



## Hypertrophic Cardiomyopathy

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# Hypertrophic Cardiomyopathy

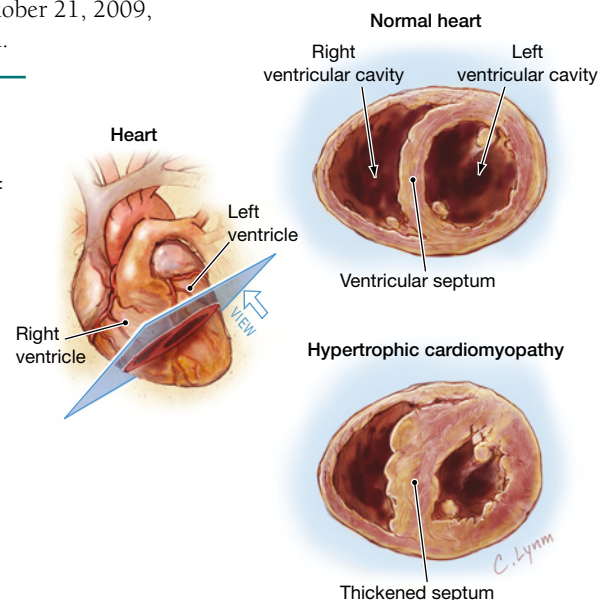
Cardiomyopathies are diseases affecting heart muscle. In **hypertrophic cardiomyopathy** (HCM, aka idiopathic hypertrophic subaortic stenosis, asymmetric septal hypertrophy, hypertrophic obstructive cardiomyopathy) **sarcomeres** (units of heart muscle) enlarge, causing heart muscle cells to **hypertrophy** (increase in size). This causes the heart muscle to thicken unevenly and changes the way different parts of the heart move, which then requires it to work harder to pump blood. In HCM, hypertrophy usually involves the **septum** (wall) between the **ventricles** (lower heart chambers), which differs from other types of cardiomyopathy. As part of the hypertrophy process, the normal pattern of muscle cells is upset and can lead to problems with electrical function of the heart, resulting in **arrhythmias** (abnormal heartbeat). The first case of HCM was diagnosed 50 years ago. It has since been found to affect 0.2% to 0.5% of the general population and appears in all racial groups. It is the most common cause of sudden death in young people, including trained athletes, and causes heart failure disability at any age. This disorder is different from left ventricular hypertrophy, which is usually caused by undertreated hypertension. The October 21, 2009, issue of *JAMA* includes an article about HCM transmitted by sperm donation.

## CAUSE

Transmission of HCM is **genetic** (inherited). HCM is transmitted to 50% of individuals in each subsequent generation. It is due to a mutation in at least 1 of 11 genes coding for sarcomere protein. In persons without a family history of HCM, the most common cause is a new mutation.

## SYMPTOMS AND DIAGNOSIS

Symptoms are shortness of breath, chest pain, dizziness, faintness, and palpitations. In children, the first sign of HCM is often sudden **cardiac arrest** (collapse and possible death) due to arrhythmias. Although it is possible to diagnose HCM through physical examination, most physicians confirm it through additional testing. The most common test is 2-dimensional **echocardiography** (ultrasonic waves directed at tissues), although magnetic resonance imaging (MRI) is becoming more widely used. Genetic testing using a blood test is the most sensitive diagnostic test. Other tests used in evaluating HCM include **electrocardiogram** (graphic tracing of electric potential caused by excitation of heart muscle) and 24-hour **Holter monitor** (monitor for heart rhythm).



## TREATMENT

Once HCM is identified in a person, all immediate family members should be tested as soon as possible. Medications such as beta-blockers or calcium channel blockers are the primary way to control symptoms. Other possible treatments include **septal myectomy** (removing a portion of the wall between the ventricles), alcohol septal **ablation** (destroying a portion of the wall), pacemakers, implantable defibrillators, and heart transplantation. **Diuretics** (water pills) should be avoided.

## PROGNOSIS

HCM can cause sudden death even in children and young adults, often during or just after vigorous exertion. However, others with HCM have no symptoms and live a normal lifespan. Remarkably, the first patient identified with HCM is still alive. He received a heart transplant in 1989.

Sources: National Institutes of Health, American Heart Association

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