I'm assuming that I'm going to be smothered with "help" when I get my results back that are a positive that I do have HD"*. The perception that the PT process was smothering played a role in this individuals' decision not to be tested.

Results session

The desire for a personalized approach is perhaps best illustrated by viewpoints on the results session. Respondents felt that the way results should be delivered is a very personal preference. Most preferred receiving results in person, as they felt it was better to receive results from someone who they knew or had a personal connection too. Respondents voiced concern that phoned results, or where you could not physically see the person, would make it difficult for the person delivering results to evaluate the well-being and emotional state of the person

receiving results. As one respondent notes: "f*or me the personal contact was crucial"* that felt "*almost like a cushion"*. However, while most respondents indicated that they felt in person result reporting was *"preferable"* many also acknowledged that for some people this would not be possible, or desirable. Some respondents said that if they had received results via phone they *"would have been just as okay as I was with it being done in person"*.

There were mixed views as to whether it was preferable to receive results from a local family physician or from the HD testing center. For example, those who said they preferred to be at the HD clinic when receiving results indicated that they could *"trust the results",* and they were getting the *"best of the best care"* and *"expertise".* For those who thought it preferable to receive their results from their local GP a number of reasons were articulated including: being in a supportive environment (i.e., close to home); the support person could easily attend; and having a good 'rapport' or history with their GP (so they felt comfortable/ trusted their GP). As one rural participant stated regarding receiving her result from her GP, *"it was better for me to be here and just come home and deal with it on my own."* However some respondents voiced concern about the knowledge and expertise of their local GP in terms of providing counseling and results for HD PT.

Some respondents would prefer to receive results alone so they could process and reflect. One respondent felt strongly about privacy when receiving results and compared it to receiving a school report: *"It's like making somebody open their report card when it's not a good one in front of somebody…[and saying] 'Oh, let's see how she's going to respond.'…Did you open your report card at school among all your friends? I sure as heck didn't… [I want to] go home, I*

want to close the door, I want to see it and then go, "Okay, I understand why I've got it or got that mark or got the positive. And okay, we'll put on a bright face and carry on."

Support person requirement

Finally, some participants voiced annoyance at the requirement that a support person be present for results (this is usually standard practice for patients undergoing PT). Some felt that by saying they would rather receive results alone they would be incorrectly characterised as being *"in denial"* or *"at risk"* when actually they were just an independent person who preferred to "*just deal with it."* As one individual who decide not to proceed with testing explains: *"I don't want my husband hugging me and telling me, "Oh, everything will be fine," until I am fine with it...I would need...a period of time to process it myself."*

5.6 Discussion

The results of the interview findings reveal that the accessibility of PT can be a barrier to testing for two major reasons: distance and the rigidity of the current testing process. The first of these, distance, is not unique to HD, and has been seen in other areas of healthcare, including other genetic services. For example, in an examination of hereditary cancer services, d'Agincourt-Canning noted that distance posed a major barrier for rural communities: "*Not only does it create an added financial burden (travel, accommodation/meals and time away from work) but people must leave their family and social supports to get the medical services they need"*(d'Agincourt-Canning, 2004, p222). The second barrier that emerged from the data is the inflexibility of the testing process - whereby the testing protocol itself may discourage testing (Kessler, 1994).

Together the barriers revealed by this study may act as a substantial impediment to individuals receiving the counseling, support and information they require. Addressing these barriers is important on several different levels. First, appropriate access to PT services allows people autonomy in their healthcare decision-making (Hawkins, Ho & Hayden, 2011). While we do not advocate all at risk for HD undergo PT, individuals at risk for the devastating condition need to be given the appropriate opportunity to discuss PT and receive information on the pros and cons of receiving results. In addition, failing to understand an individual's circumstances may have detrimental effects on the patient, as illustrated by the respondent who relayed the devastation at not being able to discuss her results with her father before he passed away.

The second fundamental reason we need to consider the accessibility of PT relates to broader considerations of equity in healthcare. While inequity in the provision of specialist services is not a new phenomenon, nor unique to genetic services, consideration of solutions that mitigate these inequities is warranted. The current testing model may be restrictive and inflexible in its requirements and discriminates against those who do not access PT services for either physical or structural reasons, or for emotional and psychological reasons. Most obviously, this may prevent comprehensive PT. However, perception of PT (whether realistic or not) is also a concern as it influences participation in PT initiation or continuation.

Finally, these findings are relevant to other jurisdictions. While BC's geography and healthcare system differ from other areas of the world, the results are important specifically because they reveal accessibility of testing can be a barrier for diverse reasons. Barriers are not

just relevant to those living in remote locations, as even those living within close proximity may encounter georgraphic, financial or other access deterrents. These results underscore the importance of understanding and exploring these factors on both a local and individual level. For example, despite the fact that BC has a relatively dispersed populus, it has one of the highest uptake rates of PT, likely due to factors such as universal healthcare coverage and the prominence and history of research and clinical care provided by the Centre for HD. These contextual factors are also important in understanding differences in uptake rates of PT in other countries and jurisdictions, and only by examining potential barriers and contextual factors on a local level can we begin to address and remove these barriers more broadly.

Solutions to improve access to PT need to be flexible and take into account individuals' circumstances and needs. For example, the concern that the process is over-burdensome and smothering can be addressed via 1) education regarding PT and 2) adapting and personalizing the testing process to an individual's circumstances (Hawkins & Hayden, 2011). In addition, that some participants felt that receiving results from their local GP had been (or would be) successful, illustrates another potential mechanism to improve access. Creating solutions also requires recognition that access barriers are complex issues that may not be resolved by, for example, simply providing travel assistance (although this did help improve access to some extent). Moreover the impact of other established reasons why people do not pursue PT should not be negated. These factors, including views about controlling the future, family attitudes and norms, financial/ insurance implications, the impact on others and the potential for discrimination are recognised as playing an important role in the decision-making process (Binedell, *et al*., 1998; Bombard, *et al*., 2009). As such, the aim of mechanisms to reduce access

barriers is to ensure that such barriers are not the only reason that individuals do not pursue testing.

PT can be provided relatively inexpensively and effectively via novel telecommunication methods such as videoconferencing. Telemedicine in genetics is not new, and has been successfully used to improve access in other areas which require complex counseling and decision-making, such as hereditary cancer (Baumanis, *et al*., 2009; Coelho, *et al*., 2005; Gattas, *et al*., 2001; Stalker, *et al*., 2006). Moreover studies suggest that telegenetics is successful and preferred by some individuals, and may also result in more cost-effective services. Telemedicine also has potential to improve access to services for those in less developed nations (Wootton & Bonnardot, 2010), where access to specialized genetic professionals may be limited, at best. As a result of this interview study, BC is now evaluating the provision of PT via telehealth, with the support of a local GP. This telehealth model is in keeping with the new international PT guidelines for HD (Macleod et. al, 2012), and preliminary analysis reveals patient satisfaction and well-being measures, throughout the PT process, maintain the quality of care and support necessary for those undergoing PT.

This study has a number of limitations. First, we acknowledge that the results may not be applicable in other healthcare regions. However, given the low number of genetics professionals, and concentration of such professionals in large, urban, academic medical centres (National Human Genome Research Institute, 2006) it is likely that these access issues are similar in other jurisdictions. Second, due to the nature of the study, recruitment could not ensure a random sample of those at risk for PT was selected, and it was particularly difficult to find participants in

rural areas who had not been tested. This constraint may also indicate the difficulty in accessing such individuals, not just to participate in research studies, but also in clinical care. Further studies may help elucidate the true meaning of access barriers in these harder to reach communities. In addition, recruitment was inherently biased toward those who self-selected to discuss these issues, and it is difficult to access individuals who have not had testing (as they are not always known) or those who had received results within the normal CAG range (as these individuals may have put HD behind them). However, snowball sampling methods (i.e., asking individuals to inform family members who were not tested or those who had received normal results about the study), and conducting interviews until saturation appeared to be met helped us to recruit a sample representing a diversity of views.

This research sought to understand the obstacles to testing in terms of access to genetic services, as well as exploring the mechanisms by which this issue may be addressed. Such research is essential to decrease inequalities in access to PT services, and other potentially beneficial research advancements in the field of HD such as involvement in clinical trials and knowledge of new therapies. What makes accessibility of PT services for HD important is not just that it may result in differences in quality of life and care, but because these differences may be addressed with creative and adaptable solutions in the delivery of PT services. The study findings underscore the need for us to rethink and personalize the way we deliver such services to improve access issues thereby preventing inequities in the healthcare system. This issue is pertinent for other areas of genetic medicine to ensure that genetic technologies and testing benefit all (Collins, *et al*., 2003). However, access barriers remain largely unaddressed, despite the increasing importance of personalized medicine and technological developments that

decrease the cost and improve portability of genetic tests and other healthcare technologies. Telemedicine, supporting GPs and other healthcare providers to assist in the provision of PT and tailoring testing protocols are just some of the ways by which we may ensure the benefits of PT are more widely realized.

6. DEVELOPING A COMPREHENSIVE, EFFECTIVE PATIENT FRIENDLY WEBSITE TO ENHANCE DECISION-MAKING IN PREDICTIVE TESTING FOR HUNTINGTON DISEASE

A version of this chapter will be submitted for publication as: Hawkins, AK, Creighton, SM, & Hayden, MR. Developing a comprehensive, effective patient friendly website to enhance decision-making in predictive testing for Huntington disease.

6.1 Introduction

As noted in previous chapters, PT for HD is recognised as being an irreversible decision of great consequence (Evers-Kiebooms, *et al*., 1987), with psychological and emotional implications such as guilt (including 'survivor guilt'), shame, fear and other effects such as unintentional risk alteration for biological relatives (Benjamin, *et al*., 1994; Bloch, *et al*., 1992; Wiggins, *et al*., 1992). However despite considerable concern about possible adverse events following predictive test results, these fears do not appear to have been realized. Instead the majority of studies suggest that people are satisfied with their decision to be tested and cope well when adequately prepared to receive an HD test result (Codori & Brandt, 1994; Decruyenaere, *et al*., 1996; Decruyenaere, *et al*., 2003; Dufrasne, *et al*., 2010; Hayden, 2000; Lawson, *et al*., 1996; Meiser & Dunn, 2000).

The decision to proceed with PT may be dynamic and unfold over time, or it may be more automatic or triggered by a particular event (Etchegary, 2006; Smith, *et al*., 2002). It is shaped by clinical or socio-political contexts (e.g., test accessibility and availability, cost, psychiatric diagnoses), as well as other personal and familial factors (i.e., relational autonomy) (Taylor, 2004). Providing adequate support during this process entails non-directive counseling to allow individuals to consider the pros and cons of such testing, and make up their own minds as to whether testing is right for them (Elwyn, *et al*., 2000; Harper, 1988). To aid in the provision of such non-directed, supported decision-making, at risk individuals should be provided with factual information on the testing process and its ramifications. One-on-one genetic counseling and education with an appropriately trained professional is a fundamental part of the informed consent process in PT (Green, *et al*., 2001). However it may be difficult to provide such one-on-

one counseling in all situations as it is time-consuming, expensive, and there are a limited number of genetics professionals able to provide such services (Edwards, *et al*., 2008; Washington State Department of Health, 2008; Holtzman & Watson, 1998). Such counseling may also not be easily available due to a number of access barriers including distance, travel time and availability of support (Hawkins, Creighton, & Hayden, 2012; Hawkins & Hayden, 2011; Hawkins, Ho & Hayden, 2011). Moreover, recognition of increased prevalence of HD (Fisher et. al, unpublished data) as well as the possibility of clinical trials for those at risk may mean that more people will consider PT for the disorder in the future, especially if a treatment becomes available.

As such, there is an imperative to develop effective and accessible tools to aid in the informed decision-making process for those considering predictive genetic testing (Touchette, *et al*., 1997). Previous research has demonstrated the importance and effectiveness of written and web-based tools in supporting complex healthcare decisions (Caulfield, *et al*., 2008; Kenny, *et al*., 1998; O'Connor, *et al*., 1999). These materials may be web or CD based and include diagrams, interactive flow-charts, decision-making trees, vignettes or personal stories, short videos/cartoons of healthcare providers and/or patients discussing their experience (Cook & Dupras, 2004). However the use of such decision-aids and websites in genetic testing is mainly confined to the hereditary cancer field (Caulfield, *et al*., 2008; Green, *et al*., 2005; Mancini, *et al*., 2006; Wang, *et al*., 2005). While some educational, written and web-based resources for HD PT do exist, these resources are often within websites providing information on HD in general and are not immediately accessible. While many of these sites are excellent sources of accurate, well-written information on HD, these more general sites may be intimidating, overwhelming

and difficult to navigate for those wishing to find information on HD PT. From a review of existing resources, as well as consultation with HCP and experts from the Huntington Society of Canada it seems that where resources do exist, they are often one dimensional (e.g., text only, or text with simple diagrams), and do not include multidimensional approaches such as interactive diagrams and visuals, short documentaries or videos, factual information, and personal perspectives. Multiple presentation methods, including narratives are thought to be the most effective in communicating information and promoting informed decision-making to a wide audience as they account for differences in learning and information processing styles (Green & Levi, 2009; Green, *et al*., 2004; Trees, *et al*., 2010; Wang, *et al*., 2005; Ziebland & Herxheimer, 2008).

The main objective of this project was to develop a patient-friendly, comprehensive, accessible web-based tool to provide accurate information about PT for HD. The aim of this website was to provide individuals at risk for the disorder a 'one stop shop' for information, support and resources that may be useful when considering PT for HD, as well as important information for family members, friends and HCPs. To ensure this resource would meet future users' needs, this project involved a mixed methods approach including a literature and existing resource review, and an interview study of those at risk for developing HD, followed by the development, pilot test and modification of a the website.

6.2 Materials and methods

6.2.1 Interview study

As noted in the previous chapter (chapter five), the first part of the interview project was designed to gain an understanding of access to PT, while the second part of the interview project sought to understand the appropriate content and format of a website geared toward predictive test decision-making. These interview questions were conducted during the same interview study described in the previous chapter and recruitment was based on a non probability sample (Babbie, 1990) which aimed to obtain a diverse sample of individuals at risk for HD, including those who had been tested and those who had not been tested (and received either an increased risk or a decreased risk results). Research Ethics approval was obtained for this study and potential interview participants were identified using the Huntington Society of Canada, British Columbia (BC) Chapter database, as well as the Centre for HD (Vancouver) database, as described in chapter five.

The 33 individuals who participated in the study were asked a series of open ended questions regarding their perspective on PT, their decision-making process surrounding PT, important considerations and components in the decision-making process and education/information needs when considering PT. Recorded interviews were transcribed and subject to a content analysis (Berg, 1998; Corbin & Strauss, 2008) with the aid of qualitative analysis software (NVIVO 9, QSR INTERNATIONAL). As noted in the previous chapter, data collection and analysis occurred concurrently, using the constant comparative method (Boeije, 2002; Glaser, 1965) so that emerging concepts, themes and ideas from the analysis could be incorporated into subsequent interviews to allow for a more detailed understanding on key issues

to be developed. The interview and analysis process was conducted until theoretical saturation was achieved (Boeije, 2002; Creswell, 2009; Strauss & Corbin, 1998).

6.2.2 Website development

Based on the results of the interview study (described below), review of existing resources and published literature on decision-making and the development of successful health-related resources, we created a website dedicated to PT for HD. Website content was developed using existing materials from the Centre for HD in Vancouver, and from the Huntington Society of Canada resources on PT. These resources were reviewed by the authors and supplemented, updated and modified to improve consistency, comprehensibility and ensure they met the content needs determined from the interview study results. In order to ensure the research section of the website was current, as well as avoid duplicating other efforts, the research section contains a direct feed from 'HD Buzz', an internet resource that aims to provide rapid dissemination of the latest HD research to the lay community. Links to other HD resources and international organisations, including a sign up section that allows viewers to receive information on clinical trials, were provided (Figure 6.1).

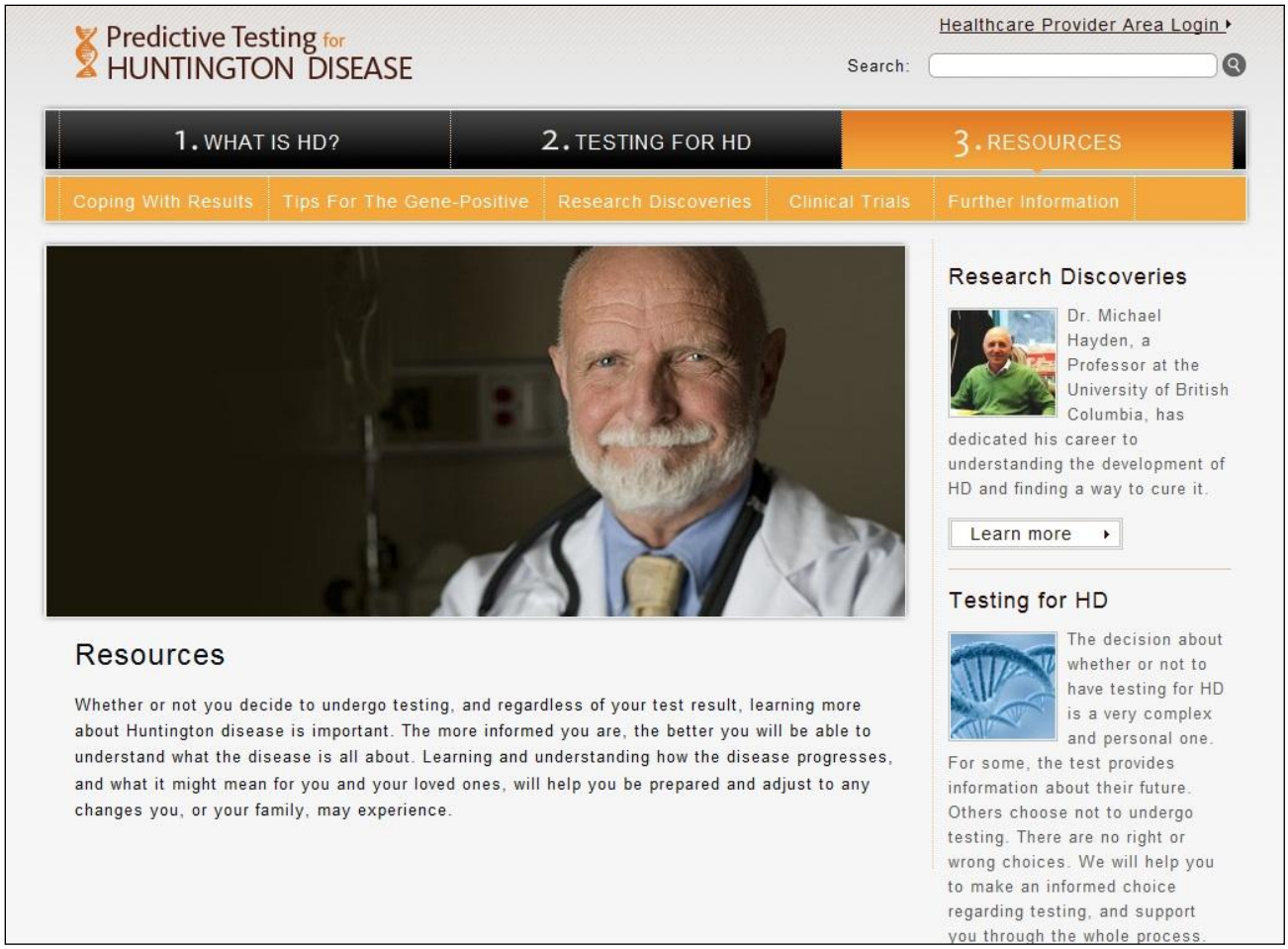


Figure 6.1: Screenshot of the resources page

In addition, interactive diagrams and images to explain complex topics, such as the CAG repeat size and results, were developed with the assistance of the website developers, (B'stro), tested and modified based on feedback from other researchers (n = 15), genetic counselors (n = 4) and lay people (n = 3) (Figures 6.2 -6.4). Four short video documentaries were developed on the following topics: 1) the decision to undergo PT; 2) interpreting and understand predictive test results; 3) coping with results; and 4) HD research. These topic areas were chosen based on the focus of the website (to help individuals understand the decision to undergo PT) and the results

of the interview study (discussed in the next section). Documentaries were uploaded on to relevant sections of the website.

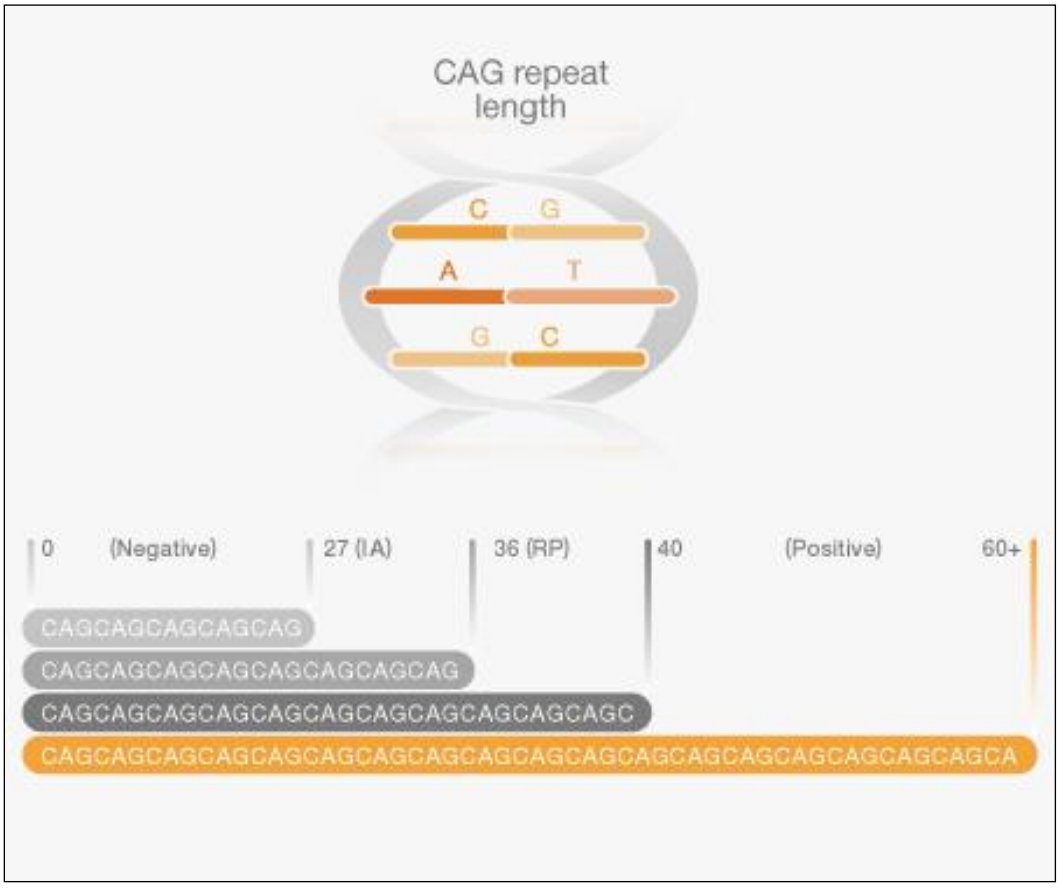


Figure 6.2: Website diagram describing Huntington disease repeat sizes

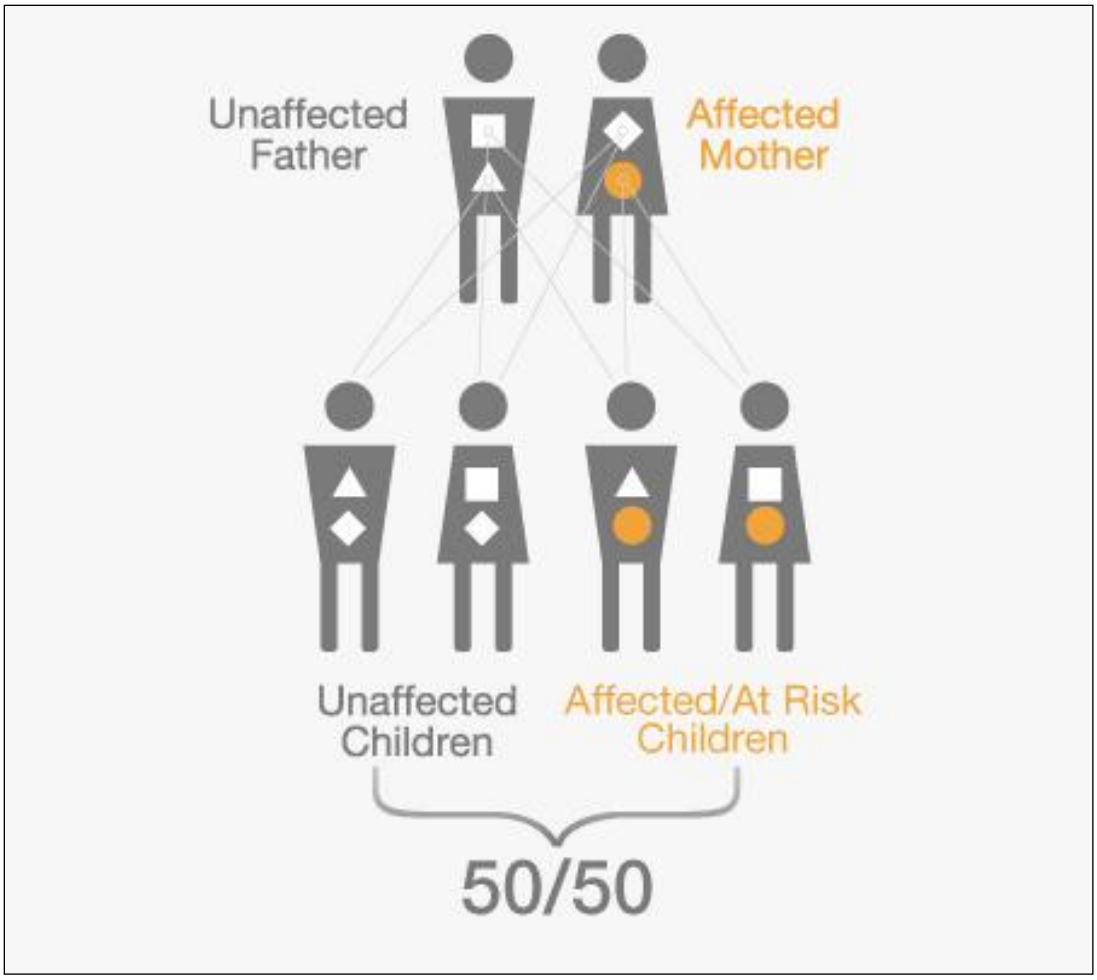


Figure 6.3: Website diagram describing the autosomal dominant inheritance pattern

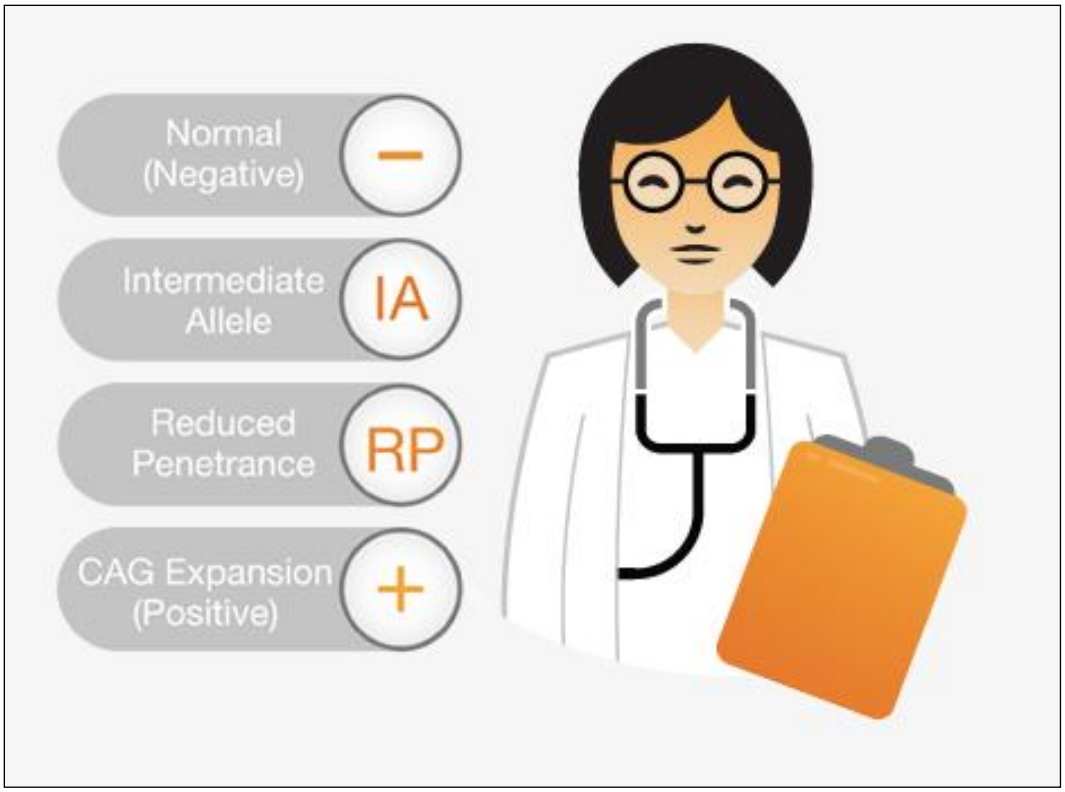


Figure 6.4: Interactive website diagram to enable interpretation of different results.
Clicking on each tab on the left hand side of the diagram brings up a full description of the meaning of the specific result.

Stories for the website were obtained, with permission, from a collection that was compiled and edited by S. Cox, in conjunction with the Huntington Society of Canada. These stories come from different sources including a series of in-depth research interviews conducted by Dr. Cox, as well as those that were submitted in response to requests for personal perspectives on genetic testing from chapters of the Huntington Society of Canada (Cox, 1999) (Figure 6.4). The personal reflections contained in the story section were divided into different sections to represent different perspectives on PT including: the decision to proceed with testing; the decision not to go through PT; the PT process; and coping with results.

