Inherited heart conditions
Sudden arrhythmic death syndrome
Introduction

You may be reading this booklet because a relative of yours – perhaps a member of your own family – has died suddenly and unexpectedly. This is not only a tragedy for the person and all your family, but a great loss for society too. You may still be asking why it happened, and how it could have happened to someone who perhaps seemed so healthy. Or maybe your doctor has suggested that you should have some tests to find out if you have inherited the same medical condition as the person who has died.

An ‘inherited cardiac condition’ means a heart condition which can run in families. It can affect one or several members of the same family. Sadly, some inherited cardiac conditions are often not diagnosed until one person dies suddenly and unexpectedly. There are several different types of inherited cardiac conditions. This booklet concentrates on one type known as Sudden Arrhythmic Death Syndrome – or SADS for short.

This booklet:

- describes how the normal heart works
- explains what the terms ‘sudden cardiac death’ and ‘Sudden Arrhythmic Death Syndrome’ mean
- explains why it is important that the close blood relatives of the person who has died should have an assessment to find out if they have inherited the same condition
- describes the tests your doctor may ask you to have, and
- offers advice on how to live a healthy lifestyle if you are found to have one of the conditions that can sometimes lead to sudden death.

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 51.

We hope that this booklet will help you and your family understand what has happened, and help you come to terms with the event. If you need further support or information, see page 57.
Introduction

The normal heart

The heart is a specialised muscle that contracts regularly and continuously, pumping blood to the body and the lungs. The pumping action is caused by a flow of electricity through the heart that repeats itself in a cycle. If this electrical activity is disrupted – for example, by a disturbance in the heart’s rhythm known as an arrhythmia – it can affect the heart’s ability to pump properly.

The heart has four chambers – two at the top (the atria) and two at the bottom (the ventricles). The normal trigger for the heart to contract arises from the heart’s natural pacemaker, the SA node (sino-atrial node), which is in the right atrium (see the diagram on page 08). The SA node sends out regular electrical impulses, which make the atrium contract and pump blood into the bottom chamber (the ventricle). 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 muscle to contract and to pump blood to the lungs and the body. The nerves that regulate the heart release certain chemicals which circulate in the blood. These chemicals alter the speed of the heart’s pacemaker and the force of the pumping action of the ventricles. For example, adrenaline increases the heart rate and the volume of blood pumped by the heart.
How the heart functions electrically
The heart’s natural pacemaker – the SA node – sends out regular electrical impulses across the top chambers (the atria) making them contract and pump blood into the bottom chambers (the ventricles). The electrical impulse then passes to the ventricles through a form of ‘junction box’ called the AV node. The impulse spreads into the ventricles, causing them to contract and to pump out the blood. The blood from the right ventricle goes to the lungs, and the blood from the left ventricle goes to the body.

Sudden cardiac death and Sudden Arrhythmic Death Syndrome (SADS)

Sudden cardiac death
Sudden cardiac death – or SCD for short – is an unexpected and sudden death that is thought to be, and usually is, caused by a heart condition.

Sudden Arrhythmic Death Syndrome (SADS)
In about 1 in every 20 cases of sudden cardiac death, no definite cause of death can be found, even after the person’s heart has been examined by an expert cardiac pathologist. This is then called Sudden Arrhythmic Death Syndrome – or SADS for short. (It used to be called Sudden Adult Death Syndrome or Sudden Death Syndrome but, because it affects children too, the term Sudden Arrhythmic Death Syndrome is now used). It is thought that cot death (Sudden Infant Death Syndrome, or SIDS) may be partly due to the same causes responsible for SADS.
Understanding your heart

What happens after an unexpected sudden death?

After an unexpected sudden death, it is usual for the coroner in the area where the death has happened to ask for a post mortem to be carried out. This involves the body being examined by a pathologist. Small samples of tissue from organs including the heart are often taken and examined under a microscope. Usually the pathologist can easily detect any abnormality like significant coronary heart disease (furring of the arteries), or pulmonary embolus (a clot on the lung). The coroner will take into account the circumstances of the death and, if necessary, will do tests for signs of any medications or drugs in the body. If it is difficult to assess the heart or to detect any abnormality in it, the pathologist may ask for the help of an expert cardiac pathologist (a pathologist who specialises in the heart) to determine the cause of death.
What causes SADS?

SADS is caused when someone has a ‘ventricular arrhythmia’ – a disturbance in the heart’s rhythm – which brings on a cardiac arrest, even though the person has no structural heart disease. (A cardiac arrest is when the heart is pumping so erratically that there is no significant blood pressure).

There is a group of relatively rare diseases called ion channelopathies that can cause life-threatening arrhythmias and are probably responsible for 4 in every 10 cases of Sudden Arrhythmic Death Syndrome. Ion channelopathies affect the electrical functioning of the heart without affecting the heart’s structure. This means that they can only be detected while a person is alive; they can’t be detected at a post mortem. There are several different types of ion channelopathies including:

- Long QT Syndrome (LQTS)
- Brugada Syndrome
- CPVT (catecholaminergic polymorphic ventricular tachycardia)
- PCCD (progressive cardiac conduction defect)
- IVF (idiopathic ventricular fibrillation), and
- Sodium channel disease.

We describe each of these on pages 14–23.

Other causes of SADS include structural heart disease and conduction disease. Structural heart disease is found to be a cause of SADS in 1 or 2 in every 10 cases. For more on this, see page 23. Conduction disease includes abnormalities in the way that the electrical impulses are conducted through the AV node (for example as in myotonic dystrophy), or when there are ‘extra’ electrical pathways in the heart, as in Wolff-Parkinson-White (WPW) Syndrome. For more information on WPW Syndrome, see the BHF booklet Heart rhythms.

