



Inherited heart conditions

Arrhythmogenic right ventricular cardiomyopathy



In association with



Authors:

Prof. William J. McKenna

Professor of Cardiology & Clinical Director, The Heart Hospital, University College London.

Dr Perry Elliott

Reader in Inherited Cardiac Disease / Honorary Consultant Cardiologist, The Heart Hospital, University College London.

Contributions from Dr Alison Muir, Dr Constantinos O'Mahony & Dr Caroline Coats, Cardiology Registrars, The Heart Hospital, University College London.

The booklet has been developed from the original version inspired by the founder of the Cardiomyopathy Association – Carolyn Biro. Published by the British Heart Foundation.

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

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

Contents

Introduction	00
Understanding your heart	
The normal heart	00
Arrhythmogenic right ventricular cardiomyopathy (ARVC)	
What is ARVC?	00
When does ARVC develop?	00
What are the symptoms of ARVC?	00
How is ARVC diagnosed?	00
Is there a cure for ARVC?	00
What other conditions can occur as a result of ARVC?	00
Testing, treatment and your family	
Implications of a diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC)	00
Assessment at a clinic for inherited heart conditions	00
Treatments for ARVC	00
Treatment for other conditions that can occur as a result of ARVC	00
Everyday life	
Living with arrhythmogenic right ventricular cardiomyopathy (ARVC)	00
Pregnancy and childbirth	00
Looking forward	
The future	00
Technical terms	00
For more information	00
References	00
Index	00

Introduction

You may be reading this booklet because you have been diagnosed with a heart condition called arrhythmogenic right ventricular cardiomyopathy (ARVC). Or maybe your doctor has suggested that you should have some tests to find out if you have inherited this condition because someone else in your family has been diagnosed with it.

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

There are three main types of cardiomyopathy:

- arrhythmogenic right ventricular cardiomyopathy (ARVC)
- hypertrophic cardiomyopathy (HCM or HOCM)
- dilated cardiomyopathy (DCM).

This booklet is about ARVC. For information on the other types of cardiomyopathy, see the other booklets in this series. See page 00 for details.

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

This booklet:

- describes how the normal heart works
- explains what ARVC is and what can go wrong if you have the condition
- explains why it is important that the close blood relatives of someone with the condition should have an assessment to find out if they have inherited the same condition
- describes the tests your doctor or cardiologist may ask you and your close family members to have
- describes the treatments you may need
- offers advice on how to live a healthy lifestyle if you are found to have ARVC.

We explain the medical and technical terms as we go along but, if you find a word you don't understand, look it up in the list of Technical terms on page 00.

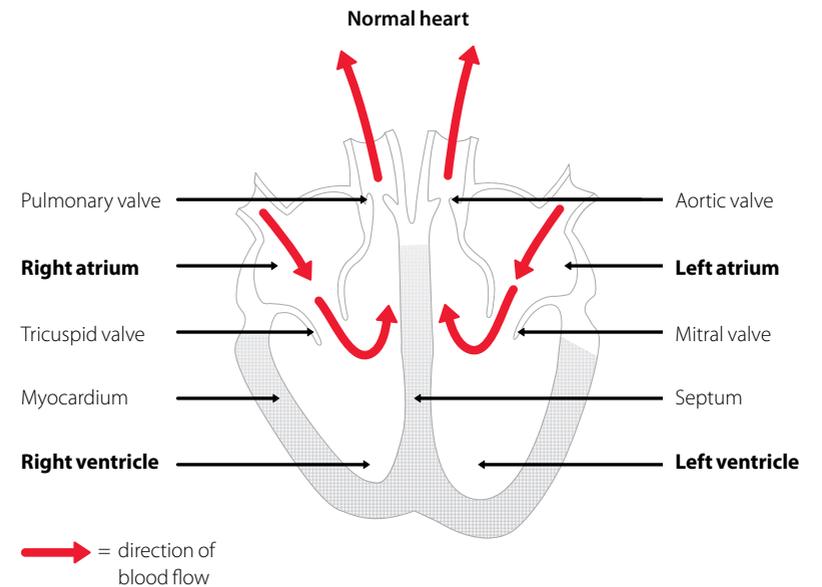
This booklet has been produced with the help of cardiologists and other health professionals, and also people who have ARVC. We hope that the booklet will help you to understand your condition and to come to terms with what it means for you and your family. If you need further support or information, see page 00.

Understanding your heart



The normal heart

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

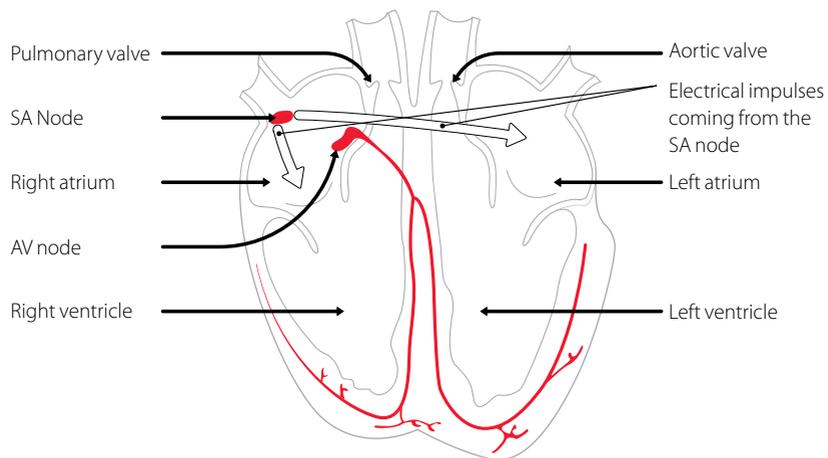


How the heart functions electrically

The pumping action of the heart is caused by a flow of electricity through the heart that repeats itself in a cycle. The normal trigger for the heart to contract comes from the heart's natural pacemaker, the SA node (sino-atrial node), which is in the right atrium (see the diagram on page 00). The SA node sends out regular electrical impulses, which make the atria contract and pump blood into the ventricles. The electrical impulse then passes to the ventricles through a form of 'junction box' called the AV node (atrio-ventricular node). This electrical impulse spreads into the ventricles, causing the heart muscle to contract and to pump

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

Normal electrical signals in the heart



Structure of the heart

The heart consists of three layers:

