

Patient Information

This booklet contains information that will help you understand more about the *FAMILION* family of genetic tests. It will also provide information about some of the conditions that your doctor may be considering when ordering any of these tests.





What are the **FAMILION**® tests?

The *FAMILION* tests are genetic tests that help healthcare providers uncover genetic alterations (known as mutations) that may contribute to cardiac conditions. Many of these conditions fall under the technical term **cardiac channelopathies**.

What are cardiac channelopathies?

Cardiac channelopathies are conditions that affect the electrical system of the heart and sometimes other organs. The heart's electrical system must be closely regulated because it helps control the heartbeat and the mechanical functions of the heart. There are a number of syndromes associated with cardiac channelopathies. You are being asked to have one of the *FAMILION* tests to help your doctor determine if you have one of these conditions and how best to treat it. The *FAMILION* tests are used in assessing conditions such as:

- Long QT Syndrome
- Catecholaminergic Polymorphic Ventricular Tachycardia
- Brugada Syndrome

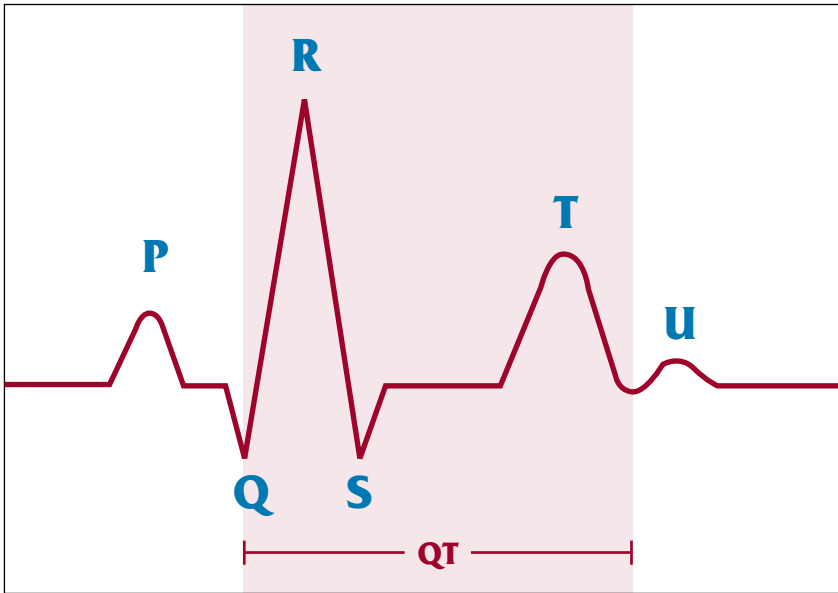
Information About Long QT Syndrome

Long QT Syndrome (LQTS) is a condition that impairs the electrical system of the heart. Symptoms of LQTS may include fainting; seizures; abnormal, very fast heartbeats; and even death. For some people, these symptoms only occur when they are stressed or taking certain medications. Most of the deaths can be prevented with early diagnosis and treatment of LQTS.

People with LQTS have hearts that look and sound normal but may have abnormal electrical activity when examined with an electrocardiogram (ECG). Some people with LQTS have a normal ECG at rest, but experience abnormal responses under certain conditions. For example, these people may have an unusual response when they are startled, experience emotional stress, or engage in vigorous physical activities.

IMPORTANT NOTE

These syndromes commonly run in families (inherited). You should discuss with your doctor whether it is appropriate to have other members of your family tested.



What is the QT interval?

Every heartbeat is started by an electrical signal that tells the heart muscle to contract. The electrical signal causes the waves that you see on an ECG; these waves are named with the letters P, Q, R, S, T, and U. The time from the beginning of the Q wave to the end of the T wave is called the QT interval. This QT interval represents the time needed by the muscle cells in the heart to “recharge” before the next beat. People with LQTS have an unusually long QT interval.

Why does it matter if the QT interval is too long?

When the QT interval is too long, your heart may not beat correctly. This can result in very fast, abnormal heartbeats, causing symptoms such as dizziness, fainting, or seizures. The severity and duration of these symptoms depend on how quickly your heart can return to a normal rhythm.

What causes LQTS?

There are pores in the heart called ion channels that help control the electrical activity of the heart. If these ion channels don't function properly, LQTS can result.

How do you get LQTS?

Familial LQTS:

This is an inherited (genetic) form of LQTS seen in about 1 of every 3,000 people. There may be a history in the family of seizures, fainting, or sudden death from unknown causes. However, at least one third of people with inherited LQTS never develop any symptoms. Some people only find out that they have inherited LQTS when they take a medication that causes them to faint or to have an unusual heart rhythm.

