

Cardiac Conditions in the Young:

FROM ARVC TO WPW

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Produced by Cardiac Risk in the Young

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Contents

Introduction	1
How the heart works and how it can cause a blackout or cardiac arrest	2
What is a Sudden Cardiac Death (SCD) & Sudden Arrhythmic Death Syndrome (SADS)? ..	4
Sudden Cardiac Death (SCD)	4
Sudden Arrhythmic Death Syndrome (SADS)	4
How does SADS relate to the cardiac channelopathies/arrhythmia syndromes?.....	4
Ion Channelopathies	5
Long QT Syndrome (LQTS)	5
Brugada Syndrome	8
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	10
Progressive Cardiac Conduction Defect (PCCD)	11
Short QT Syndrome (SQTS)	12
Early Repolarisation Syndrome	12
Sodium Channel Disease	13
Familial Atrial Fibrillation	13
Cardiomyopathies	13
Hypertrophic Cardiomyopathy (HCM)	13
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	16
Dilated Cardiomyopathy (DCM)	17
Other cardiac conditions that can affect young people	20
Wolff-Parkinson-White Syndrome (WPW)	20
Marfan Syndrome	21
Myocarditis	22
Coronary Artery Anomalies	23
Premature Coronary Artery Disease and Familial Hypercholesterolemia	24
Tests	25
Medical and family history	25
Medical examination	25
Electrocardiogram (ECG)	26
Signal averaged ECG	26
Echocardiogram	27
Exercise test	27
Cardiopulmonary exercise test	28
Holter	29
Cardiomemo and event recorder	29
Implantable Loop Recorder (ILR)	29
Provocation tests (ajmaline, flecainide, adrenaline and adenosine tests)	30
Cardiac magnetic resonance/magnetic resonance imaging (CMR/MRI) scan	30
Computed tomography (CT)	30
Other tests	31
Coronary angiography	31
Electrophysiological (EP) study	31
Ablation	31
Cardiac biopsy	31
Tilt-table testing	31
Genetic testing	32
General Lifestyle Advice	33
Exercise	33
Drugs to avoid	33
Low potassium in the blood (hypokalaemia)	34
Drugs which people with long QT syndrome should avoid.....	35
Drugs which people with Brugada syndrome should avoid.....	36
The future.....	37
Technical terms.....	37
Index.....	46

Introduction

You may be reading this booklet because you or a relative of yours has been referred to a cardiologist (a heart doctor) after experiencing symptoms. These may include chest pain, palpitations (a fast, irregular heart beat) or blackouts. Alternatively, you may not have had any symptoms but either routine tests have suggested that you should see a cardiologist or a relative may have been told that they carry a genetic condition belonging to one of a number of groups of cardiac conditions, including cardiomyopathies and cardiac ion channelopathies (also known as arrhythmia syndromes). This may have caused no problems at all or may have been responsible for a blackout or even a sudden death in the family. Some of these heart conditions can be passed on in families. Your doctor may have suggested that you should have some tests to find out if you have inherited a condition.

This booklet:

- explains how the heart works and how blackouts and cardiac arrest may be caused
- explains what the arrhythmia syndromes or channelopathies are
- explains what the cardiomyopathies are
- explains what Wolff-Parkinson-White syndrome, Marfan syndrome, myocarditis, coronary artery anomalies and coronary heart disease are
- explains why it is important that blood relatives of a person with such a cardiac condition 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 these heart conditions

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 pages 37-45.

We hope that this booklet will help you and your family understand more about these conditions. If you need further help or information please contact CRY (see back cover for contact details).

How the heart works and how it can cause a blackout or cardiac arrest

In order to understand why blackouts and cardiac arrests 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 sinoatrial node (SA node), which is in the top chamber. (See the diagram on the right.) 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 atrioventricular node (AV 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 9 (figure 3).

The mechanism of a blackout or cardiac arrest due to a channelopathy 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. If the heart's rhythm goes back to normal by itself the loss of blood pressure is only temporary and the person experiences a blackout or near-blackout. If this problem does not go back to normal this is known as a cardiac arrest and after about two minutes, and if no-one is available to begin resuscitation, the brain and heart become significantly damaged and death may follow.

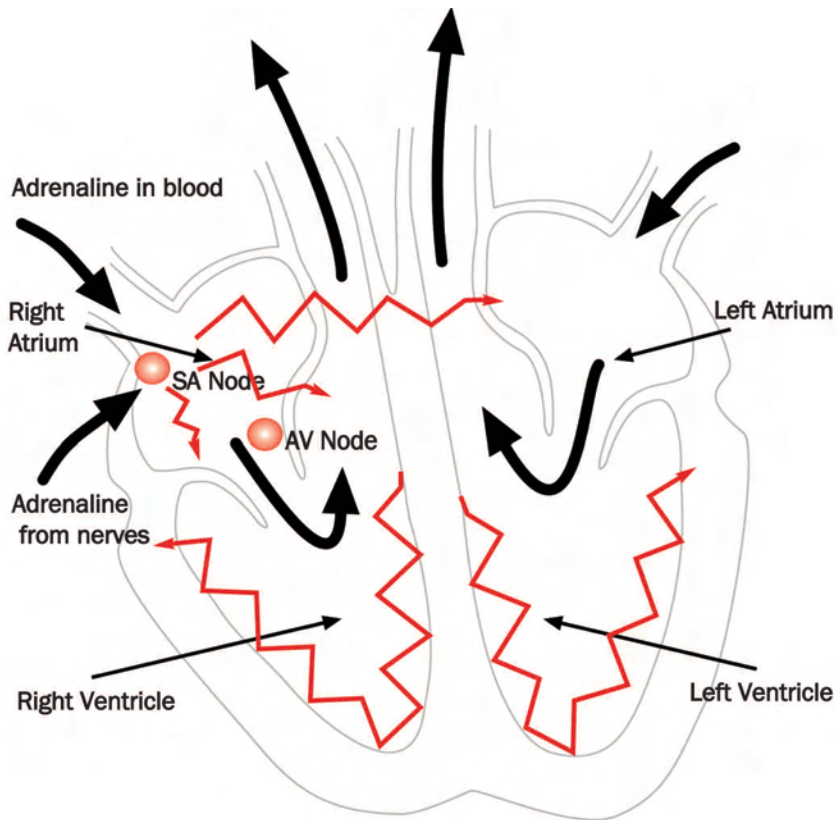


Figure 1: How the heart functions electrically

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 is a Sudden Cardiac Death (SCD) and Sudden Arrhythmic Death Syndrome (SADS)?

Sudden Cardiac Death (SCD)

Sudden death is the term used to describe an unexpected, spontaneous death. Most sudden deaths are due to a heart condition and are then called **sudden cardiac death (SCD)**. Sudden cardiac death can be a complication of some heart conditions; the risk of this occurring can be significantly reduced through lifestyle modifications, medication or surgery.

Sudden cardiac death in older people is usually due to **coronary artery disease** (blocked arteries in the heart), whereas sudden cardiac death in young people (35 years of age or less) is usually from inherited heart conditions that run in families.

Sudden Arrhythmic Death Syndrome (SADS)

In about 1 in every 20 cases of sudden cardiac death and up to 1 in 5 of young sudden cardiac deaths, no definite cause of death can be found, even after drugs have been excluded and an expert cardiac **pathologist** has examined the heart for any abnormalities of its structure. This is then called **sudden arrhythmic death syndrome (SADS)**. It is thought that cot death (sudden infant death syndrome, or SIDS) may be partly due to the same causes responsible for SADS. The conditions responsible for SADS cause a cardiac arrest by bringing on a **ventricular arrhythmia**, even though the person has no disease affecting the structure of the heart.

How does SADS relate to the cardiac channelopathies/arrhythmia syndromes?

The **cardiac channelopathies/arrhythmia syndromes** are a group of relatively rare diseases that affect the electrical functioning of the heart without affecting the heart's structure. They are often the cause of a SADS death. There are several different types of ion channelopathies, including:

- Long QT syndrome (LQTS)
- Brugada syndrome
- PVT (catecholaminergic polymorphic ventricular tachycardia)
- PCCD (Progressive Cardiac Conduction Defect)
- Short QT syndrome (SQTS)
- Early repolarisation syndrome
- Sodium channel disease
- Familial atrial fibrillation

Less frequently, SADS can be caused by other cardiac abnormalities, such as extra electrical pathways or even subtle heart muscle disease (**cardiomyopathies**).

Sudden cardiac death is a rare complication of some cardiac conditions; the risk of this occurring can be significantly reduced through lifestyle modifications, medication or surgery. If thoughts about this are worrying you, please come to a myheart meeting where you can discuss your concerns with our counsellor and a consultant cardiologist. They will be able to give you support and answers to your questions.

Ion Channelopathies

Cardiac ion channelopathies (also sometimes referred to as **arrhythmia syndromes**) affect the electrical functioning of the heart without affecting the heart's structure. They are a group of 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 person. (If they occur for the first time they are described as sporadic or 'de novo', meaning 'from new'.)

