

# DILATED CARDIOMYOPATHY

Dilated or congestive cardiomyopathy (DCM) is diagnosed when the heart is enlarged (dilated) and the pumping chambers contract poorly (usually left side worse than right). A diagram and echocardiogram comparing a normal heart and a heart with DCM are shown in figure 1a and figure 1b.

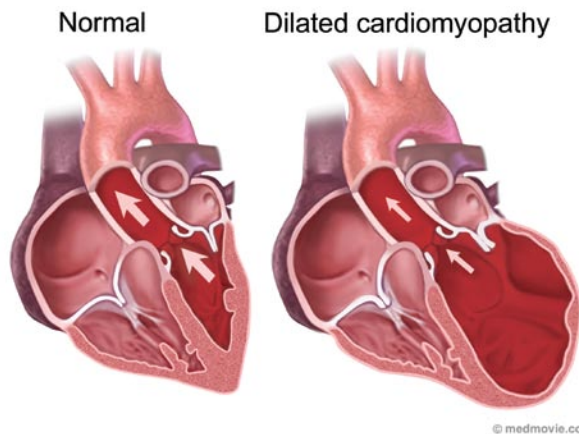


Figure 1a- A normal heart is shown on the left compared to a heart with dilated cardiomyopathy on the right. Note the increased dimensions of the left ventricle.

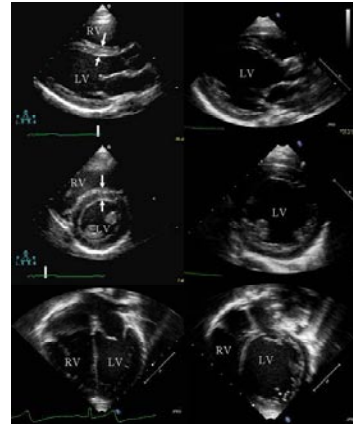


Figure 1b- Multiple echocardiographic views of a normal heart on the left and a heart with dilated cardiomyopathy on the right. Note the increased dimensions of the left ventricle with the thin walls of the left ventricle (LV).

This condition is the most common form of cardiomyopathy and accounts for approximately 55–60% of all childhood cardiomyopathies. According to the pediatric cardiomyopathy registry database, this form of myopathy is detected in roughly one per 200,000 children with roughly one new case per 160,000 children reported each year in the United States. It can have both genetic and infectious/environmental causes.

It is more commonly diagnosed in younger children with the average age at diagnosis being 2 years. Dilated cardiomyopathy can be familial (genetic), and it is estimated that 20–30% of children with DCM have a relative with the disease, although they may not have been diagnosed or have symptoms.

## Signs and symptoms of DCM

Dilated cardiomyopathy can appear along a spectrum of no symptoms, subtle symptoms or, in the more severe cases, congestive heart failure (CHF), which occurs when the heart is unable to pump blood well enough to meet the body tissue needs for oxygen and nutrients.

When only subtle symptoms exist, infants and young children are sometimes diagnosed with a viral upper respiratory tract infection or recurrent “pneumonia” without realizing that a heart problem is the basis for these symptoms. Older children and adolescents are less likely to be diagnosed with viral syndromes and more likely to present with decreased exercise capacity or easy fatigability.

With CHF, babies and young children will usually have more noticeable clinical changes such as irritability, failure to thrive (poor gain weight), increased sweating especially with activities, pale color, faster breathing and/or wheezing. In older children, congestive heart failure can manifest as difficulty breathing and/or coughing, pale color, decreased urine output and swelling, excessive sweating, and fatigue with minimal activities. Until the diagnosis is made in many children, chronic coughing and wheezing, particularly during activities, can be misinterpreted as asthma.

Some patients with DCM caused by viral myocarditis (weakened, enlarged heart muscle usually due to a viral infection) can have a rapid increase in the number and severity of CHF symptoms such that within 24–48 hours the child can become very ill requiring emergency hospitalization, and occasionally, advanced life support.

Symptoms due to heart rhythm problems (or arrhythmias, which means irregular, fast or slow heart rates) can also be either the first symptom or a symptom that appears after other symptoms have led to a diagnosis of DCM. Symptoms of rhythm problems include palpitations (feeling of funny or fast heart beats), syncope (fainting), seizures (convulsions), or even sudden cardiac arrest (heart stops beating effectively requiring resuscitation). These symptoms can occur at any age and with any stage of cardiomyopathy, even if other more severe symptoms of congestive heart failure have not yet appeared.

## Diagnosis of DCM

Once there is clinical suspicion based on the patient history and physical exam, the diagnosis of DCM is primarily based on echocardiography. With this test, your physician will be using ultrasound beams to evaluate the heart looking for dilated chambers and decreased pump function. Along with the echocardiogram, there are other tests that will likely be done to confirm the diagnosis or provide clues as to the cause.

A chest X-ray will show the heart size and can be used as a reference to follow increases in heart size that may occur over time. An electrocardiogram, or EKG, records the electrical conduction through the heart and is used to look for evidence of thickened or enlarged chambers as well as abnormal heart beats that can occur in children with this diagnosis. To more completely evaluate for the presence or absence of these abnormal heart rhythms, which may effect treatment, your doctor may also order a Holter monitor which records heart beats over a 24–48 hour period. A treadmill test can also be useful in some children (beyond age 5–7 years) who can cooperate with this study. This exercise test is used to assess the energy reserve of your child’s heart and, in cases when children do not respond adequately to medicines, this test may also help predict the need for heart transplantation.

Depending on your child’s age, a battery of blood tests may be done in order to identify treatable causes for the cardiomyopathy. This may include testing for certain viral infections such as adenovirus and the Coxsackie viruses as they have been associated with DCM especially in younger children. In many cases, no cause is discovered, and the cardiomyopathy may be referred to as “idiopathic” (cause unknown). Many heart failure specialists believe this “idiopathic” form of the cardiomyopathy is genetic. While genetic screening has not yet become a standard procedure, some physicians may send blood to molecular testing labs located in a few centers around the country so that limited genetic testing can be performed looking for possible mutations currently known to cause dilated cardiomyopathy. If your physicians believe the cause is genetic (especially common in older children and adolescents), evaluation, usually with echo, of other family members is recommended to rule out presence of this disease in other close relatives (parents, siblings).

Finally, in more advanced cases of DCM, cardiac catheterization may be performed. During this procedure, a catheter (thin plastic tube) will be slowly advanced through an artery or vein into the heart (while watching its course on a TV monitor) so that pressures within the heart chambers can be measured. A cardiac biopsy, which involves removing tiny pieces of heart muscle for inspection under the microscope, may be performed to help distinguish between infectious and genetic causes. The information provided during the catheterization may also be helpful if transplantation is being considered as one of the treatment options for your child.



## Causes of DCM

While other causes of DCM have been identified and are briefly discussed in other sections of this brochure, mounting scientific evidence is beginning to suggest that individual genetic “mutations” may be among the more common causes of DCM in children. For a greater understanding of the basics of human inheritance patterns and a more detailed discussion of the potential genetic causes of DCM, the reader is encouraged to read separate sections entitled “Overview of Inheritance” and “Genetics of Cardiomyopathies” printed elsewhere in this brochure.

## Current Treatment

Currently, there are no therapies that can “cure” DCM; however, many treatments are available that can improve symptoms and decrease risk in children with DCM. 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, current medical and non-medical therapies for DCM are summarized.

## Medical Therapy

The majority of children with DCM have signs and symptoms of heart failure. The most common types of medications used to treat heart failure include diuretics, inotropic agents, afterload reducing agents and beta-blockers.

**Diuretics**, sometimes called “water pills,” reduce excess fluid in the lungs or other organs by increasing urine production. The loss of excess fluid reduces the workload of the heart, reduces swelling and helps children breathe more easily. Diuretics can be given either orally or intravenously. Common diuretics include furosemide, spironolactone, bumetanide and metolazone. Common side effects of diuretics include dehydration and abnormalities in the blood chemistries (particularly potassium loss).

**Inotropic Agents** are used to help the heart contract more effectively. Inotropic medications are most commonly used intravenously to support children who have severe heart failure and are not stable enough to be home. Common types of inotropic medications include:

- Digoxin (taken by mouth): improves the contraction of the heart. Side effects include low heart rate, and, with high blood levels, vomiting and abnormal heart rhythm.
- Dobutamine, dopamine, epinephrine, norepinephrine (intravenous medications given in the hospital): medications that increase blood pressure and the strength of heart contractions. Side effects include increased heart rate, arrhythmias and for some, constriction of the arteries.
- Vasopressin (intravenous medication): increases blood pressure and improves blood flow to the kidneys. Side effects include excessive constriction of the arteries and low sodium.
- Milrinone (intravenous medication): improves heart contraction and decreases the work of the heart by relaxing the arteries. Side effects include low blood pressure, arrhythmias and headaches.

**Afterload Reducing Agents** reduce the work of the heart by relaxing the arteries and allowing the blood to flow more easily to the body. Common afterload reducing medications include:

- Angiotensin converting enzyme inhibitors (ACE inhibitors): captopril, enalapril, lisinopril, monoproil (taken by mouth). Side effects include low blood pressure, low white blood cell count, high potassium levels and kidney or liver abnormalities.
- Angiotensin I Blocker: Losartan (taken by mouth). Side effects include diarrhea, muscle cramps and dizziness.
- Milrinone is an inotropic agent (see above) that also relaxes the arteries.

**Beta-blockers** slow the heartbeat and reduce the work needed for contraction of the heart muscle. Slowing down the heart rate can help to keep a weakened heart from overworking. In some cases, beta-blockers allow an enlarged heart to become more normal in size. Common beta-blockers (taken by mouth) include carvedilol, metoprolol, propranolol and atenolol. Side effects include dizziness, low heart rate, low blood pressure, and, in some cases, fluid retention, fatigue, impaired school performance and depression.

In addition to improving the symptoms of heart failure, ACE inhibitors and beta-blockers have been shown to return the heart size toward normal and lessen the number of deaths and hospitalizations in adult patients with dilated cardiomyopathy without symptoms. An ACE inhibitor is recommended in children with dilated cardiomyopathy even in the absence of symptoms. Currently, no firm recommendations are available for beta-blockers in children.

## **Anticoagulation Medications**

In children with a heart that does not contract well, there is a risk of blood clots forming inside the heart possibly leading to a stroke. Anticoagulation medications, also known as blood thinners are often used in these situations. The choice of anticoagulation drug depends on how likely it is that a blood clot will form. Less strong anticoagulation medications include aspirin and dipyridamole. Stronger anticoagulation drugs are warfarin, heparin, and enoxaparin; these drugs require careful monitoring with regular blood testing. While variable, common side effects of anti-coagulants include excessive bruising or bleeding from otherwise minor skin injuries, interaction with other medications and, for warfarin, fluctuations in anticoagulation blood levels caused by changes in daily dietary intake. Information regarding which food groups can significantly affect warfarin levels can be obtained from your cardiologist.

## **Anti-arrhythmia Medications**

In some DCM patients, especially those with very dilated and poorly contractile ventricles, there may be a higher risk of an abnormal, life-threatening heart rhythm (ventricular tachycardia), and medications are used to prevent or control this abnormal rhythm which then keeps the heart beating in a regular pattern. Common anti-arrhythmia medications include: amiodarone, procainamide and lidocaine. General side effects may include slower heart rate, lower blood pressure, GI upset (nausea/constipation), headache, depressed mood, difficulties concentrating, dizziness, and skin rash among others. Consult your cardiologist for drug-specific side effects once a particular anti-arrhythmic medicine has been prescribed.

## **Pacing Therapies for DCM**

### **Pacemakers**

Pacemakers are small, battery-operated devices that are placed under the skin of the chest or abdomen and attached to electrical wires (leads) which are threaded to the heart. Depending on which type of pacemaker is used, these leads are attached to muscle tissue either on the inside or outside of the heart. The devices monitor the heartbeat and help maintain a regular rhythm in children who are prone to have abnormal heartbeats.

In some patients with DCM, the heart rate can become too slow either due to abnormally slow conduction of impulses through the heart or as a side effect of medications. In those instances, a “back-up” pacemaker can be implanted to help maintain an appropriate heart rate.

Conversely, as mentioned above, patients with DCM can develop abnormally fast, life-threatening arrhythmias (ventricular tachycardia and/or fibrillation). Implantable cardioverter defibrillators (ICDs) can be used in those children to convert these arrhythmias to a normal rhythm.

Finally, a bi-ventricular pacing system has recently become part of the standard therapy in adults with end-stage heart failure associated with DCM, and this treatment modality is currently being evaluated in children.

### **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 and in children with dilated cardiomyopathy who have serious ventricular arrhythmias.

## **Biventricular Pacemaker:**

In recent promising studies in adult patients with dilated cardiomyopathy, a special pacemaker that can pace both the right and left ventricles has been designed. This system, which has to be specially timed, allows the two ventricles to contract together and improves the synchrony of contraction between the walls of the left ventricle. Study results have shown that, when added to other medical treatments, this mode of pacing has helped some patients live longer with fewer hospitalizations and, in some cases, has decreased the need for transplantation.

At the time of this publication, use of bi-ventricular pacing in children is in the early stages of development. It is not yet known which children may benefit most from this form of pacing or the best way to implant the system. Nonetheless, this approach is considered an adjunctive therapy to medicines that patients are already receiving and should not be thought of as a medicine replacement. Over the next few years, study results in children in whom bi-ventricular pacing is being used may help cardiologists better understand which patients may benefit most from this therapy and under what circumstances.

## **Surgical Options for DCM**

No surgery has been effective in improving the heart function in dilated cardiomyopathy. In a few patients with a severely dilated left ventricle and a very leaky (regurgitant) mitral valve, surgery to repair or replace the mitral valve may help the heart function improve temporarily. Heart transplantation is the only effective surgery offered for patients with DCM who have severe heart failure that does not respond to medications or other treatments.

## **Cardiac Assist Devices (Mechanical Hearts)**

Cardiac assist devices are machines that do the work of the heart using a mechanical pump to deliver blood to the body. Cardiac assist devices are implanted when all other therapies have failed and the heart failure is severe. They improve blood flow to the body and allow other organs to recover from the stress of heart failure. These devices are typically used as bridges to transplantation. That is, they are used to support a child either until the heart function has recovered enough to effectively circulate blood through the body or until a suitable donor organ can be found. In pediatric patients, they are designed as a temporary means of support and cannot be used as a permanent alternative to the child's own heart or a transplanted heart. Potential complications of cardiac assist devices include infection, blood clots, stroke and mechanical problems with the devices themselves. There are a variety of cardiac assist devices available. The type of cardiac assist device that is used for an individual child depends on body size and also the type of assist support needed.

## **Heart Transplantation**

Dilated cardiomyopathy is one of the leading reasons for heart transplantation in children. Heart transplant is only considered in children who have such serious heart disease that there are no other medications or support devices available to sustain the child. A heart transplant offers the child with DCM 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 DCM are able to 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 DCM should be watched closely for the development of heart failure, abnormal heart rhythms, blood clots inside the heart and other signs that the heart is not able to maintain normal blood flow. Once signs of heart failure or significantly abnormal rhythms appear, aggressive therapy is begun with close monitoring for signs of worsening blood flow to the vital organs. If other organs show signs of damage or the heart failure does not respond to medications, a cardiac assist device and/or transplant is considered early, as the waiting time for a donor heart can be unpredictable. It is important that a child with cardiomyopathy be closely monitored at a center with expertise in pediatric heart failure, arrhythmias, cardiac assist devices and transplantation in order to ensure rapid response to any worsening of his or her condition.

In children with DCM, approximately 35% recover completely, 35% stabilize and the remaining may progressively worsen. Children with DCM are more prone to congestive heart failure and have a higher rate of heart transplantation compared with other forms of cardiomyopathy. However, improved medical therapy may eventually change this scenario. Current average five-year survival rates for children with DCM are 40–50%. If the cause of DCM is myocarditis, children are more likely to improve and have a better outcome than children with other causes of DCM.

## **Living with DCM**

The diagnosis of DCM 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 DCM 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 dilated cardiomyopathy and no heart failure symptoms can be allowed to perform recreational athletic activities, also known as low-dynamic or low-static sports, in a non-competitive situation. In milder cases, the child may even participate in gym class after careful discussions between the child, teacher, and parents to ensure that the level of activity does not cause overexertion (breathlessness, chest pain, excessive sweating). 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 DCM. 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 DCM to spend time with friends. The child should also be allowed to participate in recreational activities whenever 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 DCM 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 DCM 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 dilated cardiomyopathy.

## **Diet**

All children with DCM 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 DCM are associated with an inability to digest certain types of food, and in these cases, a special diet is developed in consultation with metabolic specialists. In children with dilated cardiomyopathy and heart failure, a low salt diet is recommended to avoid fluid retention.

Some children with heart failure may not grow well. In these cases, a diet that increases calories is recommended. Children who are taking some medications may have low levels of magnesium or potassium and a diet that has a higher amount of one or both of these two electrolytes may be recommended. Some children with severe heart failure can retain extra body fluid, and it may be necessary to limit the amount that a child can drink to prevent fluid from accumulating in the lungs.

## **Health Maintenance**

Routine pediatric care is important for children with DCM. Regular well child visits and standard childhood immunizations should be performed. The influenza vaccine should be administered on a yearly basis. Children under age 2 should receive Synagis for protection against respiratory syncytial virus (RSV).

A medical alert bracelet is an important safety measure for children with DCM. 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 DCM?**

Much progress has been made in our ability to diagnose DCM 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 DCM 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.