BARRIERS TO ACCESS TO AND UPTAKE OF GENETIC SERVICES FROM THE PERSPECTIVES OF GENETIC PROFESSIONALS

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by

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ABSTRACT

This research examines barriers to the provision of genetic services from the perspectives of genetics professionals working in the province of Newfoundland and Labrador, Canada. Based on open-ended and semi-structured key informant interviews, the study assesses the structure and capacity of the Provincial Genetic Services Program, the referral process and protocols followed, and the social, historical, and cultural factors shaping the utilization of genetic services from the perspectives of those who provide the services. The thesis reports on the factors that support the use of genetic services and factors that deter or decrease this use. It identifies strategies for overcoming challenges to access to and uptake of genetic services. The key findings are incorporated into recommendations to help define areas and directions for improvements in clinical genetics and to provide advice for those who develop and deliver genetic services.

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Chapter 1: Introduction

One of the promises of genetic research discovery is to identify the genetic basis of diseases and, ultimately, to improve health outcomes. In the last few decades, genetics has rapidly evolved from pure (lab) science into a new clinical discipline in the field of health care. Genetic services are the conduit for translating new genetic knowledge into clinical practice: the services help elucidate the genetic etiology of diseases and determine risk, diagnose single-gene or multi-factorial disorders and offer curative and/or preventive treatment, including population carrier screening.

The purpose of this study is to report on barriers to access to and uptake of genetic services in the Canadian province of Newfoundland and Labrador. The unique geography, history and culture of the province frames whether and how genetic services are accessed and used. This study examines the current structure and functionality of genetic services in the province of Newfoundland and Labrador (NL) and determines what factors hinder the effective and efficient delivery of genetic services from the perspectives of those who deliver these services.

1.1 State of current knowledge

This study builds on the existing body of knowledge about access to and uptake of genetic care. That research has attended to the perspectives of individual recipients of genetic care and has thoroughly investigated issues identified by clinical genetic clients (e.g., Turney, 2009; Benkendorf et al., 1997; Durfy, Bowen, McTierman, Sporleder, & Burke, 1999; Falcone, McCarthy-Wood, Xie, Siderowf, &Van Dellin, 2011; Lock,

Freeman, Sharples, & Lloyd, 2006; Peterson, Milliron, Lewis, Goold, & Merajver, 2002). That research includes examinations of client perceptions and attitudes to hereditary risk and predictive genetic testing (e.g., d'Agincourt-Canning, 2005; Cooke & French, 2008; Dahodwala et al., 2007; Calsbeek et al., 2007), the impact of genetic testing on psychosocial well-being (e.g., Graceffa et al., 2009; Edge, 2008; Vadaparampil, Miree, Wilson, & Jacobsen, 2006) and the complexities surrounding decision making about genetic testing (e.g., Cox & McKellin, 1999). Research with client recipients of genetic services has also examined intra-familial experiences of genetic risk in relation to perceived and actual kinship ties, illustrating how the flow of genetic risk information among relatives has a profound influence on, and is shaped by, by family structures and family dynamics (e.g., Forrest et al., 2003; Gaff, Collins, Symes, & Halliday, 2005).

Findings of client-based research have been used to make recommendations for improving genetic service provision. For example, Skirton, Parsons, & Ewings (2005) developed the *Audit Tool for Genetic Services,* aimed at improving the outcomes of genetic services. Beene-Harris, Wang & Bach (2007), in their call for attention to the inequalities in access to genetic services, suggested the need for proactive and novel approaches to achieving improved and effective genetic care (see also Hawkins & Hayden, 2011).

Research on public attitudes toward genetic testing has also shaped the existing knowledge about access to and uptake of genetic services. Researchers have documented a lack of public awareness about genetic screening and testing (McClaren, Delatycki,

Collins, Metcalfe & Aitke, 2008; Jonassaint et al., 2010). That lack of awareness has been attributed to deficits in the general knowledge about and understanding of basic human genetics (Christianson et al., 2010). Although members of the public have improved their interest and knowledge of genetics over the past decades (in part due to media attention surrounding the Human Genome Project) misunderstandings about genetics persist in many developed countries including the US (US National Science Board, 2008), Australia (Molster, Charles, Samanek, & O'Leary, 2009), and the UK (Voss, 2000).

A number of studies have related the challenges with uptake of genetic services to a lack of effective knowledge communication by primary health care providers and medical specialists (e.g., Geller et al., 1998; Greendale & Pyeritz, 2001; Rich et al., 2004). Physicians' knowledge deficiencies in genetics (Starfield et al., 2002, Kegley 2003), concerns about time and cost for discussing genetics with patients (Watson, Shickle, Qureshi, Emery, & Austoker, 1999) and perceptions about the relevance of genetics in their practice (Mountcastle-Shah & Holzman, 2000) are reported to contribute to the low rate of patient referrals for genetic counselling or testing (Watson, Austoker, & Lucassen, 2001).

There has been very little research on the effectiveness of genetic services from the perspectives of those who provide the services; yet their observations and attitudes are key to identifying barriers, both system- and client-related. While, there is an important body of literature focusing on the perspectives of genetic counsellors, that

research emphasises the challenges of the genetic counselling process, including dilemmas around professional obligations to members of the same family (e.g., Chan-Smutko, Patel, Shannon, & Ryan, 2008) and strategies for disclosing genetic test results to patients (e.g., Wham et al., 2010). Missing is research examining the challenges inherent within the system, from the perspectives of those who are the front-line care providers.

This study is unique in that it focuses on the perspectives of genetic professionals who are the front-line providers of genetic information and services to patients and clients[1]. Their viewpoints provide important insights into the barriers to accessing genetic services and into the factors that shape those barriers. Their perspectives are key to understanding the range of ways in which genetic care is understood, practiced, accessed, used or dismissed altogether.

This thesis reports on the factors that support the use of genetic services and factors that deter or decrease this use, in the context of the broader geographic, economic and cultural context of the province of Newfoundland and Labrador. The genetic professionals who participated in this study discuss the successes of the Newfoundland and Labrador Provincial Medical Genetics Program (PMGP),[2] as well as the barriers to accessing and using the clinical services offered through the program. Their perspectives

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[1] In the Newfoundland and Labrador context at the time of my research, the front-line genetic professionals included medical geneticists, genetic counselors, genetic nurses, and clinical genetic researchers.

[2] The Provincial Medical Genetics program (PMGP), also referred to as the "Genetic Services Department," is part of the Eastern Regional Health Authority of Newfoundland and Labrador and is mandated to deliver genetic services.

provide valuable information about how to effectively translate genetic research into genetic care and ultimately into improved health outcomes. In the context of publicly funded genetic research and genetic services, this translation piece is a measure of return on public investments.

This study also aims at suggesting strategies for overcoming the challenges identified by genetic professionals. It offers recommendations that help define areas and directions for improvements in clinical genetics. The recommendations are designed to inform policy and other genetics-related regulatory developments at regional, provincial, and national levels.

In this Chapter, I present detailed background information on the context and rationale for this study. I examine national, regional, and local milieus and explain how this study is relevant to the current state of genetic service provision in the province and how it has the potential to inform future trends and developments. Chapter 2 describes my methodology. In Chapters 3 and 4, I present a synthesis of the participants' responses and I discuss my findings, which fall into the two broad categories: systemic and psychosocial barriers. I devote Chapter 5 to a synthesis and discussion of my core findings and I provide comments on the limitations of my study. In the same chapter, I provide recommendations for future research needs as well as for system and policy improvements.

1.2 Background

1.2.1 The Human Genome Project (HGP). In the past three decades, rapid advances in human genetics and genetic technologies have brought about the promise of an improved understanding of, as well as better management of, human health and human disease. The increased possibility of enhanced health outcomes through genetic knowledge in the 1980s gave rise to the HGP – a symbolic and practical center of research activity to generate clinically significant knowledge to improve health. The HGP began formally in 1990 and was intended as a 15-year effort coordinated by the U.S. Department of Energy and the National Institutes of Health (U.S. Department of Energy, 2011). The Project aimed to identify all genes in the human genome (approximately 20,500 human genes) and the sequences of the 3 billion chemical base pairs that comprise the human DNA[3] (National Human Genome Research Institute, 2011). The initiative was followed by an explosion of technological advances, sparking the need for a parallel line of inquiry into the potential ethical, legal, and social implications (ELSI)[4] of the new genetic technologies, the information being produced, and the use (and non-use) of resulting genetic knowledge and clinical services. My study is part of that broader endeavor.

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[3] The HGP was completed in 2003.

[4] ELSI is the American acronym for the ethical, legal and social issues associated with advances in genomic and genetic research. In Canada, the term associated with the cluster of ethical and legal issues is GE[3]LS, which stands for genomics and its ethical, environmental, economic, legal, and social aspects.

1.2.1.1 Canadian context. At least 18 countries participated in the HGP and established national human genome research programs. Among those countries, Canada established itself as a leader in genomics research. The country has been recognized as having outstanding discovery potential as well as highly regarded genomics research facilities and scientists.[5] Canada's success can, to a large extent, be credited to Genome Canada, a non-profit organization established in early 2000 to develop and implement a national strategy for supporting comprehensive genomics research projects beneficial to all Canadians. These projects cover strategic areas such as agriculture, environment, fisheries, forestry, health and new technology development. Genome Canada was given a mandate by the Canadian Government to be a primary funding and information resource for human genome research with government funds allocated accordingly. The organization has cultivated a network of outstanding genetic scientists and researchers. From the outset, Genome Canada had adopted three novel approaches to supporting research: it required co-funding of projects with both domestic and international partners; it established regional focal points of expertise in genomics research across Canada; and it required the inclusion of research into the ethical, environmental, economic, legal and social (GE^3LS) aspects and potential implications of the scientific research.[6]

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[5] Burrill & Company (2007). Biotech 2007: Life sciences – a global transformation. In *Genome Canada*. Retrieved from http://www.genomecanada.ca/en/about/

[6] Genome Canada(n.d.). GE^3LS, Genomics & Society. Retrieved from http://www.genomecanada.ca/en/ge3ls/about/

There are six Canadian genome centers, situated in British Columbia, Alberta, the Prairies, Ontario, Quebec, and Atlantic Canada. These centers attract new researchers and support research activities utilizing new approaches and technologies. These hubs are also conducive to regional project development that reflects the specific needs of a given area. Genome Atlantic is one of the six regional genome centers and encompasses all four Atlantic Provinces. It is dedicated to building genomics investment and economic growth in Atlantic Canada.

1.2.1.2 Newfoundland and Labrador context. In recent years, the province of Newfoundland and Labrador[7], has become a "hot spot" for genetic research. This heightened interest is due in part to the willingness of residents to participate in scientific research (Atkinson, 2000; Greenwood, 2000; Industry Canada, 2002, 104), but primarily to the high incidence of hereditary conditions, such as cardiovascular disease, diabetes, obesity and psoriasis (Atkinson, 2000; Taubes, 2001). The region's unique geography and history are responsible for the high incidence of these conditions and their genetic underpinning. The NL population of 510,000[8] is for the most part descended from the original 20 to 30,000 founders from England (46%) and Ireland (48%) that had arrived before 1830 (Bear et al.,1987). As fish were plentiful, the inshore fishery was the primary means of livelihood. Consequently, small settlements known as outports gradually

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[7] Newfoundland and Labrador is Canada's most easterly province encompassing the island of Newfoundland as well as Labrador on the Canadian mainland.

[8] Newfoundland & Labrador Statistics Agency (2011, September). *Population by Age Groups and Sex Newfoundland and Labrador 1971-2011.* Retrieved from (http://www.stats.gov.nl.ca/Statistics/Population/PDF/PopAgeSex_BS.PDF

appeared along the coastline around natural harbours.[9] Family sizes tended to be large and, because of geographic isolation and religious segregation (English Protestant/Irish Catholic), multiple distinct genetic isolates cropped up on the island of Newfoundland.

Genetic relatedness and genetic isolation are suspected in the number of localized concentrations of inherited diseases (Bear et al., 1987). In comparison with other founder populations, in Newfoundland the founder population is relatively recent and comprised of a very limited number of founders (Rahman et al., 2003). This may explain why nowhere else in the country, including the other Atlantic Provinces, is the founder population as ethnically homogeneous and geographically stable as it is in this province. The founder effect, characterized by a decrease in genetic diversity resulting in genetic drift, has been identified for many genetic diseases. The local population displays an elevated prevalence of genetic disorders as well as elevated carrier frequencies, which makes this province a particularly attractive place for genetic research. As well, detailed and recorded information on the genealogical history of the local families (typically of large size and closely knit) is readily available or can be easily assembled. The emergence of Newfoundland as a "hot spot" for genetic research was based on the contention that such a well-documented homogenous pool makes it "easier for researchers to identify the genes associated with specific diseases" (Industry Canada, 2002, p. 103). Recognizing the substantial potential of the province as a suitable place for gene discovery, Genome Canada and Genome Atlantic co-funded genetic research in

[9] Even today, half of the population resides in communities with fewer than 2,500 inhabitants while a large proportion resides in communities of fewer than 1000 (Statistic Canada, 2006).

Newfoundland through the Atlantic Medical Genetics and Genomics Initiative (AMGGI)[10].

1.2.1.3 Atlantic Medical Genetics and Genomics Initiative. AMGGI was a unique project that aimed to systematically identify genes and genetic mutations underlying familial, monogenic disorders in the Atlantic region of Canada. One major aspect of the initiative, apart from disease gene discovery, was to transfer research results from molecular genetic discovery to clinical diagnostic laboratories. Another important aspect of AMGGI was to study the potential impact of genetic advancements on the provision of health care services – in other words, to examine the socio-economic benefits of the AMGGI research. The main AMGGI research sites were at Memorial University of Newfoundland in St. John's, NL and Dalhousie University in Halifax, Nova Scotia. The AMGGI project led to a number of significant novel discoveries, among them, the gene mutation associated with sudden cardiac death (Arrhythmogenic right ventricular cardiomyopathy [ARVC]), the gene causing rare anemia (Congenital sideroblastic anemia) as well as the gene for ataxia (Sensorineural ataxia) and the gene for a rare genetic eye disorder (Schnyder crystalline corneal dystrophy)[11].

***1.2.1.4. Genomics and its Ethical, Environmental, Economic, Legal and Social Aspects* (GE³LS).** An integral component of the AMGGI project was the innovative

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[10] Genome Canada (n.d.). Atlantic Medical Genetic and Genomics Initiative Retrieved from http://www.genomecanada.ca/medias/pdf/en/AtlanticMedicalGeneticAndGenomicsInitiative.pdf

[11] Genome Atlantic (n.d.). World class research and results. Retrieved from http://www.genomeatlantic.ca/projects/view/2-Atlantic_Medical_Genetics_and_Genomics_Initiative_AMGGI#news

study of the potential impacts of genetic discovery on the provision of health care services, including assessing the wellbeing of patients and families who are affected by genetic conditions and who are the most likely consumers of new genetic technologies. An inter-disciplinary GE^{3}LS team was formed to systematically evaluate existing and potential genetic screening programs in Atlantic Canada with a view to "facilitate effective and efficient uptake of genetic services". Utilizing qualitative methods, the aim was to analyze the range of social, historical, cultural and economic barriers to access and use of genetic services from the perspectives of patients, physicians, communities, and policy makers. The GE^{3}LS team was tasked with examining the values, beliefs and practices of physicians and genetic counselors who are the providers of genetic services, as well as those of patients, families and communities to whom these services are offered. This included assessing the genetic burden of disease at a variety of levels (personal, community, provincial, federal), along a number of dimensions (ethical, legal, psychological, sociological, and economic), in a well-defined population. The idea behind the GE^{3}LS component was that a strong collaboration between GE^{3}LS researchers, scientists and clinical investigators would ensure that the translation of genetic research from lab to clinical practice to health policy would be effective.[12]

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[12] Atlantic Medical Genetics and Genomics Initiative (n.d.). Retrieved from http://www.med.mun.ca/amggi/default.htm

1.3 Relevance of research

My study is one aspect of the GE^{3}LS subproject and it was supported in part by funding from the AMGGI project. Therefore, the rationale for my research cannot be separated from the rationale of the larger AMGGI/GE^{3}LS agenda. From the outset of this project, I continuously sought information pertaining to related GE^{3}LS/AMGGI developments. Understanding the broader context of the initiative provided me with the necessary insight to engage genetics providers in meaningful discussions. My own intellectual and personal agenda as a researcher in community health was to examine the social, historical, cultural and economic barriers to access to and use of genetic services from the perspectives of service providers.

My study focuses on what genetic service professionals (GSPs)[13] perceive as client challenges in access to and uptake of genetic services. This focus on the perspectives of GSPs, who have both insight into user perspectives and oversight of the system in general, provides: (1) descriptive accounts of the range and types of barriers to access to and uptake of genetic services, as well as the social, economic and political contexts shaping those barriers; and (2) insights into how and why potential recipients may decline or ignore genetic services in ways that do not particularly reflect "barriers" or "challenges" but rather inappropriateness or irrelevance of the services themselves, to particular individuals.

[13] This is a term I will be using interchangeably with terms such as 'genetic care providers', 'genetics practitioners', 'geneticists' through the thesis to capture the full range of genetic professionals that I interviewed.

In the following chapters, I report on what I learned from the GSPs I interviewed, about the structure and capacity of the Provincial Genetic Services Program, the referral process and protocols followed, and the social, historical, and cultural factors shaping the utilization of genetic services. I begin, in Chapter 2, with a description of the qualitative method I used to conduct my inquiry.

Chapter 2: Method

This study is based on open-ended semi-structured interviews with genetic service providers in the province of Newfoundland and Labrador. The intent of using this qualitative approach was to elicit genetics professionals' perspectives on the challenges to access to and uptake of genetic services. Due to the close-knit nature of professional relationships in the province and the sensitivity of personal genetic information, individual interviews, rather than focus groups, were chosen. This approach facilitated open and honest discussion about challenges to accessing and using genetic services, while maintaining confidentiality and objectivity.

2.1 Recruitment of genetic professionals

For the purposes of this research, I understood "genetic professional" to encompass the range of health professionals with special training in genetics who provide front-line genetic services to patients/clients[14] and their families. In the province of Newfoundland and Labrador, at the time of my research, these front-line providers of genetic information and services included medical geneticists, genetic counselors, genetic nurses, and clinical genetic researchers. Through informal discussions with members of the various genetic service professions at the time of designing the study, I was able to ascertain that there were 13 individuals who fit the criteria of "genetic professional" for the purposes of my research.

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[14] In this thesis, the terms "patient" and "client" will be used interchangeably for ease of communication. I have not chosen one term over the other, because the GSPs used both. However, I should note that although inconsistent in their wording, most GSPs indicated that "client" is be the proper term.

These genetic professionals represent a broad range of types of expertise and fields of specialization. In Newfoundland and Labrador, genetic testing is offered for a wide range of conditions that affect not only individuals but families and communities. The hereditary conditions that are commonly tested for in the province include those that were the object of the AMGGI study – ARVC (see p. 8), colorectal cancer and hereditary hearing impairment. Most genetic professionals are affiliated with the teaching hospital at Memorial University of Newfoundland and clustered in the Eastern Health region encompassing St. John's and surrounding area.[15] Medical geneticists are typically MDs who have completed training in medical genetics and are certified by the Canadian College of Medical Geneticists (CCMG). They are directly involved in cases where a patient diagnosis needs to be established. Genetic counselors have master's level training in genetic counseling and are certified by the Canadian Association of Genetic Counselors (CAGC) to practice. They provide information to patients on the inheritance of illnesses and risk occurrence; address the concerns of patients, their families, and their health care providers; and support patients and their families dealing with these illnesses. Genetic nurses are registered nurses or master's level nurses with specialized training in human genetics. Patients may be seen independently by a genetic counselor or genetic nurse unless a diagnosis needs to be established.