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, potassium or calcium - that carries an electrical charge and forms the basis of the movement of electricity through the heart muscle. Each heart muscle cell is surrounded by a membrane that separates the inside from the outside of the cell. An ion channel is the route (the **gate**) 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 may cause blackouts or cardiac arrest.

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 2,000 people. In 70% of people with LQTS, gene testing can identify the ion channels involved. 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 the LQTS diagram on page 7.) This results in an electrical disturbance in the cells of the heart called **prolonged repolarisation**. This can be seen on an **ECG** recording (we describe this test on page 26) as a lengthening of the time period, known as the **QT interval**. (We show this in the diagram on page 9 – figure 3.) This is where the name long QT syndrome comes from.

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 symptoms 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.

Are there any physical signs?

There are usually no physical signs of LQTS. However, certain rare forms of LQTS may be associated with muscle weakness, minor abnormalities of the skull, chin, fingers and toes or reduced hearing.

How is it diagnosed?

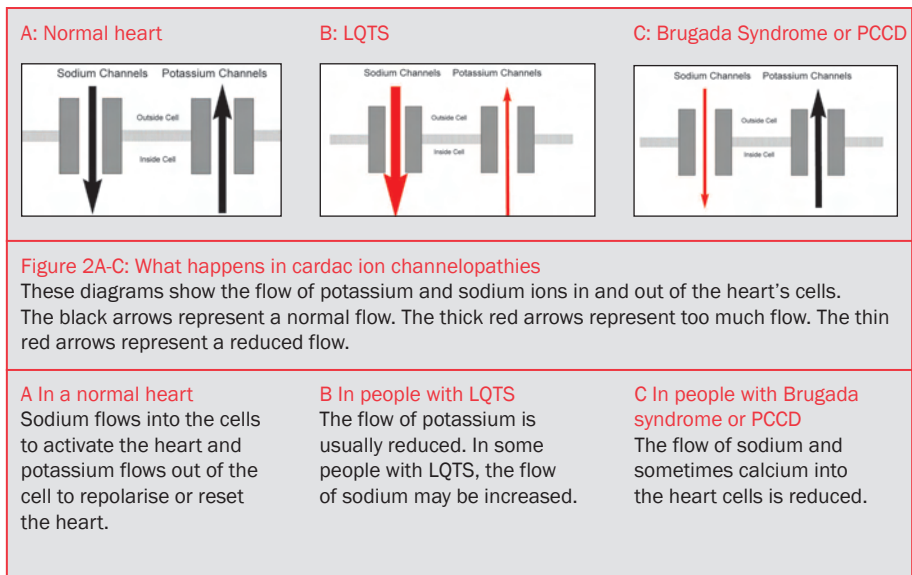
Diagnosis involves having an ECG. Sometimes it is possible to tell which ion channel has been affected just by looking at the ECG recording. Unfortunately, in a proportion of people who might be carriers of LQTS, the ECG may not show any sign of the condition. Repeated ECGs, **exercise tests** and **24-48 hour tape** monitoring may be needed before the diagnosis is established. (We describe all these tests on pages 25-32). More recently, some doctors have used slow injections of the hormone adrenaline (epinephrine) via a drip to try and improve the diagnosis of some potassium channel long QT syndrome. There is limited evidence, however, that this is any better than an exercise test.

Genetic testing can identify carriers of the LQTS gene (see page 32). 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 a mutation 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 because people with the same mutation can have no signs of it or be severely affected.

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 cardiac arrest. It is also important to avoid low blood potassium levels, known as **hypokalaemia** (see General Lifestyle Advice on page 33).



Other treatment options will vary depending on the severity of the condition. Those with one or more of the following features will likely need more intervention than those without:

- a previous cardiac arrest
- blackouts
- a very long QT interval on the ECG
- specific genetic forms of the condition
- young adult women (especially in the first year after pregnancy)
- boys before puberty

Drugs

The most commonly used drugs for LQTS 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 cardiac arrest. 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. 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 Implantable Cardiac Defibrillator (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 43, and for more on ICDs see page 41.)

Surgery

Another option for high-risk patients is to perform surgery to disrupt the nerves that release adrenaline and related chemicals into the heart. This is performed in only a very small number of people and is known as cervical sympathectomy and involves operating on the left side of the neck. (For more on this see page 39.)

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 the Brugada syndrome diagram on page 7.) 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. Other genes have been described as being involved in Brugada syndrome that produce calcium and potassium ion channels and proteins that interact with the sodium channel. They have, however, only been detected in a small number of carriers.

What are the symptoms?

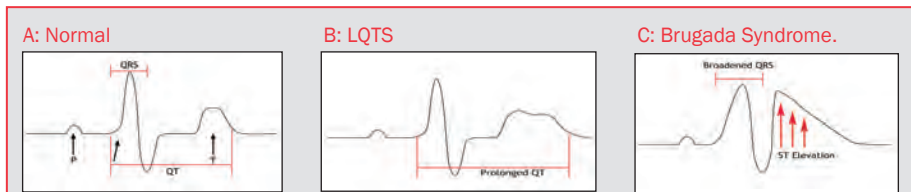
Most people with Brugada syndrome may have no symptoms at all. In others, the most common symptoms are blackouts. Some people may notice **palpitations** due to **ectopic** (extra) beats.

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 come and go, or they may not show at all. Sometimes the presence of a fever or certain medications (see page 36) can bring out the ECG changes and this may be a period when there is higher risk of blackouts or cardiac arrest. 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 30). 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.



Figures 3A-C: A simplified version of the ECG as it would appear on the ECG in the following circumstances: A: Normal B: LQTS C: Brugada syndrome.

The 'P' wave reflects the electrical activity of the SA node 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), see A. 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.

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

All carriers of Brugada syndrome should avoid certain medications (see page 36) that might worsen their condition. They are also advised to avoid low blood potassium levels, known as **hypokalaemia** (see General Lifestyle Advice on page 33), and should treat all fevers with medications that reduce their body temperature, such as paracetamol or ibuprofen. If their fever remains high they should attend hospital for an ECG or monitoring as required. It is standard practice for people who have suffered a cardiac arrest or a blackout to have an **ICD** fitted as this is a very successful form of protection. (For more information on ICDs see page 41.) The tablet quinidine has been used in some patients with severe disease and an ICD already in place but its exact role remains under investigation.

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 **electrophysiological (EP) study** may help to identify those people who do or do not need an ICD, although there is great controversy about its true value. Research has suggested that most people with normal ECGs and no symptoms should be safe without any treatment. It is unusual for children to be at high risk.

Catecholaminergic polymorphic ventricular tachycardia (CPVT)

CPVT is a rare condition that 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 calcium 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 **arrhythmias**.

What are the symptoms?

Some people with CPVT have no symptoms at all. Others may have **palpitations** (a fast or irregular heart beat), or blackouts while exerting themselves or suffering emotional stress. The condition may be found in most age groups but appears to be most severe in children and adolescents and in males.

Are there any physical signs?

There are no physical signs.

How is it diagnosed?

The diagnosis is usually made after the recording of arrhythmias that are characteristic of CPVT, while the person is exercising. The **exercise ECG** is therefore useful although some doctors use adrenaline testing (as in LQTS) as well. **Genetic**

testing is of use as 1 in 2 people with the condition carry a ryanodine receptor mutation and are more likely to have blackouts or a cardiac arrest. When a member of a family has been found to carry a mutation, testing is then useful in detecting carriers who have not yet shown any signs of the condition.

Treatment and advice

Your doctor will advise you to take beta-blockers (a type of drug), to restrict the amount of exercise you do and avoid medications that stimulate the heart. This combination can greatly improve the outlook for people with CPVT. Up to 1 in every 3 people with the condition, however, may also need to have an **ICD** fitted, particularly if they have blackouts while taking beta-blockers. (For more on ICDs see page 41). Occasionally, some patients will be given additional tablets (flecainide) to help reduce the amount of arrhythmia.

Surgery

Recently, some groups have used the **cervical sympathectomy** (already used in LQTS) to treat CPVT with some success.

Progressive Cardiac Conduction Defect (PCCD)

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 the channel similar to those found in people with Brugada syndrome (see the Brugada syndrome diagram on page 7- figure 2C).

What are the symptoms?

Dizziness and blackouts are the usual symptoms and cardiac arrest may also occur.

Are there any physical signs?

There are no physical signs usually, except if there is heart block when the doctor may feel a slow pulse.

How is it diagnosed?

The ECG abnormalities may be detected either on a standard **ECG** or with **Holter monitoring**. An **electrophysiological (EP) study** may also help the doctor make a diagnosis. (We describe all these tests on pages 26-33.) 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 tachycardias from occurring, 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 43, and for more on ICDs see page 41.) Medication alone does not help.

Short QT syndrome (SQTS)

This rare condition is similar to but distinct from LQTS. As the name suggests, the **QT interval** in carriers is shorter than in normal people. This means that the heart takes a shorter time to **repolarise** or reset itself, making it prone to **ventricular arrhythmias**. There is also an increased risk of a less dangerous arrhythmia from the top chambers of the heart (the **atria**) called **atrial fibrillation**. This is an irregular and rapid heart rhythm that may go unnoticed or causes breathlessness and **palpitations**. As in LQTS, potassium channel genes are affected, but instead of allowing less potassium through they allow through too much too quickly.

