PATIENT INFORMATION: FAMILIAL HYPERCHOLESTEROLAEMIA(FH)

Familial hypercholesterolaemia is inherited high cholesterol. It is a condition where you have a very high cholesterol level in your blood.

What does having familial hypercholesterolaemia mean?

Familial hypercholesterolaemia (FH) is called familial because it runs in the family (the other word for this is 'inherited'). Hypercholesterolaemia means cholesterol levels in your blood are too high.

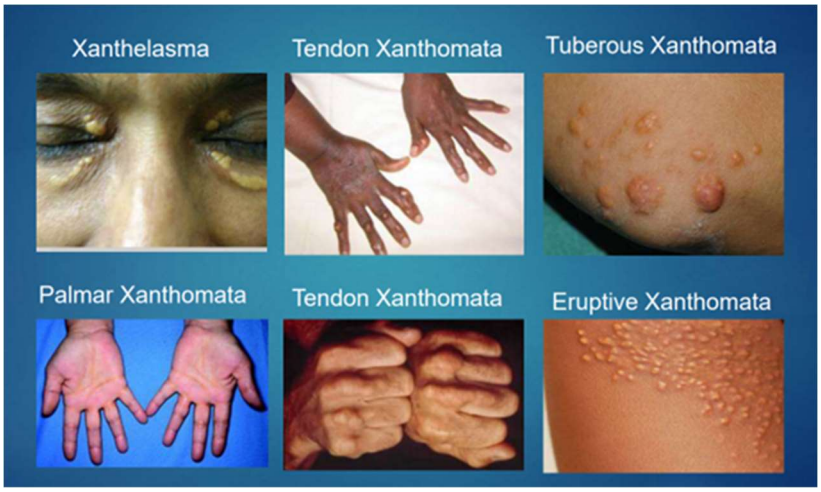
Cholesterol levels in your blood are normally controlled when cholesterol is taken back to your liver and broken down. In FH, you inherit at least one faulty gene, which means your body cannot get rid of excess cholesterol from your blood.

Without treatment, people with FH are at higher risk of heart attack than average and may have a heart attack in their 40s or 50s even if they have no other risk factors for heart disease.

How do I know if I have familial hypercholesterolaemia?

Having a high cholesterol level doesn't make you feel ill. You won't know you have it without having a blood test.

The most important feature is the development of heart disease at a young age. You might also notice:



How is familial hypercholesterolaemia diagnosed?

You may be diagnosed by chance if you go for a health screening check, or you may notice fatty deposits on the skin or around the eyes.

They may suspect FH if you have this level of raised cholesterol and:

- If you or a 'first-degree relative' - parent, brother, sister or your child - have a heart attack under the age of 60; or

- If a 'second-degree relative' - uncle, aunt, nephew, niece, grandparent - has a heart attack under the age of 50; or
- If other close relatives have cholesterol raised to the levels above.

Do I need referral if I have familial hypercholesterolaemia?

If you have suspected or definite FH, you should be referred to a specialist FH centre for DNA testing to confirm the diagnosis.

If this testing confirms that you have FH, other members of your family should also be referred for DNA testing to see if they have FH as well. This includes all first-degree (parents, brothers and sisters, children) and second-degree (uncles aunts, nephews, nieces, grandparents, grandchildren) relatives. This 'cascade testing' should be carried out in a specialist centre.

Cascade testing is very important because it can allow other members of your family who have FH to be diagnosed and treated. This can prevent them having heart attacks at an early age.

What are the treatment options for familial hypercholesterolaemia?

Being a genetic disorder, FH is not caused by an unhealthy lifestyle. However, keeping yourself in the best physical condition will help to prevent future problems. Things you can do to help yourself include:

Healthy eating: in general, it's a good idea to eat healthily. But it's important to note that what you eat makes hardly any difference to your cholesterol level.

Getting a reasonable amount of exercise. Ideally this should be at least 30 minutes of moderate-intensity or high-intensity physical exercise at least five times a week.

Avoiding smoking: this is vital if you have high cholesterol, but even more if you have FH.

Maintaining a normal weight: but slim people can have high cholesterol too. Being overweight doesn't cause high cholesterol.

You will be offered medication to help bring your cholesterol level down. The aim will be to reduce your LDL cholesterol by at least 50% compared to your pre-treatment level.

Can I live well with FH?

The outlook for people with FH is usually good if they maintain a healthy lifestyle, have regular checks and take their medication without fail.