EXPLORING THE IMPACT OF LONG QT SYNDROME:

PERSPECTIVES FROM A BRITISH COLUMBIA FIRST NATIONS COMMUNITY

by

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Abstract

There is a disproportionately high rate of hereditary Long QT Syndrome (LQTS) in Northern British Columbia First Nations people, partly due to a novel missense mutation in KCNQ1 (V205M). The effect has been previously described (Arbour et al, 2008) predisposing those affected to syncope, arrhythmia and sudden death. A community based participatory research approach has enabled over 250 community members to take part, identifying more than 40 carriers of the mutation. Although a great deal of previous research has been carried out on the biological aspects of LQTS, there has been little study into the impact of living with a mutation that predisposes one to sudden death, and no previous studies have provided cultural insights into the issues a remote First Nations community might face. The goal of this thesis was to explore what facilitates and hinders resiliency and coping for those living with LQTS. Participants were invited to partake in their choice of one to one interviews, Photovoice, and Talking Circles. Interviews were recorded, transcribed, and analyzed qualitatively using the Systematic Text Condensation method. Twelve women shared their personal experiences of living with LQTS; eight participated in individual interviews, two participated in the Talking Circle, and two participated in both. Six of the women had known mutations, one was mutation negative, and five were awaiting genetic results. Most had affected children. In general, learning about a LQTS diagnosis was perceived as traumatic, with gradual acceptance that lead to coping. The main factors that facilitate resiliency and coping were positive family relationships, spiritual faith, and knowledge about LQTS. The main factors that

hinder resiliency and coping were a poor understanding of the biological or clinical aspects of LQTS, conflicting medical advice, especially about necessary physical restrictions, and LQTS not being taken seriously by both social contacts and health care providers. It appeared that learning to live with LQTS is an ongoing process, requiring balance and interconnectedness between all aspects of wellbeing. These issues warrant further exploration. Recommendations to enhance genetic counselling within FN communities will be presented to reflect the Medicine Wheel concept.

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List of Abbreviations

ABGC	American Board of Genetic Counseling
AHP	Aboriginal Health Program
ASHG	American Society of Human Genetics
BC	British Columbia
CAGC	Canadian Association of Genetic Counsellors
ECG	Electrocardiogram
FN	First Nations
GC	Genetic Counselling
GHS	Gitxsan Health Society
LQTS	Long QT Syndrome
MW	Medicine Wheel
NSGC	National Society of Genetic Counselors
TC	Talking Circle

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My best friend, confidante, and sister

You taught me to live in the moment

I love you so much!

Co-Authorship Statement

Versions of Chapters 2 and 3 are currently being prepared for publication by Huisman, L., Arbour, L., McCormick, R., & Gitxsan LQTS Research Advisory Board. I primarily designed this project, with advice from LA, RM, and GLRAB. I collected the data, did the analysis in collaboration with LA, and personally wrote the manuscript. LA and RM supervised the research, with oversight from GLRAB. LA edited the manuscript.

A version of Chapter 4 is currently being prepared for publication by Huisman, L., Arbour, L., McCormick, R., Martin, S., & Gitxsan Nation. I primarily designed this research project, with advice from LA, MR, and GLRAB. I collected the data, did the analysis in collaboration with LA, and personally wrote the manuscript. LA and RM supervised the research, with oversight from GLRAB. LA and SM edited the manuscript.

Chapter 1 Introduction

1.1 Overview

The health of Canada's Aboriginal (First Nations, Inuit, and Metis) peoples is poorer than the general population. Health Canada is the federal department responsible for helping the people of Canada maintain and improve their health. Health Canada first became involved with First Nations and Inuit health in 1945, when Indian health services were transferred from Indian Affairs. In 1962, Health Canada provided direct health services to First Nations people living on reserves and to Inuit people living in the north. By the mid 1980s, work began to have First Nations and Inuit communities control more health services. Recently in the field of Aboriginal health there has been a move toward community based initiatives. Afterall, community members are the experts on their own health.

The Aboriginal Health Program (AHP) at the British Columbia Women's Hospital aims to empower Aboriginal women to improve their health and also aims to provide appropriate culturally sensitive services. In 2004, the AHP hosted a series of meetings with First Nations communities aimed at identifying health care concerns. In the Gitxsan community, Long QT Syndrome (LQTS), a heart condition, came up as their top health care priority.

Two large families had members from multiple generations diagnosed with LQTS and because of its hereditary nature, these families were concerned for their relatives. LQTS is a rare condition, affecting an estimated 1 in 2000 people worldwide (Schwartz, 2009). It should be noted that clusters of affected individuals have been documented in isolated populations, partly due to the

founder effect (Marjamaa et al, 2009). At the AHP meeting, the several Gitxsan women expressed their wish to address this health concern, and a community based research program was agreed upon.

A clinician researcher with considerable experience conducting health research with Inuit and First Nations communities was alerted about the Gitxsan community's concern for LQTS. A community based participatory research approach was agreed upon, and a local Gitxsan LQTS Research Advisory Board was formed and continues to guide the research. This board is comprised of board members from the Gitxsan Health Society, local health care professionals, and community members, including an Elder.

LQTS is a rare condition, characterized by prolonged ventricular repolarization. It can either be acquired or inherited. Found worldwide, it has an estimated prevalence of 1 in 2,000 (Schwartz, 2009). There is a disproportionately high rate of hereditary LQTS in a First Nations (FN) community in northern British Columbia (Arbour et al, 2008). This is partly due to a novel missense mutation, V205M, in KCNQ1, a known LTQS-susceptibility gene. The effect has been described (Arbour et al, 2008) and predisposes affected individuals to syncope, arrhythmia and sudden death.

Index Cases

Index case #1 presented in her late 30s. She was a fit, physically active mother of four. She had a history of palpitations, and had an aunt and brother who had died suddenly and unexpectedly. While coaching a sports team at the

North American Indigenous Games, she experienced a sudden, unexpected cardiac arrest; she was resuscitated and managed with an implanted cardioverter defibrilator.

Index case #2 also presented in her late 30s. Similar to index case #1, she had a histroy of palpitations and self-reported weakness. Additionally, she also had two sudden unexplained deaths in her siblings. As an infant, her youngest son had a heart murmur and further investigation revealed he had a prolonged QTc interval on electrocardiogram (ECG). Given the hereditary nature of LQTS, Index case #2 subsequently had an ECG which revealed a prolonged QTc interval as well; she was managed with an implanted cardioverter defibrilator.

These two index cases had large extended families with hereditary LQTS, but were not known to be related. Researchers from the University of British Columbia collaborated with a Gitxsan LQTS Research Advisory Board. Together they identified five research goals:

1. Confirm the molecular genetic basis and effect on ion channel to demonstrate pathogenesis
2. Characterize the clinical features, QTc interval, events, triggers (what medications trigger events), and mortality risk
3. Explore what facilitates and hinders resiliency and coping for those living with LQTS
4. Determination of community/population mutation rate in a random sample

5. Develop effective strategies for a culturally appropriate, community based genetic screening, counselling and multi-level education prograom as it applies to this condition in this community.

The community based study began in 2005. Using DNA samples from the two index cases, researchers identified a novel mutation (V205M) that was reponsible for LQTS (Arbour et al, 2008). This discovery accomplished the first goal. The research study completed for this Master's thesis stemmed from the third goal. The remainder of this chapter will provide background information regarding Gitxsan Peoples, LQTS, and genetic counselling (GC).

1.2 Pupose of the Study

Genetic testing became commercially available for LQTS in 2005. K. Staffey, Director of Patient Services and Reimbursement at PGx Health, estimated that 1800 symptomatic patients and their family members are genetically screened each year (personal communication, March 5, 2010). Research has focussed on the biological understanding of the condition, and the psychological and social implications of knowing one's genetic status is poorly understood.

Since the prevalence of LQTS is higher in the Gitxsan communities than expected (Arbour et al, 2008), research concerning the psychological and social impact of this heart condition is vital. The goal of the study was to understand the impact of living with a genetic condition that might confer risk to sudden death on the Gitxsan community. Factors that both facilitate and hinder resiliency and coping were explored. The information gathered will inform genetic counselling

and may also be useful in the development of a culturally based genetic screening program for LQTS in Gitxsan communities.

This research will contribute to the larger field of the psychological and social impact of LQTS. It provides a unique account of the impact of LQTS from a First Nations perspective, and highlights factors that help individuals, families, and the larger community to cope with a potentially fatal genetic heart condition.

1.3 Aboriginal Peoples in Canada

There are three distict groups of Aboriginal peoples living in Canada – First Nations, Inuit, and Metis. More than one million Canadian people identify as Aboriginal – roughly 4% of the population (Statistics Canada, 2006). Fifty-three percent are registered Indians, 30% are Métis, 11% are Non-status Indians and 4% are Inuit. There are 615 First Nations band in Canada, and 198 in British Columbia (Indian and Northern Affairs Canada, 2010).

1.3.1 Aboriginal Health in Canada

As mentioned, the health of Canadian Aboriginal people is much poorer than the health of the Canada's general population (Waldram et al, 1995; MacMillan et al, 1996; Smylie, 2001; Health Council of Canada, 2005). Aboriginal people have higher rates of many chronic diseases (i.e. diabetes, hypertension, cancer), infant mortality, injuries and suicide, and infectious disease (i.e. HIV/AIDS). Additionally, the life expectancies for Aboriginal men and women are 5 and 7 years less, repectively, than the general Canadian poplulation (Indian

and Northern Affairs Canada, 2006). These disparities are concerning, especially for Aboriginal people.

Recent improvements of many health indicators suggest that the health of British Columbia's Aboriginal population is improving, for example life expectancy and overall mortality (British Columbia Provinvial Health Officer, 2009), yet their overall health status continues to be lower that that of the general British Columbian population (British Columbia Provincial Health Officer, 2009 & 2001; MacMillan et al, 1996). Yet, the gap between Aboriginal and non- Aboriginal peoples' health is still evident.

The unique approach of AHP at BC Women's Hospital to meet with community members was an innovative way to uncover first hand what local health concerns are. At the 2005 meeting at Gitanmaax Hall, Gitxsan community members did not identify the prevalent chronic health concerns – they were worried about LQTS.

1.3.2 Gitxsan Peoples

In order to fully understand the information contained in this thesis and to put this study in context, we need to elaborate on the First Nation people this research focuses on, the Gitxsan Peoples. Within the traditional territory of the Gitxsan people, there are five reservations – Gitanmaax, Glen Vowell, Kispiox, Gitsegukla, and Kitwanga. There are approximately 5,400 people registered with these five bands. The two index cases introduced at the beginning of this chapter

both have First Nations ancestry, and more specifically, both have Gitxsan ancestry.

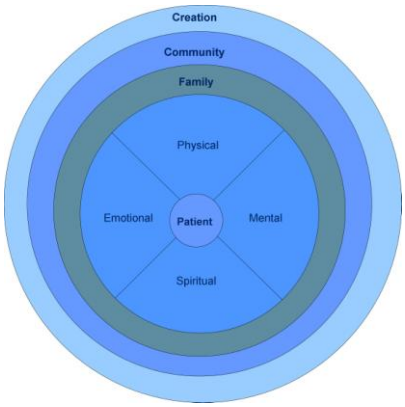
The Gitxsan people of British Columbia are have inhabited their traditional territory in the northwest part of the province for over 10,000 years. An estimated 70% of the registered Gitxsan people live on these five band reservations and two adjacent provincial municipalities – Hazelton and New Hazelton (Gitxsan Chiefs Office). Collectively, the *Hazeltons* of British Columbia are located approximately three hundred kilometers east of Prince Rupert (Figure 1.1).

Figure 1.1: Traditional Gitxsan Territory in British Columbia (http://www.gitxsan.com/html/who/people/land.htm)

The Gitxsan people have a unique worldview. A worldview is a framework of ideas and beliefs through which an individual interprets the world and interacts in it (Naugle, 2002). The Gitxsan Chief's Office says that the Gitxsan worldview affects how the people relate to the land, the way they live, the way they interact with other Gitxsan and their relationships with *Lixs Giigyet* (Gitxsan for "those who come from away"). The Gitxsan worldview also shapes how they view health and well-being. The Gitxsan people have a holistic view of health, which

encompasses four aspects of being: mental, spiritual, emotional and physical (Gitxsan Chiefs Office, 2010). Figure 1.2 illustrates the holistic view of health.

Figure 1.2: Holistic View of Health



The Medicine Wheel (MW) is a visual illustration of this holistic view of health. Aboriginal Peoples are diverse, and concepts of the MW may vary or perhaps not even exist, depending on the community that is being discussed. For the sake of clarity, the concepts of the MW are presented and the general framework that it may provide is perhaps applicable to genetic counselling with First Nations people. It has been used as a framework in several health-related fields, such psychology, social work and nursing (France et al, 2004; Dapice, 2006; MacDonald, 2008; McCormick, 2009).

An excerpt from the Native Brotherhood of BC on Health Issues quotes Rhea Joseph saying:

> *"Among the Gitksan and Wet'suwet'en, there is no mother tongue word for health. However, they do have a word for strength, which is interchangeable[with] health. They also speak of well-being.This well-being is associated with high self-esteem, a feeling of being at peace and being happy...This includes education. It includes employment. It includes*

land claims. It includes resource management. All of these lead back to wellness and well-being" (as cited in People to People, Nation to Nation, p. 96).

1.3.3 Resilience in Aboriginal Communities

Index case #1 and #2 and their respective families appear to be resilient and are coping with LQTS fairly well. Resilience is a term used to describe "the ability to recover from or adjust easily to misfortune or change" (Merriam-Webster Dictionary). To have experienced a sudden cardiac arrest (Index case #1) and to return to a self-described 'normal routine', surely epitomizes the idea of being resilient.

There is a growing body of research in the area of resiliency among Aboriginal people and communities. Much of the published research describes resliency in terms of the residential school experience, addictions, and suicide (Williams, 2010; Chandler & Lalonde, 1998).

The work of Chandler and Lalonde (1998) stresses the importance of culture in resilience. Their study of suicide in Aboriginal communities revealed a set of eight 'cultural continuity' factors that appear to protect communities from this trajedy. These factors range from community self-government, to local control of health, to availability of cultural facilities (Chandler & Lalonde, 1998 & 2008).

Williams and Lindsey (2010) reviewed how the impact of spirituality on runaway and/or homeless youth may inform the social work practice. Through

secondary analysis of interviews with runaway and homeless youth, Williams and Lindsey revealed that interventions that incorporated a focus on spirituality might be well-received by youth who have lived through considerable adversity in their lives.

A main theme that appears within the literature regarding resiliency in Aboriginal communities is the important role of culture and traditions. These studies suggest that the more an individual is involved in cultural practices and traditions, the healthier a person they are and the more resilient they become to life's adversities.

1.3.4 Genetic Research in Aboriginal Communities

Aboriginal communities have many concerns about the genetic studies that have been conducted in Aboriginal populations in the last few decades. Some concerns include lack of community involvement in the planning of the studies, insensitivity to cultural beliefs and traditions, ownership of biological samples, potential of results furthering stigmatization, and lack of feedback to the community after completion of the research projects (Dodson, 1998; Olson, 2002; Arbour & Cook, 2006).

An example of poor decision making in research happened here in Canada in 1991. Blood samples that were originally taken for health research on arthritis from the Nuu-chah-nulth First Nation group in British Columbia were also used, without consent, to isolate mitochondrial DNA and used to establish ancestry (Ward et al, 1991; Dalton, 2002). This case and others have motivated

the development of guidelines for health research in Canadian Aboriginal communities. The Canadian Institute for Health Research (CIHR) recognized the need for researchers and institutions to carry out ethical and culturally competent research with Aboriginal communities and in May 2007 released *CIHR Guidelines for Health Research Involving Aboriginal Communities.* Article 13 of these guidelines states “Biological samples should be considered ‘on loan’ to the researcher unless otherwise specified in the research agreement”. This article respects the notion that several Aboriginal people view every part and product of the body as being sacred.

