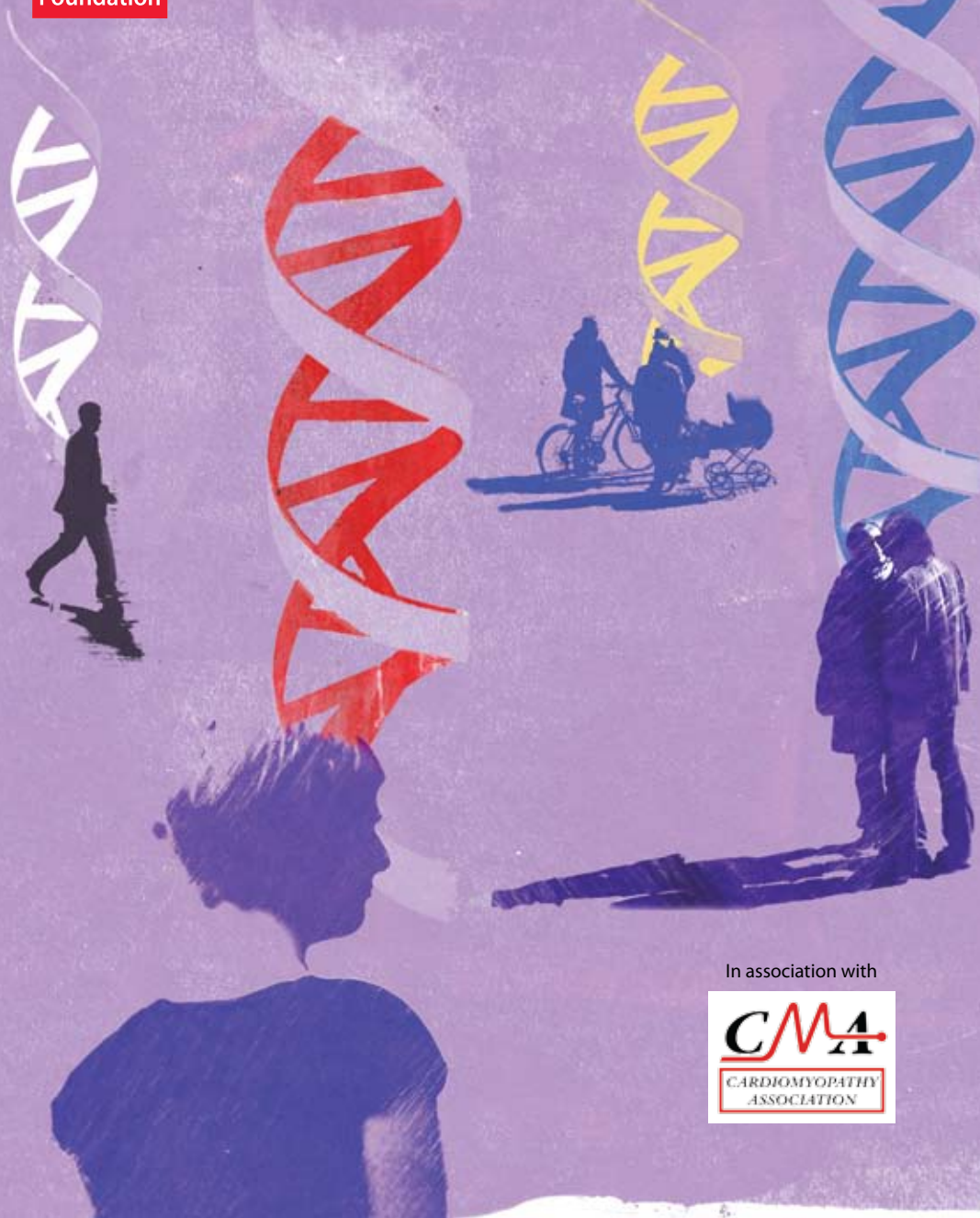




Inherited heart conditions

Hypertrophic cardiomyopathy



In association with



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This booklet is not a substitute for the advice your doctor or cardiologist (heart specialist) may give you based on his or her knowledge of your condition, but it should help you to understand what they tell you.

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

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Introduction

You may be reading this booklet because you have been diagnosed with the inherited heart condition hypertrophic cardiomyopathy. 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.

Cardiomyopathy is a disease of the heart muscle. It can run in families and can affect one or more members of a family. Some members of a family may be affected more than others. Some family members may not be affected at all.

There are three main types of cardiomyopathy:

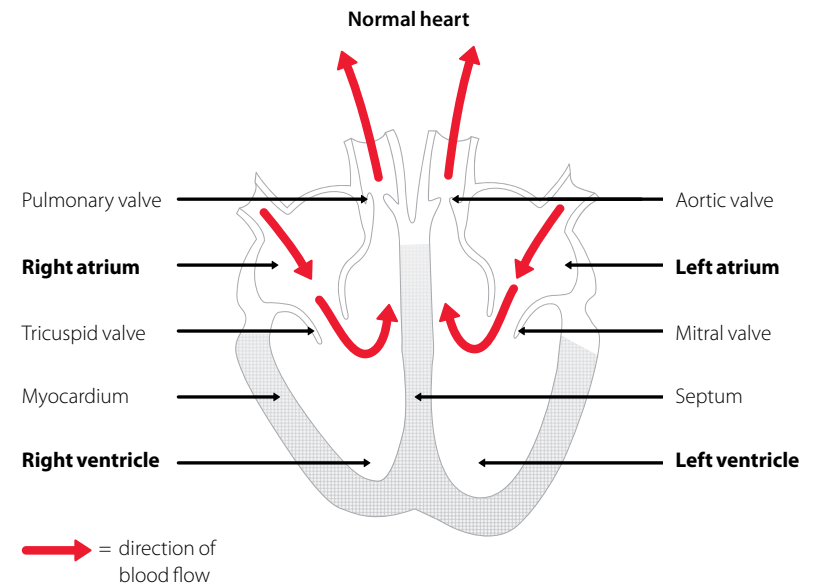
- hypertrophic cardiomyopathy (or HCM or HOCM for short)
- dilated cardiomyopathy (DCM), and
- arrhythmogenic right ventricular cardiomyopathy (ARVC).

This booklet is about hypertrophic cardiomyopathy. For information on the other types of cardiomyopathy, see the other booklets in this series. See page 64 for details.

In most cases, having hypertrophic cardiomyopathy does not affect a person's quality of life or lifespan. However, a small number of people with the condition do experience significant symptoms and could be at risk of sudden death. It is important that families affected receive an accurate assessment, diagnosis, treatment and support, from specialists in a clinic for inherited heart conditions.

The normal heart

The heart is a specialised muscle that contracts regularly and continuously, pumping blood to the body and the lungs. It has four chambers – two at the top (the atria), and two at the bottom (the ventricles). See the diagram below.

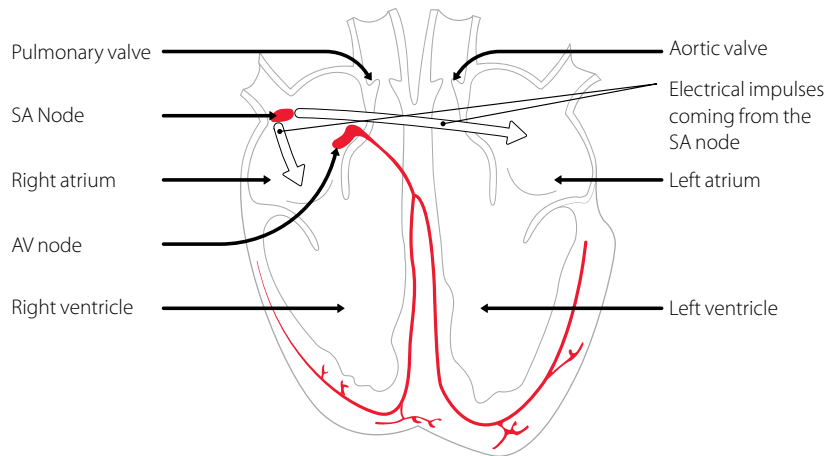


How the heart functions electrically

The pumping action of the heart is caused by a flow of electricity through the heart that repeats itself in a cycle. The normal trigger for the heart to contract comes from the heart's natural pacemaker, the SA node (sino-atrial node), which is in the right atrium (see the diagram overleaf). The SA node sends out regular electrical impulses, which make the atria contract and pump blood into the ventricles. The electrical impulse then passes to the ventricles through a form of 'junction box' called the AV node (atrio-ventricular node). This electrical impulse spreads into the ventricles, causing the heart muscle to contract and to pump

blood out of the ventricles. The blood from the right ventricle goes through the pulmonary artery and then to the lungs, and the blood from the left ventricle goes through the aorta and then around the body.