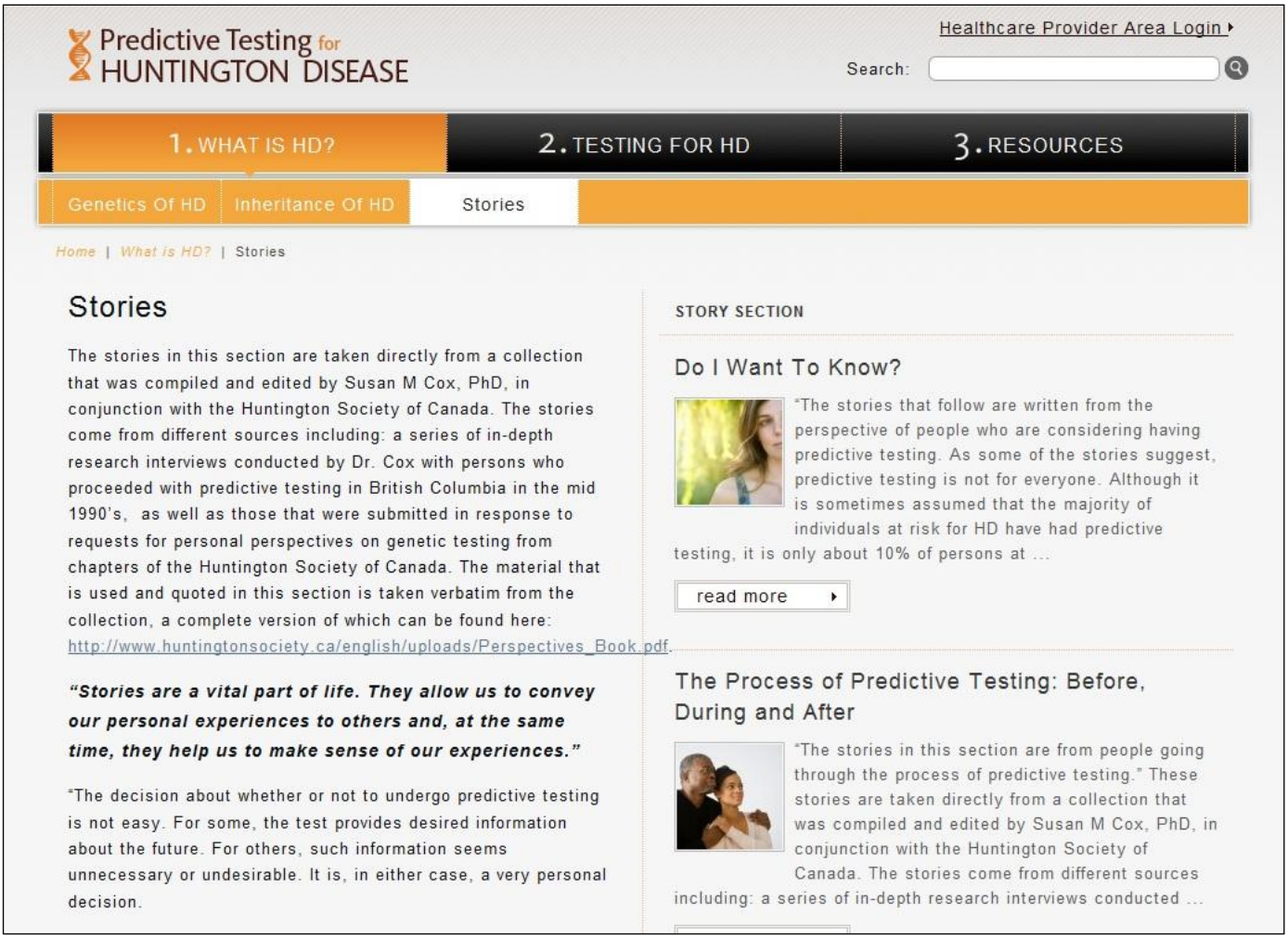


Figure 6.5: Screenshot of the stories homepage

Website structure was based on several iterations of wireframe models[19]. Based on interview feedback, overall website design and layout aimed to be welcoming, unintimidating and not institutional. Images were selected for diversity and to highlight the familial aspect of the condition. Photos on the site aimed to convey an overall tone to website users that a trusted friend or close relative was welcoming you to discuss testing (Figure 6.5). All aspects of website design, including layout, diagrams, content and documentary/diagram development were based

[19] A wireframe model is developed during the initial stage of website production and includes a visual overview of the website, including the home page, main pages, subpages and sidebars, etc.

on established principles in successful web-based education and decision facilitation tools (Cook & Dupras, 2004; Lea, *et al*., 2011; Lipkus, 2007; Lipkus & Hollands, 1999; Lobb, *et al*., 2006; Neuhauser & Kreps, 2003; Paling, 2003; Stout, Villegas, & Kim, 2001).

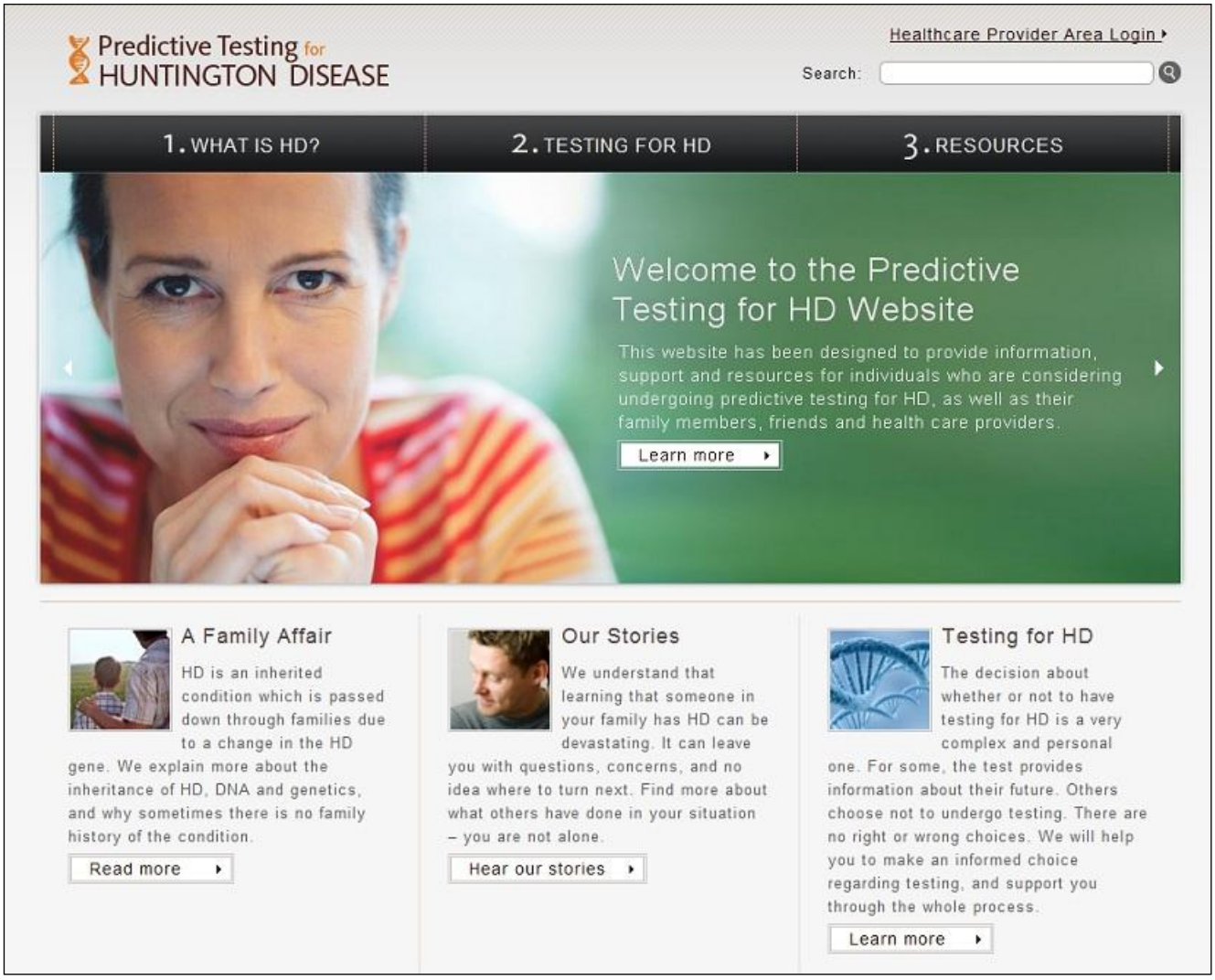


Figure 6.6: Screenshot of the website homepage

Once the website was live and fully operational, a pilot test of the site was conducted to evaluate the content and test usability. A short web-based survey was sent to 35 select individuals who had participated in the interview study (n=10), genetic counselors across North

America (n=5), HD researchers and HD experts (n=10) and lay individuals (n= 10). Pilot test respondents were recruited based on availability, convenience and snow-ball sampling as follows: 1) interview respondents from the first part of the study who had expressed willingness to evaluate the website; 2) Genetic counselors in Canada and the United States known to Alice Hawkins; 3) researchers in HD at the UBC; 4) HD experts/ HD healthcare providers; and 5) non-medical individuals with no connection to HD (i.e., lay persons, known to Alice Hawkins). These individuals received an email inviting them to view the website and participate in a short, confidential, web-based survey consisting of a series of closed-ended questions regarding the website content, usability, comprehensiveness, language level, diagrams, images and documentaries (Appendix II). Respondents were encouraged to provide comments and explain each of their responses, as well as provide suggestions for improvement. Each of these comments was reviewed several times in detail and coded for themes. Similar concepts or ideas were grouped into the same category to allow for an understanding of the diversity in comments received, as well as nuanced details and perspectives on certain issues (Creswell, 2009). Separate REB approval was also obtained for this component of the study.

6.3 Results

The following paragraphs describe the results of the interview study, followed by the website pilot-test results.

6.3.1 Interview findings

Results of the interview study revealed overwhelming support for a web-based resource to be developed which was specific to PT for HD. Interview participants noted that when people need information on a particular topic often the first place they would turn was the internet: "*a lot of people's first place of looking [sic] for answers when they have questions is the internet".* Another respondent highlights that this is particularly important for those who are in more remote or rural locations who cannot easily access resources in person: *"Having it [a predictive testing resource] online is a great idea - especially when people are remote".* Ensuring accessibility of resources also helps people who *"want to reach out, they just don't know how."* Respondents also noted that the existing websites on HD were often geared toward individuals or caregivers who had recently been informed of a HD diagnosis in their family. While these websites were helpful, they were also perceived as *"scary"* and *"intimidating"* and they were not necessarily easy to navigate.

A key finding from the interview study was the importance of personal stories and perspectives in making it easier for people to determine whether or not PT is the right choice for them and explore *"the pros and cons".* As one participant explained: "*I had a niece call me just a few months ago because of my decision not to be tested. She called me and she was calling the other one [aunt] that had tested and had been tested negative. Anyways, she's at that stage...She's a young girl deciding on a family and she's at that stage and that's what she was looking for...the stories. Why did you choose what you chose?"* Other participants emphasized the importance of hearing the story of someone who had been in a similar position, as there was a shared understanding and experience with such individuals, which could not be found in

friends or family members not at risk for HD: *"People want to know. They do want to say 'what was it like for you?' Unless you're carrying the gene or not carrying the gene, it's hard to convey the magnitude of it to someone else."*

Participants also noted that personal perspectives and stories were important in helping people feel less alone and isolated in their experience and risk of developing HD: "*Sometimes Huntington's feels like such a lone disease. Not a lot of people still know about it and, you know, you can feel like you're struggling all by yourself. [A website] just makes more people less alone, I think."* Others highlighted the importance of other peoples' stories and perspectives of going through the process and receiving results: *"If there was [somewhere] that I could read of people's experiences and kind of see how people dealt with things. Everybody has a different story and different situations you know…just kind of feeling like you are not going through some big huge hospital, wondering down a hallway where you don't even know you are going and then all of a sudden you have these amazing huge results you know…so feeling like you are kind of alone I guess."* One participant also spoke of existing HD blogs that she had read, and that these blogs had offered her a sense of comfort: *"Certainly there have been times where it's nice... to know there's a group of people [out there] and sometimes I've just gone on there to read people's stories and get a sense of not being alone in things."*

In addition to emphasizing the importance of personal stories from others at risk, interview respondents identified several other key components of a successful website on PT. These included: 1) information on the genetics and inheritance of HD, including concepts such as new mutations and reduced penetrance; 2) steps in the PT process; 3) information on where

testing could be done/ how to get tested; 4) diagrams, videos and pictures that supplemented text to aid in understanding of concepts; 5) a detailed discussion of the decision-making process involved in determining whether PT was the right choice; 6) a description of the different test results (i.e., positive, negative, reduced penetrance and intermediate alleles) and coping with results; 7) prenatal and other reproductive testing options and 8) links to other HD related resources and local support groups. As one respondent suggested: *"If there was a site that was family friendly and had lots of good information and the hard core research - a little bit for everybody - I think people wouldn't be so scared."* Many participants also noted that a section on HD related research would be helpful, especially for those who received a positive test result, even though this was not the primary goal of the site. Finally, respondents suggested that information geared toward family members/ friends of those at risk for the disease would be important as these individuals were often integral to the decision of whether or not to undergo PT.

The findings from the interview study detailed above were used to inform all stages of the website development from the overall look and feel of the site, to the content and flow of the website.

6.3.2 Pilot test results

A total of 23 of 35 potential participants completed the website pilot test survey for a response rate of 65.7%. The characteristics of these respondents are outlined in Figure 6.7. Collation of survey responses received for question one revealed that all of the respondents agreed that the website was clearly laid out and easy to navigate. Comments received included:

"One of the best-organized and clear websites I have visited" and *"it was really well laid out & beautifully designed".* Suggestions for improvement included: modifying the hover menus so they would work on touch screen devices (such as iPads); creating a separate section on prenatal testing/reproductive testing options; and creating a 'user-guide' to the website for those who were not familiar with drop down menus.

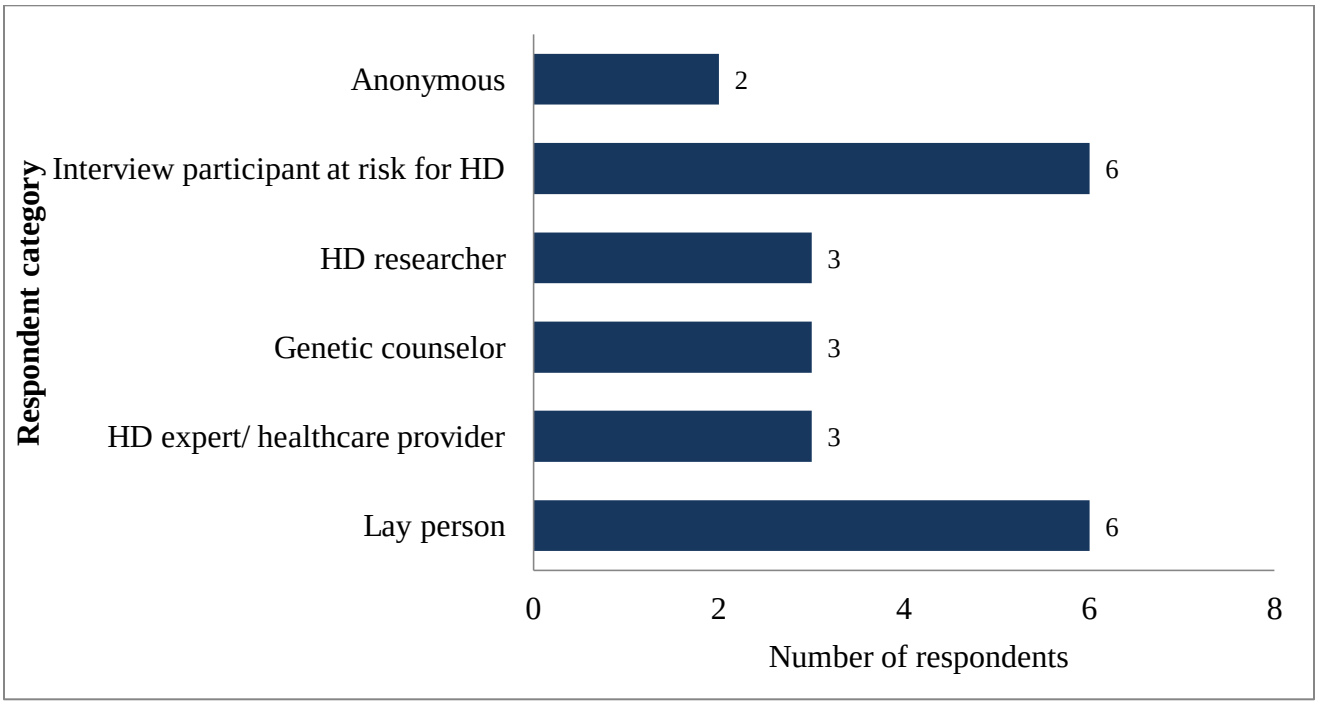


Figure 6.7: Characteristics of those who participated in the website pilot test

Participants were also asked whether the content of the website was clear and easy to understand (question 2). Again, the majority (n = 22/23, 96%) of respondents answered 'yes' to this question, and stated *"I was very impressed with how the sites information is organized and laid out...the language is at a good level for many to understand"* and "*I thought it really covered everything very well. I honestly could not think of anything that would be lacking at this point".* Suggestions for improvement in the content included: 1) some of the sections were too

text heavy; 2) subheadings needed to be reordered to make them more intuitive; 3) making the 'Research Updates' section more succinct; and 4) slowing down the speed at which the images and text on the home page moved.

The third question focused on the illustrations and diagrams developed specifically for the website which, for example, depicted the HD inheritance process; genes, chromosomes and DNA; and the different categories of CAG repeat sizes. All but one (n = 22/23, 96%) of the respondents thought the diagrams and illustrations on the site were helpful. Respondents also thought that the *"diagrams on genetics [were] useful and very clear and easy to understand without reading [the] explanation".* One respondent, a HD researcher, indicated the diagrams would be useful for lay people and that it would be a useful resource for other family members to understand the disease: "*I feel like I should give my parents this link so they can better understand what I'm working on".* Suggestions for improvement included clarifying that HD can be passed down from either a mother or father (the diagram depicted the mutation being inherited from the father) and having more ethnic diversity in the photos on the site.

The fourth and fifth questions were linked and asked respondents to consider whether the website provided adequate information on 'what is HD?' and 'testing for HD'. All respondents thought there was adequate information on the first of these, and 91% (n = 21/23) of respondents felt there was adequate information on the PT process. In particular, respondents *"loved"* the use of stories which were felt to be *"very powerful"* and *"unbiased"* as they provided a wide variety of perspectives on PT. Respondents also particularly liked the 'Important Considerations' section for undergoing HD PT. Suggestions for improvement included wording changes to improve

clarity and accuracy of the descriptions, providing further links for resources and adding more detail to the section on how testing is done.

Similarly, respondents were also asked to consider whether the website provided adequate information on the 'pros and cons' of PT. All respondents agreed that the website did provide adequate information on this topic as illustrated by the following comments: *"It does a good job of explaining what the test process is and questions to ask oneself"; "The testing is presented in a very unbiased way. It should give people some excellent issues to consider";* and *"As a website you could not do much more".* Again, people highlighted the importance of the stories in this regard: *"I got most pros/cons for testing from stories. Stories [were] long but very powerful".* Suggestions to improve this section were limited and included a proposal to include video clips of individuals discussing their reasons for and against pursuing PT.

The final section of the survey encouraged respondents to provide other comments and suggestions regarding the website. Suggestions for improvement ranged from changing the font and text spacing on the site to improve readability; addressing 'technical glitches' with the site, including difficulty viewing the site on different operating systems; and minor editing suggestions. Other comments received indicated general overall support for the site: *"The site is very important tool for people who need it"; "I can't think why we've not done this before now"; "I am impressed with this work, and would definitely refer patients to this website for accurate information and good support examples"* and *"[The] website is clear, concise, beautiful, and you have done an impressive job. With a few little 'tweaks' you will be able to confidently launch this wonderful and very informative website"* A summary of the results are provided in table 6.1.

Table 6.1: Website pilot test survey results

Question	Percentage of responses that were positive in nature (as indicated by a 'yes' response)
1. Is the website clearly laid out and easy to navigate (find your way around)?	100
2. Is the content of the website clear and easy to understand?	96
3. Are the illustrations and diagrams helpful?	96
4. Does the website provide adequate information on 'what is HD'?	100
5. Does the website provide adequate information on 'testing for HD'?	91
6. Does the website provide enough information on the pros and cons of testing and making the decision to be tested?	100

6.4 Discussion

This project illustrates the importance of a multi-method approach to developing educational resources. This methodology included 1) an extensive review of existing resources; 2) an exploration of the literature on patient education and website development; 3) interviews with people at risk for HD and experts in PT; and 4) the development and pilot testing of a website. This process drew on established methods to develop successful educational/ informational tools to develop a site which would promote informed, supported decision-making. For example, the preference for narratives is supported by empirical studies on patients needs and desires whereby people seek others' stories to help them deal with difficulty and feel comforted that they are not alone in their situation (Rozmovits & Ziebland, 2004; Trees, *et al*., 2010). Such narratives are key in helping people cope with difficult information, particularly among those with low literacy skills (Butow, Fowler, & Ziebland, 2005). Stories can add

salience to information, and help make facts and key considerations more understandable (Ziebland & Herxheimer, 2008). Moreover, these narratives can assist in the decision-making process by prompting people to reflect and consider their options more carefully, thereby helping them to make a truly informed decision (Entwistle,*et al*., 2010; Entwistle, *et al*., 2011). When constructing the website, particular attention was paid to presenting a balanced and diverse set of stories and perspectives about why people chose to have PT, or chose not to, and what the experience of the decision-making and testing process was like for them. Narratives therefore represented a broad range of perspectives to avoid the potential criticism of bias in these stories which may in turn bias decision making (Butow, *et al*., 2005; Winterbottom, *et al*., 2008). In addition, the stories were assessed in terms of their portrayal of the PT process and concepts regarding HD. This was done to avoid any potential inaccuracy in representation of the illness and disease process in the narratives which may in turn perpetuate misunderstanding and public ignorance (Segal, 2008). Pilot study responses commended the balanced nature of these stories suggesting that the selection of stories and diverse viewpoints provided a considered and unbiased set of perspectives for the reader to consider.

In addition, where possible and appropriate, suggestions for improvement received from the pilot study were incorporated into the website. For example, all grammatical and technical issues were resolved, headings and fonts were amended to improve comprehensibility, certain subsections were moved or reordered to improve intuitive flow of the website, and content was amended in places to enhance understanding. In addition, several features were integrated into the website to account for different audience needs, such as enabling text size to be changed to a larger font to assist those with visual impairments. It is possible that pilot test respondents may

have been somewhat biased in that they may have known the researchers, and by responding to the survey may have self-selected to be inherently supportive of such a website. However, honest review and feedback and candid critique was encouraged by making the survey anonymous, and by distributing the pilot study widely. The primarily positive responses received may also reflect the thorough and considered initial website development process which integrated interview findings, use of existing successful resources, and the significant evaluation and number of iterations of the website prior to pilot release.

By taking such a comprehensive, needs-assessment approach to website development, we developed a site that is specifically tailored to the needs of those considering PT. The site acts as a guide and reliable information source throughout the testing process, including the post-result period. The site has the potential to truly provide support and assistance for those making the decision of whether or not to undergo PT. The design and content of the site, as well as portability of web-based resources, allows individuals to obtain and explore information in an accessible, non-threatening manner. Such a site also improves access to reliable information, local resources and support. As illustrated by the interview study, and prior research (Hawkins, Creighton & Hayden, 2012), this resource is particularly valuable for individuals who live in remote or rural locations. To further improve access to reliable information in other areas, the site is currently being translated into 12 other languages. The site can also be used in conjunction with telehealth methods to improve access to PT for HD (Hawkins, Creighton & Hayden, 2012; Hawkins, Ho & Hayden, 2011). In conclusion, further work and evaluation is needed to determine the utility of this site for those undergoing the PT process and for their families. Such

evaluation could include pre and post measures to assess uptake of accurate information and satisfaction with the site on a variety of levels including usability, utility and comprehensibility.

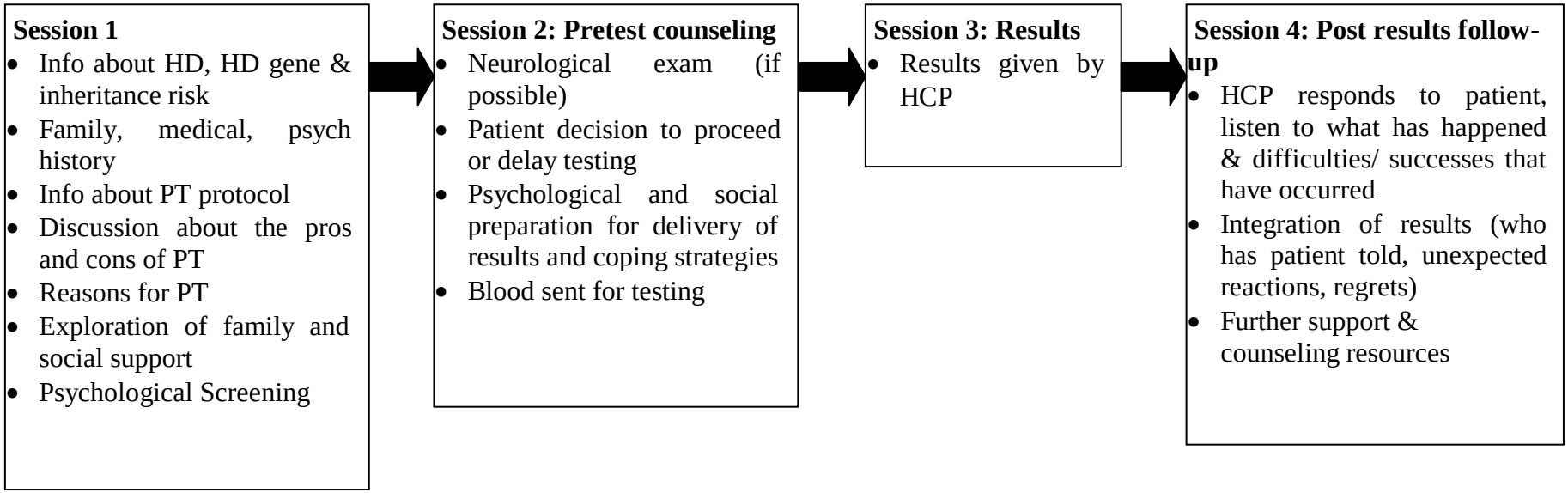
7. PROVIDING PREDICTIVE TESTING FOR HUNTINGTON DISEASE VIA TELEHEALTH: RESULTS OF A PILOT STUDY IN BRITISH COLUMBIA, CANADA

A version of the chapter will be submitted for publication as: Hawkins AK, Creighton S, Ho A, McManus B & Hayden MR. Providing predictive testing for Huntington disease via telehealth: results of a pilot study in British Columbia, Canada.

7.1 Introduction

As discussed in chapters two and three, the availability of PT for HD allows people to exercise their autonomy in terms of knowing whether or not they will develop the disorder at some point in their lives (Bortolotti & Widdows, 2011). Such information may relieve uncertainty, reduce anxiety, allow for better informed reproductive decision-making, enable access to support and allow individuals to make plans in other aspects of their lives (such as career choices, life insurance etc.) (Codori & Brandt, 1994; Decruyenaere, *et al*., 1996; Meissen & Berchek, 1987; Van der Steenstraten, *et al*., 1994; Wiggins, *et al*., 1992). The standard approach to providing predictive genetic testing for HD is outlined in Figure 7.1 and was designed to ensure individuals who decided to pursue testing made a considered, informed choice regarding the test, and were fully aware of the risks and benefits of receiving such results, including the impact on relationships and family members and the potential for genetic discrimination.

Figure 7.1: Summary of the Huntington disease predictive testing protocol



7.1.1 Predictive testing in British Columbia

The Centre for HD in Vancouver usually conducts each of the first three PT sessions in person with the patient undergoing testing. The final post results session is conducted via phone, or in person, depending on the patient's preference. In recognition of the difficulties of travelling to the Vancouver-based HD testing centre, the protocol has also been somewhat modified in recent years for those patients from geographically remote areas. After attending their first visit in person at the Centre for HD in Vancouver these individuals are given the option of completing the PT process in their local community with the support of their local HCP. In these instances their local HCP is provided with phone-based training and support, as well as written materials from the Vancouver-based HD PT team. The local HCP then conducts the pre-test counseling and results session in person with the patient, and acts as local support for any further questions or follow-up needs.

While PT must always be a carefully considered and personal choice, results of the studies reported in chapter five and six have suggested that access to PT may be a barrier to testing whereby individuals at risk for the disorder are deterred from seeking PT due to the difficulties in accessing PT services. For example, calculating PT uptake figures in rural versus non-rural areas of BC reveals that PT rates are higher in the area surrounding the Centre for HD PT service in Vancouver as compared to areas which are more remote from it (30% versus 16% uptake rates, respectively). Furthermore, the interview study discussed in chapter five, which involved in-depth, semi-structured discussions with individuals at risk for HD from all over the province who had and had not pursued PT revealed that access is a barrier to testing for two distinct reasons. First, access was viewed as a barrier in terms of the costs associated with

travelling to the PT centre including time off work, time away from family, cost of travel and ability to travel. Interestingly, the interview study also revealed a second, less obvious access barrier, namely the inflexibility of the PT protocol itself. The PT protocol, or at least the perception of the protocol, may be viewed as rigid, paternalistic, and overly burdensome in terms of counseling and spousal requirements, time and number of visits required to obtain a test result. Such a process does not take into account individual needs and does not distinguish between varying social, psychological, personal and family history or level of knowledge (Hawkins, Creighton & Hayden, 2012). Ultimately this may lead to alienation of those who wish to access information and support regarding the decision of whether to pursue genetic testing for HD.

7.1.2 Survey regarding a predictive testing telehealth service

Based on the results of the interview and mapping studies described in chapter five, a survey was conducted to better understand perceptions of and need for the PT telehealth service in BC The one page survey (see Appendix IV) was sent to 690 individuals on the Huntington Society of Canada, BC Chapter mailing list. Of the 102 responses received (response rate 15%[20]), the majority (78%) of respondents stated that they thought that providing PT in the local community via telehealth was a good idea (Figure 7.2). In addition, when asked what they believed was the ideal method of undergoing PT, 68% of respondents indicated that they felt the

[20] The relatively low response rate is likely due to a number of factors: 1) the Huntington Society of Canada BC chapter mailing list is somewhat out of date and contains contact information which in some cases was collected 10-15 years ago; 2) the list also contains the names of individuals who are not from HD families but have supported the Huntington Society of Canada in some way in the past – such individuals would be very unlikely to respond to the survey. Unfortunately, due to Research Ethics Board and Huntington Society of Canada restrictions regarding privacy of the mailing list, names and contact details of those sent the survey were not accessible and therefore it was not possible to send follow up or reminder emails to urge people to complete the survey.

ideal method was in their local community via telehealth with the support of a local healthcare provider (Figure 7.3).

Figure 7.2: Bar chart illustrating responses to the question 'do you think providing predictive testing for Huntington disease in your local community via telehealth is a good idea?'

