Genetic Counseling for Hypertrophic Cardiomyopathy

UpBeat
By **Heart Rhythm Society**

Hypertrophic cardiomyopathy (HCM) is a condition that causes the heart muscle to thicken or increase in size (hypertrophy), often in an uneven way.

WHAT ARE THE RISKS ASSOCIATED WITH HCM?

HCM may result in symptoms such as:

- **a racing heartbeat (palpitations),**
- **chest pain or pressure,**
- **shortness of breath (often during activity),**
- **loss of consciousness,**
- **stroke,**
- **and even sudden death**
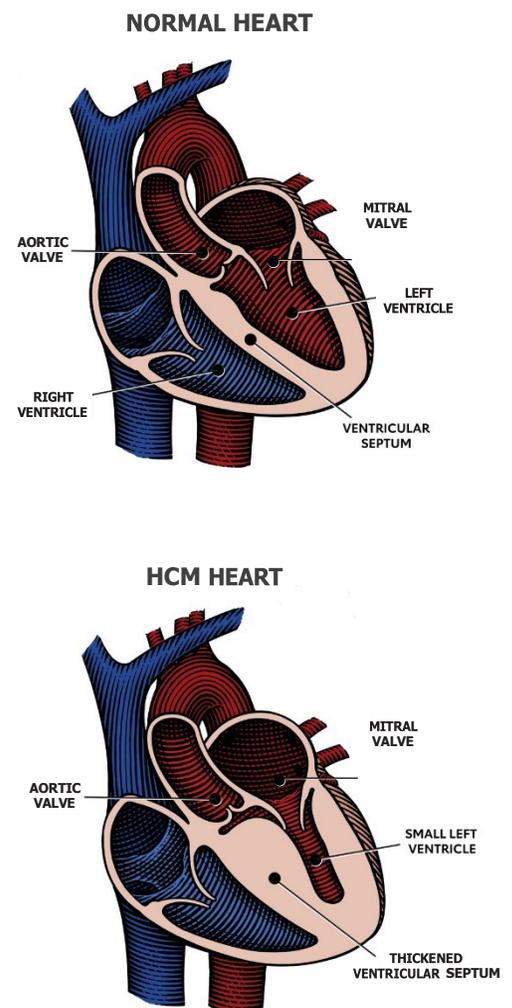
WHO IS AT RISK FOR HCM?

HCM can be passed on genetically. This means that each first-degree family member (i.e., parent, sibling, child) has a 50% chance of sharing the gene for HCM.

Individuals who inherit an HCM gene should be followed closely. They can develop any of the signs and symptoms listed above.

However, even if you have an HCM gene, you may not develop HCM symptoms. Many people who carry an HCM gene remain free of symptoms and live a normal lifespan.

For those with an HCM gene variant, family members who do not share that gene are not at heightened risk of developing HCM. No additional HCM screening or testing is needed.

Not all HCM is genetic. For families in which a primary HCM gene is not found, periodic screening for HCM by a cardiologist is recommended for family members every 3-5 years. The condition can develop at any age.

NORMAL HEART

AORTIC VALVE
MITRAL VALVE
LEFT VENTRICLE
RIGHT VENTRICLE
VENTRICULAR SEPTUM

HCM HEART

AORTIC VALVE
MITRAL VALVE
SMALL LEFT VENTRICLE
THICKENED VENTRICULAR SEPTUM

Illustrations of a normal heart (top) and a heart with hypertrophic cardiomyopathy (bottom).

Note that the heart walls (muscles) are much thicker (hypertrophied) in the heart with hypertrophic cardiomyopathy.

Image attribution: *American Heart Association*

WHAT TYPES OF TESTS ARE USED IN PATIENTS WITH HCM?

HCM can be detected during a physical exam (including hearing a heart murmur) or with an electrocardiogram (ECG). Additional tests can include genetic testing, and imaging tests such as an echocardiogram, or cardiac MRI scan. A cardiac monitor can be used to identify abnormal heart rhythms like atrial fibrillation or ventricular tachycardia.

WHAT IS THE ROLE OF GENETIC TESTING IN PATIENTS WITH HCM?

Genetic testing should be offered to all patients with HCM. For genetic testing, a blood or saliva sample is taken to test the person's DNA for gene changes that cause HCM. This testing is helpful to identify types of HCM with different risks or even specific treatments. Results of genetic testing are used to direct the care and management of patients and family members with HCM.

In about forty percent of people with a diagnosis of HCM, genetic testing can detect the presence of a disease-causing (pathogenic) HCM gene variant. If this type of gene variant is identified, first-degree family members can use genetic testing to see if they are at risk of developing HCM.

IF MY FAMILY MEMBER IS NOT EXPERIENCING ANY SYMPTOMS OF HCM, HOW IMPORTANT IS GENETIC TESTING?

If there is a known genetic cause of HCM in the family, genetic testing is very important. Some individuals may have HCM with no symptoms and may still be at risk of severe outcomes. Genetic testing can be used to identify those who are at risk.

"Some individuals may have HCM with no symptoms and may still be at risk of severe outcomes.

WILL GENETIC TESTING CHANGE MY TREATMENT PLAN IF I HAVE BEEN DIAGNOSED WITH HCM?

Genetic testing can identify the type of HCM that you have, and this can provide information about your HCM. Additionally, there are specific treatments for some types of HCM.

UpBeat
By Heart Rhythm Society

WHEN IS GENETIC COUNSELING ADVISED FOR PATIENTS WITH HCM?

Genetic **counseling** is recommended when pursuing genetic testing. Genetic counselors are specially trained healthcare providers. These counselors help patients understand the benefits and risks of genetic testing, help ensure the correct test is ordered, and help interpret the genetic test result. They are also helpful in guiding family member testing.

Where can I find more information on HCM?

- **Heart Rhythm Society (HRSonline.org)**
- **American Heart Association**
- **American College of Cardiology**
- **Hypertrophic Cardiomyopathy Association**

My questions for my doctor:

UpBeat
By Heart Rhythm Society