“Am I going to die today?”

An investigation into the psychosocial impact of Long QT Syndrome, healthcare experiences and the genetic testing process

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ABSTRACT

Background: LQTS is an inherited cardiac condition that imposes serious risks, including sudden death, and a restricted lifestyle. The limited international research on psychosocial responses to the condition, and to the genetic testing process, indicates high levels of anxiety and depression, negative effects on quality of living and significant problems accessing adequate medical care. To date, no studies on the psychosocial impact of LQTS have been completed in New Zealand.

Aims: The aims of this study were: (i) to describe the medical and psychosocial factors impacting adjustment to LQTS in New Zealand patients and families, and (ii) to compare the perspectives of clinicians and patients, of asymptomatic and symptomatic patients, and of adolescent and adult patients.

Methodology: In-depth semi-structured interviews were conducted with fourteen LQTS gene positive outpatients (three adolescent and eleven adult) and eight cardiac clinicians from New Zealand's specialist national LQTS service. These were analysed using thematic analysis.

Results: Many patients described positive experiences with the tertiary care service that the national specialist clinicians provided, while some described significant distress in relation to the healthcare they received in primary and tertiary care. Differences were identified between symptomatic and asymptomatic presentations, with symptomatic individuals describing significantly more distress in more areas of psychosocial functioning than their asymptomatic counterparts. Asymptomatic adolescents described significant impacts on their quality of life as a consequence of the restrictions imposed to reduce risk of sudden death. Most of the psychosocial and medical challenges identified by the clinicians were consistent with those described by patients. Many patients described using information and positive risk appraisals to help them cope with being at constant risk of sudden death.

Conclusion: The collective experiences described have important implications for healthcare services, especially regarding targeted psychological support and improved service implementation.

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When the heart weeps for what it has lost,
the soul laughs for what it has found.

~Sufi aphorism ~

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Chapter 1

Literature Review

Congenital Long QT Syndrome is a genetically inherited cardiac condition that causes the heart to beat irregularly, depriving the brain of oxygen resulting in fainting and seizures, and in some cases, the heart to stop and sudden death to occur. The ages that incur the highest risk for these cardiac events are vulnerable infants and children, but all ages are affected. Needless to say, Long QT Syndrome can have a profound impact on the individuals and families who are diagnosed with it.

Over the last two decades, research into the genetic origins of Long QT Syndrome has discovered that particular symptom clusters and related faults in the functionality of the individual's heart, and sometimes other systems, are caused by several localised genetic mutations. These have come to be known as Long QT Syndrome subtypes. Genetic testing is now used to identify these genetic subtypes as this information allows genotype specific treatment planning and the identification of triggers for cardiac events. For the relatives of those diagnosed with Long QT Syndrome, genetic testing plays an important role. It identifies those also at risk for a cardiac event, as not all who have Long QT Syndrome have symptoms. In fact, some never experience a symptom but despite this, are still at risk of having a cardiac event that could lead to sudden death. Once their genetic status is ascertained however, those that have a Long QT Syndrome genetic mutation identified can begin prophylactic treatment, modify their lives to minimise risk and avoid potential triggers, thereby substantially decreasing the likelihood of having a cardiac event.

With a condition such as Long QT Syndrome the individuals and families affected are required to cope with multiple psychosocial stressors, including the receipt of clinical and then genetic diagnosis and waiting for, receiving and living with the diagnoses of their close family members. They usually have to make significant lifestyle changes in order to minimise their risks and cope with the threat and/or presence of multiple cardiac events, not knowing if one may lead to their own, or their child, brother or mother's sudden death. This dissertation therefore describes research aiming to expand the limited existing knowledge on the psychosocial factors impacting Long QT Syndrome affected individuals going through this genetic testing process. It also aims to aid New Zealand's specialist

congenital cardiac service for Long QT Syndrome to understand, develop and sustain practices that meet the psychosocial needs of their Long QT Syndrome affected patients. This is the first psychosocial research of Long QT Syndrome completed on the New Zealand population.

This chapter is ordered into sections that provide a context for the research that follows. Firstly, the epidemiology and etiology of Long QT Syndrome is explained in relation to cardiac electrophysiology, genetics and inheritance. This is followed by the medical presentation and clinical management of Long QT Syndrome. The focus then changes to psychosocial factors, the first section of which delineates the research findings on the psychosocial factors that are impacted by chronic health conditions. After that, the psychosocial factors that are impacted by Long QT Syndrome are presented, followed by a discussion on healthcare and Long QT Syndrome patients' psychosocial needs. Finally, the chapter ends with the research findings on the psychosocial impact of genetic testing in general, and for Long QT Syndrome in particular.

The Epidemiology and Etiology of Long QT Syndrome

Forms of Long QT Syndrome

There are two forms of Long QT Syndrome, known as Acquired Long QT Syndrome and Congenital Long QT Syndrome. The acquired type is usually secondary to the effects of medications, such as some antipsychotics and cancer treatments, while congenital Long QT Syndrome (LQTS) is an inherited condition mostly affecting children and young adults (Camm, 2004). Congenital LQTS is the condition under investigation in the current study and is referred to as LQTS henceforth.

Prevalence

Many studies reporting epidemiological figures rely on information gathered from the LQTS International Registry. This is the single largest pool of information about LQTS available to researchers (Zareba et al, 2003; Goldenberg et al., 2008). In the most recent prevalence study LQTS was estimated to occur in one of every 2,500 people (Levine et al., 2008) with other estimates varying from 1:1000 to 1:5000 (Crotti, Stramba-Badiali & Ferrandi, 2005; Farnsworth, Fosyth, Hagland and Ackerman, 2006). Some researchers believe these figures may be underestimates as LQTS continues to frequently be unrecognized or misdiagnosed (MacCormick et al., 2009).

Distribution of LQTS

As an autosomal dominant genetic condition individuals with a relative or a family history of LQTS symptomatology, including sudden death at a young age, have a higher risk of being LQTS gene positive. Studies have not yet provided clear gender and ethnicity rates compared to the general population but information from the International LQTS Registry suggests LQTS gender rates may be consistent with general population rates (Goldenberg et al., 2008). Ethnicity distribution data is not yet available. However, for those with a gene based diagnosis, the risk of having a LQTS related cardiac event, and the frequency with which they may occur, differ depending on the individual's age and gender (Sauer et al., 2007). Cardiac events happen the most frequently in childhood. Males have an overall higher risk and earlier onset of cardiac events than females (Zareba et al., 2003). Males are also at higher risk than females during preadolescence (Sauer et al., 2007). In the post-adolescent adult period (18 to 40 years) however this reverses, with females being at greater risk. This is thought to be associated with hormones specific to women (Goldenberg et al., 2008; Locati et al., 1998; Zareba et al., 2003) and the protective role of testosterone in men (James, Choisy, & Hancox, 2007). There is a decreased risk of LQTS related cardiac events in females during pregnancy but increased risk during the 9 month post-partum period, although evidence suggests this risk is genotype specific (Heradien et al., 2006; Rashba et al., 1998; Seth et al., 2007).

Contrary to original thought, recent studies on LQTS in the over 40 age group have revealed a continued risk of cardiac events in this age group irrespective of gender (Goldenberg et al., 2008). There are different explanations for why men and women in this age group are at risk that ultimately equal each other out. That is, females have a higher risk than males of LQTS related cardiac events. Males however, have a lower risk of LQTS related cardiac events but higher frequency of developing cardiovascular disease in addition to their LQTS that causes a higher overall frequency of cardiac events than women (Goldenberg et al., 2008). These factors appear to put both genders on equal terms for overall mortality rates due to cardiac events. Females remain at higher risk, and males lower, however, after age 40 years for specifically LQTS related cardiac events (Goldenberg et al., 2008).

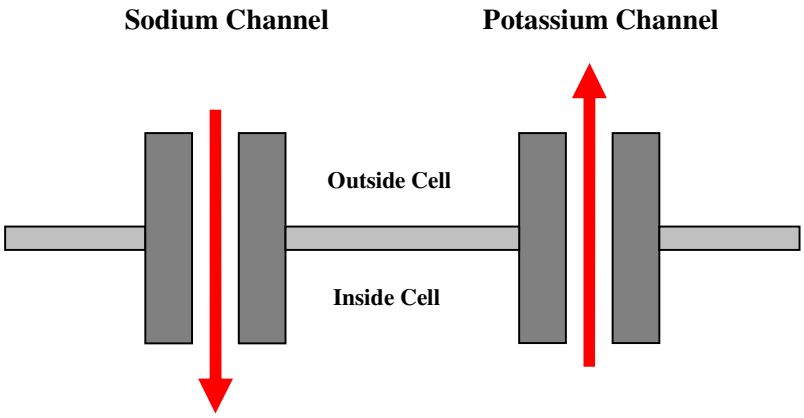
A group at particular risk of a fatal cardiac event are infants under one year of age. It is thought 4 percent of LQTS related fatalities happen in the first year of life (P. J. Schwartz et al., 2006). LQTS accounts for between 3 and 10 percent of Sudden Infant Death Syndrome (SIDS) cases (Arnestad et

al., 2007; Mitchell, 2009; D. W. Wang et al., 2007). This has led to calls for LQTS specific screening of newborns (Arnestad et al., 2007; Van Langen & Wilde, 2006).

Cardiac Electrophysiology

In human bodies all muscle tissue contracts in response to an electrical stimulus or impulse. The heart or cardiac muscle is no different. It is however unique in that while it responds to electrical impulses through its cardiac cells it also has specialised pacemaker cells that can generate electrical impulses (Camm, 2002). Important in the understanding of Long QT Syndrome (LQTS) is that each of the single cells in the cardiac muscle contain ions, such as potassium and sodium, that carry electrical impulses along ion channels in and out of the cardiac muscle cells (see Figure 1). The ion channels regulate the flow of the electrical impulses. Normally, the cells have potassium (K) on the inside and Sodium (Na) on the outside, which is known as polarization. That is, they are relaxed and the cardiac cells are ready to receive an electrical impulse. When a pacemaker cell stimulates a polarized cardiac cell with an electrical impulse most of the potassium moves to the outside of the cell and most of the sodium moves to the inside. This movement of potassium and sodium through the ion channels and cell wall causes a wider electrical impulse that is then conducted to the remaining cardiac cells of the heart. Having transmitted an electrical impulse the cardiac cells contract, which is called *depolarization*. The cardiac cells cannot respond to any further electrical impulses until the potassium has re-entered the cells and the sodium has left, causing *repolarization*. This is also known as the recovery phase, as the muscle has already contracted (depolarization) and is returning to a ready state (polarization) (Atwood, 1996; Camm, 2002).

Figure 1. Flow of potassium and sodium ions inside and outside normal cardiac cell



Cardiac Risk in the Young (2009)

Role of the Sympathetic Nervous System in LQTS

Chemicals which circulate in the blood and which are released by the nerves that regulate the heart can alter the speed of firing in the pacemaker cells and the force of the pumping action of the ventricles. For example, adrenaline in the bloodstream or from nerves in the sympathetic nervous system increases the heart rate and the volume of blood pumped by the heart. The sympathetic nervous system (part of the larger autonomic nervous system) can therefore directly affect cardiac function, such as in times of stress or emergencies. At these times the "fight or flight response" occurs causing the heart rate to become more rapid by increasing the force of cardiac contractions and blood flow and produce a state of physiological readiness to fight or flee. Other sympathetic nerves produce adrenaline consequent to experiencing anger, pain, fright (e.g. from a sudden loud noise), caffeine or medications. Each of these can cause a similar deregulating affect on the cardiac cycle (Atwood, 1996). If however, this rapidity of the cardiac cycle becomes extreme it produces what is called torsade de pointes and ventricular tachycardia. This can cause lowered blood pressure, reduced oxygenation and consequently the fainting (syncope), seizures or sudden cardiac death seen in LQTS and other conditions (P. J. Schwartz, 2006).

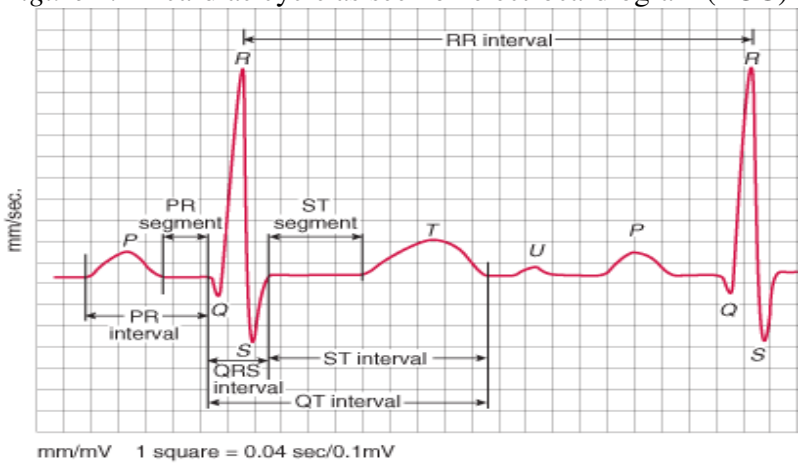
The QT Interval

A single heart beat or conduction of an electrical impulse can be viewed as a series of waves on an electrocardiogram (ECG). This combination of waves in a heart beat usually contains five major waves: P, Q, R, S, and T which together make one cardiac cycle (Conover, 1996). These ECG waveforms illustrate the following electrophysiological activity:

- The P wave represents the depolarization impulse across the atria
- The Q, R and S waves (called the QRS complex) show ventricular depolarization (the downward stroke followed by an upward stroke is the Q wave, the upward stroke is the R wave and any downward stroke preceded by an upward stroke is the S wave)
- The T wave depicts the repolarization of the ventricles

The QT interval is a specific time interval on the ECG (see Figure 2) representing the time an electrical impulse depolarizes (contracts) the heart's pumping chambers (ventricles) to the end of repolarization (the recovery phase). This coincides with the specific ion chemical activity of the potassium and sodium cardiac cells as previously mentioned.

Figure 2. A cardiac cycle as seen on electrocardiogram (ECG)

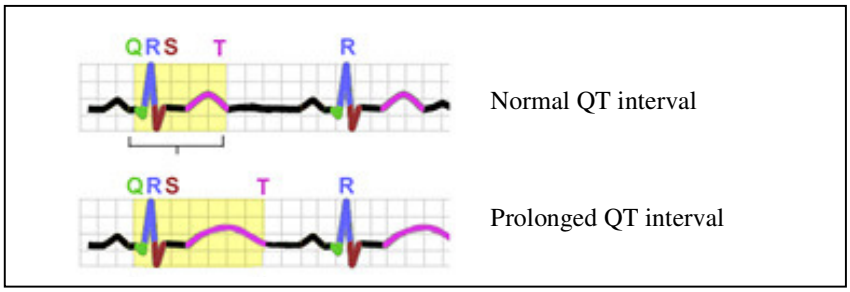


Rhythm Society, 2009)

The length of the normal QT interval is measured in milliseconds and varies with a person's heart rate, shortening as the rate increases and lengthening as the rate decreases. Just as there are inter and intra-personal differences in individual heart rates there are also differences in QT interval lengths.

In Long QT Syndrome, a prolonged QT interval typifies the condition and commonly occurs when a problem arises during the ventricular repolarization phase, usually where the cardiac muscle in the ventricles recharges sluggishly or inefficiently (Moss and Robinson, 2002). This is often best shown by a prolonged QT interval on an ECG, and can be seen in comparison to a normal QT interval in Figure 4.

Figure 3. Normal and prolonged QT interval as shown on electrocardiogram (ECG).



(Heart Rhythm Society, 2009)

Delayed electrical recovery in the cardiac muscle or heart producing a prolonged QT interval on ECG can be associated with very dangerous, rapid and chaotic heart rhythms such as torsades de pointes and ventricular tachycardia (Conover, 1996; Moss and Robinson, 2002; Schwartz, 2006).

When dysfunctional heart rhythms like this happen the heart is unable to pump sufficient blood to the brain. Inadequate blood flow to the brain causes syncope (fainting), seizures and can lead to sudden cardiac death (Schwartz, 2006). Essentially, the longer the QT interval on an ECG, the greater the risk of developing a dangerous heart rhythm and subsequent Long QT Syndrome related symptoms (Moss and Robinson, 2002).

Genes, Inheritance and the Etiology of Long QT Syndrome

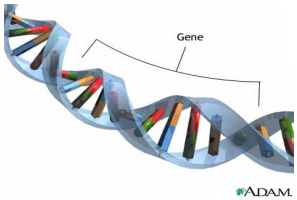
Genetics

The field of genetics and molecular biology is complex but there are some rudimentary aspects that are fundamental to understanding a condition of genetic origin such as LQTS. The human body is made up of trillions of cells which act as the basic building blocks for living. In the middle of each cell is the central nucleus and it is in here that several important elements reside. These include the deoxyribonucleic acid (DNA) coiled up inside a chromosome, the genes that represent individual sections of the DNA strand, and proteins such as amino acid sequences that complete the process of gene expression. These elements together represent the process by which the genotype becomes the phenotype and for LQTS, the process from which LQTS begins with genetic abnormalities and ends with functional ones.

Genes

DNA includes many genes in sections of varying lengths; some contain a few hundred DNA bases while others have more than 2 million (see Figure 4). Everyone inherits a copy of the same gene from each of their parents but the majority of genes are exactly the same in all people. The Human Genome Project estimates each person has between 20,000 to 25,000 genes (Delatycki, 2008) and the differences between people is estimated to be produced by less than one percent of their total genes (Nebert, 2009).

Figure 5. Gene within a strand of DNA.



(Lifespan, 2009)

The differences between individuals are usually referred to as traits and these are determined by genes found in a discrete series. Each gene has a corresponding gene (homologue) on a chromosome (one on each part) and alleles can be found at the same place in the genetic chain on both sides of the chromosome. Alleles may be identical or different, producing the variations we see in inherited characteristics such as eye colour or blood type (Pierce, 2006). If two alleles of a given gene pair are identical, the organism is called a homozygote and is homozygous with respect to that gene. If instead the two alleles are different the organism is a heterozygote and is heterozygous. Genes are said to be dominant or recessive according to the alleles and whether they are hetero or homozygous. For example, if one allele of a pair (within a chromosome) codes for blue eyes while another also codes for blue eyes (a homozygous gene) then the result will be blue eyes. If however one allele codes for blue eyes while the other codes for green eyes (a heterozygous gene) then what happens? Either blue or green eyes will be expressed and whichever one is expressed is considered the dominant gene and the one that is overpowered is considered the recessive gene.

This process can also be used to illustrate the transition between the genotype and phenotype. Generally, the *genotype* refers to the genetic makeup of an organism, either at a single locality or over all its genes collectively. This then directly or indirectly affects its molecular, physical, behavioral, and other traits, which individually or collectively are called the *phenotype* (King, Stansfield, & Mulligan, 2006). So then, it can be said that the two alleles, whether they be homozygous or heterozygous expressing a dominant or recessive allele, interact to produce the phenotype; or what we see when we look into a persons eyes.

The job of most genes is to act as a kind of blueprint and, through a process known as gene expression, to pass on the information they contain according to a set of rules within the genetic code. That is, the genetic code transcribes and then translates genetic material, producing proteins. In molecular genetic terms this process is defined as a mapping of nucleotide sequences called codons (the genes that code for proteins) with the amino acid sequences (the proteins) (Berg, Tymoczko, Stryer, & Clarke, 2002). In genetics this is considered the aforementioned fundamental level at which the *genotype* (made up of these proteins) produces the *phenotype* (made up of genotypic proteins plus environmental influences such as diet and exercise) (Hartl & Jones, 2005).

Genetic Mutations

If however a problem occurs in a gene causing a genetic mutation then this can change the protein's amino acid sequence, destabilizing the structure and function and potentially having a dramatic affect on the cell and on the person as a whole (Berg et al., 2002).

The DNA sequence of a gene can be abnormally altered in a number of ways and these gene mutations have varying effects on a person's health, depending on where they occur and whether they alter the function of essential proteins. Not all gene mutations, however, are pathogenic as some are just natural variations that are completely benign. When there is a consistently recurring mutation that is thought to be associated with a disease but it is not clear whether they are actually pathogenic they are considered unclassified variants. When looking to isolate a pathogenic mutation, however, clinical geneticists look for the rate of penetrance in the population under investigation. Penetrance in genetics is the proportion of individuals carrying a particular variation of an allele that also expresses an associated trait (phenotype). In medical genetics, the penetrance of a disease-causing mutation is the proportion of individuals with the mutation who exhibit clinical symptoms. For example, if a mutation in the gene responsible for a particular autosomal dominant disorder has 95% penetrance, then 95% of those with the mutation will develop the disease, while 5% will not (Pierce, 2006). In some cases the penetrance is low despite being clearly pathogenic.

Genetic Testing

Once inheritance was understood at a molecular level this opened up extensive avenues for research. Over the last four decades some important developments have occurred, including chain-termination DNA sequencing, in 1977, that allowed scientists to read the nucleotide sequence of a DNA molecule (Sanger, Nicklen, & Coulson, 1992). In the early eighties the polymerase chain reaction was developed, providing a quick way to isolate and amplify a specific section of a DNA from a mixture. Then, the Human Genome Project and privately owned Celera Genomics used these and other techniques culminating in the sequencing of the human genome in 2003 (Berg et al., 2002). Genetic testing followed thereafter and has become a quickly developing field offering many benefits such as knowing the relative risk one had for developing a life threatening disease and feeling a sense of control over the future and outcome (Satia, McRitchie, Kupper, & Halbert, 2006). Negative consequences too have been encountered, such as disrupted family relationships and feelings of guilt about having passed the condition on to their children (Duncan et al., 2008) and all

of this can come direct to you via the emerging direct-to-consumer internet based genetic testing market (Bloss et al., 2010).

Genetic testing is used for a variety of reasons such as forensic identity testing, screening embryos prior to implantation, and prenatal screening. For inheritable conditions newborn screening is available, as is pre-symptomatic testing for adult onset inherited disorders. That is, prior to symptom onset predictive testing can be done to predict a usually adult onset disease such as Huntington's Disease (HD), or susceptibility testing can estimate the risk of developing particular adult onset cancers, such as breast cancer, or diseases such as Alzheimer's Disease. Carrier screening is also available in the case of recessively inherited conditions to identify the unaffected individuals who each carry one of the two copies required for the disease to be expressed. Plus, genetic testing can provide certainty and confirm a diagnosis (Lerman, Tercyak, Croyle, & Hamann, 2002; Pierce, 2006).

Over the last two decades, research into the genetic origins of Long QT Syndrome has discovered particular symptom clusters and faults in the heart's function are caused by several localised genetic mutations. These have come to be known as Long QT Syndrome subtypes. Genetic testing is now used to identify these genetic subtypes. This information allows genotype specific treatment planning and the identification of triggers for cardiac events.

Generally, genetic testing in New Zealand is done either by District Health Boards (DHB) or community laboratories. Genetic testing for LQTS began in New Zealand approximately ten years ago (2001) as a research project being conducted through the University of Auckland, Medical and Health Sciences Department laboratory (Shelling, 2011). Four years later (2005), the testing was moved to a DHB hospital based laboratory to be alongside the genetic testing for other conditions (Sarfati, 2002; Shelling, 2011). According to a 2002 report to the National Health Committee (Sarfati, 2002) genetic tests are selected for limited funding by each DHB and the main reasons cited for test selection is clinical demand, interest areas within a given laboratory and level of funding required (Sarfati, 2002). Therefore, tests for less common conditions, such as LQTS, are often referred to overseas laboratories, sometimes at great cost (Sarfati, 2002). This became the case for LQTS in 2008 as funding was restricted. Unfortunately significant delays were encountered (Shelling, 2011).

There are now many genetic tests available in New Zealand (NZ) for common and rare conditions. The demand exceeds the capacity of genetic testing services however (Morgan et al., 2004). At present, any medical practitioner in NZ can order any laboratory test whether or not the test is on The Schedule Test Purchase List (the Schedule) that receives funding through the regional District Health Board (Sarfati, 2002). Also, according to the 2002 report prepared for the National Health Committee (NHC), NZ clinicians do not have easy access to clinical guidelines covering which tests to order, when to refer to local genetic services and what services they can access (Sarfati, 2002). This may prove to be a problem as the quantity and availability of tests has increased over time and will continue to do so. As this happens added pressure will be placed on primary care to respond to the demand for predictive and diagnostic genetic testing (Morgan et al., 2004).

Within the public sector internationally and in New Zealand, a team of clinicians is usually involved when individuals or families undergo genetic testing. Often included is a Clinical Geneticist; a specialist scientist who interprets the genetic material and often evaluates and diagnoses patients', a specialist doctor in the appropriate area, such as a Cardiologist for an inheritable cardiac condition such as LQTS, a Social Worker or Nurse Liaison, to provide social and medical support leading up to and after clinical contacts; and a Genetic Counselor, who helps meet information needs pertaining to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions (Harper, 2004).

Gene Determined Forms of LQTS

Since 1975 LQTS has been identified as having two hereditary variants or phenotypes, known as Romano-Ward Syndrome and Jervell and Lange-Nielson Syndrome (Jervell, Lange-Nielsen, 1957; Schwartz, Periti, Malliani, 1975; Schwartz, 2006). The Romano-Ward Syndrome variant accounts for just over 99% of the total LQTS population (I. Goldenberg et al., 2006). It is mostly inherited in an autosomal dominant fashion with each successive generation being at risk irrespective of which parent has the condition (Vetter, 2007). In the rarer cases of Jervell and Lange-Nielsen Syndrome however, it is inherited via a recessive pathway and is combined with congenital deafness (Goldenberg et al., 2006; Piippo et al., 2000; Schwartz et al., 2006).

Curiosity about the hereditary component of this condition motivated genetic studies that in 1995 famously identified genetic mutations or genotypes associated with potassium and sodium cardiac ion channel genes (Curran et al., 1995; Wang et al., 1995; Zareba et al., 2003). These genotypes

were found to cause distinct forms of LQTS of which twelve dominant Romano Ward Syndrome types are currently known, called LQT1 through to LQT12, plus two recessive forms called Jervell & Lange-Nielsen Syndrome JLN1 and JLN2 (Hendriks et al., 2008; Shimizu, 2008; Tester & Ackerman, 2008).

Since the mid 1990s research has been rapidly expanding what is known about the varying genetic origins of Long QT Syndrome. Despite initially appearing to have a single aetiology, it is now known that LQTS originates from particular mutations in multiply specific genes (Ackerman, 2005; Goldenberg, Bradley, et al., 2010) furthermore these mutations also have differing effects on the affected person's functioning, risks and treatment options (Eddy et al., 2008; Goldenberg & Moss, 2008; Shimizu, 2008). To date, more than 600 mutations in twelve distinct genes have been identified and have now come to represent the different types of LQTS (Eddy et al., 2008; Goldenberg, Bradley, et al., 2010; Napolitano et al., 2005; Tester & Ackerman, 2008).

Long-QT syndrome has been subdivided into genotypes based on the gene in which pathogenic mutations have occurred. The most prevalent forms are LQT1 and LQT2 (due to mutations in potassium channels), and LQT3 (due to a sodium channel mutation) (Crotti, Celano, Dagradi, & Schwartz, 2008). Current estimates suggest LQT1-3 make up 73-90% of the total LQTS genotyped population (Hintsa et al., 2010; Tester & Ackerman, 2008; Wilde & Bezzina, 2005). Of these, 45-50% have LQT1, 40-45% have LQT2, and 5-15% have LQT3 (Crotti et al., 2008; Levine et al., 2008). The specific chromosome and gene affected in each form of LQTS is shown in Table 1, as are the electrophysiological changes and associated additional risks. As can be seen, Sudden Infant Death Syndrome (SIDS) is a serious concern in LQT1-3 and LQT9 (Rhodes et al., 2008).

Table 1. Forms of LQTS and genetic factors involved

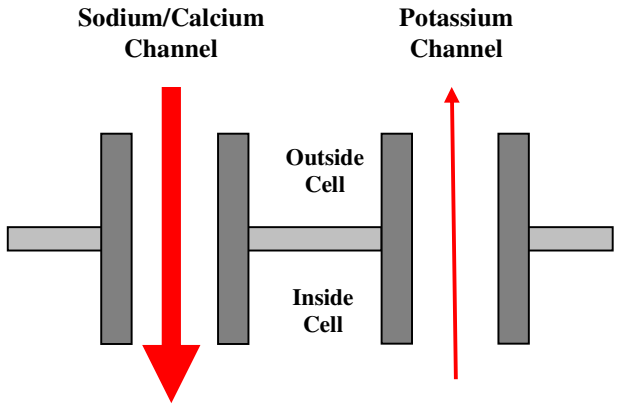
Form of LQTS	Affected Chromo-some	Gene Mutation	Electrophysiological changes in function of the Ion Channel	Additional features/risks
Romano Ward Syndrome: LQT1	**11**	**Potassium channel gene KCNQ1**	**Decrease in the outward potassium current (I_{Ks}) essential for QT shortening during increases in heart rate**	**Increased risk of Sudden Infant Death Syndrome (SIDS) (Rhodes et al., 2008)**
LQT2	**7**	**Potassium channel gene KCNH2**	**Decrease in the outward potassium current (I_{Ks}) essential for QT shortening during increases in heart rate**	**Increased risk of SIDS (Rhodes et al., 2008) and seizure activity (J. Johnson et al., 2009)**
LQT3	**3**	**SCN5A**	**Increase in the inward sodium current (I_{Na})**	**Increased risk of SIDS (P. Schwartz & Crotti, 2007) and gastrointestinal problems (Locke et al., 2006)**
LQT4	**4**	**Ankyrin-B**	**Produces calcium *overload* within a cell (Na-K ATPase, I_{Na-Ca})**	**Sinus bradycardia and paroxysmal atrial fibrillation**
LQT5	**21**	**KCNE1**	**Decrease in the outward Potassium current (I_{Ks})**	**Nil**
LQT6	**21**	**KCNE2**	**Decrease in the outward in the Potassium current (I_{Kr})**	**Nil**
LQT7 Andersen-Tawil Syndrome	**17**	**KCNJ2**	**Decrease in the inward potassium current (I_{Kl})**	**Periodic paralysis and dysmorphic features**
LQT8 Timothy's Syndrome	**12**	**CACNA1C**	**Increase in the L-type inward calcium current (I_{Ca-L})**	**Dysfunction in multiple organ systems including Congenital Heart Disease, Syndactyly, Immune Deficiency and Autism.**
LQT9	**3**	**CAV3**	**Increase in the inward cardiac sodium channel (I_{Na})**	**Increased risk of SIDS (Cronk et al., 2007)**
LQT10	**11**	**SCN4B**	**Increase in the inward cardiac sodium channel (I_{Na})**	**Nil**
LQT11	**7**	**AKAP-9**	**Increase in the outward sodium current (I_{Ks})**	**Nil**
LQT12	**20**	**SNTA1**	**Increase in the inward cardiac sodium channel (I_{Na})**	**Nil**
Jervell & Lange-Nielsen Syndrome: JLN1	**11**	**KCNQ1**	**(I_{Ks})**	**Neurosensory deafness and at 3.5 times more risk of cardiac arrest**
JLN2	**21**	**KCNE1**	**(I_{Ks})**	**Neurosensory deafness and at 3.5 times more risk of cardiac arrest**

Shimizu (2008) unless indicated otherwise

Combined Cardiac Electrophysiology and Genetic LQTS Etiology

It is thought because LQTS only arises during a specific phase of the cardiac cycle (i.e., when the ventricles are repolarizing). This implies that a form of ion channel dysfunction underlies the physiological symptoms of LQTS (Splawski et al., 1997). The commonest forms of LQTS are single gene disorders caused by mutations of the genes that provide recipes for the correct functioning of these cardiac ion channels and so are often referred to as cardiac ion channelopathies (Vincent, 2005). As mentioned earlier the cardiac ion channels and the ions that travel through them (Sodium and Potassium) (see Figure 1) are generally responsible for the correct electrical activity and rhythm of the heartbeat. The figure below (Figure 5) illustrates how the commonest ion, potassium, is usually reduced in LQTS. In the normal heart the potassium flows out of the cell to repolarise the heart and sodium flows into the cell to activate the heart. In a typical LQTS affected heart however, genetic mutations have caused alterations in the protein regulating the flow of an ion. If the potassium channel is affected, the potassium ions are allowed in too slowly. If the sodium cell is affected then too many sodium ions are allowed into the cell. Either way this results in an electrical disturbance in the cells of the heart known as prolonged repolarisation. This is illustrated on an ECG recording as a lengthening of the time period known as the QT interval (see Figure 3). The process by which a defective gene lengthens the QT interval is shown below in Figure 6. From a heartbeat-to-heartbeat perspective this repolarization problem rarely causes difficulties but if triggered by certain stimuli the heart can cause electrical cardiac chaos which is potentially life-threatening.