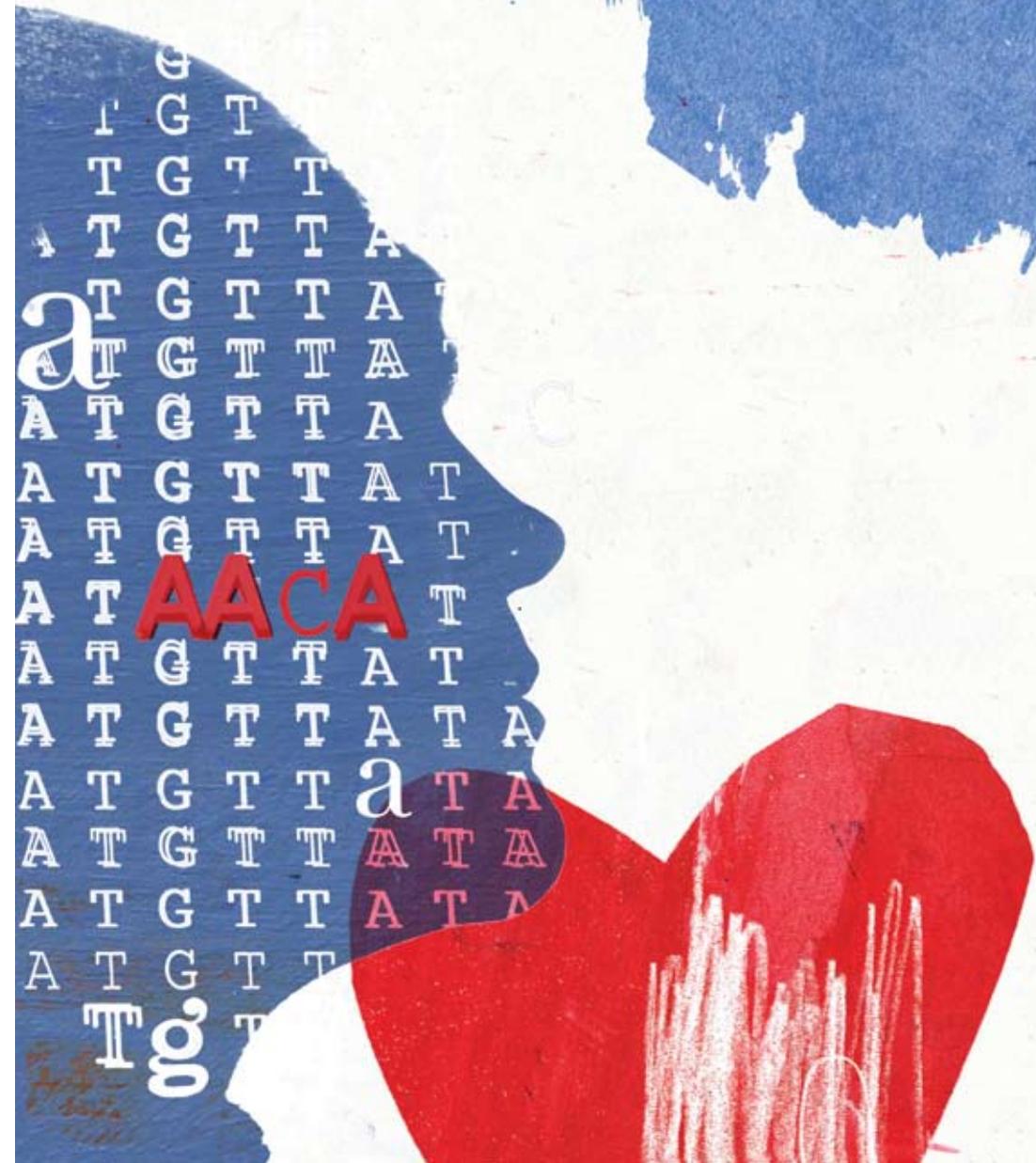
- the endocardium
- the myocardium
- the pericardium.

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

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

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

Arrhythmogenic right ventricular cardiomyopathy (ARVC)



What is ARVC?

Arrhythmogenic right ventricular cardiomyopathy – or ARVC for short – is a disease of the heart muscle. It is a genetic condition. This means that it can be passed on through families and is caused by a change or mutation in  or more genes. We explain more about how it is inherited on page 00.

When a team of cardiologists in France first identified the condition in the late 1970s, they called it arrhythmogenic right ventricular dysplasia. ('Dysplasia' means abnormal development.) Their patients had ventricular tachycardia, a type of abnormal heart rhythm which originates in the right ventricle. (For more information on ventricular tachycardia, see page 00.) They also noticed that in those patients the muscle of the right ventricle had changed in certain ways.

In the 1980s and 90s, the same changes in the heart muscle were noticed at the post-mortem examinations of some young people who had died suddenly, especially in athletes. Researchers also noticed that there was a tendency for the condition to run in families. As the problems associated with the condition were due to changes to the heart muscle, the name of the condition was changed to 'arrhythmogenic right ventricular cardiomyopathy'.

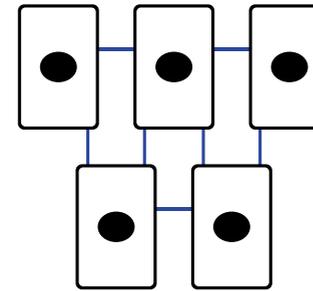
At first, ARVC was described as a disease of the right ventricle, but the changes in the heart muscle can also affect the left ventricle. ARVC does not affect the structure of the muscle of the atria (the upper chambers of the heart).

How does ARVC affect the heart muscle?

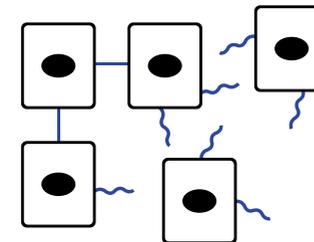
In people with a normal heart, the cells of the heart muscle are held together by proteins. It is thought that, in people with ARVC, these proteins have not developed properly. As a result, the proteins cannot keep the heart muscle cells together when under stress – such as when the heart is beating faster or working harder than normal, for example during exercise. The cells become detached and die (see the diagram on page 00). The damaged and dead heart muscle cells become fibrous and cause scarring. Fatty deposits build up, in an attempt to repair the damage. The condition is usually progressive – which means that it will get worse. The treatment for ARVC aims to control the symptoms.

As a result of these changes to the structure of the heart muscle, the walls of the ventricle become thin and stretched, which means that the heart cannot pump effectively. Also, because of the changes to the heart muscle cells, the normal passage of electrical impulses through the heart is interrupted or altered, and can cause life-threatening arrhythmias (abnormal heart rhythms) and in some cases sudden cardiac death.

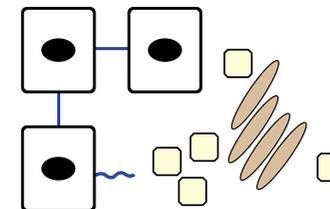
How the cells of the heart muscle are affected in people with ARVC



In a normal heart, the cells of the heart muscle are attached to each other at junctions by proteins.



In people with ARVC, the proteins have not developed properly and cannot keep the heart muscle cells together when under stress, such as when the heart is beating faster or working harder than normal, for example during exercise. Cells become detached and die.



The damaged and dead heart muscle cells become fibrous and cause scarring. Fatty deposits build up, in an attempt to repair the damage.

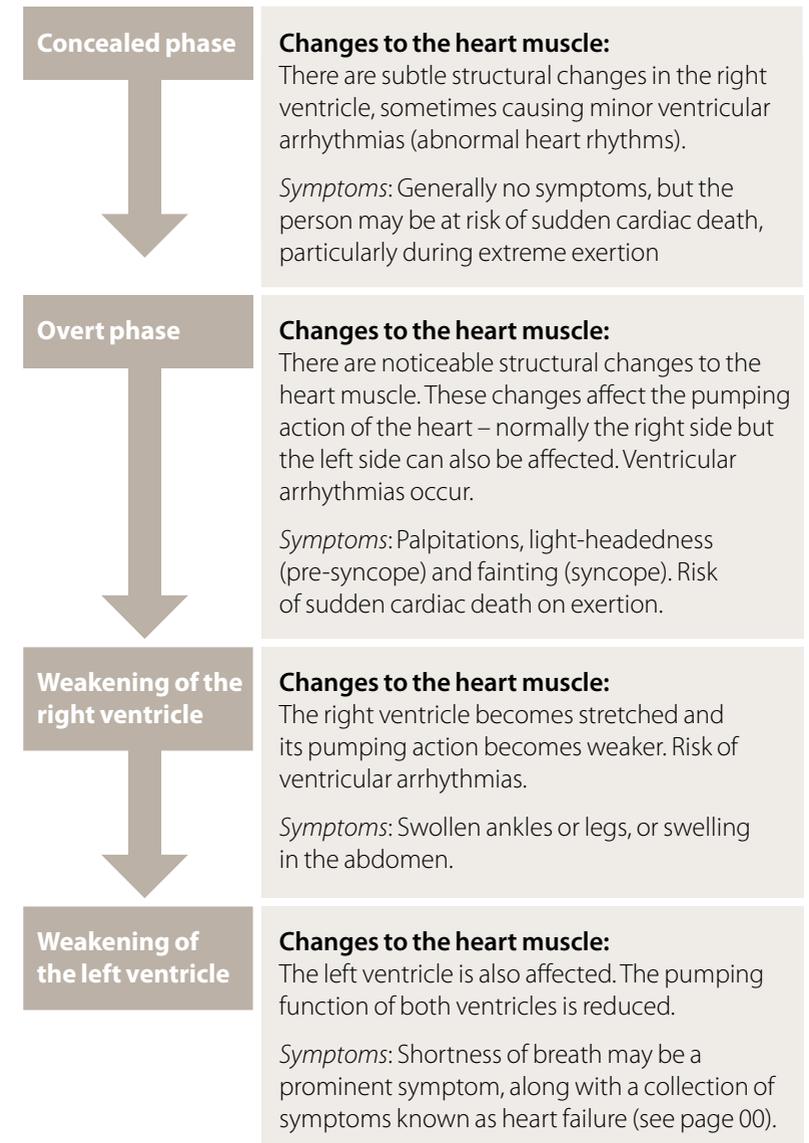
When does ARVC develop?