Also considered and included as informants for this study as part of the category "genetic professionals" were clinical genetic researchers. Although researchers do not formally provide genetic services, in the Newfoundland and Labrador context at the time

[15] See Figure 2 on page 28.

of my research they were often the initial contact that patients had with the system. Clinical genetic researchers in Newfoundland and Labrador have a rich and extensive experience studying and mapping genetic aberrations in the local communities. They serve as a conduit for translating research information to the clinical genetics program. The emphasis in the recruitment process was placed on capturing the full range of types of genetic professionals providing genetic services to patients. Therefore, a purposive sampling strategy was used. As the goal was to obtain information-rich data from as many diverse sources as possible, the study was not limited to interviewing only genetics professionals who diagnose patients with genetic conditions. The inquiry took account of all genetic professionals who provide genetic services to patients and their families. This approach was in line with reasoning by Patton (1990), who insists that "the logic and power" behind purposeful selection of informants is that a sample should be "information-rich" (p. 169).

The intention of the recruitment strategy was to engage between 7-12 genetic professionals out of all thirteen genetic providers (including the PMGP Manager) from all existing genetic services sites across the province. Thus, a maximum possible representation from both urban and rural locales as well as from diverse groups of genetic professionals was attained. Eleven genetic professionals from the province were invited via e-mail to participate in this study.[16] Table 1 (see page 17) provides additional quantitative information about the interviewees.

[16] Please refer to Appendix A: Invitation to Participate in Research Study

Table 1: Participant Profiles (N=11)

Age (in years)	Gender	Years of Experience in Role	Years of Practice in Newfoundland
20-30	Female	1-5	1-5
30-40	Female	1-5	1-5
30-40	Female	1-5	1-5
40-50	Female	1-5	1-5
30-40	Female	More than 5	More than 5
40-50	Female	More than 5	More than 5
40-50	Female	More than 5	More than 5
40-50	Female	More than 5	More than 5
40-50	Female	More than 5	More than 5
50-60	Female	More than 5	More than 5
50-60	Female	More than 5	More than 5

The reason for limiting invitations to 11 of the total 13 was that I was primarily interested in the perspectives of those GSPs who had been working with clients with the genetic conditions covered by the broader AMGGI project; my selection criteria preferentially excluded those who specialize in prenatal genetic testing.

The initial contact contained a brief description of the study, ethics approval information, my role as principal investigator in the research process and my contact information. Along with the invitation was included a copy of the consent form that had been approved through the research ethics review process.[17] Each key informant was invited to an individual interview and was asked to indicate a convenient date, time and place for the interview. The invitees were asked to respond via e-mail. The informants were invited to discuss in person and/or in a telephone interview questions about: 1) what genetic services were currently available; 2) the process of referrals and the protocols for assessment; and 3) challenges to genetic services delivery, including their perspectives on clients' challenges to access to and uptake of genetic services.

2.1.1 Interview scheduling challenges. As simple and straightforward as the recruitment process seemed at the outset, and despite it having been well designed, it presented challenges. Certainly, it was a major success that all eleven genetic professionals invited agreed in principle to participate in an interview. However, with extremely busy schedules and heavy workloads (my first insight into how under-resourced genetic services in the province are), coordinating participants' availability for interviews was a demanding task. When multiple attempts to define a precise date and time were difficult, I employed strategies such as "reminder to book" emails and follow up telephone calls. However, the most successful strategy was to tap into the cultural norms of Newfoundland – a personal encounter with the invited in the hallways of the Health Sciences Centre presented an opportunity for a chat and an impromptu casual

[17] Please refer to Appendix B: Consent Form.

reminder to schedule a time and a place for an interview. It was professionally and ethically challenging to maintain the balance between completing the interviews within a certain timeframe and not being coercive in recruitment (or worse, inadvertently deterring potential participants with repeated requests). Eleven genetics professionals were invited and agreed to participate. Of those who participated, one declined to be tape-recorded – likely out of concern for potential breach of confidentiality, although no reason was asked for or provided.

Recruitment was confined to Newfoundland because of the additional logistical and financial complexities associated with conducting research outside the province and in Labrador. Further, limiting the study to Newfoundland did provide a wide range of types of genetic services and ensured representation of different types of genetic conditions. Finally, focusing on a full range of providers within one particular province with its particular geo-socio-historical factors, rather than doing a broader but inter-provincial comparison, enabled a more in-depth look at how context shapes perspectives and decision making around genetic services. This point will be further discussed in Chapters 4 and 5.

2.2 Data Collection

Individual key informant interviews were the primary source of data collection for this study. The interviews were conducted over the course of six months and, as noted above, involved genetic service providers from the province of Newfoundland, including medical geneticists, genetic counselors, genetic nurses, and clinical genetic researchers.

Seven out of eleven interviews were conducted in person at the Health Sciences Centre in St. John's. Four interviews were conducted via telephone through the Memorial University teleconference facility. Telephone interviewing was the most cost effective method of data collection for key informants located at the satellite[18] genetics clinics across the province. Although telephone interviewing differs considerably from the in-person version, the text generated by the two methods did not reveal significant differences. The same quality and richness of data was collected from the participants who had face-to-face interviews as from those who were teleconferenced. This finding confirmed the conclusion of Sturges & Hanrahan (2004) that when distance is an issue interviewing by telephone as a data collecting method works well.

Prior to the data collection phase, I had short preliminary discussions with two of the potential informants. The intent of these encounters was to stimulate some ideas for the study design, in particular, who should be invited to participate, how to approach potential participants and how to best structure the interview process.[19]

The tape-recorded, semi-structured interviews lasted approximately one and one-half hours. At the start of each face-to-face interview, participants were provided two copies of the consent form (Appendix B). The consent form was reviewed and participants were given the opportunity to ask questions and then sign. The signed copy was returned to the investigator and the other was kept by the participants for their

[18] These are the two genetic clinics situated outside St. John's and are also referred to as outreach clinics. I provide further information on these clinics in Chapter 3.

[19] To clarify, the purpose of those conversations was not to form a hypothesis but rather to help design the study questions and streamline the interview process.

records. The informants who were situated outside St. John's were asked to fax back the signed consent form prior to the telephone interview. This procedure allowed time for the participants to pose questions about the study in advance of the scheduled interview time and confirmed that the consent process for long-distance informants was as rigorous as it was for those who participated in person.

The interview protocol consisted of 15 major question guides and probes (Appendix C). The interviews began with general background questions, including employment description, years of specialization, approximate number of clients per year, and referral process employed. These preliminary questions were followed by an open-ended, in-depth discussion of barriers to access to and uptake of genetic testing. Participants were invited to comment further on their beliefs and attitudes with regard to the process that they themselves followed for ensuring appropriate access to and uptake of genetic testing. The probing techniques employed promoted a coherent and accurate account of the participants' perspectives. The use of probing has been favoured in qualitative health research (see for example, Britten, 1995; Patton, 2002; Kvale & Brinkmann, 2009). At the conclusion phase of each interview, participants were given the opportunity to discuss additional issues that they considered relevant to the study and to pose further questions.

2.3 Ethics

Research ethics approval for this study was granted by the Human Investigation Committee, Memorial University of Newfoundland. In addition, regional health

authority approval for the study was obtained from the Research Proposal Approval Committee (RPAC) of Eastern Health for those participants who were employed by the Eastern Health Regional Authority in Newfoundland. Each interview file (paper and audio) was coded to secure the confidentiality of the information and to guarantee the anonymity of each informant. The electronic version was stored in a password protected computer file. Audio tapes, electronic back up and paper interview files (transcripts) were stored in a private locked cabinet. Signed consent forms were also securely stored in a locked cabinet separate from the audio and paper files.

2.4 Data Analysis

In order to generate findings that transform raw data into new knowledge, I engaged in active analytic processes throughout all phases of the research. The analysis phase involved the convoluted task of discerning meanings within the data. As I went through the process of analyzing the data, I found, as Thorne (2000) had observed, that there was no "sense of mystery and magic" in the process.[20] On the contrary, it was quite straightforward. In truth, I merely attempted to make a "convincing analytical claim" based on what I believed I understood the informants to mean.

All interviews were transcribed in confidence by a professional transcriptionist. The average length of an interview transcript was 18 pages, totalling 163 pages. Interview transcripts were read entirely four times: first, to acquire a broad overview of the complete interview data; second, to identify key words and phrases that define the

[20] Thorne's (2000) own observation was in reference to the language used by some authors describing the process of analysis in qualitative research.

experience described by the interviewee; third, to interpret the meanings and develop key concepts/categories; and finally, when no new insights seem to emerge, to organize the categories into main themes. As well, the transcripts were continuously reviewed to allow fine-tuning of the categories for a precise and complete account of the experiences studied.

During my initial reading of the transcripts, I was able to mentally note a number of recurring terms such as patient referrals, geography and operational capacity. In the second reading, I highlighted those and other key words and phrases. With the subsequent (third) reading, I started the coding process by penning down emerging concepts in the margins of the transcripts across from the consequential word(s) or phrase(s). Although the labeling of the categories further evolved, this was an important phase that allowed me to cluster similar concepts under a common caption, that is, to organize the categories into main themes. The process I employed followed the method for interview analysis described by qualitative research authors such as Strauss (1987) and Smith (2003).

The interview transcripts, once analyzed and organized into themes, revealed important insights about genetic professionals' opinions on genetic services and unearthed information on challenges to accessing and using genetic services. Their perspectives are presented throughout my results section in quotation marks, and include direct or indirect quotations. The intent was to allow generous room for the participants' voices to be heard and have their thoughts dominate the text. In order to keep

participants' identities confidential, given the small size of the community of genetic professionals, only identification numbers are used to describe the interviewee; any other descriptors that would normally be used to contextualize the speaker (place and type of work, role, professional training, professional status, affiliation) are not employed, as such details would inevitably expose the interviewee. As well, portions of quotes that reveal a clinic site or other details that could inadvertently identify the participant have been omitted. Finally, my own written commentaries have been carefully edited to minimize the risk of participant identification.

The analysis revealed two broad themes - systemic and psychosocial barriers. The category of *Systemic Barriers* includes aspects of the current genetic care delivery that were perceived as barriers by the genetic providers I interviewed. *Psychosocial Barriers* are attributed to the patients' experiences as perceived and interpreted by the interviewees. Economic, political, and socio-cultural contexts shape these two types of barriers.[21]

In the next two chapters, I discuss the themes raised by the informants. Systemic Barriers are presented in Chapter 3, followed by the discussion of Psychosocial Barriers in Chapter 4. Included in each theme are subthemes reflecting a wide spectrum of issues and concerns addressed during the interviews. The genetic professionals' perspectives (mainly *in vivo* quotations) lend color and authenticity to the discussion while creating focal points for my interpretative comments and analysis.

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[21] These contextual factors shaping the barriers are of course not just relevant to genetic services but rather are broadly accepted determinants of health (see National Collaborating Centre for the Determinants of Health, 2011; World Health Organization: Commission on Social Determinants of Health, 2008)

In keeping with the essence of literature review, woven into the thematic reporting are comparisons and contrasts with previous research. Although the literature review component was consolidated into a brief overview in Chapter 1 (Introduction), given the wide range of themes and sub themes debated in this study, the overview of significant literature is primarily spread throughout the two results chapters and integrated into each theme-specific discussion for more immediate context and critical assessment.

Chapter 3: Systemic Barriers

I use the term "systemic barriers" to encompass practices or situations in the current genetic care system which significantly limit or unfairly exclude certain patient groups from accessing genetic services. These systemic barriers are of course interconnected: they shape and are shaped by each other and all are embedded in the geographical, economic and political contexts of Newfoundland. In this section, however, I separate them out in an artificial way in order to explicate each. Although there was a wide range of systemic barriers identified by the genetic professionals I interviewed, there was a consistent and strong common theme – lack of economic resources.

3.1 Barrier #1: Geography

Not surprisingly, geography was emphasized as playing a dominant role in the way that health care services are allocated and delivered in the province of Newfoundland and Labrador. Historically, the natural characteristics of the Newfoundland and Labrador coastline and the overall ruggedness of the terrain had dictated the pattern of populating the province (see Figure 1 on page 27). Larger settlements were formed in areas that were most easily accessible. Predictably, the density of the population in those areas had increased over the years and transformed specific communities into geopolitical and economic centers of the province; other, more remote areas remain hugely under-populated.

Source: Based on Natural Resources Canada map[22]

Figure 1: Map – Province of Newfoundland and Labrador

[22] adaptation.nrcan.gc.ca
http://www.google.ca/imgres?q=newfoundland+and+labrador+population+density&hl=en&biw=1156&bih=478&gbv=2&tbm=isch&tbnid=IQ0z6e02JbrbaM:&imgrefurl=http://adaptation.nrcan.gc.ca/assess/2007/ch4/1_e.php&docid=qXicbflJC1rGBM&imgurl=http://www.nrcan.gc.ca/sites/www.nrcan.gc.ca.earth-sciences/files/jpg/assess/2007/ch4/images/fig1b_e.jpg&w=440&h=552&ei=v5YNT5eeEaLi2QWl_pybCA&zoom=1&iact=hc&vpx=252&vpy=109&dur=93&hovh=252&hovw=200&tx=114&ty=225&sig=114209860091680651569&page=1&tbnh=137&tbnw=109&start=0&ndsp=12&ved=1t:429,r:7,s:0

The map (Figure 1) clearly illustrates the uneven settlement patterns. In addition to the unbalanced settlement density, the populace distribution is heavily congregated around only a few centers. Naturally, the location of health care facilities and expertise follows the demographic and economic clusters.

The Regional Integrated Health Authorities[23] in the province have been established around those strategic centers. The map (Figure 2 on page 29) depicts the location, boundaries and population served for each health region. It also indicates that the configuration of the regional health authorities is intended to ensure effective servicing of each region, clustered around the major urban centers.

Nonetheless, local geography, characterized by vast and unevenly populated territory, contributes to the unequal distribution of health care resources, including human resources. For example, as of 2012, there is only one center for radiation treatment on the island portion of the province, located in the capital city of St. John's, where close to one third of the province's population resides. This is the situation with most health related services: even within the most densely populated of the health regions, the Eastern Health region servicing St. John's and area, many patients must travel long hours to appointments. Since genetic services are offered as part of the public health care system (as opposed to privately owned clinics), these services, too, are subject to resource

[23] In 2004, the 14 existing health boards of Newfoundland and Labrador were centralized into four Regional Integrated Health Authorities (Newfoundland & Labrador Department of Health and Community Services Annual Report, 2005, p. 5). The Eastern Regional Health Authority is the largest of the boards, serving a population of 290,000 (Eastern Health Newfoundland and Labrador, 2012).

limitations that the geography of the province has shaped. One research participant summarized:

> It is difficult to access [genetic care and other care] if you are outside the St. John's and Avalon Peninsula area. If you live on the Northern Peninsula, you have to go to St. Anthony or to Corner Brook. [#7]

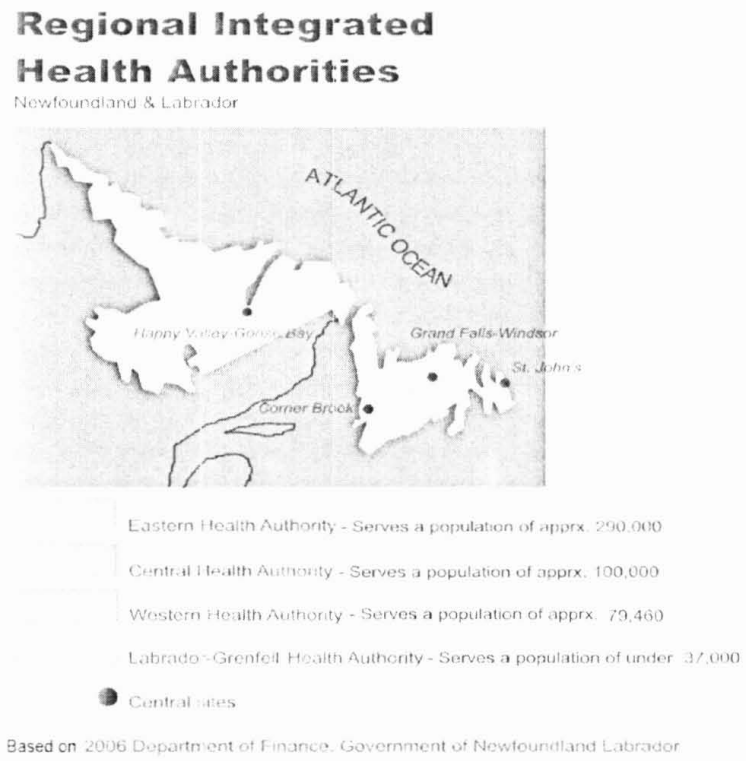

Figure 2: Map – Regional Integrated Health Authorities, Newfoundland and Labrador

The unique geography of the province, characterized by very small and remote communities located at times several hours away from the nearest urban center, is challenging for the delivery of health care. Genetic services are no exception. The map (Figure 3, below) shows the location of the PMGP genetic clinics in the province in relation to the health authorities' catchment regions.

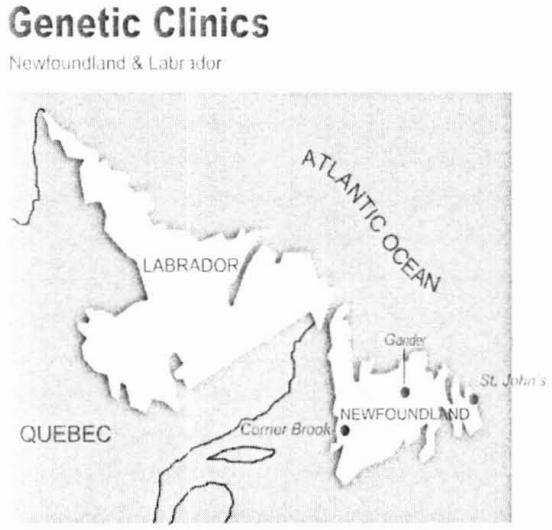

Figure 3: Map – Genetic Clinics, Newfoundland and Labrador

The main PMGP genetic clinic is situated in St. John's, while satellite[24] clinics have been set up, one in Corner Brook and one in Gander. There is a genetic research

[24]Each satellite clinic is served by a nurse trained in genetics. A medical geneticist from the main site in St. John's conducts a clinic (with a duration of one to two weeks) in the satellite (outreach) locales twice a year. These satellite clinics are referred to by the genetic professionals as "travel clinics".

nurse in St. Anthony; however, this position is not associated with the PMGP[25]. The selection of genetic clinic sites followed the same underlying principle employed with regard to the four health regions: the need for accessible and efficient health services. Map #3 (see previous page) illustrates the large and diverse geographic area covered by each genetic clinic in the province. In addition to the permanent satellite genetic facilities, travel genetic clinics (administered by medical geneticists) have been introduced as a supplementary service in these locales.

The island of Newfoundland[26] is characterized by inclement and inconsistent weather patterns for most of the year; therefore, travel genetic clinics are limited to two sessions per year. The efficiency of those clinics is additionally limited by the inability of clients to travel - poor road conditions, especially during the winter season, or lack of reliable public transportation (linked also to weather and geography) deter clients from consistently attending appointments. Genetic service providers emphasize that for patients living in remote or rural areas, traveling to and from the nearest genetic clinics also imposes arduous arrangements, including child or elder care for those left at home for short or extended periods of time. Geography places extra burdens, including financial burdens, on most individuals who need to be seen by a geneticist.

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[25] As of 2012, there are two Community Genetics Program (CGP) facilities, one in St. John's and the other in Grand Falls-Windsor, with a primary focus on colorectal cancer. Each location has two genetic nurses on site, and the two share one genetic counselor, stationed in St. John's. The CGP has been funded solely through research.

[26] The genetic clinics are established only on the island portion of the province of Newfoundland and Labrador.