Normal electrical signals in the heart



Structure of the heart

The heart consists of three layers:

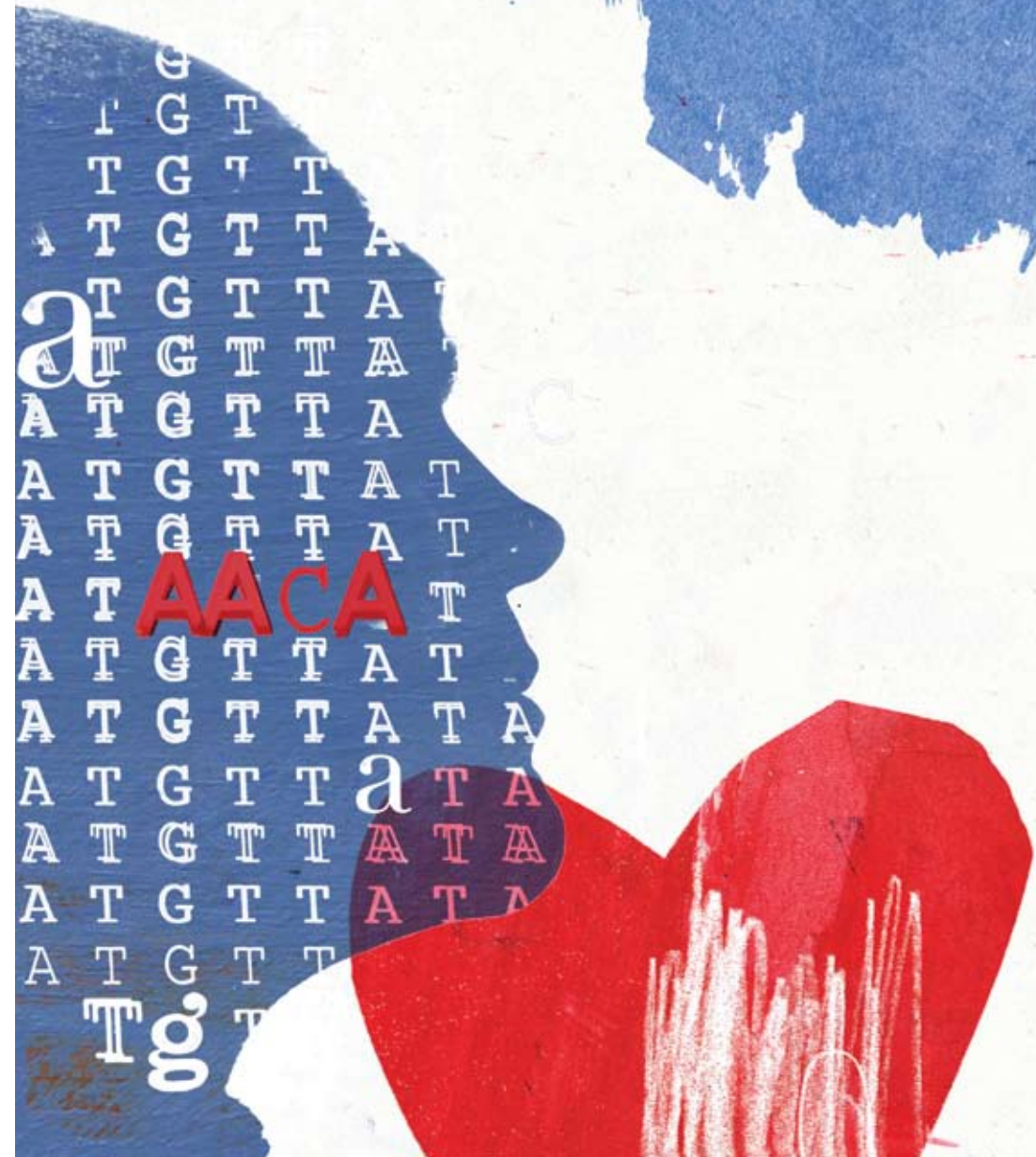
- the endocardium
- the myocardium
- the pericardium.

The endocardium is a thin layer on the inside of the heart, lining the chambers and valves.

The myocardium is the thick, muscular layer of the heart that contracts and squeezes the blood out of the heart. It is the structure of the muscle fibres in the myocardium that is affected by cardiomyopathy.

The pericardium is a thin, double layer that forms a protective sac around the outside of the heart. It contains a small amount of fluid – called pericardial fluid – which acts as a lubricant when the heart is contracting.

Hypertrophic cardiomyopathy



What is hypertrophic cardiomyopathy?

Hypertrophic cardiomyopathy is a disease of the heart muscle. It is a genetic condition. This means that it is passed on through families and is caused by a change or mutation in one or more genes. About 1 in 500 of the UK population¹ has the condition, although most people who have it have few if any symptoms. We explain more about how cardiomyopathy is inherited on page 29.

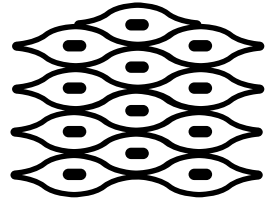
Hypertrophic cardiomyopathy was first recognised more than a century ago but was only established as an accepted diagnosis in the late 1950s. The condition has been known by a number of names including hypertrophic obstructive cardiomyopathy, (or HOCM for short), idiopathic hypertrophic sub-aortic stenosis (or IHSS), and muscular sub-aortic stenosis. The general term hypertrophic cardiomyopathy – or HCM for short – is now most widely used.

How does hypertrophic cardiomyopathy affect the heart muscle?

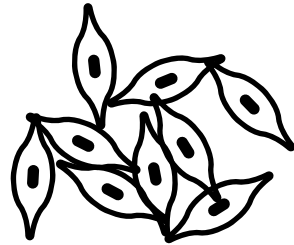
The main abnormality of the heart in hypertrophic cardiomyopathy is that the heart muscle (myocardium) has become excessively thick. How thick the muscle is, and how much of the muscle is affected, can vary from one person to another. The left ventricle is almost always affected, and in some people the muscle of the right ventricle also thickens.

In a normal heart, the cells that make up the heart muscle lie in smooth, straight lines, as shown in the diagram overleaf on the left. But in people with cardiomyopathy, the cells lie in disorganised layers (myocardial disarray), as shown in the diagram on the right. There is often also scarring of the heart muscle. The thickening and scarring of the muscle make the heart muscle stiff. Higher blood pressure is therefore needed to make the heart muscle contract and pump the blood out of the heart.

¹Watkins H, Ashrafian H, McKenna W J. 2008 The genetics of hypertrophic cardiomyopathy: teare redux. *Heart*; 94: 1264–1286.



Structure of cells in a normal heart muscle



Myocardial disarray – the structure of cells in the heart muscle of someone with hypertrophic cardiomyopathy

The heart muscle may also thicken in people who do not have cardiomyopathy. This can happen because they have high blood pressure or have been involved in intensive athletic training over a long period of time. This type of thickening is not a genetic condition that can be inherited like hypertrophic cardiomyopathy. The cells of the heart muscle in people with these conditions lie in smooth lines as in the diagram on the left.

The coronary arteries (the blood vessels that supply the heart muscle) are usually normal in people with hypertrophic cardiomyopathy. The valves in the heart are also generally normal, although the mitral valve can be affected if certain parts of the myocardium are thickened (see page 16).

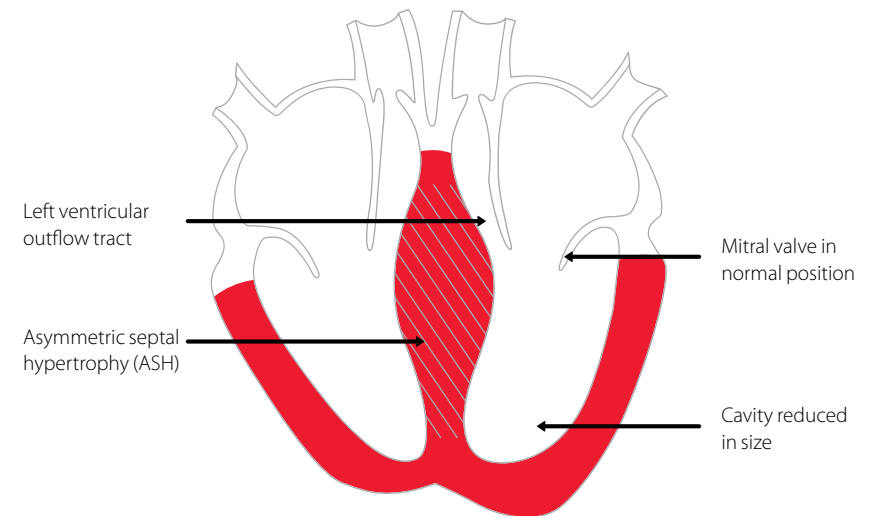
How does the muscle thickening vary?

There are four types of heart muscle thickening patterns:

- asymmetrical septal hypertrophy without obstruction
- asymmetrical septal hypertrophy with obstruction
- symmetrical hypertrophy, or concentric hypertrophy, and
- apical hypertrophic cardiomyopathy.

Asymmetrical septal hypertrophy without obstruction

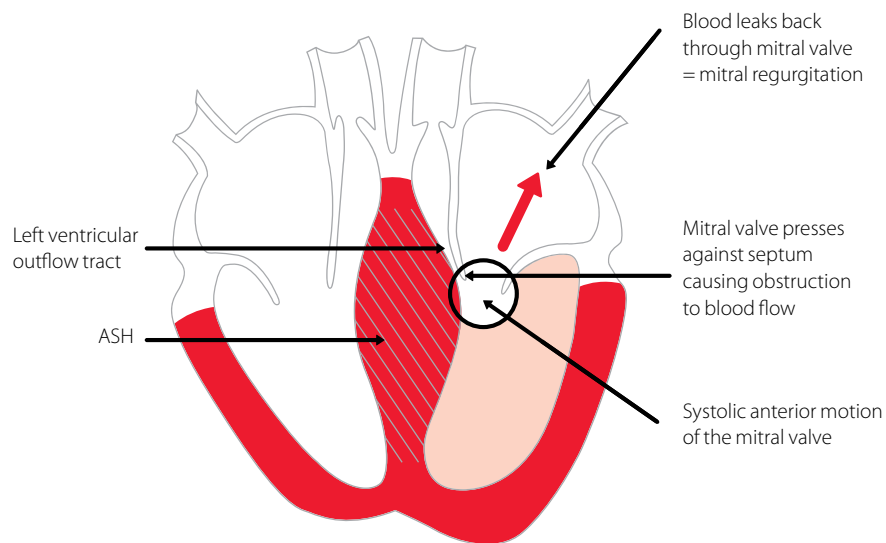
This is the most common form of hypertrophic cardiomyopathy. Here the muscle thickening occurs mainly in the septum – the muscular wall between the right and left sides of the heart (see the diagram below). The thickening occurs in the centre of the septum but does not interfere with the normal flow of blood out of the heart through the left ventricular outflow tract, so it is known as asymmetrical septal hypertrophy without obstruction. The mitral valve is not affected and is in a normal position.



Asymmetrical septal hypertrophy with obstruction

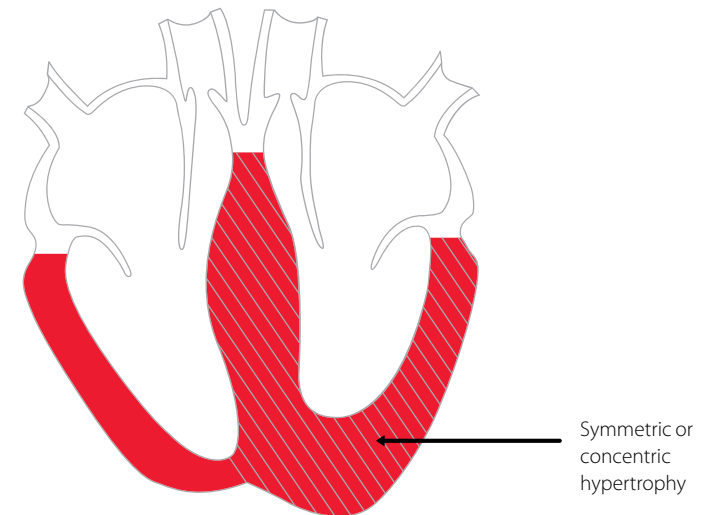
In some cases, the thickening of the septum obstructs the flow of blood through the left ventricular outflow tract when the heart contracts. This is known as asymmetrical septal hypertrophy with obstruction. The thickening of the septum has caused the outflow tract to become narrower, which means that, when the heart contracts, the mitral valve touches the septum (see the diagram below). This reduces the amount of blood that is pumped out of the heart. This is called left ventricular outflow tract obstruction. It causes a turbulent blood flow, producing a murmur (an unusual sound) that can be heard with a stethoscope.

The heart valves are designed to allow blood to move through the heart in one direction only. The mitral valve controls the flow of blood from the left atrium to the left ventricle. However, the obstruction can cause the valve to leak – that is, it lets blood flow in a backwards direction from the left ventricle back into the left atrium. This is known as mitral regurgitation.



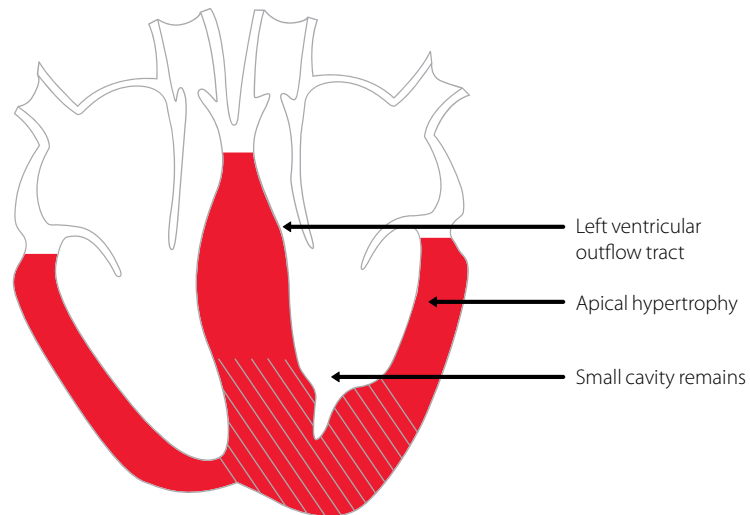
Symmetrical hypertrophy or concentric hypertrophy

This is when the muscle thickening is evenly distributed and equally severe throughout the whole left ventricle. This results in the left ventricle being smaller than normal. This condition can also lead to left ventricular outflow tract obstruction as described on page 16.



Apical hypertrophic cardiomyopathy

In about 1 in every 10 people who have hypertrophic cardiomyopathy, the thickening of the myocardium is mostly at the bottom tip, or apex, of the heart (see the diagram below). This is called apical hypertrophic cardiomyopathy. This thickening does not affect the outflow tract, so there is not usually any obstruction to the left ventricular outflow tract.



When does hypertrophic cardiomyopathy develop?

We are all unique because each one of us has our own genetic information that makes us different. Our genes make us who we are – for example, how tall we will be or what colour hair we will have. This genetic information is held in our DNA in the cells of our body. The genetic information is produced through a coding system of proteins, represented by letters, that tells all of the cells in our bodies what their function should be.

If there is a mistake in one of these codes, the cells will do something different or not develop as they should do. In hypertrophic cardiomyopathy, a mistake in the sequence of letters means that the heart muscle cells are arranged in a haphazard fashion (as shown on page 14). This is known as a genetic mutation. We explain more about genetic inheritance and coding on pages 27–31.

Although the genetic mutation responsible for hypertrophic cardiomyopathy is present at birth, the condition is not usually diagnosed at this early stage or even in infancy. Hypertrophy usually develops as children grow into adults, after puberty.

Hypertrophic cardiomyopathy is usually diagnosed when a heart murmur (an unusual heart sound) is detected. This can be when someone has a routine examination, perhaps for a medical check-up or for insurance reasons, rather than because they have any symptoms. Or, they may be seeing their doctor for another unrelated reason. Sometimes people do have symptoms (see page 20) and are sent for further tests (see page 22) so that a diagnosis can be made.

In some cases, the hypertrophy may not be detected until much later in life. This form of hypertrophic cardiomyopathy is known as 'late onset hypertrophic cardiomyopathy'.

However, sometimes hypertrophic cardiomyopathy can be misdiagnosed. For example, a doctor may think that the cardiomyopathy is caused by a condition such as high blood pressure, or that it is simply a result of 'getting older'. It is important that an accurate diagnosis is made so that families can be screened and tested for the condition.

What are the symptoms of hypertrophic cardiomyopathy?

Some people with hypertrophic cardiomyopathy don't get any symptoms. Others do get symptoms, and some of the common symptoms are listed below. Many people never have any serious problems related to their condition. In general, symptoms, whether mild or considerable, tend to be stable throughout adult life. Some people find that their symptoms get worse in later life. This may be because they develop an arrhythmia (an abnormal heart rhythm – see page 23). Or it may be because their heart muscle has become progressively stiffer, making the contraction of the heart become less forceful.

The most common symptoms are:

Shortness of breath (dyspnoea)

If shortness of breath is a symptom of hypertrophic cardiomyopathy, it generally happens when the person is exercising or being active. Most people are only mildly restricted by their shortness of breath. However, a small number of people may be more restricted in the level of exercise they are able to take, and very occasionally some people may be short of breath while they are resting.

Chest pain

Chest pain or tightness on exertion (angina) is a common symptom. The pain occurs because the heart muscle is not getting a sufficient blood supply. Although the main coronary arteries that supply the heart muscle are usually normal, the hypertrophic cardiomyopathy may have caused the microscopic coronary arteries within the heart muscle itself to become narrowed, reducing the supply of blood and causing pain.

Palpitations

This is a sensation of an extra or skipped heartbeat. In some cases, palpitations may start suddenly and feel very fast, and may be accompanied by sweating or light-headedness.

Light-headedness and blackouts

This may occur when the person is exercising, or after palpitations, or sometimes for no apparent reason.

Other symptoms

Sometimes, other conditions can develop as a result of hypertrophic cardiomyopathy and may produce other symptoms. We describe these on page 23.

How is hypertrophic cardiomyopathy diagnosed?

Your doctor may suspect that you have hypertrophic cardiomyopathy because of your symptoms, or because you have a heart murmur, or because of the results of your electrocardiogram (ECG). Or, you may be going for tests because someone else in your family has been diagnosed with the condition.