Figure 5. Flow of potassium and sodium ions in typical LQTS cardiac cell



(Cardiac Risk in the Young, 2009)

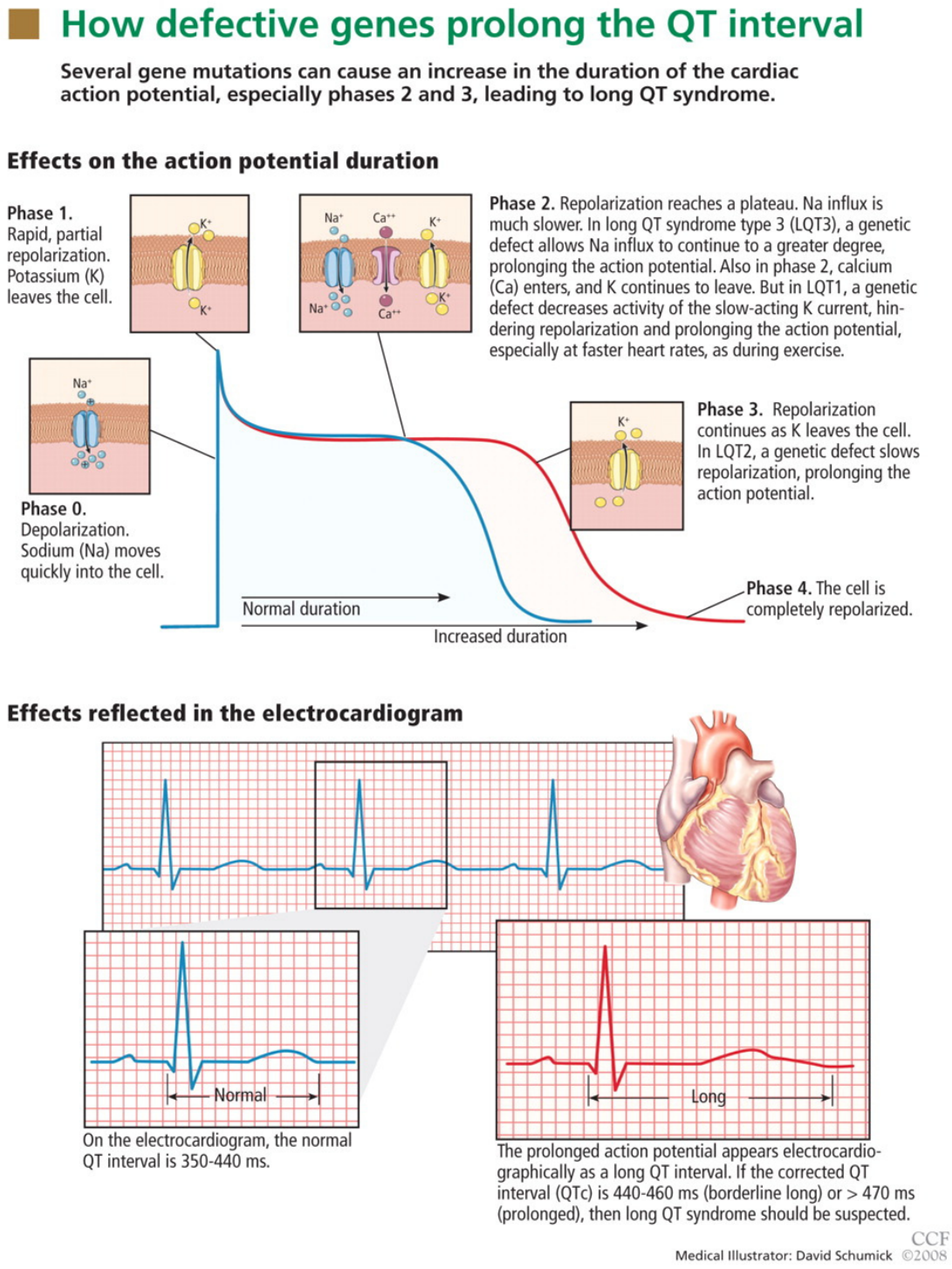
In a smaller number of cases the flow of sodium may be increased (LQTS10-12) and in rare forms of LQTS (Andersen-Tawil Syndrome/LQTS7) potassium and calcium ions are involved (Fodstad et al., 2004) (see Table 1). The ion channels have been identified in approximately 7 out of every 10 individuals with LQTS, so there is still more to learn about the channelopathies of LQTS (Vetter 2007).

The instructions for making the potassium channels come from a family of genes known as the KCN family. These potassium channels move the potassium in and out of cells that have critical roles in the body including for the nervous system, inner ear, the heart and the muscles. Mutations in KCN genes can present themselves as episodic ataxia (periods of poor coordination and balance), rare forms of epilepsy, hearing loss, cardiac arrhythmias (abnormal rhythms) and episodic paralysis (Gutman et al., 2005).

The SCN family is responsible for making the sodium channels that move sodium ions into cells. Sodium channels are found in the heart and skeletal muscles, and are essential for the normal function of the nervous system. Mutations in the SCN family of genes can cause arrhythmias, muscle weakness and episodic paralysis, several types of epilepsy and pain disorders.

The most common forms of LQTS, accounting for approximately 90% of the LQTS population (Hintsa et al., 2010), arise from these two gene families which represent the potassium and sodium ion channelopathies (see Table 1).

Figure 6. How defective genes prolong the QT interval



(Levine et al., 2008)

Currently, it is estimated that only 50-70% of the gene mutations that cause LQTS have been identified. Others are thought to be either unidentified or their location on identified genes is still to be found (Shimizu, 2008; Vetter, 2007). At present, 20% of patients with a definite diagnosis of LQTS are not positively genotyped (Schwartz, 2006; Tester & Ackerman, 2008). The most recent novel mutation discoveries were achieved through a state of the art, more complex genetic testing process and have been associated with the two aforementioned gene families and three main LQTS susceptibility genes (Tester & Ackerman, 2008). Researchers generally agree discovering a new LQTS gene is unlikely, believing instead that as technology advances so will detection of otherwise novel mutations and unclassified variants (Tester & Ackerman, 2008). The most recent New Zealand (NZ) LQTS genetic study returned a high rate of novel mutations (40%), suggesting NZ may have a unique genetic footprint for LQTS (Chung et al., 2007).

Medical Presentation & Clinical Management of Long QT Syndrome

Symptoms

Initial presentation of LQTS most commonly includes fainting (syncope), seizures or sudden cardiac death (secondary to ventricular arrhythmia) during or after physical or emotional stress, or sudden loud noises that activate the sympathetic nervous system (Cardiac Society of Australia and New Zealand, 2009; (Bloise, Napolitano, & Priori, 2002; Skinner & Group, 2007). For those who faint, the torsade de pointes rhythm that characterizes QT prolongation usually spontaneously returns to normal in less than a minute. Consciousness is commonly regained quickly, without disorientation or confusion. Only some individuals experience fatigue afterward. If the torsade de pointes rhythm persists longer, however, individuals can experience a generalized seizure. In both presentations the heart eventually reverts back to normal sinus rhythm. In a minority of patients however, the torsade de pointes rhythm persists longer still degenerating further into ventricular fibrillation, which rarely reverts back to a normal rhythm without medical intervention (electrical defibrillation). The outcome is sudden cardiac death or sudden cardiac arrest. Sudden cardiac death occurs as the first presenting symptom in 10% of children and syncope or seizures in 30-40% (Vetter, 2007).

Not all those diagnosed with LQTS experience symptoms however. In fact, approximately 50% of those with a genetically based diagnosis of LQTS *never* develop symptoms (Meulenkamp et al., 2008; Priori et al., 2003). Unfortunately, it is not yet possible to ascertain in which LQTS patients

this will occur, nor is it possible to be assured they will continue to have no symptoms so a risk remains present, especially given that a first cardiac event could be fatal (Meulenkamp et al., 2008).

Diagnosis

A diagnosis of LQTS is made based on clinical and family history, plus specific tests, including electrocardiogram (ECG), exercise induced 'provocation tests' to induce and monitor potential cardiac stress and 24 hour (Holter) recordings of the heart's activity (Schwartz, 2006; Skinner & Group, 2007). Initially, QTc prolongation on ECG was the cornerstone of diagnosis, but in the 1980s Schwartz (2006) observed that some patients were affected by LQTS despite having a normal QTc interval. This was later proven by the existence of mutation carriers with a normal QTc interval, as a consequence of low penetrance (Schwartz, 2005a). This had important practical and medico-legal implications because it was no longer possible to state that a sibling of an affected patient with a normal QTc is definitely not affected by LQTS (Crotti et al., 2008). Predictive genetic testing in Asymptomatic, normal QTc family members however, has now emerged to fill this role through the identification of mutation carriers (Goldenberg & Moss, 2008).

Cardiologists now commonly use either the Keating criteria or the Schwartz criteria to aid diagnosis (Hofman et al., 2007), with the Schwartz criteria recommended for use in Australia and New Zealand (Skinner & Group, 2007). The most recent Schwartz criteria (see Table 2) (Schwartz et al., 1993) uses patient histories and test outcome criteria that together produces a probability of LQTS.

Table 2. Diagnostic Criteria for LQTS (Schwartz, 1993)

		Points
ECG findings		
	QTc ≥ 480 ms	**3**
	QTc ≥ 460-479 ms	**2**
	QTc ≥450-459 ms (in males)	**1**
	Torsades de pointes	**2**
	T wave alternans	**1**
	Notched T wave in three leads	
Clinical history		
	Syncope with stress	**2**
	Syncope without stress	**1**
	Congenital deafness	**0.5**
Family history		
	Family members with definite LQTS	**1**
	Unexplained sudden cardiac death below age 30 among immediate family members	**0.5**

Scoring: ≥1 indicates low probability of LQTS; 2-3 points immediate probability of LQTS; ≥4 points indicates high probability of LQTS (Schwartz, 1993).

Physicians reported that these criteria are not always easy to interpret, however, especially if the probability is 'immediate' or reliable patient histories are difficult to obtain (Vetter, 2007). Doubt has also surfaced about the sensitivity of the Schwartz criteria in correctly diagnosing LQTS. Making full use of the recent advances in research into the genetic basis of LQTS Hofman and colleagues (2007) used genetic test results to scrutinise the Schwartz criteria. The results indicated the Schwartz criteria alone were not sensitive enough for clinical use and instead provided evidence that a new QT interval cut-off on ECG (QTc ≥ 430 ms) was superior in diagnosing LQTS correctly. Schwartz (2006) asserted reliable diagnosis is best when aided by the identification of a known LQTS genetic mutation. Indeed, genetic testing has become the current preferred and definitive diagnostic tool when used in conjunction with clinical and family history, and clinical findings (Hamang, Solberg, Bjorvatn, Greve, & Oyen, 2009; Hofman et al., 2007; Skinner & Group, 2007).

Genetic testing has consequently become increasingly used as a routine part of LQTS diagnosis (Crotti et al., 2008; Goldenberg, Moss, et al., 2008). Each of the twelve LQTS genes identified and associated with different phenotypes (Goldenberg, Gillespie, et al., 2010; Tester & Ackerman, 2008) present a different clinical picture, optimal management, possible responsiveness to treatment and relative risk of sudden death so it is important to assess these aspects during the diagnostic phase

(Chung et al., 2007; MacCormick et al., 2009) . Recent studies have found genetic testing uncovers the mutations and confirms the diagnosis in 70% of individuals who are the first to present for diagnostic assessment of LQTS (probands) and can identify all carriers among the relatives (cascades) of those who have tested positive (Hofman et al., 2007). Also, as nearly one third of LQTS gene carriers have ECG readings that do not indicate LQTS (QTc < 460 ms) genetic testing is important in order to identify those at risk of adverse events so preventative treatment can be put in place (Ching & Tan, 2006; Goldenberg, Bradley, et al., 2010; Goldenberg, & Moss, 2008).

Misdiagnosis

As can be seen, the diagnosis of LQTS is complex and difficult to achieve with confidence without the addition of genetic testing. Genetic testing, however, is often not available to clinicians, especially if they are rurally based. It is perhaps no surprise then that the QTc interval and syncope are often misinterpreted, especially if the QTc interval and other symptoms are unclear (Taggart, Haglund, Tester, Ackerman, 2007). Among the most common misdiagnoses are hyperventilation, epilepsy/seizure disorders, sleep dissociation state and blood pressure related vasovagal related falls (Burghaus, Liu, Eggers, Muller-Ehmsen, & Fink, 2010; Lazzara, 2008; MacCormick et al., 2009).

As previously discussed, LQTS related fainting can lead to hypoxia and generalized seizures. These seizures may appear to be the primary presenting symptom, and without further investigations or clearly contradicting clinical data (e.g., an obviously prolonged QTc interval) have been routinely misdiagnosed as seizure disorders (Ackerman, 2005; Burghaus et al., 2010; Chuang, Chuang, & Ueng, 2009; Johnson et al., 2009; Levine et al., 2008). Recent research suggests that a diagnosis of epilepsy in LQTS cases is the most common misdiagnosis in New Zealand (MacCormick et al., 2009). This misdiagnosis, coupled with treatment protocols including antiepileptic medications, has been found to correspond with an increased risk of sudden cardiac death and a delay between initial misdiagnosis and the eventual appropriate LQTS diagnosis and treatment, sometimes for up to several decades (Burghaus et al., 2010; Johnson et al., 2009; MacCormick et al., 2009).

Recent research, however, postulates that particular forms of LQTS may have a higher concordance of symptom crossover with seizure disorders (Johnson et al., 2009). In one study (Johnson et al., 2009), LQTS type 2 (LQT2) was isolated out as having the highest proclivity for seizure activity when compared to the other subtypes of LQTS. Evidence suggested a possible crossover effect of the genetic mutation responsible for LQT2 and seizure disorders, particularly temporal lobe epilepsy.

This potentially gives further credibility to the use of genetic testing as part of the diagnostic process when LQTS and perhaps also seizure disorders are being considered (Burghaus et al., 2010; Johnson et al., 2009; MacCormick et al., 2009).

Treatment

Studies have demonstrated the mortality rate in LQTS patients who remain untreated is 20% within the first year of diagnosis and 1% to 2% per year thereafter (Ching & Tan, 2006; Chuang et al., 2009). The therapies currently available however are very effective in preventing sudden cardiac death (Levine et al., 2008; Vetter, 2007). Treatment strategies for the prevention of LQTS related cardiac events include beta-blockers, implantable cardioverter defibrillators, and, more recently, left cardiac sympathetic denervation (LCSD) surgery (Atallah et al., 2008; Collura, Johnson, Moir, & Ackerman, 2009; Li et al., 2009; Wang, 2003). These treatment strategies are also utilised from genotype specific perspectives.

Pharmacotherapy

The current first line therapy of choice for LQTS is beta-adrenergic blockers (Beta-blockers) (Goldenberg, Zareba, & Moss, 2008; Vincent et al., 2009; Zareba & Cygankiewicz, 2008), unless there are contraindications (Crotti et al., 2008). These are taken orally and usually on a daily basis (Levine et al., 2008). Beta-blockers place themselves on the beta-adrenergic receptors, which are stimulated by adrenaline. The Beta-blocker "blocks" the receptors from being stimulated and when these are blocked in the heart the force and rate of the heart beat is reduced, helping to restore a regular rhythm. Beta-blockers are an appropriate treatment for Symptomatic and Asymptomatic patients and those who have never had a cardiac arrest (Vincent et al., 2009). Research indicates Beta-blockers generally improve clinical symptoms and reduce the incidence of sudden cardiac death in approximately 70% of patients (L. Wang & Feng, 2004).

A recent study asserts, however, that for those that survive a LQTS related cardiac arrest in the first year of life Beta-blockers are ineffective in preventing fatal or near-fatal cardiac events across the first ten years of life (Spazzolini et al., 2009). There is also an as yet undefined group of up to 30% of patients that do not respond to Beta-blockers and remain Symptomatic, with up to 10% still experiencing cardiac arrest or sudden cardiac death (Goldenberg, Moss, et al., 2008; Wang, 2003; Zareba & Cygankiewicz, 2008). Some believe, however, all life-threatening beta-blocker "failures"

are either due to medication non-compliance (i.e., not taking the medication at all or when they should) or using other QT-prolonging medications (Vincent et al., 2009).

Surgery

Implantation of an implantable cardioverter defibrillator (ICD) in the chest is increasingly used in high-risk LQTS patients and in those who remain Symptomatic despite the use of Beta-blockers (Goldenberg, Moss, et al., 2008; Goldenberg, Zareba, et al., 2008; Zareba & Cygankiewicz, 2008). The device is a small battery powered electrical impulse generator that is programmed to monitor the rate and rhythm of the heart and deliver an electrical shock if an irregular rhythm is detected.

The decision as to whether an ICD should be used is usually also based on the individual patient characteristics and risk factors such as the aforementioned age, sex, clinical history, ECG outcomes (including 24-hour Holter recordings), and genetic subgroup including mutation-specific features in some cases (Crotti et al., 2008). It does not appear justified in Asymptomatic patients or those who have never had a cardiac arrest or beta-blocker therapy (Vincent et al., 2009).

If an individual experiences multiple arrests and consequent shocks in clusters a left cardiac sympathetic denervation (LCSD) may be an appropriate alternative (Schwartz et al., 2004). There are some differences in opinion as to when this surgery is appropriate, however, with some physicians advocating immediate LCSD surgery if a patient is unresponsive to beta-blockers alone (Atallah et al., 2008; Crotti et al., 2008; Li et al., 2005; Wang, 2003). The surgery includes removing a portion of the nerves to the heart and consequently reducing the control the abnormally functioning sympathetic nervous system has over breathing and heart rate. LCSD appears effective in reducing the frequency of fainting and aborted cardiac arrest in high risk LQTS patients (Wang, 2003) but evidence suggests it is ineffective at reducing sudden cardiac death in the long term and it has limited popularity with cardiologists (Schwartz et al., 2004).

Management and Functional Restrictions

In addition to these specific treatments for LQTS, there are general management considerations. Irrespective of severity or symptomatology it is crucially important to avoid any medications or recreational drugs that may prolong the QT interval by causing torsade de pointes or lowering potassium levels (Skinner & Group, 2007). A constantly updated list of medications to avoid and

their gender related risk is available through www.azcert.org/medical-pros/drug-lists/CLQTS and must be considered by the patient's physicians (i.e., GP or Psychiatrist).

In the LQTS population, it is important to observe specific functional restrictions such as limiting strenuous exercise and competitive sports, especially if previous cardiac events (including fainting) have occurred either during or after exercise (Skinner & Group, 2007). Swimming is a specific form of activity associated with increased risk and also to be avoided (Schwartz, Priori, & Spazzolini, 2001). Consideration of previous triggers can prove to be very important. Possible risks include emotional distress and sudden loud noises (especially during sleep) (Kapetanopoulos et al., 2006; Levine et al., 2008; Skinner & Group, 2007). It is also important to avoid dehydration, missing meals and inadequate nutrition as these can lower potassium levels and precipitate cardiac events (Farnsworth, Fosyth, Haglund, & Ackerman, 2006). Once genetic testing has been completed the individual can be informed of the risks specific to their genotype, enabling a more accurate management plan (Levine et al., 2008; Schwartz, 2005b; Skinner & Group, 2007).

Genotype Specific Treatment and Risk Identification

The major advantage of discovering the function, type and location of the ion-channel pathogenic mutations in LQTS is the ability to tie the genotypes to their phenotypic triggers and consequently reduce risk. Directed genetic testing can confirm a diagnosis of LQTS, delineate prognosis and identify family members at high risk and allow genotype specific therapies that may reduce life threatening cardiac events (Goldenberg & Moss, 2008; Tzou & Gerstenfeld, 2009).

As LQTS subtypes LQT1-3 constitute the majority (over 90%) of LQTS cases the bulk of genotype specific therapeutic knowledge concentrates on these genotypes (Wirsching, 2010). Little appears known for the other forms of LQTS other than that Beta Blocker therapy is useful in LQT4-8, LQT12 and unknown LQTS genotypes (Shimizu, 2008). A sodium channel blocker similar to that effective for LQT3 may also be theoretically effective in LQT9-10 and LQT12 genotypes (Shimizu, 2008).

Consistent with an impairment on the potassium channelopathy, most of the cardiac events of LQT1 patients occur during strenuous exercise and physical stress, particularly swimming (Shimizu, 2008). The most recent study in NZ found a higher rate of LQT1 (60%) in the NZ sample than in the

international rate indicated by researchers (42%) (Chung et al., 2007). It was suggested the higher rate in NZ may be explained when looking at the interaction between genotype and phenotype. That is, LQT1 is commonly triggered by physical exertion and NZ is a relatively sporting nation, especially water sports (Chung et al., 2007) and especially in males (Shimizu, 2008). The treatment of choice for LQT1 is beta blockade, which is generally extremely effective; reducing risk of sudden death by 50-75% (Chung et al., 2007; Goldenberg, Bradley, et al., 2010; Skinner & Group, 2007; Skinner & Lever, 2008). Eighty percent of LQT1 patients with a history of fainting do not faint again once on Beta Blockers, and mortality reduces to under five percent (Skinner & Group, 2007). An ICD may also be indicated for LQT1 and LQT2 if cardiac events or fainting continue to occur while taking the Beta Blockers (Shimizu, 2008; Vincent et al., 2009). An important part of treatment for all of the LQTS genotypes is the removal of potential triggers and for all of them that means avoiding medications that prolong the QT interval (Goldenberg, Bradley, et al., 2010) and in LQT1 that means limiting exercise and sports. Swimming and diving are completely contraindicated (Skinner & Group, 2007). Patients with LQT1 are recommended *not* to become athletes or to compete in sports (Skinner & Group, 2007).

In LQT2 events generally occur during emotional stress, often if startled by auditory stimuli and especially if it occurs during rest (Skinner & Group, 2007). This is consistent with the genotypic involvement of the sympathetic nervous system (Crotti et al., 2008). A higher risk exists for LQT2 patients in the post partum period, although it is an increased risk for all with LQTS (Levine et al., 2008; Sauer, 2007). Removing LQT2 related triggers includes taking away loud alarm clocks and turning the telephone volume down at night (Crotti et al., 2008). For LQT2 mothers in the post partum period it is recommended partners feed infants during the night to decrease sleep interruption (Crotti et al., 2008). Beta Blocker medication is an effective treatment for LQT2, reducing the risk of sudden death by 50% (Shimizu, 2008). Pacemaker therapy may be beneficial to regulation in LQT2 (Shimizu, 2008).

LQT3 patients are at higher risk when their heart rates slow, therefore they are at highest risk during sleep (Priori et al., 2003). There are inconsistent reports regarding the effectiveness of Beta Blocker treatment in LQT3. Some contend LQT3 patients do not respond, including some reports of adverse effects, to Beta Blocker therapy and benefit more from an ICD (Welde, 2002). Others however contend there are some who benefit (Ahrens-Nicklas, Clancy, & Christini, 2009). Current NZ guidelines do not support use of Beta Blockers in LQT3. There is some evidence to support

pacemaker therapy to reduce the risk of bradycardia (abnormally slow heart beat that can cause cardiac arrest) (Levine et al., 2008). There is also evidence, based on medical trials only, some medications that inhibit the late sodium current may be beneficial once fully evaluated (Levine et al., 2008). Left Cervical Sympathectomy may be considered if an ICD cannot be placed or cardiac events continue to occur with an ICD in place (Skinner & Group, 2007).

The largest cohort of LQT1-3 patient data revealed risk factors relating genotype to gender and age (Locati, Zareba, & Moss, 1998). That is, those affected by LQT1 and LQT2 have more cardiac events over time than LQT3, but the cardiac events LQT3 patients experience are more likely to be fatal (Locati et al., 1998). Over 50% of LQT1 and LQT2 have had a cardiac event before the age of 40 with the figure for LQT3 under 30% (Shimizu, 2008). Males, especially those with LQT1, are more likely to have a cardiac event at a younger age than females, with 90% of LQT1 males having their first cardiac event before 15 years (Locati et al., 1998; Shimizu, 2008).

As is evident, LQTS is a uniquely complex inheritable condition that spans both medical and genetic fields in nearly all respects. The diagnostic processes are twofold, as are treatment specificities, and genetic testing represents a crucially important part in the medical care of LQTS.

Psychosocial Factors Impacting Health Conditions

Research tells us individuals affected by a condition with a long term or chronic course such as LQTS, especially those who are Symptomatic, have multiple challenges to negotiate (Martire, Lustig, Schulz, Miller, & Helgeson, 2005). Specific factors have been identified that mediate an individual's ability to adjust to meet these challenges. The most salient include the demands of the acute and chronic stages of illness, the coping strategies individuals have available, the effect a condition has on their quality of life, and whether specific psychological difficulties develop. Also, the impact on children as a unique group of the population are reviewed, along with the effect on their families and how support groups in particular have been found to have beneficial effects.

Adjustment Across Time and Stages of Illness

In conditions such as LQTS two stages of adjustment are commonly described in the literature: the acute state of crisis immediately after a diagnosis is made and the longer term adjustments required in response to intermittent condition related changes. The acute stage can create a sense of physical,

social and psychological disequilibrium and, if existing coping strategies are ineffective, can lead to poorer health. Exaggerated negative illness perceptions can lead to anxiety and depression. Once this stage passes individuals get a sense of the long term difficulties they will be required to cope with but, as in the acute phase, whether they adjust is somewhat dependent on the coping strategies they have available to manage the stressors (Drossman et al., 2000; Epker & Gatchel, 2000).

Coping Strategies

Research frequently divides coping approaches into problem focused and emotion focused coping, with each having implications for psychological and health outcomes (Folkman & Moskowitz, 2004). Problem focused coping describes efforts to do something in order to solve or manage the problem, and includes strategies such as gathering information, planning and decision making. Emotion focused coping involves efforts to regulate emotions, which can include seeking emotional support, avoiding emotion by distracting oneself with activities or using alcohol or drugs (Folkman & Moskowitz, 2004; Thomsen, Rydahl-Hansen, & Wagner, 2010). In adults these methods are often used successfully in unison, but are less successful on their own. Emotion focused coping has been found to help manage the initial emotional distress that might otherwise overwhelm a problem focused person during the acute phase but is ineffective in the long term, leading to increased psychological distress (Folkman & Moskowitz, 2004). Problem focused coping however, has been found to be the most effective in coping with long term difficulties, but only when something constructive is able to be done. Unfortunately, health problems can lead to more emotion focused coping because direct action is often not possible and the emotions themselves must then be managed (Penley, Tomaka, & Wiebe, 2002). In all, flexibility with and the appropriate use of both problem and emotion focused coping to meet demands leads to the most desireable psychological and health outcomes (Cheng, Hui, & Lam, 2002; Folkman & Moskowitz, 2004).

Quality of Life

Quality of life refers to the extent the condition or treatment of it interferes with daily living across four domains: physical functioning, psychological status, social functioning and condition or treatment related symptomatology. Satisfaction in each of these domains relates to an increase in quality of life that has been associated with positive health outcomes (Kahn & Juster, 2002). Conversely, when experiences in one or more of these domains, and therefore quality of life, are compromised by a condition such as LQTS this can lead to an exacerbation of symptoms, increased

risks for mortality and increased distress, which in turn increases the incidence of anxiety and depression (De Graaf & Bijl, 2002; Mittermaier et al., 2004).

Psychological Difficulties

Anxiety alone is commonly linked in the health psychology research to negative health outcomes if it is persistent (Hendriks et al., 2008). Specific times when anxiety is known to increase to distressing levels includes when waiting for a diagnosis or surgery, receiving the diagnosis, when lifestyle changes are required and by the prospect of death. Changes in health such as cardiac events or increases in heart palpitations can also increase anxiety, with all of these being linked to poorer health outcomes and coping (Rabin, Ward, Leventhal, & Schmitz, 2001). Depression is also common among those with a chronic condition and especially common in those with a cardiac condition. It often occurs later than anxiety, which tends to peak in the early or acute stage after diagnosis, and can be intermittent throughout the course of a condition (Egede, 2005). Depression also has negative health and quality of life outcomes (Anstey & Luszcz, 2002).

Effect on the Family

When an individual is affected by a condition with long term implications it also disrupts their social systems, and most frequently is their family. Illness can mean increased dependency on the part of the affected one, or ones, and, consequently, added responsibilities for others. This can cause roles to change to meet the new health and emotional needs of those affected (Pavalko & Woodbury, 2000). For the carers, this can lead to fatigue from the weight of responsibility, a sense of burden and distress about their loved one's condition (Williams et al., 2002). It can be particularly hard when the individual affected is a child as they struggle to understand the condition, treatment and restrictions placed on them. Children who do not adjust experience a variety of behavioural problems, from acting out to withdrawing (Alati et al., 2005). Family support for adults and children is especially important however, and it has been linked to improved physical and emotional functioning and treatment adherence (Martire et al., 2005).

Social Support Groups

When family supports are not available or unable to meet individuals' needs social support groups can play a valuable role. Support groups, such as those set up for other conditions such as Diabetes and Asthma, have been found to help normalize and validate common emotional reactions to conditions and in this way are beneficial in respect to many conditions (Griffiths, Calear, & Banfield,

2009; Lepore, Helgeson, Eton, & Schulz, 2003). Evaluations on the effectiveness of internet based support groups suggest positive outcomes but the quality of the research has been questioned, throwing doubt on the findings (Griffiths et al., 2009).

Psychosocial Factors Impacting Long QT Syndrome

There is limited research available on the psychosocial factors that impact those with LQTS and no research has been conducted on the psychosocial impact of LQTS, including genetic testing, on a New Zealand population. The available psychosocial research on LQTS to date has emerged from the United States of America (Farnsworth et al., 2006), Canada (Giuffre et al., 2008), the Netherlands (Hendriks, Grosfeld, van Tintelen, et al., 2005; Hendriks, Grosfeld, Wilde, et al., 2005; Hendriks et al., 2008; Meulenkamp et al., 2008), Finland (Hintsa et al., 2009) and Norway (Andersen, Oyen, Bjorvatn, & Gjengedal, 2008), all geographically, and to some extent culturally, distant from New Zealand. The recent LQTS research examining the psychosocial impact of having LQTS has largely focused on parent and child perspectives. The following section presents the salient findings from this research.

Impact of LQTS on Psychological Distress

Anxiety and Worry

Adults are generally focused on, and concerned by, the health and welfare of their children, grandchildren, or future generations, especially about whether either themselves or their children will die (Andersen et al., 2008; Grosfeld, Lips, Beemer, & Ten Kroode, 2000; Hendriks, Grosfeld, Wilde, et al., 2005). Parents of those with LQTS have been found to experience significantly higher anxiety than parents with other chronic conditions (Giuffre et al., 2008), they are also more likely to experience disease related anxiety if they are of low socioeconomic status or were not satisfied with the information they were provided (Hendriks, Grosfeld, van Tintelen, et al., 2005). It appears however, that parents are often not concerned about their own death. For some, this can be because their fears are focused on their children and grandchildren. For others having experienced resuscitated cardiac events assuages their fear of death. That is, knowing death as a result of a cardiac event would be quick and relatively painless produces less to fear. This form of coping with an increased possibility of dying has been noted not only in parents but in other adults as well (Andersen et al., 2008).

Parental anxiety about their children can become exacerbated when their child gets sick. This is because a sick child with LQTS requires additional monitoring and there is a higher risk of potentially triggering a serious cardiac event. For example, dehydration, fever and medications that prolong the QT interval are all potential triggers and commonly occur with illnesses (Farnsworth et al., 2006). Some parents attempt to reduce their anxiety by using strategies designed to keep them in closer contact with their children when they are separated from them, including providing their children with cell phones and using baby monitors in their child's bedroom to monitor breathing at night, and taking portable defibrillators to every event in which their child participates (Farnsworth et al., 2006). Educating their communities about LQTS, such as community groups, schools and local medical clinics, decreases parental fear their children might go unaided should they have an event in their absence (Farnsworth et al., 2006). Teaching their children to monitor their health and take actions consistent with good physical care, including taking medication and avoiding triggers where necessary, allowed parents and children to become more independent (Farnsworth et al., 2006).

Parents also worry for their children's future, in terms of their career, dating and marriage, and potential stigmatisation. Puberty is a particularly concerning developmental challenge to manage for parents, including the effect this could have on their ability to manage their children's demands for autonomy and treatment adherence (Hendriks, Grosfeld, van Tintelen, et al., 2005). Parents also reported trying to balance reducing cardiac risk with protecting quality of life and keeping life as 'normal' as possible. Even in the case of the Asymptomatic child, parents worry about the continuous threat that their children will develop the disease and, consequently, parents can become vigilant for symptoms.

Findings suggest that children affected by LQTS may experience less generalised fears than other chronic conditions with potential life threatening outcomes, such as asthma, with overall illness related anxiety rates between them being the same (Giuffre et al., 2008). Other findings suggest the opposite, indicating children cope quite effectively with their condition with little effect on their lives (Smets et al., 2008).

Depression

The one study identified which examined depressive symptoms in LQTS adults included findings comparing Symptomatic and Asymptomatic LQTS patients. This is the only such study where

Symptomatic and Asymptomatic outcomes have been compared. Those who were gene positive for LQTS and Symptomatic had depressive symptoms in the clinically significant range while Asymptomatic LQTS gene positive participants did not. Further, no difference has been found in clinically significant depressive symptoms between Asymptomatic LQTS gene positive and gene negative adults (Hintsa et al., 2009). This suggests the presence of cardiac events plus a LQTS gene positive diagnosis in adults is correlated with clinically significant depressive symptoms. There are currently no studies relating to depression rates in children, youth or parents.

Impact of LQTS Symptomatology

Findings indicate significant anxiety can arise in response to cardiac events, particularly in young adults. The cardiac events can lead to some people having difficulty getting to sleep as a result of replaying the event over in their minds at night (Andersen et al., 2008). For parents, the uncertain occurrence and severity of LQTS cardiac events in themselves, or their children and relatives, are also highly distressing (Andersen et al., 2008; Farnsworth et al., 2006). This is particularly so leading up to a gene based diagnosis. Once more knowledge is obtained and treatment has stabilised however, the uncertainty they felt for themselves and others is reduced (Andersen et al., 2008; Farnsworth et al., 2006). Some parents however, are unable to tolerate the distress and so opt for prophylactic surgery (e.g. ICD implantation) to decrease the risk of a random severe cardiac event leading to death, thereby also mitigating their fear for their child (Farnsworth et al., 2006).

Conversely, it has been found that parents who have never experienced symptoms themselves, or seen them in their children often, do not experience anxiety. Generally, they feel safe without having experienced cardiac events and consequently do not feel the same level of fear of dying, for themselves or for their children (Farnsworth et al., 2006).

Impact of LQTS Treatment and Management of Risk

Impact of General Treatment

Treatment can bring relief for some parents whose children are LQTS positive. Other parents, conversely, experience the treatment of their children as very burdensome (Hendriks, Grosfeld, van Tintelen, et al., 2005). Making the decision about whether to treat medically, and which treatments to choose, can be especially difficult for parents. Perhaps this is because the decision can have a direct impact on their child's quality of life, especially if they experience significant side effects after deciding to take Beta Blockers for example (Farnsworth et al., 2006). In addition, adults and parents

can feel a loss of control and subsequent increased anxiety if they experience difficulties differentiating symptoms of the condition from the side effects of the treatment (Andersen et al., 2008).

Impact of Implantable Devices

ICDs are provided to those who have already experienced a life threatening cardiac event and survived, had break through cardiac events while on medication, cannot tolerate the Beta Blockers or have LQT3 (Farnsworth et al., 2006). Already then, they have experienced potentially anxiety provoking events after which they are required to brave surgery and adapt to life with a device visible under the skin in their chest and ready to shock them at literally any time. ICDs have been found to cause distress, even though they are seen as potentially life saving devices. The shocks are feared and many report anxiety around uncertainties such as the reliability of the device to function effectively or that it might go off at an embarrassing or especially risky time (Andersen et al., 2008). Also, having experienced a cardiac event makes the threat of dying salient which may be why those who have just had an ICD implanted and have experienced a cardiac event in the past show high levels of uncertainty around their symptomatology when compared with those who have not had a past cardiac event (Carroll & Arthur, 2010). In a recent review of the literature findings indicated patients with ICD's experienced psychological distress, as measured by anxiety and depression, and consequently poor quality of life (Tagney, 2010). Discharges from the device were also associated with poorer quality of life and young adults were at highest risk for psychological distress and poor quality of life after implantation (Tagney, 2010).