How LQTS is inherited:

Many genes are known to cause LQTS. These LQTS genes are passed from one generation to the next and tell the body how to form the ion channels in the heart. If you have an altered gene, one or both of your parents, your children, and other relatives may also have this same altered LQTS gene.

Children of a parent with familial LQTS have a 50% chance of inheriting the syndrome, even though the parent may have never had any of the symptoms of LQTS. At least one parent, the children, and the brothers and sisters of an affected individual may also have the altered LQTS gene and may benefit from knowing about the diagnosis. Even if relatives have not had any symptoms, they may wish to have their QT interval measured with an ECG and possibly have genetic testing performed.



How do I know if I have LQTS?

Your doctor may suspect you have LQTS based on a history of fainting or seizures by you or a family member. Your doctor will request an ECG to measure your QT interval. If this measurement is longer than normal, then you may have LQTS. If your ECG is normal but your doctor still suspects LQTS, you may be asked to take an exercise test. These tests may tell you that you have LQTS but cannot tell you the cause of the LQTS.

How do I determine the cause of my LQTS?

Your doctor may order the *FAMILION* LQTS test, which is a genetic test for the familial form of LQTS. The *FAMILION* LQTS test will analyze your ion channel genes to see if they have any mutations that might cause LQTS. Uncovering the type of mutations you have is important to help your doctor determine the cause of your symptoms and possible treatment options.

What results can I expect from the *FAMILION*® LQTS test?

The results of your test will not necessarily be “positive” or “negative.” The *FAMILION* LQTS test will provide your doctor with information about whether you have a mutation in any of the genes that is thought to contribute to LQTS. This information will assist your doctor in deciding how best to treat your condition and/or symptoms. The test results may also suggest whether your relatives are at risk of having LQTS.



What should I do if I have LQTS?

Several treatment options are available if you are diagnosed with LQTS.

Medications

Your doctor may prescribe medications that help your heart beat correctly. It is very important that you take any medication you have been prescribed for LQTS as directed. Never stop taking a medication without first consulting your doctor.

Medical Devices

In some cases, your doctor may recommend surgery to insert an implantable cardioverter defibrillator or similar device to help control the heart.

Lifestyle Modification

You may be advised not to participate in competitive sports because symptoms can be caused by physical exertion. You might be given a list of drugs/medications that you should avoid because they are thought to cause abnormal heart rhythms in people with LQTS.

IMPORTANT NOTE

You should consider sharing your tests results with family members. Many relatives such as children, brothers, sisters, and at least one parent may also be affected with LQTS even if they have not experienced symptoms. You should discuss this with your doctor or a genetic counselor.

Information About Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)

Like LQTS, CPVT is an inherited disorder that causes abnormal electrical activity in the heart. This abnormal activity often occurs during exercise, physical activity or in stressful or emotional situations.

Causes of CPVT

Like LQTS, CPVT is caused by abnormal ion channels in the heart. This can cause ventricular tachycardia, an extremely rapid heartbeat that originates in the ventricles of the heart. This ventricular tachycardia is felt to be triggered by the release, under stress, of certain hormones, called catecholamines. Ventricular tachycardia is a potentially life-threatening arrhythmia because it may lead to ventricular fibrillation and possibly sudden death. CPVT is hereditary, so it's important for both you and your family members to be tested.

Diagnosis of CPVT

Your doctor may suspect that you have CPVT based on your symptoms and specific abnormal findings on an exercise stress test. Unlike LQTS, a resting ECG for an individual with CPVT will almost always be normal. This may lead your physician to recommend that you undergo an exercise stress test. A specific abnormal arrhythmia on the exercise stress test will often confirm a CPVT diagnosis; however, in some cases, the exercise stress test will lack this specific rhythm. The *FAMILION* CPVT test can help your doctor confirm this diagnosis for you and your family members, including those who may not show any symptoms of the disease.

Treatment of CPVT

- Medication prescribed by your doctor may help control your heartbeat. It is very important that you take your medication as directed, and never stop taking a medication without talking to your doctor first.
- Sometimes, your doctor may want to recommend implantation of a device called an implantable cardioverter defibrillator.



What if I have more questions about LQTS or CPVT?

