# Patient Information

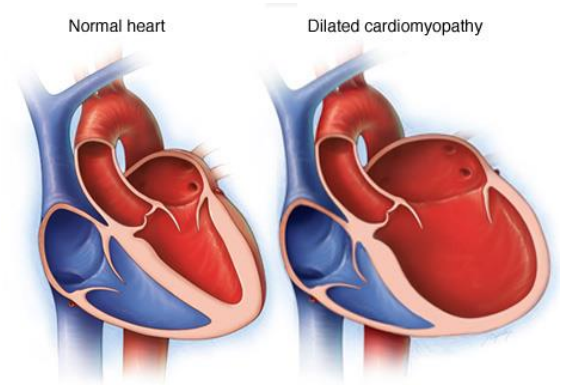
## Dilated Cardiomyopathy

### 1. The Normal Heart

The heart is a special muscle that contracts regularly and continuously, pumping blood to the body and the lungs. It has four chambers – two at the top (the atria) and two at the bottom (the ventricles). The pumping action of the heart is caused by a flow of electrical signals through the heart. These electrical signals repeat themselves in a cycle and each cycle causes one heartbeat.

### 2. Dilated Cardiomyopathy

Dilated cardiomyopathy (DCM) is a disease of the heart muscle. Having DCM means that the left ventricle (or sometimes the both ventricles) of the heart becomes dilated (enlarged). When this happens, the heart can no longer pump blood efficiently around the body and the lungs. This can lead to fluid building up in the lungs, ankles, abdomen and other organs and a feeling of being breathless. This collection of symptoms is known as heart failure. In most cases, DCM develops slowly, so the heart can be quite severely affected before a patient is diagnosed. In some cases, there may also be mitral regurgitation. This is when some of the blood flows in the wrong direction through the mitral valve, i.e. from the left ventricle to the left atrium.



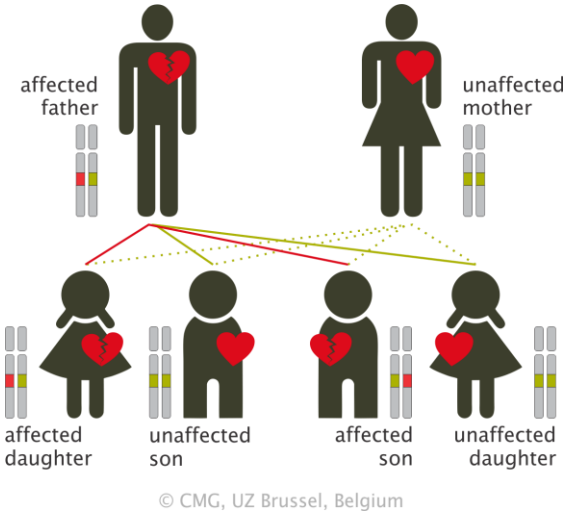

*Source: with permission from Mayo Clinics*

### 3. Prevalence & Inheritance

It is unclear how many persons are affected by DCM; i.e. the *prevalence* of DCM is unknwon. The majority of patients with DCM have a non-genetic cause like coronary artery disease, hypertension, viral infections, auto-immune disease, exposure to toxins (e.g. alcohol or certain drugs) or pregnancy. It is unknown to what extent cardiomyopathies due to non-genetic causes develop under the influence of a genetic predisposition.

In about 30% (1 in 3) of DCM cases there is a clear family history for DCM and a genetic cause. This means that in these cases DCM is caused by a defect ( a mutation) in one or more genes that can be passed on through families.

A gene is part of our DNA which contains a code for making a molecule (a protein). DCM is caused by a mutation in the genes that contain codes for specific molecules (proteins) in the heart. Every person has two copies of each gene that can be linked to DCM. A mutation in only one of the two copies of one of these genes (from the father or from the mother) is enough to develop DCM. This is called an autosomal dominant disorder and a parent who carries it has a 50% (1 in 2) chance of passing the mutation to each child. The chance that a child will not inherit the mutated gene is also 50 percent.




*Autosomal dominant inheritance*

In some cases, a new (de novo) mutation can occur in the egg or sperm cells or in an embryo. In these cases, the child's parents do not have the mutation and DCM, but the child does have DCM and can pass the mutated gene to his or her own children.