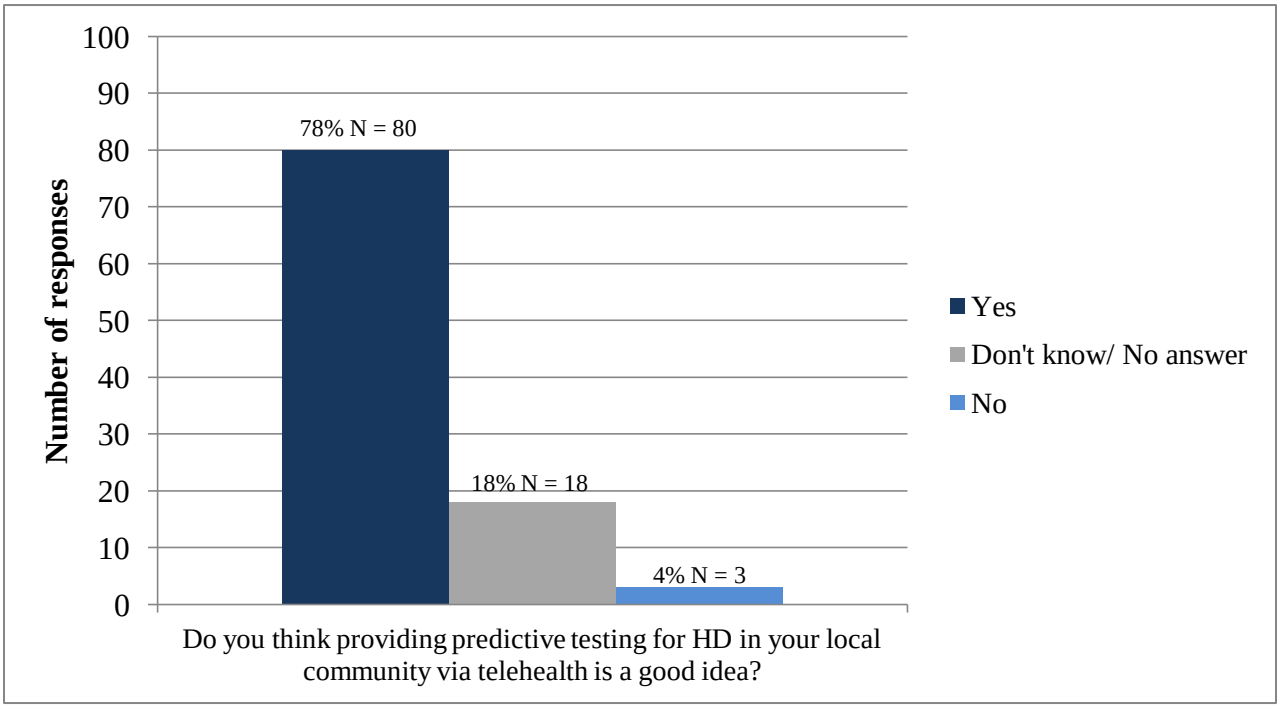
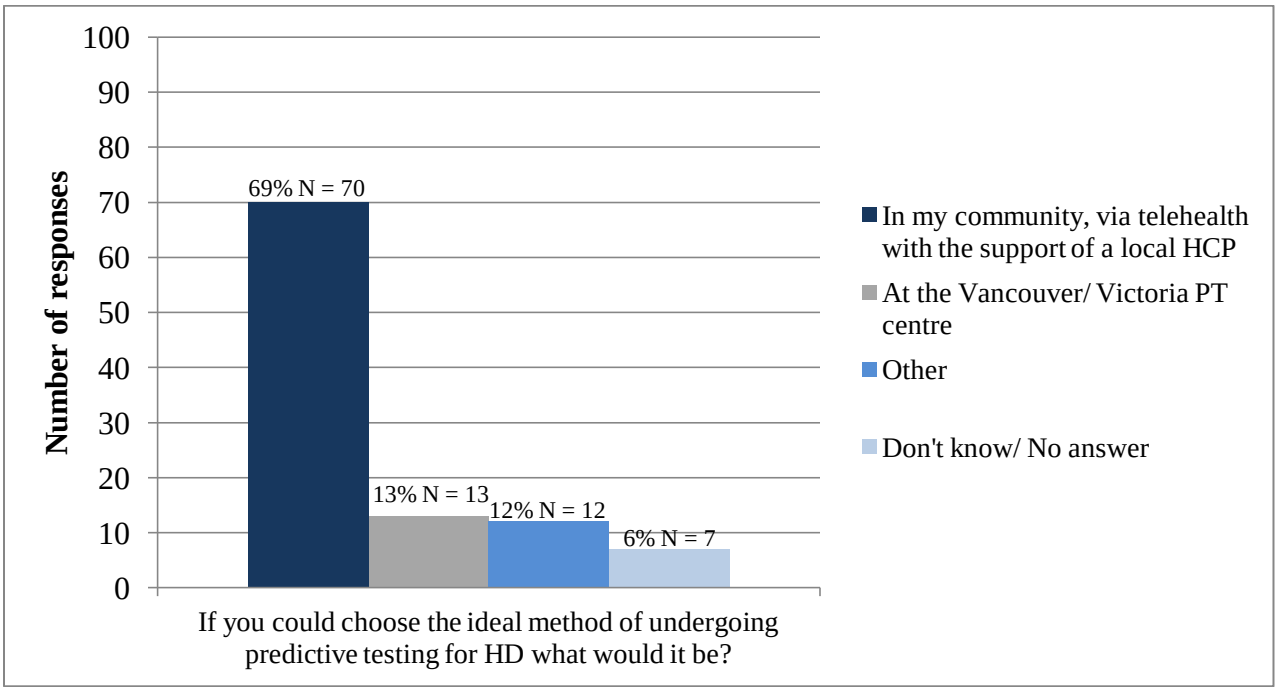


Figure 7.3: Bar chart illustrating responses to the question 'if you could choose the ideal method of undergoing predictive testing for Huntington disease what would it be?'



7.1.3 Expert workshop on predictive testing

Based on the results of the survey and interview studies, the development of a mechanism to provide PT remotely, via telehealth, seemed warranted. In order to ensure any such telehealth PT protocol maintained quality of care and support throughout the testing process, an international expert workshop, entitled 'Improving Access to HD Predictive Testing' was convened in Vancouver, on November 12th 2010. The eleven workshop participants were invited based on their expertise and knowledge of providing either PT for HD and/or novel telemedicine and telegenetics techniques as shown in Table 7.1.

Table 7.1: Area of expertise of workshop participants

Expert type	Expert knowledge area
Provides genetic services at a distance	- Telemedicine and other novel communication strategies - Non-traditional genetic counseling - Training and education needs and mechanisms
Provides a high volume of HD testing and care	- Ethical issues - Psychosocial considerations - Practical limitations

The broad aim of the day long workshop was to foster discussion and to elicit expert advice regarding the provision of PT for HD offsite, including remote and rural area service delivery. Specifically, the workshop goals were as follows:

1) To assess experts' perspectives on the key components of the PT process
2) To gather suggestions and recommendations on how to improve access to PT for HD
3) To propose and get feedback on a provisional education and testing protocol for providing HD PT remotely

4) To understand and consider mechanisms to address potential concerns related to a modification of the standard PT protocol for HD

The one-day workshop was formatted to allow short 20 minute presentations on different subtopics including: a preliminary overview of interview findings; an overview of the PT website; necessary components of the PT process; access barriers to PT; telehealth and digital technologies; evaluation and safeguards during the PT process; delivery of results and developing an implementation and roll-out strategy. After each presentation a 20-30 minute facilitated discussion between all workshop participants took place to address specific questions within each subtopic (e.g., is it acceptable to provide PT via telehealth?; is it acceptable to provide results via telehealth?, is the neurological exam a necessary component of the PT process). Key findings, decisions reached and recommendations were recorded.

Ultimately the workshop sought to provide a forum for experts' concerns, insights and knowledge to be gathered and integrated into ongoing research aimed at improving the lives of those affected with HD. This enabled a more thorough examination of the access issues to PT in other jurisdictions (including other areas of Canada, the United States and the United Kingdom). In addition, the concept of providing PT via telehealth was thoroughly critiqued, with expert attendees providing useful feedback on the preliminary protocol, provision of results and follow-up. At the conclusion of the expert workshop each of the attendees agreed that an offsite PT protocol for providing HD PT did seem a reasonable mechanism to address access issues to testing. The workshop deliberation process both enabled further development of the protocol to ensure potential criticisms and

concerns with the telehealth protocol were considered upfront and ensured that the development of the protocol was in keeping with acceptable standards of care for the provision of PT. A pilot of the offsite testing tool was recommended, and a variety of assessment mechanisms to measure success, satisfaction and safety of the novel testing tool were suggested. In addition, the participants did not feel it was necessary to conduct a neurological exam prior to testing[21].

In order to be successful, and maintain appropriate quality of care and support for individuals undergoing testing, a variety of safeguards, training and educational tools were discussed and recommended. For example, the expert workshop attendees recommended that, in most circumstances, it would be preferable for the results to be delivered in person, by a trained HCP. This HCP did not necessarily have to be a physician, but should be someone with healthcare training such as a social worker or nurse and who was capable of learning about the genetics of HD and the PT process, providing ongoing support and referral for further psychological assessment or support, as necessary. Having flexibility in choosing the appropriate HCP/ allied HCP was thought to be important as it allowed the patient to select an individual most appropriate to their circumstances, and someone with whom they had (or could have) a trusted and ongoing relationship. This flexibility also addresses rural healthcare provision limitations in which there are a high number of locum physicians, or lack of physicians (Rourke, 2008).

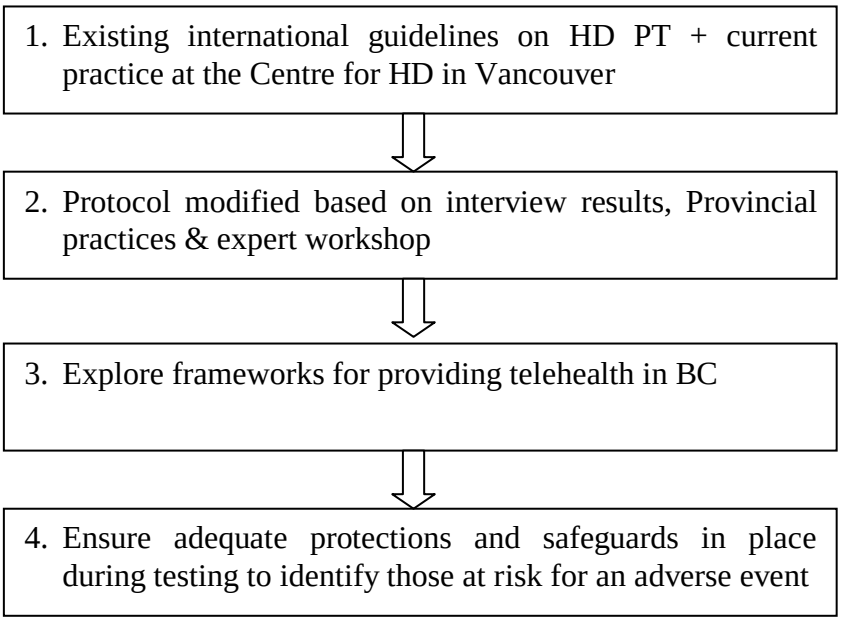
[21] If PT results were positive the expert participants agreed that the patient should receive a baseline neurological assessment at that point.

The expert workshop also allowed for further consideration of the needs of a website dedicated to PT, including evaluation of the provisional PT website model and content. The workshop also thus contributed to the development of website with the knowledge users in mind, while ensuring accurate portrayal of necessary information. Further details on the Expert Workshop are provided in the Expert Workshop Report in Appendix III.

7.1.4 Telehealth protocol development

Based on the above studies and results of the Expert Workshop, a PT protocol for HD was developed that provided testing remotely, while still maintaining quality of care and support for those undergoing the PT process. The development of the portable telehealth PT protocol was informed by a number of key factors: 1) the existing international guidelines on HD PT (MacLeod, *et al*., 2012); 2) current practice at the Centre for HD in Vancouver; 3) results of the interview study; and 4) the expert workshop. This development process is illustrated in Figure 7.4.

Figure 7.4: Flowchart illustrating the telehealth predictive test development process



Due to concerns regarding the psychological impact and potential consequences of undergoing PT for HD as previously described (Almqvist, *et al*., 1999; Benjamin, *et al*., 1994; Bloch, *et al*., 1992; Wiggins, *et al*., 1992) the fourth step of the protocol was developed to ensure that adequate protections and safeguards were in place to assure the well-being and support of those undergoing PT. Standard psychological evaluation measures were integrated into the PT protocol (BDI and SCL90) and which required timely follow-up with the Vancouver-based HD PT testing centre. In addition, to ensure that individuals undergoing PT received adequate local support, the protocol requires the assistance of a local HCP (such as a family physician). Furthermore, the protocol requires that this local HCP be identified early in the PT process. This individual agrees to participate in the process, and would be provided with written resources on HD and the PT process, including the pros and cons of testing and the psychological consequences of testing, as well as phone training and support throughout the process. The HCP would also be the one to provide testing results, and therefore would be best able to identify and

address any immediate adverse reactions, needs for support and other concerns. In addition, because the local HCP would be identified by the individual undergoing testing it was more likely that this was an individual that the patient knew, trusted and with whom they had an established relationship. This telehealth testing protocol was therefore designed to provide PT remotely while still maintaining quality of care and support for individuals undergoing testing. This process is described in Table 7.2.

Table 7.2: Stages of the telehealth predictive testing protocol

PT stage	Content of appointment
1. Referral/ request for PT	• At-risk individual (patient) goes to PT website • Reads information, requests an appointment • Appointment request (via website or phone) goes to HD clinic Genetic Counsellor (GC)/ coordinator

2. General counseling with UBC team via telehealth	• Initial telehealth consult with client re: testing, process etc
	  • Information about HD, HD gene and inheritance risk
	  • Information about PT and test protocol
	  • Reasons for PT
	  • Exploration of family and social support
	  • Psychological screening[a] (BDI, SCL 90)
	  • Family, medical, psychological history[a]
	• Local HCP identified for support during testing process

3. Pre-results session with local HC provider	• Local HCP undergoes web-based training on HD, genetics, testing process, psychosocial considerations • In person appointment between local HCP and patient and spouse/support person • Neurological exam (if possible) • Participant's decision to proceed or delay testing • Blood sent to lab for testing • Psychological and social preparation for the delivery of results and coping strategies • Schedule results session and 2 week post-results follow-up.
4. Results session	• Results given by the local HCP (in person) • Spouse/ support person present • HD clinic available via telehealth as needed
5. Post-results session	• HD clinic (via telehealth) responding to the patient; listening to what has been happening and difficulties or successes that have occurred • Integration of results (who has patient told, how have they coped, unexpected reactions, regrets) • Further support (local HCP, HD clinic, HD Resource Centre and further counselling in community)
6. Six month follow-up session	• Follow-up via teleconference (HD clinic GC)

[a] Referral for further evaluations provided as indicated (e.g., psychiatry)

[b] Participants are given the opportunity to receive results in person from the Centre for HD PT clinic, or via telehealth in exceptional circumstances (e.g., no local HCP can be identified)

7.2 Piloting the predictive testing telehealth protocol

The next stage of the project involved a 1 year pilot of the telehealth PT protocol to ensure patient satisfaction and well-being with the telehealth PT protocol using a 57 question mail-out survey. In order to examine the efficacy of the telehealth protocol as compared to the standard Vancouver-based PT protocol, the survey was also sent to patients who underwent PT in Vancouver between 2010 and 2012. The study was approved by the UBC and Children's and Women's Hospital Research Ethics Boards and written informed consent was obtained from all participants.

7.3 Methods

All Vancouver-based PT patients were identified from the Centre for HD PT clinic record based on whether they had undergone PT between July 2010 and July 2011. These individuals were mailed a letter describing the study and asking them to complete and return the questionnaire via prepaid envelope. Patients who underwent the telehealth protocol were recruited as follows: 1) an information email regarding the pilot test project was sent out via the Huntington Society of Canada BC Chapter email distribution list; 2) an information poster regarding the pilot test project was sent out via the Huntington Society of Canada BC Chapter newsletter[22]; and 3) all individuals referred to the Centre for HD for PT were informed about the availability of the pilot test project. Individuals who received the Vancouver-based and telehealth-based PT protocols underwent testing as outlined in Figure 7.5 and 7.1. All PT test

[22] This newsletter is received by approximately 700 people on the Huntington Society of Canada (HSC) B.C. chapter mailing list. Individuals on this mailing list are either families affected by HD, or individuals who have supported the HSC BC chapter in the past.

counseling was conducted by Susan Creighton +/- Dr. Michael Hayden between 2010 and 2012. For rural and telehealth participants, sessions 2 (pre-test counseling) and 3 (results) were conducted by the patient's local HCP, where possible. The questionnaire was mailed to patients who underwent PT 2-6 weeks after receiving their PT results. For those patients who attended the first PT session, but elected not to pursue testing, or postpone testing at this time, the relevant components of the survey were mailed to them within 2-6 weeks of the initial visit. Patients received a follow-up reminder phone call if the survey had not been returned within 3 weeks. At 6 weeks any remaining non-respondents were mailed a second copy of the survey, and received a second follow-up phone call.

7.3.1 Development and testing of the survey instruments

The survey instruments were developed based on the interview results (as detailed in chapter four) and in consultation with two experts in healthcare evaluation and satisfaction measurement from the BC Provincial Health Service Authority: Dr. Ruth Milner and Dr. Sema Aydede. The survey included validated questions from the BC telehealth survey and a previous HD based survey (Bombard, *et al*., 2009), as well as questions adapted from other validated and frequently-used patient satisfaction and well-being surveys (Client Satisfaction Questionnaire – CSQ8 (Larsen, Attkisson, Hargreaves, & Nguyen, 1979), Beck Depression Inventory (Beck *et al*., 1961). The survey was also reviewed for content, flow and comprehensibility by an expert reference group. Subsequent to finalization of the survey, the instrument was pilot tested by sending it to five participants from the earlier interview study (see chapter four) who indicated they would be willing to review the survey tool. These individuals were asked to evaluate the

survey instrument on the following measures: time to complete the survey, comprehension of the questions and instructions as well as survey layout.

The final questionnaire contained 57 questions for those who underwent testing via telehealth and 37 questions for those who underwent Vancouver-based testing[23]. The questionnaire sections focused on: 1) socio-demographic information; 2) information and counseling received prior to testing (including explanation of HD, discussion of the advantages and disadvantages of PT and the implication of HD results); 4) delivery of results; 5) overall support during the predictive test process; and 6) experience of the telehealth service[24]. The majority of non-demographic questions were based on a likert scale and asked participants to state whether they 'strongly agreed' = 5, 'agreed' = 4, 'neutral' = 3, 'disagreed' = 2, or 'strongly disagreed' = 1, with a statement regarding the PT process. To correspond with the previously validated BC telehealth survey, questions posed regarding the telehealth service allowed for 'yes', 'no' and 'not applicable' responses.

7.3.2 Statistical analyses

Descriptive statistics are reported for demographics, experience and satisfaction with the PT process. Experience and satisfaction with the PT process for both the telehealth-tested (TT) and Vancouver- tested (VT) participants is presented according to PT stage: 1) pretest counseling; 2) delivery of results; and 3) follow-up period and overall satisfaction with the PT

[23] The telehealth tested questionnaire contained additional questions regarding the telehealth experience.
[24] Those who underwent PT counseling via telehealth only

test process. In addition, for those who underwent testing via telehealth, experience and satisfaction with the telehealth service is reported.

The Fisher's exact test was used to investigate the majority of baseline demographic differences in the TT and VT groups. Comparisons of means to investigate differences between the two groups on satisfaction and experience scores were conducted primarily using either the Fisher's exact test (nominal data) or Mann-Whitney U tests (ordinal data). Non-parametric tests were used in these cases due to relatively small sample size and because the variable parameters were unknown. The students t-test to examine differences between the VT and TT groups was used when data was continuous and followed a normal distribution. Two-tailed significance tests were used and alpha was set at 0.05. All statistical analyses were conducted using SPSS version 19.

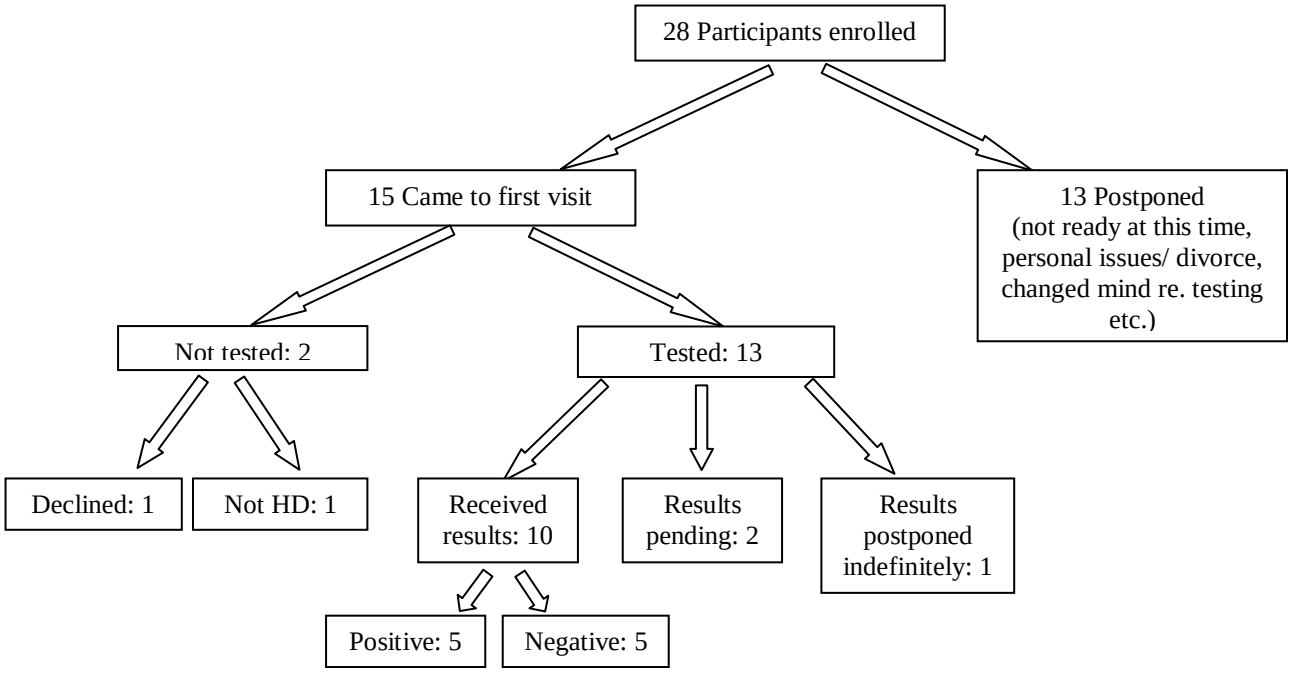
7.4 Results

7.4.1 Characteristics of the respondents

Of the 30 individuals invited to participate in the survey from the VT group, 13 people completed and returned the entire survey representing a response rate of 43.3%. All but one of these individuals completed the PT process and received results. Of the 28 individuals who initially expressed interest and consented to the telehealth PT protocol (Figure 7.10), 15 attended at least one telehealth PT session. Of these 15 participants, 14 completed at least one section of the survey, representing a response rate of 93% for the telehealth experience evaluation, pretest counseling and socio-demographic components of the survey. Of these 15 participants, 13

elected to undergo PT and are currently at various stages of receiving PT results as illustrated in Figure 7.5. Of the 10 participants who have received results, 8 completed the results and follow-up session surveys, representing a response rate to the post results survey of 80%.

Figure 7.5: Status of participants in the pilot study



Socio-demographic characteristics of those who completed the surveys are presented in Table 7.3. There were no significant differences in the baseline socio-demographic factors with respect to age (P = 0.223), education level (P = 0.670), gender (P = 0.700), marital status (P = 0.670), children (P = 1.000), HD test result status (P = 1.000) or location (P = 0.255) between the TT and VT groups. Beck Depression Inventory (BDI) scores prior to testing were similar between groups (mean BDI TT: 4.87; mean BDI VT: 3.09; Mann-Whitney U P = 0.382). The socio-demographic characteristics of the sample (age, gender, education level, children and

marital status) are similar to other HD populations in North America (Almqvist, *et al*., 2003; Bombard, *et al*., 2009; Codori & Brandt, 1994; Wiggins, *et al*., 1992).

Table 7.3: Demographic characteristics of the respondents

	Total n=28		Telehealth n=15		Vancouver tested n=13		p-value[a]
	n	%	N	%	n	%	
Gender							
Male	11	39.3	5	33.3	6	46.2	0.700[b]
Female	17	60.7	10	66.7	7	53.8	
Average Age	46.1[d]		47[d]		41[d]		0.233[c]
Marital status							
Married/ common law	21	0.75	12	0.8	9	69.2	0.670[b]
Single/ separated/ widowed	7	0.25	3	0.2	4	30.8	
Education							
Some college & above	21	0.75	12	0.8	9	69.2	0.670[b]
High school & below	7	0.25	3	0.2	4	30.8	
Children							
Have children	16	57.1	9	0.6	7	53.8	1.000[b]
No children	12	42.9	6	0.4	6	46.2	
Community/ setting							
Rural	15	53.6	10	66.7	5	38.5	0.255[b]
Non-rural	13	46.4	5	33.3	8	61.5	
Test result							
HD+	10	50	4	50	6	50	1.000[b]
HD-	10	50	4	50	6	50	

[a] Missing values are excluded, values are 2-sided
[b] Fisher exact test
[c] Students t-test (2-tailed), equal variances assumed
[d] Values are mean

7.4.2 Pretest counseling

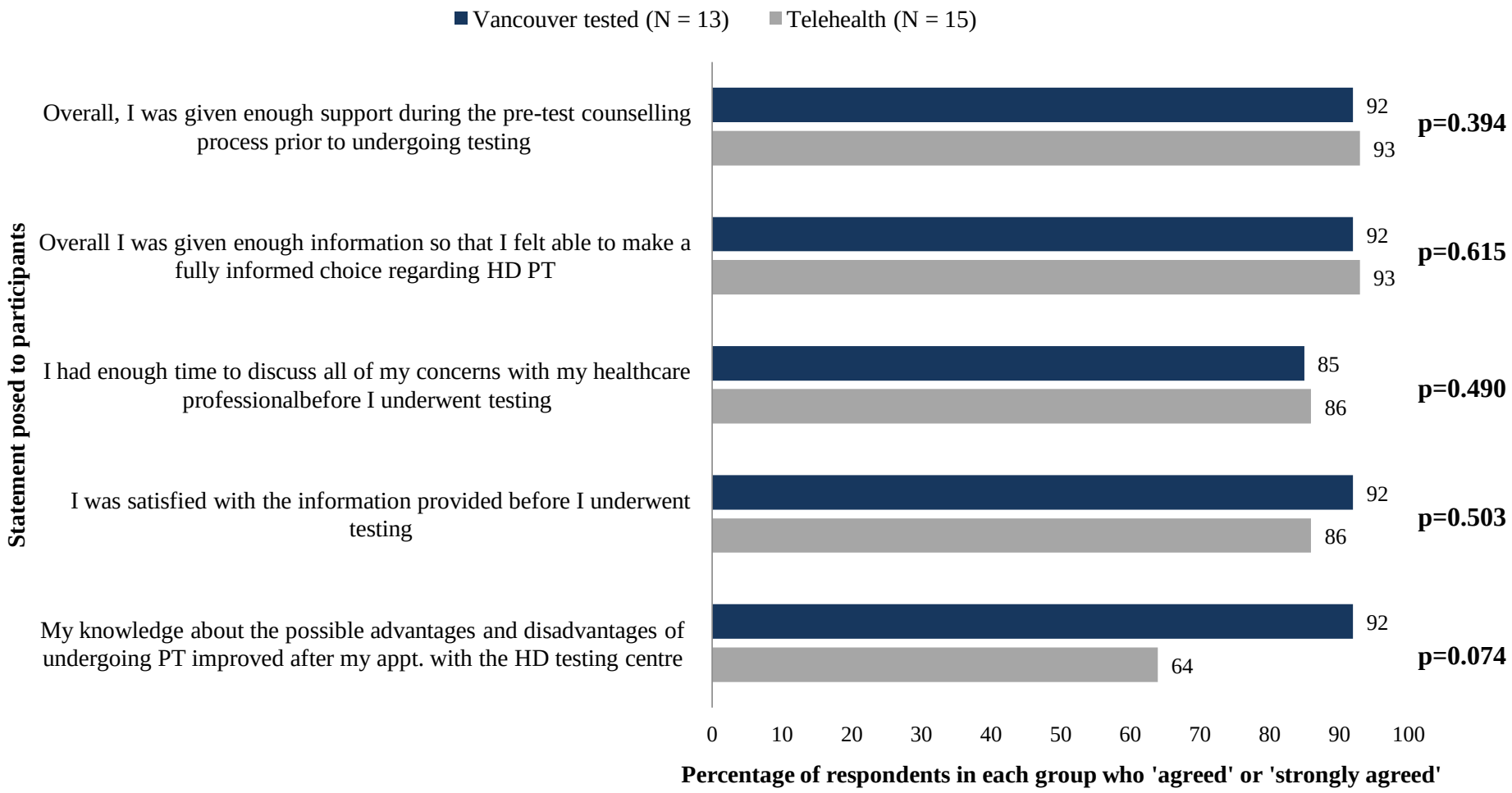
Those who underwent PT via telehealth reported a significantly lower number of hours (including travel) to attend the first PT appointment (TT mean: 2.73 hrs; VT mean: 6.69 hrs; $P = 0.002$)[25]. There were no other significant differences between the groups in terms of their satisfaction and perception of information given during the pretest counseling sessions. Both

[25] Calculated using a 2 tailed t test (equal variances not assumed).

groups reported that they either agreed or strongly agreed with statements regarding whether their knowledge of the advantages and disadvantages of PT improved during the pretest counseling process (TT median: 4; VT median: 5; Mann Whitney U *P* value = 0.074)[26] and that they were given enough information to make a fully informed choice regarding HD PT (TT median: 5; VT median: 5; Mann Whitney U *P* value = 0.615). The VT and TT groups also reported similar scores regarding whether they were able to discuss all their concerns and questions prior to undergoing PT (TT median: 4; VT median: 4; Mann Whitney U *P* value = 0.490). In terms of information provided during the pretest counseling sessions, the majority of participants in both groups either strongly agreed or agreed that they were satisfied with the information provided prior to undergoing testing (TT median: 4; VT median: 5; Mann Whitney U *P* value = 0.503) and reported similar scores for whether they were given enough support during the pretest counseling sessions (TT median: 5; VT median: 4; Mann Whitney U *P* value = 0.394). These results, reported as a percentage of 'agree' or 'strongly agree' responses on each measure for each group, are provided in Figure 7.6.

[26] The number given in brackets for each group is the median score which was reported in the survey by that group. Median figures, rather than a mean, better reflect the likert scale nature of the responses received in the survey.

