British Heart Foundation

LIFE WITH FAMILIAL HYPERCHOLESTEROLAEMIA

I love cycling and walking

I'm taking on half marathons

I'll never say no to a challenge

I live with an inherited condition

DAVID'S STORY, PAGE 10

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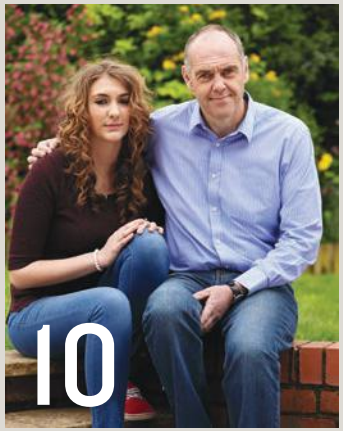
THE CHOLESTEROL CHARITY



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The illustrations used in this booklet are artistic impressions and are not intended to accurately depict the medical material that they represent.

This booklet does not replace the advice that your doctor or specialist may give, but it should help you to understand what they tell you.

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You may be reading this booklet because you've been diagnosed with the inherited condition familial hypercholesterolaemia (pronounced hyper-cholesterol-ee-me-ah) – usually called FH for short. Or maybe your doctor has suggested that you should have some tests to find out if you've inherited this condition because someone else in your family has been diagnosed with it.

FH is a condition that affects the level of cholesterol in your blood. Cholesterol is a waxy substance that's needed to help grow and repair cells in your body. FH is passed from parent to child and can affect one or more family members. Not all family members are affected, but it's very important to find out which ones are.

In most cases, having FH does not affect your quality of life and many people remain unaware that they have it. However, having too much cholesterol in your blood can increase your risk of getting coronary heart disease, which can lead to a heart attack. A heart attack can sometimes be fatal.

It's important that families who are affected by FH receive an accurate assessment, diagnosis, treatment and on-going support, from specialists at a lipid clinic.

This booklet:

- describes how the normal heart works
- explains what FH is and what can go wrong if you have the condition
- explains why it's important that close blood relatives of someone with FH have an assessment
- describes the tests your doctor may ask you and your close family members to have
- describes the treatments you may need
- provides information on how you can live a healthy lifestyle
- provides support for parents.

We explain the medical and technical terms as we go along but, if you find a word you don't understand, look it up in the list of Technical terms on page 60.

This booklet has been produced with the help of doctors and other health professionals. We hope that it will help you to understand your condition and what it means for your close family. If you need further support or information on FH, you can contact HEART UK – The Cholesterol Charity or the British Heart Foundation Genetic Information Service. (See pages 62-63 for all contact details).

UNDERSTANDING YOUR HEART

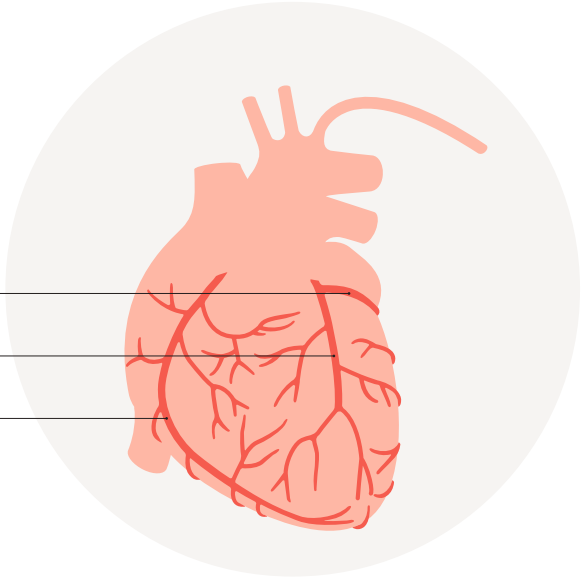
HOW YOUR HEART WORKS

To help you to understand why people with FH have an increased risk of getting heart disease, it helps to know how your heart works and what causes heart disease.

Your heart is a muscle that pumps blood around your body, delivering nutrients and oxygen to all your cells. The muscle of your heart needs its own supply of oxygen and nutrients so that it can pump blood around your body.

Your heart muscle gets its blood supply from the coronary arteries. There are three main coronary arteries – the left and right coronary arteries and the circumflex artery – on the outside of the heart. These divide many times so that the blood reaches all the parts of your heart's muscular wall.

The coronary arteries



CORONARY HEART DISEASE

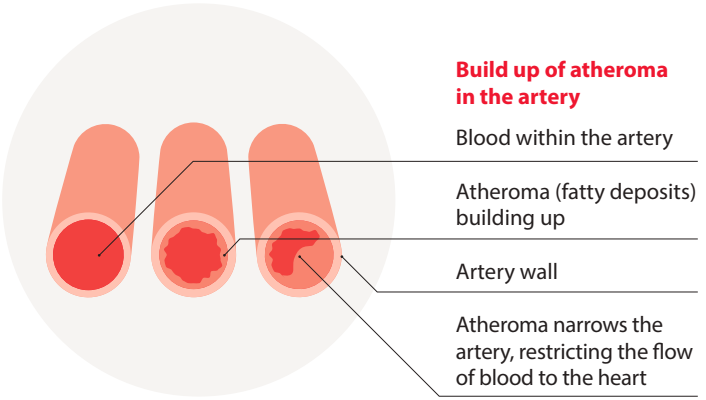
Your coronary arteries are small but play a vital role in keeping your heart healthy and pumping properly. Sometimes – either because of your lifestyle or because your family has a history of heart problems – the coronary arteries can become narrowed because fatty deposits called atheroma have built up on the inside of the artery walls. This process, shown in the diagram below, is called atherosclerosis and it is what causes coronary heart disease.

When your coronary arteries are affected in this way, they may become so narrow that they cannot deliver enough blood to your heart muscle – for example, when you are physically active.

If this happens, you may feel discomfort or pain in your chest, arm, neck, back or jaw. This may be felt in one or more of these places, or it may spread to other areas and is called angina. The amount of pain or discomfort you feel does not always reflect how badly your coronary arteries are affected.

The atheroma in the artery wall can push against the thin inner wall of the artery and cause it to rupture. When this happens, the atheroma pours out into the artery. To stop the atheroma travelling around your body, the blood forms a clot around it. However, the clot may stop the flow of blood through the artery to the heart muscle that it supplies. This is what causes a heart attack. If the blood flow cannot be restored, the muscle will die and stop your heart working properly. Sometimes a heart attack can be fatal.

Build up of atheroma in the artery



WHY DO SOME PEOPLE GET CORONARY HEART DISEASE?

There are certain things about your lifestyle or family history that can increase the risk of getting coronary heart disease. These are known as risk factors. Some risk factors you can change and others that you can't control.

Risk factors you can do something about
You are more likely to get coronary heart disease if you:

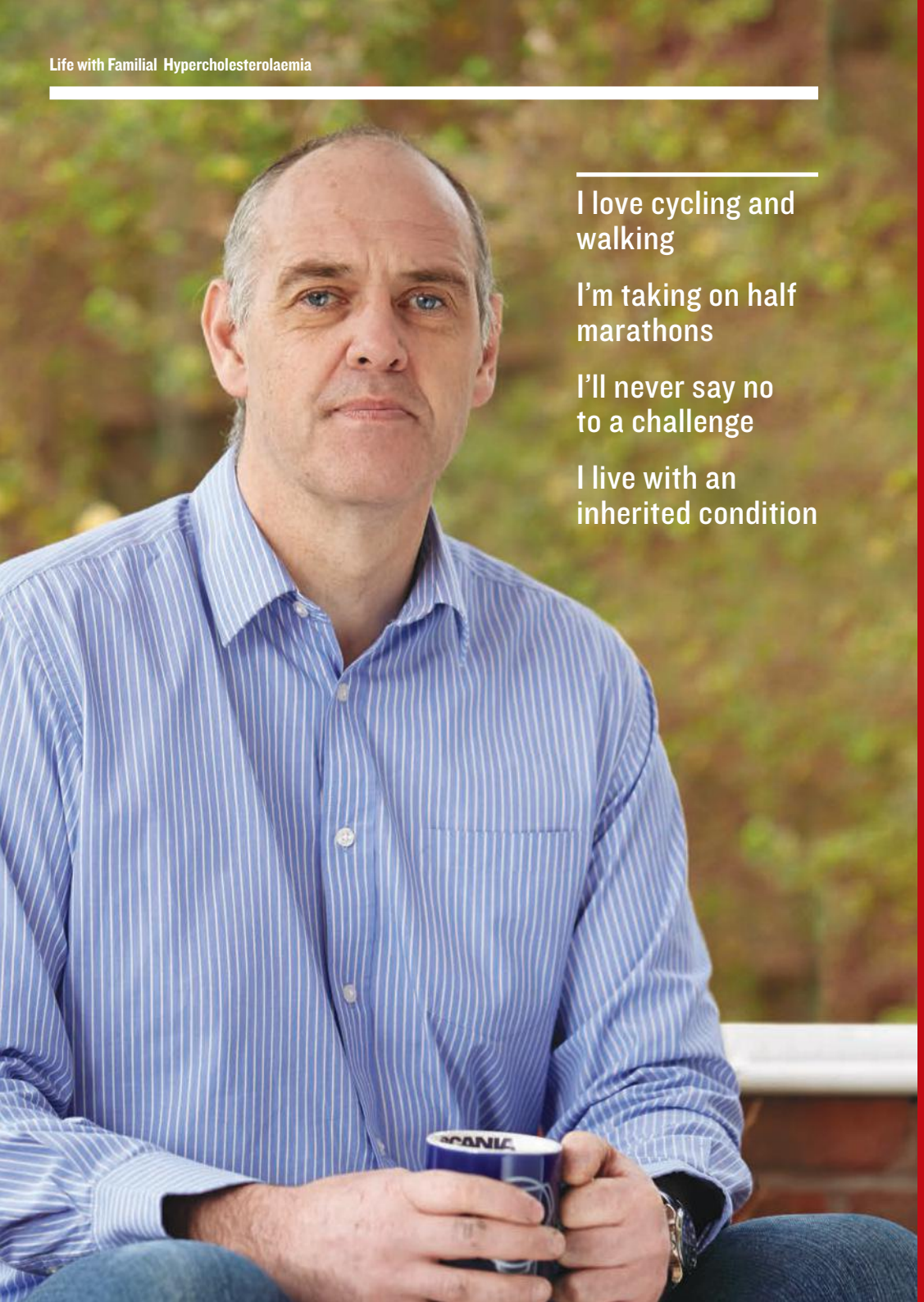
- have a high blood cholesterol level, or have been diagnosed with FH
- smoke any form of tobacco
- have high blood pressure
- don't do enough physical activity
- are overweight or obese
- have diabetes.

