

# HYPERTROPHIC CARDIOMYOPATHY

Most often diagnosed during infancy or adolescence, hypertrophic cardiomyopathy (HCM) is the second most common form of heart muscle disease, is usually genetically transmitted, and comprises about 35–40% of cardiomyopathies in children. A diagram and echocardiogram comparing a normal heart and a heart with HCM are shown in figures 2a and 2b.

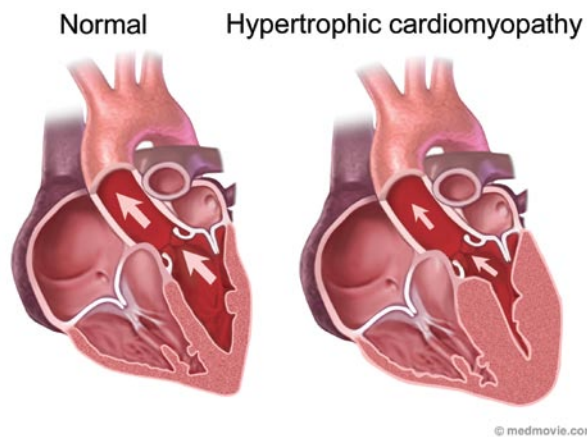


Figure 2a- A normal heart is shown on the left compared to a heart with a hypertrophic cardiomyopathy on the right. Note the increased thickness of the walls of the left ventricle.

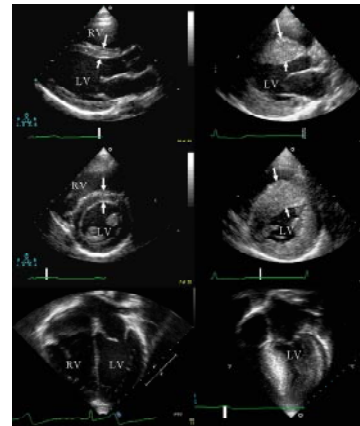


Figure 2b- Multiple echocardiographic views of a normal heart on the left and a heart with hypertrophic cardiomyopathy on the right. Note the increased thickness of the walls of the left ventricle (LV).

HCM affects up to 500,000 people in the United States, with children under age 12 accounting for less than 10% of all cases. According to the Pediatric Cardiomyopathy Registry, HCM occurs at a rate of five per 1 million children.

“Hypertrophic” refers to an abnormal growth of muscle fibers in the heart. In HCM, the thick heart muscle is stiff, making it difficult for the heart to relax and for blood to fill the heart chambers. While the heart squeezes normally, the limited filling prevents the heart from pumping enough blood, especially during exercise.

Although HCM can involve both lower chambers, it usually affects the main pumping chamber (left ventricle) with thickening of the septum (wall separating the pumping chambers), posterior wall or both. With hypertrophic obstructive cardiomyopathy (HOCM), the muscle thickening restricts the flow of blood out of the heart. Often, leakage of the mitral valve causes the blood in the lower chamber (left ventricle) to leak back into the upper chamber (left atrium). In less than 10% of patients, the disease may progress to a point where the heart muscle thins and the left ventricle dilates resulting in reduced heart function similar to that seen in DCM.

HCM is most often diagnosed during infancy or adolescence. Gene defects can be familial, and it is estimated that 50–60% of children with HCM have a relative with the disease, although they may not have been diagnosed or have symptoms.

## Signs and Symptoms of HCM

There is tremendous variation in how HCM presents and progresses. While some children have no or mild symptoms, others may have more severe symptoms including heart failure. Some patients develop abnormal heart rhythms (arrhythmias) that may put them at increased risk for sudden cardiac death. Children under 1 year of age often have symptoms of congestive heart failure whereas older children may be symptom free and, therefore, may be unaware that HCM is present. Onset of symptoms often coincides with the rapid growth and development of late childhood and early adolescence. The strenuous exercise of competitive sports has also been known to make symptoms of HCM more apparent.