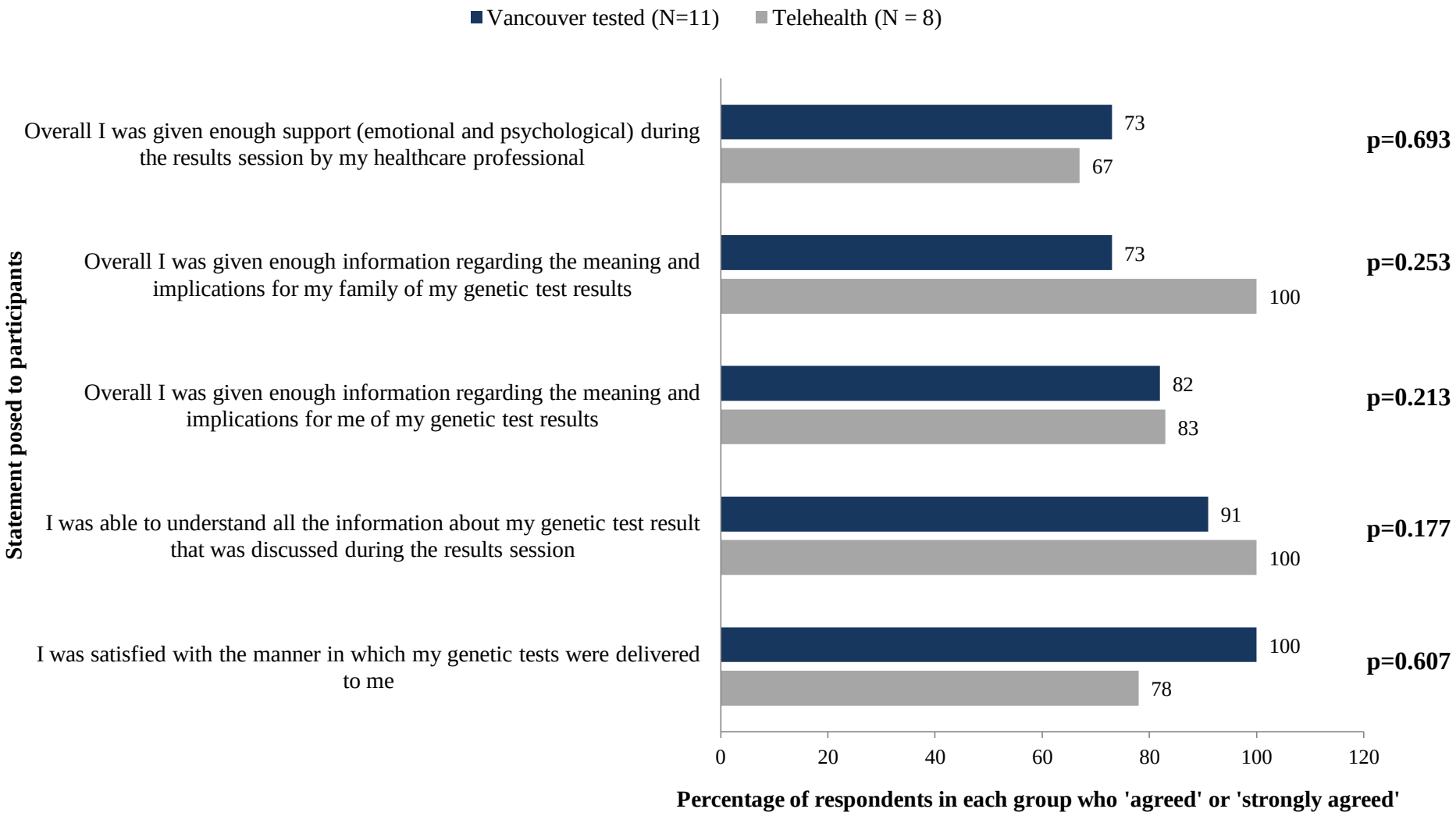
Figure 7.6: Bar chart illustrating the percentage of participants from the Vancouver Tested and Telehealth Tested groups who either strongly agreed or agreed with the statements regarding the pre-test counseling sessions.



7.4.3 Delivery of results

As Figure 7.7 illustrates, the majority of participants both groups either agreed or strongly agreed with the statement 'I was satisfied with the manner in which my test results were delivered to me', with no significant difference between the groups (TT median: 4; VT median: 5; Mann Whitney U *P* value = 0.607). There was no significant difference between the groups in terms of their scores regarding their comprehension of the information presented during the results session (TT median: 5; VT median: 4; Mann Whitney U *P* value = 0.177) and the majority of participants in both groups on average either agreed or strongly agreed that they were given information regarding the meaning of the results for themselves (TT median: 5; VT median: 4; Mann Whitney U *P* value = 0.213) and for their family (TT median: 5; VT median: 5; Mann Whitney U *P* value = 0.253). Finally, there was no significant difference found between the groups in the scores reported for the statement 'overall I was given enough support (emotional and psychological) during the results session by my HCP' (TT median: 5; VT median: 5; Mann Whitney U *P* value = 0.693). In the comments section, some participants expressed that they would have liked to have received more information in the results session regarding local support groups and ongoing research trials. One of the participants from the TT group noted that if they had lived in Vancouver they would have preferred to have received results from the Centre for HD PT team, but that receiving results locally had been easier for them.

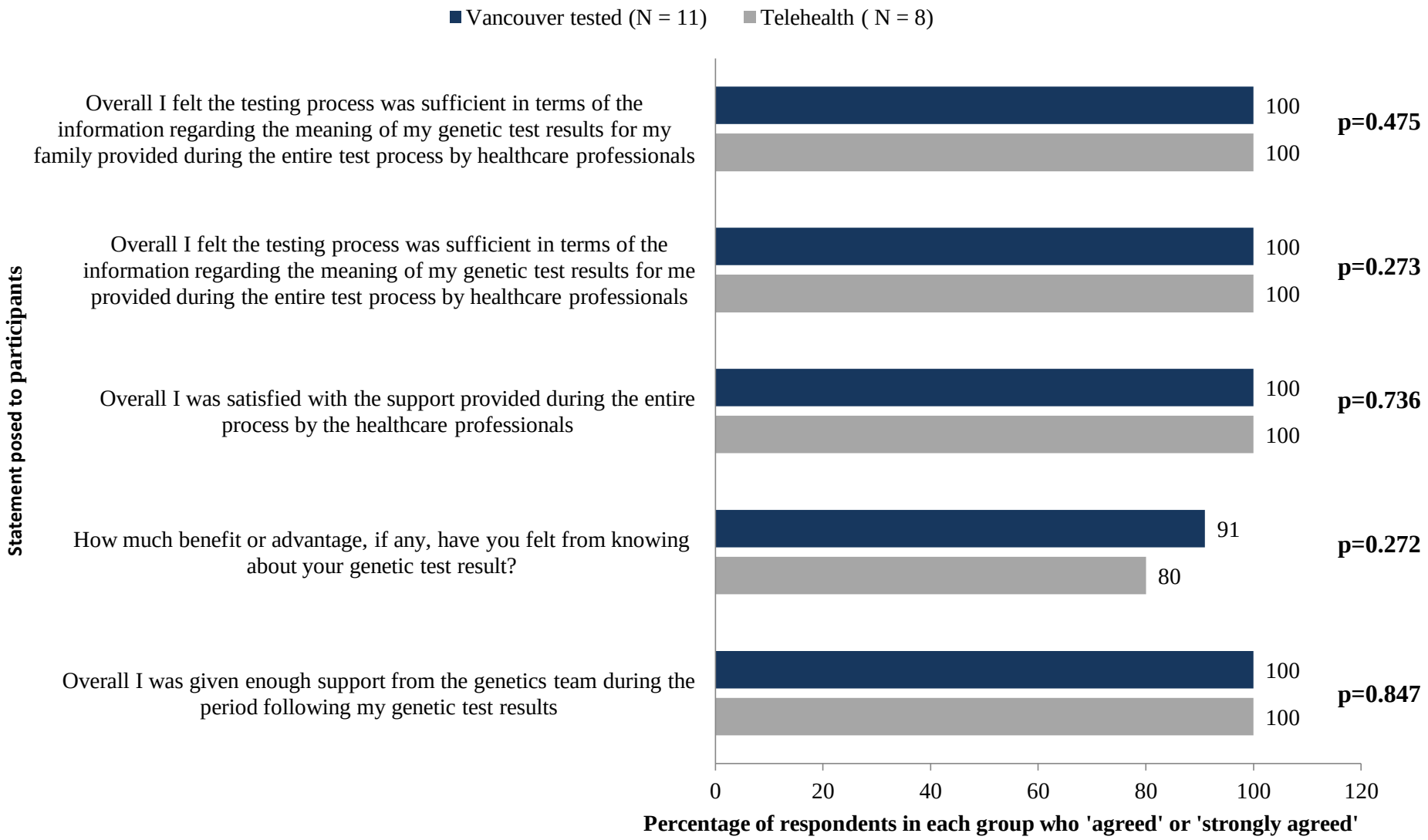
Figure 7.7: Bar chart illustrating the percentage of participants from the Vancouver Tested and Telehealth Tested groups who either strongly agreed or agreed with the statements regarding the result session.



7.4.4 Follow-up period and overall satisfaction with the predictive testing process

After completing the PT process and receiving results, participants were asked to complete a final survey regarding their overall satisfaction with the PT process (Figure 7.8). Participants in both groups were asked how much benefit or advantage they had experienced from receiving their PT result. Both the VT and TT groups reported that they believed they had received either great benefit or some benefit from receiving their HD PT result with no significant difference between either group (TT median: 5; VT median: 5; Mann Whitney U *P* value = 0.272). When asked whether they were satisfied with the support following PT results and during the entire process the majority of respondents in both the TT and VT either agreed or strongly agreed that they had received enough support during the period following delivery of results (TT median: 5; VT median: 5; Mann Whitney U *P* value = 0.847). Similarly, there was no significant difference in response score from either group to statements in relation to whether the testing process as a whole was sufficient in terms of the information regarding the meaning of the genetic test results for the patient (TT median: 5; VT median: 4; Mann Whitney U *P* value = 0.475) and their family (TT median: 5; VT median: 4; Mann Whitney U *P* value = 0.273). On the subject of whether participants would have preferred more telehealth or in-person appointments during the PT process, the majority of patients reported that they disagreed with statements regarding whether they would have preferred more telehealth sessions (TT median: 2; VT median: 2; Mann Whitney U *P* value = 0.186) or in-person sessions (TT median: 2; VT median: 2; Mann Whitney U *P* value = 0.548), suggesting that both groups felt like they had received enough support and information in the appointments they had undergone. In the final comments section two participants noted that the process had felt "*a bit touchy feely at times"* and that for some it had felt "*a little drawn out".*

Figure 7.8: Bar chart illustrating the percentage of participants from the Vancouver Tested and Telehealth Tested groups who either strongly agreed or agreed with the statements regarding predictive testing process overall.

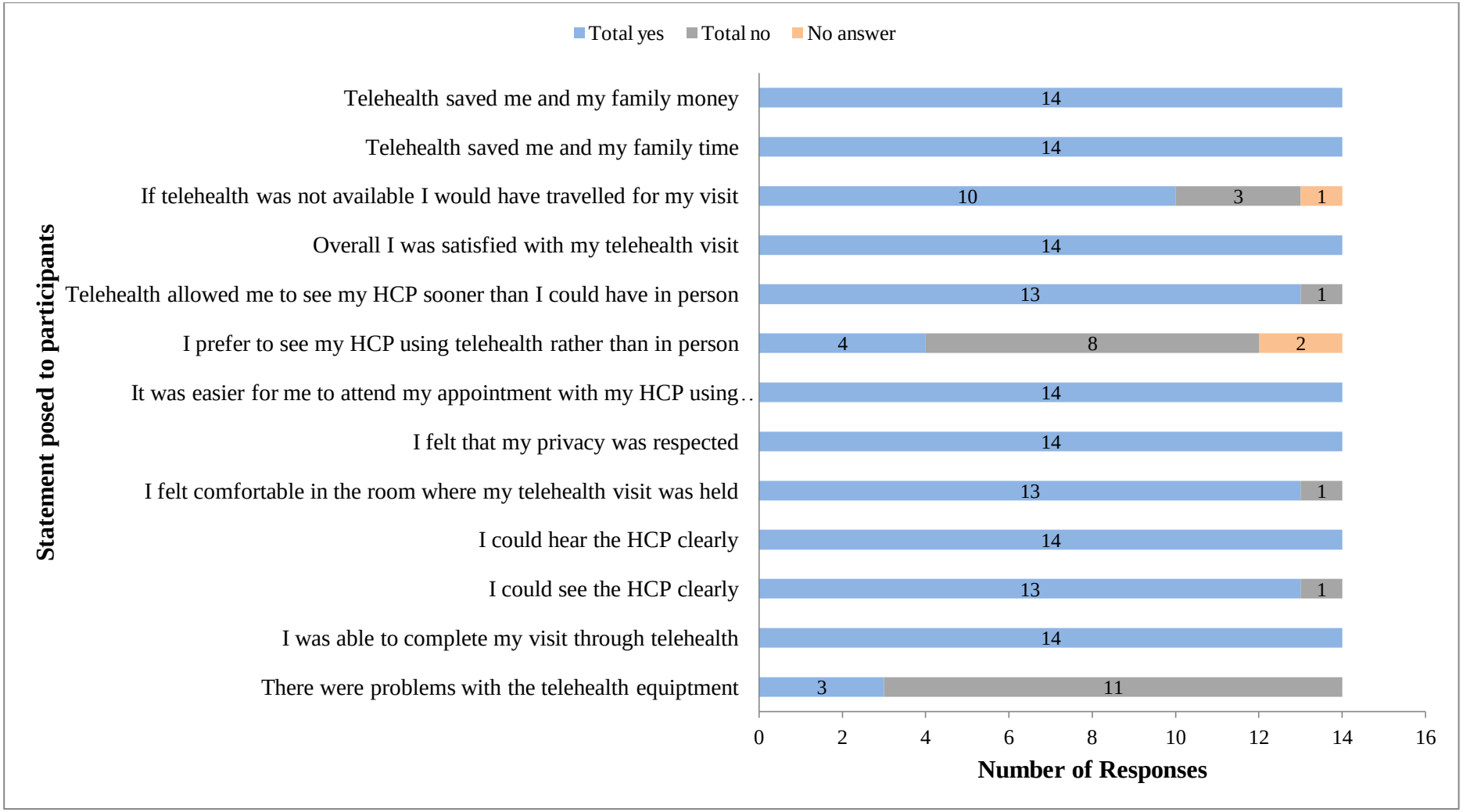


7.4.5 Telehealth service

As depicted in Figure 7.9 the majority of participants reported a positive experience with the telehealth service on a number of levels. First, from a technical standpoint, most or all respondents reported that they had been able to both see (yes = 92.9%; no = 7.1%) and hear (yes = 100%)[27] the HCP clearly during the appointment, there were no problems with the telehealth equipment (yes = 78.6%; no = 22.4%), and they were able to complete their visit via telehealth (yes = 100%). All participants reported that they believed their privacy was respected (yes = 100%) and that they were comfortable in the room where the telehealth appointment was held (yes = 92.9%; no = 7.1%). In addition participants agreed that telehealth had made it easier for them to attend their appointment (yes = 100%) and allowed them to see their HCP sooner than if they had travelled to Vancouver (yes = 92.9%; no = 7.1%). Interestingly, while the majority preferred to see their HCP in person (57.1%) some respondents (28.6%) stated they preferred to see their HCP by telehealth (na = 14.3%). Further, while the majority of participants would have travelled to Vancouver if telehealth had not been available (71.4%), some participants stated that they would not have travelled if telehealth had not been available (21.4%; na = 7.2%). Finally, all participants stated that the telehealth service had saved them both time (yes = 100%) and money (yes = 100%). To illustrate the extent of this impact, in the comments section, one participant noted that having the ability to undergo PT via telehealth had saved her over $1,000 in travel expenses and 3 days of travel to and from Vancouver.

[27] na = no answer to the question. Also note that one participant chose not to answer any of the questions on telehealth.

Figure 7.9: Bar chart illustrating responses to survey questions regarding the telehealth service experience (Telehealth Tested group)[28]



[28] One respondent chose not to answer any of the questions regarding the telehealth experience. This individual was therefore excluded from the analysis.

7.5 Discussion

7.5.1 Summary of findings from the pilot study

The results of the pilot study reveal that providing PT via telehealth, using a carefully constructed protocol is not only possible, but also warranted in terms of patient satisfaction. The telehealth PT service improves access to PT, with 17.4% (N = 3) of participants indicating that they would have not attended the visit in person if telehealth had not been available and 100% of the participants stating that telehealth made it easier to attend the PT sessions (N = 14). In addition, review of PT requests in the two year period from 2008-2009 show that there were 22 requests for PT from those in rural communities, versus 43 requests for telehealth in the two year period from 2010-2011 (when telehealth became available). This may suggest that a considerable number of individuals had not undergone testing previously due to the difficulties in travelling to the Vancouver based PT centre. In addition, nearly all TT participants stated that the telehealth service had saved them time and money, both of which were identified as barriers to PT in the foundational interview study (Hawkins, Creighton, & Hayden, 2012). The importance of this finding for some individuals is significant, and is exemplified by the participant who stated that the telehealth service had saved her approximately $1,000 and 3 days off work to travel to the appointment. While reimbursement for travel may be available, this does not solve the issue of time away from work and family. The impact of telehealth is even greater when considering that a person's spouse or support person may now be able to attend the appointment when previously they may have been prevented due to time, finances or job commitments. In fact, during the pilot project several TT participants attended the first visit

with not just one, but sometimes several family members. These family members thus had the opportunity to learn more about HD and testing for the disorder, and the broad implications of PT testing for them and their family members. Finally, the limited number of technical issues reported with the telehealth service suggests that in general, this type of service provision is both practical and feasible.

The survey results revealed that there were no significant differences between the VT and TT groups with respect to quality of care, information, counseling and support during the PT process. In both groups, the majority of participants believed that pre-test counseling had provided them with sufficient knowledge about the advantages and disadvantages of undergoing testing, the opportunity to ask questions, and the ability to make an informed, supported decision around PT. Comments received from participants on the telehealth service in this pilot study were generally positive and included: liking the flexibility and ease of set-up of the telehealth service; being able to avoid the expense and time related to traveling to Vancouver; preventing the stress of driving to and staying in the city (particularly in the winter). In addition, the telehealth service may spare unnecessary visits. For example, it was apparent during a telehealth consult for one patient referred by her local HCP for HD testing that it was very unlikely there was HD in the family. The patient was thus spared a long and unnecessary journey which would have been required had the telehealth service not been available.

In terms of the results delivery session, the majority of participants in both groups were satisfied by the manner in which results were delivered and believed they had

received sufficient information regarding the implications of these results for themselves and their families. These findings suggest that when given proper training and support, local HCPs are capable – from a patient perspective - of providing HD PT results in a supportive, knowledgeable manner and can contextualise the meaning of the results for their specific patient. It is important to note that flexibility in the delivery of results seems to be fundamental to patient satisfaction levels. For example, in their comments regarding the results session, several participants noted that they preferred receiving the results from their family physician as they had a good, long-standing and trusting relationship with their local HCP. However, one TT participant opted to have the results in person, at the Centre for HD in Vancouver, as she stated that she wanted to receive the results in person from the HD PT team, and did not have a strong relationship with her local HCP. In addition, after careful consideration, we provided one individual with PT results via telehealth at her request. This individual also reported having a poor relationship with her local HCP, had considerable travel and health restrictions, and was thought to be emotionally and psychologically stable to receive results via telehealth.

There were no significant differences among participants in either the VT or TT groups in terms of their overall experience of going through HD testing. Participants in both groups reported good overall satisfaction levels in terms of support received during the entire PT process, as well as emotional and psychological support prior to testing and during the post-results period. In addition, neither the VT nor the TT group believed that they would have benefitted or preferred more or less visits, either in person or via telehealth, suggesting that the current test protocol is appropriate in terms of numbers of

sessions. In addition, of those who completed testing and received results, there was no difference in reported level of benefit or advantage from receiving test results, with the majority of participants in both groups stating that they had either received great benefit, or some benefit from their test results. This suggests that individuals who underwent testing were adequately prepared to receive results, and had truly made an informed decision regarding PT.

In summary, the pilot study shows that providing PT via telehealth, through the protocol described above, not only equals traditional in-person testing in terms of patient satisfaction, support and education, the telehealth protocol improves upon the traditional in-person model in terms of accessibility to PT services. Providing PT via telehealth is therefore warranted both in terms of improving access to services and support, but also in maintaining quality of care and support throughout the testing process.

7.5.2 Unanticipated findings

Interestingly, since the pilot project began we have also seen a number of requests for the telehealth service from those living within the lower mainland (i.e., within two hours drive of the Vancouver HD PT clinic). Of the 28 individuals who participated in the telehealth pilot study TT group, eight were from the lower mainland. This suggests that these individuals may also prefer the option of undergoing PT via telehealth as it enables them to avoid the stress and time involved with travelling to Vancouver (which may be over three hours in rush-hour traffic), allows their spouse or other support person (s) to

accompany them to appointments, and does not require they take time off work to attend an appointment.

Another unexpected finding from the pilot project is that more people in the TT group appeared to drop-out or choose not to complete PT, even after initial discussions about the testing had been complete and consent was obtained to participate in the project. In the TT group 13/28 participants (46.4%) chose to indefinitely postpone PT for a variety of personal reasons including: they were going through a divorce; they did not feel ready; or that they had changed their mind about undergoing PT. While direct comparison with the VT group in this regard is not possible due to research ethics board restrictions and differences in timing of consent between the groups, this postponement or drop-out rate is more than double that of the 20% which is reported in the literature (Dufrasne, *et al*., 2010). While the exact reason for this seemingly higher drop-out or postponement rate among those in the telehealth group is unclear, it is possible that the availability of telehealth PT prompted individuals from more remote areas, who previously had not considered PT due to access issues, to take the first step in finding out more about PT, the PT process and the implications of receiving test results. With this in mind, the increased drop-out or postponement levels can be viewed as positive in that more people were able to access PT counseling and services. It is this improved access to support, counseling, accurate information and specialized services, not the actual uptake of PT and receiving results that determines the ultimate success of this project.

7.5.3 Limitations of the research

Due to its design as a pilot study, this study has certain limitations including the number of participants who were enrolled in both the TT and VT groups. As such, further research, involving a larger cohort, followed over a longer time frame, will be required to substantiate these findings and to assess the long term consequences of receiving PT via telehealth health or via the traditional in-person manner. Such long-term follow-up studies would also allow for comparison with published reports on the three and five year impacts of receiving test results (Almqvist, *et al*., 2003; Tibben, *et al*., 1997) as well as provide insight into whether 'drop-outs' merely postponed testing, or truly decided not to undergo PT. This study also has a number of other methodological limitations. By choosing to undergo testing via telehealth, participants in this group may be inherently more comfortable with remote communication methods or technology, and as such more likely to report a positive experience with such a service. In addition, individuals who participate in such research may be inherently more resourceful, have better health literacy to understand information and be more emotionally, psychologically and socially equipped to cope with results. As such, these self-reported surveys are subjective measures of specific groups of people (those who responded to the survey in the VT and TT groups) and may not be reflective of the PT population as a whole. Furthermore, although local HCP were all provided with the same training and educational materials about the PT counseling process and delivery of results, it was not possible in this project to ensure that standardised care was delivered by these individuals. Moreover, involving local HCPs in the PT process also means that patients are subject to different scheduling, wait times and time allotment for appointments, which may in turn lead to differing levels of satisfaction or frustration

with the PT process. Finally, this research was conducted in one province of Canada, and as such may not be generalizable to other jurisdictions.

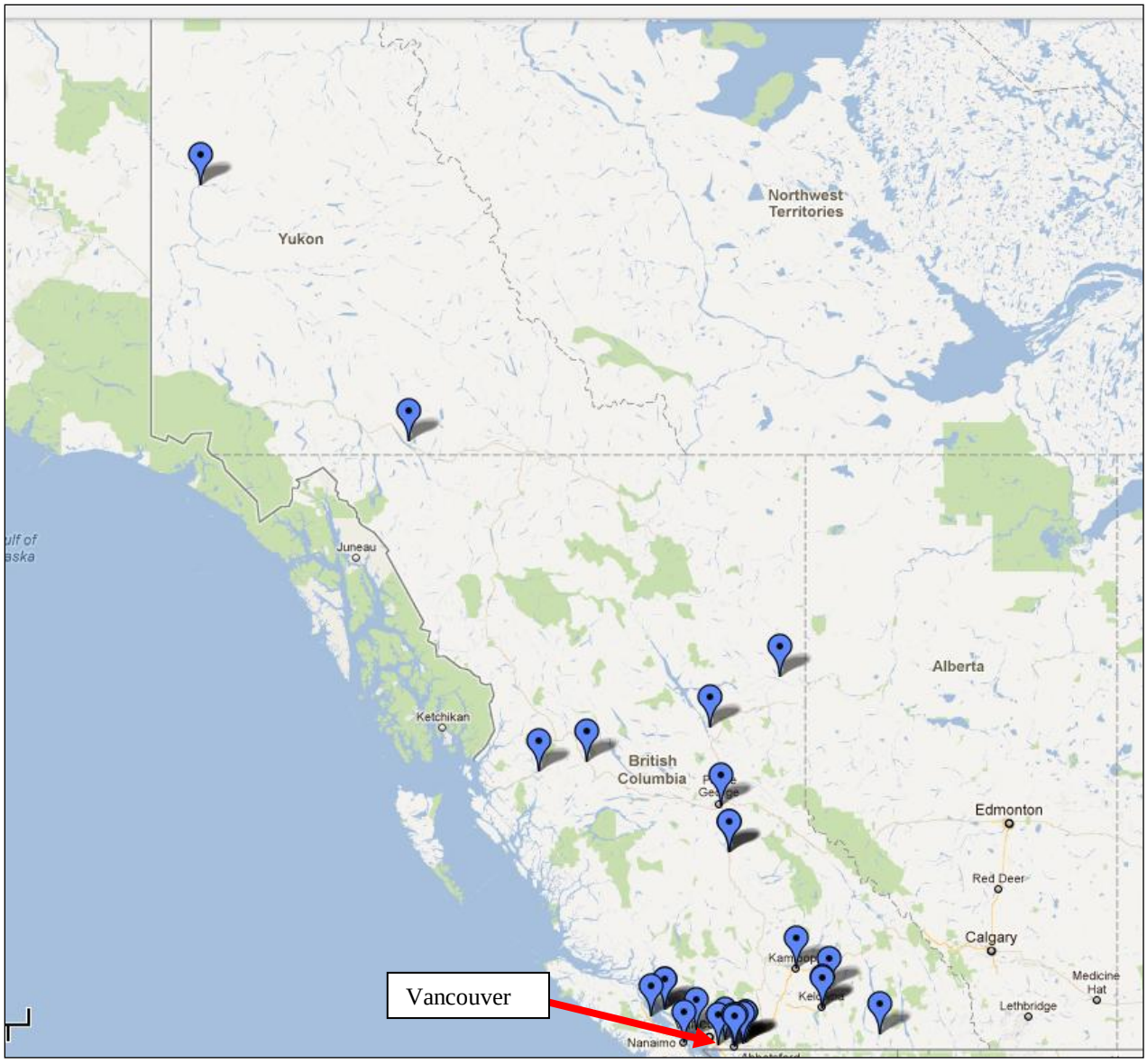
7.5.4 Implications and future directions

Telehealth in genetics and telemedicine in healthcare more broadly is not new, and has demonstrated value in other areas of complex decision making-around predictive genetic testing such as hereditary BRCA and HNPCC/ FAP cancer counseling (Buchanan, *et al*., 2011; Coelho, *et al*., 2005; Gattas, *et al*., 2001). These studies have not only demonstrated the effectiveness of such services in improving access to genetic counseling services, but have also demonstrated that such services may be cost-effective and even cost-saving to the healthcare system (Buchanan, *et al*., 2011; Mair, *et al*., 2000; Wootton & Bonnardot, 2010). Moreover a wealth of data supports the use of telehealth in communication, care provision and counseling around sensitive and complicated topics in psychiatry such as mental health and post traumatic stress disorder (Bee, *et al*., 2008; Norman, 2006; Richardson, *et al*., 2009). Given the higher penetration and growing comfort level with technological innovation and digital communication methods, particularly in younger generations, it is perhaps surprising that the uptake of telemedicine in PT for HD has been somewhat slow and there is reticence to change among care providers.

The results of this project assist in the evolution and modernizing of predictive genetic testing services in HD to address the recognised access barriers to genetic services, particularly for those from rural communities. To accomplish this goal, 1) an in-depth,

considered and multi-pronged approach to developing a supportive and feasible HD PT telehealth protocol was demonstrated, and 2) preliminary data, through a pilot study, that demonstrates the effectiveness of this service in terms of patient satisfaction, quality of care and support during the PT process was generated. As such, this is the first study to report the feasibility and value of telehealth based PT in comparison to in-person based PT for those who cannot easily access predictive genetic testing services.

Figure 7.10: Map illustrating the location of telehealth tested pilot project participants (N=28)



Each blue pointer represents the location of an individual who participated in the telehealth pilot testing project. Of the 28 individuals who participated, 8 were located within the lower mainland (i.e. within a two-hour drive from Vancouver) and are clustered in the Vancouver area on the map. The remaining 20 individuals were located throughout BC and the Yukon. Please note that those located on Vancouver Island, and the Sunshine Coast, though geographically close to Vancouver, are required to take a ferry to reach the mainland, meaning journey time to Vancouver is over two hours.

8. CONCLUSION

8.1 Summary of results and key insights

This project represents the first study of its kind to explore structural and physical access barriers to HD PT, as well as providing the first website dedicated to PT, and a comprehensive telehealth PT protocol. The first part of the project examined whether access to genetic services is indeed perceived as a barrier or deterrent to PT for HD by at-risk individuals. Results of the mapping study, survey study and interview study revealed that access to PT is a barrier to PT in rural areas, with PT rates being lower in rural areas than non-rural areas. The interview study provided a more in-depth understanding of the nature of these access barriers and found that access was an issue not just due to distance related factors but also due to the inflexible nature of the PT process. The interview study, coupled with the expert workshop, provided assessment of the key components of the PT process such as information and education provision, quality of care and support, and pre and post-test counseling from an at risk individual and expert HCP perspective. This information was used to develop a user-friendly PT protocol and website for HD, which provides remote testing for HD while still maintaining quality of care, counseling, education and follow-up support. Pilot testing of the website suggested that the site did indeed acts as a guide and reliable information source throughout the PT decision-making period and testing process, including the post-results period.

The final part of this study involved a pilot project to compare the telehealth PT protocol with the standard testing protocol on a variety of quality assurance measures to determine whether this novel protocol is equivalent to the current standard of care across established outcome measures such as satisfaction and support. Results of the pilot project

revealed that providing PT via telehealth not only equals traditional in-person testing in terms of patient satisfaction, support and education, but also surpasses the traditional model in terms of improving access to PT services. Providing PT via telehealth is therefore warranted both in terms maintaining quality of care and support throughout the testing process and importantly in improving access to services and support for those in need. The survey results indicated that there were no significant differences in terms of quality of care, information, counseling and support during the PT process between the VT and TT groups. The majority of participants in both groups believed that the counseling process had provided them with appropriate information regarding the advantages and disadvantages of PT and the ability to make an informed, supported decision around whether or not to proceed with PT. The telehealth service also appeared to improve access to PT and afforded savings in terms of patient's time and money (these amounts can be substantial). In addition, the telehealth service allowed a spouse or other support person(s) to more easily attend the telehealth appointment; indeed a number of patients also brought other family members to the initial appointment, which not only provides additional support for the person undergoing PT, it also allows these individuals the opportunity to learn more about HD, the PT process and the advantages and disadvantages of receiving test results for both their family member and potentially themselves. It is this improved access to support, counseling, accurate information and specialized services, not the actual uptake of PT, that is most important from a justice perspective. Equalizing access to such services, so that ability to undergo testing and receive support is not limited by location, allows for a more just healthcare system, particularly in countries such as Canada which espouse universal healthcare.