Making changes to these risk factors will reduce your risk of getting coronary heart disease.

Risk factors that you can't change
Not all risk factors can be controlled. These include:

- your age – your risk automatically increases as you get older
- your gender – before the age of 60, men are at greater risk than women. If you're a woman, your hormones may give you some protection against coronary heart disease before you reach your menopause. But in the years after your menopause, your risk rises significantly. And by the time you reach your 60s, the gap between men and women narrows
- your ethnic origin – people of South Asian origin living in the UK have a greater risk of getting heart disease than other people in the UK
- a family history of heart disease – this means if your father or a brother has, or had, angina or a heart attack before the age of 55, or if your mother or a sister has, or had, angina or a heart attack before the age of 65.

FAMILIAL HYPERCHOLESTEROLAEMIA (FH)



I love cycling and walking

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DAVID'S STORY

I've always had a healthy diet. I walk and cycle. So when I had a heart attack I was in complete shock. I was treated with a stent for a blocked artery, then referred by a specialist for FH testing. It turned out I had a faulty gene that causes it and that's probably why I'd developed heart disease at a young age. My high cholesterol furred up the arteries to my heart.

Six members of my family were found to have the gene, including my daughter, Bethan. We're now both on statins, we eat healthily and exercise. The good news is my cholesterol has come right down. Thanks to a simple test for FH, it's as if I've been given a second chance in life.

David & Bethan

WHAT IS FAMILIAL HYPERCHOL-ESTEROLAEMIA?

FH, which is short for familial hypercholesterolaemia, is a condition which results in exceptionally high cholesterol levels. It's an inherited condition. This means that it's passed through families and is caused by one or more altered genes. These are often referred to as 'genetic mutations' or 'gene alterations'. About one in every 200 people in the UK has FH, and it is one of the most commonly occurring inherited conditions. In people with FH, high cholesterol levels are usually present from birth and continue throughout life.

If you have FH, you have a higher risk of getting coronary heart disease at an early age. More than 300,000 people in the UK are believed to have FH. However, most of these people don't know they have the condition, putting them at risk of developing coronary heart disease and of having a heart attack. Sometimes it is only after having a heart attack or being diagnosed with coronary heart disease that a doctor may suspect that a person has FH. Diagnosing and treating people with FH early can reduce the number of people who get coronary heart disease or die prematurely.

WHAT IS CHOLESTEROL?

Cholesterol is a waxy substance which is mainly made in your body. It is one of several blood lipids (fatty substances) found in your blood.

Your body makes most of its own cholesterol in the liver. You also get a small amount of cholesterol from some foods such as eggs, liver and kidneys, and seafood such as prawns.

Cholesterol plays a vital role in how every cell in your body works. It's also the material that your body uses to make other vital chemicals such as vitamin D, bile to aid digestion, and some hormones such as cortisol, oestrogen and testosterone. It is especially important for growth when many new cells are being formed very quickly, for example, in children or during pregnancy. However, too much cholesterol in your blood can increase your risk of getting diseases of the heart and circulation.

LDL cholesterol and HDL cholesterol
Cholesterol and other fats have a special way of reaching all the cells in your body that need it. They use your blood circulation as their 'road system' and are carried on 'vehicles' made up of proteins. These combinations of fats and proteins are called lipoproteins.

There are two main types of lipoproteins – LDL (low-density lipoprotein) and HDL (high-density lipoprotein).

- Low-density lipoproteins – sometimes called LDL cholesterol or 'bad cholesterol' – carry most of the cholesterol from your liver, through the bloodstream, to where it is needed. About 70 per cent of the cholesterol in your body is carried by LDL. The lower the density of the lipoprotein, the more fats it contains, so having high LDL is harmful to you.

- High-density lipoproteins – sometimes called HDL cholesterol or 'good cholesterol' – return the extra cholesterol, that isn't needed, from your cells and your bloodstream to your liver for recycling. HDL cholesterol is a 'good' type of cholesterol because it removes cholesterol from your bloodstream. This helps prevent the cholesterol from being deposited in the arteries and causing atheroma (see page 7).

Your total cholesterol level is the total of the LDL, HDL and other fats in your blood.

Triglycerides
Triglycerides are another type of fatty substance found in your blood. You get some triglycerides from foods such as dairy products, meat and cooking oils.

Triglycerides can also be produced in your body, either by your body's fat stores or in the liver. If you are very overweight, eat a lot of fatty and sugary foods, or drink too much alcohol, you are more likely to have a high triglyceride level. Triglyceride levels increase after a meal and are normally cleared from the bloodstream over the following hours. They are usually at their lowest first thing in the morning before breakfast.

WHY IS HIGH CHOLESTEROL BAD FOR ME?

If you have raised cholesterol levels or high triglycerides, you have a higher risk of getting coronary heart disease or having a stroke than people with lower levels. The risk is particularly high if you have a high level of 'bad' LDL cholesterol and a low level of 'good' HDL cholesterol. Your risk increases if you also have other risk factors for heart disease – for example, if you smoke, or if you have diabetes or high blood pressure. Doctors agree that the lower the cholesterol level, the lower your risk of getting diseases of the heart and circulation. See page 34 for target cholesterol levels for people with FH.

IS FH THE ONLY CAUSE OF HIGH CHOLESTEROL?

FH is only one of a number of inherited conditions that cause high cholesterol, but it is one of the most common. Another inherited condition that is a common cause of high cholesterol is familial combined hyperlipidaemia – or FCH for short. FCH affects one in every 200 people in the UK. People with FCH have high levels of both cholesterol and triglycerides in their blood.

For most people, high cholesterol is not caused by inheriting a single altered gene. You may have a high cholesterol level because you:

- eat too much saturated fat
- are overweight and especially if you have a lot of fat around your waistline
- are not physically active
- have an underactive thyroid gland or long-term kidney problems.

Some medicines taken for particular conditions may also affect how your body handles cholesterol.

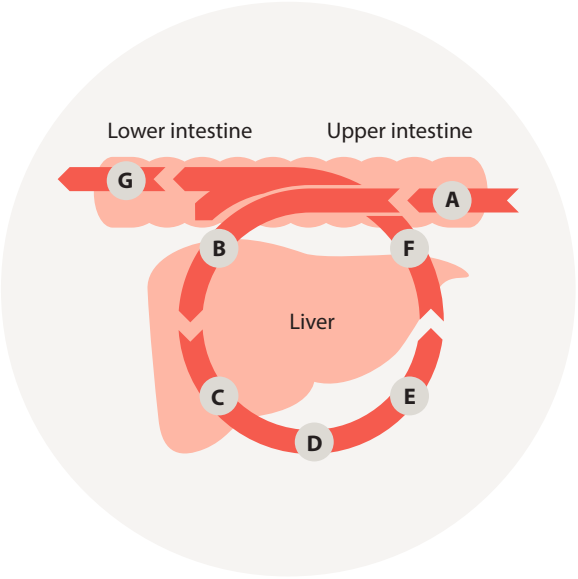
HOW IS CHOLESTEROL CONTROLLED IN YOUR BODY?

All the cells in your body are able to make cholesterol, but most is made in your liver. It's then released into your bloodstream and is transported to every part of your body where it plays a vital role in how all the cells of your body work. The liver also works to remove cholesterol from your body.

Excess cholesterol in your bloodstream is returned to your liver in the form of LDL and HDL cholesterol. To enter your liver, LDL needs to bind to proteins called LDL receptors on the surface of your liver. Once the cholesterol is inside your liver, it can either be recycled back into the bloodstream or broken down into bile acids and these, together with some cholesterol, pass into your intestines. These cholesterol-rich bile acids are essential because they help to emulsify, or mix different fats in your intestines during digestion and help the fats to be absorbed into your body. Some of the bile acids and cholesterol are re-absorbed into your bloodstream lower down the intestines, and the remainder is lost as waste. Any cholesterol that is re-absorbed from your intestine into your bloodstream is transported back to your liver to be recycled.

HDL cholesterol is unique because it is the only lipoprotein that can take excess cholesterol from your cells and transport it back to your liver for removal. Unlike LDL, HDL does not need a receptor to enter the liver.

The diagram below shows a simplified view of how cholesterol is processed in your body.



How cholesterol is processed in the body

- **A** Food enters the intestine and mixes with cholesterol-rich bile
- **B** Fat-rich particles from digestion enter the bloodstream and travel to the liver
- **C** In the liver cholesterol is made from saturated fat
- **D** Cholesterol travels from the liver to the cells, tissues, muscles, organs where it is needed
- **E** Excess cholesterol returns to the liver
- **F** Some cholesterol is broken down to bile acids and together with cholesterol re-enters the intestine
- **G** Some cholesterol-rich bile acids are re-absorbed, the rest are lost as waste

How does FH change the way that cholesterol is controlled in your body?
If you have FH, your liver can't remove enough LDL cholesterol from your blood, and so the level of LDL cholesterol in your blood remains high. High levels of LDL cholesterol can lead to an increase in the amount of cholesterol that is deposited in the linings of the artery walls, leading to atheroma, as described on page 7. While FH results in a high level of LDL cholesterol, other risk factors (see page 8) can influence the effect that LDL cholesterol has on the artery wall. For example, if you smoke, you will have higher levels of chemicals in the bloodstream that make your artery walls more permeable, which makes it easier for LDL to get stuck there.

IS THERE A CURE FOR FH?