What are the symptoms?

Palpitations, blackouts and cardiac arrest.

Are there any physical signs?

There are no physical signs usually, except if there is atrial fibrillation when an irregular rapid pulse may be felt by the doctor.

How is it diagnosed?

The **ECG** abnormalities are usually detected either on a standard ECG or a **24-hour Holter**. An **electrophysiological (EP) study** may also help the doctor make a diagnosis. (We describe all these tests on pages 26-33.) Genetic testing may find a potassium channel mutation in affected members of a family that may then also be found in other relatives.

Treatment and advice

If you have SQTS it is likely that an **ICD** will need to be fitted in order to treat dangerous arrhythmias. Quinidine has been used in some patients but it is unclear whether it is safe to rely on medication alone. Tablets may also be used to treat atrial fibrillation.

Early repolarisation syndrome

Idiopathic ventricular fibrillation (IVF) describes the group of conditions responsible for life-threatening, rapid rhythm disturbances without any signs of the heart diseases we have described above. In some of these patients changes on their ECG, known as early repolarisation, have been seen. This has become known as **early**

repolarisation syndrome and in a few cases **mutations** of potassium and calcium ion channel genes have been found. Early repolarisation can, however, be seen on the **ECGs** of many normal healthy people. The diagnosis and treatment of this condition is therefore still unclear, but an **ICD** is needed in patients who have suffered a cardiac arrest.

Sodium channel disease

There are rare and specific sodium channel mutations that can cause long QT syndrome, Brugada syndrome and/or PCCD in the same family. They can be diagnosed and treated as described above and can be identified by genetic testing.

Familial atrial fibrillation

Atrial fibrillation is the most common abnormality of the heart's rhythm. It tends to affect people as they get older and is often present with other conditions such as high blood pressure, heart disease and diabetes. The abnormality in rhythm arises from the top chambers of the heart (the **atria**) and is irregular and rapid. It may go unnoticed or cause breathlessness and **palpitations**. If left untreated, atrial fibrillation can cause weakness of the heart muscle (heart failure) and is associated with an increased risk of stroke. In a small proportion of people with atrial fibrillation the condition is clearly inherited and occurs at a young age. **Mutations** of potassium ion channel genes have been found in these families that allow through too much potassium in a similar way to **SQTS**. Treatment is no different from the usual treatment of atrial fibrillation. This involves the control of the rate of the heart (using drugs), the control of the rhythm of the heart (by giving the heart an electric shock, this is called **cardioversion**, or by using drugs), or anticoagulation (where a drug reduces the chances of a blood clot forming).

Cardiomyopathies

Cardiomyopathies affect the muscle of the heart. They are a group of relatively rare genetic conditions that are usually caused by abnormalities of the **DNA**, known as **mutations**. They are usually inherited from parents in such a way, that if a parent has a **mutation** then each child has a 50% chance of inheriting the disease. They can occur for the first time in a person and are described as sporadic or 'de novo', meaning 'from new'. Below we describe the different types of cardiomyopathy, the tests needed to diagnose them and the treatment that may be needed for each one.

Hypertrophic Cardiomyopathy (HCM)

Hypertrophic cardiomyopathy (HCM) is a condition where the heart muscle becomes thickened. Although HCM is a relatively rare heart disease, it is the commonest of the cardiomyopathies, affecting 1 in every 500 people. Traditionally, the term HCM was used for disease caused by abnormalities in **genes** which make the proteins

responsible for contraction of the heart, 'the sarcomere'. More recently, the definition of HCM has been broadened to include a number of other conditions that result in thickened heart muscle.

It is a disease that can affect both men and women of any ethnic background. Excessive muscle thickening tends to develop in puberty or early adulthood or can sometimes develop very late in life. Rarely, it may even begin before birth when the baby's heart is developing and cause problems in childhood. In a healthy adult heart, the muscle fibres are arranged in an organised fashion and the thickness of the heart's wall remains normal. In HCM, however, the heart muscle becomes excessively thick and the fibres are arranged haphazardly, making the heart vulnerable to life-threatening heart rhythms (**ventricular fibrillation/ventricular tachycardia**). The thickening may also reduce the efficiency of the heart's pumping action or even lead to the blockage or 'obstruction' of blood leaving the main pumping chamber of the heart. This can result in the symptoms described below. Diagnosing HCM may pose a significant challenge, since the heart muscle may also thicken in individuals who have high blood pressure or who participate in prolonged athletic training.

What are the symptoms?

If you have HCM you may never experience any symptoms. If you do suffer symptoms they can vary from person to person and may begin in infancy, childhood, middle or elderly life. No particular symptom or complaint is unique to HCM sufferers.

The most common symptoms are:

- shortness of breath, usually brought on by physical exertion
- chest pains, usually brought on by physical exertion
- palpitations, a rapid and/or irregular heart beat
- light-headedness, blackouts

If you suffer from any of these symptoms it does not mean that you necessarily have HCM, but if you visit your GP he/she may suggest that you undertake some tests or may refer you to a **cardiologist** (a heart specialist).

How is it diagnosed?

Diagnosis involves you having an **ECG** and an ultrasound scan of the heart, known as an **echocardiogram** or **ECHO** (We describe these tests on pages 26-27). Most people with HCM have an abnormal ECG and often the ECG may become abnormal long before thickening of the heart can be seen on an ECHO. If we suspect HCM in you, an **exercise ECG** and a **Holter monitor** will be required. In some cases, further imaging of the heart may be necessary using a **magnetic resonance imaging (MRI) scan**. (We describe these tests on pages 27-30).

Genetic testing can identify carriers of the HCM gene (see page 32). Unfortunately, this form of testing is limited at the moment, as 3 in every 10 people who are known to have HCM do not have mutations of the genes known to be associated with HCM. 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 because people with the same mutation can have different effects. However, if a well-known mutation is identified in you, it can be used to confirm the diagnosis in you or be offered to your family to screen for the condition.

Treatment and advice

There is no cure at present for hypertrophic cardiomyopathy. Treatment is aimed at preventing complications and improving symptoms.

If your tests prove positive your specialist will advise you on your lifestyle. You will probably be advised not to participate in competitive sport and strenuous activities.

For many people the condition should not significantly interfere with their lifestyle. Drugs are given when a patient has symptoms. A variety of drugs are used in the treatment of HCM and the choice will vary from patient to patient. However, if you have severe symptoms due to blockage or 'obstruction' of blood flow, and you do not improve on tablets, **surgical myectomy** or **alcohol septal ablation** may be suggested (these are described on pages 45 and 37). These can be successful in the relief of symptoms due to obstruction by removing or damaging a portion of the thickened muscle.

There are other forms of treatment, which are occasionally recommended.

- Electrical **cardioversion** may be used to treat atrial fibrillation (irregular heart beat), which is quite common (see page 38 and 40).
- A **pacemaker** may be fitted if your heart's normal electric signal fails (see page 43).
- An **Implantable Cardiac Defibrillator (ICD)** may be fitted for you if you have suffered a previous cardiac arrest or a dangerous ventricular arrhythmia (see page 45). An ICD will also be considered even if you have not had these symptoms but have other signs of risk during your assessment.

In a small number of patients the heart pump weakens eventually and symptoms of heart failure (breathlessness and ankle swelling) will require treatment which we describe in dilated cardiomyopathy (see page 17).

What should you do if you are diagnosed with HCM?

If your tests prove positive your specialist will advise you on your lifestyle and you will probably be advised not to participate in continuous strenuous activities e.g. competitive sports. Most of the time the condition should not significantly interfere with your lifestyle and can be controlled by drugs if necessary. It will be necessary for you to have at least annual check-ups. However, the severity of the disease varies from person to person and even if you have been diagnosed with HCM you may not necessarily present any symptoms and can live a normal life. Since the disease runs in families, all immediate blood relatives of affected patients have to be screened with an ECG and an ECHO.

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)

The estimated prevalence of arrhythmogenic right ventricular cardiomyopathy (ARVC), also known as **arrhythmogenic right ventricular dysplasia (ARVD)**, in the general population ranges from 1 in 1,000 to 1 in 5,000 depending on the definition of the condition. It was first recognised in the late 1970s. We think that even more information regarding ARVC will be available in the coming years, to improve our understanding of the condition. The disease affects men and women equally and has been recognised in people of diverse ethnic backgrounds. It tends to develop in early adulthood but can occasionally affect children.

ARVC is caused by a defect in the 'glue' that holds the muscle cells of the heart together, working as a unit. When stretched the 'glue' breaks down, the muscle cells separate and some die. The body then tries to repair this, resulting in replacement of the normal heart muscle cells by scar and fat tissue. This may only involve small areas of the right ventricle but may progress with time and may become more widespread and even involve the left ventricle. Some forms only affect the left ventricle.

What are the symptoms?

Often you may have no symptoms at all. You may feel **palpitations** (feeling the pulse racing or the heart pounding erratically or rapidly) which is the most common symptom. This may also be associated with light-headedness or a blackout. Unlike most cardiomyopathies, shortness of breath and chest pains are unusual symptoms and you may only experience these very late on in the condition, if at all.

How is it diagnosed?

The diagnosis can be extremely difficult and usually requires several tests undertaken by a specialist as there is no single 'diagnostic' test. In the first instance a detailed history and examination is required. Most of the tests are painless and non-invasive and include an **ECG, signal average ECG** and an ultrasound scan (**ECHO**) (we describe these tests on pages 26-27). The ECG can often pick up an abnormality but the signs on an ECHO can be very subtle in the early stages of the

condition. They are also often only seen in the right ventricle. Further imaging with a **magnetic resonance imaging (MRI) scan** may be required (see page 30). Other tests you may require include an **exercise ECG** and a **Holter monitor** (tape) to try to record any palpitations. In some specialist centres further invasive tests are performed to identify the electrical faults of the heart muscle associated with ARVC (**EP studies**) and to take a small sample (**biopsy**) of the heart muscle and examine it under the microscope (these are described on page 31). These investigations can still miss ARVC and can be associated with some risk to you.

Advances in genetics (**DNA**) means that in some hospitals, the condition may be diagnosed using a blood test. This test, however, is not available in every hospital, may take up to several months and it is not always positive in ARVC patients (we are able to identify a defective gene in 50% of patients with definite ARVC). It can be used to confirm the diagnosis or identify blood relatives who may carry the condition.

Treatment and advice

If your tests prove positive your specialist will advise you on lifestyle modifications. You will most likely be advised not to participate in competitive, strenuous activities. The majority of people with this condition are, however, asymptomatic for many years unless heart rhythm problems (**arrhythmia**) develop. Treatment aims to prevent or at least control these and tablets may be used to control the irregular heart rhythms. These may include beta-blockers, amiodarone or sotalol.

In people in whom medicine is unsuccessful in controlling rapid heart beats or where blackouts, near blackouts or even a cardiac arrest have occurred, an **implantable cardiac defibrillator (ICD)** may be fitted (we describe this on page 41). In some cases rhythm problem can be very troublesome and the only way to control them is to destroy the parts of the heart muscle causing them. This is known as **ablation** and can be performed at the time of an EP study. In a small number of individuals the heart pump weakens eventually and symptoms of heart failure (breathlessness and ankle swelling) will require treatment which we describe in dilated cardiomyopathy (see below).

It will be necessary for you to have at least annual check-ups which usually will include a repeat of the initial investigations. Since the disease runs in families, all immediate blood relatives of people with ARVC should be screened with at least an ECG and ECHO, as well as some of the additional tests we have described.

Dilated Cardiomyopathy (DCM)

In dilated cardiomyopathy (DCM) the main pumping chambers of the heart are enlarged (dilated) and contract poorly. This results in less blood being pumped around

the body which fails to meet the body's demand. This is known as heart failure. There can be a build up of fluid in the lungs and under the skin, which can cause breathlessness and swelling of the legs and abdomen.

We can find a specific cause for DCM in about half of patients. In the other half, however, the cause remains uncertain. The commonest causes of DCM in the western world include **coronary artery disease** ('furring' of arteries to the heart), which may lead to reduced blood flow to the heart muscle and a weak heart. Other common causes include longstanding high blood pressure, excessive alcohol intake and heart valve disease. Less common causes include viral infections, autoimmune diseases (i.e. the body's own defences mistakenly attack the heart muscle cells, resulting in damage to the heart muscle), deficiency of several vitamins and other rare conditions.

Pregnancy

There is a form of DCM, known as 'post-partum cardiomyopathy', which develops during late pregnancy and in the period shortly following childbirth. The cause is uncertain, but it is believed that the extra strain of pregnancy on the heart may trigger the development of the condition.

Genetics

In at least 40% of people with DCM in whom no other cause can be found, other family members can show signs of it and it has therefore been inherited. Currently, we can explain a quarter of inherited DCM by finding the underlying genetic problem (**mutation**). This does require testing for many different genes which is rarely done because of the expense. One gene, Lamin AC, is often tested for because it is found most commonly and can be associated with a higher risk of **arrhythmia**.

What are the symptoms?

Your symptoms can depend upon how severe the condition is, but you may suffer none at all yet still carry signs of DCM.

- Shortness of breath is common and can become worse with exertion. When severe, it can be present at rest or even when you lie flat in bed which may wake you up at night. You may feel more comfortable sitting-up in bed by adding extra pillows.
- Lack of energy and tiring easily.
- Ankle swelling, called oedema, which can worsen to affect the thighs, back and abdomen.
- Palpitations can occur and means that you are aware of your heartbeat due to an abnormal heart rhythm. The heart can either beat irregularly, too rapidly (tachycardia) or too slowly (bradycardia).
- Dizziness or blackouts.

How is it diagnosed?

Your doctor will take a detailed history and examine you. An ECG should be performed, followed by an ECHO. The ECHO will measure the size of your heart and how well it pumps. If there is any suspicion that you have DCM then other tests will often include an **exercise ECG**, either on a treadmill or a bicycle, or a 24-hour tape recording is also important to detect arrhythmias which may not display any symptoms. Finally, depending on each individual case, further investigations may be necessary.

Treatment and advice

Treatment aims to improve the symptoms of heart failure, prevent complications, particularly those arising from the development of arrhythmias, improve the heart function and prolong life.

Changes in your lifestyle may be important in the treatment of DCM. Your doctor may discuss the following things with you.

- Reducing all cardiovascular risk factors:
 - Stop smoking with the help of smoking cessation clinics.
 - To improve dietary habits in order to lose weight, reduce the cholesterol and the salt in the diet. An expert dietician's review can be very helpful.
 - Good control of the blood sugar if you are a diabetic.
 - To monitor and treat high blood pressure.
 - Regular exercise.
- Limiting alcohol intake to the minimum possible. We advise that no more than a small glass, preferably of red wine, should be consumed in a day.
- Limiting fluid intake according to your doctor or specialist nurse's advice.
- Daily weighings using a reliable scale. Sudden increases in weight may be a sign of fluid build-up and may act as a warning sign.

Medicines are the main treatment for heart failure. This includes water tablets (diuretics) to get rid of excess fluid, Angiotensin Converting Enzyme inhibitors (ACE-I), spironolactone and beta-blockers. Other drugs may be necessary should your symptoms persist. Please note that unless there is a reason for you not being able to take ACE-I and beta-blockers every patient with significant DCM should be on them.

If you still remain breathless or unwell, further treatment may be necessary. This may include the insertion of specialised **pacemakers** under the skin that help the heart beat more efficiently. This is known as **resynchronisation** therapy. If your heart pump function becomes badly affected, an ICD may also be offered to treat dangerous arrhythmias and prolong your life. The ICD may also be able to 'resynchronise' the heart. (Page 41 and 43 describe these procedures).

If you still do not respond to the above treatments you may be referred to a specialist centre for cardiac transplantation. Strict criteria apply to the selection of patients in order to assure a successful transplantation.

All immediate blood relatives of patients diagnosed with DCM of an unknown cause should be screened to exclude the condition.

Other cardiac conditions that can affect young people

Wolff-Parkinson-White Syndrome (WPW)

Wolff-Parkinson-White (WPW) results from an additional electrical connection between the upper chambers of the heart (**atria**) and the lower chambers of the heart (**ventricles**). This extra or 'accessory' pathway is seen in approximately 1 in every 300-500 people and sometimes it allows conduction of the electrical pulse at high speed, generating an electrical short circuit, which produces a rapid heart rate (**arrhythmia**) which can occasionally be dangerous. WPW is rarely inherited, however, and runs in families in less than 1% of cases.

What are the symptoms?

You may never experience symptoms. In fact, in the majority of people the extra pathway is completely silent. **Palpitations** are, however, the main symptom of WPW. You may find these untroubling, but they may cause chest pain, light-headedness and even blackouts. They may occur predictably or at any time and you may be able to control them. One way of doing this is by holding your breath as forcibly as possible. The palpitations may remain, however, until they can be stopped by an injection in an accident and emergency department.

How is it diagnosed?

WPW is diagnosed on an **ECG**. It is often found during a routine ECG check as part of medical insurance or cardiovascular screening or detected when testing for palpitations. Other tests may include an **ECHO**, an **exercise ECG** and a **24-hour Holter monitor** (tape).

If you are young, are a competitive athlete, have a particular job such as a pilot or a soldier or have had palpitations because of arrhythmia, more invasive tests (**EP studies**) may be recommended to assess the way the accessory pathway behaves and therefore the risk of significant arrhythmias (we describe this on page 31).

Treatment and advice

The ideal treatment if you have symptoms is to destroy the extra electrical pathway using **RF ablation** (this is described on page 44). This is done at the time of an EP

study by passing a wire into the heart and locating the pathway. This is destroyed by passing a high current through it. This takes approximately 1-2 hours and may require one night in hospital.