The best standard of research in Aboriginal communities is done collaboratively, using the principles of community-based (CB) participatory research. The three key elements of such research are collaboration, education, and action (Macaulay et al, 1999). This approach is based on communication and understanding among all research partners, and includes development of protocols outlining ethical, legal and practical aspects of the research (Hills & Mullett, 2000). The goal of participatory research is for research participants to “own” the research process and to use its results to improve their quality of life (Macaulay et al, 1999). A collaborative approach was used for this research study with the Gitxsan community.

1.4 Long QT Syndrome

1.4.1 Introduction

Given the complex nature of LQTS, a physican must take in to account several factors when considering this diagnosis – electrocardiogram (ECG), family history, and genetic test results. Typically, LQTS presents in adolescence and young adulthood (Hobbs et al, 2006).

LQTS is named for the prolonged QT interval observed on ECG. An ECG records the movement of electrical activity in the heart. The QT interval represents the time that ventricular myocytes contract (QRS complex on ECG) to the time the ventricular myocytes relax (T wave on ECG) representing the period of repolarization. The QT interval can range from 200 ms to 400 ms but the measurement will vary depending on the heart rate. Therefore, a corrected QT interval is commonly reported. Normally, QTc is less than 440 ms. LQTS commonly diagnosed with QTc greater than 470ms or 480 ms, in men and women respectively.

Family history also plays an important role in heridatary LQTS. It may go undiagnosed in a family for generations, in part because of the incomplete penetrance and variable expression of LQTS. A physician or genetic counsellor would look for other family members who have been diagnosed with LQTS, as well as any cases of sudden death within the immediate family.

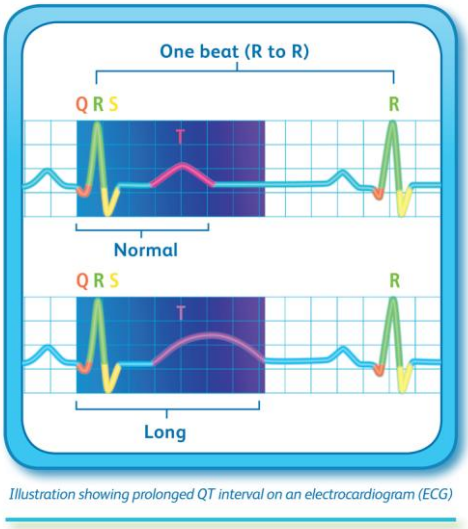
When cardiac cells are at rest, they are electrically polarized, and no electrical activity is taking place. When a cell is stimulated, ions such as potassium and sodium are exchanged across the cell membrane through ion

channels. As an electrical stimulus is passed on from cell to cell, each cell goes through depolarization followed by mechanical contraction (systole) and electrical repolarization followed by mechanical relaxation (diastole).

LQTS-affected individuals have abnormal sodium or potassium ion channels in their heart (Figure 1.4). This predisposes patients to certain cardiac arrhythmias.

LQTS is characterized by prolonged QT intervals on electrocardiogram (Figure 1.3). Symptoms include syncope, palpitation, ventricular arrhythmias, seizure, and possibly sudden death (Chiang & Roden, 2000). LQTS can be acquired or inherited. There are two inherited forms of LQTS: autosomal dominant, which is called *Romano-Ward syndrome*, and autosomal recessive, which is called *Jervel and Lange-Nielsen syndrome*. The latter form is associated with deafness.

Figure 1.3: ECG illustrating the QT interval (used with permission from Gene Dx ©)



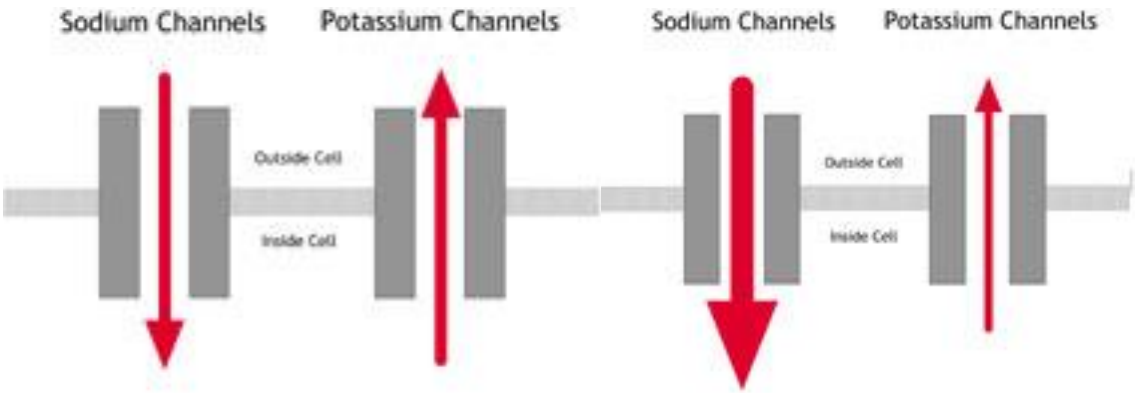
1.4.2 Discovery of Long QT Syndrome

The first account of any type of LQTS was by Jervell and Lange-Neilson syndrome in 1856 (Tranebjaerg, Bathen, Tyson, & Bitner-Glindzicz, 1999). The most common form, LQTS type 1 (described in more detail later) was first described by Romano et al. in 1963 and independently by Ward in 1964. Ward observed a brother and sister who presented with syncope due to ventricular fibrillation and who's resting electrocardiogram (ECG) showed abnormal prolongation of the QT interval. The mother was asymptomatic, but also showed a prolonged QT interval on ECG. The mother's sister had attacks of syncope and died following an episode at the age of thirty.

In the Gitxsan community, the AHP meeting was the first time both Index case families brought their concern for LQTS to the attention of the community. Index case #1 was diagnosed in 2002, and Index case #2 was diagnosed in 1999 – only three years apart.

Figure 1.4: Sodium and Potassium Flow in normal health and LQTS heart
(A) Normal heart – potassium flows out of the cell to 'repolarize' the heart, and sodium flows into the cells to activate the heart;
(B) LQTS heart - the flow of potassium is usually reduced. In some people with LQTS, the flow of sodium may be increased

(A) (B)



1.4.3 Types of Long QT Syndrome

As more genetic research is carried out, scientists are discovering variations within genes that are not associated with diseases and do not cause them. These different variants are known as polymorphisms; they cause difficulty when attempts are made to isolate pathogenic mutations. To make the interpretation of genetic tests done to diagnose LQTS easier, Splawski et al. (2000) look at the spectrum of mutations commonly found in the genes associated with LQTS (27). More recently, Kapa et al (2009) reported on distiguishing pathogenic mutations from rare benign variants.

LQTS type 1 is the most common type. The V205 mutation is in the KCNQ1 gene, which is responsible for the formation of the alpha subunit of the potassium channel.

To date, about 12 loci are associated with the different forms of LQTS, and mutations at these sites result in impaired ion channel functionality. The mutation that was found in Index case 1 of the Gitxsan community was located within the KCNQ1 gene. Specifically, a missense mutation in exon 4 changed an amino acid in the C-terminal of the voltage-gated potassium channel. Table 1.1 shows all types of LQTS and the mutations associated with impaired ion channel functionality. Type 1, 2, and 3 account for nearly 75% of all Long QT Syndrome (Tester & Ackerman, 2008).

Table 1.1: LQTS types and their associated genes

Type	Gene
LQT1	KCNQ1
LQT2	KCNH2
LQT3	SCN5A
LQT4	ANK2
LQT5	KCNE1
LQT6	KCNE2
LQT7	KCNJ2
LQT8	CACNA1C
LQT9	CAV3
LQT10	SCN4B
LQT11	ANAP9
LQT12	SNTA1

1.4.4 Inheritance of Long QT Syndrome Type 1

Long QT syndrome type 1 is the most common. It is autosomal dominant. Each individual who inherits a mutation in the KCNQ1 gene has a predisposition to LQTS. Affected individuals have a 50% chance of passing it along to their offspring. The penetrance of these mutations is still poorly understood. A significant proportion of mutation positive individuals, about 30%, will never experience any symptoms. However, a small proportion of mutation positive individuals, about 10%, may present with sudden cardiac arrest as a first event.

1.4.5 Clinical Features of Long QT Syndrome

Currently, clinical diagnosis of LQTS is made upon ECG evidence of a prolonged QT interval, greater than 480 ms in women and greater than 470 ms in men. Symptoms most often appear in adolesence and young adulthood (age 18-25). Symptoms include syncope, arrhythmia, and possibly sudden death.

Literature suggests that ECG may not be the best diagnostic tool, as reports have shown that individual's may not present a prolonged QT interval every time an ECG is performed. Additionally, at least one study reported that LQTS has been misdiagnosed as epilepsy (MacCormick et al, 2009).

1.4.6 Epidemiology of Long QT Syndrome

Long QT Syndrome is a rare condition. Current estimates in the literature range from 1 in 5000 (Goldenberg & Moss, 2008) to 1 in 20,000 (Moss & Robinson, 2002). In a prospective study of 44,596 neonates born in 18 maternity hospitals in Italy, Schwartz et al (2009) reported the first data-based prevalence estimate to be 1 in 2534. This is much higher than the previously reported estimates.

It should be noted that clusters of affected individuals are seen in ethnically homogenous populations (Piippo et al, 2001; Brink et al, 2005); this is thought to occur because of the familial nature of the condition and the founder effect, which results when a population has only a few original settlers (founders), and one or more had the gene (Chiang & Roden, 2000). This seems to be the case in the Gitxsan community, where LQTS appears to disproportionately affect individuals in comparison to the general population (Arbour et al, 2008).

1.5 Genetic Counselling

1.5.1 Healthcare in Northern British Columbia

Access to health care is a major concern around the world, particularly for people living in rural and northern areas (Strasser, 2003). This is especially true for the delivery of specialty services, such as genetic services, as these are rarely considered a health care priority (Mahowald, Verp, & Anderson, 1998).

A recent study of the access and utilization of health care services by British Columbia's Aboriginal people found that there was a lack of access to specialized medical care, especially for those who live in rural and remote areas (Wardman, Clement, & Quantz, 2005).

British Columbia is divided into five health authorities – Northern Health, Interior Health, Vancouver Island Health, Vancouver Coastal Health, and Fraser Health. The Northern Health Authority (NHA) is British Columbia's largest geographically defined health region. The NHA provides service to approximately 300,000 people, who live from Atlin to Queen Charlotte Islands and Fort Nelson to Valemount. As this region covers approximately two-thirds of British Coumbia's land mass, providing equal access to health care is a big challenge.

An even bigger challenge is to provide appropriate specialty services for health conditions that primary health care providers are unfamiliar with. For exmaple, the province's only genetics clinics are located in Vancouver and Victoria. Previously, outreach genetics outreach clinics had been occasionally held in Terrace. At the time of this study, outreach clinics were only offered in

Prince George. Gitxsan patients who are being investigated for LQTS travel 500 km to 1000 km for genetic care services.

1.5.2 Introduction to the Profession

Genetic counsellors are health care professionals with expertise in medical genetics and counselling. Genetic counselling (GC) is a relatively new profession; the first GC program was offered in 1969 at Sarah Lawrence College in New York. The first GC program in Canada was offered at McGill University in 1985.

Genetic counselling was built upon the Western values and beliefs, and was modeled after social work. The initial goal of GC was to provide medical information and genetic facts. Subsequently, it was acknowledged that emotional responses were unavoidable, and Carl Roger's *Client-Centered Counseling* techniques were integrated into the curriculum (Marks 1993, 2003).

The two main theoretical approaches to GC are the teaching model and the counselling model. More recently, a Reciprocal-Engagement model has been developed (McCarthy Veach, Bartels, & LeRoy, 2007).

1.5.3 Theoretical Approaches to Genetic Counselling

As previously mentioned, GC is a relatively new profession. As Joan Marks accepted the Award for Excellence in Medical Genetics Education from the American Society of Human Genetics in 2003, she stated:

> “The hallmark of the training of genetic counselors is based on the premise that understanding that the emotional component of genetic risk is central to providing good genetic services. That integration of medical-genetics training with psychological-counseling skills has remained at the core of all graduate programs training genetic counselors now offered in the United States and abroad”

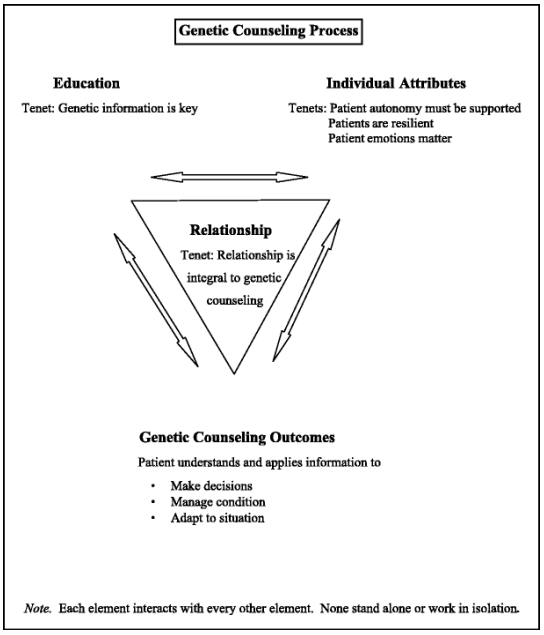
Currently, the vast majority of genetic counselling programs blend the two approaches of teaching and counselling together in the curriculum. Here they are presented separately, however, in practice these approaches are used in harmony. The teaching approach to genetic counselling fouses on the ability of a genetic counsellor to explain the biological concepts of genetic conditions to patients. This is the original approach used train genetic counsellors. The relationship is not equal; the genetic counsellor is viewed as the expert and therefore holds more power.

The counselling model of genetic counselling focuses on the ability of a genetic counsellor to support the psychological well being of a patient who is learning about genetic conditions. The relationship within this model is more equal. The genetic counsellor and the patient both hold their own expertise. The counsellor has expertise in genetic information, and the patient has expertise in his or her own life. Therefore they work together in a GC session and the relationship is reciprocal.

Four decades have passed since the first students were accepted into the first genetic counselling graduate program, yet there is no empirically

established, comprehensive model of genetic counselling practice (McCarthy Veach, Bartels, & LeRoy, 2007). McCarthy Veach et al (2007) describe a *Reciprocal-Engagement Model* of genetic counselling based on the information gathered from the directors of GC graduate programs. This model aims to encompass the current tenets and goals of genetic counselling – genetic information is key, patient autonomy must be supported, patients are resilient, patient emotions matter, and relationship is integral to genetic counselling. The model is visually represented in a triangle (Figure 1.5).

Figure 1.5: Reciprocal-Engagement Model of genetic counselling (McCarthy Veach et al, 2007)



1.5.4 Role of Culture and Spirituality in Genetic Counselling

Genetic counselling is a reciprocal process; patients need to trust genetic counsellors and other health care professionals and feel safe while sharing

personal and familial information. There is a growing body of research exploring multicultural genetic counselling.

The Western values and beliefs that genetic counselling programs were built upon may not take into consideration the values and beliefs that many First Nations people hold. Current GC programs prepare genetic counsellors to support patients and facilitate the adjustment to genetic conditions, such as LQTS.

1.5.5 Genetic Testing for Long QT Syndrome

1991 marked the first discovery of LQTS-suseptibility locus on chromosome 11 (Keating et al, 1991). 1995 marked the discovery of mutations in key cardiac potassium and sodium channels (Wang et al, 1995; Curran et al, 1995). Between 1995 and 2004, a small number of research laboratories were offering genetic testing for LQTS-suseptibility genes. In 2005, commercial genetic testing was available for 5 LQTS-suseptibility genes (KCNQ1, KCNH2, SCN5A, KCNE1, & KCNE2). In 2009, commercial screening for an additional 5 LQTS-susceptibility genes became available (ANK2, CACNA1C, KCNJ2, CAV3, & SCN4B).