Impact of Functional Restrictions

Clinicians believe it is important to help families balance the impact of LQTS, including working with schools and coaches to find ways to minimise restrictions by finding safe alternatives that don't preclude them from normal peer interactions. Not all activities need to be stopped (Vetter, 2007). While this message may be given so are the messages about the potential impact triggering a cardiac event may have and which activities should be avoided to reduce risk. Often, this includes being asked, or deciding, to give up activities from which the individual derives self esteem and enjoyment (Vetter, 2007). For example, the loss of a driving licence after a cardiac event can restrict independence that is very important to a teenager. Some people try to minimise their emotional expressiveness (including joy) to avoid becoming emotionally agitated and potentially triggering a

cardiac event (Andersen et al., 2008). Others avoid activities because engaging in them may overly concern relatives or partners (Andersen et al., 2008).

Many are also adversely affected by periodic symptoms of the LQTS, such as palpitations and dizziness and side effects of the prophylactic treatment such as fatigue, palpitations and headaches, that limit their ability to engage in their usual activities of daily living (Andersen et al., 2008). Even though many report anxiety and limitations impacting their daily lives they also wish to live as normal a life as possible (Andersen et al., 2008).

For parents, the biggest impact on their quality of life can emanate from the stress involved in making decisions for their children around managing and balancing the functional restrictions required in daily living in order to avoid triggering a cardiac event (Farnsworth et al., 2006).

Impact of LQTS on Social Functioning

LQTS is described as a lonely condition by some because of the restricted access they have to others with similar symptoms. This can mean few, or no, opportunities to socialize with others that can understand and identify with their experiences, potentially providing a sense of normality (Andersen et al., 2008). Ways to subvert this include providing support to other LQTS patients, including parents of affected children and those with ICDs (Andersen et al., 2008). Youth, in particular, can feel different and marginalised by their peers, with some even bullied for being different if they speak about their condition (Meulenkamp et al., 2008).

Healthcare of Long QT Syndrome Patients and their Psychosocial Needs

There is limited literature on what aspects of healthcare services or clinical care are helpful and unhelpful in meeting the psychosocial needs of LQTS patients, with none yet completed in New Zealand.

Two studies based in the United Kingdom endeavor to uncover adolescent and parent perspectives on when and how services could best help meet their psychosocial needs (Kendall, Sloper, Lewin, & Parsons, 2003a, 2003b). Findings consistent across both parent and adolescent perspectives identified parents as the primary information givers in the family. The parents generally did not know enough about the condition to act as reliable informants for the adolescents. It was also found

that the level of support a family gets often correlates with individual parents' ability to directly contact health professionals and consequently was influenced by how proactive, assertive and articulate they are. This has also been found in other research on chronically ill children (Beresford & Sloper, 1999; Kendall et al., 2003a, 2003b). For the adolescents, findings indicate a focus on the present as they mostly want to know about how the condition will effect their daily life (Kendall et al., 2003b).

The implications for health professionals and cardiac services from these studies is quite specific and consistent across adolescent and parent views. The amount of information individuals want and need differs and as such they recommend the amount of information given on any one occasion be flexible and individually tailored. Furthermore, the timing of information provision is important. Information given at the point of diagnosis or acute care is considered by patients as ill timed. It is preferred that information be provided at a dedicated session on a separate occasion. This way, questions can also be prepared. They believe a liaison nurse or social worker may be able to provide this service and address these needs (Kendall et al., 2003a).

The limited findings from the equally limited LQTS based psychosocial research consistently discuss the difficulties encountered by LQTS patients when receiving healthcare services. Findings from the Netherlands indicate that parents are generally dissatisfied with their ability to access information about LQTS. Some are also unsatisfied with support from physicians, especially general practitioners and referring cardiologists, feeling they had to inform them about the condition rather than the reverse (Hendriks, Grosfeld, van Tintelen, et al., 2005). Despite this and the other negative factors described, however, none of the parents regretted their decision to have their children genetically tested (Hendriks, Grosfeld, van Tintelen, et al., 2005).

In Norway, parents also feel unsatisfied with the healthcare their children receive for LQTS. Increased LQTS related symptoms in their children correlates with dissatisfaction with healthcare services (Andersen et al., 2008). Some specific difficulties are noted including parental frustration with not knowing about the risks in pregnancy and the risk of having a baby when they have LQTS.

Similar issues are encountered in the United States and Norway. The predominant problems experienced included general practitioners and other primary healthcare providers having very little or no knowledge about LQTS (Andersen et al., 2008; Farnsworth et al., 2006). In particular,

healthcare providers' lacked knowledge about symptoms of LQTS and the tests associated with it, which is essential information for accurate diagnosis (Farnsworth et al., 2006). This can, and has, resulted in the prescription of contraindicated medication, treatment or information, or being overly cautious and admitting them to hospital for minor illnesses (Andersen et al., 2008). Some parents have also experienced symptoms and concerns being dismissed by healthcare providers and incidences of missed diagnosis that have resulted in their child's death (Andersen et al., 2008; Farnsworth et al., 2006). Some people, not just parents, had also been told their symptoms were due to anxiety and their concerns had been dismissed (Andersen et al., 2008; Farnsworth et al., 2006). Individuals with ICDs have also experienced dissatisfaction, specifically regarding inadequate follow-up and maintenance of their devices (Andersen et al., 2008).

Parents indicate Cardiologists from tertiary services often give them appropriate detailed education at the time of diagnosis alongside outlined treatment options. However, when they return to their primary healthcare provider the general practitioners are often unfamiliar with LQTS or the treatment options. Parents consequently often feel solely responsible for watching out for their child's care (Farnsworth et al., 2006).

Interestingly, however, even though LQTS patients are largely unable to rely on the healthcare system to adequately meet their clinical needs, once the correct diagnosis is made a sense of safety in the healthcare system is restored; in respect to the treatment and the general clinical care provided (Andersen et al., 2008).

Psychosocial Impact of Genetic Testing

Psychosocial Risk Factors for Distress Following Genetic Testing

Research on the psychosocial impact of genetic testing for inherited diseases similar to LQTS, (i.e. colon cancer, breast and ovarian cancer and Huntington's Disease), have found particular pre-existing risk factors for psychological distress following genetic testing (Meiser, 2005; Morgan, O'Donoghue, McKenna, & Schmidt, 2008). That is, when these have been present they have predicted psychological distress in response to genetic testing. These can be divided into two areas dependent on their respective context: personal and family. Personal risk factors include:

- High levels of distress prior to testing
- A history of depression

- Gene positive and the result was inconsistent with that expected
- Those who seek threatening information are most likely to feel distressed while waiting for results
- Those who feel at higher perceived risk of death

Family context risk factors included:

- Being the first in the family to be tested and the result differs from most vulnerable siblings
- Having already lost a relative to the inherited condition
- Women with young children

Psychosocial Impact of Genetic Testing for Inherited Conditions

Recent research into the psychosocial impact of genetic testing for inherited conditions other than LQTS has extended the focus to the impact on families. They have found children, adolescents and adults often have different responses that tend to correspond to their developmental and psychosocial demands. Children, for example, are usually still emotionally dependent on their primary caregiver and so are highly influenced by the psychological wellbeing and preparedness of their mothers. That is, if a parent is anxious and avoidant of the process then it is more likely their child will also be anxious and distressed in response to the genetic testing process (Tercyak, Peshkin, Streisand, & Lerman, 2001). Children can also be affected by the psychological wellbeing of their siblings in this way too, especially if the sibling has a positive genetic result (Codori et al., 2003). If however, their mother maintains good psychological equilibrium throughout the genetic testing process, communicates openly, and facilitates education and support from appropriate health professionals for them, then the risk of psychological harm is generally ameliorated (Cameron, Sherman, Marteau, & Brown, 2009; Malpas, 2008).

Adolescents' primary focus has usually widened beyond the family. They are highly influenced by their social and peer groups relationships as they develop their identity (Ciaccio, 1971). Despite this broadening focus, research indicates the genetic testing process can still be distressing for them when they see their parents distressed. Low mood and hopelessness for the future can occur in adolescents who receive a gene positive diagnosis, especially for an adult onset disease, which can lead to further distress as their mood negatively affects their peer and familial relationships (Duncan et al., 2008).

Adults' developmental focus often shifts towards their children and grandchildren's wellbeing, along with that of their life partners (Ciaccio, 1971) (Tercyak et al., 2001). Uncertainty about the possible

implications of the genetic test result is a common experience in families with children diagnosed with chronic and potentially fatal conditions (Garwick, Patterson, Meschke, Bennet, & Blum, 2002; Santacroce, 2002; Stewart & Mishel, 2000). Parents often become distressed leading up to their children's genetic test result and for those with children who test positive these health-related worries for their children remain. This is especially so for those parents who have the condition themselves or have experienced others having negative experiences with the condition (Tercyak et al., 2001). Adults' relationships with their life partners and siblings are mutually affecting in that factors such as emotional distance, conflict or low emotional support prior to receiving a genetic test result often predict psychosocial difficulties after the result, at least in the short term (Koehly et al., 2008; Manne et al., 2004; Peterson, 2005).

In summary, psychosocial responses to genetic testing for an inherited condition often include distress, anxiety and depression. They are also highly influenced by those who are important in an individual's life. Who these are is often dependent on the developmental level of the affected person, with children being highly influenced by their parents' psychosocial responses and adolescents being affected by both parents and peers. Adults' responses are influenced by their focus on their children and life partners. These findings underscore the importance of considering the impact of the genetic testing process on the whole family alongside the developmental and psychosocial demands of the age of individual family members. It may also help explain why the research on LQTS to follow is largely centred on representing age stratified or familial role perspectives.

Differential Impact Depending on Presence of Symptoms

In 2005 a comprehensive review of the literature on the psychological impact of genetic testing for cancer susceptibility in Breast and Ovarian Cancer (BRCA1/2), Hereditary Non-Polyposis Colorectal Cancer (HNPCC) and Familial Adenomatous Polyposis (FAP) was conducted (Meiser, 2005). It was concluded that the psychosocial impact on individuals, especially in relation to their genetic status, are mediated and exacerbated by the presence of symptoms. In a study assessing the impact of two recessive disorders (cystic fibrosis (CF) and ataxia-telangiectasia (A-T)) on siblings the impact of the genetic condition was moderated by the phenotype. That is, the psychosocial impact was more severe in a visible condition (A-T) than in the less visible condition (CF) (Fanos & Johnson, 1995). This has particular relevance to LQTS as symptomatic and asymptomatic presentations are common but thus far have not received specific attention in the research.

Determinants of Decision Making in Genetic Testing

Citing concern regarding adolescents' and parents' ability to make decisions regarding genetic testing, a recent study looked into levels of knowledge and approaches to decision making about genetic testing according to age (Rew, Mackert, & Bonevac, 2010). Findings indicate younger and older individuals are limited in their knowledge of genetics, and genetic testing, while older adolescents in particular demonstrate better knowledge and consideration of a broader range of issues when making decisions about genetic testing (Rew et al., 2010). These findings suggest improved education about the benefits and harms of genetic testing is advantageous prior to testing (Rew et al., 2010).

A hypothetical study looked into parents' willingness to use predictive testing when no treatment was available. They hoped to ascertain if uncertainty regarding symptom onset or severity had an effect. It was found that approximately one third of parents indicated they would agree to predictive testing of their child irrespective of no treatment being offered. Uncertainty regarding symptom severity or onset, however, had no significant effect on parents' willingness to predictively test their child (Tarini, Singer, Clark, & Davis, 2009).

Research has also investigated the factors determining who decides to undergo genetic testing. In studies of inheritable cancers the decision to be tested has been related to psychological factors rather than socio-demographic ones. That is, findings suggest those already affected with an inheritable cancer or those who are anxious about getting it (in particular) are more likely to want to undergo genetic testing. So are those who think they are at higher risk of getting it, have children or have more first degree relatives affected (Codori et al., 1999; Esplen et al., 2007; Hadley et al., 2003; C. Lerman et al., 1999; Meijers-Heijboer et al., 2000). Reasons for undergoing testing included efforts to further research on genetic testing, to meet recommendations from clinicians, and in the case of some cancers, to know if more screening was necessary (Esplen et al., 2007). Some concerns were also raised including fear of losing health insurance (Oster et al., 2008) and psychological distress after receiving the result (Godard, Pratte, Dumont, Simard-Lebrun, & Simard, 2007). Therefore, a combination of psychological and more pragmatic reasons appear to impact decision making for genetic testing.

Ethical Implications

Ethical practice in genetic testing requires confidence that information about genetic risk and status can be provided without damaging psychological or behavioural consequences (European Society of Human Genetics, 2009). Currently ethical debate regarding genetic testing, especially as it relates to LQTS, largely concentrates on whether children in particular should receive genetic testing as there are mounting concerns regarding the impact having an early diagnosis might have on a child and their family (Delatycki, 2008; Malpas, 2008). Some believe predictive genetic testing should be delayed until a person is able to consent for themselves but opinions are divided on this issue, with the UK based Clinical Genetics Society (Clinical Genetics Society, 1994) advising testing may be necessary in the child's best interests while the European Society on Human Genetics advises against such testing (European Society of Human Genetics, 2009). The guidelines for genetic testing for Huntington's Disease, the most researched adult onset disease, advises against testing under the age of 18 years because of consistent findings of negative effects for children and families post testing (International Huntington Association and the World Federation of Neurology Research Group on Huntington's Chorea, 1994). Ethical decisions around genetic testing are traditionally based around balancing clinical benefits and risks (Wilfond & Ross, 2009) and as such it is generally considered acceptable to genetically test children if onset of the condition in question is commonly in childhood or adolescence and treatment or prevention is possible (Delatycki, 2008; Meulenkamp et al., 2008).

Decisions about whether to test children and youth, are usually made collaboratively by the clinician and parents, in the best interests of the child (Hall & Burton, 2010). Very few studies have been conducted on clinicians' perspectives on making decisions about whether to genetically test youth. One study, looked into decision making by GPs and Pediatricians to genetically test youth across age groups. Findings indicated most clinicians agreed it was appropriate to conduct predictive genetic testing for a 12 year old if treatment was available for the condition but the majority felt it was inappropriate to test a 6 months old (Plass et al., 2009).

Genetic Counselors often act as intermediaries between medical specialists, geneticists and the patient, explaining the costs and benefits as well as the condition's specific genetics. A recent study in the United States of America found genetic counselors support the use of genetic testing when the results determine disease progression, prognosis, survival rates after treatment or risk for an adverse drug reaction (Mackoff et al., 2010). They also strongly favored parents' and children's rights over doctor or insurance rights, perceiving insurance companies often as discriminatory (Mackoff et al.,

2010). Genetic Counselors did not support genetic testing for susceptibility to later disease development or when no treatment would be available (Mackoff et al., 2010).

Privacy of genetic test results in inherited conditions is another area of ethical debate, primarily because family studies/cascade testing cannot be completed without impinging on the privacy of the first family member to be identified with the condition (proband). At present, the protocol for this testing is for the proband to contact relatives and request they make contact with the appropriate service (European Society of Human Genetics, 2009). It is recognised however, that this may not be the optimal method of contacting family members as it makes it impossible then to maintain one's own privacy if that is desired. A direct approach has been recommended whereby permission is requested from the proband and once given family members are approached directly by a genetic nurse (European Society of Human Genetics, 2009; Newson & Humphries, 2005). UK case law too, is currently grappling with this issue, with cases believed to be extending the rights of patients from the currently practiced confidentiality and protection of information disclosed to a right to privacy, whereby protection is against unauthorised use as well as disclosure (Hall & Burton, 2010).

Impact of Genetic Testing on Clinicians

International research on how prepared general practitioners (GPs) in primary care are to meet the demands of modern genetic testing have consistently found GPs' knowledge is limited (Baars, Henneman, & Ten Kate, 2005; Emery, Watson, Rose, & Andermann, 1999; Escher & Sappino, 2000). A survey was conducted for the National Health Committee (NHC) on the current practice and training needs of GPs in relation to genetic testing in NZ. Findings confirmed that GPs in NZ have limited experience with genetic testing and would value both further training and access to information (Morgan et al., 2004). This mirrors international views (Baars et al., 2005; McCann, MacAuley, & Barnett, 2004). GPs felt that despite acceptance of the important role of genetic testing (Cameron, Reeve, Readings, & Winship, 2002) and there being patient specific information available to them from Genetic Services (who usually provide the genetic testing), a large proportion of GPs don't know how to access the service. They also felt genetic testing was too costly and the waiting periods restrictive for patients (Morgan et al., 2004). NZ GPs demonstrated a general lack of knowledge about inherited conditions, the appropriate terminology to use and the procedures involved in genetics (Morgan et al., 2004). Internationally too, GPs are restricted in their ability to adequately assess genetic based familial risk, recognize the inheritance patterns of different congenital conditions and confidently provide genetic advice based on genetic test results (Emery et

al., 1999; Escher & Sappino, 2000; Sarfati, 2002). GPs level of genetics based knowledge is a reflection of their training, of which many received little (Morgan et al., 2004). Medical graduates and medical students however, have demonstrated greater knowledge of genetic testing issues since the integration of more genetics training in NZ medical schools (Cameron et al., 2002), suggesting targeted learning opportunities for less recent graduates and longer practicing GPs may be advantageous.

Psychosocial Impact of Genetic Testing for LQTS

LQTS is unique among the inheritable conditions. That is, LQTS poses the most risk for the young; there are gender differences for risk; and the frequency of symptoms experienced can be highly variable, from none whatsoever to frequent sudden cardiac events involving serious risk to life. Also, genetic testing for LQTS can be predictive for family members of a diagnosed relative who often do not experience symptoms or whose clinical tests are inconclusive. The testing can also be diagnostic and confirm clinical tests for the presence of LQTS. Furthermore, subtypes of LQTS can be identified through genetic testing so that treatment can be tailored and triggers/risk factors to avoid can be specified. These factors are particular to LQTS and for this reason it is important LQTS be researched independently. Unfortunately, the psychosocial impact of genetic testing for LQTS is under researched. Psychosocial research has struggled to keep up with the rapidly developing pool of genetic and medical knowledge on LQTS. At present, what is known is largely based on responses to predictive testing, with research on diagnostic testing and comparisons of the two responses extremely limited.

Contributing Factors Leading up to Gene Based Diagnosis

Findings indicate the degree of certainty provided by a clinically based diagnosis prior to predictive genetic test results can have a direct affect on the psychological response to gene based diagnosis. That is, those who receive an uncertain clinically based diagnosis can be particularly susceptible to distress in the short term (Hendriks et al., 2008). Specifically, those with an uncertain result that go on to receive a genetically positive result have a tendency to experience higher disease related anxiety prior to genetic testing. Researchers suggest nonverbal communication from a cardiologist who has differentiated those at higher probability may contribute to their anxiety. In the long term however, distress was found to be restored, returning to normal levels irrespective of the LQTS genetic mutation outcome. That is, the clinically based diagnosis leads to distress but results from

genetic testing do not seem to increase distress any further (K. S. Hendriks et al., 2008). This is consistent with research findings on the impact of genetic testing in other inherited conditions (Duiterhof, Trijsburg, & Niermeijer, 2001; Caryn Lerman, 1997).

For parents, a clinically based diagnosis can indicate potential imminent and future risks of their child having a cardiac event. Consequently, parents often feel distressed about the possible outcome of the genetic test result from the time of the clinical diagnosis (Hendriks, Grosfeld, Wilde, et al., 2005). This may also be because treatment and LQTS management often begins at this stage and potentially means daily medication and restrictions on physical activities and/or emotional arousal. This requires significantly increased parental surveillance and the enforcement of risk related restrictions that are likely to be stressful and potentially distressing new challenges for parents and their child to manage.

Particular factors appear to put parents at further risk of developing significant distress in response to the genetic test result for their child. These include having had previous long standing experiences with LQTS prior to testing (Hendriks, Grosfeld, Wilde, et al., 2005). This may be due to the saliency around risk with prior exposure to the condition and the ability of inexperienced parents to minimize the threat, without evidence to the contrary, thereby managing their ability to cope with the distress (Hendriks, Grosfeld, Wilde, et al., 2005).

Parents, generally experience less distress after genetic test results are received if their child's predictive result is gene negative for LQTS and more distress if the result is gene positive for LQTS. The genetic status of parents, however, does not generally correlate with distress (Hendriks, Grosfeld, Wilde, et al., 2005).

However, high levels of distress are experienced by some parents when receiving a clinically based diagnosis and this distress is often the same when they receive a genetically based diagnosis for their child. That is, distress related to genetic test results doesn't increase or decrease in the short term for these parents compared to that felt at the time of initial clinically based diagnosis. In fact, experiencing high distress after receiving a clinically based diagnosis predicts high distress after a genetic based diagnosis. This may be because this subsection of parents often experience a predictive gene based diagnosis as confirmation of what they expected on the basis of a clinically based diagnosis (Hendriks, Grosfeld, Wilde, et al., 2005). This underscores the potential importance

of seeing clinically and genetically based diagnoses as part of the same process of adjustment for parents.

Impact of the Initial Acute Phase after Gene Based Diagnosis/Results

Diagnosis a Relief

For some, the process of receiving a diagnosis (clinical or genetic), however, can be a relief. Often because the waiting process has engendered a sense of uncertainty about the future, and is therefore distressing. The genetic test result can provide a sense of control; as individuals can receive medication and mediate risks themselves by avoiding triggers, that ultimately also aids adjustment (Andersen et al., 2008).

Familial Perspectives on Receiving Genetic Test Results

Parents appear particularly susceptible to high levels of anxiety and depression in the short term when undergoing predictive genetic testing themselves. Consistent with the aforementioned risk factors for problematic psychosocial outcomes of genetic testing, this is more likely to occur in parents who were highly distressed prior to genetic results and have been exposed to LQTS in a relative prior to testing. Having more affected family members and only gene positive children are additional factors that predict distress in parents, perhaps adding to the saliency of the threat to their mortality (Hendriks, Grosfeld, Wilde, et al., 2005). Receiving a diagnosis can also affect partners of those who are LQTS gene positive. Findings indicate that they experience higher levels of disease-related anxiety at clinical and genetic diagnosis points, and in the long term, compared to those with LQTS gene negative partners (Hendriks et al., 2008).

On the whole children and youth, however, tend to cope well with receiving genetic test results (Meulenkamp et al., 2008; Smets et al., 2008). A small proportion, however, may be vulnerable to distress. For example, youth who have experienced a serious cardiac event in the family immediately prior to genetic testing can show increased distress (Meulenkamp et al., 2008). Perhaps this is because they have had to deal with a loss as well as cope with finding out their own risk, or because the risk is more salient when you know someone who has experienced a serious cardiac event.

Impact of Chronic Phase after Gene Based Diagnosis/Results

Findings on the long term impact of genetic testing for LQTS are limited to the perspectives of parents. Generally, parents perceive their children and teenagers' quality and appreciation of life usually improves once they have adjusted to their genetic status over time, even though they may have experienced difficulties adjusting in the acute phase (Arteaga & Windle, 1995; D. Carroll, Hamilton, & McGovern, 1999; Farnsworth et al., 2006). This is consistent with other findings on chronic conditions (Arteaga & Windle, 1995; D. Carroll et al., 1999). It is, however, difficult to clearly identify differences between the impact of a diagnosis or genetic test results and the impact of having a particular condition.

Parents Help their Young Adjust

Particular factors have been associated with parents' ability to help their children and teenagers adjust to their gene based diagnosis. For example, adults with knowledge of, and experience with, LQTS as a parent found this benefitted their LQTS positive children and adolescents, enabling them to adjust more adaptively to their genetic status and the resulting changes (Farnsworth et al., 2006). Also, parents who were gene positive for LQTS and had children who had undergone genetic testing found one of the long term benefits of receiving their genetic test results was a better understanding of their risk of sudden cardiac death. This allowed them to take better responsibility for their own and their children or grandchildren's health, consequently generating increased feelings of control and containment in respect to their LQTS (Andersen et al., 2008).

Benefits of Understanding Developmental Differences

Understanding differences in the developmental impact of the genetic test results and treatment related changes may also aid parents in helping their children adjust. That is, children and teenagers respond differently to their genetic status results, with teenagers having a more difficult time adjusting because of the greater and more sudden impact of the restrictions. Children however, grow up with the restrictions on their activities as their norm while teenagers often have to make drastic and sudden changes, especially if heavily involved in sport or working towards a career in the Armed Forces (Farnsworth et al., 2006). It can, therefore, be beneficial for LQTS positive children to grow up understanding their condition rather than be presented with it suddenly later. Providing parents with information on their children's diagnosis as early as possible can be important in order to achieve this (Andersen et al., 2008). Parents do not feel a child knowing of their diagnosis early on will cause undue anxiety (Andersen et al., 2008). Findings on the psychosocial responses of children

and youth support this age stratified view. While children generally cope well with genetic test results some adolescents can encounter developmentally appropriate social and autonomy related problems, such as being bullied, withdrawal from peers and medication non-compliance (Meulenkamp et al., 2008).

Adverse Responses from Parents

As aforementioned, parents are at risk of anxious and depressive responses to genetic test results in the short term. A good proportion (approximately 75%) also show signs of anxiety in the long term, with those who had previously lost a relative to sudden LQTS death being especially prone (Hendriks, Grosfeld, van Tintelen, et al., 2005). Perhaps it is not surprising then that some parents struggle with parenting, particularly with being overprotective, and worried about the effect the behavioural outcomes their increased concern could have on their children (Hendriks, Grosfeld, Wilde, et al., 2005). Hendricks et al. (2005) suggested significantly increased parental focus on their children's welfare may be a reflection of feelings of responsibility for their children due to the inheritability of the condition and guilt parents may feel at having passed on the condition. In response, some LQTS children expressed frustration and insecurity as a consequence of this over-involvement by parents, with restriction of their access to unsupervised time with friends a particular problem (Meulenkamp et al., 2008; Stam, Hartman, Deurloo, Groothoff, & Grootenhuis, 2006).

Parents often experience difficulty adjusting to the results of their LQTS genetic positive children (Hendriks, Grosfeld, van Tintelen, et al., 2005). When high levels of distress are experienced shortly after receiving genetic test results the distress levels often remain the same in the long term (Hendriks, Grosfeld, van Tintelen, et al., 2005). Almost uniformly in the research, high levels of distress are correlated with high levels of depression and anxiety. This has been found to be especially so when parents are familiar with LQTS, either in themselves or their relatives and when they are dissatisfied with the information given by clinicians (Hendriks, Grosfeld, van Tintelen, et al., 2005).

In adults undergoing predictive testing, irrespective of whether they were parents, their own LQTS genetic test outcome was not predictive of generalised anxiety or depression in the long term (Hendriks et al., 2008). This is consistent with findings from research on predictive testing for other inherited conditions (Almqvist, Brinkman, & Wiggins, 2003; Decruyenaere, Evers-Kiebooms, & Cloostermans, 2003). However, specific disease-related anxiety remained somewhat increased over

time for LQTS gene positives, especially for those whose clinical diagnosis was initially uncertain, as aforementioned (Almqvist et al., 2003; Hendriks et al., 2008).

Youth often take their medication irregularly which can be because they doubt the medication's effectiveness (Hendriks, Grosfeld, van Tintelen, et al., 2005; Hendriks, Grosfeld, Wilde, et al., 2005). Feeling in control is often paramount for coping in youth (Meulenkamp et al., 2008) and perhaps making independent decisions about the medication and adherence is a process through which youth can exert some control.

Recommendations from Findings

Recommendation consistent across studies is for careful genetic counselling before and after receiving results, including providing information that is individually tailored to meet the needs of different age groups, differing levels of genetics knowledge, and to support information seeking coping. Psychological support is also recommended to contribute to the assessment and management of risk and the ongoing well being of at risk individuals in the short and long term (Andersen et al., 2008; Hendriks, Grosfeld, van Tintelen, et al., 2005; Meulenkamp et al., 2008).

Summary

The literature on genetic testing demonstrates it is extremely useful for the identification and treatment of LQTS, and has been increasingly used in New Zealand over the last decade. Relatively little is known about the psychosocial impact of this process, however, as research thus far has been largely restricted to responses to predictive genetic testing and the perspectives of parents. What is clear from this research is that most cope well with the genetic testing process and are often impacted more by the initial clinical diagnosis. Some have significant, often age or familial role related, psychosocial difficulties however. These include being influenced by the distress of other family members, peers and partners; the presence of pre-existing psychological difficulties; prior exposure to LQTS cardiac events; negative healthcare experiences; and the presence of symptoms. Many of these factors continue to be problems when living with LQTS in the long term. These problems have been linked to high levels of illness related anxiety and depression, leading to increased overall distress for some and an exacerbation of symptoms.

Conclusion

This literature review has had several purposes. The full complexities within the condition of LQTS were intended to be captured, and conveyed, as these may contribute to our understanding of what people experiencing LQTS are required to adjust to. As may be seen, it is a potentially complicated condition to understand, encompassing both complex physical and genetic processes, especially for those unfamiliar with specialist cardiology or genetics. For those experiencing the condition it may, therefore, be hard to create a framework from which to understand one's self, as someone with LQTS, and to adequately explain the condition to others. This may be equally the case for primary or secondary care clinicians, such as general practitioners. It is intended that by giving such a thorough account of the condition, and the multiple processes within it, those reading this thesis, including those experiencing LQTS and the many professionals involved in their care, may have an opportunity to develop this understanding, as I have.

The literature provided on LQTS also sought to demonstrate the differing condition-specific factors that may impact on individuals with LQTS. For example, those diagnosed with LQTS must make multiple changes to their lives once the diagnosis is made. Everyday activities that may have been an important part of their social or personal lives may need to be restricted or completely ceased. The person experience a range of restriction from giving up being part of an important sports group, to no swimming, to no longer being able to wake and feed your infant in the night. These potentially required changes may have a real life impact on the individuals, and families, of those who are asked to make them. These experiences may serve as a constant reminder of their vulnerability or to isolate them from others, for example. This research provides an opportunity to explore these experiences so that we might better understand the potential implications of such condition-specific factors affecting those with LQTS.

In addition, the process of getting to the diagnosis, both clinical and genetic, and the treatments, include clinical experiences that in all likelihood also need to be psychologically and socially managed. Within the diagnostic and treatment processes there are a number of specific procedures that may be experienced as stressful by those undergoing this. For example, investigations such as the provocation test and the implantation of cardiac devices may be emotionally demanding for patients.

A LQTS patient may also have to go through up to two diagnostic processes, clinical and genetic. As aforementioned, the research postulates there is likely to be a psychosocial impact from the diagnoses, and of genetic testing, but there is little information about the first hand experiences of adults and adolescents with LQTS. Also, the services that patients receive have been demonstrated in the literature to potentially interplay with this clinical and psychosocial process, such as when misdiagnosis is made, for example. It has not yet known, however, the role New Zealand health care services have to play in mitigating the LQTS patients' psychosocial experiences of clinical care.

Then there is also the fact that approximately fifty percent of those experiencing LQTS experience absolutely no symptoms and no apparent signs of having a condition. This raises questions about how this absence of symptoms impacts their psychosocial adjustment. In addition, the other fifty percent are required to manage the physical and psychosocial impact of cardiac events, which the literature suggests may be a difficult process for some. It may, therefore, be interesting to see accounts of these different experiences through the lens of psychosocial adjustment theories such that we might better understand any differences in experiences between 'symptomatic' and 'asymptomatic' patients. Developmentally too, what theory tells us is there are differences between the psychosocial focus and needs of adolescence and adulthood that might also help us begin to better understand any differences between these adolescent and adult groups.

In addition, a section of the research to date on the psychosocial impact of LQTS has separated out those undergoing predictive testing, or those 'at-risk' of having LQTS, from those who have already been 'diagnosed' and confirmed through genetic testing. As can be seen in the description of the diagnostic clinical processes provided, however, this is often not that clear cut. The immediate families of the first person identified in a family (the 'proband') are usually clinically tested (e.g. ECG and/or Holter Testing), and sometimes clinically diagnosed and/or given a probability of having LQTS, *before* predictive genetic testing is conducted. This potentially means there are some who undergo predictive testing, and are consequently considered at-risk for LQTS, when they already have received a probable diagnosis while others, at the other end of the spectrum, may have been told it is very unlikely given the results of the clinical tests. It is, therefore, perhaps unclear the role the different diagnoses take and whether the 'at-risk' predictively tested group is entirely distinct, and appropriate to be asserted as such, from those who are already 'diagnosed'.

Theories that contribute to our understanding of what it takes to adjust to a health condition include those on coping, quality of life, the emotional support we receive through social and familial supports and psychological responses to each stage of a chronic condition. But as we have seen through the description of the condition, LQTS has a unique set of clinical and psychosocial experiences that we might seek to understand in this context. Given what we know about coping, for example, how do individuals with LQTS, and their families, cope with these experiences? What happens when they do or don't cope effectively? What do they experience at these times? What we know at this time is there are different ways to cope, and by overlapping these theories; as well as those on quality of life, the phases of a condition, social supports and psychosocial responses to chronic conditions, with the experiences of those with LQTS we might better understand the LQTS patient. By understanding their experiences alongside these theories of adjustment we might begin to get a picture of their psychosocial needs.

Given the potential severity of the psychosocial difficulties encountered by some, and the paucity of research on the impact of both living with and being genetically tested for LQTS, this study aims to extend on the existing research, using theories of adjustment and what is currently known about the challenges facing those with LQTS or a chronic condition to explore and hopefully further understand the psychosocial factors impacting those with LQTS, including those undergoing the genetic testing process. Secondly, the study aims to explore whether and how experiences may differ according to the different subgroups (e.g. 'symptomatic' and 'asymptomatic') identified in the literature. The outcomes of this research are intended to contribute to both the international pool of research and to provide New Zealand specific knowledge that is intended to aid those working with LQTS patients in New Zealand healthcare services.

Chapter 2

Consultation

Common criticisms of some psychological research, and indeed research in general, is that it is *about* the people rather than *for* them (Seymour & Davies, 2002). This study was initiated by the Cardiac Inherited Diseases Group (CIDG) in order to help them to better understand the adjustment processes of their patient group, evaluate the practices within their service from a pragmatic perspective and to provide feedback and results in such a way that positive change may be achieved, benefiting clinicians, individuals and families with LQTS. The following chapter outlines the process through which the research project developed, the consultation that occurred leading to the development of the specific research methodology and design, the relevant ethics involved, and finally, the rationale and aims of the research project are described.

The Setting

The following section describes the group that requested the research and the youth and adult medical services they work through. Next, the process a LQTS patient typically goes through is illustrated and explained from both youth and adult perspectives.

The Cardiac Inherited Diseases Group (CIDG)

CIDG is a nationwide multidisciplinary group whose main aim is to "prevent sudden cardiac death due to inherited heart conditions in the young" (Cardiac Inherited Diseases Group, 2010). There are representatives from numerous professions including specialist Cardiologists, Electrophysiologists, Molecular and Clinical Geneticists, Genetic Counsellors, Pathologists and associated professional staff, from eight different regions around New Zealand. In Auckland, members of CIDG specialising in Pediatrics are based in the Department of Pediatric and Congenital Cardiac Services (Cardiac Services) at Starship Children's Health and adult services are at the Greenlane Clinical Centre and in Auckland City Hospital. The Chairperson for CIDG and the Coordinator of the LQTS Registry, which was in the development phase when this research project began, are both based in the Department of Pediatric and Congenital Cardiac Services (Cardiac Services) at Starship Children's Health, Auckland.

