



HEART UK
THE CHOLESTEROL CHARITY



Familial Hypercholesterolaemia

An educational booklet for people
with familial hypercholesterolaemia (FH)

HEART UK – The Cholesterol Charity
providing expert support, education and influence

Contents

Familial Hypercholesterolaemia

What is familial hypercholesterolaemia (FH)?	03
The family tree	04
What is cholesterol?	05
What causes FH?	06
Why does my doctor think I have FH?	08
How is FH diagnosed?	10
How early can FH be diagnosed?	10
More about FH and genetics	11

Living with FH

How can LDL-cholesterol be reduced?	12
A healthy lifestyle	13
A guide to healthy eating	14
Healthy fat swaps	15
Cholesterol lowering foods	16
Medicines to lower cholesterol	18

Understanding Cardiovascular Disease (CVD)

What is cardiovascular disease?	19
Cardiovascular risk factors	20
What are lipoproteins?	21

Guide to medical terms

HEART UK - The Cholesterol Charity	24
------------------------------------	----

Familial Hypercholesterolaemia

What is familial hypercholesterolaemia (FH)?

Familial hypercholesterolaemia is an inherited condition, where an altered gene causes high blood cholesterol. It is usually referred to as FH.

Familial means that it runs in families and is passed from a parent to their child. Usually it is possible to trace FH over several generations.

Hypercholesterolaemia means high blood cholesterol. The type of cholesterol that is specifically increased in FH is Low Density Lipoprotein-cholesterol (LDL-cholesterol). LDL-cholesterol floats in the blood stream and transports cholesterol around your body to the cells that need it. We talk a lot about LDL-cholesterol in this booklet.

FH is one of the most commonly inherited conditions. As many as one in 250 people may have an altered gene that causes FH.

FH is linked to a higher risk of very early cardiovascular disease. Cardiovascular disease means problems of the heart or blood vessels caused by a build-up of fatty deposits inside the arteries and a higher risk of blood clots. The risk from FH varies from family to family and from person to person within the family.

Your risk of cardiovascular disease depends upon your cholesterol level, other inherited factors, diet, smoking, level of physical activity, and whether you are male or female.

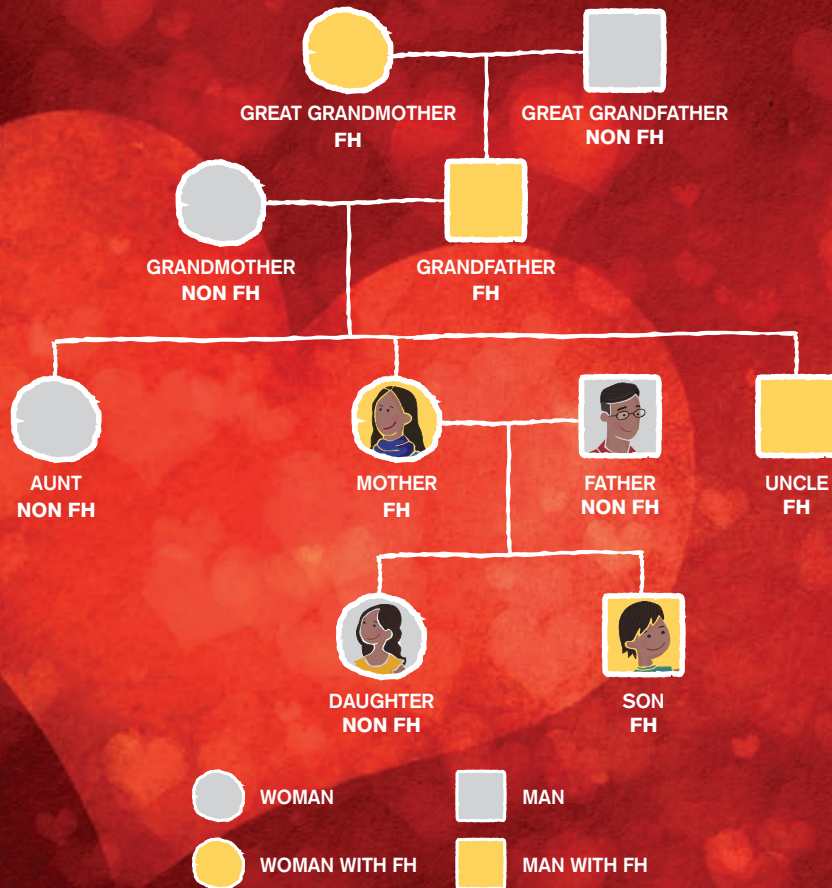


IMPORTANT

Because FH is inherited, it runs in families. It causes high blood cholesterol, high LDL-cholesterol, and an increased risk of early heart and blood vessel disease caused by fatty deposits.

The family tree

FH is an inherited condition which can usually be traced over several generations



What is cholesterol?