There isn't a cure for FH, but it can be managed very successfully. Treatment can significantly reduce your risk of getting coronary heart disease, having a heart attack or needing other treatments. See page 31-42 for more information.

IS THERE A RISK OF SUDDEN DEATH WITH FH?

If you or a member of your family has a heart attack, it's possible that it could be fatal and may be recorded as a sudden death. However, not everyone who has a heart attack dies.

Sometimes a sudden death in a family may alert family members to the possibility of FH or other inherited conditions. If someone has died suddenly – and particularly if they had previously been well – a coroner may encourage close relatives of the person to speak to their own doctor or to the British Heart Foundation Genetic Information Service (see page 62 for details) about being tested for an inherited heart condition.

TESTING, DIAGNOSIS AND YOUR FAMILY

RACHEL'S STORY

I believe my son John and I are alive because I was tested for FH.

A few years ago John had high blood pressure and cholesterol and at times he was passing out. Because he was fit, and only 42, doctors didn't suspect it could be his heart.

My younger sister had a heart attack aged 48. I'd never had a problem with my heart, but being a dairy farmer I know how genes work. I asked my sister's cardiologist if there could be a faulty gene in the family that I had passed to John. He checked me for signs of high cholesterol and sent me to be tested for FH. It came back positive.

I had blocked arteries and needed surgery. It was a huge shock, but now I'm managing my FH and I feel fit and healthy. My results also went to John's doctors and he had more tests and emergency bypass surgery. So the FH test may have saved both our lives.

HOW CAN YOU INHERIT A CONDITION?

Your body is made up of trillions of cells. Each cell has a nucleus, which contains information that makes you unique. This information is your genes. We each have between 20,000 and 25,000 different genes. Genes give the instructions that are needed for development and growth of all the cells in your body, and they determine characteristics like hair colour, eye colour, height and blood type. Each gene provides a code, a set of instructions to produce a specific protein or part of a protein, which will decide on a characteristic or control a particular function in your body.

Genes are arranged end to end along threadlike structures called chromosomes and are made up of a chemical substance called DNA. Each cell usually carries 46 chromosomes arranged in 23 pairs.

You inherit one copy of each chromosome pair from your mother and the other copy from your father. As the chromosomes contain your genes, this means that you also inherit one set of genes from each of your parents. This is why you get certain characteristics from your mother and others from your father.

Your body is made of trillions of cells

Each cell has a nucleus

Each nucleus has 46 chromosomes, in 23 pairs

Each chromosome is made up of a long spiral of DNA

The DNA spiral is divided into genes. You have between 20,000 and 25,000 genes.

It may help to think of your DNA as a book:

- The chromosomes are the chapters. There are 23 pairs of chromosomes, so 23 chapters.
- Each gene is like a paragraph in the chapter. Genes provide the code for proteins, which decide characteristics like hair and eye colour or the ability to control cholesterol levels.
- The code in each gene is determined by a string of DNA. The DNA is like the letters in the words that make up the paragraph. The way these 'letters' are arranged can influence your risk of developing conditions such as high blood pressure or heart disease. For example, the words BARE and BEAR have the same letters, but they're arranged differently so they mean different things. In the same way, if the codes in the DNA are arranged differently, they may work in different ways.

If one of your genes has a 'mistake' in it, a bit like a spelling mistake in a word, it could lead to an abnormal protein being produced in a particular type of cell. The same mistake could be passed on to the next and following generations. This 'mistake' is known as a gene alteration or a genetic mutation. Some 'mistakes' have little or no effect, but others can result in inherited conditions such as FH.

HOW DOES FH DEVELOP?

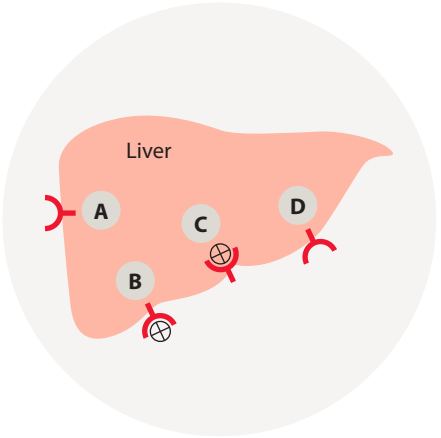
DNA provides the instructions to make the special proteins that help control the amount of cholesterol in your body. FH develops when there is a gene alteration in one of these DNA codes. These gene alterations are present from birth, and result in exceptionally high cholesterol levels. The level of cholesterol in your blood is measured in millimoles per litre, also called 'mmol/l'. In people with FH cholesterol levels are usually between 7.5 and 12mmol/l in adults, but it can be as high as 20mmol/l.

Gene alterations can occur at any point in the code of a gene. So far, more than 200 different gene alterations that cause FH have been found in people from the UK, and more than 1,000 gene alterations have been identified in people with FH around the world. There may be other gene alterations that cause FH that we have not yet identified.

How do the altered genes affect cholesterol levels?
If you've been diagnosed with FH, you may have a gene alteration in one of these three genes:

- the LDL-receptor gene – this gene makes the protein that helps to remove cholesterol from your blood. This is the most common gene alteration in people with FH.
- the APOB gene – this gene makes a protein that helps hold cholesterol-carrying lipoproteins (LDL) together in your blood. If there is an alteration in this gene, the LDL does not bind well to LDL receptors on the surface of your liver. This means that it is only removed slowly from your blood and your LDL level stays high. This happens in two or three in every 100 people with FH.
- the PCSK9 gene – this gene makes an enzyme that controls the removal of cholesterol by breaking down the LDL receptor protein. If there is an alteration in this gene, it means that more LDL receptors are broken down in your liver. This means there are fewer LDL receptors on the surface to remove LDL from your blood, so the level of LDL cholesterol in your blood remains high. This happens in only a small number of people with FH.

These gene alterations mean that your liver is less able to take up excess cholesterol from your blood. In turn, this means that less is passed into your intestines, from where it can be removed from your body. The diagram below shows how normal cholesterol control is affected in someone with FH.



LDL receptor

LDL particle wrapped up by protein

How having FH affects cholesterol levels

A LDL receptor on surface of liver cell – most people with FH have fewer LDL receptors

B LDL protein binds to LDL receptor – in some people with FH the protein binds poorly to the receptor

C LDL removed from blood

D LDL receptor is recycled and it returns to the surface – in some people with FH, LDL receptors are poorly recycled

HOW IS FH INHERITED?

Drawing a family tree will allow your doctor to see if there is anyone else in your family who may have the same condition. The family tree, known as a pedigree, can show a particular inheritance pattern.

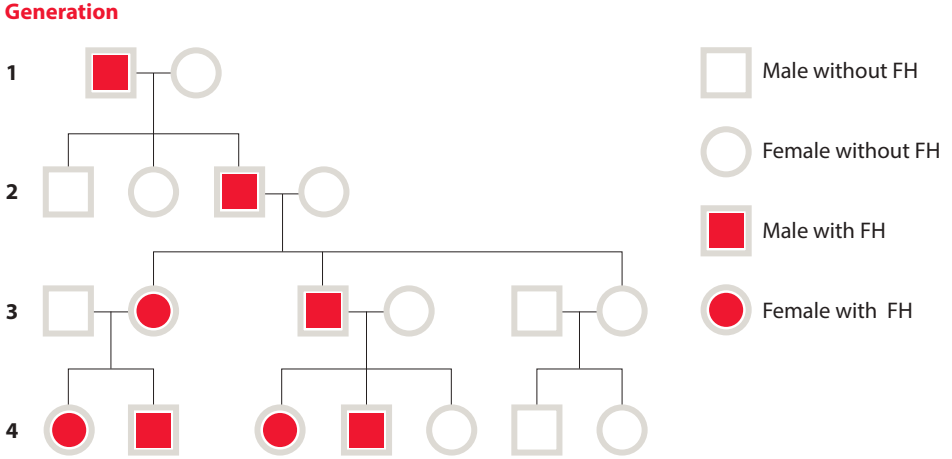
The inheritance pattern for FH is autosomal dominant. This means that each child of a parent with FH has a 50:50 or one in two chance of inheriting the condition. It can affect boys and girls equally and cannot skip a generation. So a child cannot inherit FH if neither parent has it.

As you have two copies of each gene, one from each of your parents, it is possible to inherit an altered gene from either your mother or your father, and in some rare cases from both your parents.

There are three types of FH:

- Heterozygous FH
- Compound heterozygous FH
- Homozygous FH.

Heterozygous FH
Almost all people with FH have heterozygous FH. It happens when you inherit an altered gene from one parent and a normal gene from the other. Each child has a 50:50 or one in two chance of inheriting the altered gene from a parent who has FH. See the diagram below.



Compound heterozygous FH and homozygous FH
Around one in every one million people has compound heterozygous or homozygous FH. If you have these types of FH, you will usually have a much more severe form of the disease than someone with heterozygous FH. Your cholesterol level is often higher than 12mmol/l and, if it's not treated, you may develop heart disease as a child.

Compound heterozygous FH is very rare and only occurs if you inherit two different types of gene alterations, one from each parent. These alterations may be different alterations in the same gene, or alterations in two different genes associated with FH. This type of FH can only happen in a family where both parents have FH. The probability of each child inheriting an altered gene from both parents is almost always one in four, or 25 per cent.

Homozygous FH is also very rare and only happens if you inherit two copies of exactly the same gene alteration from each parent. This is most likely to happen when the parents are blood relatives and have both inherited the original altered gene from a common ancestor.

WHY DOES MY DOCTOR THINK I HAVE FH?

It's not easy to diagnose FH. Your doctor or specialist may suspect FH if:

- a routine blood test shows you have high cholesterol
- you have a heart attack, especially if it happens at a young age
- other members of your family have a history of coronary heart disease as a child (see page 8)
- other members of your family have been diagnosed with FH, or
- you or your doctor have noticed other changes to your body that may suggest you have high cholesterol – for example, tendon xanthomata or corneal arcus. For examples of some physical signs of FH, see pages 26-27.