Disease severity and symptoms are related to the extent and location of the hypertrophy and whether there is obstruction to blood leaving the heart or valve leakage from the left-sided pumping chamber back into the left atrium above. The first sign of HCM may be a murmur, although this is usually absent in the non-obstructive form of HCM. Symptoms in children with HCM can include dyspnea (shortness of breath during exercise), angina (chest pain), presyncope (light-headedness or dizziness), syncope (fainting), exercise intolerance or palpitations/arrhythmias (irregular heartbeats). Symptoms in infants may be more difficult to detect but include difficulty breathing, poor growth, excessive sweating (diaphoresis) or crying and agitation during feeding thought to be due to chest pain.

Children with severe HCM may have symptoms of heart failure such as difficulty breathing, swelling around the eyes

and legs (edema), tiredness or weakness, coughing, abdominal pain and vomiting. Mild symptoms of heart failure can also resemble asthma. Children with HCM may also develop an abnormal heartbeat (arrhythmia), either beating too fast (tachycardia) or too slow (bradycardia). Symptoms resulting from rhythm problems can appear without a child having congestive heart failure or other more obvious symptoms of HCM. The risk of sudden death from arrhythmia is higher with this form of cardiomyopathy compared with other forms of pediatric myopathy especially among adolescent patients. Finally, in some cases of HCM (especially those with extreme wall thickness), the disease evolves towards progressive wall thinning and LV chamber dilation until the heart appears to have all the features of DCM. In these cases, a family history of HCM, a prior echocardiogram that showed HCM or a cardiac biopsy may help differentiate between the two.

## **Diagnosis of HCM**

Once suspected, the diagnosis of hypertrophic cardiomyopathy is established with an echocardiogram (or ultrasound of the heart) looking for abnormally thick walls predominantly in the left pumping chamber (left ventricle). In addition, the extent of obstruction or muscular narrowing through the outlet of the left ventricle to the aorta (main vessel which carries blood to the body) will be assessed. This diagnosis can only be made after other potential causes of abnormal wall thickening (i.e., aortic valve stenosis, coarctation of the aorta, high blood pressure, etc.) are eliminated either by physical exam or echocardiogram.

An electrocardiogram, or EKG, which records the electrical impulses sent through the heart, may show evidence of thickened pumping chambers. Many cardiologists will order a Holter monitor to record your child's heartbeats over a 24–48 hour period. This will allow your physician to check for abnormal, and sometimes life-threatening, heart rhythms, which can occur more often in children with HCM. To further estimate your child's risk for developing these abnormal heart rhythms, some cardiologists may ask, in children old enough to cooperate, to perform an exercise treadmill test.

Since the cause of this form of cardiomyopathy varies and depends on your child's age, additional laboratory testing may be requested. In some cases, an accumulation of abnormal proteins or sugars (glycogen) may occur in the heart causing the increased wall thicknesses. In others, genetic mutations or abnormalities in the mitochondria (powerhouses of the cells) produce this effect. Blood testing and, in some cases, a muscle biopsy from the leg or arm can help identify the cause or help your doctor formulate a prognosis.

Genetic testing is available for some but not all forms of HCM. In the next decade, as more genetic bases for HCM are identified, genetic testing may become a routine part of the HCM evaluation.

Finally, because most patients with this diagnosis have inherited the disease, once confirmed in your child, your cardiologist will likely recommend screening (usually with echocardiogram) of the child's parents, siblings and perhaps other close relatives to discover whether this disease is present in any other family members.

## **Causes of HCM**

Mounting scientific evidence is beginning to suggest that individual genetic "mutations" may be among the more common causes of HCM in children. For a greater understanding of the basics of human inheritance patterns and a more detailed discussion of the potential genetic causes of HCM, the reader is encouraged to read separate sections entitled "Overview of Inheritance" and "Genetics of Cardiomyopathies" printed elsewhere in this brochure.

