



# LIFE WITH SUDDEN ARRHYTHMIC DEATH SYNDROME

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I fundraise  
to honour my  
brother

I remember  
Mark and  
help others

An inherited  
heart condition  
changed my life

**JASON'S STORY,  
PAGE 06**

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

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

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You may be reading this booklet because a relative of yours – perhaps a close member of your family – has died suddenly and unexpectedly. You may still be asking why this tragedy happened, and how it could have happened to someone who perhaps seemed so healthy. Maybe your doctor has suggested that you should have some tests to find out if you have inherited the same condition.

An ‘inherited heart condition’ is a condition which can run in families. It can affect one or several members of the same family. Sadly, some inherited heart conditions are often not diagnosed until one person dies suddenly and unexpectedly – this is called ‘sudden cardiac death’ and, if the death is unexplained, it is known as **Sudden Arrhythmic Death Syndrome** – or **SADS** for short. There are several different types of inherited heart conditions which can cause SADS.

This booklet:

- Describes how the normal heart works.
- Explains what the terms 'sudden cardiac death' and 'Sudden Arrhythmic Death Syndrome' mean and describes the conditions that can cause SADS.
- Explains why it's important that close blood relatives of someone who has died should have an assessment to find out if they've inherited the same condition.
- Explains what happens when you go to an inherited heart conditions clinic and the healthcare professionals that may be involved in your family's care.
- Describes the tests your doctor may ask you to have.

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 35.

We hope that this booklet will help you understand why a close family member may have died unexpectedly and help you and your family come to terms with what has happened. For information on where to get further support, see page 39.

At the **British Heart Foundation** we're fighting for every heartbeat. The research we fund has helped push the boundaries of our understanding of genetics, and given us tools we can use to find and help people at risk of dangerous heart conditions and sudden arrhythmic death syndrome. Join the fight at **bhf.org.uk**

**Cardiac Risk in the Young** is a charity that aims to reduce the frequency of young sudden cardiac death by working with cardiologists and family doctors to establish good practice, appropriate screening facilities and by promoting and protecting the cardiac health of young people. Help support us by visiting **www.c-r-y.org.uk**

# UNDERSTANDING YOUR HEART

# JASON'S STORY

My brother Mark died in his sleep from an inherited heart condition.

It's never really the same after someone dies, and it can be hard to accept. My way of coping has been through fundraising as a way of honouring Mark. I did the Bournemouth Pier to Pier swim, amongst other charity events which personally was such a great thing, remembering Mark whilst helping others.

After Mark's death we were advised to go as a family and get checked out. Luckily, I was given the all clear, but I still go for regular check-ups. My doctor still wants to keep an eye on me, which is brilliant.

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I fundraise to honour my brother

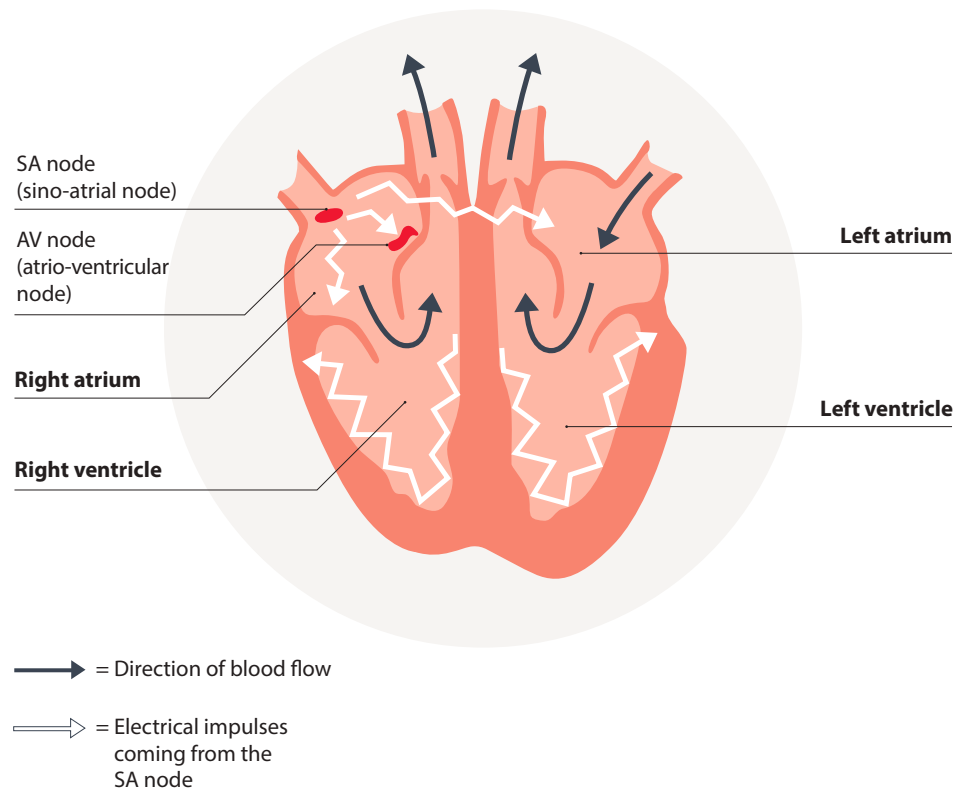
I remember Mark and help others

An inherited heart condition changed my life



## THE NORMAL HEART

The heart is a specialised muscle that contracts regularly and continually, pumping blood around your body. The pumping action of your heart 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 an arrhythmia – it can affect your heart’s ability to pump properly.



