

FH in Children

High cholesterol is present in people from birth. It is recommended that children at risk (because of an affected parent, brother or sister) should be tested from the age of 10 years. Children should be encouraged to exercise regularly, eat a healthy diet (ensuring there are sufficient calories and nutrients to support normal growth) and strongly discouraged from smoking. Cholesterol-lowering medication can safely be taken from 10 years onwards and should be prescribed by a specialist clinic.

FH Family Forum

The FH family forum is a group of individuals and families in Wales who are affected by FH. The aims of the forum are:

- To provide a network for individuals and their families with inherited high cholesterol.
- To develop and promote services for FH within Wales.
- To provide a forum for sharing advances in this area.

To be included in the mailing list for newsletters and information about future events, please contact fhwales@gmail.com or find them on facebook by searching for "FH for short".

Further information

www.wales.nhs.uk/FHservice

Wales FH Project

www.heartuk.org.uk

HEART UK, The Cholesterol Charity

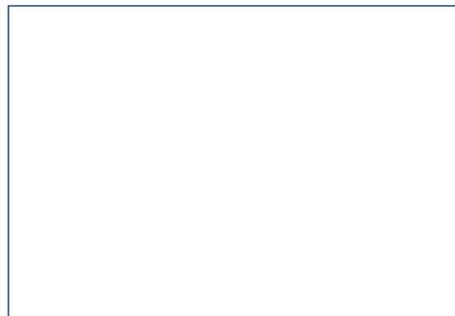
www.bhf.org.uk

British Heart Foundation

www.nice.org.uk/cg71

NICE Guideline for FH

Your local FH contact is:



Information Leaflet for Patients

Familial Hypercholesterolaemia (FH)

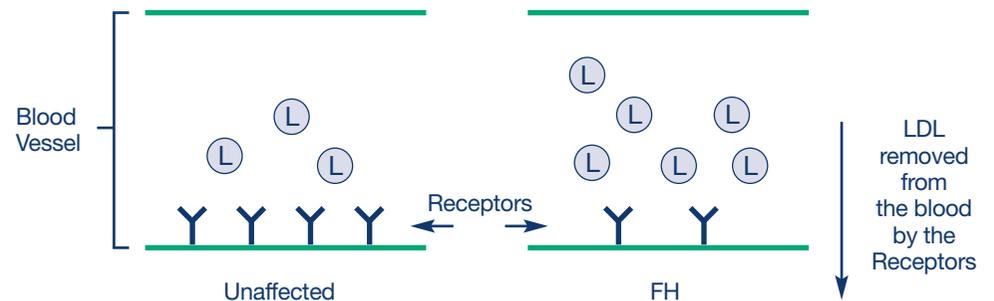
What is FH?

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 coronary heart disease. However, there are now effective treatments available and, combined with a healthy diet and lifestyle, the risk of heart disease is much improved. In particular, it is important not to smoke. People with FH who are treated can expect to live a normal and healthy life.

What causes FH?

FH is caused by increased levels of a specific type of cholesterol - Low Density Lipoprotein (LDL) Cholesterol. In most people, LDL is safely removed from the blood by receptors. People with FH have less of these receptors and this leads to high levels of LDL in the blood.



Diagnosis of FH

A diagnosis is based on a combination of high cholesterol levels, clinical signs and a family history of heart disease or high cholesterol. DNA testing can be helpful to confirm the diagnosis.

Signs of FH



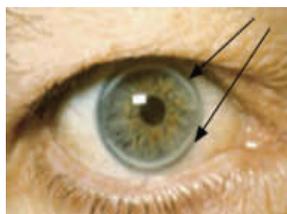
Tendon Xanthomata

Pronounced Zan-tho-mata. These are cholesterol deposits which show up as fatty lumps on the Achilles tendons, knuckles or knees.



Xanthelasmas

Pronounced Zan-thel-as-mas. These are cholesterol deposits which show up as small lumps near the inner corner of the eye. They are usually yellow in colour.



Corneal Arcus

This is a white ring around the iris. It can occur naturally as people get older (over the age of 60) and it doesn't always mean that someone has FH. If present, this is a reason to have your cholesterol measured.

However, many people with FH do not have these tell-tale physical signs as they may not appear until middle age, if at all. For this reason, people are often unaware that they have FH until they (or a family member) suffer from a heart attack at a young age.

Treatment for FH

FH is a very treatable condition which can be controlled with the use of cholesterol-lowering medication, combined with a healthy diet and lifestyle.

Diet - People with FH should follow a generally healthy diet similar to other individuals at risk of heart disease. Guidance can be gained from a dietician experienced in cholesterol management.

Smoking - It is particularly important that individuals do not smoke. In people with FH, smoking approximately trebles the risk of a heart attack.

Medication - Statins are the most commonly used form of medication, but other types of medication may also be helpful. This can be discussed with your doctor.

It is now known that people with FH who are treated can expect to have a life expectancy which is the same as the general population. Therefore, it is important to diagnose the condition early, so that the right treatment can be started and heart attacks prevented.

Genetics of FH

FH occurs in around 1 person in 500 and is one of the most frequently occurring inherited conditions.

FH is caused by an alteration in a single gene. It is inherited in such a way that immediate blood relatives of individuals with FH (parents, brothers, sisters, children) have a 50:50 chance of also having FH.

DNA Testing

This is a relatively recent development in the diagnosis of FH. It is a type of blood test where a person's DNA is examined in a laboratory to find out if they carry a particular alteration (mutation) in one of the genes which cause FH.

This type of test can be used in some individuals to make a definite diagnosis of FH. If a DNA diagnosis has been made in an individual, DNA testing for FH can then be offered to family members to see whether or not they also have the condition. This is more reliable than cholesterol testing alone, as cholesterol levels can be variable.

Cascade Testing

Cascade testing is another name for family testing. It is used to identify those with FH within families. When an individual is diagnosed with the condition, cascade testing is recommended to identify others in the family who may have FH, so they can be offered treatment to lower cholesterol (and prevent heart attacks).

Immediate blood relatives are offered testing and then, if any of these relatives are found to have FH, their relatives are also offered testing, and so on, in a cascade fashion.