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 gures 2a and 2b.

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 ve per 1 million children.

“Hypertrophic” refers to an abnormal growth of muscle bers in the heart. In HCM, the thick heart muscle is stiff, making it dif cult for the heart to relax and for blood to ll the heart chambers. While the heart squeezes normally, the limited

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 identi□ed, genetic testing may become a routine part of the HCM evaluation.

Finally, because most patients with this diagnosis have inherited the disease, once con□rmed 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 scienti□c 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 speci□c 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 dif□culty 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 □ll more completely when the thick muscle in the ventricular septum narrows the out□ow of blood from the heart. These medications can cause excessive slowing of the heart rate, low blood pressure, dizziness, and in some cases, □uid retention, fatigue, impaired school performance 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

Many children with HCM can lead relatively normal lives once the diagnosis has been established and appropriate

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 in□uenza 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 □eld. 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 identi□cation 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 scienti□c research 5(.)]TJ 0 -2.