There are a number of organizations and groups that can assist you. Here is a list of resources:

Sudden Arrhythmia Death Syndromes Foundation

800.STOPSADS (800.786.7723)

www.sads.org

Cardiac Arrhythmias Research and Education Foundation

800.404.9500

www.longqt.org

National Society of Genetic Counselors

610.872.7608

www.nsgc.org

American Heart Association

800.242.8721

www.americanheart.org

Heart Rhythm Society

508.647.0100

www.HRSonline.org

PGxHealth

1.877.2.PGX.Health (877.274.9432)

www.pgxhealth.com

Information About Brugada Syndrome

Like LQTS, Brugada Syndrome causes abnormal electrical activity in the heart. The most common symptom is fainting. Brugada Syndrome is very similar to LQTS, but the ECG may look different. Not everyone with Brugada Syndrome will have symptoms. The prevalence of Brugada Syndrome is not known and, although it is more common in men, it is also seen in women.

What causes Brugada Syndrome?

As described elsewhere in this booklet for LQTS, abnormal ion channels also cause Brugada Syndrome. The abnormal function may result in a very fast heartbeat called ventricular fibrillation. This is a very disorganized beating of the heart that compromises blood flow to the body. If not corrected, ventricular fibrillation can result in death. Since Brugada Syndrome can be inherited, it is important for you and your family members to be tested.

How do I know if I have Brugada Syndrome?

Your doctor may suspect you have Brugada Syndrome based on your symptoms and specific abnormal findings in your ECG. The doctor may recommend that you undergo a “provocative test.” During this test, you would be given a drug to produce the typical ECG findings. The *FAMILION* Brugada Syndrome test can also assist your doctor in confirming this diagnosis in you and your family members.

Is there any treatment for Brugada Syndrome?

There are no specific medications for Brugada Syndrome. Your doctor may advise surgical insertion of a special implantable cardioverter defibrillator, which will reset the heart if ventricular fibrillation occurs. Your doctor may also prescribe medication.



What if I have more questions about Brugada Syndrome?

There are a number of organizations and groups that can assist you. Here is a list of resources for you:

Ramon Brugada Senior Foundation

www.brugada.org

American Heart Association

800.242.8721

www.americanheart.org

National Society of Genetic Counselors

610.872.7608

www.nsgc.org

PGxHealth

1.877.2.PGX.Health (877.274.9432)

www.pgxhealth.com

Frequently Asked Questions

Q. How are the *FAMILION* tests conducted?

A. You will be asked to provide a blood sample. This sample will be collected by your doctor or at a laboratory of his/her choosing. The blood sample will be sent to the laboratories of PGxHealth for analysis. In about 6 weeks, your doctor will receive test results. Your doctor will then use these results to determine the best course of action for you and your family.

Q. Are there different *FAMILION* testing options?

A. Yes. The *FAMILION* family of genetic tests includes four different testing options that your physician may order. These are:

- LQTS Test (Long QT Syndrome)
- CPVT Test (Catecholaminergic Polymorphic Ventricular Tachycardia)
- BrS Test (Brugada Syndrome)
- Family Specific Test

Q. What is the Family Specific test?

A. The gene mutations associated with cardiac channelopathies can be passed from one generation to the next. When a patient is confirmed by means of genetic testing to have a gene mutation associated with a cardiac channelopathy, it is important that family members are tested for the same mutation. Even if family members do not exhibit any symptoms, they can still be carriers of the gene mutation found in their family member and should still be tested.

Q. Will anyone else know the results of my tests?

A. Your tests' results are strictly confidential. PGxHealth only communicates test results to physicians you have authorized. Even if an insurance company has paid for the tests, the results remain strictly confidential.

Q. How many times will I need to have this test?

A. Since your genetic makeup never changes, the *FAMILION* test option ordered by your physician should only need to be performed once. Instances may arise where your physician may opt to order more than one of the *FAMILION* testing options.

Q. Will my health insurance pay for the *FAMILION* test ordered by my doctor?

A. PGxHealth provides a service to help you determine if your insurer will cover the cost. Call PGxHealth at 1.877.2.PGX.Health (877.274.9432) for assistance between 9:00 am and 5:00 pm Eastern time.

Q. Will the results of my test affect my ability to get health insurance?

A. Your *FAMILION* test results will only be released to your doctor. PGxHealth will not send these results to your insurance carrier.

Q. What if I have more questions about any of the *FAMILION* tests?

A. You may contact PGxHealth at 1.877.2.PGX.Health (877.274.9432) between 9:00 am and 5:00 pm Eastern time. After hours, please leave a message, and a customer service representative will return your call as soon as possible.

You may also e-mail the customer service department at familioninfo@pgxhealth.com. We will respond to your e-mail in a timely manner.

All inquiries are confidential.



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