Southeast of Scotland Genetic Service
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Hypertrophic Cardiomyopathy (HCM) Predictive Testing

Information for Patients

A gene alteration has been identified in your family which predisposes to Hypertrophic Cardiomyopathy (HCM). This means that a test is available for you, to see if you have the gene alteration and have an increased risk of developing HCM.

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

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

Please see below and overleaf for more information about HCM

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 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:		

Hypertrophic Cardiomyopathy is a disease of the heart muscle and can be a genetic condition. It can run in families and can affect one or more members of a family. It is very variable. Some members of the 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, most people with the condition live a normal life.**

Although the genetic change is present from birth, the condition develops and is usually diagnosed as children grow into adults, after puberty. About 1 in 500 of the UK population has the condition.

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

The main abnormality in this condition 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, such as echocardiogram (ultrasound of the heart) and ECG (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 Implantable Cardiac Defibrillator (ICD) – 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.

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 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.