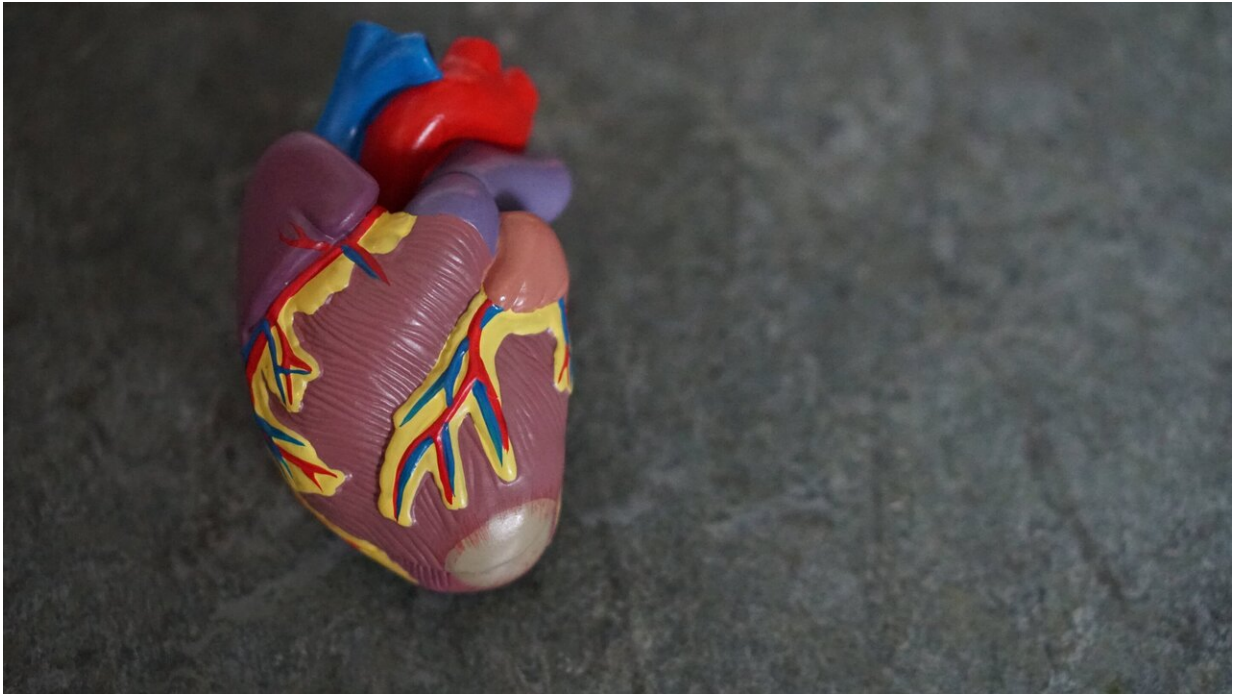


Q&A: Hypertrophic cardiomyopathy

February 8 2021, by Mayo Clinic News Network



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My sister was just told she has hypertrophic cardiomyopathy. I've heard of that causing sudden death in athletes. She says she feels fine and doesn't need treatment now, so is she at low risk? What does her diagnosis mean for me and my family?

Hypertrophic [cardiomyopathy](#) is a fairly common [heart](#) condition, affecting about 1 in 500 people. In [patients](#) with hypertrophic

cardiomyopathy, the heart walls become too thick because of excess heart muscle tissue, or hypertrophy.

Heart wall thickening can occur in other conditions, as well, such as hypertension, where the heart thickens to pump against higher blood pressure.

In hypertrophic cardiomyopathy patients, heart thickening is not a response to high blood pressure. Rather, it is most commonly related to abnormalities in the genes related to heart muscle cells. Hypertrophic cardiomyopathy usually is diagnosed using an ultrasound of the heart, called a transthoracic echocardiogram.

Many patients with hypertrophic cardiomyopathy have no symptoms, and the condition often is discovered when a provider hears a murmur on cardiac examination and orders a transthoracic echocardiogram to investigate. Other patients may discover that they have hypertrophic cardiomyopathy when they present with symptoms like shortness of breath with activity, chest pain while exerting, lightheadedness or a feeling of abnormal heartbeat.

About two-thirds of patients with hypertrophic cardiomyopathy have "obstruction," where blood flow through the heart becomes more turbulent related to the thickening of the heart walls and crowding of the inside of the heart. This type of turbulent blood flow is one cause of a heart murmur.

In asymptomatic patients with hypertrophic cardiomyopathy, like your sister, new medications are not needed. However, patients experiencing symptoms related to obstruction may need to have their heart medicines adjusted.

The most commonly prescribed medicine to treat symptoms from

hypertrophic cardiomyopathy is a class of drugs called beta blockers. Some patients can require heart procedures to treat severe symptoms that don't respond to medications. Infrequently, patients with hypertrophic cardiomyopathy who have medically refractory or severe symptoms can require heart transplant.

Oftentimes, one of the main concerns of a patient or family member of a patient with newly diagnosed hypertrophic cardiomyopathy is an increased risk of sudden cardiac [death](#). Although hypertrophic cardiomyopathy is one of the main causes of sudden cardiac death in [young patients](#), most patients with hypertrophic cardiomyopathy have a normal life span. Your sister should work with a multidisciplinary hypertrophic cardiomyopathy center, such as Mayo Clinic, where providers with extensive experience evaluating and treating patients with hypertrophic cardiomyopathy can provide appropriate and consistent monitoring to determine her risk for sudden cardiac death.

Sudden death in hypertrophic cardiomyopathy is caused by an abnormal heart rhythm, called ventricular tachycardia or ventricular fibrillation. A person's risk of sudden cardiac death is unrelated to symptoms. That means that patients who feel well can have high [sudden death](#) risk, and very symptomatic patients can have low risk.

When people are diagnosed with hypertrophic cardiomyopathy, testing is performed to understand their risk of sudden death. This testing can include a heart rhythm monitor, a heart MRI and an exercise stress test. It's also important to understand [family history](#) when determining sudden death risk. In patients found to be at high risk for sudden death, an implantable cardioverter defibrillator can be implanted to lower that risk.

Because hypertrophic cardiomyopathy can be inherited, screening of [family members](#) is needed, even if they have no symptoms. The reason

to screen asymptomatic family members is because sudden death risk does not correlate with symptoms.

Current guidelines recommend screening first-degree family members of patients who are diagnosed with hypertrophic cardiomyopathy. For your family, this would mean that if your sister has children, they should be screened prior to the onset of puberty. This screening can help them understand their risks and guide what level of activity participation is advised. In addition to being screened yourself, your parents and any other siblings should be screened.

Screening can occur in two ways: via genetic testing or echocardiography. Genetic testing is a blood test that would first be performed in your sister. If the genetic cause of her hypertrophic cardiomyopathy is identified, this can be used to screen you and other family members.

Use a genetic counselor to help with the process of genetic screening. If [genetic testing](#) is not pursued or if a genetic cause of hypertrophic cardiomyopathy is not identified in your sister, periodic screening with an echocardiogram for you and the [family](#) members discussed would be recommended.

An echocardiogram can be arranged by a primary care provider or a cardiologist. In adults, this should occur every three to five years. In children or adolescents, this should occur every one to three years.

While it's great to hear that your sister doesn't have symptoms, she should be aware of any changes and talk with her health care provider. Most patients with hypertrophic cardiomyopathy have a normal life span, but it is important that your sister maintain care with a cardiologist who is comfortable navigating [hypertrophic cardiomyopathy](#).

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