CONDITION

Hypertrophic cardiomyopathy

Hypertrophic cardiomyopathy is a condition in which the heart muscle becomes unusually thick.

WHAT IT IS
Abnormal thickening of the heart muscle

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Breathlessness, chest pain, palpitations, fainting

WHAT CAUSES IT
Usually inherited, sometimes unknown

HOW IT IS DIAGNOSED
Heart ultrasound or cardiac MRI

MAIN TREATMENT
Medicines, heart procedures, and implanted defibrillators

CAN IT BE CURED?
No - treatment controls symptoms and risks

IS IT AN EMERGENCY?
Sometimes - fainting or severe chest pain

LONG-TERM OUTLOOK
Many live well with monitoring; risks differ by heart findings

Overview

Hypertrophic cardiomyopathy, or HCM, is a condition in which part of the heart muscle becomes unusually thick. The thickening most often affects the wall between the heart's two lower chambers, called the septum.

The thick muscle may be stiff, making it harder for the heart to relax and fill with blood. In some people, it also narrows the path through which blood leaves the heart. This is called obstructive HCM. HCM without this narrowing is called non-obstructive HCM.

HCM affects people differently. Some have no symptoms and learn about it during a heart test or family screening. Others have breathlessness, chest discomfort, fainting, or abnormal heart rhythms.

The condition matters because it can occasionally lead to heart failure, stroke, or a dangerous heart rhythm. However, most people are not at high risk of sudden death. Regular assessment helps identify the treatment and monitoring appropriate for each person's findings.

Symptoms

HCM may cause no symptoms, especially early in life. Symptoms can begin at almost any age and do not always match the amount of muscle thickening.

Possible symptoms include:

Breathlessness can happen because a stiff heart does not fill normally, raising pressure in the heart and lungs. Chest discomfort may occur when the thickened muscle needs more oxygen than its blood supply can provide.

Palpitations may come from extra beats or an abnormal rhythm. One important rhythm is atrial fibrillation, in which the upper chambers of the heart beat irregularly. It can increase the chance of a blood clot and stroke.

These symptoms also occur with many other conditions. Tests are needed to find the cause.

- Shortness of breath, particularly during activity
- Chest pain or pressure, often with exertion
- A racing, pounding, fluttering, or irregular heartbeat
- Dizziness or light-headedness
- Fainting, especially during or soon after exercise
- Unusual tiredness or reduced ability to exercise
- Swelling of the ankles or legs

Causes and risk factors

HCM is usually caused by a change in a gene that affects proteins inside heart muscle cells. It commonly runs in families. A child of someone with a disease-causing HCM gene change may inherit that change, although the severity and age of onset cannot be predicted from inheritance alone.

Some people with HCM have no identified gene change or family history. A negative genetic test does not always rule out an inherited cause because not every relevant gene change is known or detectable.

Having a parent, sibling, or child with HCM increases the chance of having the condition. Close relatives may be offered heart screening even when they feel well.

Other conditions can also cause a thickened heart muscle, including long-standing high blood pressure, heart valve problems, and certain metabolic or storage conditions. Athletic training can enlarge or thicken the heart in a different way. Part of diagnosis is checking whether another explanation fits better.

Diagnosis

An echocardiogram is the main test used to diagnose HCM. This ultrasound scan shows the thickness of the heart walls, how the valves move, and whether blood flow out of the heart is narrowed. Testing may be repeated during exercise or while standing if narrowing is suspected but not present at rest.

An electrocardiogram, or ECG, records the heart's electrical activity. It can show patterns associated with HCM or an abnormal rhythm, but an ECG alone does not confirm the condition.

Other tests may include:

A diagnosis includes more than confirming thickened muscle. Assessment also looks for obstruction, valve leakage, abnormal rhythms, and features linked to sudden cardiac death. These findings guide follow-up and treatment.

If a disease-causing gene change is found, close relatives can be tested for that specific change. When genetic testing is negative or not performed, relatives may still need periodic ECGs and echocardiograms. The timing depends on age, family history, and previous results.

- Cardiac MRI for detailed pictures of the muscle and areas of scarring
- A portable heart monitor to record rhythms over one or more days
- An exercise test to assess symptoms, blood pressure, and rhythm
- Blood tests to look for other causes of heart thickening
- Genetic testing when an inherited cause is suspected

Treatment and management

Treatment aims to reduce symptoms, improve blood flow, manage abnormal rhythms, and lower the risk of serious complications. People without symptoms may not need medicine, but they still need periodic review.

Medicines can slow the heartbeat and help the heart fill more effectively. Beta blockers like metoprolol or propranolol are often used. Some people receive certain calcium-channel blockers or other medicines that reduce obstruction and symptoms.

A cardiac myosin inhibitor may be considered for some adults with symptomatic obstructive HCM. This type of medicine reduces excessive contraction of the heart muscle. It requires regular echocardiograms and careful review of medicine interactions because it can weaken the heart's pumping ability. It can also harm a developing pregnancy.

If severe obstructive symptoms continue despite medicine, a procedure may reduce the thickened septum. Septal myectomy is surgery that removes part of the muscle. Alcohol septal ablation uses alcohol delivered through a small blood vessel to create a controlled reduction in part of the septum. These procedures are usually planned through a centre experienced in HCM.

An implantable cardioverter-defibrillator, or ICD, may be recommended when particular findings suggest a higher risk of a life-threatening rhythm. An ICD monitors the heartbeat and can deliver treatment if such a rhythm occurs. It is not routinely needed by everyone with HCM.

