



Cardiomyopathy 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.



HCM and ARVC Testing



What are the **FAMILION**® tests?

The *FAMILION* family of genetic tests help healthcare professionals uncover genetic alterations (known as mutations) that may contribute to cardiac syndromes. These syndromes fall under the following technical terms:

- Cardiac Channelopathies (see booklet titled *Cardiac Channelopathy Patient Information*)
- Cardiomyopathies

What are cardiomyopathies?

Cardiomyopathies are a group of inherited diseases that cause the heart muscle to become abnormally enlarged, thickened and/or stiffened. There are five main types of cardiomyopathies:

- Hypertrophic Cardiomyopathy (HCM)
- Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)
- Dilated Cardiomyopathy (DCM)
- Left Ventricular Non-compaction (LVNC)
- Restrictive Cardiomyopathy (RCM)

All five cardiomyopathies may make it difficult for your heart muscle to function properly.

You are being asked to have one of the *FAMILION* tests for cardiomyopathies. Information gained from this test may help your physician determine treatment options for you or if your family members need to be tested. Even those first-degree (blood relatives) family members who are not experiencing the symptoms of a cardiomyopathy may need to be tested.

Information About Hypertrophic Cardiomyopathy (HCM)

HCM is the most common form of heart muscle disease, affecting one in every 500 people. It is the most common cause of sudden cardiac death in people under 30 years of age.

HCM is a disease in which the heart muscle (called the myocardium) becomes abnormally thick or “hypertrophied,” most often affecting the left ventricle. The left ventricle is responsible for supplying oxygen-rich blood to the rest of the body. HCM may make it difficult for the heart to pump enough blood throughout the body, especially during physical exertion. It may also affect the heart’s electrical system, leading to problems with heartbeat regulation and mechanical function. Patients may ultimately develop abnormal heart rhythms (arrhythmias) that put them at increased risk for sudden cardiac death.

Some people with HCM lead normal, healthy lives. Unfortunately, for others, it can cause a wide variety of symptoms from shortness of breath to severe problems like heart failure.



Normal



Hypertrophic Cardiomyopathy



In HCM, the walls of the left ventricle may thicken.

What causes HCM?

HCM is a genetic disease that is passed on from one generation to the next. It is most often triggered by genetic mutations that cause the heart muscle to grow abnormally thick. Most HCM is “obstructive,” commonly referred to as hypertrophic obstructive cardiomyopathy. When HCM becomes obstructive, blood flow from the heart is restricted, which can cause significant complications.

How is HCM diagnosed?

Making a diagnosis of HCM can be challenging because there are many diseases that may cause thickening of the heart muscle. Your doctor may suspect that you have HCM based upon hearing a heart murmur, the symptoms you’re experiencing and/or specific abnormal findings on diagnostic tests such as an echocardiogram or electrocardiogram. Typical symptoms of HCM include shortness of breath, chest pain and/or fainting, especially during exercise or physical exertion.

Currently, the most commonly used test to diagnose HCM is called an echocardiogram. The echocardiogram uses sound waves to produce images of your heart that may help your doctor to see if there is any abnormal thickness and if blood flow is being obstructed. It is very important to identify the disease as early as possible to help guide your physician toward the most appropriate treatment options as well as possibly prevent complications.



What results can I expect from the **FAMILION® HCM Test**?

The results of your test will not necessarily be “positive” or “negative.” The *FAMILION* HCM Test will provide your doctor with information about whether you have a mutation in any of the genes that is thought to contribute to HCM. 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 HCM.

What are the treatment options for HCM?

Currently, there are no therapies that can prevent or stop the progression of HCM. Treatments are available that can improve symptoms and help better manage HCM. Some treatment options include:

- medications that may help relax the heart muscle and slow your heart rate so that your heart can pump more effectively
- implantation of a device called an implantable cardioverter defibrillator (ICD) or a pacemaker
- surgical options to reduce the size of the obstruction.

The **FAMILION® HCM Test**

The *FAMILION* HCM Test may help your doctor accurately distinguish HCM from other diseases that cause thickening of the heart muscle. Because HCM is an inherited cardiac syndrome, there are distinct genetic signs that can help confirm a diagnosis of HCM. Once the first person in your family is identified by genetic testing as having an HCM gene mutation, other family members should be tested. Family testing is important because others may be at risk regardless of whether or not they are experiencing any symptoms.

Information About Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)

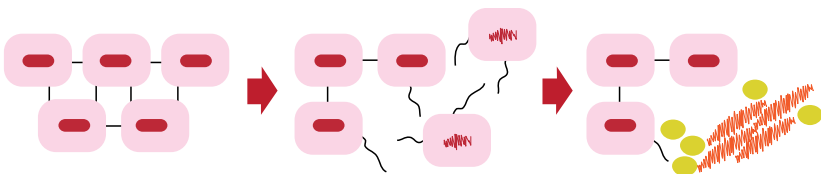
ARVC is a potentially lethal and progressive disease of the heart muscle. The exact prevalence is unknown, but estimates range from 1 in 5,000 to 1 in 1,250 people. Approximately 20% of sudden cardiac deaths in people under 35 years of age are attributed to ARVC.