If your doctor suspects that you may have thickening of your heart muscle, he or she will send you to the hospital, usually as an outpatient, for a series of tests. Your doctor will also refer you to a cardiologist for specialist advice. The most common tests for hypertrophic cardiomyopathy are:

- a physical examination
- an electrocardiogram (ECG)
- an echocardiogram
- exercise testing
- an angiogram.

We explain more about these and other tests on page 32.

Is there a cure for hypertrophic cardiomyopathy?

At present there is no cure for hypertrophic cardiomyopathy but treatment aims to improve symptoms and prevent complications. The need for and choice of treatment will be different for everyone. Your treatment may stay the same for many years, or may change more frequently. There are many forms of treatment available which may help to reduce the risk of developing symptoms, or help to relieve your symptoms. We explain more about these on page 36.

What other conditions can occur as a result of hypertrophic cardiomyopathy?

In some people with hypertrophic cardiomyopathy, a number of other conditions can develop as a result of having the condition. These may include the following.

Arrhythmias

The abnormality of the heart muscle in people with hypertrophic cardiomyopathy can sometimes interfere with the normal electrical activity of the heart. In the affected parts of the heart muscle, the electrical impulse may become disrupted as it crosses the areas of disarrayed cells and scarring. This can lead to fast or erratic heart rhythms known as arrhythmias. Arrhythmias are a common complication in people with hypertrophic cardiomyopathy. The two most common arrhythmias in people with this condition are **atrial fibrillation** and **ventricular tachycardia**.

In **atrial fibrillation**, the atria (the two upper chambers of the heart) beat irregularly and very fast. This can lead to feelings of palpitations or fluttering in the chest. The condition can usually be controlled with medication (see page 36). For more information on atrial fibrillation, see our booklet *Atrial fibrillation*. (To order our booklets see 'More information' on page 64).

Ventricular tachycardias are arrhythmias that affect the ventricles – the lower pumping chambers of the heart. The ventricles take over the heartbeat independently of the SA node, leading to a rapid heartbeat. Ventricular tachycardias can be controlled with medication (see page 36), but they can sometimes lead to more life-threatening arrhythmias, and the risk of sudden death. For more information on sudden death, see overleaf.

Heart block

A small number of people with hypertrophic cardiomyopathy may get heart block. This is when the electrical impulse travels down to the ventricles slowly, or may even be completely blocked. This affects the way that the heart contracts. For information on treatment for heart block, see page 44.

Endocarditis

This is an infection of the endocardium, the lining of the heart. It is a rare but serious condition, but it can be treated. For more information on endocarditis see page 44.

Is there a risk of sudden death with hypertrophic cardiomyopathy?

Hypertrophic cardiomyopathy is a common disease, and the majority of affected people remain well and have few or no symptoms. Research has shown that, with proper treatment and follow-up, most people with the condition live a normal life. However, because there is a very small risk of getting a life-threatening arrhythmia, a small proportion of people with hypertrophic cardiomyopathy are at risk of sudden cardiac death.

Sudden cardiac death is the result of an arrhythmia (when the heart beats too fast and chaotically), which may eventually cause the heart to stop beating. This is called a **cardiac arrest**. (This is different to a 'heart attack', which happens when one of the coronary arteries that supplies the heart with blood becomes blocked and the heart muscle which it supplies may be starved of oxygen).

For information on treatment for people with hypertrophic cardiomyopathy who may be at risk of sudden death, see page 44.



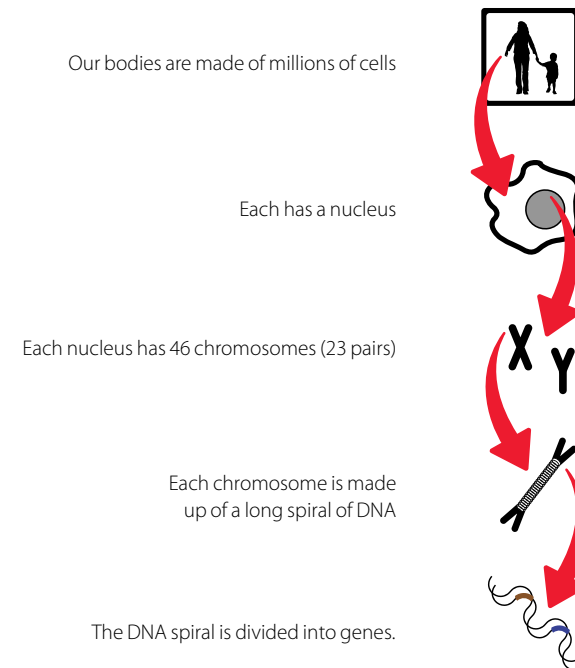
Testing, treatment and your family

Implications of a diagnosis of hypertrophic cardiomyopathy

How do people get hypertrophic cardiomyopathy?

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. Your genes give the instructions that are needed for development and growth of all the cells in your body, and they determine things like hair colour, eye colour, height and blood type.

Genes are arranged end to end along a threadlike structure called a chromosome. 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.



We inherit one set of chromosomes from each of our parents. As the chromosomes are made up of genes, this means that we inherit one set of genes from each parent. This explains why we inherit certain characteristics from our parents.

It may help to think of yourself as a book:

- The chromosomes are the chapters. There are 22 pairs of chromosomes and one pair of sex chromosomes, (men have an X and a Y chromosome XY, and women have two X chromosomes XX), so 23 chapters.
- The genes are coding for proteins that dictate characteristics like hair and eye colour or your height. The coding is like the words in the chapters.
- The code in your DNA is like the letters in the words. 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 that particular type of cell. The same mistake will be passed on to the next and following generations. This is known as a genetic mutation. Some changes have little or no effect, but others can result in heart problems such as hypertrophic cardiomyopathy, or other genetic conditions such as cystic fibrosis (a disease affecting the lungs and pancreas).

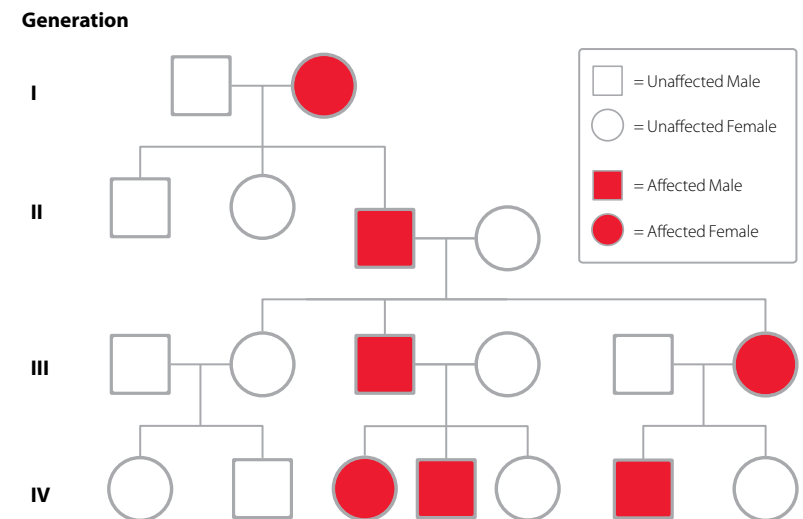
Research has shown that about 5 or 6 in every 10 families with hypertrophic cardiomyopathy have mutations in one of 12 genes that are important in the development of heart muscle cells. The discovery of where and how these gene mutations happen has helped us understand how hypertrophic cardiomyopathy develops. If someone is found to have hypertrophic cardiomyopathy and a mutation is identified, this means that their close family members can be tested to see if they have the same mutation.

How is hypertrophic cardiomyopathy inherited?

Drawing a medical 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 an inheritance pattern. The inheritance pattern for hypertrophic cardiomyopathy is known as autosomal dominant. This means that each child of a person who has hypertrophic cardiomyopathy has a 50:50 or 1 in 2 chance of inheriting the condition (see the diagram below). The condition may be passed on from an affected male or female. It does not skip generations.

A family tree

This family tree shows four generations affected by hypertrophic cardiomyopathy. Each child of an affected person has a 50:50 chance of inheriting the condition.



If you inherit the condition, there is no way of knowing how severe it might be. Two people in the same family may have different symptoms, and the symptoms may be mild in one person but severe in the other. However, if you don't inherit the mutation, you cannot pass it on to your children, even if other close members of your family have it.

If you don't know your medical family history, or if this is the first time that hypertrophic cardiomyopathy seems to have been diagnosed in your family, any children of the affected person probably has a 50:50 or 1 in 2 chance of having the condition.

What if something is found in you?

If your doctor thinks that you may have hypertrophic cardiomyopathy, it is important that you have an assessment (tests) to find out whether you do have the condition. We describe all the tests that you may need to have as part of this assessment on page 32. Confirming a diagnosis will help the doctors to decide what 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 you to live a normal life.

It may also be possible to have a genetic test to identify the specific genetic mutation that has caused your condition. Genetic testing needs to be done at a specialist clinic for inherited heart conditions. Genetic counsellors will explain, to people who have a genetically determined disease, how likely it is that they will pass the abnormal gene on to their children. Brothers and sisters of the affected person can also be affected and may be tested.