CIDG are interested in inherited cardiac conditions, including Long QT Syndrome, Brugada Syndrome, Dilated Cardiomyopathy, Hypertrophic Cardiomyopathy and Arrythmogenic Right Ventricular Cardiomyopathy. As a group, CIDG states its aims are to facilitate screening of at-risk individuals and their families, to provide education about inherited cardiac conditions to the public, patients and medical professionals alike, and to develop best practice guidelines to enhance professional practice in the field (Cardiac Inherited Diseases Group, 2010). CIDG aim for this screening and educating body to one day become a government funded national service and intend to make their case by demonstrating its utility in saving lives through clinical best practice and the clinical application of molecular genetics, better known as genetic testing (Cardiac Inherited Diseases Group, 2010).

Typical Process of CIDG Service Delivery

Pediatric Cardiology

The Pediatric Cardiology department, in which the Pediatric Congenital Cardiac Service operates, is a tertiary referral centre for NZ, therefore they receive all complex case referrals and all cardiac inherited diseases go through CIDG (see Figure 7 below for example of typical referral process). The CIDG Chairperson is also the only Pediatric Electrophysiologist in NZ so he sees all the complex pediatric electrical arrhythmia cases including the congenital or inherited cardiac conditions such as LQTS. These inherited cardiac cases are allocated to the Pediatric Congenital Cardiac Service and the CIDG Chairperson is also the head of this service.

Adult Cardiology

The process from referral to adult services is different to the pediatric cases only in terms of the initial presentation and referral. This is primarily because potential cardiac patients are typically seen as inpatients after having had a cardiac arrest. It is highly atypical for a patient to be referred as an outpatient. The process from referral onwards is the same as with Pediatrics (see Figure 8) except they are seen at Auckland City Hospital in the Cardiac Care Unit (CCU) (rather than at Starship Children's Health).

Figure 7. Referral process from GP to Pediatric Congenital Cardiac Service

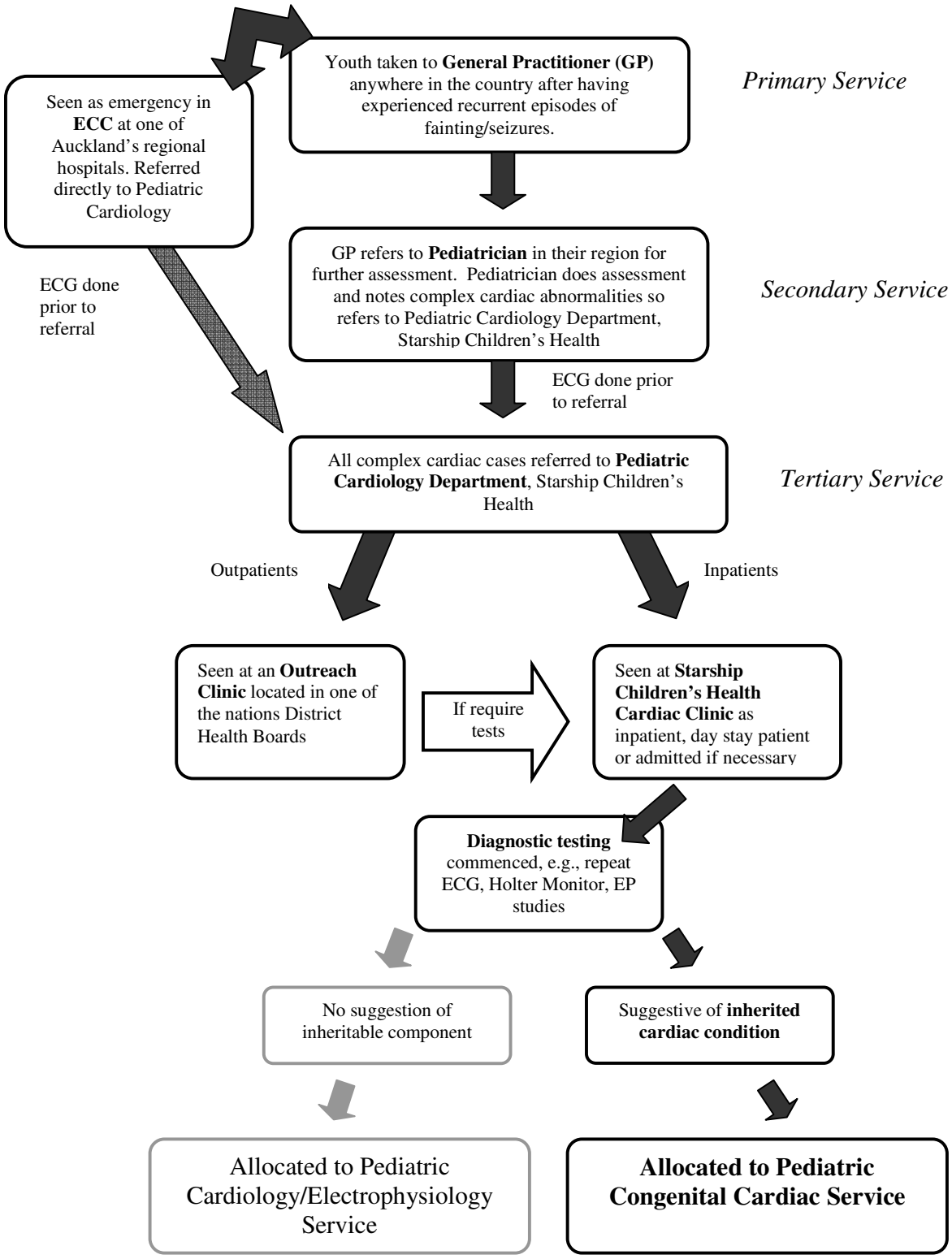
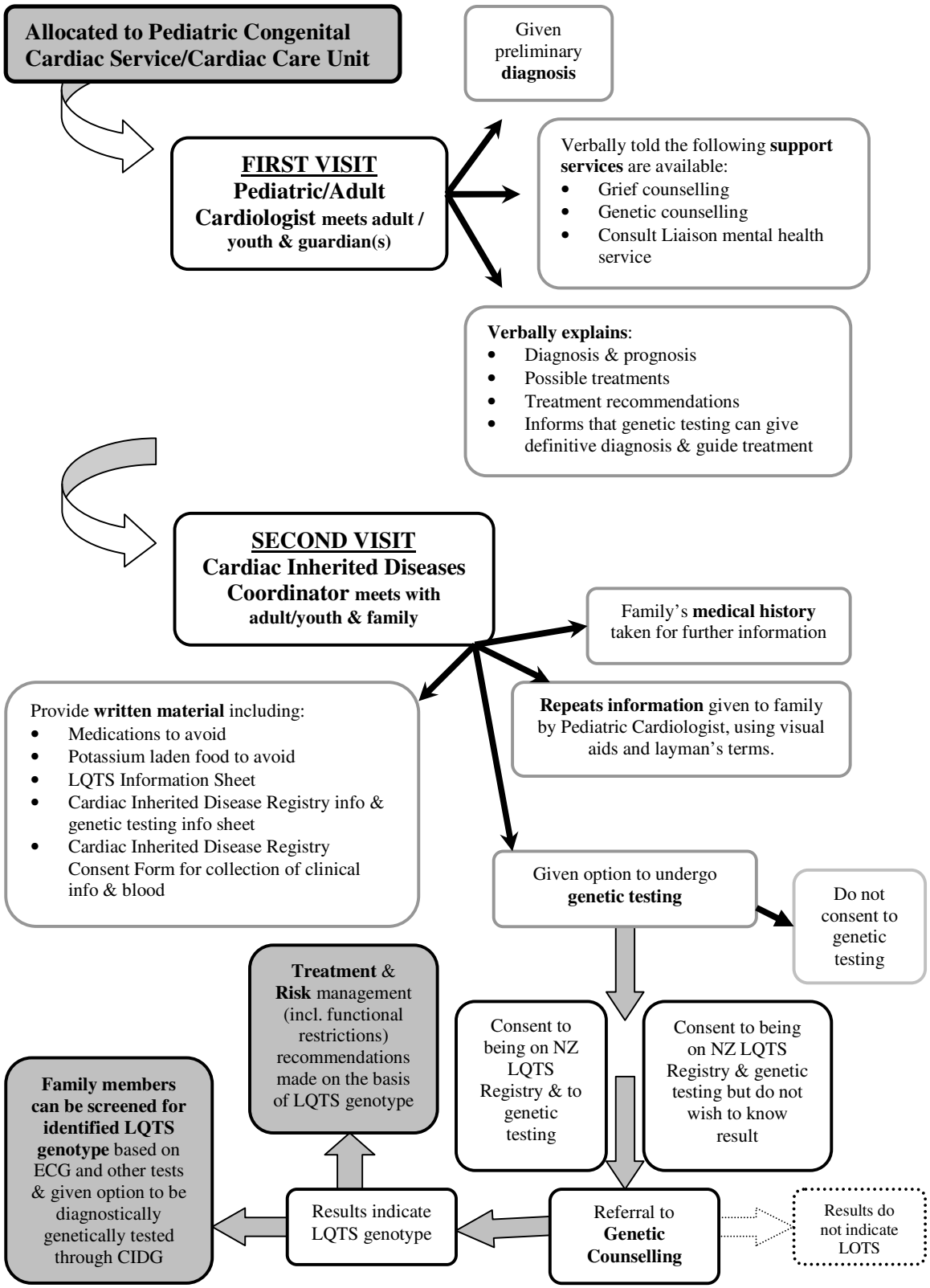


Figure 8. Referral process from Pediatric or Adult service to genetic testing



Origin of the research with CIDG

The CIDG Chairperson and Consultant Pediatric Cardiologist from Cardiac Services first approached a faculty member of the Population Health Department at the University of Auckland to conduct research on the psychosocial impact of sudden death in families in 2004. A research proposal was developed and put forward to the department but was not pursued after encountering difficulties organising the research project's practicalities with CIDG. Difficulties included the lack of a system for organising information relevant to the LQTS patients seen by the service and difficulties liaising with key administrative staff.

In 2005 CIDG referred a group of LQTS patients to the Consult Liaison Team (CLT), the mental health service available to patients at Starship Children's hospital. CIDG clinicians reported some concern they may not be adequately identifying LQTS patients that could benefit by having mental health support and the two services together discussed the possibility of doing a piece of research aimed at identifying LQTS patients' psychological needs and whether CIDG staff were currently meeting them. At the time, two clinicians from CLT held joint positions with the University of Auckland within the Psychology and Medical Health Sciences departments. At the end of 2005 a Doctorate of Clinical Psychology Candidate (the principle investigator and writer) was approached by one of these faculty members and asked to lead the project.

Given the difficulties initially experienced careful consultation with CIDG was advised and undertaken and the cardiac clinic process and Cardiac Services structure was reviewed with an evaluative and pragmatic eye. This was in an effort to ensure the same difficulties would not be encountered again. At the time, a database was being developed that would hold all the data pertaining to the LQTS affected and at-risk patients. This database was projected by CIDG to be up and running before participant recruitment was expected to begin the following year. A good relationship was also established with key administrative staff and avenues of communication were specifically defined. This being in place, a research proposal was consequently developed and accepted by all relevant parties; including CIDG, the Department of Psychology at the University of Auckland, the Ministry of Health National Ethics Committee and Auckland District Health Board.

Consultation Process and Outcomes

In the following section the consultation process with CIDG is described, including information about who was consulted and the outcomes of this. Defining researcher roles is reviewed next and, finally, the ethical and cultural consultation process is described.

CIDG clinicians involved in the genetic testing for LQTS process were consulted at regular intervals throughout the research project and especially in the development phase. The author first attended meetings with the CIDG team as a whole in order to become familiar with the system, the clinicians and the clinical process involved for those experiencing LQTS and potentially undergoing genetic testing. The author then conducted a series of individual semi-structured interviews from key clinicians identified by CIDG. The purpose of these interviews was to ascertain common areas of research interest and establish the research aims. The following clinicians were identified and consequently interviewed for approximately 30-90 minutes:

- Consultant Pediatric Cardiologist & Electrophysiologist, Department of Pediatric & Congenital Cardiac Services, Starship Children's Health & Auckland City Hospital
- Cardiac Inherited Disease Registry Co-ordinator & Senior Technologist, Department of Clinical Physiology, Starship Children's Health & Auckland City Hospital who arranged and supported the cardiac clinics and follow ups
- Clinical Registrar and Fellow, Department of Pediatric & Congenital Cardiac Services, Starship Children's Health & Auckland City Hospital whom assisted the Consultant Pediatric Cardiologist
- Child & Adolescent Psychiatrist, Consult Liaison Team, Starship Children's Health & Senior Lecturer, Department of Psychological Medicine, University of Auckland who saw families referred by Cardiac Services with concerns about their mental health
- Geneticist & Associate Professor, Head of the Medical Genetics Group, School of Medical Sciences, University of Auckland and head of laboratory initially conducting LQTS genetic tests

Consultation Outcomes

The CIDG clinicians reported a primary interest in learning more about how LQTS patients psychologically adjust to, or cope with, having LQTS after they leave the hospital clinic following diagnosis. Specifically, they were interested in the impact of receiving genetic test results and how

this may differ for those who experience symptoms, and therefore usually already have an initial LQTS diagnosis, and those who are Asymptomatic and may only have a tentative initial LQTS diagnosis, but have a family member who has been diagnosed for example.

CIDG clinicians were also seeking clarification around whether LQTS patients' experience is that it's better to know about their LQTS genetic standing or to remain unaware, as some patients appeared to prefer. Also, what was the impact on family relationships when family members received a positive or negative genetic test result? Specifically, they wondered whether guilt and blame affected parental, spousal and sibling dyads when looked at in the context of positive versus negative LQTS genetic results. For example, did parents with a positive genetic test result feel guilty for passing it on? Also, did siblings with a negative genetic test result feel guilty for not having the condition when another sibling received a positive test result and did anyone blame a genetically positive parent or spouse? Also of interest was identifying which aspects of resiliency and coping differentiated those that ultimately adjusted, coped and found benefits during the process.

The CIDG clinicians also wanted to know how well the service they currently provided matched the emotional needs of their LQTS patients, specifically in terms of the way the results were given, by whom and how, plus whether they were providing enough and the right supports to the children, adolescents and adults that made up the LQTS affected families.

The CIDG clinicians thought obtaining feedback and consequently learning about these factors could allow them to more adequately prepare and counsel LQTS patients about what they might expect during the genetic testing for LQTS process, make suggestions about what can help based on what they know has helped others in the past, and identify those families that may benefit from additional support.

Australasian Collaboration Initiated but Suspended

The primary researcher was alerted by CIDG to an Australian based project investigating the psychosocial impact of genetic testing for LQTS. Both NZ and Australian parties could see the potential benefits of having a larger data pool thereby adding statistical power to correlations, and enabling comparisons between the samples. A quantitative part of the present study was consequently designed to identify the psychological and demographic factors that distinguished those that struggled to psychologically adjust to their genetic test results from those who did not. This

paralleled the study being conducted in Australia and, as consistency was required for direct comparisons, it was decided both would be longitudinal, over the course of the genetic testing process, and questionnaire based. Unfortunately, it was not possible to proceed with this collaboration or the quantitative portion of the research project, however, as data collection problems beyond the control of the primary researcher were encountered by the NZ study. These precluded continuing with the longitudinal project.

Research Roles

The primary researcher in the NZ study was a representative from the University of Auckland and as such was a separate party otherwise uninvolved with CIDG and the cardiac clinic process. While this was seen as an advantage in terms of providing an objective point of view for the research it was potentially also a disadvantage. Access to potential participants could be more difficult, as might accessing participant information in a timely manner. In an attempt to address these potential problems two members of the cardiac team agreed to be co-researchers with one specifically appointed as the Liaison for the study. The role of the Liaison was defined and primarily consisted of facilitating access to information about the clinical process of genetic testing for participants, assisting in accessing potential participants and advising the primary researcher immediately after participants received their genetic test results so further questionnaires could be sent out. The other co-researcher was the Chairperson of CIDG and a Consultant Pediatric Cardiologist and Electrophysiologist on the cardiac team at Starship Children's Hospital. His primary role was to help the project maintain a high profile among the Cardiologists at both Starship Children Hospital and Auckland City Hospital so that they would give potential participants research invitations.

At the time of initially designing the study the laboratory conducting the genetic testing for LQTS in New Zealand was a University of Auckland research facility, but plans were in place for the testing to shortly be taken over by a hospital based laboratory.

Ethics and Cultural Safety

The study received ethics approval from the Health and Disability Ethics Committees, Northern X Regional Ethics Committee, Ministry of Health (NTX/06/12/158) and ADHB Human Participants' Research Committee (3653). To ensure cultural appropriateness a review was provided by a Maori staff member at the University of Auckland, Department of Psychology familiar with conducting research with Maori participants, and the ADHB Maori ethics committee (3653). The Maori Clinical

Advisor, Werry Centre, Child & Adolescent Workforce Development also agreed to being a co-investigator with his role being to support the cultural safety of Maori participants. That is, he could attend and facilitate introductions and co-facilitate the explanation of the research project and the potential use, storage and collection of the information when necessary. If preferred by the participant he could co-facilitate the interviews or collect the quantitative data verbally.

The Rationale

As identified in the literature and CIDG consultation reported above, there was a strong rationale for investigating the psychosocial experiences of those affected by, and undergoing the genetic testing process for, LQTS. The literature published to date illustrates the medical LQTS body of knowledge is developing quickly and it is therefore important the psychosocial factors, especially those relating to patient care, are also investigated and disseminated with equal pace. It is expected this information will be immediately useful to CIDG clinicians and the services in which they work. Hopefully it will also be useful for other services like it and contribute to the international body of knowledge from which the aims also arose. These aims were developed in consultation with CIDG and the research co-investigators.

The Aims of the Study

1. To describe the medical and psychosocial factors impacting adjustment to LQTS in New Zealand patients and families
2. To compare the experiences of (i) clinicians and patients, (ii) asymptomatic and symptomatic patients, (iii) diagnosed and at-risk patients and (iv) of adolescent and adult patients.

Chapter 3

Methodology

The following chapter firstly describes the qualitative research design, followed by the procedure for recruiting and conducting the interviews, the interview outline and the participant demographics. Lastly, the thematic analysis process is illustrated.

Qualitative Design of Present Study

To explore the experiences of those affected by LQTS a qualitative general inductive approach was used, involving semi structured interviews. This was to allow a picture of LQTS patients' specific emotional and psychological needs, and whether the clinical care they received met these needs, to emerge independently from both the LQTS affected and clinician perspectives. An advantage of qualitative research's inductive methodologies is the ability to generate knowledge of complex processes from which conceptualisations, themes and models of the topic of interest can emerge (Connolly, 2003). This is especially useful when little is yet known about the topic as is the case with the psychosocial factors associated with LQTS (Ezzy, 2002). Also, potentially complex and emotionally fraught issues and experiences can be explored and answers clarified in a relaxed and conversational way (Ezzy, 2002). It was hoped this would also be a more culturally appropriate medium for Maori participants than a structured interview or paper and pencil questionnaires.

Epistemologically, a 'realist paradigm' was used whereby experiences, meanings and the reality of participants is reported (Braun & Clarke, 2006). That is, the experiences of the participants are represented as reality, 'their reality', and as such quotations are used in the Results section with the intention of reflecting these personal participant realities. This does not mean, however, that no interpretation has been conducted during the process of data analysis. In the process of analysing the data, the writer/researcher has interpreted what quoted experiences might mean in the context of the existing literature in this area. The researchers has also been active in deciding on categories and themes, according to her theoretical ideas and beliefs about what each portion of information represents and means in this process. Inevitably, this process is also a product of the researchers subjectivity, beliefs and values. So, while attempting to illustrate the realities of the participants there has also, of course, been an element of theoretical interpretation and subjectivity on the writer's behalf.

The interviews were digitally audiotaped and transcribed verbatim. The transcriptions were analysed using a thematic analysis approach, a method of identifying, analysing and reporting themes within data (Braun & Clarke, 2006). This method of analysing data was chosen for several reasons. In the case of this research project, it was important to the services that the outcomes of the research reflect the 'realities' of the participants as much as possible, reflecting their words, which was interpreted to align with a 'realist paradigm'. Braun and Clarke (2006) assert one of the strengths of thematic analysis as a method is that it can "work both to reflect reality and to unpick or unravel the surface of 'reality'" (p. 81). Also, thematic analysis can differ from other methodologies that attempt to describe patterns across qualitative data (e.g. Grounded Theory and Thematic Discourse Analysis), in that thematic analysis needn't be attached to a phenomenological or social constructionist epistemology (Braun & Clarke, 2006), which may have again been at odds with the realist paradigm requested. Also, this method has the flexibility to be used within almost all theoretical frameworks (Braun & Clarke, 2006) allowing the researcher to make sense of the data within the context of the wide range of available theories.

Eligibility

All individuals 16 years or over who had received a genetic test result identifying a Long QT genetic mutation, and who felt they could recall key events (e.g. receiving genetic test results) and their emotional reactions to them, were eligible for inclusion in the study. They also needed to be being medically cared for by clinicians at Auckland District Health Board (ADHB). Any clinician that was medically or clinically involved in the care of LQTS affected patients and working for ADHB, and at least partly based at either Auckland City Hospital, Starship Childrens Health or Greenlane Clinical Centre, were eligible to participate in the study as a clinician participant.

Procedure

Potential patient participants were sent a Participant Information Sheet, Consent Form, prepaid self-addressed envelope and a covering letter of introduction from the Cardiac Inherited Diseases Registry Coordinator and the department of Paediatric and Congenital Cardiac Services (see Appendices I, II, III respectively). If no response was received within two weeks another set was posted out, for a maximum of two times. When a reply indicating interest was received, the researcher telephoned to go over the study again and answer questions. Potential participants were asked questions to ascertain if they met eligibility criteria (see previous section). If so, times and locations for the interviews were arranged according to their availability and preference. The Maori

Advisor and Social Worker was available to co-interview Maori participants if they chose. This was offered to three participants who each declined the option.

At the beginning of each interview anonymity and confidentiality was discussed again to provide clarity and ensure participants were well informed. They were also reminded they could withdraw at any time and did not have to answer every question. The interviews themselves were begun by orienting participants to the first time they became aware of LQTS impacting them personally. Often this was responded to with a narrative of either their own or a relative's distressing Symptomatic experience. From there the interview attempted to follow the journey in time towards and including the genetic testing process, results and any psychologically salient experiences afterwards. This worked well in practice and the interview outline reviewed in the following section was used throughout as prompts. The outline's main purpose was to offer participants opportunities to speak to these factors at natural points in their narratives rather than losing access to their depth of experience by imposing too much structure with sequential or disruptive direct questioning.

Potential clinician participants were recruited from their respective medical departments directly by telephone and interviewed at a place and time of their convenience. All were consequently conducted at a hospital. Clinicians were first asked to describe their role before being asked to discuss the questions in the clinician interview outline.

Patient Interview Outline

The interview outline was developed to meet the aims of the study and further the literature in the area. The questions, which can be found in Appendix IV, covered topic areas relating to:

1. Factors specific to the condition, including diagnosis, symptomatology, treatments and procedures
2. Experience of the genetic testing process from waiting for the results through to the impact and how they were received
3. Potential psychological indicators of difficulties adjusting or coping from time of initial knowledge of LQTS through to present.
4. The significant supports used and whether they were helpful
5. Experience of CIDG and other healthcare plus level and quality of support provided
6. Recommendations to medical services and peers

Clinician Interview Outline

The clinician interview outline was developed to provide clinician perspectives on the following (see Appendix V):

- The psychological and emotional challenges of those affected by LQTS
- How clinicians saw themselves helping or mediating these challenges and needs, including their view on the best and worst methods of doing so
- What additional factors are still required to better meet the emotional and psychological needs of LQTS patients

Participants

In all, twenty seven names of individuals over 16 years who had received a genetically based diagnosis of LQTS were provided by CIDG. Six chose not to participate, two had incorrect contact details, two lived too far out of the region to access and three felt they would not be able to adequately recall pivotal events in the genetic testing process and so were excluded. Fourteen LQTS affected people consented, representing a response rate of 52%. These 14 were interviewed for an average of one and a half to two hours each (ranging from 55 to 185 minutes) at a place of their choice, predominantly their homes.

Eleven clinician names were provided by CIDG as those working within the ADHB with LQTS patients. Three declined to be interviewed, one because she was overseas at the time. Eight clinicians were interviewed, representing a response rate of 73%. The eight provided a cross section of all the major occupational roles within the Paediatric and Adult Cardiology services. The clinicians were based at Auckland City Hospital (Adults), Starship Children's Health (Children) and Greenlane Clinical Centre (Specialist Outpatient). The medical clinicians (i.e. Electrophysiologists, Pediatric Cardiologists, & Consultant Cardiologists) also provided specialist services to regional services throughout the country. The genetic specialists (i.e. Clinical Geneticist & Genetic Counsellor) also provided some services to national regions but on a more irregular basis. The clinicians were all involved in the care of LQTS patients but to varying degrees; some regularly as part of specialist practice while others only occasionally during a general cardiac clinic. All were practicing at Auckland City Hospital or Starship Hospital and therefore eligible to be part of CIDG. This included the CIDG Registry Coordinator (n=1), Adult & Pediatric Cardiologists (n=4), an Electrophysiologist (n=1), a Genetic Counsellor (n=1) & Clinical Geneticist (n=1) in ADHB.

Interviews were conducted at their respective hospital and were on average 30 minutes duration (ranging from 20 to 45 minutes).

Patient Demographics

Demographic details of the patients are provided in the next table, reflecting their diversity with respect to age, gender and ethnicity. New Zealand population demographics indicate females represent 51% of the population while males represent 49% (Statistics New Zealand, 2002), not dissimilar to the rates found in the current study. In terms of ethnicity, Maori represent 15%, European 79%, Pacific Island 7%, and Asian 7% of the general population (Statistics New Zealand, 2002). The current study is not consistent with these ethnicity representations.

Table 3. Summary of Basic Demographics of Patients

		Number interviewed (n=14)	Percentage of Sample
Age	**16-20 years**	**3**	**21%**
	21-25 years	**2**	**14%**
	26-30 years	**0**	**-**
	31-35 years	**0**	**-**
	36-40 years	**1**	**7%**
	41-45 years	**2**	**14%**
	46-50 years	**2**	**14%**
	51-55 years	**2**	**14%**
	55+ years	**2**	**14%**
Ethnicity	**Maori**	**1**	**7%**
	Maori/NZE	**1**	**7%**
	NZ European (NZE)	**9**	**64%**
	NZE/Eastern European	**3**	**22%**
Gender	**Male**	**6**	**43%**
	Female	**8**	**57%**

Table 4. Clinical Demographics for Patients

	Number interviewed (n=14)	Percentage of Sample
Symptomatic	**5**	**36%**
Adolescents	**2**	**40% (of Symptomatics)**
Adults	**3**	**60% (of Symptomatics)**
Asymptomatic	**9**	**64%**
Adolescents	**1**	**11% (of Asymptomatics)**
Adults	**8**	**89% (of Asymptomatics)**
Diagnosed - Received clinically based LQTS diagnosis prior to genetic testing	**7**	**50%**
At-risk - Received genetic based LQTS diagnosis	**7**	**50%**
Parents	**10**	**71% (of total)**
Symptomatic parents	**2**	**20% (of parents)**
Asymptomatic parents	**8**	**80% (of parents)**
Parents participating with LQTS gene positive child	**6**	**60% (of parents)**
Symptomatic parent with LQTS positive child	**1**	**10% (of parents)**
Asymptomatic parent with LQTS positive child	**5**	**50% (of parents)**
ICD or Pacemaker implanted	**5**	**36%**
Taking LQTS related medication	**12**	**86%**
Average length since received genetic test result	**2.5 years (1.2 yrs - 4 years)**	

Some other potentially important factors are worthy of note. Two of the parents had children with the Jervell and Lange-Neilsen subtype of LQTS that includes congenital deafness and a higher overall risk than subtypes LQTS1-3. Two of the patients had LQTS unclassified variants; i.e., as yet unclassified to a LQTS subtype, plus another cardiac problem. Also, eleven of the fourteen patients had experienced cardiac events and/or witnessed immediate family members' events. The three who had never witnessed an event were asymptomatic themselves and had no Symptomatic immediate family members. Of the fourteen patients seven had experienced the loss of an immediate family member to suspected or confirmed LQTS. A further two had lost a second degree relative such as a grandfather or aunt. Only four had experienced no known, or likely, losses to LQTS.

In respect to the genetic testing specifically, five patients were classified as *probands* as they were the first member in their families identified with a LQTS pathogenic mutation, and nine were classified *cascades,* Asymptomatic family members who underwent predictive testing to ascertain if they carried the LQTS mutation and were therefore at risk. Two did not fit this pattern. One was an exception within the cascades as she was Symptomatic but had been misdiagnosed and was cascade tested after post-mortem testing of a first degree relative revealed a LQTS mutation. The other was an exception within the probands as she was Asymptomatic but a prolonged QT interval had been identified during a routine check up.

In terms of routes of referrals, four of the five Symptomatic patients were referred to CIDG for proband testing by Cardiologists, and one by Accident and Emergency clinicians. Five of the nine Asymptomatic patients were referred for cascade testing after a proband was identified through CIDG, two were referred by Pediatricians, one was referred by a GP, and one was referred after post mortem testing of a relative had revealed a LQTS mutation.

Clinician Demographics

In the process of completing the interviews each clinician was provided an opportunity to describe their role in caring for LQTS patients. In order to retain their descriptions rather than interpreting them, quotes are presented in the table below (see Table 5). It is hoped that by providing the context from which each respective professionals operates we might be able to understand some of the differences that emerge from the data that might relate to the clinicians' roles in caring for their patients. In order to maintain anonymity and confidentiality as far as possible, (especially as there are few clinicians in the data set and in the service), representative quotes from each profession have been used throughout rather than from identified individuals.

Of the eight clinician participants seven identified themselves as of New Zealand European descent (87.5%) and one as British (12.5%). Five were male (62.5%) and three were female (37.5%). Their ages were spread from two between 31-35 years, one 36-40 years, two 41-45 years, one was 46-50 years, one was 51-55 years and another was over 55 years.

Table 5. Professional Roles of Clinicians

Name	What is your role in LQTS patient care?
Genetics	**"My role is to discuss the genetics of LQT, the inheritance, the implications for family members. The pros and cons of testing possible if they are going to have genetic testing what the possible outcomes may be so that we'll either find a mutation, we won't find anything, or we might find an unclassified variance. The implications of testing such as the insurance implications and psychological implications of testing which isn't so much for the affected, clinically affected people, but I guess for the people who aren't, don't have a kind of diagnosis but are found to carry the family mutation."**
	"So I guess my job is to try and help (pause) give my opinion as a geneticist about general policy things, and specific cases. General policy things such as should we be contacting family members directly? Should we be contacting them through the people we've already had contact with and specific cases as in, is this mutation positive or not, and how can we try and improve it, whether it is or not. (pause) I guess it's a learning experience for all of us."
Cardiology	**"So, my sort of expertise on LQT is really just patients who have come through the General Paediatric Cardiology clinics and are having a diagnosis made of Long QT. And then they would move on to the in-house disease service, so it would be at the early stages in diagnosis. So it's, I guess our involvement would probably generally be 'You might have this'. (laughter) We'll do some more tests. And then if you've got it, you'll see somebody else."**
	"I see some LQT patients as the first specialist they're referred to, probably more now through private practice and occasionally now through clinical practice, because often now they'll either be picked up through the Paediatric Service cause they're young people or they may come in because of a collapse and then they may get seen with regard to a possible ICD by other members of the EP service."
	"So I'm one that we obviously have six of us who are Paediatric Cardiologists. Two of us actually also see adults born with Congenital heart disease and then I do quite a lot of cardiac catheterisation and other kinds, I do interventions and that sort of stuff as well. So where I would come across the Long QT people would be usually that I would see a child in the clinic. Sometimes they would come in because they have a parent or another relative who someone knows has Long QT or suspects has Long

	QT and they're interested in looking at other members of the family. And I guess the other alternative is that a child comes in and they have had some symptom that suggests to have Long QT, so we start the investigation with them for Long QT and then, you know, move out to the family." **"Yep we all do visiting clinics around the country so I go from here to [other regions]. "**
	"I work in the adult service, the adult congenital service and then a lot of their Genetic Cardiac Diseases as well, of which LQT is one, but is probably the one that I have the least to do with on a clinical level because they tend to get funnelled towards Electrophysiologists."
Clinical	**No account of role given during interview. LQTS Registry Coordinator, clinical administrator and technician.**

Thematic Plan of Analysis

Braun and Clarke (2006) propose a model of thematic analysis that facilitates the identification, analysis and reporting of patterns or themes that emerge from the data. The process used and described in the following sections is based on this model and includes several important phases, including repeated rereading, review and refinement until clarity and homogeneity within the thematic data is achieved. First outlined is the process for the patient data, followed by that for the clinician data.

Data Familiarization

This involves the researcher becoming familiar with the data through reading and re-reading the patient and clinician interview transcripts. To achieve this, each interview was digitally recorded and transcribed in full by a transcription service that maintained confidentiality. The transcripts were all read and cross referenced with the audio versions to check for any errors. These errors were corrected and some segments initially missed in the transcripts were added.

After reading through the whole data set it became apparent that it made sense to treat the data derived from patients and clinicians separately. That is, the patients primarily centred on their own adjustment to having LQTS and their experiences of being clinically cared for. The clinicians however focused on what adjustment they perceived was required by their patients and what clinical care they provided to aid this adjustment. So, while the broad themes appeared similar (clinical care

and adjustment) the perspectives of providers and receivers differed. Also, one of the aims of this research was to address clinicians' ability to meet the emotional and psychological needs of LQTS patients. To do this the LQTS patient and clinician data sets needed to be analysed separately to enable examination of similarities and differences. The following describes the patient data analysis followed by the clinician data analysis process, for which the same process of analysis was used.

Generating Initial Codes

Each segment of text was systematically and manually evaluated until a broad theme emerged from the main points. These were recorded (coded) in a separate electronic document for later cross-referencing. According to Morse (1994), the above familiarisation and code generating phases facilitate a researcher's maturing *comprehension* of the data. In the following two phases, the researcher begins merging the data in the process of *data synthesis*.