Cholesterol is an essential fatty substance that is needed by your body to build cells, make some hormones and to produce bile acids that are needed for digestion.

Cholesterol is carried around the body inside small round parcels of fat and protein. These are called lipoproteins (see page 21).

Most of our cholesterol is carried in Low Density Lipoproteins; doctors refer to these as LDL. The cholesterol that is carried inside LDLs is called LDL-cholesterol.

We all have LDL-cholesterol, travelling around our bodies in our blood, but too much is not good for our arteries (blood vessels). This is because excess cholesterol can become trapped in the walls of blood vessels making them much narrower, a process called atherosclerosis. Atherosclerosis can lead to cardiovascular disease. Find out more about this process on page 19.



IMPORTANT

FH is an inherited condition.

If you have FH it is likely that other people in your family will have FH too.

If a parent has FH there is a 1 in 2 chance that each of their children will have FH too.



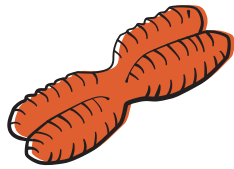
IMPORTANT

LDL transports cholesterol in your blood to your cells. Cholesterol is needed to build new cells, make hormones and bile acids. If there is too much LDL-cholesterol it can get stuck inside the walls of your blood vessels and cause narrowing. This process is called atherosclerosis.

What causes FH?



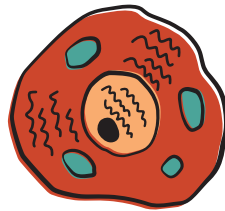
DNA



Chromosome



Cell nucleus



Cell

Before we explain what causes FH it is helpful to know how genetic conditions are passed down through families.

Our genetic code

The features that we inherit from our parents are passed down through DNA; our genetic code. DNA is arranged in genes which sit on chromosomes. Chromosomes live inside the cell nucleus.

Genes give instructions for how our bodies are built, what they look like and how they work. A small change in just one of the building blocks of DNA, within a specific gene, can result in a medical problem.

What is different in FH?

In FH there is a change in one of four genes, all of which give instructions to make the proteins involved in the removal of LDL-cholesterol (by LDL receptors) from the blood.

This means that people with FH find it more difficult to remove surplus cholesterol from their blood.

LDL receptors

LDL receptors play a vital role in keeping our cholesterol levels in balance. These receptors sit on the outside of many kinds of cells but especially liver cells. They pick up cholesterol rich LDL particles from the blood and take them into the cell. Once inside the cell, LDLs are broken down to release their cholesterol. The cholesterol is then either used by the cell, stored, or removed (excreted) by the liver in the bile. After the LDL receptors release their LDL, they are recycled back to the cell surface to pick up more LDLs.

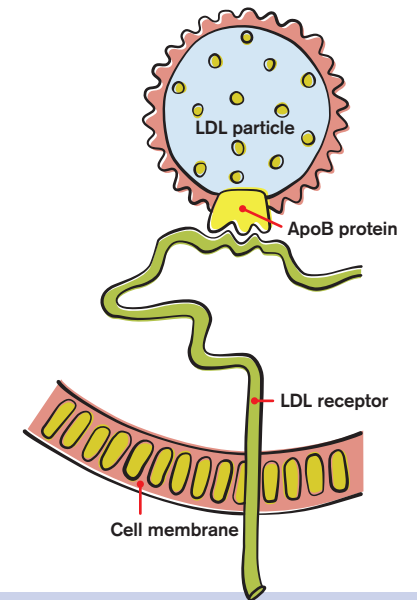
LDL receptors and FH

In FH there are fewer working LDL receptors. This means that people with FH cannot remove cholesterol from the blood as well as people who do not have FH.

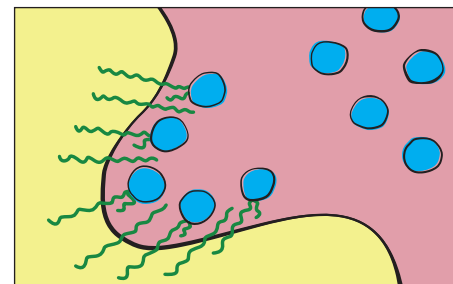
Genetics and FH

Most people with FH have an altered LDL receptor gene. Only a few people have FH because of a problem with the APOB, PCSK9 or APOE gene.

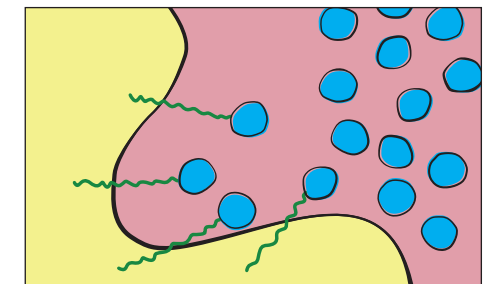
These “genetic spelling changes” result in LDL receptors that either don’t work very well or don’t work at all. This, in turn, limits the amount of cholesterol that can be removed from the blood.