Tablets may be preferable to an ablation if you are asymptomatic or if an EP study shows that you have no risk for dangerous arrhythmias or if the accessory pathway is difficult or dangerous to treat. The pathway can also disappear as you get older.

Marfan Syndrome

Marfan syndrome affects many parts of the body, including the heart, blood vessels, skeleton, and eyes. 1 in 5,000 people in the United Kingdom have Marfan syndrome and both men and women can be affected.

People with Marfan syndrome produce abnormal 'connective tissue'. Connective tissue helps hold the body together, binding tendon to muscle and muscle to bone and is made of microscopic fibres. This tissue provides the stretchy strength of tendons and ligaments around joints and in blood vessel walls. In Marfan syndrome a fibre called fibrillin is made abnormally which makes weaker tissue. Marfan syndrome is inherited from parents in 75% of cases usually due to a **mutation** in the fibrillin gene. In the remaining 25% of cases it is caused by a new or 'de novo' mutation. The condition is inherited and, as such, children of a person with Marfan syndrome have a 50% chance of developing the condition.

What are the symptoms?

The severity and pattern of disease are variable in Marfan syndrome, even within families. Diagnosis requires a careful physical examination. You may be very tall and suffer from spine problems. Your arms, legs and fingers may be long and your arm span may even be longer than your height. The joints of the hands and wrist may also be very flexible. The roof of your mouth can be high and arched and your teeth may seem overcrowded. You may also have problems with your vision caused by problems with the lens of the eye.

One of the main concerns in Marfan syndrome is that the main blood vessel in the body, the **aorta**, can become stretched, enlarged and may tear (aortic dissection) or burst (aortic rupture). This can be dangerous to you. You can also develop leaking heart valves that can be heard by a doctor as a heart **murmur**.

Not everyone with Marfan syndrome has all of these problems and a **genetic test** can provide a definite diagnosis in at least 90% of cases.

Treatment and advice

The complications of Marfan syndrome can be corrected. The importance of

diagnosis is to allow careful observation so that the life-threatening risks of aortic dissection or rupture can be avoided. Enlargement of the aorta usually occurs very slowly, so an **ECHO** once a year will identify patients with this problem. Tablets called beta-blockers can slow the enlargement down. If dangerous enlargement occurs then plans can be made for an operation to replace that part of the aorta.

Enlargement of the aorta can also cause the **aortic valve** to become leaky (aortic regurgitation). The function of the aortic valve is to prevent blood from flowing back into the heart once it has been pumped out. Leakage of blood back into the heart can cause breathlessness. In these situations the aortic valve can be replaced at the same time as the aorta is repaired.

Leaking of the **mitral valve** can also be caused by Marfan syndrome. Constant leaking of the mitral valve causes the main pumping chamber to work harder and may again cause breathlessness sometimes with **palpitations**. In this situation the mitral valve can be repaired or replaced during an operation.

Eye, mouth and skeleton problems are not life-threatening and each problem can be corrected if necessary. There are only four rare complications requiring emergency care:

- **Tear of the aorta**
This may cause severe chest and/or back pain and even a collapse.
- **Collapse of lung**
This may occur during periods of exertion and presents with severe breathlessness. You may become blue and complain of chest pain.
- **Glaucoma**
The lens problem may produce a sudden increase of pressure in the eye and cause acute eye pain.
- **Dislocations**
Loose ligaments do not provide adequate support for joints, and can cause a joint to become separated, known as dislocation.

Myocarditis

Myocarditis refers to inflammation of the heart muscle. It is most often due to a virus. Other causes include drug abuse and conditions such as systemic lupus erythematosus (SLE). Myocarditis due to a virus is relatively common, but most cases are very mild and are never seen by a doctor. However, some cases are severe and can lead to considerable damage to the heart pump, resulting in severe heart failure and even dangerous **arrhythmia**.

What are the symptoms?

You may feel feverish and have generalised aches and pains as with any other viral illness. When the heart is severely affected you may feel tired and breathless. Chest

pain and **palpitations** are relatively common under these circumstances. Apart from a rapid heart rate, the doctor may not find any other abnormalities if you have only mild myocarditis. However, if it is severe there may be evidence of heart failure and/or arrhythmia.

How is it diagnosed?

The presence of a flu-like illness and fever may raise the suspicion of myocarditis in someone with symptoms. Simple blood tests may, however, show signs of inflammation and heart damage. The **ECG** may show changes which can come and go, are non-specific and can occur in other heart diseases. An **ECHO** can show an enlarged heart, which pumps poorly. The diagnosis can be confirmed by a **biopsy** (a small sample) taken from the heart. An **MRI scan** can be useful for diagnosing inflamed heart muscle.

In very mild cases, both the ECG and the ECHO may be normal, so when symptoms of a flu-like illness are present you should be cautious and not take part in any exercise or sports until you have recovered.

Treatment and advice

You should rest until symptoms settle. Patients with heart failure and arrhythmia must stay in hospital for treatment of these (see the section on dilated cardiomyopathy). Steroids may be used in specific forms of myocarditis.

You should not take part in any exercise or sports until all symptoms have settled and the ECG and ECHO are back to normal. Over three quarters of people will improve within two weeks without any complications. Around 1 in 10 patients may then develop dilated cardiomyopathy and require lifelong treatment for heart failure (see the section on dilated cardiomyopathy).

Coronary Artery Anomalies

The heart muscle requires a regular supply of oxygen and food. This is supplied by blood carried by two blood vessels known as the left and right coronary arteries which come from the left and right side of the body's main artery, the **aorta**. In 1 in 100 people both coronary arteries come from the same side of the aorta. This is called an 'anomalous coronary origin' or 'coronary artery anomaly'. The unusual origin of the artery means that the artery must take a different route to supply the heart muscle. In the majority of people this causes no problems. However, in a small number the abnormal route can cause the anomalous coronary artery to be squashed or kinked during exercise. If the blood supply to the heart is disturbed then it can cause chest pains or the dangerous **arrhythmias**. The cause of coronary artery anomalies is not known; however, in most cases they do not appear to be inherited.

What are the symptoms?

If you have a dangerous coronary artery anomaly you may experience warning symptoms, including chest discomfort or blackouts that usually happen during exercise.

How is it diagnosed?

Looking for coronary artery anomalies can be difficult because an **ECG** and even an **exercise ECG** are usually normal, even if you are at high risk. The artery origin can be seen using an **ECHO**. If both arteries are not clearly seen then a more detailed heart scan, such as cardiac **MRI** or **computed tomography** (CT), can be used.

Treatment and advice

If you have a low-risk anomaly you may be treated with beta-blockers and advised to avoid high levels of physical exertion. If high-risk anomalies are found then surgery to reconnect the artery to the right side of the aorta can be lifesaving.

Premature Coronary Artery Disease and Familial Hypercholesterolemia

The heart muscle requires a regular supply of oxygen and food. This is supplied by blood carried by a network of blood vessels known as the coronary arteries. The coronary arteries can become narrowed by 'furring' of the artery wall with cholesterol, called atherosclerotic plaques. Blood clots can form on these plaques and block the artery, causing chest pains. The heart muscle supplied by the blocked artery is starved of oxygen and dies soon afterwards, known as a heart attack. This can also cause dangerous **arrhythmias**. The build-up of plaques in the coronary arteries increases with age and most people who have significant narrowing are usually over 50 years of age.

There is a genetic risk for the development of coronary artery disease, but many factors are involved, including smoking, high blood pressure, diabetes and cholesterol levels. Some families, however, carry **gene mutations** that make them likely to have a very high cholesterol levels. This is known as **familial hypercholesterolemia** or **FH**. Families affected usually have members who have suffered from heart attacks, sometimes at very young ages.

What are the symptoms?

If you have narrowed arteries you may experience heavy or tight chest pain, called angina, or breathlessness, most commonly during exertion, as a warning. If you have FH you may develop fatty lumps in the skin around the eyes or in the muscle tendons, but, usually, the only way of telling is to have a blood test.

How is it diagnosed?

An **ECG** at rest will often be normal. An **exercise ECG** may show signs that the heart is being starved of oxygen (see pages 26-27 for a description of these tests). Depending

on how high your doctor thinks your risk of having narrowed coronary arteries is, a range of different tests may also be offered, such as an ECHO during exercise, a special **MRI scan** or a **CT scan** of the coronary arteries. If he or she thinks your risk is high then you may be offered **coronary angiography** to diagnose the condition (see page 31 for a description of this).

Treatment and advice

If you have coronary artery disease you should receive advice on your life-style, including stopping smoking, a healthy diet and regular exercise. If your cholesterol levels are high they can be treated with tablets, known as statins, to reduce the risk of further artery furring. Tablets can also treat symptoms of chest pain and reduce the chance of a heart attack. If your angiogram suggests a very tight narrowing in one of the coronary arteries then a catheter can be used to open up the narrowing by blowing up a balloon at the narrowing (known as angioplasty) and inserting a metal tube (stent) to keep the artery open. If your angiogram shows more severe furring, especially if both the right and the left coronary arteries are affected, then a bypass operation may be required to improve the blood supply to the heart.

