

When a young person dies suddenly



Information for the family and relatives of
a young person who has died of Sudden
Arrhythmic Death Syndrome (SADS)



by Dr Elijah R Behr MD

Produced by Cardiac Risk in the Young



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Introduction

You may be reading this booklet because a young 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 so young and 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.

This booklet outlines the possible causes of sudden death in young people and children. It concentrates on the medical conditions responsible for a sudden unexpected death where a definite cause cannot be found, even after a post-mortem. This is called Sudden Arrhythmic Death Syndrome, or SADS.

The booklet:

- describes what happens after someone has died suddenly and unexpectedly
- explains how the heart works and how it can cause sudden death
- explains what causes SADS and why it is important that the close blood relatives of the person who has died should have a medical examination and tests to find out if they have inherited the same condition
- describes the tests your doctor may ask you to have
- 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 have tried to explain medical and technical terms as we go along but, if you find a word you do not understand, you can look it up in the list of technical terms on page 28.

We hope that this booklet will help you and your family understand what has happened, and hopefully help you come to terms with the event. If you need further help or information see page 27.

What is the difference between Sudden Arrhythmic Death Syndrome (SADS) and Sudden Cardiac Death (SCD)?

Sudden Cardiac Death (SCD)

Sudden cardiac death is a dramatic and/or spontaneous 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 heart has been examined by an expert cardiac pathologist. This is then called Sudden Arrhythmic Death Syndrome. (In the past it has also been 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.

What happens after an unexpected sudden death in a young person?

After an unexpected sudden death it is usual that the coroner of the area where the death has happened will ask for a post-mortem to be performed. 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 artery 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 (one who specialises in the heart) to determine the cause of death. The pathologist or coroner may suggest that small samples of heart tissue, spleen and/or blood are kept, sometimes in frozen form, for further testing in case a genetic or inherited cause of sudden death is suspected. This is known as 'molecular autopsy' and will only take place with agreement from the family.

How the heart works, and how it can cause sudden death

In order to understand why sudden death can happen, it helps to understand how the heart works.

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, which is in the top chamber (see the diagram opposite). The SA node sends out regular electrical impulses causing the atrium to contract and to 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. Chemicals which circulate in the blood, and which are released by the nerves that regulate the heart, alter the speed of the 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.

The electrical activity of the heart can be detected by doing an 'electrocardiogram' (also called an ECG). An ECG recording looks something like the ones shown on page 11 (figure 3A-C).

A death is described as sudden when it occurs unexpectedly, spontaneously and/or even dramatically. Some will be unwitnessed; some may occur during sleep or during or just after exercise. Most sudden deaths are due to a heart condition and are then called sudden cardiac death (SCD). Up to 95 in every 100 sudden cardiac deaths are due to disease that causes abnormality of the structure of the heart. The actual mechanism of death is most commonly a serious disturbance of the heart's rhythm known as a 'ventricular arrhythmia' (a disturbance in the heart rhythm in the ventricles) or 'ventricular tachycardia' (a rapid heart rate in the ventricles). This can disrupt the ability of the ventricles to pump blood effectively to the body and can cause a loss of all blood pressure. This is known as a cardiac arrest. If this problem is not resolved in about two minutes, and if no-one is available to begin resuscitation, the brain and heart become significantly damaged and death follows quickly.

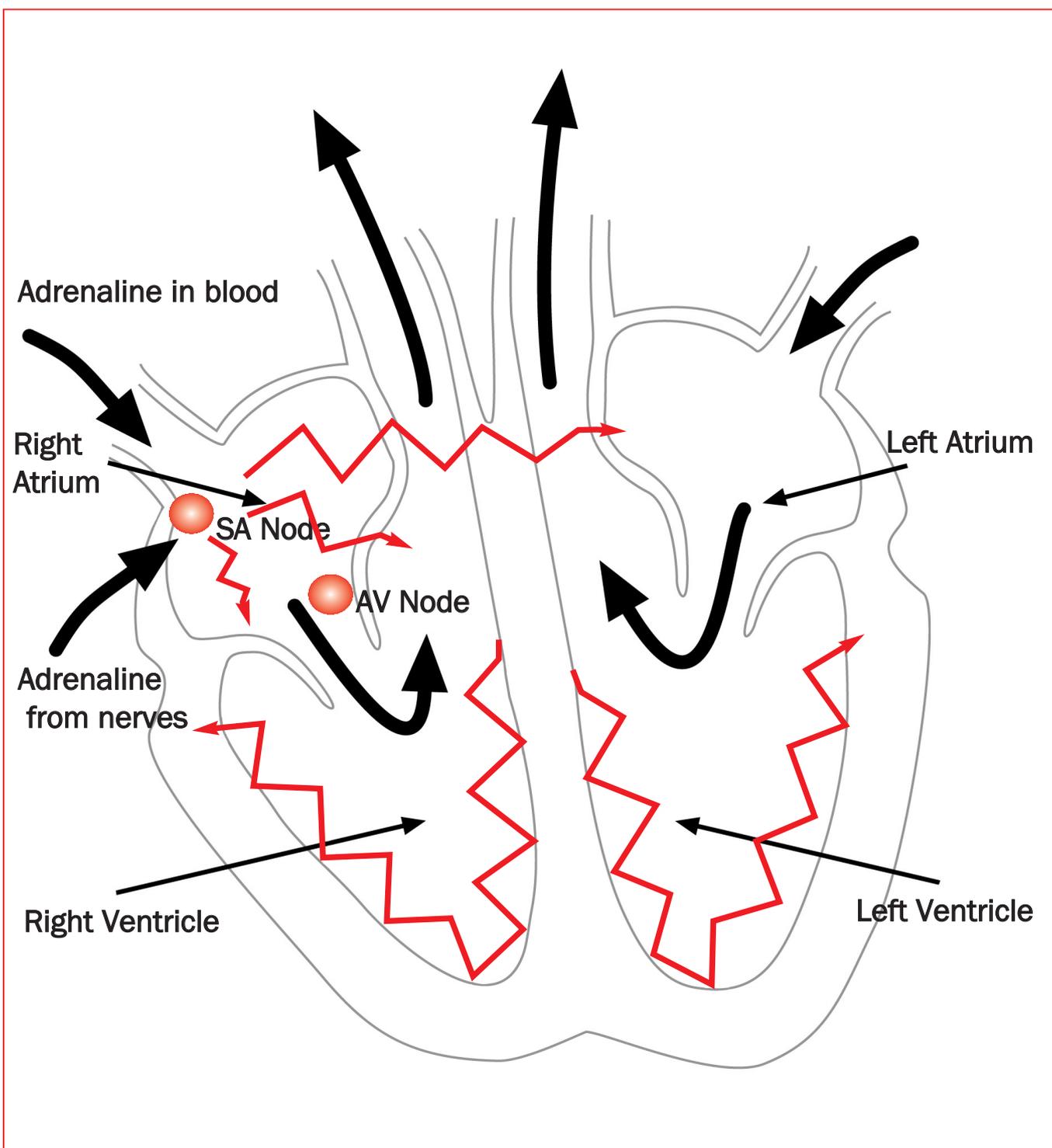


Figure 1: How the heart functions electrically The heart's natural pacemaker - the SA node - sends out regular electrical impulses from the top chamber (the atrium) causing it to contract and pump blood into the bottom chamber (the ventricle). The electrical impulse is then conducted to the ventricles through a form of 'junction box' called the AV node. The impulse spreads into the ventricles, causing the muscle 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.

What causes sudden death in young adults and children?

A sudden death in a young person can be caused by:

- **heart disease**, including cardiomyopathy, congenital heart disease, myocarditis, genetic connective tissue disorders, mitral valve prolapse or conduction disease
- **medication-related causes**
- **other causes**. We explain these below.

