

Arrhythmogenic Cardiomyopathy

An introduction to ACM or ARVC (Arrhythmogenic Right Ventricular Cardiomyopathy)

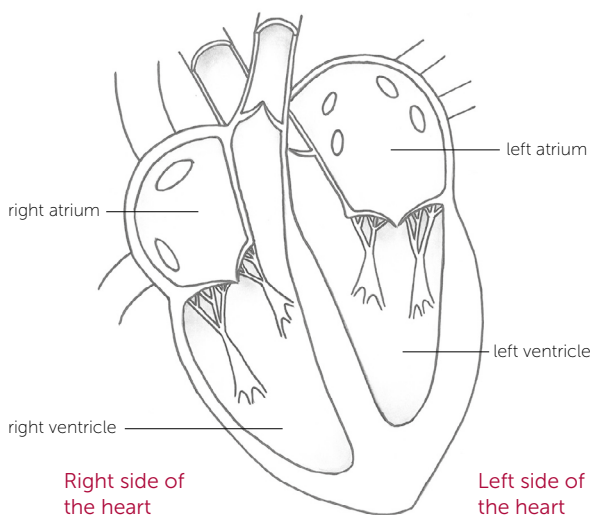
- A problem with the proteins that hold heart muscle cells together causes these cells to be lost and replaced by fibrous scar tissue and fatty cells, causing the ventricle walls to become thin.
- ACM can affect the electrical activity of the heart and causes arrhythmias.
- ACM is a genetic heart muscle condition which affects the right ventricle, left ventricle or both.

What is ACM?

Arrhythmogenic cardiomyopathy is a type of cardiomyopathy that affects the ventricles (lower pumping chambers) of the heart and may cause arrhythmias (abnormal heart rhythms). It can affect the right ventricle, the left ventricle or both. For this reason it is sometimes called arrhythmic cardiomyopathy (as the main symptoms are arrhythmias). It doesn't affect the atria (upper chambers) of the heart.

In ACM there is a defect in the proteins that join the cells of the heart muscle (myocytes) together. This means that the proteins do not develop properly and cannot keep the muscle cells together. When this happens the muscle cells detach and die, the area of the heart becomes inflamed, and the lost cells are replaced with fibrous scar tissue and fat deposits.

The structure of the heart



This affects the structure of the heart muscle, and it becomes thin and stretched. This can cause two main problems:

- the electrical pathways through the heart that coordinate the heart beat may be affected, causing arrhythmias;
- the thin walls of the ventricles of the heart are unable to pump blood as effectively as normal.

For more about the role of the heart see our factsheet 'About the heart'.

What causes ACM?

ACM is often a genetic condition (caused by an altered or 'mutated' gene) and can be inherited (passed from parent to child). However, the genetics of ACM are complicated.

ACM is often 'autosomal dominant'. This means that the mutated gene is found on one of the non-sex chromosomes (called the autosomes). A child of an affected parent will have a 50% chance of inheriting the gene mutation. In some cases it is 'recessive' (and it therefore only develops into the condition if both parents pass on the gene).

Who gets ACM?

ACM is rare compared to some other types of cardiomyopathy (such as dilated and hypertrophic cardiomyopathy). Around 1 in 10,000 people are thought to have ACM, although it is likely that there are more people living with the condition than this figure suggests.

What are the symptoms of ACM?

ACM can be a progressive condition, and in some people the symptoms may become worse over time. The symptoms are related to the electrical activity of the heart as well as the structure of the heart and how well it pumps.

Symptoms can include the following.

- **Palpitations (feeling your heart beating too fast, too hard or like it is 'fluttering')** – this is caused by arrhythmias (when the electrical messages which control the heart's rhythm are disrupted).
- **Light-headedness and fainting (loss of consciousness)** – reduced oxygen levels or blood



flow to the brain, due to arrhythmias, can cause light-headedness or dizziness and, in some cases, loss of consciousness.

- **Swollen legs, ankles and tummy** – build-up of fluid in the tissues, because the heart isn't pumping effectively, can cause swelling (called 'oedema').
- **Breathlessness (or dyspnoea)** – fluid builds-up around the lungs, making it harder to breathe.