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

If there is a mistake in one of these codes, the cells will do something different or not develop as they should do. In people with ARVC, there is a mistake in the sequence of letters that are needed to form the proteins that hold the heart muscle cells together (as described on page 00). This 'mistake' is known as a genetic mutation. The mutation is present from birth, but the changes to the heart muscle rarely occur before adolescence. Explain more about genetic inheritance and coding on pages 00-00.

ARVC can have four phases, although not everyone will develop all of the phases or be affected in the same way. Many people with the condition never develop any serious problems. We describe how the condition can progress in the diagram below, and we explain more about the symptoms on page 00.

Phases of ARVC



What are the symptoms of ARVC?

Some people with ARVC don't get any symptoms. Others do get symptoms but many people never have any serious problems related to their condition. Some of the common symptoms are listed below. The symptoms will depend on how the disease progresses in the person and whether the condition affects one or both ventricles.

People with ARVC may get one or more of the following symptoms.

Palpitations

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

Light-headedness (pre-syncope)

This may occur when you are exercising, or after palpitations, or sometimes for no apparent reason. It may feel as if you are almost passing out, or that your head is spinning.

Fainting (syncope)

This is when you lose consciousness. This is often described as collapse, fainting, passing out, having a blackout, or being 'out for a few seconds'.

Swelling of the ankles or legs (oedema), or of the abdomen (acites)

Swelling of the ankles or legs or in the abdomen could indicate that the pumping action of your heart is poor. This can happen because the right ventricle cannot pump blood to the lungs properly. This is sometimes known as right-sided heart failure. It can also happen if the left ventricle is unable to pump blood out. Your doctor will do some tests to find out how your heart is affected. For more information on tests, see page 00.

Shortness of breath (dyspnoea)

A minority of patients with ARVC get short of breath when they are being physically active. This is normally caused by damage to the left ventricle or because the person has an arrhythmia. Most people are only mildly

restricted by their shortness of breath. However, a small number of people may be more restricted in the level of exercise they are able to take, and very occasionally some people may be short of breath while they are resting or lying down.

Any of the symptoms described above may be caused by an **arrhythmia**, which often occurs during strenuous activity. We explain more about the different types of arrhythmia below. Chest pain, breathlessness, sweating and nausea may also accompany the arrhythmia. All of these symptoms need to be investigated. If the person loses consciousness, they should be taken to an accident and emergency department at a hospital.

If your heart muscle is severely affected and the pumping action of your heart is significantly reduced, your doctor may tell you that you have heart failure. We explain more about **heart failure** on page 00.

Arrhythmias

The abnormality of the heart muscle in people with ARVC can sometimes interfere with the normal electrical activity of the heart. In the affected parts of the heart muscle, the electrical impulse may become disrupted as it crosses the areas of damaged and scarred heart muscle cells. This can lead to slow, fast or erratic heart rhythms known as arrhythmias. Arrhythmias are common in people with ARVC. We describe some of the different types of arrhythmia below. For information on treating them, see page 00.

Ventricular premature beats (VPB)

Also called premature ventricular contractions (PVC) or ventricular ectopics
Ventricular premature beats occur when an electrical impulse starts in one of the ventricles. The ventricles respond by contracting before the normal impulse from the atria arrives at the ventricles, which creates the feeling of an extra heartbeat. People with ventricular premature beats may experience an awareness of the heart beating out of rhythm or fluttering, known as palpitation.

Ventricular premature beats are not usually dangerous and are relatively common in healthy people. They can occur because of simple things like drinking too much coffee, smoking, lack of sleep, anxiety or drinking alcohol. However, ventricular premature beats may also be a sign of an underlying heart condition, particularly if they happen irregularly or if there are lots of them.

Ventricular tachycardia (VT)

Ventricular tachycardia is when you have a series of ventricular premature beats in rapid succession. The beats can occur in short bursts, or they may last longer than 30 seconds. Ventricular tachycardia can produce a fluttering feeling in the chest, or a sensation of your heart beating out of rhythm. You may feel as if you are about to faint, or may feel light-headed or dizzy. This is known as pre-syncope.

Ventricular tachycardia is a serious arrhythmia. It can cause a sudden fall in blood pressure and may cause loss of consciousness known as **syncope**.

Ventricular fibrillation (VF)

Ventricular fibrillation is a life-threatening arrhythmia and requires urgent hospital treatment. It happens when the electrical impulses of the heart become completely disorganised, causing the ventricles to quiver or fibrillate. When this happens, the heart cannot pump the blood out of the ventricles, and the heart stops beating. This is known as a **cardiac arrest**. If a person suffers a cardiac arrest, prompt resuscitation and defibrillation treatment (giving an electrical shock) may help the heart to return to a normal rhythm and prevent sudden cardiac death. We explain more about sudden cardiac death on page 00.

Atrial fibrillation (AF)

In atrial fibrillation, the atria (the two upper chambers of the heart) beat irregularly and very fast. This can lead to feelings of palpitations or fluttering in the chest. This may happen just occasionally (**paroxysmal atrial fibrillation**), or it may be there all the time (**persistent atrial fibrillation**). Atrial fibrillation is not generally life-threatening, but when the atria do not contract normally, this increases the risk of a clot forming in

the atria, which can sometimes lead to a stroke.

For more information on arrhythmias, see our booklets *Heart rhythms and Atrial fibrillation*. (To order our booklets, see 'More information' on page 00.)

How is ARVC diagnosed?

It can be difficult to diagnose ARVC, as the changes to the heart muscle may be very subtle. Many of the changes and symptoms can also be caused by other conditions, making diagnosis even more difficult.

Your doctor may suspect that you have ARVC because of your symptoms, or because of the results of a chest x-ray. Or, you may be going for tests because someone else in your family has been diagnosed with the condition.

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

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

We explain more about these and other tests on page 00.

Is there a cure for ARVC?

At present there is no cure for ARVC but there are many treatments available which aim to reduce the risk of developing symptoms or to help relieve your symptoms, and to prevent complications. We explain more about these treatments on page 00.

What other conditions can occur as a result of ARVC?

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

Heart block

A small number of people with ARVC may get heart block. This is when the electrical impulse in the heart travels down to the ventricles slowly, or may even be completely blocked. This affects the way that the heart contracts. For information on treatment for heart block, see page 00.

Heart failure

Heart failure is a collection of symptoms – as described on page 00 – caused by the reduced pumping action of the heart. If you have ARVC, and if your heart muscle is badly affected, your doctor may tell you that you have heart failure. There are four stages of heart failure. Stage four is the most severe. There is heart failure for heart failure but the symptoms can be controlled – see page 00.