## 4. Symptoms

Persons who are affected with DCM may remain well (without symptoms). Some patients will have a few symptoms and others may develop problems which need complex treatment.
If patients with DCM suffer from symptoms, these symptoms can often be controlled with medication or other therapies. The symptoms of DCM are similar to those of heart failure. Heart failure is a term used to describe a group of symptoms that can occur when the heart muscle becomes less efficient at pumping blood around the body. The symptoms of heart failure include: shortness of breath, swelling of the feet, ankles, abdomen and of the lower back, tiredness, and also palpitations due to arrhythmias (an abnormal heart rhythm). Some arrhythmias can even lead to sudden death; this is especially the case for patients in whom the pumping ability of the heart muscle is severely decreased.

## 5. Diagnosis

The most common tools to make the diagnosis of DCM are the medical and family history, physical examination, a heart electrical tracing (the electrocardiogram or ECG), a heart ultrasound scan (echocardiogram), exercise testing, heart rhythm monitor (Holter), a heart magnetic scan (magnetic resonance imaging or MRI), and blood testing (e.g. renal function, sodium potassium, and NT-proBNP which is a protein that is released into the blood by the heart when heart failure occurs).

### 5.1. ECG (electrocardiogram)

This is the most basic test. Small sticky patches (electrodes) are put onto the chest and sometimes to arms and legs. These are connected by wires to an ECG recording machine, which picks up the electrical activity for a few seconds that makes the heartbeat. Sometimes additional or repeated ECG-tests are necessary.

### 5.2. Echocardiogram (echo)

Echocardiogram uses ultrasound waves to look at the structures of the heart. An echocardiogram can detect different types of structural changes in the heart, for example heart muscle diseases like DCM and heart valve abnormalities. Areas of thinning of the heart muscle can be identified.

### 5.3. Exercise test (stress test)

Exercise test is the same as the ECG described above, but is recorded before, during and after exercising on a treadmill or an exercise bike.This records any changes in the electrical patterns that occur with exercise.

### 5.4. MRI

An MRI scan uses a magnetic field to create images of the heart. The scanner itself is a large tube with a table in the middle, allowing the patient to slide into the tunnel. The test takes about one hour. An MRI is very good at showing the structure of the heart and blood vessels, showing the condition of the heart muscle, and identifying any scarring (fibrosis) within the heart.

### 5.5. Electrophysiology study (EPS)

This test involves having a long tube called a catheter inserted into a blood vessel and fed up to the heart. Electrical signals are sent through the catheter to the heart which makes it beat at different rates, which is recorded. This can be used to find where in the heart arrhythmias are starting (and can be used to identify treatment options).

### 5.6. Genetic testing

In about 30-40% of the DCM families, a mutation can be found in one of the genes that can cause DCM. Because not all genes that can cause DCM are known, a negative result of the genetic testing (i.e. when no mutation is found) does not rule out an inherited cause of the DCM.

## 6. Therapy

Although there is no cure for DCM, treatments help to control symptoms, and to decrease the long-term risk. Treatment depends on symptoms and (if available) the specific gene-mutation. If patients are at high risk of sudden death (for example after a previous cardiac arrest) or if symptoms cannot be controlled by medication, implantation of an internal cardiac defibrillator (ICD) can be considered. An ICD can correct most life-threatening arrhythmias.

## 7. Lifestyle & Sports

There are key recommendations for patients (and families) that are diagnosed with DCM, to prevent them from arrhythmias:

- avoidance of strenuous exercise - especially intense, competitive sport and heavy weight lifting;
- regular check-ups to monitor any changes in the disease;
- encouragement of relatives to be screened.

The diagnosis of DCM and the ability to pass on the condition can lead to anxiety and many other questions. Medical social workers or psychologists have experience with this and may be helpful for the patient and the family members.

## 8. Follow-up

The heart doctor (cardiologist) will advise on how often follow-up is needed depending on symptoms, age, and treatment.

## 9. Family Screening

If a mutation in a gene is found in a patient with DCM (*see genetic testing*), family members of this patient (to start with the first-degree family members: mother, father, brothers, sisters, and children) can have genetic testing at a specialist genetic heart clinic. Family members in whom the same mutation (familial) is found, are called mutation carriers and will be followed by a cardiologist. Family members in whom the familial mutation is not found can be reassured. If there is not a mutation identified in a patient with DCM, family members of this patient (to start with the first-degree family members) are advised to visit a cardiologist for heart tests.
DCM usually develops after puberty, as children grow into adults. However, some children have symptoms of the disease at earlier age. Therefore the recommendation for children to be examined is from the age of ten years.

## 10. DCM and Pregnancy

Before becoming pregnant it is important to discuss any potential risks, any medication changes and care during the pregnancy.

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