Thematic Searching

Using Braun and Clarke (2006) as a guide, labels representing potential themes were written in pencil on a thematic map and were erased and moved around until a clear picture emerged. Thematic searching aimed to be broad and inclusive, accounting for significant themes while not discarding sub-themes. This level of analysis quickly generated multiple themes with two overarching broad themes, 'Clinical Care' and 'Adjusting to LQTS'. To check the integrity of the coding, consultation was sought for these two main categories at this stage. Forty uncoded statements that I had allocated to either of the two broad themes were randomly selected and provided to an independent researcher (my primary supervisor). Most were coded consistently by the two researchers. One statement was coded contrary to my coding but after discussion it was concluded that the particular statement selected provided insufficient context to be coded correctly. That is, the statement needed to include a sentence more of the rest of the participant's story in order to be interpreted and coded appropriately.

After this process was completed the thematic map was divided into the two broad themes, with a radial diagram consequently drawn for each theme and the corresponding subthemes within each. The labels were reviewed repeatedly, frequencies noted and candidate subthemes developed in each map. The entire data set was then re-read twice to fine tune the themes and to make sure they reflected all the main points covered in the transcripts as a whole. Adjustments to candidate themes

and subthemes were made throughout this process and definitions were also developed to bring clarity.

Thematic Review

A deeper level of synthesis then began, whereby data was reviewed and refined at two levels. Firstly, all coded data extracts were reviewed, such that all the collated extracts for each theme were checked to ascertain a meaningfully coherent pattern. This was depicted graphically on a more developed thematic map. Several iterations of this process ensued. Next, the entire data set was re-read to evaluate the validity of the candidate themes against the raw material.

During the process of reviewing and refining themes, internal homogeneity and external heterogeneity were assessed, as suggested by (Patton, 2002). This was done in discussion with my primary supervisor. One factor discussed was how to best present data from the different subgroups of participants. Having analysed the data a pattern had developed whereby parents mostly spoke about their affected children rather than themselves, irrespective of their own symptomatology or diagnosis. Participants sometimes came from different perspectives depending on whether they had experienced symptoms such as fainting or cardiac arrests (Symptomatic) or had never had a symptom (Asymptomatic). The impact of the diagnosis and genetic test results also appeared different for those Diagnosed (who had already been diagnosed with LQTS based on clinical tests) than At-risk (who mostly did not already have a LQTS diagnosis). As the patient group is relatively small (n=14) it was recognised that some problems could arise in developing robust themes should the sample subgroups be analysed separately (as had been discussed prior to beginning the thematic analysis). Another option discussed was to report the data as a series of separate case studies, but this would significantly increase the likelihood the participants could be individually identified. Participants who were Diagnosed (D), At-risk (AR) or Parents (P) would have an identifier after their name to allow comparisons to be made, especially with respect to the frequency each group was represented in particular subthemes.

In regards to the differing Asymptomatic and Symptomatic perspectives in particular. It was noted these subgroups were sometimes significantly divergent, more so than the other subgroups. The decision was made to trial reanalyzing the themes from these separate sample group perspectives. After some time re-categorising themes separately it became clear, however, that there were more

commonalities than originally thought. The process was again reviewed and it was decided that in order to represent the data and participants effectively the data would best be reported as a whole set but their status as Symptomatic or Asymptomatic would also be identified (A) or (S) in each quote, much the same as the other sample groups. This would allow comparisons between the two groups without losing the commonalities and taking away the validity these give to the data as a whole.

Next, with the themes and subthemes identified, a coding check was conducted that this time included both the two broad themes and the subthemes. Quotes from each of the 15 subthemes under the two themes were selected randomly and put into a separate word document. Definitions for the themes were given to my primary supervisor with this document, including the knowledge that some quotes fit into two themes. Together we analysed the quotes where there were disagreement and identified several types of differences:

1. Three quotes had been coded by each researcher to different subthemes. After discussion, the researchers agreed the quote fit into both themes.

2. In three instances one of the researchers did not have access to the full transcript and information therein that would have provided additional meaning. For example, the following quote wasn't coded to the subtheme regarding Clinicians 'meeting social and emotional needs'. However, had the second researcher had additional information from a later excerpt in that interview referring to this event as having met the participant's emotional need for validation around her own emotional responses it was agreed it would have also been coded to this subtheme.

   > And I'm thinking "oh Christ, thank Christ it's just grommets, there's nothing wrong". And we took her back down and then in walks (the clinician) and you could just tell she had tears in her eyes because it was the second child she had to tell us in the family that was profoundly deaf. *A–D–P*

3. Three were due to simple differences. A subtheme was simply missed or the definition clearly misread by either myself or the second researcher. On review agreement was achieved.

4. Two differences were due to the second researcher having been told that quotes could be coded into two subthemes, when a clearer instruction would have been 'more than one subtheme' (as two had been coded into three subthemes). Changing this instruction and reviewing the relevant quotes produced general agreement in each instance.

5. Two quotes were coded to a particular subtheme due to a lack of clarity with the definition. That is, the definition for 'diagnostic difficulties' had included failures to 'respond to concerns'.

> **DIAGNOSTIC DIFFICULTIES:** Factors relating to difficulties encountered from clinicians/centres for medicine during the diagnostic stage of clinical care, including misdiagnosis and failure to diagnose or respond to concerns

This was too broad and caused over-inclusions from one researcher and potential overlap to other subthemes. It was intended to only refer to **diagnostic** concerns. The definition was consequently refined to that shown below, after which the data was reanalysed using the new definition.

> **DIAGNOSTIC DIFFICULTIES:** Factors relating to difficulties encountered from clinicians/centres for medicine during the diagnostic stage of clinical care, including misdiagnosis and failure to diagnose or respond to *diagnostic* concerns

Clinician Data

The clinician interview transcripts were treated as a separate data set from the patient data so the differing perspectives could be preserved and compared.

The same process of thematic analysis was followed for the clinician data as utilised for the patient data. When reviewing the themes and subthemes in consultation with my primary supervisor, there was agreement for the majority of themes. However during discussion it was agreed some themes were always coding together. For example, 'Challenges to Care Provision: Aspects of providing care the clinicians identify as challenging or problematic when working with LQTS patients' and 'Improving services: Aspects of service provision that are identified by clinicians as needing improvement' were often coded together. This suggested it would make sense to merge these and other subthemes, which led to a discussion and consequent review of the overall theme structure. The data was then recoded to the new theme structure and further consultation on all the clinician data was undertaken. The outcome of this second check revealed a higher level of agreement. Five differences were encountered, resembling those made in the patient data set:

1. Two quotes differed because a theme was missed by one of the researchers.

2. Two quotes had less access to the transcript and contextual knowledge to accurately code to the same subthemes. For example, with the following quote one researcher was aware from the transcript that the 'uncertainties' referred to were clinical in nature and that a 'Genetic Counsellor' provides expert knowledge rather than psychological support (unlike the usual role of a 'Counsellor'). Without this common knowledge/information the two researchers coded to the same subthemes ('Challenges' & 'Solutions') but in different over-arching themes ('Clinical Care' & 'Psychological Adjustment').

   > There are things that might make Long QT different or the same as giving other pieces of information but, you know, I can certainly think of parallels, you know, situations where you have sort of *uncertainties* that you're trying to explain. But I think sometimes the details of the genetics particularly of Long QT are better explained by a *Genetic Counsellor*, you know, I think you need to be very familiar with that information and I think Genetic Counsellors probably have, well I know they do have more skills. *Cardiologist*

3. Raters put one quote into different subthemes. After discussion, it was agreed it should be coded into both.

In summary, these processes produced two themes for the patient data with nine and six subthemes within each. It also produced two themes for the clinician data, with three subthemes within each. These will be presented in the following chapter, before findings will be synthesised and recommendations for CIDG and other Cardiac service providers in general (including GP's) are outlined in the final chapter of this dissertation.

Chapter 4

Results

The main purpose of this study was to better understand (i) how people psychologically adjust to LQTS and receiving genetic test results and (ii) how staff in New Zealand's LQTS specialist service perceive and meet their psychosocial needs. In analysing the interview data it became clear an individual's response and adjustment to their genetic test result depended upon several factors beyond genetic testing. These factors are reflected in the two overarching themes and the subthemes that emerged from the twenty two interviews.

The Themes

The thematic analysis of the data from the 14 LQTS patients produced two themes:

Clinical Care

Aspects related to the service or care provided by centres for medicine or clinicians

Adjusting to LQTS

Aspects that are challenges to, or impact upon, potential psychological adjustment to the condition

The two themes produced seven and nine subthemes, respectively (see Table 6). These will be discussed later in this section. The same two themes also emerged from the data provided by the clinicians. The definitions of the clinician's two themes both incorporate the three subthemes for each (see Table 6):

Clinical Care

(i) Aspects of providing care the clinicians identify as challenging or problematic when working with patients affected by LQTS, (ii) what the clinicians report they do to try and alleviate these difficulties and (iii) what further aspects of service provision would be necessary for improvements in clinical and medical care to take place for Patients

Adjusting to LQTS

(i) What clinicians perceive are the emotional and psychological challenges LQTS patients are affected by, including (ii) what clinicians do to support resolution of these

problems and aid adjustment, and (iii) what the clinicians identify would be helpful for the adjustment process but are not currently provided.

Table 6. Participant Themes and Subthemes

	Clinical Care	**Adjusting to LQTS**
Patients	**Appreciation** **Clinical Systems** **Information Provided** **Genetic Testing** **Meeting Social & Emotional Needs** **Diagnostic Difficulties** **Recommendations**	**Response to Symptoms** **Impact of Diagnosis** **Functional Restrictions** **Coping** **The Family** **Confusion & Uncertainty** **Response to Clinical Interventions** **Burden on Others** **Mortality**
Clinicians	**Challenges** **Solutions** **Recommendations**	**Challenges** **Solutions** **Recommendations**

To represent the prevalence of the patient and clinician themes, the descriptors recommended by Braun and Clarke (2006) of *all, many* or *several* participants have been used. For the patient data, descriptors of the prevalence are also sometimes given for Symptomatic (S) or Asymptomatic (A), at risk (AR) or diagnosed (D) and parent (P) or dependent free (DF) when/if the sample group is over-represented or it is especially relevant to a theme (see Table 7). These descriptors are also used at the end of each quotation to provide additional context. The clinician data also has quote identifiers reflecting professional roles (i.e. Cardiology, Genetics, Clinical) for context. The quotations are used to illustrate the meaning of each of the themes and subthemes, and are drawn from a cross section of participants. Patient names or pseudonyms have not been provided in order to protect the confidentiality of this specific patient group.

Table 7. Patient subgroups and in text abbreviations

Patient Subgroups identified by		Abbreviation in text		Abbreviation in text
Symptomatology	**Asymptomatic**	**A**	**Symptomatic**	**S**
Diagnostic status prior to genetic testing	**At Risk**	**AR**	**Diagnosed**	**D**
Parental status	**Parent**	**P**	**Dependent Free**	**DF**

As the themes from the patient and clinician data sets are similar, the remainder of the chapter reports them in order of theme first, with the patient and clinician data sets reported separately therein (see Table 6). This makes it possible to conceptualise the themes and subthemes both across patient and clinicians and within. The variations between the participant data sets, and subgroups within each participant group (e.g. Symptomatic patients), are commented on at the end of each theme's section.

Clinical Care

Patient Themes

The seven subthemes that emerged from the patient perspectives were Appreciation, Clinical Systems, Information Provided, Genetic Testing, Meeting Social and Emotional Needs, Diagnostic Difficulties and Recommendations.

Appreciation

This subtheme was centred on the aspects of clinical care appreciated by patients. Many patients were very appreciative and grateful to clinicians and to the wider health system for covering the costs, their advocacy and potentially saving their or their relatives' lives.

> And I'm glad you know. It's so nice that it's happening here in New Zealand like where you do get the help. I mean they've just been great, the Hospital and all that eh. I mean I talked to the Cardiologist when she got this defibrillator put in, and I said to him how much would

this cost in this operation and he said oh probably about fifty thousand I seem to remember and you know it was absolutely free eh, you know public health. *A-AR-P*

I'm just so grateful that hey there was something we had and the family was being looked at, even though it did take, in the end its worked out alright, no one's died you know. *A-AR-P*

I think through their, through their talk and recommendations and the fact that the (CIDG) specialist was pushing for my daughter to have a defibrillator. You know he, it did, it wasn't done privately. It did come through the public system. So he was, he was you know pushing for it. So no he was he, I don't think he could've done any better for us to be honest. *A-AR-P*

Clinical Systems

Relates to aspects encountered as a function of effective or ineffective clinical systems. Many patients were impressed with the efficiency of CIDG clinic systems and processes.

That was actually very, very good. I was just in and out of there. In the old days, golly, you were there for half a day, you know, waiting in this massive line and then you know. I'd see a different doctor every time I went in. And yeah I do remember the last time I went in. It was very, very efficient. *S-D-DF*

The, they didn't keep me waiting and you know I went straight in and you know I was made to sort of feel that you know, that they cared there as well. *A-D-P*

However several encountered delays due to documentation difficulties within CIDG, demonstrated by losing files, samples and forms.

Well that was done relatively recently I'd sent the bloods off, oh no actually when I was in [specific] hospital they took blood. They told me about the study, filled in the forms, they took blood and went away. I never heard anything about it, so I asked my Cardiologist and he followed up and they said "Oh we seemed to have lost that one" so they didn't have any records of me having filled in the forms or anything so I had to do it all again and wait for months and months. *S-D-DF*

> This is all, I should tell you that all this should be on file. I did have a file for everything but that was all lost. All my files were lost. They had to do everything again cause they lost the files. *S-D-DF*

Several were unimpressed with the system in place for Symptomatic LQTS patient births. All three female Symptomatic participants that gave birth after receiving their LQTS diagnosis had a planned Specialist assisted birthing strategy that the on-call Specialist failed to get to in time for the delivery. Two of these patients had implanted devices that were recommended to be switched off during delivery to prevent harm to mother and baby.

> I told them I said "look you need to get the Obstetrician and I feel like I'm really close to having this baby". And they said "oh no we don't get the Obstetrician in until, you know, right before you're going to have the baby and you're, you know, hours away". Ten minutes later I had her and she just came like a shot out of nowhere with no pushing and I was like I told them "get the Obstetrician, get the Obstetrician". *S-AR-P*

> And she's going "hang on, hang on you can't just have it" and I said "sorry!". And by the time you know the Heart team came down, too late, I had already delivered him. They go "oh my God you're not supposed to do this". And they were afraid because something might have happened to him while I was having him you know when the device did go off. *S-D-P*

Information Provided

This subtheme is concerned with the quality of information provided to patients in written materials and in explanations about concepts or processes, including instances showing insufficient clinician knowledge of the condition. Four Asymptomatic patients who were exclusively provided information by CIDG found the quantity and quality of information provided was entirely adequate for their purposes.

> No, the information that (CIDG) sent out was pretty extensive and as long as you read it, you know you, there was the understanding of it was pretty clear really. *A-AR-P*

Conversely, many patients found the information they received somewhat insufficient, augmenting it with additional information from the internet. This pertained to gaining an understanding of the condition, risk perceptions and treatment from CIDG or primary and secondary regional services.

> Yeah, yeah I think I've been given information. I've looked at a lot myself on the Internet and things as well. I think initially when I was diagnosed there was nothing offered. Like I was pretty much was told you know "you've got this condition see you later". *S-D-P*

> I sort of kept doing a bit of research on the internet and looking up things and printing off lists of stuff I shouldn't have. You know medications I shouldn't have. *A-D-P*

Many were concerned clinicians outside CIDG had insufficient knowledge of LQTS, especially General Practitioners (GPs).

> Yeah I think definitely the Doctors, I have to say your GPs, GPs just don't know anything about this condition. *S-AR-P*

> So not all the GP's know really. They don't have a clue about what it is. But she didn't kill me anyhow, so that was alright (laugher). *A-D-P*

Genetic Testing

This theme related to genetic testing, including deciding whether to get tested, waiting for and receiving genetic test results.

All the patients reported frustration with having to wait a long time to receive their genetic test results but many understood the delays as they had received an explanation.

> I don't know whether that happened first, but certainly the results, the genetic results, took a long time to come through. And my sister and her family found it was taking a hell of a long time. Yeah, they were really frustrated about the whole process actually, and I mean if there's one complaint I guess it's that it took a long time for the test results to come through, but I understand it's really expensive and it's hard to do so you know. *A-AR-P*

> But it was, yeah like I say it was a good while later than they said till we got them back and at that time it appeared I think that funding had been made available and things could happen at a lot more faster pace. *A-AR-P*

Several others however reported feeling uninformed during the genetic testing process, expressing their dissatisfaction.

> Yeah, quite a while for the genetic blood results, I can't remember exactly but it was quite a while, yeah it was about six or eight months. I rang them up a couple of times to see how long they would take, yeah, but I didn't actually speak to these people here I always got the Lab people. I just wanted to know why it took so long. *A-AR-P*

Several reported they were glad they had decided to be genetically tested, as it provided clarity about the condition that affected them.

> I'm glad I did it. At least then I knew what I was dealing with. *A-AR-P*

Several parents discussed concerns associated with deciding whether to proceed with predictive genetic testing of their children. They discussed preferring to avoid and delay knowing the diagnosis for fear it would be positive and beyond their or their children's ability to cope.

> I'm like I keep going to get it done I'm like "oh I'm, I'm just not ready yet" cause I keep thinking "what if they come back and they say that she's got it". *S-D-P*

> I thought I'll let my kids make those choices later on, when they're older. And with the reaction of my younger one who feels he's got it and no one else has got it, you know, just the way his whole, you know, just the way his whole personality's changed really quickly. I thought, oh God, I'm not going to put that on him either. *S-D-P*

Several parents discussed how fear of being told you could die, or would have your options limited in the future, were reasons why their children didn't want to be genetically tested.

> So, you know like if my son wanted to go into the army or the air force, he couldn't. If he was tested and it proved positive so he said he'd rather not do that. *A-D-P*

> Like my twenty year old when he goes 'I don't want to know, I don't want to know that I might have what you've got and could die'. You know, what can you say to that? *S-D-P*

Meeting Social and Emotional Needs

This theme was concerned with the care provided by clinicians in meeting individuals' social and emotional needs, either through larger collective clinical/hospital systems, referrals or individual clinician factors and communication styles.

Many felt CIDG specifically did a very good job at meeting their emotional needs, several identifying the Registry Coordinator or a specific Cardiologist as trusted, approachable and responsive supports.

> And I used to tell [Registry Coordinator] and them, cause she was always there when they, I've always sort of felt comfortable with [Registry Coordinator] because she was there and I'd go "oh good". She's someone I trust. *S-D-P*

> What worked best was being to able to contact [Cardiologist] for a start, you know, because he was the person that knew exactly what the condition was. We had lots of faith in him. *A-AR-P*

All the patient participants stated preferences in terms of the communication style that best suited their needs. Many valued an approach where they were related to in an honest and open way.

> But [clinician] actually said to me one day "Well, you know, we're really scraping the bottom of the barrel with you". And I thought I like this guy, you know, he's very honest and, you know, he kind of wasn't bullshitting me about "Oh don't worry we've got plenty of things we can try" he just said "shit" and I quite liked that about him. *S-D-P*

> So he was he was really good to talk to I really enjoyed that experience cause he was, he wasn't like, so much like a Doctor in that he's an older guy, experienced and just you know it was like just like talking to a bloke you know what I mean? Which is quite good. *A-AR-P*

Many did not value an overly direct, inflexible or dismissive approach that came across as detached; lacking warmth and empathy. In one case this was combined with denying access to an alternative professional opinion and felt like bullying.

> So he was very, he was a lot more direct and, and a little bit more frightening if you know what I mean. *A-AR-P*

> Like I was pretty much was told you know "you've got this condition see you later". And I remember I had a big breakdown to [clinician] and he's, I mean he's a great man but he was like looking at me like 'what are you so sad for, you know, what's your problem?'. *S-AR-P*

> That would be my sort of feeling that he might have been a bit abrupt and a bit sort of dismissive (pause), and our daughter doesn't like him at all. I've always found [clinician]'s attitude, I didn't warm to him at all eh? *A-AR-P*

> With [my daughter] we felt quite bullied like you know. No, not by [clinician] I guess by [another clinician] because you know we weren't allowed to talk to anybody, we weren't allowed to talk to a cardiologist. Everything had to go through [clinician], she needs to have this medication you know basically whether you like it or not. *A-AR-P*

Part of providing clinical care is making referrals where appropriate so that is included in this section. Psychological support itself was assigned to the 'Adjustment to LQTS' theme and reported therein. Many spoke of being offered a referral to someone to talk to for psychological support if they opted for it, which many did not because they felt it wasn't needed, even though several described having experienced significant psychological difficulties at another point in the interview.

> I think they just said like if there's anyone you need to talk to just tell us and we'll got someone in to talk to you and stuff like that. Yeah, they were real nice. *S-D-DF*

> Well they did say did I want grief counselling or something at the time, but I said I didn't. I mean yeah, I didn't, I didn't need that so I guess you know they offered me something and I refused it. *A-AR-P*

One Asymptomatic patient who'd received genetic testing four years ago said they would have liked to have spoken to someone if a referral had been offered.

> I reckon if they had someone, they made an appointment for me to talk to someone, it would've been good. You know somebody to talk about things and how it affects me, emotionally and, you know, and cause I think it affected me quite a lot in that, I changed. I wasn't the same person anymore. *A-D-P*

Several felt there was a lack of follow up support provided from CIDG and other regional services.

> I mean you're sort of told in a very clinical kind of setting and then you're like "okay see you later" and that's it and that supports not extended out into the community or into the home. And yeah, that's where the downfall is I think they're great at what they do medically but yeah socially it's, the supports not there. *S-D-P*

Diagnostic Difficulties

This theme reflected the factors relating to difficulties encountered from clinicians/centres for medicine during the diagnostic stage of clinical care, including misdiagnosis and failure to diagnose or respond to diagnostic concerns.

Many recounted intense frustration that primary and secondary service clinicians didn't appear to believe their health concerns, failing to diagnose and dismissing them.

> Yeah so about 8 years ago just over 8. And so I went to the Doctor and they just checked me over and they said "oh you seem okay now". And you know and I told them about my sister but they just kind of, hypochondriac, you know, "you don't have that". *S-AR-P*

> …and you know once one professional tells another professional that I was a bit paranoid, that there's something wrong with [me] then, you know, its always believed amongst the medical profession and never the mother, you know, and I think that's just wrong. It should never have been missed. The whole hearing thing should never have been missed. *A-AR-P (Failure to complete an adequate hearing assessment for Jervell Lange-Neilsen Syndrome diagnosis despite parents repeatedly expressing concerns their toddler could not hear).*

Several others reported feeling angry at having received prior medical checks and LQTS not being identified.

> No, there was one in [suburb]. I remember specifically and she said, and it actually read it on there that I had this Long, Long QT and when I went back to her later on and said 'why didn't you do *something* about it then'. Because you know they'd been giving me medications and all sorts that I shouldn't have been having. She said 'I didn't know what it was so I just left it'. I was so angry. I never went back again. *A-D-P*

> I was crying all over the place, angry, I was really angry. I think I was angry because the Doctor never picked it up years back. *S-D-P*

Several found it distressing that their primary or secondary service level clinician had failed to immediately inform them of their LQTS diagnosis.

They do their rounds and I said to her "so do we have a diagnosis, do we know what it is" and she said "of course we do, we've always known what it is, its Long QT Syndrome". That was the first thing LQTS, the first thing heard about and then after a while you know cause then I just sat there and bawled you know cause you know it was quite emotional. *A-AR-P*

Many of the patients and/or their relatives had been misdiagnosed. For several this had caused significant medical complications and for two resulted in tragedy. All had been misdiagnosed with epilepsy.

The [hospital] couldn't pick it up you know they kept saying either I was on drugs and that. I was an epileptic and, you know, sent me home. Another baby came along and of course with each baby I had they seemed to get longer. *S-D-P*

Through when our younger daughter [name], when she collapsed and died, fifteen years ago and then of course she was on life support for a week. That's how we found out what she was wrong with her. An ECG was done for her you see whereas she, they, she was diagnosed as being an epileptic. *A-AR-P*

She'd experienced seizures pretty much from the age of 8 and they diagnosed her with epilepsy, and put her on to epilepsy medication which she kept having them, and then she passed away. And they still thought it was epilepsy at the time, and then they did an autopsy and said that it was probable LQT which we didn't know anything about it. And it was a few months later the Doctors sent some information and things and said this is what we think she died from we need your family to have ECGs done. So we had them done and sort of nothing came back. *S-AR-P*

Yeah and we got there and they were absolutely disgusted with how [a hospital] had handled it the whole, the whole thing. And they said that the shot that she had been given or whatever she had been given to relax her body for the plane trip should have been given right at the beginning to stop the fitting. Because what had happened was her brain had swelled, all her organs had swelled from the fitting just because she hadn't rested that time. And so they had her on drugs and a ventilator and things to keep her alive and yeah about 3 o'clock in the morning or so they said she's not getting any better she's not reacting and they said that the chances were that she wouldn't come out of it herself. She was in a coma but that she would

be brain damaged and have extensive organ damage as well so if she would live, she would be in a wheelchair for the rest of her life and that she would most probably be in a state of vegetation as well. So they gave us the option of turning off the machines and drugs and things. Yeah once they turned off everything it only took about 10 minutes for her to. It was when they turned off the respirator and that that. It was more the treatment that she'd had leading up to it that had caused the damage and of course the medication cause she was on [epilepsy medication] or something, it was quite a, which is something you can't have when you've got LQTS so. *S-AR-P*

Recommendations

This subtheme concentrated on the additional aspects of clinical care patients feel they would benefit from primary and secondary centres of medicine and CIDG specifically.

Many of the patients reported they would have benefitted by a speedier genetic testing process because of the adverse emotional toll the waiting took on them and their families.

> I think if they had had the time, you know, and the funding you know, the time to just shorten it all up. *A-AR-P*

> So it would have been nicer to have it happen in the timeframe it did from when I was diagnosed to when the children got tested and that was quite quick, well quick as far as the hospital was concerned, like a month or so. *A-AR-P*

Several young people suggested it would be good if clinicians organised LQTS patients to meet together and talk.

> Maybe just someone, like, doctors or someone give their input and, like, maybe ask someone else that's had the experience to talk to you. To talk to other people because I'm sure other people haven't had that opportunity that's had the same thing happen to them so, yeah it's good to talk to someone who has it. *S-D-DF*

Several also suggested an improved organisational system; that prevents files or forms from being lost, to prevent additional delays.

> Yeah, so, all my files were basically lost so they need to sort that out. Get some better systems happening so they don't lose stuff like that. *S-D-DF*

Many Asymptomatic patients spoke about how useful it would be to have a diagram to help explain the condition to others.

> A simple diagram. Rather than just words cause you know or you know what I'm saying? If they say this is what happens or three or four, this is your heart, this is what normally happens; this is what happens with you. *A-AR-P*

Several patients suggested GPs in particular would benefit by further education on the condition, in terms of diagnosis, medical management and seeking support when necessary.

> Yeah they'd be better educated but also in following up. I mean like people like myself coming in and talking about their family history. When they know it's there but they dismiss it. I think they need to take it a bit more seriously and you know even if they think 'well it's really not that' to be, it's better to be safe than sorry. So yeah just more education to GPs for that initial diagnosis because they're the ones who refer you on to the Specialist. So if you can't get past your GP to be seen then you're not going to get any treatment. *S-A-P*

> And a misdiagnosis I mean it still goes on that people are diagnosed with epilepsy so I guess just not to for there not to be a fear for the GPs you know that they're seen as being incompetent if they say "well I'm not sure what's going on". Rather than having to give a diagnosis if they just say "well I don't know what it is" and having you know having the network of people that can support them. *S-AR-P*

Clinician Themes

Three subthemes that emerged from the Clinician data have been defined as the 'Challenges' they face in caring for those affected by LQTS, the 'Solutions' they currently have to assist or combat these and 'Recommendations' they make for improved service provision in the future.

Challenges

This subtheme concentrates on the aspects of providing clinical and medical care which the clinicians identify as challenging or problematic when working with LQTS patients.

Many Cardiologists reported that diagnosing LQTS appropriately is a problem outside CIDG, potentially because it is a complex & rare diagnosis that can often resemble other diagnoses, especially epilepsy if seizures are present.

> There is a lot of misdiagnosis and think that's, I think that some of that is understandable because it's a difficult diagnosis to make. *Cardiology*

> I think there is a little bit of, when you deal with a rare diagnosis you became a sort of expert in terms of that and then when you look back along somebody's history it can all seem very you know obvious to you, but. But it is a rare condition and for people like, you know, family practitioners or people who don't deal with this all the time I think it can be really rare, you know, really hard. *Cardiology*

Several clinicians also held reservations about the potential impact of educating the community, especially in relation to current resource limitations and projected figures should more referrals be made.

> Yeah so more awareness in the community and more information out there would actually sort of help it as well. But having said that if you put all the awareness and information out into the community you've got to have the hospital system sorted out ready to go to take the influx when it comes in. We've been so scared of that, we haven't really ever wanted to go there with the other stuff too readily to be honest. *Clinical*

Several clinicians were concerned there was insufficient follow up of patients, potentially putting patients at risk, especially if they didn't adhere to their medication.

> Like many of my colleagues, and I think this has been an international habit, make the diagnosis of Long QT Syndrome and say "Take the Beta-blockers, off you go, bye-bye". And leave it with them and the GP maybe. Few months later they're off the Beta-blockers and it's all forgotten. *Cardiology*

Many were concerned about possible ethical issues inherent in genetically testing for LQTS, often stating conflicting opinions around whether it should be done and at what age. The argument appeared to hinge on the interpretation of 'medical benefit' and whether genetic testing for an individual would provide this.

> Now, around the western world, we tend to try and not to do genetic testing in people where there may not be a medical benefit, and till they're adults and can make up their own mind. I haven't really decided with LQT, at what age, you know? If you got a competitive 12 year old, it is not unreasonable to test there. You actually need to get as many young people as you can tested [pause] because it's quite a contentious area. *Genetics*

> You can't be absolutely certain that what you've done was the right thing, you obviously do what you think is the right thing, but sometimes it's not, it wasn't the right thing. It's more controversial rather than a clear cut disease where the disease is going to be this, this and this and death is going to be at this age. *Cardiology*

Many clinicians spoke of limited time hindering their ability to emotionally support Patients.

> I mean I guess the difficulties that people have, you know, in terms of the impact of the diagnosis they, there's sort of acute moments when we're talking to them in the clinic, if you need to change that much it'd be nice to have a bit more time you could talk to them in a bit more detail. *Cardiology*

> The problem is that person [psychological support] hasn't been in the meeting with them, so hasn't sort of gone through the process of being involved in the testing, so bringing in another new person just to say "How are you doing?" is probably not ideal, you really want it to be someone who's been through the whole thing with them, and then we're onto the next patient. And the clinicians are going to want to, and the clinicians have no spare time to be hanging around while all this other social stuff is going on. *Genetics*

Several also suggested even if the time were available to provide emotional support Cardiologists may not be the ideal people to do it.

> They like interventional procedures, they like helping people in that way but I haven't met one cardiologist who enjoys outpatient clinics. *Clinical*

> The cardiologist's got fingers in many different pies, and he and the other cardiologists are just too busy to spend time and to be honest, I don't think they have the patience. I'm not sure that I will want to be going through all that kind of stuff either, even if there was time, there's going to be some set of things generally that should be done separately. *Genetics*

Solutions

This subtheme centres on what the clinicians report they do to try and ameliorate the problematic aspects of providing clinical and medical care to patients. A lot of these are mirrors of the aforementioned problems as often the clinicians came up with potential solutions to the problems they discussed. Many clinicians talked about the importance of helping patients, and local medical professionals potentially involved, understand the complexity of the condition and manage it appropriately by sending comprehensive follow up letters.

> Yeah you're sort of highly likely to be affected or likely to be affected you know there's all these things that have been quite challenging to get my head around so you think oh gosh these poor patients how do they … so we, I explain what an unclassified variant is and I think most people get their head around that and then. And I write everything in a follow up letter so you know if they go away and try and explain it to their family and get themselves all tied up in knots they'll get a letter from me within a couple of weeks so I think that helps. *Genetics*

> And then again keeping other people in the loop, because although this would be a sub-specialty service you've also got their own GP, who's not gonna be all that familiar with things and needs to be involved. Their own cardiologist, they might live in Hamilton or Wellington, or could live anywhere really, who's not that familiar with this diagnosis. So correspondence, there's dealing with the Pathologists to get the diagnosis right and so forth. So, there's quite a lot of issues there, but yeah being open and available and repeatedly available. *Cardiology*

Many spoke of the advantages there are with CIDG having a multi-disciplinary team of complementary specialist clinicians in place to respond to patients' medical needs. This includes being effectively diagnosed and treated by Cardiologists; coordinated, supported and followed up by clinical staff; and having the genetic results and ramifications explained by the Geneticist and/or Genetic Counsellor.

> I mean a lot of this has already happened in the last few years, so for example having people available that are specifically dedicated to it. So even having the Electrophysiologists (EP) service that doesn't exist in many other hospitals for example is good. Then having this EP

> service that sub-specialises with certain individuals with particular interest in LQTS. *Cardiology*
>
> I think us being involved as a Genetic Service, so that cascade testing of these unclassified variants doesn't happen without our involvement or yeah, is a good thing. I think maybe yeah we're kind of used to dealing with unclassified variants where other Hospital Services aren't really. *Genetics*
>
> But you know who else would someone ring anyway, you know and to try and actually sort out a school camp or something like that. You sort of, these roles I think are necessary, they're sort of interlinking cause you can get stuff done quickly and efficiently. *Clinical*

Many reported CIDG now provides a specific pathway for referrals of patients through their newly established Arrhythmia Clinics. This allows the use of a multi-disciplinary specialist team and an efficient clinic process for patients and referring doctors alike.

> Yeah, we're booking everyone just in the Arrhythmia Clinic (CIDG) now. I think before that I may have tested one, two people in the general clinic but you want to be testing them and also have an ECG that's performed on them and read by someone who's experienced in reading the ECGs. And I think there are not that many people in New Zealand who, my understanding is you need someone who really knows what they're looking at. So it's just the perfect place to do it if they can have an ECG then we see them with the ECG report and the cardiologists can work out the QT interval and then you talk about testing knowing their QT interval rather than not. *Genetics*

Several clinicians discussed the role of genetic testing as a useful diagnostic tool that can add certainty around a clinical diagnosis and therefore determine which treatment recommendations can be made.

> If they've had an ECG and it's been reassuring that they're already reassured because of that as they should be because that's one of the biggest indicators about whether they're going to have a problem or not. And if it's not well then that's almost diagnostic in itself if it's in the affected range and I guess if it's in the grey zone then again the QT, the genetic test results will help give them some certainty around that. And I guess then if, if the cardiologist is concerned enough about something about their QT and they've got a positive gene test result

> then they may or may not medicate them on Beta Blockers depending on whether they think it's warranted or not. *Genetics*

Several spoke of measures CIDG have taken to improve processes within communities outside the main centres for medicine. This has been primarily done by gaining national ethical approval to make the genetic testing process standardised across NZ.

