Southeast of Scotland Genetic Service
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Genetic Testing – Hypertrophic Cardiomyopathy

Information for patients

Patient Label

Hypertrophic Cardiomyopathy (HCM) is a disease of the heart muscle and can be a genetic condition. It can run in families and can affect one or more individuals. It is 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 such families as a small number of people with the condition do experience significant symptoms and could be at risk of sudden death. Relatives of people with HCM should be seen by specialists to provide accurate assessment and treatment if necessary (please see overleaf).

There are a number of genes that when altered, predispose towards the development of HCM. Although the genetic alteration is present from birth, the condition develops and is usually diagnosed as children grow into adults, after puberty. It may not appear until later life.

We are able to offer you a genetic test to see if you have alterations in these genes. The following is a summary of the outcome of this test:

- **If a genetic alteration is identified in your sample:** This offers an explanation for your HCM development. It also means we can offer a genetic test to your blood relatives. In most cases, your children, siblings and parents would each have a 50:50 chance of having this gene change. Their gene test would ensure their cardiac screening recommendations are appropriate.
- **If a genetic alteration 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 of 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 an alteration in a gene where it is not known if it was significant in causing you to have HCM or if it is just a natural variation that 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.

Further information

Hypertrophic cardiomyopathy (HCM) affects around 1 in 500 people. The main abnormality is that the heart muscle (myocardium) has become excessively thick. How thick the muscle is and how much of the muscle is affected can vary. The left ventricle (chamber) may become thickened and in some people the muscle of the right ventricle may also thicken. The thickening and scarring of the muscle make the heart muscle stiff. This prevents the heart from pumping enough blood, especially during exercise and higher blood pressure is needed to make the muscle contract and pump the blood out of the heart.

Symptoms vary from none at all, to mild or considerable and may not appear until later life. The symptoms may be any of the following:

- Shortness of breath during exercise (dyspnoea)
- Chest pain (angina)

- Palpitations (sensation that the heart has skipped or added an extra beat)
- Light headedness and blackouts or seizures
- Abnormal heart rhythm (arrhythmia).

Treatments should be determined by any symptoms/presentation. At present there is no cure for HCM, but treatments are available to help control symptoms and prevent complications. Some people with HCM are at risk of getting an abnormal heart rhythm that puts them at increased risk of sudden cardiac death. A cardiology specialist will organise regular cardiology tests. These may include an echocardiogram (ultrasound of the heart) and ECG (Electrocardiogram- that measures electrical impulses sent through the heart). These tests monitor and assess whether treatments and risk reducing measures should be considered. These include:

- Medicines – to help control blood pressure, abnormal heart rhythms or other symptoms
- A pacemaker – to control the heart rate
- An ICD (Implantable Cardiac Defibrillator) – if there is risk of having a life-threatening abnormal heart rhythm. The device constantly monitors the heart rate and would deliver 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 HCM 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. However, they will not be able to drive an HGV or commercial passenger vehicle and may have to reconsider manual jobs which involve strenuous activity. The cardiology service provides individual advice about activity or lifestyle implications.

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 a 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.*

For more information and links to support organisations you can look at the website: Network for Inherited Cardiac Conditions Scotland (NICCS) at www.niccs.scot.nhs.uk/

Plan of action going forward

Please tick as appropriate		Enter details
	You have decided to proceed with testing and we have made the following arrangements for you to receive the result:	
	You have decided to postpone testing at this time. You are aware your first degree relatives may still be eligible for clinical screening because of your HCM diagnosis.	

We will write to your referring clinician/GP with details of the plan above.

Following testing, **results are usually available within 3 months**. Results are confirmed in writing to you and copied to your referring clinician/GP.

Seen by	Genetic Counsellor (contact number at top of first page)	Date: