Inherited heart conditions
Familial hypercholesterolaemia
You may be reading this booklet because you have 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 have 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 is needed to help grow and repair cells in the body. FH is passed from parent to child and can affect one or more family members. Not all family members are affected, but it is very important to find out which ones are.

In most cases, having FH does not affect a person’s 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 is 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 is important that the close blood relatives of someone with the condition have an assessment to find out if they have inherited FH
• 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 to have a healthy lifestyle.

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 63.

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 or on other inherited heart conditions, you can contact HEART UK – The Cholesterol Charity or the British Heart Foundation Genetic Information Service. (See pages 68–69 for all contact details).
How the heart works

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

Your heart is a muscle that pumps blood around your body, delivering nutrients and oxygen to all the 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.
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 had 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. (See the diagram below.) This process 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. You may feel the pain in one or more of these places, or it may spread to other areas. This pain 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 the thinner inner wall may rupture. When this happens, the atheroma pours out into the artery. To stop the atheroma travelling around the 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 called a heart attack. If the blood flow cannot be restored, the muscle will die and this could lead to heart failure. Sometimes a heart attack can be fatal.

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. There are some risk factors that you can change or do something about, and others that you can’t change.

Risk factors that you can change or 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
• take too little physical activity
• are overweight or obese
• have diabetes.

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

Familial hypercholesterolaemia (FH)

Risk factors that you can’t change
These are the risk factors that you can’t change:

- 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.
What is familial hypercholesterolaemia (FH)?

FH – which is short for familial hypercholesterolaemia – is a condition which results in exceptionally high cholesterol levels. It is a genetic condition. This means that it is passed on 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 500 people in the UK has FH, and it is one of the most commonly occurring genetic conditions. In people with FH, high cholesterol levels are usually present from birth and continue throughout life. We explain more about how conditions are inherited on page 23.

If you have FH, you have a higher risk of getting coronary heart disease at an early age. More than 120,000 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 liver makes cholesterol mostly from the saturated fats in food. (See page 54 for more information on fats.) You also get a small amount of cholesterol from some animal foods such as eggs, liver and kidneys, and seafood such as prawns. Plant foods (cereals, fruit, vegetables, pulses, nuts and seeds) don’t contain any cholesterol.

Cholesterol plays a vital role in how every cell in your body works. It is 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 (for example, in children or during pregnancy) when many new cells are being
formed very quickly. 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 ‘transport system’ for 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). The lower the density of the lipoprotein, the more fats it contains. So having a high LDL is harmful to you.

- Low-density lipoproteins – sometimes called LDL cholesterol or the ‘bad’ cholesterol – carry most of the cholesterol from your liver, through the bloodstream, to where it is needed. About 70% of the cholesterol in your body is carried by LDL. Having a high LDL is harmful to you.
- High-density lipoproteins – sometimes called HDL cholesterol or the ‘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 10).

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.

**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. It is often hard to detect people with FCH until they are in their 20s and 30s or even older, as this is often when triglyceride levels are seen to increase.

For most people, high cholesterol is not caused by inheriting an altered gene. People may have a high cholesterol level because they eat too much saturated fat, smoke, are overweight – especially if they have a lot of fat around their waistline – or because they are not physically active. Other people may have high cholesterol levels as a result of an underactive thyroid gland or long-term kidney problems. And sometimes medicines which people take for particular conditions may affect how their body handles cholesterol.

**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 LDL (‘bad’) cholesterol and a low level of HDL (‘good’) 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.

The average total blood cholesterol level of adults living in England and Scotland is 5.3mmol/l. Doctors agree that the lower the cholesterol level, the lower your risk of getting diseases of the heart and circulation. See page 39 for target cholesterol levels for people with FH.
How is cholesterol controlled in the body?

All of the cells in your body are able to make cholesterol, but most is made in your liver. It is 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 acts as the ‘central clearing house’ for cholesterol, as it is the only organ that can 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 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 (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 accept excess cholesterol from the cells and transport it back to the liver for removal. Unlike LDL, HDL does not need any LDL receptors to enter the liver.