> Yeah no, so yeah, a lot of the cases are like that and then I get odd ones where somebody doesn't want to take consent and upset the patient, so they just would rather test without going through the proper process, and a lot of the stuff I get from out of town is not good, and that's why now we've got national ethics where we actually want to tighten up the processes and make everyone do the same thing. Because we can enforce it now and because they can actually use the forms, they can do the same as everyone else. *Clinical*

Many clinicians also spoke of how useful the Registry Coordinator's role was in being available to do follow-up support and for CIDG patients to access and have concerns addressed responsively, directly and quickly.

> But I think you know, I mean, now that the CIDG system if you like is in place, and particularly with the coordinator, I mean that's an enormous support to people too and so a lot of people now will ring the coordinator up and so it's obviated, I mean there wasn't anything like that before. So I think that deals with a huge amount of day to day concerns. *Cardiologist*

Several Cardiologists reflected on a particular personal communication style that they've found most helpful to patients, one that involves responding respectfully.

> I can very easily by the tone of my voice or a very slight inflection say to you "That's a fairly stupid question you've asked". I don't have to say that, but a perceptive person will pick up on that straight away and you might think it's a stupid question, but you have to just step back a bit and say, "Hang on, I'd better not, I think this is a stupid question but let's just deal with this and sort of take a little bit of time and sort that problem out". *Cardiology*

Recommendations

This subtheme represents the aspects of service provision that would be necessary for improvements in clinical and medical care to take place for patients.

Many clinicians discussed the need for a speedier, localised genetic testing process.

> I guess one of the other things that I haven't touched on is that with the testing thing. Initially the genetic testing for LQT was going to be done in Auckland and then now it's been done in Denmark but we haven't had results back in eight months. And I think that's the other reason we haven't had continuity yet. People that I saw to consent to testing last year we haven't had a result back yet. And so yeah and Denmark hasn't communicated well to us and now that we're going to use Melbourne and then hopefully eventually the testing will be up and running in Auckland. So I think one thing is you know we want a service where we can offer people tests and have results back in a timely manner so maybe three months for the proband, six weeks for the cascades and that they are actually done in that time frame. Because I know that it really it's awful for these patients when you say your results will be three months and nine months later the results aren't back. *Genetics*

Several also spoke of needing more specialists to meet demand, especially a Genetic Counsellor

> I'd like to have more free availability of genetic counsellors. *Cardiology*

Several also suggested psychological support is required, especially in identifying those at risk of psychological difficulty.

> I think like in the adult Congenital Heart Disease service they rock up through the door and they sit down and they fill in their questionnaire, which is sort of a stress and anxiety type of score. And the ones that score high get offered the counselling. I would love to have that as a routine. That's probably the biggest gap. *Cardiology*

Several recommended more regular Arrhythmia Clinics to meet demand and provide more time.

> I'd like to see more arrhythmia clinics. *Clinical*

Several recommended additional training be given to general medical staff, including GPs.

> Yeah and more education, more education for Doctors and nursing staff but also GPs dealing with families like these. *Clinical*

> I would like to have junior doctors who are being trained up properly in inherited Heart Disease and there's no, if you look at the way Cardiology Services are developed you've got Arrhythmia Specialists. You've got Interventional Specialists, you know, people in stents and things. You've got Echocardiography Specialists and then you've got Congenital Heart Specialists. But there is no such thing as an Inherited Heart Disease Specialist, there's no such thing as an Inherited Heart Disease training programme in Australasia. So, we need junior doctors and senior doctors who are trained and understand genetics, genetic heart disease and how to manage them and are prepared to work with a team. *Cardiology*

Several Cardiologists recommended a nationalised service that was centrally funded to provide a consistent service to all areas in NZ.

> I would have a nationally funded resource. I would have it centrally funded. It wouldn't be something that would be up to the local hospital to manage it, you know. Timaru is not going to have the resources to do that, so there should be a nationally funded resource so when the Cardiologist in Timaru phones ups, you say "Sure, I'm going to send you a counsellor, off you go". And the counsellor pops over and sees them in their car, you know what I mean? *Cardiology*

Similarities and Differences

Information

Many patients spoke of having to look elsewhere for additional information. Asymptomatic patients particularly indicated the information provided was sufficient. This would indicate the information was insufficient for those perhaps needing to do more risk evaluations and cope, for example, with the additional stresses of cardiac events. This was an area of service provision CIDG was interested in hearing about during the consultation phase interviews. It was not raised however, during the clinicians' interviews.

LQTS in Regional Centres and GP Surgeries

Both cardiologists and patients discussed their experiences and concerns at the level of current knowledge about LQTS and misdiagnosis in general, and of epilepsy specifically, in New Zealand medical communities outside the specialist services of CIDG. Clinicians were aware of the lack of knowledge of LQTS given the rarity of LQTS in the population and the limited exposure clinicians from smaller centres would have to it. However, the consequences spoken of by the patients powerfully illustrate the significant risks. It is understandable that some of the patients expressed extreme anger at not receiving an accurate diagnosis initially if ignorance, and perhaps a lack of diligence, is a reason for it, and an even higher risk of sudden death is the potential consequence. The clinicians identified that the best solutions were education and training in the community and supporting their community colleagues by sending comprehensive follow-up letters to help ameliorate any lack of knowledge. They have already gained national ethical approval to implement standardised systems for genetic testing nationwide. Both clinicians and patients recommended additional training on LQTS for medical staff outside CIDG, especially GPs. Several Cardiologists also recommended a nationalised service to provide continuity and consistency in standards between centres for medicine irrespective of their local funding opportunities. Pragmatically, the Cardiologists also voiced some concerns that should nationalised education occur CIDG resources would be stretched beyond capacity with the influx of referrals.

CIDG Systems

Many patients were impressed with the efficiency of the CIDG Arrhythmia clinics. The clinicians reported focusing on trying to make these effective and efficient; utilising the skills of the multidisciplinary specialist CIDG team. It appears CIDG are achieving their goals for the clinic and that their recommendation that they have more clinics may only improve things further. However no mention was made by CIDG about improving the system for safely storing files and forms that patients indicated was a problem.

Specialist Assisted Births

Symptomatic patients indicated the services for those giving birth were ineffective as the Specialist failed to arrive. The clinicians did not comment on this, perhaps because they are not involved in this aspect of patient care.

Clinicians Providing Social and Emotional Support

Many patients preferred a particular approach when it came to feeling supported and comfortable with their clinician. The preferred honest and open approach patients could relate to was generally also consistent with what clinicians described as the most effective. Some clinicians however did not come from this approach; instead behaving less empathetic, being busy and detached, which was distancing and experienced as unsupportive. Many clinicians suggested Cardiologists were perhaps neither the best suited to, nor interested in, spending time behaving in an emotionally supportive way. Some Cardiologists felt they had insufficient time to provide emotional support to patients. Perhaps if their recommendation for additional Arrhythmia Clinics was met this would improve their ability to allocate time to supporting those affected by LQTS. Many patients felt CIDG did a good job at meeting emotional needs and the Registry Coordinator was consistently identified as an important individual support for many, as was also identified by several clinicians. Many patients and some clinicians felt continuing this support after leaving the clinic through community services would be very helpful, such as GPs or Community Social Workers.

Accessing Psychological Support

Many patients reported routinely being offered psychological or grief counselling support if they chose to accept the offer. Many chose not to accept support, however, despite illustrating a need. The patients who did accept psychological support found it very helpful, reporting decreased distress after using techniques learnt from the Psychologist referred to. Clinicians agreed psychological or counselling services were needed for LQTS patients. Ideally, they would have psychological support in the form of a psychologist as part of the CIDG multidisciplinary team and include psychological screening to identify those at risk of, or experiencing, significant distress when in contact with patients.

Genetic Testing

All of the patients and many of the clinicians understood the adverse effects of extensive waiting times for genetic test results and agreed there is a significant need to speed up the process in New Zealand. Clinicians suggested this may be best achieved by localising the genetic testing process. Patients reported being able to reconcile the delays somewhat but only if they were kept informed of delays, which only some were.

Several Symptomatic patients indicated they experienced some difficulties deciding whether to have their children undergo predictive genetic testing, preferring to avoid or delay testing for fear they or their children wouldn't be able to cope with a gene positive result. Some children feared the potential negative consequences of the condition and its restrictions should they be genetically tested. Many clinicians were also conflicted about whether children should be offered genetic testing but on ethical grounds. Both parents and clinicians were concerned about the potential ramifications a gene based diagnosis could have beyond just the medicine. Several clinicians reported genetic test results can be useful in adding certainty to an uncertain clinical diagnosis and guiding treatment recommendations. Some patients agreed with this. Several clinicians also recommended an additional Genetics Counsellor to ensure adequate genetics consultation.

New Method of Explaining LQTS

Several Asymptomatic patients found LQTS a difficult condition to explain to others and recommended a simple diagram depicting the problem be provided when diagnosed. This could be used to explain the condition to them in the clinic which they could then take away and use to describe the condition to others. Some clinicians discussed providing follow up letters with information to help patients so an opportunity exists for it to be incorporated.

Summary of Similarities and Differences in 'Clinical Care'

Table 8 provides a summary of the similarities and differences described in the 'Clinical Care' theme.

Table 8. Summary of the similarities and differences within clinical care

Similarities	
Areas of agreement between patients and clinicians	There is insufficient LQTS related knowledge and misdiagnoses within the regional centres, primary and secondary service clinicians, including GPs
	Training is needed for primary and secondary clinicians, particularly GPs
	Improved follow up for patients would be beneficial
	An open, honest interpersonal style from clinicians is ideal
	The extensive waiting period for genetic test results is unsatisfactory with both offering solutions
Areas of agreement between parent patients and clinicians	Concerns about children undergoing predictive testing, with many parents preferring to delay or avoid testing in response to their and their children's fears
Similarities between patient subgroups	No differences were found between patients 'at-risk' who underwent predictive testing from those were 'diagnosed' and received diagnostic genetic testing
Differences	
Patients view only	Lack of management of paperwork within CIDG
	Insufficient Specialist support during LQTS patient births
	The provision of a visual aid during explanation of LQTS to improve understanding, and be used by patients to explain condition to others
	Not informed sufficiently during wait for genetic test results
Clinicians view only	Clinicians report Cardiologists may not be the most suitable for providing emotional support
Patient subgroup differences	Symptomatic and Asymptomatic patients differed in their information requirements, with Symptomatic patients generally needing more than was provided by services.

Adjusting to Long QT Syndrome

This category centres around patients' perspectives on aspects that are challenges to or impact upon psychological adjustment to the condition, and what clinicians perceive are the emotional and psychological challenges or difficulties LQTS patients are affected by during the adjustment process.

Patient Themes

Nine subthemes emerged from the patients. These were Response to Symptoms, Impact of Diagnosis, Functional Restrictions, Coping, The Family, Confusion & Uncertainty, Response to Clinical Interventions, Burden on Others and Mortality (see Table 6, p. 72).

Response to Symptoms

This subtheme concentrates on the responses individuals have had to symptoms associated with LQTS, including symptoms arising from medications. All of the Symptomatic patients reported feeling anxious about their symptoms, with many describing significant anticipatory anxiety about experiencing future cardiac events. Three spoke of being especially fearful of losing consciousness (LOC).

> One thing I struggled with was that although I can't remember the event itself I must have had some residual memory of the feeling of the passing out, because for a while afterwards when I was just dozing off to sleep that kind of feeling you get where you're just kind of losing consciousness would make me go "Ohhh, don't like that" and so as I was going off to sleep I'd kind of jolt awake again and think "Oh, I don't like that feeling" cause it reminded me of passing out. *S-D-DF*

> I hardly ever went out during those times because of that fact and because of the fact that I was still dealing with, still dealing with the fear of losing unconsciousness. That's an unreal feeling. It's an unreal feeling, it's like you know it's happening but there's no way in hell that you can control it, you know, it's just. It's yuk, I wouldn't put it on anybody it's just a yuk feeling. *S-D-P*

Several also described agoraphobic feelings, being reluctant to leave the perceived safety of home or feeling vulnerable in public out of fear they may have a cardiac event.

> I did feel I suppose a little bit agoraphobic after being stuck at home for quite a while. *S-D-DF*

> I was still having, still afraid of going out because I was scared that each time I went out I'd have an episode, and every time I had an episode the defib would go off. And that used to kick me back for a six, you know, it used to be, drain the hell out of you. I hated the sensation of going, losing consciousness, I dreaded it. *S-D-P*

Many are perhaps recounting conditioned fear responses to symptoms that had become classically conditioned stimuli. That is, because LOC, dizziness or heart palpitations (the symptom) was repeatedly paired with cardiac events that produced extreme fear of dying, eventually a cardiac event was no longer needed to produce the same level of fear response to the symptom as they had become conditioned to co-occur.

Impact of diagnosis

This subtheme centres on the circumstances under which individuals first realise they have LQTS and the effect this has on them, including fear for self or one's child, grief, depression and minimal impact. Many Asymptomatic patients described minimal impact from the diagnosis.

> Oh, I wasn't too fussed about it really. *A-AR-P*

Perhaps the threat to mortality is less salient in the absence of symptoms. All the Asymptomatic parents however spoke of their fear and grief if their child was diagnosed; with it being especially distressing for all of the Asymptomatic parents of Symptomatic children.

> And so for myself I didn't, it didn't it didn't sort of worry me. I felt you know confident that, that but I was more worried, I didn't, you know, I was more worried for my kids and things so that, yeah, that I would've preferred if they didn't have the gene. *A-AR-P (Parent of Asymptomatic child)*

> There was one day where [our daughter] was, when she was still in NICU that we just went for a drive, you know, and I felt really stink about leaving her but she was sound asleep and we went home and we just cried. We just cried, you know, this little baby; it was really awful, really, really awful. *A-AR-P (Parent of Symptomatic child)*

Several Symptomatic patients described feeling minimal concern about the initial diagnosis; being relieved by the treatments offered.

> I think if I'd been told you've got this thing and you're just going to have to sit there and, yeah, then it would have been different but because it was like, okay I'm getting this great ICD and I'll be able to do whatever then I wasn't too concerned. *S-D-DF*

Several other Symptomatic patients, however, discussed becoming fearful of dying, with each cardiac event and symptom exacerbating and maintaining the fear.

> I think initially when I was diagnosed there was nothing offered. Like I was pretty much was told you know "you've got this condition see you later". And I remember I had a big breakdown to (specific clinician) and he's, I mean he's a great man but he was like looking at me like "what are you so sad for you know what's your problem". And I guess when you're told you know you've got this condition and I was like "oh my goodness, I'm going to die" cause you know my Sister died and that was my initial thing was "I'm going to die". And so each day I'm thinking "am I going to die today?" Each time I fainted, I freaked out more. *S-D-P*

> But I think, if you'd interviewed me, you know, when it was all relatively fresh I think I would have been a lot more fearful and a lot more, I probably would have been balling by now cause I was quite emotional about it. *S-D-DF*

The several Symptomatic patients further described feeling a significant loss of control and ability to cope with additional stressors; the hopelessness that ensued after receiving a LQTS diagnosis; and the sense they were waiting for death. They described feeling depressed. In several cases, a significant safety concern to their self and their children was described at that time.

> I suppose I was a bit depressed for a while, I was quite teary for a while and I, and I used to have quite, for a little while, mostly after that work episode cause after that I felt, oh this is a part of my life and it is going to keep happening and the drugs that I'm on haven't stopped it and, you know, I had two in one day and how often is it going to happen and am I going to die. *S-D-DF*

> You know really that's freaking harsh to say but you kind of yeah, it's kind of like it would be a blessing if something fatal happened because I could get out of how I'm feeling you

know. But then again I wouldn't want to be without them but it's just like sometimes because you just, yeah you're just waiting for them to die. *A-AR-P*

I was angry at my Doctor. I was angry at the Doctors at the Hospital. I was angry because at myself cause I couldn't cope, which was unusual for me. I could do anything. I was invincible, you know. I was angry cause my family started treating me differently. It's not just that they tried to help yeah I was just and I was also depressed I think. I don't know how you could do that at one time but I was quite depressed to the stage that I was willing to kill myself and my kids cause no one would be able to look after my kids more than I could. So instead of killing myself I'd kill them first and then myself. It affected me a lot. *S-D-P*

Many Asymptomatic patients spoke of coming to terms with the fear of dying, often through being able to balance the potential threat with the probability of death.

For me, being post 40 you know it's not so much of an issue the way I understand it, especially where I sit in the, in my QT sort of gap or whatever you call it. So it's less of a concern for me. *A-AR-P*

I just felt that if anything was gonna happen it was gonna happen, you know. And not much use worrying about it. I'd probably worry more about skin cancer than that. You know, stuff like that or, or things that are gonna affect you today. And, and also the fact that the information they gave us is that once you've reached a certain age which is you know I'm around that or above it, I think it's forty and I'm above that. The percentage the likelihood of having it is quite remote, down to about 5% or something like that. *A-AR-P*

Patients who experienced diagnosis related distress did so in response to the first diagnosis they received, irrespective of whether it was clinical or gene based. That is, for those who were first clinically diagnosed this had the most impact with the following genetic test result less so, and vice versa for those who received a gene based diagnosis first. As is consistent with at-risk predictive testing Asymptomatic patients then experienced more impact after gene based diagnosis and diagnostically tested Symptomatic patients experienced more impact after clinical based diagnosis. While this trend was noted, the patients did not directly discuss this.

Functional Restrictions

This subtheme reflects the impact of restrictions on functioning. Patients most affected by the restrictions on functioning were those who were physically active with competitive sports or swimming leading up to diagnosis. Of these, it appeared hard for many Asymptomatic patients and all the adolescents in particular; perhaps because there was no discernable sign why this sometimes drastic change in lifestyle was necessary and it was most salient to those developmentally at a physical peak.

> They basically put it down, well you know I had to stop I had to stop all sports. They said I couldn't do anything whatsoever, all competitive sport that is. So again that, that was pretty tough for a young fella who absolutely loved going out Saturday mornings playing rugby and also cricket and that with my friends. Everything had to stop. I couldn't do anything, no more swimming, no nothing. I was gutted. *S-D-DF*

> But, and again it's because it's something that doesn't really affect you physically. It doesn't, you know, you don't feel short of breath, you don't feel anything. You know it really feels normal. And so it's hard to explain to kids too, the seriousness of it you know so. Because they just don't feel anything. *A-AR-P*

Several Symptomatic participants also found the changes extremely distressing.

> And that was it my worst ever, my worst time that anybody could have been and that was dealing with the grief of that I had a disorder that you know it limited what I could do. Cause oh, I was in the peak of my rugby (laughter). I couldn't play rugby, I couldn't play. I thought everybody alienated me because I had, because I had this, you know these devices. So I couldn't play sport or do any, I was very active, very active, mother, you know. Family orientated, sports, kohanga, school, you know, I was quite and then all of a sudden 'bang'. Part of your life was like, "oh sorry you can't do this anymore and, you know, you've got to take it easy now" and I thought 'my God, I'm at my peak and I can't do this and that!'. So that was where all the anger had stemmed up, and depression at the same time of course, and I felt useless. *S-D-P*

Many of the older patients found it possible to balance their desire for physical activities with the relative risk they felt they were taking.

> Yeah. Cause you can go one way, you can be, you can be "poor me" or you can be "well okay I'm still gonna… what can I do" what, what can I do that's safe and without being, I've never tried a cocoon but you know still I, I've just re-diverted my thing from diving to, to saying I'm, I do a lot of you know go to poker, fishing and manage that and try and balance you know, cause that's my passion. *A-AR-P*

All of the parents with a LQTS gene positive child spoke of feeling guilty at having to restrict their children's activities and consequently their risk, however several parents mostly chose not to adhere to medical advice so as not to disrupt their children's lives.

> She's sort of a bit resentful about the whole thing. Particularly when the doctor says oh you can't have this or can't have that or you can't go swimming. She does go swimming, much to the Cardiologist's horror. We just think its part of life eh. *A-AR-P*

> And you can't you know he loves his rugby too and so you, you've got to try and keep a balance. You can't just sort of say "no, you can't do that." *A-AR-P*

Several patients found themselves with little impact from the functional restrictions as it fitted with their existing lifestyles.

> But I haven't felt like I've had any big struggles. There's nothing I've had to give up that has been a real loss to me so I've been very lucky. *S-D-DF*

Coping

This subtheme relates to methods of coping, including the strategies and supports used. Many participants were able to cope with their fear of dying and procedures using coping strategies such as seeking information from the internet to understand, but for several people this was unhelpful.

> I did a lot of reading about Long QT and I did a lot of, you know, Internet searching which probably drives the doctors nuts cause half of its good and half of its crap. *S-D-DF*

> But your point about sourcing information I suppose I have filtered out some too that I didn't find helpful like the websites and that kind of thing, if it was just getting me down and I found myself sitting reading websites and crying at the computer and I thought "Well this isn't doing me any good. Get this out, get out of doing this". *S-D-DF*

Several sought information from the internet to support a positive outlook or risk appraisal.

> I've just sort of basically just got on with things and just, it was good to get information and read it through. Like, certain information I've got is is things like certain things you can do that can help you know like I've stopped diving. I try to, you know the, the things that can bring this on, I try not to, you know, the extreme competitive stuff I don't do or anything like that. It's just I, you know, it's about balance. *A-AR-P*

Several advocated for positive reframing of what could otherwise be seen as negative.

> So, so what it does is it gets you out of you know, it gets you into pro-active stuff, positive stuff you know. And I think that's you know, and, and in that same way I could think "poor me I've got this, bloody hell and Dad gave it to me" and you know and go on and 'oh shivers'. Everything you do is gonna be negative you know. So you can do you can go that way but I've always gone the positive way. *A-AR-P*

Several said not worrying about it helps.

> It's just being is talking to yourself and saying hey you know not worry about it because that promotes, before you know it everything's affecting you. You know. *A-AR-P*

Several Symptomatic patients suggested developing support groups or having meetings to go over some educational material plus have an opportunity to meet others in a similar position is or would be very beneficial.

> I think you know meetings with people and actually sort of you know whether some group sessions or you know where you can get a whole heap of people together, whether it would be in here or wherever. I'd be quite happy to travel for something like that. *S-D-DF*

> And I know this is not the case for everybody who has this condition, I don't know, although, you know, a lot of people I haven't met many but I have met some with Long QT through the ICD support group at [particular hospital]. But most of them are, you know, relatively young, fit people and I suppose similar to me they're just getting on with it. *S-D-DF*

Many of the Symptomatic patients described a stage of abusing substances to numb themselves, avoid and thereby cope.

> That's how I dealt with it was the booze. It'd numb it, it numbed the situation. So what I used to do was drink and I'd just say to (my husband) "I'm out I'm going" and he goes "where are you going" and I say "I'm going out, I just need a beer", you know. And I'd just drink to where I actually dropped, or drunk where people had brought me home and I had no idea where I'd been or I'd blacked out, you know. *S-D-P*

Many patients described comparing their situation or risk factor with one worse than their own in order to reduce anxiety and see the positives.

> You know I've inherited it, I've got to manage it and, and I'm just and I'm just, I look at it this way I'm blessed that I didn't have it worse. (pause) You know, you know. And, and hey the chance of me having a bloody car accident sometimes if I'm being silly is well like if I'm dying from that you know or going out in the boat when not safe, or different things you know or if I don't manage my your lifestyle, well you could die from heart disease, whatever. You know. *A-AR-P*

> I mean cause sometimes you think "oh why me" and but then you've got to think well you know why not me and what can I do with it and you know there's people who are far worse off as well than having I mean yeah. *S-AR-P*

Several also spoke about contributing to others in a similar situation to both normalise their experience and as a way of feeling useful and ultimately better.

> So I'm kind of, I feel a bit more at peace about that now so that had triggered a few emotions. And I guess I've found too the thing that helps I started a Support Group here. *S-AR-P*

Several spoke of incompatible coping mechanisms in effect within key relationships that had or were breaking down as a consequence. This commonly had a detrimental effect on each individual's ability to cope as they felt alone and invalidated in their attempts to adjust.

> And my Aunty was trying to get me to go to Counselling and things and at the time I just wasn't ready for it. And so yeah there was lots of tension. I guess the way I handled it was I, I would come home from school and things and shut myself in my room listening to my angry music and write my angry poems. And she's like "oh that's not good, that's not good" but that was how I expressed and she tried to take, I guess take those things away from me and that was my coping mechanism for the time. *S-AR-P*

Several opted to undergo counselling/therapy and reported it was immensely helpful in reducing their distress and developing their coping skills.

> Yeah, I'm not so bad now, like I've been seeing [person] as well, just for help with that. I think too because I'm coming up for another operation within the next year to have the ICD replaced and so it's brought back those initial feelings and fears of I guess death is the ultimate thing. She's been so much help. *S-AR-P*

> Got through the counselling cause they were on my doorstep in a matter of 24 hours, bang they were there and oh I did a lot of crying and I think it was they came twice a week to my doorstep, I didn't have to go outside my home. They helped me through it. *S-D-P*

The Family

This subtheme relates to the social and emotional relationships and roles within families. All parent participants with LQTS positive children felt to blame for passing LQTS on to their children.

> Oh, sort of, I mean there's sort of a little bit of blaming in you quite sort of involuntary, you kind of blame yourself that you've done this to your daughter eh? But then you know my mother did it to me without knowing and then T will probably do it to her kids and it won't be her fault would it? So you sort of rationalise it out pretty quickly. But you know I do sort of feel sometimes I've brought this on the family. *A-AR-P*

> There's definitely guilt involved in thinking that it came down my family, side of the family so it's like and I know that dad went through that thinking you know well if I've gone and passed it on to my children. *A-AR-P*

Several of those without children (DF) and parents (P) reflecting back prior to having children, spoke of having grave reservations about having children for fear they would pass on LQTS.

> For a long time I did think well maybe I won't have children for that reason I was worried about passing on the condition to them. *S-AR-P*

Several discussed apportioning blame to their partners perhaps as a way of deflecting potentially intolerable feelings of guilt should they be the originator of the LQTS gene.

> But then to me it was it was if it was Romano-Ward it was a dominant gene so it had to come from one of us and I was hoping it wasn't me I was hoping it was my husband. And it's a horrible thing to even you know think about but you kind of think "I hope it's him" it will be him it won't be me. *A-AR-P*

Confusion and Uncertainty

This subtheme concentrates on instances in which the patient struggles to understand or is uncertain, such as when trying to understand the condition, the genetic test results, the risk or explaining this to others. All the participants found LQTS difficult to describe to others effectively as it is a complex condition with an atypical presentation, especially as you look otherwise fit and healthy.

> Yeah and people look at [my daughters] and I explain you know even the people from the [charity organisation] that I'm on they had no idea that they are deaf because of this heart condition you know, they're just like "well what's that?". Cause you know normally it's something else is why you're deaf and not this. So yeah they see them they this particular group sees them as just being deaf children which I think is great and when you say that the heart condition they say "well they don't have blue lips" you know they don't get really tired they look quite healthy they appear to be quite healthy. They don't understand it. *A-AR-P*

> People find that quite difficult to understand my condition because, and I find it difficult to explain myself to other people like workmates and friends and that, because I look normal, feel normal, act normal but then every now and then this thing happens that does freak you out but then after half an hour I can kind of get up and get going again. And so the way I describe it to people is it's like "Yes, I'm alive, I'm alive, oops, no I'm dead, no I'm alive again". *S-D-DF*

Many also struggled with understanding their genetic test results and none described a LQTS subtype when recounting their result or diagnosis. It is unclear whether this is because they forgot, hadn't been informed or just didn't mention it.

> To my knowledge it was quite, it wasn't high, high but it was medium to high, it was quite, it was, I don't know. But they put it in numbers but I've forgotten what number it was but yeah. I don't know. *A-AR-P*

> Oh the genetic, oh okay. Basically that I had the gene. And that I was the one that passed it on to my kids. And that, that's came from my my side of the family and they found you know when they tested, the gene was in me. *A-AR-P*

Several described the risk factors for all the subtypes as things to avoid for them and confused these with other possibilities, often leaving them confused and potentially over restricting themselves.

> But, you know, what I'm saying is just the fright. I don't know, that's just what, what I believe is it's, it's an extreme fright, doing too much or extreme cold or something, some chemical. I've heard grapefruit, I don't know, something in grapefruit, and milk, too much milk, so bugger if I know what. *A-AR-P*

Responses to Clinical Interventions

This subtheme reflects responses to the procedures required during clinical assessment or treatment for LQTS, including tests, surgery, medication and implantable devices. Several found the diagnostic provocation tests highly distressing, especially when experienced at a young age.

> And then I went through some pretty regular testing as a young fella which was pretty sort of traumatising. So they actually took me through some tests actually trying to make me my heart go up into, you know. Put me on treadmills and basically making me run as fast and as far as I could. Yeah and there was another one where I had to hold my head under water in a bucket of ice water until I blacked out as well. Yeah I would've been nine or ten. Yeah, yeah, it was pretty traumatising. *S-D-DF*

Many Symptomatic participants had ICDs implanted and feared dying while having surgery or feared having a foreign object inside their bodies. Parents of Symptomatic children had the same concerns.

> I'm sort of dreading when she has to go back for another, you know, operation you know to get the tube it needs, you know what it's like eh, as she grows they sort of stretch inside her. *A-AR-P*

> Yeah because after the pacemaker (laughter) after the pacemaker they thought well you know not long later they thought "I think you need a defibrillator". I go "oh God is that another

machine" and they're "yeah it's another machine implant" so I have one pacemaker and one defibrillator inside. I was terrified of having all that inside me. *S-D-P*

Many participants spoke about how taking medication each morning was a constant unwelcome reminder of the condition.

> Yeah, they don't pop into my head totally un-triggered, but for example because I have to take medication everyday what really annoys me about that I don't mind taking medication is that every morning I have to be reminded that there's something wrong with me. *S-D-DF*

> You know if you just, I think about it every time probably when I take my medication you know. Every day it's there. *A-AR-P*

Burden on Others

This subtheme concentrates on instances where the burden for one's safety and care is either given to, or is taken on by, others. Many Symptomatic participants spoke about the impact of their spouses taking on the burden of their care, including worrying and reviving them should they go into arrest.

> And but it had a big psychological impact on Grant who was very fearful of being round anybody who he might be responsible for after he had to save me. So if he, ever since then he hates being in the water with little kids or we've got a boat up at the beach. He hates taking little kids out on the boat because he feels responsible and that he might have to save them. And he had some counselling about it, not at that time but after I had a second cardiac arrest. But it had much more impact on him. *S-D-DF*

> I was being very selfish but I said to him "Look, you know, this has happened to me as well and you're shutting me out because you won't talk about what happened and what we went through and I want to know because it was me that, you know, collapsed on the street". And he kind of said "Well, it's alright for you, you know, you weren't there and you haven't had to live through it but I've had days of watching you in intensive care and wondering if you're going to die and having to resuscitate you". And I thought actually he's got a good point there, you know, he's been through a hell of a lot more than I have psychologically, so. *S-D-DF*

Several Symptomatic patients also described attempts to cope in public by alerting others to their condition and requesting they take the burden of their care.

> For a while I kind of went around wanting to have a neon sign over my head saying "I've got a heart condition please watch out for me" and I felt. And so when I did go into my first class that I taught, you know, I did actually tell my students, it was a Masters class, I did say "Look, I have got a heart condition and I could possibly pass out and I've got this thing and it could give me a shock and it will look like I'm kind of, you know, getting the paddles, if you watch ER that's what it will look like but don't worry, you know, I'm fine and I'll come round very quickly". And they all just sat there like. (laughter) *S-D-DF*

Several spoke of behaving stoically in order to help their spouses cope.

> So initially I was, I suppose kind of, I don't know, putting on a bit of a brave face to help (husband) and trying to kind of, you know, jolly it up a bit. *S-D-DF*

Mortality

This subtheme refers to instances when mortality is considered, one's own or others'. Three Symptomatic participants described a sense of foreshortened future when comparing own mortality with relatives who died young.

> I'd already had in my mind that well maybe I'm not going to live that long cause if I'm going to take after my mother. You know, at that time I was thirty, thirty one and Mum was only fifty six when she died so I was already thinking "Oh maybe I'm only going to last til I'm fifty six". *S-D-DF*

Several others spoke of the role repeated cardiac events played in making mortality more salient.

> I have had times when I've been quite teary and felt, you know, am I ever going to be normal again because I did have a long period where I was quite fearful and especially after I had the shocks and stuff they really upped my Beta block dose and I was really dizzy and light headed for about a month or so and I couldn't even have a walk around the house without touching the walls to kind of feel that I was kind of touching something solid. *S-D-DF*

All those who had lost a close relative to LQTS found it adds saliency to the risk of dying and to their or their children's mortality.

> I guess it's, it's more so that I've seen you know I saw my sister die from that and so that I know, I know it can kill you like this condition. *S-D-P*

Several described having come to terms with being at high risk of dying suddenly, finding acceptance in their own way and no longer fearing death.

> I guess when you get a bit phobic about death (laughter) you think "I don't want to die cause I like life so much". But and you worry about it I guess you know about death. But then when I think about my faith and I think well I know that there's something after this life so it's not, it's not that you're life is over it's that you're moving on to something else. And when you think of it that way as well it doesn't seem such a big, a big deal having a heart condition yeah. *S-AR-P*

> Yeah, I worry a lot more about, it's a very selfish thing because I'm not worried about dying at all, I'm not religious at all so I don't believe in an afterlife or anything but I do kind of think well, I've had almost the dying experience on a number of occasions now and if that's how I die then hey, it's not that bad. You just faint, that's it, you're gone. *S-D-DF*

Clinician Themes

Challenges

This subtheme relates to the emotional and psychological challenges (positive and negative) patients experience during the adjustment process. Many clinicians spoke about how difficult they believe it is for patients to understand all the information provided by the clinics; especially as they feel the results and prognosis are potentially uncertain.

> So, yeah, (pause) I have to say in the last couple of years, I expect that it's somewhat easier for me to deal with the uncertainty of results, because we deal with it quite often and whether the patients find it easier is a different matter. I think a lot of the time they're just bowled over with information, to the point where they're not sure how much they understand. *Genetics*

> I think you just have to accept that it's a huge diagnosis for a family to take on board, mostly because of the vagueness and the uncertainties. A lot of time you cannot say "You have the diagnosis and you're gonna die at 2pm next Thursday", you can't say that. And you can just say "You're at risk. You might never die from it, you might. It could be when you drive home this afternoon. It might never be. *Cardiology*

The genetics clinicians further discussed how this uncertainty must be difficult for individuals and families to adjust to.