Heart disease

Heart disease is the most common cause of an unexpected sudden death in all age groups. In people aged 30 or over, the heart disease is usually due to 'furring' or 'blockages' of the blood vessels that supply the heart, i.e. coronary artery disease. But in younger people and in children the cause is much more often something other than coronary artery disease. The main causes are listed below. Some of these are inherited conditions. Some are detected easily while the person is alive, while others may go unnoticed until a tragic sudden death occurs.

Cardiomyopathies

These are abnormalities of the heart muscle and are usually inheritable.

Hypertrophic cardiomyopathy (HCM) The walls of the heart become abnormally thick without any other cause being identifiable. Even if there is not any thickening, the arrangement of the heart's muscle cells (myocytes) are disorganised and disrupted.

Arrhythmogenic right ventricular cardiomyopathy (ARVC) This condition causes the heart's muscle to become thin because of an abnormal amount of fat and scar tissue in its wall. It affects mainly the right side of the heart but can also affect the left side and frequently causes heart rhythm disturbances.

Dilated cardiomyopathy (DCM) The left and right sides of the heart become enlarged and pump less efficiently, sometimes progressing to heart failure when the heart cannot meet the body's requirements.

Congenital heart disease

This group includes abnormalities of the structure of the heart which have been present since birth. Some of them may be inherited conditions. They include:

Valvular and more complex disease Abnormality of the heart's valves that can be associated with other abnormalities of the heart's structures such as a 'hole in the heart' (for example, Fallot's Tetralogy).

Anomalous coronary arteries

When there is an abnormal arrangement of the arteries that supply blood to the heart muscle.

Myocarditis

Myocarditis is inflammation of the heart's muscle. It is usually due to a viral infection although it can be a complication of other medical conditions or exposure to drugs. It is not inheritable.

Genetic connective tissue disorders

These are inheritable conditions affecting the structures that give support, strength and elasticity to the walls of the major blood vessels and, to a lesser extent, the heart muscle - for example Marfan's syndrome and Ehler-Danlos. These can occasionally cause sudden death by arrhythmias or, more commonly, due to the sudden rupture of a major blood vessel such as the aorta (the major blood vessel that leaves the left side of the heart and supplies blood to the body).

Mitral valve prolapse

The mitral valve can sometimes be 'floppy' in appearance. This will show up on an echocardiogram (see page 18). This is very common and affects around 1 or 2 in every 20 people. It is usually an asymptomatic and benign condition. In some rare cases mitral valve prolapse can be inherited in a family and can then be associated with arrhythmias and sudden death.

Conduction disease

This includes abnormalities in the way that the electrical impulses are conducted through the AV node due to disease (for example as in myotonic dystrophy), or because there are additional or 'accessory' pathways as in Wolff-Parkinson-White (WPW) Syndrome.

Medication-related causes

Prescription, over-the-counter and illegal drugs can have potentially dangerous but usually rare side effects, particularly if too much is taken (an overdose). These effects include arrhythmias (disturbance in the heart's rhythm) and sometimes a sudden death.

Other causes

Research suggests that sudden death may be caused infrequently by conditions such as fits (epilepsy) and severe asthma attacks. Pulmonary embolus (a clot to the lungs) has become better known recently due to its association with staying immobile for long periods during air travel (see page 32). It can cause a sudden collapse and a rapid death.

Sudden Arrhythmic Death Syndrome (SADS)

In around 1 in every 5 cases of sudden cardiac death under 35 - up to 600 every year in the UK - no cause can be found, despite examination of the heart by an expert cardiac pathologist. The cause of death is therefore described as 'unascertainable'. This is called Sudden Arrhythmic Death Syndrome, or SADS.

In the next section we describe some of the conditions responsible for SADS.

What causes SADS?

The conditions responsible for SADS cause a cardiac arrest by bringing on a 'ventricular arrhythmia' (a disturbance in the heart's rhythm), even though the person has no structural heart disease.

There is a group of relatively rare diseases called **ion channelopathies** that affect the electrical functioning of the heart without affecting the heart's structure. This means that they can only be detected in life and not at post-mortem. Ion channelopathies are probably responsible for 4 in every 10 cases of SADS. 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)
- Early repolarisation syndrome
- Mixed sodium channel disease
- Short QT syndrome

We describe each of these on pages 8-15.

Structural heart disease is sometimes found to be a cause of SADS (between 1 and 2 in every 10 cases). For more on this see page 15.

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 cardiac 'ion channels'. 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 do not 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.

Below 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 5,000 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 cell 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 people with potassium channel associated LQTS, the channels do not behave as efficiently as normal. They let potassium ions into the cell too slowly. If the sodium channel is affected, too many sodium ions are allowed into the cell (see figure 2B on page 9). This results in an electrical disturbance in the cells of the heart called 'prolonged repolarisation'. This can be seen on an ECG recording as a lengthening of the time period known as the 'QT interval' (shown in the diagram on page 11 - figure 3). This is where the name long QT syndrome comes from.

Rare forms of LQTS known as Andersen's and Timothy syndromes have been associated with potassium and calcium channel abnormalities respectively.

What are the symptoms?

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 events that put them at risk.

The most common symptom of LQTS is blackouts. Sometimes palpitations due to extra or 'ectopic' heartbeats can be a problem.

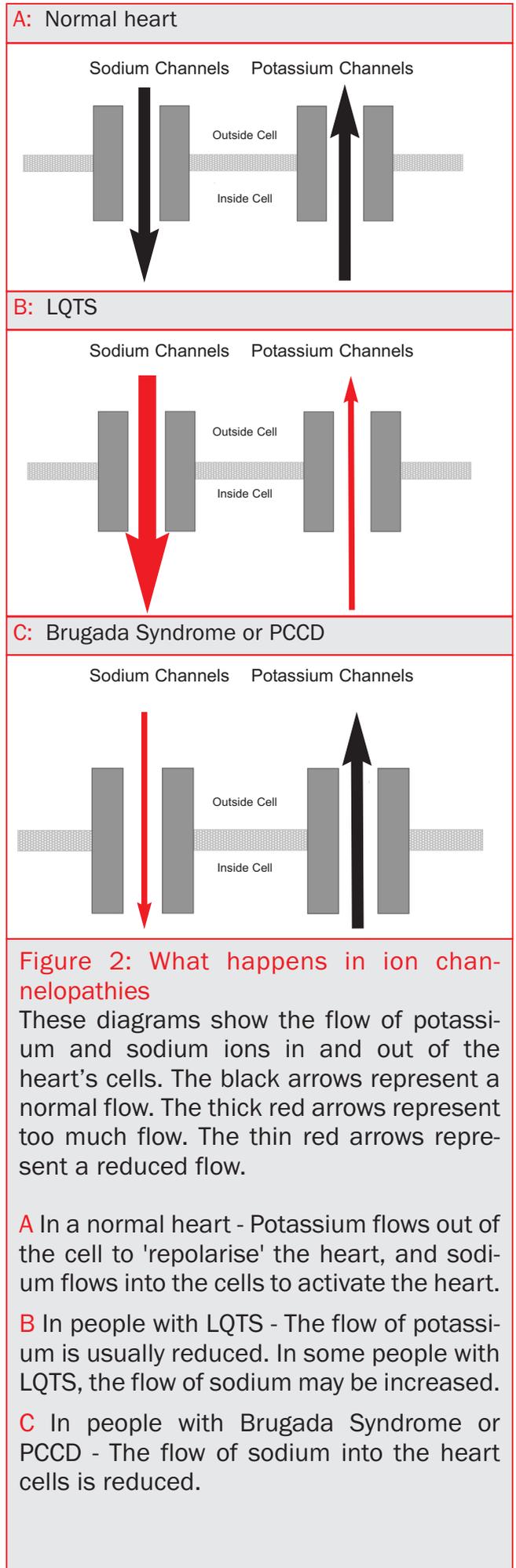
Potassium channel LQTS is associated with sudden death which is related to exercise or when the person has been startled or awoken suddenly ('sudden arousal'). The sodium channel form is associated with death while asleep.

