Familial hypertrophic cardiomyopathy

What is familial hypertrophic cardiomyopathy?

Familial hypertrophic cardiomyopathy (FHC) is a condition in which part of your heart muscle (usually the main pumping chamber of your heart, called the ‘left ventricle’) is thicker than normal.\(^1\)

‘Familial’ means that this condition is inherited from one or both of your parents, and that it runs in your family.

The degree and distribution of thickening (called ‘hypertrophy’) varies. Hypertrophy mainly happens in the muscle between the right and left ventricles, known as the ventricular septum. In this case, the septum is usually the thickest part of the left ventricle and the term ‘asymmetric hypertrophy’ (ASH) is often used.

FHC may affect around 1 in 500 Australians (approximately 36,000).\(^2\)

What causes FHC?

FHC is a genetic condition, meaning that there is a mutation (a change) in one of the genes that was passed on to you from one of your parents.

Just as you have genes that determine your eye and hair colour, you also have genes that determine your heart muscle development and function. This gene defect affects the structure, size and function of your heart muscle.

We each have about 38,000 pairs of genes. Through the sperm or egg, each parent passes on a copy of one gene from each of their gene pairs to give their child a complete set (i.e. one gene copied from the father, one gene copied from the mother, to make a new pair of genes).

If a parent has a gene mutation that causes FHC, one gene in that gene pair will have the mutation and the other gene in that pair will be normal. Therefore there is a 50% chance of passing on the abnormal gene to each child.

If a child receives the abnormal gene, we say that they have ‘inherited’ hypertrophic cardiomyopathy – or FHC. Abnormal genes can be passed from generation to generation.

Genetic research over the last 10 years has identified at least 12 genes that contain mutations that lead to hypertrophic cardiomyopathy.\(^2\)

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\(^1\) ‘Hyper’ means greater than normal; ‘trophic’ means growth; ‘cardio’ means heart; ‘myo’ means muscle; ‘pathy’ means disease – so literally hypertrophic cardiomyopathy means a ‘disease where there is greater than normal growth of your heart muscle’.

How is FHC diagnosed?

Your doctor can do several things to find out if you have FHC, including:

- a thorough medical history, symptom history and family history
- a complete physical examination
- an electrocardiogram (a tracing of the electrical activity of your heart, also called ‘ECG’)
- an echocardiogram (an ultrasound scan of your heart that gives ‘pictures’ of your heart from which muscle wall thickness can be measured).

Sometimes clinical tests can’t give a definite diagnosis of FHC. In this case, your doctor will need to find evidence of left ventricular hypertrophy and rule out other known causes for it, such as high blood pressure, heart valve disease, advanced athletic training and some metabolic disorders.

Clinical screening for FHC can start from birth, but the age at which echocardiographic features may develop can’t be predicted. You should be tested regularly up to the age of 30 years (every three to five years in the first decade, and then every two to three years in the second and third decades). If you don’t show any signs of FHC by the age of 30 years, it is less likely that you have it. However, you should still have tests every five years up to the age of about 60 years.

The age for when hypertrophy develops in people with the gene varies. The most common time that FHC is visible by echocardiogram is the teenage years. Some adults may develop hypertrophy much later in life, or even not at all, even though they have the FHC gene.

Even though hypertrophy doesn’t develop in some people with the FHC gene, there may still be microscopic changes in their hearts. This means these people may still be at risk of the same complications experienced by people with FHC.

Your doctor can test for the genetic mutation that causes FHC. However, it may be different between families and can’t always be found. Genetic testing from a simple blood sample is still in the experimental stages, but may eventually become the standard for testing for FHC. Genetic testing can now also be done before birth at 10 to 12 weeks into a pregnancy.

If you are thinking about having genetic testing, talk to a genetic counsellor first.

What are the symptoms of FHC?

The symptoms of FHC can vary from none at all through to heart failure or premature, sudden death. They may be different for different people, and may even differ within the same family.

Common symptoms of FHC include chest pain, shortness of breath, palpitations, lethargy, dizzy spells and blackouts.

These symptoms may start at any age, and can occur after meals and during or just after exercise.

A small number of people with FHC experience significant symptoms and are at risk of sudden death.

Symptoms may occur for a number of reasons.
‘Outflow tract obstruction’ (or ‘hypertrophic obstructive cardiomyopathy’) is when the thickening (hypertrophy) of your heart muscle that occurs with FHC partially obstructs normal blood flow out of your heart. If blood can't flow out easily, there is increased pressure within your heart. Over time, the top chambers of your heart (the atria) may get bigger and cause a type of abnormal heart rhythm (or arrhythmia) called 'atrial fibrillation' (AF). AF doesn't always have symptoms, but may cause palpitations, chest pain, shortness of breath, dizziness and a blood clot in the atrium of your heart.