> I think the biggest issue we have is geography. If we have somebody in the Northern Peninsula in Trout River with a population of, I don't know, 112 or whatever it is, to try and get those individuals down to Corner Brook or Gander for a genetic counseling session is very, very difficult. This means that person who's in Trout River has to spend two or three hours traveling to the clinic and two or three hours traveling home. [#6]

> I guess a big barrier is that there are so many people in Newfoundland who don't live close to St. John's and getting there is a big deal for them. [#2]

> Just the distance - that's another barrier. You know, there will be always people who live far away. [#1]

The impact of geography on access to genetic services is of course not unique to the province of Newfoundland and Labrador. For example, in the Canadian context, d'Agincourt-Canning and colleagues have identified the same concern for rural and remote areas of British Columbia (d'Agincourt-Canning et al., 2008, p.554).[27]

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[27] Geography also has an impact on the provision of medical care in general. For example, in the United States, Wennberg & Wennberg (2000) have identified geography as a challenge to provision of medical services for the state of Michigan and concluded that the pattern is prevalent across the United States as a whole. They report, too, that the majority of Americans tend to avail to services of physicians, whose practices are nearby to provide appropriate level of care. Cromley & McLafferty (2002) also demonstrate that geography can impede access to and use of health care services.

My findings suggest that access to efficient genetic care as well as uptake of the services in a consistent manner is immensely influenced by geography. The extent of this influence may gradually diminish over the next few decades due to the process of urbanization as well as demographic changes taking place in the province. Meanwhile, further capacity building of the clinical genetic services across the province is needed to ease the burden of distance and travel time on access to genetic care.

To summarize, geography is a powerful barrier to access to and uptake of all health and social services, and genetic services are no different. Geography permeates the range of challenges identified by the research participants. Although I artificially differentiate geography as a barrier, for clarity in discussion, in fact it underlies all of the barriers identified through the interviews.

3.2 Barrier # 2: Lack of Family Physicians

The principle foundation of the public health care system is that resources will be justly distributed and easily accessible. In reality, however, there are individuals and families who are unnecessarily excluded in a context of limited health care resources.

Insufficient access to family physicians was emphasized by genetic professionals as being a significant barrier to referral to, and therefore access to, genetic services.

> It would be family doctors who recognize that several members of the family probably have the same condition. And very frequently, I find

> patients do not have a family doctor anymore because the doctor left [the community]. [#1]

A portion of the population in the province does not have a family physician, either by "choice or by circumstance" (Primary Care Advisory Committee, 2001, p. 10). Residents of rural Newfoundland and Labrador are more likely not to have a regular doctor compared to residents of urban areas (Mathews & Edwards, 2004, p. 166). Genetic professionals reported that this is a major barrier to accessing specialty care, including genetic services, since access is largely achieved through a referral mechanism. Family physicians are strategically positioned to connect patients with the needed health care expertise and service. If family physicians are not in place, the "orphaned patients" end up seeing a variety of health care providers, which may result in inconsistent medical records and disconnected care (Primary Care Advisory Committee, 2001, p. 10). Because of the geographic and social isolation associated with physician practices in the remote areas of the province[28], the turnover rate of medical professionals is consistently high. To minimize it, the provincial government has invested in and implemented various recruitment and retention strategies; however, ensuring their long-term sustainability is an issue[29]. The province is characterized by low birth rate and outmigration and diminishing economic vitality[30]. Supplying and retaining physicians in remote and rural

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[28] Communities in those areas are scattered and often average 200-500 people.

[29] Newfoundland and Labrador Department of Health and Community Services (2007, January 16). *News Release*. Retrieved from http://www.releases.gov.nl.ca/releases/2007/health/0116n02.htm

[30] These characteristics are especially pronounced after the collapse of the traditional fishery in the province in the 1990s.

areas poses challenges not only in this province (Mathews, Edwards, & Rourke, 2007) but in other Canadian provinces (d'Agincourt-Canning et al., 2008) as well as in other developed counties such as Australia (Kamalakanthan & Jackson, 2008), Norway (Straume & Shaw, 2010), the USA (Hawkins & Hayden, 2011) and worldwide (World Health Organization, 2010).

A related issue reported by participants was the lack of consistency of family physicians - that is, even for patients who did have access to a family physician, the high turnover rate of physicians can mean that short-term physicians are not sufficiently informed about the specific genetic disorders for which families of the region may be at risk.

> I think one of the difficulties is that there is such a turnover of rural doctors. Members of the family, you know, have been so pleased to have a screening protocol and they've taken it to their family doctor. And then, the next time they come in – six months later – the family doctor is now a new family doctor. Could they have another copy of the recommendations I just sent to the previous doctor? So, this turnover of health care personnel is a difficulty. Due to the very nature of genetic diseases, the clusters tend to be in the smaller communities. If there is a new person [physician] there, the family members have a difficult time receiving a good follow-up, and they are the ones who need a family doctor who

> knows what the problem is in their family, but the doctors keep changing. [#5]

Interestingly, the literature on the knowledge deficit of family physicians has emphasized lack of sufficient education in genetics (Prochniak, Martin, Miller, & Knapke, 2012; Klitzman, 2009); by contrast, my research found that the greater concern was the lack of personal knowledge of the families of the given region and of the genetic diseases, which characterize the community.

A second concern expressed by genetic professionals was the potential for a lack of appropriate referrals by other specialists. Referrals to genetic services are not only made by family physicians, but are also made by specialists. For example, if a family physician refers a patient to an oncology specialist and the oncologist suspects a genetic mutation, he/she may further refer the patient or family to genetic services.

> Well, I think there is a big subset of specialists who don't refer. [#3]

> So in Newfoundland, a lot of times it's the family doctors who refer, and probably less often, it's the specialists. [#2]

While some specialists have experience with referring patients to genetic services, there remain significant problems with the level of knowledge about, as well as attitudes

toward, population genetics among non-genetic medical professionals serving rural and remote areas.

3.2.1 Location of genetic clinics. According to the genetic professionals I interviewed, the way genetic care is situated in the province results in imbalanced access. For example, as of 2011 there are no genetic clinics in Labrador. Patients residing in that region of the province have to travel to the Corner Brook genetic site (see Figure 3 on page 30) at those times when the travel genetic clinic is being held. In order to see a medical geneticist during the twice-yearly genetic clinics offered at the Corner Brook or Gander site, patients must travel either by ferry and road or by air and road.

> You know, [in terms of] access to the person who lives 10 hours drive away -- service is not available at the same level to you or I who get our appointment and walk in to have it done. If you have to have all services equal, access should include that fact that Newfoundlanders are spread out over a huge area. So, there has to be some way to equalize this cost to do the visits. I think this is one thing that has to be worked into how you deliver services. [#5]

This lengthy commute in inclement and at times unpredictable weather conditions is not always a feasible or sensible option for patients living in rural and remote areas.[31] Clients categorized as semi-urgent or urgent typically need to travel long distances and

[31] Nor is it always a sensible or feasible option for the two medical geneticists serving the province. They are stationed in St. John's and travel twice a year to Corner Brook and Gander respectively, for a week-long travel clinic.

frequently, whether to the genetic site servicing their area (with choices of schedule limited to times of the medical geneticist's visit) or to the central site in St. John's. That is why the participants felt that "there's not equal access to everybody for genetic consults and care." [#8]

Presently, a permanent clinic in Labrador seems unlikely to be feasible because, as research participants explained, the number of genetics professionals in the province is limited and their workload is quite sizable. This means that even if resources were available for infrastructural support to set up a clinic in Labrador, there are insufficient personnel.

Concerned about the unbalanced access to their services, the genetic professionals interviewed brought forward ideas about improved access: establish travel clinics to Labrador and other remote regions and increase the number of medical geneticists at the St. John's site to allow more frequent outreach clinics. Reflecting on the issue, one participant pointed out that, as a start, a reasonable solution would be to have a mobile facility offering genetic services in the Labrador region of the province over the course of several days, at least once a year.

> I am thinking that for instance, instead of having the people of Labrador come to us, may be we could travel to Labrador. That way, we could do a clinic in the community. [#9]

Another participant suggested,

> It's almost easier if we had a traveling clinic, you know, and the traveling clinic would do the Northern Peninsula at this point in time and the west coast at another. [#6]

However, the province remains a place that has "too few people scattered out over such a huge territory." [#1]

> "I am not sure that we're ever going to [find] a way...there's always going to be a section of the population that's going to drive or spend time getting in to appointments." [#8]

This observation echoes the findings of other researchers (Evans, Whitehead, Diderichsen, Bhuiya, & Wirth, 2001; Starfield, 2006; Hawkins, & Hayden, 2011) reporting on the inevitability of unfavorable health outcomes for certain pockets of society, especially those residing in rural locales. Given the fact that the rural component comprises 42% of the population of Newfoundland and Labrador (Statistics Canada, 2006), compromised access to health care seems to be the norm rather the exception. Irreversible trends toward urbanization combined with out-of-province migration have plagued the rural communities of Newfoundland and Labrador in many ways, and limited access to health care is increasingly the reality. Reflecting on the challenges of providing genetic services in rural and remote communities, Hawkins & Hayden (2011) labeled access to genetic care "a major barrier" to the democratic distribution of health benefits effective clinical genetics can bring (p. 197).

3.2.2 Technological innovation and rural and remote health service delivery.

In an effort to remedy the imbalanced access to genetic care, an innovative service delivery method has been instituted - telemedicine genetic sessions with patients from rural and remote areas of the province. For example, genetic consults for clients who reside in Labrador are occasionally conducted by telephone. Patients from Labrador travel only to a designated hospital office in Goose Bay (Labrador) where a nurse facilitates the telecommunication session with a medical geneticist in St. John's. Asked to describe their telemedicine service experience, one participant commented:

> I wouldn't say it is equal to seeing patients. It's way better in person. Connection with the patients is a lot more difficult over the phone. So I would definitely say face to face is better: but for financial reasons, we will do it over the phone. [#1]

Novel technologies such as the use of telemedicine are a welcome advancement and, in the context of clinical genetics, they bring the promise of improved access to services and better health outcomes.[32] Ironically, the validity of genetic tests and the safety and effectiveness of new therapies are improving while access to them remains disproportionate.

3.2.3 Accessing clinical care through research. Clinical genetic care in the province was originally initiated through research and was operated solely on research

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[32] Hawkins & Hayden (2011) forecast a "pervasive" access problem amplified by the introduction of new genetic therapies and technologies. While the association between new genetic technologies and access to care was not explicitly discussed by the participants, it was a subtext running through accounts of the centralization of genetic technologies.

money. Currently, the delivery of the services is fully funded by the province and available at no charge to clients; however, it remains closely associated with genetic research activity. Some of the interviewees reported that access to genetic care and related medical and/or genetic tests are significantly expedited if clients become part of a research study. In other words, a genetic research study can facilitate "queue-jumping" for its research participants. Despite the fact that there are notoriously long wait lists (and wait times) for many patients, swift service is possible for those who access testing as research subjects, because research (rather than the provincial medical care plan) pays for the testing services and requires timely results in order to meet project deadlines and justify funding. In fact, one genetic professional mentioned that for clients who are research subjects there may be no wait time at all to avail of the testing. Similarly, another respondent commented,

> It's probably unfair, in that people who participate in research studies generally get the test done faster. They generally get information back faster. So, if we need to have a test, like an MRI, done then we will probably do that after hours. You don't have to wait six or eight months in queue with everybody else. Our patients will actually have their testing done a little faster because we're being billed for it and not MCP. And I often say to people – find a genetic study, get into it, which is not fair, but. [#6]

It is worth noting that not every individual with a genetic condition is aware of the existence of genetic research, including the "benefit" described above. A client may have a genetic condition that needs attention at a time when no appropriate genetic research is carried out. Alternatively, a client may not be willing to participate in a research study. In other words, expedited access to genetic services through research may offer certain benefits; however, it is not an option for everyone.

It was apparent from the interviews that despite the customary argument about deficient health care resources, timely, even expedited, access to care is achievable if patients participate in research. A research study with sound funding, which allows the research subjects to be paid for participation, can be especially alluring, and participants may complete a medical procedure (for which the usual wait time is months, if not years) in a much shorter period of time. Of course, it could be argued that the number of genetic research participants is insignificant in comparison to the overall number of those requiring genetic testing and that the benefit to the society produced by the genetic research can justify these practices. Although this practice – encouraging clinic patients to enter into a research study to expedite results – may be beneficial for the researchers and definitely for some research subjects, it is a departure from the principle of universal and just access to medical care. Moreover, this practice may have effects that are worth monitoring: most obviously, patients may be being coerced into research. Furthermore, as beneficial and expedient as the practice of research-related entry in the genetic care system may be, there are some troubling aspects to this practice that were highlighted by genetic professionals. One participant succinctly summed this up:

> Lots of genetic stuff goes on as part of research projects that does not provide right clinical care for the patients. [#7]

This genetic professional explained that once patients are seen for the purposes of a research study, they may become forgotten. The genetic condition, however, remains with the individual or the family and needs to continue to be addressed beyond the completion of the research study. In order to fill in the gap in clinical care, the patient must secure a genetic service that is able to pick up those clients and follow up with them.

In contrast to this observation, another genetic professional stated that the cardiac genetic clinic, which has been created as a result of genetic research, provides follow up care to all cardiac patients regardless of the way they have accessed the system – through genetic research, or through the conventional referral mechanism by family physician or medical specialist. This comprehensive genetic care however is not available to those with other genetic conditions.

One respondent highlighted the mechanics of enrolling patients in clinical genetic research:

> If there is nothing else available clinically, we send it to research. But that's not quite true. If there is nothing else available clinically or if it is too expensive for our budget to pay for, then we'll offer them [patients] research. Whenever we assign someone to research genetic testing, we tell the client that it's probably going to be about three months, but could be

years or never. You know, we never give guarantees with research testing. [#1]

The respondent further explained that usually clinical testing is offered where there is a gene identified in the family. However, if a gene is newly identified or rare and consequently not available in a clinical laboratory, or if available but extremely expensive, then testing is arranged through research. Nonetheless, this participant also noted that "not a very high percentage of their patient population" [#1] is offered this arrangement.

Echoing her colleague's comments, another participant noted that "research patients" become well aware that genetic testing via research is the only clinical alternative they have at that moment. In those cases, the genetic professionals make sure that their patients fully understand that research testing is likely associated with undetermined results. This means that although some clients may have queue-jumped to access genetic services (and even have their genetic test completed) the results of this test may not be received expeditiously and the wait time can be indefinite. In other words, while queue jumping via research is an attractive (and often irresistible) option that can expedite being seen in genetic clinic and may expedite the testing process (non-genetic and genetic testing), receiving genetic test results is far from swift. Understandably, this is something frustrating for patients/clients who had entered the system and jumped the queue via research participation.

> And they [patients] know that that's [genetic testing, not treatment] the only option they have. So it's better than nothing kind of thing. [#2]

These comments made it apparent that there are some negative effects of blending research and clinical care. The genetic service providers I spoke with raised concerns about the quality and efficiency of genetic care when offered under the framework of genetic research. They expressed concern that the perceived promise of quick access to care (that is, the queue jumping which clients assume to be a benefit of participating in research), does not in fact translate into the expedited receipt of genetic test results or more efficient genetic care in terms of treatment or cure. On the contrary, the wait for results can be tedious and frequently indefinite, as it is dependent on whether or not appropriate genetic testing methods and techniques become available. In other words, genetic research may offer a quicker access to the system, but it does not always guarantee quick results.

> Sometimes you have to wait for the research to get better to be able to actually give them [patients] results. I can think of one participant, who I think I saw first in 1998 and eight years later we were able to give her definite result. [#7]

These findings support the view that the presumed duty to communicate genetic research results to participants is problematic[33] (Knoppers, Joly, Simard, & Durocher, 2006). The challenge stems from the nature of the human genetic research, as its results

[33] Unless the information is reliable and clinically significant.

are usually of unknown or uncertain predictive value, and it is not meant to address the genetic status or other health issues of individual patients (Bioethics Advisory Committee, 2005). The genetic professionals I interviewed noted that those issues are addressed in the consent process, but 'research patients' have various levels of understanding and expectations concerning their research participation.

Yet another concern with using research as a way to expedite access to testing for individuals is that research funding is not secure. As with any research, genetic research is dependent on the availability of funding, and securing continuous financial support for research is not guaranteed. This means that the clinical needs of clients can be met through research only as long as money is available. Once research funding is utilized, clients who have been enrolled in a study may be "left in the lurch" [#7], waiting for years to receive genetic test results. When asked if patients who waited for a long time for genetic test results received any treatment or other services in the span of those years, this respondent [#7] explained that those patients continued to have regular clinical appointments, but those appointments are merely to keep communication going while awaiting results, a frustrating experience for patients/clients. The genetic professional then took the opportunity to argue for the importance of blending clinical practice with research:

> That is why the research has to link with clinical [care], because if you were doing research per se and it wasn't anchored with clinical [care], those patients would be sitting out there for eight years not knowing. They may be affected. You just don't know because you did not have the right

> results. You could not work it out. You can't leave people out there worrying that they might have it [a defective gene] because the lab can't decide because of just the way the testing is. You have to provide something. [#7]

This example was provided by the participant to highlight the importance of communicating clinically significant research results to clients. However, the example also suggests that much of that communication may entail regularly bringing patients into the clinic to provide them with an update, even when that update is repeatedly on nothing else but incomplete research results. Blending research and clinical care may divert valuable resources, which can be utilized for other, more pressing cases. Miller, Giacomini, Ahern, Robert & de Laat (2008) report similar findings in the province of Ontario and further add that eligibility criteria for participation in research do not always coincide with clinical criteria. Potentially, the entry-through-research practice may inadvertently filter out individuals who are a clinical priority. While it may seem that it makes sense to link research and clinical care (since that way research knowledge is directly translated into clinical practice and issues arising in clinical care can be directly addressed by research (Hodgkinson et al., 2009), the observations of some of the genetic processionals contradicted this logic. Samuels et al., (2008) argue that the line between research and clinical diagnosis is necessarily 'fundamentally blurry' and 'fluid' and extend this understanding to their patient practice (p. 386; see also Pullman & Hodgkinson, 2006). In arguing for the importance of keeping those boundaries blurry, however, they distinguish between genetic research findings that are unquestionably

related to clinical genetic care, those that are 'possibly genetic care' and those that are 'definitely research'. My findings suggest that, in keeping with that distinction, careful attention must be paid to ensuring that clients themselves are not inadvertently misled by blurred distinctions between genetic testing conducted primarily for research purposes and testing conducted primarily for the clinical care of the individual being tested. The concern expressed by genetic professionals I interviewed is that patients are using genetic research as a means to queue-jump and receive expedited clinical care. Patients are disappointed when they find out that there is not definitive answer about their risk status or when wait times for receiving results are undetermined. Still unexplored is the question of whether the perceived promise of quicker results may in fact be a kind of implicit coercion to participate in research, a topic that deserves study.

3.3 Barrier # 3: Cost

There is a high cost associated with establishing and operating genetic clinics. The PMGP is no exception, especially given the resource implications of access to rural and remote communities. Importantly, however, the costs to the public health care system of providing genetic services are not the only financial implications of a provincial genetics program.[34]

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[34] A thorough examination of the costs (and benefits) to society as a whole, and in particular to a province or health region, that result from the establishment of a genetic service program within a system of socialized medicine such as Canada's, is an important topic that deserves careful study. Such an account is beyond the scope of this thesis, which touches on the topic only in terms of "overall costs to society" being raised as an issue by the genetic professionals I interviewed.

The provision of genetic services requires a significant amount of time and resources. Services are relatively new and time intensive; they include genetic testing, genetic screening and genetic counseling (pre- and post-test). In the clinical setting, genetic tests can help detect gene variations associated with a specific disease or condition. Genetic tests can be performed to validate a suspected diagnosis. Predictive genetic testing highlights the possibility of future illness or an individual's response to therapy. It can also be used as a tool to determine the carrier status of unaffected individuals, indicating whether their children may be at risk. There is a range of costs associated with genetic care: individual cost, cost to the province, societal cost, and cost for research and development. Although the literature on cost is limited, there is a consensus that the delivery of genetic services requires considerable expense (Lawrence et al, 2001; Phillips, Veenstra, Ramsey, van Bebber & Sakowski, 2004). While the cost of genetic testing is high, it is relatively small compared with the other aspects of genetic care services such as surveillance, prevention and treatment costs (Morgan, Hurley, Miller & Giacomini, 2003). Cost of providing genetic testing services was a key theme running through the interviews I conducted.

