



What is dilated cardiomyopathy?

Dilated cardiomyopathy (DCM) is the most common form of heart muscle disease affecting 50 to 60 percent of diagnosed children. "Dilated" refers to the **enlarged or stretched muscle fibers** of the heart. DCM usually starts in the left ventricle, the chamber that pumps oxygenated blood to the body. Like an overstretched rubber band, the heart muscle becomes weak and is unable to pump blood efficiently.

This forces the heart to work harder and the heart chamber to enlarge. In some cases, the heart becomes so weak that it cannot pump enough blood to meet the body's needs, limiting the amount of physical activity that can be performed. Fluid may also build up in the lungs and other organs once the disease progresses to congestive heart failure.

How many children are affected?

According to the Pediatric Cardiomyopathy Registry, DCM occurs at a rate of **6 per million children**. It is more commonly diagnosed in younger children with the average age of diagnosis at 2 years old.



Education

What is the prognosis?

In children with DCM, approximately **one-third recover**, a third stabilize and the remaining third get worse. If the cause of DCM is myocarditis, children are more likely to improve and have better outcomes than those who have DCM from other causes.

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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childrenscardiomyopathy.org

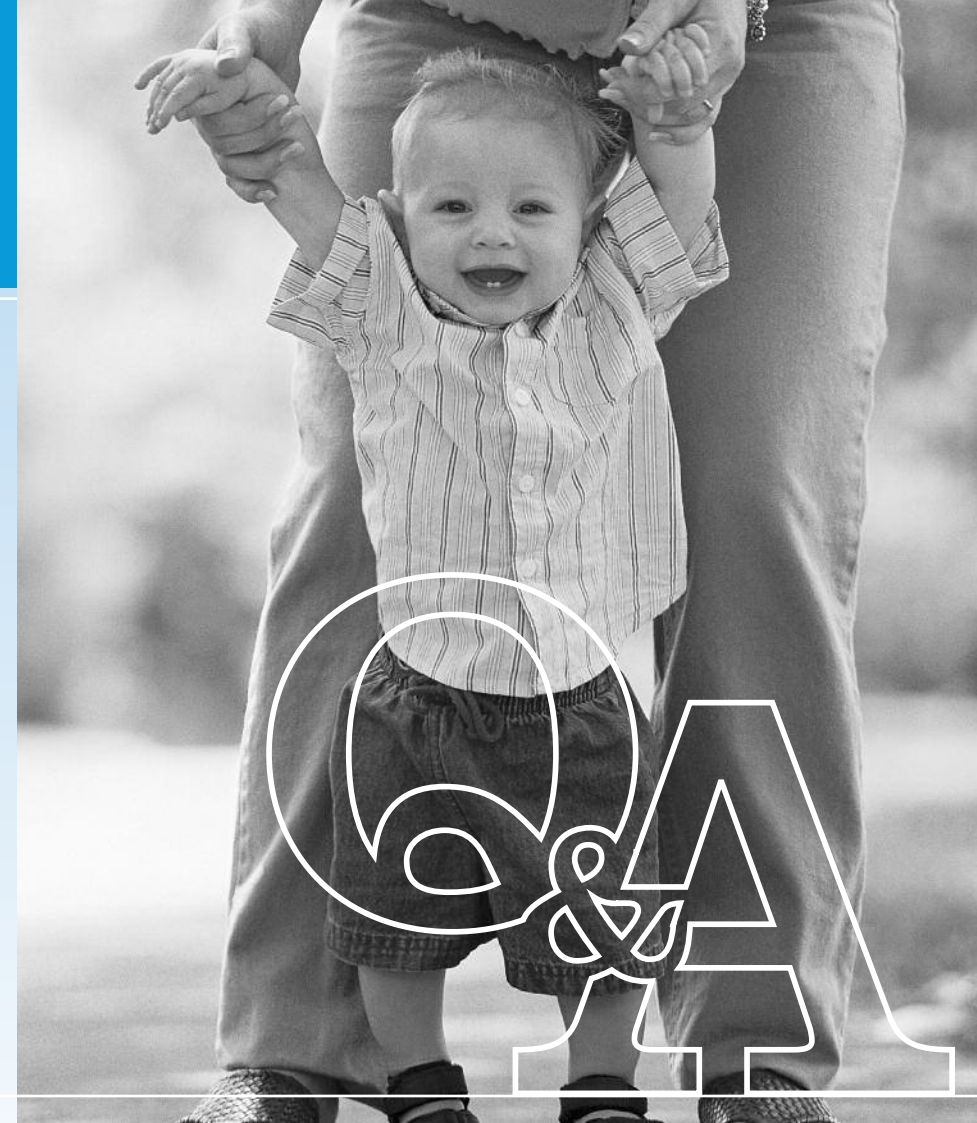


Children's Cardiomyopathy Foundation
P.O. Box 547 | Tenafly, NJ 07670
Tel: 866.808.CURE | Fax: 201.227.7016
Email: info@childrenscardiomyopathy.org

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Understanding Dilated Cardiomyopathy



What causes dilated cardiomyopathy?

Neuromuscular disorders (Duchene muscular dystrophy, Becker muscular dystrophy) and myocarditis are the most common causes of DCM in children. **Myocarditis** is a viral infection that inflames and damages the heart muscle cells. Viruses associated with myocarditis include coxsackie virus B, adenovirus, echovirus and HIV. Other causes include inborn errors of metabolism; diseases of the immune system; exposure to chemicals, toxins or powerful medications (chemotherapy drugs); excessive alcohol use; severe anemia and nutritional deficiencies.