In British Columbia at the time of this study, genetic testing for LQTS was offered on a service basis. At the beginning of this study, testing was done through Familion ® and Boston University in the United States. If a physician suspects LQTS in a patient, the physician would refer the patient to a genetic counsellor if the patient's family history is suggestive of herditary LQTS. The

patient would meet with a genetic counsellor, who would review the family history, and provide the patient with information about what his or her history suggests about the their risk for LQTS. A genetic counsellor would take into account the patient's family history and clinical presentation to determine if the patient is a candidate for genetic testing. At the time of this study, there were no specific criteria for genetic testing for LQTS, and testing was considered on a case by case basis.

1.6 Gaps in Existing Research

To date, there is only one academic paper regarding genetic counselling an American Indian tribe. There is no literature regarding GC in Canada's Aboriginal population. Today there is more diversity in Canadian society, as multiculturalism continues to increase. Each culture has a unique worldview, and it would not be uncommon for a genetic counsellor to hold a different worldview than their patient. Culture is so embedded in everything that we do, and can be described as "a group of people's total way of lie: the way they act and think, organize themselves, relate and communicate, make or build things, express feelings and emotions, and respond to the world" (Kroeber & Kluckhohn, 1952). The majority of research on LQTS has focussed on the biological and clinical aspects.

1.7 Rationale for the Study and Research Objectives

Numerous studies have looked at different aspects of Long QT Syndrome, with the majority focusing on the ion channel genes and the mutations that have been found to cause LQTS. Yet, to date, only five studies have looked at the psychological aspects of this potentially lethal syndrome (Hendriks et al, 2005; Farnsworth et al, 2006; Smets et al, 2007; Giuffre et al, 2008; Muelenkamp et al, 2008; Anderson et al, 2008). All six of these studies were conducted in Caucasian populations, and with the exception of one (Anderson et al, 2008) all have focussed on the psychological impact on parents who have children diagnosed with LQTS.

My research project explored the personal impact of living with LQTS, with the understanding that results will inform genetic counselling. This study will provide the cultural insights into the issues that a remote First Nations community faces with regard to LQTS. This information will ultimately contribute to the continued improvement of health care available to First Nations people living with LQTS in Northern British Columbia. It is important for all health care providers to understand how the LQTS and the genetic test affects the individuals who use it.

As stated earlier, the overall goal of this study was to set out by the Gitxsan LQTS Research Advisory Board. They wanted to “explore what facilitates and hinders resiliency and coping for those living with LQTS”. This goal was broken down into these three objectives for this study:

1. From a cultural context explore what facilitates and hinders resiliency and coping for individuals and families.

2. To explore issues of barriers to effective physical and psychological management.
3. To understand the multigenerational impact of LQTS.

The next three chapters of this thesis are presented as independent manuscripts, each with intention of publication. This manuscript-based format means that there will be some redundancies. The last chapter will combine tie all aspects of the study together with an overview of the salient findings.

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Chapter 2 Understanding the Personal and Community Impact of Long QT Syndrome: Perspectives from Gitxsan Women [1]

2.1 Introduction

There is a considerable gap between the health status of Canada's Aboriginal Peoples (First Nations, Inuit, and Métis) and the general Canadian population (Waldram, Herring, & Young, 1995). Cardiovascular disease has been one of the leading causes of death in Canada over the past 25 years (Statistics Canada, 2010). In the Canadian Aboriginal population, the incidence of cardiovascular disease is estimated to be 1.5 times higher than the general population (First Nations & Inuit Health Branch, 2010). Despite recent improvements on some measures of health, for example life expectancy and infant mortality (FNIHB), the gap still exists between Aboriginal and non-Aboriginal Canadian populations. In 2004, the Aboriginal Health Program (AHP), at the British Columbia Women's Hospital, hosted a series of meetings in First Nations communities aimed at identifying local health care concerns. Health care professionals were expecting to hear about concerns for chronic diseases, such as diabetes, cancer, and tuberculosis. Yet, the meeting at Gitanmaax Hall with the Gitxsan community identified Long QT Syndrome (LQTS), as their top health care priority. LQTS, which is a rare cardiac condition, came up as their top health care priority.

Two families mentioned having been recently diagnosed with this potentially fatal condition. LQTS is a rare condition, with an estimated prevalence

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of 1 in 2000 (Schwartz, 2009) to 1 in 2500-10,000 people (Lu & Kass, 2010). So, in a small community of approximately 5000, researchers were alarmed to discover more than 40 individuals affected with LQTS. The families were deeply concerned that other relatives could also have LQTS and just not know it yet.

LQTS is a treatable heart condition that affects the rhythm of the heart, as a result of delayed repolarization of cardiac ventricular action potentials. It is characterized by a prolonged QT interval observed on an electrocardiogram (ECG). It can be either acquired or inherited. Gitxsan community members elected to address their concern for LQTS through a university-community collaboration. A disproportionately high rate of hereditary LQTS was revealed (Arbour et al, 2008).

People with LQTS typically experience their first symptom in adolescence and young adulthood (Hobbs et al, 2006), with events including syncope, *torsades de pointes*, seizures, and possibly sudden cardiac death (Goldenberg & Moss, 2008). Treatment options range from beta-blocker therapy to implantable cardioverter defibrillators to left cardiac sympathetic denervation (Goldenberg & Moss, 2008).

Genetic testing revealed that LQTS in the Gitxsan community was caused largely by a mutation (V205M) in the KCNQ1 gene (Arbour et al, 2008). To date, more than 500 mutations in 12 genes are associated with the different forms of LQTS, and mutations at these sites result in impaired ion channel functionality. Types 1, 2, and 3 account for 70%-75% of all inherited LQTS (Tester & Ackerman, 2008). Mutations in the KCNQ1 gene (a KQT-like voltage-gated

potassium channel-1 gene) cause LQTS type 1 (OMIM # 192500). The variable expression and incomplete penetrance of these mutations in LQTS makes it difficult to determine the prognosis of a given mutation carrier (Crotti et al, 2009; Lu & Kass, 2010). An estimated 50% of mutation carriers will ever develop symptoms (Zhang et al, 2004).

Currently, people with LQTS are diagnosed based on abnormal ECG findings. If a physician were suspicious that his or her patient has hereditary LQTS, that patient would be referred to a genetic counsellor. A genetic counsellor would provide individuals and families with information on the nature, inheritance, and implications of genetic disorders (such as LQTS) to help them make informed medical and personal decisions (McCarthy Veach, Bartels, & LeRoy, 2005).

Genetic counsellors involved with the community-based study provided participants with individual results, either in person or over the telephone. The Gitxsan LQTS Research Advisory Board (whose composition is outlined in chapter 1) anticipated that current genetic counselling practice might not be culturally specific for Gitxsan people.

There is a scarce amount of research about the psychological and social impact of LQTS. All but one of the published studies have focused on the impact on parent's and children's concerns (Hendriks et al, 2005; Farnsworth et al, 2006; Smets et al, 2007; Giuffre et al, 2008; Muelenkamp et al, 2008). To date, the only study to investigate adult's ability to cope with LQTS was conducted in Norway

(Anderson, Oyen, Bjorvatn, & Gjengedal, 2008). None have offered insights from any Canadian Aboriginal perspective.

Susceptibility screening for LQTS became available in 2005 through PGx Health ®. Like any genetic screening, it anticipates early detection and prevention recommendations for individuals and families at high risk. It will be important for genetic counsellors and other health care professionals who offer genetic testing to be aware of the psychological and social impact of LQTS. This study attempts to provide insight and add to a more comprehensive understanding of the psychological and social impact of LQTS, as well as how genetic testing is perceived. The purpose of this study was to explore what facilitates and hinders resiliency and coping for those living with LQTS.

2.2 Methods

2.2.1 Research Design

Three qualitative research methods were offered to all participants. The first option was to be interviewed individually. Second, we invited participants to the option of Photovoice and the third option was a Talking Circle. Qualitative research methods were selected because they allow participants to share stories and experiences about the impact of LQTS.

Offering three methods allowed participants to guide the direction of the information that was shared. This paper focuses solely on results from the individual interviews, specifically looking at the factors that facilitate and hinder

coping and resiliency. Results from the Talking Circle and Photovoice are discussed in chapters 4 and 5, respectively.

Participants were recruited through the larger core LQTS community based study, which looked at mutation status for LQTS (*KCNQ1* V205M & R591H). Posters were placed throughout the community (health stations, hospital, community boards), as well the research assistants for the core study invited participants individually by phone or in person. The present study was approved by the University of British Columbia's Behavioural Research Ethics Board and supported by the Gitxsan Health Society.

2.2.2 Interview Protocol and Procedures

Participants were given the option of where they would like the interview to take place. This allowed a safe and respectful space for each interview. An explanation of the study and what is involved was given at the outset of the interview. After providing informed consent, I interviewed each participant in person. As the interviewer I was a graduate student in the Genetics Graduate Program, at the University of British Columbia, whose course-work included a foundation of human genetics, counselling psychology, qualitative research methodology, and Aboriginal health. The aim of the interviews was to facilitate participants to share their personal experience of LQTS, while encouraging them to guide the process discussing whichever topics they viewed to be most relevant.

The interview script was developed in consultation with a local research advisory committee, and was based on the available studies that have previously explored the psychological impact of LQTS (Appendix A). The interview guide consisted of eleven open-ended questions. Questions explored a range of experiences from their first encounter with LQTS to what they understand of it today, to specific factors that both help and hinder their ability to cope with this information and diagnosis.

Following the interview, participants were given the option of obtaining further follow-up with the genetic counsellors involved in the study. Participants were also given a list of local counselling resources (public school system, Gitxsan Health Society, hospital) they could access if they needed additional support.

2.2.3 Data Analysis

Systematic Text Condensation was selected as the analysis method. This qualitative analysis method was first described by Giorgi in 1985, and modified by Malterud in 2001. It is a four-part analysis. First, interview transcripts were read for holistic understanding of the text. Second, transcripts were re-read to identify meaning units. Third, the meaning units were explored to abstract insights. And fourth, all the transformed meaning units were synthesized into a consistent statement regarding the participants' experience.

2.3 Results

Ten open-ended interviews were performed for this study. The analysis discussed here involved ten participants who all were interviewed individually. Interviews took place in the participants' home (7) or the local health station (3), depending on the participants' preference.

2.3.1 Study Participants

All participants were either affected by LQTS personally or had family members who were affected. These ten participants came from a total of four extended families. All participants were female, and all of them were mothers. Ages ranged from early 20s to late 60s. Five interview participants had been diagnosed with LQTS and had tested positive for the V205M mutation. The other five participants were being tested for the V205M and R591H mutations, but had unknown mutation status at the time of the interview. There was variation in the amount of time that had passed since participants were first introduced to LQTS, ranging from one year to fourteen years. Three participants had an implantable cardioverter defibrillator. Eight participants had been prescribed beta blocker therapy. All ten women had received genetic counselling in connection with the larger core community-based LQTS study.

We aimed to recruit individuals with widely varying experience with LQTS, as we anticipated that this would result in a thorough representation of the impact of LQTS in this community. Having a large wealth of information and experiences is a goal of qualitative research. Despite their differences in terms of how long

they have lived with the knowledge of LQTS, and the symptoms they have experienced (personally or through family members), there was obvious similarity in LQTS-related experience when we reviewed the interviews and transcripts. We recognize that this is a relatively small sample size of ten. We considered data saturation when the stories and experiences shared became redundant.

Table 2.1: Sociodemographical and Medical Characteristics of the Participants

	Participants (n = 10)
Age (years)	
20-30	2
31-50	7
50+	1
Sex	
Female	10
Male	0
V205M test result	
Positive	5
Negative	0
Unknown at time of interview	5
Treatment of participants	
No treatment	0
Medication	7
ICD	3
Years since learning of LQTS	
< 1 year	1
1 - 5 years ago	2
6 - 10 years ago	6
> 11 years	1
Sudden cardiac death or sudden unexpected death in siblings	
Yes	4
No	0
Unknown	6

2.3.2 Prominent Themes

The Systematic Text Condensation analysis identified several themes and their components reflecting the psychological and social impact that LQTS has on individuals and families. This section of the chapter characterizes the details of each theme as they relate to coping.

The ten women interviewed in this study are referred to as “participants”. It was perceived that learning about a LQTS diagnosis as traumatic, even life altering. When asked to recall the first time they learned about LQTS, participants used words such as *emotional, restricted, unsure*, and *concern*. Each participant was able to vividly recount the first time they learned about LQTS. Participants recounted experiences of gradual acceptance that led to continuing coping. Each theme is presented, and where possible, direct quotes are used to illustrate the components of each theme. Quotations are also used to give the participants a voice, realizing and respecting that they are sharing a personal journey of LQTS.

2.3.3 Theme 1 – Positive Family Relationships Facilitates Coping

All ten participants shared stories about the support they receive from their families. Participants discussed the conversations they had with family members, reporting that support came from both formal and informal gatherings. For example, one participant reported having regular Talking Circles, bringing the entire family together to share how they have been feeling lately, providing an opportunity to validate important events that had happened in their lives. Another participant reported having several low-key family celebrations for birthdays,

anniversaries, and small accomplishments (such as kindergarten graduation or winning a sports tournament).

One participant, who had family members clinically diagnosed with LQTS, but unknown genetic status herself, recounted the support she received from her family:

> *"I believe in the strength in families. I believe in the strength of communities. And in nations when they have joined together…Right after when we have our gathering and we share a meal, we say our prayers, we sing our songs and we also have gratitude for all the blessings that we've had…and we feel like there's a sense of relief – um no a bit of peace. And also have hope."* (Participant D)

Another participant, whose genetic test result confirmed her clinical diagnosis of LQTS, described the role her family plays and the inspiration they give to help keep everything in perspective:

> *"I'd say family is a big support to me…I think it's my kids too, you know that keep me going. Because they're always busy with hockey, soccer, basketball, you know, ahh…I would say they're the light in my life that keeps me going "* (Participant F)

Overall, families appear to provide unconditional support or as much support as they are able to individuals who are impacted by LQTS. Families help individuals to overcome the initial shock of a LQTS diagnosis, grounding them in traditions and values, helping to keep everything in perspective. Knowing that

their family members would provide support as needed left the participants feeling loved, cared for, and included in a group.

2.3.4 Theme 2 – Spiritual Faith Facilitates Coping

Most participants (8/10) spoke about spirituality and faith that enabled them to cope with the impact of LQTS. Participants spoke about relying on the faith that their parents had instilled in them from childhood. Responses ranged from personal reliance on meditation and prayer, to participating in organized religion services and traditional Gitxsan ceremonies.

One participant, who had a sister that died at a young age, and she herself has an implanted defibrillator, explained her immediate response to a doctor's original clinical diagnosis eleven years ago:

> *"in the hospital, I made a choice to… I actually just talked to God and I thanked God for allowing him to allow me to find – well for the doctors to find the Long QT, so I could live longer. And I could be around to raise my children and watch my grandchildren grow up. So I was very, very thankful. I was very appreciative"* (Participant A)

Another participant, who did not have a clinical diagnosis of LQTS, said this about the role of spirituality in initial reaction to discovering LQTS "ran in her family":

> *"I found that when my sister was diagnosed, there was a lot of impact at an emotional level, as well as at a spiritual level. I found that we really had*

to have a lot of faith and lots of prayers so that way she could be well"

(Participant D)

Having an awareness of their spiritual self helped some participants cope with the initial shock of LQTS, and continues to help other participants day to day.

2.3.5 Theme 3 – Knowledge about LQTS Facilitate Coping

Many participants shared stories about how the initial genetic counselling session had facilitated their ability to cope with LQTS. Learning the basics of what LQTS is, the signs and symptoms to look out for, the treatment available, and what triggers (pharmaceuticals, intense physical activity, etc) to avoid, all helped the participants feel equipped to face daily life.