> Financially, genetic testing can only be done through the main site which has a budget for this. MCP[35] doesn't necessarily pay for it. You can't just bill MCP. It has to come through the St. John's site. So, the test has to be

[35] MCP is the acronym for Medical Care Plan, the term used in Newfoundland and Labrador to refer to the provincial Medicare program.

> deemed appropriate. That's why everyone has to be seen by someone in St. John's before they are eligible to get that genetic testing done. [#2]

> The genetic testing for the cancer gene is extremely expensive. It's only offered through one laboratory in the US, which patented the gene, so no other lab can do it. [#1]

> It would be cheaper if we could do them [genetic tests] in-house but we don't have lab resources. We do not have the technologies. Our diagnostic lab does genes that are commonly taken from our population. At least 50 percent of our molecular genetic test is outsourced to other Canadian labs, American or European labs, wherever we can get the cheapest test. [#8]

As mentioned earlier, the PMGP is closely associated with genetic research activities. Often, the type of genetic testing that is required and the specific genes being tested dictates whether testing in a research setting is possible. The majority of participants indicated that they have collaborated on a number of research projects carried out by molecular or clinical genetic researchers. This alliance proves beneficial, especially when genetic testing may constitute part of gene hunting. The respondents also commented that although the research-related testing has no immediate clinical benefit to patients, it could lead to gene discovery. The respondents also commented that the

process of searching for a gene is time consuming and costly, with no guarantee that there will ever be information of clinical relevance coming out of the research. One of the interviewed genetic professionals indicated,

> No genetic service really has enough of time and money and certainly Newfoundland does not. [#6]

However, when gene hunting is successful and the benefits to individuals are obvious, then the relationship between research and clinical care is valorized and its importance re-confirmed. A good example is the identification of the gene causing ARVC[36]. Local families known to carry a mutation are now tested and provided follow-up care, thanks to a successful research study.

The respondents also explained that once it is established that there is a certain mutation in a particular family, it is easy to test other family members to determine whether they have inherited the same mutation or not. This process is relatively uncomplicated and not as costly in comparison with cases where the exact mutation in a family with hereditary conditions is not identified. Although less costly, the latter process still requires significant amount of funding, especially long term funding, to secure its smooth operation. The genetic professionals commented that it may take years before a mutation is "worked out."[37]

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[36] In 2008 the genetic research team at Memorial University of Newfoundland led by Dr. Terry-Lynn Young identified the gene causing the condition (see Merner et al., 2008). The discovery effort was part of the AMGGI objectives.

[37] To clarify, this is the term used by key informants.

As treatment for specific genetic conditions is not always available, genetic screening and/or testing are not always cost effective. Petersen, Brensinger, Johnson & Giardiello (1999) discuss the significant cost associated with genetic testing for hereditary forms of colorectal cancer, in particular. They suggest that the cost advantage for gene testing increases as the size of the pedigree[38] and number of at-risk members increases. In the case of colorectal cancer, genetic testing is not relied on for screening or diagnostic purposes. Nevertheless, it may be considered appropriate for high risk families to establish the possibility for developing certain forms of colorectal cancer.

Genetic testing is labor intensive and requires expensive equipment. As well, the testing has to be appropriate for the patients who qualify. The genetic professionals I spoke with pointed out that the high cost prevents them from offering timely service to everyone eligible for genetic testing:

> Due to cost, we do not have access to timely testing. [#1]

> Sometimes the criteria are there, but we have to find families that will most benefit from the testing. Do you run the risk of missing a few families? Yeah, you probably do, and it's a constant. [#3]

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[38] A pedigree is a diagram of the genetic relationships and medical history of a family, using standardized symbols and terminology. The pedigree shows the relationships between family members and indicates which individuals express or silently carry the genetic trait in question.

Although genetic professionals are vigilant in creating and refining their criteria for testing, they are limited by cost and lacking resources in their practice. These limitations are passed on to their eligible clients.

> We generally sign up people to research projects when there is no clinical testing available. As we can't buy the test our next best option is to enroll them in a research project. [#1]

Research labs tend to perform testing for individual patients when ordered though a genetic professional. However, a test may provide inconclusive results and may require multiple family members to participate. In addition, if genetic testing is done in the context of a research study, testing results may not be available for many months or years, and sometimes they do not become available at all.

> If we are trying to find the mutations in a family and if there is no known mutation out there ... we are looking for that needle in a haystack, those are the ones that take the longest. [#6]

Participants also explained that there are not enough people working in genetic research labs, due to scarce or inconsistent funding for searching for gene mutations. Funding is more likely to be available to perform tests once a genetic mutation is known.

An important aspect of the cost of genetic services to patients is the cost of genetic counseling. Genetic professionals invest significant time in providing counseling to clients as part of the pre-test orientation as well as the disclosure of test results. The

professionals I interviewed emphasized the time consuming nature of their service, which includes triaging, preparation and delivery of a consult session, communicating genetic information to clients, conducting follow up by telephone or in person, and documenting the consults. The professionals discussed the time and effort involved in tailoring individual counseling sessions to the specifics of each case, even for counseling individuals within the same family.

> ...you look at the referral and you you'll think – I need to run after this a bit more and you will call them [the clients] and you'll get some more information over the phone and you'll hand hold a bit more. [#3]

> And so, you just take any person as they come and try to see what their feelings are and try to help them. [#2]

These excerpts are in line with previous findings demonstrating that providing indepth information that is specific to each presenting member of a family is an important part of the counseling process; however it is associated with time and cost (Petersen et al., 1999).

The genetic professionals I interviewed also emphasized that for some genetic professionals it is not only the time taken to see clients, but also the time to travel to see clients in remote and rural areas, which adds to the overall cost of providing genetic counseling services.

In their quantitative study, (Lawrence et al., 2001, p. 479) demonstrate that genetic counseling is costly in terms of personnel time, as it averages three to four hours per client for a counseling session. Their findings further reveal that this cost, although high, is insignificant compared to the cost of genetic testing and disclosing results.

Only one research participant felt uncertain about the advantages of providing patients with information about genetics and genetic care in relation to the overall high cost of the care. One aspect of the dilemma revolved around the fact that certain diseases, cancers in particular, are not always genetic-based. The concern was that the cost associated with genetic services delivery is consequential, yet an insignificant percentage of clients can benefit from those services. The comments specifically referred to breast cancer cases seen at the genetic clinic.

> A genetic condition is only a very, very small part of that whole illness profile that any one person might have. Here's an example: Breast cancer is only 15 percent of cancers that have a genetic reason. Yet, you know, we don't see that 15 percent. We might see, well, 1 or 2 percent. [#4]

The research participants did not mention anything about the cost related to patients who, due to a poor or incomplete preliminary assessment and referral process (including self-assessment and self-referral), present themselves at the genetic clinic and undergo further investigation when not necessary. Although the issue was not brought up during the interviews, it is worth noting the findings of Reis et al. (2006), who emphasize the "substantial cost" associated particularly with low-risk patients (true low-risk, not

false) referred for genetic consults or screening procedures. These are classic cases of "little return" that add up to the already high cost of genetic care.

The genetic professionals reiterated that the lack of adequate resources infuses every aspect of their services. They admitted that at times it is a challenge to keep operations aligned with nationally accepted standards. Although they did not provide specifics, their commentary is direct and clear.

> I think the problem is that in some cases we really do not keep up with the national standards because we don't have the money. We have a fixed amount of money in our budget. So, that's our big thing. It's not not-knowing what to do. We know what to do. We just don't have the money. [#8]

Although provided at no direct cost to patients, genetic services entail significant financial sacrifice to individuals and to communities. One of the financial burdens emphasized by genetic professionals was the significant costs of transportation and related costs incurred because of the time that transportation entails. Recipients of genetic services, particular those living in rural and remote areas, incur significant costs related to transportation.

> Somehow that cost of that flight or that drive or the fact that you are not working for three days - the day you are driving across the island, the day you are having your appointments and day you are driving back-something has to equalize things to cover those aspects. [#5]

Other significant costs to patients occur when commuting to the nearest, yet still significantly distant, genetic centre.

> It is so expensive with gas and lodging and they are coming in just to have a conversation, right? So a lot of people don't see that as necessary. [#2]

Accommodation is an additional expense if patients have no option to stay with relatives or friends. For the duration of a visit, eating in restaurants is usually the only option, and one that is more costly than eating food prepared at home. Very often patients travel with one or more family members or friends who provide emotional support, which further increases the out-of-pocket expenses.

It is not unusual to have a last minute cancellation of the genetic consult if the geneticist is sick or, for other unforeseen reasons, becomes unavailable for the appointment. This can contribute to further costs for the patient and those who accompany them. It is, of course, artificial to separate out the issue of these financial costs related to transportation (a systemic barrier) from the psycho-social experiences of individual patients and families bearing those costs (to be discussed in Chapter 4).

3.4 Barrier # 4: Limited Capacity

The primary focus of the PMGP services is to assess and manage patients that have been referred to genetic care. The program also looks at the strengths and needs of clients in order to determine other service needs. For instance, family needs are addressed in order to help create an appropriate support environment that enables both clients and

their family to better cope with possible distress. In some cases, genetic professionals also relay information to the extended family.[39] In effect, the genetic care is individualized (tailored) in a way that meets the unique needs of clients and their families. Services are available in three locations in the province – St. John's, Gander and Corner Brook (see Figure 3) – and patients are given a choice to be seen in the most convenient location. The three sites, as explained earlier, were set up as permanent genetic assessment sites (however with visiting medical geneticists) in the relatively large urban centres in the province.

3.4.1 Lack of personnel. At the time of the interviews, there were only two medical geneticists serving the entire province. Typically, they offer consultations at the St. John's site as well as through satellite (travel) clinics[40]. The genetic clinics established outside St John's follow the triaging protocols adhered to at the main centre, however, those clinics "barely meet the demands" as one informant [#4] commented. The satellite clinics lack permanent medical geneticists to provide administrative, diagnostic or supervisory duties on site. Unlike the St. John's site, where the medical geneticists are stationed together with a team of genetic counselors, the lack of other permanent staff in the outreach clinics makes it cumbersome for the genetic nurse-counselor[41] in the outreach clinics; they frequently need to consult with the St. John's site. Although

[39] Clients are asked to circulate a family letter among their relatives to inform them about a possible genetic risk or about a genetic test, if available, as well as other relevant information.

[40] The satellite clinics (also referred to as outreach clinics or travel clinics) are permanent sites, visited by medical geneticists who travel to the sites.

[41] The two genetic nurses/counselors at the satell te clinics are well trained and experienced genetic professionals. However, they are not certified genetic counselors.

communication with the main site occurs relatively regularly, the contact is not immediate and only via telephone[42]. As well, when the medical geneticists complete an outreach visit, they routinely take the dictations from the consults to St. John's to have them typed. According to one participant, the typing may not be completed right away as it is added to the already high volume of paperwork that is processed in St. John's. This creates a backlog of patient files at the St. John's site that, according to the participants, translates into delays of at least six months. As well, all other paperwork is sent to St. John's for approval and then forwarded back to the outreach. Although having the St. John's site as a hub for centralized management of client files may be a sensible approach in terms of efficiency (in fact, there is no feasible alternative), it does not ease the already time- and labour-intensive process of genetic care delivery. Because this process of having patient files processed in a central location is under-resourced, it is burdensome and lengthy and thus adds to the wait time for patients as well as to the overall cost.

Each of the two medical geneticists stationed in St. John's is assigned an outreach centre. They are scheduled to travel to their assigned area once or twice a year to provide service for two weeks at each clinic. This presents a very limited window of time for patients to be seen, and translates into only 18 patients a year according to the participants. This limited timeframe exposes the genetic outreach structure to a wide range of vulnerabilities:

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[42] Staff meetings are also held via teleconference and their regularity depends on the availability of a medical geneticist whose attendance is sine qua non.

> You know, with only two geneticists in Newfoundland, if someone gets sick, then everything falls down. [#9]

The PMGP addresses those vulnerabilities by attracting locums from other provinces. Typically, a locum is a semi-retired medical geneticist who is recruited to cover the duties of the local medical geneticist when that geneticist is unavailable. Typically, locums work for three to six weeks per year at the main genetic site. This practice, initiated relatively recently at the time of the interviews, helps with both case management and wait times; however its scope is limited, as locums do not have administrative or supervisory responsibilities, and their availability on an as-needed-basis is not certain. So far, only one locum has had a clinic in the outreach, which lasted for one week. Budgetary and physical space constraints contribute to the decision to attract locums instated of hiring permanently additional genetic professionals.

Given the increased demand for genetic care and the large geographic area served, the number of medical geneticists is far from adequate. So is the number of genetic counselors, especially outside St. John's. To offset this shortage, nurses with genetic training[43] provide counseling services in the outreach. The issue of limited availability of genetic professionals has arisen across Canada (Silversides, 2007) as well as in other countries and has been reflected in previous studies (Hawkins, & Hayden, 2011; Yoon, Thong, Taib, Yip, & Teo, 2011; Klitzman, 2009; Vig et al., 2009).

[43] The practice of hiring genetic nurses has been discussed by Lee et al. (2006).

According to the research participants, the challenges associated with insufficient personnel extend to the genetic labs as well, which compounds the time and workforce constraints associated with genetic care.

In general, the interviews revealed that while the current structure (that is, a system dependent largely on one central and two satellite/travel genetic clinics) facilitates access to genetic care, the outreach model is far from meeting the desired standard of care. Having only two medical geneticists serving the entire province is seen as an unsatisfactory arrangement; however, increasing the number may not be an immediate solution due to limited resources. Reflecting on the issue, one research participant noted:

> So, I think in a perfect world (pause), we would have unlimited number of genetic counselors and geneticists working in this clinic. I wish we had more people. I think financial resources are always an issue. [#3]

3.4.2 Heavy workload. Dealing with a heavy workload is another issue closely related to the limited number of genetic professionals in the province. Not surprisingly, when invited to comment on the barriers to providing genetic care, participants indicated that heavy workload is an on-going issue.

> We're running pretty much on full cylinders.... [#3]

One aspect of the heavy workload borne by the genetic service providers is the cumbersome process of obtaining and reviewing important (medical and family related) information from clients as well as documenting that information in the appropriate

format. Participants noted that the number of patients they see is not as high as those a medical specialist or family physician would attend to. However, they emphasized the overwhelming amount of labour involved in delivering genetic care, which more than one participant described as a "different style of medicine"– genetic professionals are required to spend a significant amount of time preparing before they see a patient and genetic consultations are consistently lengthy.

> We estimated that to see our patients takes us about eight hours, and that's eight hours after a lot of assistance is given in terms of collecting or reviewing patient information.... [#8]

A considerable amount of paperwork needs to be assembled for both genetic diagnostic and counseling purposes. As family history information is key and the process of attaining it is complex as well as time consuming, there is a designated person (on a rotation basis) at the main PMGP site in St. John's who is responsible for collecting medical history from various sources, including personal and family medical records.

Based on the family history information provided[44], a genetic counselor constructs the patient's pedigree.[45] Although centralized management[46] of these documents is a prudent approach, it does not translate into less paperwork. On the contrary, the workload is consistently high especially at the satellite clinics:

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[44] When completed, the adult family questionnaire contains information about relatives in three generations on both the father's and mother's side of the family.

[45] At the time of the interviews, it was reported that 20-30 % of pedigrees are drawn by undergraduate student volunteers.

[46] The completed family history questionnaires are received in St. John's unless it is a direct referral to the outreach clinic.

> Well, they kind of have to do all that themselves in the outreach, so I think paper work is probably a little bit more extensive in the outreach than it would be in a major center. [#9]

Each satellite clinic has one permanent genetic nurse/counselor who resides in the area. Having knowledge about the families in the community is of professional advantage to the nurse/counselor in configuring pedigrees and helping clients to fill out family history forms. Being part of a particular community also helps the genetic nurse/counselor in establishing rapport and trust with clients. While being familiar with clients and their relatives from the area facilitates the work of the genetic professionals to some degree, it takes considerable time and effort to address the clinical as well as the emotional needs of each client:

> In terms of the consult, reading the person's emotions and everything and making that really good personal contact is important. [#2]

> The assessments are a lot longer and for a new consult we take an hour and a half and we address the needs not just for the person sitting in front, but those of at least the first-degree relatives, if it's that type of genetic risk. [#8]

In addition to communicating with patients and medical geneticists (in-person, via telephone or in writing), the genetic counselors are involved in obtaining price quotes

from genetic labs, scheduling genetic tests and, in the case of satellite clinics, even performing some preliminary tests[47]. Both medical geneticists and genetic counsellors also prepare and review a high volume of paperwork preceding and following the genetic consult. All these activities are both labour and time intensive, especially when only limited clerical help is available.

> Well, too much paper work. And probably not enough support. I don't mean genetic support; but I need, like I said, a person helping me with my work. [#9]

For the outreach clinics, the work processes are even more convoluted as access to both professional and clerical support is not as immediate as at the St. John's site. For example, generally uncomplicated procedures such as approval of correspondence to patients or typing geneticists' dictations become tortuous as the papers are sent to the main centre and processed there (depending on availability of personnel), and then mailed back.

The literature on the work of genetic professionals has described well the time-consuming nature of conveying information about the basics of genetics to patients (see especially Yoon et al., 2011): Pedigree configuration, risk assessment, diagnosis, interpretation of test results, as well as preparing and communicating other relevant information to clients take additional time. My research has shown, however, that the

[47] These are not genetic tests.

"hidden" work of managing patient files is equally significant in terms of time commitment.

3.4.3 Continuity of care. Care related to a genetic disease cannot be provided by a geneticist alone. Medical services for hereditary conditions involve other medical specialists as well. For example, in the case of hereditary colorectal cancer, gastroenterologists and /or surgeons are involved. During the interviews, participants reported that there are not enough gastroenterologists in the province to perform colonoscopies, nor surgeons to carry out intervention when appropriate.[48] Research participants commented that coordination of care is a huge barrier.

> Care related to a genetic disease is not just by a geneticist. If it is a neurological disease, then your neurologist and maybe a neurosurgeon will be involved. And so, you have to have access to that part of genetic care. It is not just the geneticist and genetic counselor; it is what the geneticist and genetic counselors recommend keeping you healthy. I think that's the thing that sometimes is not considered and often that can be a barrier.... The people have to be followed up. That's what's difficult – to put that whole pathway in place. [#5]

According to this participant, it can be a challenge to secure follow up for patients, both by genetic professionals and other specialists. As van Maarle, Stouthard, & Bonsel

[48] This point is not specific to colorectal cancer. I refer to this condition specifically for illustration purposes.

(2002) note, strengthening the link between genetic diagnosis and follow up care is critical in achieving high quality genetic care.

In contrast, another participant provided an example of successful continuity of care – a cardiac genetic clinic, which utilizes a multidisciplinary approach and provides a full range of necessary genetic services, cardiac care and follow up. In the particular clinic described, patients with a genetically inherited cardiac disorder are ascertained through genetic research or the PMGP and are treated by cardiac specialists in communication with their family physician.

> This clinic works extremely well because it has cardiac services involved, genetic services involved, genetic research involved, and all the information goes to the right physicians who are involved with the patients, so all bases are covered, basically. [#7]

Part of the problem with providing continuity of care is geography and its associated barriers, as already discussed. For example, for patients with ARVC,[49] St. John's is the tertiary centre for cardiac diseases and the only place in the province where patients can be treated for cardiac conditions. Although cardiac patients who reside outside St. John's may be in close proximity to a satellite genetic clinic, the only service they can obtain at the outreach clinic is genetic information. For cardiac-related testing, including genetic testing, they have to travel to St. John's. They also have to travel to St.

[49]Arrythmogenic Right Ventricular Cardiomyopathy is an inherited form of heart disease characterized with degradation of the heart muscle, which is replaced by scar tissue and fat. The first symptom is death, hence, it is referred to in lay terms as sudden cardiac death.

John's for the treatment itself. Hence, although continuity of care exists in theory, issues related to geography and its related social and economic costs are significant.

Lee et al. (2006) comment that bringing together professional expertise in a multidisciplinary team is a challenging task in genetic care. This holds true even where genetic services are delivered in a public hospital. Beene-Harris et al. (2007) suggest that there is an apparent need for other health professionals to join genetic specialists in the provision of follow-up and supportive care; they call for improved coordination between diagnostic and follow up care by installing an infrastructure that encompasses all the necessary health expertise. However, in the Newfoundland and Labrador context, the issue is not one of lack of engagement of specialists in genetic care; rather, the issue is how to access the continuity of care that exists, given the challenges of geography and limited resources.

In their commentaries, the participants openly pointed to the need for changes to the system to transform genetic care into more accessible, meaningful, and efficient arrangements for both patients and providers. The nature of genetic disease is such that visiting the genetic clinic is only the first specialty care stop for patients and family members; the genetic clinics cannot resolve all aspects of care needed. Securing access to other specialists within the continuum of care is crucial, and this issue demands further investigation and resolution.

3.4.4 Managing client volume. One research participant in particular elaborated on the lack of sufficient resources necessary to handle the volume of people accessing

genetic services. A concern was expressed that raising people's awareness about genetics may bring more clients to the clinic, and that the clinic does not have the capacity to deal with any increase in client volume.

> We do not have a lot of time and resources to deal with a lot more referrals efficiently. So, it becomes one of those things that you're sort of balancing how aggressively you want to go out and find these other referrals. [#3]

One research participant felt that "direct advertising" might be key to improved public knowledge about genetics and genetic diseases. However, the participant expressed concern that this information may generate unnecessary fear about genetic conditions that people may or may not want to be screened for.

> We generate fear, you know, by telling them [the public] that these conditions exists when in fact, you know, if you think of illness as a whole, a genetic condition is only a very, very small part of that whole illness profile. [#4]

When it comes to promoting genetic services, "marketing of fear[50]" is not an acceptable technique; however it could be quite effective in augmenting client volume. Because of fear for their health and the health of their offspring, people seek genetic services and want to be tested. A spike in fear results in escalated demand for genetic testing, which, in turn, endorses the establishment of services.

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[50] This is a phrase used by critical theorists such as Lupton (1994, p. 142) to refer to the way in which medicine (or genetics in this case) markets itself by creating a fear of disease so that health consumers will actively seek out the new medical (or genetic) technology to avoid the disease.

> So, that's [whether or not to raise public awareness] always a dilemma, you know, that I struggle with, because we do not have the resources to handle, for instance, an influx of patients that we could generate into our department by just making people aware of genetic conditions. [#4]

The demand-spawned-by-fear phenomenon has been well documented in the literature since the concept of "geneticization[51]" (Lippman, 1991, p.18-19) was first introduced.

3.4.5 Wait times. The issue of how to manage the volume of patients accessing the PMGP services is intertwined with both the shortage of genetic professionals and the labour and time-intensive workloads they have. The professionals I interviewed distinguished between wait times for an appointment with a genetic counselor or clinical geneticist, and wait times to have a genetic test completed. Further, they were of the opinion that wait time, especially for appointment with a medical geneticist, is directly related to the number of geneticists and genetic counselors. They explained that if a physician refers a patient for genetic assessment, it may take a year or so before that patient is called for a genetic appointment.

> The wait list for medical geneticists right now is four months to a year and a half, to see them. [#1]

[51] "Geneticization" is a term coined by Lippman (1991, p. 18-19) to offer a critique of the process by which disorders, behaviours, and physiological variations are increasingly understood as being genetic in origin.

The participants reported that the PMGP has made attempts to address wait times. For example, at the time of my research, the model of client intake had been improved by adopting a centralized triage system based on three priority categories of cases: urgent, semi-urgent and routine. On a monthly rotation basis, a genetic counselor in St. John's is assigned to client intake duties and allots each case to a category as well as to a colleague according to his/her "specialization"[52]. Triaging is dependent on diagnostic criteria as well as client age and involves professional judgment. One participant stated that clients could always access the system, but whether their case will be expedited depends entirely on the category one is triaged to [#5]. In general, an urgent case is a high risk case that requires clinical management of the disease; it is processed fairly quickly with access to testing "in a matter of a couple of hours to 72 hours" [#3]. This generally includes in-patient hospital referrals and unusual metabolic cases. Semi-urgent cases, which most clients fall into, are supposed to take three to six months waiting for testing. Such semi-urgent cases may include instances in which, based on family history, there is a strong indication that a family member may carry a mutation. Non-urgent cases are referred to as routine cases. These are instances in which the genetic risk is low and where the clients do not require clinical management (for example, a client is eager to know, out of curiosity, if their particular condition has a genetic component[53]). As routine cases are prevalent, they are often left to "languish a few years" [#7], as disease management is not an issue.

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[52] For example, certain genetic counselors specialize in neonatal, cancer, or cardiac care. All other cases fall into the general case category and are handled by counselors without specialization.

[53] Having the genetic influence on disease risk determined may also alter family members' attitudes toward the disease trait and genetics in general.

> With the triage system in place, the goal is to give clients – including non-urgent (routine) cases – a fair chance to be seen within a year. [#1]

However, the participants indicated that the wait time for an appointment with a medical geneticist could sometimes be up to five years; for a genetic counselor specializing in cancers, it could be three years; and for a general genetics consult, one year to 18 months.

In terms of wait times for obtaining the results of genetic testing, these depend on the type of test needed.

> That can range....two weeks would be the shortest turnaround time to get a blood test back, up to may be four months for a clinical DNA test.... I can't think of any that is longer than that amount of time. [#2]

Participants indicated that they also order genetic testing for the purposes of research. As explained earlier, genetic research is an integral part of the genetic services system, and research-based testing is an option when no clinical testing is available. Obtaining research testing results may take years.

> We send off a DNA sample to some research lab that we are not paying for [occasionally, research labs may not require a fee for tests performed as part of a research project] and that can take a month-to-never to come back. So it could be indefinite in terms of how long it would take to get results back from our research laboratory because they have no obligation

> to finish that study or whatever they are doing, or their study is so long that, you know, they may never get to our sample we sent them until eight years later or something. [#1]

> So if the DNA sample was collected maybe 10 years ago, it was only recently, relatively recently, tested because the genetic test has only been recently available. [#4]

Another participant commented on the uncertainty associated with wait times for test results when clinical patients are tested through research.

> For our hereditary colon cancer families, a lot of testing is being done through research. And with research, you never have a set timeline. [#3]

While there is a triage system to establish case priority, the set timelines are not always met. In fact, those triage-based timelines are somewhat arbitrary – they are not based on any standard guidelines. According to one research participant, the triage process recently implemented at the PMGP at the time of the interviews was conceived locally and may not be in keeping with the process at other genetic centres across the country. One participant commented that there is not really an infrastructure in place to evaluate if in fact the PMGP is meeting the objectives they set themselves in terms of following timeframes. When back-ups occur due to a high volume of client files, then locums are brought in to assist with the workload and to reduce the wait times.

Evidently, the wait time for a client to be seen by a medical geneticist can be fairly long: it may take a year, two years, or even five years. Likewise, genetic testing may at times take years to produce results if any[54]. One informant summed up the relationship of this particular barrier (wait times) to systemic barriers in general:

> If you are going to reduce the wait times, then there has to be more funding for more personnel to do the work. [#3]

3.5 Barrier # 5: Genetic Literacy of Physicians who are not Geneticists

The participants made a strong argument that, in addition to the need for adequately resourced genetic centres, an important determinant of the quality of genetic services is the knowledge that medical professionals (non-geneticists) have about genetics and clinical genetics in particular. The participants reported that there are inconsistencies in terms of the degree of genetic knowledge held by family physicians and medical specialists (non-geneticists). One genetic practitioner elaborated:

> There are some people that are what I would call our regular referrals, who understand what we do over here and send a lot of patients our way, and those people know very well what's going on. There are other physicians, I think, who have less of an idea of what we're doing and sometimes will only refer maybe because the patient has asked for that referral, and then

[54] Sometimes results of significance can take years, or never materialize, because it takes time to develop a reliable genetic test or to make a gene discovery with clinical relevance.

there's...clearly there's physicians who do not know anything about what we do here and do not refer. [#2]

So I guess awareness among physicians in the specialty group is really what generates referrals of clients into our services... [#4]

It was suggested that targeted information directed to family physicians would make that particular group of non-geneticists more aware of what the PMGP is doing:

Ideally, it would be nice if every family doctor's office had, you know, a poster that addressed the question: do you have this and this in your family and here are the people that can help you with that. [#3]

Participants also alluded to the gate-keeping role that family physicians play in the process of genetic care delivery.

The family physician has the power to move them [patients] along the system. [#6]

We rely now on family doctors to assimilate the fact that there is screening program available. We find that there's not enough uptake of patients that come in through that mechanism. [#4]

Reflecting on the importance of the knowledge level of other health providers, and medical specialists in particular, one participant commented:

> It depends on the information the specialist has. So, really it's how informed is your specialist who's treating you – your attending physician – whether or not they promote genetics as being part of a service that's available to you. So that's haphazard. [#4]

However, as mentioned previously, the program's capacity to handle more clients is a huge hurdle to the desire to increase effective referrals. When family physicians or other non-genetics specialists choose not to move patients through the system, the bottleneck point (that is, the wait list of clients to receive genetic services) is eased considerably; yet keeping patients from being referred to genetic care is counterintuitive to the goal of the PMGP. Hence, recognizing the strategic importance of the referring physicians and reducing or eliminating any obstacle they face in their role of referring patients to genetic care is essential for the steady operations of the Program and its effectiveness.

> If we have every single doctor in the province understanding genetics, it would be five years before you can have any appointments because the referrals would be too many. However, they have where to send people and some knowledge is going to move along. [#5]

Genetics awareness and adequate knowledge are central in achieving the balance between appropriate referrals and under- or over-use of referrals to genetic care. It seems,

then, that deficiencies in genetic competencies seem to have a dual role: they present a challenge to the genetic services by limiting or diverting access to genetic care for those who would benefit from it, while at the same time, the lack of appropriate referrals provides a benefit to the current system, given that the current system can handle only a limited number of referrals. As a prerequisite for efficient genetic services Starfield et al., (2002) recommend that family physicians be knowledgeable and confident to deal with genetic problems. However, genetics education for physicians (non geneticists) is only minimal and needs continuous update given the rapid advances in genetics (Caulfield, 1999; Metcalfe, Hurworth, Newstead, & Robins, 2002; Klitzman, 2009). Contrary to what the majority of the participants stated and what the literature affirms, some participants commented that physicians (non-geneticists) have "much more perspective about genetics than they are given credit for" [#5]. According to one participant [#5], the majority of physicians are willing to refer patients to the genetic clinic, however, they do not choose to because of concerns regarding continuum of genetic care. Another participant commented:

> I feel like the majority of physicians in Newfoundland and Labrador are really aware of the genetics in the province and are very good at making referrals. I've worked other places where doctors out there don't even know that the genetic department exists and the referrals aren't appropriate. But I'd say the majority of physicians just make their referral. However, I do not get the feeling that patients are being educated on the reason why they're being referred. [#2]

This observation is not surprising, as physicians have notoriously busy practices where physician-patient relation time is limited. Moreover, awareness of genetic services does not always translate into adequate genetic competencies; medical professionals (non-geneticists) may not be fully confident in discussing genetics with patients and may choose to defer such conversations to the genetic specialist to whom the patient is being referred. These findings parallel what has been described in the literature. Results from a U.S. survey revealed that more than 90 per cent of medical professionals had no training on common genetic disorders (Maradiegue, Edwards, Seibert, Macri, & Sitzer, 2005). Research on a U.S. population has also shown that patients have concerns and are dissatisfied when doctors fail to direct them to proper genetic information or treatment options, suggesting that there is a need for further education (Beene-Harris et al., 2007). Metcalfe et al. (2002) conducted research on Australian medical professionals' views on their own level of knowledge of genetics. The physicians self-assessed their competencies as inadequate, indicating that they underutilize genetic services and claim low relevance of genetics to their practice. Similar are the findings of Suther & Goodson (2003), who reviewed published literature on primary care physicians' perspectives on the barriers they experience in providing genetic care. They discuss inadequate genetic knowledge of primary care physicians as a barrier to genetic services and describe the low confidence level of physicians in assessing and referring patients to genetic care.

Unmistakably, the majority of participants I interviewed felt that there is a gap in physicians' genetic knowledge and that this lack of knowledge is a barrier to access to and up-take of genetic care. Their views are well supported in the literature, suggesting

that there is a need in Newfoundland and Labrador for doctors to obtain continuing education in clinical genetics.

3.6 Barrier # 6: Post Referral Attrition

One research participant elaborately described a number of scenarios arising with already referred patients.

3.6.1 Communication with attending physician. According to that genetic professional, some patients feel uncomfortable saying "no" to their physician when referred to the genetic clinic, even when they are hesitant about availing of genetic services. These patients subsequently do not present to the clinic, or discontinue care, situations that produce the same result as a non-referral.

3.6.2 The burden of completing lengthy information forms. Several interviewees suggested that the family history questionnaire which clients are required to complete is a huge disincentive to pursuing genetic assessment. The form is a fairly extensive document and some patients find the questionnaires overwhelmingly complex. Nevertheless, it is essential to have the family history completed, as genetic professionals use it to configure a pedigree necessary to determine genetic risk.

> We send out family history questionnaires to get the pedigrees. We lose a certain portion of referrals that way. People are just not willing to fill in these forms. [#3]

The turn-around time for a patient/client to send the required information back to the genetic professionals determines to a great extent when that patient can be clinically ascertained. The PMGP has a three month turnaround deadline for receiving both the family history and the release of information form (which consents to contact with other family members). To ensure the timeliness of responses, the genetic clinic follows up with patients via telephone every 30 days, up to three times, as a reminder to send back the completed documents. Patients are offered help with completing the forms if it becomes clear they need it (that is, if they request help or return the forms with incomplete information).

> Part of the problem is that people are overwhelmed by all the paperwork that needs to be done. Maybe we could devote more resources to getting the information over the phone rather than doing it by a form. [#5]

The more recently established colorectal cancer screening clinics use an improved version of the PMGP questionnaire, where certain questions are consolidated and better articulated to alleviate clients' confusion and save time. It is premature to compare attrition levels in terms of the two versions of the questionnaire.

3.6.3 Communication between family members. Once a patient pedigree is configured, genetic professionals need to obtain additional information regarding diseases that have occurred in the family. Having family members sign a release form enables the collection of this information. This requires communication with family members, a task that patients may find burdensome. Genetic professionals reported that some patients are

not willing to communicate with relatives to ask them to sign the release of information form, causing at least a 15% client drop out rate[55]. As one participant put it,

> If they [patients] go through sending in their paperwork and they talk to all their family members, by that time they are committed. Otherwise, we lose a certain sub-set of people. [#3]

Participants explained that the PMGP staff is well aware of the attrition problem. The Program keeps track of "lost clients" and has been working to find ways to overcome this hurdle.

More research is clearly needed on post referral attrition as a barrier to uptake of genetic care. Having patients referred to the genetic clinic for assessment and losing those patients partway through the process is costly for the system and possibly for the patient if extensive travel or other arrangements were incurred. More importantly, there is a lost opportunity to provide and receive genetic care that may be time sensitive for the patients and their families, and desired health outcomes may not be achieved.

[55] It is not clear whether these "lost clients" cases are also associated with self-referrals, where patient self-motivation is the driving force for availing of genetic assessment.

Chapter 4: Psychosocial Barriers

In this Chapter, I focus on how genetic professionals have understood and interpreted the psycho-social challenges experienced by individuals and family members. The privileged position of genetic professionals as providers of services is key to a full assessment of the challenges to accessing and using genetic services. Genetic professionals have direct contact with patients and families and thus have an in-depth understanding of the broad range of experiences of individual patients and families with various symptoms and clinical needs.

Genetic professionals' interactions with patients and families are influenced by individual patient characteristics as well as their familial and other circumstances. Understanding patients' beliefs about inheritance as well as their sense of vulnerability associated with perceptions of risk is a fundamental part of genetic professionals' work, and indeed is imperative for effective genetic counseling (Walter, Emery, Braithwaite, & Marteau, 2004). Geneticists must be constantly cognizant of and receptive to what their clients are saying and not saying (believing, assuming, valuing) because doing so is an essential strategy for identifying any stumbling blocks that clients are likely to experience in the process of receiving genetic care. In fact, genetic professionals are in the best position to prevent, mitigate or help overcome some of the challenges to access by first recognizing them and then exercising direct influence to overcome the challenges.

Psychosocial issues associated with genetic testing have been well examined and described in the literature (e.g., Meiser, 2005; Braithwaite, Emery, Walter, Prevost, &

Sutton, 2006; McAlister, 2007; van Oostrom et al., 2007). The majority of research focuses on the perspectives of individuals who pursue genetic care and includes a broad spectrum of issues, including difficulties discussing genetic risk within the family (e.g., MacKenzie, Patrick-Miller, & Bradbury, 2009; Featherstone, Atkinson, Bharadwaj, & Clarke, 2006); willingness or lack of willingness to undergo genetic testing and learn the test results (e.g., Lacour et al., 2008); social stigma and discrimination related to testing (Smith, 2007); and anxiety and depression associated with undergoing genetic testing (Douma, Aaronson, Vasen, & Bleike, 2008; Shalowitz, & Miller, 2008; Dixon-Woods, Jackson, Windridge, & Kenyon, 2006; d'Agincourt-Canning, 2001; Lippman, 1991).

Beyond what the patients and families say about their experiences of genetic care (amply described in the literature), genetic professionals' perspectives are key to understanding psychosocial barriers to effective and efficient genetic care. According to the genetic professionals I spoke with, there are a number of psychosocial factors influencing patients' ability to access genetic services. It is worth noting that genetic providers' comments varied in terms of whether or not psychosocial barriers were raised spontaneously: only two participants talked spontaneously about the difficulties their clients face; others provided commentary when specifically asked.

4.1 Barrier #1: Lack of Client Awareness of Genetic Services

Many of the genetic professionals I spoke with were concerned that the general public does not know that there is a genetic clinic and what its mandate is. In fact, genetic

professionals saw lack of awareness as a leading challenge in appropriate access to genetic care. As one participant succinctly put it,

> I think that the biggest barrier is probably just awareness that genetic services exist. [#3]

Interestingly, the level of awareness of what genetic services are and can do does not increase even after the patient is referred to the genetic clinic. The assumption that patients, in particular those referred by a medical professional (non-geneticist), are being advised on what to expect from the genetic clinic and have acquired basic information about the services they are going to receive, simply does not hold true.

> I would say that most people, when they're referred, don't really have a good idea of what we can do or what we're going to do. [#2]

> I always joke that they [the public] think we're cloning people down here (laughs) because they have no idea what we are. [#3]

> If they are new families I find that they have absolutely no idea what we do here. [#6]

> ...the bulk of people are pretty clueless. [#1]

Those commentaries overwhelmingly illustrate genetic providers' concerns about the lack of understanding about genetic care. The PMGP clinic has engaged in some publicity efforts that predictably have generated additional interest:

> We've done a little bit...we can call it some publicity: I was on the radio with one of my patients. So we got a couple of phone calls after that that I can think of. [#3]

Efforts to augment public awareness through newsletters, public seminars and other public forums were discussed by the genetic professionals. Raising patient awareness about the genetics clinic and the services it provides was seen by the respondents as a desirable move forward; however, the lack of resources to accommodate an increased volume of clients (as discussed previously) was described as one of the main reasons public education about genetic services continues to be insufficient.

Participants reported that public interest about genetic services is increasing; the number of referrals is swelling, and the waitlist is longer in comparison with waitlists in the recent past. Importantly, genetic professionals feel that the heightened demand for genetic services is not paralleled by an increased understanding of what genetic services are and what they can offer to clients.

