



Cardiac Inherited Disease Registry N.Z.

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Familial Dilated Cardiomyopathy (DCM) Information for families

1) What is Dilated Cardiomyopathy (DCM)?

Dilated Cardiomyopathy (DCM) is a medical term used to describe a condition in which the heart becomes enlarged beyond normal limits and therefore cannot perform as effectively as a pump. The word 'cardio' simply means 'the heart', 'myo' means 'muscle', and 'pathy' means a disease state. The word 'dilated' in this context means abnormally enlarged. Taken together these terms mean that as the heart muscle becomes enlarged it progressively weakens as a pump.

Dilated Cardiomyopathy in New Zealand is commonly caused by coronary heart disease, (Coronary disease is caused by a build up of fatty plaque in the coronary arteries, the arteries that feed the heart muscle its oxygen and nutrients). There are other known causes of DCM including alcohol induced forms, and other idiopathic forms that may come on after a viral illness or pregnancy. About 25% of cases were thought to be 'idiopathic' that is, no identifiable cause could be found. In recent years doctors have found that approximately 30% of affected individuals seem to have other family members who are also affected by the disease.

Inherited diseases are those that are carried in a person's genes (the blueprint we are born with that is derived from our parent's genes). Since genes are passed down through generations by parents to children, the genes (research has shown that there may be more than one gene involved), responsible for the familial form of dilated cardiomyopathy are also passed down through a family. A diagnosis of Familial Dilated Cardiomyopathy can be made when idiopathic Dilated Cardiomyopathy is found in two or more blood relatives in the same family.

Familial dilated Cardiomyopathy may be inherited from one or rarely from both parents. This means that every child born to a parent who carries the genes responsible for Familial Dilated Cardiomyopathy will have a 50% chance of inheriting the disease, although the severity of the condition may vary.

2) What are the symptoms of Dilated Cardiomyopathy?

Part of the reason why it has taken some time to identify that Dilated Cardiomyopathy can be inherited, is that the age of onset and the symptoms may be very different in two related family members. Doctors are still unsure why the disease is mild in one person and severe in another. For example one person may die suddenly, early in life whilst another family member may have few symptoms until late in life. There is no way to predict which family members are at greater risk, or how, when or in whom symptoms will arise.

As the disease worsens the heart becomes weaker so that physical activities become harder to do. Sufferers' of familial DCM may have great difficulty doing any exercise or effort related activity. Symptoms of dilated cardiomyopathy range from mild to severe, and include shortness of breath, rapid and or irregular heart rhythms (often described as palpitations), chest discomfort and extreme fatigue. Together these symptoms' may be collectively referred to as 'heart failure'.

In addition to heart failure symptoms, patients with Dilated Cardiomyopathy may also suffer from heart rhythm disorders, commonly called "arrhythmias". Arrhythmias are not the normal heart rhythm they are abnormal heartbeats (which may arise from various areas inside the heart, not however from the normal electrical conduction system that keeps the heart in a normal rhythm).

Arrhythmia's are caused by the abnormal changes that are occurring at a cellular level inside the heart, due to muscular degeneration, and a lack of vital of blood supply to some areas' of the heart especially during activity. Heart rhythm problems can cause additional stress on an enlarged heart with a reduced blood supply. Heart rhythm disturbances may cause fast heartbeats or slow heart beats. Fast heart rhythms can be particularly dangerous and cause blackouts or collapse. Slow heart rhythms can also cause blackouts, as well as tiredness and dizziness. Symptoms of arrhythmia can be, rapid or forceful heart beat, a fluttering feeling in the chest, irregular heart beats, feeling of missed beats, and collapse, or dizziness, shortness of breath, chest pain, or sudden collapse requiring resuscitation (called sudden cardiac death).

3) What Tests are available to diagnose Familial Dilated Cardiomyopathy?

Screening tests are recommended for first degree family members, parents, brothers/sisters and children. Timing of screening tests in small children is still under discussion, however usually a baseline echocardiogram is obtained and routine surveillance is maintained every 3-5 years throughout adulthood.

Diagnostic testing includes a physical examination, an ECG (electrocardiograph) recording of your heart activity, and an Echocardiogram (a non invasive test, which uses an ultrasound beam to view the heart beating and is able to be used to measure the size of the heart and its chambers and valves). Some further tests such as Exercise tests, Holter monitors, etc may be required, and however your specialist will arrange any tests that are necessary in order to monitor your heart's function. Screening tests may need to be repeated every couple of years, and if any symptoms develop screening tests will be used to decide if the symptoms are due to any changes in the heart caused by DCM.

4) Research Activities

During the 1990's much has been learned about the genetic mutations that contribute to the condition. At present these genetic analyses (diagnosis) are time consuming and very expensive, and not yet complete or available as a diagnostic laboratory test. Doctors researching this condition hope one day to offer a simple test. This would ensure early diagnosis, and treatment. A plan for managed care with ongoing screening tests so those individuals at risk of inheriting this condition can be investigated and monitored from an early age. Research into finding the genes that are responsible for Familial Dilated Cardiomyopathy is continuing, as is the development of genetic tests (to diagnose the condition). Research may also help in the future to produce gene specific therapies, (such as medications that work to control the abnormal genes, to stop them from being 'expressed' and thus stop the heart enlarging and causing the symptoms of the disease). There is currently no diagnostic genetic test available to screen for Familial Dilated Cardiomyopathy.

5) What is the Treatment for Dilated Cardiomyopathy?

There is no cure available for Familial Dilated Cardiomyopathy, at this time. However it can be treated. There are many medications such as Beta-blockers and ACE inhibitors (Angiotensin converting enzyme inhibitors) that help to reduce the load on heart muscle and reduce blood pressure to improve heart muscle function.

Implantable defibrillators (implantable devices which are able to deliver a shock to restore normal heart rhythm, used to prevent sudden cardiac death in high-risk patients) are also options for some individuals. If symptoms and cardiac function worsens then a cardiac transplant may be the treatment of choice. It is hoped research efforts will enhance treatment options available, by providing information on which treatments are more effective at treating the symptoms of the condition. Your doctor may advise lifestyle modifications such as avoidance of activities that cause severe symptoms like heavy manual work, and strenuous exercise. Substances known to worsen the condition such as cigarette smoking and alcohol consumption should also be avoided.

6) Further Information

The Cardiomyopathy Association of Australia Ltd is a non-government organisation providing information, support and advocacy for people living with cardiomyopathy and their families and supporting research into Cardiomyopathy. www.cmaa.org.au. New Zealand Representative:

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7) Participation in Cardiac inherited disease registry:

If you would like more information prior to deciding whether you would like to join the Cardiac inherited disease registry, please feel free to contact us.

If you have any queries or concerns regarding your rights as a participant in the Cardiac Inherited disease registry you can contact an independent Health and Disability Advocate. This is a free service provided under the Health & Disability Act; through the offices of the Health and Disability Commissioner.

Telephone (NZ wide): 0800 555 050

Free Fax (NZ wide): 0800 2787 7678 (0800 2 SUPPORT)

Email: advocacy@hdc.org.nz

This N.Z Registry has received Ethical Approval from the N.Z. Multi-centre Ethics Committee: AKX/02/00/107