South East Of Scotland Genetic Service
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FAMILIAL HYPERCHOLESTEROLAEMIA (FH)

VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)

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.

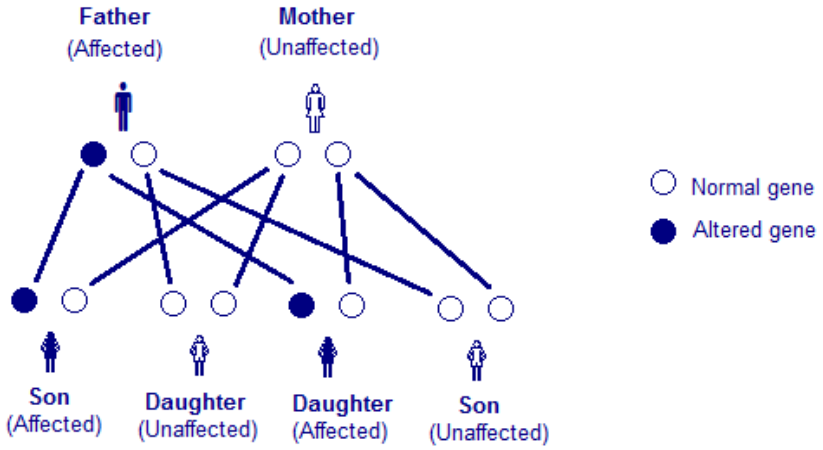
Fortunately high cholesterol responds extremely well to medical treatment (using a medicine called a “statin”) and this greatly reduces the risk of heart attacks at an early age. Identifying affected family members and offering this simple treatment can be life saving.

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.

This leaflet has been given to you because you have been clinically diagnosed with FH. In some families we are able to identify gene alteration causing FH. Your genes have been tested and although the lab has identified a gene alteration, we do not know if this is the cause of your FH. This type of change is called a **Variant of Uncertain Significance** (VUS).

Testing for family members is not usually indicated when a **Variant of Uncertain Significance** (VUS) has been identified. However, depending on the type of **Variant of Uncertain Significance** (VUS) it may be that the lab can offer family studies, in which case this will be discussed with you at your appointment.

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.



Your cholesterol levels and medication will be monitored by a Specialist doctor (Lipidologist) and your GP. Although family genetic testing is not indicated for the **Variant of Uncertain significance (**VUS) we would suggest that close family members (children, brothers, sisters and parents) have their cholesterol levels checked. This is usually advised from around age 16 years.

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.

SUPPORT AND INFORMATION

"Heart UK" is a patient support group and publishes a regular newsletter.
Address: 7, North Road, Maidenhead. Berkshire. SL6 1PE
Website: www.heartuk.org.uk

LINK TO LEAFLET LINK TO LEAFLET:

- https://www.bhf.org.uk/informationsupport/publications/heart-conditions/m111f-inherited-heart-conditions---familial-hypercholesterolaemia