The LDL receptor is attached at one end to the cell membrane. The other end binds to a special protein on the outside of the LDL particle called apolipoprotein B or ApoB.



Normal liver cell



A liver cell in someone with FH. In FH there are fewer working LDL receptors.



IMPORTANT

Genes give instructions for how our bodies are built and work. LDL receptors catch and remove cholesterol from the blood. In FH there are fewer working LDL receptors.



IMPORTANT

FH is caused by a change in one of four genes which are responsible for proteins that remove LDL-cholesterol from the blood. The most common change is in the LDL-receptor gene. A small number of people with FH have an alteration in the APOB, PCSK9 or APOE gene.

Why does my doctor think I have FH?

Your doctor may suspect FH if you, or other close family, have very high cholesterol; especially if there is unexplained early heart disease in your family. Your doctor may ask you more questions about your family, arrange for you to have a blood test called a lipid (blood fat) profile and do a physical examination. They may arrange for you to have a genetic test.

A lipid profile is needed to assess the levels of various lipids (blood fats) and lipoproteins in your blood. The results tell you and your doctor how much total cholesterol, LDL-cholesterol, HDL-cholesterol, non-HDL cholesterol and triglycerides you have in your blood.



Your doctor may think you have FH if:

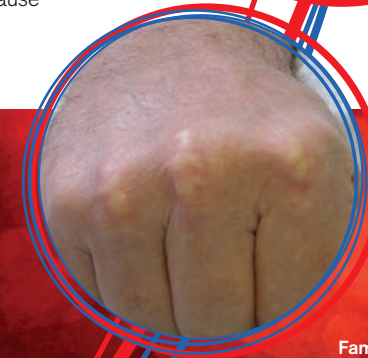
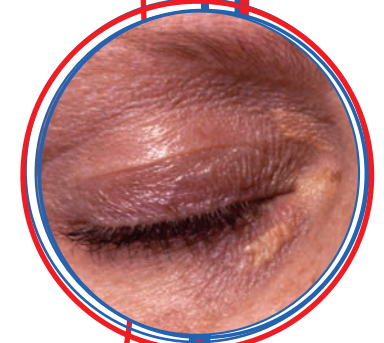
- there is a history of early cardiovascular disease in close family members
- you are a male and have had a heart attack before the age of 50, or female and have had a heart attack before the age of 60
- your cholesterol is very high
- other family members have been diagnosed with FH
- you have any physical signs of high cholesterol (see below)

Signs of FH

Some people with FH have physical signs on the outside of their body. These include swollen tendons on the heels and knuckles of the hands (tendon xanthoma) or yellowish patches around the eyes (xanthelasma). A white deposit of cholesterol in the shape of an arc (corneal arcus) may also be seen at the edge of the coloured part of the eye. Tendon xanthoma only occur in people who have inherited a condition that causes high cholesterol. Corneal arcus and xanthelasmas may happen for other reasons.

Diagnosing FH early

Diagnosing FH at a young age is important. Treatment works better when started early and before the high levels of cholesterol can cause damage to your blood vessels.



IMPORTANT

FH is suspected in people who have early cardiovascular disease and high cholesterol levels and who have close relatives with these conditions. Brothers, sisters and children of people already diagnosed with FH should all be assessed.

How is FH diagnosed?

Doctors diagnose FH by looking for unusually high LDL-cholesterol, finding fatty bumps under the skin and by checking for early heart disease in you or your close family. Together these can suggest definite or possible FH.

FH can also be confirmed by a genetic test, but this is not available everywhere. The test looks for a “disease causing” alteration in the LDL receptor, APOB, PCSK9 or APOE genes. If you are the first person in your family to have a genetic test for FH it can take about 3 months to get a result.

If you are diagnosed with FH, it is likely that some close family members (parent, brothers, sisters, children) will have FH too and they will need to be checked. Doctors refer to this as cascade testing - it is one of the ways we can find FH before it causes a problem.

How early can FH be diagnosed?

Babies born to a parent with FH have a 1 in 2 chance of inheriting FH. These children should be tested for FH at the earliest opportunity and ideally before their 10th birthday.

Diagnosing FH at a young age is essential. Eating well and being active are important from a young age and can help reduce the impact of FH in later life.

Looking for a genetic alteration in a family member is much easier to do because doctors already know the gene change they are looking for. The results are usually available within two to three weeks.

What are the benefits of genetic testing

FH cannot skip a generation. However sometimes a person with the altered gene will not show the typical pattern of raised cholesterol we would expect of someone with FH. This may be because background genetic factors and a healthy diet and lifestyle mask the condition. However because they have the altered gene they can still go on to have a child with FH. This is why experts prefer to be able to confirm a diagnosis with genetic testing and why genetic testing is so helpful as it allows you to track FH through the family.