If you don't know where to go for the assessment, call the **BHF Genetic Information Service** on **0300 456 8383** to find out where your nearest clinic for inherited heart conditions is, and for information and support about genetic testing.

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. Buying genetic tests without

the appropriate support and guidance could give you misleading and inaccurate information.

What about screening for your family?

The majority of people with hypertrophic cardiomyopathy have at least one other affected first-degree relative. (A first-degree relative means a parent, brother, sister or child). All first-degree relatives should be provided with sufficient information to allow them to decide whether they wish to be screened for the condition. Screening usually involves having some tests at a hospital as an outpatient. We describe these tests on page 32.

The children of affected parents should be screened every three years until puberty, and then every year until they reach the age of 20. Screening usually involves having an ECG and an echocardiogram (see page 33). If there is no evidence of hypertrophic cardiomyopathy in their early adulthood, it is unlikely that the condition will develop in later life.

In most adults where the tests show no abnormalities, it is most likely that the person does not have hypertrophic cardiomyopathy. However, in a small number of people, the abnormal gene may still be present even though there is no physical evidence of the disease.

If another family member with hypertrophic cardiomyopathy has had a genetic test which has found a particular mutation to be the cause of the condition, it may be possible to screen other family members to see if they have the same genetic mutation.

Assessment at a clinic for inherited heart conditions

Below we describe what happens when someone has an assessment at a clinic for inherited heart conditions to find out if they have inherited a particular heart condition. These clinics are usually in a hospital.

Medical history

Your doctor will ask you lots of questions about your medical history. They will also ask you about your family, including your parents and possibly your grandparents. They may ask you if you have ever had symptoms such as blackouts or palpitations, or if there have been any sudden deaths in your family, including any cot deaths. If there is a history of sudden death, any coroner's or pathologist's reports that you may have could be helpful.

Medical examination and tests

Most people with hypertrophic cardiomyopathy don't have any visible physical signs of the condition. Your doctor will take your pulse. If this feels 'jerky' or quite strong, this could suggest that the heart muscle is thickened and that your heart is working harder than normal. A heart murmur is found in 3 or 4 in every 10 people with hypertrophic cardiomyopathy. The murmur may be due to an obstruction in the outflow tract or a leaking mitral valve. See page 16.

Your doctor will do an ECG to look at the electrical rhythm of your heart. You will also need to go to a hospital for a chest x-ray and an echocardiogram to confirm whether you have hypertrophic cardiomyopathy. You may also need further tests to find out how thick the muscle is and how much of it is affected. And you may need to have other tests to measure how well you can exercise. We explain more about all of these tests on the next pages.

NON-INVASIVE Tests marked below with this symbol are 'non-invasive', which means that the test does not involve penetrating the skin or body.

ECG **NON-INVASIVE**

Also called an **electrocardiogram**

This is the most basic test. It involves taping electrical leads onto your legs, arms and chest and taking readings of the electrical activity of your heart. These are printed out onto paper for the doctor to examine.

Exercise test **NON-INVASIVE**

Also called an **exercise ECG**

This test is the same as the ECG described above, but is recorded before, during and after a period of time spent exercising on a treadmill or an exercise bike. This allows the doctor to examine any changes in the electrical patterns that occur with exercise, and analyse any abnormalities.

24-hour ECG monitoring **NON-INVASIVE**

Also called **Holter monitoring**

This test involves using a digital recording device in the shape of a pager. You wear the device on a belt round your waist. Four or six ECG leads from the device are taped to your chest. The device records the electrical activity of your heart for 24 to 48 hours, or for up to seven days. The doctor can then analyse the electrical activity and rhythm of your heart to find out if you have any arrhythmias, such as atrial fibrillation or ventricular tachycardia.

Echocardiogram

Also called an **echo** **NON-INVASIVE**

This test uses ultrasound waves to look at the structure of the heart. It produces a picture of the heart and allows doctors to measure the heart muscle and identify areas of abnormal thickness. It will also look at the heart valves and identify if there is any regurgitation (see page 16). Additional equipment, called Doppler ultrasound, can produce a colour image of blood flow within the heart and provide information on how well the heart is working.

Cardiopulmonary exercise test NON-INVASIVE

Some hospitals may also ask you to do a cardiopulmonary exercise test. This test analyses the efficiency of the heart muscle by measuring the amount of oxygen your body uses during exercise. You will be asked to breathe into special equipment while you are exercising. If the efficiency of your heart is low, this may suggest that you have cardiomyopathy.

Coronary angiogram and electrophysiological study

If you have chest pain or an abnormal heart rhythm, your doctor may suggest that you have other tests such as a coronary angiogram or an electrophysiological study (EPS). Both these tests are performed in an X-ray laboratory. The tests allow doctors to see parts of the body, and any medical items such as cardiac catheter tubes or pacing wires, using an X-ray camera. You will be asked to lie down on a special table and will be given a local anaesthetic in your groin. The doctor will then place fine tubes, called cardiac catheters or electrodes, into blood vessels in your groin. These are gently passed through to the heart.

During a **coronary angiogram**, the coronary arteries (the arteries that supply blood to the heart muscle) are injected with a dye to reveal any narrowing that could be caused either by cardiomyopathy or coronary heart disease.

An **EPS (electrophysiological study)** involves placing electrical leads inside the heart to analyse its electrical properties and to bring on arrhythmias. This test can be useful for diagnosing and treating abnormal heart rhythms.

Magnetic Resonance Imaging (MRI)

For this test, you lie in a short 'tunnel', around which there is a large magnet. Short bursts of magnetic fields and radio waves from the MRI scanner allow images to be created, processed and analysed. You must lie still while the scan is done. The whole test takes about an hour. An MRI is not painful or uncomfortable. However, some people with claustrophobia (fear of enclosed spaces) may find they cannot cope with having this type of scan. An MRI scan uses a magnetic field to create images of the heart. An MRI is very good at showing the structure of your heart and blood vessels. It can also measure the flow of blood through the heart and some of the major arteries. An MRI scan can also show where the heart is working abnormally in conditions such as cardiomyopathy. And it can identify defects in the structure of the heart.

You cannot have an MRI scan if you have a pacemaker or an ICD, because it can interfere with the way these devices work.

Radionuclide tests and CT scans

Radionuclide tests and CT scans give more detailed information than the exercise ECG test (see page 33). They are less common than electrocardiograms (ECGs) or echocardiograms, because the specialised equipment and staff are only available at some hospitals in the UK.

For more detailed information about these and other tests, and for information about levels of radiation with these investigations, see our booklet *Tests for heart conditions*. (To order our booklets see 'More information' on page 64).

Treatments for hypertrophic cardiomyopathy

Hypertrophic cardiomyopathy is not curable, but most symptoms caused by the condition can be controlled by using medicines. Some people may need to have other treatments as well. You will find information about these on page 40. People with hypertrophic cardiomyopathy with obstruction may need additional treatment (see page 42).

Medicines

Below we describe the medicines most commonly used for people with hypertrophic cardiomyopathy. Different people need to take different medicines, depending on their condition and symptoms.

Beta-blockers

Beta-blockers act by slowing the heart rate. This reduces the amount of work the heart has to do, so that it needs less oxygen, blood and nutrients. Some beta-blockers can help control abnormal heart rhythms.

Many preparations of beta-blockers are available, but they differ only slightly from each other.

Unwanted effects – Serious side effects are rare if beta-blockers are used carefully. Minor side effects are common but they tend to lessen as time goes by. The minor side effects include tiredness, fatigue, and cold hands and feet. Other less frequent effects include feeling sick, diarrhoea, skin rashes, impotence, nightmares and dizziness.

You should not stop taking beta-blockers suddenly without medical advice, as coming off them too quickly can make angina worse.

Calcium channel blockers (calcium antagonists)

The heart needs a regular flow of calcium for the muscle cells in the heart to work properly. Calcium channel blockers reduce the amount of calcium entering the muscle cells of the arteries (including the coronary arteries) and cause them to relax and widen. As a result of this, the 'resting phase' of the heart's pumping cycle lasts longer. (The resting phase is when the heart rests in between heartbeats and the coronary arteries fill up and

supply the heart muscle with blood). This means that the heart receives a better supply of blood and has to do less work to pump enough blood around the body.

Some calcium channel blockers may increase or reduce the heart rate while you are resting, while others may have no effect on the heart rate. So different calcium channel blockers are used for different heart conditions.

Unwanted effects – Serious side effects are not common. Minor effects include flushing, headache, dizziness, feeling faint or fainting, swollen ankles, indigestion, feeling sick and vomiting.

Anti-arrhythmic medicines

Anti-arrhythmic medicines are medicines for controlling the rhythm of the heart. Beta-blockers, and the calcium channel blocker verapamil, are often used to treat disturbances of the heart rhythm (arrhythmias), but there are several other medicines that are used almost exclusively for this purpose. One of these is amiodarone.

Amiodarone

Amiodarone is very effective in controlling disturbances of the heart rhythm, especially atrial fibrillation (see page 23).

Unwanted effects – At low doses, amiodarone is well tolerated, but it has important side effects. It may produce headache, flushing, dizziness and stomach upsets. More seriously, and more rarely, it may cause disorders of the thyroid gland, lungs and liver. To avoid these complications, you will need to have regular blood tests. You may also have chest X-rays and tests to see how your lungs are working.

Amiodarone tends to make the skin very sensitive to sunlight so, if you are taking this medicine, you should use a powerful sunscreen cream when you are in strong daylight or bright sunshine, and wear a hat.

Anticoagulants

Some people with hypertrophic cardiomyopathy develop atrial fibrillation (see page 23). Atrial fibrillation carries a risk of blood clots forming. Blood clots are made up of platelets (tiny blood cells) clumped together, and a protein called fibrin. Anticoagulants prevent fibrin from forming and so prevent clots from forming too. However, in doing so, they may cause internal bleeding or make bleeding from a minor injury worse. There are two main types of anticoagulants – heparin and warfarin.

Heparin is injected into a vein and has an immediate effect in preventing further blood clots from forming. This is known as intravenous heparin and is only given in this way if you are in hospital. Another form of heparin called 'low molecular weight heparin' may be given by injection just under the skin, over a longer period of time. This can be given to you either in hospital, or by a nurse at home.

Warfarin (or another oral anticoagulant) is given when long-term prevention of clotting is needed. This medicine is most often used for people with disease of the heart valves, especially those who have been given an artificial heart valve, or for some people who have an irregular heart rhythm such as persistent atrial fibrillation.

Because the desired effect of anticoagulants is to make the blood thinner so that it doesn't clot so easily, you will need regular blood tests to make sure that the clotting activity of the blood is within safe but effective levels. At the start, these tests may be carried out almost every day, but later on they will be done only every four to eight weeks. If you have been taking warfarin for a long time and your condition is stable, you may be able to get a home-testing kit and do the tests yourself.

If you are taking oral anticoagulants, you should check with your doctor or pharmacist before taking any other medicines – both over-the-counter and prescription medicines. This is because oral anticoagulants interact with many medicines including antibiotics, aspirin and cimetidine, and with some medicines that are used to treat arthritis, gout, epilepsy, high blood cholesterol and heart rhythm disorders.

Alcohol increases the effect of warfarin, so it is important to avoid excessive drinking or binge drinking.

If you are taking oral anticoagulants, you should also avoid drinking cranberry juice as this can affect the amount of time it takes for your blood to clot. Your anticoagulation clinic should be able to give you advice on what changes you may need to make to what you eat and drink.

If you are taking anticoagulants, you should always carry an **Anticoagulant card** and remember to tell any doctors, dentists and nurses who are treating you that you are taking anticoagulants.

Any of the following symptoms could mean that your dose of anticoagulants may be too high:

- prolonged bleeding from cuts
- bleeding that does not stop by itself
- nose bleeds that last for more than a few minutes
- bleeding gums
- severe bruising
- red or dark brown urine
- red or black stools
- for women, heavier bleeding during periods, or other vaginal bleeding.

If you are worried, contact your GP or anticoagulant clinic or the casualty department at your local hospital. Make sure that you have your dosage record card and any other medications with you.

Diuretics

Diuretics, or water tablets, increase the output of water and salt in the urine. They are particularly valuable in reducing the workload of the heart by making sure that the body does not hold too much salt or water.

Diuretics can also help to control your blood pressure. If you have too much fluid in your body, your heart has to work harder to pump it around the body and the extra workload increases your blood pressure.

There are three main types of diuretic – thiazide diuretics, loop diuretics and potassium sparing diuretics. Thiazide diuretics (such as bendroflumethiazide) and loop diuretics (such as furosemide and bumetanide) can cause you to lose potassium, so your doctor will arrange a blood test a few weeks after you start taking your tablets, to check the potassium level in your blood. If this is getting low, you may be given potassium supplements or a potassium sparing diuretic instead, to correct the problem with the potassium level.

If you are taking a diuretic, you should not have too much salt in your food, as this will counteract the effects of the diuretics. Don't add any salt to food during cooking or at the table, and avoid salty foods. Many processed foods and ready meals contain high levels of salt. It is also important to avoid using salt substitutes as these contain potassium which may have an effect on your blood test results.

Unwanted effects – People with diabetes may find that diuretics raise their blood sugar. People with gout may find that diuretics make their condition worse. For more information about medicines see our booklet *Medicines for the heart*. (To order our booklets see 'More information' on page 64).

Other treatments for hypertrophic cardiomyopathy

Pacemakers and ICDs

If you are at high risk of sudden death (for example, if you have already had a cardiac arrest), or if medicines have failed to control your symptoms, your doctor may advise you to have a pacemaker or an ICD fitted, as well as taking your medication. ICD stands for 'implantable cardioverter defibrillator'.

A pacemaker and an ICD both consist of a very small box containing a battery, and special electrode leads. The box is inserted under the skin and attached to the heart by the electrode leads.

A **pacemaker** controls the heart rate and stops any excessive slowing of the heart that could trigger an arrhythmia. The pacemaker is usually

implanted just under your left collarbone. The procedure usually takes about an hour and is normally done with a local anaesthetic and sedation. You will need to have follow-up checks every three to twelve months. The pacemaker battery usually lasts between six and ten years (and sometimes even longer). When a new battery is needed, the box containing it can be replaced easily. For more information on pacemakers, see our booklet *Pacemakers*. (To order our booklets see 'More information' on page 64).

An **ICD** acts in the same way as a pacemaker, but it can also identify any dangerous arrhythmias and deliver an electrical shock to 'reset' the heart. Some people have described the shock as feeling like having a 'kick in the chest'. An ICD is slightly larger than a pacemaker and is usually positioned under the chest wall muscle below the left shoulder. The procedure may take between one to three hours. Most people have a local anaesthetic as well as sedation, but some may have a full (general) anaesthetic. You will need to have check-ups at the ICD clinic once every three to six months. The battery lasts between four and eight years. When a new battery is needed, the box containing it can be replaced easily. For more on ICDs, see our booklet *Implantable cardioverter defibrillators (ICDs)*. (To order our booklets see 'More information' on page 64).

Heart transplantation

For a very small number of people, heart transplantation may be considered. For more information, see our booklet *Heart transplantation*. (To order our booklets see 'More information' on page 64).

Treatment for hypertrophic cardiomyopathy with obstruction

About 1 in every 4 people with hypertrophic cardiomyopathy has an obstruction to the outflow of blood from the heart (see page 16). This is known as left outflow tract obstruction. It can cause chest pain, breathlessness and fainting. It can also make it more likely that you will develop arrhythmias, in particular atrial fibrillation.

Treatment with medications

Medicines are used to reduce the force of the contraction of the heart. The medicines that are often used are beta-blockers and calcium channel blockers (see page 36).

Sometimes a medicine called **disopyramide** is used as well. This medicine was developed mainly for treating abnormal heart rhythms, but is also very useful for treating symptoms relating to hypertrophic cardiomyopathy with obstruction, such as chest pain and shortness of breath. If you need to take this medicine, your doctor will start you on a low dose, and gradually increase the dose while keeping you under medical supervision. Common side effects of disopyramide include dry mouth, blurred vision, difficulty passing urine, and constipation. Once you have been taking this drug for a short while, you will need to have an ECG to monitor the effect of the disopyramide on your heart rhythm, as a small number of people can develop arrhythmias as a result of taking this drug.

Other treatments

A small number of people who have hypertrophic cardiomyopathy with obstruction will still have symptoms even though they are taking the maximum amount of medicines. So they may need further treatment to help reduce their symptoms and improve their heart muscle function.

There are two main types of treatment:

- surgical myectomy
- alcohol septal ablation.

Both of these treatments carry a small risk, and are therefore only used for people who have hypertrophic cardiomyopathy with obstruction, and who have severe symptoms.

Surgical myectomy

A myectomy is an open-heart surgical procedure to remove part of the thickened muscle that is causing the obstruction, allowing the blood to circulate more freely. It involves removing a portion of the thickened muscle from the septum, thus widening the outflow tract and relieving the obstruction (see the diagram on page 16). Usually the mitral valve is normal but in some people this valve may need to be repaired or replaced. If this is necessary, it is done during the same operation.

What are the risks?

As with any open-heart surgery, there are risks associated with the surgery itself and with having an anaesthetic. You should discuss the specific risks of this procedure with your cardiologist. For more information, see our booklet *Having heart surgery*. (To order our booklets see 'More information' on page 64).

Alcohol septal ablation

This procedure reduces the thickening of the top part of the septum without the need for open-heart surgery. It involves inserting a catheter into the groin in the same way as for an angiogram (see page 34). A small amount of alcohol solution is then injected into a minor branch of the coronary artery that supplies the upper part of the septum. The alcohol destroys the part of the thickened heart muscle that is causing the obstruction. This makes the muscle thinner and relieves the obstruction.

What are the risks?

In a small number of cases, the alcohol injection may damage the electrical system of the heart. If this happens, you may need to have a pacemaker fitted after this procedure (see page 40). You should discuss this with your cardiologist.