The diagram on the next page shows a simplified view of how cholesterol is processed in your body.

How does FH change the way that cholesterol is controlled in the 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 10. While FH results in a high level of LDL cholesterol, the other risk factors mentioned on pages 10–11
can influence the effect that LDL cholesterol has on the artery wall. For example, if you have high blood pressure, this will increase the risk of LDL particles being forced into the artery wall and causing atheroma. 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 particles to get stuck there.

**Is there a cure for FH?**

There isn’t a cure for FH, but it can be treated very successfully. The treatment can significantly reduce your risk of getting coronary heart disease, having a heart attack or needing other treatment. We tell you more about the treatment for FH on pages 39–48.

**Is there a risk of sudden death with FH?**

If you or a member of your family has a heart attack, it is 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 heart 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 68 for details) about whether they (the close relatives) need an assessment (tests) to find out if they may have an inherited heart condition.
How are certain conditions inherited?

Our bodies are made up of millions of cells. Each cell has a nucleus, which contains information that makes each one of us unique. These are our genes. We each have around 23,000 different genes. Genes give the instructions that are needed for development and growth of all the cells in our body, and they determine characteristics like hair colour, eye colour, height and blood type. Each gene provides the instructions to produce a specific protein (or part of a protein), which will decide on a characteristic or control a particular function in the body.

Genes are arranged end to end along threadlike structures called chromosomes. The chromosomes and genes are made up of a chemical substance called DNA. Each cell usually carries 46 chromosomes arranged in 23 pairs. See the diagram below.
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 have 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 regulates or controls the removal of cholesterol by breaking down the LDL receptor protein. 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 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 excreted into the intestines, from where it can be removed from your body. The diagram on page 26 shows how normal cholesterol control is affected in someone with FH.
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, will 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 1 in 2 chance of inheriting the condition, and it can affect boys and girls equally. It cannot skip a generation so, if neither parent has it, a child cannot inherit it. As we all have two copies of each gene, one from each parent, it is possible to inherit an altered gene from either your mother or your father, and in some rare cases from both your parents.

We explain the three types of FH on the next few pages.

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 1 in 2 chance of inheriting the altered gene from a parent who has FH. See the diagram below.

A family tree

This family tree shows four generations affected by FH.
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%.

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 (for example, cousins) and have both inherited the original altered gene from a common ancestor.

If you have homozygous FH or compound heterozygous 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 is not treated, you may develop heart disease as a child. You are also more likely to have some of the other symptoms we have described on page 31.

Around one in every 1 million people has compound heterozygous or homozygous FH, so around 60 people in the UK are expected to have one of these forms of FH. We currently know of only around 30 such patients – mostly children and young adults.

Why does my doctor think I have FH?

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

- a routine blood test shows a high cholesterol level
- you have a heart attack, especially if it happens at a young age
- other members of your family have a history of premature coronary heart disease (see A family history of heart disease on page 12)
- 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 that you may have FH – for example, tendon xanthomata or corneal arcus (see page 31).

On the next few pages we explain what happens if your doctor thinks you might have FH.

What will happen if my doctor suspects I have FH?

If your doctor thinks that you may have FH, it is important that you have an assessment (tests) to find out whether you do have the condition. On pages 30–32, 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 will 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 need to 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 a 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 32.
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 page 68 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 a high cholesterol level, or if they have had a heart attack or have heart disease. In particular, they will want to know what age they 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 syringe and needle and sent to a laboratory for analysis, or
- a finger prick (capillary sample) is taken and analysed on a desktop analyser in the clinic.

To make a formal diagnosis of FH, the doctor usually needs 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 the full range of blood fats (lipids). 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 32).

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 the 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, he or she will also be looking for physical signs of FH. However, if you don’t have these, it does not mean you do not have FH, as they are not always found. They are also very easy to miss, so you may not have noticed them yourself. The main signs they 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 is believed that tendon xanthomata are only found in people with FH.
- **Xanthalasmas** – Xanthalasmas (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.

Xanthalasmas and corneal arcus can also occur for reasons other than FH.
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 screening. The test is simpler because the geneticists will know which gene alteration they need to look for.

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. (For more information about inheritance, see page 26.) 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 and screening after you have had advice from a specialist team who can make sure that the right test is 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 68–69 for all contact details.)

**What if an altered gene is found?**

If you have a genetic test and a gene alteration is found, 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 29–31.

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

If you are 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.)

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 is important that relatives who could be affected are given proper counselling and are screened for FH. This is what is meant by cascade screening.

**What if an altered gene is not found?**

If you are the first person in your family to have genetic testing
If all your tests suggest that you may have FH but an altered gene is not found in you, it does not necessarily mean that you do not have FH. It just means that the doctors have not been able to identify a known gene alteration in your DNA. It is possible that you are one of the one in four people who has 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 screening
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 a high cholesterol you may still need treatment to lower your cholesterol.

**What about my children?**

If you have FH, any children you have should be screened and tested before the age of 10 years 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.

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 that may be present.

If an altered gene has been found in you or your partner, it should be possible to test your child for the same altered gene. Confirming a diagnosis early in childhood is important, and starting any necessary treatment will 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.
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 screened 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 are 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 expert at dealing with inherited conditions and have trained counsellors who can listen and explain the process of screening to you and your family members. Speaking to a trained professional does not mean that you have to commit to a screening programme or any treatment, but it can provide you with the information you need to make the right decision for you.

The benefits of having screening, and having treatment if you are diagnosed with FH, are

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

What treatment will I need?

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

FH is usually treated using a medicine called a statin, which will help to lower cholesterol levels. As well as taking a statin, you will 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. We talk about other treatments for FH on page 47.

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. For example, if your LDL cholesterol is now 6mmol/l, the aim would be to reduce it to 3mmol/l or less.

For 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, it may be better to try to reduce total cholesterol to 4mmol/l or below, and to 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 atheroma in children and adolescents. If FH is diagnosed early and effective treatment is given, most people with FH 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?**

As you have FH, your liver can’t remove excess cholesterol from your blood in the same way as someone who doesn’t have FH. So the cholesterol levels in your blood rise as less cholesterol can be removed from the body. Taking a medicine called a statin will help to remove some of the excess cholesterol.

Most people with FH have one altered gene and one normal gene. The altered gene means that the cholesterol removal process is not working as it should. Statins help ‘switch on’ the normal gene to make it more effective than normal. So, the normal gene will be doing some of the work of the altered gene as well, which helps clear the excess cholesterol from your blood.

Statins work by blocking an enzyme in the liver (HMG-CoA reductase) which is needed to produce cholesterol. So statins reduce the amount of cholesterol made by your liver. As your liver is making less cholesterol, your liver will try to collect more cholesterol from your blood by ‘switching on’ the LDL-receptor genes. This increases 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 the next page.
Statins also help to stabilise the atheroma (the build-up of fatty deposits) within the lining of the 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.

Can children take statins?
Children with FH should be seen on a regular basis in a child-friendly environment within a lipid clinic. Diet is an essential and important part of treating FH in young children. Statins and other medicines that are licensed for children may be prescribed over the age of 10 years. Although there is good evidence that statins are safe, starting lifetime medication in children will be made using clinical judgement in each case.

How do I know which statin is best for me?
Your doctor will prescribe the type and amount of statin that suit you best, given your needs and medical history. Most people are prescribed Simvastatin to begin with. Other statins that are available are Atorvastatin, Fluvastatin, Pravastatin and Rosuvastatin.

Some statins affect LDL, HDL and triglycerides, while others only affect LDL significantly. Over time, your doctors may change the statin you are prescribed. This will depend on how well the statin is lowering your cholesterol, other medicines you are taking, and any other medical conditions you may have.

