

Inherited high Cholesterol (FH and FCH)

Everybody's cholesterol levels are affected by the genes they inherit from their parents. Why? Because there are many genetic traits that influence the amount of cholesterol and other blood fats in the blood stream and how these are transported around the body and ultimately disposed of. However there are two main forms of inherited high cholesterol – familial hypercholesterolaemia (FH) and familial combined hyperlipidaemia (FCH) which can significantly increase the risk of cardiovascular disease. Together they affect about 1 in 100 of the population. It is important to identify people with FH and FCH so that they can be treated effectively.

Familial Hypercholesterolaemia (FH)

FH occurs in about one person in every 500 and is one of the most frequently occurring inherited conditions. It is caused by an abnormal gene resulting in exceptionally high cholesterol levels, usually between 7.5 and 12mmol/l but sometimes in excess of 20mmol/l. Triglycerides are generally not increased, or if they are, only moderately. High cholesterol levels start from birth and are present throughout life. People with FH are at high risk of early cardiovascular disease (CVD).

More than 120,000 people in Britain have the problem, a similar number to those who need insulin to control their diabetes. However, unlike that type of diabetes, many people with FH go undetected with tragic consequences.

People with FH may develop heart problems or die from cardiovascular disease early in life, often in their 30s, 40s or 50s, sometimes earlier, especially if they are not treated.

Family Matters

FH is a genetic problem and can be passed from parent to child. Each family member has an even chance of inheriting the problem (like tossing a coin). Whenever FH is diagnosed, it is essential that all close blood relatives have their cholesterol levels measured so that other family members with FH can be traced and they too can start preventative treatments.

Signs of FH

As well as a very high cholesterol level and a strong family history of CVD, FH can sometimes be recognised by outward signs, 'lumps and bumps', which need the expert eye of a doctor for accurate diagnosis. Not everyone with FH has these signs. They may result from cholesterol deposited in the tendons at the back of the hands overlying the knuckles and in the Achilles tendon at the back of the ankles. The resulting swellings are called tendon xanthomata (pronounced zan-tho-mata). Cholesterol may also be deposited in the skin around the eye or eyelid. These deposits are usually yellow and are called xanthelasmas (pronounced zan-thel-as-mas). Another visible sign often seen in people with FH is a pale or white ring around the inside of the outer rim of the iris, the coloured part of the eye. The ring is called corneal arcus. Only tendon xanthomata are specific to FH. Xanthelasmas and corneal arcus can occur for other reasons as we get older.

Testing of FH in children

It is important to identify FH in childhood, particularly in families where CVD occurs early in adult life. Children should be tested before the age of ten, but generally not before age two, although it is possible to test for FH soon after birth. A diet low in saturated fat can be followed by children over two years of age, provided there is sufficient energy and nutrients to support normal growth. Parents should be referred to a registered dietitian experienced in lipid management. Whilst opinions differ regarding the treatment of FH in children, it is recognised that they will be in a position to benefit early in adult life if their FH is diagnosed in childhood. Lipid lowering drugs for children should only be prescribed by a specialist clinic.

This fact sheet includes images of xanthelasmas, tendon xanthomata on the Achilles tendon and on the back of the hand and corneal arcus.

Familial Combined Hyperlipidaemia

Familial Combined Hyperlipidaemia (FCH) is another inherited disorder of cholesterol. It is characterised by having a high triglyceride level as well as high cholesterol. This condition is not as well understood as FH, but is also associated with premature CVD. The elevated blood fats may not be present in childhood, and may not appear until the 20s or 30s. Tendon deposits are not present in FCH, and cholesterol levels tend to be not quite as high as in FH. Treatment generally consists of healthy lifestyle practices and medication as discussed below.

Treatment of FH and FCH

Diet is an essential element for the treatment and management of FH and FCH, although medication is generally also necessary at some stage. There are several cholesterol lowering drugs available on prescription that are effective in treating these conditions. It is important to maintain a heart healthy diet and lifestyle, even when prescribed medication, as the two complement each other.

Foods fortified with plant sterol and stanols can be used as part of a healthy diet to help improve cholesterol lowering. These products work by blocking cholesterol absorption in the gut and can lower LDL cholesterol by 10-15%.

Drug treatment

Drug treatment is generally reserved for people at high risk of CVD for whom diet and lifestyle measures have failed to reduce blood lipids to acceptable levels. This will include many people with FH and FCH and most individuals who have already developed CVD. Increasingly, other groups of people, particularly those who combine more than one risk factor, such as those with diabetes, are also treated with lipid-lowering drugs. The following are the more commonly prescribed classes of drugs for treating lipid disorders.

Statins

These come in tablet form and work by slowing down production of cholesterol in the liver, which is where most of the body's cholesterol is made. They do this by stimulating the receptors in the liver to get rid of excess cholesterol.

Statins are thoroughly tested and very effective at lowering cholesterol, LDL in particular. They are generally well tolerated, and reassuringly have been shown to extend life by preventing CVD. If generalised muscle aches (flu like aches and pains) occur, this should be reported to the doctor (although it is rare for statins to be the cause of muscle inflammation). Statins should not be used in people with liver disease, pregnant women or women who might become pregnant. Use of statins in children should be under supervised specialist care.

Fibrates

Fibrates come in tablet form and are useful when both triglycerides and cholesterol are raised. These drugs tend to be well tolerated. Fibrates should not be used during pregnancy or by individuals with liver or kidney disease.

Resins

Also known as bile acid sequestrants, these come as powder, granules or tablets. The powder and granules are taken by mixing with water, fizzy drinks, fruit juice or yoghurt. Resins prevent re-absorption of bile salts in the intestine, which means that more cholesterol is used up in replacing them. Resins are safe for children because they are not absorbed into the body, although extra folic acid and fat soluble vitamins may be recommended. Many people may experience side effects such as excessive wind and constipation which can limit the usefulness of resins.

Selective cholesterol absorption inhibitors

These drugs block the absorption of dietary and biliary cholesterol in the gut. They work at the brush border of the small intestine. They can help to reduce cholesterol levels by up to 18%. However, when they are combined with a statin drug they can greatly enhance the cholesterol lowering effect.

Omega-3 fish oils

Eating two portions of fish each week, one of which should be oily is recommended for all groups of the population in the UK. In addition, following a heart attack, people are advised to take 2-4 portions of oily fish per week for up to 4 years or alternatively are prescribed a medicine containing 1g of omega 3 fish oils per day.

Fish oils (2-4g daily) have also been shown to reduce plasma triglyceride levels. Although fish oils can be prescribed, for cost reasons there may be some reluctance to do so. The active ingredients in fish oils are EPA and DHA. Many over the counter supplements contain less EPA and DHA, than the equivalent fish oil medicines.

Before opting to take supplements it is important to discuss this with your doctor or dietitian especially if you are taking other medicines such as anticoagulants like warfarin.

Other treatments

Apheresis

Apheresis is a treatment similar to kidney dialysis. A person is connected to a machine that removes the LDL or bad cholesterol from their blood. The 'cleaned' blood is then returned to them. The process takes about 3 hours to perform and has to be repeated at fortnightly intervals. The procedure is quite expensive and is only available at specialist centres. It is used when people still have a higher than desirable cholesterol level despite diet and tablets.

Liver transplantation

In very extreme cases, complete or partial liver transplantation can be undertaken. However, the procedure is extremely difficult and the long term effect of taking anti rejection drugs such as cyclosporine can shorten life expectancy.