What will happen if my doctor suspects I have FH?
If your doctor thinks that you may have FH, it's important that you're tested for the the condition. On pages 25-30, we describe all the tests that you may need to have as part of this assessment. Confirming a diagnosis will help the doctors to decide which treatment is best for you and how often you'll need to be followed up.

They will also be able to advise you on what you can do to help yourself and your family to live a normal life.

Your doctor will ask you detailed questions about your family history, arrange for blood tests, and do a physical examination. Your GP may have been the first person to suspect you have FH, but your final diagnosis is most likely to happen at a lipid clinic.

Most lipid clinics take place in a hospital outpatient department and are run by a specialist, sometimes called a lipidologist or cardiologist. Some lipid clinics are held in GP surgeries and are run by a GP with a special interest in diseases of the heart and circulation. Your specialist doctor will be able to make a diagnosis of FH using the family history you provide, and the results of your tests and physical examination. To do this, they will use a set of guidelines called the 'Simon Broome criteria'. We explain this on page 27.

To find out about lipid clinics near you, visit the HEART UK website or call their helpline. You can also call the British Heart Foundation Genetic Information Service. (See pages 62-63 for all contact details).

What do I need to know about my family history?
Your doctor will want to know if any of your blood relations have high cholesterol, or if they have had a heart attack or heart disease. In particular, they'll want to know what age your relatives were when they were first diagnosed with heart disease, developed angina, had their first heart attack or needed other treatment. Your doctor will also want to know if any of your relatives have died from coronary heart disease and at what age.

What blood tests will I need to have?
Measuring your blood cholesterol involves a simple blood test. It can be done in two ways:

- either a blood sample is taken with a needle and syringe and is sent to a laboratory for analysis or,
- a finger prick, also known as a capillary sample, is taken and analysed using a small machine that displays the results after a few minutes.

To make a formal diagnosis of FH, your doctor will usually need a 'full fasting lipid profile'. This means that they will take a sample of your blood using a needle and syringe and send it to be tested for a full range of blood lipids, the fatty substances in your blood. You will be asked not to eat anything, and to drink only clear fluids such as water, or tea and coffee without milk, for 12 hours before your blood is taken for the test. Your doctor will check your total cholesterol level and your LDL cholesterol level against the Simon Broome criteria (see page 27).

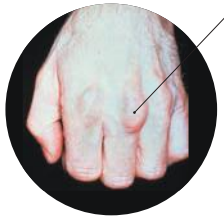
Your doctor may also ask for other blood tests to be done, such as a blood sugar level test to check for diabetes, and also liver and kidney function tests.

Can I use a home cholesterol testing kit?

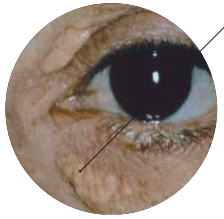
Home cholesterol testing kits are not suitable for diagnosing FH because they are not sensitive enough to measure the different types of cholesterol in your blood, particularly in people with FH. It is important that you are assessed properly in a lipid clinic or by your GP.

What other tests will I have?

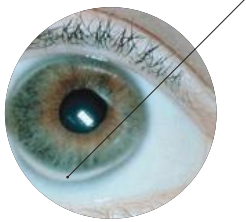
When your doctor examines you, they will also be looking for physical signs of FH. These signs are very easy to miss, so you may not have noticed them yourself. However, if you don't have any physical signs, it doesn't mean you don't have FH. The main signs your doctor will be looking for are:



Tendon xanthomata – Tendon xanthomata (pronounced zan-tho-mata) – sometimes called 'tendon xanthoma' – are swellings in the tendons on the back of your hands and your achilles tendon at the back of your ankle. They are caused by excess cholesterol being deposited in these areas. It's believed that tendon xanthomata are only found in people with FH.



Xanthelasmas – Xanthelasmas (pronounced zan-thal-as-mas) are deposits of cholesterol, usually yellow in colour, in the skin around the bottom of your eye and on your eyelid. If you have these, it may be an indication that you have high cholesterol, but it doesn't mean that you definitely have FH.



Corneal arcus – Corneal arcus is a pale white ring around the outside of your iris, the coloured part of your eye. If you have corneal arcus and you are under 50 years old, it is a strong indication that you may have FH.

Xanthelasmas and corneal arcus can also occur for reasons other than FH.

Simon Broome criteria for diagnosis of familial hypercholesterolaemia

Definite FH is defined as:

a) total cholesterol above 6.7mmol/l or LDL cholesterol above 4mmol/l in a child (a child is aged less than 16 years)

b) total cholesterol above 7.5mmol/l or LDL cholesterol above 4.9mmol/l in an adult

Plus either

c) tendon xanthomas in the patient or close relative (parent, brother, sister, child, grandparent, aunt, uncle)

Or

d) DNA based evidence of an altered gene.

Possible familial hypercholesterolaemia is defined as:

a) total cholesterol above 6.7mmol/l or LDL cholesterol above 4mmol/l in a child

b) total cholesterol above 7.5mmol/l or LDL cholesterol above 4.9mmol/l in an adult

Plus either

c) family history of heart attack below the age of 50 in a second degree relative (grandparent, aunt, uncle) or below the age of 60 in a first degree relative (brother, sister, parent, child)

Or

d) family history of raised cholesterol:

- above 7.5mmol/l in an adult
- above 6.7mmol/l in a child, brother or sister under 16.

WHAT IS GENETIC TESTING FOR FH?

Why am I being asked to have a genetic test for FH?
You may be invited for genetic testing to:

- find the gene alteration that has caused your FH
- find out if you have the gene alteration that has been identified in other members of your family.

It can be helpful to know more about the type of FH you have. This will help your doctor prescribe the right treatment for you and can also help find other close family members with the same condition. If this is the case, your doctor may refer you to a specialist genetics clinic to have a test to identify the specific gene alteration that has caused your condition. The test will be carried out by a clinical geneticist – a doctor who specialises in diagnosing and counselling people with inherited medical conditions. A gene alteration can usually be identified in about three out of every four people with FH. However, genetic testing may not be available at all clinics. It usually takes about two months to get your results.

If someone in your family has already been diagnosed with FH and has already had an altered gene identified, your doctor may suggest that you have a test to see if you have the same gene alteration. This is called cascade testing. The test is simpler because the geneticists will know which gene alteration they need to look for.

Why should I be tested for FH?
The benefits of being tested, and having treatment if you are diagnosed with FH, are:

- appropriate treatment can be started quickly
- your cholesterol levels can be monitored and managed
- your risk of getting coronary heart disease and having a heart attack is greatly reduced
- close members of your family can be tested and treatment can be started if necessary
- starting treatment early, before coronary heart disease is established, greatly reduces your risk of dying prematurely.

What is genetic counselling?
Genetic counselling will help you understand what happens when you have a genetic test and what the results might mean for you and your family. A trained counsellor can explain what the tests mean and how having FH may affect you. They will also talk to you about how the results may affect any children you have or may consider having in the future. You can find out more on being a parent with FH on page 55. You don't have to have a genetic test if you don't want one, and the counsellor will be able to advise you and support you in making your decision.

You should only have genetic testing after you've had advice from a specialist team who can make sure that the right tests are done and that the results are interpreted correctly. Having a genetic test without the appropriate support and guidance could give you misleading and inaccurate information.

Your doctor should be able to refer you for an assessment at an appropriate clinic. Or, you can visit the HEART UK website or call their helpline to find out where your nearest specialist lipid clinic is. You can also call the BHF Genetic Information Service for information and support about genetic testing and other inherited heart conditions. (See pages 62-63 for all contact details.)

What if an altered gene is found?
If you're found to have a gene alteration, your specialist will be able to tell you which gene is affected and how this alters your cholesterol levels. We explain more about this on pages 21-22.

Everyone is different, so even if you have the same gene alteration as someone else in your family, your cholesterol levels may not be affected in the same way. You may have higher, lower or the same cholesterol levels as them.

If you're told that you have FH, your specialist may ask for your help in identifying close relatives – such as brothers, sisters or cousins – who may also have inherited the altered gene. Most people with FH have at least two other affected first-degree relatives. A first-degree relative means a parent, brother, sister or child. For more information about inheritance, see page 23.

Your specialist should be able to identify those at risk in your family and may ask you to contact them. Or they may arrange to contact them directly with your permission. Either way, it's important that relatives who could be affected are given proper counselling and are tested for FH. This is what is meant by cascade testing.

What if an altered gene is not found?

If you are the first person in your family to have genetic testing

If your blood tests suggest that you may have FH but you don't have an altered gene, it does not necessarily mean that you don't have FH. It just means that your doctor has not been able to identify a known gene alteration in your DNA. It's possible that you are one of the one in four people who have FH for which a gene alteration has not yet been identified.

If an altered gene has already been found in your family and you are invited for cascade testing

If you have a genetic test and the gene alteration which has already been identified in your family is not found in you, it means that you have not inherited that form of FH from your family and you cannot pass it on to any children you may have. However, if you have high cholesterol, you may still need treatment to lower your cholesterol.

TREATMENT FOR YOU AND YOUR FAMILY

DONNA'S STORY

My dad, granddad and uncle all had heart attacks young. At a yearly check-up for diabetes, my nurse Delyth spotted my cholesterol was high and suggested I get tested. It was positive for FH. I found out it runs in my family. Delyth was fantastic in helping me to contact family members. Now 24 of us have been tested and 13 of us have FH.

That includes one of my sons, Rhys. But we take statins and I've cut down on eggs and cheese. It's helped bring down my cholesterol. And it hasn't stopped Rhys. He was extremely sporty before he was tested and he still is. Football is his passion and nothing is stopping him doing the sport he loves.

My family are super sporty

My boys love rugby and football

We make sure they can follow their passion

We live with an inherited condition

WHAT TREATMENT WILL I NEED?

If you or your child are diagnosed with FH, your doctor will discuss and agree a treatment plan with you.

FH is usually treated using medicines called statins, which help to lower your cholesterol levels. As well as taking a statin, you'll need to make changes to your diet and lifestyle which will also help to improve your cholesterol levels. We tell you more about these changes in the next chapter on Everyday life and about being a parent with FH on page 55. We talk about other treatments for FH on page 39.