More about FH and genetics

Homozygous FH

Rarely (up to 1 in 250,000 births), someone inherits an altered gene from both parents. This results in very, very high cholesterol levels and a severe form of FH. Medicines and dietary change may not be enough to reduce the extremely high cholesterol levels. One way to treat this form of FH is by mechanically removing LDL-cholesterol from the blood using a dialysis-like cleansing technique called LDL apheresis.

If you have inherited two identical copies of the same altered FH causing gene doctors refer to this as homozygous FH. Compound heterozygous FH is when you have inherited one copy of two different FH causing genes.

Variants of unknown significance (VUS)

Sometimes a genetic test for FH may find a “variant of unknown significance” or VUS. This means that a piece of the LDL receptor, APOB, PCSK9 or APOE gene looks different from the way it is expected to look, however researchers don't yet know if this change is harmless or can cause FH. If you have a VUS your health professional will talk to you about what this means for you and your family. Usually a few affected family members will need to be identified with the same VUS before it is possible to agree whether this new change is disease causing or not.

What if my genetic test is negative for FH?

Sometimes a gene alteration cannot be found in someone with unusually high cholesterol levels. It is important to understand what this means. It may mean that you do not have FH. Alternatively, it might mean that the laboratory has just not been able to identify a known gene alteration in your DNA.

Any risk factors you have will still need to be managed. So if the genetic test was recommended because of unusually high cholesterol levels, your doctor will talk to you about what to do to lower these.



IMPORTANT

FH is diagnosed by finding a combination of unusually high cholesterol levels, cholesterol lumps under the skin and family history of early heart disease and where possible through genetic testing. Genetic testing is not available everywhere.



IMPORTANT

In families where FH has been diagnosed, children should be checked for FH as early as possible and before they are 10 years old. All children, but especially children diagnosed with FH, should be encouraged to eat healthily from an early age.

How can LDL-cholesterol be reduced?

You can keep your cholesterol levels low by eating well, being active and by taking any medicines or treatments that your doctor prescribes. It is also important not to smoke.

There are three main steps:

Step 1: A healthy lifestyle

Step 2: A healthy diet

Step 3: Medication

Keeping a healthy body weight

Many people struggle with their weight, yet we know that being a healthy weight has many health benefits. In practice it is much easier to prevent weight gain than to lose it.

People who have a lot of fat around their waist (apple shaped) are particularly at risk of heart disease. Excess weight can also increase your cholesterol and triglyceride levels and your chances of developing type 2 diabetes.

If you gain weight easily try to:

- Opt for smaller portions
- Fill half of your plate with vegetables
- Use a smaller plate, bowl, glass
- Be more active
- Avoid sugary and fatty snacks
- Choose foods that fill you up
- Check food labels
- Get help, advice or support



A healthy lifestyle



If you are an adult with FH it is OK to have a moderate amount of alcohol. This means no more than 14 units per week and having some alcohol free days each week. Alcohol should be consumed with caution if your blood triglyceride level is high.



For adults at least 150 minutes of activity (or 75 minutes of very vigorous activity) is recommended each week. Children should be active for at least an hour a day.



On average smokers die 15 years earlier than non-smokers. People with FH who smoke are at very high risk of cardiovascular disease.



IMPORTANT

There is a lot you can do to help keep you and your family healthy. This includes eating healthily, being active and taking any medicines your doctor prescribes.



IMPORTANT

On average smokers die 15 years earlier than non-smokers. People with FH who smoke are at very high risk of cardiovascular disease.

A guide to healthy eating

Healthy eating is important for everyone, not just people with FH. It gives you all the energy you need for everyday activities, the nutrients you need for growth and repair and helps you to stay strong and healthy.

The diet we recommend for people with FH is the same as we recommend to everyone. It's about eating fewer processed foods and having more fruit, vegetables, wholegrains, nuts, and low fat dairy foods. It is also important to have the right balance of fats and to avoid too much sugar, salt and alcohol.

One key step is to replace foods that are high in saturated fats with foods that are either low in fat or contain heart healthy unsaturated fats. See page 15 for how to do this.



Small simple changes

For most people it is best to start by making small changes to your diet and building these up over time. It can take a lot of effort and you may need regular help and advice from a dietitian or from your doctor.

What you eat on a regular basis is what matters. Remember a healthy diet for people with FH is good for everyone whatever their age, so try involving the whole family in any changes you make.

Do I need to limit cholesterol in food?

Cholesterol is only found in foods that we get from animals, so there is none in fruit, cereal grains, vegetables, seeds, nuts and pulses.

For most people it is not necessary to limit your intake of cholesterol containing foods like egg yolk and shellfish, but you should discuss this with your doctor or dietitian. Some foods - offal, fatty meat, full fat milk, cream, cheese and butter - also contain cholesterol. If you are making some of the fat swaps opposite you will already be limiting many of these foods.