Table 8.1: Summary of key results

Key results
1. Predictive test uptake rates in BC are lower in rural areas as compared to non rural areas (16% and 30% respectively).
2. Access to PT can be a barrier to receiving testing and counseling due to distance related factors and due to inflexibility in the PT process.
3. A specially developed PT website, which includes educational information, stories, diagrams and short documentaries, may aid decision-making in PT and provide a reliable information source throughout the PT process.
4. Telehealth PT is equal to traditional in-person PT in terms of patient satisfaction with the PT process, support and education.
5. Providing PT via telehealth improves access to PT for those from both rural and non-rural areas.

Delivering PT via telehealth represents a paradigm shift in the delivery of genetic services strategy that is more individualized, accessible and readily available. This project adds to a growing body of literature on the utility of telehealth services in genomic medicine within a broader context of increasing technological innovation and comfort with novel communication mechanisms (Buchanan, *et al*., 2011; Mair, *et al*., 2000; Wootton & Bonnardot, 2010; Coehlo *et al*., 2005). In addition, improving accessibility to counseling and information on HD PT increases the number of individuals who are aware of research advances and clinical trials in HD, ultimately leading to greater number of potential research participants and research beneficiaries. As such, this project has relevance and utility for both HD and other genetic conditions.

8.2 Strengths and limitations of the project

While previous studies have explored emotional and psychological barriers and reasons for not undergoing PT (Cox, 1999; Cox 2003; Benjamin *et al*., 1994; Bloch *et al*.,

1992; Wiggins, *et al*., 1992), and some have suggested that structural barriers may exist (Kessler, 1994), there has not been a thorough and systematic exploration of the nature of these barriers, or how these barriers might be addressed. The research presented here not only represents an in-depth qualitative analysis of the nature of these access issues, it also informed the development and subsequent pilot-testing of a novel telehealth protocol for providing global telehealth PT for HD.

Until this point, no such protocol has been systematically developed, using a rigorous mixed-methodology approach, and then pilot-tested to reveal that telehealth PT improves access while maintaining quality of information, care and support. Employing a mixed methodological approach, drew on the strengths of different research approaches, ultimately achieving a more rigorous, complete and comprehensive understanding of 1) the nature of access barriers to PT, and 2) the development and evaluation of the proposed solutions to these access barriers (Flick, 1998). The initial qualitative interview component of the study, coupled with the mapping study (which compared PT uptake in rural and non-rural areas in B.C.) and survey of at-risk individuals allowed for exploration and understanding of the complexities pertaining to access issues, revealing important factors that may have been missed by quantitative measures alone. For example, the interview study revealed that time, distance, financial and other opportunities were factors that may act as disincentives to PT. While these factors could perhaps have been surmised from prior literature in other areas of genetics (d'Agincourt-Canning, 2004; Abrams & Geier, 2006), the interview analysis resulted in some unexpected findings which may be specific to HD. For example the inflexibility of the current PT protocol, in and of itself (whether

real or perceived), was identified by some as a barrier to PT. As such, the initial qualitative data collection and analysis, coupled with the mapping study that revealed lower PT rates in rural communities, and survey study that revealed support for PT via telehealth, provided a strong foundation for the next stage of this project.

Building on the findings from part one of the research project (the mapping study, the interview study and the survey of at-risk individuals), the expert workshop involved international attendees with experience and expertise in the provision of PT, HD, and telehealth. Such a workshop allowed us to discuss our initial findings from part one of the project and then present initial ideas regarding the provision of PT via telehealth. This enabled a more complete examination of access issues to PT in other jurisdictions (including other areas of Canada, the U.S. and the U.K.). In addition, the concept of providing PT via telehealth was thoroughly critiqued, with expert attendees providing useful feedback on the protocol, provision of results and follow-up. For example, the expert workshop informed us that, in most circumstances, it would be preferable for the results to be delivered in person, by a trained HCP. This process both enabled further development of the protocol by ensuring potential criticisms and concerns were considered *a priori*, and by ensuring that protocol development was guided and in keeping with acceptable standards of care for the provision of PT. The expert workshop also facilitated an exploration of the needs of a website dedicated to PT, including evaluation of the provisional PT website model and content. The workshop also thus contributed to the development of a website with the knowledge-users in mind, while ensuring accurate portrayal of necessary information.

By conducting such a thorough analysis of the issue at hand – namely access to PT for HD – through the first part of the project, the next stages of this research could draw on the strengths and breadth of these results to develop a robust telehealth PT protocol. This process was informed by the results of the first part of the study and by existing guidelines and practices, including the availability of local telehealth facilities. The resultant protocol considers the wide range of issues, concerns, practical limitations and criticisms that might arise, resulting in a more robust, considered and useful protocol than would have otherwise been developed.

Evaluation of the pilot component of the project involved predominantly quantitative surveys, which allowed for some objective quantification of the new protocol in comparison to standard in-person testing. The survey questions employed were based on established validated tools where possible. Where new or modified questions were included, these questions were first pilot-tested to ensure they were comprehensible and adequately captured the factors to be measured and examined. Using this quantitative methodology, built on the findings of the initial predominantly qualitative stages of research, allowed for in-depth examination of the telehealth protocol to test preliminary theories and ideas developed in the first two stages of the project. Overall, the mixed methodologies employed in this research project allowed for exploration of the research questions from diverse perspectives, to maximize learnings and test emergent ideas (Chenail, 2000; Denzin & Lincoln, 2000; Creswell, 2009).

While the research project did have numerous strengths, in terms of novelty of the research and findings, as well as the methodological approach, the project also had some limitations. First, the project was conducted in BC, and it is unclear whether access barriers would be similar in other jurisdictions. For example, access barriers in systems with non-universal health coverage, such as the US, may differ and may also relate to insurance coverage and ability to pay for health services (Freeman & Corey, 1993). In addition, there are some areas, such as countries in Europe, where the population concentration is denser, and countries are smaller, requiring less travel time to reach large urban centres. Access in these areas may be less of a concern than in countries such as Canada where some sectors of the population reside many hours from the nearest medical genetics centre. Nonetheless, our results revealed that even for individuals who lived relatively close to Vancouver access was still a barrier to testing (Hawkins, Creighton & Hayden, 2012), so it is likely that our findings are also applicable in other areas. We propose that rather than a weakness, questions regarding the applicability of this research in other jurisdictions represent an opportunity for further research and comparison, as will be discussed in the section on future research opportunities (section 8.4)

Another limitation of the current study relates to recruitment of participants which occurred at various stages of the project. Due to the nature of the study, financial constraints and privacy considerations, the different components of the study had limited sample sizes. For example, the pilot study involved 15 people who went through the telehealth protocol, in comparison to 13 people who went through the protocol via in-person testing. As such, only preliminary comparisons between the two groups can be

made, and a larger study is required to confirm the findings. In addition, it is difficult to know the exact reasons why some people chose not to participate in various stages of the project, such as responding to surveys or interview invitations. Due to ethical and privacy concerns follow-up with non-responders was not usually possible. As such there may have been a significant selection bias. It is not possible to know whether non-respondents would have had significantly different viewpoints and perspectives on the various topics examined during the research project. Strategies to address this issue, including incentives, may have aided response rate, and would have been useful if resources were available, yet may have also introduced other selection biases.

The preliminary nature of the pilot project and constraints of doctoral research meant that long term data on individuals who underwent PT via telehealth could not be collected. Such data would be useful to determine the longer term safety and satisfaction with the telehealth protocol. Collection of 3 and 5 year follow-up data on individuals who underwent telehealth PT could be compared to other studies that have looked at long-term well-being of individuals who have undergone PT in other jurisdictions and for other disease (Tibben *et al*., 1997; Almqvist *et al*. 2003).

Finally, this study was interested in exploring structural and physical access barriers to PT. As such, the study questions focused on these issues, but did not examine other potential impediments to access, including psychological and resource based barriers. Previous research has indicated that psychological barriers may be another important consideration in understanding why people decide whether or not to pursue testing (Cox,

1999; Cox 2003; Benjamin *et al*., 1994; Bloch *et al*., 1992; Wiggins, *et al*., 1992); this was beyond the scope of this research. Further, resource access barriers may be quite different in other jurisdictions. For example, in South American countries, one of the major access issues to PT is the availability of trained personnel to provide PT, and support during the entire process. Moreover, PT may not even be available in some regions. In such cases, the only method of obtaining PT may be through private testing via a genetic laboratory in another country. Finally, education regarding the availability and potential advantages of PT may exist, and some at risk individuals might not even know that PT is an option[29]. Further research in these areas may help elucidate other access barriers to PT.

8.3 Practice recommendations

The results of this pilot study show that providing PT via telehealth can improve access to testing without compromising patient well-being or support. Based in part on this work, the newly revised international HD PT guidelines allow for predictive test counseling to be provided by telehealth in situations where access to a specialized centre is not possible (see MacLeod *et al.* Recommendations for the Predictive Genetic Test in Huntington's Disease. 2012). Specifically these guidelines state that: "*Wherever possible, support should be available close to the person's community, and on a remote basis, by phone or telehealth where necessary"* (MacLeod *et al.*, 2012, p5).

[29] These access barriers in South America are based on discussions with Sonia Margarit (Universidad del Desarrollo, Santiago, Chile) and Mario Cornejo (CH San Pablo Sede Surco, Neurología-Neurogenética INCN, Lima, Peru). Sonia Margarit is a genetic counselor working in HD in Chile, and Mario Cornejo is a neurologist working in HD in Peru.

The results of the pilot study also have implications for the provision of PT worldwide. As a result of the pilot study, the Centre for HD in Vancouver now routinely offers PT via telehealth to those individuals who are considering PT in B.C. and the Yukon. In addition, the Huntington Society of Canada has also recognised the importance and potential impact of the research and is currently developing a strategy to roll-out the telehealth PT service in other areas of Canada.

Finally, this research has significant implications for personalized medicine and other genetic diseases. This project focused on the translation of clinically relevant and meaningful genetic knowledge in an accessible manner. This mixed methods, multistep telehealth model development and testing process provides a reliable model that may be adapted and implemented in other areas of genetic services. While telehealth in genetics, and telemedicine more broadly, is not new there remains limited provision of such services despite demonstrated value in other areas of complex decision making in genetic testing such as hereditary cancer counseling (Buchanan, *et al*., 2011; Coelho, *et al*., 2005; Gattas, *et al*., 2001). These studies have established the effectiveness of such services in improving access to genetic services, but have also indicated the cost-effectiveness of such services (Buchanan, *et al*., 2011; Mair, *et al*., 2000; Wootton & Bonnardot, 2010). In addition, there is considerable data to support the use of telehealth in communication, care provision and counseling around sensitive and complicated topics in psychiatry such as mental health and post-traumatic stress disorder (Bee, *et al*., 2008; Norman, 2006; Richardson, *et al*., 2009). It is hoped that the results of this project will add to the growing body of literature supporting the utility of telehealth in the provision of genetic services, ultimately

improving access to testing, counseling, support and research for those at-risk for and impacted by all genetic diseases.

8.4 Future research

There are a number of key related areas of research that that are beyond the scope of this research project. As noted above, further longitudinal research and involving a larger and more diverse cohort is required to confirm the pilot study findings. Such research is also necessary to substantiate to determine the long-term consequences of receiving PT via telehealth. Such long-term studies would allow for comparison with published literature on the three and five year impact of receiving HD predictive test results using methods similar to those presented in this research (Almqvist *et al*., 2003; Tibben, 1997). In addition, longer term follow up would enable a better understanding of the time frame in which people make decisions regarding testing. This would elucidate whether the larger 'drop-out' rate for those undergoing PT via telehealth indicated postponement versus a true decision not to pursue testing. Longer term studies would also allow assessment of PT uptake rates in the rural versus non-rural population in BC This would enable an understanding as to whether differences in uptake rates observed in the mapping study are decreased over time with the availability of PT via telehealth, or whether other potential barriers or factors influencing the uptake rates in the two groups need to be explored. Finally, larger longitudinal studies may help answer broader justice questions, such as the health, psychological and other impacts of differing levels of access to genetic services.

An additional avenue for future research involves exploring the perspective and satisfaction of the local HCP involved in the telehealth PT protocol. Assessment of their understanding of the process, the disease, meaning of results, advantages and disadvantages of testing and psychosocial aspects of the process is important in ensuring standardized and high quality of care for individuals undergoing PT. In addition, evaluation of local HCPs' level of comfort and satisfaction with support from the Centre for HD-based team will also be necessary if the growth is to be realized over the longer term. Further evaluation and updating of the PT website developed for this project is essential. Such evaluation could include a pre-and post website viewing survey to determine whether the website adequately meets the goals of aiding the PT decision making process as well as providing accurate and relevant information regarding HD, the PT process, the advantages and disadvantages of PT, the meaning of results, and so on.

Further work is needed to translate the telehealth model to other regions in Canada, as well as evaluate the effectiveness of the model elsewhere. Successful global roll-out of the telehealth PT model requires exploration of service translation into other areas of the world to establish the effectiveness of the telehealth model in terms of quality of care, accessibility, patient satisfaction and support. For example, South American countries have more and less well developed infrastructures, with varying degrees of literacy, telehealth connectivity and access to predictive genetic testing services. There are also important differences among healthcare system (e.g. universal health care versus insurance-based), and country-specific privacy and licensing considerations. For example, the US has a primarily insurance-based healthcare system, with different privacy and discrimination

laws that must be taken into account in the set up of a telehealth service. Conducting roll-out and ongoing evaluation in these different areas is necessary to determine the region-specific challenges to providing PT via telehealth in terms of feasibility, acceptability, obstacles and barriers such as:

- attitude toward PT (including the influence of culture/ religion, knowledge of genetics/ inheritance, etc.)
- familiarity and comfort with telehealth and technology in general
- healthcare system diversity and variability in HCP knowledge
- communication mechanisms (including health literacy, access to internet, etc.)
- connectivity (i.e., availability of a telehealth network, or secure internet based telehealth platform)

Understanding and devising solutions to resolve challenges that arise is essential to achieve one of the primary objectives of this study, namely developing a telehealth PT model that is accessible from anywhere. Replication of this study in other jurisdictions is needed to test the robustness, efficacy, feasibility and accessibility of the telehealth protocol in diverse geographical areas and healthcare infrastructures. Cost-effectiveness analyses will be an essential part of such studies, and ultimately a fundamental component of the success and availability of any telehealth PT service.

From a broader ethics and policy perspective, cost-effectiveness analyses will be also be important in answering difficult resource allocation questions, such as whether we should provide access and develop capacity for telehealth genetics services versus invest

societal dollars in other areas of healthcare which may realize greater utility. Such analyses are imperative specifically because they may help to answer the issues raised at the beginning of this thesis (chapter two) regarding justice and equity in healthcare provision. Fundamental questions regarding the impact and utility of improving access and the provision of genetic services remain unanswered. Measuring this impact will likely be difficult. As we have seen, the impact may be primarily psychological, and result in a reduced (or increased) burden of stress for individuals and families at risk for HD. Subsequent differences in career choice, education, insurance and other factors may then result in social and economic effects. The impact of PT availability may also result in cost-savings or cost-expenditures for the healthcare system. For example, individuals receiving results may choose expensive reproductive options, such as in vitro fertilization and preimplantation genetic diagnosis, and these decisions may have long-term healthcare system ramifications.

The precise consequences of improving access to PT, and genetic services more broadly, are difficult to quantify, and are further complicated by the need to compare the effects of genetics services to other areas of healthcare. Moreover, as with other areas of healthcare, genetics is a constantly evolving field, with new treatments, technologies and diagnostic techniques on the horizon. For example, if a cure or treatment for HD is found in the coming years, the argument for improving access to PT will likely shift significantly and become clearer. Yet even then consideration of other factors, including resource allocation in healthcare, will result in further discussion regarding the necessity, utility and importance of access to HD PT, and genetic services more generally. As such there is need

for ongoing examination of the questions presented in this thesis, as well exploration of dynamic and creative solutions to access issues. Ultimately this will enable a more just and equitable healthcare system, which is responsive to the changes in our health knowledge and the needs of our population.

BIBLIOGRAPHY

Abrams, D., & Geier, M. (2006). A comparison of patient satisfaction with telehealth and on-site consultations: a pilot study for prenatal genetic counseling. *Journal of Genetic Counseling,* 15(3), 199.

Acheson, D., Baker, D., & Illsley, R. (1998). Inequalities in health. *British Medical Journal,* 317(7173), 1659.

Adair, A., Hyde-Lay, R., Einsiedel, E., & Caulfield, T. (2009). Technology assessment and resource allocation for predictive genetic testing: A study of the perspectives of Canadian genetic healthcare providers. *BMC Medical Ethics, 10*(1), 6.

Adelson, N. (2005). The embodiment of inequity. *Canadian Journal of Public Health*. 96(s2): 545.

Alkire, S., & Chen, L. (2004). Global health and moral values. *The Lancet,* 364(9439), 1069.

Allin, S. (2008). Does equity in healthcare use vary across Canadian provinces? *Healthcare Policy,* 3(4), 83.

Almqvist, E., Brinkman, R., Wiggins, S., & Hayden, M. (2003). Psychological consequences and predictors of adverse events in the first 5 years after predictive testing for Huntington's disease. *Clinical Genetics,* 64(4), 300.

Almqvist, E.W., Bloch, M., Brinkman, R., Craufurd, D., & Hayden, M. (1999). A worldwide assessment of the frequency of suicide, suicide attempts, or psychiatric hospitalization after predictive testing for Huntington disease. *The American Journal of Human Genetics,* 64(5), 1293.

Anderson, J.M., Rodney, P., Reimer-Kirkham, S., *et al.* (2009). Inequities in health and healthcare viewed through the ethical lens of critical social justice: Contextual knowledge for the global priorities ahead. *Advances in Nursing Science.* 32: 282.

Andrew, S.E., Goldberg, Y.P., Kremer, B., Telenius, H., Theilmann, J., Adam, S., *et al*. (1993). The relationship between trinucleotide (CAG) repeat length and clinical features of Huntington's disease. *Nature Genetics,* 4(4), 398.

Arber, S., & Ginn, J. (1993).Gender and inequalities in health in later life. *Social Science & Medicine.* 36(1): 33.

Arbour, L., & Cook, D. (2006). DNA on loan: Issues to consider when carrying out genetic research with aboriginal families and communities. *Public Health Genomics,* 9(3), 153.

Asada, Y., & Kephart, G. (2007). Equity in health services use and intensity of use in Canada. *BMC Health Services Research,* 7(1), 41.

Association of Genetic Counselors and Nurses (UK), Accessed June 1, 2011 from: http://www.agnc.org.uk/About%20us/aboutagnc.htm.

Babbie, E. (1990). *Survey Research Methods* (2nd ed.). Belmont, CA: Wadsworth.

Baumanis, L., Evans, J., Callanan, N., & Susswein, L. (2009). Telephoned BRCA1/2 Genetic Test Results: Prevalence, Practice, and Patient Satisfaction. *Journal of Genetic Counseling,* 18(5), 447.

Baylis, F., Kenny, N.P., & Sherwin, S. (2008). A relational account of public health ethics. *Public Health Ethics*. 1(3): 196.

Beauchamp, D. E. (1976). Public health as social justice. *Inquiry,* 13(1), 3.

Beauchamp, T., & Childress, J. (2009). *Principles of Biomedical Ethics*. New York, NY: Oxford University Press.

Beck, A.T., Ward, C,. & Mendelson, M. (1961). Beck Depression Inventory (BDI). *Archives of General Psychiatry* 4(6): 561.

Bee, P., Bower, P., Lovell, K., Gilbody, S., Richards, D., Gask, L., *et al*. (2008). Psychotherapy mediated by remote communication technologies: a meta-analytic review. *BMC Psychiatry,* 8(1), 60.

Bell, T. S., Branston, L. K., Newcombe, R. G., & Barton, G. R. (1999). Interventions to improve uptake of breast screening in inner city Cardiff general practices with ethnic minority lists. *Ethnicity and Health,* 4(4), 277.

Benjamin, C. M., Adam, S., Wiggins, S., Theilmann, J. L., Copley, T. T., Bloch, M., *et al*. (1994). Proceed with care: Direct predictive testing for Huntington disease. *American Journal of Human Genetics,* 55(4), 606.

Berg, B. (1998). *Qualitative Research Methods for the Social Sciences* (6th ed.). Boston, MA: Allyn and Bacon.

Bernhardt, C., Schwan, A. M., Kraus, P., Epplen, J. T., & Kunstmann, E. (2008). Decreasing uptake of predictive testing for Huntington's disease in a German centre: 12 years' experience (1993–2004). *European Journal of Human Genetics,* 17(3), 295.

Binedell, J., Soldan, J. R., & Harper, P.S. (1998). Predictive testing for Huntington's disease: II. Qualitative findings from a study of uptake in South Wales. *Clinical Genetics,* 54(6), 489.

Bloch, M., Adam, S., Wiggins, S., Huggins, M., & Hayden, M. (1992). Predictive testing for Huntington disease in Canada: The experience of those receiving an increased risk. *American Journal of Medical Genetics,* 42(4), 499.

Boeije, H. (2002). A purposeful approach to the constant comparative method in the analysis of qualitative interviews. *Quality & Quantity,* 36(4), 391.

Bombard, Y., Penziner, E., Decolongon, J., Klimek, M., Creighton, S., Suchowersky, O., *et al*. (2007). Managing genetic discrimination: Strategies used by individuals found to have the Huntington disease mutation. *Clinical Genetics,* 71(3), 220.

Bombard, Y., Veenstra, G., Friedman, J., Creighton, S., Currie, L., Paulsen, J., *et al*. (2009). Perceptions of genetic discrimination among people at risk for Huntington's disease: A cross sectional survey. *British Medical Journal,* 338, b2175.

Bonelli, R., Wenning, G., & Kapfhammer, H. (2004). Huntington's disease: present treatments and future therapeutic modalities. *International Clinical Psychopharmacology,* 19(2), 51.

Bortolotti, L., & Widdows, H. (2011). The right not to know: the case of psychiatric disoders. *Journal of Medical Ethics,* 37, 673.

Braithwaite, D., Emery, J., Walter, F., Prevost, A.T., & Sutton, S. (2004). Psychological impact of genetic counseling for familial cancer: a systematic review and meta-analysis. *Journal of the National Cancer Institute.* 96,122.

Brandt, J., Quaid, K.A., Folstein, S.E., Garber, P., Maestri, N.E., Abbott, M.H., *et al*. (1989). Presymptomatic diagnosis of delayed-onset disease with linked DNA markers. The experience in Huntington's disease. *Journal of the American Medical Association,* 261(21), 3108.

Broadstock, M., Michie, S., & Marteau, T. (2000). Psychological consequences of predictive genetic testing: a systematic review. *European Journal of Human Genetics,* 8(10), 731.

Buchanan, A., Datta, S., Adams, M., Hollowell, G., Beresford, H., Boling, J., *et al*. (2011). *Telemedicine vs. in-person cancer genetic counseling in rural oncology clinics: a randomized controlled trial of cost and patient satisfaction*. Paper presented at the 2011 American College of Medical Genetics Annual Education Conference in Vancouver, Canada.

Burgess, M. & d'Agincourt-Canning, L. (2002). Genetic testing for hereditary disease: Attending to relational responsibility. *Journal of Clinical Ethics.* 12,361.

Burgess, M. Proposing modesty for informed consent. (2007). *Social Science & Medicine.* 65,2284.

Burke, S., Martyn, M., Stone, A., Bennett, C., Thomas, H., & Farndon, P. (2009). Developing a curriculum statement based on clinical practice: genetics in primary care. *British Journal of General Practice,* 59(559), 99.

Butow, P., Fowler, J., & Ziebland, S. (2005). Section E: Using personal stories. A.O'Connor, H., Llewelyn-Thomas, & D. Stacey (Eds.), *International Collaboration Document,*

International Patient Decision Aids Standards (IPDAS) Collaboration, 24. Accessed June 24, 2012 from: http://ipdas.ohri.ca/IPDAS_Background.pdf

Canada Health Act. (2005). *Canada Health Act FAQs.* Accessed April 7, 2010, from http://www.hc-sc.gc.ca/hcs-sss/medi-assur/res/faq-eng.php#a6.

Canadian Association of Genetic Counselors. Accessed October 17, 2011 from: https://cagc-accg.ca.

Casey, M., Thiede, C., & Klingner, J. (2001). Are rural residents less likely to obtain recommended preventive healthcare services? *American Journal of Preventive Medicine,* 21(3),182.

Caulfield, T. (2000). Underwhelmed: hyperbole, regulatory policy, and the genetic revolution. *McGill Law Journal, 45*, 437.

Caulfield, T., Burgess, M., & Williams-Jones, B. (2001). Providing genetic testing through the private sector: a view from Canada. *ISUMA: Canadian Journal of Policy Research.* 2,72.

Caulfield, T., McGuire, A., Cho, M., Buchanan, J., Burgess, M., Danilczyk, U., *et al*. (2008). Research ethics recommendations for whole-genome research: Consensus statement. *PLoS Biology,* 6(3),73.

Central Intelligence Organization, (2009). World Fact Book. Accessed November 30, 2009, from: https://www.cia.gov/library/publications/the-world-factbook/fields/2212.html.

Charmaz, K. (2006). *Constructing Grounded Theory: A Practical Guide through Qualitative Analysis.* Thousand Oaks, CA: Sage Publications.

Chenail, R. (2000). Practicing Qualitative Research: Navigating the "Seven C's": Curiosity, Confirmation, Comparison, Changing, Collaborating, Critiquing, and Combinations. *The Qualitative Report,* 4(3 & 4).

Coburn, D. (2004). Beyond the income inequality hypothesis: Class, neo-liberalism, and health inequalities. *Social Science & Medicine*, 58(1), 41.

Codori, A.M., & Brandt, J. (1994). Psychological costs and benefits of predictive testing for Huntington's disease. *American Journal of Medical Genetics,* 54(3), 174.

Coelho, J., Arnold, A., Nayler, J., Tischkowitz, M., & MacKay, J. (2005). An assessment of the efficacy of cancer genetic counselling using real-time videoconferencing technology (telemedicine) compared to face-to-face consultations. *European Journal of Cancer,* 41(15), 2257-2261.

Colin, L. (2007). Statistics Canada: Census Snapshot. Accessed November 30, 2009, from: http://www.statcan.gc.ca/pub/11-008-x/2007004/pdf/10313-eng.pdf.

Collins, F.S., Green, E.D., Guttmacher, A. E., Guyer, M.S., *et al*., (2003). A vision for the future of genomics research. *Nature* 422(6934), 835.

Cook, D. A., & Dupras, D. M. (2004). A practical guide to developing effective web-based learning. *Journal of General Internal Medicine,* 19(6), 698.

Cookson, R., & Dolan, P. (2000). Principles of justice in healthcare rationing. *British Medical Journal,* 26(5), 323.

Corbie-Smith, G., Thomas, S., & St George, D. (2002). Distrust, race, and research. *Archives of Internal Medicine,* 162(21), 2458.

Corbin, J., & Strauss, A. (2008). *Basics of qualitative research: Techniques and procedures for developing grounded theory*. Thousand Oaks, CA: Sage Publications.

Cox, S. (1999). *Personal Perspectives on Genetic Testing for Huntington Disease*: Huntington Society of Canada. Accessed Feb. 5 2012 from: http://www.huntingtonsociety.ca/english/uploads/Perspectives_Book.pdf.

Cox, S.M., & McKellin, W. (1999). 'There's this thing in our family': Predictive testing and the construction of risk for Huntington disease. *Sociology of Health & Illness.* 21(5): 622.

Cox, S.M., (2003). Stories in decisions: How at-risk individuals decide to request predictive testing for Huntington disease. *Qualitative Sociology*. 26(2): 257.

Craig, J. (2008). Complex diseases: Research and applications. *Nature Education,* 1(1).

Craufurd, D., Kerzin-Storrar, L., Dodge, A., & Harris, R. (1989). Uptake of presymptomatic predictive testing for Huntington's disease. *The Lancet, 334*(8663), 603.

Creighton, S., Almqvist, E., MacGregor, D., Fernandez, B., Hogg, H., Beis, J., *et al*. (2003a). Predictive, pre-natal and diagnostic genetic testing for Huntington's disease: the experience in Canada from 1987 to 2000. *Clinical Genetics,* 63(6), 462.

Creswell, J. (2009). *Research Design: Qualitative, quantitative, and mixed method approaches* (3rd ed.). Thousand Oaks, CA: Sage Publications.

d'Agincourt-Canning, L. (2004). Genetic testing for hereditary cancer: Challenges to ethical care in rural and remote communities. *Healthcare Ethics Committee Forum,* 16(4), 222.

Dahlgren, G. (2008). Neoliberal reforms in swedish primary healthcare: For whom and for what purpose? *International Journal of Health Services,* 38(4), 697.

Daniels, N. (1985). *Just Healthcare*. West Nyack, NY: Cambridge University Press.

Daniels, N. (2000). Accountability for reasonableness. *British Medical Journal,* 321(7272), 1300.

Daniels, N. (2001). Justice, health, and healthcare. *American Journal of Bioethics,* 1(2), 2.

Daniels, N., & Sabin, J. (1997). Limits to healthcare: fair procedures, democratic deliberation, and the legitimacy problem for insurers. *Philosophy and Public Affairs,* 26(4), 303.

Davis, J., Furstenthal, L., Desai, A., Norris, T., Sutaria, S., Fleming, E., *et al*. (2009). The microeconomics of personalized medicine: today's challenge and tomorrow's promise. *Nature Reviews Drug Discovery, 8*(4), 279.

Decruyenaere, M., Evers-Kiebooms, G., Boogaerts, A., Cassiman, J. J., Cloostermans, T., Demyttenaere, K., *et al*. (1996). Prediction of psychological functioning one year after the predictive test for Huntington's disease and impact of the test result on reproductive decision making. *British Medical Journal,* 33(9), 737.

Decruyenaere, M., Evers-Kiebooms, G., Cloostermans, T., Boogaerts, A., Demyttenaere, K., Dom, R., *et al*. (2003). Psychological distress in the 5-year period after predictive testing for Huntington's disease. *European Journal of Human Genetics,* 11(1), 30.

Demyttenaere, K., Evers-Kiebooms, G., & Decruyenaere, M. (1992). Pitfalls in counseling for predictive testing in Huntington disease. *Birth Defects Original Article Series,* 28(1), 105.

Denton, M., Prus, S., & Walters, V. (2004). Gender differences in health: a Canadian study of the psychosocial, structural and behavioural determinants of health. *Social Science & Medicine.* 58(12): 2585.

Denzin, N., & Lincoln, Y. (Eds.). (2000). *The Handbook of Qualitative Research* (2nd ed.). Thousand Oaks, CA: Sage Publications.

Dufrasne, S., Roy, M., Galvez, M., & Rosenblatt, D. S. (2010). Experience over fifteen years with a protocol for predictive testing for Huntington disease. *Molecular Genetics and Metabolism,* 102(4),494.

Eckman, M., Rosand, J., Greenberg, S., & Gage, B. (2009). Cost-effectiveness of using pharmacogenetic information in warfarin dosing for patients with nonvalvular atrial fibrillation. *Annals of Internal Medicine,* 150(2), 73.

Edwards, J., Greenberg, J., & Sahhar, M. (2008). Global awakening in genetic counseling. *Nature Precedings.* Accessed July 31, 2012 from: http://hdl.handle.net/10101/npre.2008.1574.1

Elwyn, G., Gray, J., & Clarke, A. (2000). Shared decision making and non-directiveness in genetic counselling. *Journal of Medical Genetics,* 37(2), 135.