> I think this, the figures I've heard from from the Cardiologist, you know if it's a good going family history 50% of the time we'll find something, but half of those of results will be a new change, not previously described. So, a lot of the doc time we'll probably going to be dealing with situations where we can't get a definitive answer, even if we do find something in genetic testing. So, I guess if you're looking at it from a family perspective, there's all sorts of stresses eh? This is what's happened to them, what's potentially happened to their extended family. The uncertainty around the clinical evaluation, cause it's often not black and white, that you can tell people sometimes "You're definitely not affected" or "You definitely are." *Genetics*

Many of the Cardiologists spoke about the effect the inheritability of LQTS must have on a family, especially young adults considering starting a family.

> I mean you know you're doing the things that are so different to dying than in an otherwise chronic progressive disease of the elderly, if you will. That guilt factor's not there. The passage of a genetic condition is not there. The fact that you may have a child as a teenager or a bit older who wants to get married and then find out that they have it, that clearly will impact on that child. I mean not to be judgemental that you're dating someone you want to get married and then get told that they've got a 50/50 chance of passing on a fatal illness that only presents itself as sudden death, that's quite a big one. So, you can see why there's some people who don't want to know. And for those who do want to know it's a big deal to find out. *Cardiology*

Several also felt parents were challenged by feelings of guilt in having passed LQTS on.

> But that side of things with the guilt factor, the fact that it's genetic, the fact that there's uncertainty, the fact that it's gonna continue generation to generation is quite an important concept. *Cardiology*

Many clinicians discussed difficulty placing functional restrictions on an individual and how hard it must be for parents to enforce this with their children. They felt it could potentially change a

person's life detrimentally and were concerned they therefore only genetically tested those that would medically benefit from the outcome.

> An unfortunate young chap who was only in his 20s I think who his one interest in life was driving a big rig, and he'd bought a big truck and he'd invested, got his long, his particular license to do it and he was deemed positive and he'd had an episode when he had this unexplained collapse. And so we had to tell him that he wasn't able to drive commercially and that really totally wrecked his life. *Cardiologist*

> It impacts the life a lot because of what you say you should do, shouldn't do, swimming alone, driving, occupation, pilots. I mean there's a lot of spin-offs. *Cardiologist*

Several clinicians were acutely aware of the impact the diagnosis could have on some people and especially the anxiety it could evoke.

> Depending on the personality of the patient you then expose the patient to anxiety that you can't take away, because on the face of it you tell someone you're at risk of getting something really bad happening to you if you get overexcited or you swim or whatever. Then some people will get very introspective and they can get pretty paralysed about that. *Cardiologist*

Several clinicians also spoke about specific maladaptive coping strategies they believe could caused additional difficulties, namely denial or avoidance.

> In bringing it all back to the surface and explaining what the diagnosis was it was then that she hit the wall, so you know, I mean people can I think cover things up and choose not to deal with them or they don't know what they are so they put them to one side but when you unearth and say hey come into this clinic we've got something to tell you and you do that then it's what happens when they leave and that's what always worries me. *Clinical*

Solutions

This subtheme concentrates on what the clinicians report they currently do to try and alleviate or reduce the impact of the emotional and psychological difficulties patients' experience, including how effectively they feel they are at this.

The clinicians spoke of a variety of ways they endeavour to reduce the anxiety they see patients often overwhelmed with. Many Cardiologists offer risk evaluations, using real life comparisons, to ease anxiety. Several said listening was important.

> On the face of it you tell someone you're at risk of getting something really bad happening to you if you get overexcited or you swim or whatever. Then some people will get very introspective and they can get pretty paralysed about that. So, you know, that's a problem and I've tried to put it in perspective that, you know, there's always some risk in life, so there's a risk that you can, I often use car for example, I say "Well you came up here in the car, there's a small chance you could have a fatal accident". "You could've been electrocuted in your home, that doesn't stop you using electricity". Just so that people know that you can't take away all risk, but maybe the risks are commensurate with other everyday activities for which they take risks but it doesn't end up paralysing them or being perceived. *Cardiology*

> Often times I don't, you know I'll be seeing somebody and I try not to actually get into any sort of situation where I'm doing anything that's remotely like counselling but sometimes you do have to listen to people. *Clinical*

Many Cardiologists spoke of giving reassurance to ease anxiety. Primarily by asserting themselves as experts with a breadth of knowledge to help from, potentially adding a sense of containment by giving the condition a name and maintaining there are actions to take that will help.

> I think that one of the ways to reassure people to start with is just to actually talk about, you know, what you know. And the fact that you've come from a situation where there is anxiety and now at least we have something definite to work with and that this is a known condition. And that there are a lot of experts who are putting a lot of time into, you know, figuring out what we know now and what we're going to know in the future. So that instead of having a, you know, something that's totally unquantifiable you come to something that at least it has a name and there are, there is an approach to that to help with that problem. *Cardiology*

> Whenever you introduce some bad news to somebody the first you have to say is "Look, there is something I can do to help". So, advice that there are things which are going to help the condition, Beta-blockers or practical things to do. *Cardiology*

Several Cardiologists also spoke of encouraging the use of existing adaptive coping strategies.

> Particularly some families that have developed a dark sense of humour to help themselves get through it and people have talked about how the parents used to argue between each other as to which should go and check the children at night, for example. You know, cause they're just frightened the child might be dead when you go there. So, how they, how each individual family works through that fear, they, you know, they bring it into their life and make jokes of it. I encourage that. *Cardiology*

Furthermore, several Cardiologists felt teenagers commonly coped by denial and while several were accepting of the strategy all also strongly advocated prophylactic medication and behaviours.

> The other thing with teenagers is to offer them as much as possible the opportunity to deny their condition if they wish to. One of my phrases is "You can deny it if you like, but please don't do these things, come and see me again. Take the medicine and avoid these other medications and then for all other intents and purposes you can deny you've got it". It's a good coping strategy, just get on with your life but take the Beta-blockers, don't forget to take the Beta-blockers. *Cardiology*

Information was considered by many clinicians to help patients cope with the complexity and uncertainty of LQTS, as well as being necessary to clearly discuss the condition with supports.

> So yeah it's you never talk about 'definites' it's 'almost certain'. So we provide information both to the patient and for the patient's supply to the extended family. That's helpful, really helpful as well cause I think even if you understand it in Clinic it can be really hard to go away and relate the information to other people. We will talk about some of the psychological implications of testing so I guess that's information and support. *Genetics*

Several clinicians spoke of referring to Psychologists or Grief Counsellors to assist with psychological difficulties, including excessive anxiety or restricted functioning due to low mood.

> When you're talking to somebody you can't really differentiate what it is your doing. Sometimes it's dealing with the pain they've got, sometimes it's God I've passed it onto my children or you know, those sorts of things and I just try and refer people. If I see somebody who I don't thinks coping you know I actually use a Grief Counsellor and Clinical Psychologist a lot. *Clinical*

Several clinicians gave them as much time as possible to process the information in the clinic, making it clear by restricting jargon, and providing opportunities for them to re-contact with any questions.

> And also give the patient time to actually say if they don't' understand and checking that the information you're giving them is actually, what you think you're telling them is what they're hearing. And of course I mean, we know, there's plenty of states people walk out and they've only taken a fraction of it, but you can at least help by not using confusing language and that's always a trap cause it's easy to keep dropping into that jargon. And some of your LQT is laden with that, laden with that sort of thing, and to you this is the, you know, 500th patient you've seen, but to them it's something brand new and it's bloody scary. *Cardiology*

Several clinicians spoke of arranging for mutually affected patients to get together, perhaps as an opportunity to normalise and validate their experience of the condition with others in a shared position.

> I, you know, in that situation what I probably would do is, you know, is go to the Coordinator and say "Look, have you got a family who you'd be happy for these, like to talk to?" Because I think sometimes you can hear from a Clinician but actually to talk to a parent whose facing the same problem is really helpful. *Cardiology*

Many spoke of how important it is, as a clinician, to adequately understand the uncertainty involved with LQTS in order to be able to provide accurate advice and care to patients.

> You've got to be understanding about the implications and what it is that you're saying about a genetic disease, it's not just LQT but it's like any inherited cardiac disease there's usually bad news and it's usually filled with uncertainty. So, if you don't fully understand the uncertainty that comes with a lot of what you say, you can't be very specific, you can't, you know, even if you tell someone you have LQT you don't know if they're gonna die from it or not and it's a sudden death. So it's not like somebody might or might not get cancer, not that that's great thing to get, but usually if you get it there's a period of getting it, a period of having the diagnosis, and later a period of getting used to it, a period of hearing what your options are, hearing or seeing an Oncologist. *Cardiology*

Recommendations

This subtheme is concerned with Clinicians perspectives on what further would be helpful to support patients' emotional and psychological adjustment. Several clinicians recommended having a multidisciplinary team there to meet patients multiple needs.

> Have the right people there to do the discussion with, so again it's not just me between cases it's yourself, a Geneticist, the Psychologist, the Clinician for example offering contact details so they can ring you back whether it'd be an email or a cell phone or written appointment, make another appointment. *Cardiology*

All the clinicians recommended a Psychologist be part of the CIDG multidisciplinary team permanently, so patients could either access them during and after clinics or after they had been flagged by significant scores on anxiety and depression psychometric assessment tools.

> And then yeah being able to refer someone there and then and say hey do you want to talk to someone, they're here, would be just the icing on the cake, it would be fabulous. And I think, well not only fabulous I think it's so necessary and it's overdue. *Clinical*

> To me, I mean if in a perfect world you would have the availability of a Clinician like a Cardiologist or a, potentially a Paediatrician who had skills that, you know, an appropriate medical practitioner and a Genetic Counsellor and a Psychologist. *Cardiology*

> We would really like to have a Psychologist attached to our clinic, sitting there available if somebody wanted to see them. That's the goal. *Genetics*

Many clinicians also recommended either peer support through the development of support groups, using existing groups more or appointing a dedicated Social Worker to follow up with individuals in their homes. Perhaps also those that have chosen not to contact the service regarding their decision to be genetically tested could be included as several clinicians spoke of concern they are potentially not meeting their needs.

> I think patient support organisations and parent support organisations have a really big role to play for people as well. To help give people support and that's something in terms of paediatric cardiology when you use the sort of heart children group as a model, you know, having local family support people. I think sometimes we think, we do a reasonable job in

terms of taking care of the medical care, but you hear really interesting stories from the family support people when people go home with these little fragile babies and are trying to cope, you know. And I think to have someone who is not a professional, whose not but whose actually someone like you whose been in this situation to come in and have a cup of tea with you, sit with you and hold your hand is actually, I mean that's the kind of emotional practical support that I thinks incredibly valuable. *Cardiology*

Firstly, those who we never get to see, and obviously they're the hardest to study or to contact, but it would be nice to have an, to sit down to try and work out why they don't want to see us, and we were talking this morning, some centres have like, social workers who care for them, really, to try and contact these families. So, I think it would be nice to have money to try and increase that contact with those families who are reluctant to be involved with us, it's tricky, it may be too pushy but at the same time, you want them to understand why we want to get them in. *Genetics*

Similarities and Differences

Anxiety and Depression

All the Symptomatic patients experienced anxiety, many in relation to dying, cardiac events and losing consciousness. Several also described feelings of fear around having a cardiac event unsupported in public and this restricted them from leaving the perceived safety of their home. Many of the clinicians were aware Symptomatic patients could experience this anxiety and of the significant and paralysing effect it can have. Several Cardiologists offer risk evaluations to try and decrease the risk related anxiety, which is a coping strategy commonly used by patients. Some Cardiologists also attempt to reassure with their expertise and knowledge, offering containment by giving a quantifiable name or diagnosis and proposing options that will potentially help, something patients also report aids their coping. Several clinicians also report trying to listen to concerns and several refer to Psychologists or Grief Counsellors for additional support in these instances. Some Symptomatic patients experienced depression after receiving their or their children's diagnosis, a difficulty not discussed by clinicians. They also described hopelessness and in two cases to the extent that significant safety concerns for themselves and their children were described.

Involvement in Sports and Age Affect Impact of Functional Restrictions

The psychological reactions to having to make significant restrictions on activity levels, in order to reduce risk of a cardiac event and potential sudden death, appeared to differ according to age and level of involvement in sports. All of the patients that described considerable involvement in sports prior to diagnosis reported significant distress at having to make a drastic life change for themselves at what they felt was the peak of their physical life. They also reported it had a significant impact on their access to their usual sporting social communities. Many Asymptomatic patients also found the change difficult to comprehend in the absence of symptoms. The several who were not involved in sports prior to diagnosis however reported minimal difficulties and the several older who were involved in sport prior to diagnosis described being able to transfer their interests to less risky activities without reporting much distress. Many clinicians were aware that placing restrictions on a person's activities could be devastating but examples were restricted to potential occupationally related problems. Both the clinicians and those affected by LQTS discussed how difficult it is, or must be, to enforce the restrictions on children, particularly Asymptomatic children as they too find it difficult to understand why it is necessary when they felt and looked healthy.

LQTS as Confusing and Uncertain

The genetics clinicians discussed how little certainty they can provide when giving a diagnosis for LQTS, especially if it's an unclassified variant, and the detrimental impact they felt this must have on patients. The patients however did not directly refer to difficulties grappling with an uncertain result but many did describe confused accounts of their diagnosis, demonstrating a lack of clarity, and many described avoiding all subtype risk factors rather than just those pertaining to their LQTS type for instance. Patients also reported having difficulties describing the condition theoretically and conceptually to others as its complex and their condition is not physically obvious to others. Several clinicians understood this and tried to provide information for family and friends at the Arrhythmia Clinics to help them understand better. Both genetic clinicians and many Symptomatic patients agreed it is difficult to cope with the uncertainty about when a cardiac event might occur and potentially kill you.

Prominent Coping Strategies

Both the patients and clinicians referred to problem focused coping strategies such as gaining information and making risk evaluations as effective, with several clinicians encouraging these and

other adaptive strategies. Several Cardiology clinicians were concerned adolescents use of maladaptive coping techniques such as denial would have an adverse effect on their medication compliance but adolescent patients did not describe this. Several clinicians described concern when patients employed maladaptive emotion focused coping strategies such as denial or avoidance. This is consistent with the actions described by several patients who reported a period of time when they used emotion focused coping strategies such as avoiding or numbing the emotional impact by heavily abusing substances in attempts to cope.

Affect of LQTS on the Family

Clinicians were concerned the inheritability of LQTS was difficult for families to reconcile and especially those looking to have a family, which was consistent with several patients' reports. In addition, several clinicians' concerns were consistent with all the parent patients' reports of feeling guilty and to blame for passing LQTS on to their children. Several affected by LQTS also reported blaming or hoping their partners were to blame for passing on the LQTS gene, which clinicians did not report.

Psychological Support

Several patients reported psychological support was good in developing coping skills and several clinicians spoke of referring to Psychologists or Grief Counsellors to assist with coping. All the clinicians recommended a Psychologist be made part of the CIDG multidisciplinary team to ensure this happened. Patients reported Psychological support was helpful.

Support Groups Recommended

Both Symptomatic patients and clinicians recommended developing Support Groups or educational events with an opportunity to meet others with LQTS. Several Clinicians hope this might have the added benefit of accessing those who traditionally don't approach or engage with services.

Perceptions on Responses to Interventions and Medication Differed

Many patients discussed the negative impact provocation tests and implanted devices had on them but these were not points raised by the clinician group. Both discussed taking medication however, but in different ways. Several clinicians felt it would be possible to be in denial about your symptoms while also taking the medication everyday but patients reported taking medication is a daily reminder of the presence of the condition, and so not consistent with denial.

Symptomatic Patients, Burden on Others and Mortality

Several Symptomatic patients discussed ways others took on, or were asked to take on, the burden of their condition, these factors were not discussed by clinicians. Several of those affected by LQTS also described considering their or their children's mortality in the process of adjusting to LQTS, including the impact of having someone close die as a result of the same condition. Clinicians did not discuss this factor.

First Diagnosis has most Psychosocial Impact

For those distressed by the diagnosis, patients generally appeared to express diagnosis related distress in respect to the first diagnosis they received, irrespective of whether it was clinical or gene based. This was not noted by clinicians.

Parents of Symptomatic Children and Symptomatic Patients Experienced More Significant Impact of Diagnosis

Many Asymptomatic patients described minimal concern at receiving a LQTS diagnosis or of quickly accepting it after evaluating their relative risk. When it came to receiving a diagnosis for a Symptomatic child however, parents described fear and grief responses. Symptomatic patient responses were quite different. While several Symptomatic patients felt minimally concerned about the diagnosis after experiencing relief by the treatments offered, many other Symptomatic patients had significantly adverse responses. Several described feeling a loss of control, hopelessness, depression and a decreased ability to cope with additional stressors. For some, this led to significant safety concerns toward themselves and their children. Many also reported being afraid of dying on a daily basis after receiving the diagnosis, with each cardiac event exacerbating and maintaining the fear. As aforementioned, many clinicians recognised how anxiety provoking a diagnosis of LQTS can be and they take several measures to try and help alleviate this aspect. The clinicians did not discuss the differences in reaction to diagnosis between Symptomatic and Asymptomatic, however.

Summary of Similarities and Differences

Table 9 provides a summary of the similarities and differences between patient and clinician perspectives, and other subgroups, described in the 'Adjusting to LQTS' theme.

Table 9. Summary of similarities and differences in 'Adjusting to LQTS'

Similarities	
Areas of agreement between patients and clinicians	Symptomatic patients experience illness related anxiety, particularly in response to cardiac events
	Aware the affect of LQTS on the family includes fear of, and guilt for, passing the condition on to other generations, and blame toward those who have passed the condition on
	Psychological support is needed and useful for patients
	Particular coping strategies are helpful, such as information seeking and making informed risk appraisals, which clinicians help develop when possible. Avoidance, especially through the abuse of alcohol or drugs, is not helpful.
	LQTS is a difficult condition to understand and explain to others
	Support groups and educational events are recommended
Similarities between patient subgroups	Symptomatic and Asymptomatic patients feel similarly distressed by the functional restrictions, albeit for different reasons
Differences	
Patients view only	Some Symptomatic patients experienced hopelessness and depression after receiving a diagnosis
	The first diagnosis received by patients has the most impact, irrespective of whether clinical or genetic
	Appeared unclear about which form of LQTS they had and avoided all possible triggers, irrespective of form of LQTS
	Negative impact of provocation tests and implantable devices
	Larger adverse impact of functional restrictions for patients with high involvement in sports prior to diagnosis, especially adolescents
Clinicians view only	Patients found unclear results difficult to understand
Patient and clinician views differed	Clinicians believed patients could take daily medication while in denial of condition however patients reported taking medication was a reminder, and therefore incompatible with attempts to deny the existence of the condition

	Adolescent age group have a harder time adjusting to the functional restrictions
Differences between patient subgroups	Increased distress for Symptomatic patients in relation to anxiety and depression
	Symptomatic patients tended to place more burden on loved ones
	Symptomatic patients and parents experienced more impact of their or their children's diagnosis than Asymptomatic patients
	Functional restrictions difficult to reconcile for Asymptomatic patients because appear well to themselves and others
	Symptomatic patients place more value on support groups

In addition, no consistent differences emerged between At-risk and Diagnosed subgroups other than those that aligned with Asymptomatic and Symptomatic subgroups respectively. That is, when Symptomatic patients' perspectives dominated so did Diagnosed and when Asymptomatic patients' perspectives dominated then so did At-risk. This remained true even though two patient participants were atypical in that they belonged to the Diagnosed and Asymptomatic groups. That is, the perspectives of the two who were Asymptomatic but received a clinical diagnosis of LQTS prior to genetic testing aligned with the Asymptomatic group when differences were noted. This perhaps suggests symptomatology may play a larger role in differentiating some psychosocial factors than whether they undergo diagnostic or predictive testing (for at-risk patients).

Chapter 5

Discussion

This final chapter begins by discussing the general aims of the research. The limitations of the study are then identified followed by a discussion of the major findings in relation to previous research. Patient and clinician perspectives are discussed together along with any subgroup differences. The implications for future research and the implications for clinical practice and CIDG are then discussed. The chapter concludes with personal reflections on the research process.

The Aims of the Research

This research study had the following two aims:

1. To describe the medical and psychosocial factors impacting adjustment to LQTS in New Zealand patients and families
2. To compare the experiences of (i) clinicians and patients, (ii) asymptomatic and symptomatic patients, (iii) diagnosed and at-risk patients and (iv) of adolescent and adult patients.

Limitations

The qualitative findings of the present study provide valuable and important information about the experiences of the LQTS patients and clinicians in the study. They are limited, however, in their ability to demonstrate causal relationships between factors, especially when related to the impact of psychosocial factors over time. A mixed method design incorporating a quantitative methodology would provide points of comparison from baseline measures prior to testing and time sensitive information about psychosocial reactions to genetic testing for LQTS that retrospective in-depth interviews are limited in their capacity to do. Comparisons over time, especially from the point prior to diagnosis as a baseline, may also provide valuable information regarding any changes or predisposing psychosocial factors impacting individuals during the acute and chronic phases of the condition.

The study partly aimed to understand the psychosocial factors specific to the genetic testing process, but this was difficult to achieve when using a qualitative retrospective method of information collection. The method used meant patients had to recall their psychosocial responses rather than

report their current responses to a current situation. This can be problematic as automatic or unconscious psychological processes may occur to restore emotional equilibrium after a distressing event, such as receiving a LQTS genetic diagnosis (Horowitz, Wilner, & Alvarez, 1979). Some information salient to the initial psychological impact evident at the time may therefore have been lost or forgotten in this process.

In addition, the clinician interviews were significantly shorter in duration, and therefore content, from the patient interviews. This was largely because the clinicians had limited time available to be interviewed and perhaps also because the interview schedule was less structured than that used for the patients (see Appendices IV and V). The clinician data is consequently not equal to that of the patients in depth and breadth and as such limits comparisons between the two. The clinicians may have discussed more topics in more detail, should they have had more time or structured questions.

The study also has a relatively small sample size, especially when making between group comparisons, which has implications for generalising the results to the larger LQTS population. The findings are also based on the experiences of individuals and healthcare services in New Zealand and as such are limited in the extent they can be generalised to other nationalities. Currently little data exists on demographics in the New Zealand LQTS population so it is unclear how often LQTS occurs in specific ethnicities, genders, ages and genotypes in the New Zealand population. When the research was proposed it was expected a database with past and present demographic and clinical information would be available to fulfil this role. Unfortunately this was not the case. Participant selection therefore was completed according to the current system. This did not hold separate, easily accessible data on LQTS patients from which a cross section of all patients already identified with a genetic mutation could be taken. The gender rates of the present study (females 57% and males 43%) were broadly consistent with the general population (females 51% and males 49%) (Statistics New Zealand, 2002), which is indicated to also be representative of the LQTS population (Goldenburg et al., 2008). Previous research indicates Symptomatic and Asymptomatic patients represent approximately fifty percent of the LQTS population each whereas the current study rates somewhat under represented Symptomatic patients (36%) and Asymptomatic patients were overrepresented (64%) (Meulenkamp et al., 2008). The participants were also not as ethnically diverse, and there were fewer adolescents, than would be ideal for generalising purposes. Nevertheless the response rates (patients, 52%) and (clinicians, 73%) were reasonably high.

Discussion of Major Findings in Relation to Previous Findings

Psychosocial Impact of LQTS

Findings from the present study indicate there are differences between Symptomatic and Asymptomatic LQTS patients, with the psychosocial impact of LQTS generally more psychologically significant for Symptomatic LQTS patients. This was also the case for parents whose children were Symptomatic, which will be discussed further below. Primarily Symptomatic patients found the condition hard to adjust to because of the changes required to their (or their children's) daily life and having to psychologically adjust to living with the anxiety produced by cardiac events and the constant risk of sudden death. This is consistent with previous research findings specific to parental responses to their symptomatic children (Andersen et al., 2008; Farnsworth et al., 2006; Grosfeld et al., 2000; Hendriks, Grosfeld, Wilde, et al., 2005; Rabin et al., 2001) and with research on other inherited conditions (Fanos & Johnson, 1995; Meiser, 2005), but has not been identified in the general population of Symptomatic LQTS adults. Some Symptomatic patients experienced significant distress, particularly during the initial or acute phase of the condition, which led to high levels of illness related anxiety, agoraphobic responses and depression; a finding consistent with literature regarding acute phase responses to other chronic conditions (Drossman et al., 2000; Epker & Gatchel, 2000) and LQTS research on depression (Hintsa et al., 2010). This restricted their capacity to function and was beyond their ability to cope which, for some, posed a safety concern for themselves and their children. Psychological support was not offered in many of these cases. Asymptomatic patients, on the other hand, generally did not experience distress, other than that associated with their Symptomatic children. This is consistent with previous research findings for adults undergoing predictive testing, whom may be assumed to be Asymptomatic (K. S. Hendriks et al., 2008). Some Asymptomatic patients, however, found LQTS a difficult condition to reconcile and explain to others in the absence of symptoms, they also were adversely affected by the functional restrictions placed on them. Perhaps this was because no tangible symptoms were present to use as a frame of reference for understanding why it was necessary. Previous research has not investigated Asymptomatic responses and therefore this is a new finding.

While the difficulties of the Symptomatic group of patients has been described in previous research, mostly in relation to parent and child perspectives (Andersen et al., 2008; Farnsworth et al., 2006;

Hendriks, Grosfeld, Wilde, et al., 2005), a distinction between Symptomatic and Asymptomatic presentations and the affect this has on the psychosocial impact of LQTS has not been previously investigated and is a new finding. The anxiety and depression experienced by some adults, irrespective of their status as parents, is also a new finding in the LQTS literature. Previous research on other chronic inherited conditions, however, has found anxiety and depression to be prevalent, and more so in the acute phase of the condition (Egede, 2005; Rabin et al., 2001). Unfortunately the extent of any pre-morbid anxiety or depression was unclear so it was not possible to confirm previous research which indicated that pre-existing psychological distress, for example, may be a risk factor for psychological difficulties after receiving genetic test results (Hendriks, Grosfeld, van Tintelen, et al., 2005; Meiser, 2005; Morgan et al., 2008).

Findings indicate there were no differences between Symptomatic and Asymptomatic LQTS patients in respect to the specific psychosocial impact of the functional restrictions however. That is, the functional restrictions were difficult to deal with, irrespective of symptomatology, but mostly in those who were previously active in sports and for adolescents. Over and above having to give up activities previously enjoyed these individuals also found their inability to affiliate with the sport related social groups to which they once belonged produced a sense of social isolation and incompetence. The participants in this study may not have had physically active or adversely affected occupations and so did not discuss this potential difficulty. The clinicians understood that restricting an individual's functioning could be distressing but their explanations largely focused on the impact on occupational options. It may be that they were unaware of the effect on this group of previously sporty patients. While the difficulty of having to make the functional restrictions has been noted in previous research on LQTS in relation to parents (Andersen et al., 2008; Vetter, 2007) the specific factors potentially relating to age and pre-existing sporting involvement have not previously been identified.

Coping Strategies

Coping strategies included a combination of problem and emotion focused strategies. Included were strategies such as seeking information in order to educate oneself and make informed risk appraisals and finding emotional supports in family and others affected by LQTS. Others described using primarily emotion focused strategies such as denial and avoidance, sometimes through the abuse of alcohol and drugs. Differing needs for information between Symptomatic and Asymptomatic patients was also found, with Symptomatic patients requiring more than was provided by services.

Perhaps because Symptomatic patients have more salient reminders of the potential risks, and are generally considered to be at higher risk, more information is sought in attempts to reach a point whereby risk appraisals feel satisfactory or coping is possible. This has not been noted in previous research. The high use of problem focused coping such as information seeking is consistent with previous research on coping with chronic conditions (Penley et al., 2002).

What helped many patients cope was the support of their family, including their partners, which is consistent with previous research (Martire et al., 2005). When disparate coping strategies were used by partners, however, this had a detrimental affect on their relationship and feelings of mutual support. This has not been found in previous research and is therefore a new finding. Other factors that helped patients cope included positive reframing of potentially negative thoughts or events, not focusing on worries as this may become overwhelming, comparing one's situation with those worse off and promoting competence and effectiveness in one's self by helping others. Helping others has also been found to be beneficial in previous LQTS related research (Andersen et al., 2008). The other techniques used are, however, new findings regarding coping with LQTS.

Quality of Life

Previous research into the quality of life of patients with a chronic condition indicated negative psychosocial outcomes occur when four specific domains of functioning are affected, namely: physical functioning, psychological status, social functioning and condition or treatment related symptomatology (Kahn & Juster, 2002; Mittermaier et al., 2004). Findings from this study are broadly consistent with this premise, as those who found it the most difficult to adjust to LQTS and described being in significant distress experienced difficulties in all four domains. That is, they were Symptomatic and therefore experienced condition related symptomatology; these symptoms and the functional restrictions detrimentally impacted their ability to physically function; many withdrew from their social circles as they were sports based or because they struggled to cope with the fear of having a cardiac event in public; and illness related anxiety was common.

Impact of Genetic Testing

Genetic testing for LQTS per se did not lead to distress for many. Patients instead referred to the first diagnosis they received, whether it was clinical or gene based, as that which affected them the most. This time specific factor appeared to be more relevant to patients than their symptomatology or the type of testing used to provide the diagnosis (i.e. predictive or diagnostic), which dictated the

At-risk and Diagnosed subgroups (respectively). This is a new finding in the literature but requires further, preferably longitudinal, investigation to ascertain a causal relationship. A difference in psychosocial responses was again noted between Symptomatic and Asymptomatic patients with Asymptomatic patients experiencing little adverse impact, except in relation to a LQTS diagnosis in their child, especially if their child was symptomatic. These findings are consistent with previous research on predictive testing of adults (Hendriks et al., 2008); with other inherited conditions (Almqvist et al., 2003); and with parents in response to their child's diagnosis (Hendriks, Grosfeld, Wilde, et al., 2005). Symptomatic patients, however, tended to become fearful of dying after diagnosis, with each cardiac event maintaining and exacerbating this fear, and a feeling of loss of control and hopelessness for their future. A minority, however, were comforted by the treatments made immediately available to them, such as implantable devices. Both of these findings are consistent with those found in parents of children found to be gene positive for LQTS after predictive testing (Andersen et al., 2008; Hendriks, Grosfeld, Wilde, et al., 2005). Since no previous research is available regarding adults undergoing diagnostic testing or receiving a clinical diagnosis, as is the case for most Symptomatic LQTS patients, this is a new finding.

A difference was found between Symptomatic and Asymptomatic LQTS patients in the extent of difficulty they described when deciding whether to undergo predictive genetic for their children. Symptomatic parents often preferred not to gene test their children, for fear they or their children would not cope should they be identified as having a LQTS positive genetic status. This has also been suggested by previous research into inheritable conditions (Godard et al., 2007). The children of some of these parents had reported their desire not to be genetically tested, which may have influenced their parents. This has not been found in the literature. Clinicians also had difficulty deciding whether to genetically test children. They found themselves ethically unsure about the potential repercussions of such testing, but generally decided on the basis of whether the child medically benefitting from the testing would outweigh any potential adverse effects; a finding also found in previous literature (Wilfond & Ross, 2009).

Other Group Differences

On the whole, clinicians and LQTS patients' perspectives were largely consistent, although some factors discussed by patients were not indicated by clinicians. It is not clear whether this is, in part, because clinicians had less time and a less structured interview as aforementioned. Differences were found between Symptomatic and Asymptomatic LQTS patients and have been identified throughout,

however no specific differences emerged between At-risk and Diagnosed groups that wasn't already accounted for by Symptomatic and Asymptomatic differences. Commonly, the At-Risk group underwent predictive testing and were Asymptomatic. The Diagnosed group were usually those who underwent diagnostic testing and were Symptomatic. Some exceptions emerged in the current study, with some participants not aligning to these groups, and it was noticed that in these cases the presence or absence of symptoms appeared to mediate psychosocial responses more than the form of genetic testing or method of diagnosis used. This apparent trend requires targeted research before it can be conclusively asserted. Perhaps it can be hypothesised that to patients the presence or absence of symptoms has a powerful influence on their daily lives that is more significant and tangible than the reasons for their genetic testing.

Psychosocial Impact on the Family

As has been found in previous research parents were largely concerned about their children and grandchildren (Hendriks, Grosfeld, Wilde, et al., 2005). Parents worried about deciding whether to genetically test their children, about the ability of children to cope with genetic status, and about the effect of functional restrictions. They were also concerned for their children's future. These are all findings which are consistent with previous research (Godard et al., 2007).

Within a family, some partners felt burdened and responsible for the care and safety of their LQTS affected partner. This was also the case for some parents, who felt the weight of responsibility for caring for their LQTS positive children burdensome at times. This has not been reported in the LQTS research thus far and is another new finding. It has, however, been found in research relating to chronic conditions (Williams et al., 2002). Some looked to blame others for passing the condition on or blamed themselves for passing it on to their children, a finding consistent with previous research (Andersen et al., 2008). Close or mutually affected family members were consistently identified as the best supports for LQTS patients, a finding consistent with previous research on chronic conditions (Martire et al., 2005). All of these findings underscore the importance of addressing the familial needs alongside those of individuals, and of the capacity of the family to be it own best resource for psychosocial care.

Clinical and Healthcare Experiences

LQTS patients' experienced different qualities of healthcare. The positive experiences were mostly noted in response to the care patients received from CIDG and included being provided with an

efficient service and having clinicians who were well informed, supportive and approachable. However, many of those that experienced significant distress were not identified by clinicians, including CIDG, and consequently not supported.

When patients spoke of their healthcare experiences, many described a particular style of interaction between themselves and their clinician that afforded them a sense of emotional safety. LQTS patients and most clinicians generally found an honest, open and respectful approach were the most amenable to the psychosocial needs of the patients. A direct and detached style was not found helpful for most people, and generally considered inconsistent with their psychosocial needs. Some clinicians believed being direct was helpful, and perhaps it is in some instances, but on the whole it was not preferred. No previous research was found on the preference for this interpersonal communication style aspect of psychosocial care for LQTS patients.

The negative patient experiences were largely in relation to services provided by GPs, referring Cardiologists and Specialists assisting births. Some LQTS patients were unsupported by their appointed Specialist during what were planned to be Specialist supported births because their implantable devices required monitoring and needed to be made inactive during the final stages of birth. It is fortunate no harm came to mother or baby during these unassisted births. It is unclear which specific Specialists were required, but may include Cardiac clinicians and/or Obstetricians. Difficulties have been found in the international research on the healthcare provided by GPs and referring Cardiologists (Andersen et al., 2008; Hendriks, Grosfeld, van Tintelen, et al., 2005; Farnsworth et al, 2006) but not in respect to Specialists assisting births and as such this is a new finding.