What level of cholesterol should I aim for?
Everyone is different, so your doctor will discuss the best treatment plan for you and agree target levels for your cholesterol. For most adults with FH, the aim is to at least halve your LDL cholesterol level.

The average total blood cholesterol level of adults living in the UK is 5.1mmol/l. Adults who have FH, and who have already been diagnosed with coronary heart disease or who have had a stroke or other forms of arterial disease, should try to reduce total cholesterol to 4mmol/l or below, and reduce LDL cholesterol to 2mmol/l or below.

Your doctor will arrange for you to have blood tests each time you go to the clinic. The results of these tests will show how well your cholesterol is being controlled and help your doctor decide if any changes need to be made to your medicines. A follow-up appointment also helps your doctor to monitor any other changes in your general health. Your doctor will probably see you more often when you are first diagnosed with FH – perhaps once every two to three months. Once your treatment is stable, your visits will be less frequent – perhaps just once a year.

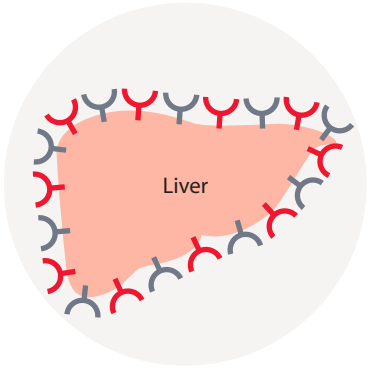
Why do I need to lower my cholesterol?
High cholesterol is a significant risk factor for coronary heart disease. There is good evidence that if you reduce your cholesterol levels, you can reduce the risk of getting heart disease, having a heart attack or needing other treatment. Treatment and careful monitoring of people with FH can help prevent the development of fatty deposits within the walls of arteries, known as atheroma, in children and adolescents. If your FH is diagnosed early and you receive effective treatment, you can live a normal life.

If you already have disease of the heart or circulation, treatment can help to stop it getting worse and help reduce the risk of having a heart attack or stroke.

TAKING STATINS TO LOWER CHOLESTEROL

How do statins work?
If you have FH, your liver can't remove excess cholesterol from your blood in the same way as someone who doesn't have FH. Cholesterol levels in your blood rise as less cholesterol can be removed from your body. Taking medicines called statins will help to remove some of the excess cholesterol.

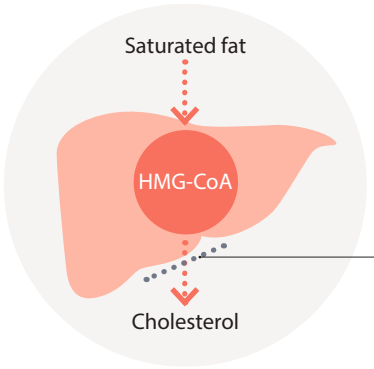
Statins work by blocking the enzyme HMG-CoA reductase in the liver which is needed to produce cholesterol. So statins reduce the amount of cholesterol made by your liver. As your liver is making less cholesterol, it will try to collect more cholesterol from your blood by increasing the number of LDL receptors on the surface of your liver. The extra LDL receptors take more cholesterol out of your blood, and the level of cholesterol in your blood falls. See the diagram on page 36.



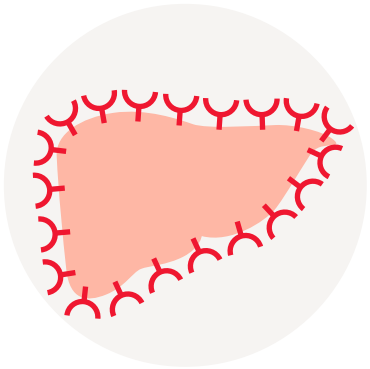
Someone with FH before treatment
In someone with FH, there are either fewer LDL receptors on the surface of liver cells or they do not work as well.

Functional LDL receptor

Dysfunctional or missing LDL receptor



How statins affect cholesterol production in the liver
Statins stop normal cholesterol production in the liver by blocking the action of the enzyme HMG-CoA reductase.



Someone with FH treated with statins
Statins reduce the level of cholesterol in the liver. This switches on more LDL receptors which remove cholesterol from the bloodstream and LDL cholesterol levels are lowered.

Statins also help to stabilise the build-up of fatty deposits within the lining of your arteries and so can reduce your risk of developing heart disease or having a heart attack. This is why most people who are at high risk of coronary heart disease, stroke or peripheral arterial disease, or who have diabetes, are prescribed a statin, even if they have a normal cholesterol level.

Statins reduce total cholesterol levels and LDL levels very effectively. Research shows that, if you have FH and you take statins, your life expectancy will increase compared with someone who has FH but does not take statins. If you have FH and take statins but don't have coronary heart disease, your life expectancy should be the same as people who don't have FH.

How do I know which statin is best for me?
Your doctor will prescribe the type and amount of statin that suits you best, given your needs and medical history. Most people with FH require high intensity statin therapy and are prescribed atorvastatin. Other statins such as simvastatin, pravastatin and rosuvastatin may also be effective.

Over time, your cholesterol levels may rise and your doctors may change the statin you are prescribed. This will depend on the results of your blood tests, other medicines you are taking, and any other medical conditions you may have. Your statin treatment may need to be adjusted or interrupted if you are prescribed courses of certain antibiotics or other treatments.

Grapefruit juice can affect how some medicines work in your body. If you're taking simvastatin, you should avoid drinking grapefruit juice. However, if you're taking atorvastatin or another statin, you can have small quantities of grapefruit juice.

Most statins should be taken in the evening, because your body makes most of the cholesterol it needs at night. Some can be taken at any time of the day. Information about each statin, and about when and how you should take it, is in the patient information leaflet that comes with your medicine. If you have any questions about statins, talk to your doctor or pharmacist.

How safe are statins?
All statins used in the UK are tested to look at their long-term effects and are generally safe. However, statins are not suitable for:

- people who have liver disease
- women who are trying to become pregnant, or who are pregnant or breastfeeding
- children under the age of 10 years (See page 56).

Possible side effects of statins
Clinical trials have shown that statins are relatively free of side effects. Occasionally, you may get side effects such as:

- stomach upset
- skin rashes
- disturbed sleep.

These side effects will often resolve themselves, but if they don't go away, it's important that you report them to your doctor.

Very occasionally, statin medicines may cause muscle aches and pains, and it's important that you report these to your doctor or nurse. If you get these side effects, your doctor will probably advise you to stop taking statins and the muscle problems will usually go away.

To reduce the likelihood of side effects, it's important to let your doctor know about any other medicines and supplements you're taking, and also to tell them about any other relevant medical history.

What about over-the-counter statins?
There are some low-dose statins that you can buy from your local pharmacist without a prescription from a doctor. They are unlikely to help someone with FH, as the dose is usually too small. If you have FH, it's important that all your cholesterol-lowering medicines, including statins, are prescribed by the doctors at your lipid clinic or by your GP, so that they can give you careful, regular follow-up and adjust your medicines if necessary.

What if I'm planning to start a family?
It's important that women do not try to conceive a baby while they're taking statins. Tell your doctor about any plans you might have to start a family, as they can advise you about any changes you may have to make before you conceive and the importance of preventing an unplanned pregnancy.

Cholesterol levels naturally increase during pregnancy, probably because cholesterol is needed for growth. It's usually safest to stop taking statins at least three months before you try for a baby. There's limited information about the effects that statins might have on a developing baby but most women who have conceived while on cholesterol-lowering medicines have gone on to have normal babies, although a few babies have been born with developmental abnormalities. During this time, your doctor may consider giving you other cholesterol-lowering medicines that don't affect a developing baby. You can start taking your statin again once your baby is born and you've stopped breastfeeding. For more information on being a parent with FH, see page 55.

OTHER MEDICINES

There are several types of medicines which can be used to lower blood cholesterol levels either instead of, or as well as, statins. These are:

- medicines which bind bile acids
- ezetimibe
- fibrates.

Many of these medicines work by helping to prevent your intestines from absorbing cholesterol either from your food or from bile acids. This in turn prevents it entering your bloodstream and raising your blood cholesterol levels.

Medicines which bind bile acids
These medicines, which are also called 'bile acid binding medicines' or 'bile acid binding resins', include cholestyramine, colestipol and Cholestagel. They work by stopping the bile acids, which are made using cholesterol, from being re-absorbed from your intestine back into your bloodstream. This means that, when your liver needs to make more bile acids, it has to take more LDL from your bloodstream and the level of cholesterol in your bloodstream falls.

Cholestyramine and colestipol come in powder form, in sachets. You have to soak some types in fruit juice before you take them. Others are already mixed with fruit flavouring and you just need to add water. Cholestagel comes in a tablet. You should take all these medicines immediately before or during a meal. They may make you feel fuller than usual at first, but most people gradually get used to this. Cholestyramine and colestipol are not absorbed into your body, so they can be used safely by children and pregnant women.

The safety of Cholestagel in pregnant women and children has not yet been established. You may get heartburn or constipation when taking these medicines, but this is more likely with larger doses.

Ezetimibe
Ezetimibe is another type of cholesterol-lowering medicine. Ezetimibe helps to lower your blood cholesterol levels by preventing your small intestine from absorbing cholesterol. It can be taken with a statin or, if you can't take statins for any reason, it can be taken on its own.

Fibrates
Fibrates such as bezafibrate, ciprofibrate, fenofibrate and gemfibrozil are useful if you have high levels of cholesterol and triglycerides, and your HDL cholesterol is low. You will not usually be prescribed fibrates if you are also taking statins, except under strict medical supervision – for example, if you have FH and you have a very high level of triglycerides. You shouldn't take fibrates if you're pregnant, or have liver or kidney disease.

Medicines which reduce triglyceride levels
Fish oils
If you regularly eat oily fish, control your weight, and limit how much alcohol you have and yet you still have a high triglyceride level, your doctor may prescribe fish oils for you. If you are taking fish oils that have not been prescribed for you, tell your doctor about them so that they can make sure that they don't interfere with any other medicines you're taking, such as warfarin.