Tests

Because the channelopathies, cardiomyopathies and Marfan's may be inherited, if you are a blood relative in the immediate family of someone who has a channelopathy or cardiomyopathy or has died of SADS, it is important that you are evaluated for signs of these diseases. Below, we explain what is involved in the evaluation and describe the tests you may need to have.

Medical and family history

It is vital that a clear history of the family and any carriers is established. For example, fits brought on by exercise can be due to an underlying channelopathy such as LQTS or CPVT or a cardiomyopathy such as HCM, or a sudden cardiac death during sleep may have been caused by an undiagnosed LQTS or Brugada syndrome. It is also important to find out about any medications and any potentially dangerous drugs that may interact with the channelopathies. 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 identify any disease of the heart's structure. For example, if there is a valve leakage this will cause a **murmur** that a doctor can hear through a **stethoscope**. HCM that causes a blockage to blood flow (obstruction) may also cause a murmur.

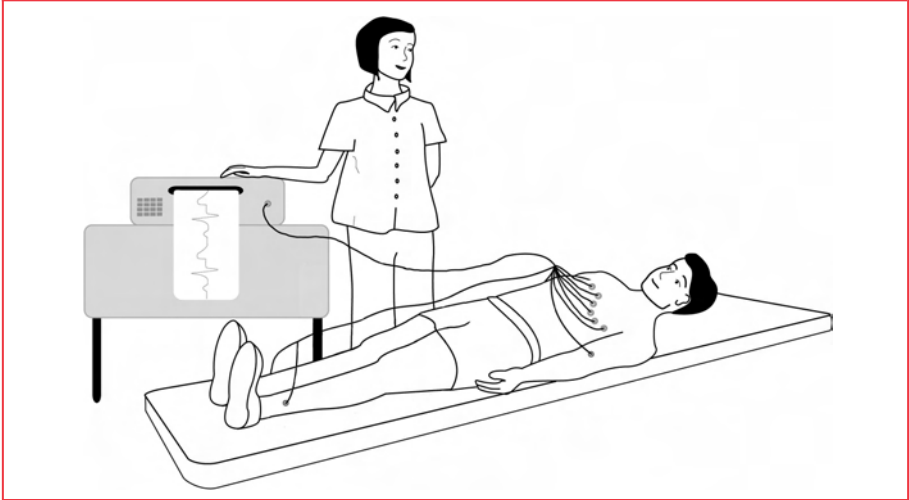
Your doctor may suggest that you have some of the tests described below.

Electrocardiogram (ECG) *

This is the most basic test.

Figure 4: Electrocardiogram (ECG)

Electrical leads from the ECG machine are taped to the chest, legs and arms and a recording is made of the electrical activity of the heart.



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.

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 ARVC, Brugada syndrome or PCCD.

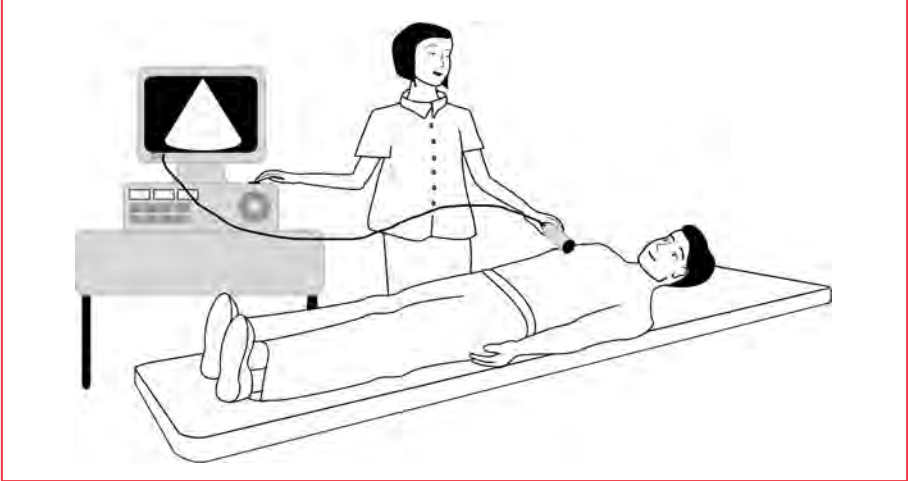
* Tests are non-invasive. Non-invasive means that it does not involve penetrating the skin or body

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 cardiomyopathy or by heart disease that has damaged the heart - for example, a previous heart attack that you may not have even been aware of. It can also monitor the heart for changes and is very useful for keeping an eye on any enlargement of the aorta in Marfan syndrome. Occasionally, an injection of 'contrast' at the time of an echo (known as a contrast echo) can improve the image quality and improve the ability to detect these conditions. An ECHO can be performed while someone is exercising and help in the diagnosis of coronary artery disease.

Figure 5: Echocardiogram

The operator puts some clear gel on your chest and then places an ultrasound probe on it. The probe sends ultrasound beams into your body and their reflections are detected and used to generate images of the heart. You can see different parts of your heart on a screen as the probe is moved around on your chest. The test is similar to the ultrasound scan that is used to examine a pregnant woman's unborn baby. It is completely painless.



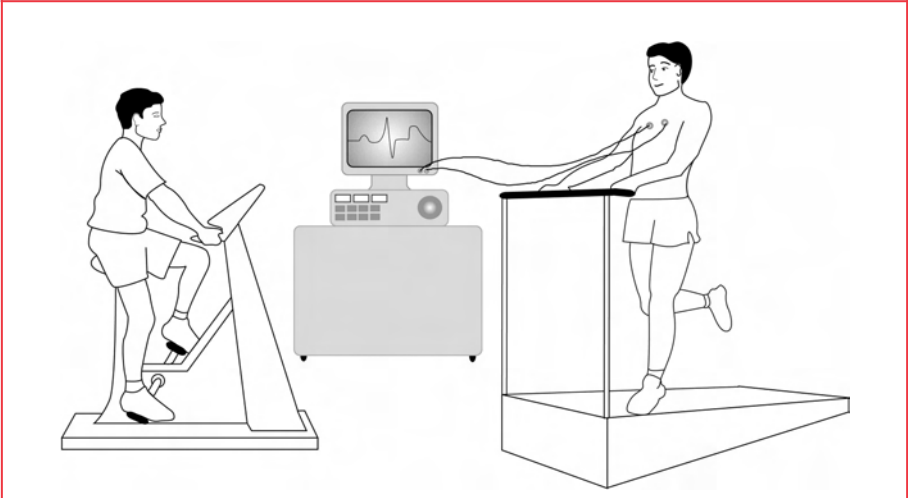
Exercise test * (Also called an exercise ECG.)

This test is the same as the ECG described on page 26 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 ARVC, LQTS or CPVT, diagnosing coronary artery disease or assessing risk in HCM.

** Tests are non-invasive. Non-invasive means that it does not involve penetrating the skin or body.*

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 amount of oxygen your body uses during exercise. You will be asked to breathe into special equipment while you are exercising. If the efficiency of your heart is low, this may suggest that you have a cardiomyopathy (inefficient pumping action of the heart).

** Tests are non-invasive. Non-invasive means that it does not involve penetrating the skin or body.*

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. Anything between three to twelve 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 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 or other features which may be present in certain conditions such as LQTS and Brugada syndrome.

Cardiomemo and event recorder*

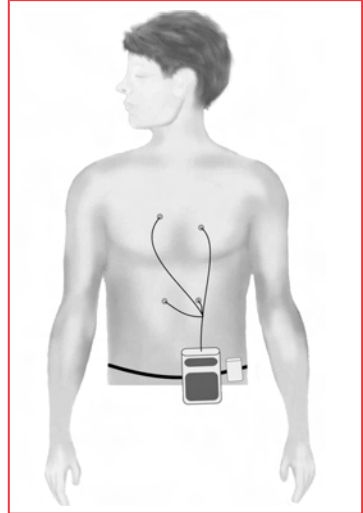
These are more sophisticated versions of the basic Holter. Whenever you have an attack of symptoms, you can activate the device to record your heart's rhythm. (You can also do this with the digital Holter.) The advantage of the cardiomemo is that it doesn't have any leads, so you can just place it on your chest when you get symptoms, without having to put any leads in position.

Implantable loop recorder (ILR)

When it is difficult to assess or record a symptom because it only happens infrequently - as with blackouts - an implantable loop recorder 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 ILR. 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.

Figure 7: Holter

The Holter monitor is attached by anything between 3 to 12 electrical leads to your body. It monitors your heart's electrical activity over a period of time.



* Tests are non-invasive. Non-invasive means that it does not involve penetrating the skin or body.

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 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.

Recently slow adrenaline (epinephrine) injections have been given in a similar way to try and improve the diagnosis of certain forms of long QT syndrome by uncovering longer QT intervals. The same approach has been used for CPVT where the test tries to reveal specific forms of **ventricular arrhythmia**. The place of these tests over and above more usual tests such as an **exercise ECG** is unclear.