Many also had negative experiences with their primary and secondary care physicians (GP's and referring Cardiologists or Pediatricians). This is consistent with international findings (Andersen et al., 2008; Farnsworth et al., 2006; Hendriks, Grosfeld, van Tintelen, et al., 2005). For example, some patients experienced their concerns not being believed or put down to anxiety, a finding also consistent with international literature (Andersen et al., 2008; Farnsworth et al., 2006). Incidences of missed diagnosis and misdiagnosis from general practitioners and referring cardiologists were common. For several, this had caused significant distress, physical complications and, for two, resulted in tragedy. This is consistent with experiences internationally (Andersen et al., 2008; Farnsworth et al., 2006) and underscores the need for education of general practitioners and

cardiologists. Consistent with findings from NZ based research (MacCormick et al., 2009) and international sources (Burghaus et al., 2010) all had been misdiagnosed with epilepsy. The clinicians interviewed in this study were aware misdiagnosis was a significant problem and the first research investigating this concern was conducted through CIDG in New Zealand (MacCormick et al., 2009). This is perhaps testament to CIDG's commitment to highlight the problem within the national and international arena.

In defence of these primary and secondary physicians, the international literature and the CIDG clinicians in this study (who are experts in LQTS) agree it is a difficult condition to diagnose, especially without genetic testing (Schwartz, 2006). In addition, as CIDG clinicians point out, it is a relatively rare condition affecting approximately 1:1000 at most (Crotti et al., 2008) and as such is probably not something primary and secondary clinicians see frequently enough to develop a thorough understanding of it. New Zealand and internationally based studies have demonstrated GPs in particular have limited knowledge about inherited conditions, genetic testing, and assessing genetic based familial risk. This is perhaps a reflection of gaps in their training (Emery et al., 1999; Morgan et al., 2004; Sarfati, 2002). More recently trained physicians in New Zealand, however, demonstrate more knowledge of gene based conditions (Cameron et al., 2002).

Research Implications

Longitudinal Quantitative Research into Psychosocial Impact of Genetic Testing for LQTS

A longitudinal, largely quantitative, research project into the psychosocial impact of genetic testing for LQTS over time has recently been conducted in Australia and is in the process of reaching publication. Conducting a New Zealand quantitative study of a similar nature (as originally planned for the current study, see p. 56) may enable cause and affect relationships between factors to be demonstrated, especially in relation to the time specific factors impacting LQTS and the genetic testing process. Comparisons over time, especially from the point prior to diagnosis as a baseline, may have also provided valuable information regarding any changes or predisposing psychosocial factors impacting individuals during the acute and chronic phases of the condition. Findings may also be able to be compared with outcomes from this study and represent the beginning of an Australasian LQTS psychosocial knowledge base.

Longitudinal Research Targeting Psychosocial Impact of Clinical Compared to Gene Based Diagnosis for LQTS

Findings in the present study suggest that the first clear diagnosis (clinical or gene based) may be the most salient to LQTS patients irrespective of the form of genetic testing they undergo. Longitudinal research measuring and comparing the psychosocial impact at clinical and gene based diagnosis points may further clarify this finding and potentially demonstrate a cause and effect relationship.

Clarification of Group Differences

As aforementioned, there appeared to be a difference in responses depending on the presence or absence of symptoms but no differences between At-risk and Asymptomatic patients, or between Diagnosed and Symptomatic patients. It may be that looking at the absence or presence of symptoms in LQTS is more meaningful in understanding psychosocial responses than researching the method of genetic testing. Research investigating the differences in the psychosocial adjustment factors identified in this research between (a) Symptomatic and Asymptomatic and (b) those undergoing predictive testing compared to diagnostic testing, while controlling for presence and absence of symptoms, may be useful in clarifying and confirming the current findings.

Research Targeting Psychosocial Impact of Specific Forms of LQTS with Additional Physical and Functional Difficulties

Some participants had relatives with a rare and unique form of LQTS (Jervell and Lange-Neilsen Syndrome) that includes additional physical and functional difficulties. While comparisons were not made in the present study, it was observed that the psychosocial impact of this form of LQTS may differ from that associated with the more frequently occurring forms of LQTS (LQT1-3). This issue may, therefore, benefit from targeted research to uncover any unique psychosocial responses and needs; as it would for other unique forms of LQTS with additional physical and functional difficulties, including Timothy Syndrome and Anderson Tawil Syndrome.

Best Method of Educating Primary and Secondary Clinicians

The need for GPs and referring secondary clinicians to be further educated regarding LQTS is clear. This may be particularly so for less recent graduates. An investigation into how this might best be achieved may be highly advantageous in ensuring the information is presented in a way that meets their needs.

Specialists Attend Births

If a Specialist is part of a birthing plan then it is assumed the Specialist is required to arrive in time for the birth. The current study found examples of this not happening. It is recommended, therefore, that a review of Specialists' birth attendance record in LQTS cases be conducted in New Zealand to ascertain whether these were isolated cases and, if not, measures be taken to ensure Specialists are there in time to perform their potentially life saving role. Perhaps the views of the experienced birthing mother may also be used an indicator of the approach of the final stage alongside that of the midwife.

Do Parental Information Requirements Differ According to the Presence of Symptoms

Information requirements differ between Symptomatic and Asymptomatic patients. Future research is needed to ascertain whether children's information needs also differ in respect to the presence of symptoms, and whether parents respond and meet these needs.

Familial Dominant Coping Strategies

It was observed that parents who reported predominantly coping through avoidance reported their children also preferred to avoid finding out their genetic status; thereby avoiding having to cope with the distress of a potential positive gene status. This may suggest families have mutually reinforcing dominant coping strategies that in the case of LQTS may put families at risk for psychological distress or at-risk for cardiac events; as avoiding genetic testing may also be a missed opportunity for potentially life saving prophylactic treatment. Larger studies are needed to ascertain whether this finding is replicated.

Clinical Implications

Symptomatic Patients Require More Support

Findings from the current study indicate Symptomatic patients require more support in terms of both the provision of additional information and psychological assessment for signs of illness related anxiety or depression. Parents of Symptomatic children, and partners of Symptomatic adults, may also require additional support should they become burdened by caring for their children or partners. It is recommended that clinicians be aware of the potentially adverse impact cardiac events can have on both individuals and families and be vigilant for signs of distress so appropriate supports can be put in place.

Educate Primary and Secondary Level Clinicians

The education of primary clinicians such as GPs and secondary clinicians might best include a focus on the importance of taking action on patients concerns, differential diagnosis and follow up care of LQTS. It may also be beneficial to clearly identify how to access specialist LQTS services for clarification of diagnosis, to obtain further information or to ascertain how to make a referral for genetic testing when necessary.

Utilise Multidisciplinary Team Where Possible

Clinicians suggested Cardiologists may not be the ideal providers of emotional support. While this may be the case for some Cardiologists, especially those who are perhaps less comfortable with patient interaction with emotional content, findings indicate some clinicians who perform other roles within a multi-disciplinary team are able to meet these needs and frequently do. The multidisciplinary team approach therefore may help meet patients' psychosocial needs by providing multiple opportunities for emotional support from different members of the team. However, this relies on one of the team having these skills by chance and that they are able to take on this role. This may not always be possible and, as time is frequently a factor, may not be a priority. It may be beneficial, therefore, for members of the team less skilled with emotional content, such as Cardiologists, to receive emotion focussed or psychological training and support. It may also be beneficial for a specific counsellor or psychologist to be part of the multidisciplinary team for the dual purpose of supporting clinical staff and patients, and providing psychological assessment and interventions when patients indicate they are in significant distress.

Establish Support Groups and Education Sessions

This may provide patients with a space to share experiences and thereby normalise their own; to validate concerns and feelings; and to develop their own feelings of competence and effectiveness by providing them a space in which to help others. Providing education sessions as a shared activity may also provide opportunities for more vocal members to ask questions which the more reticent may be reluctant to ask, and to foster further mutual support. It may also enable the provision of accurate information to a wider base while also being an opportunity to provide group based follow up and education around new developments or issues of long term care.

Provide Psychological Support

Psychological support by a professional skilled in psychological assessment and interventions, such as a Psychologist, may identify those at risk individually, as a family, and provide psychological interventions for those in distress. This might be best completed within the acute period of the condition between the point of diagnosis and long term follow up during an annual check up for instance. It may also be useful to include those avoiding genetic testing when it is indicated to be of potential medical benefit. Particular psychosocial factors requiring intervention may include lack of supports, social isolation, previous high involvement in sports, feeling burdened by the care of others, lack of information or understanding of the condition and high levels of illness related anxiety or depression. A psychological assessment for safety to self and others (e.g. their children) is imperative for those showing distress (as evidenced by the two participants who reported suicidal and homicidal ideation in relation to their children). For the family, an additional assessment of partners' or parents feelings of burden for caring for their affected partner/child and the degree of distress this is causing is also indicated and intervention in these instances may help the family as a whole cope more effectively. Studies on psychological therapy have found it to be effective in reducing psychological distress for patients with a variety of illnesses, including cardiac conditions, HIV, breast cancer and rheumatoid arthritis (Astin, Beckner, Soeken, Hochberg & Berman, 2002; Beltman, Oude Voshaar, & Speckens, 2010; Himelhoch, Medoff & Oyeniyi, 2007; Linden, Phillips & Leclerc, 2007; Tatrow & Montgomery, 2006). Cognitive behaviour therapy and mindfulness-based interventions have been demonstrated to be effective in decreasing psychological distress in acute and chronic illnesses (Beltman, Oude Voshaar, & Speckens, 2010; Kabat-Zinn, J., Lipworth, L. & Burney, R., 1985).

Develop a Diagram to Explain LQTS

Being a difficult condition to understand and explain, developing a diagram and brief explanation that can be used to explain the condition to patients would be beneficial. This may act as a visual reminder of how to construct a clear understanding and may be useful if taken home and used to explain the condition to potential supports. It may be especially useful for Asymptomatic patients if the brief explanation includes reference to the absence of symptoms in some people with LQTS.

Provide Individually Tailored Information

For many, gathering information enabled them to cope more effectively. The quantity required differed, however, according to the presence or absence of symptoms; with Symptomatic patients requiring more. This underscores the need for multiple levels of breadth and depth in the information provided from healthcare services. The internet was often cited as a resource for additional information and therefore it may be beneficial to patients if a list of useful websites be provided. Cites included might best be a cross section of informative options and support options such as provider supervised forums. In addition, parents often act as the informants for their affected children (Kendall et al., 2003a, 2003b) and although it is unclear whether parents of Symptomatic children also require more information it is equally important they be given adequate information to meet the needs of their family.

Recommendations to CIDG

In the consultation phase of this study CIDG principally wanted to know how well the service they provided met the psychosocial needs of their LQTS patients. In summary, most patients had wholly positive experiences of the tertiary specialist CIDG service in New Zealand. This positive appraisal of tertiary services is echoed in the international literature (Farnsworth et al., 2006). This may in part be due to the ability of a tertiary service to provide a multidisciplinary team of genetic, medical and support professionals to meet LQTS patients' multiple needs. Perhaps this is also because of the commitment CIDG clinicians demonstrate by their recommendations and the solutions they have developed in addressing their impression of the difficulties faced by LQTS patients.

There are some areas that are potentially in need of addressing, however. These are provided here as recommendations. The following recommendations to CIDG are over and above the clinical implications discussed above, which should also be taken into consideration. In some instances, recent changes in CIDG systems have to some extent already addressed difficulties and as such are also reported to provide an up to date impression.

Psychologist in Multidisciplinary Team

Currently, the services of a Psychologist can be utilised by CIDG when a need is identified by clinicians. Patients living within the region of the national hospital are also routinely asked if they would like to see a Psychologist. Findings from the current study suggest that some patients benefit

from psychological intervention when utilised, a finding which is consistent with previous research on the efficacy of psychological treatments for people with various conditions, including cardiac conditions. Interventions such as cognitive behavioural therapy have been shown to be effective in reducing distress in patients with these and other chronic conditions (Beltman, Oude Voshaar, & Speckens, 2010; Linden, Phillips & Leclerk, 2007). Many patients, however, described a reluctance to engage in psychological services, suggesting that to do so may be tantamount to admitting a weakness in their ability to cope that is unacceptable. This may be attributable in part to a social culture that perhaps continues to evaluate psychological intervention as stigmatising and only for those at the extreme end of adverse psychological functioning. Recent New Zealand wide media campaigns are in progress to demystify, destigmatise and normalise psychological difficulties and thereby encourage the use of psychological interventions but until the time that this becomes socially acceptable in New Zealand culture getting people to opt in for therapeutic intervention may remain a problem. It is recommended therefore that efforts be made by CIDG to demystify and normalise psychological services by discussing what's involved, the potential benefits and what others just as 'normal' as themselves have found beneficial during a clinic consultation. Having a Psychologist present from the point of contact in clinics may allow individuals to see who they will be meeting, and especially if a rapport is developed with this person and an invitation is given directly from them, rather than as a referral to an unknown service, they may be more likely to engage. In addition, having a Psychologist present at clinics might better facilitate the recommendations for assessment and intervention discussed earlier (p. 134).

One of the advantages of a multidisciplinary team discussed earlier (p. 133) is its ability to utilise the skills of all its members to best meet the needs of its patients. Findings from the current study indicate some CIDG clinicians struggle to attend to their patients' psychosocial needs during clinics and the task of providing psychosocial support has largely fallen to the Registry Coordinator. This has potentially put the Registry Coordinator under a lot of pressure to fulfil dual roles which as the service and Registry grows will probably become unmanageable, if it is not already. In addition, findings from the current study show some patients can experience significant psychological difficulties that require specialist psychological assessment to identify and intervene when necessary. A specialist Psychologist as part of the multidisciplinary team is recommended, therefore, to compliment the multidisciplinary team in providing psychosocial support during clinics, to allow the Registry Coordinator to focus on further developing the Registry and service, and to provide risk assessments and psychological interventions when necessary.

Decreased Waiting Time for Genetic Test Results

A speedier genetic testing process is required, because of the adverse emotional toll the waiting took on them and their families. Patients and clinicians discussed the need for a speedier, localised genetic testing process. This has been achieved since this research was conducted. Genetic testing is now done in New Zealand and the wait time for results is significantly reduced. It now takes approximately three months for a proband and approximately two months or less for a cascade; provided the sample is of good quality.

Increased Follow Up

Patients in the current study indicated several problems associated with follow up after receiving genetic test results through CIDG. The principal problem was the lack of knowledge on the part of the primary or secondary care physician they were referred back to. It is recommended therefore that an educational pack, in addition to a clinical letter, be provided to these physicians and access avenues to CIDG be provided to ensure patients receive appropriate follow up care. Should it be possible through the Registry or database, it would be desirable to maintain a list of these physicians so that regular updates can be sent regarding contraindicated medications and new LQTS related developments. This may be able to be achieved electronically and thereby have less associated cost. Another problem was regarding follow up leading up to the dissemination of genetic test results. While the wait is much less now, should delays be encountered it is recommended patients be contacted directly and the reason, and expected length of delay, be explained. The current findings indicate patients benefit from these explanations and are consequently more able to adapt to the wait.

Increased Time in Clinics

Several clinicians recommended more regular LQTS consultation clinics to meet demand and to provide more time for patients to ask questions and voice concerns. It is understood clinics are now happening on a more regular basis and that these are more adequately meeting demand for clinics. It is unclear however whether adequate provision has been made for patients to ask questions and voice concerns and, perhaps more importantly, for clinicians to feel less time pressured and therefore to be more able to respond to patients' psychosocial needs.

Reduce Loss of Files, Samples and Documents

Delays in getting genetic testing completed were encountered due to the loss of forms and samples, although it was unclear whether these originated from CIDG. CIDG and referring services' organisational systems may therefore benefitted from some improvement. It is understood, however, CIDG may now have addressed this problem as they have a database established as part of the National LQTS Registry for organising and holding LQTS related information. In addition, since this study was conducted CIDG, having obtained national ethics approval, have standardised the consent, collection and genetic testing process for LQTS samples nationwide. This will hopefully prevent the loss of files, samples and forms in the future.

Meet Psychosocial Patient Needs Across Team and Nationally

One Cardiologist recommended a nationalised service that was centrally funded to provide a consistent service to all areas in NZ. This may increase the likelihood that clinicians who are appropriately trained about LQTS and its medical management care for LQTS patients. This might be best achieved using a multidisciplinary team approach that includes a specialist psychological role as aforementioned.

Dissemination

The final step for this study is to present the findings to CIDG and the LQTS patients. The report will include a summary of the research findings and recommendations that emerged from the study. It is considered the researcher's responsibility to present findings to key stakeholders so as to potentially facilitate implementation of the findings. Also to this end, it is intended the results will be published in a peer reviewed journal accessible to primary and tertiary care clinicians, genetic counsellors and psychologists.

Conclusions and Reflections

The current study aimed to broaden the previous research on the impact of LQTS that has thus far concentrated almost exclusively on the impact of predictive genetic testing from the perspective of parents and their children or youth. This research has endeavoured to be broad, providing an insight into the psychosocial impact of adults and adolescents living with LQTS and undergoing both diagnostic and predictive genetic testing for LQTS. This has allowed comparisons to be made that have not yet been identified in the developing LQTS psychosocial literature. Most notable in the

new findings is the apparent distinction in the psychosocial responses between Symptomatic and Asymptomatic patients, with Symptomatic patients experiencing more psychosocial adverse responses to genetic testing, and to living with the condition, than their Asymptomatic counterparts. These responses can be significant and include anxiety, depression and agoraphobia. These responses occur mostly within the acute phase of the condition and are impacted more by the first diagnosis provided by clinicians rather than the form of genetic testing undergone. Perhaps this is because patients look to a diagnosis for clarity, irrespective of whether it is clinical or gene based.

Some Symptomatic patients were able to cope with the diagnosis, however, especially when using both emotion and problem focused coping strategies. More specifically, the problem focused strategy of gathering information in order to validate and normalise their psychosocial experiences, and to make risk appraisals that action could be taken to reduce the risk of cardiac events, seemed most successful in the long term. Family support was also important for coping, irrespective of symptomatology. However disparate coping strategies between partners, and a sense of burden in some carers, had a detrimental effect.

While Asymptomatic patients may not experience the same level of distress, they did experience distress with respect to their Symptomatic children. They also had difficulty reconciling for themselves, and explaining to others, the severity of their condition in the absence of symptoms. They also experienced the adverse effects of the functional restrictions in the same way as those who were Symptomatic, especially if either group had been involved in sports prior to receiving a diagnosis. Adolescents, who are perhaps more active in sports groups, also found the functional restrictions difficult to manage.

The clinical care received by patients had a significant impact on LQTS patients. Overall, patients were impressed with the clinicians and systems in place at CIDG but those that experienced significant distress had not been identified and were consequently largely unsupported by CIDG, perhaps underscoring the need for a psychology specialist as part of the multidisciplinary team. The most significant concern however, is in respect to the unassisted specialist births and high rate of misdiagnosis, missed diagnosis and consequent tragic outcomes of relatives of those diagnosed with LQTS. Each of these demonstrates the urgent need for training of primary and secondary physicians and a review of specialist assisted births to ascertain if these were isolated cases.

In the process of completing this research I have been struck by the commitment of CIDG clinicians to provide a service that meets both medical and psychosocial needs. These clinicians really do go above and beyond what is probably expected in their roles to provide the best they can. This cannot help but make one proud to be completing research that may contribute to this process. The commitment of CIDG clinicians is demonstrated by their initiation of this research and it is hoped the findings go some way to answering their questions.

During the course of this research I have also been training to become a Clinical Psychologist and in doing so have developed a burgeoning fascination with the responses of those coming to terms with having a chronic illness. Little did I know at the time that this would become particularly poignant for my family as our son developed a chronic condition immediately after the interviews were completed. We felt the impact for ourselves. The patients who allowed me into their lives and shared themselves so openly helped me and are an inspiration to us all. Without their participation there would be no way of deepening our understanding or improving our services to benefit people like my family, son and I. For this I thank them and as it seems only fitting, I leave the last words to those who know the psychosocial impact of LQTS the best:

> I guess when you're told you know you've got this condition and I was like "oh my goodness, I'm going to die".
>
> I mean cause sometimes you think 'oh why me?' and but you've got to think 'well, you know, why not me?' and 'what can I do with it?'.

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APPENDIX I

THE UNIVERSITY OF AUCKLAND
NEW ZEALAND
Te Whare Wānanga o Tāmaki Makaurau

Department of Psychology
University of Auckland
Private Bag 92019, Auckland
New Zealand

Building 721,
200 Morrin Road,
Glen Innes
Reception: Level 3
Ph: 3737599 ext 84990
Fax: 373-7450

PARTICIPANT INFORMATION SHEET

We would like to invite you to participate in a study exploring the psychological impact of genetic testing for Long QT Syndrome has on adults, adolescents, children and their families. Your participation is entirely voluntary (your choice). You do not have to take part in this study, and if you choose not to take part, this will not affect any future care or treatment by Paediatric and/or Congenital Cardiac Services or at the Cardiac Clinic. If you do agree to take part, you are absolutely free to withdraw from the study at any time, without having to give a reason.

Project Title:
The psychological impact of genetic testing for Long QT Syndrome in children, adolescents, adults and their families: A New Zealand study.

Researchers:
Senior Lecturer and Senior Clinical Psychologist Dr Heather McDowell, Doctorate of Clinical Psychology candidate Sarah Watson, Lecturer Dr Jeanne Reeve and Associate Professor Linda Cameron, Department of Psychology, University of Auckland. Leah Andrews, Child and Adolescent Psychiatrist, Starship Children's Health and Senior Lecturer, Department of Psychological Medicine, University of Auckland. Consultant Paediatric Cardiologist/Electrophysiologist Jonathan Skinner, Cardiac Inherited Disease Registry Co-ordinator/Senior Technologist Jackie Crawford, Clinical Registrar/Fellow Judith MacCormick and Maori Advisor/Social Worker/Therapist Matthew Shepherd, Auckland City Hospital & Starship Children's Health

About the study:
In this part of the study, we are exploring the experiences of those undergoing genetic testing for Long QT Syndrome (LQTS), looking at what areas were the most difficult and how Paediatric and/or Congenital Cardiac Services (ADHB) at the Cardiac Clinic helped or not during this time.

We are inviting individuals who have received genetic testing results indicating the presence of a LQTS genetic mutation under the care of Paediatric and/or Congenital Cardiac Services (ADHB) at the Cardiac Clinic Auckland City Hospital or Starship Children's Health to participate in this part of the study. We aim to recruit up to 15 participants over the age of 16 years. The Clinical Registrar or Cardiac Inherited Diseases Registry Co-ordinator will already have told you briefly about this study in their letter to you enclosed and/or when they telephoned you. Being still a fairly new initiative, we believe that the findings of this study will provide a basis for understanding factors that impact those undergoing genetic testing for LQTS and isolating what currently helps and what else might help better in the future. This information will be useful in developing informational materials that will

improve people's understanding of LQTS and the genetic testing process, as well as possibly identifying additional services that may be helpful at particular key times.

About your participation:

There are some initial questions which should take a couple of minutes to complete and are asking for some general information about you, your LQTS and your family. This will happen after you first meet the interviewer (Sarah Watson and possibly also Matthew Shepherd) at a time and place of your choice. An interview that is expected to take between 20 minutes to an hour will then be conducted. The questions will be about how it has been for you since you first became aware of LQTS affecting you through to now, including when you received the genetic test results. There are also questions about what things were helpful or not in terms of the service provided by Paediatric and/or Congenital Cardiac Services and CIDG.

At the end of the study, you will receive a report of the study findings. This report will be sent to your preferred address or we can arrange to meet with you and discuss these in person, please indicate your preference on the Consent Form.

Participants who have not received a genetic test result identifying the presence of the genetic mutation for Long QT Syndrome under the care of Paediatric and/or Congenital Cardiac Services will be excluded, as well as those under the age of 16. Lastly, as the study requires answering questions in languages known to the interviewers, potential participants will also be excluded if they are not able to communicate verbally in English or Maori. Please note Auckland DHB Maori Ethics Committee do not support tissue banking in any form, given collective whanau, hapu, iwi, whakapapa implications. However they have noted individuals have the right to make their own decisions.

Your participation is entirely voluntary; that is, it is by your own choice. You do not have to take part in this study. If you do agree to take part, you are free to withdraw from the study at any time without giving a reason, including stopping the interview at any point and not continuing. You do not have to answer all of the questions.

There are no known risks caused by this study. However, there is a chance that you may experience increased worry about your genetic test result, cardiac condition, or treatment experience. In the unusual circumstance that you were to experience significant worry and emotional distress, then a clinical psychologist at the Consult Liaison Team, Starship Children's Health, is available for consultation and assistance. Please contact Sarah Watson or Heather McDowell in this instance, the contact details are to follow. In addition, though you may not benefit directly from participation, it is possible that the study findings will lead to a better understanding of what factors impact those going through the Long QT Syndrome clinic process the most, potentially identifying areas for future improvement and/or additional services, information etc. As a client of Paediatric & Congenital Cardiac Services at the Cardiac Clinic, your decision to participate or to not participate in the study *will not* affect your relationships with the clinical staff. We understand and are grateful for the positive contribution you may be making to knowledge.

All of your responses will be dealt with in ways that prevent anyone other than the researchers from identifying you as their source. Code names will be used through the study so that your name will not appear in any electronic datasets or research reports. Any data published in research journals will be presented as group averages and percentages so that it will not be possible to identify you as the source of any of the information. Data will be stored in a password-protected database on a password-protected computer or in a locked cabinet in a locked office at the University of Auckland.

The data will be deleted or shredded 10 years after the publication of the study findings. You may have a friend, family or whanau support to help you understand the risk and/or benefits of this study and any other explanation you may require.

In the unlikely event of a physical injury as a result of your participation in this study, you may be covered by ACC under the Injury Prevention, Rehabilitation and Compensation Act. ACC cover is not automatic and your case will need to be assessed by ACC according to the provisions of the 2002 Injury Prevention Rehabilitation and Compensation Act. If your claim is accepted by ACC, you still might not get any compensation. This depends on a number of factors such as whether you are an earner or non-earner. ACC usually provides only partial reimbursement of costs and expenses and there may be no lump sum compensation payable. There is no cover for mental injury unless it is a result of physical injury. If you have ACC cover, generally this will affect your right to sue the investigators. If you have any questions about ACC, contact your nearest ACC office or the investigators.

Please Contact Us For More Information

Please contact us if you have any questions or concerns about the survey, if the study raises any questions or concerns about your test result or care, or if you would like more information about the study.

Please contact one of the following members of the University based research team:

Heather McDowell (Senior Lecturer and Senior Clinical Psychologist), Sarah Watson (Doctorate of Clinical Psychology Candidate and Trainee Clinical Psychologist), or Jeanne Reeve (Lecturer), Department of Psychology, University of Auckland, Private Bag 92019, Auckland. Phone: (09) 373-7599 ext. 88556 (for Heather), ext. 84990 (for Sarah) or ext. 83873 (for Jeanne).

You may also contact the head of the Psychology Department:

Associate Professor Fred Seymour, Department of Psychology, The University of Auckland, Private Bag 92019, Auckland, Phone: (09) 373-7599 ext. 88414

For Maori health support, or to discuss any concerns or issues regarding this study, please contact Mata Forbes RGON, Maori Health Services Co-ordinator / Advisor, 5th Level, GM Suite, Auckland City Hospital. Phone: 307 4949 extn. 23939 or Mobile 021 348 432.

If you have any queries or concerns regarding your rights as a participant in this study, you may wish to contact an independent Health and Disability Advocate on 0800-555-050, free fax 800 2787 7678 (0800 2 SUPPORT), or email advocacy@hdc.org.nz .

This study has received ethical approval from the Northern X Regional Ethics Committee on 6th December 2006 for two and a half years. Reference Number NTX/06/12/158.

APPENDIX II

THE UNIVERSITY OF AUCKLAND
NEW ZEALAND
Te Whare Wānanga o Tāmaki Makaurau

Building 721,
200 Morrin Road,
Glen Innes
Reception: Level 3
Ph: 3737599 ext 84990
Fax: 373-7450

Department of Psychology
University of Auckland
Private Bag 92019, Auckland
New Zealand

INTERVIEW CONSENT FORM

Project Title:
The psychological impact of genetic testing for Long QT Syndrome in children, adolescents, adults and their families: A New Zealand study.

Researchers:
Sarah Watson, Dr Heather McDowell, Dr Jeanne Reeve, Dr Leah Andrews, Mr Jonathan Skinner, Jackie Crawford, Dr Judith MacCormick and Matthew Shepherd.

- I have read and understand the information describing the aims and content of the study designed to explore the experience and psychological impact of genetic testing for Long QT Syndrome.
- I have had the opportunity to ask questions and have them answered. I have had the opportunity to use family/whanau support or a friend to help me ask questions and understand the study.
- I understand that, by submitting this consent form, I agree to take part in this research under the terms indicated in the Participant Information Sheet.
- I understand that taking part in this study is voluntary, and my participation or non-participation will not affect my treatment or my clinical relationships with the employees at the Paediatric and/or Congenital Cardiac Services or Cardiac Clinic in any way.
- I understand that my participation in this study is confidential and that no material which could identify me will be used in any reports on this study.
- I have had time to consider whether to take part in the study.
- I agree for the interview to be audiotaped and my data stored in a locked cabinet and a password-protected database for a period of 10 years.
- I understand that I have the right to withdraw my information up to two weeks after the interview without giving a reason, but that my study responses cannot be withdrawn from the study after this date.
- I know who to contact if I have any questions about the study.
- I agree to take part in this research.

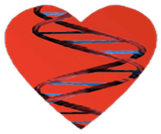
Participant Name:_____

Participant Signature:_____ Date: _____/ _____/ _____

At the end of the study, I prefer to receive the group results by post to my preferred address ☐ or I prefer this be done in person ☐ (please tick which applies).

At the end of the study I wish to have a copy of the audiotape posted to my preferred address ☐ or I prefer to have the audiotape destroyed ☐ (please tick which applies).

Appendix III



DEPARTMENT OF PSYCHOLOGY
FACULTY OF SCIENCE
THE UNIVERSITY OF AUCKLAND

Cardiac Inherited Disease Group

INVITATION TO HELP IN A RESEARCH ON THE IMPACT OF LONG QT SYNDROME

Hello,

Long QT Syndrome is one of the many cardiac inherited diseases that the Cardiac Inherited Diseases Group (lead by Dr Jon Skinner at Auckland City Hospital) monitors.

As part of our work with Long QT Syndrome and commitment to increasing knowledge, we are interested in finding out how Long QT impacts individuals and families. In order to answer these questions, we have asked Sarah Watson, from the Department of Psychology at Auckland University, to undertake this study. This part of the study includes an interview with Sarah Watson to provide potentially important information that may contribute to the development of future services.

The decision to participate in this study is entirely voluntary (your choice). If you are interested in finding out more, further information is included with this letter in the 'Information Sheet'. Should you then be happy to be contacted by Sarah Watson to either have any questions answered and/or arrange a time for interview at a place of your convenience, either fill in your details in the section below and mail it to Sarah at the following address or email her your contact details.

Thank you,

Jon Skinner
Cardiologist

Jim Stewart
Cardiologist

Jackie Crawford
Coordinator: Cardiac Inherited Disease Registry

Sarah Watson
Department of Psychology
University of Auckland
Private Bag 92019
Auckland
spay001@ec.auckland.ac.nz

Name: _____

Address: _____

Phone Number: _____

Mobile Number: _____

E-mail address:_____

Appendix IV

Patient Interview Outline

LQTS:

1. Orient to 1st awareness of LQTS e.g., when did you first become aware you may have Long QT Syndrome?
2. Ascertain which services involved
3. What was the impact at that time
    - Were Clinicians/services aware of the impact this was having
    - Is there anything Cardiac Services could have done to help
4. Understanding of what LQTS is e.g., what causes Long QT?
5. Status e.g., what symptoms of Long QT do you have?
6. Treatment receiving
    - Medication adherence
    - If not, what got in the way?
    - How could Cardiac Services help with this?
7. Risk appraisals
8. Medical care before receiving the genetic test results?
    - What useful
    - What could have helped
9. Family history of LQTS?
    - Experiences
    - Have relationships changed as a consequence – good or bad

Genetic Testing:

10. Concerns or worries about being genetically tested
    - Were Cardiac Services aware of these?
    - How did/could Cardiac Services have helped with this?
11. Enough information about being gene tested given at the time? (want more or less)
    - What else might have been helpful?
12. Length of wait for results
    - What was wait like
    - Were Services/Clinicians aware of impact
    - What hospital staff offered at this time
    - What would have been helpful
13. Understanding of genetic test result
14. Positives and negatives about the way results were given
    - What Services/Clinicians could do to help with this
15. Impact of the results
    - Service/Clinicians aware of impact?
    - Did the impact change over time
    - What helped/would have helped
16. Illness perceptions

17. Benefits re genetic testing e.g., what do you feel are the good things about being genetically tested for Long QT? What good things came out of it?
18. Negatives re genetic testing
    a. What do you find helps? - coping
    b. How do you think Cardiac Services could help with this?
19. How do you feel about your decision to be tested now?

Psychological Factors:

20. Worries/Anxiety
    - Preoccupying intrusive thoughts e.g., do you spend time thinking about Long QT? Nature, frequency, duration, intensity. Check restrictions on functioning or enjoyment, sleep disturbance, physical symptoms, safety behaviours(avoidance)
    - Coping e.g. how do/did you make yourself feel better?
21. Trauma
    - Re-experiencing, numbing. Frequency, duration, intensity. Check restrictions on functioning or enjoyment, sleep disturbance, re-experiencing, numbing
    - How do you make yourself feel better?
2. Depression
    - Mood, weight/appetite changes, negative bias, sleep disturbance, decreased interest, harm
    - Risk/safety

Supports:

22. Key supports during the time between being aware of possibly having Long QT through to having received the genetic test results?
23. Who in family talks to about Long QT, why them? When?
24. Talk to friends or colleagues about having Long QT?
    - If yes, how is that?
25. Talked to anyone else outside your family who has Long QT?
    - If so: How many and how was that?
    - If no: Would you like to?

Recommendations

26. Service recommendations

27. Peers - What would you say to other people going through the same thing? (about to undergo genetic testing for Long QT)

Appendix V

Clinician Interview Outline

Discuss anonymity, confidentiality

What do you think are the significant psychological and emotional challenges LQTS patients' experience?

What do you think are the significant psychological and emotional challenges LQTS patients' experience when they go through the genetic testing process?

What are you able to do to help mediate your LQT patients' emotional or psychological experience? That is, what works to support them emotionally and psychologically?

What kind of things have you found or seen that don't work?

If you had additional money and resources available and you were asked to design a service that would best meet your LQT patients' physical and psychological needs. What things, additional to what you have now, would you implement?