Clinical Genetic Service leaflet reference: 065

Southeast of Scotland Genetic Service
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Familial Hypercholesterolaemia (FH) Predictive Genetic Test

Information for patients

Thank you for attending the Genetic Clinic to discuss your family history of Familial Hypercholesterolaemia (FH).

A gene alteration has been identified in your family and is thought to be the cause of your family history of Familial Hypercholesterolaemia (FH). This means that a test is available for you, to see if you have inherited the gene alteration and whether or not you need to be referred to the Lipid Clinic for cholesterol monitoring and treatment.

- ***If you carry the gene change***, you are likely to develop high levels of cholesterol in the blood because you will be less able to remove the form of cholesterol called LDL. If you have FH and are not on treatment, you will be at a much greater risk of heart attack than other people. Modern treatments can restore cholesterol levels to normal or near normal for most people with FH. For example, if you have FH and take "statins", and don't already have coronary heart disease, you will have the same life expectancy as people who don't have FH. Your children or future children will be at a 1 in 2 (50%) risk of inheriting the gene alteration.
- ***If you do not carry the gene change,*** your risk of high levels of cholesterol is similar to other people in the general population and your children would not be at risk of inheriting the gene alteration.

Please see overleaf for more information.

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:	
	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. If you have inherited the gene alteration you will be referred to your local Lipid Clinic for ongoing management.

You were seen by:	Genetic Counsellor	Patient Label
Date:		

Additional leaflets provided:

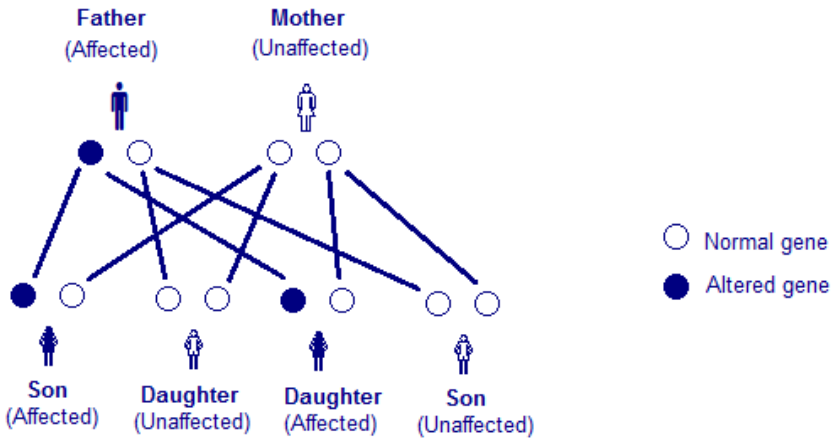
- “Familial Hypercholesterolaemia: Your quick guide”. British Heart Foundation
- “Life with Familial Hypercholesterolaemia”. British Heart Foundation

Familial Hypercholesterolaemia (FH)

Hypercholesterolaemia is the medical term for high cholesterol in the blood. Cholesterol is a major contributor to heart attacks. Narrowing of the coronary arteries in the heart results from cholesterol laid down in the wall of the artery. Although this can take many years to occur before it becomes serious, the process is accelerated if blood cholesterol is high. Fortunately high cholesterol responds extremely well to medical treatment (using a medicine called a “statin”) and this greatly reduces the risk of heart attacks at an early age.

In some families there is a tendency to have very high blood cholesterol , even if they follow a healthy diet and this is then associated with heart attacks at a much earlier age than usual. The high cholesterol arises because affected family members carry a gene alteration which is responsible for Familial Hypercholesterolaemia (FH). People with FH have an alteration in one of their two copies of a gene that is critical for removing cholesterol from the blood. Identifying affected family members and offering treatment can be life saving.

About one in every 200 people in the UK has FH, and it is one of the most commonly occurring inherited conditions. FH can be inherited from a parent. We each have two copies of every gene (one from our father and one from our mother). FH is passed on in the family in an autosomal dominant manner. This means that only one copy of the gene needs to be altered for someone to have the condition. If a parent carries an alteration in one of their FH genes, their child (male or female) has a 50% (or 1 in 2) chance of inheriting the gene alteration and having FH.



Treatment for FH:

Medication: to lower the level of cholesterol in the blood. “Statins” are most commonly used, but others may also be helpful. This is discussed at the Lipid Clinic.

Smoking: it is especially important for people with FH not to smoke. Those who do are 3 times more likely to have a heart attack than people who do not smoke. Support is available to stop.

Diet: advice should be available to follow a cholesterol lowering diet.

Alcohol: it’s recommended to follow the same advice on moderate drinking as given to everyone else.

Modern treatments can restore cholesterol levels to normal or near normal for most people with FH. People who are adequately treated for FH are expected to have a normal lifespan.