

Hypertrophic cardiomyopathy

An introduction to hypertrophic cardiomyopathy or 'HCM'

- HCM is a condition where areas of heart muscle become thickened and stiff.
- The thickening makes it harder for the heart to contract and pump blood out to the body.
- Depending on where the thickening is, it can affect how blood flows out of the heart (referred to as 'HCM with obstruction' or 'HOCM').

What is HCM?

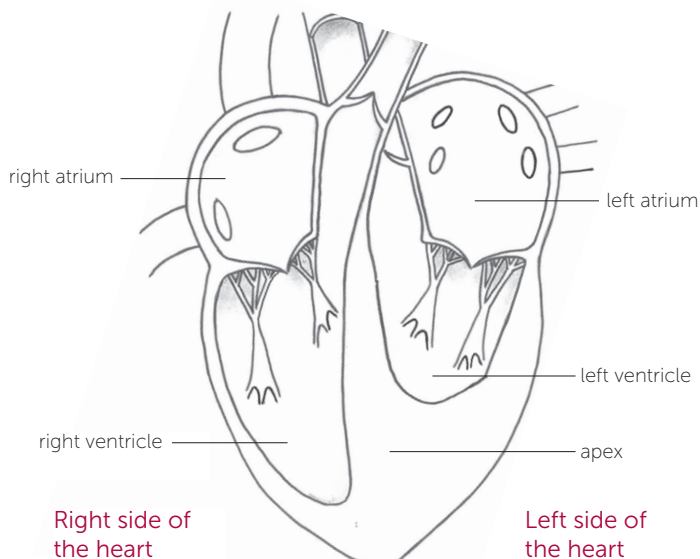
In HCM the muscular walls of the heart's ventricles (lower pumping chambers) become thickened. In the area of thickened muscle, the muscle cells can appear disorganised (or in 'disarray'). HCM generally affects the left ventricle (the main pumping chamber), and particularly the septum (area of muscle in the middle of the heart, which separates the right and left sides). However, it can also affect the right ventricle. The average thickness of the left ventricle wall in an adult is 12mm, but in HCM the thickness is 15mm or more.

The thickening of the muscle makes the ventricle stiffer, which makes it harder for the heart to relax and fill with blood, and contract to pump blood out. Depending on the area and amount of thickening, the volume of the ventricle (and how much blood it can hold) may be normal, or may be reduced (and hold less blood than normal).

Types of HCM?

Different types of HCM are described according to where in the heart the thickened area of muscle is. This also affects what symptoms someone might experience and what treatment is needed.

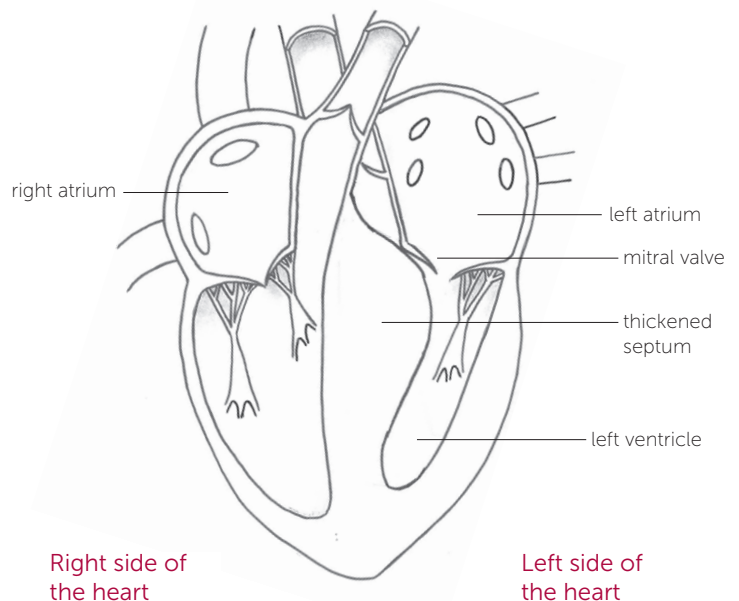
Apical hypertrophy – the thickened area is at the bottom (apex) of the heart. This usually reduces the volume of the ventricle.



Symmetrical hypertrophy – the thickening affects the whole of the left ventricle, reducing its volume. This is sometimes called 'concentric' hypertrophy.

Asymmetrical septal hypertrophy without obstruction – the thickening affects the heart's septum. 'Without obstruction' means that the thickening doesn't affect or restrict the flow of blood leaving the heart.

Asymmetrical septal hypertrophy with obstruction – the thickening affects the heart's septum. However, unlike the type above, the thickened area causes an obstruction that affects the flow of blood leaving the heart. When this happens, the ventricle has to work harder to pump out blood. The mitral valve can also be affected and is unable to close properly. This means that blood leaks back into the atrium (called mitral regurgitation) which can cause a murmur (sound that can be heard through a stethoscope).



How many people have HCM?

HCM is thought to be the commonest inherited cardiac condition, and affects between 1:200 - 1:500 people in the UK. It can affect adults and children, and affects men and women.



What causes HCM?

HCM is usually caused by a genetic mutation, and usually by a single mutation (rather than several mutations happening together). It is often inherited as an autosomal dominant genetic trait. 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 mutation. Mutations in different genes can affect when the signs of HCM develop. So although someone is born with the mutation, they may not develop the condition, or any symptoms, until teenage years or their early 20s. Some people have minor or even no symptoms. In some cases, the condition occurs later in life, known as 'late onset HCM'. Also, there are some genetic conditions, such as Anderson-Fabry disease, that can cause HCM.

For the reasons above, it is important that first-degree relatives (children, siblings and parents) of the person with HCM are also screened for HCM. This usually involves having an echocardiogram and an ECG (see page 3), under the care of a cardiologist. Genetic testing of the relatives may also be considered. This involves looking at *their* genes for the same mutation that causes HCM in their relative.

Although more than one person in a family can have the same genetic mutation, how it presents can vary (in where the thickening is and the severity of any symptoms it causes).

What are the symptoms of HCM?

There is wide variation in the symptoms of HCM, with some people having few or no symptoms, to people who have very severe symptoms.

Common symptoms 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).
- **Chest pain** – caused by reduced oxygen levels getting to the heart.
- **Dizziness or fainting** – caused by reduced oxygen levels or blood flow to the brain, most commonly due to arrhythmias.
- **Breathlessness** (or dyspnoea) – fluid builds-up around the lungs, making it harder to breathe.
- **Tiredness** – as the heart's function is reduced, less energy is delivered to the tissues, causing excessive tiredness.

Arrhythmias

HCM can cause arrhythmias (abnormal heart rhythms) as the disrupted muscle cells may affect the normal electrical signaling in the area known as the Sinus Node that controls the rhythmic pattern of the heart beat. This disruption causes a change in the heart's rhythm, and it beats too fast, too slow or erratically. These arrhythmias can include the following:

- **Atrial fibrillation (AF)** – caused by disruption of the electrical messages that normally cause the heart muscle to contract. In AF multiple electrical impulses are generated from the atrial walls, which causes them to beat very quickly and be uncoordinated. This can make the flow of blood around the atrium 'turbulent', and the heart less efficient at pumping out blood. Some of these impulses pass to the ventricles and can cause a typical fast and irregular heart rhythm. AF can increase the risk of blood clots forming, which can increase the risk of a stroke. It is important that most people with AF are on blood-thinning drugs to prevent this (see page 3)
- **Ventricular tachycardia (VT)** – VT starts due to abnormal electrical activity in the ventricle walls, where the heart contracts abnormally quickly (over 100 beats per minute). This fast rhythm can stop the heart from filling effectively with blood, which can lead to loss of consciousness.
- **Ventricular fibrillation (VF)** – the heart's system for regulating the heart beat is interrupted by multiple abnormal impulses coming from the ventricle walls. This causes the ventricles to contract in an uncoordinated way, and they 'quiver' rather than contract normally, so blood is not pumped out of the heart effectively. This condition is life-threatening as it can lead to a cardiac arrest and requires urgent treatment with a defibrillator.

A note on left ventricular outflow tract obstruction

Left ventricular outflow tract obstruction – or LVOTO – is when the thickened area of heart muscle obstructs the flow of blood leaving the left ventricle. Symptoms include fatigue, breathlessness and chest pain. Some people have symptoms when they are resting (not doing activities) but others only have symptoms when they are exercising (and the heart is under pressure).

What are the complications of HCM?

HCM can lead to additional complications.

- **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. It describes a collection of symptoms caused by a heart that is struggling to work effectively, such as weight gain (due to fluid retention), shortness of breath, a cough, oedema, palpitations, dizziness and tiredness.

♥ See our factsheet 'About heart failure'.



- **Stroke** – there is a small increased risk of a stroke in people who have AF. This is usually controlled with medication (see treatment below).
- **Sudden cardiac death (SCD)** – SCD risk factors are evaluated individually. These risk factors include non-sustained ventricular tachycardia, syncope, abnormal blood pressure response to exercise, family history of sudden death, and massive left ventricular hypertrophy. Having at least 2 of these risk factors will make you high risk. (see 'ICD guidelines').

A note on the progression of HCM

For some people with HCM, their condition progresses to a stage known as a 'burn-out phase'. This is when the thickened left ventricle starts to lose muscle cells and develops scarring, and it becomes dilated (enlarged). This can cause the heart to appear to have dilated, rather than hypertrophic, cardiomyopathy. Additional therapy might be needed to support the hearts function.

How is HCM diagnosed?

There are several tests that might be used to diagnose HCM, including the following.

- **Medical history** – to look at any previous symptoms and whether there might be other family members with this condition (as it can be genetic). This will often involve drawing the individual's family tree.
- **Physical exam** – to look for any signs of the condition and any other health problems.

The results of the medical history and physical exam determine which of the following tests are needed.

- **ECG** (electrocardiogram) – this looks at the electrical activity of the heart and whether arrhythmias (abnormal heart rhythms) are happening. An ECG might also be done during exercise. Many people with HCM have abnormal ECGs.
- **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. Tests look at how the heart works during exertion (under increased pressure to work), how your blood pressure responds to exercise, and measures your oxygen use 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. A 'holter' is an ECG device which is worn on the waist or in a pocket which makes it possible to move and walk around.
- **MRI** (magnetic resonance imaging) scan – this scan produces high quality images and is used to look at the structure and function of the heart and any presence of fibrous tissue within the heart muscle walls.

www.cardiomyopathy.org

Helpline 0800 018 1024 Mon-Fri 8.30am-4.30pm (Free from a landline, mobile costs vary)

How is HCM treated and managed?

When someone is diagnosed with HCM it is important to consider how it affects them individually, and their individual risk of having dangerous arrhythmias. Treatment aims to control symptoms and reduce complications. Some people may not need treatment.

Medication

- **Beta-blockers** – these are given to prevent arrhythmias and may also be used to reduce symptoms of obstruction.
- **Calcium channel blockers** – these reduce the force at which the heart contracts and reduce left ventricular outflow tract obstruction (see page 2).
- **Anti-arrhythmic medication** – these reduce abnormal heart rhythms and help to control the normal rhythm and help to improve symptoms in people with LVOTO.
- **Anticoagulants** (blood thinners) – may be used in people with arrhythmias to reduce the risk of blood clots forming, which could lead to a stroke.
- **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. Although these are not always given to people with HCM, they might be helpful in some cases.

Devices

Some people, when medication is not working or they have dangerous arrhythmias, might have an implanted device to reverse these arrhythmias. On rare occasions a pacemaker may be used to treat the symptoms of LVOTO (see page 2).

- **ICDs** (implantable cardioverter defibrillator) – this device detects and corrects any dangerous arrhythmias, which could otherwise lead to a cardiac arrest and sudden cardiac death.
- **Pacemaker** – pacemakers either send electrical impulses on demand, or continuously, to take over the electrical signaling of the heart and keep a normal rate.

ICD guidelines

The European Society of Cardiology developed guidelines to determine whether an individual should be considered for an ICD. This determines the level of risk of sudden cardiac death (SCD) for an individual. An individual's risk is calculated by considering a number of risk factors. Some of these factors are:

- their age;
- the thickness of their left ventricle;
- whether they have a family history of SCD;
- whether they have syncope (loss of consciousness) due to arrhythmias;
- whether they experience episodes of ventricular tachycardia (see arrhythmias);
- measurement of their left atria (one of the top chambers of the heart); and
- measurement of pressure of blood leaving the heart in LVOTO.



People at high risk of SCD will usually be offered an ICD, which will reduce the risk of SCD.

Surgery

Surgery for HCM involves reducing or removing the area of thickened heart muscle to reduce the obstruction and help the blood to flow through the heart. This is done using one of two methods.

- **Septal alcohol ablation** – this involves carefully injecting an alcohol directly into the area of thickening in the heart. This shrinks an area of the heart muscle directly around the injected site, and reduces the thickening. This is a very specialised form of treatment and isn't suitable for everyone.
- **Surgical myectomy** – this involves surgery to remove part of the thickened muscle to reduce the obstruction. This is a form of open heart surgery.

Heart transplant – a small number of people may have a transplant if their heart is in severe failure and not responding to treatment. This involves removing the failing heart and replacing it with a heart from a donor.

Lifestyle management

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

- **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'.*

- **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.
- **Minimising salt** – reducing salt intake can help to reduce water retention (which can cause swelling in the ankles and tummy) and blood pressure, which helps with heart function. Your specialist can give you guidance on your salt 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.

- **Minimise caffeine** – some people are more sensitive to the effects of caffeine than others, and it can cause palpitations in some people. For anyone who *is* sensitive to caffeine, limiting tea and coffee with caffeine, and avoiding items high in caffeine (such as energy drinks, strong coffee and high cocoa content chocolate), may be helpful.

If you are not sure whether caffeine might affect you, you might like to talk to your specialists about how to manage this.

A note on exercise

Exercise is often recommended for people with a heart condition. Moderate exercise can be important for people with HCM, depending on what symptoms they have.

If you have HCM it is important to talk to your doctors about what exercise is suitable for you. You might also like to ask whether you can have cardiac rehabilitation, which offers practical advice about exercise.

♥ *For more information about exercise see our factsheet '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.

Cardiomyopathy^{UK}

the heart muscle charity

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