ARVC is a heart muscle disease (cardiomyopathy) in which the heart muscle is gradually replaced with fat and scar tissue. The fat and scar tissue interferes with heart's electrical system and leads to abnormal heart rhythms (arrhythmias). The word 'arrhythmogenic' simply means to cause abnormal heart rhythms. As suggested by the name, ARVC was once believed to be a disease exclusively of the right ventricle, one of the heart's two lower chambers. However, we now know that either one or both ventricles can be affected. ARVC may also cause the heart to become enlarged and contract poorly, reducing the ability of the heart to pump blood to the rest of the body. Common symptoms associated with ARVC include: dizziness, fainting and/or heart palpitations.

An early and accurate diagnosis of ARVC is important. The prognosis for patients diagnosed early and started on appropriate therapy is very good.

What Causes ARVC?

ARVC is a genetic disease that is passed from one generation to the next. It is most often caused by mutations in genes important for producing proteins that help hold heart muscle cells together. Although the disease process is not fully understood, defects in these proteins are believed to gradually allow muscle cells to detach from one another and die. The heart is unable to replace these cells and instead, scar and fat tissue replaces the missing muscle cells. ARVC can worsen over time or in response to stress placed on the heart muscle.



1. Normal heart cells allow electrical signal to go cell-to-cell.

2. Heart cells disconnect and electrical signal is disrupted.

3. The body replaces the no longer functioning heart cells with scar tissue and fat.

How is ARVC Diagnosed?

Making a diagnosis of ARVC can be challenging, and ordinarily, several diagnostic tests are needed. Your doctor may suspect that you have ARVC based upon the symptoms you're experiencing and/or specific abnormal findings on diagnostic tests such as an electrocardiogram (ECG), stress testing, Holter monitor, echocardiogram or cardiac magnetic resonance imaging (CMRI).

The progression of ARVC may be associated with 'hot phases'. These are times when a patient's disease is progressing and they are commonly experiencing symptoms associated with active disease. 'Hot phases' are unpredictable and may put the patient at increased risk of potentially lethal arrhythmias.

What results can I expect from the *FAMILION*® ARVC Test?

The results of your test will not necessarily be "positive" or "negative." The *FAMILION* ARVC Test will provide your doctor with information about whether you have a mutation in any of the genes that is thought to contribute to ARVC. 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 ARVC.

What are the Treatment Options for ARVC?

Currently, there are no therapies that can prevent or stop the progression of ARVC. Treatments are available that can improve symptoms and help better manage ARVC. Some treatment options include:

- Medications that may help relax the heart muscle and slow your heart rate so that your heart can pump more effectively (beta-blockers)
- Insertion of a device called an implantable cardioverter defibrillator (ICD)
- Surgical options to alter rhythm abnormalities

The *FAMILION*® ARVC Test:

The *FAMILION* ARVC Test may help your doctor accurately distinguish ARVC from other diseases with similar symptoms. Because ARVC is an inherited cardiac syndrome, there are distinct genetic signs that can help make or confirm a diagnosis of ARVC. Once the first person in your family is identified by genetic testing as having an ARVC gene mutation, other family members should be tested. Family testing is important because others may be at risk regardless of whether or not they are experiencing any symptoms.

For More Information

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

Sudden Arrhythmia Death Syndromes Foundation

800.STOPSADS (800.786.7723)

www.sads.org

Hypertrophic Cardiomyopathy Association

973.983.7429

www.4hcm.org

Cardiomyopathy Association

www.cardiomyopathy.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

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 4-6 weeks, your doctor will receive the 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 five different index testing options that your physician may order. These are:

- LQTS Test (Long QT Syndrome)
- CPVT Test (Catecholaminergic Polymorphic Ventricular Tachycardia)
- BrS Test (Brugada Syndrome)
- HCM (Hypertrophic Cardiomyopathy)
- ARVC (Arrhythmogenic Right Ventricular Cardiomyopathy)

Q. What is the Family Specific test?

A. The *FAMILION* LQTS, BrS, CPVT, HCM and ARVC Tests all have a Family Specific option. When the first person in a family is identified as having a genetic mutation associated with LQTS, BrS, CPVT, HCM or ARVC, first-degree (blood) relatives should also be tested. The *FAMILION* Family Specific Tests look only for the mutation found in the person already tested. 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 test 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, Monday thru Thursday, 8:30 a.m. - 6:30 p.m. ET and Friday, 8:30 a.m - 5:00 p.m. ET.

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), Monday thru Thursday, 8:30 a.m. - 6:30 p.m. ET and Friday, 8:30 a.m - 5:00 p.m. ET. 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.

PGxHealth Customer Service:

(P) 1-877-2-PGX-Health (877-274-9432)

(F) 203-786-3418 • www.pgxhealth.com

The **FAMILION**® Family of Genetic Tests

Cardiomyopathies



Cardiac Channelopathies

