



LONG QT SYNDROME

More about Long QT Syndrome (LQTS)

- Long QT predominately affects children and young adults.
- Patients are usually identified during evaluation of an unexplained fainting episode, or as part of a family evaluation when one family member has been affected and diagnosed.
- Individuals with LQTS may be completely free of symptoms, or have recurrent syncope (fainting).

Recent Genetic Discoveries:

In a landmark series of scientific articles in 1995, Dr. Mark Keating and colleagues at the Howard Hughes Medical Institute at the University of Utah reported the discovery of two genes which cause Long QT Syndrome. Subsequently, four more affected genes were identified. These genes are responsible for maintaining electrical balance in the heart and specifically affect the electrical recovery of cells. You only need to inherit one of these gene defects to have Long QT Syndrome. There are still some families with LQTS who are not linked to any of the six affected genes, which means there are more genes yet to be identified. The most common mutations for the Long QT syndrome are found on the following genes and their associated triggering events.

- LQT1: (KVLQT1 on chromosome 11) Exercise, especially swimming
- LQT2: (HERG gene on chromosome 7) Emotions or emotional stress; startle event
- LQT3: (SCN5A on chromosome 3) During sleep or at rest with or without arousal

These scientific breakthroughs have led to a simple commercially available blood test for the diagnosis of LQTS. Until a gene-based cure is developed, there are effective therapies available to stabilize the electrical activity in the heart of patients with LQTS including beta blocker medications and implantable pacemakers and defibrillators. As with any medical condition, please discuss your treatment options directly with your primary healthcare provider and/or cardiologist.

Safety Tips for LQTS Patients

- Use the same pharmacy and advise the pharmacist that you have Long QT Syndrome.
- Establish a contact with a primary care physician and a cardiologist.
- Do not participate in competitive sports which may elevate emotions and physical exertion.
- If surgery is required, be sure to inform both the surgeon and anesthesiologist that you (or your child) have LQTS.
- Try to avoid environments with acute loud noises that could produce a "startle response", such as alarm clocks or shrill telephone rings.
- Do not participate in rapid weight-reduction programs, and do not use weight-reduction medications.

The International Long QT Registry

The Long QT Registry is based at the University of Rochester under the direction of Arthur J. Moss, M.D. This world-wide registry was started in 1979 by Dr. Moss as a research group who has provided follow-up of patients affected with this Long QT Syndrome. Since 1985, the registry has been supported by research funding from the National Institutes of Health to study the clinical, cardiac and genetic aspects of LQTS.

As of July 1, 2003, the LQTS Registry had enrolled over 1200 families with this disorder. The information gained from the Long QT Registry has advanced our understanding of this disorder and permitted more accurate diagnosis and treatment of those with LQTS.

If you have LQTS and are not already enrolled in the LQTS Registry, please contact the C.A.R.E. office which will refer patients for genetic screening for LQTS.

For the latest information on the Long QT Syndrome, call the C.A.R.E. Foundation at (800) 404-9500, e-mail care@longqt.org, or visit the website at www.longqt.org