Treatment for other conditions which can occur as a result of hypertrophic cardiomyopathy

On page 23 we explained that certain other conditions can occur as a result of hypertrophic cardiomyopathy. These are the treatments you may need if you have developed one of those conditions.

The two **arrhythmias**, ventricular tachycardia or atrial fibrillation, usually require treatment with medicines (see page 23).

Heart block can be treated by implanting a pacemaker (see page 40).

If you get **endocarditis** (an infection of the lining of the heart), you will need to go into hospital for several weeks and have antibiotics through an injection in a vein. In the past, people who had ever had endocarditis were advised to take antibiotics before having dental treatment, but this advice has now changed and you don't need to take antibiotics unless you are specifically told to do so.

If your doctor thinks you are at **increased risk of sudden death**, you may be offered treatment which could include taking medication, or having an ICD (implantable cardioverter defibrillator) fitted (see page 41), or both.

Everyday life



Living with hypertrophic cardiomyopathy

Who should be involved in routine medical care?

Your GP should be involved with your general everyday care. He or she can discuss your condition with your cardiologist and make any changes to your treatment. You will probably see your cardiologist once a year, but this will depend on your overall condition and your symptoms.

Do I have to make any changes to my everyday life?

For many people, hypertrophic cardiomyopathy does not interfere with their lifestyle. However, there are some things that could make your symptoms and condition worse. On the next few pages we talk about the everyday things that concern people with hypertrophic cardiomyopathy.

Exercise

Your heart is a muscle and needs exercise to stay healthy. However, some people with hypertrophic cardiomyopathy become short of breath or have chest pain (angina) when they are physically active. Most people with hypertrophic cardiomyopathy will be advised not to take part in competitive sports or other strenuous physical effort. This can be difficult for younger people who have previously taken part in this type of activity. You should always discuss with your doctor the type and level of physical activity that you should do.

Diet, alcohol and weight

Being overweight places an extra strain on the heart. Eating a healthy, balanced diet will help you to maintain a healthy weight.

Drinking more than the recommended amount of alcohol can damage your heart health and can lead to high blood pressure. See our booklet *Keep your heart healthy* for information on sensible limits, or ask your doctor what is a sensible limit for you. (To order our booklets see 'More information' on page 64). Alcohol is also a depressant, so it can make feelings of anxiety or depression worse.

If you have left ventricular outflow tract obstruction, you may find that alcohol makes your symptoms worse.

Weight gain

If you are eating a healthy diet but you notice that you seem to have been putting on weight over a few days (about a half to 1 kilo, or 1 or 2 pounds) you will need to see your doctor. The weight gain could be due to fluid retention which can be treated by adjusting your medicines.

Smoking

Although tobacco smoking is not directly associated with hypertrophic cardiomyopathy, smoking will increase the risk of developing coronary heart disease and of reducing the flow of blood to your heart muscle. It also reduces the amount of oxygen being carried in the blood and makes your heart work harder. For information on giving up smoking see our booklet *Smoking and how to give up*. (To order our booklets see 'More information' on page 64).

Sex

Sexuality, sexual intercourse, pregnancy, erectile dysfunction (when a man is unable to get or maintain an erection), loss of sex drive, and safe sex are some of the concerns that people with hypertrophic cardiomyopathy have. Feeling embarrassed and not talking about your concerns can lead to feelings of inadequacy and delays in finding appropriate solutions.

Knowing your limitations and what brings on your symptoms can help you to enjoy a full and sexually active life. Like any other physical activity, having sex can increase the heart rate and blood pressure. This increases the work of the heart and, for some people with a heart condition, sex may bring on symptoms such as breathlessness or chest discomfort. However, sex is just as safe as other equally energetic forms of physical activity or exercise.

To reduce the chance of having angina symptoms during sex, avoid having sex after a heavy meal, and try not to be too energetic at the start of your sexual activity.

Loss of sex drive is not uncommon in people with a heart condition. Some men may experience impotence. This may be the result of taking certain medicines, such as beta-blockers, which can affect your sex drive. Or it may

be the result of the emotional stress you are feeling, or the result of poor blood circulation or diabetes.

Other common causes of loss of sex drive are:

- depression
- concerns and fear about how safe it is for you to have sex
- anxiety about the possibility of your ICD delivering an electrical shock during sexual activity
- lack of communication between you and your partner.

Impotence is a common problem so, if you are having difficulties, talk to your doctor about it. Talking to your partner can also help to lessen your concerns and fears and help your partner to understand your situation.

If you have a heart condition, you should be cautious about taking PDE-5 inhibitors such as Viagra. You should not take these medications if you are taking GTN or any other medication containing nitrates. PDE-5 inhibitors can also interfere with other medicines for your heart, so always check with your doctor beforehand.

If you have hypertrophic cardiomyopathy with left ventricular outflow tract obstruction you should not take this type of medication as it can make your symptoms worse.

General health

Do I need to have a flu vaccination?

Having a flu vaccination will not prevent you from getting the flu viruses, but it will reduce the severity of the flu if you do get it. Every year there is a different strain of the flu virus and a new vaccine is developed. If your doctor recommends that you have the flu vaccine, you will need to have a vaccination each year.

What if I need an anaesthetic?

In most people with hypertrophic cardiomyopathy, having a general or local anaesthetic is not associated with any additional risk. Your anaesthetist will monitor your blood pressure and heart rate carefully during your surgery or procedure. Some spinal blocks or epidurals can cause a drop in blood pressure, so they should be used with caution.

Anxiety and depression

Finding out that you have a diagnosis of cardiomyopathy can be distressing. In the early days it is normal to feel shock, anger and disbelief. We all have different ways of adjusting, and there is no right or wrong way. Stress, fear and anxiety can sometimes help us to re-examine and to change our lifestyles. However, when emotions like this persist for long periods of time, or begin to overwhelm you, they can lead to anxiety and depression that may need medical treatment.

Talking to someone can be very helpful. Friends and relatives may be able to understand how the condition is affecting you, but sometimes it is easier to talk to someone who's not so closely involved. Talking to other people with cardiomyopathy may help you come to terms with your condition and help you to see that you are not alone. There are also many organisations and charities that can help you to cope. See page 67 for details.

Recognising and understanding the cause of your anxiety can help you to deal with it. Using relaxation techniques can help. If simple techniques don't help, or if you feel overwhelmed with your anxiety, you should speak to your doctor who will be able to decide if you need medical treatment.

Depression

It's normal to feel fed up or miserable from time to time, and feelings like this usually don't last for long or interfere significantly with our lives. However, if these feelings persist and severely interfere with your life, you may have clinical depression. Clinical depression can last for months and can affect you in a variety of ways, both physically and psychologically. You may be unable to sleep properly, lose your appetite, or frequently think that life is not worth living. Depression usually comes on gradually. Some people may not recognise that they are becoming depressed because they are paying more attention to their physical symptoms than their psychological state.

Having a routine and remaining active can help to deal with mild depression. It can help you to feel in control and get back to a natural sleeping pattern. Your doctor may suggest that taking sleeping tablets for a short time will help you. Although hypertrophic cardiomyopathy often restricts vigorous exercise, it shouldn't stop you being active. (See page 47 for more on exercise).

Alcohol is associated with a feeling of being happy but it is actually a depressant. You should not exceed the recommended levels of alcohol. See our booklet *Keep your heart healthy* for information on this, or ask your doctor. (To order our booklets see 'More information' on page 64).

If you have severe or chronic depression, you may benefit from taking anti-depressants. They help by altering the transmission of chemical substances in the brain, thereby improving your mood. It can take several weeks for you to feel the benefits of anti-depressants. Your doctor may also suggest that you would benefit from speaking to a clinical psychologist.

Driving

The UK Driver and Vehicle Licensing Agency (DVLA) gives the following advice for people with hypertrophic cardiomyopathy:

- If you have an ordinary driving licence, you can continue driving unless there are distracting or disabling symptoms, in which case you should notify the DVLA.
- If you have a licence to drive a heavy goods vehicle or a passenger-carrying vehicle, you will not be allowed to drive these vehicles.

For more details, contact the DVLA on 0870 240 0009.

Work

Most people who are diagnosed with hypertrophic cardiomyopathy will be able to continue in their normal job. However, if you have a heavy manual job which involves strenuous activity, you should discuss this with your doctor.

For some occupations – such as airline pilots and heavy goods vehicle drivers – there are strict guidelines about whether you can continue in your job if you have hypertrophic cardiomyopathy. Whatever your job, your doctor may be able to provide additional information for your employer which could mean that you may be able to continue in your job. If your employer (or potential employer) asks you to fill out a medical questionnaire, you must tell them about your condition.

Holidays and travel insurance

If you have hypertrophic cardiomyopathy, there are no restrictions on flying provided that your symptoms are well controlled. If you think you might need oxygen during the flight, speak to your doctor about this and then to your travel company. If you have heavy bags, make sure there is someone who can lift them for you. Remember to leave enough time to get to the departure gates without rushing. Many of them are a long way from the security gates. If you get breathless easily, you may also want to organise in advance to have help at the airport. The airlines can often provide transport to a departure gate or a wheelchair for you to use.

