What is hypertrophic cardiomyopathy?

Hypertrophic cardiomyopathy (HCM) is the second most common form affecting about 40 to 50 percent of children with cardiomyopathy. "Hypertrophic" refers to the **abnormal growth of muscle fibers** in the heart. With HCM, the pumping function is normal but the heart muscle is thickened and stiff, making it difficult for the heart to relax and for blood to fill the heart's chambers. This limited filling prevents the heart from supplying enough blood to the body, especially during exercise. HCM usually affects the heart's main pumping chamber (left ventricle), and the muscle wall (septum) that separates the two lower chambers of the heart is thickened.

There are different types of heart muscle thickening patterns. **Asymmetric septal hypertrophy** is when the septum is thicker than the rest of the wall; **concentric hypertrophy** is when the thickening is evenly distributed in the entire ventricle; and **apical hypertrophy** is when there is localized thickening at the tip of the heart.

With asymmetric septal hypertrophy, it can occur with obstruction or without obstruction. **Hypertrophic obstructive cardiomyopathy** (HOCM), also known as idiopathic hypertrophic subaortic stenosis (IHSS), is when the thickened heart muscle restricts blood flow out of the heart. Obstruction can cause the heart's mitral valve, located between the heart's two left chambers, to leak. In mitral regurgitation, the blood flows backwards from the heart's lower chamber (left ventricle) back into the upper chamber (left atrium).

Information

What is the prognosis?

The outcome of HCM is **highly variable**, with some children remaining asymptomatic and some developing mild, moderate or severe symptoms. Heart failure and sudden death may be a risk factor for some children. In less than 10 percent of patients, the disease may progress to a point where the heart muscle thins and stretches (dilates), and the pumping performance deteriorates. In general, a heart transplant is less common for children with HCM and is only recommended when there is unmanageable heart failure.

The Children's Cardiomyopathy Foundation (CCF) is a national non-profit organization dedicated to finding causes and cures for pediatric cardiomyopathy through the support of research, education, and increased awareness and advocacy.

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Understanding Hypertrophic Cardiomyopathy

How many children are affected?

HCM affects up to 500,000 people in the U.S., with children under the age of 12 accounting for less than 10 percent of all cases. According to the Pediatric Cardiomyopathy Registry, HCM occurs at a rate of **5 per million children** and is most often diagnosed during infancy and adolescence.

What causes hypertrophic cardiomyopathy?

HCM is often caused by genetic defects (mutations) that run in a family. Mutations are changes in the DNA of a gene that can be **inherited or occur spontaneously** during fetal development for unknown reasons. It is estimated that 50 to 60 percent of children with HCM have identifiable genetic causes. Many inherited HCM cases are caused by mutations in the sarcomeric genes that affect the proteins responsible for contraction of the heart muscle. The effect of this type of HCM is isolated to the heart. Mutations in the sarcomeric genes are inherited in an autosomal dominant manner in which one parent contributes the defective gene and there is a 50 percent chance their child will inherit the condition. Because the impact of these genetic mutations can differ, the severity of the disease can vary widely among family members.

Sarcomeric mutations are the most common cause of HCM, but the disease can also be caused by systemic disorders (fatty acid defects, glycogen storage diseases, organic acidemias, lysosomal storage disorders, mitochondrial defects) that affect many areas of the body. Other possible causes include genetic malformation syndromes (Noonan syndrome) and neuromuscular disorders (Friedreich's ataxia). To date, more than 40 genes with more than 800 mutations have been identified to cause HCM.

Infants and young children may develop HCM as a result of an inherited **metabolic or mitochondrial condition**. In these cases, the body is unable to break down foods to produce energy. The resulting accumulation of fats or sugars (glycogen) cause the heart walls to become thicker. These rare disorders are usually inherited in an autosomal recessive manner in which both parents contribute a defective gene and there is a 25 percent chance their child will inherit the condition.

What are the common symptoms?

There is **tremendous variation** in how the disease presents and progresses. In general, children under one year often present with a more serious form of HCM whereas some older children may show mild to no symptoms. The onset of symptoms usually coincides with the rapid growth and development of late childhood and early adolescence. Competitive sports also have been known to make symptoms of HCM more pronounced.