What are ion channelopathies?

Ion channelopathies are rare genetic conditions that are caused by abnormalities of the DNA, known as ‘mutations’. They are usually inherited from parents, although they can occur for the first time in a family. If they occur for the first time they are described as ‘sporadic’.
The mutations affect certain genes – specific segments of the DNA that are responsible for the production of ‘ion channels’ in the heart. An ion is a chemical substance – such as sodium or potassium – that carries an electrical charge and forms the basis of the movement of electricity through the heart muscle. An ion channel is the route that the ions take in and out of the heart muscle cells to allow the movement of electricity. The ion channels regulate the flow of electrical charge. If these channels don’t behave normally, the electrical function of the heart becomes abnormal. The person can then be prone to arrhythmias (disturbances in the heart’s rhythm) that can cause blackouts, cardiac arrest and in some cases sudden death.

In the following section we describe the different types of channelopathies, the tests needed to diagnose them and the treatment that may be needed for each one.

**Long QT Syndrome (LQTS)**

LQTS is the most common and best understood type of channelopathy. It occurs in about 1 in 2,000 people. In 7 in every 10 people with LQTS, the ion channels involved have been identified. In most cases, two of the potassium channels that regulate the movement of potassium ions from the inside to the outside of the cells are affected. In a small proportion of people with LQTS, a sodium channel that regulates the flow of sodium ions from the outside to the inside of cells is affected. In a small proportion of people with LQTS, a sodium channel that regulates the flow of sodium ions from the outside to the inside of cells is affected.

In people with potassium channel associated LQTS, the channels do not behave as efficiently as normal. They let potassium ions out of the cell too slowly. If the sodium channel is affected, too many sodium ions are allowed into the cell (See the LQTS diagram on page15). This results in an electrical disturbance in the cells of the heart called ‘prolonged repolarisation’. This can sometimes be seen on an ECG recording as a lengthening of the time period of a particular part of the heartbeat cycle, known as the ‘QT interval’. This is where the name Long QT Syndrome comes from.

There are two rare forms of LQTS: Andersen’s Syndrome is associated with potassium channel abnormalities, and Timothy Syndrome is associated with calcium channel abnormalities.

These diagrams below show the flow of potassium and sodium ions in and out of the heart’s cells. The thick arrow represents too much flow. The thin arrow represents a reduced flow.

**Normal Heart**

In a normal heart, potassium flows out of the cell to ‘repolarise’ the heart, and sodium flows into the cells to activate the heart.

**LQTS**

In people with LQTS, the flow of potassium is usually reduced. In some people with LQTS, the flow of sodium may be increased.

**What are the symptoms of LQTS?**

LQTS varies greatly in severity. Symptoms vary according to the type of channel involved, whether the person is male or female, their age, and the length of the QT interval on the ECG. Males are more likely to have symptoms before puberty, while females are more likely to have them in adolescence and early adulthood. Relatives from the same family who have inherited the same mutation may have very different experiences. For example, some may have a normal QT interval and not have any symptoms; some may have a very abnormal QT interval but no symptoms; and some may have a very abnormal QT interval and have many episodes of abnormal heart rhythm that put them at risk.

The most common symptom of LQTS is blackouts caused by a heart rhythm disturbance. Sometimes palpitations due to extra or ‘ectopic’ heartbeats can be a problem.
Potassium channel LQTS is associated with sudden death which is related to exercise or when the person has been startled or awoken suddenly (‘sudden arousal’). The sodium channel form of LQTS is associated with death while asleep.

Are there any physical signs of LQTS?
There are no physical signs of LQTS. However, people with Andersen’s Syndrome may also have muscle weakness or minor abnormalities of the skull, chin, fingers and toes.

How is LQTS diagnosed?
Diagnosis involves having an ECG (see page 29). Sometimes experts can tell which ion channel has been affected just by looking at the ECG recording. Unfortunately, in many people who might be carriers, the ECG doesn’t show any sign of the condition. Repeated ECGs, exercise tests and 24–48 hour ECG monitoring may be needed before any hint of the condition is seen, and even then there may be no clear sign of it. We describe all these tests on pages 29–34.

Genetic testing can sometimes identify carriers of LQTS (see page 33). Unfortunately, this form of testing is of limited use at the moment, as 3 in every 10 people who are known to have LQTS do not have mutations of the genes known to be associated with LQTS.

Many families who do have the mutations appear to have a specific change to the DNA code which is not found in other families. This change is known as a ‘private’ mutation. This can make it difficult to decide whether a mutation is causing the disease or not. Things are further complicated by the fact that people with the same mutation can have effects that vary greatly in severity. All of this can make it difficult for doctors to decide on the best way to treat people with this condition. That is why you need to get an expert medical opinion from a clinic that specialises in inherited cardiac conditions.

How is LQTS treated?
The level of risk of sudden death helps decide on the need for treatment. Those who are statistically at greatest risk of sudden death are people with one or more of the following features:

- a previous cardiac arrest
- blackouts
- a very long QT interval on the ECG
- sodium channel mutations
- young adult women

Children who are most at risk tend to be young boys before puberty, and girls who are passing into puberty.

Drugs
The first line of treatment is with drugs. The most commonly used drugs are beta-blockers. These block the effects of adrenaline and associated natural chemicals in the body that make the heart pump harder and faster. They therefore also block the effects of exercise on the heart. They are effective in the most common forms of LQTS, as they reduce symptoms and the risk of sudden death. However, they are less effective in people with the sodium channel form of LQTS.

There are other more recent trends in drug treatment that look promising, but their long-term benefits are unknown. These involve using different anti-arrhythmic drugs. These drugs block disturbances in the heart rhythm that can cause sudden death. Potassium supplement pills have also been tried with occasional success.

Pacemaker or ICD
If you are at high risk (for example if you have already had a cardiac arrest), or if drugs have failed to control your symptoms, your doctor may advise you to have a pacemaker or an implantable cardioverter defibrillator (ICD) fitted, as well as taking your medication. A pacemaker and an ICD both consist of an electronic box containing a battery, that is inserted under the skin and attached to the heart by special electrical ‘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 3 to 12 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 the BHF booklet Pacemakers.

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 may have to be 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 the BHF booklet Implantable cardioverter defibrillators (ICDs).

Surgery
Another option is to perform surgery to disrupt the nerves that release adrenaline and related chemicals into the heart. This is known as ‘cervical sympathectomy’. It involves operating on the left side of the neck and removing the nerves.

Advice on physical activity for people with LQTS
If you have LQTS, your doctor will advise you to avoid excessive exercise or strenuous athletic activities. See General lifestyle advice, on page 37.

**Brugada Syndrome**
This condition was first identified in the early 1990s. It is not a common condition in the western world, but seems to be much more common among young men in South East Asia. In the western world it affects mainly young and middle-aged adult men.

Brugada Syndrome has been associated with mutations in the sodium channel, but this appears to account for only 1 in every 10 people with the condition. The sodium channel behaves abnormally in that movement of sodium ions into the cells is restricted (See the Brugada Syndrome diagram below). This results in particular changes on the ECG, but there are no abnormalities in the structure of the heart.

These diagrams below show the flow of potassium and sodium ions in and out of the heart’s cells. The thin arrow represents a reduced flow.

**Normal Heart**
In a normal heart, potassium flows out of the cell to ‘repolarise’ the heart, and sodium flows into the cells to activate the heart.

**Brugada Syndrome or PCCD**
In people with Brugada Syndrome or PCCD, the flow of sodium into the heart cells is reduced.

What are the symptoms of Brugada Syndrome?
Some people with Brugada Syndrome may have no symptoms at all. In others, the most common symptoms are blackouts caused by a ventricular arrhythmia (a disturbance in the heart’s rhythm). Some people...
may notice palpitations due to ectopic beats. Sudden death may occur. If it does, it usually happens while the person is sleeping.

Are there any physical signs of Brugada Syndrome?
There are no associated physical signs.

How is Brugada Syndrome diagnosed?
Diagnosis involves having an ECG (see page 29). The changes characteristic of Brugada Syndrome may appear on the ECG continuously or intermittently, or they may not show at all. Having a high temperature can sometimes bring out the ECG changes.

If the changes don’t show up on the ECG, there are certain tests that can make the ECG changes visible. These are called ‘provocation tests’. A provocation test involves having an injection of a small amount of an anti-arrhythmic drug while you are having an ECG. The drugs most commonly used for this are ajmaline and flecainide. However, there is some controversy about how much reassurance a negative ECG result from a provocation test should give. Researchers have found that, in some carriers who have already been identified by genetic testing, changes on the ECG are not seen even with a provocation test. However, in these people the level of risk does appear to be low.

Genetic testing is not very useful for diagnosing Brugada Syndrome, because mutations have been found in only a small proportion of people known to have the syndrome.

How is Brugada Syndrome treated?
Without appropriate treatment, the outlook for people with Brugada Syndrome can be poor, especially in people who get symptoms or who have already had a cardiac arrest. The highest rates of sudden cardiac death are found among young male adults. It is therefore standard practice for high-risk carriers to have an ICD fitted, as this is a very successful form of protection. For more information on ICDs, see page 18. So far, medication has not been shown to help protect people with Brugada Syndrome.

If you have an abnormal ECG but don’t have any symptoms, it can be very difficult to decide what treatment to give you. An EPS (an electrophysiological study) may help your doctors decide whether you need an ICD or not. Research suggests that people who have a normal ECG and no symptoms should be safe without having any treatment. It is unusual for children to be at high risk.

If you are a carrier and you get a fever or high temperature, you should take paracetamol or ibuprofen to help lower it. This is because having a fever seems to increase the risk of having the changes characteristic of Brugada Syndrome.

CPVT (catecholaminergic polymorphic ventricular tachycardia)
CPVT is a rare condition found in young people and children. It causes a particular type of arrhythmia (a disturbance in the heart’s rhythm). It has been associated with two genes that make proteins found inside the cell – the human ryanodine receptor (a calcium ion channel) and calsequestrin (a protein that interacts with the channel). These regulate the release of calcium ions into the rest of the cell. If these do not function normally, the level of calcium inside the cell becomes too high, resulting in the arrhythmias characteristic of CPVT.

What are the symptoms of CPVT?
Some people with CPVT have no symptoms at all. Others may have blackouts. Sudden death may occur while the person is exerting themselves or suffering emotional stress.

The condition can affect children and seems to cause more blackouts in males than in females.

Are there any physical signs of CVPT?
There are no physical signs.
Conditions that cause SADS

How is CVPT diagnosed?
The diagnosis is usually made after the chance recording of arrhythmias that are characteristic of CPVT, while the person is doing exercise. Genetic testing is also useful in cases where a member of the same family has already been found to carry a mutation and is showing the signs of the condition.

How is CVPT treated?
Your doctor will advise you to take beta-blockers (a type of drug) and to restrict the amount of exercise you do. This combination greatly improves the outlook for people with CPVT. About 1 in every 3 people with the condition will also need to have an ICD fitted (For more on ICDs, see page 18).

PCCD (progressive cardiac conduction defect)
PCCD is a rare condition. In people with PCCD, the heart’s electrical impulses are conducted very slowly and this results in the gradual development over time of ‘heart block’. (Heart block is a failure of the heart’s electrical impulses to conduct properly from the top chambers – the atria – to the bottom chambers – the ventricles. The severity of the condition and its associated risk can vary). PCCD can cause arrhythmias – either because the heart’s rhythm is too sluggish (bradycardia and asystole), or because rapid rhythm disturbances (tachycardia) arise from parts of the heart that have escaped normal regulation. In some people, PCCD has been associated with sodium channel mutations that cause changes in channel behaviour similar to those found in people with Brugada Syndrome (see the diagram on page 19).

What are the symptoms of PCCD?
Dizziness and blackouts are the usual symptoms. Sudden death may also occur.

Are there any physical signs of PCCD?
There are no physical signs.

How is PCCD diagnosed?
The ECG abnormalities characteristic of PCCD may be detected either on a standard ECG or with 2-hour ECG monitoring. An electrophysiological study (EPS) may also help the doctor make a diagnosis (We describe all these tests on pages 29–34). If a sodium channel mutation is identified in affected members of a family, it may also be found in other close blood relatives.

How is PCCD treated?
If you have PCCD, you will need to have a pacemaker fitted in order to stop dangerous bradycardia (slow heart rates) from occurring. This may not prevent ‘escape tachycardias’ (a type of fast heart rhythm) so you may also need to take anti-arrhythmic drugs. Some people may need to have an ICD fitted instead of a pacemaker (For more information on pacemakers and ICDs, see page 18). Medication alone does not help.

IVF (idiopathic ventricular fibrillation)
This term describes the group of conditions responsible for life-threatening, fast heart rhythm disturbances in people who do not have any signs of heart disease. Brugada Syndrome and CPVT form part of this group, but there have been reports of people with IVF who do not have the ECG changes characteristic of the Brugada Syndrome but who do have sodium channel mutations. Treatment includes having an ICD fitted (see page 18), and can be successful in protecting the person.

Sodium channel disease
There are very rare and specific sodium channel mutations that can cause Long QT Syndrome, Brugada Syndrome and/or PCCD in the same family. They can be diagnosed and treated as described on pages 14–23 and can be identified by genetic testing.

Structural heart disease
In some cases, the pathologist examining the person who has died cannot confirm that the person had structural heart disease – either because
Conditions that cause SADS

there is no evidence of it, or because there is not enough evidence and the heart is felt to be relatively normal. So the cause of death will be recorded as SADS. This may happen even in cases where evidence of inherited structural heart disease is subsequently detected in other members of the victim’s family. The presence of very subtle structural heart disease in the victim may, however, have been enough to cause sudden cardiac death.

In these circumstances the most common causes of death are:

- arrhythmogenic right ventricular cardiomyopathy (ARVC)
- dilated cardiomyopathy (DCM)
- hypertrophic cardiomyopathy (HCM)
- mitral valve prolapse (MVP – see below)
- Wolff-Parkinson-White Syndrome (WPW)

For more information on all types of cardiomyopathy, contact the Cardiomyopathy Association (For contact details, see page 57).

Mitral valve prolapse

Mitral valve prolapse is when the mitral valve is ‘floppy’ in appearance. This will show up on an echocardiogram (see page 32). Mitral valve prolapse is very common and affects about 1 or 2 in every 20 people. In most cases, people with this condition do not have any symptoms, and the condition is not harmful. In some rare cases, mitral valve prolapse can be inherited in a family and can then be associated with arrhythmias and sudden death. For more information on mitral valve prolapse, see the BHF booklet Valvular heart disease.
Implications of a SADS death for close blood relatives of the person who has died

If you are a close blood relative of a person who has died of SADS, it is important that you have an assessment (tests) at a specialist clinic for inherited cardiac conditions to find out if you have inherited the same medical condition as the SADS victim. If there have been any other sudden or suspicious deaths in your family, including cot death, this further suggests that there may be an underlying inherited condition. On pages 29–34 we describe all the various tests that you may need to have as part of the assessment.

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 cardiac conditions is.

What if nothing is found in your family?

In families where someone has died of SADS and the remaining family members are tested, about 1 in every 2 families show no sign of inherited heart disease. This can be due to two reasons:

The SADS victim may not have inherited any abnormality from his or her parents. Either the victim was the first to suffer a mutation in the family (and therefore the victim’s children are the only relatives who are at risk), or there was another cause which has not been identified and which is not inherited.

or

Some family members may be carriers but show no signs of any disease. It is impossible to give 100% reassurance that a relative is not a carrier, except in cases where a mutation has been identified in the person who has died and the victim’s relative has had genetic testing which shows that he or she doesn’t have the same mutation. However, people who do not have any symptoms or signs are at low risk of sudden death, so in these cases the doctor can give some reassurance. The people at highest risk
Medical history
It is vital that a clear medical history of the victim and his or her death is established, using the family's and friends' recollections as well as the reports of the coroner, pathologist, GP and police. For example, fits brought on by exercise can be due to an underlying channelopathy such as LQTS or CPVT, or a sudden cardiac death during sleep may have been caused by sodium channel LQTS or Brugada Syndrome. It is also important to find out about any medications and any potentially dangerous drugs that the person may have taken before they died. Your doctor may ask you if you have ever had symptoms such as blackouts or palpitations, as these may suggest underlying heart disease. The doctor may also ask about any other sudden or suspicious deaths in your family, including cot deaths, that may suggest an underlying inheritable condition.

Medical examination and tests
A medical examination may help to discover if there is an inheritable structural heart disease in your family. For example, if there is mitral valve prolapse with leakage from the valve, this will cause a 'murmur' that a doctor can hear through a stethoscope. Your doctor may also suggest that you have some of the tests we describe below.

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

ECG
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. If the first ECG does not show any sign of a channelopathy, the test can be repeated later.

Signal-averaged ECG
This is an ECG that adds together the electrical readings from at least 250 heartbeats so that any very subtle variations can be seen – for example, if the electrical impulses in the heart are being conducted more slowly. It is useful for diagnosing Brugada Syndrome, PCCD or ARVC.

Assessment at a clinic for inherited cardiac conditions
Below we describe what happens when the relative of a person who has died of SADS has an assessment at a clinic for inherited cardiac conditions to find out if they may have the same condition as the person who has died (see page 27).
Exercise test
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. This test is particularly useful in detecting some of the features that are characteristic of LQTS or CPVT.

Cardiopulmonary exercise test
Some hospitals may also ask you to do a cardiopulmonary exercise test. This test analyses the efficiency of the heart muscle by measuring the amounts 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 (inefficient pumping action of the heart).

24-hour ECG monitoring
Also called Holter monitoring
This test involves using a recording device that comes in two different forms: either a small portable tape recorder (like a walkman), or a small digital device the shape of a pager. You wear the device on a belt around 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 if a digital one is used. The doctor can then analyse the electrical activity and rhythm of your heart to find out if you have any arrhythmias (for example, the arrhythmias typical of LQTS and CPVT), or some of the other features characteristic of LQTS.

Cardiomoemo and cardiac event recorders
These are more sophisticated versions of the 24-hour ECG monitoring device described above. Whenever you have an attack of symptoms, you can activate the device to record your heart's rhythm. The advantage of the cardiomoemo is that it doesn't have any leads, so you can just place it on your chest when you get symptoms, without having to put any leads in position.

Implantable loop recorder
Also called an ILR
When it is difficult to assess or record a symptom because it only happens infrequently – as with blackouts – an implantable loop recorder (or ILR for short) can be used. The device, which is the size of a packet of chewing gum, is placed under the skin at the left shoulder. You will need to go into hospital as a day case to have this done. A small cut about 2cm long (just under one inch) is made and the device is inserted. The device monitors the heart’s rhythm and can record any abnormal events that it is programmed to detect. If anything happens, you just place a special small box on the surface of your skin over the ILR. You can then activate the device by pressing a button on the box, which makes the ILR record the preceding 1 minutes of the heart’s activity. The device can then be ‘interrogated’ by a computer at the hospital and the doctor can examine the recording. The device has a battery that can last up to two years if necessary.

Provocation test
Also called ajmaline, flecainide or adenosine tests
You may be asked to have this test if your doctor suspects Brugada Syndrome. While you are having an ECG test, you will be given an injection of ajmaline or flecainide (anti-arrhythmic drugs). The test may show changes on the ECG that are typical of one of the channelopathies. A fine plastic tube is inserted into a vein at the front of your elbow. The drug is injected through the tube for about  to 10 minutes and you will be monitored for 20 minutes or a few hours afterwards, depending on the drug used. There is, however, a risk as 1 in every 200 Brugada Syndrome carriers or their immediate blood relatives have a potentially life-threatening arrhythmia during the injection. The test is therefore always performed with appropriate facilities to protect patients from this risk. Ajmaline is preferable as it lasts for a shorter period of time in the circulation. Adenosine (another short-acting chemical) is used if Wolff-Parkinson-White Syndrome (WPW) is considered a possible diagnosis.
**Echocardiogram**

Also called an **echo**

This test uses ultrasound waves to look at the structure of the heart. It is useful for people whose ECG shows changes that could be caused either by a channelopathy or by uninherited heart disease that has damaged the heart – for example, a previous heart attack that you may not have even been aware of. An echocardiogram can also detect inheritable conditions such as cardiomyopathy and mitral valve prolapse.

**Cardiac magnetic resonance scan**

Also called an **MRI scan**

This is a special kind of scan used to examine the structure of the heart and the nature of its muscle. It uses a magnetic resonance scanner that creates intense fluctuating magnetic fields around your body while you are inside the scanner. This generates the signals that make up the pictures produced. It may be useful for detecting the presence of fat and scarring in the heart muscle that is associated with cardiomyopathies.

**Coronary angiogram and electrophysiological study (EPS)**

Depending on the results of the tests described above, 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 that allows the body, and any medical items such as cardiac catheter tubes or pacing wires, to be seen 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 furring or blockages – coronary heart disease. (The ECG changes that are characteristic of Brugada Syndrome or LQTS can sometimes be caused by coronary heart disease).

**Tilt-table test**

If you get episodes of fainting or blackouts, your doctor may recommend that you have a tilt-table test to find out more about what is causing you to faint. The fainting can be caused by common conditions – such as vasovagal syndrome (see page 56) – which tend to particularly affect young women and girls but have a very low risk of causing sudden death. Or it may be caused by a more rare but potentially life-threatening channelopathy. It is important to find out what is causing the fainting or blackouts so that your doctor can give the appropriate treatment.

A fine plastic tube is inserted into a vein at the front of your elbow as a precaution in case the doctor needs to give you any drugs during the test. While you lie flat on a special table, your blood pressure, pulse and ECG are monitored. The head of the table is then raised to an angle of 0 to 7 degrees and monitoring is continued. If nothing happens, a spray of a substance called GTN is given under your tongue as a stimulus and you will be monitored for another 10 to 15 minutes. The table will then be returned to the flat position and the leads disconnected. The whole test takes around 45 minutes. If your blood pressure falls at the same time as you suffer your usual symptoms, this means that you have vasovagal syndrome or a related condition.

**Genetic testing**

In most of the inherited conditions known to cause SADS, mutations of specific genes have been detected and are thought to cause a specific disease. So in principle, if we could identify these mutations, we would be able to make a diagnosis from any DNA sample including any obtained from SADS victims at their autopsy, or from their relatives who have given blood. Unfortunately this can only be done in some families at the
moment because we don’t have complete knowledge of all the genes and mutations involved in any condition. For example, only 7 in every 10 people known to have LQTS have mutations of known identified genes. Also, many variations in the DNA code are found in a large number of people and do not necessarily cause any disease. Many families with LQTS have mutations which are specific to them (‘private’ mutations), which can also make it difficult to decide whether it is the mutation that is causing the disease or not. Although our understanding is still limited, in some cases a genetic test can suggest what treatments might be best and how risky the condition is. As research progresses, more genes will be identified and there will be better tools to decide whether a mutation causes a disease and how best to treat it.
**General lifestyle advice**

**Exercise**
The majority of conditions that can cause sudden cardiac death appear to be worsened by exercise. So doctors usually advise people with these conditions to avoid competitive sports and unrestricted severe exertion. This can be especially difficult for younger people who may be unwilling to stop sport. It is important to get a balance between the benefit of restricting exercise and the negative impact the restrictions may have on the person. Hopefully the person can come to terms with the changes he or she needs to make.

This advice is complicated by the fact that SADS deaths often occur at night and during sleep – as with the Brugada Syndrome and sodium channel LQTS. If you have one of these conditions, your doctor can advise your partner what to do if anything happens, and may encourage you to buy a home ‘defibrillator’. (If someone has a cardiac arrest, this machine may be able to return the heart to a normal rhythm by delivering an electrical ‘shock’ through the chest wall).

**Vomiting and diarrhoea**
In people with Long QT Syndrome or Brugada Syndrome, a drop in the levels of potassium in the blood can cause a serious deterioration in the person’s condition. Any prolonged vomiting or diarrhoea (lasting more than a day) can cause a significant loss of potassium in the blood – a condition called hypokalaemia. If this happens, it is important that you get rehydrated, using a salt and sugar preparation such as Dioralyte. (You can buy this at your local pharmacy, or you may be able to get a prescription for it from your doctor). If vomiting prevents you taking this, you should go to hospital to receive appropriate fluids through a drip. It is best to get your GP to arrange this, but if this is not possible you should go to the Accident and Emergency department of your local hospital.
Drugs to Avoid

Anyone with a condition affecting the heart that can cause sudden cardiac death needs to take extra care with medicines. All medicines, both
• those prescribed by your doctor and
• any you buy over the counter
must be checked by your GP or cardiologist as some can increase the risk of sudden death.

If you have Long QT Syndrome
For people with Long QT Syndrome there are specific medications that can have a serious effect by further prolonging the QT interval. We give a list of these medicines on page 39. This list includes drugs that can stimulate and irritate the heart by causing adrenaline-like effects. You must always check with your GP or cardiologist before taking any new medication, as this list will change with time.

If you have Brugada Syndrome
In people with Brugada Syndrome, the number and range of drugs that may make the condition worse are unknown and caution must be used. For example, certain anti-arrhythmics, all beta-blockers and some anti-depressants are known to interact badly with it. On page 45 we give a list of the medicines that people with Brugada Syndrome should avoid.

Drugs which people with Long QT Syndrome should avoid
Below is a list of the drugs that people with Long QT Syndrome should avoid. This list may not be complete so, if your doctor prescribes any new drugs for you, check with your GP or pharmacist that the drug is suitable for people with Long QT Syndrome.

*Drugs which have been unlicensed, withdrawn or suspended in the UK market.