Entwistle, V.A., Carter, S.M., Cribb, A., & McCaffery, K. (2010). Supporting patient autonomy: the importance of clinician-patient relationships. *Journal of General Internal Medicine,* 25(7), 741.

Entwistle, V. A., France, E. F., Wyke, S., Jepson, R., Hunt, K., Ziebland, S., *et al*. (2011). How information about other people's personal experiences can help with healthcare decision-making: A qualitative study. *Patient Education and Counseling*, 85(3), e291.

Epstein, C. (2004). Genetic testing: Hope or hype? *Genetics in Medicine,* 6(4), 165.

Ernst, F., & Grizzle, A. (2001). Drug-related morbidity and mortality: updating the cost-of-illness model. *Journal of the American Pharmacists Association,* 41(2), 192.

Etchegary, H. (2006). Genetic testing for Huntington's Disease: how is the decision taken? *Genetic Testing,* 10(1), 60.

Evers-Kiebooms, G., Cassiman, J. J., & Van den Berghe, H. (1987). Attitudes towards predictive testing in Huntington's disease: a recent survey in Belgium. *Journal of Medical Genetics,* 24(5), 275.

Evers-Kiebooms, G., & Decruyenaere, M. (1998). Predictive testing for Huntington's disease: a challenge for persons at risk and for professionals. *Patient Education & Counseling,* 35(1), 15.

Flick, U. (1998). *An Introduction to Qualitative Research: Theory, Method and Applications*. London, UK: Sage Publications.

Food and Drug Administration. (2007). *FDA Approves Updated Warfarin (Coumadin) Prescribing Information.* Accessed 2010, March 9, from: http://www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/2007/ucm108967.htm.

Fox, S., Bloch, M., Fahy, M., & Hayden, M. (1989). Predictive testing for Huntington disease: I. Description of a pilot project in British Columbia. *American Journal of Medical Genetics,* 32(2), 211.

Frueh, F., & Gurwitz, D. (2004). From pharmacogenetics to personalized medicine: a vital need for educating health professionals and the community. *Future Medicine,* 5(5), 571.

Freeman, H.E., & Corey, C.R. (1993). Insurance status and access to health services among poor persons. *Health Services Research* 28(5): 531.

Futter, M., Heckmann, J., & Greenberg, L. (2009). Predictive testing for Huntington disease in a developing country. *Clinical Genetics,* 75(1), 92.

Garber, J., & Offit, K. (2005). Hereditary cancer predisposition syndromes. *Journal of Clinical Oncology,* 23(2), 276.

Garner, R., Carrière, G., & Sanmartin, C. (2010). The Health of Inuit, Métis and First Nations adults living off-reserve in Canada: The impact of socio-economic status on inequalities in health. *Statistics Canada, Health Research Working Paper Series*. Accessed July 31, 2012 from: http://www.statcan.gc.ca/pub/82-622-x/82-622-x2010004-eng.htm

Gattas, M., MacMillan, J., Meinecke, I., Loane, M., & Wootton, R. (2001). Telemedicine and clinical genetics: establishing a successful service. *Journal of Telemedicine & Telecare,* 7(s2), 68.

Genetics Education Center. *Genetic Counseling Programs,* Accessed June 30, 2012 from: http://www.kumc.edu/gec/prof/gcprogs.html.

Giacomini, M., Kenny, N., & DeJean, D. (2009). Ethics frameworks in Canadian health policies: Foundation, scaffolding, or window dressing? *Health Policy*. 89 (1), 58.

Ginsburg, G., & Willard, H. (2009). Genomic and personalized medicine: foundations and applications. *Translational Research*, 154(6), 277

Glaser, B.G., (1965). The constant comparative method of qualitative analysis. *Social Problems,* 12(4), 436.

Goizet, C., Lesca, G., & Dürr, A. (2002). Presymptomatic testing in Huntington's disease and autosomal dominant cerebellar ataxias. *Neurology, 59*(9),1330.

Green, M.J., Biesecker, B.B., McInerney, A.M., Mauger, D., & Fost, N. (2001). An interactive computer program can effectively educate patients about genetic testing for breast cancer susceptibility. *American Journal of Medical Genetics,* 103(1),16.

Green, M.J., & Levi, B.H. (2009). Development of an interactive computer program for advance care planning. *Health Expectations,* 12(1),60.

Green, M.J., Peterson, S.K., Baker, M.W., Friedman, L.C., Harper, G.R., Rubinstein, W.S., *et al*. (2005). Use of an educational computer program before genetic counseling for breast cancer susceptibility: effects on duration and content of counseling sessions. *Genetics in Medicine,* 7(4), 221.

Green, M.J., Peterson, S.K., Baker, M.W., Harper, G.R., Friedman, L.C., Rubinstein, W.S., *et al*. (2004). Effect of a computer-based decision aid on knowledge, perceptions, and intentions about genetic testing for breast cancer susceptibility. *Journal of the American Medical Association,* 292(4), 442.

Grosse, S., & Khoury, M. (2006). What is the clinical utility of genetic testing? *Genetics in Medicine,* 8(7), 448.

Gupta, P., & Lee, K. (2007). Genomics and proteomics in process development: Opportunities and challenges. *Trends in Biotechnology,* 25(7), 324.

Hamburg, M.A., & Collins, F.S. (2010). The path to personalized medicine. *New England Journal of Medicine*. 363(4): 301.

Harper, P., Gevers, S., de Wert, G., Creighton, S., Bombard, Y., & Hayden, M. (2004). Genetic testing and Huntington's disease: issues of employment. *The Lancet Neurology,* 3(4), 249.

Harper, P., Lim, C., & Craufurd, D. (2000). Ten years of presymptomatic testing for Huntington's disease: the experience of the UK Huntington's Disease Prediction Consortium. *Journal of Medical Genetics,* 37(8), 567.

Harper, P. (1988). *Practical Genetic Counseling*. Oxford, UK: Butterman-Heineman.

Harper, P. (1992). The epidemiology of Huntington's disease. *Human Genetics,* 89(4), 365.

Hart, T. (1971). The inverse care law. *The Lancet,* 297(7696), 405.

Hawkins, A.K., Creighton, S., & Hayden, M.R. (2012). When access is an issue: Exploring barriers to pt for Huntington disease in BC, Canada. *European Journal of Human Genetics.* Advanced online publication: http://www.nature.com/doifinder/10.1038/ejhg.2012.147

Hawkins, A.K., & Hayden, M.R. (2011). A grand challenge: Providing benefits of clinical genetics to those in need. *Genetics in Medicine,* 13(3), 197.

Hawkins, A.K., Ho, A., & Hayden, M.R. (2011). Lessons from predictive testing for Huntington disease: 25 years on. *Journal of Medical Genetics,* 48(10), 649.

Hayden, M. (1981). *Huntington's Chorea*. New York, NY: Springer-Verlag.

Hayden, M. (2000). Predictive testing for Huntington's disease: the calm after the storm. *The Lancet,* 356(9246), 1944.

Held, D. (2006). *Models of Democracy* (3rd ed.). Cambridge, MA: Polity Press.

Holtzman, N., & Watson, M. (1998). *Promoting safe and effective genetic testing in the United States: Final report of the Task Force on Genetic Testing*. Baltimore, MD: Johns Hopkins University Press.

Ho, A. (2008). Relational autonomy or undue pressure? Family's role in medical decision making. *Scandinavian Journal of Caring Sciences.* 22(1),128.

Hogarth, S., Javitt, G., & Melzer, D. (2008). The current landscape for direct-to-consumer genetic testing: legal, ethical, and policy issues. *Annual Review of Genomics & Human Genetics* 9,161.

Huggins, M., & Hayden, M. (1992). Predictive testing for Huntington disease. *Journal of Medical Ethics,* 18(1), 47.

Hunter, D. (2005). Gene-environment interactions in human diseases. *Nature Reviews Genetics,* 6(4), 287.

Ioannidis J.P. (2009). Personalized genetic prediction: Too limited, too expensive, or too soon? *Annals of Internal Medicine,* 150(2), 139.

International Society for Equity in Health. (2005). *Definitions*. Accessed April 1, 2010, from: http://www.iseqh.org/workdef_en.htm.

Iredale, R., Jones, L., Gray, J., & Deaville, J. (2005). The edge effect: An exploratory study of some factors affecting referrals to cancer genetic services in rural Wales. *Health & Place,* 11(3), 197.

Judge, K., Mulligan, J.A., & Benzeval, M. (1998) Income inequality and population health. *Social Science & Medicine*. 46(4-5),567.

Kawachi, I., & Kennedy, B. (1997). Socioeconomic determinants of health: Health and social cohesion: Why care about income inequality? *British Medical Journal,* 314(7086), 1037.

Kenny, T., Wilson, R., Purves, I., Clark Sr, J., Newton, L., Newton, D., *et al*. (1998). A PIL for every ill? Patient information leaflets (PILs): a review of past, present and future use. *Family Practice,* 15(5), 471.

Kenny, N., & Joffres, C. (2008). An ethical analysis of international health priority-setting. *Health Care Analysis.* 16(2),145.

Kenny, N.P,, Sherwin, S., & Baylis, F. (2010). Re-visioning public health ethics: a relational perspective. *Canadian Journal of Public Health,* 101(1), 9.

Kessler, S. (1994). Predictive testing for Huntington disease: a psychologist's view. *American Journal of Medical Genetics,* 54(3), 161.

Kessler, S., Field, T., Worth, L., Mosbarger, H., Opitz, J. M., & Reynolds, J. F. (1987). Attitudes of persons at risk for Huntington disease toward predictive testing. *American Journal of Medical Genetics,* 26(2), 259.

Khoury, M. (2003). Genetics and genomics in practice: the continuum from genetic disease to genetic information in health and disease. *Genetics in Medicine,* 5(4), 261.

Khoury, M., Berg, A., Coates, R., Evans, J., Teutsch, S., & Bradley, L. (2008). The evidence dilemma in genomic medicine. *Health Affairs,* 27(6), 1600.

Khoury, M., Gwinn, M., Yoon, P., Dowling, N., Moore, C., & Bradley, L. (2007). The continuum of translation research in genomic medicine: How can we accelerate the appropriate integration of human genome discoveries into healthcare and disease prevention? *Genetics in Medicine,* 9(10), 665.

Khoury, M., Jones, K., & Grosse, S. (2006). Quantifying the health benefits of genetic tests: the importance of a population perspective. *Genetics in Medicine,* 8(3), 191.

Knight, J. (2009). Genetics and the general physician: insights, applications and future challenges. *QJM: An International Journal of Medicine*. 102(11), 757.

Knoppers, B., & Chadwick, R. (2006). Human Genetic Research: Emerging Trends in Ethics. *Focus: The Journal of Lifelong Learning in Pyschiatry*, IV(3), 416.

Kremer, B., Goldberg, P., Andrew, S. E., Theilmann, J., Telenius, H., Zeisler, J., *et al*. (1994). A worldwide study of the Huntington's disease mutation: the sensitivity and specificity of measuring CAG repeats. *New England Journal of Medicine*, 330(20), 1401.

Krugman, P. (2012). The real winners. *New York Times*. Accessed June 28, 2012 from: http://www.nytimes.com/2012/06/29/opinion/the-real-winners.html.

Laccone, F., Engel, U., Holinski–Feder, E., Weigell–Weber, M., Marczinek, K., Nolte, D., *et al*. (1999). DNA analysis of Huntington's disease. *Neurology*, 53(4), 801.

Larsen, D.L., Attkisson, C.C., Hargreaves, W.A., & Nguyen, T.D. (1979). Assessment of client/patient satisfaction: Development of a general scale (CSQ). *Evaluation and Program Planning*, 2(3),197.

Lauerman, J. (2009, Feb 5, 2009). Complete genomics drives down cost of genome sequence to $5,000 *Bloomberg News*. Accessed April 10, 2012 from http://www.bloomberg.com/apps/news?sid=aEUlnq6ltPpQ&pid=20601124.

Lauridsen, S., & Lippert-Rasmussen, K. (2009). Legitimate allocation of public healthcare: beyond accountability for reasonableness. *Public Health Ethics*. 2(1), 59.

Lawson, K., Wiggins, S., Green, T., Adam, S., Bloch, M., & Hayden, M. (1996). Adverse psychological events occurring in the first year after predictive testing for Huntington's disease. The Canadian Collaborative Study Predictive Testing. *British Medical Journal*, 33(10), 856.

Lea, D., Kaphingst, K., Bowen, D., Lipkus, I., & Hadley, D. (2011). Communicating genetic and genomic information: health literacy and numeracy considerations. *Public Health Genomics*, 14(4-5), 279.

Leinsalu, M., Vagero, D., & Kunst, A. (2003). Estonia 1989-2000: enormous increase in mortality differences by education. *International Journal of Epidemiology*, 32(6), 1081.

Lerman, C., Hughes, C., Benkendorf, J., Biesecker, B., Kerner, J., Willison, J., *et al*. (1999). Racial differences in testing motivation and psychological distress following pretest education for BRCA1 gene testing. *Cancer Epidemiology Biomarkers & Prevention*, 8(s1), 361.

Lipkus, I.M. (2007). Numeric, verbal, and visual formats of conveying health risks: suggested best practices and future recommendations. *Medical Decision Making*, 27(5), 696.

Lipkus, I. M., & Hollands, J. (1999). The visual communication of risk. *Jounral of the National Cancer Institute Monographs*, 1999(25), 149.

Lobb, E., Butow, P., Moore, A., Barratt, A., Tucker, K., Gaff, C., *et al*. (2006). Development of a communication aid to facilitate risk communication in consultations with unaffected women from high risk breast cancer families: a pilot study. *Journal of Genetic Counseling,* 15(5), 393.

Longstaff, H., Schuppli, C.A., Preto, N., Lafrenière, D., & McDonald, M. (2009). Scientists' perspectives on the ethical issues of stem cell research. *Stem Cell Reviews and Reports*. 5(2),89.

Lundberg, O. (1991). Causal explanations for class inequality in health--an empirical analysis. *Social Science & Medicine,* 32(4), 385.

Maat-Kievit, A., Vlis, M., Zoeteweij, M., Losekoot, M., Van Haeringen, A., & Roos, R. (2000). Paradox of a better test for Huntington's disease. *Journal of Neurology, Neurosurgery & Psychiatry,* 69(5), 579.

MacDonald, M.E., Ambrose, C.M., Duyao, M.P., Myers, R.H., Lin, C., Srinidhi, L., *et al*. (1993). A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. *Cell,* 72(6), 971.

MacLeod, R., Tibben, A., Frontali, M., Evers-Kiebooms, G., Jones, A., *et al*. (2012). Recommendation for the predictive genetic test in huntington's disease. *Clinical Genetics*. Advanced online publication: DOI: 10.1111/j.1399-0004.2012.01900.x.

Mair, F., Haycox, A., May, C., & Williams, T. (2000). A review of telemedicine cost-effectiveness studies. *Journal of Telemedicine and Telecare,* 6(s1), 38.

Mancini, J., Noguès, C., Adenis, C., Berthet, P., Bonadona, V., Chompret, A., *et al*. (2006). Impact of an information booklet on satisfaction and decision-making about BRCA genetic testing. *European Journal of Cancer,* 42(7), 871.

Markoff, J. (2012). Cost of gene sequencing falls, raising hopes for medical advances. *New York Times*. Accessed June 8, 2012 from: http://www.nytimes.com/2012/03/08/technology/cost-of-gene-sequencing-falls-raising-hopes-for-medical-advances.html?pagewanted=all.

Marmot, M.G. (2005). Social determinants of health inequalities. *The Lancet.* 365(9464),1099.

Marmot, M.G. (2003). Understanding social inequalities in health. *Perspectives in Biology and Medicine.* 46(3),s9.

Martens, P., Fransoo, R., Burland, E. *et al.* (2003). The Manitoba RHA indicators atlas: population-based comparisons of health and health use. *Centre for Health Policy*. Accessed July 31, 2012 from: http://mchp-appserv.cpe.umanitoba.ca/reference/RHA03_Atlas_web.pdf

Mastromauro, C., Myers, R.H., Berkman, B., Opitz, J.M., & Reynolds, J.F. (1987). Attitudes toward presymptomatic testing in Huntington disease. *American Journal of Medical Genetics,* 26(2), 271.

McLeod, C., & Sherwin, S. (2000). in *Relational Autonomy: Feminist Perspectives on Autonomy, Agency and the Social Self.* eds C. Mackenzie & N. Stoljar. Oxford, UK: Oxford University Press.

McCarthy, M., & Hirschhorn, J. (2008). Genome-wide association studies: potential next steps on a genetic journey. *Human Molecular Genetics,* 17(r2), r156.

McCarthy, M., Abecasis, G., Cardon, L., Goldstein, D., Little, J., Ioannidis, J., *et al*. (2008). Genome-wide association studies for complex traits: consensus, uncertainty and challenges. *Nature Reviews Genetics,* 9(5), 356.

Meiser, B., & Dunn, S. (2000). Psychological impact of genetic testing for Huntington's disease: an update of the literature. *British Medical Journal,* 69(5), 574.

Meissen, G.J., & Berchek, R.L. (1987). Intended use of predictive testing by those at risk for Huntington disease. *American Journal of Medical Genetics,* 26(2), 283.

Milnerwood, A., Gladding, C., Pouladi, M., Kaufman, A., Hines, R., Boyd, J., *et al*. (2010). Early increase in extrasynaptic NMDA receptor signaling and expression contributes to phenotype onset in Huntington's disease mice. *Neuron,* 65(2), 178.

Morrison, P., Harding-Lester, S., & Bradley, A. (2011). Uptake of Huntington disease predictive testing in a complete population. *Clinical Genetics,* 80(3), 281.

Morse, J. (1991). Approaches to qualittaive-quantittaive methodological triangulation. *Nursing Research,* 40(1), 120.

National Center for Biotechnology Information. (2009). Accessed November 30, 2009, from http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gnd&part=huntingtondisease.

National Human Genome Research Institute. (2006). The Future of Genomic Medicine: Policy Implications for Research and Medicine. Accessed Feb 12, 2010, from: http://www.genome.gov/17516574.

National Society of Genetic Counselors. (2008). NSGC Professional Status Survey 2008. Accessed December 7, 2009, from: http://www.nsgc.org/_private_files/members_only/PSSsurveys/NSGCPSSReport2008Final.doc.

Neuhauser, L., & Kreps, G.L. (2003). Rethinking communication in the e-health era. *Journal of Health Psychology,* 8(1), 7.

Norman, S. (2006). The use of telemedicine in psychiatry. *Journal of Psychiatric and Mental Health Nursing,* 13(6), 771.

Norrgard, K. (2008). Genetic variation and disease: GWAS. *Nature Education,* 1(1).

Nozick, R. (1974). *Anarchy, state, and utopia*. New York, NY: Basic Books.

Nunes, R. (2003). Evidence-Based Medicine: A new tool for resource allocation? *Medicine, Healthcare and Philosophy,* 6(3), 297.

O'Connor, A.M., Rostom, A., Fiset, V., Tetroe, J., Entwistle, V., Llewellyn-Thomas, H., *et al*. (1999). Decision aids for patients facing health treatment or screening decisions: systematic review. *British Medical Journal,* 319(7212), 731.

Obama, B. (2008). *Speech to the American Medical Association*. Barack Obama's plan for a healthy America: lowering health care costs and ensuring affordable, high-quality health care for all, 2009. Accessed November 3, 2010 from: http://www.barackobama.com/pdf/HealthPlanFull.pdf..

Okamoto, S., Pouladi, M., Talantova, M., Yao, D., Xia, P., Ehrnhoefer, D., *et al*. (2009). Balance between synaptic versus extrasynaptic NMDA receptor activity influences inclusions and neurotoxicity of mutant huntingtin. *Nature Medicine*. 15(12), 1407.

Pagon, R. (2006). GeneTests: an online genetic information resource for healthcare providers. *Journal of the Medical Library Association,* 94(3), 343.

Paling, J. (2003). Strategies to help patients understand risks. *British Medical Journal,* 327(7417), 745.

Pauly, B., MacKinnon, K., & Varcoe, C. (2009). Revisiting "Who gets care?": Health equity as an arena for nursing action. *Advances in Nursing Science,* 32(2), 118.

Pauly, B. (2008). Shifting moral values to enhance access to health care: Harm reduction as a context for ethical nursing practice. *International Journal of Drug Policy*. 19(3),195.

Personalized Medicine Coalition. *The Case for Personalized Medicine*, Accessed July 30, 2009 from: http://www.personalizedmedicinecoalition.org/communications/TheCaseforPersonalizedMedicine_5_5_09.pdf.

Peter, F. (2001). Health equity and social justice. *Journal of Applied Philosophy,* 18(2), 159.

Peter, F. (2004). Health equity and social justice. *Public Health, Ethics, and Equity*, 93.

Peter, F., Evans, T., Whitehead, M., Diderichsen, F., Bhuiya, A., & Wirth, M. (Eds.). (2001). *Challenging Inequities In Health: From Ethics To Action*. New York, NY: Oxford University Press.

Pong, R., DesMeules, M., Heng, D. *et al.* (2011). Patterns of health service utilisation in rural canada. *Chronic Diseases and Injuries Canada.* 31(s1), 1.

Powers, M., & Faden, R. (2000). Inequalities in health, inequalities in healthcare: four generations of discussion about justice and cost-effectiveness analysis. *Kennedy Institute of Ethics Journal,* 10(2), 109.

Provincial Advisory Committee on New Genetic Technologies. (2001). *Genetic Services In Ontario: Mapping The Future; Working Together For A Healthier Ontario*. Accessed May 4, 2011 from: http://www.health.gov.on.ca/english/public/pub/ministry_reports/geneticsrep01/genetic_report.pdf

Rae, D., & Simpson, S.A. (2012). A standard of care for Huntington's disease: who, what and why. *Neurodegenerative Disease Management,* 2(1), 1.

Rawls, J. (1971). *A Theory of Justice*. Cambridge, MA: Harvard University Press.

Richardson, L.K., Christopher Frueh, B., Grubaugh, A.L., Egede, L., & Elhai, J.D. (2009). Current directions in videoconferencing tele-mental health research. *Clinical Psychology: Science and Practice,* 16(3), 323.

Rosenblatt, A., Liang, K.Y., Zhou, H., Abbott, M., Gourley, L., Margolis, R., *et al*. (2006). The association of CAG repeat length with clinical progression in Huntington disease. *Neurology,* 66(7), 1016.

Ross, C., Katzov, H., Carleton, B., & Hayden, M. (2007). Pharmacogenomics and its implications for autoimmune disease. *Journal of Autoimmunity, 28*(2-3), 122.

Rourke, J. (2008) Increasing the number of rural physicians. *Canadian Medical Association Journal.* 178(3), 322.

Rozmovits, L., & Ziebland, S. (2004). What do patients with prostate or breast cancer want from an Internet site? A qualitative study of information needs. *Patient Education and Counseling,* 53(1), 57.

Ruger, J.P. (2004). Health and social justice. *The Lancet,* 364(9439), 1075.

Sassi, F., Luft, H.S., & Guadagnoli, E. (2006). Reducing racial/ethnic disparities in female breast cancer: screening rates and stage at diagnosis. *American Journal of Public Health,* 96(12), 2165.

Scheuner, M., & Rotter, J. (2006). Quantifying the health benefits of genetic tests: a clinical perspective. *Genetics in Medicine,* 8(3), 141.

Segal, J.Z. (2008). Breast cancer narratives as public rhetoric: genre itself and the maintenance of ignorance. *Linguistics and the Human Sciences,* 3(1), 3.

Sen, A. (1993). Capability and Well-Being. In M. Nussbaum & A. Sen (eds.) *The Quality of Life.* Oxford, UK: Clarendon Press.

Sen, A. (2002). Why health equity? *Health Economics,* 11(8), 659.

Sheinfeld Gorin, S., & Heck, J.E. (2005). Cancer screening among Latino subgroups in the United States. *Preventive Medicine,* 40(5), 515.

Sherwin, S. (1988). A relational approach to autonomy in health care. In S. Sherwin (ed.), *The Politics of Women's Health: Exploring Agency and Autonomy* (p 19-47). Philadelphia, PA: Temple University Press.

Sherwin, S. (1992). *No Longer Patient: Feminist Ethics And Health Care*. Philadelphia, PA: Temple University Press.

Sherwin, S. (1996). in *Philosophical Perspectives on Bioethics* (eds. L.W. Sumner & J. Boyle) (p187-209). Toronto: University of Toronto Press.

Sherwin, S. (2000) A relational approach to autonomy in health care. *Readings In Health Care Ethics*. Peterborough, ON:Broadview Press.

Singer, P.A., Martin, D.K., Giacomini, M., & Purdy, L. (2000). Priority setting for new technologies in medicine: qualitative case study. *British Medical Journal,* 321(7272), 1316.

Smith, G., Ebrahim, S., Lewis, S., Hansell, A., Palmer, L., & Burton, P. (2005). Genetic epidemiology and public health: hope, hype, and future prospects. *The Lancet,* 366(9495), 1484.

Smith, J.A., Michie, S., Stephenson, M., & Quarrell, O. (2002). Risk perception and decision-making processes in candidates for genetic testing for Huntington's disease: an interpretative phenomenological analysis. *Journal of Health Psychology,* 7(2), 131.

Stalker, H., Wilson, R., McCune, H., Gonzalez, J., Moffett, M., & Zori, R. (2006). Telegenetic medicine: improved access to services in an underserved area. *Journal of Telemedicine and Telecare,* 1*2*(4), 182.

Statistics Canada. (2010). Accessed February 12, 2010, from http://www.statcan.gc.ca/start-debut-eng.html.

Starfield, B. (2006). State of the art in research on equity in health. *Journal of Health Politics, Policy & Law,* 31(1), 11.

Starks, H., & Brown Trinidad, S. (2007). Choose your method: A comparison of phenomenology, discourse analysis, and grounded theory. *Qualitative Health Research,* 17(10), 1372.

Stout, P.A., Villegas, J., & Kim, H. (2001). Enhancing learning through use of interactive tools on health-related websites. *Health Education Research,* 16(6), 721.

Strauss, A., & Corbin, J. (1998). *Basics of Qualitative Research* (2nd ed.), Thousand Oaks, CA: Sage Publications.

Sutherns, R., & Bourgeault, I. (2008). Accessing maternity care in rural Canada: There's more to the story than distance to a doctor. *Healthcare for Women International,* 29(8), 863.

Tabrizi, S., Langbehn, D., Leavitt, B., Roos, R., Durr, A., Craufurd, D., *et al*. (2009). Biological and clinical manifestations of Huntington's disease in the longitudinal TRACK-HD study: cross-sectional analysis of baseline data. *The Lancet Neurology,* 8(9), 791.

Takahashi, H., Wilkinson, G., Nutescu, E., Morita, T., Ritchie, M., Scordo, M., *et al*. (2006). Different contributions of polymorphisms in VKORC1 and CYP2C9 to intra-and inter-population differences in maintenance dose of warfarin in Japanese, Caucasians and African-Americans. *Pharmacogenetics and Genomics,* 16(2), 101.

Tassicker, R.J., Teltscher, B., Trembath, M.K., Collins, V., Sheffield, L.J., Chiu, E., *et al*. (2008). Problems assessing uptake of Huntington disease predictive testing and a proposed solution. *European Journal of Human Genetics,* 17(1), 66.

Taylor, S.D. (2004). Predictive genetic test decisions for Huntington's disease: Context, appraisal and new moral imperatives. *Social Science & Medicine,* 58(1), 137.

Tibben, A. (2007). Predictive testing for Huntington's disease. *Brain Research Bulletin,* 72(2-3), 165.

Tibben, A., Timman, R., Bannink, E.C., & Duivenvoorden, H.J. (1997). Three-year follow-up after presymptomatic testing for Huntington's disease in tested individuals and partners. *Health Psychology; Health Psychology,* 16(1), 20.

Touchette, N., Holtzman, N.A., Davis, J.G., & Feetham, S. (1997). Toward the 21st century: incorporating genetics into primary healthcare. Cold Spring Harbor, NY: Cold Spring Harbor Laboratory Press.

Trees, A.R., Koenig Kellas, J., & Roche, M.I. (2010). Family Narratives. In C. L. Gaff & C. L. Bylund (Eds.), *Family Communication About Genetics: Theory and Practice*. New York, NY: Oxford University Press.

UN Department of Economic and Social Affairs Population Division, (2005). *World Urbanization Prospects*, Accessed November 30, 2009 from: http://www.un.org/esa/population/publications/WUP2005/2005wup.htm.

UN Statistical Division, (2012). *Population Density and Urbanization*, Accessed July 31, 2012 from: http://unstats.un.org/unsd/demographic/sconcerns/densurb/densurbmethods.htm

Van der Steenstraten, I., Tibben, A., Roos, R., Van de Kamp, J., & Niermeijer, M. F. (1994). Predictive testing for Huntington disease: nonparticipants compared with participants in the Dutch program. *American Journal of Human Genetics,* 55(4), 618.

Veatch, R. (1981). *A Theory of Medical Ethics*. New York, NY: Oxford University Press.

Wadelius, M., & Pirmohamed, M. (2006). Pharmacogenetics of warfarin: current status and future challenges. *The Pharmacogenomics Journal,* 7(2), 99.

Walker, F. (2007). Huntington's disease. *The Lancet,* 369(9557), 218.

Wallace, S.P., & Villa, V.M., (2003). Equitable health systems: cultural and structural issues for Latino elders. *American Journal of Law & Medicine* 29(2-3): 247.

Wang, C., Gonzalez, R., Milliron, K. J., Strecher, V. J., & Merajver, S. D. (2005). Genetic counseling for BRCA1/2: a randomized controlled trial of two strategies to facilitate the education and counseling process. *American Journal of Medical Genetics Part A,* 134(1), 66.

Washington State Department. of Health, (2008). Genetic Services Policy Project Final Report. Accessed June 10, 2010 from: http://depts.washington.edu/genpol/docs/AppD.pdf.

Watt, I., Franks, A., & Sheldon, T. (1994). Health and healthcare of rural populations in the UK: is it better or worse? *British Medical Journal,* 48(1), 16.

Wellcome Trust Case Control Consortium, (2007). Genome-wide association study of 14000 cases of seven common diseases and 3000 shared controls. *Nature* 447(7145), 661.

Went, L. (1994). Guidelines for the molecular genetics predictive test in Huntington's disease. *Journal of Medical Genetics,* 31(7), 555.

Wexler, N. S. (2004). Venezuelan kindreds reveal that genetic and environmental factors modulate Huntington's disease age of onset. *Proceedings of the National Academy of Sciences of the United States of America,* 101(10), 3498.

Whitehead, M., & Dahlgren, G. (2007). Concepts and principles for tackling social inequities in health: Levelling up Part 1. *Copenhagen: World Health Organisation.* Accessed July 31, 2012 from: http://www.enothe.eu/cop/docs/concepts_and_principles.pdf

WHO. (2007). *The Top Ten Leading Causes of Death*. Accessed Feb. 15, 2010 from http://www.who.int/mediacentre/factsheets/fs310.pdf.

WHO. (2010). *Genomic Resource Centre.* Accessed Feb 15, 2010, from: http://www.who.int/genomics/public/access/en/index.html.

WHO. (2012). *World Health Statistics*. Accessed June 3, 2012 from: http://www.who.int/healthinfo/EN_WHS2012_Full.pdf.

