

# Patient Information

## Long-QT Syndrome

### 1. The Normal Heart

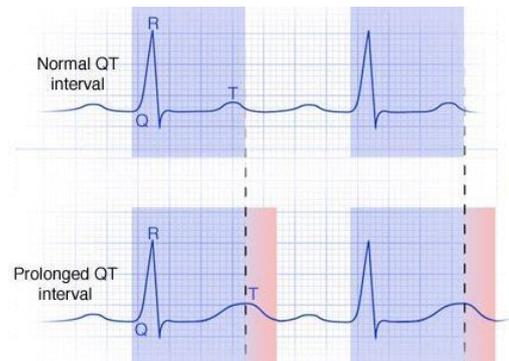
The heart is a special muscle that contracts regularly and continuously, pumping blood to the body and the lungs. It has four chambers – two at the top (the atria) and two at the bottom (the ventricles). The pumping action of the heart is caused by a flow of electrical signals through the heart. These electrical signals repeat themselves in a cycle and each cycle causes one heartbeat. When the electrical activity of the heart is disturbed, known as an arrhythmia, it can affect your heart's ability to pump properly.

### 2. Long-QT Syndrome

Long-QT syndrome (LQTS) is a disease that affects the electrical activity of the heart. The QT interval is the measurement of one part of the heartbeat on an ECG. During each heartbeat, an electrical signal through the heart causes the heart muscle to contract and to pump blood. Once the heart muscle has contracted, it must have time to recover and relax before the next electrical signal is received. The length of time this relaxation takes is called the QT interval. In people with LQTS, the QT-interval is longer than normal (as the name suggests). If the next signal arrives too early (i.e. when the muscle has not fully recovered from the last contraction) it can cause the heart to beat abnormally fast, leading to dizziness, black-outs or even death.

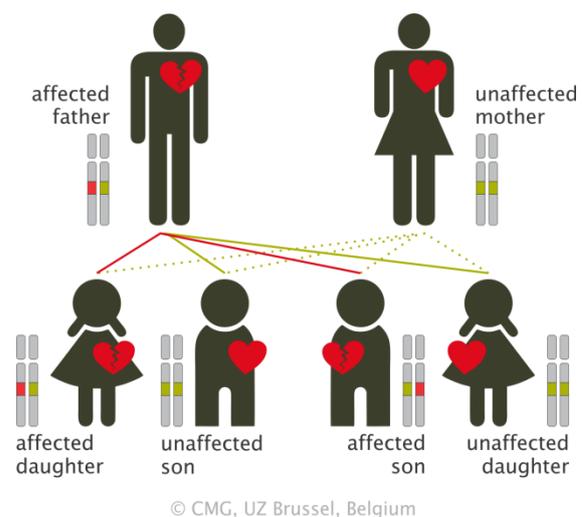
### 3. Prevalence & Inheritance

About 1 in every 2000 persons has LQTS (the prevalence of the disease). LQTS is a genetic disease. This means that LQTS is caused by a defect (a mutation) in a gene that can be passed on through families. A gene is part of our DNA which contains a code for making a molecule (a protein).



Source: with permission from Mayo Clinics

Every person has two copies of each gene that can be linked to LQTS. LQTS is caused by a mutation in the genes that contain codes for molecules (proteins) in the heart. A mutation in only one of the two copies of one of these genes (from the father or from the mother) is enough to develop LQTS. This is called an autosomal dominant disease and a parent who carries it has a 50% (1 in 2) chance of passing the mutation to each child. The chance that a child will not inherit the mutated gene is also 50 percent.



*Autosomal dominant inheritance*

Sometimes, LQTS can also be an autosomal recessive disease. This means that you need

For more information: <https://guardheart.ern-net.eu>



mutations on both copies of a gene (from both father and mother) to develop LQTS. Whether LQTS is an autosomal dominant disease or an autosomal recessive disease depends on the types of involved gene and mutation. In some cases, a new (de novo) mutation can occur in the egg or sperm cells or in an embryo. In these cases, the child's parents do not have the mutation and LQTS, but the child does have LQTS and can pass the mutated gene to his or her own children.

## 4. Symptoms

Long QT syndrome most often affects children and young adults. The most common symptom is fainting or collapse. These symptoms often occur during activities that increase the heart rate and the adrenaline level in the body, such as exercise (particularly swimming), emotional situations and sudden loud noises. Diagnosing LQTS can be difficult, as many people often do not display symptoms. However, once LQTS is diagnosed, adequate therapies are available.

## 5. Diagnosis

The most common tools to make the diagnosis of LQTS are the medical and family history, physical examination, a heart electrical tracing (the electrocardiogram or ECG), Holter monitoring and exercise testing. Unfortunately, diagnosis can be very difficult because many people with the condition can have a normal ECG.