Arrhythmias

Arrhythmias are caused by the disruption of the normal electrical signalling in the heart that controls the heart beat. This disruption causes a change in the heart's rhythm, which means it may beat too fast, too slow or erratically. Types of arrhythmias that can happen in ACM include:

- **Atrial fibrillation (AF)** – caused by disruption of the electrical messages that normally cause the heart muscle to contract. In AF the atria beat very quickly and are uncoordinated. This can make the flow of blood around the atrium 'turbulent', and the heart less efficient at pumping out blood. The erratic blood flow can increase the risk of blood clots forming, which can increase the risk of a stroke. Anticoagulant drugs are often prescribed to reduce this risk.
- **Ventricular premature beats (VPB)** – this is an extra heart beat which occurs when electrical impulse starts in one of the ventricles, and it contracts before it receives the normal signal via the atria. ACM can sometimes cause an increase in the number and frequency of these types of extra beats.
- **Ventricular tachycardia (VT)** – VT starts due to abnormal electrical activity in the ventricles, where the heart contracts abnormally quickly (over 100 beats per minute). It may lead to loss of consciousness due to the output of blood from the heart being reduced.
- **Ventricular fibrillation (VF)** – in VF the contraction of the ventricles is uncoordinated, and they 'quiver' rather than contract normally, so blood is not pumped out of the heart effectively. This condition is life-threatening and requires urgent treatment with a defibrillator (see treatment below).

What are the complications of ACM?

ACM can be serious as it can cause complications:

- **Heart block** – this is where the normal electrical activity that controls the heart beat is slowed or stopped, and stops the heart contracting normally. Heart block might require a pacemaker if the heart is unable to keep a normal rhythm.
- **Heart failure** – when the heart is not working effectively and it 'fails' to pump enough blood, at the right pressure, to meet the body's needs.

The term 'heart failure' describes a collection of symptoms caused by a heart that is struggling to work effectively, these include weight gain (due to fluid retention), shortness of breath, a cough, oedema, palpitations, dizziness and tiredness.

See our factsheet on heart failure

- **Sudden cardiac death (SCD)** – this can happen due to dangerous arrhythmias, such as ventricular fibrillation (VF), where the contraction of the ventricles is uncoordinated, and they 'quiver' rather than contracting normally, so blood is not pumped out of the heart effectively. Although it is relatively rare, if VF is not controlled (using a defibrillator to shock the heart back into normal rhythm), it can cause the heart to stop beating (a cardiac arrest).

How is ACM diagnosed?

ACM can be difficult to diagnose as the changes in the heart can be subtle and the fat deposits can be hard to see. Because the heart chambers can become enlarged, it can sometimes be misdiagnosed as dilated cardiomyopathy.

- **Medical history** – to look at symptoms and whether other family members have this condition (as it can be genetic).
- **Physical exam** – to check for any physical symptoms.
- **ECG (electrocardiogram)** – this looks at the electrical activity of the heart, recording the passage of electrical signals through the heart and whether arrhythmias (abnormal heart rhythms) are happening. A 'signal-averaged ECG' might be used to look for particular electrical signals that are common to ACM. An ECG might also be done during exercise.
- **Echo (echocardiogram)** – this is a type of ultrasound scan, which uses sound waves to create echoes when they hit different parts of the body. This looks at the structure of the heart and how it is working.
- **Exercise ECG tests** – these are tests done during exercise, such as on an exercise bike or a treadmill, to look at how the heart works during exertion (where it is under increased pressure to work) and measure oxygen consumption during exercise.
- **Holter monitoring** – this is when an ECG is recorded over a period of time (such as a few days), while carrying on with normal activities. The 'holter' is a ECG device which is worn on the waist or in a pocket which makes it possible to move and walk around. It can record any abnormal rhythms that may occur.
- **Electrophysiology study (EPS)** – this test involves having a fine catheter inserted into a blood vessel



at the top of the leg and fed up to the heart. Electrical signals are sent through the catheter to the heart which makes it beat at different rates, which is recorded. This can be used to find where in the heart arrhythmias are starting (and can be used to identify treatment options).

