THE SCOTTISH FAMILIAL HYPERCHOLESTEROLAEMIA WORKING PARTY

Familial Hypercholesterolaemia – Patient Information Leaflet

What Is Familial Hypercholesterolaemia (FH)?
Hypercholesterolaemia is the medical term for high cholesterol in the blood. In some families, very high blood cholesterol is passed from parent to child. This condition is called familial hypercholesterolaemia. It affects about 1 in 500 people.

About FH
Everyone has cholesterol in the blood. People with FH are less able to remove the form of cholesterol called LDL from the blood. This is because they have a change in a gene that is important for removing cholesterol.

FH and Heart Disease
Heart disease is caused by narrowing of the heart arteries. This happens because cholesterol is laid down in the arteries of the heart. If someone has FH and is not on treatment they are at a much greater risk of heart attack than other people. High blood pressure and smoking also increase the risks of heart attack. Treatment from an early age removes this extra risk.

How is FH Diagnosed?
There are strict criteria to diagnose FH. These include blood tests and examination.
DNA blood tests may be able to help clarify the diagnosis and are very useful for testing family members.

What Causes FH?
Genes are tiny “packages” of information which influence how our bodies grow, develop and function. We each have two copies of every gene (one from our father and one from our mother).
FH is caused by changes in genes that control cholesterol levels. People with FH have a change in one of the two copies of a gene. When they have a child there is a 50:50 or 1 in 2 chance that FH will be passed on.

How Do Doctors Use FH Genetic testing?
This is used to confirm a diagnosis and then to test other members of a family.
If the test shows a gene change, then we can make sure that the patient is on the right treatment, and the patient’s family is then offered testing.
If no genetic fault is detected, the patient should still remain on their medication for a raised cholesterol.

Will a Genetic Test for FH Affect My Insurance?
When a genetic fault is detected in a family member, we can offer the genetic test to other members of the family who are not affected. In these cases we call it a predictive test (i.e. a test that tells you whether you are at risk of developing a condition in the future). As with all types of medical test, if you take a predictive test after buying an insurance policy, this will not affect the premium you are paying; an insurer's assessment of your application for insurance can only be made once, on the information available at the time. At present there is a moratorium, due for review in 2011, stating

that insurance companies are not supposed to discriminate on the grounds of gene test results. For more information see:
www.londonideas.org/internet/public/insurance.htm, www.abi.org.uk

Treatment for FH

MEDICATION – FH patients need lipid lowering medication. Most commonly used are "statins" but others may also be helpful. These will be discussed by the doctor at the Lipid Clinic.
SMOKING – people with FH who smoke are 3x more likely to have a heart attack than those who do not smoke.
DIET – is important. People with FH should see a dietician.
ALCOHOL – moderate amounts of alcohol are acceptable.

Benefits of treatment

Modern treatments can restore cholesterol levels to normal or near normal for most FH patients. People who are treated for FH have been shown to have a normal life expectancy.

Further information

"Heart UK" is a patient support group with a regular newsletter. 7 North Road, Maidenhead, Berks, SL6 1PE. 01628-628638 www.heartuk.org.uk