## **Current Treatment for HCM**

Currently, there are no therapies that can "cure" HCM; however, many treatments are available that can improve symptoms and potentially decrease risk in children with HCM. The choice of a specific therapy depends on the clinical condition of the child, the risk of dangerous events and the ability of the child to tolerate the therapy. In the following sections, treatments for HCM are summarized.

## **Medical Therapies**

Medications are used to treat children with HCM who have symptoms such as difficulty breathing, chest pain, decreased activity tolerance or fatigue and generally include beta-blocking and calcium channel-blocking medicines. Beta-blocking medications are used to slow the heartbeat and allow the heart to fill more completely when the thick muscle in the ventricular septum narrows the outflow of blood from the heart. These medications can cause excessive slowing of the heart rate, low blood pressure, dizziness, and in some cases, fluid retention, fatigue, impaired school performance and depression. Calcium channel blockers improve the filling of the heart by reducing the stiffness of the heart muscle, and

are used in patients with chest pain or breathlessness. Side effects can include excessive slowing of the heart rate and lower blood pressure. Common calcium channel blockers are verapamil and diltiazem. Diuretics, which must be used with caution in this disease, are used to decrease fluid accumulation (when present) from the lungs in children with severe hypertrophic cardiomyopathy. In the case of HCM which has progressed into a myopathy with DCM features (occurs relatively infrequently as a later stage of HCM - see previous discussion of HCM), therapies used to treat HCM will no longer be effective, and standard medicines for the treatment of DCM will be substituted.

## **Pacing Therapies for HCM**

### **Implantable Cardioverter Defibrillator (ICD)**

ICDs are designed to prevent sudden death from a serious arrhythmia known as ventricular tachycardia or fibrillation. An ICD constantly monitors heart rate, and when ventricular tachycardia or fibrillation is detected, the ICD delivers a shock to the heart that restores normal rhythm. ICDs are used in patients with hypertrophic cardiomyopathy who are felt to be at high risk for a sudden death or have documented serious ventricular arrhythmias.

### **Surgery for HCM**

Septal myomectomy is a surgery that is performed in patients with hypertrophic cardiomyopathy when symptoms of heart failure have developed because the thickened muscle of the septum (the wall dividing the two sides of the heart) has narrowed (obstructed) the blood flow out of the heart, and medical therapy has not been effective. The surgery involves removing the muscle that has obstructed the blood flow. While septal myomectomy is effective in controlling symptoms, it does not stop hypertrophy from progressing, nor does it treat the life-threatening abnormal rhythms associated with HCM.

### **Heart Transplantation**

Currently, transplantation for HCM is not routinely performed. Two exceptions include medically refractory ventricular arrhythmias, and HCM which has developed features of DCM not responsive to standard DCM therapy. A heart transplant will offer that child the chance to return to a normal lifestyle. While a donor heart can cure the symptoms of heart failure and greatly improve survival, it is a major operation with considerable risks and long-term complications. Once a transplant is done, other concerns arise, such as infection, organ rejection, coronary artery disease, and the side effects of medications.

### **Prognosis**



Many children with HCM can lead relatively normal lives once the diagnosis has been established and appropriate therapies have been started. The long-term prognosis varies depending on the cause, severity and degree of functional impairment. Children with HCM should be watched closely for increasing symptoms of activity intolerance and the development of DCM features with heart failure and symptoms of palpitations or syncope which might suggest abnormal heart rhythms that require more thorough investigation. It is important that a child with HCM be closely monitored at a center with expertise in pediatric cardiomyopathy, arrhythmias and transplantation in order to ensure rapid response to any worsening of his or her condition.

The outcome of HCM is highly variable with some patients remaining asymptomatic, some developing symptoms and possibly progressive heart failure, and all remaining at risk for sudden death. Heart transplant is performed uncommonly and is primarily reserved for patients with severe, uncontrollable heart failure. Average 10-year survival rates for older children with HCM are > 90%, although infants with HCM typically do far more poorly.

