Children's Cardiomyopathy Foundation™

Frequently Asked Questions About Pediatric Cardiomyopathy

What is cardiomyopathy?
Cardiomyopathy is a chronic heart disease that involves the deterioration of the heart muscle (*myocardium*). Eventually the heart no longer pumps effectively and cannot supply the body with enough blood to function. In severe cases, cardiomyopathy can lead to heart failure or sudden death.

There are five different forms of cardiomyopathy and these include: dilated cardiomyopathy, hypertrophic cardiomyopathy, restrictive cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy and left ventricular non-compaction cardiomyopathy.

What is the difference between cardiomyopathy and congenital heart disease?
Congenital heart disease is a problem with the heart's structure and function that is present at birth. Problems include holes in the heart and misplaced, malformed and/or missing valves, vessels and heart chambers. Cardiomyopathy differs in that it is a disease involving changes to the heart muscle that affects the heart's main pumping function.

Cardiomyopathy may or may not be present at birth since it can either be inherited or acquired (e.g. myocarditis, chronic drug and alcohol exposure, cancer chemotherapy, AIDS) at any stage in life. Additionally, congenital heart defects can often be repaired through surgery, whereas for cardiomyopathy there are no surgical procedures that can "cure" or restore the heart to normal.

Who gets cardiomyopathy?
Cardiomyopathy predominantly affects adults, but in rare instances does affect infants and children. Cardiomyopathy can occur in any child regardless of age, gender, race or socio-economic background. Recent studies show that children are about 10 times more likely to develop cardiomyopathy during the first year of life than ages 2 to 18 combined. Studies show that boys and African Americans are at a slightly higher risk for certain forms of cardiomyopathy.

How common is cardiomyopathy in children?
According to the Pediatric Cardiomyopathy Registry, a nationwide patient database funded by the National Institutes of Health, one out of 100,000 children in the U.S. is diagnosed with cardiomyopathy each year. However, this incidence is a conservative number and does not include all forms of secondary cardiomyopathies such as those caused by cancer-treating drugs or by diseases affecting other parts of the body besides the heart.

How many children are affected?
While there is no published number, it has been estimated that 30,000 children in the U.S are living with some form of primary and secondary cardiomyopathy. This is comparable to the number of people affected with cystic fibrosis in the U.S. This number may be underestimated as children without obvious symptoms or those who die of sudden cardiac arrest have not been properly diagnosed and are therefore not accounted for.

What causes pediatric cardiomyopathy?
There are many causes of cardiomyopathy in children, including some that are not fully understood. Cardiomyopathy can be inherited from one or both parents, or it can be acquired from a viral infection, toxins affecting other organs or use of certain chemotherapy drugs. It can result from metabolic, mitochondrial or other multi-system disorders. Despite advances in understanding in the last decade, the exact cause remains unknown in two-thirds of cases; these are said to be *idiopathic.*

How serious is cardiomyopathy?
Cardiomyopathy is an extremely variable disease. It can present quite differently in young children, teens and adults, and even among family members. Some people live a normal life without knowing they have cardiomyopathy because they have no symptoms. In some cases symptoms remain at a steady level for years, while in others symptoms may worsen over time. For this group of patients, cardiomyopathy can be a serious, progressive disease leading to heart failure or sudden cardiac death when undetected.

What are common symptoms of cardiomyopathy?
Symptoms will vary based on the type of cardiomyopathy and the stage of the disease. Some children have few or no symptoms, while others may have severe symptoms due to the heart's inability to pump blood efficiently. Common symptoms may include shortness of breath; chest pain; fatigue; irregular heartbeats that feel rapid, pounding or fluttering; dizziness, light-headedness and fainting; swelling of the legs, ankles and feet; and abdominal bloating due to fluid buildup.

How is cardiomyopathy diagnosed?
Diagnosis is often confirmed through an echocardiogram (*echo*) and electrocardiogram (*EKG, or ECG*) performed by a pediatric cardiologist. In some cases, other non-invasive and invasive tests are necessary. Genetic testing may be recommended to verify the cause of the disease. Since pediatric cardiomyopathy is rare, it is important to locate a children's medical center with a wide range of experience in treating children with cardiomyopathy.

Why is early diagnosis critical?
Early diagnosis and treatment of the disease is essential to preventing complications and rapid progression to heart failure. Early identification of the disease and accurate diagnosis of its cause, especially when related to a systemic disorder, can prevent an acute crisis and result in a more targeted treatment.

How is pediatric cardiomyopathy treated?
The first treatment option is usually medication to improve the functioning of the heart. Children with irregular heart rhythm (*arrhythmia*) may need an implantable defibrillator to regulate their heartbeats. For children with hypertrophic cardiomyopathy, a surgical procedure called a myectomy may be necessary to relieve some symptoms. If medical management is ineffective in controlling symptoms and preventing heart failure, a heart transplant may be recommended.

Is there a cure for cardiomyopathy?
While there is no cure for the disease, the disease is treatable. Symptoms and complications can be effectively controlled by medications, surgery, implantable devices, or in severe cases, a heart transplant. Treatment is determined by the type of cardiomyopathy and how serious it is.

How will cardiomyopathy affect a child's lifestyle?
Although cardiomyopathy is a chronic condition many children with cardiomyopathy can lead a relatively normal life with few lifestyle restrictions. A diagnosis will most likely mean more frequent doctor visits for monitoring the condition and daily intake of several cardiac medication. Depending on the cause, type and stage of the disease, other modifications may involve diet, restriction from competitive and contact sports, and minor school accommodations.

Should others in the family be screened?
Cardiomyopathy often has a genetic origin, so there is a possibility that other family members may have the same genetic mutation that causes the disease. It is important to assess a family's risk and discuss family screening. The child's cardiologist and geneticist should work together to formulate a long-term screening plan and discuss genetic testing options.

Can cardiomyopathy be prevented?
There is no reliable method of preventing the disease. If a family member has the disease and a genetic mutation has been identified, other siblings or relatives can be screened for the same genetic mutation. For family planning purposes, a couple may consider pre-implantation genetic diagnosis (PGD) to identify a known genetic mutation in embryos created through in vitro fertilization (IVF) cycles.

What is the expected outcome for this disease?
While there have been improvements in diagnosing the disease, outcomes have not improved much in the past three decades. Prognosis is still unclear for many children. Pediatric cardiomyopathy is a highly variable disease with many factors involved. Depending on the cause of the disease and the stage at which the child is diagnosed, the outcome may vary considerably. Some research indicates that 33 percent will recover, 33 percent will stabilize and 33 percent will worsen.