Healthy fat swaps

EAT LESS	SWAP FOR
Butter, ghee, coconut and palm oil, lard, suet, hard margarines.	Vegetable spreads, vegetable oils such as olive, rapeseed, sunflower and soya.
Fatty meat and processed meat products such as sausages, fatty bacon, salamis, canned meat.	Lean meat, chicken or turkey with skin removed, white fish, oily fish at least once a week. Have meat-free days - try dishes based on beans, pulses, Quorn, tofu, nuts or soya meat alternatives.
Full fat dairy foods (milk, yogurt, cream, cheese).	Lower fat milks such as semi-skimmed, 1% fat or skimmed milk and calcium fortified soya alternatives to milk. Low fat yogurts. Low fat cheese such as half fat cheddar and cottage cheese.
Cakes, sweet, filled or coated biscuits Coconut - fresh, dried and desiccated.	Plain buns such as currant/hot cross buns, scones, semi-sweet biscuits.
Cream or pastry based desserts.	Fresh, baked or poached fruit, milk puddings and custard made with low fat milk, low fat yogurts/desserts, fruit crumbles made with unsaturated spread.
Pastry, sausage rolls, savoury pies.	Potato topped pies.
Cream-based curries e.g. kormas. Cheese and cream based pasta dishes. Extra cheese or meat topped pizzas, sandwiches with cheese fillings.	Tomato and vegetable based curries and pasta dishes. Thin crust pizzas with vegetable toppings, sandwich fillings such as hummus, lean chicken, egg salad and falafel.
Roasting or frying with butter, lard, other animal fats or coconut oil.	Use small quantities of vegetable oil or try other cooking methods e.g. casseroles, boiling, grilling, steaming, roasting bags.
Milk chocolate, toffee, fudge, crisps and fried salty snacks.	Dark chocolate, chewing gum, nuts, seeds, popcorn. Lower fat crisps or baked savoury snacks.



IMPORTANT

Five important guidelines for a heart-friendly diet:

1. Replace saturated fats with unsaturated fats
2. Eat more wholegrains, nuts, pulses, vegetables and fruit every day



3. Choose meat free meals (fish, nuts, pulses, soya, quorn) more often
4. Limit food and drinks high in sugar or alcohol
5. Cook more meals from scratch and limit takeaway and processed foods

Cholesterol lowering foods

Cholesterol lowering foods

There are some key foods that can help to lower cholesterol. You can read more about these on our website and in our special cholesterol lowering plan called the UCLP® or Ultimate Cholesterol Lowering Plan®.

These include:

Nuts – all kinds, such as almonds, Brazils, hazelnuts, peanuts, pecans, walnuts

Oats and Barley – porridge, oat based cereals, pearl barley, healthy recipes including oats or barley

Do I need to buy foods fortified with plant sterols and stanols?

Plant sterols and stanols are very similar to cholesterol in their shape, size and make up but they are only found in plant foods. They have been shown to lower cholesterol if eaten in the right amounts and every day. They work by reducing the amount of cholesterol our bodies can recycle. We get a small amount of plant sterols from the plant foods we eat but this is not enough to lower our cholesterol. So food companies have developed special foods that have added plant sterols or stanols in them. These foods can usually be found in the chiller cabinet as mini yogurt drinks, spreads, milks and yogurts.

If you take these foods remember to have them with a meal and aim for around 2-3g per day.

Salt

Most of us are eating too much salt, which increases blood pressure and the risk of a stroke, heart attack and heart disease. Salt is found in many of the foods we buy, including ready meals, processed foods, cooking and table sauces.

Sugar

The sugar in fruit juice, smoothies, fizzy and soft drinks, confectionary, syrups and added as an ingredient to foods and drinks or in recipes like cakes, biscuits and desserts is best limited. This kind of sugar is now referred to as “free sugars”.

Experts recommend that adults should have no more than 30g of free sugars per day. Children should have less than this.

Your doctor, or dietitian, may suggest you limit sugar and sugary foods as much as possible if the level of triglycerides in your blood is found to be too high.

Is diet enough to lower my cholesterol?

A change in diet is the first step to reducing cholesterol levels. Most people will see a drop in their cholesterol but the amount will depend on the individual and how many changes they can make to their diet.

We talk about medicines and treatments for FH on page 18.



IMPORTANT

Oats, barley and nuts have been shown to actively reduce cholesterol if eaten regularly. Foods fortified with plant sterols or stanols can also lower cholesterol but they are not a replacement for medicines.

Most people with FH do not have to avoid eggs or shellfish.



Dietary changes can lower LDL- cholesterol by 10-15%. For most people with FH this will not be enough and a change in diet should be combined with cholesterol-lowering medication.

Understanding Cardiovascular Disease (CVD)

What is cardiovascular disease?