Adenosine (another short-acting chemical) is given under the same circumstances if Wolff-Parkinson-White syndrome (WPW) is considered a possible diagnosis.

Cardiac magnetic resonance/magnetic resonance imaging (CMR/MRI) scan*

This is a special kind of scan used to examine the structure of the heart and the nature of its muscle. It uses a magnetic resonance scanner that creates intense fluctuating magnetic fields around your body while you are inside the scanner. This generates the signals that make up the pictures produced. It may be useful for detecting and assessing cardiomyopathy, especially if it is very subtle and may mimic a channelopathy (for example, ARVC). It can also be useful to assess myocarditis, coronary artery anomalies or the signs of coronary artery disease.

Computed tomography (CT)

CT is a special type of X-ray technique where three-dimensional images are produced using a series of two-dimensional X-ray images. It is mainly used to look at the coronary arteries.

* Tests are non-invasive. Non-invasive means that it does not involve penetrating the skin or body.

Other tests

Coronary angiography, electrophysiological (EP) study, ablation and cardiac biopsy

Depending on the results of the above tests, your doctor may suggest that you have other tests, such as coronary angiography or an **electrophysiological (EP) study**. 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 EP study involves placing electrical leads inside your heart to analyse its electrical properties and induce **arrhythmias**. It may be useful in diagnosing Wolff-Parkinson-White syndrome (WPW) and progressive cardiac conduction defect (PCCD) and deciding on what treatment to give people with Brugada syndrome. If the extra pathway seen in WPW is detected at EP study it can be treated there and then by 'burning' it away using high frequency radio waves. This procedure is called **RF ablation**. This may also be useful in treating arrhythmia if medicines are unable to control them, particularly if you have ARVC.

If your doctors suspect a heart muscle condition (cardiomyopathy or myocarditis) then a small sample, a **biopsy**, can be taken from the inside of the heart in a procedure similar to or even at the time of a coronary angiogram or an EP study. The biopsy can then be examined under a microscope for any signs of cardiomyopathy and help in diagnosis.

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 hospitals.

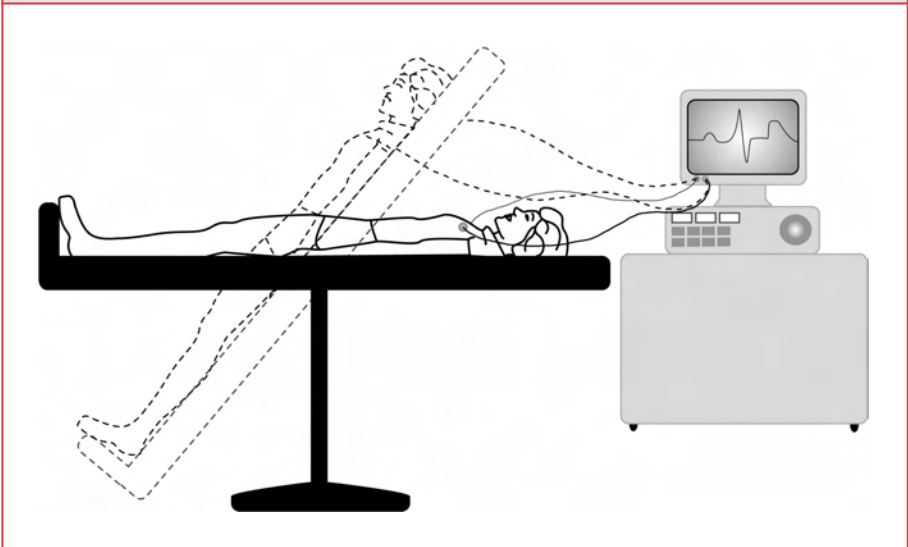
Tilt-table testing

Tilt-table testing is used to identify other common conditions that can cause blackouts - such as vasovagal syndrome (see page 45) or simple fainting - that tend to particularly affect young women and girls. These symptoms are very similar to the symptoms of 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 channelopathies, cardiomyopathies, Marfan syndrome and familial hypercholesterolaemia, **mutations** of specific **genes** have been detected that are thought to cause a specific disease. So, in principle, if we could identify these mutations, we would be able to make a diagnosis in any **DNA** sample, including any obtained from **SADS** or sudden death victims at their autopsy or from 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 every 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.

General Lifestyle Advice

Exercise

The majority of conditions that we have discussed 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 very difficult if you are used to doing a lot of sport. It is important to get a balance between the benefit of restricting exercise and the negative impact the restrictions may have on you.

In certain conditions, such as Brugada syndrome and sodium channel LQTS, a cardiac arrest may occur at night and during sleep. If you have one of these conditions, your doctor can advise your family or 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.)

Drugs to avoid

Anyone with a condition affecting the heart 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 cardiac arrest.

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 35 and another comprehensive list is available online at www.qtdrugs.org. 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 less clear and caution must be used. We give a list of these medications on page 36 as they currently stand, although the evidence for some drugs is very limited. Another comprehensive list is available online at www.brugadadrugs.org.

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. This is less of a problem in the other heart conditions but may still occur. 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 intravenous fluids.

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.

* = unlicensed, withdrawn or suspended in the UK market. Derived from Yap et al 1999, reports from www.qt drugs.org up to January 2008, case reports from www.pubmed.com up to January 2008, the British National Formulary edition 54 at www.bnf.org.

Drug Group	Avoid Completely	Close Monitoring and Professional Supervision Required if Drug is Absolutely Necessary
Antiarrhythmics:		
Class I:	quinidine* procainamide disopyramide ajmaline* dihydroquinidine*	flecainide propafenone pirmenol* cibenzoline* mexiletine
Class III:	amiodarone sotalol d-sotalol* dofetilide* azimilide* ibutilide* sematilide* ersentilide* almokalant* nifekalant* terikalant* dronedarone*	
Anti-anginals & vasodilators:	prenylamine* terodiline* lidoflazine* bepridil*	Vardenafil
Anti-hypertensives:	indapamide	nicardipine isradipine moexipril/hydrochlorothiazide
Antihistamines:	terfenadine* astemizole*	ebastine* diphenhydramine
Serotonin agonists & antagonists:	cisapride* ketanserine* dolasetron	ondansetron, granisetron
Antimicrobials:		
Macrolides:	erythromycin clarithromycin spiramycin	azithromycin roxithromycin* telithromycin
Fluoroquinolones:	sparfloxacin* moxifloxacin	gatifloxacin* grepafloxacin* levofloxacin gemifloxacin* ofloxacin
Antifungals:		ketoconazole fluconazole itraconazole voriconazole
Antimalarials:	chloroquine halofantrine* pentamidine	quinine
Others:		foscarnet amantadine cotrimoxazole trimethoprim sulfa
Psychiatric:		
Tricyclic antidepressants:		amitriptyline nortriptyline desipramine* clomipramine imipramine trimipramine doxepin trazodone protriptyline* amoxapine*
Serotonin reuptake		fluoxetine paroxetine venlafaxine sertraline zimidine* citalopram
Phenothiazines:	thioridazine* chlorpromazine	trifluoperazine prochlorperazine fluphenazine pimozide mesoridazine quetiapine
Others:	haloperidol droperidol* sertindole	risperidone ziprasidone maprotiline lithium clozapine
Anti-cancer:	arsenic trioxide	tacrolimus tamoxifen geldanamycin* octreotide sunitib
Others:	probutol domperidone levomethadyl* methadone	vasopressin tizanidine alfuzosin amantidine felbamate* fosphenytoin chloral hydrate organophosphates* galantamine solifenacin clobutinol* Perflutren lipid microspheres (echocardiographic contrast)

Drugs which people with Brugada syndrome should avoid:

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

Adapted from: Brugada syndrome. Napolitano C, Priori SG. Orphanet J Rare Dis. 2006 Sep 14;1:35. Brugada Syndrome: Report of the Second Consensus Conference Charles Antzelevitch et al. Circulation. 2005;111:659-670

Drug Group	Drugs to be avoided
Class I antiarrhythmics:	all to be avoided including flecainide, propafenone, disopyramide, pilsicainide, ajmaline, procainamide, cibenzoline
Local anaesthetics (non antiarrhythmic):	bupivacaine
Alpha adrenergic agonists:	methoxamine, noradrenaline
Beta-blockers:	all to be avoided especially propranolol
Calcium channel blockers:	verapamil, nifedipine, diltiazem
Nitrate:	isosorbide dinitrate, nitroglycerine
Potassium channel activators:	pinacidil
Parasympathetic agonists:	acetylcholine
Ergot alkaloids:	ergonovine
Tricyclic antidepressants:	all to be avoided including amitriptyline, nortriptyline, desipramine, clomipramine
Tetracyclic antidepressants:	maprotiline
Phenothiazine:	perphenazine, cyamemazine
Selective serotonin reuptake inhibitors:	fluoxetine
Opioid analgesics:	propoxyphene
First-generation antihistamines:	dimenhydrinate
Others:	propofol, lithium, cocaine, alcohol intoxication

The future

Research in the channelopathies and cardiomyopathies is progressing rapidly and in the future it is expected that the majority of 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. It is crucial that, when a heart disease such as a channelopathy or cardiomyopathy is diagnosed, all immediate blood relatives should be evaluated by a specialist **cardiologist** to find out if they have an inherited heart disease.

