Southeast of Scotland Genetic Service
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Arrhythmogenic Cardiomyopathy (ACM) Predictive Testing

Information for Patients

A gene variant linked to an increased risk of Arrhythmogenic Cardiomyopathy (ACM) has been identified in your family. Genetic testing is now available to determine whether you have this variant and assess your risk of developing the condition.

- **If you have the gene variant**, your lifetime risk of developing ACM is significantly increased. Additionally, each of your children have a 1 in 2 (50%) chance of inheriting this gene variant.
- **If you do not have the gene variant**, your risk of developing ACM is similar to that of the general population and your children would not be at risk of inheriting the gene variant.

If you have the gene variant, we will refer you to a cardiologist for regular monitoring and advice. There are several treatments and interventions available to manage the condition if signs of ACM develop.

Please see below and overleaf for more information about ACM

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:		

Arrhythmogenic Cardiomyopathy (ACM), sometimes still referred to as Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), is caused by a problem with a protein in the heart muscle. This protein acts like glue, holding the heart muscle cells together. When this “glue” doesn’t work properly, the walls of the heart can become weakened.

ACM is usually a genetic condition that can run in families, affecting one or more relatives. However, its impact can vary widely.

The condition is caused by a variant in a gene. Although the gene variant is present from birth, symptoms often do not appear until adulthood.

Identifying those at risk within a family is very important because a small number of people with ACM may experience serious symptoms or be at risk of sudden cardiac death. It is essential that affected individuals are evaluated by cardiology specialists, who can provide an accurate diagnosis and arrange appropriate treatment if needed.

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:

- Palpitations (or arrhythmias) feeling your heart beating too fast or 'fluttering'
- Light headedness or blackouts/fainting
- Shortness of breath on exercise
- Swollen ankles, legs or tummy area
- Chest pain (angina)

Treatments should be determined by any symptoms. At present there is no cure for ACM, but treatments are available to help control symptoms and prevent complications. Some people with ACM 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. Possible treatments 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 rhythm 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 ACM 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 the 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 about ACM:
https://www.bhf.org.uk/informationsupport/conditions/arrhythmogenic-cardiomyopathy

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