Is there a risk of sudden cardiac death with ARVC?

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

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

For information on treatment for people with ARVC who may be at risk of sudden cardiac death, see page 00.



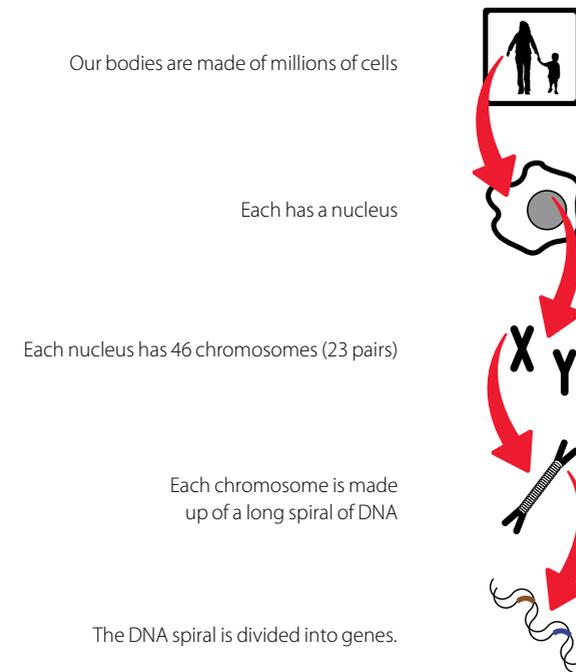
Testing, treatment and your family

Implications of a diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC)

How do people get ARVC?

Our bodies are made up of millions of cells. Each cell has a nucleus, which contains information that makes each one of us unique. These are our genes. Your genes give the instructions that are needed for development and growth of all the cells in your body, and they determine things like hair colour, eye colour, height and blood type.

Genes are arranged end to end along a threadlike structure called a chromosome. The chromosomes and genes are made up of a chemical substance called DNA. Each cell usually carries 46 chromosomes arranged in 23 pairs. See the diagram below.



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

It may help to think of yourself as a book:

- The chromosomes are the chapters. There are 22 pairs of chromosomes and one pair of sex chromosomes, (men have an X and a Y chromosome XY, and women have two X chromosomes XX), so 23 chapters.
- The genes are coding for proteins that dictate characteristics like hair and eye colour or your height. The coding is like the words in the chapters.
- The code in your DNA is like the letters in the words. The way these 'letters' are arranged can influence your risk of developing conditions such as high blood pressure or heart disease. For example, the words BARE and BEAR have the same letters, but they're arranged differently so they mean different things. In the same way, if the codes in the DNA are arranged differently, they may work in different ways.

If one of your genes has a 'mistake' in it – a bit like a spelling mistake in a word – it could lead to an abnormal protein being produced in that particular type of cell. The same mistake will be passed on to the next and following generations. This is known as a genetic mutation. Some changes have little or no effect, but others can result in heart problems such as ARVC, or other genetic conditions such as cystic fibrosis (a disease affecting the lungs and pancreas).

Research has enabled scientists to identify genetic mutations that may indicate if a person is at risk of developing certain conditions. In some genetic conditions such as Huntington's chorea (a condition that affects muscle coordination and cognitive function), if the genetic mutation is identified in a person, the person will go on to develop the condition. However, in other genetic mutations – such as ARVC – the condition does not always develop in people who carry the abnormal gene. This is known as incomplete penetrance. (We explain more about this on page 00.) This

makes it hard to predict if a person who has the genetic mutation will go on to develop the condition.

The discovery of where and how these genetic mutations happen has helped us understand how ARVC develops. If someone is found to have the condition and a mutation is identified, this means that their close family members can be tested to see if they have the same mutation.

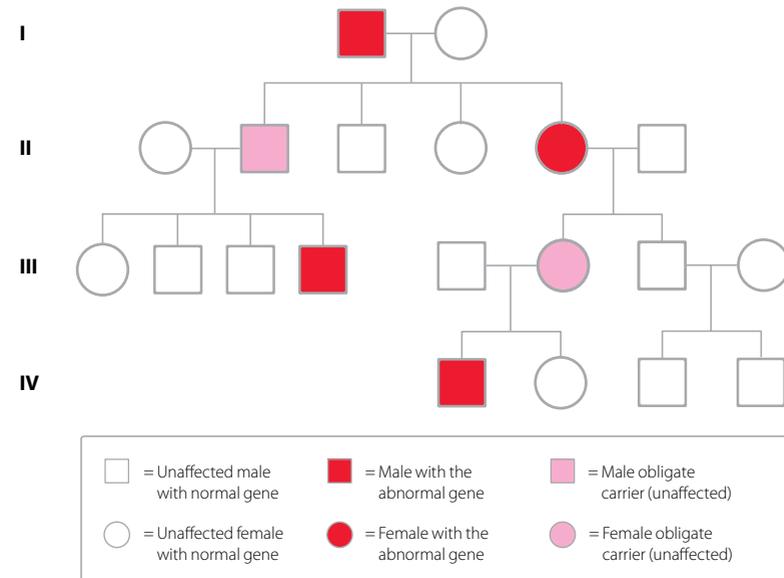
How is ARVC inherited?

Drawing a medical family tree will allow your doctor to see if there is anyone else in your family who may have the same condition. The family tree, known as a pedigree, will show an inheritance pattern.

The most common inheritance pattern for ARVC is known as autosomal dominant. Autosomal dominant means that each child of a person who has ARVC has a 50:50 or 1 in 2 chance of inheriting the genetic mutation. The genetic mutation may be passed on from an affected male or female, but not everyone who inherits the mutation will go on to develop the condition. This is known as incomplete penetrance. A person who has the genetic mutation but who does not develop the condition is known as an obligate carrier of the genetic mutation. However, the child of an obligate carrier could inherit the genetic mutation and has a 50:50 or 1 in 2 chance of developing the condition. So, in this case it may seem that the mutation has skipped a generation. See the diagram on page 00.

A family tree – ‘autosomal dominant with incomplete penetrance’ inheritance pattern

Generation



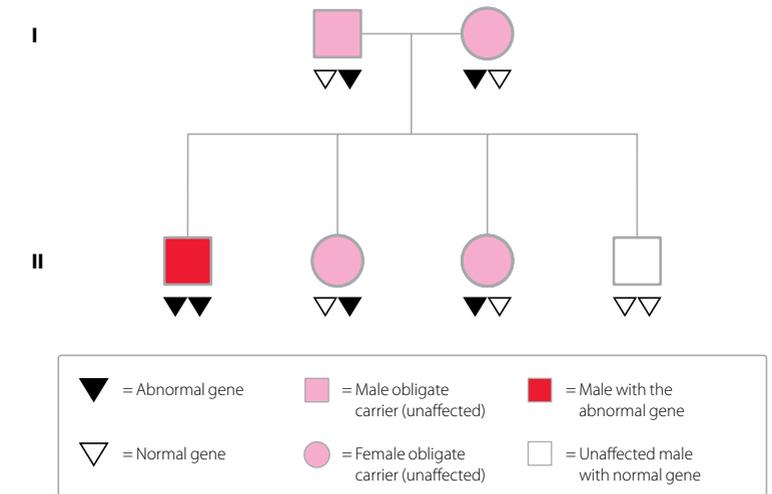
If you inherit the genetic mutation for ARVC, there is no way of knowing if you will develop the condition or how severe it might be. Two people in the same family may have different symptoms, and the symptoms may be mild in one person but severe in the other. However, if you don't inherit the mutation,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

If you don't know your medical family history, or if this is the first time that ARVC seems to have been diagnosed in your family, any child of an affected person probably has a 50:50 or 1 in 2 chance of being an obligate carrier or an affected person.

Sometimes, ARVC is inherited in a different pattern known as autosomal recessive. This means that a person only develops the condition if he or she inherits two copies of the genetic mutation – one from each parent. In other words, both parents must be carriers. Any child then has a 1 in 4 or 25% risk of developing the condition (two copies of the mutation); a 1 in 2 or 50% chance of being a carrier (one copy of the genetic mutation); and a 1 in 4 or 25% chance of not inheriting the genetic mutation.

A family tree – ‘autosomal recessive’ inheritance pattern

Generation



What if something is found in you?

If your doctor thinks that you may have ARVC, it is important that you have an assessment (tests) to find out whether you do have the condition. We describe  the tests that you may need to have as part of this assessment on page 00. Confirming a diagnosis will help the doctors to decide what treatment is best for you and how often you will need to be followed up. They will also be able to advise you on what you can do to help you to live a normal life.

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

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

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

What about screening for your family?

If you have ARVC, it is possible that other first-degree relatives may also either have the condition or be obligate carriers. (A first-degree relative means a parent, brother, sister or child.) All first-degree relatives should be provided with enough information to allow them to decide whether they wish to be screened for the condition. Screening usually involves having some tests done at a hospital as an outpatient. We describe these tests on page 00.

ARVC is a progressive disease. So, even if a person is found not to have the condition but other first-degree relatives do have it, or if the person has been identified as having the genetic mutation, he or she could still develop the condition in the future. Therefore the families of affected individuals may be asked to go for regular screening. How often they need to have screening will vary for each family. Screening  usually involves having an ECG and an echocardiogram. See page 00.

If someone with ARVC has had a genetic test that has found a particular genetic mutation to be the cause of the condition, it may be possible to test other family members to see if they have the same genetic mutation.

- If a person has a negative genetic test, this would mean that he or she does not have the same genetic mutation and would no longer have to go for screening, and that they could not pass on the condition to their children.
- If someone is found to have a genetic mutation, it is not possible to say whether they will develop the condition or if they are an obligate carrier. So they would be asked to go for regular screening and check-ups.

Assessment at a clinic for inherited heart conditions

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

Medical history

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

Medical examination and tests

Most people with ARVC don't have any visible physical signs of the condition. The doctor will take your pulse to assess for any abnormal heart rhythms and will ask you questions about how you feel when you are physically active. He or she will also ask you if you have ever had any symptoms such as unexpected light-headedness or fainting.

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

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

ECG **NON-INVASIVE**

Also called an **electrocardiogram**

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

Signal-averaged ECG **NON-INVASIVE**

This is a special type of ECG which detects certain electrical signals in the heart that are sometimes produced by people with ARVC.

Exercise test **NON-INVASIVE**

Also called an **exercise ECG**

This test is the same as the ECG described above, but is recorded before, during and after a period of time spent exercising on a treadmill or an exercise bike. This allows the doctor to examine any changes in the electrical patterns that occur with exercise, and analyse any abnormalities. In people with ARVC, strenuous activity can trigger abnormal heart rhythms. It is important for your doctor to know if this happens to you, so that they can treat any abnormal rhythms that they find.

24-hour or 48-hour ECG monitoring **NON-INVASIVE**

Also called **Holter monitoring**

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

Implantable loop recorder

This is a small device which is implanted, usually under the left collarbone, and can stay in place for several months. You are given a hand-held device which you press when you feel any symptoms such as light-headedness or syncope or if you feel palpitations. When you press the device, it stores the information about your heart rhythm. You will need to go to hospital as a day case to have the device implanted.

Echocardiogram **NON-INVASIVE**

Also called an **echo**

This test uses ultrasound waves to look at the structure of the heart. It produces a picture of the heart and allows doctors to measure the heart muscle and identify any abnormal areas. In particular, the doctors will look at how the heart muscle wall moves, whether there is any thinning of or bulges in the muscle wall, or if the walls of the ventricles have become stretched. Additional equipment, called Doppler ultrasound, can produce a colour image of blood flow within the heart and provide information on how well the heart is working.

Cardiopulmonary exercise test **NON-INVASIVE**

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

Electrophysiological study (EPS)

An electrophysiological study involves placing electrical leads inside the heart to analyse its electrical properties and to bring on arrhythmias. This test can be useful for diagnosing and treating abnormal heart rhythms. You will be asked to lie down on a special table and will be given a local anaesthetic in your groin. The doctor will then place fine tubes, called cardiac catheters or electrodes, into blood vessels in your groin. These are gently passed through to the heart.

Magnetic Resonance Imaging (MRI)

For this test, you lie in a short 'tunnel', around which there is a large magnet. Short bursts of magnetic fields and radio waves from the MRI scanner allow images of the heart to be created, processed and analysed. You must lie still while the scan is done. The whole test takes about an hour. An MRI is not painful or uncomfortable. However, some people with claustrophobia (fear of enclosed spaces) may find they cannot cope with having this type of scan. An MRI scan uses a magnetic field to create images of the heart.

An MRI is very good at showing the structure of your heart and blood vessels. It can also measure the flow of blood through the heart and some of the major arteries. An MRI scan can also show where the heart is working abnormally in conditions such as cardiomyopathy. And it can identify defects in the structure of the heart.

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

Radionuclide tests and CT scans

Radionuclide tests and CT scans give  detailed information than the exercise ECG test (see page 00). They are less common than electrocardiograms (ECGs) or echocardiograms, because the specialised equipment and staff are only available at some hospitals in the UK. There are two main types of tests. These are **myocardial perfusion scans** and **CT scans**.

Endomyocardial biopsy

In some cases, the doctors may decide to take a small piece of heart tissue and examine it under a microscope to see if there have been any changes in the structure of the heart muscle cells. However, in people with ARVC, not all areas of the heart muscle are affected. So, even if you have a negative result – that is, one that shows that the heart muscle is normal – it is still possible that other areas of the heart may be affected. This test is becoming less common as other tests develop.

For more detailed information about tests, and for information about levels of radiation involved in the tests, see our booklet *Tests for heart conditions*. (To order our booklets, see 'More information' on page 00.) 

Treatments for ARVC

ARVC is not curable, but most of the symptoms caused by the condition can be controlled by using medicines. Some people may need to have other treatments as well.  Give information about these medicines and treatments on pages 00–00. The need for and choice of treatment will be different for everyone. Your treatment may stay the same for many years, or may change more frequently.

The three main aims of treatment for ARVC are:

- to identify people at risk of sudden cardiac death and offer protective measures
- to prevent or control arrhythmias (abnormal heart rhythms)
- to improve the pumping action of the heart muscle.

Identifying and reducing the risk of sudden cardiac death

It is not possible to accurately predict if someone is at risk of sudden cardiac death. **Most people with ARVC are at low risk of sudden cardiac death.** However, if you have experienced any of the following, it may increase your risk:

- previous cardiac arrest
- episodes of syncope (fainting)
- recorded evidence of ventricular tachycardia on an ECG or exercise test
- ECG evidence of slow conduction of electrical impulses to the right ventricle
- stretching of the wall of the right ventricle and changes to the left ventricle.

There are certain times – known as ‘hot phases’ – when the disease process becomes more active, increasing the risk of sudden cardiac death. For example, unexplained dizzy spells, sustained palpitations, or blackouts can be a sign of a hot phase. If you experience any of these symptoms, you should see your doctor or go to an accident and emergency department at a hospital. Hot phases may come and go, but they could be a sign that the disease is progressing. This is why regular monitoring and follow-up with your doctor are important.

What can be done to reduce the risk of sudden cardiac death?