If you have FHC, there is a 20 to 40% chance that you may get AF with or without outflow tract obstruction.

Ventricular tachycardia (VT) occurs when hypertrophy of your heart muscle creates scar tissue between heart muscle cells. This scar tissue disturbs the normal electrical activity of your heart, causing a type of arrhythmia called 'ventricular tachycardia' (VT). VT causes your heart to beat very fast. It is a serious type of arrhythmia that can cause chest pain, shortness of breath, dizziness and blackouts.

In severe cases, VT can become ventricular fibrillation (VF), a very serious type of arrhythmia that causes a cardiac arrest. If not treated immediately, it can lead to sudden death.

About 10 to 20% of individuals with FHC are at increased risk of sudden cardiac death from VT or VF.

Reduced blood supply to the heart muscle occurs because more blood is needed to ‘feed’ the extra muscle in hypertrophied (thicker) heart muscle. If the blood vessels to your heart are not big enough to supply the extra heart muscle, you may have chest pains and arrhythmias.

How is FHC treated?

There is no cure for FHC, but there are medicines and surgical procedures that may reduce your symptoms and risk of the serious problems associated with FHC. Medicines and surgical procedures can also help you to lead a relatively normal life.

You will need to see your cardiologist regularly for a check-up and an echocardiogram. This will help your cardiologist to see if there are any changes in your heart and take action to prevent more changes.

Medicines

You might need to take one or more medicines to manage FHC, including:

- beta blockers and calcium channel blockers – to improve problems of 'stiffness' in the left ventricle of your heart, decrease the degree of hypertrophic obstructive cardiomyopathy, and reduce chest pain and shortness of breath
- anti-arrhythmic medicines – to treat or prevent arrhythmias
- anti-coagulants – to prevent clots forming in your heart, especially if you have AF.

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You should avoid taking diuretics. Diuretics reduce your body fluids, including blood volume, which may increase obstruction of the left ventricle of your heart.

**Surgical procedures**

If your symptoms aren’t reduced by taking medicine, your cardiologist may recommend that you have one or more of the following procedures:
- implanting a ‘bi-ventricular pacemaker’ – this may reduce the degree of obstruction and reduce your symptoms
- a ‘myectomy’ – this is an operation in which a small part of the hypertrophied muscle that is causing the obstruction is removed
- localised injections of alcohol into parts of your heart muscle via your coronary arteries (the arteries that supply blood to your heart) – this thins your heart muscle and may reduce the obstruction.

**Heart transplant**

In rare cases, the hypertrophied heart muscle can thin over time and the left ventricular chamber may enlarge. If this happens, your heart may stop pumping properly and you may eventually get chronic heart failure. If you have severe chronic heart failure that doesn’t respond to medicine, you may need a heart transplant.

**Electrical cardioversion**

Some arrhythmias may not respond to medicines, so your heart may need a small electrical shock (an ‘electrical cardioversion’). Electrical cardioversion is done under a general anaesthetic and helps your heart rhythm return to normal.

**Implantable defibrillators**

If you have a serious arrhythmia, your cardiologist may recommend that you get an implantable defibrillator. An implantable defibrillator is about the size of a pacemaker and delivers a small electrical shock inside your heart to help return your heart rhythm to normal.

**Can lifestyle changes help to reduce FHC?**

Making some simple lifestyle changes may help to reduce your risk of serious FHC symptoms.

- It’s important that you talk to your cardiologist about the type of physical activity that is safe for you to do. You will need to avoid intense, strenuous physical activity, because it may lead to blackouts and, in rare cases, sudden death. Types of physical activity to avoid include competitive sports, such as squash and football; endurance sports, such as marathon running; and isometric exercise, such as weight lifting.

- Try to do regular, gentle physical activity every day – but check that it’s safe with your cardiologist first.

- Avoid taking very hot baths or showers, standing for long periods in hot weather, and becoming dehydrated from vomiting or diarrhoea. These things may cause a drop in your blood pressure, which may lead to light-headedness or blackouts.

- Enjoy healthy eating and achieve and maintain a healthy body weight.

- Be smoke-free – don’t smoke and avoid other people’s smoke (second-hand smoke).
What other tests may be performed?

Your cardiologist can do tests to find out what is causing your FHC symptoms, and if you are at high risk of blackouts or serious arrhythmias.

There is no precise single way to find out who will have severe symptoms. However, information from all of the tests described here may help your cardiologist to work out if you are at high or low risk of severe symptoms.