## **Living with HCM**

The diagnosis of HCM affects many areas of a child's life. The following sections outline the general approaches to living with this disease. It is important that specific recommendations are developed by the team caring for the child.

### **Physical Activity**

Children with HCM are not allowed to play competitive sports because of the possibility of a sudden collapse or increased heart failure. A competitive sport is an organized team activity for which training is required.

A child with hypertrophic cardiomyopathy may be allowed to perform low-impact activities based on their clinical status, degree of potential outflow obstruction and relative risk of arrhythmias. However, specific activity recommendations should be individualized by the treating cardiologist.

### **School**

The intellectual, psychological and social benefits of attending school cannot be overestimated in the child with HCM. Adjusting medication schedules so they do not interfere with school activities, discussing safe activity levels with school personnel, and providing tutoring to maintain academic performance are important interventions that can help a child to stay in school and keep up with their peers. Often close communication between the parents, medical care team, and the school nurse can help to keep a child up to date in school.

### **Friends**

Every effort should be made to allow a child with HCM to spend time with friends. Depending on the specific recommendations of the treating cardiologist, the child may be allowed to participate in light activities when possible. However, an effort should be made to avoid contact with those who are acutely ill with fever, even though many children with this disease are able to tolerate upper respiratory tract illnesses (common colds) well.

### **Psychological Issues**

Adjusting to having a chronic illness is stressful not only for the child but for the entire family as well. The child's reaction to having HCM often depends on the stage of the child's development. Discussions about the disease should be tailored to the specific concerns of the child. Child-life professionals and pediatric psychiatrists are important resources to help children cope, and their services are often available through the treating center.

### **Family**

The impact of a diagnosis of HCM is felt throughout the child's immediate and extended family. It is important for parents and other caregivers to realize that they are not alone in feeling the weight of responsibility that comes with taking care of a child with a chronic illness. Anticipating and/or preventing the stress imposed by an illness is an important part of caring for the child and family, and personnel at the treating medical center can help identify issues that can lead to increased stress.

Practical solutions to problems giving medications, keeping track of appointments, and maintaining normal family life

can often be found through discussions with nurse clinicians, the social worker, psychiatrist, and other parents of children with hypertrophic cardiomyopathy.

## **Diet**

All children with HCM should follow a healthy diet. The recommendations published in 2005 by the United States Department of Agriculture (USDA) can be found at the following website address: <http://www.mypyramid.gov/>. Certain types of HCM may require alteration of dietary intake, and in these cases, a special diet may be developed in consultation with metabolic specialists.

## **Health Maintenance**

Routine pediatric care is important for children with HCM. Regular well child visits and standard childhood immunizations should be performed. The influenza vaccine should be administered on a yearly basis. Children under age two should receive Synagis for protection against respiratory syncytial virus (RSV).

A medical alert bracelet is an important safety measure for children with HCM. In the event of an emergency, these bracelets allow medical personnel to know details about a child's illness, especially if a family member is not available.

## **What Does the Future Hold for HCM?**

Much progress has been made in our ability to diagnose HCM in both the clinical and molecular arenas. However, much additional research is needed in this field. Areas of research to be highlighted over the next decade include: 1) better understanding of HCM as a disease process and the characteristics of the disease as they relate to outcome, which will lead to better management strategies; 2) increased clinical trials which will lead to new drug development and more effective therapies; and 3) molecular identification of novel genetic mutations as well as more precise diagnostic genetic testing/screening which will result in more accurate diagnosis.

It is the expectation of the medical community that the data derived from exploring these avenues of scientific research will translate into a clinician's ability to tailor medical therapy based on a given child's precise diagnosis. Achieving this goal over the next couple of decades will represent a large milestone in the field of pediatric cardiomyopathy research and will, hopefully, improve the ongoing care and prognosis of children afflicted with this heart muscle disease.