Participants found comfort in the knowledge that LQTS can be treated effectively with beta-blocker therapy or an implantable cardioverter defibrillator. One participant, who has several family members with LQTS, said:

"So I feel a bit more um relief and a bit more joy knowing that there is somewhat of a cure if you could say those little pills are regulating your heartbeat so that way you can continue to breathe and to enjoy the day"

(Participant D)

Another participant, whose sister was clinically diagnosed with LQTS, shared a similar opinion:

"I was so worried when she said that she had [LQTS], and I thought it was incurable…and after she told me that they gave her some medications –

*the whole – my life – everything lifted up…and I felt so light and I was so glad because I didn't think they would have medications for this. It gave me hope that my brothers and sister would have a longer life" (*Participant H*)*

One participant, whose genetic testing revealed a positive mutation status, explained how receiving the initial genetic counselling session helped her cope:

*"'[It] was when [the genetic counsellors] came and explained all of this [LQTS]. That was very helpful" (*Participant G*)*

Overall, it appeared that learning about the biological concepts of what LQTS is helped facilitate coping for the participants.

2.3.6 Theme 4 – An Incomplete Understanding of the Biological or Clinical Aspects of LQTS Hinders Coping

Almost half of the participants (4/10) shared stories regarding the confusion they felt surrounding exactly what having LQTS means, and what they should expect. Though many participants spoke about how the initial genetic counselling session had helped in their ability to cope with LQTS, participants felt ill equipped to explain to others what the condition is. One participant, who was diagnosed with a predisposition to LQTS, shared:

"It's still not clear because you know I don't spend a lot of time researching it [LQTS]. But the explanation I got at that time [from the genetic counsellors] was very helpful…it helped clear a lot of things up"

(Participant G)

2.3.7 Theme 5 – Uncertainty Regarding Appropriate Levels of Physical Activity Hinders Coping

Many participants (7/10) shared concerns for the medical advice they had received, especially surrounding the restrictions in physical activity. Intense physical activity and competitive sports are often discouraged, as it significantly increases the risk of prolonging the QT interval, possibly leading to torsades de pointes. All ten participants reported the changes in their exercise routine that they have had to make. Yet, none of the participants had a clear picture of what level of activity was appropriate.

One participant, who had two affected children, recalled the physical activity restrictions placed during her children's elementary school years:

> *"my son and daughter couldn't take PE in school. They were ousted out of that"* (Participant A)

This participant went on the explain the difficulty she had restricting her son's physical activity outside of school as well:

> *"he was taken out of sports and all physical activity when he was a very active boy. So, he got very, very angry. And I felt very, very guilty. But at the same time I'm like no, you're not going to join the hockey team"* (Participant A)

2.3.8 Theme 6 – LQTS not being taken seriously by both social contacts and health care providers hinders coping

Some participants (4/10) shared negative experiences with health care providers (local physicians, hospital staff, cardiologists, etc). Several participants talked about the frequent turn over of health care professionals at the local hospital, and having to explain to a new doctor what LQTS is, reminding them of the list of medications they need to avoid.

One participant, who was diagnosed with LQTS nearly ten years ago, shared a story of a physician whose professionalism she questioned after she had this encounter at the local hospital:

> *"it sort of got to the point where ah there are so many people thinking that they have Long QT…I felt they [the local doctors] were turning it into a joke. Um, when I would go into the emergency, you know, with bronchitis or whatever, and they'd say, do you have anything else? I'd say I've got the Long QT. Oh, another Long QTer and they'd laugh. And I felt very, very bad…you know so it's like if I had cancer, would they say, Oh, another cancer patient. You know, I felt um like they were making a joke of it by that time"* (Participant A)

This participant showed tremendous coping abilities and resiliency. She voiced concern that her children and relatives may have a similar and completely unnecessary experience if they ever required emergency care.

Another participant, who had survived cardiac arrest, shared an unfortunate conversation she had with a physician:

"Oh, you're the one who um caused this epidemic for Long QT at our hospital. I said, if that's what you're calling it and if that's what it takes for people to stand up and notice and actually do something about it, then yeah it is. I said I am that person" (Participant C)

Yet another participant, who was clinically diagnosed with LQTS as a young adolescent, commented on the frustration she felt when her sports coach failed to recognize the serious risk that competitive sport placed on her heart:

"with my high school coaches. Like basketball. Because they just try pushing and pushing and pushing and push you, hey and … them not knowing the effects that it could have" (Participant B)

Cardiology is a specialty medical service that is not provided locally, so patients travel over 1000 km to Vancouver once or twice a year for care. Some participants disclosed frustrations with the cardiology appointments. One participant, who had a borderline QTc and more than one sibling with an ICD, said this about her experience with a cardiologist in Vancouver:

"I wasn't too - too impressed with - with [the cardiologist's] time and understood that it's a long ways, but we [his patients] travel too and then when you go there you wait for over a hour – two hours and then when you finally see him it seemed he wanted you to get out of the office. And so (laughing) he's yawning and being really bored. And then and not really taking it as if it was that important. So, after a while I go there to for a couple of years and then I just thought – I just run out of my pills and then I quit going there. And then they just forget about it and then I just didn't

really take too much interest in it. Because maybe I do have it, but maybe I don't" (Participant D)

Some people also talked about the frustration they felt when social contacts did not understand what it is like to have LQTS. Because LQTS is not a visible illness, especially when patients are compliant with the treatment, other people will not know an individual is 'sick'.

One participant, who had several family members affected with LQTS, voiced her frustration that the serious nature of LQTS was not recognized by people she knew (such as teachers and peers):

"Like people don't understand how it can affect you and so – like if they don't have it like they don't really see a big deal with somebody else who has it [LQTS]" (Participant B)

Another participant, whose genetic status was unknown at the time of the interview, had observed a lack of support from community members who may not be familiar with LQTS and how it can affect one's daily life:

"sometimes I feel that in a very subtle way, [other families] kinda don't really support it or they [don't understand] it…[there's a] lack of communication…a lot of people don't know and so when you're not feeling well and don't want to participate in some of the um social things that are happening then…the support is not always there" (Participant D)

Participants cited difficulty and frustration because of the general lack of knowledge about LQTS in the community. It is a condition that is managed

effectively, and therefore individuals do not appear ill. Yet, they live everyday with the possibility of a sudden cardiac event to occur at any moment.

2.4 Discussion

The goal of this study was to understand the impact of living with a genetic condition found in the Gitxsan community that might confer risk to sudden death. Factors that both facilitate and hinder resiliency and coping were explored. We anticipated that the results from this study would have both similarities and differences to that of previous reports yet would provide details that are unique to First Nations culture.

The present study suggests that participants' reaction of feeling *overwhelmed, emotional, and scared* to the initial diagnosis of LQTS is comparable to groups in the Netherlands, Norway, and United States (Hendriks et al, 2005; Farnsworth et al, 2006; Smets et al, 2007; Giuffre et al, 2008; Muelenkamp et al, 2008). This study also revealed some coping mechanisms that may be unique to Gitxsan culture, such as emphasis on spirituality and faith. Perhaps spiritual beliefs are what it takes to get through uncertainty. Faith is not necessary when you know how things are going to work out, that is knowledge. It is in the time of unknowing that having faith is what sees you through to the other side. Faith is what gives you strength.

Learning that you or a family member has LQTS can be troubling. Participants most frequently cited family support as a mechanism of coping. Additionally, GC was seen as an important step for participants' ability to cope

with LQTS. Many stressed that the knowledge conveyed during the result-giving GC session about what LQTS is and how to interpret their result helped participants to understand how LQTS will affect their lives and what to expect in the future.

As mentioned, learning about Long-QT Syndrome appears to be a distressing event. This result is consistent with other genetic counselling studies that have explored genetic testing for autism (Selkirk et al, 2009), and various other genetic conditions (Michie, Marteau, & Bobrow, 1997). This is common within many GC research studies. The amount of information presented in a GC session is great, and participants in this study reported that genetic counsellors did a great job of breaking the information down into manageable bits. However, research has demonstrated that patients do not retain much of the information that is presented in a session (Hallowell et al, 1997; Hopwood, Howell, & Evans, 2003) Therefore, follow up care is essential, as many individuals may require more than one explanation of LQTS before they have a complete understanding.

The most important factor that facilitates an individual's ability to cope with LQTS appears to be the support provided by families. In Gitxsan culture, family and notions of kinship are fundamental. Literature surrounding Aboriginal resilience suggests that it is collective, rather than individual, and encompasses spirituality, family strength, Elders, ceremonial rituals, oral traditions, identity, and support networks (HeavyRunner & Marshall, 2003).

Spirituality also played a key role in an individual's ability to cope. Many participants spoke about relying on prayer to help them through the struggles

they experienced in coping with LQTS. In a survey of 127 North American genetic counsellors, only 60% reported having asked about a patient's spirituality in the past year, and of those who did, the majority only brought up the topic after the patient had mentioned it (Reis et al, 2007). Given the results of this study, it may be beneficial for genetic counsellors to ask Gitxsan patients who are being investigated for LQTS to ask if they consider themselves spiritual.

It was informative how many participants reported inconsistent medical advice, especially with regard to appropriate levels of physical activity. As most participants were mothers themselves, the main concerns were for their children's opportunity in sports. Participants were living in a small rural town, where school and recreational sports leagues are a favorite pastime. With the frequent turnover of hospital staff, participants were unsure of appropriate levels of activity.

2.4.1 Expansion of Genetic Counselling Issues from a Cultural Perspective

A literature search for the terms "genetic counselling", and "First Nations", "Native", "Indigenous", "Indian" or "Aboriginal" resulted in one published peer-reviewed paper. As genetic counselling is a relatively new health care profession, it is just now beginning to develop a model of practice. Many of the concepts detailed in the proposed *Reciprocal-Engagement Model* (McCarthy et al, 2009) overlap with core values and beliefs shared by many Indigenous Peoples. These issues are explored in further detail in chapter three.

2.4.2 Limitations of the Study

The participants in this study were all being tested for two mutations (V205M and R591H) that were commonly found in near their community. Varying amounts of time had passed with regard to when each participant had first learned about LQTS – some had only learned about this condition within the past year, whereas other had known for ten years or longer. Additionally, each participant arrived at genetic testing for LQTS dependent upon their unique familial experience with LQTS. More stringent inclusion criteria may have been beneficial. For example, only interviewing people who have demonstrated an indentified measurable coping level. However, the diversity that is reflected in this study, in terms of when diagnosis was received and their health care experience, provides a bigger picture of the impact LQTS can have on an individual's life.

Another limitation is that only women volunteered to participate in this study. However, LQTS affects men as well. It is unfortunate that no male perspectives represented in this study. We can only speculate reasons why men who are at risk for LQTS are not coming forward for this LQTS study. The issue of gender is explored further in chapter four.

It is difficult to know if the study participants are representative of the entire population of community members who are impacted by LQTS. This is a common limitation of qualitative research, where recruitment relies on self-selection. It is likely that the people who volunteered for this study are coping well with LQTS, as it takes courage and strength to discuss some of the issues surrounding this potentially fatal condition.

This study was designed to give a voice to the participants whose lives are impacted by LQTS. The information gathered in this research study helps to provide the beginnings of a rich understanding of the psychological and social impact of LQTS in a First Nations community. These results can be useful to improve the quality of genetic counselling and overall health care, as well as stimulate and promote further research in this field. It should be noted that the results from this study cannot be generalized to the entire population.

2.5 Conclusion

All ten women who participated in this study appeared to be coping well with LQTS. Having knowledge about the biological and clinical aspects of LQTS greatly increased their ability to cope. Additionally, participants' families were able to provide the support needed and create a safe environment where an individual could share their symptoms, experiences, and discuss their vulnerabilities.

A noteworthy factor contributing to an individuals' inability to cope was conflicting medical advice from various health care practitioners. Participants noted the frequent turnover in hospital staff at their rural hospital, and the poor understanding of LQTS by other prominent people in the community (public school teachers, particularly physical education teachers, as well as sports coaches).

All participants were mothers, and their main concern was not for themselves, but for their children, grandchildren, and for the generations yet to

come. The multigenerational impact of LQTS is the focus of chapter 4. The stories revealed in this study indicate that LQTS has a major impact on the daily lives of participants and also highlights the various strategies they use to cope with this potentially fatal disease. Further investigations into the psychological and social issues experienced by this patient group are necessary.

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Chapter 3 Long QT Syndrome Genetic Counselling: Experiences within a British Columbia First Nations Community[2]

3.1 Introduction

"Aboriginal peoples today experience the kinds of health problems most closely associated with poverty, yet they also suffer from problems linked to their historical position within the Canadian social system" (Waldram, Herring, & Young, 1995). In the spring of 2004, the Vice President of BC Women's Hospital hosted a meeting at the Gitanmaax Community Hall to identify local concerns regarding priorities for health and how they should be managed. Instead of listing the anticipated concerns for conditions such as diabetes, heart disease, or tuberculosis, the group of Gitxsan women who gathered at this meeting voiced their concern for a rare cardiac condition, called Long QT Syndrome (LQTS).

Today there are approximately 1.1 million Aboriginal people living in Canada (2006 Census, Statistics Canada). There are three distinct groups of Aboriginal people living throughout the country – 61% are First Nations, 34% are Métis, and 5% are Inuit (Indian & Northern Affairs Canada). Each group has unique local geographic and linguistic heritages, cultural practices, and spiritual beliefs. It is important to avoid generalizations.

There are 198 registered First Nations bands in British Columbia (BC) (Indian and Northern Affairs Canada). There is wide diversity among these groups. British Columbia has a vast wealth of First Nations languages. It is home

[2] A version of this chapter will be submitted for publication. Huisman, L., Arbour, L., McCormick, R., & Gitxsan LQTS Research Advisory Board. (2010). Long QT Syndrome Genetic Counselling: Experiences within a British Columbia First Nations community.

to 60% of First Nations languages in Canada with 32 languages and about 59 dialects (First Peoples' Heritage, Language and Culture Council, 2010). However, there are some historic roots, as well as some beliefs and traditions that are common. For example, the Medicine Wheel is a philosophy shared by several First Nations groups.

A clinician researcher with considerable experience conducting health research with Inuit and First Nations communities was alerted about the Gitxsan community's concern for LQTS. A community based participatory research approach was agreed upon, and a local research advisory committee was formed and continues to guide the research.

LQTS is a rare condition, characterized by prolonged ventricular repolarization. It can either be acquired or inherited. Found worldwide, it has an estimated prevalence of 1 in 2,000 (Schwartz, 2009). There is a disproportionately high rate of hereditary LQTS in a First Nations (FN) community in northern British Columbia (Arbour et al, 2008). This is partly due to a novel missense mutation, V205M, in KCNQ1, a known LTQS-susceptibility gene. The effect has been described (Arbour et al, 2008) and predisposes affected individuals to syncope, arrhythmia and sudden death.

Patients being investigated for hereditary LQTS are typically seen by a genetic counsellor. Genetic counsellors are health care professionals with expertise in medical genetics and counselling. They work with patients to facilitate the adjustment to genetic conditions. Genetic counselling (GC) is a relatively new health care profession. Genetic counsellors often work as

members of an multidisciplinary healthcare team, providing both individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions (NSGC; CAGC). This definition describes the many roles that a genetic counsellor plays within their profession. The first North American program opened in 1969 at Sarah Lawrence College (Bronxville NY), while the first Canadian GC program opened in 1985 at McGill University.

Modeled after social work, genetic counselling was built upon the values and beliefs of dominant Western culture. The original curriculum focused on the ability to teach medical genetic information to patients. In reviewing the original GC practice (Marks, 1993) recognized the role that emotional responses played when patients learned genetic information. The curriculum was expanded to include a larger focus on counselling skills and techniques, using Carl Roger's *Client-Centered Counseling* approach to address the arising psychosocial issues.