### How the heart functions electrically

- Your heart has four chambers – two at the top (the atria) and two at the bottom (the ventricles).
- The normal trigger for your heart to contract starts in 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 atria contract and pump blood into the ventricles.
- The electrical impulses then pass to the ventricles through a form of ‘junction box’ called the AV node (atrio-ventricular node). This causes the ventricles to contract and pump blood out of your heart.
- The blood from the right ventricle goes through the pulmonary artery to your lungs, and the blood from the left ventricle goes through the aorta and then around your body.

The speed of your heart’s pacemaker and the force of the pumping action of the ventricles are also controlled by nerves that regulate your heart by releasing certain chemicals that circulate in your blood. For example, adrenaline increases your heart rate and the volume of blood pumped out of your heart.

## 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 one in every 25 cases of sudden cardiac death, no definite cause of death can be found. This is then called **Sudden Arrhythmic Death Syndrome** – or **SADS** for short. It’s 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?

After an unexpected sudden death, it's usual for the coroner to ask for a post-mortem or autopsy to be carried out to determine the cause of death. The next of kin will be contacted by the coroner's officer to obtain information and communicate with the family.

The autopsy involves a pathologist examining the body to see if there is a cause for the unexpected death. They will look for conditions such as coronary heart disease (furring of the arteries) or pulmonary embolus (a clot on the lung). The pathologist will inform the coroner if there is an established cause of death or not and may suggest that other tests are required. The coroner will take into account the circumstances of the death and, if necessary, will approve further tests, including:

- Examining small samples of tissue taken from organs, including the heart, under a microscope.
- Tests for signs of any medications or drugs in the body.
- Assessment of the heart by an expert heart pathologist.

When all the information is collected, the coroner will decide on the cause of death and write the death certificate.

If the death is unexplained or if it's thought that it may be caused by a faulty gene (see page 14), it's best practice for the pathologist and the coroner to recommend that the family be seen in an inherited heart condition clinic (see page 23). In addition, a sample of tissue from the initial autopsy may be kept and can be used for post-mortem genetic testing sometimes known as 'molecular autopsy' (see page 33).

## CONDITIONS THAT CAN CAUSE SADS

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Mark's memorial fund supports young people in sport

I want to support children like my daughters

An inherited heart condition changed my life



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# JOANNE'S STORY

When my husband, Mark, died it left a massive hole in a lot of people's lives.

He died of an inherited heart condition, so it was important to get our children checked over.

Our daughters Rebecca and Victoria now have regular cardiology appointments. Because they've done it since they were young it's just part and parcel of their lives.

Mark was a great sports fan – he was a huge fan of the local rugby team. And so we started a memorial fund to support young players. It helped pay for the training, kit and exams for the kids whose families couldn't afford it.

There are photos of Mark around in the house and we talk about him. He's still very much around.



## WHAT CAUSES SADS?

SADS happens when a certain underlying condition causes someone to have a dangerous arrhythmia – a disturbance in the heart’s rhythm – which brings on a cardiac arrest.

A cardiac arrest is when the heart stops pumping blood around the body. Someone who is having a cardiac arrest will suddenly lose consciousness and will stop breathing or stop breathing normally. Unless treated immediately, this leads to death within minutes.

A group of uncommon diseases called **ion channelopathies** can cause life-threatening arrhythmias and are probably responsible for about three in every ten cases of Sudden Arrhythmic Death Syndrome. Ion channelopathies affect the electrical functioning of the heart without affecting the heart’s appearance or structure. This means that the problem can only be detected while a person is alive; it can’t be detected at a post-mortem. There are several different types of ion channelopathies including:

- **Long QT syndrome (LQTS)**
- **Brugada syndrome**
- **Catecholaminergic polymorphic ventricular tachycardia (CPVT)**
- **Progressive cardiac conduction defect (PCCD).**

We describe each of these on pages 15-21.

Other causes of SADS include **structural heart disease** and **conduction disease**.

For more on structural heart disease, see page 22. Conduction disease includes abnormalities in the way that the electrical impulses are conducted through the heart, or because there are ‘extra’ electrical pathways in the heart, as in Wolff-Parkinson-White (WPW) syndrome. For more information on conduction disorders and WPW syndrome, see the BHF booklet *Heart rhythms* and our medical information sheet *Wolff-Parkinson-White (WPW)*.