Cardiovascular disease is often shortened to CVD.

It is the name for diseases of the heart and blood vessels such as heart disease, heart attack, stroke and angina.

CVD is caused by atherosclerosis

Atherosclerosis involves a build up of cholesterol rich fatty deposits (called plaques) inside your blood vessels. These deposits cause your blood vessels to narrow and lead to reduced blood flow.

How does it happen?

Atherosclerosis is a slow process, but it can begin very early in life. The speed at which it progresses depends upon the number of risk factors that you have. It starts when cholesterol-filled cells become trapped in the inner wall of damaged blood vessels (see illustration on page 20). This causes inflammation, the build up of more cells, further deposits of cholesterol, the formation of scar tissue and hardening, resulting in the formation of "plaque".

Angina

Plaques can narrow the blood vessels and reduce the flow of blood to the heart and other organs. Plaques that form in the small arteries that supply the heart muscle can limit the blood supply to the heart. This can result in pain in the chest, arms, shoulders and neck especially when you exercise or are stressed. It can be uncomfortable or painful. This is angina.

A heart attack

Plaques can burst resulting in damage to the inside of the blood vessel, and the formation of a blood clot, which can severely restrict or block the blood supply. This causes an immediate lack of oxygen to the part of the organ supplied by the blood vessel. When this happens in the heart it is called a myocardial infarction, MI or heart attack.

It is important to restore blood flow as quickly as possible after a heart attack to limit any damage. This can be achieved with blood clot-dissolving drug treatment, a direct mechanical removal of the blood clot using a catheter (a small tube designed to go into blood vessels) and by increasing the size of the blood vessel with a small balloon that is expanded inside the vessel, followed by insertion of metal netting in the blood vessel (stenting). These methods are often combined.

Medicines to lower cholesterol

Statins

Most people with FH will also need to take a medicine, called a statin, to help keep their cholesterol levels low. Doctors usually aim to lower the amount of cholesterol in the blood by half; but you should discuss this with your doctor. Your target cholesterol levels will depend upon when you were first treated, how old you are, how high your cholesterol is, your family history and any other risk factors you have.

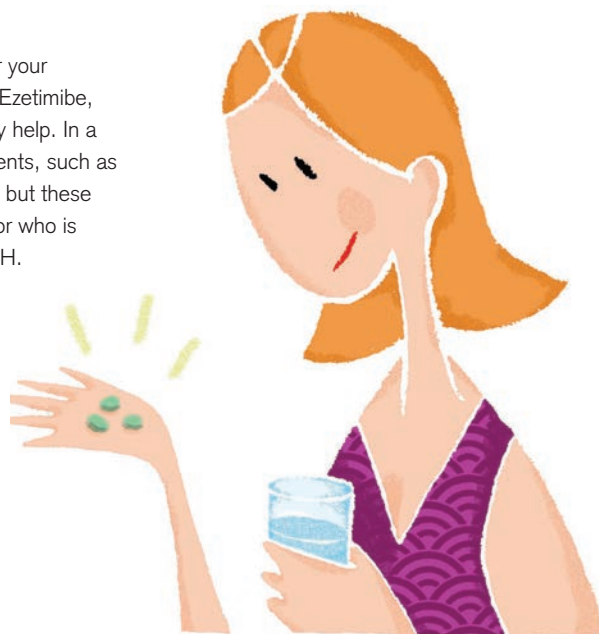
Ezetimibe

In some cases a statin may not lower your LDL-cholesterol enough and adding Ezetimibe, a cholesterol absorption inhibitor, may help. In a small number of people other treatments, such as resins or fibrates may be considered, but these are usually only prescribed by a doctor who is experienced in treating people with FH.

PCSK9 inhibitors

A new medicine called a PCSK9 inhibitor is now available for some people with FH. It is usually only given if statins and other medicines fail to lower your cholesterol enough.

Cholesterol can also be lowered mechanically by removing it from the blood using a dialysis-like cleansing technique called LDL apheresis. Only a few people with FH will need this type of treatment.



IMPORTANT

Medicines that lower cholesterol include statins, cholesterol absorption inhibitors, resins and PCSK9 inhibitors. Your doctor might prescribe one or more of these types of medicines. Your medication, a healthy lifestyle, and a heart friendly diet need to be continued throughout life.



IMPORTANT

Heart attack and stroke are the end result of atherosclerosis. Atherosclerosis is a hardening and narrowing of the blood vessels caused by a build up of cholesterol and inflammation to form plaque. Plaque decreases the size of the inside of a blood vessel. A damaged plaque can cause a blood clot, with very rapid narrowing or blockage of the blood vessel.