Are there any physical signs?

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 it diagnosed?

Diagnosis involves having an ECG. Sometimes it is possible to tell which ion channel has been affected just by looking at the ECG recording (see figure 3 on page 11). Unfortunately, in many



people who might be carriers, the ECG does not show any sign of the condition. Repeated ECGs, exercise tests and 24-48 hour tape monitoring may be needed before any hint of the condition is seen, and even then there may be no sign of it (we describe all these tests on pages 17-23).

Genetic testing can sometimes identify carriers of LQTS (see page 23). Unfortunately, this form of testing is limited 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. An additional problem is that many families who do have the mutations appear to have a specific change to the DNA code which is not found in other families (known as a 'private' mutation). This sometimes makes 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 makes it very difficult for doctors to decide on the best way to treat people with this condition.

Treatment and advice

If you have LQTS, your doctor will advise you to avoid excessive exercise or strenuous athletic activities. He or she will also advise you to avoid certain drugs that can make the condition worse and which could increase the risk of blackouts and sudden cardiac death. It is also important to avoid low blood potassium levels, known as hypokalaemia (see General Lifestyle Advice on page 24).

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
- young adult women
- specific genetic forms of the condition.

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 antiarrhythmic drugs. These drugs block disturbances in the heart rhythm that can cause sudden death. Potassium supplement pills and 'potassium sparing' water tablets (meaning that potassium is not lost in the urine as with most water tablets) 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 cardiac defibrillator (ICD) fitted, as well as taking your medication. A pacemaker and an ICD both consist of an electronic box 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. 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. (For more information on pacemakers see page 31, and for more on ICDs see page 30).

Surgery

Another option is to perform surgery to disrupt the nerves that release adrenaline and related chemicals at the heart. This is known as 'cervical sympathectomy' and involves operating on the left side of the neck. (For more on this see page 29).

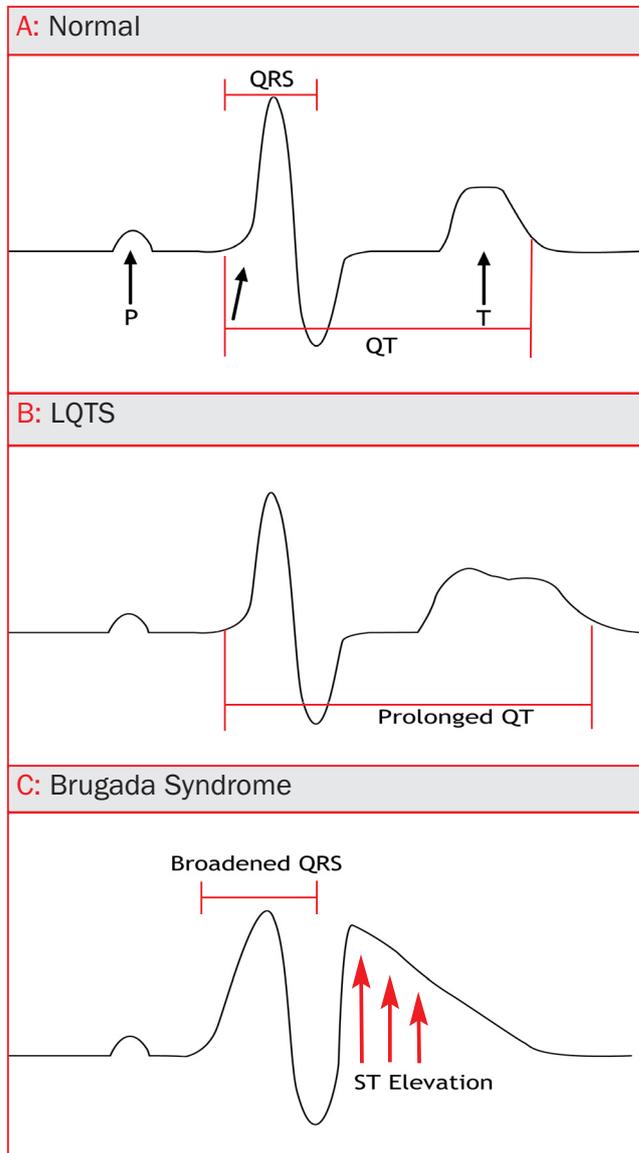


Figure 3: A simplified version of the ECG as it would appear in one lead in the following circumstances:

- A:** Normal
- B:** LQTS
- C:** Brugada Syndrome.

The 'P' wave reflects the electrical activity of the pacemaker and atrium while the 'QRS' reflects the electrical activity of the ventricle pumping. The 'T' wave represents the ventricle resetting itself ready for electrical activation again. This is also known as 'repolarisation' and is measured using the 'QT' interval (the beginning of the QRS to the end of the T wave). In B the repolarisation is prolonged and hence so is the QT interval. In C the area of the ECG tracing connecting the end of the QRS and the T wave (the ST segment) is elevated abnormally with some broadening of the QRS itself (right bundle branch block).

Brugada syndrome

This condition was first identified in the early 1990s. It is an uncommon 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. It has been associated with mutations in the same sodium channel that is affected in LQTS, but this appears to account for only 1 in every 5 people with the condition. The sodium channel behaves abnormally in that movement of sodium ions into the cells is restricted. (See figure 2C on page 9). This results in particular changes on the ECG (as shown in the diagram on the right - figure 3C) but no abnormalities in the structure of the heart.

What are the symptoms?

Some people with Brugada syndrome may have no symptoms at all. In others, the most common symptoms are blackouts. Some people may notice palpitations due to ectopic beats. Sudden death may occur. If it does, it usually happens while the person is sleeping or doing very little.

Are there any physical signs?

There are no associated physical signs.

How is it diagnosed?

Diagnosis involves having an ECG. The changes characteristic of Brugada syndrome may appear on the ECG continuously or intermittently, or they may not show at all. Sometimes the presence of a fever can bring out the ECG changes. If they do not show up on the ECG, there are tests that can make the ECG changes visible. These are called 'provocation tests' and involve having a short injection of an antiarrhythmic drug while you are having an ECG (see page 20). The drugs most commonly used for this are ajmaline and flecainide. There is some controversy, however, about how much reassurance a negative result 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.

Treatment and advice

The outlook for people with Brugada syndrome can be poor, in people who get symptoms or have already had a cardiac arrest, the highest rates of sudden cardiac death being found among young male adults and particularly those who originally come from South East Asia. It is therefore standard practice for high risk carriers to have an ICD fitted as this is very successful form of pro-

tection. (For more information on ICDs see page 30). To date medication has not been shown to be protective in people with Brugada syndrome although there is research into this possibility.

Unfortunately it can be very difficult for doctors to decide how to treat those people who do not get symptoms but who have an abnormal ECG. An EPS (an electrophysiological study) may help to identify those people who do or do not need an ICD but its usefulness is increasingly disputed. Research has suggested, however, that adults with normal ECGs and no symptoms have such low risk that an ICD should not be needed. It is highly unusual for children to be at high risk. Carriers are advised to take fever lowering drugs such as paracetamol or ibuprofen due to the potential effect of fever on the ECG. They are also advised to avoid certain medications and low blood potassium levels, known as hypokalaemia (see General Lifestyle Advice on page 24).

CPVT (Catecholaminergic polymorphic ventricular tachycardia)

CPVT is a rare condition found in young people and children. It causes a particular type of arrhythmia. 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?

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?

There are no physical signs.

How is it 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 useful to help make a diagnosis and 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.

Treatment and advice

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 30).

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 impulse 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 of rapid rhythm disturbances (tachycardia) arising 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 figure 2C on page 9).

What are the symptoms?

Dizziness and blackouts are the usual symptoms. Sudden death may also occur.

Are there any physical signs?

There are no physical signs.

How is it diagnosed?

The ECG abnormalities may be detected either on a standard ECG or with Holter monitoring. An electrophysiological study may also help the doctor make a diagnosis. (We describe all these tests on pages 17-23). If a sodium channel mutation is identified in affected members of a family then it may also be found in other relatives.

Treatment and advice

If you have PCCD you will need to have a pacemaker fitted in order to stop dangerous bradycardia from occurring. This may not prevent 'escape tachycardias' so you may also need to take antiarrhythmic drugs. Some people may need to have an ICD fitted instead of a pacemaker. (For more on pacemakers see page 31, and for more on ICDs see page 30). Medication alone does not help.

Early repolarisation syndrome

Early repolarisation is seen on the ECGs of around 5% of all normal people but it has recently been associated with an increased risk of sudden death. In particular some survivors of a cardiac arrest who have no signs of structural heart disease, long QT and Brugada syndromes or CPVT, have clear early repolarisation changes on their ECG that have worsened dramatically just before their cardiac arrest. This has been described as early repolarisation syndrome and in a few cases mutations of potassium and calcium ion channel genes have been found. The diagnosis and treatment of this condition is still unclear but an ICD is needed in patients who have suffered cardiac arrest.

Mixed sodium channel disease

There are 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 above and can be identified by genetic testing.

Short QT syndrome

Short QT intervals are seen on the ECGs of some families and can be associated with blackouts and sudden death due to ventricular arrhythmias. It is a very rare condition and as in long QT syndrome, the potassium channels are affected, except that too much potassium is allowed to flow out of the cells. If you have short QT syndrome, your doctor may prescribe quinidine or even an ICD to treat your condition.

Structural heart disease

In some cases, the pathologist cannot confirm a diagnosis of structural heart disease - either because there is no evidence of it, or because there is not enough evidence and the heart is felt to be relatively normal. So the 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)
- Wolff-Parkinson-White Syndrome (WPW).

We explain each of these on pages 5-6.

If you are a close blood relative of someone who has died of SADS

Why you need to have tests?

If you are a close blood relative of the person who has died of SADS, it is important that you have tests to find out if you have inherited the same medical conditions as the SADS victim. (We describe all the tests on pages 17-23). If there have been any sudden or suspicious deaths in your family, including cot death, this further suggests that there may be an underlying inherited condition.

What if nothing is found in your family?

In families where someone has died of SADS and the remaining family members are tested, about 4 in every 10 families show no sign of inherited heart disease. This can be due to two reasons:

1. 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.

2. 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 is genetically tested to see if he or she has 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 are those who have symptoms, or have already had a cardiac arrest, or have significant abnormalities on their ECG.

In the meantime, there is little evidence that repeated testing of relatives of someone who has died of SADS is helpful - unless the relative develops new symptoms, or the technology for detecting these conditions improves.

What if something is found in your family?

If you are a relative of someone who has died of SADS and you have been diagnosed with one of the conditions described in 'What causes SADS', you will need regular follow-up - whether you receive treatment or not - unless your doctor believes that you are at very low risk.

If you are young and have been tested, and your doctor thinks that you are not affected, you should have another review in the future. This is because ECG changes can become more obvious with age in children and young adults or they may show up on some ECG recordings but not on others. So, for example, children will need some follow-up until they pass through puberty.

If a recognised mutation was found in the person who died of SADS (or if another relative with signs of inherited heart disease is found to carry one), and if you are found not to have the mutation, then you can be fully assured that you are not affected.

Tests

Because the conditions that cause SADS can be inherited it is important that, if you are a blood relative in the immediate family of someone who has died of SADS, you are evaluated for signs of these diseases, particularly the ion channelopathies. There may also have been other sudden or suspicious deaths in your family, including cot deaths, suggesting that there may be an underlying inheritable condition. Below, we explain what is involved in the evaluation and describe the tests you may need to have.

Molecular autopsy

When and if DNA from the SADS victim has been kept from the post-mortem it can be tested for mutations that cause genetic heart disease. This may help in two ways. Firstly, it can confirm that a mutation found in blood relatives caused the sudden death of the SADS victim. Secondly, if a comprehensive range of genes is tested then the genetic test may be able to diagnose the actual cause of death. This currently only happens in a research setting but may be available routinely in the future.

Medical history

It is vital that a clear 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.

Medical examination

A medical examination may help to discover if there is an inheritable structural heart disease in the 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.

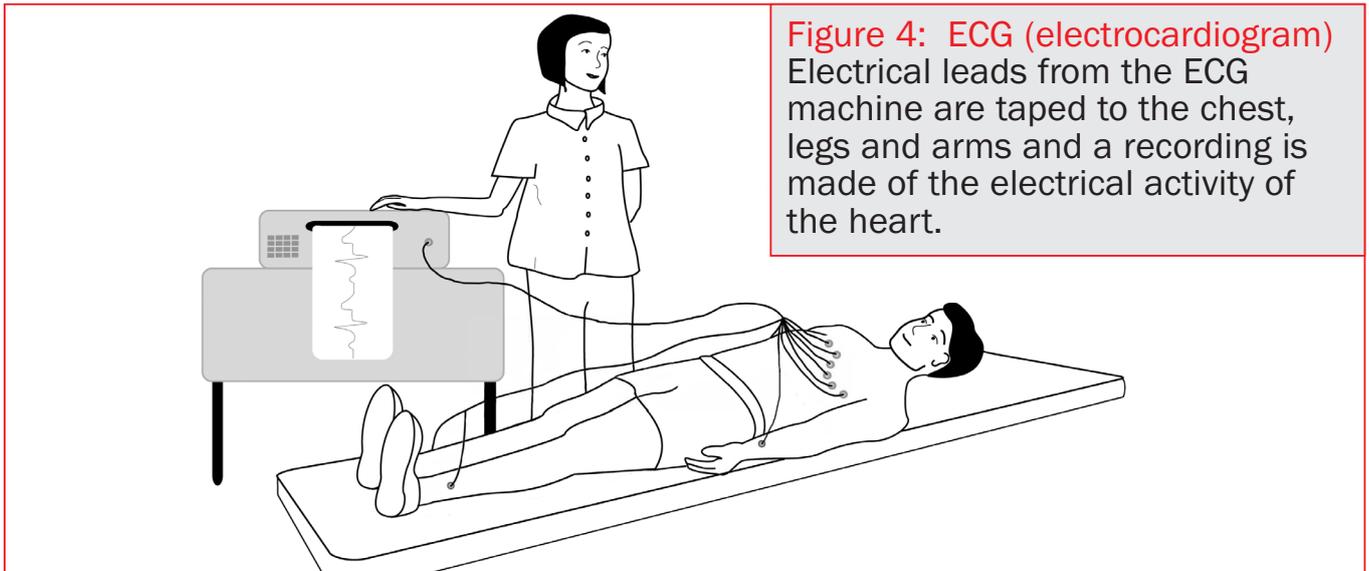
Your doctor may suggest that you have some of the tests we describe below.

ECG (electrocardiogram) *

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

** Tests are non-invasive.*

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

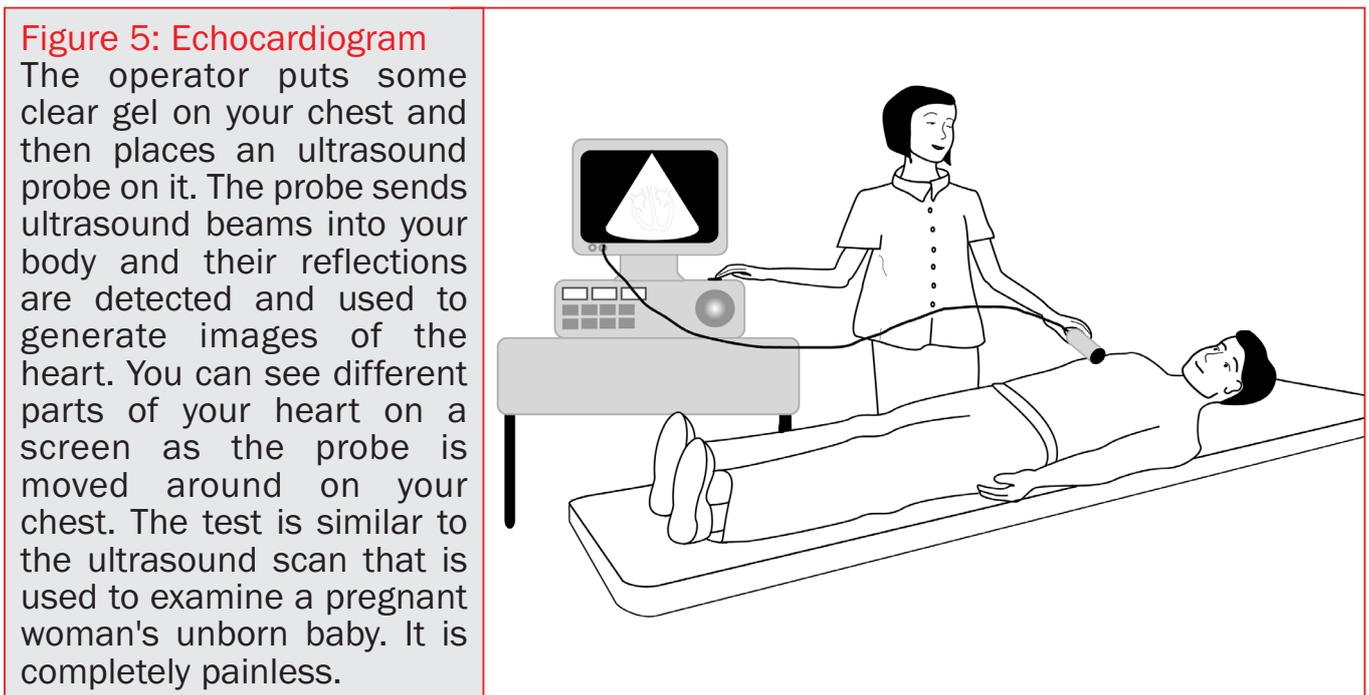


Signal averaged ECG *

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.

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.



* Tests are non-invasive.

Exercise test * (Also called an Exercise ECG)

This test is the same as the ECG described on page 17 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.

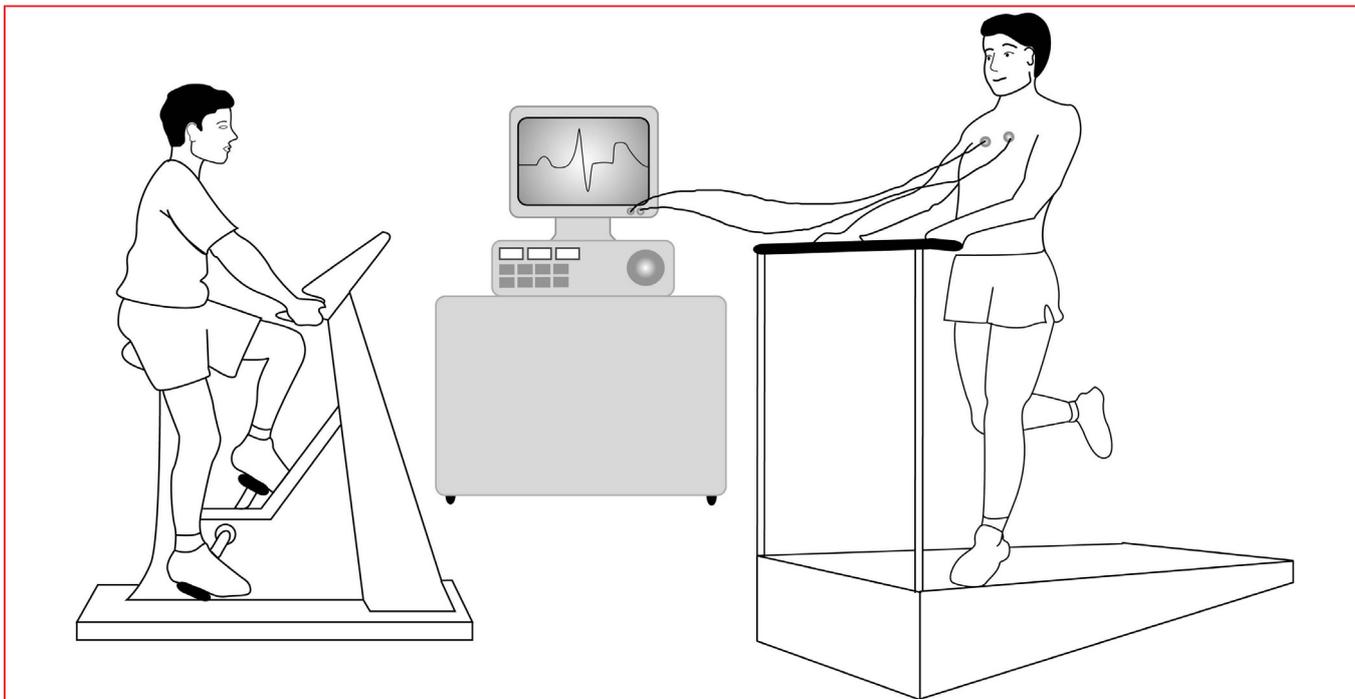


Figure 6: Exercise test.

Electrical leads from the ECG machine are taped to your body and you are monitored while you exercise either on an exercise bike or treadmill. If you are having a 'cardiopulmonary exercise test', your doctor will ask you to breathe in and out of a special piece of equipment while you are doing the exercise, in order to monitor how efficiently your body uses oxygen.

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).

Holter *

The Holter is a recording device that comes in two different forms:

- 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 round your waist. Four or six ECG leads from the

** Tests are non-invasive.*

device are taped to your chest. The device records the electrical activity of your heart for 24 to 48 hours, or for up to 7 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.

Cardiomemo and event recorder *

These are more sophisticated versions of the basic Holter. Whenever you have an attack of symptoms, you can activate the device to record your heart's rhythm. (You can also do this with the digital Holter). The advantage of the cardiomemo 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.

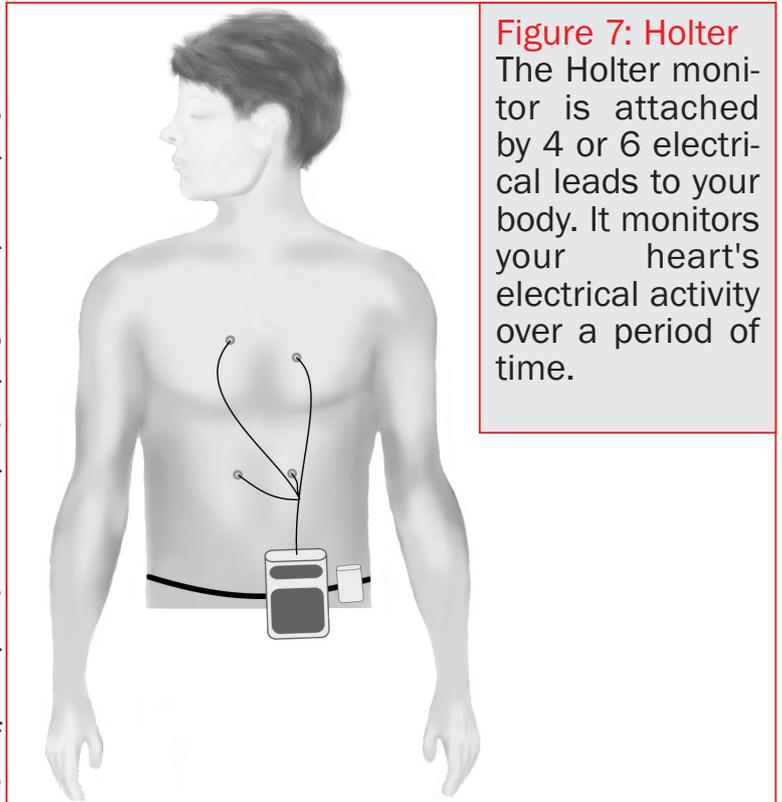


Figure 7: Holter
The Holter monitor is attached by 4 or 6 electrical leads to your body. It monitors your heart's electrical activity over a period of time.

Reveal© device

When it is difficult to assess or record a symptom because it only happens infrequently - as with blackouts - a Reveal© device 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 2 cm 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, a small box with a button can also be placed on the surface of the skin over the Reveal© device. The device may then be activated by pressing the button, causing it to record the preceding 15 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 tests (Ajmaline, flecainide, adrenaline and 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 (antiarrhythmic 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

* Tests are non-invasive.

injected over a short period of time (5-10 minutes) and you will be monitored for 20 minutes or a few hours afterwards, depending on the drug used. There is, however, a risk in 1 in 200 Brugada syndrome carriers or their immediate blood relatives of causing 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 a shorter period of time in the circulation.

Under the same circumstances adrenaline may be given to try and diagnose LQTS or CPVT while adenosine (another short-acting chemical) is given if Wolff-Parkinson-White syndrome (WPW) is considered a possible diagnosis.

Cardiac Magnetic Resonance (CMR) 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 ARVC.

Other tests

Coronary angiography and electrophysiological study (EPS)

Depending on the results of the above tests, your doctor may suggest that you have other tests such as coronary angiography or an electrophysiological study (EPS). Both these tests are performed in an X-ray laboratory that allows the body and any medical tools (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 moving 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 coronary angiography the coronary arteries (the arteries that supply blood to the heart muscle) are injected with a dye to reveal any furring or blockages - coronary artery disease. (The ECG changes that are characteristic of Brugada Syndrome or LQTS can sometimes be caused by coronary artery disease).

An EPS (electrophysiological study) involves placing electrical leads inside the heart to analyse its electrical properties and induce arrhythmias. It may be useful in diagnosing Wolff-Parkinson-White Syndrome (WPW) and PCCD and deciding on what treatment to give people with Brugada Syndrome. If the extra pathway seen in WPW is detected at EPS it can be treated there and then by 'burning' it away using high frequency radio waves. This procedure is called 'RF ablation'.

** Tests are non-invasive.*

There are other tests that may be used to provoke ECG features in LQTS such as 'cold pressor tests'. A stimulus such as placing your hands in ice-cold water can bring out the ECG features of the condition. This does not appear to increase significantly the likelihood of making a diagnosis but is still used at some centres.

Tilt-table testing

Tilt-table testing is used to identify other common conditions that can cause blackouts - such as vasovagal syndrome (see page 33) or simple fainting - that tend to particularly affect young women and girls but have a very low risk of causing sudden death. These symptoms are very similar to the symptoms of more rare and potentially life-threatening conditions like the channelopathies, so it is important to discover the cause of the blackouts so that the doctor can give appropriate treatment. While you lie flat on a table, your blood pressure, pulse and ECG are monitored. The table is then tilted to an angle of 60 to 75 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 10-15 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.

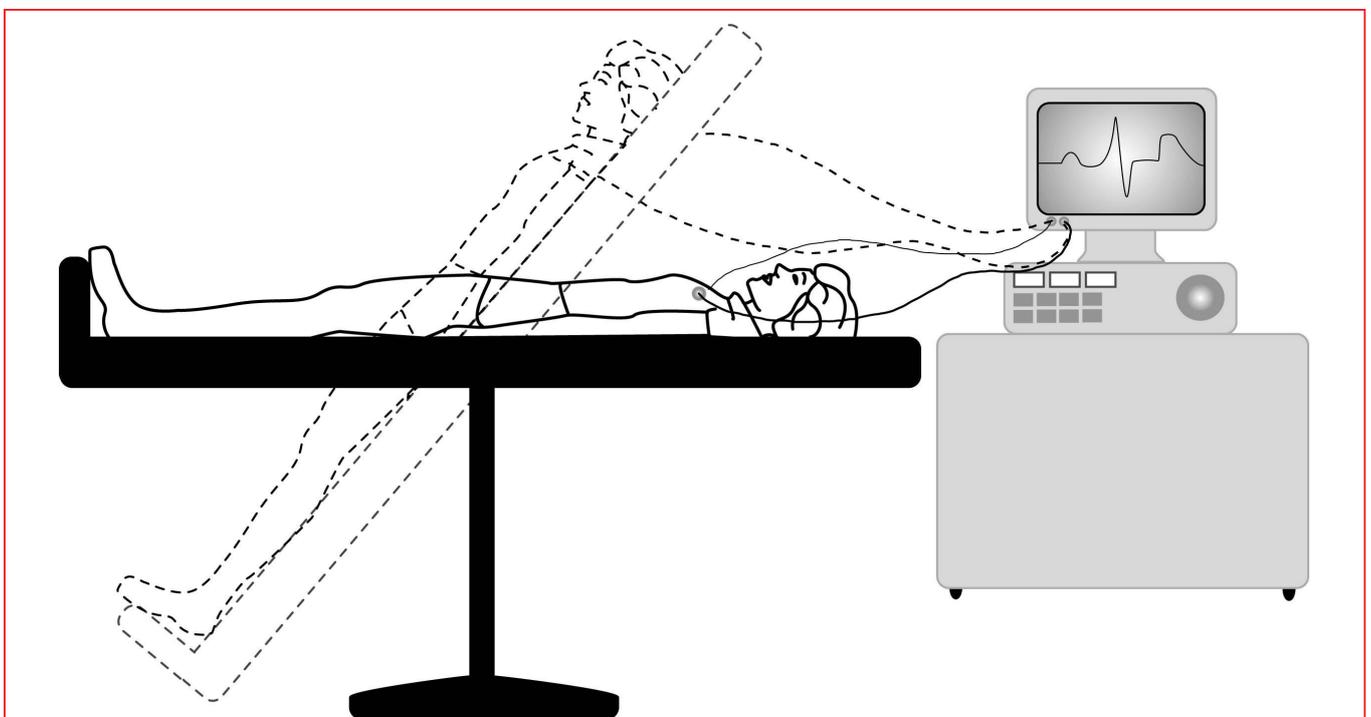


Figure 8: The tilt table test

The tilt table test involves monitoring the ECG, pulse and blood pressure while you are lying flat on a table, then when the table is tilted to 60-75 degrees, and then lying flat again.

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. If we could identify these mutations we would be able to make a diagnosis in any DNA sample, including any obtained from SADS victims at their autopsy or from their relatives who have given blood. Unfortunately this cannot be done at the moment because we don't have complete knowledge of all the genes involved in any condition. For example, 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 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. As research progresses, more genes will be identified and there will be better tools to decide whether the impact of a mutation causes a disease.

* Tests marked with a * are non-invasive.

'Non-invasive' means that it does not involve penetrating the skin or body.

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).

Low potassium in the blood - hypokalaemia

In both the long QT and Brugada syndromes a drop in the levels of potassium in the blood can cause a serious deterioration in the condition. Any prolonged vomiting or diarrhoea (more than a day) can cause a significant loss of potassium. If this occurs it is recommended that rehydration is undertaken using a salt and sugar preparation such as 'dioralyte'. If vomiting prevents this then it is recommended to go to hospital to receive appropriate fluids through a drip.