If you are taking Simvastatin, you should avoid drinking grapefruit juice or eating grapefruit. However, if you’re taking Atorvastatin or another statin, you can have small quantities of grapefruit juice (or grapefruit). 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.

Possible side effects of statins
Statins are generally safe and well tolerated, and are widely used. Clinical trials have shown that they are relatively free of side effects. Occasionally, people report side effects such as a stomach upset or disturbed sleep. These side effects will often resolve themselves, but if they don’t go away, it is important that you report them to your doctor.

Very occasionally, statin medicines may cause muscle aches and pains, and it is 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 is important to let your doctor know about any other medicines and supplements you are taking, and also to tell him or her about any other relevant medical history.

How safe are statins?
There are several statins available in the UK. All of these have been tested in long-term, large clinical trials that have looked not just at the results the statins produce but also at their effect on health and their long-term safety. Statins are not suitable for people who have liver disease or for women who are trying to become pregnant, or who are pregnant or breastfeeding. (See Statins and pregnancy, on page 44.)
Treatment for you and your family

What about over-the-counter statins?
These are low-dose statins that you can buy from your local pharmacist without a prescription from a doctor. They are unlikely to help someone who has FH, as the dose is smaller than is usually needed. If you have FH, it is 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 am planning to start a family?
It is important that women of child-bearing age do not try to conceive a baby while they are taking statins. Tell your doctor about any plans you might have to start a family, as he or she can advise you about any precautions you should take 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 is usually safer to stop taking statins at least three months before trying for a baby. There is 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 do not affect a developing baby. Women can start taking their statin again once the baby is born and they have stopped breastfeeding.

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:
- fibrates
- medicines which bind bile acids
- nicotinic acid medicines
- ezetimibe.

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

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 should not take fibrates if you are pregnant, or have liver or kidney disease.

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 the intestine back into the bloodstream. This means that, when the liver needs to make more bile acids, it has to take more LDL from the bloodstream and the level of cholesterol in the 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 the 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. Some people who take these medicines may get heartburn or constipation, but this is more likely with larger doses.
Nicotinic acid medicines
Nicotinic acid medicines, which include Tredaptive and Niaspan, stop the release of fatty acids from fat cells in the body, leading to a reduction in LDL and an increase in HDL cholesterol.

If you are also taking medicines to reduce your blood pressure, or if you have low blood pressure, taking nicotinic acid can lower your blood pressure further.

Ezetimibe
Ezetimibe is another type of cholesterol-lowering medicine. Ezetimibe helps to lower 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.

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 which have not been prescribed for you, tell your doctor about them so that he or she can make sure that they don't interfere with any other medicines you are taking, such as warfarin.

Will taking Co-enzyme Q10 help me?
Co-enzyme Q10 (ubiquinone) is normally found in 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 some pain relief 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 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 is 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 are 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.

Other ways of reducing your cholesterol
Along with the medicines and treatments that your doctor prescribes to control your cholesterol, it is 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.

Our bodies make cholesterol from the saturated fats in foods, so reducing the amount of saturated fats you eat will help to reduce your cholesterol levels. We tell you more about fats on pages 54–55. Your doctor may
refer you to a dietitian who could help you identify changes you can make. Eating a healthy diet that is low in saturated fat can reduce your cholesterol by up to 10%.

You can also increase the amount of HDL cholesterol in your body by being physically active, stopping smoking and replacing saturated fats with healthy fats found in fish, seeds, nuts and olives.

**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. One experimental treatment being tested at the moment in people with homozygous FH aims to lower the amount of LDL produced by the liver. It requires weekly injections. It seems to be effective, although we don’t yet know if it will be safe in the long term.

In the future, pharmacogenetic testing may be used to work out the best type and dose of statins or other medicines that should be used for each individual with FH. This form of testing looks for gene alterations that affect how your body uses and breaks down different medicines.