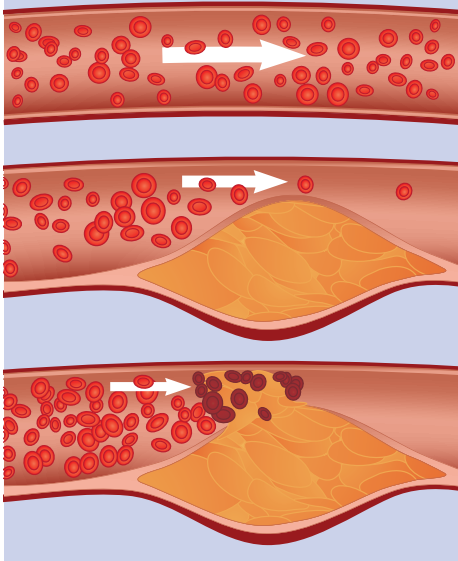
Cardiovascular risk factors

Cardiovascular risk factors are things that we know increase the chances of a person developing cardiovascular disease.

Some people are more likely to develop cardiovascular disease during their lifetime than others. This is because they have more risk factors.

The development of atherosclerosis

Cross-section of blood vessels in three stages showing the build up of cholesterol and the development of atherosclerosis.



Some risk factors such as your age or being male cannot be changed.

Other risk factors such as high blood pressure, high cholesterol, smoking, being overweight, having diabetes or being inactive can be changed or managed.

People who are diagnosed with FH should be seen by a specialist doctor. They will talk to you about how best to manage your high cholesterol and any other risk factors that you have. It can be helpful to see a dietitian too; they will be able to offer advice and guidance on everyday eating.

Is it possible to reduce the risk of cardiovascular disease in FH?

YES! Studies have shown that a drop in LDL-cholesterol can reduce the risk of cardiovascular disease. And, people already showing signs of early cardiovascular disease can reduce their risk of further cardiovascular disease.

It is important to start reducing your LDL-cholesterol as soon as possible, and to reduce any other risk factors you might have. Stopping smoking is crucial for reducing the risk of cardiovascular disease.

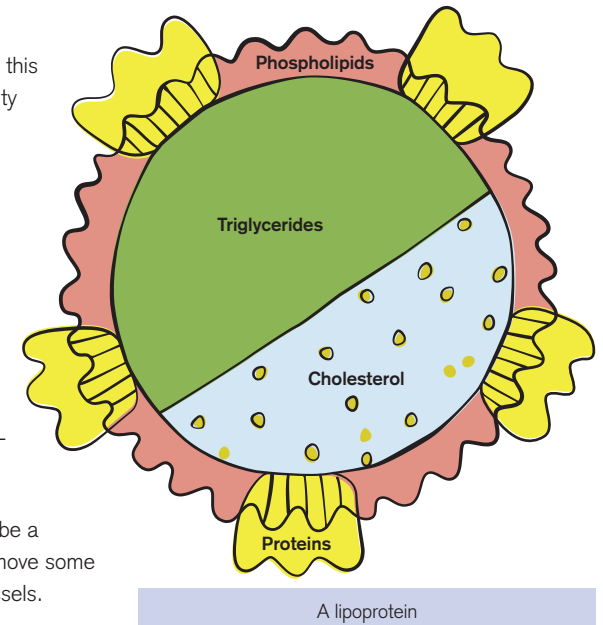
What are lipoproteins?

Lipoproteins are particles that transport fat around in the blood, see illustration. Fatty substances called triglycerides and cholesterol cannot be dissolved in the blood and depend on this transport system to take them from the organs that absorb or produce them (the gut and liver) to the cells.

The two most important lipoproteins in this transport system are called Low Density Lipoprotein (LDL) and High Density Lipoprotein (HDL). The cholesterol carried on these lipoproteins is referred to as LDL-cholesterol (LDL-C) and HDL-cholesterol (HDL-C). Chylomicrons and VLDL are two other lipoproteins.

The cholesterol transported in HDL is often called “good” cholesterol. One of the important functions of HDL is to transport excess cholesterol from the cells and tissues back to the liver. So having a low HDL-cholesterol can be a problem. HDL may also be able to remove some cholesterol from the walls of blood vessels.

LDL-cholesterol is often referred to as “bad” cholesterol. One of the aims of treatment is to keep the levels of LDL-cholesterol as low as possible.



IMPORTANT

Risk factors such as high LDL-cholesterol, smoking and high blood pressure increase the risk of developing atherosclerosis and heart disease. You can reduce your risk factors by eating a healthy diet, being more active and by taking the medicines your doctor prescribes.



IMPORTANT

Lipoproteins transport fatty substances, such as cholesterol and triglycerides, in the blood. Two important lipoproteins are LDL and HDL. The cholesterol that is carried on LDL is often referred to as “bad” cholesterol.

Guide to medical terms

ApoB: The LDL particle has a specific protein attached to it named Apolipoprotein B or ApoB. ApoB acts as a bridge between the LDL particle and the cells in your body that carry the LDL-receptor.

