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N.Z. Cardiac Inherited Disease Registry
LONG QT SYNDROME

INFORMATION FOR FAMILIES

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 causing frequent faints or collapse resulting in 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 heartbeat 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 heartbeat 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.

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. There are several different types of Long QT syndrome, and many yet to be discovered. Some of these have different triggers for instance Long QT type 1 may cause symptoms during swimming, or exercise whereas Long QT type 3 can cause symptoms during rest or sleep. Since the discovery of Long QT types 1-3, there have been some other genes discovered that also cause the QT interval to prolong, although these genes appear to be rare in the New Zealand population. 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 (the procedure is called a left cardiac sympathectomy). Everyone with LQTS should avoid medications that are known to prolong the QT interval. A constantly updated list can be obtained from the website www.qtdrugs.org or contact the co-ordinator (details above) with your details and copies can be posted or emailed to you.

Screening family members

Long QT syndrome is carried in our genes which form proteins or blueprints for life that we are born with. Since these genes may be inherited, 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 word. Centralised registries – such as the International LQTS Registry based in Rochester, New York and the registry we have established in New Zealand - are vital. Registries help achieve successful family screening programmes, and allow dissemination of new information to affected family members and their clinicians, and allow collection of population incidence data and trends. They also permit useful research studies into this uncommon yet widespread condition.

Research

Scientific breakthroughs – including the discovery of 13 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 70% of affected families will have a gene defect that has thus far been identified.

Genetic diagnosis

Until recently, genetic diagnosis has only been available by sending blood overseas. These genetic analyses are time consuming and very expensive. Now a significant proportion of families will be able to receive a genetic diagnosis within New Zealand. Laboratories testing for LQTS exist in Auckland and Wellington. Your supervising physician, after discussion with yourself, will send your blood for genetic testing to the facility that seems most appropriate for your case at that time.

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. At www.cidg.org we update information regularly about many aspects of inherited heart diseases, an Australian family support site can be found atwww.sads.org.au. Other good sites include www.SADS.org; www.qtdrugs.org; and www.cry.org.uk

If you have any other questions you would like to ask, please do not hesitate to contact the registry co-ordinator and/or discuss your queries with your Specialist.

If you have any queries or concerns regarding your rights as a participant in the Cardiac Inherited disease registry you can contact an independent Health and Disability Advocate. This is a free service provided under the Health & Disability Act; through the offices of the Health and Disability Commissioner.

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The N.Z. Cardiac Inherited Disease Registry has received Ethical Approval from the N.Z. Multi-centre Ethics Committee: AKX/02/00/107