If you are at high risk of sudden cardiac death, your doctor may advise that you have an ICD fitted. ICD stands for implantable cardioverter defibrillator. An ICD consists of a very small box containing a battery, and special electrode leads. The box is inserted under the skin and attached to the heart by the electrode leads.

An ICD can be used to control abnormal heart rhythms (arrhythmias), but its main purpose is to identify any life-threatening arrhythmias and deliver an electrical shock to ‘reset’ the heart’s rhythm. Some people have described the shock as a feeling of having a ‘kick in the chest’.

An ICD is usually positioned under the chest wall muscle, below the left shoulder. The procedure to implant the ICD may take between one and three hours. Most people have a local anaesthetic as well as sedation, but some may have a full (general) anaesthetic. You will need to have check-ups at the ICD clinic once every three to six months. The battery lasts between four and eight years. When a new battery is needed, the box containing it can be replaced easily. For more on ICDs, see our booklet *Implantable cardioverter defibrillators (ICDs)*. (To order our booklets, see ‘More information’ on page 60.)

If you have been diagnosed with ARVC, you will be advised not to take part in activities that require a high level of endurance such as competitive sports. This is because there is a higher risk of arrhythmias happening during these activities and a higher risk of sudden cardiac death.

Preventing and controlling arrhythmias

The most common arrhythmias that occur in people with ARVC are described on page 60. We describe the different types of treatment below.

Medicines

Medicines are normally used to help prevent or treat arrhythmias. We describe the medicines most commonly used for people with ARVC on pages 60-60. The medicines your doctor prescribes for you will depend on your condition and symptoms.

Cardioversion and catheter ablation

As well as prescribing medicines, your doctor may decide to treat your arrhythmia using two other forms of treatment. These are cardioversion and catheter ablation. You may be advised to have one or both of these treatments.

Cardioversion

Occasionally a rhythm disturbance becomes continuous and does not respond to medication. If this happens, your doctor may recommend that you go into hospital for a treatment called cardioversion. You will be

given a light general anaesthetic and a controlled, low-dose electric shock is then applied across your chest. This can return the heart to its normal rhythm. You may need to have more than one cardioversion to treat the arrhythmia.

Some people may need to have an emergency cardioversion. This would be done if the rhythm disturbance came on suddenly or became severe, and seemed to reduce the pumping efficiency of the heart, causing a fall in blood pressure.

Catheter ablation

If your arrhythmia is not controlled with medication, or was not suitable for or has not responded to cardioversion, or if the side effects of the arrhythmia are unacceptable, your doctor may suggest that you have a catheter ablation. This procedure is similar to the EPS described on page 00. During the procedure, if the doctors can identify the electrical pathway in the heart that is causing the abnormal rhythm, they will use radio waves to destroy the pathway to try and prevent the abnormal rhythm from happening again. However, the arrhythmia may occur again in a different part of the heart muscle, and so the treatment may need to be repeated.

Pacemaker

A pacemaker may be inserted if you have heart block. Very occasionally a pacemaker may be used to help to control atrial fibrillation. For more on this, see page 00.

Improving the pumping action of the heart muscle

As ARVC progresses, more of the heart muscle is affected. This results in the wall of the heart muscle becoming weaker and the heart becoming less able to pump effectively. As the condition normally affects the right ventricle, this can lead to the symptoms of swollen ankles, legs or abdomen as described on page 00. However, if the condition also affects the left ventricle, the symptoms can include shortness of breath and pulmonary oedema (when fluid collects in the lungs). Medicines such as ACE inhibitors and angiotensin II receptor antagonists can help to improve

the pumping action of the heart (see page 00).

Pulmonary oedema is a serious condition and needs to be treated in hospital. Most people who have this condition will have severe breathing difficulties and need to be given oxygen. They will also need to have diuretics given by injection or through a drip. (See page 00 for more information on diuretics.)

Medicines used for people with ARVC

Many of the symptoms of ARVC can be controlled by using medicines. You may need to take one or more of the following medicines.

ACE inhibitors (angiotensin converting enzyme inhibitors)

ACE inhibitors are very effective in reducing and controlling blood pressure. In people with ARVC, they can help to control the symptoms of heart failure and prevent symptoms getting worse.

Unwanted effects – ACE inhibitors can cause a rapid fall in blood pressure, leading to episodes of feeling dizzy and fainting. Some people develop a dry, persistent cough, but if this happens your doctor can prescribe a different medication, called angiotensin II receptor antagonists, for you.

Angiotensin II receptor antagonists

Angiotensin II receptor antagonists act in a similar way to ACE inhibitors, but do not cause a cough.

Unwanted effects – Light-headedness.

Beta-blockers

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

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

Unwanted effects – Serious side effects are rare if beta-blockers are used carefully. Minor side effects are common but they tend to lessen as time

goes by. The minor side effects include tiredness, fatigue, and cold hands and feet. Other less frequent effects include feeling sick, diarrhoea, skin rashes, impotence, nightmares and dizziness.

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

The beta-blocker sotalol is often used to treat people with ARVC. It is usually well tolerated and effective, but it does carry the additional risk of possibly inducing an abnormal heart rhythm known as **Torsades de pointes**. Your cardiologist will decide if it is alright for you to take sotalol.

Anti-arrhythmic medicines

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

Amiodarone

Amiodarone is very effective in controlling disturbances of the heart rhythm, especially atrial fibrillation (see page 00).

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

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

Anticoagulants

Some people with ARVC develop atrial fibrillation (see page 00). Atrial fibrillation carries a risk of blood clots forming inside the heart. Blood clots are made up of platelets (tiny blood cells) clumped together, and a protein

called fibrin. Anticoagulants prevent fibrin from forming and so prevent clots from forming too. However, in doing so, they may cause internal bleeding or make bleeding from a minor injury worse. There are two main types of anticoagulants – heparin and warfarin.

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

Warfarin (or another oral anticoagulant) is given when long-term prevention of clotting is needed. This medicine is most often used for people with disease of the heart valves, especially those who have been given an artificial heart valve, or for some people who have an irregular heart rhythm such as persistent atrial fibrillation. Because the desired effect of anticoagulants is to make the blood thinner so that it doesn't clot so easily, you will need regular blood tests to make sure that the clotting activity of the blood is within safe but effective levels. At the start, these tests may be carried out almost every day, but later on they will be done only every four to eight weeks. If you have been taking warfarin for a long time and your condition is stable, you may be able to get a home-testing kit and do the tests yourself.

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

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

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

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

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

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

Diuretics

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

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

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

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

Unwanted effects – People with diabetes may find that diuretics raise their blood sugar. People with gout may find that diuretics make their condition worse. For more information about medicines see our booklet *Medicines for the heart*. (To order our booklets see 'More information' on page 00).

Treatment of other conditions that can occur as a result of ARVC

On page 00 we explained that certain conditions can occur as a result of ARVC.

Heart block

People who have heart block (when the electrical impulses that travel through the heart muscle are slowed down or completely blocked – see page 00) usually need to have a pacemaker fitted. However, not all types of heart block need to be treated.

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

A pacemaker controls the heart rate and stops any excessive slowing of the heart that could trigger an arrhythmia. The pacemaker is usually implanted just under your left collarbone. The procedure usually takes about an hour and is normally done with a local anaesthetic and sedation. You will need to have follow-up checks every 3 to 12 months. The pacemaker battery usually lasts between six and ten years (and sometimes even longer). When a new battery is needed, the box containing it can be replaced easily. For more information on pacemakers, see our booklet *Pacemakers*. (To order our booklets, see 'More information' on page 00.)