### What causes an ion channelopathy?

Ion channelopathies are caused by abnormalities in an individual’s genetic make-up. Each one of us has our own genetic information that makes us unique. Your genes make you who you are, for example what colour your hair is, your blood type and your gender. This genetic information is held in your DNA, in the cells of your body. Your genetic information acts as a code that allows a system of proteins to be created. This tells all of the cells in your body what their function should be.

If there’s a mistake in one of these codes, your cells may not work as they should do. These mistakes are known as **gene mutations** or **gene faults**. Certain gene mutations will cause an ion channelopathy. They are usually inherited from your parents, although they can occur for the first time in a family. If they occur for the first time in a single family member they are described as ‘sporadic’. In some cases, a person may inherit a gene mutation, but not show any symptoms of the condition. However, these people are still able to pass the gene mutation on to their children and are known as ‘carriers’ of the condition.

Some mutations affect certain genes that are responsible for the production of ‘ion channels’ in your 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 your heart muscle.
- An ion channel is a gateway that allows ions to pass in and out of your cells.

For example, a potassium channel is an ion channel that allows potassium ions to pass in and out of your cells. In your heart, the movement of ions in and out of heart muscle cells controls the heart’s electrical impulses. This allows you to produce a heartbeat.

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. For more information on each of these conditions, see our booklet *Life with Inherited abnormal heart rhythms*.

## LONG QT SYNDROME (LQTS)

LQTS is the most common and best understood type of ion channelopathy.

- It occurs in as many as one in 2,000 people.
- For around seven in every ten people with LQTS, the ion channels involved have been identified.
- In most cases, two potassium channels that regulate the movement of potassium ions from the inside to the outside of cells are affected.
- In a smaller proportion of people with LQTS, sodium or calcium channels are affected.

There are currently 13 known types of LQTS and each type is associated with different ion channel abnormalities. The type of LQTS you have depends on which gene mutations you inherit.

In people with potassium channel-related LQTS the ion 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. This results in an electrical disturbance in the cells of your heart, which means that your ventricles take longer to recharge after each heartbeat.

This can sometimes be seen on an electrocardiogram (ECG) recording as a lengthening of a part of the heartbeat cycle known as the QT interval. This is where the name long QT syndrome comes from. For more information on having an ECG test, see page 30.

### What are the symptoms of LQTS?

The most common symptom of LQTS is blackouts, caused by an arrhythmia. Sometimes palpitations due to extra or ectopic heartbeats can be a problem. Some types of LQTS are associated with sudden death related to exercise, when a person is asleep or when the person has been startled or awoken suddenly.

Symptoms vary from person to person. 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.

### How is LQTS diagnosed?

Diagnosis involves having an ECG (see page 30). Sometimes experts can tell which ion channel has been affected just by looking at your ECG recording. In some cases, diagnosis can be very difficult and many people with the condition have a normal ECG. Repeated ECGs, exercise tests, 24–48 hour Holter monitoring and adrenaline testing may also be needed before the condition is diagnosed. We describe all these tests on pages 30–33. However in some cases, there may be no clear sign of the condition.

Genetic testing can also be used to identify carriers of LQTS (see page 32).

### How is LQTS treated?

The treatment you'll need depends on the risk of dying suddenly. People at the greatest risk of sudden death are those with one or more of the following features:

- a previous cardiac arrest
- blackouts
- a very long QT interval on the ECG
- young adult women
- certain gene mutations
- having more than one gene mutation.

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

### Medicines

The first line of treatment is with medication. The most commonly used medicines are beta-blockers. These block the effects of adrenaline and associated natural chemicals in your body that make your heart pump harder and faster. They are effective in the most common forms of LQTS, as they reduce your symptoms and most importantly your risk of sudden death.

### Cardiac devices

If there's a high risk of sudden death (for example if you have already had a cardiac arrest) or if medication has 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 medicine.

A pacemaker and an ICD consist of:

- a small metal box called a pulse generator, containing a battery
- one, two or three electrical leads known as electrodes that deliver electrical impulses to the heart muscle.

### Pacemaker

A pacemaker can prevent your heart from beating too slowly by 'pacing' your heart to make your heart rate faster. A pacemaker works by:

- monitoring and storing information about your heart rhythm and heart rate
- sending electrical impulses to your heart that stimulate it to contract. This helps to control your heart rate and stop any excessive slowing of your heart that could trigger an arrhythmia.

For more information on pacemakers, see our booklet *Pacemakers*. To order our booklets see *More information on page 39*.

**ICD**

An ICD is slightly larger than a pacemaker and monitors your heart rhythm through electrodes placed into your heart. If it detects a dangerous arrhythmia it can deliver a small electrical shock to restore your heart's normal rhythm. This is called shock therapy. An ICD can deliver the following treatments:

- sending electrical impulses to your heart that stimulates it to contract (in the same way as a pacemaker)
- cardioversion – one or more small electric shocks to restore your heart's normal rhythm
- defibrillation – one or more larger electric shocks to get your heart back into a normal rhythm.

For more on ICDs, see our booklet *Implantable cardioverter defibrillators (ICDs)*. To order our booklets see More information on page 39.

**Surgery**

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

**Advice on physical activity for people with LQTS**

If you have LQTS, your doctor is likely to advise you to avoid excessive exercise or strenuous athletic activities.

For more information on LQTS, see our booklet *Life with Inherited Abnormal Heart Rhythms*.

**BRUGADA SYNDROME**

Brugada syndrome affects mainly young and middle-aged adult men. It is not a common condition in the western world, but seems to be much more common in South East Asia.

Brugada syndrome can be caused by mutations in one of several different genes. In some cases these mutations affect the function of sodium channels. This can affect how sodium ions pass in and out of your cells, which can affect the way your heart beats.

**What are the symptoms of Brugada syndrome?**

Most people with Brugada syndrome have no symptoms at all. Others may have palpitations, blackouts or even a sudden cardiac arrest (which can lead to sudden death).

**How is Brugada syndrome diagnosed?**

Diagnosis involves having an ECG (see page 30). The changes characteristic of Brugada syndrome may appear on your ECG continuously or come and go, or they may not show at all.

If your ECG appears normal, a provocation test can be used to make the ECG changes associated with Brugada syndrome visible (see page 31). During the test, you'll receive an injection of a small amount of an anti-arrhythmic medicine while having an ECG. The medicines most commonly used for this are ajmaline and flecainide. However, ECG changes may still not be seen and you may need to have further tests.

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

**How is Brugada syndrome treated?**

There is a risk of dying suddenly for people with Brugada syndrome, which is greatest in adult males and most often happens during sleep or when resting. It's therefore common practice for people at high risk of sudden death to have an ICD fitted. For more information on ICDs, see page 18. Medication to treat Brugada syndrome may be possible in the future, thanks to promising research on two drugs, quinidine and hydroquinidine. However, these drugs are not currently licensed for use in the UK.

If you have an abnormal ECG but don't have any symptoms, it can be very difficult to determine your risk of dying suddenly and decide what treatment you should have. An EPS (electrophysiological study) may help your doctors decide whether you need an ICD or not (see page 32). However 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're 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 your risk of developing an arrhythmia. If the fever persists you may need further treatment and close monitoring in hospital until it settles.

For more information on Brugada Syndrome, see our booklet *Life with Inherited Abnormal Heart Rhythms*.

## CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT)

CPVT is a rare condition which can cause a particular type of dangerous arrhythmia – known as ventricular tachycardia. It's found mainly in children and young people, and is associated with gene mutations that affect the levels of calcium inside your cells. Having too much calcium in your cells can result in ventricular tachycardia.

### What are the symptoms of CPVT?

Some people with CPVT have no symptoms at all. Others may have palpitations or blackouts. In some cases, sudden death may occur when 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.

### How is CPVT diagnosed?

If you experience symptoms, your doctor will refer you to a specialist who will do an exercise test and possibly an adrenaline test. These tests use an ECG that records your heart's electrical rhythm whilst you are exercising or receiving a slow adrenaline injection (see provocation tests on page 31).

Genetic testing is also useful if a member of your family has been found to carry a gene mutation and is showing the signs of the condition (see page 32). This can identify members of the same family that carry the same gene mutation and are at risk of developing CPVT.

### How is CPVT treated?

Your doctor will advise you to take beta-blockers (a type of medicine) and to restrict the amount of exercise you do. This combination greatly improves the outlook for people with CPVT. Sometimes another medicine called flecainide is useful if beta-blockers cannot control your arrhythmia.

If you're at high risk of dying suddenly, you may need to have an ICD fitted (for more on ICDs, see page 18). You may also be considered for cervical sympathectomy or cardiac denervation surgery to disrupt the nerves that release adrenaline into your heart (see page 36).

For more information on CPVT, see our booklet *Life with Inherited Abnormal Heart Rhythms*.

## PROGRESSIVE CARDIAC CONDUCTION DEFECT (PCCD)

PCCD – also known as Lev-Lenègre's syndrome - is a rare condition. In people with PCCD, the heart's electrical impulses pass through the heart very slowly and over time this can lead to 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 also cause other types of arrhythmias, including:

- slow heart rhythms (bradycardias)
- fast heart rhythms (tachycardias).

In some people, PCCD has been associated with sodium channel mutations that cause changes in ion channel behaviour similar to those found in people with Brugada syndrome.

### What are the symptoms of PCCD?

Some people with PCCD have no symptoms at all. Others may have episodes of dizziness or blackouts, and sudden death may also occur.

### How is PCCD diagnosed?

Arrhythmias characteristic of PCCD may be detected either on a standard ECG or with 24-hour Holter monitoring. An electrophysiological study (EPS) may also help your doctor make a diagnosis. (We describe all these tests on pages 30-33.)

If a sodium channel mutation is identified in affected members of a family, genetic testing may also be used to find the same gene mutation 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 your heart rate from dropping too low. However, this does not prevent your heart from beating too quickly, so you may also need to take anti-arrhythmic medicines. Some people may need to have an ICD fitted. (For more information on pacemakers and ICDs, see page 17.) Medication alone does not appear to help.

For more information on PCCD, see our booklet *Life with Inherited Abnormal Heart Rhythms*.

## STRUCTURAL HEART DISEASE

Structural heart disease is when there's a problem with part of the heart's structure, for example the heart muscle or the valves of heart. In some cases it's difficult to know if the person who has died had structural heart disease. This can be because there was no diagnosis of it, or because when the heart is examined after death, it looks relatively normal. In these cases the cause of death is likely to be recorded as sudden arrhythmic death. The presence of very subtle structural heart disease in the person who's died may be enough to cause sudden cardiac death. In these cases the cause of death will also be recorded as SADS.

In these circumstances the most common causes of death are:

- arrhythmogenic right ventricular cardiomyopathy (ARVC)
- dilated cardiomyopathy (DCM)
- hypertrophic cardiomyopathy (HCM), and sometimes
- mitral valve prolapse (MVP – see below).

For more information, see our booklets on cardiomyopathy or contact Cardiomyopathy UK. (For contact details, see page 39.)

### Mitral valve prolapse

Mitral valve prolapse is when the mitral valve (the valve which controls the blood flow between your heart's left atrium and ventricle) is 'floppy' in appearance. This will show up on an echocardiogram (see page 31). Most people with mitral valve prolapse don't have any symptoms, and the condition is not harmful. In rare cases, mitral valve prolapse can be inherited in a family and can then be associated with arrhythmias and sudden death.

# THE INHERITED HEART CONDITIONS CLINIC

# DONNA'S STORY

My son Adam was 23 when he died. We found out that he'd died from an inherited condition called Brugada syndrome.

It was important for me and my two other children, Ash and Rhianna, to get tested - thankfully we were given the all clear.

Adam's death was a really terrible tragedy for us. I understood what Ash and Rhianna were going through in losing their brother. I too lost my sister when I was younger. But one positive is that they are extremely close now.

For a time after Adam died everything got on top of me. But then I thought I've got to get a grip, I've got two other children who rely on me.

I have to be careful not to wrap them up in cotton wool and just let them live their lives.

It will affect your life forever, but you have to find normality again. You owe it to yourself and to those closest to you.

I raised money to help support young people like Adam

I want to stop anyone else going through what I did

An inherited heart condition changed my life



## ATTENDING THE CLINIC

### Why have you been referred to an inherited heart conditions clinic?

You may have been referred to a clinic which specialises in inherited heart conditions because a close family member has died very suddenly – possibly from an inherited heart condition. When one family member dies of an inherited heart condition, other close relatives may also be at risk from the same condition.

### What is an inherited heart conditions clinic?

An inherited heart conditions clinic is where a team of specialists investigate, treat and support families who have an inherited heart condition. Assessment at a clinic can help to tell which other family members may be at risk and then offer treatment if necessary.

### What happens before you go to the clinic?

When your referral has been received you will be contacted and sent an outpatient appointment. You may also be offered a telephone assessment before your outpatient appointment. Your appointment letter will explain what will happen on the day and the tests you may need to have. You can have an individual appointment but some services can see families together.

Many centres see young children or otherwise they will be referred to the nearest specialist centre.

### What happens during the appointment?

During the appointment you'll be asked questions about yourself, your health and any symptoms that you have. You'll be asked about your family history – so it's helpful to bring any information that you know about your family; their ages, any health or heart problems and if relevant, the cause and age of any deaths. The healthcare team will be particularly interested in information about anyone who has collapsed, blacked out, suffered from fits, or if there have been any sudden deaths, including cot death. If there's been a sudden death in the family, it's useful to bring a death certificate or a coroners or post-mortem report, if you have it.

From the information you provide, the nurse or doctor will draw your family tree; this can help to clarify if there is an inherited condition running in your family and can also help to identify which members of your family may also benefit from screening.

### What tests will I need to have?

You'll have a physical examination and your doctor will want to listen to your chest and heart and take your blood pressure. It's likely that you will need to have some tests. We describe the tests you may have on pages 30-33. Sometimes these tests are carried out on the same day or you may need to return for a separate appointment. If you have lots of tests on the same day your visit may last for several hours. After your tests you will see a consultant or a member of their team to go through your results and discuss your care and treatment. You may be asked to return for ongoing appointments.

If you require a genetic test, some clinics will be able to offer this, along with genetic counselling, or you may be referred to the nearest Regional Genetics Centre that can provide the tests.

### Who will I meet at the clinic?

An inherited heart conditions clinic is run by a team of different healthcare professionals. The following may be involved in your care:

- **Consultant cardiologist** – who specialises in inherited heart conditions. They will help to rule out a condition, or give you a diagnosis. They are responsible for your ongoing management and treatment. For children attending the clinic, a children's cardiologist will also be involved.