- **Holter monitoring** is a continuous period of ECG recording over 24 to 72 hours. Your heartbeat and heart rate are measured during normal daily activities and abnormalities not seen on a standard ECG may be detected. Event monitoring may be used to record the ECG over a longer period of time (up to weeks) if you have intermittent symptoms.

- **Exercise testing** (monitoring your heart while you walk on a treadmill or pedal a bicycle) helps to detect arrhythmias that happen when you are doing physical activity. Exercise testing may also pick up abnormal blood pressure responses to physical activity. It is also used to check if medicines help to relieve symptoms.

- **An ultrasound or echocardiogram** provides 'pictures' of your heart, and measures your heart muscle's wall thickness and the pumping function of its left ventricle.

- **Cardiac catheterisation and coronary angiography** show your cardiologist how narrow your coronary arteries are. During coronary angiography, you will be given a local anaesthetic and then your cardiologist will insert a fine tube ('catheter') into your groin, arm or wrist and guide it to your heart. Dye is injected into your coronary arteries so that special X-rays can be taken of your arteries and heart. Dye is also injected into the left ventricle of your heart to show your cardiologist if your heart is pumping properly. These tests may help your cardiologist to find out if chest pain (angina) is being caused by FHC or another disease, like coronary heart disease.

- **A ‘pacing study’** is done to see if a dual chamber pacemaker will relieve your symptoms. Your cardiologist will insert a temporary pacemaker wire into your heart using similar procedures and equipment to those used in cardiac catheterisation. Your heart will be artificially paced at different intervals, and measurements will be made to see if the obstruction is reduced.

- Other tests that are performed less often include:
  - **electrophysiology studies** – to check your heart's electrical activity in more detail
  - **radionuclide studies** – to check if your heart is pumping properly and how blood flow is being distributed to your heart.

What about my family?

If you have FHC, your immediate family (parents, siblings and children) should be tested by a cardiologist to see if they also have FHC – even if they don’t have any symptoms.

A person with FHC won't necessarily have the same symptoms as other members of their family with FHC.

Some relatives may choose not to be tested for a variety of reasons. This decision should be respected. Because not everyone shows symptoms of FHC, you can have it and still lead an
active, physically demanding life. However, it’s good to find out if you have it so that you can take steps to prevent it getting worse and causing serious problems in the future.

Before deciding whether or not to be tested for FHC, you and your family should talk to an experienced health professional who has knowledge of FHC and the implications of a negative or positive result.

**What else should I be aware of?**

**Medical and life insurance**

Talk to your insurance company about whether or not they provide coverage for people with FHC and if FHC affects the costs of premiums.

**Employment**

If you have FHC, you may not be able to do some types of jobs, because they are too strenuous or risky. This includes jobs as commercial airline pilots and some defence force jobs. Talk to your doctor if you are not sure if your job is suitable for you.

Some employers may ask you to undergo a medical examination and to disclose your medical history before they will accept you for employment.

**Resuscitation training**

Anyone with a heart condition is at a higher risk of suffering a cardiac arrest than someone with a normal heart. Therefore, if you or someone in your family has FHC, it’s important that you and your family know how to do cardiopulmonary resuscitation (CPR).

Call our Health Information Service on 1300 36 27 87 for more information about CPR.

**Antibiotics**

Tell your doctor and dentist that you have FHC, and remind them about it before you have any surgical or dental procedures.

Your doctor and dentist may prescribe prophylactic antibiotics for you to help to prevent bacteria from entering your bloodstream and causing an infection in your heart (endocarditis).

**Pregnancy**

Many women who have FHC have no problems during pregnancy and labour, but every case is different and should be assessed individually.

Epidural anaesthetics can sometimes cause a sudden drop in blood pressure, so you may need to consider alternative pain relief.

Some medicines for FHC may also cause problems in the foetus. If you have any concerns, talk to your doctor before you become pregnant.
Medicalert

If you collapse away from home and on your own, it is helpful to have information about your condition and treatment, and the name of your doctor. A 'Medicalert' bracelet or pendant has a contact number on it that health professionals and police can call to get this information if you can’t provide it.

Further information

If you think you might have FHC, talk to your doctor or a cardiologist.

If you or a family member have FHC and need specific advice and support, contact:

Cardiomyopathy Association of Australia
Phone: 1300 552 622
Email: info@cmaa.org.au
Website: www.cmaa.org.au.

If you'd like to know more about FHC or how to improve your heart health, call our Health Information Service on 1300 36 27 87 (for the cost of a local call) or email health@heartfoundation.org.au.

The Heart Foundation would like to thank Ms Julie French, the principal author of this document, who was supported in this task by many clinicians, scientists and families involved with the HCM Clinic at the Royal Prince Alfred Hospital, Sydney.