If you want to travel within the European Union, you should get a European Health Insurance Card (EHIC) and take it with you when you're travelling. This card ensures EU nationals obtain the same level of health care as a local resident in a member country when travelling. You can apply for this card at most post offices.

If you are diagnosed with hypertrophic cardiomyopathy, an insurance company may charge more for your travel insurance. For a list of insurance companies who are sympathetic to people with heart conditions, contact the British Heart Foundation or the Cardiomyopathy Association. Their contact details are on page 64–65.

Life insurance and mortgages

If you have hypertrophic cardiomyopathy, you may have difficulty obtaining life insurance or a mortgage.

The Association of British Insurers (ABI) says that insurers will take into account a family history of all medical conditions. They will not ask people to take genetic tests when applying for life insurance. However, if you wish to take out a new life insurance policy, you will be required to report the results of any genetic tests you have already had, unless otherwise indicated by the life insurance company.

Financial support

Some people with hypertrophic cardiomyopathy may be able to apply for Disability Living Allowance. This is a social security benefit for people who have an illness or disability and who need help getting around or help with personal care. There are other benefits and allowances available for those people whose symptoms result in severe restrictions.

To find out more about the benefits you are entitled to, call the Benefit Enquiry Line on 0800 882 200 (a freephone number), or visit your social security office, citizens advice bureau or local social services department.

For information about Tax Credits, contact the Inland Revenue helpline on 0845 300 3900.

Pregnancy and childbirth

Pregnancy is usually safe for the majority of women with hypertrophic cardiomyopathy. However, as for women with any heart condition, pregnancy carries a slight additional risk for women with hypertrophic cardiomyopathy. This is because pregnancy increases the workload of the heart. If you know you have hypertrophic cardiomyopathy, you may be concerned about taking medications during pregnancy and the effect that they may have on your baby. If you are planning a family, you should discuss these concerns with your doctor before becoming pregnant.

If you are thinking of having an epidural during your labour, you should discuss this with your doctor early on in your pregnancy, as an epidural can cause a significant fall in blood pressure.

For some women, becoming pregnant produces symptoms of cardiomyopathy for the first time. This may be difficult to deal with, but your obstetrician will liaise closely with your cardiologist to look after you and your baby.

Should I have children?

You will find information about the risk of passing on the condition to your children on page 29. The decision about whether to have children is one that you should make only after discussing it with your partner and at a clinic for inherited heart conditions. To find out where your nearest clinic is, call the **BHF Genetic Information Service** on **0300 456 8383**.



Looking forward

The future

Research is being carried out into many different aspects of hypertrophic cardiomyopathy, in many countries.

We have already identified 12 of the common genetic mutations that cause this condition, and ongoing research aims to identify the other gene, or genes, involved, and to understand how these gene mutations result in the changes to the heart muscle.

Other research aims to discover the reasons for the symptoms of hypertrophic cardiomyopathy and the causes of sudden death.

Developments in genetic testing have enabled screening and testing of close family relatives to find out if they carry the same gene mutation. The results of this research can help improve the quality of life, treatment and monitoring of people with hypertrophic cardiomyopathy, including those who do not yet have symptoms.

Technical terms

A

Angina

Heaviness or tightness in the centre of the chest, which may spread to the arms, neck, jaw, back or stomach. Or it may affect just the neck, jaw, arms or stomach.

Angiogram

An X-ray picture of the blood vessels which shows whether the arteries are narrowed and, if so, how narrow they have become. An angiogram can be used to examine the coronary arteries (a coronary angiogram) or other arteries in your body.

Anticoagulation

Treatment with medicine, to thin the blood and reduce the risk of clots.

Aorta

The large artery (blood vessel) leading out of the left side of your heart and supplying the whole body with blood.

Apical hypertrophy cardiomyopathy

Thickening of the heart muscle at the tip (or apex) of the left ventricle.

Arrhythmia

A disturbance of the heart's rhythm.

Atrial fibrillation

A fast, irregular heart rhythm.

Atrium

One of the two top chambers of the heart.
(The plural of 'atrium' is 'atria')

Autosomal inheritance

Where a condition is passed on in a family from one generation to the next without skipping any generations.

C**Cardiac arrest**

The state of the heart when it is pumping so erratically or ineffectively that there is no significant blood pressure to supply blood to the heart and brain. If basic life support is not started within two minutes, there could be permanent brain damage, and if left untreated the person will quickly die.

Cardiologist

A doctor specialising in diseases of the heart.

Cardiomyopathy

Any disease of the heart muscle that is not caused by narrowings in the coronary arteries, valve disease or high blood pressure.

Cardioversion

A procedure to restore a regular heart rhythm.

Chromosome

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

Concentric hypertrophy

A condition where thickening occurs equally throughout the wall of the ventricle.

D**Diuretics**

Also known as 'water tablets'. Diuretics increase the output of water and salt in the urine.

DNA

The genetic code from which proteins – 'the building blocks of life' – are made. We all receive a copy of half of each of our parents' DNA when the egg and sperm meet to conceive a new human being.

Doppler ultrasound

A test usually combined with an echocardiogram to produce a colour-coded image of blood flow within the heart.

E**Electrophysiological study (EPS)**

A technique for detecting and analysing abnormal heart rhythms.

Endocarditis

An infection of the inner lining of the heart, usually affecting the valves.

G**Gene**

The segment of DNA responsible for the production of a specific substance such as a protein, which in turn forms the basis for the body to exist and function.

H**Heart block**

A failure of the electrical system in the heart to conduct electrical impulses properly from the top chambers (atria) to the bottom chambers (ventricles) via the atrio-ventricular (AV) node. The severity of the condition and the risk associated with it can vary.

Heart failure

When the pumping action of the heart is inadequate.

Hypertrophy

Thickening of muscle or tissue.

I**ICD**

A metal electronic device similar to a pacemaker (see Pacemaker). It is implanted under the chest wall muscle below the left shoulder. It can regulate the rhythm of the heartbeat and, if a dangerous arrhythmia occurs, it can deliver an electrical shock to the heart to restore the normal heart rhythm.

M**Mitral regurgitation**

When the blood flows in a backward direction through the mitral valve.

Mitral valve

The valve on the left side of the heart, between the atrium and the ventricle.

Murmur

The sound of the turbulent flow of blood in the heart, sometimes due to leakage through a valve, or narrowing of valves. It can be heard through a stethoscope.

Mutation

An abnormality 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.

Myectomy

A surgical operation to remove thickened muscle and relieve outflow tract obstruction.

Myocardial disarray

When the cells of the heart muscle are lined up in a disorganised way.

Myocardium

The heart muscle.

O**Outflow tract**

The short channel below the aortic valve, through which the blood flows from the ventricle into the aorta.

P**Pacemaker**

A metal electronic device which can regulate the rhythm of the heartbeat. It is usually implanted just under your left collarbone.

Palpitation

When you become aware of your heartbeat – for example, when it feels as if it is beating abnormally fast or slowly, or irregularly or heavily.

Pulmonary artery

Artery carrying blood from the right side of the heart to the lungs.

S**Septum**

The thick, muscular wall between the left and right sides of the heart.

T**Tachycardia**

A fast heart rate.

V**Ventricles**

The two bottom chambers of the heart.

For 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, on where to go for an assessment. The service is staffed by specialist cardiac nurses and a bereavement counsellor.

BHF publications

Publications in the *Inherited heart conditions* series:

Dilated cardiomyopathy

(Due October 2009)

Arrhythmogenic right ventricular cardiomyopathy

(Due October 2009)

Sudden arrhythmic death syndrome

Inherited heart rhythm disturbances

Other BHF publications

Atrial fibrillation

Implantable cardioverter defibrillators (ICDs)

Keep your heart healthy

Medicines for your heart

Pacemakers

Tests for heart conditions

Smoking and how to give up

Losing someone to heart disease

Offers help and support in coping with the loss of someone due 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 cardiomyopathy

Cardiomyopathy Association

Unit 10 Chiltern Court, Asheridge Road, Chesham, Bucks HP5 2PX

Freephone Helpline 0800 0181024 (Monday–Friday, 8.30am–4.30pm)
Website: www.cardiomyopathy.org
Email: info@cardiomyopathy.org

The Cardiomyopathy Association (CMA) is a registered charity that helps people who are affected by cardiomyopathy. It provides support and information on the different types of cardiomyopathy on its website, in booklets, and in DVDs and videos.

It also offers support through a nationwide network of support groups and people affected by the condition, and through regional information days where cardiologists can provide members with information on the latest research in cardiomyopathy. To become a member of the association, call the helpline above.

For support on coping as a family where there has been a sudden cardiac death

Cardiac Risk in the Young – CRY

Unit 7, Epsom Downs Metro Centre Waterfield Tadworth Surrey KT20 5LR

www.c-r-y.org.uk
www.sads.org.uk
www.cry-csc.org.uk

Phone: 01737 363222
Fax: 01737 36344
Email: cry@c-r-y.org.uk

CRY offers help, support and counselling to families where there has been a sudden cardiac death of an apparently fit and healthy young person.

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