Clinical Genetic Service leaflet reference: 067

Southeast of Scotland Genetic Service
Western General Hospital, Crewe Road, Edinburgh, EH4 2XU
Tel: 0131 537 1116

Dilated Cardiomyopathy (DCM) Predictive Testing

Information for patients

A gene change has been identified in your family which is thought to be the cause of your family history of Dilated Cardiomyopathy (DCM). This means that a gene test is available that can show if you have an increased risk of developing this condition.

- If you carry the gene change, your lifetime risk of developing DCM is likely to be significantly increased, and your children or future children would be at a 1 in 2 (50%) risk of inheriting the gene change.
- If you do not carry the gene change, your risk of developing DCM is likely to be similar to other people in the general population and your children or future children would not be at risk of inheriting the gene change.

If you are found to have the gene change, we would refer you to cardiology for regular follow up. There are a number of ways that they can help you to manage the increased risk.

Please see overleaf for more information.

Plan of action going forward

Please tick as appropriate		Enter details
	You have decided to proceed with testing. We have made the following arrangements for you to receive the results.	
	A follow up appointment has been arranged on	
	You have decided to get back in touch when you feel ready to proceed with testing.	

Following testing, results are usually available within 6 weeks. Results are confirmed in writing to you and copied to your referring clinician/GP.

You were seen by:	Genetic Counsellor	Patient label
Date:		

Dilated Cardiomyopathy is disease of the heart muscle. It can run in families and can affect one or more members of a family. Some members of a family may be more affected than others. Some family members may not be affected at all or may not develop symptoms. **In most cases, with proper diagnosis, treatment and follow-up, people with the condition live a normal life.**

DCM can be caused by a change (mutation) in one or more genes. Although the genetic change can be present from birth, the condition usually develops in adulthood. About 1 in 250 of the UK population has the condition.

It is important to identify families where DCM is inherited as a small number of people with the condition experience significant symptoms and could be at risk of sudden death. It is important that families affected are seen by **cardiology specialists** so that accurate diagnosis, treatment and support can be arranged if necessary.

The main abnormality in this condition is that the left ventricle of the heart becomes stretched (dilated). When this happens, the heart muscle becomes weakened and is unable to pump blood around the body effectively. This can lead to fluid building up in the ankles, abdomen, lungs and other organs and a feeling of being breathless. This collection of symptoms is known as heart failure.

Symptoms of DCM vary from none at all to considerable and may not appear until later life:

- Shortness of breath (dyspnoea)
- Swelling of the ankle and abdomen
- Excessive tiredness
- Palpitations (sensation that the heart has skipped or added an extra beat)
- Abnormal heart rhythm of beating too fast or too slowly
- Light headedness, blackouts or seizures
- Blood clots.

In most cases the condition develops slowly, so without cardiac screening, people can have quite severe symptoms before they are diagnosed. With cardiac screening, there is the opportunity to minimise symptoms and risk.

Treatments are determined by the results from cardiac screening and symptoms. At present there is no cure for DCM, but treatments are available to help control symptoms and prevent complications. If you are found to have the genetic change a cardiology specialist would organise regular tests for you, such as echocardiogram (an ultrasound of the heart) and ECG (that measures electrical impulses sent through the heart). These are to assess whether treatments should be considered, such as:

- Medicines – to reduce symptoms and the workload for the heart, to help control blood pressure, and minimise the risk of abnormal heart rhythms or stroke
- A pacemaker – to control the heart rate
- An ICD – if there is risk of having a life-threatening abnormal heart rhythm. The device constantly monitors the heart rate and delivers a shock to the heart to restore normal rhythm if necessary.

Healthy living advice includes regular gentle exercise, maintaining a healthy weight by eating a balanced diet (plenty of fresh fruit and vegetables), avoiding smoking and not drinking excessive amounts of alcohol. The standard childhood immunisations are recommended.

People with DCM may have to make some changes to their lifestyle, such as avoiding competitive sports, but most people are able to continue to work and drive a car. The cardiology service would provide individual advice about whether you could drive an HGV or commercial passenger vehicle and if manual jobs which involve strenuous activity should be reconsidered.

A limitation of the genetic test is that just like much of medical science, we cannot yet know with certainty all the factors that influence health. There is very small chance that current classification of a gene variant may change as more information becomes available. This may change the clinical diagnosis, prognosis or treatment you receive.