### 5.1. ECG (electrocardiogram)

This is the most basic test. Small sticky patches (electrodes) are put onto the chest and sometimes to arms and legs. These are connected by wires to an ECG recording machine, which picks up the electrical activity for a few seconds that makes the heartbeat. Sometimes additional or repeated ECG-tests are necessary.

### 5.2. Exercise test (stress test)

Exercise test is the same as the ECG described above, but is recorded before, during and exercising on a treadmill or an exercise bike. This records any changes in the electrical patterns that occur with exercise.

### 5.3. Holter monitoring

Holter monitoring involves a small digital machine, which can be worn on a belt round the waist. Four or six ECG electrodes from the machine are taped

to the chest. It then records the electrical activity of the heart for 24-48 hours, or for up to seven days. During the monitoring all activities are listed in a 'diary'.

### 5.4. Cardiomemo and cardiac event recorders

These are more complicated versions of the Holter monitoring test described above. During any symptoms, the device can be triggered to record the heart's rhythm. The advantage of the cardiomemo is that it doesn't have any electrodes, so it just can be placed on the chest while having symptoms.

### 5.5. Echocardiogram (echo)

Echocardiogram uses ultrasound waves to look at the structures of the heart. An echocardiogram can detect different types of structural changes in the heart, for example heart muscle diseases and heart valve abnormalities. Areas of thinning of the heart muscle can also be identified. Patients with LQTS don't have structural problems, but often an echo is performed once to confirm this.

### 5.6. Genetic testing

There are several types of LQTS. Each type is caused by mutations in a different gene. In about 70% (7 in 10) of the patients with LQTS, the cause of the disease can be detected in these genes. Most of the patients, in whom a mutation is identified, have the mutation in one of the following three genes: KCNQ1, KCNH2, or SCN5A. These three genes cause types 1, 2 or 3 of the LQTS.

## 6. Therapy

There is no cure for inherited LQTS, but treatment helps prevent symptoms and minimizes the risk for fainting or cardiac arrest. The treatment depends on symptoms, age, gender, and the specific gene-mutation. Often a medicine known as a beta blocker is prescribed to reduce arrhythmias. These drugs are believed to be successful in 80-90% of patients. Beta blockers do not shorten the QT interval but block the effects of adrenaline and other similar natural substances in the heart, and lead to a slower heart rate. In some patients, other medications on top of beta blockers may be indicated. In patients where medications do not work or in patients after a cardiac arrest, an internal cardiac defibrillator (ICD) or cervical sympathectomy may be considered. An ICD can correct most life-threatening arrhythmias. Cervical sympathectomy (also called cardiac denervation) is a surgical procedure to damage the nerves that



release adrenaline and similar natural substances in the heart.

## 7. Lifestyle & Sports

There are key recommendations for patients (and families) who are diagnosed with LQTS, to prevent them from arrhythmias:

- in general avoid competitive and strenuous sports
- sporting is permitted only after advise from an expert heart specialist.
- consequent use of beta blockers (if prescribed)
- avoid drugs that might prolong the QT interval and therefore worsen disease. This list of drugs to avoid can be found at <http://crediblemeds.org>
- encourage relatives to be screened

The diagnosis of LQTS and the ability to pass on the condition can lead to anxiety and many other questions. Medical social workers or psychologists have experience with this and may be helpful for the patient and the family members.

## 8. Follow-up

The heart doctor (cardiologist) will advise on how often follow-up is needed depending on symptoms, age, and treatment.

## 9. Family Screening

If a mutation in a gene is found in a patient with LQTS (see genetic testing), family members of this patient (to start with the first degree family members: mother, father, brothers, sisters, and children) can have genetic testing at a genetic heart clinic. Family members in whom the same mutation is found, are called mutation carriers and will be followed up by a cardiologist. Family members in whom the mutation is not found can be reassured. If there is not a mutation identified in a patient with LQTS, family members of this patient (to start with the first degree family members) are advised to see a cardiologist. LQT-patients can experience symptoms in childhood. Therefore genetic and heart testing, and timely treatment of the family members who are diagnosed with LQTS is important even in the first years of life.

## 10. LQTS and Pregnancy

During pregnancy it is important to continue the treatment with a beta blocker. Sometimes it's necessary to switch the type of beta blocker, because not all types are suitable for use during pregnancy. When beta blockers are used during pregnancy, it is advised to plan for delivery in hospital, because of a possible lower heart rate in the baby. In the first nine months after delivery extra follow-up is advised because of an increased risk of arrhythmias in the mother in this period (this is especially the case for patients with type 2 LQTS).



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