Will taking Co-enzyme Q10 help me?
Co-enzyme Q10 (ubiquinone) is normally found in your muscle cells and is used in energy production in your body. It is similar in structure to cholesterol and is made in your body in a similar way to cholesterol. Because statins reduce cholesterol production, it is thought that they may also partially block the production of co-enzyme Q10.

A few people who experience muscle aches and pains while taking a statin, have reported improvement in their symptoms from taking a co-enzyme Q10 supplement at the same time. However, so far, clinical trials have shown limited evidence of any clinical benefit from taking co-enzyme Q10 supplements. Before deciding to take co-enzyme Q10, you should speak to your doctor.

OTHER TREATMENTS TO LOWER YOUR CHOLESTEROL LEVELS

LDL-apheresis
LDL-apheresis is a procedure where your blood is regularly 'cleaned of cholesterol' outside of your body. The process is similar to having dialysis if you have kidney disease. Your blood is passed through a special machine that filters out the cholesterol before the blood is returned to your body. Each treatment involves staying in hospital for up to four hours at a time. The treatment needs to be repeated at regular intervals. How often you need this treatment will depend on the type of FH you have and how quickly your levels of cholesterol rise, but it is usually between two and four times a month.

Very few people need LDL-apheresis. It's of most value if your LDL cholesterol level remains higher than the target set for you by your doctor, despite having the maximum amount of treatment with medicines and a cholesterol-lowering diet. You may also have LDL-apheresis if you're unable to take cholesterol-lowering medicines. Almost everyone who has homozygous FH or compound heterozygous FH will need LDL-apheresis.

Although LDL-apheresis is very effective, starting the treatment needs careful consideration as it may involve long journeys to a specialist centre on a regular basis for the rest of your life. There are only a few specialist centres in the UK that offer this type of treatment.

Lomitapide (Lojuxta)
Lomitapide has recently been licensed in Europe for adults with the homozygous form of FH and this can be used alongside of, or instead of apheresis. It works by partially blocking a protein responsible for fatty substances forming in the gut and liver. By blocking this protein, lomitapide helps reduce how much fat is absorbed and lowers the level of LDL cholesterol in your blood. If you take lomitapide you should consume less than 20 per cent of your calories from fat as this has been shown to help reduce unwanted gastrointestinal (stomach related) side effects. This is lower than normally recommended for a heart healthy diet, but advice and support is available to help you make this change.

OTHER WAYS OF REDUCING YOUR CHOLESTEROL

Along with the medicines and treatments that your doctor prescribes to control your cholesterol, it's vital that you also make changes to your lifestyle. Taking statins on their own without making any changes to your diet and lifestyle will limit how much you can reduce your cholesterol levels overall. For more information on making lifestyle changes, see the next chapter on Everyday life.

WILL THE TREATMENT FOR FH CHANGE IN THE FUTURE?

Pharmaceutical companies are continually looking for better and more effective medicines to treat conditions such as FH, and it is very likely that new medicines that can help reduce cholesterol more effectively will be available in the future. New areas of research include:

- weekly injections that aim to lower the amount of LDL produced by the liver in people with homozygous FH.
- fortnightly injections of an antibody that blocks the PCSK9 enzyme. This appears to be effective in lowering LDL in heterozygous FH and is in the late stages of development.
- looking at gene alterations that affect how your body uses and breaks down different medicines to work out the best type and dose of medicines that should be used for each individual with FH.
- using cardiac regenerative medicine to look at ways in which we may be able to help the heart to repair itself after a heart attack. It may also be possible to repair damage to blood vessel walls caused by high cholesterol levels, helping to prevent or reduce the risk of a heart attack.

EVERYDAY LIFE

I'm a busy working Mum

My son is always on the go

He keeps me active and on my toes

I live with an inherited condition

SUZANNE'S STORY

My father died aged 41 and now I realise he probably had FH, like me.

I was tested for FH because, while I didn't have an unhealthy lifestyle, my cholesterol levels were excessively high. It was a relief to know what was going on. Now I keep my heart healthy by taking statins, watching my diet and making sure I'm active.

My son, Cameron, will be screened when he's old enough. If he does test positive I'll be happy knowing he'll get the healthiest start possible, with the same great care and support I've had. The FH test wasn't created early enough for my dad, but it's giving my son and I a brighter future.

Suzanne & Cameron

HOW MIGHT HAVING FH AFFECT MY LIFE?

It is normal to have concerns about being diagnosed with FH. For example, you may find it difficult to explain the importance of being tested for FH to other members of your family. Some family members may find it easier to deal with the uncertainty of not knowing, rather than learning to live with a diagnosis of FH. If you're a parent, you may feel guilty about having passed the altered gene to one or more of your children.

It may help you or other members of your family to talk to someone about this – either a close friend, your doctor, or someone at HEART UK - The Cholesterol Charity. Specialist genetics clinics are experts at dealing with inherited conditions and have trained counsellors who can listen and explain the testing process to you and your family members. Speaking to a trained professional does not mean that you have to commit to being tested or receive any treatment, but it can provide you with the information you need to make the right decision for you.

MAKING POSITIVE LIFESTYLE CHANGES

Making changes to your lifestyle will help to protect your heart health and help reduce your cholesterol levels, whether you have FH or not.

You can reduce your cholesterol levels and protect your heart health by:

- stopping smoking
- eating a healthy diet
- drinking no more than the recommended limits of alcohol
- taking regular physical activity
- being a healthy body weight and shape.

Stopping smoking
Smoking is a risk factor for heart disease in everyone who smokes, but is particularly bad for people with FH who are already at higher risk because of their very high LDL cholesterol levels. Smoking can lower your levels of HDL cholesterol as well as make LDL cholesterol more likely to damage healthy blood vessels.

Stopping smoking can quickly reduce your risk of getting coronary heart disease and can also reduce your risk of getting other conditions such as lung cancer. It can be difficult to stop smoking, so speak to your doctor or practice nurse about the type of help and support that is best for you.

Healthy eating
People with FH should follow the same general advice on healthy eating as for other people in the UK. By eating a healthier diet, you can help reduce your cholesterol levels by around 5 – 10 per cent. Some people may find that healthy eating has a greater effect on their cholesterol levels than other people. But it's important to remember that as well as helping to lower your cholesterol, making healthy changes to your diet can have additional heart health benefits such as controlling your blood pressure, helping to prevent diabetes and helping you to keep to a healthy weight.

Your doctor should arrange for you to see a dietitian so that you and your family can get some one-to-one advice and help you make some gradual changes.

The Mediterranean diet
Research suggests that following a Mediterranean-style diet may help to lower your risk of developing cardiovascular disease, reduce your chances of weight gain and help to lower your levels of LDL cholesterol.

A typical Mediterranean diet has lots of vegetables, fruits, beans, cereals and wholegrain foods like wholegrain bread, pasta and brown rice. It also contains moderate amounts of fish, white meat and some low-fat dairy produce, and in some cases nuts and a small amount of red meat and sweet desserts. Unsaturated fats such as olive oil are used instead of saturated fats such as butter, lard or ghee. This combination of different foods, along with a Mediterranean lifestyle and eating pattern, appears to provide heart health benefits.

Choose the right type of fats
Fat is an important and vital part of our diet. It's rich in energy and contains essential nutrients. You also need a certain amount of fat to help repair and maintain your body. However, there are good and bad types of fat.

Oils and fats are made up of a combination of saturated fats, monounsaturated fats and polyunsaturated fats. To help reduce your cholesterol levels it's important to cut down on saturated fats and replace them with unsaturated fats. However, all fats are high in calories, so you should eat only modest amounts. Try to avoid highly processed foods which may also contain harmful trans fats.

What are saturated fats?
Saturated fats affect how your body manages cholesterol, so you should try to reduce the amount that you eat. Saturated fats are usually hard at room temperature and are mostly found in animal foods such as red meat and dairy products, but also in some vegetable oils such as coconut and palm oil.

What are unsaturated fats?
Unsaturated fats are usually liquid at room temperature. Some are also present in spreads made from vegetable oils. Make sure that most of the fats in your diet come from these sources. Unsaturated fats are found in vegetable oils, nuts and seeds, oily fish, and spreads based on sunflower, olive or rapeseed oil. It's best to have a mixture of monounsaturated and polyunsaturated fats in your diet (see table on page 49).

What are trans fats?
Trans fats occur naturally in small amounts in dairy foods and red meat and these are considered safe to eat. However industrially produced trans fats have been linked to an increased risk of cardiovascular disease. These trans fats are formed when vegetable oils are made into solid fat through a process called hydrogenation. Trans fats made in this way have a similar effect to saturated fat as they can increase your LDL cholesterol. However, unlike saturated fats they can also reduce HDL cholesterol.

Most of us don't eat a lot of trans fats, and many food manufacturers have reduced the amount of hydrogenated vegetable fats they use in the products they make. At the moment, the nutritional information on food packaging doesn't have to include information about trans fats, but foods that have 'hydrogenated fat or oils' in the ingredients list are likely to contain trans fats. It's best to avoid these foods where possible.

Unsaturated fats

Monounsaturated fats

Found in:

- olive oil and rapeseed oil
- avocado
- nuts and seeds (almonds, cashews, hazelnuts, peanuts and pistachios).

Some spreads are made from monounsaturated fats.

Polyunsaturated fats

Found in:

- corn oil, sunflower oil and soya oil
- nuts and seeds (walnuts, pine nuts, sesame seeds and sunflower seeds).

Some spreads are made from polyunsaturated fats.

Omega-3 fats

Found in:

- fish oil
- oily fish such as herring, mackerel, pilchards, sardines, salmon, trout and fresh tuna.

See page 52 for more about omega-3 fats.

Saturated fats

Saturated fats

Found in:

- butter
- hard cheese
- whole milk
- fatty meat
- meat products
- biscuits
- cakes
- cream
- lard
- dripping
- suet
- ghee
- coconut oil
- palm oil
- pastry.

Trans fats

Found in:

- pastries
- cakes
- biscuits
- crackers
- fried foods
- takeaways
- hard margarines.