Wiggins, S., Whyte, P., Huggins, M., Adam, S., Theilmann, J., Bloch, M., *et al*. (1992). The psychological consequences of predictive testing for Huntington's disease. *New England Journal Of Medicine,* 327(20), 1401.

Wilkinson, R. (1997). Socioeconomic determinants of health: Health inequalities: relative or absolute material standards? *British Medical Journal*, 314(7080), 591.

Williams-Jones, B., & Burgess, M. (2004). Social Contract Theory and Just Decision Making. *Kennedy Institute of Ethics Journal*, 14(2), 115.

Williams-Jones, B., & Burgess, M. (2006). Democratising Access to Genetic Services. *Familial Cancer*, 5(2), 117.

Williams, D., & Collins, C. (1995). US socioeconomic and racial differences in health: patterns and explanations. *Annual Review of Sociology*, 21(1), 349.

Williams, J.K., Schutte, D.L., Holkup, P.A., Evers, C., & Muilenburg, A. (2000). Psychosocial impact of predictive testing for Huntington disease on support persons. *American Journal of Medical Genetics*, 96(3), 353.

Winterbottom, A., Bekker, H.L., Conner, M., & Mooney, A. (2008). Does narrative information bias individual's decision making? A systematic review. *Social Science & Medicine*, 67(12), 2079.

Woolf, S. (2008). The meaning of translational research and why it matters. *Journal of the American Medical Association*, 299(2), 211.

Wootton, R., & Bonnardot, L. (2010). In what circumstances is telemedicine appropriate in the developing world? *Journal of the Royal Society of Medicine ShortRreports*, 1(5), 37.

Young, I. (1990). *Justice and the Politics of Difference.* Princeton, NJ: Princeton University Press.

Young, I. (2002). Equality of whom? Social groups and judgments of injustice. *Journal of Political Philosophy*, 9(1), 1.

Zhang, P., Tao, G., & Anderson, L. (2008). Differences in access to healthcare services among adults in rural America by rural classification categories and age. *Australian Journal of Rural Health*, 11(2), 64.

Zhang, P., Too, G., & Irwin, K. (2000). Utilization of preventive medical services in the United States: a comparison between rural and urban populations. *The Journal of Rural Health*, 16(4), 349.

Ziebland, S., & Herxheimer, A. (2008). How patients' experiences contribute to decision making: illustrations from DIPEx (personal experiences of health and illness). *Journal of Nursing Management*, 16(4), 433.

APPENDICES

Appendix I: Interview guides

Interview guide A: Individuals who have undergone testing (rural and non rural)

1. Have you undergone predictive testing for HD?
2. When did you undergo testing for HD?
3. Where were you living at the time you underwent testing for HD?
4. Why did you decide to undergo predictive testing for HD?
    a. PROMPT: why that specific time? How long had you known you are at risk?
5. Please walk me through what you can remember of the HD predictive testing process
    a. PROMPT: who you spoke to, how long it took to get results, who gave you the results, how many visits/ conversations/ phone calls you had
6. What do you think were the most important/ necessary/ helpful aspects of the testing process?
    a. PROMPT: why?
7. What do you think were the unnecessary aspects of the testing process?
    a. PROMPT: why?
8. Did you feel you received adequate information regarding the advantages and disadvantages of HD predictive testing prior to undergoing testing?
9. Did you have any follow-up discussions after receiving your test result?
    a. PROMPT: Were these research related or clinical?
10. Did you feel you received adequate support during the testing process?
    a. PROMPT: prior to and after results
11. What was your overall experience of the predictive testing process (prior to and after)?
    a. PROMPT: negative/ disadvantages
    b. PROMPT: positive aspects/ advantages
12. Do you have any suggestions as to how to improve the testing process?
13. Are you glad you underwent predictive testing for HD?
    a. PROMPT: Why/ why not?
14. What are your overall feelings about the testing process and your result?

15. Are you involved in research regarding HD?
    a. PROMPT: type/ how long/ contact with researchers

Interview guide B: Individuals who have not undergone testing

1. When did you first learn you were at risk for HD?
2. Have you ever considered undergoing predictive testing (provide definition as necessary) for HD
    a. PROMPT: why/ why not? what have you heard about it?
3. What are the main reasons you decided not to undergo testing for HD?
4. Do you know anyone who has undergone testing for HD?
    a. PROMPT: what is your understanding of why they decided to undergo predictive testing
5. What is your understanding of what is involved with the HD testing process?
    a. PROMPT: is there any aspect of the testing process which you think would be particularly difficult for you?
6. From your knowledge of the HD predictive testing process, what do you think could be done to improve the process?
7. What do you think are the most important components of the HD predictive testing process?
    a. PROMPT: Why?

Appendix II: Website pilot test survey

1. Is the website clearly laid out and easy to navigate (find your way around)?

   ☐ Yes

   ☐ No

   If not, which sections are unclear?_____

   _____

   _____

   _____

2. Is the content of the website clear and easy to understand?

   ☐ Yes

   ☐ No

   If not, which sections are unclear?_____

   _____

   _____

   _____

3. Are the illustrations and diagrams helpful?

   ☐ Yes

   ☐ No

   If not, what are your suggestions for improvement?_____

   _____

   _____

4. Does the website provide adequate information on ‘what is HD’?

   ☐ Yes

   ☐ No

   If not, what are your suggestions for improvement?_____

5. Does the website provide adequate information on ‘testing for HD’?

   ☐ Yes

   ☐ No

   If not, what are your suggestions for improvement?_____

6. Does the website provide enough information on the pros and cons of testing and making the decision to be tested?

   ☐ Yes

   ☐ No

   If not, what are your suggestions for improvement?_____

   _____

   _____

   _____

   _____

7. Any other comments and suggestions?_____

   _____

   _____

   _____

   _____

8. Please check which box best describes you:

   ☐ I participated in the interview study

   ☐ I am a member of the Hayden Lab

   ☐ I am a genetic counselor

   ☐ I am a family member or friend with limited knowledge of HD

   ☐ I prefer to remain anonymous

Appendix III: Expert workshop report

EXPERT WORKSHOP:

Improving access to HD predictive testing

November 12TH, 2010

Introduction

The 'Improving Access to HD Predictive Testing' Expert Workshop was held in Vancouver, Canada, on November 12th, 2010. The workshop attendees were invited to attend based on their expertise and knowledge of providing predictive testing for Huntington Disease (HD) and/or novel telemedicine and telegenetics techniques. The broad aim of the workshop was to foster discussion and elicit expert advice regarding the provision of predictive testing for HD offsite, including remote and rural areas.

Specifically, the workshop goals were as follows:

- To assess experts' perspectives on the key components of the predictive testing process
- To gather suggestions and recommendations on how to improve access to predictive testing for HD
- To propose and get feedback on a provisional education and testing protocol for providing HD predictive testing remotely
- To understand and consider mechanisms to address potential concerns related to a modification of the standard predictive testing protocol for HD

Ultimately the workshop sought to provide a forum for experts' concerns, insights and knowledge to be voiced and integrated into ongoing research aimed at improving the lives of those affected with this devastating disorder.

Attendees

Experts were invited on the basis of their ability to provide broad insights, experiences and recommendations in one of two broad areas:

1. The provision of predictive testing for HD
2. Novel telemedicine and telegenetics tools and strategies

In order to ensure a variety of perspectives were considered and achieve diverse discussion, workshop participants were invited from a number of different countries, practice areas and backgrounds, as detailed in table III.1.

Table III.1: List of expert workshop participants:

Name	Role	Affiliations
Michael Bidu, MBA	President, Digi BC	The Digital Media and Wireless Association of BC, Vancouver
Robi Blumenstein, LLB, MBA	President, CHDI	CHDI, New York
Jeff Carroll*, PhD	Post Doctoral Fellow and HD advocate	Boston University, Massachusetts
David Craufurd, MB, BS, MSc, FRCPsych	Consultant and Senior Lecturer in Neuropsychiatric Genetics	University of Manchester, UK
Mark Guttman*, MD	Director of the Centre for Movement Disorders; Board Member, Ontario Telehealth Network; and Assistant Professor in Neurology and Psychiatry	Centre for Movement Disorders, Markham, Ontario; Ontario Telehealth Network; Division of Neurology and Psychiatry, Department of Medicine, University of Toronto
Alice Hawkins, MA (Oxon), MS, MPH	PhD Candidate and Genetic Counselor	Centre for Molecular Medicine and Therapeutics and Centre for Applied Ethics, University of British Columbia, Vancouver
Michael Hayden, MB, ChB, PhD, FRCP (C), FRSC	Canada Research Chair in Human Genetics and Molecular Medicine University Killam Professor, Department of Medical Genetics	Centre for Molecular Medicine and Therapeutics; Child and Family Research Institute, University of British Columbia, Vancouver
Kendall Ho, MD	Associate Professor, Emergency Medicine and Director, eHealth Strategy Office	University of British Columbia, Vancouver
Richard Kohl, PhD	Strategy Consultant	Learning and Leading for Large Scale Change LLC, Portland
Martha Nance, MD	Director of the HDSA Center of Excellence Clinic at HCMC; Neurologist and Geneticist	Hennepin County Medical Center and Park Nicolette Methodist Hospital, Minnesota
Thoren Young	Patient advocate	None
Tyler Willman	Website Developer	B'stro, Vancouver

Outline of events

The full day workshop included a number of diverse presentations by workshop attendees, followed by group discussion and comments, as detailed in the agenda below. Four topic areas formed the framework for the day's presentations and discussions. The workshop was chaired by Dr. Michael Hayden.

Thursday, November 11, 2010	
All day	Out of town participants arrive and check in to the Sutton Place Hotel
Friday, November 12, 2010	
8:00 am to 8:30 am	Continental Breakfast in the Cezanne Board Room, Sutton Place Hotel
8:30 am to 9:00 am	**Welcome, Introductions, Outline and goals for the day: MRH**
9:00 am to 9:20 am	**TOPIC I: ACCESS TO PREDICTIVE TESTING** **Presentation: Alice Hawkins** *Project Outline and Rationale* - Access to predictive testing - Justice/ ethical arguments - *Predictive testing in BC* - Summary data and map - Interview/ survey results
9:20 to 10:30 am	**Group discussion:** - Barriers to predictive testing - Strategies/ testing process in other areas
10:30 am to 10:45 am	Break
10:45 am to 11:00 am	**TOPIC II: eHEALTH STRATEGIES & SCALING UP IN BC** **Presentation: Richard Kohl** *Strategies for 'scaling up'*
11:00 am to 11:15 am	**Presentation: Michael Bidu** *Digital media and wireless solutions to telehealth*

11:15 am to 11:30am	**Presentation: Kendall Ho** *Providing telehealth in BC*
11:30 am to 12:45 pm	**Group Discussion:** - Successful telehealth strategies - Telehealth concerns
12:45 pm to 1:30 pm	Lunch
1:30 pm to 1:50 pm	**TOPIC III: COMMUNICATION & INFORMATION** **Presentation: Alice Hawkins and B'stro** *Website Outline and Overview (rationale, design, wireframe, initial content)*
1:50pm to 3:00pm	**Group discussion:** - Feedback on the website/ concerns - Strategies to implement and test website
3:00 pm to 3:15 pm	Break
3:15 pm to 4:30 pm	**TOPIC IV: TRAINING & EDUCATION** **Group discussion** - How to involve and train rural healthcare providers - Neurological exam and psychiatric assessment - Reimbursement
4:30pm to 5:15pm	**Wrap up/ summary/ next steps: MRH**
6:30 pm to 9:00 pm	Dinner at ORU
Saturday, November 13, 2010	
Morning	Out of town participants depart

Workshop outcomes

Discussions during the workshop were recorded via note taking. Recommendations reached and important considerations discussed, are summarized below:

Topic I: Access to predictive testing

1. Barriers to predictive testing were discussed and thought to include:

- Delivery of testing and physical barriers (e.g., travel costs, time off work, leaving family support etc)
- Local healthcare provider knowledge (does not have accurate information about HD, testing etc)
- At-risk population knowledge (regarding testing, testing process and risk)
- Anonymity and confidentiality
- Other psychological barriers (e.g., lack of treatment)

Improving access to predictive testing involves addressing one or more of these barriers

2. Counseling: Participants of the workshop felt that the word and need for 'counseling' may itself be a deterrent to testing.

3. Gender differences in access to testing: It was suggested that the current testing process, including in-person counseling, may, in part explain higher testing rates in women than men, as women are more amenable to undergoing counseling. Providing testing offsite may thus lead to an increase in male participation in predictive testing.

4. Anonymity and confidentiality: participants reported that some individuals who requested testing were concerned about anonymity and confidentiality, illustrated by reports of individuals requesting testing in different countries and under pseudonyms. It was also suggested that some individuals may not participate in testing as they do not want to go to the HD clinic, where they might be seen by a family member also attending clinic, or where the clinic team may let something 'slip' to relatives that testing had taken place.

   Anonymity and confidentiality is also an issue in rural areas, where family physicians may be known socially, and where test result or at risk status knowledge may slip, inadvertently or purposefully, to other family members, employers or other community members. It was suggested that the ability to provide testing offsite, through a chosen healthcare professional, may diminish the issue of anonymity and confidentiality resulting in increased uptake of predictive testing both in remote and urban settings.

5. Neurological exam: the neurological exam as a component of the predictive testing process was discussed in some detail. Many predictive testing sites are no longer performing the exam as part of the predictive testing process, and will only offer it if an individual receives a positive test result. Neurological exam was also thought to be a costly and time consuming part of the testing process, which some patients found frustrating. The workshop participants felt that neurological exam is not indicated for every individual undergoing predictive testing.

Topic II: eHealth strategies & scaling up in BC

1. Successful electronic/ internet models are user-centered can be grouped into 3 categories, each of which help to build trust in a website:
   a) Content rich, so people become engaged in the content of the site
   b) 'People like me', so people feel personal connection to a site
   c) Recommendation through peers (may also be done through social networking)

   Giving people the option to stay in touch with people, through a newsletter, social networking and so on also helps people feel connected to a website. The extent to which a website is interactive is also important; interactive components may include podcasts, video clips, blogging and so on.

2. The 'stages of change' model was presented and discussed as a method to bridge the knowledge – action gap. Stages of change: i) pre-contemplative, ii) contemplative, iii) preparation, iv) action, v) maintenance of change. Technology can assist in motivating and maintaining change (in this case change of the standard predictive testing model).

3. A 'hub' model may help in 'scaling-up' the offsite predictive testing model. For example, Vancouver could act as a hub for Canada in providing the offsite testing, training and support to other Provinces.

4. When piloting the offsite testing protocol it will be important to try and understand when, and why, some individuals postpone or stop the testing process. This will help us better understand the needs and concerns of at-risk individuals as they go through testing.

5. Privacy of results was also discussed, and given careful consideration in terms of how to preserve privacy of the result. The local healthcare provider should receive results only shortly prior to seeing the patient. A variety of mechanisms to ensure this was done appropriately were discussed (e.g., a two-way 'key' that both the local healthcare provider and at-risk individual had to access prior to release of results)

6. Sources of potential criticism of a modified predictive testing process that is provided offsite were identified:
   - No improved reach or increase in access to predictive testing
   - Serious adverse event of an individual who goes through the offsite testing process
   - Novel offsite predictive testing model is perceived as 'pushing' testing

7. Evaluation of the offsite testing method needs to include measurements of both positive and negative outcome measures. Such measurements include:
   - Satisfaction surveys and questionnaires after each stage of the offsite predictive testing process
   - Measuring adverse events (e.g., follow-up surveys and BDI)
   - Pre and post tests of the website to understand the effectiveness of the website as an educational tool (see Topic III:4, below)

- Predictive testing rates
- Cost-effectiveness
- Usability of system

Topic III: Communication & information

1) Translation: The website will need to be translated into other languages when it is expanded to other regions.

2) Population diversity and access to the internet needs assessments need to be conducted in the future in order to ensure the offsite testing will reach a significant proportion of those in need.

   Most of BC has internet coverage (may not be wireless)

3) Website accessibility and marketing also need to be considered. For example, the website should be easily found through internet search engines, and can also be advertised via regional HD newsletters. Links to other HD sites will also help. Political and policy considerations also need to be taken into account.

4) Evaluation of knowledge transfer and training tools is important to understanding the success of the website and telehealth sessions. Pre and post test measures were discussed as a potential mechanism to do this.

5) Avoid duplication: While a website dedicated to predictive testing is important, it should not attempt to recreate information already available on existing sites.

Topic IV: Training & education

1) Individuals undergoing offsite testing still need to see a local healthcare to support them through the testing process, provide results, and ensure that the individual receives appropriate care and follow-up during and after testing. The individual undergoing testing should have some discretion over who this support individual should be, however this individual should be a trained healthcare professional of some sort (such as a family physician, nurse, social worker). Ensuring this individual is a qualified healthcare professional will help satisfy concerns about integrity and adequacy of this support person.

2) Training of this individual, via internet education and support from the HD testing centre is essential. This individual needs to know the basics of HD testing, psychosocial factors, importance of confidentiality, as well as being able to identify the need for further referral as necessary (e.g., for psychiatric evaluation).

3) Payment for this healthcare support person will likely be determined by standard billing practices for that region.

Conclusion

At the conclusion of the expert workshop each of the attendees agreed that an offsite predictive testing protocol for providing HD predictive testing did seem a reasonable mechanism to address access issues to HD predictive testing. In order to be success, and maintain appropriate quality of care and support for individuals undergoing testing, a variety of safeguards, training and educational tools were discussed. In addition, a pilot of the offsite testing tool was recommended, and a variety of assessment mechanisms to measure to success, satisfaction and safety of the novel testing tool were suggested. If the pilot is successful, the offsite testing model may be used and adapted for other areas of the world in order to improve access to predictive testing and services worldwide.

Each individual who attended was asked to consider being an advisory board member for the offsite predictive testing project.

Appendix IV: British Columbia Huntington disease predictive testing survey

Dear Friend of the Huntington Society of Canada, BC Chapter,

RE: Invitation to participate in a study regarding a novel predictive testing process for Huntington disease (HD) in your home community.

You are receiving this letter because you have previously expressed an interest in receiving information from the Huntington Society of Canada (HSC) or have participated in HSC activities in the past. The HSC is assisting in the mailing and communication about this study and is not involved in conducting the study or anyway promoting the actual research project.

Please note that your privacy was protected and your address was not shared with the BC Study team. HSC forwarded this information to you on behalf of the BC Study team to ensure confidentiality.

We are sending you this letter to inform you about a research study which looks at a new, offsite predictive testing process for HD. Under this new testing process, all appointments involved in HD predictive testing are done in your local community via telehealth (video-conferencing) with HD clinic at UBC in Vancouver and with the support of your local healthcare provider. This study is being conducted by the Centre for Huntington Disease at the University of British Columbia. Further information on this study and what it involves is contained on the back of this letter.

In addition, we have included a brief survey regarding predictive testing in BC to help us provide better services. This survey is completely anonymous. *If you do agree to participate in this survey please complete and return the completed survey in the enclosed pre-paid envelope.* **By returning this questionnaire you are consenting to participate in this research study.**

Thank you for your consideration.

Michael R. Hayden MB, ChB, PhD, FRCP (C), FRSC
Canada Research Chair in Human Genetics & Molecular Medicine
University Killam Professor, Department of Medical Genetics

Alice Hawkins, MA, MS, MPH
PhD Candidate, UBC

Please assist us to improve our predictive testing service in BC please answer the following questions and return in the envelope provided.

This survey is anonymous

By returning this questionnaire you are consenting to participate in this research study

1. Have you undergone predictive testing for HD?
   - ☐ Yes *(proceed to question 4)*
   - ☐ No *(proceed to question 2)*
   - ☐ Don't know *(proceed to question 2)*

2. Are you interested in undergoing predictive testing for HD?
   - ☐ Yes ☐ No ☐ Don't know

3. Are you more likely to undergo predictive testing for HD now that is available in your local community via telehealth?
   - ☐ Yes ☐ No ☐ Don't know

4. Did/has the **distance** to the predictive testing centre in Vancouver/ Victoria prevented you from undergoing predictive testing in the past?
   - ☐ Yes ☐ No ☐ Don't know

5. Did/has the **travel time** to the predictive testing centre in Vancouver/ Victoria prevented you from undergoing predictive testing in the past?
   - ☐ Yes ☐ No ☐ Don't know

6. Did/has the **financial cost** of going to the predictive testing centre in Vancouver/ Victoria prevented you from undergoing predictive testing in the past?
   - ☐ Yes ☐ No ☐ Don't know

7. Did/has the **stress** of going to the predictive testing centre in Vancouver/ Victoria prevented you from undergoing predictive testing in the past?

   ☐ Yes ☐ No ☐ Don't know

8. Do you think providing predictive testing for HD in your local community via telehealth is a good idea?

   ☐ Yes ☐ No ☐ Don't know

9. If you could choose the **ideal** method of undergoing predictive testing for HD what would it be?

   ☐ In my local community, via telehealth, with the support of local healthcare provider

   ☐ At the Vancouver/ Victoria predictive testing centre

   ☐ Other:_____

10. Please indicate the approximate driving time (one way) from where you live to your **nearest** predictive testing site:

☐ Less than 2 hours ☐ 2-4 hours ☐ 4-8 hours ☐ More than 8 hours

11. Any comments?_____

_____

_____

_____

Appendix V: Pilot test surveys

Vancouver tested survey

Survey regarding the predictive testing protocol for Huntington disease

Your participation in this survey is completely voluntary and your answers will be kept confidential

HOW TO COMPLETE THIS QUESTIONNAIRE

- Most questions require that you simply TICK the box (☐) indicating the appropriate answer(s)
- There are no right or wrong answers; we are interested in your personal opinions and experiences
- Some questions may appear repetitive, but please note that there are some important and subtle differences
- If you need more space when answering a question, please use additional pages at the back of the questionnaire
- If you have any questions or would like to talk to any of us about your opinions and experiences, please call us at: 6048752000 x 4638

By returning this questionnaire you are consenting to participate in this research study.

Section 1: Respondent details

This section contains questions about you in order to describe the people that have participated in this study.
Please check or fill in the answer that applies

1.1 Are you...? ☐ Male ☐ Female

1.2 What is your marital status? (please check ONE)

- ☐ Never married (single)
- ☐ Married and living with spouse
- ☐ Separated
- ☐ Divorced
- ☐ Widowed
- ☐ Common law/ live in partner
- ☐ Other (please specify

1.3 Do you have children? (please check ONE) ☐ Yes ☐ No

1.4 People living in Canada come from many different cultural and racial backgrounds. To which ethnic or cultural group do you belong? (Please check ONE)

- ☐ White or European descent
- ☐ Aboriginal (North American Indian, Metis, or Inuit)
- ☐ Black or African American
- ☐ Latin American
- ☐ Chinese
- ☐ Japanese
- ☐ Korean
- ☐ South Asian (i.e., East Indian, Pakistani, Sri Lankan)
- ☐ South East Asian (i.e., Cambodian, Indonesian, Laotian, Vietnamese)

- ☐ West Asian (i.e., Afghan, Iranian)
- ☐ Filipino
- ☐ Arab
- ☐ Don't know
- ☐ Other (please specify)_____

1.5 What is the highest grade or level of education you have attained? (Please check ONE)

- ☐ No schooling
- ☐ Some elementary
- ☐ Completed elementary
- ☐ Some secondary
- ☐ Completed secondary
- ☐ Some community college, technical college, CEGEP or Nursing training
- ☐ Completed community college, technical college, CEGEP or Nursing training
- ☐ Some university or teacher's college
- ☐ Completed university or teacher's college
- ☐ Other (please specify)_____

_____

1.6 What is your current employment status (please check ONE)

- ☐ Full-time
- ☐ Part time
- ☐ Unemployed and seeking work
- ☐ Unemployed or retired and not seeking work

1.7 Please describe your occupation_____

1.8 What is your best estimate for your total household income in the last 12 months before taxes and deductions? Please tick ONE

- ☐ Less than $15,000
- ☐ $15,000 – 29,999
- ☐ $30,000 – 44,999
- ☐ $45,000 - 59,999
- ☐ $60,000 – 74,999
- ☐ $75,000 – 89,999
- ☐ $90,000 – 104,999
- ☐ Over $105,000
- ☐ Don't know
- ☐ Other (please specify)_____

1.9 Which of the following best describes your experience with HD (Please tick ONE)

- ☐ I have had no prior experience with people who had HD
- ☐ I have known people who have had early symptoms of HD
- ☐ I have known people who have had severe disease or have died

Section 2: Pre-test period and testing process

The section refers to all the appointments, conversations and visits you had regarding predictive testing for HD prior to receiving your results. This will help us understand your experience of the testing process, as well as what you found beneficial/ helpful and difficult/ unnecessary.

2.1 What was format of your pre-test counseling appointment(s) (please check all that apply)

- ☐ Over the phone with the genetic counsellor (Susan Creighton) at the HD clinic at UBC
- ☐ In person with the genetic counsellor (Susan Creighton) at the HD clinic at UBC

2.2 What was the entire time it took to attend the pre-test counseling appointment (s) (including travel and appointment time)?[30]

☐ 1-4 hours ☐ 4-6 hours ☐ 6-10 hours ☐ 10-24 hours ☐ Other:_____

Section A: Reasons for undergoing testing for HD

2.3 What were your reasons for undergoing HD testing (please rate how strongly you agree or disagree with EACH of the following statements by placing a tick in the appropriate box)

Reason	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. Knowing is better than not knowing, even if it can be stressful					
b. Knowing can help you understand the role of genetics in your or your family's lives					
c. Knowing the result can give you a greater sense of personal control and					

[30] Note exact time in hours to attend appointment (rather than range) was clarified with each respondent subsequent to returning the survey. This was done so that statistical analysis of the difference between the groups could be established.

certainty					
d. Knowing can help you plan your future					
e. Knowing can help you make choices about having children					
f. Knowing can help in your communication and relationships with your family					
g. Knowing can help in your communication and relationships with others					
h. Knowing can give you the opportunity to take preventive actions to control or monitor the development of symptoms					
i. Knowing can help you share the family history with others at risk in your family					
j. Knowing can give you a reliable scientific basis to use in explaining your or your family's situation to others					
k. Knowing can allow you to support and/or be supported by others					

l. Other (please specify)_____

_____

2.4 What were your reservations about undergoing predictive testing for HD?

Reason	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I had concerns about coping with the results					
b. I had concerns about family members coping with the results					
c. I had concerns about the implications of					

the test results for family members					
d. I had concerns about privacy and confidentiality					
e. I had concerns about insurance					
f. I had concerns about the cost of testing					
g. I had concerns about employers/ employment					
h. I had concerns about how others would treat me if they knew I had a gene mutation					
i. There is no cure for Huntington disease					

2.5 Why did you decide to undergo testing at this time?

☐ I had recently (within 3 months) learnt about the condition in a family member and decided to undergo testing

☐ I had known about the condition for some time (more than 3 months) but decided to undergo testing for the reasons outlined above (in question 2.3)

Please provide details on your exact reasons:_____

_____

_____

_____

_____

☐ I had wanted to undergo testing for sometime but had been unable to attend the appointments at UBC (go to question 2.6)

☐ Other_____

_____

2.6 If you checked the third box in question 2.4 please rank the reasons you were unable to make the appointments (1 being the biggest reason)

- ☐ Could not take time away from work _____
- ☐ Did not want/ could not take time away from family _____
- ☐ Cost of travel to the HD clinic _____
- ☐ No available transportation to get to the HD clinic _____
- ☐ Other:_____

_____

_____

_____

2.7 Please complete the following table to indicate the amount of time you spent communicating with healthcare professionals prior to deciding to undergo predictive testing

	Amount of time
a. How much time **in total** did you spend communicating with the **Geneticist or Genetic Counsellor at UBC before** you decided to undergo testing?	hours
b. How much time **in total** did you spend communicating with your **family physician before** you decided to undergo testing?	hours

2.8 Before undergoing testing would you have preferred more appointments/conversations sessions to discuss HD testing with any of the above mentioned healthcare professionals?

- ☐ Yes
- ☐ No

2.9 Please read each of the following statements and put a check mark in the chart which best corresponds to your situation.

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. **Prior to my first contact** with the HD genetic testing centre or other healthcare provider I had a good knowledge of the possible advantages and disadvantages of undergoing predictive genetic testing for HD					
b. **After my appointment (s) or discussion (s)** with the HD genetic testing centre or other healthcare providers I had a good knowledge of the possible advantages and disadvantages of undergoing predictive genetic testing for HD					
c. My knowledge of the possible advantages and disadvantages of undergoing predictive genetic testing for HD significantly improved after my **after my appointment (s) or discussion (s)** with the HD genetic testing centre or other healthcare providers					
d. I was satisfied with the amount of information provided **before** I underwent testing					
e. I was able to understand all the information that I discussed with my healthcare professional (including the HD predictive testing centre) before I underwent HD testing					
f. I had enough time to discuss all of my concerns with my healthcare professional (including the HD predictive testing centre) before I underwent HD testing					
g. All of my questions were answered by my					

healthcare professional (including the HD predictive testing centre) before I underwent HD testing					
h. Overall, I was given enough information so that I felt able to make fully informed choice regarding HD predictive genetic testing					
i. Overall, I was given enough **support** during the pre-test consultation and counselling process (prior to undergoing HD predictive testing)					

2.10 What other information would you have liked to have received from your healthcare professional before you underwent HD testing?_____

_____

_____

_____

2.11 Overall, what do you think about the amount of time of the pre-testing consults and counselling visits (prior to undergoing HD predictive testing)?

a. ☐ Too much

b. ☐ Just right

c. ☐ Too little

Section 3. Results session

This section refers to how and when you received your HD predictive test results.

3.1 What was your predictive test result?

- ☐ Positive (I did inherit the genetic mutation for HD and will develop symptoms of HD during my lifetime)
- ☐ Negative (I did not inherit the genetic mutation for HD)
- ☐ Don't know
- ☐ Prefer not to mention

3.2 How were your genetic testing results delivered?

- ☐ In person by the genetic counsellor/ geneticist at UBC
- ☐ In person by my family physician
- ☐ In person by a social worker
- ☐ Over the phone by the genetic counsellor/ geneticist
- ☐ Over the phone by my family physician
- ☐ Over the phone by a social worker
- ☐ Mailed letter_____ from_____(please insert)
- ☐ Other_____

3.3 What do you believe would have been the best method of delivering your genetic testing results?

- ☐ In person by the genetic counsellor/ geneticist
- ☐ In person by my family physician
- ☐ In person by a social worker
- ☐ Over the phone by the genetic counsellor/ geneticist
- ☐ Over the phone by my family physician
- ☐ Over the phone by a social worker
- ☐ Mailed letter_____ from_____(please insert)
- ☐ Other_____

3.3 For each of the following questions please check the answer that best applies to your situation

Question	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I was satisfied with the manner in which my genetic test results were delivered to me					
b. I was able to understand all the information about my genetic test result that was discussed during the results session					
c. I had enough time to discuss all of my concerns about my genetic testing results with my healthcare professional during my genetic test result session					
d. All of my questions about my genetic testing results were answered by my healthcare professional					
e. Overall, I was given enough **information** regarding the meaning and implications **for me**					

of my genetic testing results by my healthcare professional					
f. Overall, I was given enough **information** regarding the meaning and implications **for my children** of my genetic testing results **for me** by my healthcare professional					
g. Overall, I was given enough **information** regarding the meaning and implications **for my extended family** of my genetic testing results **for me** by my healthcare professional					
h. Overall, I was given enough **support** (emotional & psychological) during the results session by my healthcare professional					

3.4 What other information would you have liked to receive about your genetic testing results by your healthcare professional?_____

_____

_____

3.5 What other support would you have liked to receive when you were given your genetic testing results?_____

_____

_____

3.6 Overall, what do you think about the amount of time of the results session with your healthcare professional? (please check)

a. ☐ Too much

b. ☐ Just right

c. ☐ Too little

Section 4. Follow-up since results

This section refers to the six month follow-up appointment from receiving your test result. This section is also about your overall satisfaction with the entire predictive testing process including the pre-test period, the results period, and the follow-up period.