The majority of genetic counsellors are Caucasian females under 40 years of age, ranging from 84.6% – 94.2% of the total profession (CAGC, 2007; Koltz, 2009; Parrott & Del Vecchio, 2007). Correspondingly, the National Society of Genetic Counselor's (NSGC) 2006 Professional Status Survey reported that Native Americans/Alaska Natives consistently represent less than 1% of NSGC membership. Current efforts to increase the diversity in the GC profession have been disjointed, sporadic, and lack realistic goals and effective implementation and evaluation components (Mittman & Downs, 2008).

3.1.1 Rationale for an Aboriginal Approach to Genetic Counselling

Western values and beliefs differ from those of many Aboriginal[3] people. For example, many Aboriginal Peoples value a sense of collectivity, reflecting their belief of the interconnectedness of all beings, whereas Western culture values a sense of individual autonomy. This clash in values contributes partly to the ineffectiveness of current GC practices and care with Aboriginal patients. Table 3.1 compares Western and Aboriginal values and beliefs.

Table 3.1: Comparison of Cultural Values and Beliefs

Western Cultural Values & Beliefs	Aboriginal Cultural Values & Beliefs
Individual Autonomy	Sociocentric (connection to family, community, natural & spirit world)
Individualism	Collectivism
Fragmentation of mental, physical, emotional & spiritual dimensions	Balance & interconnectedness of mental, physical, emotional & spiritual dimensions
Linear	Circular

Diversity among ethnic groups is not so much biophysical as it is socially inherited cultural differences. Culture can be defined in many ways, the most universally agreed upon definition being “a group of people’s total way of life: the way they act and think, organize themselves, relate and communicate, make or build things, express feelings and emotions, and respond to the world” (Kroeber & Kluckhohn, 1952).

Canada prides itself on being a multicultural country. This diversity is reflected in the wide variety of cultural heritages, with many Canadian people identifying with more than one (Multicultural Canada). The diversity of worldviews

[3] The term Aboriginal is meant to be all inclusive of First Nations, Métis, & Inuit.

contributes to complexity of applying the current mainstream genetic counselling paradigm to patients and families who identify with minority heritages.

Recently in the field of Aboriginal health, there is an emphasis on the need for programs to be both culturally competent *and* culturally safe. Both cultural competence and safety have been identified as key components in developing services to Aboriginal People (NAHO, 2009). The US Department of Health and Human Services defines *cultural competence* as “a set of congruent behaviors, attitudes, and policies that come together in a system, agency, or among professionals and enable that system, agency, or those professionals to work effectively in cross cultural situations”. While *cultural safety* is described as “[moving] beyond the concept of cultural sensitivity to analyzing power imbalances, institutional discrimination, colonization, and colonial relationships as they apply to health care” (NAHO).

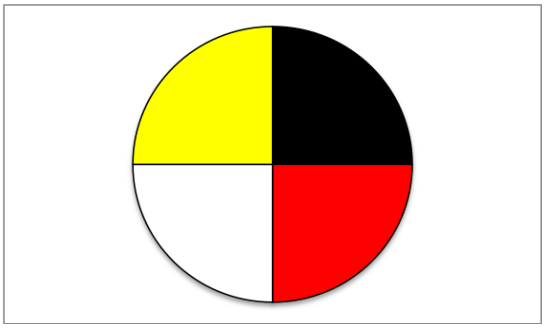
3.1.2 Medicine Wheel Literature in Health related Fields

Many Indigenous Peoples of North America use the Medicine Wheel (MW) to teach various concepts related to balance in all natural systems. The circular nature of the MW (with no beginning and no end) emphasizes relationship, interconnectedness, harmony, and balance (Hart, 2002). The concept of the MW has been around for thousands of years, and was first introduced to mainstream public knowledge in 1984 when the book “The Sacred Tree” was published.

Most medicine wheels follow the basic pattern of having a circle divided into equal quadrants (Figure 3.1).

"The Medicine Wheel concept from Native American culture provides a model for who we are as individuals. We have an intellectual self, a spiritual self, an emotional self, and a physical self. Strength and balance in all quadrants of the Medicine Wheel can produce a strong, positive sense of wellbeing, whereas imbalance in one or more quadrants can cause symptoms of illness. Addressing issues of imbalance can potentially diminish your patient's symptoms and enrich their quality of life." (Montour, 1996)

Figure 3.1: Medicine Wheel



There is an obvious lack of research in this area. The concept of the Medicine Wheel has been applied to various other helping professions, such as counselling psychology, social work, and nursing (McCormick, 1996; France et al, 2004; Dapice, 2006; Verniest, 2006; MacDonald, 2008). Notably, the foundational concepts of the MW are in line with and complement some of the concepts proposed in the *Reciprocal-Engagement Model* – relationship and reciprocity.

3.2 Methods

3.2.1 One Research Example: Impact of Long QT Syndrome

A community-based participatory research approach has enabled over 250 Gitxsan community members to take part in a study that identified more than 40 mutation carriers. Although a great deal of previous research has been carried out on the biological and clinical aspects of LQTS, there has been little study into the impact of living with a mutation that increases risk for syncope, seizures and sudden cardiac death.

In a sub-study of LQTS in the Gitxsan community, qualitative research methods were used to explore the personal and familial impact of living with a condition that increases the risk of sudden death (described in chapter 2). To our knowledge, this sub-study was the first study to provide cultural insights into the issues that a remote First Nations (FN) community might face. The concepts and ideas expressed in this chapter have developed from a need for culturally safe and culturally competent genetic counselling (GC).

The qualitative research methods were offered for this study were individual interviews, Photovoice, and a Talking Circle. These methods are explained in detail in elsewhere in this thesis; interview procedure can be found in chapter 2, Photovoice procedure can be found in Appendix B, and Talking Circle procedure can be found in chapter 4. The results from the individual interviews are presented in this chapter.

3.2.2 Data Analysis

The data gathered from the interviews was originally analyzed using the Systematic Text Condensation method (Giorgi, 1985; Malterud, 2001). The results from that analysis were presented in chapter 2. At the outset of this research study, we anticipated that the information gathered would be used by genetic counsellors who work with FN patients and families. It became apparent to us that the impact of LQTS touched on all aspects of participants' wellbeing. Therefore, the four quadrants of the MW (mental, physical, emotional, and spiritual) were used as a framework to analyze the information gathered during this study.

3.3 Results

Ten women participated in the interview portion of this study. A sociodemographical description of the participants can be found in chapter 2.

Each of the realms of the MW is equally important. Additionally, singular events, practices, or factors, may fall into more than one realm, as they are not completely exclusive. For the ease of clarity and explanation, this paper will highlight the impact in each realm, and direct quotations are used where possible illustrate components of each realm. One participant, who was diagnosed with LQTS after a cardiac arrest, addressed how the impact touched on all aspects of her wellbeing:

> *"Um, physically, emotionally, spiritually… it was quite, um, quite a kick in the ass. (laughing) To say the least"* (Participant C)

3.3.1 Emotional Dimension

The emotional dimension of the Medicine Wheel represents feelings, the activities and potentials of the heart (Hart, 2002).

Participants of a LQTS study reported feeling very emotional when learning about their diagnosis. As one participant recounted her experience:

> *" my emotions were when the, um, doctor initially told me that there was something wrong with my heart…there was something they need to investigate and I needed to be put into [the hospital's] heart unit. Um, I remember thinking okay, that's fine. And I went to the phone in the hospital and totally broke down on the phone…I think I was just devastated for that moment. Ah thinking that there was something wrong with me"* (Participant A)

All ten participants were able to vividly recall the date that they learned about LQTS. Many described what they were doing right before learning the news of LQTS, then describing their immediate reaction. Some of the words that participants used in recounting their experience were: *devastated, sad, emotional, scary, overwhelming, afraid, anxiety, traumatic.*

One participant, whose mutation status was unknown at the time of the interview, described her reaction to her sister's apparent cardiac arrest:

> *"The emotions were very overwhelming to note how serious and how um of being afraid. Um having lots of anxiety. Um not knowing if people would be alive the next day."* (Participant D)

Another participant, whose genetic results confirmed her clinical diagnosis, shared her reaction to learning which parent she inherited the V205M mutation from:

> *"It was mixed emotions and ah shocking to find it was my dad that was the one that passed it along to us…we really honestly thought it was mom until the day [the genetic counsellors] came by. When they said it was dad. We were – we were just floored"* (Participant A)

This participant also shared her feelings surrounding passing on the gene from one generation to the next:

> *"Somewhere I had to um assure dad that it was okay that you know that he was the one that passed the gene on to me. And I'm glad that I had that opportunity…so he didn't feel guilty. Um, you know I said it's okay dad. I said you know you've lived a long life and I said you give me hope. I said you are my hope now"* (Participant A)

Participants recalled a vast range of emotions during their personal journey with LQTS. All participants shared emotions that can be perceived as traumatic, which is not unexpected considering the potentially fatal nature of this condition.

3.3.2 Intellectual Dimension

The intellectual dimension of the Medicine Wheel represents activities related to the mind, often referred to as the thinking self. Several participants

relied on the knowledge and understanding of LQTS as coping mechanism. One participant recalled the date she received her results, saying

> *"when the [researchers] came and explained all of this. That was very helpful. In understanding what [long qt syndrome] is"* (Participant G).

This participant went on to explain that learning this information gave her a sense of relief, reporting that she had always had a feeling that something was wrong but never knowing the reason for the mild symptoms she had experienced.

Another participant stated that *"[the genetic counselling session] helped to clear a lot of things up"* (Participant G).

Additionally, a lack of knowledge regarding long qt syndrome was reported to hinder participants' ability to cope. One participant, who has an implanted defibrillator and whose genetic results confirmed her clinical diagnosis said

> *"I really haven't had any educational information or you know counselling or anything about this Long QT and today I still don't really, you know, understand"* (Participant G)

3.3.3 Physical Dimension

The physical dimension of the Medicine Wheel represents the body, the shell that protects the spirit. All ten participants described the physical symptoms that they had experienced.

One participant commented,

> *"ever since I was a child, my heart would always flip. Now, I don't know, that's the only way I can describe it. It just jumped around in my chest.*

Ah, for as long as I could remember. And then after I was diagnosed, and had the um the ICD, the defibrillator … the pacemaker implanted, my heart no longer does that. So, physically my heart doesn't jump around" (Participant A).

Another participant, whose genetic status was unknown at the time of the interview, communicated:

"my heart [was] pounding…yeah sometimes it would feel like it would just pop right out of my chest" (Participant G)

In addition to recounting the symptoms that participants have experienced (either personally or in family members), participants also discussed the limitations and reservations they have surrounding physical activity. One participant, who has siblings diagnosed with LQTS but whose mutation status was unknown at the time of the interview, spoke about weighing the benefits of exercise against the potential harm of triggering a cardiac event:

"It really puts a limit on their potential, particularly when they want to strive for sports. You kind of have to find a little balance, so that way they don't harm themselves" (Participant D)

Another participant, who was diagnosed with LQTS as a teenager, shared the advice she received:

"*I wasn't allowed to play outdoor sports. And I love softball-soccer. It was a big change. I had to kick it back a little bit."* (Participant I)

3.3.4 Spiritual Dimension

The Spiritual dimension of the Medicine Wheel represents the characteristics of relationship, unity, honor, balance, and healing (Lane et al, 1984). Spirituality means different things to different people. It is a difficult concept to define, and is equally difficult to quantify for the sake of research.

Participants in the LQTS sub-study all reported at least one measure of spirituality or faith. The most frequently reported spiritual practice was prayer. One participant talked about the role of prayer in her family's life:

> *"My grandmother was a prayer warrior and she really, really let us see and bring us to places where we can see the miracles and to know that power. But when um you see and feel things differently and I don't want to be hurting myself by taking all this hurt and pain and putting it inside. And I find that sometimes it's easier just to let it go to him and he'll take care of it"* (Participant A)

Participants relied on the faith that their parents had instilled in them from childhood. Believing in the Creator enabled them to let go of the need to act stoic and allowed them to feel comfort, protection and calmness.

> *"And our Creator, I know that protected my sister and there was a lot of um family gatherings in our home. We did a lot of smudging and lots of – lots and lots of prayers"* (Participant D).

Another participant spoke of the spiritual practices she and her family did during her mother's recovery from an apparent sudden cardiac arrest:

"the [the Medicine Man] did like songs and stuff and um we did a – like – a circle where we all just did a prayer for her. To help her get through her coma and stuff…I think we did a smudge too" (Participant I)

Some of the other participants shared stories of using other traditional Gitxsan customs.

"*we'll go to sweat lodge and - and - and we'll pray and so that way the extended House group gave us a lot of support, which really meant a lot to me"* (Participant A)

3.4 Discussion

The goal of this study was to explore what facilitates and hinders resiliency and coping for those living with LQTS. The overall impact this condition has on peoples' lives was examined in within the context of the MW. To our knowledge, this is the first time that the impact of a genetic condition has been described using this approach.

3.4.1 Current Approaches to Genetic Counselling

There are two main approaches to genetic counselling. The original approach used was the teaching approach. With this approach, the genetic counsellor is viewed as the expert and the goal is to teach the patient relevant medical and genetic information. An alternative approach is the counselling approach. With this approach, the genetic counsellor is viewed as a counsellor

and the goal is to understand the patient, provide support, and facilitate problem solving (McCarthy Veach, LeRoy, & Bartels, 2003).

Today most GC graduate programs use a blend of both teaching and counselling approaches, but there is no universally defined *Model of Practice*. The American Board of Genetic Counseling (ABGC) has developed practice-based competencies and the *Scope of Practice* that help guide genetic counsellors in defining the profession and the parameters of how the service is delivered. In 2008, McCarthy Veach et al proposed a *Reciprocal-Engagement Model* based on genetic counselling tenets and goals that were identified by the GC graduate program directors.

3.4.2 Culturally Competent Genetic Counselling with First Nations Peoples

A literature search for the terms “First Nations” or “Aboriginal” or “Indian” or “Indigenous” or “Native” and “genetic counselling” revealed one single result. In 1996 an account of genetic counselling in a Navajo kindred was published (Lynch et al, 1996). Genetic testing for nonpolyposis colorectal cancer was performed on 51 family members, and DNA-based genetic counselling was provided to 23 individuals. Based on the participants’ reaction to the genetic information present in the GC session, Lynch et al (1996) noted “those individuals who were given positive results were for the most part stoic” (p. 32). This is a similar result to the findings of the present study.

The number four is a sacred number to many Aboriginal. There are four seasons – spring, summer, autumn, winter; four directions – east, south, west,

north; four “races” – black, white, red, yellow; and four aspects of wellbeing – mental, physical, emotional, and spiritual.

The number four is also prominent in the field of genetic counselling. The helping relationship can be divided into four phases – preparation for case, engagement, follow up, and conclusion. The responsibilities of genetic counsellors can be divided into four roles – educator, interpreter, counsellor, and facilitator. There are four medical ethical principles – autonomy, beneficence, nonmaleficence, and justice. As well, there are four domains of genetic counsellor competencies – communication skills, critical-thinking skills, interpersonal/counselling/psychological assessment skills, and professional ethics and values.

It has been reported that genetic counsellors do not consistently assess a patient’s spirituality. A cross-sectional study of a convenience sample of 127 genetic counsellors investigated the current spiritual assessment practices of genetic counsellors and their reaction to the HOPE spiritual assessment tool (Reis et al, 2007). Most participants self-reported being mildly, moderately, or strongly spiritual (81.7%). Sixty percent of participants indicated that they had performed a spiritual assessment within the past year; yet only 8.7% reported using them for more than half of their genetic counselling sessions. Primary reasons cited for not performing spiritual assessment included: limited time during GC session, thinking the client would be uncomfortable discussing spirituality, and not knowing what to do with the information attained from an assessment (Ries et al, 2007).