> There are people out there that have pretty dramatic family histories of – you name it – whether it's cancer or heart disease or such-and-such disease. I think letting them know that there is a clinic here – that our job is to sort these things out – is the way to improve the service. [#3]

The perception held by the genetic professionals that there is a lack of public awareness about, what genetic services offer as well as lack of knowledge of genetics is in keeping with other studies that have revealed the lack of public awareness and the critical need for public education (McClaren et al., 2008; Jonassaint et al., 2010).

4.2 Barrier #2: Client Knowledge of Genetics

Many of the genetic professionals I spoke with felt that clients' lack of awareness about the role of the genetic services was coupled with a lack of knowledge about genetics and inheritance in general. All genetic professionals unconditionally stated that their clients' understanding of genetics is minimal. They expressed frustration about the fact that although their clients receive a vast amount of genetics-related information through the clinic, verbally and in writing in multiple iterations, the information remains incomprehensible for the majority of clients. They emphasized that genetics is based on probabilities and genetic risk is an abstract concept that proves very difficult for many patients to understand. One participant observed:

> There are several barriers that I have found, and the first barrier is the level of knowledge. Patients really find it difficult to conceive of that they pass on something that doesn't....how they pass it on and you know, that it doesn't go to one sex or the other sex, and they have a lot of old wives' tales, inherent sorts of thought processing around it anyway. So to try to explain that to them and to be able to break it down to them so they

> understand it, that's the first level. That's the theme that I've come across most. [#7]

> It's strange because people will say, especially if they have two or three sisters they'll say – oh, my mother really lucked out because there are no boys in our family and you can't pass it onto boys. So there is a real confusion about it. [#6]

Although the participants did not specifically discuss whether certain genetic concepts were better understood than others, the examples they provided alluded to difficulties understanding inheritance patterns. Their comments substantiate findings of previous research that have demonstrated that the general public and patients have poor knowledge of genetics (e.g., Christensen, Jayaratne, Roberts, Kardia, & Petty, 2010; Falcone et al., 2011; US National Science Board, 2008; Molster et al., 2009). Genetic professionals were especially concerned that people may be holding and "passing on" misconceptions about genetics and inheritance patterns that deter them from seeking genetic care, an observation that is also supported in the literature (McClaren et al., 2008).

4.2.1 Education through the genetic clinic. Individual clients and family members are offered detailed genetic information about genetics and genetic services by the genetic practitioners. One respondent explained that clients seen in the clinic arrive, with or without a diagnosis, and the genetic clinic supplies most of the information about

their disease and its genetic basis (provided the gene is known). In cases where the referring physician mentions a genetic condition as a possibility, depending on the education level of the patients and their internet access, some clients might very well gain some prior knowledge of genetics and genetic services. However, the majority of clients approach the genetic clinic with little or no awareness and typically everything is novel to them.

> Many of my patients wouldn't know they had a genetic disease until they saw me, in which case I am the one that's supplying at least the initial information. [#8]

Once a client leaves the clinic, they are already furnished with a great deal of information and support materials, including printed materials and referrals to websites. Subsequent to the visit, a follow up letter is sent out to the client summarizing the discussion that had occurred during the consult. In fact, the mandate of the genetic counselors is mainly to provide education and support to clients with genetic conditions and to address their psychosocial issues. One informant argued that this is the reason why genetic counselors should be referred to as genetic educators. In the same vein, another informant attested that their clients' understanding is usually minimal in the beginning of the contact and, depending on other variables[56], they will have an enhanced level of understanding at the end. The 'education' component of the genetic service is vital as

[56] Variables include, for example, the client's level of formal education, as will be discussed in the next section.

clients may be able to obtain better understanding about their actual risks, as opposed to their perceived risks, which according to one informant are oftentimes "inflated".

> First, I will explain to them how genes work and how an illness or syndrome is passed on to children, and the difference between recessive disorders and genetic disorders. I say to somebody - you have to have two genes for this, so your mom and your dad had to have this in order for you to have it, like that sort of thing. It's very difficult to explain that to them. [#6]

> You know, most cases of Down's syndrome are not inherited: and then people have a family history of Down's syndrome and they think they are very much at risk of having a child with Down's syndrome themselves, just an example. [#8]

> I think we are giving the patients a variety of sources. It's actually the genetic counselor that's working with them and would give them pamphlets on genetics, information on patient support or recommend websites that might help with a particular disease. [#1]

Efforts to augment public vigilance through newsletters, public seminars and other public forums and lectures where appropriate were discussed by the respondents.

These practices are especially relevant when the genetic professionals are involved in long-term genetic research projects.

> So we've made a conscious effort to make sure people are informed. We'll have a newsletter going out. I mean it's just a brief note but we will describe what we've done in the past, where we hope to go in the future. If there's been challenges to it, we'll put that into it to say "staffing has been an issue" or "funds have been cut" – something like that, so they understand. We had a full seminar day and invited the public and all the people who participated to come here to the Health Sciences. We had speakers, we provided lunch. They had the opportunity to get up close with surgeons, geneticists, genetic counselors and that sort of thing. So that's the way to we try to build bridges and maintain patients in studies. [#6]

The genetic practitioners I interviewed emphasized their educational role. They explained that providing information to their patients is part of the supportive role they play for individuals and families. Those efforts appear to be even more streamlined when related to genetic research through which clinical genetic care is accessed. Through the interviews it became evident that the current environment is one of balancing public awareness of genetics with the desire to prevent the creation of a "genetic need" or unnecessary fear (as discussed in Chapter 3).

Paradoxically, it appears that while the clients and the public seem to have limited genetic knowledge, the participants reported that, presently, the interest in their services is more pronounced than ever. This trend is consistent with the findings of Wang & Watts (2007), who also note that the genetic profession may not have adequate resources for increasingly dealing with adult clientele.

An important counter-point to studies on the knowledge deficit of the general public and genetic clients is research that argues that the emphasis on clients' interpretation of statistics is a moot point. While the genetic service providers I interviewed did not speak to this point, critical scholars of genetics have arguedthat clients' understanding of genetic risk will necessarily be reinterpreted in the context of the riskiness of daily life, and that genetic professionals therefore are better advised to attend to the meaning (rather than the statistical interpretation) of genetic risk (e.g., Peterson, 1999). "The public are not passive consumers of health education messages, but active participants in their interpretation and social construction" (Sanders, Campbell, Donovan, & Sharp, 2007, p. 519).

4.2.2 Education through family experience. While discussing the inadequate level of clients' genetic knowledge, the genetic professionals reported that patients who had family members with prior experience with genetic services had a much better understanding of genetics and what genetic services can offer.

> It makes a difference what families know about genetics and whether or not they have heard about a gene or genetics before. [#1]

Another participant used the example of a family with many young people dying from colon cancer:

> They [family members] recognize that there is something going on in the family that is not common throughout the whole population" [#5].

In that example, if a family member mentions that because of early detection through colonoscopy they had a polyp removed and thus did not have to undergo chemotherapy or radiation treatment, then other family members follow suit and pursue genetic care. In other words, the presence of a genetic condition in a given family and especially its successful treatment can mobilize better understanding of genetics and inheritance. As well, the management of a pre-existing condition by some within a family may motivate members of the affected family to avail of genetic counseling and screening, including through self-referral. Genetic professionals reported that, within families with hereditary colon cancer, the uptake of genetic testing is high because individuals want to know if they are in the high-risk category: the test results determine the level of rigor of the screening protocol to be followed. For that particular condition, given that the screening for colorectal cancer is invasive and unpleasant, patients prefer to know for sure how frequently they need to subject themselves to the procedure. By contrast, the uptake of testing for other conditions may follow different patterns. For example, relatively higher numbers of individuals in families with breast cancer choose not to have genetic testing even when it is available to them. The complexity of decision-making around genetic testing for hereditary breast cancer has been documented in the

literature (e.g., d'Agincourt-Canning, 2006). The relatively low uptake rate can only in part be explained by the fact that the benefit of screening for hereditary breast cancer is a little less clear than for colorectal cancer and some other conditions.

The genetic professionals with whom I spoke also pointed out that gaining knowledge through the family[57] may lead to misunderstandings and misconceptions, but for the most part, prior familial exposure to genetic care was deemed likely to have a positive effect on clients' knowledge and understanding of genetics and the genetic condition. This is in contrast to the findings of Sermijn et al. (2004), who illustrate that even in cases where individuals convey genetic information to relatives, this communication is unlikely to augment family members knowledge and awareness of the genetic trait.

4.2.3 Education through media. A number of participants mentioned the role of the media in educating the public about genetics, primarily in terms of the media's role in misleading the public:

> ...you listen to TV, or read magazines or newspapers about a new gene for something, and sometimes this information is given in rather glowing terms. [#5]

[57] Kelly, Sturm, Kemp, Holland, & Ferketich, 2009 demonstrate that learning from a family member about genetic risk is favored to a more authoritative source of information.

> ....when genetics gets talked about in the media it's sort of as if it's here and it's for everybody and it's easy and so on, and sometimes there is misunderstanding.... [#4]

Participants explained that many clients have great difficulty in grasping the concept that a genetic test is a test for a particular family and has to be "worked out" in an affected family member first. With a known diagnosis, the genetic test is first performed in the proband[58] – if a mutation is detected, the proband receives accurate information about their disease risks as well as genetic risk information about family. This means that if there is a test identified for one family with hereditary cancer, for example, the same test is not available to another family that may appear to have the same condition, but for which the genetic basis remains unknown.

> Those people kind of have the impression from the media that, you know, there is a genetic test for everything, or they hear about one family having a genetic test – like the stomach cancer family – and they don't understand why their family can't just have the test; and you have to explain to them that there's mutations and different genes, and so on... regardless, they just want a test so badly. [#3]

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[58] A proband is an affected individual through whom a family with a genetic disorder is ascertained; a proband is usually the first affected individual in a family who brings a genetic disorder to the attention of the genetic service.

> The media is not talking about all those other difficult families that you can't figure out the basis. [#5]

The concern is that the media is giving an inaccurate impression that there is genetic test for every condition that appears to be hereditary. At the same time, the commentary provided by the genetic professionals suggests that the media has an important role to play in the effort to increase public knowledge about genetics.

Two interviewees spoke about the role of the internet as a source of information about genetics.

> My personal belief is that after we give them our own information, pretty much anyone who has access to the internet will look to see what they can find.[#8]

While not expressed as a significant concern by the genetic professionals I interviewed, there has been a great deal of attention paid in the literature to the potential harms of accessing genetic information as well as genetic testing through the internet, including the lack of oversight of test validity and utility (Caulfield, Ries, Ray, Shuman, & Wilson, 2010), the lack of licensed physician involvement, as well as the lack of consumer understanding of test results and interpretation (Robertson, 2009). Annes, Giovanni, & Murray (2010) address the mounting unplanned costs to the system and, most importantly, the questionable health value of tests accessed via the internet. Direct-to-consumer genetic testing is especially debatable as genetic counseling is typically absent in this type of service. A report issued by the US Government Accountability

Office (2010) argued that companies offering direct-to-consumer genetic testing engage in misleading marketing practices in an environment lacking consistency of results. These tests can be especially detrimental to pre-symptomatic individuals who need to be thoroughly post-test counseled and clinically managed.

In general, the genetic professionals I interviewed were concerned that public knowledge of genetics is inadequate and, even with information intervention through the genetic clinic (and possibly other sources), the level of understanding of genetics is insufficient for informed decision making about genetic testing. This confirms what Sturgis, Brunton-Smith, & Fife-Schaw, (2010) found about the effect of supplying the public with information about genetics and inheritance. Those authors demonstrate that providing such information does not generally translate into improved public knowledge and interest in the science of genetics. More importantly, it does not significantly alter fundamental values and beliefs. Patients' knowledge (or lack of knowledge) was found to be on par with the knowledge of the public at large (see also Calsbeek et al., 2007).

4.3 Barrier # 3: Client Attitudes

From the interviews, it became clear that genetic professionals invest a lot of time and effort in providing as much individualized information as needed to each client. Although some clients may contact the genetic clinic with further questions and requests for additional information, the majority of potential clients do not. Unsurprisingly, only some are interested in pursuing genetic care. Genetic professionals' perspectives on who seeks genetic care suggest that there are two main groups of clients: those who are very

much pro-genetics and those who are very hesitant. A subset of the first group of clients believes that genetic tests are able to provide definite answers. This subset of clients has difficulty coming to terms with the limitations of genetic testing. In contrast, those who are more hesitant to begin with are quicker to not follow up with potential genetic services, either because they remain unconvinced of the benefits of genetic testing or because they are reluctant to commit to engaging with family members as part of the process. These clients are also most likely to fail to show up for subsequent appointments.

The observations of the medical geneticists I spoke with are congruent with the work of Cooke & French (2008) who conclude that there is a direct connection between patient attitudes and their intentions to avail of genetic screening. These authors also underscore the importance of creating positive attitudes among patients, which translates into positive intentions that, in turn, enhance the odds for client uptake of genetic services.

An important observation made by the genetic professionals I interviewed is that the decision of potential clients about whether to pursue genetic testing is neither straightforward nor predictable – factors such as the level of understanding about genetics, perceived severity of the illness, and family dynamics are not in themselves predictors of clients' attitudes and behavior. Rather, some potential clients are simply not attracted to the idea of genetics and genetic care.

> Some don't like the idea of genetics. Those patients just don't show up for their appointments, right? [#8]

> My sense is that people, in general, that don't send in the forms and don't make the effort are people that end up not coming for their appointment often. It's a pretty good predictor of whether they are going to come. [#1]

> ...you look at the referral and you'll think - I need to run after this a bit more and you'll try to call them [clients] and you'll get some of the information over the phone, and you'll hand hold a bit more: and those are the people that more often than not...when you give them an appointment and then they don't come in. [#3]

These observations by the genetic professionals are insightful in that they reflect, rather than challenge, the meanings of genetic testing in the context of everyday life. As the limited qualitative research with those who decline genetic testing has begun to illustrate, decision-making is complex and not necessarily correlated in a straightforward way with a "knowledge deficit"[59] of genetic information (Cox & McKellin, 1999; Lock et al., 2006; Duncan et al., 2008).

[59] That is, the notion that users of genetic services or members of the general public are not sufficiently educated in genetic science and therefore make decisions that are somehow irrational with regard to using genetic services.

Unmistakably, the above excerpts exemplify the added effort genetic professionals invest with each client to ensure that clients are genuinely committed to and interested in genetic testing. Although none of the three participants cited above mentioned the link between patients' knowledge about genetic testing and attitudes to genetic testing (that is, clients' "attraction to" genetics), they seem to operate on the implicit assumption that full knowledge does not imply an attitude of attraction to testing. The genetic professionals reported that regardless of patients' formal education and intellectual ability, they only see individuals who are interested and have an attitude of "attraction to" genetic testing; experience has taught them that only the truly committed follow through with the whole process despite its complexities and challenges.

> It really varies. You have very, very smart people who are onboard the genetics train so to speak - and you have the opposite as well. [#3]

The participants explained that a good predictor for a client's commitment is the completion of the paperwork, combined with thorough family communication. None of the respondents was able to provide concrete numbers to better illustrate the client attrition rate after a contact with the genetic professionals had been initiated. The clinic does not keep statistics on how many clients receive information but do not follow through with a full range of genetic testing and follow up services.

4.3.1 Preparedness for and concerns about genetic testing. The genetic consultation process determines whether or not a client qualifies for genetic testing. Usually, the presence of a strong family history indicates that a person meets the high risk

criteria that identify them as eligible for testing. As discussed above, conveying eligibility information to the individual does not automatically result in a decision to pursue genetic care. In fact, the decision process is complex; it involves individual and family members, takes time, and can be emotionally charged.

4.3.1.1 Pre-symptomatic testing. Genetic professionals explained that pre-symptomatic genetic testing is offered to clients who are clinically healthy but deemed at risk for developing a particular genetic disorder. For those unaffected by a genetic condition, this means that a diagnosis is reached before the patient has clinically experienced disease symptoms.

Pre-symptomatic genetic testing may cause considerable distress, especially if no treatment is available (Graceffa et al., 2009). In those cases, thorough pre- and post-test genetic counseling and support is imperative regardless of the test results. For example, first degree relatives of patients with genetic disease may not be particularly interested in predictive testing if therapy is not available (Dahodwala et al., 2007).

There is a vast amount of research documenting the psychosocial distress associated with genetic testing (Cohen, 1998; Skirton, Frazier, Calvin, & Cohen, 2006; Duncan et al., 2008; Edge, 2008; Fanos et al., 2011). For example, Fanos and colleagues (2011) have described patients' unease when it comes to testing (in the absence of symptoms) and disclosure of test results. They have demonstrated that at each stage of the test process, patients have to cope with psychological issues, during decision making about whether to undergo testing, as well as during the process of deciding whether or not

to know the test results. The authors also describe the emotional distress at the stage when results are disclosed, even if the results are favorable. Patients may feel anguish because they have to alter their life long perception of self and revise plans to include or exclude genetic disease (see also Cox & McKellin, 1999; d'Agincourt-Canning, 2005, on the complexity of decision making around genetic testing).

The genetic professionals I spoke with did not raise the issue of distress related to "favorable" test results, but rather emphasized patients' experiences with a positive diagnosis. The participants referred to late-onset disorders where genetic diagnosis, although beneficial for patient treatment, brings a whole gamut of changes on the personal and familial level.

> Some patients are very anxious because they are at that stage [of life] where there is a complete sense of loss. [#1]

> Some people that are referred have been living with the disability for awhile, whether it's physical or cognitive and, you know, it had taken them time to adjust to the fact that they are different. [#8]

Respondents also raised the issue of patient distress associated with receiving an indeterminate test result – that is, a result that could not provide clear answers to the client in terms of their status as a carrier of a mutation. It is not unusual that a genetic test may have been completed on time, with clear-cut results, however it may not be clear

what those results mean and even more unclear what clinical management can be recommended based on those results.[60]

> I think there was one patient who was very angry with me recently, because she did not understand how her siblings had got the result and she hadn't. She was angry because she thought her test hadn't been run: in other words, we didn't do it. Again, it's often communication that's the problem. Her test had been run but we couldn't put it to one side of the fence or the other. It's just that she didn't have an answer. [#7]

The respondents indicated that in some cases, patients' concerns about the distress associated with learning the results are very strong and, despite discussions with the genetic practitioners about the benefits of testing, those clients remain averse to the idea of knowing their genetic risk status.

The interview data suggest that clients' attitudes about genetic care vary, but that those attitudes determine whether or not clients will avail of genetic care or will make an effort to communicate with relatives and "spread the word" about possible genetic risk within the family. This finding supports previous work arguing that personal attitudes towards DNA testing combined with adequate knowledge are major determinants of optimal utilization of genetic testing (Calsbeek et al., 2007).

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[60] Withholding genetic test result information for the benefit of the patient and their family may be ethically and professionally justified. These situations likely arise where genetic research is blended with clinical genetic care.

4.4 Barrier #4: Family Communication

Communication with the family and within the family is critical not only in terms of producing accurate pedigrees and assessing risk but also in terms of informing and possibly recruiting at-risk relatives. Genetic professionals reported that, although important, genetic information (including genetic risk) is not always communicated from clients to family members.

> Some people then are not willing to contact their relatives to ask them to sign the release form and things. So then we lose a certain subset of people this way. [#1]

> There are families that do not pursue things because the families just do not communicate well, and we all know families like that (chuckles). Yeah, they've lost touch with their family because, what happens a lot of times is the person referred may not even be a person who's had cancer themselves. It is because of their family history of cancer, but they're not able to go and contact their family members because either they've lost touch often because of cancer or because they've cut off contact. [#3]

While talking about the importance of communicating among relatives, one participant noted the distress that genetic professionals may experience when family members do not contact the clinic. The reasons can vary, however, not conveying the

information can be a key barrier to uptake of service by family members who are eligible for genetic testing.

> In terms of barriers, there is always this population within every family who never come to see me, and it is not because they live in another province: they live here. That makes me nervous about whether they're getting our information or not. If the relatives aren't telling them for whatever reason, then they are in the dark about it. [#2]

While discussing possible reasons for this reluctant behaviour, the participants mentioned that families do not always communicate well. Family dynamics vary, depending on degree of kinship, the closeness of relationships, and how often contact occurs. These observations confirm others' findings (Claes et al., 2003; Gaff et al., 2005; Koehly et al., 2003), that first-degree relatives are more likely to be informed compared to second and third degree relatives (Vos et al., 2011; Clarke et al., 2005).

Inevitably, staying in touch with families is not always achievable. Geneticists reported that lost ties, due to various family dynamics or geographical distance, were frequent scenarios in which a client may not be in a position to, or is not willing to convey essential genetic information to family members. Consequently, only a certain percentage of patients contact their relatives, even if the relatives live in the same or nearby communities. Interrupting the flow of genetic risk information provided by the geneticists compromises the effectiveness of genetic care for both the patient and their relatives and creates a barrier to the uptake of genetic advice and care. The challenges in

communicating genetic information to family members as reported by the participants came as a surprise in light of the fact that the population of the province is known to have very strong family and community attachments.

> I guess one thing I always wonder about is when I see a family that we've identified a dominant gene in, and say there are 20 siblings in that family, they all live in Newfoundland, and yet, I never see all 20 of them. They never come in. I am not sure whether they [patients] have conveyed the information properly to the other siblings or they are mad at their siblings and they don't talk to them....And we say, "Tell them [your siblings] by phone or in person or copy the letter that I sent you and just give it to all your relatives". But you know, not everyone is doing that and so this is a barrier. [#2]

The participants alluded to the difficulties their patients may have in communicating complex genetic information due to a lack of appropriate formal training. As previous studies have demonstrated, patients' lack of knowledge about the science of genetics may exacerbate barriers already in place due to family dynamics, creating an unfavourable context for conveying important genetic information to relatives (Mesters, Ausems, Eichhorn, & Vasen, 2005).[61] Other published studies have brought to light a wide range of factors influencing patients' ability to communicate genetic information to relatives: "In understanding why, and where, information is likely to be passed on,

[61] To counterbalance the impact of the deficiencies in circulating information within the family, Forrest, Delatycki, Skene, & Aitken (2007) suggest enhancing the support that genetic professionals provide to their clients to enable them to more easily communicate with family members.

account needs to be taken of cultural, familial and individual factors" (Forrest et al., 2003, p. 324).

4.5 Barrier #5: Level of Clients' Formal Education

Providers perceive the educational level of patients to be a serious barrier, especially as genetic information is complicated and understanding it can be confusing even for the well educated. Historically, the low rate of adult literacy in the province has been notable (Newfoundland and Labrador Royal Commission, 2003). One participant mentioned they have come across a number of clients who are either undereducated or illiterate; this participant was quick to note that these clients are nevertheless intelligent and that illiteracy should not be viewed as a deterrent to providing full education about genetic services.

> Though these clients did not learn to read and write, they should not be barred from having the right treatment and the right care. [#7]

Another informant confirmed that a client's understanding of genetic information depends to a large degree on the individual's level of formal education. Genetic professionals attested that, typically, they rely on written correspondence with clients, which may also include information pertaining to family members. During a consult, the genetic professionals provide clients with written pamphlets containing information about genetics and suggest that clients access additional information sources including websites. The issue of literacy was illustrated in a number of scenarios:

> We write to everybody and send a family history to everybody. But if you can't read your letter and your family history or be able to follow it or know exactly who a first cousin is.....I think that's a major barrier to genetic services and genetic testing here. [#7]

> We contacted this patient three times and they didn't come in. So therefore, we assume then they don't want to, when we don't have a clue whether they actually can read the letter in the first place.[#3]

It was also noted that if clients are contacted in the context of clinical genetic research, then the clients' education is not such an obvious barrier despite the fact that illiteracy is a particularly serious problem in the province. This is because clinical genetic researchers travel to various communities and establish personal contact with members of the affected families: direct verbal contact lessens the importance of attention to written correspondence with those patients. One genetic professional described this as a "very intensive, verbal connection." [#7]

> If they [clients] are illiterate, which quite a few people are in this province, the researchers can do that [provide information] verbally and they can do that so they know themselves what has been understood [by clients]. [#7]

Genetic researchers, then, have the opportunity to learn through direct contact with a range of family members about the family history without relying on written

communication between family members. Through these personal encounters within the family, genetic professionals acquire a sound understanding of the relationship among family members, even without asking specific questions. As well, researchers can directly supply information about genetics and genetic risk to clients and more immediately recognize whether or not the information has been understood and to what extent.

In the context of clinical genetic research, the challenges posed by client literacy and educational level may be less pronounced, but they still exist. Genetic professionals who are researchers report needing resources and time to travel in order to pay home visits to families and provide each member with genetic information specific to their family. This requires the presence of sufficient and continuous funding. One genetic professional explained that, for financial reasons, they increasingly communicate with clients over the phone although they fully realize the advantages of face-to-face contacts in providing an optimum environment for building rapport and facilitating interaction.

The literature has paid particular attention to the positive role of higher education in understanding genetic information (e g., Calsbeek et al., 2007). Fewer studies have addressed the role of adult illiteracy in the context of genetics (e.g., Erby, Roter, Larson, & Cho, 2007; Lubitz et al., 2007)[62]. The ability to read and write is particularly important in terms of the efficient collection of family history in creating accurate pedigrees.

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[62] A number of publications have shown that patient inability to read poses a quandary in the provision of clinical care in general (e.g., Grimes & Snively, 1999).

4.6 Barrier # 6: Client Fears

Genetic professionals reported that the need to respond to a client's emotional vulnerability by providing emotional support is as important as the effort to ensure that information is understood and retained and that the client's interest in pursuing genetic care is not compromised.

> You just take each person as they come and try to see what their feelings are and if you can help them. [#3]

Not surprisingly, the participants reported that clients may experience anxiety and fear that their genetic test results are going to be positive. They emphasized that clients often have unsettling thoughts about how their lives are going to be affected and how they are going to cope with a positive result. This was seen as particularly true for individuals with adult-onset genetic conditions, some of which can be life threatening.

> If you find out about your genetic condition at 50, there's not much you can do about it. There is no doubt that's a huge area of concern and a difficult issue to come to terms with. [#8]

Two of the participants reported that, typically, fear was not common among their clients and described their clients as generally fairly optimistic. They emphasized that the majority of clients believe they are in control of their own health regardless of test outcomes and that they are prepared to avail of screening to "avoid something catastrophic from happening based on that gene". [#4]

Genetic professionals explained that due to their training, they are constantly aware that the information they are giving to a client may cause emotional distress. When a patient is tested through the cardiac clinic, for example, and is found to carry the defective gene (which immediately puts their children at 50% risk), they typically experience anxiety and fear in relation to their children's health. Forrest et al. (2003) refer to this as "generational responsibility," whereby affected parents are eager to inform their children and even their nieces and nephews to ensure the timely disclosure of information to allow adjustments in life-course plans. Other studies (Tibben, Timman, Bannick, & Duivenvoorden, 1997) have demonstrated that carrier partners[63] with children were significantly more distressed than those without offspring. In keeping with these findings, the genetic professionals I spoke with reported that individuals without children may choose to not even have genetic testing.

There is a compelling body of evidence on the emotional impact that genetic testing may have, included elevated anxiety and clinical depression associated with genetic care (e.g., Jones & Clayton, 2012; Douma et al., 2008). The interviews in this research did not discuss this.

4.6.1 Role of gender. Only one participant identified gender as a barrier to accessing genetic services. The informant observed that Newfoundland men are very reticent about coming in for genetic treatment. According to the informant, women are considerably more receptive to accessing genetic services. Women also provide a great

[63] This refers to couples in which at least one of the partners is a carrier of a defective gene.

amount of encouragement and support to men, aiding the men to pursue genetic advice and care.

> The women try and make them come in and they don't want to come in. And that's sort of tied a bit with their education as well, you know, if they left school again at 12 and didn't really sort of get into the health care system. [#6]

> And he sort of grunted a yes because his wife was sitting next to him, you know, and… anyway, so on the day he was supposed to come in for this treatment the lab downstairs rang and said, "Your patient hasn't arrived". I know this is strange. So I rang his wife, who was at work, and she said, "The bugger! He's not turned up! I'll make sure he turns up. [#6]

The participant went on with the story to explain that the wife then alerted and mobilized the entire community and they located the husband, which turned out to be important, as the man had an affected status and bringing him in for the procedure in fact saved his life. This example may present an extreme case from a number of perspectives, however it serves to illustrate the point that men in general are deemed to be particularly disinclined and distressed when urged to seek genetic care. This respondent's amusing and lively observations are in agreement with the literature on the subject of gender and genetic care. A number of studies have shown that the genders engage differentially in

genetic testing for conditions where hereditability is gender neutral (Taylor, 2005; Creighton et al., 2003; Hayden, 2003).

4.6.2 Confidentiality. Genetic professionals commented that confidentiality is often something that their patients worry about. They reported that the concern is with whether or not one's genetic information is going to remain in the genetic clinic and, if not, to whom it might be disclosed. One participant described how genetic professionals typically respond to a patient's anxiety on the issue of confidentiality:

> Well, the family doctor doesn't get the genetic result. The genetic results are released to that patient and not to the family doctor. So right now, it's how we deal with that. I don't know if that's a perfect system or not. That's how we deal with it. [#4]

Another genetic professional explained that they ask their clients if they prefer to have their genetic information included in their hospital chart. If clients approve, only then does the information become part of the record. However, the participant admitted that they recommend that the family history remain part of the record in case a client is in a medical emergency and cannot speak for themselves. Alternatively, releasing genetic information, including results from genetic testing, to family members is accomplished only through a valid consent.

One participant added that observing confidentiality is challenging in cases where they see multiple members of the same family.

> You can imagine how that becomes difficult when I've already seen 10 people from his family and I am seeing the 11th one here and I am trying to explain to them why they're here, and I have to say, like, your aunt and uncle have the disease. [#2]

> I say like, yeah, there's a reason why you're here. And have to explain it in general terms and usually they know…obviously, they know that these people are affected, but every once in awhile they they're like--oh, I didn't know he had it. You know, like there is a bit of info that they may not know, and it's always this balance of trying to be…trying not to give too much info about their family but enough so that they understand the significance of the disease in their family and how it's affecting people. I mean, technically, that's a breach of confidentiality, but you have to decide what and how much. [#3]

Evidently, the difficulty is around weighing the commitment to confidentiality against the benefit of releasing important information to a relative to better illustrate their need to pursue further genetic assessment and treatment.

> In extreme circumstances, you can breach confidentiality for some very good reason. Then you have to have a whole ethics meeting about it and get more than one person to agree with you that you really need to go that route and breach that confidentiality. [#1]

This concern expressed by the genetic providers echoes the ethics debate on the issue of when and where a confidentiality/privacy breach is acceptable (Sudell, 2001; Knoppers, 2002; Pullman & Hodgkinson, 2006). Interestingly, only one informant observed that confidentiality and privacy are expressed in a rather peculiar way in Newfoundland, especially in rural areas. In the participant's view, everyone in a given community "knows everything about everyone" and to substantiate the statement, provided an example where an entire family history had been presented to the geneticist, without a request, by a person who was not even related to that particular family.

> I do not know, but these stories are very interesting because, you know, they would never happen anywhere else I've ever worked. [#7]

It appears that, in rural Newfoundland, the sense of community and belonging to a community is generally stronger than the sense of being an autonomous individual with privacy rights. In those remote areas where everybody looks after everybody, confidentiality of genetic information may be a non-issue for some individuals. Undoubtedly, rural Newfoundland has its own distinct culture which filters through the way genetics is practiced there. The genetic professionals I spoke with emphasized that the lack of confidentiality among community members and family members is not deliberate or with the intention to harm; it is part of the mutually supportive culture of the small isolated outport communities.

4.6.3 Discrimination. Informants reported that the most important psychosocial barrier to the uptake of genetic services is patient awareness and knowledge, but the

second major concern is fear of discrimination, whether related to employability or insurability. Concerns about insurance discrimination have been well described in the literature (Cameron, Sherman, Marteau, & Brown, 2009; Morren, Rijken, Baanders, & Bensing, 2007). The genetic professionals I spoke with provided concrete examples of how insurance and employment discrimination have discouraged their patients to further pursue genetic services.

> [T]he biggest problem with genetic testing is the fact that there might be problem with insurance. A far as I am concerned, that's the very biggest thing. That's what turns people off and that's what worries people. [#6]

> The biggest concern that people have is whether they won't be able to get a mortgage because of the insurance...whether, you know, there's job discrimination or won't be able to get life insurance. If they weren't that worried, then far more people would be able to make a decision on whether they want a genetic test. Because they don't know what might happen with that insurance kind of issue, they may put it off even though they, for their medical, would like to have a genetic test. [#1]

> If people are going to say no, in my experience, it's mostly because of this concern about what's going to happen with insurance. [#2]

> The other big issue is about the insurance. What I say to them is that their charts do not merge with their medical chart. But the insurance companies are now getting smarter and they say, "Have you ever seen a genetic professional? Have you ever had a genetic testing for a genetic disorder?" So that's the other big issue that … you think you're one step ahead and then the insurance will just get one step ahead faster. [#7]

Other examples given by the interviewees demonstrate that some clients hear about denial of insurance from others, usually relatives. Because of prior, although indirect, exposure to insurance implications, clients are concerned.

> They'll say, you know, "My brother — he can't get insurance because he has polycystic kidney disease", you know, or "My brother told me that he couldn't get insurance because he told them that he got this test done". So, yes, because more and more people are getting into insurances and stuff — then, yeah, it's…they're becoming more aware of it. [#5]

The examples provided by the genetic professionals have certainly shaped their own beliefs that the fear of discrimination is a roadblock for clients and is a potent deterrent to the uptake of genetic care. This is in concert with findings in other published studies on the issue of fear of genetic discrimination among patients (e.g., Powell, Chandrasekharan, & Cook-Deegan, 2010; Peterson et al., 2002).

Contrary to what the majority of participants reported, some researchers in

Canada argue that insurance discrimination based on genetic status is a non-issue. While they acknowledge that there is a fear of genetic discrimination, they point out that the fear is not substantiated and there is no need for a genetics-based antidiscrimination law in Canada[64] (Lemmens, Pullman, & Rodal, 2010; Lemmens, 2003). Regardless of the debate on whether or not Canada needs legislative protection against genetic discrimination, the perspectives of the genetic professionals I interviewed clearly indicate that clients' fear of insurance or employment related genetic discrimination is a barrier to the use of genetic testing.

4.6.4 Social stigma. One genetic professional raised the issue of social stigma related to genetic status. The respondent shared a story of a woman who had reported that the community had singled out her family because of their particular genetic condition.

> She said, "All my children are grown and they're moving away and I don't want them involved with this, because", she said, "it used to be a joke in the community -- they would say, 'don't get involved in so and so because they're going to die on you anyway'". So that was really difficult. So she said, like, "I don't want to be involved in this [genetic testing]. I know the

[64] In counterpoint to that argument, health reporter Theresa Boyle (May 18, 2011) has described the case of a healthy 25 year old Canadian chiropractor who had been consistently denied mandatory insurance needed for her practice because her father has Huntington's disease. To provide coverage, the insurers insisted on proof that the young professional did not have a carrier status. Boyle's article highlights the view of the Canadian Coalition for Genetic Fairness that "Canadian jurisdiction lags behind jurisdictions of other developed countries in acting against genetic discrimination." Retrieved from http://www.healthzone.ca/health/newsfeatures/article/992995--young-woman-faces-insurance-hoops-due-to-father-with-huntington-s

information might be good, but like we've had enough with this already". [#7]

When asked how frequently the clients express fear of stigmatization, the respondent commented that this scenario is an isolated occurrence. Although other participants did not discuss stigma as a barrier to availing of genetic testing, it is worth mentioning that stigma related to genetic conditions has been described in the literature (Williams et al., 2010; Smith, 2007). Smith (2007), citing Meiser, Mitchell, McGirr, Van Herten, & Schofield (2005), suggests that the heightened awareness about the genetic basis of diseases and particularly genetic testing may lead to labeling, stigma and further discrimination.

Chapter 5: Synthesis of Findings and Recommendations

This chapter provides a précis of key findings that emerged from the commentaries of the genetic professionals whom I interviewed. The synthesis is followed by recommendations for enhancing clinical genetics practices within the province of Newfoundland and Labrador. The Newfoundland and Labrador case study is then used as an heuristic device for informing genetic services more generally. Attention is given to pertinent policy considerations. Limitations of the study are also discussed together with recommendations for further research.

5.1 Synthesis

The unique geography, history and culture of the province of Newfoundland and Labrador frame whether and how genetic services are accessed and used. The genetic professionals who participated in this study discussed both the successes of the PMGP and the barriers to accessing and using the clinical services offered through that program. Participants also underscored the link between clinical genetics and genetic research. Participants expressed hope that the findings of this study will give voice to their concerns and also help bring about changes for improved professional practice and efficient service delivery to patients.

Issues raised by the genetic professionals illuminated the complex and multi-directional relations and transactions involved in the provision of genetic service: between them and their clients, between them and clients' family members, between them and the system, between clients and their family members, between clients and the

system, and so on. Because of this intricacy, the underlying themes derived from the data are diverse yet never distinct. The themes that emerged about access-uptake roadblocks to genetic services were grouped into two broad categories — systemic and psychosocial barriers — each comprised of subcategories. The separation between the two types of barriers and between subcategories was intentionally and artificially constructed as a means of organizing the findings and clarifying ambiguity in the analysis and translation of what the medical professionals conveyed.

5.1.1 Systemic barriers. Systemic barriers refer to practices or situations in the current genetic services which were reported by the genetic professionals to limit or exclude certain patient groups from access-uptake of those services.

Geography was identified by the genetic professionals as a powerful barrier to accessing and using genetic services. Although discussed as a separate barrier, geography permeates a whole range of challenges identified by the research participants. The findings of this study confirm previous research suggesting that remoteness from major urban centers poses significant barriers to accessing efficient genetic care and achieving better health outcomes.

Access to genetic services is not equal for everybody. The genetic professionals I spoke with reported that a significant proportion of the population in the province does not have a family physician and that the turnover rate of medical professionals in the rural/remote areas of the province is high compared to the rest of the province. They commented that the lack of family physicians is a major barrier to accessing their

specialty care, which is largely dependent on the referral mechanism. They also expressed concern about the levels of genetic competence, as well as attitudes towards population genetics, among non-genetic medical professionals.

Participants explained that access to genetic care and related medical tests is accelerated if patients are enrolled in a research study.

There is a consensus that the delivery of genetic services requires considerable expense, including individual costs, costs to the province, societal costs, and costs to research and development. Although the genetic professionals did not explicitly categorize costs into types, particular emphasis was given to the high cost of running a genetic clinic and performing genetic tests with limited material and human resources. The participants discussed the high cost to individuals particularly where curative or preventive intervention is not available.

Limited personnel at the genetic clinics and heavy workload of genetic professionals were discussed by the participants as principle reasons for long wait times. According to the respondents, the number of geneticists in the province is far from adequate and it is exigent to secure continuum of care for patients by genetic professionals and/or other specialists.

Not all participants agreed that there are inconsistencies in the genetic knowledge held by family physicians and medical specialists (non-geneticists). Although there was a divergence of views, the participants who reported inconsistencies reiterated that those inadequacies are a barrier to access-uptake of genetic care. Some of those views are well

supported in the literature, suggesting that family physicians and medical specialists should seek continuing education in clinical genetics. However, lack of knowledge about patients and families within a community was emphasized as a barrier.

Genetic professionals also discussed challenges to their services associated with already referred patients (*post-referral attrition).* The participants explained several mechanisms through which the genetic clinic loses referred patients, however they emphasized two main ways: patient reluctance to decline a referral from their family physician or medical specialist (even when they have no intention of following through with the appointment); and the complexities of completing family history questionnaires in relation to managing difficult family dynamics.

