LONG QT SYNDROME - INFORMATION SHEET

Background
Long QT syndrome (LQTS) is an uncommon hereditary disorder characterised by abnormal electrical activity in the heart. It affects mostly children and young adults with frequent faints or even sudden death. The most common form of LQTS is inherited in a dominant pattern, which means that each child of an affected parent has a 50% chance of inheriting the disorder. Once a diagnosis is made, treatment is available. However, an accurate diagnosis can be difficult, because not all people affected by LQTS become ill and display symptoms.

Usually, LQTS can be seen on an electrocardiogram (ECG). Every heart beat is triggered by an electrical signal that tells the heart’s muscle cells to contract. After contracting, these cells must recover – or relax – before the next heart beat is initiated. The amount of time needed by these cells to recover can be measured on an ECG and this is called the QT interval. During the last part of this interval, the heart is vulnerable and electrically unstable. People with LQTS have an abnormally long QT interval. If the next electrical signal arrives before the muscle cells have completed their recovery period a dangerously fast heart rate can occur leading to a fall in blood pressure and loss of consciousness.

![ECG - QT Interval](image)

Schematic diagram of the electrical activity of one heart beat recorded by an ECG. The electrical discharge (depolarization) of the ventricle is represented by the QRS complex, with the T wave reflecting electrical recovery (repolarization). The QT is the time interval from the beginning of the QRS complex to the end of the T wave, and is generally less than 0.45 seconds in normal individuals. In subjects with the long-QT syndrome, the QT is usually longer than 0.45 seconds. There is considerable overlap between those with and those without long QT syndrome.
People with LQTS are sometimes identified after an unexplained fainting episode. These episodes are usually associated with surges of adrenaline, such as with sudden loud noises, intense emotional reactions, awakening from sleep or during intense physical activity, especially swimming. Currently, the primary treatment for LQTS is a beta blocker medication, which inhibits the effects of adrenaline on the heart. Some patients (not the majority) may benefit from pacemakers or implantable defibrillators or surgical resection of a group of nerves connecting the brain and the heart. Everyone with LQTS should avoid medications which are known to prolong the QT interval. A list will be provided with this information sheet.

Research
Scientific breakthroughs – including the recent discoveries of six groups of genes that cause LQTS – may soon lead to better therapies and ultimately a cure. It seems likely that different forms of LQTS, caused by the different genes, may benefit from “gene-specific” therapies yet to be developed. Much of the current research into LQTS aims to improve the reliability of the tests we use to make the diagnosis – some people who carry the abnormal genes can have a normal QT interval on the routine ECG. Exercise tests and 24 hour ECG monitoring show considerable promise and are already useful clinically. It is likely your physician may request either or both of these during evaluation of your family. Other research is at a molecular level – identifying the gene defects and the effects these have on the heart muscle cells. Only about 60% of affected families will have a gene defect which has thus far been identified.

Screening family members
Since the gene runs in families, screening of family members is very important, yet this is complex and time consuming. Not only are the tests difficult to interpret, but the family members may be spread across New Zealand and the rest of the world. Centralised registries – such as the International LQTS Registry based in New York, a national registry such as we are establishing in New Zealand – are vital. These help achieve successful family screening programmes, and allow dissemination of new information to affected family members and their clinicians. They also permit useful research studies into this common yet widespread disease.

Other sources of information
There are a number of physicians in New Zealand who have a special interest in the management of families with long QT syndrome and some of these are listed below. There are many web sites to visit, of varying quality. A good start is www.SADS.org and www.cidg.org.

National Long QT Registry in New Zealand:
A registry is based in Auckland co-ordinated by Jackie Crawford (Senior Cardiac Technician) and Dr Jon Skinner as part of the inherited cardiac disease service. A molecular diagnostic service is available at (www.cidg.org) labplus Auckland and a research programme is based at Auckland University (Associate Professor Andrew Shelling).
**Genetic diagnosis:**
Until very recently genetic diagnosis has only been available by sending blood overseas. Now a significant proportion of families will be able to receive a genetic diagnosis within New Zealand. Laboratories testing at present exist in Wellington and Auckland. Your supervising physician, after discussion with yourself, will send your blood for genetic testing to the place that seems most appropriate for your case at that time.

The importance of the genetic test is two-fold. A genetic diagnosis can be found in about 60%.

1. It defines the sub-type of Long QT syndrome, so treatment and advice can be adjusted appropriately.
2. It enables screen of family members to see who may be at risk, and if they could pass it on to their children.

There are three common types:
Type 1 and 2 respond well to betablocker treatment, because the triggers for type 1 (60%) is exercise; swimming is especially dangerous. For type 2 (30%) the commonest triggers are sudden and loud noise (especially at night), and emotional excitement. In type 3 (10%) events usually occur at night, and defibrillator pacemakers are most commonly needed in this group.