Cragun et al. (2009) investigated religious measures of genetic counsellors and compared them to a representative sample of the U.S. population. This study found that genetic counsellors were significantly more likely to report having no religious affiliation (nearly ¼), less likely to believe in god (OR = 0.540), significantly less likely to frequently attend religious services (OR = 0.690), significant less likely to pray frequently (OR = 0.297), and significantly less likely to believe in immortality (OR = 0.314).

Interestingly, in the present LQTS sub-study, participants reported faith and spirituality as the second most common factor that enabled their ability to cope with LQTS. Seven out of ten participants reported a reliance on prayer. These results suggest that it would be helpful to Gitxsan patients if genetic counselors would address spirituality during a GC session.

A genetic counselor can help a patient describe and identify their feelings as they go through the process of genetic counselling. The *counsellor* role is especially important during the follow up phase of the counselling relationship.

A genetic counsellor can help a patient understand the results of their genetic testing and understand the implications this will have on his or her life. This *educator* role is especially important during the pre genetic testing phase of the counselling relationship. Being an educator at this phase is crucial in preparing a patient for the information that they are going to learn about themselves, it cannot be unlearned.

A genetic counsellor can help a patient understand how a genetic condition will affect them physically and help facilitate any transitions they might

have to make in their daily life. A genetic counsellor can be an *interpreter* and translate the results of the genetic testing so the patient understands the meaning of their results. Table 3.2 demonstrates the four roles a genetic counsellor plays and gives examples of the relation to the MW.

Table 3.2: Examples of how the MW relates to each role a GC plays

Genetic Counselor Role[2]	Example of the Relation to Medicine Wheel
Interpreter Interpret family & medical histories	• help patient understand familial impact of disease – *emotional, mental & spiritual* • help patient understand symptoms & prognosis – *physical & mental*
Educator Educate about inheritance, testing, management, prevention, resources, treatment & research	• help patient understand genetic test results & impact – *mental, physical, emotional, & spiritual* • give information about disease management – *mental, physical, & spiritual*
Facilitator Facilitate the decision making process, family discussion, sharing results	• help patient identify their own values & priorities – *mental, emotional, & spiritual* • present information in a culturally sensitive way - *mental, physical, emotional, & spiritual*
Counsellor Help patient adapt to the risk of the condition, listen to patient, communicate clearly, establish rapport	• refer patients to local counsellors & support groups – *mental, emotional, & spiritual* • help patient adjust to idea of genetic risk – *emotional & spiritual*

3.5 Conclusion

Genetic counsellors play a crucial role in the health care team for patients with genetic conditions. In order to best serve First Nations patients, the current genetic counselling paradigm must be re-visited and perhaps modified to reflect unique First Nations worldviews.

The Medicine Wheel concepts appear to provide an excellent framework to base a First Nations approach to genetic counselling on. During a genetic counselling session, it is important to address all four aspects of well-being – the mental, physical, emotional, and spiritual. Further research into GC with First Nations patients will enable the appropriate modification the current GC paradigm to suit patients need best.

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Chapter 4 The Multigenerational Impact of Long QT Syndrome: A Gitxsan Perspective[4]

4.1 Introduction

In 2004, heath care providers and a clinician researcher were alerted to a British Columbia (BC) First Nations community's concern for Long QT Syndrome (LQTS). A community-based research program has enabled over 300 participants to undergo genetic testing for LQTS. Approximately 60% of the participants were women and 40% men. These participants represented more than 50 families.

LQTS is a potentially fatal disease, and can be acquired or inherited. It is a complex condition and affects people all over the world at an estimated prevalence of 1 in 2000 (Schwartz, 2008). It can be caused by a mutation in one of the genes that code for imperative ion channel sub units. These mutations potentially cause disruptions in the heart's conduction system because of altered ion exchange, and may result in a prolonged QT interval observed on electrocardiogram and ventricular arrhythmias. Clinical signs most often appear in adolescence and include predisposition to palpitations, syncope, and seizures without warning (Hobbs et al, 2006). Effective treatments to prevent these cardiac events include the use of beta-blockers, an implantable cardioverter defibrillator, and left cardiac denervation. As many as 30% of individuals with

[4] A version of this chapter will be submitted for publication. Huisman, L., Arbour, L., McCormick, R., Martin, S., & Gitxsan LQTS Research Advisory Board. (2010). The Multigenerational Impact of Long QT Syndrome: A Gitxsan Perspective.

LQTS will never experience a symptom, and for roughly 10% sudden cardiac death may be the first event (Goldenberg et al, 2008).

To date, an estimated 12 genes are thought to be responsible for congenital Long QT Syndrome (LQTS). Genetic screening for these susceptibility genes first became commercially available in 2005 (Familion ®). The early detection of genetic factors contributing to LQTS can lead to successful treatment and prevention recommendations. The community study with a BC First Nation, identified a novel mutation (V205M) which was partially responsible for the disproportionately high rate of LQTS type 1 in this population.

In British Columbia (BC), physicians refer patients with a medical and family history of LQTS to medical genetics. There is one provincial medical genetics program, and five affiliated clinical genetics clinics in BC. Five clinics are located in Vancouver and one in Victoria.

British Columbia, especially the lower mainland is a diverse, multicultural environment. It is therefore common for genetic counsellors and geneticists to be from an ethnic background different than their patients. This trend is observed nation wide and there is a growing body of research in the field of multicultural genetic counselling. While major scientific discoveries are uncovering the biological reasons for genetic diseases, much less attention has been paid to how people perceive genetic information and what they do with it. It is important for genetic counsellors, and all health care providers, to acknowledge the role and influence of culture. Brunger and Bassett (1998) eloquently stated that:

“People do not make decisions as members of ethnic communities, or as autonomous Individuals. Instead, decisions are usually based on a variety of factors: educational opportunities, societal expectations and norms, family obligations, and relationships, and so on. All of these factors are shaped by culture. Therefore, responses to testing are neither uniform nor predictable” (p. 21).

To date however, only one study (Lynch et al, 1996) has been published that explored genetic counselling issues in an American Indian tribe. The Canadian Association of Genetic Counsellor’s has recognized the lack of research concerning GC in Aboriginal groups and their 2009 annual education conference marked the first symposium that focused on current genetic counselling practice and research within First Nations communities. A common theme among recommendations for multicultural GC is the need for a genetic counsellor to be respectful of other worldviews and to be aware of their own worldview.

Notions of family and kinship are integral to the Gitxsan people. Johnson (2000) explains:

> “Gitksan society is hierarchical, with matrilineal corporate groups called Houses (*Wilp*) headed by Chiefs. The Gitksan relationship to land differs from that of most Western peoples; for the Gitksan, people are part of the land, in an inextricable and even social relationship with it. The health of the land and that of the people are intertwined, and there is, as we have

seen, a spiritual value to land and the relationship to other species" (p. 303)

The overall goal of the present study was to gain a more comprehensive understanding about LQTS genetic testing in a First Nations community, and the impact it has on individuals, their families, and the community at large. Three objectives were introduced at the end of chapter one; this chapter is concerned with the third objective – to understand the multigenerational impact of LQTS.

4.2 Methods

In consultation with the local research advisory committee, three qualitative methods were selected in order to address the objectives set out for this study. Participants had the option in partake in one or all three of the methods – individual interviews, Photovoice, and Talking Circles. Participants were recruited from the core study "Exploring the impact of Long QT Syndrome in northern British Columbia" (Arbour et al, 2008). This chapter will focus on the data collected during the Talking Circle. The other two methods and results are presented elsewhere (interview findings discussed in chapters 2 and 3; Photovoice discussed in chapter 5).

We held a Talking Circle in January 2009. All participants who volunteered for the interview portion of this research project were telephoned and hand delivered a thank you note, which included an invitation to take part in the Talking Circle (Appendix B). Of the ten personal invitations extended, five participants

expressed interest, and two actually attended the Talking Circle. Posters and advertisements were placed around the community to invite everyone.

4.2.1 Talking Circle

Talking Circles are similar to focus groups that are often used in qualitative research. However, the main difference is the protocol used in a traditional Talking Circle (TC). A TC is a sacred place; it is a space of respect, harmony, and balance. Traditionally a Talking Stick is passed around, and whoever is holding the stick speaks their truth, without interruption. The rest of the people actively listen. There are different types of circles: healing circles, mediation circles, and sharing circles.

Four women participated in the TC, and are referred to as participants in this chapter. Two of the TC participants also took part in individual interviews, and two of the TC participants only took part in this research method.

We held a Sharing/Caring Circle. The theme chosen to reflect on was “The impact of Long QT Syndrome”. It was a time for the participants to come together in a safe place and reflect on whatever was in their hearts. It was also an opportunity for participants to either confirm or contest the preliminary findings of the interviews.

The Talking Circle (also referred to as a Sharing Circle) was held on a Friday and began at twelve o’clock. A local Elder was invited to lead the circle. As the eldest female in the group, she provides the natural leadership. As is custom,

she opened with a prayer. Next, we discussed how to address the topics that were proposed.

4.2.2 Case Family

Four generations of the *L family* came to the Talking Circle. The eldest in attendance was the great-grandmother, Linda. Linda's daughter (Lori) was also at the TC. Lori had three children; from eldest to youngest they are Larry, Lisa, and Luke. Lisa participated in the TC. Lisa has one daughter (Lily). Lisa brought her preschool daughter Lily to the TC. Lily was too young to contribute to the discussion, but she listened and observed the TC, which was her role as a child. Participation in the TC was completely voluntary. It was a coincidence that four generations of the same family participated. This reinforces the strong role of family support in the L family.

As an infant, Luke's pediatrician was concerned about an 'innocent heart murmur'. There was particular vigilance to the family, since Luke's aunt (Lori's sister) had died suddenly, just after child birth. Luke was referred to a pediatric cardiologist who performed an electrocardiogram (ECG). Luke's ECG revealed an abnormally prolonged QT interval, and was diagnosed with LQTS. This initiated cascade testing for LQTS in this family. Lori was clinically diagnosed with LQTS in 1999.

Lori was one of the index cases of the larger core community study on LQTS, which identified the V205M mutation. Many of the family members of the *L family* enrolled in the community study. Linda had never experienced any

symptoms or cardiac events, but had suspicions that she was a mutation carrier. Lori's two youngest children (Lisa and Luke), who were living with her at the time of the study, also enrolled in the community study. Lisa was also clinically diagnosed when her ECG showed a prolonged QTc interval.

Linda, Lori, Lisa, and Lily (four generations of the same family) all attended the Talking Circle. This paper presents a unique perspective from the *L family* into the multigenerational impact that LQTS may have on a family.

Upon consultation with the local research advisory committee, Linda was invited as a local Elder to lead the Talking Circle. The Talking Circle was used as an opportunity for the interview participants to review and validate the preliminary findings from the interview transcripts. Five questions were posed to the group (Table 4.1).

Table 4.1: Questions addressed during the Talking Circle

What is the first thing that comes to mind when I mention LQTS?
What is the most helpful event/incident that helped you accept/cope with knowing about LQTS in you/your family?
What is the least helpful event/incident that helped you accept/cope with living with LQTS?
Is there a need for a support group for LQTS? If yes, what exactly would it look like? What does the community need?
Men who may be at risk for LQTS don't seem to be coming forward for testing. Why do you think this is?

Since the Talking Circle is a sacred space, out of respect for the participants and the cultural meaning, there is no audio recording or verbatim transcript developed. With permission, notes were taken and have been used in conjunction with transcripts from the interviews to piece together the important concepts and ideas that were discussed. The information has been thematically analyzed and the data is presented in categories, using direct quotes where possible to illustrate the significance of each category.

4.3 Results

Several stories and personal experiences were shared regarding each participant's journey with LQTS. There were five questions set out by the researchers; however, a Sharing Circle is a safe space for individuals to share their truth at that moment in time. Therefore, the anecdotal information is presented in this chapter.

4.3.1 Role of Family

During the TC participants stressed that family support was crucially important to an individual's ability to cope with LQTS. Family members were able to provide love and compassion. The *L family* spoke about how helpful it was to receive their genetic test results all together. Typically, individual patients receive their genetic test result privately, and a genetic counsellor may assist a patient to share their result with family members if they wish. At the Sharing Circle, the *L*

family recollected their choice to receive their genetic results all together in Linda's home. Having the entire family together to receive the news helped them each "to be strong for each other".

Both Lori and Lisa had clinical diagnoses before enrolling in the community study, and each shared that they hoped the DNA result would prove that they had incorrectly been diagnosed with LQTS. They wished that "it would all have been wrong" and they wouldn't have to worry about LQTS anymore. Neither Lori or Lisa were surprised when their DNA results came back positive for the V205M mutation, but they both had to let go of that hope that they would be "normal" again.

Linda on the other hand, shared the shock and disappointment of her negative result. During the circle she shared stories of her relatives experiencing LQTS-like symptoms, including fainting history and palpitations in her siblings and maternal aunts. The more events Linda could recall, the more she was convinced her family carried the "bad gene", since they could trace symptoms of LQTS in her family members and not in her husband's relatives. Linda had prepared herself for getting a positive result. It was important for the L family to communicate their feelings.

4.3.2 Importance of Laughter

In the *L family*, it was also obvious that laughter was viewed as a coping mechanism. The three adult women at the Talking Circle used humour and laughter as a way to "keep things in perspective".

The L family recalled how Linda's husband reacted to the LQTS result. Linda's husband had never experienced any symptoms or cardiac events. When he learned that in fact he did carry the V205M mutation, he and the rest of the *L family* were in complete shock. While the L family was waiting for results, Linda's husband used to joke with his family about the nature of LQTS in the community saying, *"I think it's because ah you know maybe relatives were going with relatives"*.

In response to the first time LQTS was diagnosed in the family, Linda shared this about the support she received from her parents and family:

> *"I would say my family has been a big help. They've been um positive about this whole thing. I can't remember um my parents never ever broke down. They were just um so strong in their faith that I was going to be okay. So that's probably where I got that strength from – that faith. Um, so yeah my parents have a lot to do with you know influencing me. And their strength also I turned it around and it was my strength for my kids, then"*

4.3.3 Concern for the Next Generation

The strong family ties in the *L family* and their Gitxsan cultural heritage placed large value and importance for the youngest children of this generation and for the generations yet to come. The three adult women in the *L family* disclosed at the Talking Circle that it was easier to "deal" or "cope" with their *own* result than it was to "deal" or "cope" with *their child(ren)'s* result(s).

Interestingly, when Lori's youngest grandchild (her eldest son's daughter) was later tested for the V205M mutation, Lori commented *"I [realized] that I did not have to emotionally hold up [my son], his girlfriend and their daughter, because they had each other and she has strong family ties from her side"*

Lori talked about guilt that she felt for having passed along "bad genes" to her children. As mentioned, Linda's husband tested positive for the V205M mutation. Anticipating her father would have a similar reaction to herself, Linda shared her need to let her father know it was okay to have passed along the mutation:

> *"somewhere I had to assure dad that it was okay that you know that he was the one that passed the gene on to me. And I'm glad that I had that opportunity…I said you know you're – I said you've lived a long life and I said you give me hope. I said you are my hope now"*

This quote demonstrates the burden of knowing one's genetic status on an individual with LQTS.

4.3.4 Men at risk for LQTS

When asked to speculate reasons why men who may be at risk for LQTS are not coming forward for testing, the participants smiled and laughed. Initial discussion revolved around the observed stereotype that men appear less interested in health in general – that this is not unique to LQTS. Linda shared a story of how difficult it was to get her husband to see a doctor for anything. Lori shared a story that echoed that sentiment. When first invited to enroll in the core

LQTS study, Lori's eldest son expressed no interest. When Lori's genetic result confirmed her clinical diagnosis, she approached him again. He still was not at all interested in knowing his genetic status, and found comfort in his borderline QTc interval on ECG and the fact that he had not experienced any symptoms. However, when his girlfriend became pregnant, he felt a responsibility to his child and enrolled. Because several family members had been diagnosed with LQTS he was not too surprised with his positive genetic result for the V205M mutation, and shortly after birth, his daughter also tested positive.