- **Other cardiologists** – for example cardiologists who specialise in the electrical function of the heart and devices to treat heart rhythm problems.

- **Nurse specialist** – they offer information, advice and support, including psychological support to you and your family. They assist with the practical aspects of living with an inherited heart condition and often act as a link between you and other healthcare professionals within the team.

- **Geneticist** – specialists in genetics and the science of genes and familial disease. They will review your family history and decide which genetic tests you need to have.

- **Genetic counsellor** – provides information to help you make a decision about genetic testing. They help you to understand how genetic information may impact your future and your family. In some clinics, the nurse specialist also does this role.

- **Clinical psychologist** - provides support to you and your family, and can help you deal with the psychological impact of an inherited heart condition.

- **Cardiac technicians/physiologists** - carry out some of the tests that you might need and are specialists in obtaining precise readings and images of your heart and its function.

## IMPLICATIONS FOR YOUR FAMILY

When someone has died from SADS, close family members should be seen in the clinic - because the condition can be shared within the family. The clinic team will explain how your relatives can be referred for screening.

### What if your family shows no signs of an inherited heart condition?

Sometimes, in families where someone has died of SADS and the remaining family members are tested, there are no signs of inherited heart disease. This can be due to two reasons:

Reason 1: **The person who has died may not have inherited any abnormality from his or her parents.** Either the person was the first to suffer a gene mutation in the family (and therefore their children are the only relatives who are at risk), or there was another cause which has not been identified and which is not inherited.

Reason 2: **Some family members may be carriers but show no signs of any disease.** It's impossible to give 100% reassurance that a relative is not a carrier, except where a gene mutation has been identified in either the person who has died or another relative, and genetic testing has shown that other family members don't have the same gene mutation. However, people who do not have any symptoms or signs are at low risk of sudden death. 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 an inherited heart condition is diagnosed in your family?

If you're a relative of someone who has died of SADS, and you have been diagnosed with one of the conditions described on pages 14-22, you will need regular follow-up unless your doctor believes that you are at very low risk.

If you're young and have been tested, and your doctor thinks that you are not affected, you should still be assessed again in the future, unless a genetic test has ruled out that you have the condition. 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 regular follow-up especially while they are going through puberty.

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

## EMOTIONS YOU MAY BE FEELING

This may be an extremely difficult time in your life. You may be worried that you or a member of your family might have an inherited heart condition. Or you may have recently lost a close member of your family. Some people don't feel ready to go to a clinic appointment after a recent bereavement – but the earlier you are seen, the sooner a heart problem can be ruled out or diagnosed. If an inherited condition is found, it can be monitored and treated to help prevent serious complications.

The nurse specialist in the clinic can give you ongoing care for your emotional needs. Some services may also be able to offer a referral to a bereavement counsellor for extra support.

Ideally it's best to openly talk within your family when it comes to inherited heart conditions, because of the impact it may have on others. However, family relationships are sometimes difficult, so dealing with this issue with your family can be an added burden. There are many ways to inform family members and the nurse specialist and genetic counsellor can help you with this.

It's natural to feel anxious and worried whilst waiting for tests. If you're a parent you may be concerned about your children inheriting a heart condition. Keep in mind that this is something beyond your control and if a diagnosis is made, a lot can be done to prevent complications. Many of those affected are able to lead normal, good quality lives.



**The Genetic Information Service (GIS)**

The British Heart Foundation's GIS can provide you with information and support if someone in your family has been diagnosed with, or has died from, a suspected inherited heart condition. This includes supporting you and your family by helping you get an expert assessment in a specialist clinic that deals with inherited heart conditions.

Call our GIS on **0300 456 8383** to speak to one of our specialist nurses for information about inherited heart conditions and how they can affect you and your family. For more details, see page 39 and our booklets ***Losing someone to heart disease*** and ***Heart to heart - heart disease and your emotional wellbeing***.

**TESTS FOR INHERITED HEART CONDITIONS**

In this section we describe the tests that relatives of a person who has died of SADS may have as part of an assessment at an inherited heart conditions clinic. These tests help to identify any family members that may have the same condition as the person who has died.

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

**ECG** **NON-INVASIVE**

Also called an **electrocardiogram**

Small sticky patches called electrodes are put onto your chest, and sometimes your arms and legs. These are connected by wires to an ECG recording machine which records the rate, rhythm and electrical activity of your heart.

**Signal-averaged ECG** **NON-INVASIVE**

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.

**Exercise test** **NON-INVASIVE**

Also called an **stress test**

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. This test is particularly useful in detecting some of the features that are characteristic of LQTS or CPVT.

**Cardiopulmonary exercise test** **NON-INVASIVE**

This test analyses the efficiency of your heart muscle by measuring the amounts of oxygen your body uses during exercise. You will be asked to breathe into special equipment while you're exercising.

