Southeast of Scotland Genetic Service
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Tel: 0131 537 1116

Genetic Testing – Dilated Cardiomyopathy (DCM) Information for patients

Patient Label

Dilated Cardiomyopathy (DCM) is a disease of the heart muscle. It can be caused by:

- Medical conditions such as uncontrolled high blood pressure, coronary artery disease, heart valve problems, skeletal muscle disorders, auto-immune disease or pregnancy. It can also be caused by excessive alcohol or other toxins.

Or

- A genetic condition that can affect one or more members of a family. It can be very variable. Some members of the family may be more affected than others. Some family members may not be affected at all. **In most cases, with proper diagnosis, treatment and cardiac follow-up, most people with the condition live a normal life.**

  It is important to identify families where it is caused by a genetic condition as a small number of people with the condition experience significant symptoms and could be at risk of sudden death. Relatives of people with DCM should be seen by cardiac specialists (please see overleaf).

There are a number of genes that when altered, predispose towards the development of DCM. Although the genetic change is present from birth, the condition usually develops later. **We are able to offer you a genetic test to see if you have alterations in these genes.**

Your possible results:

- ***If a genetic change is identified in your sample,*** this offers an explanation for your DCM development. It also means we can offer a genetic test to your blood relatives. Your children, siblings and parents would each have a 50:50 chance of having this gene change. They would be able to have a gene test to ensure their cardiac screening recommendations are appropriate.
- ***If a genetic change is not identified in your sample,*** we will not have a blood test to offer your relatives. Cardiac screening advice for your relatives would be on the basis of the family history. All your children, siblings and parents would be recommended to have cardiac screening. We would store your DNA in case in the future, further gene tests for this condition become available.
- **Occasionally we find an unclassified variant**. This is a change in a gene where it is not known if it had a role in causing you to have DCM or if it is just a natural genetic variation that is does not cause the disease. If you obtain this result, the cardiac screening advice for your blood relatives would be the same as NOT finding a gene change in your sample.

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 influence the clinical diagnosis, prognosis or treatment you receive.

For more information and links to support organisations you can look at the website; Network for Inherited Cardiac Conditions Scotland (NICCS) here:

www.niccs.scot.nhs.uk/

SCAN ME

Please tick as appropriate	Plan of action going forward	Enter details
	You have decided to proceed with testing and we have made the following arrangements for you to receive the results:	Letter / phone call /appointment
	You have decided to get back in touch when you feel ready to proceed with testing.	

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

You were seen by:	Genetic Counsellor (0131 537 1116)	Date:

Further Information

About 1 in 250 of the UK population has dilated cardiomyopathy. The main abnormality in this condition is that the left ventricle of the heart becomes stretched (dilated). When this happens, the heart muscle becomes weak, thin or floppy and is unable to pump blood around the body efficiently. This can lead to fluid building up in the ankles, abdomen, lungs and other organs of the body and a feeling of being breathless. This collection of symptoms is known as heart failure. Symptoms of DCM vary from none at all, to mild or 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

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

Treatments would be 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. Cardiology specialists 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 would be recommended.

People with DCM may have to make 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.