<table>
<thead>
<tr>
<th>Drug group</th>
<th>Avoid completely</th>
<th>Close monitoring and professional supervision required if drug is absolutely necessary</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anti-anginals and vasodilators</td>
<td>bepridil*</td>
<td>vardenafil</td>
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<tr>
<td></td>
<td>lidoflazine*</td>
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<td></td>
<td>prenylamine*</td>
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<tr>
<td></td>
<td>terodiline*</td>
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<tr>
<td>Anti-arrhythmics Class I</td>
<td>ajmaline*</td>
<td>cibenzoline*</td>
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<tr>
<td></td>
<td>dihydroquinidine*</td>
<td>flecaïnide</td>
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<td></td>
<td>disopyramide</td>
<td>mexiletine</td>
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<td></td>
<td>procaïnamide</td>
<td>pirmenol*</td>
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<tr>
<td></td>
<td>quinidine*</td>
<td>propafenone</td>
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<tr>
<td>Drug group</td>
<td>Avoid completely</td>
<td>Close monitoring and professional supervision required if drug is absolutely necessary</td>
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<tr>
<td><strong>Anti-arrhythmics</strong> Class III</td>
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<td>Almokalant*</td>
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<td>Amiodarone</td>
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<td>Azimilide*</td>
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<td>Dofetilide*</td>
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<td>Dronedarone*</td>
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<td>D-sotalol*</td>
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<td>Ersentilide*</td>
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<td>Ibutilide*</td>
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<td>Nifekalant*</td>
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<td>Sematilide*</td>
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<td>Sotalol</td>
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<tr>
<td>Terikalant*</td>
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<tr>
<td><strong>Anti-cancer</strong></td>
<td>Arsenic trioxide</td>
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<td><strong>Anti-histamines</strong></td>
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<td>Terfenadine*</td>
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<td>Ebastine*</td>
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<td>Indapamide</td>
<td>Isradipine</td>
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<td>Moexipril/ Hydrochlorothiazide</td>
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<td>Nicardipine</td>
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<td><strong>Anti-microbials</strong></td>
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<td><strong>Macrolides</strong></td>
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<td>Azithromycin</td>
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<td>Erythromycin</td>
<td>Roxithromycin</td>
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<td>Levofloxacin</td>
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<td>Ofloxacin</td>
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<tr>
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<td>Halofantrine*</td>
<td>Itraconazole</td>
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<td>Pentamidine*</td>
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<td>Voriconazole</td>
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<td>Quinine</td>
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<tr>
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<td></td>
<td>Cotrimoxazole</td>
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<td>Foscarnet</td>
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<td></td>
<td>Trimethoprim sulfa</td>
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<td>Drug group</td>
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<td><strong>Psychiatric</strong></td>
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<td>Serotonin reuptake inhibitors</td>
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<td></td>
<td>venlafaxine</td>
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<td>zimeldine*</td>
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<td>clomipramine</td>
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<td>quetiapine</td>
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<td>risperidone</td>
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<td>ziprasidone</td>
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<td>Serotonin agonists and antagonists</td>
<td>cisapride*</td>
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<td>dolasetron</td>
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<td>levomethadyl*</td>
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<td>methadone</td>
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<td>probucol</td>
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<td>alfuzosin</td>
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<td>amantadine</td>
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<td>chloral hydrate</td>
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<td>clobutinol*</td>
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<tr>
<td></td>
<td>felbamate*</td>
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<td>fosphenytoin</td>
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<td></td>
<td>galantamine</td>
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<td></td>
<td>organophosphates*</td>
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<td></td>
<td>perflutren lipid</td>
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<td>microspheres</td>
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<td>(echocardiographic contrast)</td>
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<td>solifenacin</td>
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<td>tizanidine</td>
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<td>vasopressin</td>
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</tbody>
</table>
Stimulant drugs
Some cold remedies contain some of the following drugs, which should also be avoided, so it is important always to check the label:

- adrenaline (epinephrine)
- cocaine
- dobutamine
- dopamine
- ephedrine
- fenfluramine
- isoprenaline (isoproterenol)
- metaproterenol
- midodrine
- norepinephrine (noradrenaline)
- phentermine
- phenylpropanolamine
- pseudoephedrine
- ritodrine
- salbutamol (albuterol)
- salmeterol
- sibutramine
- terbutaline


Drugs which people with Brugada Syndrome should avoid
Always check new medication before it is taken as this list cannot be guaranteed to be complete.

<table>
<thead>
<tr>
<th>Drug group</th>
<th>Drugs to avoid</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Alpha adrenergic agonists</strong></td>
<td>methoxamine, noradrenaline</td>
</tr>
<tr>
<td><strong>Anti-arrhythmics:</strong></td>
<td>All to be avoided including:</td>
</tr>
<tr>
<td>Class I</td>
<td>ajmaline, cibenzoline, disopyramide, flecainide, pilscainide, procainamide, propafenone</td>
</tr>
<tr>
<td><strong>Anti-depressants</strong></td>
<td>All to be avoided, including:</td>
</tr>
<tr>
<td>Tetracyclic anti-depressants</td>
<td>amitriptyline, clomipramine, desipramine, nortriptyline</td>
</tr>
<tr>
<td>Tricyclic anti-depressants</td>
<td>All to be avoided, especially propranolol</td>
</tr>
<tr>
<td><strong>Beta-blockers</strong></td>
<td>diltiazem, nifedipine, verapamil</td>
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<tr>
<td><strong>Calcium channel blockers</strong></td>
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### Drug group

**Drugs to avoid**

<table>
<thead>
<tr>
<th>Drug group</th>
<th>Drugs to avoid</th>
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<tbody>
<tr>
<td>Ergot alkaloids</td>
<td>ergonovine</td>
</tr>
<tr>
<td>First-generation anti-histamines</td>
<td>dimenhydrinate</td>
</tr>
<tr>
<td>Local anaesthetics</td>
<td>bupivacaine</td>
</tr>
<tr>
<td>Nitrates</td>
<td>isosorbide dinitrate</td>
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<td></td>
<td>nitroglycerine</td>
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<tr>
<td>Opioid analgesics</td>
<td>propoxyphene</td>
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<tr>
<td>Parasympathetic agonists</td>
<td>acetylcholine</td>
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<tr>
<td>Phenothiazine</td>
<td>cyamemazine</td>
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<td></td>
<td>perphenazine</td>
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<tr>
<td>Potassium channel activators</td>
<td>pinacidil</td>
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<td>Selective serotonin reuptake inhibitors</td>
<td>fluoxetine</td>
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<tr>
<td>Other drugs to avoid</td>
<td>alcohol intoxication</td>
</tr>
<tr>
<td></td>
<td>cocaine</td>
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<td></td>
<td>lithium</td>
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<td>propofol</td>
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</table>

Looking forward

The future

Research into channelopathies is progressing rapidly and in the future it is expected that all the genes involved will be discovered.

In the future, it may also be possible to diagnose all carriers easily – even in those people who have a normal ECG reading. It may also be possible to choose the best treatment based on the type of mutations involved, and the treatment may even be designed based on this knowledge.

