Treatment

Beta-blockers are often very effective in preventing ventricular arrhythmias for LQT1 and LQT2 sufferers.

Patients with LQT3 may respond to sodium channel-blocker drugs or the implantation of a device (pacemaker or ICD). Patients with LQT3 are most susceptible to a slow heart rhythm, a pacemaker is thus often needed to stop the onset of *torsade de pointes*.

Implantable cardioverter defibrillators (ICDs) are increasingly used for patients that have been given beta-blockers or pacemakers but have recurrent symptoms in which case an ICD with combined beta-blocker therapy may be recommended.

Competitive sport should be discouraged but recreational sports that do not cause an increased heart rate may be acceptable.

Patients should be instructed how to respond to excessive electrolyte loss, for example, during diarrhoea or excessive perspiring in which case the advice is to take drinks that contain minerals such as potassium and present to the emergency medical services.

Implantable Cardioverter Defibrillator (ICD) & Pacemaker

The implanted defibrillator is a small automatic device containing a microprocessor and battery that are connected to a very thin lead that has one end positioned within the heart. The ICD constantly monitors the heart and can detect tachycardia (fast heart rhythms); some ICDs may also detect bradycardia (slow heart rhythms). The ICD may also treat ventricular tachycardia (an unstable irregular heart beat). When an abnormal rhythm is detected the ICD delivers small electrical signals via the thin lead to the heart muscle to restore the normal heart rhythm. The ICD will greatly reduce the chance of cardiac arrest but if this corrective treatment is not successful and cardiac arrest follows, the ICD will deliver a strong signal (defibrillation shocks) to restart the heart beat.

Normally the signals from the ICD are not noticeable, although the patient may notice some of the symptoms from the onset of the abnormal heartbeat such as dizziness. If the ICD needs to restart the heart then the patient will feel what has been described as a "kick in the chest".

The ICD provides treatment for the heart rhythm disorder (arrhythmia), it does not provide a cure for cardiac arrhythmias.

Modern ICDs will store information about the treatment it has given, The Physician can interrogate this information with the ICD in position and undisturbed.

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A pacemaker is very similar to the ICD but it only treats bradycardia, which is a slow heartbeat. The pacemaker monitors the heart and delivers an electrical signal via the thin wire to speed up the heart rate. The increased circulation increases the oxygen supply to the body generally providing the patient with more energy and improved shortness of breath.

Influence of Age and Sex

The LQT interval is shorter in men than women, and QT prolongation can be diagnosed more readily in women than in men. Clinical studies show a predominance of LQT in females, but in men the risk of their first cardiac event is in childhood, diminishing after the age of 15 years. In females the possibility of the first cardiac event continues throughout life. Women are at risk of experiencing their first cardiac event immediately after childbirth, probably due to altered levels of female hormones.

Brugada Syndrome

Brugada syndrome has similar symptoms to LQTS, the patient suffers syncopal episodes due to ventricular fibrillation or ventricular tachycardia, there is also the possibility of sudden death. The heart structure is normal but there is a distinctive ECG trace resembling right bundle branch block and ST segment elevation but there is a variability of the ECG pattern associated with Brugada that can make the diagnosis difficult.

Beta-blockers and other antiarrhythmic drugs have little effect in the treatment of Brugada thus an implantable cardioverter defibrillator is often necessary.

This disease has been associated with sodium channel genes that are mutated and is thus hereditary.

Wolff Parkinson White Syndrome (WPW)

The symptoms of WPW are episodes of rapid heartbeat called paroxysmal supraventricular tachycardia (PSVT), the heart rate rises to between 180 and 240bpm. Tachycardias may be present from birth or childhood but are not normally present until later in life. Fainting and palpitations typical of LQT and Brugada syndromes may be experienced. Sudden death is a possibility but very rare in this condition.

PSVT is caused by the electrical signal that initiates the heartbeat, arriving at the ventricles too soon. The signal travels via a bypass (an extra piece of tissue) between the atria and the ventricles. This is a congenital condition and the bypass is thus present from birth.

Definitive treatment in those with symptoms is with radiofrequency ablation that is curative. Drug treatment usually with flecainide suppresses symptoms until ablation is completed.

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SADS UK

The Long QT Syndrome

Different genetic types explained

Chelsea is living with the Long QT Syndrome and being appropriately treated

IF YOU SUFFER FROM FAINTING SPELLS WHEN TAKING EXERCISE OR WHEN STRESSED, PLEASE CONSULT YOUR DOCTOR

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The Long QT Syndrome (LQTS)

The heart is made up of millions of cells that form the muscle of the heart. Each of these cells have pores in the cell membrane termed ion channels, these allow the movement of chemicals into and out of the cells providing an electrical signal that stimulates the cells causing the heart to beat and thereby pump blood. The heart normally beats at between 60 and 100 times per minute. The movement of sodium, potassium and calcium through the pores of the cells, produces the electrical signals. Long QT Syndrome is caused by a disorder of the pores that affects the production of the electrical signal. This delays the recovery of the signal before the next beat can start and this extends the QT interval shown on a typical ECG trace. This extended QT interval can be associated with the onset of rapid chaotic heart rhythms that cause improper pumping of the blood with resultant abrupt fainting episodes (syncope) without forewarning and can also cause sudden unexpected death. These syncopal episodes may occur at any age but are most likely to occur between the ages of 10 to 25 years.