Research into cardiac regenerative medicine is looking at ways in which we may be able to help the heart to repair itself. For example, at the moment, if you have a heart attack, the heart muscle that has been affected dies and heart attack survivors often develop heart failure as a result. The research is looking at ways in which we may be able to reverse some of this damage. The research also aims to identify how we can teach our blood vessels to repair themselves. This means that, in the future, it may be possible to repair damage to blood vessel walls caused by high cholesterol levels (as in FH), helping to prevent or reduce the risk of a heart attack.

All this research is currently in its early stages, but if it is successful it could save millions of lives.
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 level 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 levels of HDL cholesterol.

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.

It is important to discourage all children from smoking, 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

Making sure that the food you eat is low in saturated fat and high in fibre, fruits and vegetables, can help reduce your cholesterol levels by around 5–10%. Some people may find that healthy eating has a greater effect on their cholesterol levels than other people. People with FH should follow the same general advice on healthy eating as for other people in the UK, but we give some additional tips below.
Your doctor should arrange for you to see a dietitian, so that you and your family can get some one-to-one advice, which will help you to make some gradual changes. It’s important to remember that making healthy changes to what you eat can have additional heart health benefits for both you and your family – such as controlling your blood pressure, helping to prevent diabetes and helping you to keep to a healthy weight.

It is 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 other family members as well (whether they have FH or not) and prevents a child from feeling they are being treated differently.

What is a healthy diet?

Eating a varied and balanced diet with few processed foods helps to keep you healthy. You should have foods from the four main food groups:

- fruit and vegetables
- bread, rice, potatoes, pasta and other starchy foods
- milk and dairy foods
- meat, fish, eggs, beans and other non-dairy sources of protein.

Cutting down on the amount of saturated fat you have is vital for reducing your cholesterol levels. Check and compare food labels to find out how much saturated fat the food contains. Foods that contain more than 5 grams of saturated fat per 100 grams, or more than 6 grams per portion, are high in saturated fat. We explain more about saturated fat in What type of fat should I eat? on page 54.

Here are some other changes you can make to have a healthier diet:

- have starchy foods – such as cereals, pasta, rice, chapattis, potatoes, noodles and bread – as the basis of most meals. Choose wholegrain or wholemeal varieties where possible,
- choose foods rich in soluble fibre – such as oats, beans, peas and lentils – several times each week,
- eat plenty of fruits and vegetables – at least five portions a day,
- have at least two portions of low-fat dairy foods or soya alternatives every day,
- eat a handful (about 1 ounce or 30 grams) of unsalted nuts every day.

If you need to lose weight, you may need to eat more, or less, of certain food groups. Ask your doctor to refer you to a dietitian who may be able to help you.

What about foods that contain plant sterols and plant stanols?

Plant sterols and plant stanols can help to reduce cholesterol levels. They work by preventing cholesterol and cholesterol-rich bile acids being absorbed by your intestines.

Plant sterols are naturally present in small quantities in many fruits, vegetables, nuts, seeds, cereals, pulses and vegetable oils. However, the amount of plant sterols that you would get from these foods – around 150 to 350 milligrams a day (0.15 to 0.35 grams) – does not result in meaningful reductions of cholesterol levels. To make a difference, you need to eat foods that are enriched – that is, foods that have plant sterols or stanols added to them.

Foods enriched with either sterols or stanols can help to reduce cholesterol levels. Eating between 2 grams and 2.5 grams of plant sterols (or stanols) every day can lower cholesterol by an extra 10% on top of having a healthy diet low in saturated fat and taking a statin.

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, because 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.
Can I eat foods that contain cholesterol?
On page 15 we explained that most of the cholesterol in your body is made from the saturated fat found in foods. Many foods 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 (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 them. However, people with FH should have only modest amounts of these foods. A sensible amount is no more than three or four eggs a week, and shellfish no more than once or twice a week. You should avoid offal (such as liver and kidney) altogether. Shellfish such as cockles, mussels, oysters, scallops and clams are all low in cholesterol and in saturated fat and can be eaten freely. Talk to your dietitian for more advice on foods that are high in cholesterol.