In the meantime, better understanding of these conditions and improvements in methods for diagnosis should still result in better management.

However it is crucial that, when an unexplained and unexpected sudden death occurs, all immediate blood relatives should have an assessment by a specialist cardiologist to find out if they have an inherited heart disease such as a channelopathy.
Technical terms

Anti-arrhythmic drugs
A group of medicines used to regulate and control the heart’s rhythm.

Aorta
The major blood vessel that leaves the left side of the heart. It supplies blood to the body.

Aortic valve
The valve through which blood passes from the left ventricle into the aorta.

Arrhythmia
A disturbance of the heart’s rhythm. A ‘ventricular’ arrhythmia can be life-threatening.

Asystole
When the heart’s rhythm stops completely because there is no electrical activity.

Atrium
One of the two top chambers of the heart. The plural of ‘atrium’ is ‘atria’.

Autopsy
A post mortem examination of a dead body.

AV block
See ‘heart block’.

AV node
Atrio-ventricular node. The part of the heart that lies between the top chambers (‘atria’) and bottom chambers (‘ventricles’). It regulates the transmission of electrical impulses from the heart’s natural pacemaker in the atrium to the ventricle. If the impulses from the atrium become too fast, the AV node helps to prevent the heart from pumping too fast.
**Technical terms**

**B**

**Bradycardia**
A slow heart rate.

**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 the problem is not resolved within two minutes there will be permanent brain damage, and if left untreated the person will quickly die. This is the mechanism by which the channelopathies can cause sudden death.

**Cardiologist**
A doctor who specialises in the heart.

**Cardiomyopathy**
Disease of the heart muscle, which is usually inheritable.

**Cardiopulmonary exercise test**
An exercise test that monitors the consumption of oxygen, using a set of breathing tubes.

**Catheter ablation**
See ‘RF ablation’.

**Cervical sympathectomy**
A form of surgery that is useful for some people with LQTS. It reduces the amount of adrenaline and related chemicals delivered to the heart by certain nerves (the left cervical ganglia).

**Coroner**
The government-appointed legal person responsible for ensuring that no foul play has occurred when an unexpected death happens.

**D**

**Defibrillator**
A device used if a person has a cardiac arrest. It may be able to return the heart to a normal rhythm by delivering an electrical ‘shock’ through the chest wall.

**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.

**E**

**Ectopic beat**
An ‘extra’ beat which occurs when the heart activates prematurely, disrupting its normal rhythm. The heart’s natural pacemaker resumes its normal control after a brief pause. Most of the time the person does not notice these extra beats but, if they do become aware of them, the sensation depends on how close the ectopic beat occurs to the preceding normal beat. If it is close, only the pause might be noticed. If it occurs further away, it might be felt as an extra beat from the heart, making the rhythm feel irregular or erratic.

**ERSCD**
Exercise-related sudden cardiac death. See ‘sudden cardiac death’.

**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.
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. A 'sporadic' mutation is not inherited from a parent's DNA but occurs due to damage to the DNA after the egg or sperm that forms a human embryo is made.

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

Pathologist
A doctor trained to examine the body, and samples of its organs, after a person has died, in order to diagnose any abnormalities.

Post mortem
The examination of a dead body by a pathologist.

Prognosis
A patient’s outlook. In this context it means the likelihood of any life-threatening events.

QT interval
A lengthening of the time period of a particular part of the heartbeat cycle, as seen on an ECG.

RF ablation
The use of high-frequency radio waves to destroy small areas of heart tissue such as the extra or 'accessory' pathways seen in Wolff-Parkinson-White Syndrome. Also called 'catheter ablation'.
Technical terms

S

SCD
See ‘sudden cardiac death’ below.

Sino-atrial node
The heart’s natural pacemaker.

Sudden cardiac death
A death is described as sudden when it occurs unexpectedly, spontaneously and/or even dramatically. If the death is due to heart disease it is called sudden cardiac death (or SCD). Some will be unwitnessed or occur during sleep, while others occur during or immediately after exercise (exercise-related sudden cardiac death or ERSCD).

Syndrome
A collection of medical features of an illness that make it a distinctive condition.

T

Tachycardia
A fast heart rate.

V

Vasovagal syndrome
A disorder of the nerves supplying the blood vessels and heart that can result in dizzy episodes or blackouts. This is due to sudden drops in blood pressure because of rapid opening up (dilatation) of the arteries with or without sudden slowing of the heart rate. It is usually harmless, although blackouts may place the person in dangerous situations. Treatment can involve tablets and/or a pacemaker.

Ventricles
The two bottom chambers of the heart.

Ventricular
From, or belonging to, the ventricle.

For more information

For information on your nearest clinic for inherited cardiac 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 assessment. The Service is staffed by specialist cardiac nurses and a bereavement counsellor.

BHF publications
Implantable cardioverter defibrillators (ICDs)
Pacemakers

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 support on coping as a family with SADS
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 363444
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.

For more information on cardiomyopathy
The Cardiomyopathy Association
Unit 10, Chiltern Court
Asheridge Road, Chesham
Bucks HP 2PX
Phone: 0800 018 1024
Fax: 01494 797199
info@cardiomyopathy.org
www.cardiomyopathy.org

The Cardiomyopathy Association provides support and information on the different types of cardiomyopathy.
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About the British Heart Foundation

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

What you can do for us
We rely on donations to continue our vital work. If you would like to make a donation to the BHF, please ring our Supporter Services team on 0844 847 2787 or contact us through our website at bhf.org.uk/donate or send it to us at the address on the back cover.

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