

What are the possible results of predictive testing?

Testing can reveal one of two results:

You **have not** inherited the altered gene:

- Although an altered FH gene is present in your family, you have not inherited it.
- Your own risks of developing coronary heart disease are not significantly increased with regards to FH.
- As you do not have the altered FH gene, you cannot pass it on to your children.
- You may still have raised cholesterol due to other factors, so cholesterol testing may still be appropriate.

You **have** inherited the altered gene:

- Effective treatment is now available for people diagnosed with FH which significantly reduces the risk of heart attacks in both men and women. Preventative treatment with cholesterol lowering drugs (including statins) in combination with a healthy lifestyle is effective in delaying or preventing the onset of coronary heart disease.
- Each of your children will have a 50% chance of inheriting the altered FH gene. Genetic testing can be offered to children at risk of FH from the age of 10 years.

Follow up appointments and support:

Once you have received your predictive test result, your geneticist or genetic counsellor will arrange to phone you in the following week and if appropriate follow up appointments can be arranged. Of course we are available at other times and you are welcome to contact us.

Understandably, predictive testing and receiving the results can be a challenging time. Some people would like further opportunities to talk through their feelings, coping strategies and more information.

Contact numbers

If you have any further questions please contact your genetic counsellor or geneticist.

Genetics Department Tel No.

Your Consultant Geneticist is:

Tel No.: _____

Your Genetic Counsellor is:

Tel No.: _____

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Predictive Testing for Familial Hypercholesterolaemia (FH) Genes



This leaflet has been given to you by one of the genetics team to provide you with some information about predictive testing for FH. It also highlights some of the important things to think about before having the test.

What is Familial Hypercholesterolaemia (FH)?

FH is an inherited condition which results in high cholesterol levels in the blood from birth. "Familial" means that it is inherited through families. "Hypercholesterolaemia" is the medical term for high blood cholesterol. "Hyper" means raised and "aemia" means in the blood. It can cause the arteries to narrow and clog and can lead to early angina and heart attacks (coronary heart disease). FH is usually diagnosed using a combination of factors including raised blood cholesterol, fatty deposits in tendons (xanthoma), a family history of high cholesterol and/or coronary heart disease. In some families it is possible to confirm the diagnosis by testing your genes (genetic testing).

What are genes?

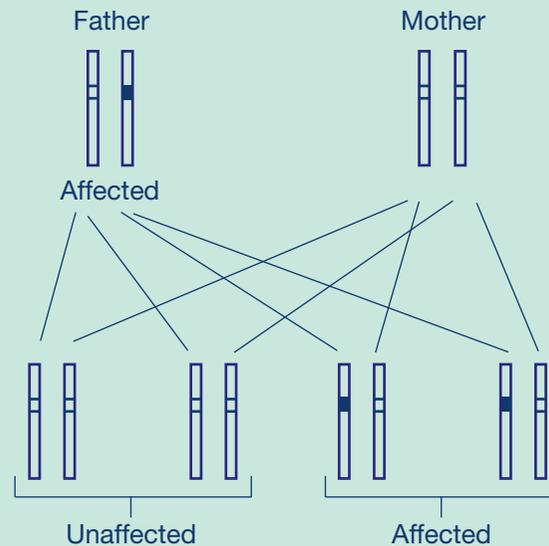
Genes are coded messages which give instructions for how cells in our body grow and function. Genes come in pairs and we inherit one copy from each parent.

How are genes are involved in FH?

We know of at least three genes involved in FH, called LDLR (low-density lipoprotein receptor), APOB (apolipoprotein B-100) and PCSK9 (proprotein convertase subtilisin/kexin 9). Genetic testing has shown that a change (alteration) in one of these genes has been found in a member of your family. This alteration leads to increased levels of cholesterol and a higher risk of angina and heart attacks (coronary heart disease) in both men and women.

How are the FH genes inherited?

We have two copies of our FH genes. If someone is affected with FH, they will have one copy with an alteration and one unaltered copy. Only one copy of each gene pair is passed on to a child. If your parent has an altered FH gene then you have a 50% chance of inheriting the altered gene. If you have an altered FH gene then each time you have a child there is a 50% chance that you will pass on this gene alteration.



Occasionally the inheritance of FH can be more complicated, but this will be explained if appropriate.

What is a predictive genetic test?

As someone in your family has an FH gene alteration, you could have inherited this. We can offer you a genetic test to see if this alteration is present in you. We call this a predictive test and it involves taking a blood sample.

Before we take a blood sample from you we will discuss what the test could mean for you and your family. You will have the opportunity to ask questions, so that you can make sure that this is something you want to do. This test is not something that you need to rush into, and you will be offered further appointments if necessary.

Your geneticist or genetic counsellor will help you to consider:

- How this test result could affect you and your future
- If this could affect your employment and insurance
- How this test result could affect your family, children and relationships
- Who you plan to talk to about this test and the result
- Who you would like to share your result with

Receiving your result:

Test results are usually given in person and we will arrange an appointment for you to come to clinic approximately 4 to 6 weeks after we take your blood sample. We advise that you bring someone with you to this clinic appointment for support.