Heart failure

Heart failure is a condition that has four stages. Stage one is the mildest stage and stage four is the most severe. Your condition can be controlled with medicines and changes to your lifestyle. To find out more about the condition, see our booklet Heart failure. Or, if you have been diagnosed with heart failure, our guide The heart failure plan can help you to control and cope with  symptoms. (To order our booklets, see 'More information' on page 00.)

If you are at increased risk of sudden cardiac death

If your doctor thinks you are at increased risk of sudden cardiac death, you may be offered treatment which ould include taking medication, or having an ICD fitted (see page 00), or both.

Everyday life



Living with arrhythmogenic right ventricular cardiomyopathy (ARVC)

Who should be involved in routine medical care?

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

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

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



Exercise

Your heart is a muscle and needs exercise to stay healthy. However, people with ARVC may be at risk of sudden cardiac death if they exercise vigorously. Most people with the condition will be advised not to take part in competitive sports or other strenuous physical activity. This can be difficult for younger people who have previously taken part in this type of activity. You should always discuss with your doctor the type and level of physical activity that you can do.

Diet, alcohol and weight

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

Drinking more than the recommended amount of alcohol can damage your heart health and can lead to high blood pressure. Alcohol and caffeine can also stimulate arrhythmias. You can eat chocolate (which contains caffeine), drink coffee and alcohol, but in moderation. Alcohol is also a depressant, so it can make feelings of anxiety or depression worse. See our booklet *Keep your heart healthy* for information on sensible limits

for alcohol, or ask your doctor what is a safe limit for you. (To order our booklets, see 'More information' on page 00).

Although ARVC causes fat deposits to be laid down in the heart muscle to repair it, this is not caused by the fat in the foods you eat. See page 00 for an explanation of why this happens.

Weight gain

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

Smoking

Although tobacco smoking is not directly associated with hypertrophic cardiomyopathy, smoking will increase the risk of developing coronary heart disease and of reducing the flow of blood to your heart muscle. It also reduces the amount of oxygen being carried in the blood and makes your heart work harder. For information on giving up smoking see our booklet *Smoking and how to give up*. (To order our booklets see 'More information' on page 00).

Recreational drugs

Recreational drugs – such as cocaine, marijuana and ecstasy – act as stimulants. They increase the heart rate and blood pressure and can increase the risk of life-threatening arrhythmias and sudden cardiac death in otherwise healthy people. The risk for people with ARVC is even greater, so anyone with the condition should avoid using recreational drugs.

Other medications

Some over-the-counter medicines – such as those bought for coughs, colds and hay fever – contain pseudoephedrine. This acts as a stimulant and can increase the risk of arrhythmias, and it should therefore be avoided. You should always check with your pharmacist or doctor before taking any other medications, including herbal remedies.

Sex

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

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

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

Loss of sex drive is not uncommon in people with a heart condition. Some men may experience impotence. This may be the result of taking certain medicines, such as beta-blockers, which can affect your sex drive. Or it may be the result of the emotional stress you are feeling, or the result of poor blood circulation or diabetes.

Other common causes of loss of sex drive are:

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

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

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

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

General health

Do I need to have a flu vaccination?

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

What if I need an anaesthetic?

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

Anxiety and depression

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

Talking to someone can be very helpful. Friends and relatives may be able to understand how the condition is affecting you, but sometimes

it is easier to talk to someone who's not so closely involved. Talking to other people with cardiomyopathy may help you come to terms with your condition and help you to see that you are not alone. There are many organisations and charities that can help you to cope. See page 00 for details.

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

Depression

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

Having a routine and remaining active can help to deal with mild depression. It can help you to feel in control and get back to a natural sleeping pattern. Your doctor may suggest that taking sleeping tablets for a short time will help you. Although ARVC often recommends vigorous exercise, it shouldn't stop you being active. (See page 00 for more on physical activity.)

Alcohol is associated with a feeling of being happy but it is actually a depressant. You should not exceed the recommended levels of alcohol. See our booklet *Keep your heart healthy* for information on this, see your doctor. (To order our booklets see 'More information' on page 00).

If you have severe or chronic depression, you may benefit from taking

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

Driving

The UK Driver and Vehicle Licensing Agency (DVLA) gives the following advice for people with ARVC.

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

For more details, contact the DVLA on 0300 790 6801.

Work

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

For some occupations (such as joining the police or armed forces, becoming an airline pilot or heavy goods vehicle driver) and for some competitive sports, there are strict guidelines about whether you can continue in your job or sport if you have ARVC. Whatever your job or sport, your doctor may be able to provide additional information for your employer which could mean that you may be able to continue in your job or sport. If your employer (or potential employer) asks you to fill out a medical questionnaire, you must tell them about your condition.

Holidays and travel insurance

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

If you are diagnosed with ARVC, an insurance company may charge more for your travel insurance. For a list of insurance companies which are sympathetic to people with heart conditions, contact the British Heart Foundation  the Cardiomyopathy Association. Their contact details are on page 00.

Life insurance and mortgages

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

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

Financial support

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

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

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

Pregnancy and childbirth

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

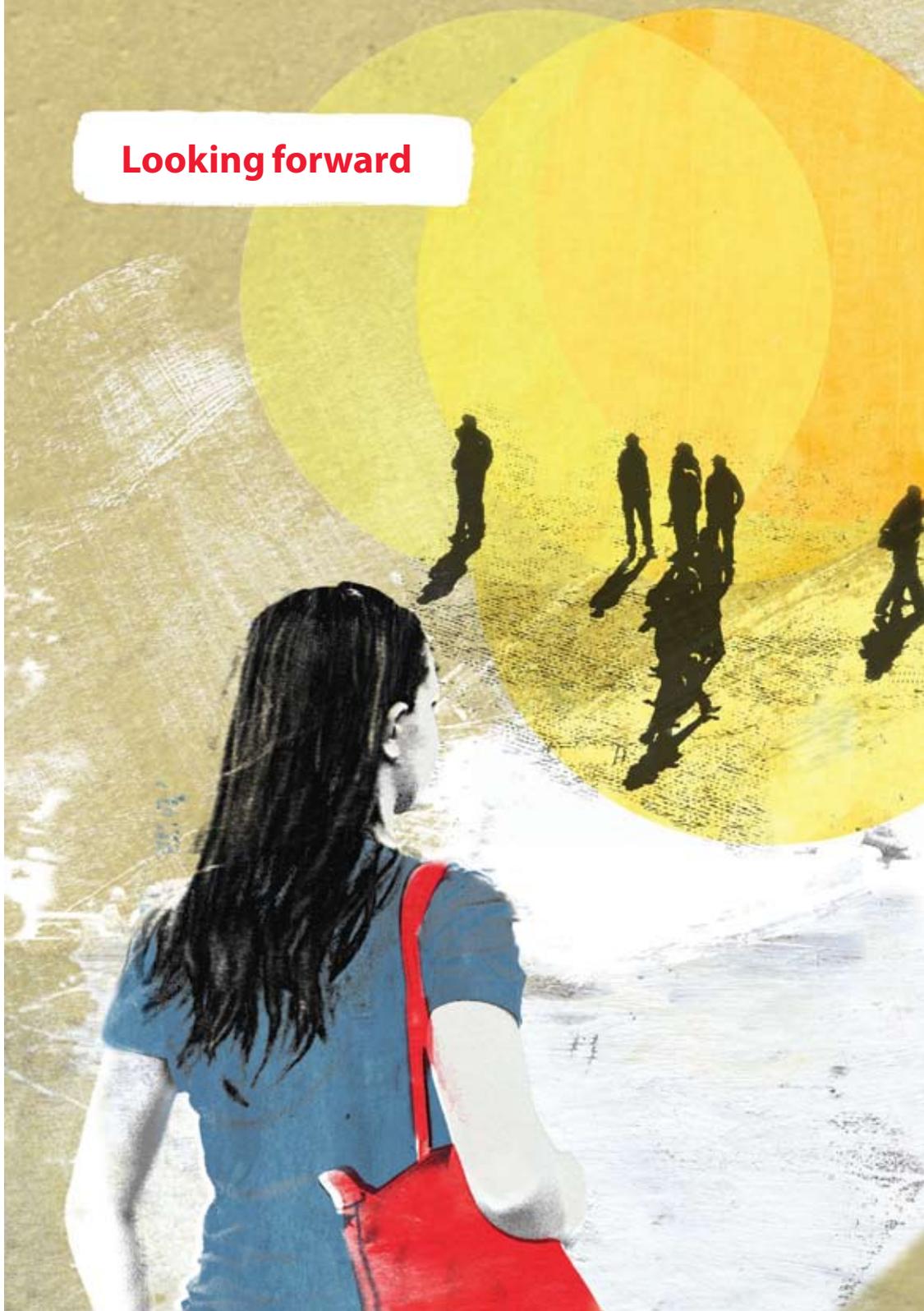
Severe morning sickness during pregnancy can lead to dehydration and chemical imbalance, which increases the risk of arrhythmias. It can also reduce the effectiveness of any medication that you are taking to control your symptoms. If your symptoms of morning sickness are severe, you should see your doctor.