Bile and bile acids: The liver produces bile acids from cholesterol. These bile acids are excreted into the gut (as bile) when we eat. Bile is needed to help us digest the fat in our food.

Cell: Cells are the building blocks of the body and can be compared to building bricks. The body is made up of approximately 100,000,000,000,000 (100 billion) cells.

Cholesterol is an essential fatty substance used to build cell walls. It is also needed to make hormones, vitamin D and bile acids. All cells can produce cholesterol but most cholesterol is made in your liver and gut. The liver is also the main organ for breaking down cholesterol by converting it to bile acids. If your body makes excess cholesterol, absorbs too much, or cholesterol is broken down too slowly there will be excess cholesterol in your blood.

Chromosomes: The hereditary material present in the nucleus of each cell.

Chylomicrons: Large lipoprotein particles that transport lipids from the gut to cells.

Dietitian: A health professional who is an expert in diet and health. They will have undergone 4–5 years of university study.

DNA: A string of molecules. Genes are made from DNA.

Familial hypercholesterolaemia (FH): A condition passed down through families. It results in high blood cholesterol.

Gene: A section of DNA that gives instructions for building a certain protein.

HDL-cholesterol: Also referred to as “good” cholesterol. It can be a problem if you have too little of this in your blood.

Trans fat/partially hydrogenated fat: A type of fat found in food products. This fat starts as an unsaturated liquid fat and then is industrially hardened. The UK food industry uses less of this fat than it used to. Trans fats increase bad cholesterol and lower good cholesterol.

LDL-cholesterol: Otherwise referred to as “bad” cholesterol. It is best to only have a small amount of LDL-cholesterol in the blood.

Lipids: Lipids are the name given to fatty substances, such as cholesterol and triglycerides, in the body. Your doctor can find out your levels of cholesterol and triglycerides by doing a blood test.

Lipoproteins: Lipoproteins are small transport packages made up of cholesterol, triglycerides and proteins which circulate in the blood. There are various kinds of lipoproteins, the most important being HDL and LDL.

Myocardial infarction (MI): A heart attack. Usually this happens suddenly when the blood supply to the heart is blocked. Causes of myocardial infarction include high cholesterol and smoking.

Receptors: Receptors are located on the outside of the cells. They catch substances in the blood needed by cells. There are special receptors for lipoproteins. People with familial hypercholesterolaemia (FH) often have too few working receptors for LDL (low density lipoproteins). This means that the cholesterol from LDL remains in the blood. Excess circulating cholesterol can then become trapped in the walls of blood vessels, eventually forming plaques which cause narrowing.

Saturated fat: Found mainly in animal foods such as butter, ghee, lard, suet, meat and full fat dairy foods and foods made from these such as cakes, biscuits, pies, pastries, puddings and confectionary. Coconut oil and palm oil are also saturated fats. Eating too much saturated fat results in an increase in blood cholesterol.

Unsaturated fat: Found in vegetable sources such as nuts, seeds and vegetable, nut and seed oils, oily fish and avocado. We should replace saturated fats with these.

Triglycerides: Another word for fats. They are found in foods and in the blood. It is good to have low levels of triglycerides in the blood.

VLDL: When fat from the gut reaches the liver it is packed into large, fat rich particles, which are called VLDL (very low density lipoprotein).



IMPORTANT

HEART UK – The Cholesterol Charity – provides expert support, guidance and education for people with FH and their families. Please call our Cholesterol Helpline or visit our website.

T: 0345 450 5988 E: ask@heartuk.org.uk W: www.heartuk.org.uk



IMPORTANT

HEART UK is working hard to improve the number of younger people who are diagnosed with FH so that they can benefit from early treatment.

HEART UK – The Cholesterol Charity

We hope you have learnt more about familial hypercholesterolaemia (FH), an inherited condition that runs in families and is caused by an altered gene.

HEART UK is the only charity in the UK dedicated to providing expert support, guidance and education for people with FH.

Call/email our Cholesterol Helpline

0345 450 5988/ask@heartuk.org.uk

A free and confidential service offering information on cholesterol and an opportunity to talk things over with qualified nurses and dietitians.

Donate

Please help to fund our work. All our materials are free, but you can make a donation towards our work via our website.

Raise funds

There are many ways you can raise funds for HEART UK. Visit our website for more information.

National Cholesterol Month

October is National Cholesterol Month – a whole month devoted to raising awareness and funds for HEART UK.

Literature orders

Call 01628 777046 or email us on literature@heartuk.org.uk

General enquiries

01628 777046

Go online

Advice and lots more information is available on our website. Check out our frequently asked questions, recipe ideas, what your cholesterol numbers mean and sign up to our free monthly e-newsletter for more facts and tips to help manage cholesterol.

Visit our website

You can find out more on our website at www.heartuk.org.uk

Written by HEART UK with support from our wonderful patient ambassadors.