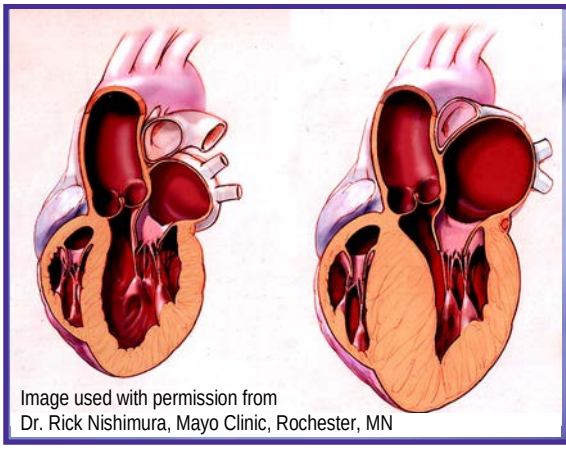

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Dr. Rick Nishimura, Mayo Clinic, Rochester, MN

Genetic testing for Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is a complex type of heart disease. It can include:

- Extra thick heart muscle (especially the bottom part of the heart called the left ventricle)
- Stiff heart muscle
- Mitral valve changes
- Heart muscle cells that are not arranged correctly (myocardial disarray)

These changes in the heart make it harder to pump blood and can cause arrhythmias (irregular heartbeats). About 1 in 500 people have HCM.

The muscles in the heart can get enlarged when they work harder than usual. This could be due to having high blood pressure or a narrow blood vessel (aortic stenosis), or even heavy exercise ("athlete's heart"). When heart muscles thicken without a clear reason, it is called hypertrophic cardiomyopathy (HCM).

How is HCM diagnosed?

HCM is diagnosed by a doctor based on your medical history, your family history, a physical exam, an ultrasound of your heart (echocardiogram), and heart rhythm testing (electrocardiogram or EKG). Thick heart walls can be seen by ultrasound and an irregular heartbeat may be found by EKG.

What causes HCM?

HCM is often due to changes in the genetic code (DNA) that cause the heart muscle to slowly thicken over time. There are many different genes that can cause HCM. These genes are the instructions for making different parts of the heart muscle. When one of the HCM genes is not working right, the heart muscle is not formed correctly. That makes the heart work harder and get thicker over time. Some people with HCM do not have a disease-causing change in any of the HCM genes we know about. It is very likely that there are other genetic causes of HCM that have not yet been discovered.

How is HCM inherited?

HCM can run in the family. If a person has a disease-causing change in one of the HCM genes, others in the family may have inherited the same change. These changes are called pathogenic variants. HCM is usually an autosomal dominant condition. This means that both men and women can have the condition and there is a 50% (1 in 2) chance to pass the genetic change to any child. Rarely, HCM is due to a gene with a different type of inheritance.

GATHERING YOUR FAMILY HISTORY

It is not always easy to recognize HCM. Many people with HCM are not aware of the condition. In some families, there may be only one or two people who develop heart symptoms that need medical attention. It is helpful to ask family members about medical problems that might be related to HCM.

Ask about:

- Unusual shortness of breath, especially during exercise
- Fainting or dizziness
- Palpitations (irregular fast heart beats)
- Chest pain or pressure during or after exercise
- Heart enlargement
- Sudden cardiac death or unexplained death
- Early heart failure

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How is genetic testing done?

A blood or saliva test can be done to look for pathogenic variants (genetic changes that can cause HCM). Genetic testing works best when there is a family history of HCM, and testing is done on a person who has HCM. Current testing finds a pathogenic variant in about 50% of people who have HCM and a history of HCM in their family. You can learn more about genetic testing by talking with a genetic counselor.

What are the benefits of genetic testing?

When a pathogenic variant is found in a person with HCM, other people in the family can be offered genetic testing. Testing can identify family members at risk for HCM, even if they have no heart symptoms. Healthy people with a pathogenic variant can lower the risk for heart problems by having regular cardiac screening. A family member who did not inherit the variant can be reassured.

What if testing does not find a genetic change that causes HCM?

Current testing is unable to find all genetic causes of HCM. Many people with HCM will get a test result with no pathogenic variant found in any of the tested genes. Cardiac screening would still be recommended for close relatives. There may be a genetic cause for HCM even with a negative (normal) result. A genetic cause is more likely when there is a strong family history of HCM.

What happens if I inherit a pathogenic variant in an HCM gene?

A person who inherits a pathogenic variant is more likely to develop HCM during their lifetime. However, some people with a pathogenic variant never develop HCM or any related heart problems. Regular cardiac screening is recommended for anyone with a genetic change that can cause HCM.

SCREENING FOR HYPERTROPHIC CARDIOMYOPATHY

Screening recommendations for those at risk for HCM include echocardiogram (heart ultrasound) and EKG (electrocardiogram). The frequency of screening depends on your age and medical history.

Echocardiogram is a heart ultrasound. It is the most common test used to diagnose hypertrophic cardiomyopathy. This test uses sound waves to produce images of the heart. Your doctor can see the thickness of your heart muscle, how the blood flows through your heart and how your heart valves move.

EKG (Electrocardiogram) is a test that checks the rhythm of your heartbeat. It records the electrical activity of your heart as it beats. An EKG can detect abnormal electrical signals that may result from thickened heart muscle. A Holter monitor or ZIO patch is a portable form of EKG that records your heartbeat for a much longer period of time. These monitors may be on the body for 24 to 48 hours or as long as 14-30 days.

What happens if I develop HCM?

Most people with HCM are able to live a normal life without severe heart problems. However, some people with HCM will have symptoms that can include breathing trouble, chest pain, fainting, dizziness, fatigue, and heart palpitations or arrhythmias. Less often, problems with blood flow through the heart or irregular heart rhythms can lead to sudden cardiac death. Treatment can help reduce or prevent symptoms and lower the risk of complications. Treatment includes regular follow-up, lifestyle changes, medications, and procedures, as needed. An automatic implantable cardiac defibrillator (AICD) may be placed when there are concerns about dangerous heart rhythms.

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