Which types of fat should I eat?
Fat is an important and vital part of our diet. It is 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.

Foods containing fat are made up of a combination of saturated fats, monounsaturated fats and polyunsaturated fats. On pages 56–57 we give examples of foods that contain these different types of fats. Where possible, it is important to cut down on saturated fats and replace them with monounsaturated fats and polyunsaturated fats. Omega-3 fats are good for your heart too. All fats are high in calories, so you should eat only small amounts, as this will help you to maintain a healthy weight and shape. Also, avoid highly processed foods which may contain harmful trans fats.

What are saturated fats?
Saturated fats are usually hard at room temperature and are found in animal foods such as red meat and dairy products. Your body produces cholesterol from saturated fats, so it is best to reduce the amount of saturated fats that you eat.

What are unsaturated fats?
Unsaturated fats are usually liquid at room temperature. Some are also present in spreads made from vegetable oils. It is important to make sure that most of the fats in your diet come from these sources. Unsaturated fats are found in vegetable oils, nuts and seeds, and spreads based on olive oil or sunflower oil.

What are trans fats?
Trans fats occur naturally in small amounts in dairy foods and red meat. It is industrially produced trans fats, which have been linked to an increased risk of coronary heart disease. Industrially produced trans fats are formed when vegetable oils are processed 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 and reduce HDL.

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 as a food ingredient in the products they make. At the moment, the nutritional information on food packaging doesn’t have to include information about trans fats, but the ingredients list should state if the food contains any hydrogenated fat. So if you see ‘hydrogenated fat’ – especially ‘partially hydrogenated fat’ – on a label, it is best to avoid that food.

Fish and oily fish
Aim to eat at least two portions of fish a week. One of these portions should be oily fish such as mackerel, salmon, sardines or fresh tuna. All fish, but especially oily fish, provide the richest source of the beneficial ‘omega-3’ fatty acids, known as DHA and EPA (docosahexaenoic acid and eicosapentaenoic acid). A regular intake of these – either from eating oily fish or taking fish oil supplements if they have been recommended by your doctor – can help to lower blood triglyceride levels, help prevent your blood from clotting, and help to regulate your heart rhythm. And they can help to improve your chance of survival if you have a heart attack.
Unsaturated fats

**Monounsaturated fats**

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

Spreads made from olive oil

**Polyunsaturated fats**

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

Spreads made from seed oils

**Omega-3 fats**

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

See page 57 for more about omega-3 fats from sources other than fish.

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/fats’ or ‘partially hydrogenated oils/fat’ in the list of ingredients are likely to contain trans fats.

Vegetarians and people who don’t eat fish can get a form of omega-3 called alpha linolenic acid (ALA) from plant-based foods such as dark green leafy vegetables, nuts, linseed and soya and fortified products. However, this form of omega-3 may be less effective than the EPA and DHA found in oily fish or fish oil supplements.

**Should I follow a low fat diet?**

No, it is not necessary to reduce all sources of fat, just limit saturated fat. Eating a low fat diet reduces the amount of healthy cholesterol or HDL cholesterol in the blood so it is important to include some foods that contain unsaturated fats (see page 56). If you are trying to lose weight then you will need to reduce your total calorie intake. You can do this by limiting saturated fats, alcohol and sugary foods and by being more active. If you are also feeding a family 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.

**Salt**
Cutting down on salt will help lower your risk of getting high blood pressure – which is a risk factor for diseases of the heart and circulation. If you already have high blood pressure, cutting back on salt may help improve it. When you are cooking, try adding 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**
Drinking moderate amounts of alcohol has been shown to offer some benefit to heart health and can help raise HDL cholesterol (the protective type of cholesterol). However, alcohol is high in calories and also drinking over the recommended limit can increase your risk of heart 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 the same benefits in other ways, such as eating a healthy diet and being physically active. Men should drink no more than 3 to 4 units of alcohol each day, and women no more than 2 to 3 units each day.

