Published on *My46* (https://www.my46.org) 01/20/2022

My46 Trait Profile

Long QT syndrome

Other Names: Romano-Ward syndrome

Long QT syndrome is a genetic condition characterized by a high risk of ventricular arrhythmias (i.e., an abnormal heart beat) and sudden death. It can be caused by mutations in any one of many different genes.

Characteristics of Long QT syndrome

Long QT syndrome (LQTS) is an inherited arrhythmia (irregular heart rhythm) condition. People with LQTS have structurally normal hearts, but the irregular heart rhythm can cause episodes of fainting (syncope) often during strenuous exercise (such as running a marathon or swimming) or when experiencing a strong emotion like fear or anger, and sudden cardiac death. People with LQTS can also have episodes of ventricular tachycardia (a type of fast heart beat), including torsade de pointes (TdP). Most people with LQTS start experiencing symptoms (syncope or ventricular tachycardia) in the pre-teen years or 20s; however, some people with LQTS never have any symptoms.

Long QT syndrome can be inherited (passed from parent to child) or acquired (meaning that the features of LQTS appear after taking a certain medication or another illness). Acquired LQTS can be caused by taking medications that lengthen the QT interval or cause TdP; low levels of potassium, calcium, or magnesium in the blood; hypothermia (low body temperature); or myocardial ischemia (damage to heart muscle cells due to a lack of oxygen).

Diagnosis/Testing

LQTS is most frequently diagnosed when specific patterns, including a prolonged QT interval and/or T-wave abnormalities, are seen on an electrocardiogram (also called an ECG or EKG -- a test that checks for problems with the electrical activity of the heart). There are at least 12 different genes that are known to cause LQTS. Mutations in any one of these genes are thought to cause LQTS by disrupting channels within the hearts muscle cells that are responsible for maintaining the hearts rate and rhythm. Approximately 25% of people with LQTS will not have a mutation in any of the known LQTS genes. This likely means that there are other genes involved in causing LQTS.

Management/Surveillance

It is recommended that individuals with LQTS are followed by a cardiologist who specializes in irregular heart rhythms also known as an electrophysiologist. Management of LQTS includes medications called beta-blockers. Some individuals with LQTS will need to have an implantable cardioverter defibrillator (ICD). An ICD is a small device placed under the skin that detects arrhythmias and keeps track of the heart rhythm. It can correct an abnormal beat in the heart.

People with LQTS should avoid drugs or medications that prolong the QT interval or cause torsade de pointes, as well as competitive sports or other activities that are strenuous or cause emotional stress (such as roller coasters, scary movies, and alarm clocks).

Mode of inheritance

LQTS is inherited in an autosomal dominant manner. This means inheriting one mutation is enough for an individual to be affected and show signs of LQTS. The mutation can be inherited from an affected parent or it can rarely

occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not a person with LQTS has a parent with LQTS. If a parent also has LQTS, the risk of having a child with LQTS is 50% with each pregnancy. If a parent does not have LQTS, the risk that other siblings will also have LQTS is very low.

Special considerations

Syndromic forms of LQTS have also been described including Anderson-Tawil syndrome that also causes periodic paralysis and characteristic facial features, and Timothy syndrome that also causes birth defects such as heart defects and webbing of the fingers or toes.

Resources

CredibleMeds
http://www.qtdrugs.org
Sudden Arrhythmia Death Syndromes (SADS) Foundation
http://www.sads.org
Genetics Home Reference: Long QT syndrome
http://ghr.nlm.nih.gov/conditionGroup/long-qt-syndrome
National Heart, Lung, and Blood Institute: Long QT Syndrome
http://www.nhlbi.nih.gov/health/health-topics/topics/qt/

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Created: 08/2013 **Created by:** Courtney Eddy, MS, CGC, LGC, MT(ASCP), Heather MacLeod, MS, CGC
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC