Dilated Cardiomyopathy (DCM)

Definition
Cardiomyopathy is a disease that affects the heart muscle. In dilated cardiomyopathy (DCM) the heart muscle becomes thin, the left ventricle (lower left chamber of the heart) becomes enlarged (dilated) and the heart is unable to squeeze efficiently, reducing the amount of blood that is pumped to the body.
Symptoms
Many individuals with DCM experience no symptoms. For those who have heart failure, symptoms can include:

Older children and adults
- Shortness of breath and fatigue
- Feeling dizzy or light-headed
- Fainting or passing out (syncope)
- Persistent cough
- Feeling abnormal heart beats (pulsitations)
- Unexplained weight gain or swelling

Newborns and babies
- Difficulty feeding or poor growth
- Excessive sweating with feeds or activity
- Excessive fussiness
- Fast breathing

Causes
There are many causes of DCM. Around 20 to 50 percent of individuals with DCM have a familial form. For these individuals, an underlying genetic cause results in a condition that runs in a family and affects only the heart. It is possible that parents, siblings, and even aunts/uncles or cousins of an individual with DCM may also have a heart muscle condition.

In children, the most common acquired cause of DCM is myocarditis. Adults can have DCM as a result of myocarditis as well. Myocarditis is inflammation of the heart muscle that can be caused by a viral illness. These viruses are the same illnesses that may cause a common cold but in some individuals can lead to a heart muscle problem. Other causes of DCM include chemotherapy exposure and metabolic diseases. In many cases, the cause of DCM in an individual cannot be identified. For these individuals, their heart muscle disease is called idiopathic DCM, which means DCM with an unknown cause.

Diagnosis
Diagnosis of DCM is based on cardiac testing, family history, medical history, and a physical exam.

- **Cardiac testing:** An echocardiogram is the most common test used to diagnose DCM, as the dilation of the ventricle and overall squeeze of the heart can be measured with this test. Other tests may include blood tests, electrocardiogram (EKG), exercise stress echo test, cardiac catheterization and magnetic resonance imaging (MRI).

- **Family history:** A family tree should be constructed with specific attention to a history of cardiomyopathy, rhythm problems, sudden cardiac or unexplained death, cardiac surgery or presence of other cardiac disease in relatives.

- **Medical history:** A history of heart failure symptoms including feeding issues, heart rhythm problems, and exercise intolerance.

- **Physical exam:** A thorough physical exam will be done to evaluate for signs of a cardiac problem. Assessment of skeletal muscle strength will be performed since problems with skeletal muscle may occur with cardiac muscle disease.

Occasionally, a myocardial biopsy may be performed in the cardiac catheterization laboratory to help determine the cause of the cardiomyopathy. A tiny instrument will be inserted into the heart via a vessel in the leg and a tissue sample will be taken from the heart, examined under a microscope and tested for viruses to help to determine the cause of the cardiomyopathy.

Management and Treatment
The treatment for individuals with DCM is different for each person. Treatment is aimed at the cause of the heart failure if known. The primary goal is to improve cardiac function and prevent symptoms. Individuals with DCM need to be followed by a cardiologist on a regular basis. Medications are used to treat symptoms, reverse heart damage and to prevent further damage to the heart muscle. Medications such as beta-blockers and ACE inhibitors will be used in most cases. Other medications may be prescribed to manage symptoms or to prevent abnormal heart rhythms.

Lifestyle changes are recommended for some individuals. Exercise is generally important, however, individuals with DCM may experience fatigue and shortness of breath with exercise. It’s important for these individuals to limit their activity if they become tired, dizzy or have difficulty breathing. In some cases, exercise is not recommended and participation in competitive sports may be discouraged.

Some people with DCM are at increased risk for sudden cardiac arrest, which results from a sudden loss of heart function caused by a fast heart rhythm called ventricular tachycardia. Unless emergency treatments are initiated immediately, including CPR and defibrillation, sudden cardiac death can occur. For individuals at risk for sudden cardiac arrest, an implantable cardioverter-defibrillator (ICD) may be recommended. In some patients with DCM, a pacemaker (cardiac resynchronization therapy) may be combined with the ICD. Mechanical pacing from different areas of the heart may improve the heart’s ability to supply blood to the body.

If cardiac function continues to worsen despite medications, a mechanical device may be necessary to help the heart pump and supply blood to the body. The device that would be used for young children is called the Berlin heart device. For older children and adults there are multiple devices that could be surgically placed to provide cardiac support.

In some cases, if the condition is severe, a heart transplant may be considered.
Genetic Testing

Genes are packages of genetic information that tell our bodies how to develop and function. DCM can be caused by a change or mutation in a gene that encodes proteins important for the contraction or squeeze of the heart muscle. Mutations in these genes result in familial DCM, which is inherited in an autosomal dominant manner. A parent that carries a gene mutation will have a 50 percent chance of passing it on to each child.

Genetic testing is available to look for mutations in the genes known to cause DCM. This type of testing is done by a blood sample. It is best to begin genetic testing in a family with an individual who is known to have DCM. If a gene mutation is found in someone with DCM, genetic testing for the same mutation can be offered to close family members. Finding a gene mutation in other family members will help identify family members who may be at risk to develop heart muscle disease and who should have cardiac screening.

A genetic counselor and/or geneticist can assess the family history and provide information about the chance that there is a genetic predisposition to cardiomyopathy. A genetics professional can also explain and facilitate genetic testing and interpret results for patients and families.

Screening for Family Members

All first degree relatives of an individual who has familial or idiopathic DCM should undergo routine cardiac evaluation. This includes parents, brothers, sisters, and children. If a genetic mutation causing DCM in an individual is known, screening is recommended for those family members proven to also carry the gene mutation. Routine cardiac screening would not be recommended for family members who did not inherit the gene mutation.

The timing of routine cardiac screening for family members should be discussed with a cardiologist and genetics professional who have experience in caring for individuals with cardiomyopathy. How frequently an individual should be evaluated is based on published guidelines and the individual’s age, personal medical and family history.