4.4 Discussion

In the present study I examined the mutigenerational impact that LQTS has on one Gitxsan family. The *L family* is a tightly knit group. Each family member knows that they can rely on the others when the stress and emotional toll of LQTS becomes overwhelming. It is important for health care providers to recognize the pivotal role of *family*.

Clinical practice and research in the field of genetics challenges the conventional principle of patient autonomy. In this case study, we clearly see that one person cannot make a decision without it affecting the rest of their family.

4.4.1 Gitxsan Concept of Family

The central role of family may not be unique to this Gitxsan family, but it is important that health care providers understand the roles of family and kinship in

a First Nations community. The traditional, hereditary system of the Gitxsan nation is still upheld today:

> "It is a matrilineal system with members of a Wilp, or House, tracing their lineage through their mothers. All Gitxsan belong to a Wilp, which is the basic unit for social, economic and political purposes. The Wilp is a collection of closely related people. It consists of one to several families and membership can number from 20 to more than 250 people. Each Wilp has a hereditary chief. A hereditary chief may have several wing chiefs who perform particular functions for House members" (Gitxsan Chiefs Office, 2010).

Understanding the social structure of Gitxsan society will help genetic counsellors in a multicultural GC session. Many genetic counsellors report concerns about culture, especially when it comes to obtaining informed consent from minority groups. With the majority of genetic counsellors being Caucasian females, it is not uncommon for the genetic counsellor to be from another ethnic background compared to the patient. In order to provide effective GC services to patients from different cultures and ethnicities, genetic counsellors need to become effective multicultural counselors: they need to increase their knowledge of relevant cultural groups while, at the same time, becoming more aware of their own cultural roots and beliefs (Weil, J. 2001).

4.4.2 Genetic Counselling and Families

Dr. Sheldon C. Reed coined the term genetic counselling in 1974. Reed (1955) reported three requirements for genetic counselling: 1) knowledge of human genetics, 2) respect for sensitivities, attitudes, and reactions of clients, and 3) teach and provide genetic information to the full extent known. GC, by nature, challenges the biomedical principle of autonomy. When you are dealing with genetic issues, you are dealing with family issues. Weil (2000) discussed the notion of family beliefs:

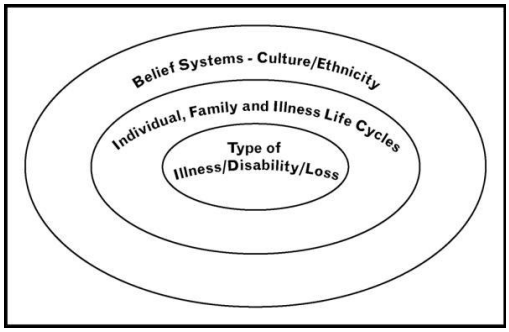
> "Every family functions with a complex set of beliefs, perceptions, values and rules concerning the nature of the world, appropriate responses to situations and problems that are encountered, and the resources and degree of control that are available to meet life challenges" (p. 34).

Family beliefs are inherently tied to an individual's culture and worldview. Weil (2000) also described aspects of family beliefs that are relevant in the genetic counselling setting:

1. Beliefs concerning the normal and acceptable range of human variability.
2. The personal and social meanings of the particular situation or disorder.
3. Beliefs concerning the ability to affect the course of life events.
4. Beliefs concerning the cause of genetic disorders, birth defects, and other situations relevant to genetic counselling.

The Family Systems Illness model (Rolland & Williams, 2005) may offer a framework to build family resilience. This model "emphasizes the interaction of illness, individual, and family development; multigenerational patterns of coping with illness, loss, and other adversity; and belief systems, including influences of culture, spirituality, and gender" (p. 6).

Figure 4.1: Family System-Illness Model (Rolland, 1994)



This model is similar to the "Philosophy Wheel of the Gitxsan" (Smith, 2004). Both models are represented in a circular fashion, and symbolize the illness at the centre with family and cultural factors as outer realms, which influence the person at the centre.

When the term resilience is used in an Aboriginal context people generally think about residential schools. However, the term is also used in various other contexts, used most basically to describe the ability to "bounce back from adversity". The family resilience perspective is "grounded in a systemic orientation, it looks beyond the parent-child dyad to consider broader influences in the kin network, from sibling bonds to couple relationships and extended family ties" (Walsh, 2003). Walsh's definition of family resilience describes the coping style of L family. It will be important for genetic counsellors working with Gitxsan

people to recognize that collective decisions may be the norm, rather than typical Western individualized autonomy.

Without having information directly from the men who decline genetic testing, we can only speculate reasons why they are not coming forward. Different cultures express feelings and emotions differently. Gitxsan culture varies from Western culture in terms of when and how emotions are appropriately expressed. Gitxsan people, especially men, value stoicism. Though QTc interval may not be the best indicator of whether an individual has the V205M mutation, and thus a predisposition to LQTS, Gitxsan men may find comfort in knowing that their QTc interval was within normal range. We speculate that another reason men may not be coming forward for testing is that men, in general, exhibit self-care behaviour differently than women.

Gitxsan people have a “complex, matrilineal, kinship-based social and political structure” (Napoleon, 2002, p. 156). There are approximately 60 Wilps (Houses) from the four Clans mentioned earlier. Napoleon goes on to state “There is no one authority over the Houses; rather, each has an internal authority system and reciprocal relationships with other Houses...[and] within Gitxsan society, individual rights are expressed and acted on as collective rights” (p. 165). The central role of family provides for the natural protection of elders and children.

4.4.3 Limitations of the study

One limitation of this case study is whether the *L family* is a representative example of a typical family impacted by LQTS is debatable. This is a common limitation to qualitative studies, and to case study designs in particular. Most likely, we cannot tease out general factors and principles, which will apply to every family that is impacted by LQTS. However, it is notable from the data generated from this Talking Circle is that other families will likely have strengths and resources they can access which will greatly help the emotional challenges a LQTS brings to a family.

4.5 Conclusion

There has been little research on people's wider experiences of LQTS and their health care trajectories. To our knowledge, this may be the first account of the multigenerational impact of LQTS in a First Nations family. Given the oral tradition of the Gitxsan people, it may be beneficial to take this study one step further and construct a story based on the L family's experience. Further research is required to explore this further, so that new families who are identified with LQTS can benefit from the best support available and help ease the significant life adjustments that accompany LQTS.

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Chapter 5 Discussion and Conclusion

5.1 Psychological and Social Impact of LQTS Assessed in this Thesis

Through sharing their stories, the participants have voiced their personal experience and wisdom concerning their journey with LQTS. The findings from this study are substantiated in the literature. Information gathered from this study may contribute knowledge to genetic counselling theory, policy, practice, and the community. This information also provides an increased understanding of the needs of First Nations people diagnosed with LQTS and offers guidance for creating culturally relevant strategies for genetic counsellors and geneticists to incorporate into their wellness plans when working with First Nations patients and their families.

The research presented in this thesis provides the first account of the psychological and social impact of LQTS in a First Nations community. We can see several similarities and differences when comparing the results of this study to the published literature in this field.

The introductory chapter provided an overview of the Gitxsan Peoples, LQTS, and genetic counselling. Together these sections all led to the rationale behind this research study – Gitxsan Peoples want to understand the factors that contribute to resiliency and coping with regard to LQTS. The second chapter of this thesis focused on the factors that affect an individual's ability to cope. The third chapter addressed the scope of the psychological and social impact of LQTS in terms of the Medicine Wheel. And the fourth chapter presented a family as a case study, demonstrating the multi-generational impact LQTS can have.

Together, these three chapters provide an all-encompassing glimpse of the impact LQTS has had on the Gitxsan community.

Here I will briefly review the findings of this research, discuss the strengths and weaknesses of this study, summarize current knowledge, suggest future investigations required in this field, and highlight the significance of this research.

At the outset of this study, only one peer-reviewed paper had been published in the literature (Hendriks et al, 2005). A Dutch research team measured anxiety (both disease-related and state) and depression in a group of 37 parents who had children undergoing genetic testing for LQTS. Researchers reported that the continuous threat of developing LQTS-related symptoms affects the psychological wellbeing of the parents for at least eighteen months (Hendriks, 2005). Several participants in our study raised concerns for their child(ren). Participants voiced particular concern for the child(ren)'s involvement with physical activity.

Since this principal study, five other research teams have published academic papers concerning some aspect of the psychological impact of LQTS (Farnsworth et al, 2006; Smets et al, 2007; Giuffre et al, 2008; Anderson et al, 2008; Meulenkamp et al, 2008).

One of the striking similarities between previous studies of parents' concerns and the stories participants in this study shared is parents' concern for their children. Farnsworth, Forsyth, Haglund, & Ackerman (2006) described the first qualitative study on American parents with young children who have been diagnosed with LQTS. Interviews with 31 parents revealed that parents do in fact

have a fear of their young children dying. Some of the actions parents have taken to avoid this tragic possibility include providing cell phones for their children and checking their messages often, using a baby monitor during the night in the child's room, and taking a portable defibrillator to events that their child takes part in (Farnsworth, 2006). A few participants in the present study voiced concerns similar to this. One participant had similar expression of fear:

> "*I was fearful for them [my kids]. I had the faith for me, but for some reason it – I kept reading in the [Sudden Arrhythmia Death Society] website that so many thousands of children die each year from this, so I was in panic state when I found that my kids had it. I don't think I had a gotten a decent night's sleep for a good year. I'd be checking up on the constantly throughout the night, to see if they were breathing. It just freaked me right out"* (Participant A).

5.2 Strengths and Weaknesses of this Study

One of the most notable strengths of this research study is its collaborative nature. There has been concern for the nature and intent of conducting research within Aboriginal communities, especially with regard to genetic research (Nuu chah nulth and Havasupai cases). The Canadian Institutes of Health Research, Institute of Aboriginal Peoples Health, have developed guidelines for researchers who wish to explore Aboriginal Peoples health in a research setting. In line with these guidelines, the core community study followed a community-based

approach, and the sub study presented in this thesis followed a collaborative approach.

A second notable strength of this research study was the research design. It was a first attempt to define the psychological and social impact of LQTS in a First Nations community. The three methods (individual interviews, Photovoice, and Talking Circle) enabled participants to voice their experience. Participation in this study also proved to validate each participant's experience with LQTS. Additionally, the design adhered to the Kirkness & Barnhardt's 4 R's (1991) – respect, relevance, reciprocity, and responsibility. The research respected the Gitxsan people and their culture. The study was relevant to the community's health care concerns. The research relationships within the project were of a collaborative nature and reciprocal. And, the study was conducted in a responsible manner. Interestingly, although Photovoice methodology is a recognized way of recognizing individual voices within communities, no individuals opted for it. The reason is unclear, but we recognize the importance of introducing choice of methods of expression. Offering three methods enabled participants to voice their experience in a manner in which they felt most comfortable.

While the study yielded several fascinating results, it was limited by a number of factors. One of the weaknesses of this study was the small sample size of 12 participants – 8 women participated in the interview only, 2 women participated in the TC only, and 2 women participated in both research methods. This is a common limitation to qualitative studies. Additionally, only women are

represented in our sample population. This was not unexpected, as only a few men have enrolled in the core community study, only 9 of the 42 (21%) V205M carriers were male. Without speaking to the men who have not participated, I can only speculate reasons that might explain this imbalance. As mentioned in chapter four, it may be partly due to the culture value on stoicism. Also, it may be partly due to the fact that I am a young female researcher who grew up outside of the community. I might have been viewed as an outsider, or perhaps it is a cultural norm for men to speak only to other men about health-related issues.

Another weakness was the relatively broad inclusion and exclusion criteria. The study may have been stronger if more stringent criteria were laid out, for example, interviewing only participants who have demonstrated an identified and measurable coping skill. However, having a wide range of coping and resilience levels represented in this sample provides a wide range the overall impact of LQTS.

5.3 Current Knowledge and Future Research on the Psychological and Social Impact of LQTS

This thesis adds to the growing body of research regarding the psychological and social impact of LQTS. It offers a unique perspective from First Nations women. The stories shared during this study reveal that receiving a LQTS diagnosis is an overwhelming event.

Since GC is still a relatively new profession, and given that it is currently developing an official model of practice, it is an ideal time for studies like this one

to contribute in creating a multicultural model rather than a monocultural model as it has been in the past. The proposed *Reciprocal Engagement Model* (Figure 1.5) shares some similarities with the Philosophy Wheel of the Gitxsan (Smith, 2004). Both models acknowledge that each element of the model is complementary to the other elements. Both models also suggest ideally relationships are based on mutual respect and each partner benefits from the reciprocal nature of the relationship.

Additionally, it has been noted "the *complexity* of genetic information is not what makes it so difficult for patients to comprehend, rather its *emotional impact* makes understanding difficult" (McCarthy et al, 2007, p. 725). Participants in this study cited family support, spirituality and faith as being central for resiliency and coping with the knowledge of LQTS. This result echoes the importance of personal beliefs, religious and cultural traditions and their influence on general attitudes toward illness and medical treatment. The profound impact these beliefs should always be explored with patients.

Future studies on the psychological and social impact of LQTS might focus on creating a population specific, validated questionnaire which addresses quality of life interactions with spirituality, and includes a pre-post test format to allow new patients to experience the GC session and then elaborate on their spiritual needs following their session.

5.4 Significance

We anticipated that the psychological impact of LQTS would be similar in the Gitxsan community compared to the general population. This hypothesis is partly true. There were several similarities, such as parents concern for their children, dissatisfaction with health care practitioner's knowledge of LQTS, anxiety and worry for serious cardiac events. However, some of the factors that participants cited as being helpful in their ability to cope were unique to the Gitxsan Peoples.

Notions of family and kinship are at the heart of Gitxsan culture. This was demonstrated in the results that are discussed in chapter 2, 3, and 4. Gitxsan society is built upon relationships. It was expected that LQTS would impact the physical and emotional realms of a patient. Yet, this study revealed that knowledge of LQTS also impacted the mental and spiritual realms of these Gitxsan women. The Medicine Wheel has been used to provide a framework within other helping professions, and I propose that it is indicated for use in the GC setting as well.

The information presented in this thesis has potential application for genetic counsellors who are working with First Nations patients and families. Many valuable teachings can be derived from the stories shared by the twelve participants in this study.

Additionally, the factors that facilitate resiliency and coping for LQTS may be transferable to similar genetic conditions, such as hypertrophic cardiomyopathy and familial hypercholesterolemia.

In addition to the rich information this study will provide for the field of GC, it also has facilitated the healing process in the Gitxsan community. In talking with one participant, she shared with me these beautiful words:

> *"You know I always give thanks to the Creator for allowing me to find the Long QT because unfortunately my sister never had that chance. Um, she just died suddenly. In hindsight tells us she must have had the Long QT as well, but we just didn't know. Um, so that that was the downfall. Knowing that [my sister] never got the chance to find out that she had Long QT and she could have worked with it. So this is why this [the research study] is exciting, because the word is getting out there…And there may be a thousand more [sisters] that, you know, we could help live"* (Participant A)

5.5 Conclusion

LQTS is a relatively rare cardiac condition that disproportionately affects the Gitxsan communities. The V205M mutation is thought to have a significant genetic contribution to this initial observation (Arbour et al, 2008). However, little is known about the psychological and social implications of LQTS. In this thesis several factors were examined for their influence on an individual's ability to cope. The findings have clarified some aspects of this field and may ultimately have implications for patient counselling, however perhaps more significantly this research has illuminated new areas of research in this field. This provides background for future research into genetic counselling with First Nations patients. The psychological and social impact of many genetic conditions,

including LQTS, is poorly understood. This is increasingly concerning given the advent and promises of personalized genomic medicine.