Technical terms

Alcohol septal ablation

If you have HCM that makes you breathless, dizzy or suffer chest pain, this may be due to blockage of the flow of blood from the main pumping chamber in the heart due to thickening of the heart muscle. An injection of alcohol into the blood vessel supplying this bit of muscle can help to reduce the obstruction and improve symptoms. This can be done at the time of coronary angiography (see page 31).

Angiography and angioplasty

During coronary angiography the coronary arteries (the arteries that supply blood to the heart muscle) are injected with a dye to reveal any narrowing or blockages. This can then be treated with a catheter to open up any narrowing by blowing up a balloon at the narrowing (known as angioplasty) and inserting a metal tube (stent) to keep the artery open (see pages 25 and 31).

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). 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. The aortic valve is composed of three cusps. 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 life-threatening.

Asystole

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

Atrial

Of or belonging to the atria.

Atrial fibrillation

An arrhythmia involving the atria (the two upper chambers of the heart).

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'.

Atrioventricular (AV) 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.

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.

Channelopathy

Conditions characterised by defective ion channels in the cells of the heart.

Computed tomography (CT)

An imaging technique where three-dimensional images are produced using a series of two-dimensional X-ray images.

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.

Electrical cardioversion

A cardioversion is a procedure that can help your heart rhythm, get back to its normal rhythm (called sinus rhythm) if you have persistently abnormal rhythm, such as atrial fibrillation or atrial flutter.

Fibrillation

The fast irregular contraction of muscle fibres in the heart

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.

Heart failure

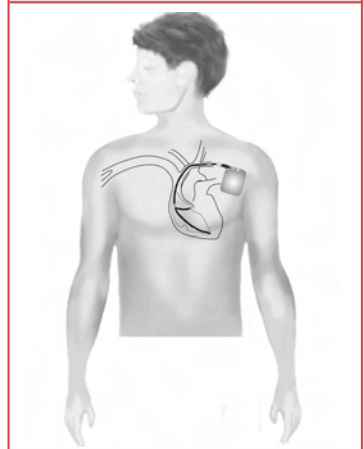
The reduced pumping function and efficiency of the heart muscle resulting in the body not being supported properly by the heart muscle. This can result in breathlessness and oedema.

Implantable cardiac defibrillator (ICD)

A metal electronic device similar to a pacemaker (see below). It can regulate the rhythm of the heartbeat, 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 1½ 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.

Figure 9: ICD (implantable cardiac 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.



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.

Magnetic resonance imaging (MRI)

An MRI uses a magnetic field and radio waves to create pictures of tissues, organs and other structures inside the body.

Mitral valve

This is 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 100 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 arrhythmia.

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.

Oedema

Swelling of the ankles, legs and even abdomen due to the collection of fluid under the skin in patients with severe cardiomyopathy, particularly DCM. It is caused by increased pressure of blood returning to the heart from the body but not being pumped efficiently by the heart due to heart failure.

Pacemaker

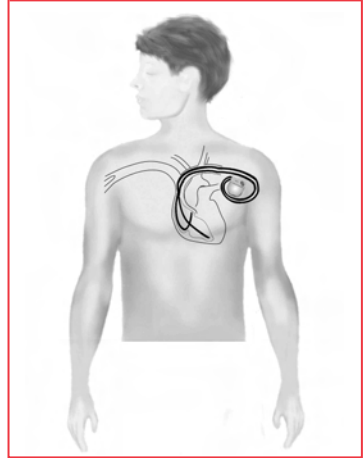
A small metal electronic device with internal batteries. It sits under the skin at the left shoulder. It is attached to the top and bottom chambers of the heart by two electrical leads that are inserted via the large veins near the shoulder. These leads both monitor the heart rhythm and allow treatment 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.

Figure 10: Pacemaker.

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



Palpitations

A fast heart beat that may be felt as the heart pounding regularly or irregularly.

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.

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.

Radiofrequency (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 or areas that cause arrhythmia in ARVC.

Repolarisation

The electrical resetting of the heart muscle ready for its next activation. The time taken is measured by the QT interval.

Sinoatrial (SA) node

The sinoatrial node generates the normal or sinus rhythm of the heart. It is impulse-generating tissue that acts as a pacemaker. It is located in the right atrium of the heart.

Sudden cardiac death (SCD)

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.

Surgical myectomy

This is a treatment for HCM that involves removing small amounts of over developed heart muscle to reduce any blockage in blood flow in the heart.

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. It can be diagnosed with a tilt-test or implantable loop recorder (see pages 29 and 31).

Ventricles

The two bottom chambers of the heart that pump blood to the body and the lungs.

Ventricular

From, or belonging to, the ventricle.

Ventricular fibrillation

In ventricular fibrillation (VF) the muscle fibres in the ventricles contract completely randomly so the ventricles cannot perform their pumping action.

Index

ablation: 17, 21, 31, 37, 44
angiogram: 25
arrhythmia syndromes: 4-5
arrhythmogenic right ventricular cardiomyopathy: 16-17, 26, 27, 31
arrhythmogenic right ventricular dysplasia: 16-17
ARVC: 16-17, 26, 27, 31
ARVD: 16-17
Autopsy: 32, 38
AV node: 2-3, 38, 41
Biopsy: 31
Brugada syndrome: 4, 7-10, 13, 25, 30-31, 33-34, 36
cardiac arrest: 2-5, 8, 33, 39, 40
cardiac biopsy: 31
cardiac channelopathies: 4-5
cardiac event recorder: 29
cardiac magnetic resonance scan: 17, 30
cardiomemo: 29
cardiomyopathy: 13, 25, 27, 28, 30, 39, 42
cardiopulmonary exercise test: 28-39
catecholaminergic polymorphic ventricular tachycardia: 4, 10-11
cervical sympathectomy: 8, 39
channelopathies: 4-5, 7, 25, 30, 31, 32-33
CMR scan: 30
computed tomography scan: 30, 39
congenital heart disease: 39
coronary angiogram: 25, 31
coronary artery anomalies: 23-24, 30
coronary artery disease: 4, 18, 24-25, 27, 30, 31, 39, 40
coroner: 40
CPVT: 4, 10-11
CT scan: 30, 39
Defibrillator: 8, 33, 40, 41
drugs to avoid: 33, 35-36
early repolarisation syndrome: 4, 12-13
ECG: 2, 26-28
ECHO: 27
Echocardiogram: 27
ectopic beat: 40
electrocardiogram: 2, 26-28
electrophysiological study: 31
EP study: 31
ERSCD: 44
event recorder: 29
exercise: 33
exercise test: 27-28, 39
familial atrial fibrillation: 4, 13
familial hypercholesterolemia: 24-25
family history: 25
genetic testing: 32-33
heart disease: 13, 25, 27, 39, 44
heart / how the heart works: 2-3
HCM: 13-16, 27, 37, 45

Index

Holter: 29
hypertrophic cardiomyopathy: 13-16, 27, 37, 45
hypokalaemia: 7, 10, 34
ICD: 8, 41
idiopathic ventricular fibrillation: 12-13
ILR: 29, 45
implantable cardiac defibrillator: 8, 41
implantable loop recorder: 29, 45
ion channel: 5, 7, 41
ion channelopathies: 4, 5, 7
IVF: 12-13
lifestyle advice: 33
long QT syndrome: 5-8, 9, 25, 27, 29, 31, 32-33, 35
LQTS: 5-8, 9, 25, 27, 29, 31, 32-33, 35
low blood potassium levels: 34
magnetic resonance imaging scan: 30, 42
Marfan syndrome: 21-22
medical examination: 25
mitral valve prolapse: 42
MRI scan: 30, 42
Mutation: 5, 13, 32-33, 42
MVP: 42
Myocarditis: 22-23
Pacemaker: 2-3, 8, 38, 43, 44
Pathologist: 43
PCCD: 11-12
post-mortem: 38, 43
private mutations: 32-33
progressive cardiac conduction defect: 11-12, 31
premature coronary artery disease: 24
provocation test: 9, 30
QT interval: 6-7, 9, 12, 30, 33, 44
SA node: 2-3, 9, 44
SADS: 4-5, 25, 32
SCD: 4, 44
short QT syndrome: 12
sodium channel disease: 4, 13
SQTS: 12
signal averaged ECG: 26
sudden arrhythmic death syndrome: 4-5
sudden cardiac death: 4-5, 25, 44
surgery: 4, 8, 11, 24, 39
technical terms: 37-45
tests: 25-33
tilt-table test: 31-32
Wolff-Parkinson-White Syndrome: 20-21, 30, 31, 40
WPW: 20-21, 30, 31, 40



Cardiac Risk in the Young is a charity which:

- offers support to those young people diagnosed with cardiac conditions
- publishes and distributes medical information written by leading cardiologists for the general public
- 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
- 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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