Clinical Genetic Service leaflet reference: 005

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Familial Hypercholesterolaemia (FH)

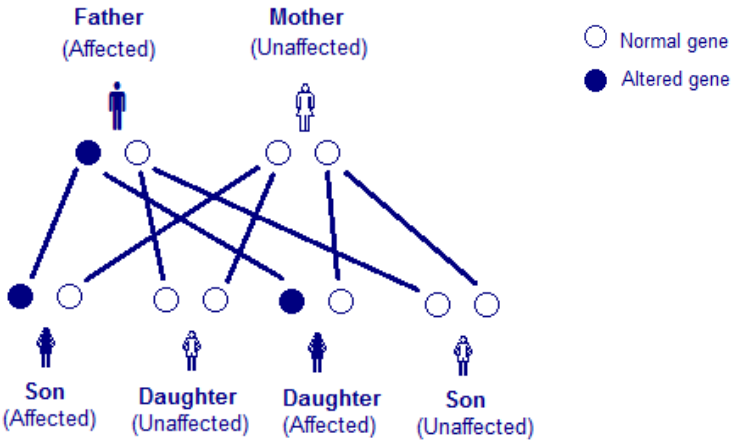
Information for patients with a confirmed diagnosis

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.

The laboratory has identified the underlying genetic alteration responsible for your Hypercholesterolaemia. We have documented your family history and identified other family members at risk of FH. As the genetic cause for your FH has been confirmed, there is now an informative test available for family members to determine if they have inherited the same gene alteration and are at risk of high cholesterol and early heart disease.

Your relatives at risk are your first-degree relatives (children, brothers, sisters and parents). They have a 1 in 2 (50%) risk of inheriting the gene alteration. 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.

The diagram below shows how FH is inherited.



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, like you, carry a gene alteration which is responsible for Familial Hypercholesterolaemia (FH). About one in every 200 people in the UK has FH, and it is one of the most commonly occurring inherited conditions.

Genetic tests are very important for identifying other family members at risk of FH once an affected family member has been diagnosed.

The genetic test is the best way to know whether or not your relatives have FH. It provides a much more definite result than just a blood cholesterol test; this is especially so in FH families where some cholesterol measurements fall in a "grey" area leaving some uncertainty as to the diagnosis.

A cholesterol test is **not** the best way and your relatives should **not** just rely on a cholesterol test, even though they believe or have been told that their cholesterol is "normal".

We would like you to pass your information to other family members at risk, so they can have a genetic test.

If your relatives live within Lothian, Fife or the Borders, they can return the supplied *"To whom it may concern"* reply slips to request an appointment. Otherwise they can seek referral to their local genetic service through their GP.

Your cholesterol levels and medication will be monitored by a Specialist Doctor (Lipidologist) and your GP.

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:

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