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

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

Should I have children?

You will find information about the risk of passing on the condition to your children on page 00. The decision about whether to have children is one that you should make only after discussing it with your partner and at a clinic for inherited heart conditions. To find out where your nearest clinic is, call the **BHF Genetic Information Service** on **0300 456 8383**.



Looking forward

The future

Research is being carried out into many different aspects of Arrhythmogenic right ventricular cardiomyopathy (ARVC), in many countries.

Identifying the genetic mutations that cause this condition, and understanding more about the inheritance patterns, will help to identify more people who may be at risk of sudden cardiac death. This will allow doctors to treat those people and reduce this risk.

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

Basic scientific research is focused on the study of how the genetic abnormalities cause arrhythmia and heart failure. The research will also allow doctors to understand how and why the changes to the heart muscle happen and may help them to predict how the condition develops and progresses.

Technical terms

A

Anticoagulation

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

Aorta

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

Arrhythmia

A disturbance of the heart's rhythm.

Atrial fibrillation

A fast, irregular heart rhythm.

Atrium

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

Autosomal dominant inheritance

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

Autosomal recessive inheritance

Where a genetic mutation can be passed on through families but a person only develops the condition if they inherit two copies of the genetic mutation – one from each parent.

Autosomal inheritance

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

AV node

The point in the heart where the electrical impulses from the atria pass through to the ventricles.

C**Cardiac arrest**

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

Cardiologist

A doctor specialising in diseases of the heart.

Cardiomyopathy

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

Cardioversion

A procedure to restore a regular heart rhythm.

Chromosome

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

D**Diuretics**

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

Doppler ultrasound

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

E**Electrophysiological study (EPS)**

A technique for detecting and analysing abnormal heart rhythms.

EPS

See 'Electrophysiological study'.

G**Gene**

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

H**Heart block**

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

Heart failure

When the pumping action of the heart is inadequate.

Holter monitor

A 24-hour or 48-hour recording of an ECG (electrocardiogram).

I**ICD**

A metal electronic device, similar to a pacemaker, which can deliver an electrical shock to the heart to restore the normal heart rhythm.

Implantable cardioverter defibrillator

See 'ICD'.

Incomplete penetrance

An inheritance pattern where a gene mutation is passed on to a person in the next generation but the person does not develop the condition. This person may pass on the mutation to the next generation who may or may not develop the condition.

M**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.

Myocardium

The heart muscle.

O**Obligate carrier**

A person who is carrying a gene mutation but does not develop the condition. Their children may or may not inherit the genetic mutation, and may or may not develop the condition.

P**Pacemaker**

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

Palpitation

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

Pre-syncope

Symptoms of light-headedness and dizziness with a feeling of being likely to faint, but with no loss of consciousness.

S**Septum**

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

Sinus node

A collection of specialised cells found in the right atrium that creates electrical impulses that form the heartbeat. Sometimes known as the heart's natural pacemaker.

Syncope

Fainting or loss of consciousness.

T**Tachycardia**

A fast heart rate.

V**Ventricles**

The two bottom chambers of the heart.

Ventricular

From, or belonging to, the ventricle.



For more information

For information on your nearest clinic for inherited heart conditions

BHF Genetic Information Service

Greater London House
180 Hampstead Road
London NW1 7AW

Phone: 0300 456 8383
Website: bhf.org.uk

The BHF Genetic Information Service provides information, for families affected by an inherited heart condition, on where to go for an assessment. The service is staffed by specialist cardiac nurses and a bereavement counsellor.

BHF publications

Publications in the *Inherited heart conditions* series:

Hypertrophic cardiomyopathy

Dilated cardiomyopathy

Sudden arrhythmic death syndrome

Inherited heart rhythm disturbances

Other BHF publications

Atrial fibrillation

Heart failure

The heart failure plan

Heart rhythms

Implantable cardioverter defibrillators (ICDs)

Keep your heart healthy

Medicines for your heart

Pacemakers

Tests for heart conditions

Losing someone to heart disease

Offers help and support in coping with the loss of someone due to heart disease.

To order any of these booklets, call the BHF Orderline on **0870 600 6566**, or email orderline@bhf.org.uk, or visit bhf.org.uk/publications

For more on cardiomyopathy

Cardiomyopathy Association

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

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

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

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

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

Cardiac Risk in the Young – CRY

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

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

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

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

Index

acites
AF
angiogram
anticoagulants
arrhythmias
assessment
atrial fibrillation
autosomal dominant
autosomal recessive
biopsy
breathlessness
cardiac arrest
cardiac catheterisation
cardiopulmonary exercise test
chest pain
clinic for inherited heart conditions
CT scan
death
diagnosis
driving
dyspnoea
ECG
echocardiogram
electrocardiogram
electrophysiological study
EPS
everyday life
exercise test
family
future
heart block
heart failure
Holter monitoring
ICD
implantable loop recorder
incomplete penetrance
inheritance patterns
Magnetic Resonance Imaging
medicines
MRI
myocardial perfusion scan
normal heart
obligate carrier
oedema
pacemaker
palpitations
pedigree
pulmonary oedema
radionuclide tests
septum
shortness of breath
signal-averaged ECG
sports
sudden death
symptoms
tests
travel
treatment
ventricular fibrillation
ventricular premature beats
ventricular tachycardia
VF
VPB
VT
Work

About the British Heart Foundation

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

What you can do for us

We rely on donations to continue our vital work. If you would like to make a donation to the BHF, please ring our Supporter Services team on 0844 847 2787 or contact us through our website at bhf.org.uk/donate or send it to us at the address on the back cover. There are lots of other ways that you can help us, go online at bhf.org.uk to find out how.

Have your say

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

About the Cardiomyopathy Association

The Cardiomyopathy Association (CMA) is a national UK charity which provides information and support to families affected by cardiomyopathy and promotes excellence in clinical practice through education and direct funding. To learn more about the CMA please call 0800 0181024 or visit our website www.cardiomyopathy.org

GENETIC INFORMATION SERVICE

Information and support on inherited heart conditions



0300 456 8383

A local rate number



bhf.org.uk

Phone lines open 9am
to 6pm Monday to Friday

Cardiomyopathy Association

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

www.cardiomyopathy.org

Phone: 01494 791224

Registered charity number 803262



British Heart Foundation

Greater London House

180 Hampstead Road

London NW1 7AW

Phone: 020 7554 0000

Fax: 020 7554 0100

Website: bhf.org.uk

M11E 1209