- **Implantable loop recorder** – this is a small device, implanted under the skin, that records the electrical activity of the heart to identify any arrhythmias. Implantable loop recorders can be in place for up to a couple of years, if this is required.
- **MRI (magnetic resonance imaging) scan** – this scan produces high quality images and is used to look at the structure and function of the heart.
- **Radionuclide test and CT test** – this involves having a radioactive dye (called a contrast dye) injected into a blood vessel. A CT scan, which uses x-rays, is done to produce images of the heart. The dye helps to see the structure of the heart and how blood flows around it.

How is ACM treated and managed?

Although there is no cure for ACM, treatments are used to help reduce and control symptoms and reduce the risk of complications. Treatment focuses on improving the pumping of the heart, controlling arrhythmias and reducing the risk of cardiac arrest.

Medication and devices

- **ACE inhibitors (angiotensin-converting enzyme inhibitors)** – relax the smooth muscle around the blood vessels to reduce the workload on the heart, and reduce the volume of the blood, making it easier for the heart to work.
- **Anti-arrhythmic medication** – reduces abnormal heart rhythms and helps to control the normal rhythm.
- **Anticoagulants (blood thinners)** – may be used in people with arrhythmias to reduce the risk of blood clots forming, which could lead to a stroke.
- **Angiotensin II Receptor Blockers (ARBs)** – dilate (enlarge) the blood vessels which helps to reduce blood pressure and may be used if the person is not able to tolerate ACE inhibitors.
- **Beta-blockers** – reduce the rate and force of the heart's contraction, by reducing stimulation of adrenaline (which would normally make the heart beat faster).
- **Diuretics (water tablets)** – reduce the build-up of fluid on the lungs or the ankles by encouraging the kidneys to get rid of water as urine.

- **Pacemaker** – may be recommended for people who have heart block (which makes the heart rate slow down). Pacemakers control the electrical signalling of the heart to keep a normal heart rhythm.
- **Cardioversion** – this is a when an electric shock is given to the heart under anaesthetic, to try and control arrhythmias (most commonly atrial fibrillation) and put the heart back into a normal rhythm. This is similar to what an ICD does (see below) but is a procedure done in hospital.
- **Catheter ablation** – this uses radio waves to treat areas of the heart where the electrical pathways cause arrhythmias. It stops the transmission of electrical signals that affect the normal heart rhythm.
- **ICDs (implantable cardioverter defibrillator)** – this may be recommended due to the risk of life-threatening arrhythmias. ICDs detect and correct any dangerous arrhythmias which could otherwise lead to a cardiac arrest.

Lifestyle management

In addition to medication and devices, there may be ways to reduce the effect of ACM through lifestyle. The following are examples of what might help:

- **Healthy eating** – a balanced diet can help to keep a healthy weight, which will reduce the impact on the heart as well as helping with general health.
- **Keeping a healthy weight** – as this can help to reduce any extra pressure on the heart and lungs.
- **Minimise alcohol** – alcohol can raise your heart rate and increase blood pressure. You may not need to completely avoid it, but keeping within recommended guidelines can reduce any potential affects.

The Chief Medical Officer reviewed these guidelines in 2016. You can read them at www.gov.uk and search 'alcohol guidelines'.

- **Minimise caffeine** – caffeine can raise your heart rate and increase blood pressure. As everyone is different in how they react to it, you might like to talk to your specialists about how to manage your caffeine intake.
- **If you smoke** – stopping smoking is important to help your overall health as well as your heart and lung function (as it can reduce oxygen levels in the blood as well as narrowing blood vessels). Your GP or an NHS stop smoking service may be able to help.



A note on exercise

Although exercise is often recommended for people with a heart condition, it can trigger arrhythmias and heart failure symptoms, and can be dangerous if the condition is unstable. Exercise for people with ACM needs to be considered carefully, and be part of a discussion between the individual and their cardiologist or specialist nurse.

See our factsheet on [Cardiomyopathy and Exercise](#)

We're here for you

At Cardiomyopathy UK we offer help and support for you and your family. We have information about each type of cardiomyopathy as well as diagnosis, treatment and lifestyle issues. Look on our website or call us for more information. Call our helpline to talk to our cardiomyopathy support nurses. We can put you in contact with other people affected by cardiomyopathy through our support groups, peer support volunteers and social media. Contact us for more about our services, or look online at www.cardiomyopathy.org

Send your feedback to contact@cardiomyopathy.org

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