Foods that have 'hydrogenated oils or fats' or 'partially hydrogenated oils or fats' in the list of ingredients are likely to contain trans fats.

Can I eat foods that contain cholesterol?
Many foods that are rich in saturated fat are also rich in cholesterol. So reducing your saturated fat intake will reduce your cholesterol intake as well. Some foods, however, are low in saturated fat, but do contain some cholesterol. These include eggs, offal, such as liver and kidney and shellfish including prawns, crabs and lobster. Apart from containing cholesterol, these are otherwise very healthy foods, so most people with moderately high cholesterol don't need to restrict all of them.

People with FH should have only modest amounts of foods containing cholesterol. A sensible amount is no more than three or four eggs a week, and shellfish no more than once or twice a week. Some shellfish such as cockles, mussels, oysters, scallops and clams are all low in cholesterol and in saturated fat and you can eat them freely. You should avoid offal altogether. Talk to your dietitian for more advice on foods that are high in cholesterol.

Foods that can help lower cholesterol
Some foods such as nuts, oats, soya foods and plant stanols/sterols have been shown to have blood cholesterol-lowering benefits. If you include these foods daily, they could help to further reduce your cholesterol, even more than just cutting down on the amount of saturated fat you eat and following a healthy diet. The Ultimate Cholesterol Lowering Plan (UCLP) by HEART UK shows how to include these foods in your daily diet. See page 63 for more information.

Soluble fibre also helps to lower cholesterol by reducing the amount that is absorbed and recycled into the bloodstream from your intestine. Good sources of soluble fibre include oats, oatbran, whole barley and pulses such as baked beans, kidney beans, soya beans, peas, lentils and chickpeas.

Plant sterols and plant stanols
Plant sterols and plant stanols can help to reduce your cholesterol levels. In a similar way to soluble fibre, they work by preventing cholesterol and cholesterol-rich bile acids being absorbed by the body from your intestines.

Plant sterols and stanols are naturally present in small quantities in many fruits, vegetables, nuts, seeds, cereals, pulses and vegetable oils. However, the amount that you get from these foods is too little to help reduce cholesterol levels. To make a difference, you need to eat foods that have plant sterols or stanols added to them, like some types of spreads, yoghurts, and mini drinks.

Although the effect varies between individuals, there is evidence to show that plant sterols and stanols can help to reduce LDL cholesterol by 7 – 10 per cent when 1.5 to 2.4g per day is regularly consumed as part of a healthy balanced diet. Whilst this may lower the risk of heart attack, clinical trials have not yet been done to support this.

If you have FH, you could benefit from using these products on a daily basis. However, if you are taking the medicine ezetimibe, you are unlikely to benefit from them, as the medicine works in a similar way to the plant sterols and stanols.

Plant sterol or stanol products are not recommended for adults who don't have a high cholesterol level or FH, and they are not suitable for women who are pregnant or breastfeeding. These products can be taken by children with FH, but first discuss with your doctor or dietitian whether they are suitable for your child.

Have some fish
Fish is naturally low in saturated fat and is a good source of protein. Oily fish are also a good source of vitamins A and D and provide the richest source of the beneficial 'omega-3' fatty acids, known as DHA and EPA (docosahexaenoic acid and eicosapentaenoic acid).

These fatty acids can help to lower blood triglyceride levels, however to have an effect you would need to eat several portions of oily fish a week. If you have high blood triglycerides your doctor may recommend a fish oil supplement or other medication to help lower your blood triglyceride level. Aim to eat at least two portions of fish a week, one of which should be an oily fish such as mackerel, salmon, sardines or fresh tuna.

Cut down on salt
Cutting down on salt will help lower your risk of getting high blood pressure – a risk factor for cardiovascular disease. If you already have high blood pressure, cutting back on salt may help improve it. When you're cooking, try adding spices and herbs to your food instead of salt, and don't add salt at the table. Try to avoid ready meals and processed foods, as these often contain a lot of salt.

Alcohol and your heart
Drinking moderate amounts of alcohol can offer some benefit to heart health as it can raise 'good' HDL cholesterol. However, alcohol is high in calories and drinking over the recommended limit can increase your risk of cardiovascular disease, heart failure and other health problems such as liver disease. We do not recommend that anyone starts drinking to protect their heart health, as you can achieve greater benefits from eating a healthy diet and being physically active. Men should drink no more than three to four units of alcohol each day, and women no more than two to three units each day. It's important to have one or two alcohol free days each week.

Physical activity
Being physically active will help reduce your risk of getting diseases of the heart and circulation.

- Adults should aim to do at least 150 minutes of moderate intensity activity every week
- Children and young people under the age of 18 should aim to do at least one hour of activity every day
- Children under the age of five that are capable of walking unaided should be physically active daily for at least three hours, spread throughout the day.

This will help to:

- increase your HDL cholesterol
- control your body weight and shape
- control your blood pressure
- reduce stress levels.

One way of reaching 150 minutes a week is by being active for 30 minutes at least five days a week. If you find it difficult to do 30 minutes all in one go, you can break it down into two lots of 15 minutes or three lots of 10 minutes. Your activity should make you feel warm and slightly out of breath, but you should still be able to hold a conversation. Try to vary the type of activity you do and include children and the rest of your family so that everyone's heart health will benefit. If you're not used to doing this amount of activity, talk to your doctor first and start slowly, building up gradually over time.

Keeping a healthy body weight and shape
Being overweight, and particularly carrying too much weight around your middle is linked to having higher LDL and lower HDL cholesterol levels. Too much weight around your middle means having a waist size of:

- more than 94 centimetres or 37 inches for men
- 90 centimetres or 35½ inches for men of South Asian origin
- 80 centimetres or about 31½ inches for women.

If you're not sure whether you are overweight or if your body shape puts your health at risk, ask your doctor or practice nurse.

Managing stress
A small amount of stress from time to time helps you to cope in difficult situations. However, there is evidence that exposure to prolonged periods of stress can have a damaging effect on your health. In stressful situations, your body produces the hormones adrenaline and cortisol. These hormones help you to respond to situations that require action – sometimes known as the 'fight or flight' response. However, being exposed to stressful situations for prolonged periods of time often leads to unhealthy coping strategies such as smoking, drinking more than the recommended limit of alcohol, and unhealthy eating habits. All of these increase your risk of getting diseases of the heart and circulation.

Try to find ways of managing your stress. Take time out for a break, take up a hobby, make time for yourself and your friends, try yoga, or practise relaxation techniques. Whatever works for you, make sure you plan it into your daily routine.

You may find that being diagnosed with FH is stressful.

You may worry about the impact it might have on your life and your family. Or, you may experience feelings of guilt that you could pass FH on through your family. Talk to your GP or genetic counsellor about how you feel. You can also read the advice on being a parent with FH on the next page.

BEING A PARENT

FH is an inherited condition, meaning it's passed down through a family. Some people may have no physical signs or symptoms of FH (see pages 26-27) and only learn that they have it after suffering a heart attack or being diagnosed with heart disease. If your child has inherited FH from you or your partner, neither of you should feel guilty – you can't change your DNA or the genes your child inherits. If your child is diagnosed and treated for FH early, they can live a normal, healthy life.

When should my children be tested for FH?
If a history of early heart disease or high cholesterol runs in your family, or you or your partner have been diagnosed with FH, it is highly recommended that your children are tested for FH. Getting an early diagnosis and adopting a healthy lifestyle, including a healthy diet and plenty of physical activity and cholesterol-lowering medication can all greatly reduce the chances of your child developing heart disease later on in life.

If you have FH, your children should be tested between two and ten years old or as early as possible after that. People with FH have high levels of cholesterol from birth, so if your child's cholesterol level is within an acceptable range, it is unlikely that your child will have FH.

Diagnosing FH in children
If your child has a moderately high total cholesterol level between 5.5 and 6.7mmol/l, this makes it difficult to rule out or diagnose FH. Under these circumstances, your lipid clinic will suggest that your child has tests at regular intervals as he or she grows up. How frequently your child will need to be tested will depend on your child's cholesterol levels and any other physical signs of FH that they may have.

If an altered gene has been found in either parent, it will be possible to test your child for the same altered gene. On average, half the children of a parent with FH will inherit the altered gene and have FH. The other half will inherit the normal gene and will not have FH. A genetic test will show if your child has inherited the altered gene or not.

Confirming a diagnosis early in childhood is important, and starting any necessary treatment will greatly reduce the risk of heart disease caused by FH in later life. You and your child may find it easier to start to make changes to your diet and lifestyle together, and starting at this early age will help them keep up the changes in the future.

Can my child take statins?
Children with FH should be seen on a regular basis in a child-friendly environment within a lipid clinic. Statins and other medicines that are licensed for children may be prescribed for children over the age of ten years. Although there is good evidence that statins are safe, the decision to start medication in children will be made using clinical judgement in each case. As there is no cure for FH, even after your child's cholesterol levels are lowered, they will need to continue taking cholesterol-lowering medications throughout their life. It can sometimes be hard to get your child to take their medication and stick to a routine. It's vital that your child takes their medication to reduce their risk of heart disease in the future. Taking your medication together as a family will help your child form a routine.

What if my child has compound heterozygous or homozygous FH?
Your child can only inherit these forms of FH if both parents have FH. Compound heterozygous FH is very rare and occurs when a child inherits two different gene alterations associated with FH, one from each parent. If two of the same gene alterations are inherited from each parent, then the child will have homozygous FH (see page 23 for more on how FH is inherited). This is also very rare. Having either of these forms of FH will mean a child has a much more severe form of FH, usually with much higher cholesterol levels.

Children with compound heterozygous or homozygous FH are often diagnosed at an early age, as physical signs of FH often develop quickly and require immediate cholesterol-lowering medication. Some children may also need LDL-apheresis to remove cholesterol from their blood (see page 41). This procedure is very effective, but you should seek advice from your lipid specialist before starting this kind of life-long treatment.

Supporting your family
Making healthy lifestyle changes will protect your family's health. You can support your family and help keep your cholesterol levels and the risk of heart disease lower by making gradual lifestyle changes such as:

- eating a healthy diet
- taking regular exercise
- reducing the amount of alcohol you drink
- stopping smoking.

