



## Hypertrophic Cardiomyopathy (HCM)

### CONTACT US

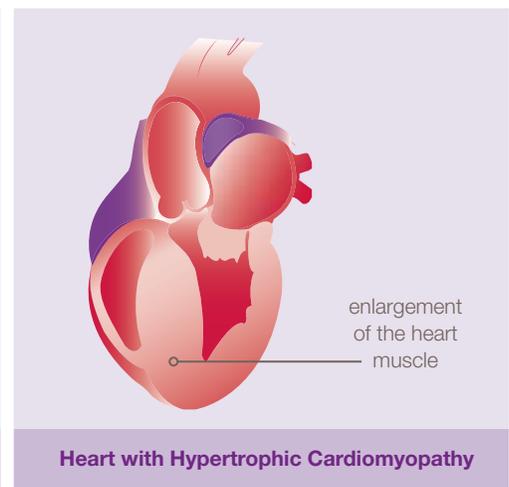
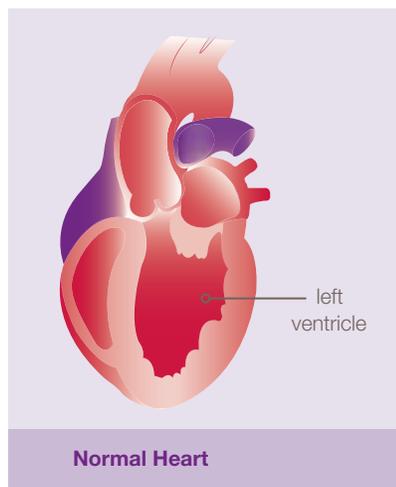
To make an appointment or to learn more about treatment of Hypertrophic Cardiomyopathy at Cincinnati Children's Hospital Medical Center, please call **513-803-1746**.

The Cardiomyopathy/Heart Failure Clinic cares for children and adults and has a focus on family centered care.

### Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is a condition of heart muscle disease in which the muscle is thickened (hypertrophic). This thickening typically occurs in the lower left chamber of the heart, called the left ventricle. Thickening of the heart muscle can occur at the septum (muscular wall that separates the left and right side of the heart), the posterior wall or free wall (outside wall of the left ventricle), the apex (the bottom of the heart) or throughout the entire left ventricle. When the muscle becomes thickened, it may make it difficult for an efficient amount of blood to flow into and out of the heart, especially during exercise. In some cases, the thickening of the muscle can block blood flow from the left ventricle to the aorta. This is called "outflow tract obstruction."

The cardiomyocytes (heart muscle cells) in individuals with HCM have characteristic features. Under a microscope, the cells appear disorganized and irregular (myocyte disarray). This disarray may cause changes in the electrical signals traveling through the lower chambers of the heart and lead to ventricular arrhythmias (abnormal heart rhythm). Hypertrophic cardiomyopathy affects an estimated 600,000 to 1.5 million Americans, or one in 500 people.



## Symptoms

Many individuals with HCM experience no symptoms. For those who have heart failure or ventricular arrhythmias, symptoms can include:

### Older children and adults

- Shortness of breath and fatigue
- Feeling dizzy or light-headed
- Fainting or passing out (syncope)
- Chest pain
- Palpitations (feeling abnormal heart beats)

### Newborns and babies

- Excessive sweating
- Difficulty feeding or poor growth

Even though some people can have HCM and experience no symptoms, they may still be at risk for complications of the condition. The most serious complication of HCM is sudden cardiac arrest. Sudden cardiac arrest is a sudden loss of heart function caused by a dangerously fast heart rhythm called ventricular tachycardia. Unless emergency treatments, including CPR and defibrillation, are initiated immediately, sudden cardiac death can occur. Most people with HCM have a low risk for sudden cardiac death. However, HCM is the most common cause of sudden cardiac death in people under age 30. For this reason, it is important for anyone at risk to be followed by a cardiologist with experience in caring for individuals with HCM.

## Causes

HCM is largely a genetic condition. When an individual is diagnosed with HCM, there is a 40 to 70 percent chance that an underlying genetic cause will be identified. Most commonly, the genetic cause results in a condition that runs in a family and affects only the heart. It is possible that parents, siblings, and even aunts/uncles or cousins of an individual diagnosed with HCM may also have this heart muscle condition. If they haven't had cardiac screening, family members may not know they are affected. There are other genetic causes of HCM which can be associated with other health problems. These include genetic syndromes, metabolic, and mitochondrial disorders.

Hypertrophy can be acquired and is not always genetic. For example, hypertrophy can result from high blood pressure that has gone untreated or poorly controlled for a period of time. Individuals who are endurance athletes can also develop hypertrophy. In some people, the cause of HCM is unknown.

## Diagnosis

The diagnosis of HCM is based on family history, medical history, physical exam, and cardiac testing.

- **Family history:** A family tree should be constructed with specific attention to cardiomyopathy, rhythm problems, sudden cardiac or unexplained death, cardiac surgery or presence of other cardiac disease in relatives.
- **Medical history:** A history of heart failure symptoms, rhythm problems and passing out is important.
- **Physical exam:** Special attention should be paid to the cardiac and skeletal muscle systems.
- **Cardiac testing:** An echocardiogram is the most common first test used to diagnose HCM, as the characteristic thickening of the heart walls is usually visible on the echocardiogram. To further define the muscle abnormality and to determine if there is scar in the heart a cardiac magnetic resonance imaging (MRI) may be ordered.