For each of the following questions please check the answer that best applies to your situation

4.1 Have you undergone follow-up contact with the genetics team at UBC since receiving your genetic testing results?

☐ Yes ☐ No

4.2 When did this follow up contact with the genetics team at UBC take place?

☐ 2-4 weeks after I received my test results

☐ 1-2 months after I received my test results

☐ 2-4 months after I received my test results

☐ 4-6 months after I received my test results

☐ More than 6 months after I received my test results

4.3 Were you satisfied with the amount of time it took for the follow-up to take place?

☐ Too much ☐ Just right ☐ Too little

4.4 How did this follow-up contact with the genetics team take place?

☐ The genetics team called me on the phone

☐ The genetics team sent me an email

- ☐ I had an in person visit with the genetics team
- ☐ The genetics team left me a voicemail and I called them back
- ☐ The genetics team left me a voicemail and I did not call them back

If you did not call the genetics team back please explain why_____

_____

_____

_____

- ☐ I called the genetics team
- ☐ I called the genetics team but they did not call me back
- ☐ Other:_____

4.5 Please complete the following table regarding your follow-up contact with the genetics team by placing a check mark next to each statement indicating your level of agreement with the statement

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I was satisfied with the follow-up contact I received from the **genetics team**					
b. I was able to understand all that was discussed with the **genetics team** during my follow up contact					
c. I had enough time to discuss all of my concerns with the **genetics team** during my follow up contact					
d. All of my questions were answered by the **genetics team** during my follow up contact					
e. Overall, I was given enough **information** from the **genetics team** during the period following my genetic test results					

f. Overall, I was given enough **support** from the **genetics team** during the period following my genetic test results					

4.6 Overall, I think the follow-up contact from the genetics team during the period following my genetic test results was worthwhile:

☐ Yes ☐ No

4.7 Please provide any other comments regarding the post-test result period:_____

_____

_____

_____

_____

_____

_____

4.8 How much <u>benefit or advantage,</u> if any, have you felt from knowing about your genetic test result?

☐ Great benefit ☐ Some benefit ☐ Neutral ☐ Little benefit ☐ No benefit

4.9 Do you think knowing about your genetic test result has bought benefit because....(please rate how strongly you agree or disagree with EACH of the following statements by placing a tick in the appropriate box)

Question	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. Knowing is better than not knowing, even if it can be stressful					
b. Knowing has helped you understand the role of genetics in your or your family's lives					
c. Knowing the result has given you a greater sense of personal control and certainty					
d. Knowing has helped you plan your future					
e. Knowing has helped you make choices about having children					
f. Knowing has helped in your communication and relationships with your family					
g. Knowing has helped in your communication and relationships with others					
h. Knowing has given you the opportunity to take preventive actions to control or monitor the development of symptoms					
i. Knowing has helped you share the family history with others at risk in your family					
j. Knowing has given you a reliable scientific basis to use in explaining you or your family's situation to others					
k. Knowing has allowed you to support and/or be supported by others					

l. Other: _____

_____

_____

_____

4.10 Please rate how strongly you agree or disagree with EACH of the following statements by placing a tick in the appropriate box:

Statement	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. Overall, I was satisfied with the **support** provided during the entire testing process by healthcare professionals					
b. Overall I felt that testing process was sufficient in terms of the **information regarding** the predictive testing process provided during the entire testing process by healthcare professionals					
c. Overall I felt that the testing process was sufficient in terms of the information regarding the **meaning of my genetic test results for me** provided during the entire testing process by healthcare professionals					
d. Overall I felt that the testing process was sufficient in terms of the information regarding the **meaning of my genetic test results for my family** provided during the entire testing process by healthcare professionals					
e. Overall I would have preferred to have **more in person visits** at the HD testing centre during my predictive testing process					
f. Overall I would have preferred to have **more telephone/ telehealth appointments with** the HD testing centre during my predictive testing process					
g. Overall I would have preferred to have **less in person visits** with my family physician during my predictive testing process					
h. Overall I would have preferred to have **more in person visits** with my family physician during my predictive testing process					

4.11 Please provide any additional comments on your testing experience and suggestions for improvements in the space below:

_____

_____

_____

THANK YOU FOR TAKING THE TIME TO COMPLETE THE QUESTIONNAIRE!

Your time and efforts are greatly appreciated

Telehealth tested survey

Survey regarding the novel predictive testing protocol for Huntington disease

Your participation in this survey is completely voluntary and your answers will be kept confidential

HOW TO COMPLETE THIS QUESTIONNAIRE

- Most questions require that you simply TICK the box (☐) indicating the appropriate answer(s)
- There are no right or wrong answers; we are interested in your personal opinions and experiences
- Some questions may appear repetitive, but please note that there are some important and subtle differences
- If you need more space when answering a question, please use additional pages at the back of the questionnaire
- If you have any questions or would like to talk to any of us about your opinions and experiences, please call us at: 6048752000 x 4638

ID:

Section 1: Respondent details

(To be administered after pre-test counseling appointment)

This section contains questions about you in order to describe the people that have participated in this study.
Please check or fill in the answer that applies

1.1 Are you...? ☐ Male ☐ Female

1.2 What is your marital status? (please check ONE)

- ☐ Never married (single)
- ☐ Married and living with spouse
- ☐ Separated
- ☐ Divorced
- ☐ Widowed
- ☐ Common law/ live in partner
- ☐ Other (please specify

1.3 Do you have children? (please check ONE) ☐ Yes ☐ No

1.4 People living in Canada come from many different cultural and racial backgrounds. To which ethnic or cultural group do you belong? (Please check ONE)

- ☐ White or European descent
- ☐ Aboriginal (North American Indian, Metis, or Inuit)
- ☐ Black or African American
- ☐ Latin American
- ☐ Chinese
- ☐ Japanese
- ☐ Korean
- ☐ South Asian (i.e., East Indian, Pakistani, Sri Lankan)

- ☐ South East Asian (i.e., Cambodian, Indonesian, Laotian, Vietnamese)
- ☐ West Asian (i.e., Afghan, Iranian)
- ☐ Filipino
- ☐ Arab
- ☐ Don't know
- ☐ Other (please specify)_____

1.5 What is the highest grade or level of education you have attained? (Please check ONE)

- ☐ No schooling
- ☐ Some elementary
- ☐ Completed elementary
- ☐ Some secondary
- ☐ Completed secondary
- ☐ Some community college, technical college, CEGEP or Nursing training
- ☐ Completed community college, technical college, CEGEP or Nursing training
- ☐ Some university or teacher's college
- ☐ Completed university or teacher's college
- ☐ Other (please specify)_____

_____

1.6 What is your current employment status (please check ONE)

- ☐ Full-time
- ☐ Part time
- ☐ Unemployed and seeking work
- ☐ Unemployed or retired and not seeking work

1.7 Please describe your occupation_____

1.8 What is your best estimate for your total household income in the last 12 months before taxes and deductions? Please tick ONE

- ☐ Less than $15,000
- ☐ $15,000 – 29,999
- ☐ $30,000 – 44,999
- ☐ $45,000 - 59,999
- ☐ $60,000 – 74,999
- ☐ $75,000 – 89,999
- ☐ $90,000 – 104,999
- ☐ Over $105,000
- ☐ Don't know
- ☐ Other (please specify)_____

1.9 Which of the following best describes your experience with HD (Please tick ONE)

- ☐ I have had no prior experience with people who had HD
- ☐ I have known people who have had early symptoms of HD
- ☐ I have known people who have had severe disease or have died

Section 2: Pre-test period and testing process

(To be administered after pre-test counseling appointment(s))

The section refers to all the appointments, conversations and visits you had regarding predictive testing for HD prior to receiving your results. This will help us understand your experience of the testing process, as well as what you found beneficial/ helpful and difficult/ unnecessary.

2.1 What was format of your pre-test counseling appointment(s) (please check all that apply)

- ☐ Over the phone with the genetic counsellor (Susan Creighton) at the HD clinic at UBC
- ☐ Via telehealth with the genetic counsellor (Susan Creighton) at the HD clinic at UBC
- ☐ Via telehealth with the genetic counsellor (Susan Creighton) and geneticist (Michael Hayden) at the HD clinic at UBC
- ☐ With my local GP/ healthcare provider_____(please specify)

2.2 What was the entire time it took to attend the pre-test counseling appointment (s) (including travel and appointment time)?[31]

☐ 1-4 hours ☐ 4-6 hours ☐ 6-10 hours ☐ 10-24 hours ☐ Other:_____

Section A: Reasons for undergoing testing for HD

2.3 What were your reasons for undergoing HD testing (please rate how strongly you agree or disagree with EACH of the following statements by placing a tick in the appropriate box)

[31] Note exact time in hours to attend appointment (rather than range) was clarified with each respondent subsequent to returning the survey. This was done so that statistical analysis of the difference between the groups could be established.

Reason	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. Knowing is better than not knowing, even if it can be stressful					
b. Knowing can help you understand the role of genetics in your or your family's lives					
c. Knowing the result can give you a greater sense of personal control and certainty					
d. Knowing can help you plan your future					
e. Knowing can help you make choices about having children					
f. Knowing can help in your communication and relationships with your family					
g. Knowing can help in your communication and relationships with others					
h. Knowing can give you the opportunity to take preventive actions to control or monitor the development of symptoms					
i. Knowing can help you share the family history with others at risk in your family					
j. Knowing can give you a reliable scientific basis to use in explaining your or your family's situation to others					
k. Knowing can allow you to support and/or be supported by others					

l. Other (please specify)_____

_____

2.4 What were your reservations about undergoing predictive testing for HD?

Reason	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I had concerns about coping with the results					
b. I had concerns about family members coping with the results					
c. I had concerns about the implications of the test results for family members					
d. I had concerns about privacy and confidentiality					
e. I had concerns about insurance					
f. I had concerns about the cost of testing					
g. I had concerns about employers/ employment					
h. I had concerns about how others would treat me if they knew I had a gene mutation					
i. There is no cure for Huntington disease					

2.5 Why did you decide to undergo testing at this time?

☐ I recently (within 3 months) learnt about the condition in a family member and decided to undergo testing

☐ I had known about the condition for some time (more than 3 months) but decided to undergo testing for the reasons outlined above (in question 2.3)

Please provide details on your exact reasons:_____

_____

_____

_____

_____

☐ I had wanted to undergo testing for sometime but had been unable to attend the appointments at UBC (go to question 2.6)

☐ Other_____

_____

2.6 If you checked the third box in question 2.5 please rank the reasons you were unable to make the appointments (1 being the biggest reason)

☐ Could not take time away from work _____

☐ Did not want/ could not take time away from family _____

☐ Cost of travel to the HD clinic _____

☐ No available transportation to get to Vancouver _____

☐ Nowhere to stay in Vancouver_____

☐ Do not like going to Vancouver_____

☐ Other:_____

_____

_____

_____

2.7 Please complete the following table to indicate the amount of time you spent communicating with healthcare professionals prior to deciding to undergo predictive testing

	Amount of time
a. How much time **in total** did you spend communicating with the **Geneticist or Genetic Counsellor at UBC before** you decided to undergo testing?	hours
b. How much time **in total** did you spend communicating with your **family physician before** you decided to undergo testing?	hours

2.8 Before undergoing testing would you have preferred more appointments/conversations sessions to discuss HD testing with any of the above mentioned healthcare professionals?

- ☐ Yes
- ☐ No

2.9 Please read each of the following statements and put a check mark in the chart which best corresponds to your situation.

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. **Prior to my first contact** with the HD genetic testing centre or other healthcare provider I had a good knowledge of the possible advantages and disadvantages of undergoing predictive genetic testing for HD					
b. **After my appointment (s) or discussion (s)** with the HD genetic testing centre or other healthcare providers I had a good knowledge of the possible advantages and disadvantages of undergoing					

predictive genetic testing for HD					
c. My knowledge of the possible advantages and disadvantages of undergoing predictive genetic testing for HD significantly improved after my **after my appointment (s) or discussion (s)** with the HD genetic testing centre or other healthcare providers					
d. I was satisfied with the amount of information provided **before** I underwent testing					
e. I was able to understand all the information that I discussed with my healthcare professional (including the HD predictive testing centre) before I underwent HD testing					
f. I had enough time to discuss all of my concerns with my healthcare professional (including the HD predictive testing centre) before I underwent HD testing					
g. All of my questions were answered by my healthcare professional (including the HD predictive testing centre) before I underwent HD testing					
h. Overall, I was given enough information so that I felt able to make fully informed choice regarding HD predictive genetic testing					
i. Overall, I was given enough **support** during the pre-test consultation and counselling process (prior to undergoing HD predictive testing)					

2.10 What other information would you have liked to have received from your healthcare professional before you underwent HD testing?_____

_____

_____

_____

2.11 Overall, what do you think about the amount of time of the pre-testing consults and counselling visits (prior to undergoing HD predictive testing)?

a. ☐ Too much

b. ☐ Just right

c. ☐ Too little

Section B: Telehealth (videoconferencing) experience

2.12 This was my first Telehealth visit:

☐ Yes ☐ No

2.13 There were problems with the Telehealth equipment:

☐ Yes ☐ No

2.14 I was able to complete my visit through Telehealth:

☐ Yes ☐ No

2.15. Do you agree with the following statements?

I knew what to expect during my Telehealth visit	Yes ☐	No ☐	N/A ☐
I could see the healthcare provider clearly	Yes ☐	No ☐	N/A ☐
I could hear the healthcare provider clearly	Yes ☐	No ☐	N/A ☐
I felt comfortable in the room where my Telehealth visit was held	Yes ☐	No ☐	N/A ☐
I felt that my privacy was respected	Yes ☐	No ☐	N/A ☐
It was easier for me to attend my appointment with my healthcare provider using Telehealth rather than in person	Yes ☐	No ☐	N/A ☐
I prefer to see my healthcare provider using Telehealth rather than in person	Yes ☐	No ☐	N/A ☐
Telehealth allowed me to see my healthcare provider sooner than I could have in person	Yes ☐	No ☐	N/A ☐
Telehealth allowed me to have more regular follow-up for my condition	Yes ☐	No ☐	N/A ☐
Overall, I was satisfied with the Telehealth visit	Yes ☐	No ☐	N/A ☐
I would use Telehealth again	Yes ☐	No ☐	N/A ☐

If Telehealth was not available, I would have travelled for my visit	Yes □	No □	N/A

2.16 Telehealth saved me and my family time

☐ Yes ☐ No

2.17 If yes, please complete the following table:

Time off work	Yes □	No □	N/A □
Travel	Yes □	No □	N/A □
Other:			

2.18 Telehealth saved me and my family money

☐ Yes ☐ No

2.17 If yes, please complete the following table:

Airfare	Yes □	No □	N/A □
Bus	Yes □	No □	N/A □
Gas	Yes □	No □	N/A □
Childcare	Yes □	No □	N/A □
Hotel	Yes □	No □	N/A □
Meals	Yes □	No □	N/A □
Other:			

Section 3. Results session

(to be administered 1-2 weeks after results session)

This section refers to how and when you received your HD predictive test results.

3.1 How were your genetic testing results delivered?

- ☐ In person by the genetic counsellor/ geneticist at UBC
- ☐ In person by my family physician
- ☐ In person by a social worker
- ☐ Over the phone by the genetic counsellor/ geneticist
- ☐ Over the phone by my family physician
- ☐ Over the phone by a social worker
- ☐ Mailed letter_____ from_____(please insert)
- ☐ Other_____

3.2 What do you believe would have been the best method of delivering your genetic testing results?

- ☐ In person by the genetic counsellor/ geneticist
- ☐ In person by my family physician
- ☐ In person by a social worker
- ☐ Over the phone by the genetic counsellor/ geneticist
- ☐ Over the phone by my family physician
- ☐ Over the phone by a social worker
- ☐ Mailed letter_____ from_____(please insert)

☐ Other_____

3.3 For each of the following questions please check the answer that best applies to your situation

Question	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I was satisfied with the manner in which my genetic test results were delivered to me					
b. I was able to understand all the information about my genetic test result that was discussed during the results session					
c. I had enough time to discuss all of my concerns about my genetic testing results with my healthcare professional during my genetic test result session					
d. All of my questions about my genetic testing results were answered by my healthcare professional					
e. Overall, I was given enough **information** regarding the meaning and implications **for me** of my genetic testing results by my healthcare professional					
f. Overall, I was given enough **information** regarding the meaning and implications **for my children** of my genetic testing results **for me** by my healthcare professional					
g. Overall, I was given enough **information** regarding the meaning and implications **for my extended family** of my genetic testing results **for me** by my healthcare professional					
h. Overall, I was given enough **support** (emotional & psychological) during the results session by my healthcare professional					

3.4 What other information would you have liked to receive about your genetic testing results by your healthcare professional?_____

_____

_____

3.5 What other support would you have liked to receive when you were given your genetic testing results?_____

_____

_____

3.6 Overall, what do you think about the amount of time of the results session with your healthcare professional? (please check)

a. ☐ Too much

b. ☐ Just right

c. ☐ Too little

Section 4. Follow-up since results

This section refers to the period after you received your genetic test result.

For each of the following questions please check the answer that best applies to your situation

4.1 Have you received any follow-up contact from the genetics team at UBC since receiving your genetic testing results?

☐ Yes ☐ No

If you answered no to question 4.1 please proceed to question 4.2

4.2 When did this follow up contact with the genetics team at UBC take place

☐ In the first week after I received my test results

☐ 2-4 weeks after I received my test results

☐ 4-8 weeks after I received my test results

4.3 Were you satisfied with the amount of time it took for the follow-up to take place?

☐ Too much ☐ Just right ☐ Too little

4.4 How did this follow-up contact with the genetics team take place?

☐ The genetics team called me on the phone

☐ The genetics team sent me an email

☐ I had an in person visit with the genetics team

- ☐ I had a video conferencing (telehealth) appointment with the genetics team
- ☐ The genetics team left me a voicemail and I called them back
- ☐ The genetics team left me a voicemail and I did not call them back

  If you did not call the genetics team back please explain why_____

  _____

  _____

  _____

- ☐ I called the genetics team
- ☐ I called the genetics team but they did not call me back
- ☐ Other:_____

4.5 Please complete the following table regarding your follow-up contact with the genetics team by placing a check mark next to each statement indicating your level of agreement with the statement

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I was satisfied with the follow-up contact I received from the genetics team					
b. I was able to understand all that was discussed with the **genetics team** during my follow up contact					
c. I had enough time to discuss all of my concerns with the **genetics team** during my follow up contact					
d. All of my questions were answered by the **genetics team** during my follow up contact					
e. Overall, I was given enough **information** from the **genetics team** during the period following my genetic test results					
f. Overall, I was given enough **support** from the **genetics team** during the period following my					

genetic test results					

4.6 Overall, I think the follow-up contact from the genetics team during the period following my genetic test results was worthwhile:

☐ Yes ☐ No

4.7 Have you received any follow-up contact from your local healthcare provider (family physician) since receiving your genetic testing results?

☐ Yes ☐ No

If you answered no to question 4.7 please proceed to question 4.11

4.8 When did this follow up contact with your local healthcare provider (family physician) take place

☐ In the first week after I received my test results

☐ 2-4 weeks after I received my test results

☐ 4-8 weeks after I received my test results

4.9 Were you satisfied with the amount of time it took for the follow-up to take place?

☐ Too much ☐ Just right ☐ Too little

4.10 How did this follow-up contact with the genetics team take place?

☐ My local healthcare provider (family physician) called me on the phone

☐ I had an in person visit with my local healthcare provider (family physician)

☐ I had a video conferencing (telehealth) appointment with my local healthcare provider (family physician)

☐ My local healthcare provider (family physician) left me a voicemail and I called them back

☐ My local healthcare provider (family physician) left me a voicemail and I did not call them back

If you did not call the genetics team back please explain why_____

_____

_____

_____

☐ I called my local healthcare provider (family physician)

☐ I called my local healthcare provider (family physician) but they did not call me back

☐ Other:_____

4.11 Please complete the following table regarding your follow-up contact with your local healthcare provider (family physician) team by placing a check mark next to each statement indicating your level of agreement with the statement

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I was satisfied with the follow-up contact I received from the genetics team					
b. I was able to understand all that was discussed with the **genetics team** during my follow up contact					
c. I had enough time to discuss all of my concerns with the **genetics team** during my follow up contact					
d. All of my questions were answered by the					

genetics team during my follow up contact					
e. Overall, I was given enough **information** from the **genetics team** during the period following my genetic test results					
f. Overall, I was given enough **support** from the **genetics team** during the period following my genetic test results					

4.12 Overall, I think the follow-up contact from the genetics team during the period following my genetic test results was worthwhile:

☐ Yes ☐ No

4.13 Please provide any other comments regarding the post-test result period:_____

_____

_____

_____

_____

_____

_____

Section 5. Overall satisfaction with testing process

(to be administered 6 months after receiving test result)

This section refers to the six month follow-up appointment from receiving your test result. This section is also about your overall satisfaction with the entire predictive testing process including the pre-test period, the results period, and the follow-up period.

For each of the following questions please check the answer that best applies to your situation

5.1 Have you undergone 6 month follow-up contact with the genetics team at UBC since receiving your genetic testing results?

☐ Yes ☐ No

5.2 When did this follow up contact with the genetics team at UBC take place?

☐ 5-6 months after I received my test results

☐ 6-7 months after I received my test results

☐ More than 7 months after I received my test results

5.3 Were you satisfied with the amount of time it took for the follow-up to take place?

☐ Too much ☐ Just right ☐ Too little

5.4 How did this follow-up contact with the genetics team take place?

☐ The genetics team called me on the phone

☐ The genetics team sent me an email

☐ I had an in person visit with the genetics team

- ☐ I had a video conferencing (telehealth) appointment with the genetics team
- ☐ The genetics team left me a voicemail and I called them back
- ☐ The genetics team left me a voicemail and I did not call them back

If you did not call the genetics team back please explain why_____

_____

_____

_____

- ☐ I called the genetics team
- ☐ I called the genetics team but they did not call me back
- ☐ Other:_____

5.5 Please complete the following table regarding your follow-up contact with the genetics team by placing a check mark next to each statement indicating your level of agreement with the statement

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. I was satisfied with the follow-up contact I received from the **genetics team**					
b. I was able to understand all that was discussed with the **genetics team** during my follow up contact					
c. I had enough time to discuss all of my concerns with the **genetics team** during my follow up contact					
d. All of my questions were answered by the **genetics team** during my follow up contact					
e. Overall, I was given enough **information** from the **genetics team** during the period following my genetic test results					
f. Overall, I was given enough **support** from the **genetics team** during the period following my					

genetic test results					

5.6 Overall, I think the follow-up contact from the genetics team during the period following my genetic test results was worthwhile:

☐ Yes ☐ No

5.7 Please provide any other comments regarding the post-test result period:_____

_____

_____

_____

_____

_____

_____

5.8 How much <u>benefit or advantage,</u> if any, have you felt from knowing about your genetic test result?

☐ Great benefit ☐ Some benefit ☐ Neutral ☐ Little benefit ☐ No benefit

5.9 Do you think knowing about your genetic test result has bought benefit because....(please rate how strongly you agree or disagree with EACH of the following statements by placing a tick in the appropriate box)

Question	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. Knowing is better than not knowing, even if it can be stressful					
b. Knowing has helped you understand the role of genetics in your or your family's lives					
c. Knowing the result has given you a greater sense of					

personal control and certainty					
d. Knowing has helped you plan your future					
e. Knowing has helped you make choices about having children					
f. Knowing has helped in your communication and relationships with your family					
g. Knowing has helped in your communication and relationships with others					
h. Knowing has given you the opportunity to take preventive actions to control or monitor the development of symptoms					
i. Knowing has helped you share the family history with others at risk in your family					
j. Knowing has given you a reliable scientific basis to use in explaining you or your family's situation to others					
k. Knowing has allowed you to support and/or be supported by others					

l. Other: _____

_____

_____

_____

5.10 Please rate how strongly you agree or disagree with EACH of the following statements by placing a tick in the appropriate box:

Statement	Strongly agree	Agree	Neutral	Disagree	Strongly disagree
a. Overall, I was satisfied with the **support** provided during the entire testing process by healthcare professionals					
b. Overall I felt that testing process was sufficient in terms of the **information regarding** the predictive testing process provided during the entire testing process by healthcare professionals					
c. Overall I felt that the testing process was sufficient in terms of the information regarding the **meaning of my genetic test results for me** provided during the entire testing process by healthcare professionals					
d. Overall I felt that the testing process was sufficient in terms of the information regarding the **meaning of my genetic test results for my family** provided during the entire testing process by healthcare professionals					
e. Overall, undergoing testing via testing via the offsite testing protocol (i.e., not having to go to Vancouver) saved me time					
f. Overall, undergoing testing via testing via the offsite testing protocol (i.e., not having to go to Vancouver) saved me money					
g. Overall I would have preferred to have **more in person visits** at the HD testing centre during my predictive testing process					
h. Overall I would have preferred to have **more telephone/ telehealth appointments with** the HD testing centre during my predictive testing process					
i. Overall I would have preferred to have **less in person visits** with my family physician during my					

predictive testing process					
j. Overall I would have preferred to have **more in person visits** with my family physician during my predictive testing process					

5.12 Please provide any additional comments on your testing experience and suggestions for improvements in the space below:

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THANK YOU FOR TAKING THE TIME TO COMPLETE THE QUESTIONNAIRE!

Your time and efforts are greatly appreciated

Beck depression inventory

For each question, please check the box number for each statement that fits best with how you are feeling right now

Question A

☐ 0) I do not feel sad

☐ 1) I feel sad or blue

☐ 2) I am sad or blue all the time and can't snap out of it **or** I am so sad and unhappy that it is very painful

☐ 3) I am so sad or unhappy that I can't stand it

Question B

☐ 0) I am not particularly pessimistic or discouraged about the future

☐ 1) I feel discouraged about the future

☐ 2) I feel I have nothing to look forward to **or** I feel that I won't ever get over my troubles

☐ 3) I feel that the future is hopeless and that things cannot improve

Question C

☐ 0) I do not feel like a failure

☐ 1) I feel I have failed more than the average person

☐ 2) I feel I have accomplished very little that is worthwhile or that means anything **or** as I look back on my life all I can see is a lot of failures

☐ 3) I feel I am a complete failure as a person

Question D

☐ 0) I am not particularly dissatisfied

☐ 1) I feel bored most of the time **or** I don't enjoy things the way I used to

☐ 2) I don't get satisfaction out of anything anymore

☐ 3) I am dissatisfied with everything

Question E

☐ 0) I don't feel particularly guilty

☐ 1) I feel bad or unworthy a good part of the time

☐ 2) I feel quite guilty **or** I feel bad or unworthy practically all the time now

☐ 3) I feel as though I am very bad or worthless

Question F

☐ 0) I don't feel I am being punished

☐ 1) I have a feeling that something bad may happen to me

☐ 2) I feel I am being punished or will be punished

☐ 3) I feel I deserve to be punished **or** I want to be punished

Question G

☐ 0) I don't feel disappointed in myself

☐ 1) I am disappointed in myself **or** I don't like myself

☐ 2) I am disgusted with myself

☐ 3) I hate myself

Question H

☐ 0) I don't feel I am any worse than anybody else

☐ 1) I am very critical of myself for my weaknesses or mistakes

☐ 2) I blame myself for everything that goes wrong **or** I feel I have many bad faults

Question I

☐ 0) I don't have any thoughts of harming myself

☐ 1) I have thoughts of harming myself but would not carry them out

☐ 2) I feel I would be better off dead **or** I have definite plans about committing suicide **or** I feel my family would be better off if I were dead

☐ 3) I would kill myself if I could

Question J

☐ 0) I don't cry anymore than usual

☐ 1) I cry more now than I used to

☐ 2) I cry all the time now and I can't stop it

☐ 3) I used to be able to cry but now I can't cry at all even though I want to

Question K

☐ 0) I am no more irritated now than I ever am

- ☐ 1) I get annoyed or irritated more easily than I used to
- ☐ 2) I feel irritated all the time
- ☐ 3) I don't get irritated at all at the things that used to irritate me

Question L

- ☐ 0) I have not lost interest in other people
- ☐ 1) I am less interested in other people now than I used to be
- ☐ 2) I have lost most of my interest in other people and have little feeling for them
- ☐ 3) I have lost all my interest in other people and don't care about them at all

Question M

- ☐ 0) I make decisions about as well as ever
- ☐ 1) I am less sure of myself now ant try to put off making decisions
- ☐ 2) I can't make decisions any more without help
- ☐ 3) I can't make any decisions at all any more

Question N

- ☐ 0) I don't feel I look any worse that I used to
- ☐ 1) I am worried that I am looking old or unattractive
- ☐ 2) I feel that there are permanent changes in my appearance and they make me look unattractive
- ☐ 3) I feel that I am ugly or repulsive looking

Question O

- ☐ 0) I can work about as well as before
- ☐ 1) It takes extra effort to get started at doing something **or** I don't work as well as I used to
- ☐ 2) I have to push myself very hard to do anything
- ☐ 3) I can't do any work at all

Question P

- ☐ 0) I can sleep as well as usual
- ☐ 1) I wake up more tired in the morning that I used to
- ☐ 2) I wake up 1-2 hours earlier than usual and find it hard to get back to sleep
- ☐ 3) I wake up early every day and can't get more than 5 hours sleep

Question Q

- ☐ 0) I don't get any more tired than usual
- ☐ 1) I get tired more easily than I used to
- ☐ 2) I get tired from doing anything
- ☐ 3) I get too tired to do anything

Question R

- ☐ 0) My appetite is no worse than usual
- ☐ 1) My appetite is not as good as it used to be
- ☐ 2) My appetite is much worse now

☐ 3) I have no appetite at all any more

Question S

☐ 0) I haven't lost much weight, if any, lately

☐ 1) I have lost more than 5 pounds

☐ 2) I have lost more than 10 pounds

☐ 3) I have lost more than 15 pounds

Question T

☐ 0) I am no more concerned about my health than usual

☐ 1) I am concerned about aches and pains *or* upset stomach *or* constipation *or* other unpleasant feelings in my body

☐ 2) I am so concerned with how I feel or what I feel that it's hard to think of much else

☐ 3) I am completely absorbed in what I feel

Question U

☐ 0) I have not noticed any recent change in my interest in sex

☐ 1) I am less interested in sex than I used to be

☐ 2) I am much less interested in sex now

☐ 3) I have lost interest in sex completely

BDI SCORE

Please add up the numerical value you circled for each question and enter here: _____