

HCM GENETIC TESTING INFORMATIONAL GUIDE

Hypertrophic cardiomyopathy (HCM) is often caused by a genetic change that makes the heart muscle wall (the wall of the heart's main pumping chamber) thicken and stiffen, reducing blood flow into and out of the heart in some cases. HCM can be a chronic and progressive disease that gets worse over time, resulting in serious complications that leave you less able to do the activities you love or increasing your risk for atrial fibrillation, stroke, heart failure and sudden cardiac death. However, with appropriate modern management, it has been proven that HCM can be completely compatible with a normal life span and normal quality of life.

WHY SHOULD I CONSIDER GENETIC TESTING?

HCM can be caused by a change in your genes, which is passed on from generation to generation within a family. Genetic testing looks for these changes, sometimes called variants, in your DNA. Genetic testing is useful in many areas of medicine and can change the medical care you or your family member receives. Early identification for HCM is important to any treatment you may need and can also inform and guide the monitoring of your at-risk family members.

WHAT IS GENETIC COUNSELING?

Genetic counseling gives you information about how genetic conditions might affect you or your family. The genetic counselor will educate you on the process and potential results, as well as the potential risks and uncertainties related to testing. Counseling is also critical after genetic testing so the counselor can explain the results and potential consequences for your health and the health of your family members including children.

Genetic counselors can also advise and support patients on the best ways to communicate the news of any genetic variants they may discover to other family members. Although privacy laws restrict the ability of health care professionals to disseminate information directly to potentially affected relatives, they can provide written letters that explain the genetic findings, which you can give to your family members.



HOW SHOULD MY FAMILY BE TESTED?

If HCM is genetic, children of an affected parent each have a 50% chance of inheriting it. HCM is the most common form of inherited heart disease and can affect people of any age. Many people with HCM show no outward sign of the disease, so it's important for your children, siblings and parents to have their hearts checked. This can be done with an EKG and echocardiogram (ultrasound of the heart). Early identification of HCM in a family member may lead to earlier treatment and better outcomes. This is important because sometimes sudden cardiac death is the first indication that HCM is present in a family.

If the specific genetic cause of your HCM is found by genetic testing, your family members can be tested for this specific change. If they inherited the abnormal genetic change, they are at risk to develop HCM at some point in their life. If they did not inherit the abnormal change, they are not at risk to develop HCM or to pass it on to their children.

Genetic testing is also important if any family members have experienced these typical triggers for HCM evaluation:

- Cardiac event like arrhythmia or atrial fibrillation
- Heart murmur
- Abnormal EKG
- A diagnosis of “thick heart walls” or “athlete’s heart”

Once a genetic variant is identified within a family, all first-degree relatives should consider undergoing genetic testing and counseling for that specific variant whenever possible. Your genetic counselor can work with you through shared decision-making to determine additional family members who may be candidates for genetic testing.

HOW HCM GENETIC TESTS WORK

Genetic tests are minimally invasive and conducted using a blood or saliva sample. This sample is analyzed for 30-50 genes where mutations are linked to HCM. The patient's genes are compared to the genes of normal and HCM-affected patients from a database. Results are usually ready in a few weeks.

WHERE CAN I GET TESTED?

Genetic testing for rare and undiagnosed conditions should be ordered by a qualified health care professional. Your health care professional can provide information on available genetic testing and counseling services.

HOW MUCH WILL IT COST?

Many insurance companies including commercial plans, Medicare, and Medicaid will cover genetic testing. It's important to talk to your health care team and health insurance company for details. Prior authorizations may also need to be obtained. Your health care professional may be able to provide additional information.

As part of your genetics evaluation, the genetic counselor may be able to help you understand the costs.

IF GENETIC TESTS AREN'T CLEAR

Family screening can still take place even if the results of your genetic testing aren't helpful or if you choose not to pursue genetic testing. Ask your medical provider about these options for family screening.

ADDITIONAL INFORMATION

The Genetic Information Nondiscrimination Act, or GINA, was passed in 2008 to prevent medical insurers from raising your rates or dropping your coverage based on genetic test results. It also prevents people from employment discrimination based on genetic testing.