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, as some can increase the risk of sudden death.

For people with LQTS 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 25. 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.

In people with Brugada syndrome the number and range of drugs that may make the condition worse is unknown and caution must be used. Antiarrhythmics, beta-blockers and some antidepressants are known to interact badly with it. Some medicines are listed on page 26.

Drugs which people with long QT syndrome should avoid

Below is a list of the drugs that people with long QT syndrome should avoid. Please check with your GP and pharmacist if your doctor prescribes any new drugs for you, as this list may not be complete.

* = *Drugs which are no longer available*

Anti-anginals/vasodilators

bepidril, lidoflazine*, prenylamine*, ranolazine, terodiline*

Antiarrhythmics

Class 1: ajmaline, disopyramide, encainide*, flecainide, procainamide, propafenone quinidine
Class 3: almokalant*, amiodarone, azimilide, bretylium, dofetilide, d,l-sotalol*, ibutilide, nifekalant (Japan), sotalol

Anti-cancer

arsenic trioxide, tacrolimus, tamoxifen

Anticonvulsant

felbamate, fosphenytoin (prodrug of phenytoin)

Anti-hypertensives

indapamide, isradipine, moexipril / hydrochlorthiazide, nifedipine

Antihistamines

astemizole*, azelastine, diphenhydramine, ebastine, hydroxyzine, terfenadine*

Antimalarials

amantidine, chloroquine, halofantrine, quinine

Antimicrobials

Macrolide antibiotics: azithromycin, clarithromycin, erythromycin, roxithromycin, spiramycin, telithromycin
Quinolone antibiotics: ciprofloxacin, gatifloxacin, gemifloxacin, grepafloxacin*, levofloxacin, moxifloxacin, ofloxacin, sparfloxacin
Antifungals: cotrimoxazole, fluconazole (caution with itraconazole), ketoconazole, voriconazole
Others: pentamidine, trimethoprim sulfa (bactrim)

Antiviral: foscarnet (HIV)

Anti-migraine

naratriptan, sumatriptan, zolmitriptan

Psychiatric drugs

Tricyclic antidepressants: amitriptyline, amoxapine, clomipramine, desipramine, doxepin, imipramine, nortriptyline, protriptyline
Phenothiazines: chlorpromazine, fluphenazine, prochlorperazine, thioridazine*, trifluoperazine
Others: atomoxetine, citalopram, clozapine, droperidol*, fluoxetine, haloperidol, levomethadyl, lithium, maprotiline, mesoridazine, methadone, paroxetine, pericycline, pimozide, quetiapine, risperidone, sertindole, sertraline, trazodone, venlafaxine, zimelidine, ziprasidone

Serotonin agonists and antagonists

cisapride*, dolasetron, granisetron, ketanserin*, ondansetron

Others

alfuzosin, chloral hydrate, domperidone, galantamine, octreotide, organophosphates, probucol, tizanidine, tolterodine, vardenafil, vasopressin

Stimulant drugs

Some cold remedies contain these drugs so it is important always to check the label.

adrenaline (epinephrine), amphetamine, cocaine, dexamethylphenidate, dobutamine, dopamine, ephedrine, fenfluramine, isoprenaline (isoproterenol), metaproterenol, methylphenidate, midodrine, norepinephrine (noradrenaline), phentermine, phenylephrine, phenylpropanolamine, pseudoephedrine, ritaline, salbutamol (albuterol), salmeterol, sibutramine, terbutaline

Drugs which people with Brugada syndrome should avoid

Below is a list of medications and drugs that people with Brugada syndrome should avoid. Always check new medication before it is taken as this list cannot be guaranteed to be complete.

Alpha adrenergic agonists

methoxamine, noradrenaline

Antiarrhythmics

Class 1: all to be avoided including ajmaline, cibenzoline, disopyramide, flecainide, pilsicainide, procainamide, propafenone

Beta-blockers

all to be avoided especially propranolol

Calcium channel blockers

diltiazem, nifedipine, verapamil

Ergot alkaloids

ergonovine

First-generation antihistamines

dimenhydrinate

Local anaesthetics (non antiarrhythmic)

bupivacaine

Nitrates

isosorbide dinitrate, nitroglycerine

Opioid analgesics

propoxyphene

Parasympathetic agonists

acetylcholine

Potassium channel activators

pinacidil

Psychiatric drugs

Tricyclic antidepressants: all to be avoided including amitriptyline, clomipramine, desipramine, nortriptyline,

Tetracyclic antidepressants: maprotiline

Phenothiazines: cyamemazine, perphenazine

Selective serotonin reuptake inhibitors: fluoxetine

Others: lithium

Others

alcohol intoxication, cocaine, propofol

The future

Research in the 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 be evaluated by a cardiologist to find out if they have an inherited heart disease such as a channelopathy.

For more information

Cardiac Risk in the Young - CRY
1140B The Axis Centre
Cleeve Road, Leatherhead
Surrey. KT22 7RD
www.c-r-y.org.uk www.sads.org.uk
Phone: 01737 363222
Fax: 01737 363444
E-mail: cry@c-r-y.org.uk



Cardiac
Risk *in the*
Young

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

This booklet can also be accessed through the CRY website.

Technical terms

Antiarrhythmic drugs

A group of medicines used to regulate and control the heart's rhythm. They may be of use in specific situations in people with LQTS (e.g. mexiletine) although they are not of proven benefit in preventing sudden death. Ajmaline and flecainide are used in tests used to diagnose Brugada syndrome.

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 heart into the aorta. A bicuspid valve (a valve that has only two cusps or 'flaps') fails to open properly due to the absence of the third cusp and can narrow up due to repetitive damage.

Arrhythmia

A disturbance of the heart's rhythm. A 'ventricular' arrhythmia can be lifethreatening.

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

Atrioventricular 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 natural pacemaker in the atrium to the ventricle. It helps to prevent the heart from pumping too fast if the impulses from the atrium become too rapid.

Bradycardia

Slowing of the heart rate.

Cardiac

'Of the heart' or 'belonging to the heart'.

Cardiac arrest

The state of the heart when it is pumping so erratically or ineffectively that there is no significant blood pressure to supply 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.

Cervical sympathectomy

A form of surgery that is useful for some people with LQTS. It reduces the amount of adrenaline and its by-products produced and delivered to the heart by certain nerves (the left cervical ganglia). It involves operating on the left neck and removing or blocking these nerves, which are not essential to normal function. The procedure can be relatively short but it does need a general anaesthetic.

Congenital heart disease

Disease of the heart, present from birth.

Coronary artery disease

Narrowings and blockages of the arteries supplying the heart muscle, due to 'furring of the arteries'. Also known as 'arteriosclerosis'.

Coroner

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

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.

Delta wave

The ECG feature characteristic of Wolff-Parkinson-White (WPW) syndrome.

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.

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 'SCD'.

Gene

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

Heart attack

When the heart muscle is damaged by an artery becoming blocked and depriving part of the heart of oxygen. This is caused mainly by coronary artery disease. (A heart attack is also called a 'myocardial infarction').

Heart block

A failure of the heart's electrical impulses to conduct properly from the top chambers (atria) to the bottom chambers (ventricles) via the atrioventricular (AV) node. The severity of the condition and its associated risk can vary.

Hypokalaemia

Low blood potassium levels.