5.1.2 Psychosocial barriers. This category was constructed to synthesize genetic professionals' understandings of client psychological and social barriers to accessing genetic care. Lack of awareness about genetic services was identified by the participants as an important reason why patients may not avail of genetic care – some professionals described it as being the most important barrier to the effective uptake of genetic services. Interviewees indicated that the PMGP approaches the issue of public awareness of genetics with caution – the rationale is to curb increased demand for the services and avoid generating unnecessary fear of genetic disease among members of the public. In the same vein, all genetic professionals I interviewed unreservedly acknowledged that their clients' understanding of genetics is minimal. The respondents were concerned that the concept of genetics remains abstract and difficult to understand for clients, even after

information intervention. They also emphasized the challenge of education about genetics given some of the deeply rooted misconceptions about disease and inheritance that tend to be transmitted within families or communities.

Patient attitudes toward genetics and genetic care were also identified as a barrier. The genetic professionals I spoke with underscored the importance of these attitudes and highlighted that patient mistrust of the benefits of genetic testing is evident even among those who make a conscious effort to learn more about genetics. The observation that patient attitudes affect decisions about whether to pursue genetic care in a manner that is neither straightforward nor predictable concurs with prior research.

Lack of communication within the family also poses a barrier to access to and uptake of genetic care. Genetic information obtained through the genetic clinic is not always consistently communicated between and among siblings and other family members. The genetic professionals I interviewed expressed worry that patient relatives may not be adequately informed about their genetic risk. Lack of communication within families may be due to family dynamics, disease or death of a relative, lack of personal motivation on the part of the client, and myriad other reasons. However, an inconsistent flow of information among relatives may also be explained by deficiencies in patients' genetic knowledge and skill level to convey complex genetic information or fear that (not) conveying genetic risk status within families may further complicate difficult family relations.

Genetic providers indicated that the formal educational level of patients is a serious barrier to the pursuit of genetic care. The participants noted that given the low rate of adult literacy in the province, they do not exclude illiteracy as a factor when clients fail to respond to written communication. Communicating with those clients in person or via telephone may, to a certain extent, ensure that the information is received and possibly understood; however, this technique is time- and resource-intensive. Genetic professionals discussed patient fears as a stumbling block to accessing and using genetic services. Some pointed out that the fear of discrimination, particularly insurance-related discrimination, is the second largest barrier (geography being the largest) to genetic testing. Participants spontaneously spoke about their patients' concerns regarding confidentiality -- specifically, having their genetic information disclosed (inadvertently or intentionally) for purposes unrelated to health.

Gender was also identified as a barrier to actively seeking and persisting with genetic care. And, finally, one genetic professional mentioned the fear of potential social stigmatization and explained that, because of this fear, availing of genetic testing to confirm disease is perceived by some patients to be an upsetting experience and is likely to be avoided.

5.2 Recommendations: Local Context

The results of this research point to specific policy and process changes that could enhance access to and uptake of genetic services in the province. As emphasized throughout this thesis, the unique social, economic, political, and cultural contexts of the

province shape how genetic services are currently used; these contexts also necessarily shape whether and how genetic services can be made more effective.

Recommendation #1: Careful consideration should be given to human resource projection and planning to ensure an adequate number of genetic professionals is employed within the province to help ease the current workload of geneticists. Sufficient professional human resources are also likely to reduce wait times, better respond to underserved areas, and make work processes more efficient both within the clinic and in outreach with clients and families.

Recommendation #2: The current PMGP should rethink ways of expanding its services to additional geographic areas, such as the Labrador portion of the province. By utilizing novel or existing structures and resources, a number of possible options can be considered such as additional travel clinics, new permanent clinics, or services through community based centers. Service delivery through less traditional means, e.g., tele- and video-conferencing can be utilized more frequently and in a way that facilitates patient access. Whatever form is selected, close proximity to home will ensure uncomplicated access to genetic services and will make it easier for patients to seek genetic care.

Recommendation #3: A provincial education strategy in genetics is needed to target both medical students and practicing physicians. It should include programs and processes for improved and continuous genetic education, including updated guidelines for referral to genetic services, and clinical skills training on how to effectively discuss genetics with patients. This strategy will help enhance the delivery of genetic services,

and genetic counseling in particular, to ensure that adult clients do understand complex genetic information and are comfortable conveying it to their family if they choose to do so. Along with a provincial education strategy, enhancing the undergraduate and postgraduate medical school curricula to include further training in genetics may be a desirable goal.

Recommendation #4: A second facet of a provincial education strategy should address public education in genetics, starting with age appropriate information through the school system[65]. This way, members of the public will have an opportunity to learn about genetics early in life, allowing them to gradually develop familiarity with genetic care. This approach is likely to spark discussions (spontaneous as opposed to on demand) about genetics with and among family members. This in turn may help overcome fears of or hesitancy with availing of genetic care.

5.3 Implications: General Context of Genetic Services Policy and Planning

This research points to several important and innovative directions for theory and methodology on knowledge translation with regard to genetic research. First, the research results underscore the importance of attending to the voices of front-line service providers. This group of key informants, with the vantage points of proximity to end-

[65] Currently, the K-12 science curriculum in Newfoundland and Labrador (guided by the pan-Canadian science framework) introduces genetics to grade 9 students (Intermediate Science, Reproduction). Genetics-related themes such as stem cell research and genetic disorders are included in Biology 3201 for grade 9 students who self-select this course. Although these options provide a valuable glimpse into the captivating world of molecular genetics, the broader implications of genetics, genetic technologies and genetic services are not part of the discussion.

users and meta-level gaze of the system in general, enables a perspective that is both unique and valuable.

Second, the research points to the need for attending to simple and cost-effective changes to the system as a means of improving service delivery and uptake. For example, the cumbersome nature of family history questionnaires clearly impacts post-referral attrition rates in the province of Newfoundland and Labrador. On this point, the interview data raise interesting questions about whether and how restructuring questionnaires can facilitate both intra-familial communication and improvement of client abilities to "stick with" genetic services available to them. Further research on the understanding and use of family history questionnaires is needed. Locally, an analysis of the effects of introducing the improved version of the family history questionnaire by the PMGP could serve as a useful case study to this end.

Third, the research raises interesting questions about the "service-marketing vs. fear mongering" dilemma. Literature in the field of genetics has highlighted the problem of "geneticization" and the "marketing of fear"; the literature has also emphasized problems associated with the "knowledge deficit" about genetic information among the lay public. Yet the relationship between the two phenomena (knowledge deficit and marketing of fear) has not been adequately addressed. The interview data point to the need for research on the economic and social implications of having a population that seeks out genetic testing services. Specifically, the data point to the need for analysis of

the tension between the goals of public education about genetics, and the reality of scarce resources to offer genetic services in a publicly funded health care system.

The study raised serious questions about whether and how insurance discrimination results from genetic testing. Further research is clearly needed and, if discrimination is indeed found to be occurring, it is important to introduce regulations that prohibit genetic discrimination in Canada. While setting clear rules for the insurance industry, it will guarantee protection and will alleviate fears of discrimination among clients. It will also lessen hesitance among clinicians who may refrain from referrals for fear their patients may be discriminated against.

Participants only touched on some cost issues related to the delivery of genetic services, but their concern about cost clearly underscores the need for cost-effectiveness/cost-utility analyses. Those analyses should include not only the costs to the health care system, but also personal and societal (non-health sector) financial gains and losses attributed to genetic services. Considering those factors will provide a valuable perspective on the impact that genetic services have on clients, families, communities and the province.

5.4 Study Limitations

There are a number of limitations associated with this study and it is expected that future research will address them. Although I conducted interviews with a diverse range of genetic professionals, nonetheless the data is necessarily drawn from a relatively small and homogeneous group of genetic professionals. Secondly, the genetic services I discuss

in this study are provided from and within one system and are all part of the public health system. Comparative work with views of genetic professionals in private settings or with those employed in other provinces or countries may provide insights on the (in)efficiencies of various types of systems and draw attention to similar or dissimilar challenges. Another limitation is that all genetic professionals who fit the recruitment criteria were women, reflecting the fact that the majority of the genetic professionals in the province are female. Subsequent research may highlight differences between male and female genetic practitioners' perspectives on barriers.

When interpreting the findings of this study, caution should be applied to the fact that the statements of the genetic professionals are only snapshots in time and ensuing changes in their practice and the way they recruit, diagnose, counsel, follow up or treat clients are not reflected in this study. Regardless of those limitations there are lessons learned about the challenges in accessing and availing of genetic services. Those lessons can, and in fact should, be considered in designing an improved framework for the delivery of genetic care within NL and more broadly.

Research on patient's perspectives was intentionally not part of this study, as the published research has attended well to this perspective. Within the local context, however, research is still needed to juxtapose the data on providers' perspectives on barriers to access-uptake of genetic services with those of the patients' themselves, in order to inform service delivery in the province. The extent to which the two points of view are consistent or diverge will be key information for understanding how to provide

socially and economically appropriate genetic services in a way that responds to the infrastructural and psychosocial challenges faced by those who choose to avail of genetic services.

5.5 Final Notes

This study contributes to the existing knowledge by providing an authentic account of genetic professionals' perspectives on how the services they provide are organized, accessed and delivered and what access-uptake challenges their clients face on both systemic and psychosocial levels. This research also contributes to our knowledge about why certain individuals choose not to avail of genetic testing services, a challenging perspective to access.

Adequately resourced and better organized genetic care has the potential to facilitate appropriate access to and uptake of genetic service. The study provides a source for strategic direction to healthcare decision makers and health policy makers regarding short and long-term investments in genetic screening and testing in this region and beyond.

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APPENDICES

Appendix A
Invitation to Participate in Research Study

Dear XXX,

My name is Valerie Darmonkow. I am a Master's student in the Division of Community Health and Humanities, Memorial University. I am conducting research on *Genetic Professionals' Perspectives on Barriers toward Access to and Uptake of Genetic Services*. This study is part of the Atlantic Medical Genetics and Genomics Initiative [AMGGI], (Terri-Lynn Young, PI). The AMGGI project involves a significant qualitative research component, which seeks to understand the social, historical, cultural, and economic barriers to access to and use of genetic services from the perspectives of patients, providers, and the public. My subproject involves individual interviews with 7-12 genetic professionals.

As you are a genetic professional working in the Atlantic region, I am inviting you to participate in an interview that will take approximately 45 minutes to one hour. Your professional experience, knowledge, and insights would be of great assistance in understanding the barriers to access to and uptake of genetic services in our region.

To aid you in the process of deciding whether to participate, attached are a short description of the study together with a *Consent to Take Part in Health Research* form. Confidentiality issues are taken into consideration and thoroughly explained in item 8 of the form. Should you choose to take part in this interview, please reply to this email, and indicate:

(a) Possible dates and times for the interview over the next 6 weeks (see attached calendar)
(b) The location most convenient to you for the interview

I would like to thank you for taking time from your busy schedule and responding to this invitation. I am looking forward to hearing from you.

Sincerely,

Valerie Darmonkow
MSc in Medicine Candidate
Division of Community Health & Humanities
Faculty of Medicine
Memorial University of Newfoundland
Tel: (709) 754-8740 or (709) 777-7284 (Dr. Fern Brunger, Supervisor)
Email: mdarmonkow@nf.sympatico.ca

Appendix B
Consent Form

Faculty of Medicine, School of Nursing and Pharmacy of Memorial University of Newfoundland; Eastern Health; Newfoundland Cancer Treatment and Research Foundation

Consent to Take Part in Health Research

TITLE: Genetic Professionals' perspectives on barriers towards access and uptake of genetic services

INVESTIGATOR(S): Valerie Darmonkow

SPONSOR: Genome Atlantic

You have been invited to take part in a research study. It is up to you to decide whether to be in the study or not. Before you decide, you need to understand what the study is for, what risks you might take and what benefits you might receive. This consent for explains the study.

The researchers will:

- discuss the study with you
- answer your questions
- keep confidential any information which could identify you personally
- be available during the study to deal with problems and answer questions

If you decide not to take part or to leave the study this will not affect your relations with researchers involved with the Atlantic Medical Genomic and Genetics Initiative.

1. Introduction/Background:

   You are being asked to be interviewed about your perspective on barriers to access and uptake of genetic services. This research will constitute a Master's of Science thesis and it part of a broader research project on ethics and genetics which aims to understand the social, historical, cultural and economic barriers to access and use of genetic services. That project is itself part of a large-scale science project, AMGGI, funded by Genome Canada.

Initials: ____

2. Purpose of study:

This research examines barriers to uptake of genetic services from the perspectives of genetic professionals working within Eastern Regional Integrated Health Authority of Newfoundland and Labrador. The objective of this research is to identify and provide analysis of barriers towards access and uptake of genetic services from the perspectives of genetic professionals. The study will be based on qualitative interviews with 7-12 genetic professionals who provide information, counseling and support to families at risk. Three representative genetic conditions, the focus of the broader AMGGI study, will be emphasized in the qualitative inquiry: Arrhythmogenic right ventricular cardiomyopathy (ARVC), hereditary hearing impairment, and colorectal cancer.

3. Description of the study procedures and tests:

You will be interviewed by the investigator, Valerie Darmonkow. The interview will take approximately one to one and a half hours and it will be audiotape-recorded. You will be invited to describe and reflect on issues, concerns and memories that you identify as important in understanding the barriers to access and uptake of genetic services in the Atlantic region. In addition, you will be invited to reflect on the effectiveness of genetic screening and testing.

You may refuse to answer and question and can turn the tape-recorder off or have any portion of the tape deleted if you wish. If, following the interview, you find that you have additional information to convey, or if you feel you may not have expressed your beliefs adequately during the interview, you may contact us to have a follow-up telephone interview scheduled at your request. You may be re-contacted after the interview for clarification or further information.

4. Length of time:

Your interview will last approximately 1½ hours. It is possible that you will be invited to participate in a follow-up interview if I require additional information or wish you to explain on points you have discussed.

5. Possible risks and discomforts:

Due to the small number and high profile or genetic professionals in this region, I cannot guarantee anonymity. If you choose to participate in this study you may be at risk of having your identity inadvertently known by colleagues who read publications arising from this research. This may lead to social harm to you, should you be expressing points of view that may be at odds with the broader community of genetics/genomics researchers and clinicians.

Initials: _____

If any comments are attributed to you as an identifiable individual in resulting presentations/publications, you will have prior opportunity to accept or reject that attribution. As well, you will be given the opportunity to review publications and affirm or correct or correct any statement that may be directly or indirectly attributable to you. That is, any information to be used in publications that reflects or quotes your perspective will be offered for review and approval by you; and the information will be adjusted accordingly prior to publication.

You can choose whether or not you wish to be *explicitly identified* as having participated in this research. At any time you can reverse this decision.

Even if you choose to participate in this interview as an *unidentified* source of information, I cannot guarantee complete anonymity.

You will not be asked to disclose confidential information about yourself or your clients. You can refuse to answer any question and can request that I turn the tape-recorder off or have any portion of the tape deleted.

6. Benefits:

It is not known whether this study will benefit you.

7. Liability Statement:

Signing this form gives us your consent to be in this study. It tells us that you understand the information about the research study. When you sign this form, you do not give up your legal rights. Researchers or agencies involved in this research study still have their legal and professional responsibilities.

8. Confidentiality:

You may choose whether or not to be identified as having participated in this research; and at any time you can reverse your decision. To minimize the risks of misattribution or risks to reputation, when any comments are attributed to you as an identifiable individual in resulting presentations/publications, you will have prior opportunity to accept, reject or correct that attribution.

Should you choose to participate in this interview as an unidentifiable source of information, I will treat you as such. However, due to the small size of the interviewee community (genetic professionals working within Eastern Health of NL), it is essential that you fully understand that in the process, there could be a potential loss of confidentiality. Given that you, as a participant, may be identifiable even with full precautions about privacy and confidentiality, you are

Initials: ____

asked to choose whether you wish to be explicitly identified as having participated in this research.

Interview tapes and transcripts will be identified by number, and will be accessibly only to the interviewer, her supervisor and transcriptionist (listed below). The interview data may be used in future studies by Valerie Darmonkow or Dr. Fern Brunger in the next five years. It will constitute part of the data for the broader AMGGI project. Interview data will be destroyed at the end of five years.

Transcripts will not contain identifying information. A code number will be assigned, and that information will be securely stored in a locked filing cabinet apart from the transcripts themselves. As well, once the tape is transcribed, the investigator will block out/remove any potentially identifying information contained within the text of the transcript.

9. Questions:

If you have any questions about taking part in this study, you can meet with the investigator who is in charge of the study at this institution. That person is:

Valerie Darmonkow: (709)754-8740 or mdarmonkow@nf.sympatico.ca

Also, if you have any questions about the study or want further information you can contact Dr. Fern Brunger (supervisor, co-investigator on GE^{3}LS component) at the Faculty of Medicine, Memorial University, at 777-7284/ or fbrunger@mun.ca

Or, you can talk to someone who is not involved in the study at all, but can advise you on your rights as a participant in a research study. This person can be reached through:

Office of the Human Investigation Committee (HIC) at (709)777-6974
Email: hic@mun.ca

Initials: ____

Signature Page

Study Title: Genetic Professionals' perspectives on barriers towards access and uptake of genetic services.

Name of principal investigator: Valerie Darmonkow

To be filled out and signed by the participant:

	Please check as appropriate:
I have read the consent [and information sheet].	Yes { } No { }
I have had the opportunity to ask questions/to discuss this study.	Yes { } No { }
I have received satisfactory answers to all of my questions.	Yes { } No { }
I have received enough information about the study.	Yes { } No { }
I have spoken to Valerie Darmonkow and she has answered all my questions.	Yes { } No { }
I understand that I am free to withdraw from the study - at any time - without having to give a reason	Yes { } No { }
I understand that it is my choice to be in the study and that I may not benefit.	Yes { } No { }
I agree to be audio-taped.	Yes { } No { }
I agree to take part in the study.	Yes { } No { }

_____ _____

Signature of Participant Date

_____ _____

Signature of Witness Date

To be signed by the investigator:

I have explained this study to the best of my ability. I invited questions and gave answers. I believe that the participant fully understands what is involved in being in the study, any potential risks of the study and that he or she has freely chosen to be in the study.

_____ _____

Signature of Investigator Date

Telephone number: _____

Initials: ____

Appendix C
Interview Guide

Section A: Introduction

- Thank you for agreeing to participate in the study. Ice breaker.
- Offer to review study information: I hope you have read the information about this study that was sent out to you.
- Provide opportunity to ask questions: Do you have any questions about the research?
- Brief interviewee about ethics/confidentiality and review consent form; obtain verbal answers to the signature page; interviewee signs the consent form (Those interviewed via phone have already returned the consent form via fax prior to the interview).

Section B: Background information

- I understand that you are working as a genetic professional within the Provincial Medical Genetic Program. Tell me something about the Program.
- Please describe the nature of your work. What roles and responsibilities do you have? How long have you been practicing? What are the qualification requirements for your role?
- In approximate terms, how many adult clients do you see at the clinic in a course of one year?
- Please explain where your clients reside? Are they all from St. John's?

Section C: Specific experiences regarding various aspects of service delivery

- Would you please discuss the way individuals are referred to the clinic?
- How efficient is this mechanism? What could be improved?
- Would you please describe what guidelines and protocols are followed in the clinic?

- Please provide a sense of how long the wait times for genetic services are. What are the issues?

- According to you, what challenges, if any, do genetic professionals encounter in serving their clients? Tell me about strategies you apply (or you are aware of) for overcoming these constraints.

Section D: Perceptions about clients

- What do your clients know about genetic services?

- Where do your clients obtain information about genetics and genetic diseases?

- What are your clients' attitudes toward genetic services?

- Please help me understand what concerns or fears, if any, clients have regarding genetics and genetic testing. Tell me about strategies you apply (or you are aware of) for overcoming these constraints.

- What other challenges do your clients experience in pursuing genetic care?

Section E: Wrap-Up and Closing

- I wonder whether, based on your observations or experiences, you would like to share any additional thoughts or provide further comments regarding barriers to access-uptake of genetic services.

- Thank you.

- Mention about possible post-interview contact (note if in agreement): In case further clarification is needed, I hope you do not mind if I contact you. Likewise, if you want to share additional thoughts that may come up with after the interview, you are welcome to contact me.

- De-brief.