Atrial fibrillation may require rhythm or heart-rate treatment. Blood-thinning medicine is commonly used to reduce stroke risk when HCM and atrial fibrillation occur together.

Management also includes reviewing other medicines and conditions that affect blood pressure or fluid balance. Becoming dehydrated can worsen obstruction and symptoms in some people, but fluid advice may need adjustment if heart failure is present.

Medicines commonly used for this include aficamten and mavacamten . Which one suits you, if any, is a decision for you and your doctor.

Complications

Most people do not develop every complication. Possible problems include:

Risk is not judged from one feature alone. Previous cardiac arrest, certain episodes of fainting, family history, heart thickness, pumping function, heart rhythm recordings, and MRI findings may all contribute to an individual risk assessment.

- Atrial fibrillation and blood clots that can cause a stroke
- Heart failure due to poor filling or, later, weaker pumping
- Leakage through the mitral valve
- Ventricular tachycardia or ventricular fibrillation, which are dangerous rhythms

- Sudden cardiac death, which is uncommon but important to assess
- Ongoing chest pain, breathlessness, or limited exercise capacity

Prevention

Inherited HCM cannot currently be prevented. Healthy habits do not remove the gene change or reverse established muscle thickening.

Early detection can help prevent or reduce some complications. This includes attending follow-up, taking prescribed medicine, reporting new symptoms, and completing recommended rhythm or imaging tests.

Family screening can find HCM before symptoms appear. It does not prevent inheritance, but genetic counselling can explain how the condition may be passed on and discuss reproductive options without directing personal decisions.

Smoking avoidance and management of blood pressure, cholesterol, diabetes, and weight support overall heart health. These measures are worthwhile, but they do not replace HCM-specific care.

Living with it and what to expect

HCM is usually a lifelong condition. The amount of thickening may remain stable for years or change over time. Symptoms may also stay mild, appear later, or become more noticeable. Regular follow-up is needed even when you feel well.

Many people with HCM continue to work, study, travel, and have active family lives. Some need adjustments because of breathlessness, rhythm problems, medicines, or an ICD. Others have few day-to-day limitations.

Physical activity is usually encouraged rather than avoided completely. Recreational exercise of mild to moderate intensity is suitable for many people. Decisions about vigorous or competitive sport require an individual assessment because heart findings, symptoms, rhythm history, and the type of activity all matter. Sudden restriction from all exercise can have its own physical and emotional costs.

Pregnancy is tolerated by many people with HCM, but heart symptoms and medicines should be reviewed before pregnancy when possible. Care may involve both cardiology and maternity teams. Some HCM medicines are unsuitable during pregnancy.

Follow-up commonly includes discussion of symptoms, an ECG, heart imaging, and periodic

rhythm monitoring. The schedule is based on age, symptoms, previous results, and treatment. New symptoms should be reported rather than waiting automatically for the next routine appointment.

Living with an inherited heart condition can also bring worry about exercise, children, or sudden cardiac death. Clear risk assessment and family screening can reduce uncertainty. Psychological support or an HCM patient group may also be useful.

When to get medical help

Arrange a routine medical review if you develop new mild breathlessness, reduced exercise tolerance, recurring palpitations, ankle swelling, or chest discomfort. Also seek review if established symptoms are becoming more frequent or medicines are causing problems.

Contact your cardiology team promptly after an unexplained fainting episode, particularly if it happened during exercise. New sustained palpitations or a newly irregular pulse also need timely assessment.

Call emergency services for:

If someone collapses and is not breathing normally, call emergency services, begin CPR if able, and use an automated external defibrillator if one is available.

- Severe or persistent chest pain
- Severe difficulty breathing
- Fainting with chest pain, palpitations, or exercise
- A fast or irregular heartbeat with marked weakness or breathlessness
- Signs of stroke, such as facial drooping, arm weakness, or speech difficulty
- Collapse, unresponsiveness, or absent normal breathing

Common questions

Is hypertrophic cardiomyopathy fatal?

It can cause life-threatening rhythms, but sudden death is uncommon for most people with HCM. Risk is higher with certain personal, family, imaging, and rhythm findings. An ICD can reduce the risk of death from dangerous rhythms in people selected as being at increased risk.

Can hypertrophic cardiomyopathy go away?

HCM does not usually go away. Medicines and procedures can improve symptoms and blood

flow, but they do not remove an inherited tendency. Septal reduction can remove or shrink part of the thickened muscle without curing the underlying condition.

Can I exercise with hypertrophic cardiomyopathy?

Many people can exercise safely, and complete inactivity is not routinely advised. The suitable type and intensity depend on symptoms, obstruction, rhythm history, treatment, and personal goals. Vigorous or competitive exercise needs a specialist discussion rather than a single rule for everyone.

Can I pass HCM to my children?

Many forms of HCM follow an autosomal dominant inheritance pattern. This means each child of a person with the relevant gene change has a 1 in 2 chance of inheriting it. Inheriting the change does not predict when HCM will appear or how severe it will be.

Is HCM the same as an athlete's heart?

No. Intensive training can cause a healthy increase in heart size and sometimes wall thickness, known as athlete's heart. HCM involves abnormal heart muscle and may include scarring, obstruction, or characteristic family and genetic findings. Imaging and exercise history help distinguish them.

Can I drink alcohol if I have HCM?

Alcohol may trigger palpitations, contribute to dehydration, or worsen atrial fibrillation in some people. It can also interact with medicines. Advice should reflect your symptoms, heart rhythm, treatment, and any heart failure.

Sources

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