Disease severity and symptoms are related to the location and extent of the hypertrophy and whether there is obstruction or mitral regurgitation. Children with HCM may experience shortness of breath (dyspnea), chest

Symptoms

pain (angina), **light-headedness or dizziness** (presyncope), fainting (syncope), difficulty exercising and irregular heartbeats (palpitations). Symptoms in infants are more difficult to detect but include difficulty breathing, slow weight gain, excessive sweating (diaphoresis) and agitation during feeding due to **chest pain**. Children with severe HCM may show symptoms of heart failure such as difficulty breathing; extreme fatigue; a persistent cough; abdominal pain; vomiting; and swelling of the abdomen, legs and ankles (edema).

Some children may develop **abnormal heart rhythms** called **arrhythmia** where the heart beats too slow (bradycardia) or too fast (tachycardia). Signs include feeling irregular, rapid and forceful heartbeats (palpitations), fainting (syncope) and convulsions (seizures). Arrhythmia may occur at any stage of the disease regardless of congestive heart failure.

How do you diagnose hypertrophic cardiomyopathy?

HCM may be diagnosed in a physical examination when a heart murmur is detected, although this may be absent with non-obstructive HCM. To formally diagnose HCM, cardiologists rely on noninvasive cardiac tests such as the **echocardiogram** and **electrocardiogram** (EKG). An echocardiogram indicates the location and extent of the muscle thickness, the heart's pumping efficiency, and the severity of any obstruction and mitral regurgitation. From this test, the speed at which blood flows into the heart (velocity) and the percentage of blood ejected from the heart with each beat (ejection fraction) can be calculated. A reduced velocity and higher than normal ejection fraction of 70 to 90 percent confirms a diagnosis of HCM. An EKG provides information on the heart's electrical activity, abnormal heart rhythms and heart size. This test nearly always shows higher than normal voltages associated with thickened pumping chambers.

Other tests may be ordered to access the heart's condition and determine a treatment plan. These tests include an **exercise stress test** to see how the heart responds to exercise, a **Holter monitor** to look for abnormal heartbeats, and **magnetic resonance imaging** (MRI) to measure muscle thickness and evaluate heart and blood vessel function.

In some cases, more invasive cardiac tests may be necessary. A **cardiac catheterization** may be performed to evaluate the heart's pumping ability, the degree of obstruction and the need for a heart transplant. Because HCM patients are more susceptible to fatal arrhythmias, an **electrophysiology** (EP) **study** may be done at the same time to study the heart's electrical system. During the EP test, the physician will try to induce a fast heart rhythm (tachycardia) to determine whether anti-arrhythmic medication or an automatic implantable cardioverter defibrillator (AICD) are needed to protect the child from sudden cardiac arrest.

What are the treatment options?

For children with HCM, medical therapy aims to control symptoms related to heart obstruction, improve filling of the heart chambers and prevent arrhythmias. With optimal treatment, symptoms can be improved or eliminated in many children with HCM.

Several types of **medications** are used to control HCM in children. Beta-blockers (atenolol, metoprolol, propranolol) and calcium channel blockers (verapamil) may be prescribed to children with obstructive HCM to reduce the heart's workload by slowing the heart rate and decreasing the force and contraction of the heart muscle. Diuretics (furosemide, spironolactone) help to reduce excess fluid in the body and may be recommended in advanced stages of heart failure. For children with heart rhythm problems, anti-arrhythmic medications (amiodarone, digoxin, disopyramide, procaineamide, verapamil) may be used to keep the heart beating at a regular rate. In some cases, an anti-coagulant (aspirin, dipyridamole, enoxaparin, heparin, warfarin) may be used to reduce the risk of stroke associated with HCM and atrial fibrillation.

An **automatic implantable cardioverter defibrillator** (AICD) or a **pacemaker** may be surgically implanted to control arrhythmias that do not respond to medication and to prevent sudden cardiac death. Children considered high risk include those who have experienced fainting, cardiac arrest, show a drop in blood pressure during exercise, have a family history of cardiac arrest or show signs of severe arrhythmia. An AICD automatically corrects life-threatening arrhythmias (ventricular fibrillation, ventricular tachycardia) that interfere with the heart's pumping ability. A pacemaker normalizes other arrhythmias and may be used in special circumstances to relieve obstruction associated with HOCM.

In patients with severe obstruction or mitral regurgitation, surgery may be recommended to lessen the obstruction and improve symptoms associated with HCM. During a **septal myectomy**, excess heart muscle is removed to relieve obstruction and, if necessary, a leaky mitral valve is repaired or replaced.

Treatment