What is dilated cardiomyopathy?

Dilated cardiomyopathy (DCM) is the most common form of heart muscle disease affecting children. DCM is seen in 50 to 60 percent of children with a heart muscle disease. “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, making it hard to perform physical activity. For babies with DCM, they can be too tired to take a bottle or breastfeed. For older children, they can have a harder time exercising or have stomach pain when they eat. Fluid may also build up in the lungs making it hard to breathe or hard to lie down flat to sleep, which is called congestive heart failure.

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 (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.

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.

What is the prognosis?

In children with DCM, approximately one-third recover, one-third stabilize, and the remaining one-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. Children with myocarditis can become very sick quickly, but they can also recover quickly with medical care and treatment of the symptoms.

LOUIS

This booklet, featuring real children with cardiomyopathy, was created to provide families and caregivers with a broad overview of cardiomyopathy and is for general information only. The material presented is not intended to be complete or serve as medical advice. The information should not be a substitute for consultation with a qualified health care professional who is more familiar with individual medical conditions and needs.

The Children’s Cardiomyopathy Foundation (CCF) is dedicated to finding causes and cures for pediatric cardiomyopathy through the support of research, education, and increased awareness and advocacy.

Register with CCF for additional resources: childrenscardiomyopathy.org

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UNDERSTANDING

DILATED CARDIOMYOPATHY

BENJAMIN

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How is DCM diagnosed?

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 echocardiograms and electrocardiograms (ECG/EKG). An echocardiogram measures the size of the heart and how well the heart pumps. An ejection fraction (EF) can be calculated by measuring the percentage of blood ejected from the heart with each beat. A result of less than 30 percent usually indicates severe DCM. An ECG/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. Cardiac catheterization is the most useful and accurate tool for diagnosing cardiovascular problems. It can check for artery blockages, measure heart and lung pressures, evaluate the heart's pumping ability, and perform a heart (endomyocardial) biopsy. This biopsy removes a small piece of heart muscle for analysis under a microscope to inspect for myocarditis, metabolic disease, or structural abnormalities.

ALESSANDRA

Diagnosed with DCM shortly after her first birthday, **Alessandra** was prescribed multiple medications to treat her condition. Despite her diagnosis, she continues to thrive with her own heart and has not required any surgeries. An accomplished athlete, Alessandra competed in the Junior Olympics for water polo in 2024.

What causes DCM?

Neuromuscular disorders (Duchenne 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 parvovirus, enterovirus, 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 30 to 50 percent of children with DCM inherit the disease from their parents. This can occur even though the parent may not exhibit symptoms, and even when the abnormal gene has not been discovered. 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 the abnormal gene comes from one parent, and it only takes one abnormal gene to cause the heart muscle disease. In autosomal dominant cases, there is a 50 percent chance that the child will inherit the condition from the parent.

Children can have DCM associated with other problems such as muscle weakness throughout the body. These include metabolic disorders (mitochondria defects) or a genetic malformation syndrome (Kearns-Sayre syndrome). These tend to be rare disorders that are either passed down from the mother, called X-linked, or caused by two abnormal genes (one from each parent). In X-linked cases, male children primarily have the disease or have the most severe form of the disease. Disorders such as Barth syndrome or Duchene or Becker muscular dystrophy can be inherited in an X-linked manner, in which the mother passes down the mutation. In recessive conditions, the disease is only seen when two genes are abnormal. This is how the parents can be healthy, but they can each pass one abnormal gene to their child. There is a 25 percent chance their child will have two abnormal genes which causes the condition. If a genetic cause is found, ask your child's doctor whether close family members (like parents or siblings) should also be tested.

How many children are affected?

According to the CCF-supported Pediatric Cardiomyopathy Registry (PCMR), DCM occurs at a rate of 6 per 1-million children. It is more commonly diagnosed in younger children with the average age of diagnosis at 2 years old.

What are 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 (thromboses) 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, spironolactone) 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 also used to reduce stress on the heart muscle. Some children with severe heart dysfunction may also require anticoagulation medications (aspirin, dipyridamole, enoxaparin, heparin, warfarin) to prevent blood clots from forming.

For children with heart rhythm problems, antiarrhythmic medications (amiodarone, lidocaine, procainamide, sotalol) 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. In some patients, a cardiac resynchronization therapy (CRT) device, which involves implanting a combination pacemaker and ICD to coordinate the heart's pumping action and improve blood flow to the body, may be recommended.

Close monitoring of a child with DCM is important since heart function can deteriorate rapidly. In severe cases, a machine to help the heart rest and to move blood through the body may be needed. This is called a ventricular assist device (VAD). 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.

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