**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 30 minutes of moderate-intensity physical activity a day on five days a week. (Children should aim to do one hour of activity a day on seven days of the week.) This will help to:
- increase your HDL cholesterol
- control your body weight and shape
- control your blood pressure
- reduce stress levels.

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. The activity you choose 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, so that you don’t get bored. Include children and the rest of your family in your activity so that everyone’s heart health will benefit. If you’re not used to doing this amount of activity, start slowly and build up gradually.

**Keeping to a healthy body weight and shape**
Being overweight, and particularly carrying too much weight around your middle – having a waist size of more than 94 centimetres (about 37 inches) for men, 90 centimeters (35½ inches) for men of South Asian origin, or 80 centimetres (about 31½ inches) for women – is linked to having higher LDL cholesterol levels. 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 is good for us and helps us to cope in difficult situations. However, there is evidence that exposure to prolonged periods of stress can have a damaging effect on our health. In stressful situations, our bodies produce the hormones adrenaline and cortisol. These hormones help us 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.

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 the FH on through your family. Talk to your GP or genetic counsellor about how you feel.

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.
How will having FH affect other parts of my life?

**Prescription charges**
If you have FH, you will 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 local pharmacy about prescription charges for people with long-term conditions, as many pharmacies have subscription systems which mean that you will pay less for your prescriptions.

**Insurance**
If you are 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 are treated when you apply for life insurance or travel insurance. It is also possible it may affect you if you apply for a mortgage.

Some insurance companies may decide that you are at higher risk of getting coronary heart disease and may charge you higher premiums. Some insurance 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 a 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 are 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, 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 is 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 is 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**  
Combine two liquids or substances which do not normally mix easily.

### 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.

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.

HDL
High-density lipoprotein. The ‘good’ cholesterol.

Hypertension
High blood pressure.

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.

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.

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

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.

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.

Keep your heart healthy
- Angina
- Heart attack
- Eating well
- Get active, stay active
- Stop smoking
- Publications in the Inherited heart conditions series
  - Hypertrophic cardiomyopathy
  - Dilated cardiomyopathy
  - Sudden arrhythmic death syndrome
  - Inherited heart rhythm disturbances
  - Arrhythmogenic right ventricular cardiomyopathy
- Other booklets
  - Losing someone to heart disease

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

For more on FH
HEART UK – The Cholesterol Charity
7 North Road
Maidenhead
Berkshire SL6 1PE
Helpline: 0845 450 5988
(Monday to Friday, 10am to 3pm. Calls are charged at a local rate.)
Website: www.heartuk.org.uk
Email: ask@heartuk.org.uk

HEART UK helps people who are diagnosed with FH and their families to come to terms with their condition. We offer help and support via our website, booklets, a membership scheme and a dedicated helpline.

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

HEART UK’s National Cholesterol week is run each year to raise awareness of the dangers of raised cholesterol. To find out more or get involved contact info@teamredlaces.org.uk or call the Teamredlaces hotline on 0845 873 9597.

HEART UK’s annual conference takes place each July and people with FH and their families are welcome to attend. Call HEART UK or visit the website for more information about this or other educational events run to support people with raised cholesterol.

You can register to receive regular newsletters and information about high cholesterol or become a member of HEART UK. Visit our website call our helpline or email ask@heartuk.org.uk
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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.

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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 on the back cover. There are lots of other ways that you can help us. Go online at bhf.org.uk/support us to find out how.

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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 information, help and support through our website, booklets factsheets, helpline, events and conference.

HEART UK has campaigned on behalf of people with FH to influence the production and uptake of national guidelines on the detection and treatment of FH. And we have worked to raise awareness of the condition, and to develop and provide resources for those with FH, families and health care professionals.

If you would like to know more about the work of HEART UK, receive regular news about inherited high cholesterol, or find out how you can get more involved, email us at ask@heartuk.org.uk or call our Helpline.
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