Implantable cardioverter defibrillator (ICD)

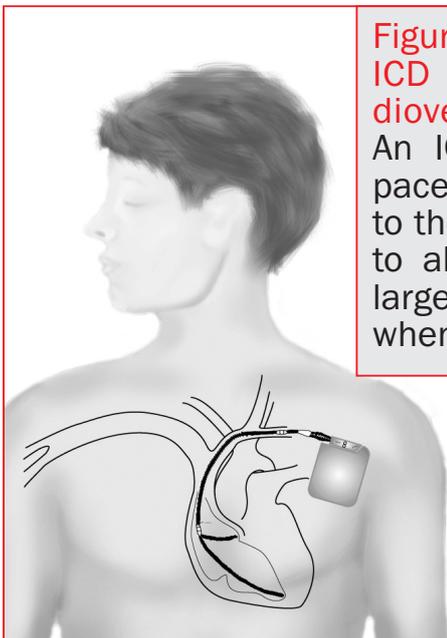


Figure 9:
ICD (implantable cardioverter defibrillator).
An ICD is similar to a pacemaker but the lead to the ventricle is larger, to allow it to deliver a larger shock to the heart when necessary.

A metal electronic device similar to a pacemaker (see left). It can regulate the rhythm of the heart-beat, like a pacemaker does. If a dangerous arrhythmia occurs, the ICD can deliver a shock to the heart. Some people have described the shock as feeling like having a 'kick in the chest'. An ICD is larger than a pacemaker and may have to be positioned under the chest wall muscle at the left shoulder. The procedure usually takes 1 to 2 hours and may require a general anaesthetic. The ICD clinic

checks are needed once every 3 to 6 months. The battery lasts up to 5 years. When a new battery is needed, the box containing it can be replaced easily.

Ion

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.

Ion channel

The route that ions take in and out of the heart muscle cells to allow movement of electricity.

Mitral valve

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

Mitral valve prolapse (MVP)

When the heart is seen on an echocardiogram, the mitral valve can appear 'floppy'. This is very common, and affects around 1 or 2 in every 20 people. It can become more severe and the valve can become thickened and leaky. Only in rare cases it can be inherited in a family and may be associated with arrhythmias and sudden death.

Murmur

The sound of the turbulent flow of blood in the heart, sometimes due to leakage through 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. 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

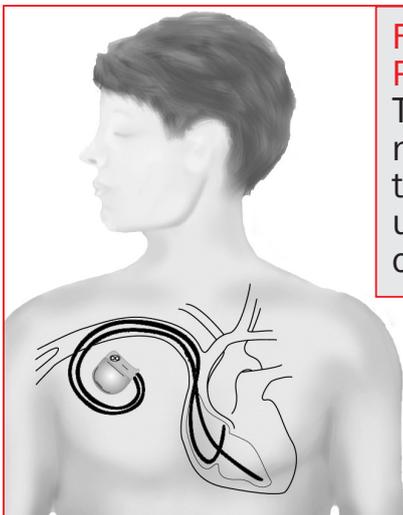


Figure 10:
Pacemaker.

The leads are connected both to the top chamber (atrium) and the bottom chamber (ventricle).

A small metal electronic device with internal batteries. It sits under the skin in the left or right shoulder. It is attached to the top and bottom chambers of the heart by two electrical leads that are inserted via the large veins near the shoulder. These leads both monitor the heart rhythm and allow treatment to be delivered to the heart. Sometimes only one chamber (the ventricle) is connected. A pacemaker can be inserted under local

anaesthetic through a small 2-inch cut in the skin. The procedure takes between 45 minutes and 1 hour.

The device is programmed to prevent the heart from slowing down too much by giving tiny imperceptible shocks that activate the heart, independently from the heart's natural pacemaker. The pacemaker's battery, the leads and the programming are monitored once every 6 to 12 months in a Pacing clinic, using a special magnet and computer software. The battery lasts 5-10 years. When a new battery is needed, the metal box is replaced - a simple procedure that can be performed through the old scar.

Pathologist

A doctor trained to examine the body after death, and samples of its organs, 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.

Prolonged repolarisation

When repolarisation is slower than normal, the time taken for it to occur is described as prolonged. This can be represented on the ECG by abnormalities of T waves and an increase in the QT interval (see figure 3B on page 11).

Pulmonary embolus (PE)

In certain circumstances a large clot can form in the deep veins of the legs - for example after long periods of immobility. The clot can dislodge and travel through the veins to the heart where it can block the arteries supplying the lungs and stop the flow of blood to the body. This can cause a sudden collapse and a rapid death. (Also called 'thromboembolism').

QT interval

An ECG measure of repolarisation from the beginning of the QRS to the end of the T wave.

Repolarisation

The electrical resetting of the heart muscle ready for its next activation. The time taken is measured by the QT interval (see figure 3A on page 11).

RF ablation

The use of high frequency radio waves to 'burn' away small areas of heart tissue such as the extra or 'accessory' pathways seen in Wolff-Parkinson-White syndrome.

Right bundle branch block

A delay of the electricity flowing to the right ventricle of the heart.

SCD 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 (SCD). Some will be unwitnessed or occur during sleep, while others occur during or immediately after exercise (exercise-related sudden cardiac death or ERSCD).

Stethoscope

A piece of equipment which a doctor uses to listen to the heart and chest.

Syndrome

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

Tachycardia

A rapid heart rate.

Thromboembolism

See 'pulmonary embolus' above.

Toxicology

The scientific study of the effects of substances (drugs and chemicals) on the body and mind.

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.

Wolff-Parkinson-White syndrome (WPW)

In this condition there is an abnormal pathway electrically connecting the top (atrium) and bottom (ventricle) chambers of the heart that can be extremely difficult to detect at an autopsy. It can, however, be diagnosed on the ECG as a 'delta wave' although it may not always be present and may require an 'adenosine test' (see page 20) and/or an 'electrophysiological study' (see page 21) to confirm its presence. It can by-pass the usual electrical regulation of the AV node and cause an abnormally rapid conduction of electrical impulses from the atria to the ventricles. In a small proportion of cases this can be severe enough that it leads to a cardiac arrest. It is rare that this condition is inherited but if so it is usually accompanied by other conditions such as unusual forms of hypertrophic cardiomyopathy.

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Coping with a young sudden cardiac death

The death of a child or young adult is so totally out of order with the sequence of life that it can have devastating consequences within the family. With a sudden death, not only has there been no preparation for such a death as in terminal illness, nor is the death accidental when there is an obvious and direct link between an occurrence and the tragic consequences. This can lead to those closest to the one who has died blaming themselves for overlooking possible symptoms. Dealing with their terrible loss is then compounded by feelings of guilt.

Devastating grief is not just something that will affect your emotions. It can also have physical consequences that leave you exhausted, feeling sick and unable to eat or sleep. When there has been a young death from a heart disorder - particularly if there is a possibility that this may have been a genetic condition - family members can subsequently start suffering from breathlessness, chest pains and dizziness, all of which are recognisable cardiac symptoms which can in themselves be frightening.

Sharing the way you feel about what has happened is very important, but it is not always easy to do this with others who are suffering directly from the same loss.

CRY is a charity that offers help, support and counselling to families where there has been a sudden cardiac death of an apparently fit and healthy young person. Please call us on 01737 363 222 if you want any further help or information, or if you would like to be put in touch with someone who has gone through a similar experience to yourself.

A handwritten signature in black ink, appearing to read 'Alison Cox', with a horizontal line underneath.

Alison Cox
Founder and Chief Executive of CRY



Cardiac Risk in the Young is a charity which:

- aims to reduce the frequency of young sudden cardiac death by working with cardiologists and family doctors to establish good practice and appropriate screening facilities to promote and protect the cardiac health of our young
- offers bereavement support to those who have suffered a loss, through a network of affected families and counselling
- offers supports to those young people diagnosed with cardiac conditions
- publishes and distributes medical information written by leading cardiologists for the general public
- promotes and develops heart screening programmes and funds medical research, and
- funds fast track services at the CRY Centres for Sports Cardiology, Cardiac Pathology and Inherited Cardiovascular Conditions at leading London Hospitals.

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