5.6 References

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Appendix A: Interview Guide

1. Please take a moment to recall or remember the day you/someone you know found our you/they had LQTS. What emotions/thoughts did you have when you were told this information?
2. What does that result/information mean to you now?
3. As soon as possible after you received this information, please try to recall an incident or event that occurred that **helped** you in your ability to cope with the LQTS diagnosis. Tell me about that event.
    a. What exactly happened? (e.g. what did the person say or do, etc)?
    b. How did you feel afterwards (e.g. what was the outcome of the event)?
4. As soon as possible after you received this information, please try to recall an incident or event that occurred that **hindered** you in your ability to cope with the LQTS diagnosis. Tell me about that event.
    a. What exactly happened? (e.g. what did the person say or do, etc)?
    b. How did you feel afterwards (e.g. what was the outcome of the event)?
5. Has knowing you/they have LQTS changed your daily life? If yes, how? If not, how do you keep things the same?
6. How often do you think about LQTS compared to other things in your life? Per month? Week? Day?
7. Did you choose to receive genetic test results? Why? Why not? Are you glad that you have them? Why?

8. How has the knowledge of the condition affected your relationships (husband, children, etc)? do you children understand that you have this? How did they react when told? How do you feel about potentially passing this on to your child?
9. How has your culture/spirituality influenced your reaction to LQTS?
10. How satisfied are you with the support provided by health care providers locally? How could they provide better support for you?
11. Is there anything else you think I should know about how LQTS impacts your life?

Appendix B: Photovoice

Introduction

The second phase of this research project was to use the participatory action approach of Photovoice (1996). Photovoice is a well-recognized Participatory Action Research (PAR) method. The emphasis of this approach is on personal voice and facilitative teaching methods in a group setting (Wang *et al*, 1996; Wang, 1999). This approach uses still photos along with written narrative to tell a personal story. Photographs are used as a catalyst to engage participants. This is a participatory action research strategy that is increasingly valued by health professionals (Wang et al, 1996). This qualitative research method has also been modified to explore notions of health in an Indigenous community (Castleden, Garvin, & Huu-ay-aht First Nation, 2008). This technique is appropriate in this research setting because it goes along with the storytelling tradition of the Gitxsan people. This approach also upholds the principles of CB research, which is consistent with the new CIHR guidelines.

Proposal

Disposable cameras will be bought and distributed to participants for use. Each participant will be given one camera and allowed 1-2 weeks in order to capture their photographs. An introductory session will introduce the Photovoice technique and guide participants to create their story using digital photos and written narration. The concept of Photovoice will be introduced and participants will be asked to take photographs of places and activities that represented the

impact that LQTS has had on their daily lives. When participants return the cameras, I will develop the film and meet each participant individually to discuss their

Result

No one participated in this method of conveying the impact of LQTS on their life.

References

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Appendix C

Talking Circle

First of all I would like to thank each of you for participating in this research project. Secondly, I would like to invite you to take part in a talking circle. This talking circle will be an opportunity for us to reflect back on the common themes I have drawn from the interviews I conducted last summer (2008). Below are the questions I would like to discuss at the talking circle.

Questions

1. What is the first thing that comes to mind when I mention LQTS?
2. What is the most helpful event/incident that helped you accept/cope with knowing about LQTS in you/your family?
3. What is the least helpful event/incident that helped you accept/cope with living with LQTS?
4. Is there a need for a support group for LQTS? If yes, what exactly would it look like? What does the community need?
5. Men who may be at risk for LQTS don't seem to be coming forward for testing. Why do you think this is?

Talking Circle – Exploring the impact of long qt syndrome

Agenda – January 23rd 2008 – 12pm

- Welcome and Prayer – Elder
- Review agenda for session, and ground rules – Lee-Anna Huisman & Elder
- Introductions
- Explain the main objectives of the session – why are we all here?
- Questions
    1. What is the first thing that comes to mind when I mention LQTS?
    2. What is the most helpful event/incident that helped you accept/cope with knowing about LQTS?
    3. What is the least helpful event/incident that helped you accept/cope with living with LQTS?
    4. The majority of participants so far have been women. Men who may be at risk for LQTS don't seem to be coming forward for testing. Why do you think this is?
    5. Is there a need for a support group for LQTS? If yes, what exactly would it look like? What does the community need?
- Closing and Prayer – Elder

Appendix D: UBC Behavioural Research Ethics Board Certificates of Approval

The University of British Columbia
Office of Research Services
Behavioural Research Ethics Board
Suite 102, 6190 Agronomy Road,
Vancouver, B.C. V6T 1Z3

CERTIFICATE OF APPROVAL - FULL BOARD

PRINCIPAL INVESTIGATOR:	INSTITUTION / DEPARTMENT:	UBC BREB NUMBER:
Laura Arbour	UBC/Medicine, Faculty of/Medical Genetics	H07-02249

INSTITUTION(S) WHERE RESEARCH WILL BE CARRIED OUT:

Institution	Site
N/A	N/A

Other locations where the research will be conducted:
Hazeltons' Community Health Centre Subject's home UBC Island Medical Program

CO-INVESTIGATOR(S):
N/A

SPONSORING AGENCIES:
Canadian Institutes of Health Research (CIHR)

PROJECT TITLE:
Exploring the Impact of Long QT Syndrome: A closer look at the factors that facilitate and hinder resiliency.

REB MEETING DATE:	CERTIFICATE EXPIRY DATE:
November 22, 2007	November 22, 2008

DOCUMENTS INCLUDED IN THIS APPROVAL: **DATE APPROVED: December 17, 2007**

Document Name	Version	Date
Protocol:		
Exploring the Impact of Long QT Research Proposal	5	November 7, 2007
Consent Forms:		
Exploring the impact of Long QT Consent Form	8	December 10, 2007
Exploring the Impact of Long QT Photo Consent Form	1	December 10, 2007
Assent Forms:		
Exploring the Impact of Long QT Assent Form	4	December 4, 2007
Advertisements:		
Exploring the Impact of Long QT Recruitment Poster	3	December 4, 2007
Questionnaire, Questionnaire Cover Letter, Tests:		
Exploring the Impact of Long QT Sample Interview Questions	3	October 25, 2007
Other Documents:		
Support Services in Hazelton BC	2	October 25, 2007
Support letter from Gitxsan Health Society	N/A	October 30, 2007

The application for ethical review and the document(s) listed above have been reviewed and the

procedures were found to be acceptable on ethical grounds for research involving human subjects.

Approval is issued on behalf of the Behavioural Research Ethics Board and signed electronically by one of the following:

Dr. M. Judith Lynam, Chair
Dr. Jim Rupert, Associate Chair
Dr. Laurie Ford, Associate Chair

Appendix E: Consent and Assent Forms

All Subjects Informed Consent Form

Exploring the Impact of Long QT Syndrome: A Closer Look at the Factors that Facilitate and Hinder Resiliency

Principal Investigator: Dr. Laura Arbour, UBC Department of Medical Genetics, 1 Hospital Way, Victoria, BC

Co-Investigators (researchers): Dr. Rod McCormick, UBC Department of Education, Education Counseling and Special Education; Sarah McIntosh, UBC Department of Medical Genetics; Lee-Anna Huisman, Masters student, UBC Genetics Graduate Program

Funding: The Canadian Institutes for Health Research – Institute for Aboriginal Peoples' Health is funding this research project.

Introduction:
You/your child are/is invited to participate in this study if Long QT Syndrome (LQTS) is part of your life.

The participation in this study is strictly voluntary. Your participation or refusal of participation will not alter your care or the care of your relatives. Please take the time to review this information, think about it, talk about it with others if you wish, and decide whether you want to participate.

Who can participate in this study? We are inviting all First Nations people in and around the Hazeltons who have been impacted by Long QT Syndrome to participate in this study. This includes people who are at risk for LQTS, diagnosed with LQTS, have a friend/relative with LQTS, or care for someone with LQTS.

Purpose:

You are invited to participate in a research project that will provide information to health care providers (doctors, nurses, genetic and other counselors) and others with the condition about what its like to live with Long QT Syndrome.

What does the study involve?

We will use three different methods in this research study. You are invited to participate in the interview and will also have a choice of 2 other opportunities to communicate your views and feelings. Your involvement in this study will take

about one (1) to five (5) hours, depending on how much you would like to be involved.

1) We will interview you and ask you questions about your experience living with LQTS.

2) If you would like another way of communicating, in addition to the interview we will help you tell your personal story of being impacted with LQTS through the creation of Photovoice. Photovoice is research method that involves giving participants access to a digital camera for a week. They use the camera to capture images that help them express themselves. They use the photos to write a story about their experiences and feelings.

3) Thirdly, we invite you to take part in traditional talking circles, where you can share your experiences and feelings regarding LQTS with others in your community. *The group will decide whether they want the group discussion used in the report of the study.*

Although the study reports will use the information learned from the interview, photo-voice and possibly the talking circle (only with consent). All identities will be kept confidential. Participants will not be named in any reports.

Results:

Under no circumstances will your personal information from this study be given to anyone else without your explicit direction, except when required by law. This information will be used only for the purpose of this research study. Any publication or discussion of these research results will use anonymous codes instead of your name.

It is expected that this study will be completed in about one (1) year. A copy of the report will go to the Gitxsan Health Society. No individual information about genetic predisposition will be included in the report to the Gitxsan Health Society.

Any publications will be reviewed by the Community Research Advisory Committee and the Gitxsan Health Society before being finalized.

Risks:
Potential risks of participating in this study are that it may be upsetting to recall and speak about your feelings. We have made arrangements for support services in your community if they are needed, and you can decide not to discuss any issues that make you uncomfortable.

Benefits of research:
Benefit from this study cannot be guaranteed, but the information may be useful to others who are impacted by LQTS, to health care providers caring for people with LQTS, and to others impacted by similar conditions.

Confidentiality:

You/your child's confidentiality will be respected. No information that discloses you/your child's identity will be released or published without your specific consent to the disclosure. However, research records identifying you/your child may be inspected in the presence of the Investigator or his or her designate by representatives of Health Canada, and the UBC Research Ethics Board for the purpose of monitoring the research. However, no records which identify you/your child by name or initials will be allowed to leave the Investigator's offices.

Signing this consent form in no way limits your/your child's legal rights against the investigators, or anyone else.

Contact:

If you have any questions or desire further information with respect to this study, please contact Dr. Laura Arbour or one of her associates or Lee-Anna Huisman.

If you have any concerns about your treatment or rights as a research subject you may contact the Research Subject Information Line at the University of British Columbia at 604-822-8598.

Consent:

I understand that participation in this study is entirely voluntary and I may refuse to participate or may withdraw from this study at any time without any consequences to me, or my family's continuing medical care. As well, I do not have to give any reason for refusing to participate or deciding to withdraw. If I withdraw from the study, the information collected up until that time will be used in the analysis of the study, unless otherwise stipulated by me.

A copy of this consent form is provided for my records.

I agree to participate in this study as discussed with me and described above. If I decide to also participate in photo-voice or talking circle, I will sign the bottom of this form at that time.

_____ _____
Subject printed name and signature Date

_____ _____
Witness printed name and signature Date

_____ _____
Investigator's printed name and signature Date

Addendum:
I agree to participate in photo-voice

_____ _____
Subject printed name and signature Date

_____ _____
Witness printed name and signature Date

_____ _____
Investigator's printed name and signature Date

I agree to participate in talking circles.

_____ _____
Subject printed name and signature Date

_____ _____
Witness printed name and signature Date

_____ _____
Investigator's printed name and signature Date

All Subjects Assent Form:

For those who are too young to consent themselves (ages 10-13), but capable of assent

Exploring the Impact of Long QT Syndrome: A Closer Look at the Factors that Facilitate and Hinder Resiliency

Long QT Syndrome (LQTS) is a condition that affects the way the heart beats. Some people with LQTS become sick, but many people with the condition are healthy and never know they have it.

Many people in the Hazeltons have LQTS.

People in Hazelton and at the University of British Columbia are doing research to find out more about LQTS and what its like for people to live with LQTS.

You are invited to be a part of this research, either because you have LQTS or because one of your relatives has this condition. This study involves three (3) different activities. The first is an *interview*, where we will ask questions about what it is like to have or know someone with LQTS. The second is *Photovoice,* where you create your own story using pictures you take. The third is *Talking Circles*, where Hazelton families affected by LQTS have the chance to get together and share how they are feeling. You can choose to be involved in any or all of these activities.

The information you share with us about living with LQTS will be used to help us (doctors, nurses, counselors) understand what its like to live with LQTS. This, in turn, may help us to provide better care and support for families with this condition.

Your identity will be kept secret, and your name will not be used in any research reports.

It is up to you to decide whether you want to be in the research.

I agree to participate in the interview.

Name

Date

Addendum:

I agree to participate in photo-voice

Name

Date

I agree to participate in talking circles.

Name

Date

All Subjects Photo Consent Form

Exploring the Impact of Long QT Syndrome: A Closer Look at the Factors that Facilitate and Hinder Resiliency

Principal Investigator: Dr. Laura Arbour, UBC Department of Medical Genetics, 1 Hospital Way, Victoria, BC 250-727-4461

Co-Investigators (researchers): Dr. Rod McCormick, UBC Department of Education, Education Counseling and Special Education; Sarah McIntosh, UBC Department of Medical Genetics; Lee-Anna Huisman, Masters student, UBC Genetics Graduate Program

Thank you for your participation in the creation of a Photovoice project. After analysis of your story, and only with your permission, we would like to use your story in publications and presentations.

I agree to the use of my work in the following:

- ☐ Public talks and meetings
- ☐ Academic presentations
- ☐ Published in an academic journal
- ☐ None of the above – I prefer to keep my work private

Additionally,

- ☐ I will like to know before these presentations
- ☐ I will not be identified by name
- ☐ I will be identified by this name:_____

Consent:

A copy of this consent form is provided for my records.

I agree to participate in this study as discussed with me and described above.

_____ _____

Subject printed name and signature Date

_____ _____

Witness printed name and signature Date

_____ _____

Investigator's printed name and signature Date

Appendix F: Gitxsan Health Society Letter of Support

GITXSAN HEALTH SOCIETY

BOX 223 HAZELTON, B.C. V0J 1Y0
TEL: (250) 842-5165 FAX: (250) 842 - 0079

October 30th, 2007

Behavioral research Ethics Board

Re: Lee-Anna Huisman – Graduate Thesis Research Proposal

Dear Board Members,

My name is Neil Belanger and I am the Executive Health Director for the Gitxsan Health Society operating in Hazelton, located in Northwestern British Columbia.

The Gitxsan Health Society provides services to three, semi-isolated communities' within the Gitxsan First Nation. These communities are; Gitanmaax, Kispiox and Sik-e-dakh (Glen Vowell), in addition to members of those communities residing off reserve, living between Smithers and Terrace, collectively over 2,000 people.

The Gitxsan Health Society, more accurately the Gitxsan Nation, continues to have an expressed interest in the past, current and continued research of Long QT Syndrome (LQT) within our communities, which was begun by Dr. Laura Arbour. The importance of this research, its findings and furthering our understanding of it, can not be accurately conveyed. As a direct result of the LQT research completed to date, our organization has committed considerable financial resources to purchase mobile defibrillators for each of our communities and in doing so, assisting to ensure the health and welfare of our people.

Lee-Anna's proposed research, as an extension of the continuing clinical research on LQT, is not only consistent with the Gitxsan Health Society's mission; it is a crucial component of our updated health plan, builds on existing strengths, programs and services and builds capacity for our people, it is vital that this work continues.

On behalf of the Gitxsan Health Society, our Board of Directors and the communities we serve, I would ask that you accept this letter as our endorsement and support for the proposed research programme that Lee-Anna Huisman wishes to undertake within our communities.

In closing, I would like to thank you for your time and attention, should you require further information, please do not hesitate to contact my office.

Respectfully,

Neil Belanger
Executive Health Director
Gitxsan Health Society