DCM can also be 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 20 to 30 percent of children with DCM inherit the disease from their parents, even though the parent may not exhibit symptoms. To date, more than 40 genes have been identified to be associated with familial DCM. Many inherited DCM cases are caused by mutations in the cytoskeletal genes that maintain the structure of the heart muscle cells or by mutations in the sarcomeric genes

that regulate how heart muscle cells generate energy. **Familial DCM** is usually 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.

Children may have DCM associated with a **metabolic disorder** (mitochondrial defects) or a genetic malformation syndrome (Kearns-Sayre syndrome). In these cases, the contraction of the heart muscle is affected. Other gene defects can limit heart growth and lead to the abnormal stretching of muscle cells. 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. Disorders such as Barth syndrome, Duchene or Becker muscular dystrophy can be inherited in a X-linked manner in which the mother passes down the mutation.

What are the common symptoms?

A very common symptom is **difficulty breathing**. Often this is noticed with increased activity (feeding, exercise) or when there is an upper respiratory infection. In babies and young children, additional symptoms include irritability, difficulty breathing, poor appetite and slow weight gain. In older children, additional symptoms include extreme fatigue, difficulty exercising, a persistent cough, abdominal pain, nausea and vomiting. When the heart becomes weaker and progresses to congestive heart failure, the child may have pale or ashen skin; decreased urine output; excessive

sweating (diaphoresis); and swelling of the abdomen, legs and ankles.

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.

Children with DCM caused by **metabolic abnormalities** may also have low blood sugar (hypoglycemia), excessive acid in the blood (metabolic acidosis) or neurological abnormalities such as decreased muscle tone (hypotonia) and changes in mental status or behavior (encephalopathy). These symptoms may arise when the body's metabolic demand exceeds supply or when the body cannot break down accumulated toxins.

How do you diagnose dilated cardiomyopathy?

DCM is most often detected when there are signs of congestive heart failure such as excessive tiredness, fluid in the lungs (crackles), an enlarged liver, a galloping heartbeat or a heart murmur. To formally diagnose DCM, cardiologists rely on noninvasive cardiac tests such as the **echocardiogram** and **electrocardiogram** (EKG). An echocardiogram measures the size of the heart and how well the heart pumps. An ejection fraction can be calculated by measuring the percentage of blood ejected from the heart with each beat. A result of less than 40 percent usually indicates severe DCM. An EKG provides

information on the heart's electrical activity, abnormal heart rhythms and heart size.

Other tests may be ordered to assess the heart's condition and determine a treatment plan. These tests include a **chest x-ray** to check the heart's shape and size and to look for fluid in the lungs, **computed tomography** (CT scan) to observe the structure and function of the heart and blood vessels, **magnetic resonance imaging** (MRI) to evaluate heart and blood vessel function, an **exercise stress test** to see how the heart responds to exercise, and a **Holter monitor** to look for abnormal heartbeats.

In some cases, more invasive cardiac tests may be necessary. A **cardiac catheterization** is the most useful and accurate tool for diagnosing cardiovascular problems. It can be used to check for blockage in the arteries, measure heart and lung pressures, evaluate the heart's pumping ability and perform a heart (endomyocardial) biopsy. A heart biopsy involves removing a small piece of heart muscle for laboratory analysis. The tissue sample is inspected under microscope to check for signs of myocarditis, metabolic disease or structural abnormalities of the heart muscle.

What are the treatment options?

For children with DCM, medical therapy aims to control the symptoms of congestive heart failure, improve heart function and prevent complications such as blood clots or arrhythmias. With appropriate treatment, symptoms can be improved or eliminated in many children with DCM.

Several types of medications are used in children with DCM to treat heart failure symptoms and to stop the heart from deteriorating. Common **medications** include angiotensin converting enzyme (ACE) inhibitors (captopril, enalapril, lisinopril) to relax the arteries and decrease the heart's workload, diuretics (bumetanide, chlorothiazide, furosemide, spirono-lactone) to reduce excess fluid in the lungs or other organs, and digoxin to improve the heart's ability to pump blood through the body. More recently, beta-blockers (atenolol, carvedilol, metoprolol) are being recommended to reduce stress on the heart muscle. Some children with severe heart dysfunction may also require anti-coagulation medications (aspirin, dipyridamole, enoxaparin, heparin, warfarin) to prevent blood clots from forming.

For children with heart rhythm problems, anti-arrhythmic medications (amiodarone, lidocaine, procaineamide) may be prescribed to keep the heart beating at a regular rate. A **pacemaker** or **automatic implantable cardioverter defibrillator** (AICD) may be surgically implanted to control arrhythmias that do not respond to medication. Dual chamber or biventricular pacing, which involves pacing the lower heart chambers to contract together, may be suggested to help a failing heart pump more efficiently.

Close monitoring of a child with DCM is important since heart function can deteriorate rapidly. A **heart transplant** may be necessary when a child does not respond to medical treatment or shows signs of severe heart failure, poor growth or high lung pressure (pulmonary hypertension). Children with DCM are more prone to congestive heart failure and therefore have a higher rate of heart transplantation compared to other forms of cardiomyopathy. Unfortunately, a heart transplant is not an option if the DCM is caused by a disease that affects the entire body system or other organs.