Other tests: Blood tests, electrocardiogram (EKG), exercise stress echo test and cardiac catheterization may also be recommended.

## Management and Treatment

The treatment for individuals with HCM is different for each person and is based on a number of features including the presence of outflow tract obstruction (blood isn't able to leave the left ventricle because of the thickened muscle), the function of the heart (how well it is squeezing), the presence of any symptoms, the age and activity level of the patient, the presence of abnormal heart rhythms and the family history. Treatment is aimed at minimizing or preventing symptoms and reducing the risk of complications such as heart failure and sudden cardiac arrest.

Individuals with HCM need to be followed by a cardiologist on a regular basis. Medications are used to treat symptoms and prevent further complications. Medications such as beta-blockers and calcium channel blockers relax the heart muscle, allowing it to function more efficiently. Other medications may be prescribed as needed to control heart rate or decrease the occurrence of arrhythmias.

Lifestyle changes are recommended for some individuals. Specifically, participating in competitive or endurance sports places individuals at higher risk for heart rhythm problems and are not recommended for individuals with HCM. A medical procedure may be needed if the heart muscle becomes too thick or if a rhythm problem is detected.

Most people with HCM have a low risk for sudden cardiac death. However, it is important to identify the small number of patients with HCM who do have a higher risk for sudden cardiac arrest, so preventive measures can be taken. A physician can assess an individual's risk and may prescribe preventive treatments such as antiarrhythmic medications or an implantable cardioverter defibrillator (ICD) to reduce the chance of a sudden cardiac death.

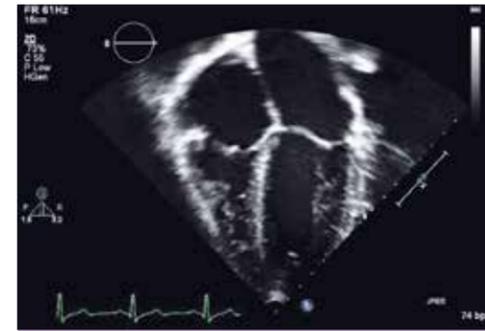


Fig 1.1

Normal Heart

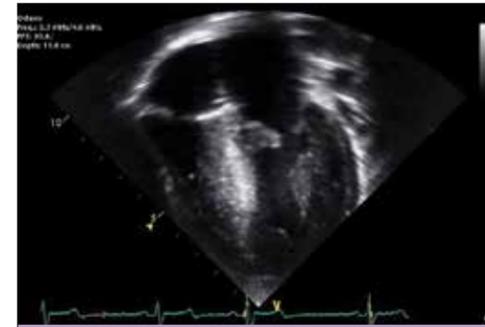


Fig 1.2

Heart with Hypertrophic Cardiomyopathy

## RESOURCES FOR FAMILIES

### The Cardiomyopathy Association:

[www.cardiomyopathy.org](http://www.cardiomyopathy.org)

### Children's Cardiomyopathy Foundation:

[www.childrenscardiomyopathy.org](http://www.childrenscardiomyopathy.org)

### Hypertrophic Cardiomyopathy Foundation:

[www.4hcm.org](http://www.4hcm.org)



Cincinnati Children's is ranked #8 in Cardiology and Heart Surgery, and third among all Honor Roll hospitals in the 2012–13 *U.S. News & World Report* listing of Best Children's Hospitals.

## Screening for Family Members

All first degree relatives of an individual who has HCM should undergo routine cardiac evaluation. This includes parents, brothers, sisters, and children. If a gene mutation causing HCM in an individual is known, screening is recommended for those family members proven to also carry the gene mutation. Routine cardiac screening would not be recommended for family members who did not inherit the gene mutation.

The timing of routine cardiac screening for family members should be discussed with a cardiologist and genetics professionals who have experience in caring for individuals with cardiomyopathy. How frequently an individual should be evaluated is based on published guidelines and the individual's age, personal medical and family history. The best screening tools for HCM include echocardiography and magnetic resonance imaging (MRI).

## Genetic Testing

Genes are packages of genetic information that tell our bodies how to develop and function. HCM is often caused by a change or mutation in a gene that encodes proteins important for the contraction or squeeze of the heart muscle. Gene mutations in more than 20 genes have been identified that cause HCM. HCM is most often inherited in an autosomal dominant manner. A parent that carries a gene mutation will have a 50% chance of passing it on to each child.

Genetic testing is available to look for mutations in the genes known to cause HCM. This type of testing is done on a blood sample. It is best to begin genetic testing in a family with an individual who is known to have HCM. If a gene mutation is found in someone with HCM, genetic testing for the same mutation can be offered to close family members. Finding a gene mutation in other family members will help identify family members who may be at risk to develop HCM.

A genetic counselor and/or geneticist can assess the family history and provide information about the chance that there is a genetic predisposition to cardiomyopathy. A genetics professional can also explain and facilitate genetic testing and interpret results for patients and families.

For immediate consultations, physician referrals and patient care follow-up, call the National Physician Priority Link at **888-987-7997**.

[www.cincinnatichildrens.org](http://www.cincinnatichildrens.org)