It's important to discourage all children from smoking and alcohol, but it's particularly important to make children and young people who have FH aware of the increased risks of smoking for people with FH.

Healthy eating for your family
Diet is an essential and important part of treating FH in young children and by following a healthy eating plan, your entire family can benefit. It's important to encourage your children to learn about healthy eating and how the food and snacks they eat can affect their cholesterol levels, so that they grow up being able to make healthy choices and look after their own health. Making changes as a family will benefit all members, whether you have FH or not, and prevents a child from feeling they are being treated differently.

When planning your healthy diet, remember that children, especially the under fives, will have different dietary needs to adults as they need calories to help them develop and grow. Ask your doctor to refer you to a dietitian for more advice.

HOW WILL HAVING FH AFFECT OTHER PARTS OF MY LIFE?

Prescription charges
If you have FH, you'll have to pay for your prescriptions, unless you live in a part of the UK where you don't have to pay prescription charges or you don't have to pay them for some other reason. You may find it helpful to speak to your doctor or pharmacist about schemes to reduce the cost of prescription charges for people with long-term conditions.

Insurance
If you're being treated for a medical condition, you usually have to declare it to your insurance company; otherwise it could invalidate your insurance. Having a diagnosis of FH may affect how you're treated when you apply for life insurance or travel insurance. It's also possible it may affect you if you apply for a mortgage.

Some insurance companies may decide that you're at higher risk of getting coronary heart disease and may charge you higher premiums. Some companies don't differentiate between high cholesterol as a result of poor diet and other lifestyle factors, and the high cholesterol which is caused by an inherited condition such as FH.

HEART UK has been working with insurance companies to help them understand that FH is a treatable condition. Now a few better-informed, sympathetic insurers accept that, if you're diagnosed and treated and you don't already have coronary heart disease, FH does not reduce your life expectancy compared to someone that does not have FH.

To get the best possible insurance cover, you should always use an insurer that specialises in pre-existing medical conditions. For travel insurance, a single-trip cover policy may be more cost-effective than an annual policy with some insurers, so make sure you ask about both.

Consider getting a letter from your specialist about your condition and how it's managed, to support your insurance application. The insurance company may ask you to consent to a medical report being obtained from your GP. If they do, ask to see the medical report before it's sent, as you may want to ask your doctor to remove any information which is not relevant to your application and which you do not want to share.

TECHNICAL TERMS

A

Atheroma
Fatty deposits that can build up within the walls of the arteries.

Atherosclerosis
The build-up of fatty deposits within the walls of the arteries.

B

Blood lipids
Fatty substances found in the blood.

C

Cholesterol
A fatty substance mainly made in the body by the liver.

Chromosome
A threadlike fibre which is in all cells and which carries genetic information.

Coronary heart disease
When the walls of the arteries become narrowed by a gradual build-up of fatty deposits called atheroma.

E

Emulsify
Mixing two or more liquids or substances that do not normally mix easily.

Enzyme
An enzyme is a protein which helps to speed up chemical reactions in your body.

F

Familial combined hyperlipidaemia
An inherited condition in which the levels of cholesterol and triglycerides in the blood are raised.

Familial hypercholesterolaemia
An inherited condition in which the blood cholesterol level is very high.

Familial hyperlipidaemias
A collective term for inherited conditions that affect fat levels in the blood.

G

Gene
A segment of DNA responsible for the production of a specific substance such as a protein, which in turn is essential for a particular characteristic or function in the body.

H

HDL
High-density lipoprotein. The 'good' cholesterol.

Hypertension
High blood pressure.

L

LDL
Low-density lipoprotein. The 'bad' cholesterol.

LDL-apheresis
A hospital-based treatment for people with extremely high blood cholesterol levels.

Lipids
Fatty substances in the blood.

Lipoproteins
Combinations of cholesterol and proteins which transport lipids (fats) in the blood.

M

Mutation
An alteration or 'mis-spelling' of the DNA code that causes its eventual product (usually a protein) to function abnormally, which in turn is responsible for a disease or condition.

mmol/l
A unit measuring concentration. It means millimoles per litre and is the unit often used to describe the levels of cholesterol in your blood.

O

Omega-3 fat
A type of polyunsaturated fat found in certain types of fish.

T

Trans fats
A type of fatty acid that acts like saturated fat.

Triglycerides
A type of fatty substance found in the blood.

MORE INFORMATION

For information on your nearest clinic for inherited heart conditions

BHF Genetic Information Service
Greater London House
180 Hampstead Road
London NW1 7AW

Phone: 0300 456 8383
Website: bhf.org.uk

The BHF Genetic Information Service provides information for families affected by an inherited heart condition, including information on where to go for an assessment. The service is staffed by specialist cardiac nurses and a bereavement counsellor.

At the British Heart Foundation we're fighting for every heartbeat. We support a ground-breaking genetic testing programme for FH by funding FH nurses, ensuring a cascade testing service is extended to eight NHS Trusts across England and Scotland. This will help to identify the estimated one in 200 families with FH in the UK.

To find out more about our work on FH and the research we fund, visit **bhf.org.uk/research**

BHF publications

The following booklets will give you more information on making changes to your lifestyle and reducing your risk of diseases of the heart and circulation.

Angina (HIS6)

Eating well (G186)

Get active, stay active (G12)

Heart attack (HIS7)

Keep your heart healthy (HIS25)

Losing someone to heart disease (G419)

Reducing your blood cholesterol (HIS3)

Stop smoking (G118)

To order any of these booklets:

- call the BHF Orderline on 0870 600 6566, or
- email orderline@bhf.org.uk or
- visit bhf.org.uk/publications

Our resources and services are free of charge, but we rely on donations to continue our vital work. If you'd like to make a donation, please call our donation hotline on **0300 330 3322** or visit our website at **bhf.org.uk/donate**

FOR MORE ON FH

HEART UK – The Cholesterol Charity
7 North Road
Maidenhead
Berkshire SL6 1PE

FH and Cholesterol Helpline
Phone: 0345 450 5988
(Monday to Friday, 10am-3pm calls charged at local rate)
Email: ask@heartuk.org.uk
Website: www.heartuk.org.uk

HEART UK helps people who are diagnosed with FH and their families to come to terms with their condition. We offer expert support, guidance and education via our dedicated helpline, our website, Cholesterol News, our resources, patient meetings and online patient forum.

A dietitian who speaks a number of Asian languages is available to offer advice on Friday mornings.

Children's FH Resources
HEART UK has produced resources specifically for children with FH, including:

- The Story of Hope – a 4 minute film about a family with FH, and
- Buddy's FH adventure – an e-book aimed at children 7 years and above.

These resources are free of charge from HEART UK.

To find out more, visit www.heartuk.org.uk/FHchildrensresources

National Cholesterol Month
This event runs each year to help raise awareness of the dangers of raised cholesterol and to raise much needed funds for HEART UK. To find out more or to get involved contact development@heartuk.org.uk or call HEART UK on 01628 777046

HEART UK supports the education of health professionals who diagnose and treat FH and we campaign long and hard to keep FH at the top of the Government's health agenda.

Call HEART UK or visit our website for more information.

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About the British Heart Foundation

The British Heart Foundation is the nation's heart charity, saving lives through pioneering research, patient care and vital information.

What you can do for us
We rely on donations to continue our vital work. If you would like to make a donation to the British Heart Foundation, please call our donation hotline on **0300 330 3322**, or visit **bhf.org.uk/donate**, or post it to us at the address below. Thank you for supporting our fight.

There are lots of other ways that you can help us. Go online at **bhf.org.uk** to find out how.

Have your say
We would welcome your comments to help us produce the best information for you. Why not let us know what you think? Contact us through our website at **bhf.org.uk/contact** or write to us at the address below.

British Heart Foundation
Greater London House
180 Hampstead Road
London NW1 7AW
bhf.org.uk

Heart Helpline
0300 330 3311
(a similar cost to 01 and 02 numbers)
For information and support on anything heart-related.

Genetic Information Service
0300 456 8383
(a similar cost to 01 and 02 numbers)
For information and support on inherited heart conditions.

About Heart UK–The Cholesterol Charity

HEART UK helps people who have a high cholesterol level or who are diagnosed with FH to come to terms with their condition. We offer expert support, guidance and education through our website, resources and **FH and Cholesterol Helpline**.

HEART UK also works closely with the doctors, nurses, dietitians and other professionals who treat people with FH as well as the families themselves. HEART UK continually campaigns on behalf of people with FH to influence the production and uptake of national guidelines on the detection and treatment of FH. We have worked to raise the awareness of the condition, and to develop resources for people with FH, their families and health care professionals.

Get in touch – Get involved
We're always keen to hear from people with an interest in cholesterol and to discuss ways in which we can support each other. If you would like to know more about the work of HEART UK, receive regular news about FH, or find out about how you can get more involved, email us at **ask@heartuk.org.uk** or call our **FH and Cholesterol Helpline** on **0345 450 5988**.

Support us
HEART UK are a small charity with a big heart. We receive no government funding, so rely on the generosity of our kind donors to continue our work. If you would like to make a donation to HEART UK, please visit our website **www.heartuk.org.uk/donate**, call us on **01628 777046** or send a cheque made payable to HEART UK to the address below. Thank you for your support.

HEART UK – The Cholesterol Charity
7 North Road
Maidenhead
Berkshire SL6 1PE
www.heartuk.org.uk

British Heart Foundation

HEART UK
THE CHOLESTEROL CHARITY

Coronary heart disease is the UK's single biggest killer.

At the British Heart Foundation, we've pioneered research that's transformed the lives of people living with heart and circulatory conditions. Our work has been central to the discoveries of vital treatments that are changing the fight against heart disease.

HEART UK – The Cholesterol Charity helps people who have high cholesterol or who are diagnosed with FH. We offer expert support, guidance and education and aim to prevent premature deaths caused by high cholesterol and cardiovascular disease.

But so many people still need our help.

Join the British Heart Foundation and HEART UK in our fight for every heartbeat in the UK. Every pound raised helps to make a difference to people's lives.

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