A history of unexplained syncope in a child or young adult, especially during physical exertion or emotional stress, or a history of unexplained drowning/near drowning or sudden death within a family, should provoke a consideration of the LQT syndrome.

The exact numbers of individuals affected is unknown although it is estimated there are around 1 in 5,000 people who have LQT. As conditions of the electrics of the heart cannot be detected by standard post mortem a death may be recorded as unascertained; it is therefore difficult to know how many npeople are affected.

Inherited Long QT Syndrome

Most patients do not experience symptoms of the disorder but when symptoms do occur these include fainting, irregular heart rhythm or sudden death. Sudden death may occur without previous symptoms.

The symptoms of LQT differ depending upon the gene inherited, three most common LQT genotypes are the following:-

Genotype LQT1 – Gene mutated KCNQ1 (=KvLQT1); cardiac events are triggered by exercise and stress. Diving and swimming are a typical trigger of LQT1.

Genotype LQT2 – Gene mutated KCNH2 (=HERG); cardiac events are triggered by both exercise and rest. Events provoked by noise such as an alarm clock are almost exclusive to LQT2.

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Genotype LQT3 – Gene mutated SCN5A; cardiac events are triggered by rest and sleep, this is because the LQT interval is excessively prolonged at slow heart rates when the body is at rest. Patients with LQT3 are at high risk of slow heart rates.

LQT4, LQT5 and LQT6 are much less common. Most LQT syndrome genes are responsible for carrying the information responsible for the assembly of the potassium channels, the exception is LQT3 which is responsible for the sodium channels. New LQT genes continue to be discovered.

LQT1 and LQT2 are the most common variants with syncope, and seizure-like activity being the usual clinical signs. Approximately eighty five percent of events are related to physical activity or emotional stress. Female LQT2 patients have a high incidence of syncope occurring during menstruation and in the postpartum period after childbirth.

The number of non fatal cardiac events that occur before the age of 40 are significantly higher in patients with LQT1 and LQT2 than in patients with LQT3. The likelihood of sudden death during a cardiac event is much higher in patients with LQT3.

The classification "LQT Syndrome" therefore covers several different genetic diseases caused by mutations in cardiac ion channels. These all have the common effect of prolonging ventricular repolarization and thereby limiting the ability to initiate the next electrical signal to start the following heart beat.

Romano-Ward & Jervell and Lange-Nielsen Syndromes

Terminology has changed but the two major clinical LQT syndromes characterised on the basis of the genetic transmission of genes still carry the names they initially acquired and are:-

The Romano-Ward syndrome (autosomal dominant disorder without deafness) describes all forms of LQTS without deafness.

The Jervell and Lange-Nielsen syndrome (autosomal recessive disease with congenital deafness) is characterised by the presence of LQTS and deafness. This is much less common and affecting mainly young children.

In patients with LQT syndrome, episodes of sudden loss of consciousness are almost always due *to torsade de pointes* arrhythmias. An important characteristic of *torsade de pointes* is its potential to self-terminate or to deteriorate into ventricular fibrillation. This explains why affected patients often survive several syncopal attacks before potentially succumbing to a fatal one. In patients who experience syncope only, the *torsade de pointes* rhythm has spontaneously returned to normal, usually within one minute, and the patient then regains consciousness without much in the way of disorientation or confusion. On the other hand, in a minority of patients the rhythm persists and then degenerates into the heart rhythm know as ventricular fibrillation. The second rhythm abnormality rarely reverts back to normal rhythm without medical intervention and may cause sudden death.

Methods of detection – ECG tests

The standard ECG test may not identify members of families with congenital LQT syndrome, which may be a difficult diagnosis even for the most experienced physician. An exercise ECG may enhance the accuracy of diagnosis. During medium levels of exercise patients with LQT1 may have a reduced heart rate, LQT2 patients may have a normal heart rate.

During recovery from exercise, patients with LQT1 show a more exaggerated QT interval prolongation compared to LQT2 patients. Patients with LQT3 display a high degree of QT interval shortening in response to increased heart rate. Abnormal responses with altered repolarization in LQTS at different times in different regions of the heart may be manifest in beat-to-beat alterations of the T wave polarity or amplitude. That may be observed briefly at rest but most commonly appears during emotional or physical stress and may herald *torsade de pointes* and is a marker of high risk patients. Sinus node dysfunction such as sinus bradycardia or sinus pauses cause a lower than expected heart rate during exercise. Slow heart rates (bradycardia) are particularly striking in younger children. Sinus pauses may initiate *torsade de pointes* arrhythmias.

Family History

LQTS is usually inherited by autosomal dominant transmission. This means that it affects boys and girls equally and that each child has a 50% chance of inheriting the gene. Once a family member is identified as having LQTS, it is extremely important that all family members are examined for evidence of the syndrome. It is especially important to know which parent and grandparent has evidence of the disease, since brothers, sisters, aunts, uncles, nephews, nieces and cousins, on the affected side of the family are potentially at risk. Constructing your family tree and identifying the causes of death within the family, looking for a history of sudden death can be very helpful. Causes of death such as epilepsy, pneumonia and asthma should be considered as these may have been used to classify death in the past before a better knowledge of the LQTS emerged. Cases of drowning and other unexplained accidents should also be considered as being potentially due to LQTS.

Please support the work of the charity by sending a donation to:
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