**Holter monitoring** **NON-INVASIVE**

Also called **ambulatory ECG**

This test involves using a small digital device that you wear on a belt round your waist. Four or six ECG leads from the device are taped to your chest. The device records the electrical activity of your heart for 24 to 48 hours, or for up to seven days. The doctor can then analyse the electrical activity of your heart to find out if you have any arrhythmias.

**Cardiomemo and cardiac event recorders** **NON-INVASIVE**

These are more sophisticated versions of the Holter monitoring test described above. Whenever you have an episode of symptoms, you can activate the device to record your heart's rhythm. 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.

**Implantable loop recorder**

Also called an **ILR**

An implantable loop recorder (or ILR for short) can be used when it's difficult to assess or record a symptom because it only happens infrequently. The device, which is the size of a packet of chewing gum or smaller, is placed under the skin around your left shoulder under local anaesthetic. You'll need to go into hospital as a day case to have this done. The device monitors your heart's rhythm and can record any abnormal events.

**Provocation test**

Also called **ajmaline, flecainide or adrenaline tests**

You may be asked to have an ajmaline or flecainide test if your doctor suspects Brugada syndrome or an adrenaline test if your doctor suspects CPVT or LQTS. During these tests, you'll have an ECG whilst being given an injection of ajmaline or flecainide (antiarrhythmic medicines) or adrenaline. These medicines can unmask ECG changes associated with some ion channelopathies.

**Echocardiogram** **NON-INVASIVE**

Also called an **echo**

This test uses ultrasound waves to look at the structures of your heart. It is similar to the ultrasound scan used in pregnancy. An echocardiogram also gives information on your heart's function and pumping action. It can be used to detect structural heart disease such as cardiomyopathy and mitral valve prolapse.

**Cardiac magnetic resonance scan** **NON-INVASIVE**

Also called an **MRI scan**

This is a special kind of scan used to examine the structure of your heart and the nature of its muscle. At the start of the test you may need to have a special dye (contrast medium) injected into a vein in your arm. It may be useful for detecting heart muscle changes that may be 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).

A **coronary angiogram** involves having a local anaesthetic in your wrist or groin, where a catheter (a thin flexible tube) will be passed into your artery, directed through your blood vessels and into your heart. A special dye will then be passed through the catheter and a series of X-rays taken. This can show up any narrowed areas or blockages in your arteries that could be caused by coronary heart disease.

An **EPS (electrophysiological study)** uses the same technique to place electrical leads inside your heart. These leads analyse your heart's electrical activity and can help trigger an arrhythmia under safe controlled conditions. This test may be useful in diagnosing PCCD, and for deciding what treatment to give people with Brugada syndrome.

### **Tilt-table test**

If you get episodes of dizziness or blackouts, your doctor may recommend that you have a tilt-table test to find out if this is caused by one of the ion channelopathies mentioned in this booklet.

This test can bring on your symptoms under controlled and safe conditions to help your doctor make a diagnosis. You will lie flat on a special table, while your blood pressure and ECG are monitored. The head of the table is then raised to an angle of 60 to 90 degrees and monitoring is continued. If nothing happens, a medicine called GTN is sprayed under your tongue. This will make your heart rate a little faster and your blood pressure a little lower to help reproduce your symptoms. You'll continue to be monitored and after another 10 to 15 minutes, the table will be returned to the flat position. The whole test takes around 45 minutes.

### **Genetic testing**

In most of the ion channelopathies and structural heart diseases, mutations of specific genes have been detected and are thought to cause a specific disease. Identifying one of these gene mutations can help diagnose an ion channelopathy or structural disease. However, as we don't know all of the genes and gene mutations involved in each condition, genetic testing does not always help.

Ongoing research is helping to identify new gene mutations that cause inherited heart conditions. By identifying these gene mutations, more people can be diagnosed by genetic testing and treated for their condition. If a genetic test is not currently available, some clinics may offer to store a small blood sample in case this research leads to a genetic test being available in the future.

Genetic testing is not usually offered straight away, as your healthcare team will need to collect information from your other tests first. This helps them know which condition to test for using a genetic test.

There are many implications of having a genetic test. Your healthcare team will talk to you about the benefits and risks of having the test. They will give you support and help you understand the implications of the results for both you and your family.

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

### **Molecular autopsy**

After an unexpected sudden death, a post-mortem or autopsy may be carried out by a pathologist to determine the cause of death (see page 10). After the autopsy, the pathologist may keep a piece of tissue or amount of blood suitable for genetic testing. It may be possible to test this sample for gene mutations in the genes that cause inherited heart conditions. This is known as molecular autopsy and it may help in diagnosing the cause of death and/or inform the genetic testing of family members at risk.



**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 your heart by certain nerves (the left cervical ganglia).

**Coroner**

An official who holds inquests into violent, sudden, or suspicious deaths.

**D****Defibrillator**

A device that can be 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 early (premature) or extra heartbeat. The majority of people who have ectopic beats are unaware of them.

**G****Gene**

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

**H****Heart block**

When the electrical impulses sent by the atria to the ventricles are delayed or are blocked.

**I****Implantable cardioverter defibrillator (ICD)**

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

**Ion**

A chemical substance (such as sodium or potassium) that carries an electrical charge and forms the basis of the movement of electricity through your heart muscle.

**Ion channel**

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

**Ion channelopathy**

A condition caused when ion channels in the cells of your heart do not work properly. They can affect the electrical functioning of your heart and may be life-threatening if not treated.

**M****Mitral valve**

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

**Mitral valve prolapse**

When a mitral valve is floppy and may leak.

**Murmur**

The sound of the turbulent flow of blood in the heart, which may be caused by an underlying problem such as a heart valve defect. A murmur can be heard through a stethoscope.

**Mutation**

A mutation or 'mistake' in the DNA code that causes its eventual product (usually a protein) to function abnormally, which in turn is responsible for a disease.

**P****Pacemaker**

An electronic device which takes over the role of your heart's natural pacemaker and regulates the rhythm of your heartbeat. It is usually implanted just under your 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 to determine the cause of death.

**S****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. Some deaths will be unwitnessed or occur during sleep, while others occur during or immediately after exercise (exercise-related sudden death).

**Syndrome**

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

**T****Tachycardia**

A fast heart rate.

**V****Ventricles**

The two bottom chambers of your heart.

**Ventricular**

From, or belonging to, the ventricle.

## MORE INFORMATION

For information on your nearest clinic for inherited heart conditions

**BHF Genetic Information Service (GIS)**

Greater London House  
180 Hampstead Road  
London NW1 7AW

**Phone:** 0300 456 8383

**Website:** [bhf.org.uk](http://bhf.org.uk)

The BHF GIS can provide you with information and support if someone in your family has been diagnosed with, or has died from, a suspected inherited heart condition. This includes supporting you and your family by helping you get an expert assessment in a specialist clinic that deals with inherited heart conditions.

To find out more about our work on inherited heart conditions and the research we fund, visit [bhf.org.uk/research](http://bhf.org.uk/research)

**BHF publications**

The following booklets provide more information on inherited heart conditions:

**Life with Inherited Abnormal Heart Rhythms (M111B)****Life with Dilated Cardiomyopathy (M111D)****Life with Hypertrophic Cardiomyopathy (M111C)****Life with Arrhythmogenic Right Ventricular Cardiomyopathy (M111E)**

For anyone who has lost a loved one to a heart condition, see our booklet [Losing someone to heart disease \(G419\)](#). It aims to help you understand how grief and bereavement might affect you and offers help and support in coping with your loss.

Visit our website [bhf.org.uk](http://bhf.org.uk) for more information.

To order any of these booklets:

- call the BHF Orderline on 0300 200 2222, or
- email [orderline@bhf.org.uk](mailto:orderline@bhf.org.uk), or
- visit [bhf.org.uk/publications](http://bhf.org.uk/publications)

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

## FOR MORE SUPPORT AND INFORMATION

### Cardiac Risk in the Young - CRY

1140B The Axis Centre  
Cleeve Road  
Leatherhead KT22 7RD

**Phone:** 01737 363222

**Email:** cry@c-r-y.org.uk

#### Websites:

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

### Cardiac Risk in the Young myheart Network

CRY's myheart network offers help, support and information to young people who are coping with a diagnosis of a heart condition.

**Website:** www.myheart.org.uk

Contact details as above.

## FOR MORE INFORMATION ON CARDIOMYOPATHY

### Cardiomyopathy UK

Unit 10 Chiltern Court  
Asheridge Road  
Chesham  
Bucks HP5 2PX

**Freephone Helpline:** 0800 018 1024  
(Monday to Friday, 8.30am to 4.30pm)

**Website:** www.cardiomyopathy.org

**Email:** info@cardiomyopathy.org

Cardiomyopathy UK is a registered charity that helps people who are affected by 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 British Heart Foundation, please call our donation hotline on **0300 330 3322**, visit **bhf.org.uk/donate**, or post it to us at the address below. Thank you for supporting our fight.

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

### Have your say

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

### British Heart Foundation

Greater London House  
180 Hampstead Road  
London NW1 7AW  
**bhf.org.uk**

### Heart Helpline

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

### Genetic Information Service

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

## About Cardiac Risk in the Young

Cardiac Risk in the Young is a charity that:

- 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 support 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.
- Funds fast track services at the CRY Centres for Sports Cardiology, Cardiac Pathology and Inherited Cardiovascular Conditions at leading London hospitals.

### Cardiac Risk in the Young

1140B The Axis Centre  
Cleeve Road  
Leatherhead KT22 7RD  
**www.c-r-y.org.uk**



**British Heart  
Foundation**



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

Cardiac Risk in the Young supports young people diagnosed with potentially life-threatening cardiac conditions. We promote and develop heart screening programmes and fund medical research to help reduce the risk of young sudden death.

But so many people still need our help.

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