Patient Information

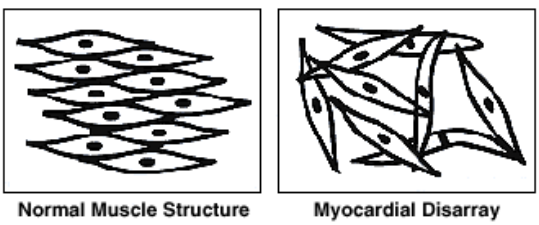
Hypertrophic Cardiomyopathy

1. The Normal Heart

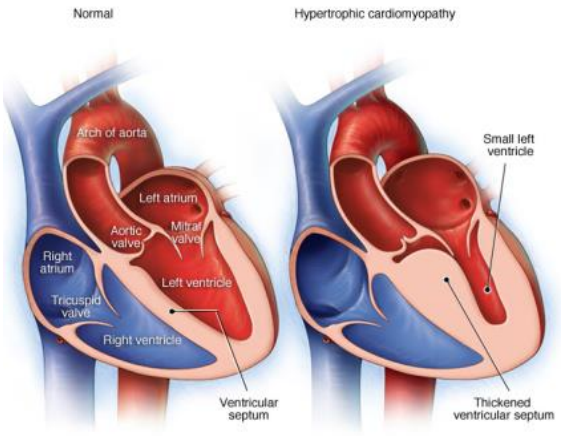
The heart is a special muscle that contracts regularly and continuously, pumping blood to the body and the lungs. It has four chambers – two at the top (the atria) and two at the bottom (the ventricles). The pumping action of the heart is caused by a flow of electrical signals through the heart. These electrical signals repeat themselves in a cycle and each cycle causes one heartbeat.

2. Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is a disease of the heart muscle. Having HCM means that the heart muscle can become excessively thick, most commonly in the interventricular septum (i.e. part of the heart muscle between the left and the right ventricle). How thick the muscle is, and how much of the muscle is affected, can vary from person to person. The left ventricle is almost always affected, and in some individuals the muscle of the right ventricle also thickens. In some patients the thickening of the heart muscle causes an obstruction for the blood flow out of the heart; in this situation the term Hypertrophic Obstructive CardioMyopathy (HOCM) is used. In a normal heart, the cells that make up the heart muscle lie in smooth, straight lines, as shown in the left picture below. In contrast, in patients with HCM, the cells lie in disorganized, jumbled layers (known as 'myocardial disarray'), as shown in in the right picture below. The heart muscle can also become progressively stiffer, making it more difficult for the heart to pump.



Normal Muscle Structure | Myocardial Disarray



Source: with permission from Mayo Clinics

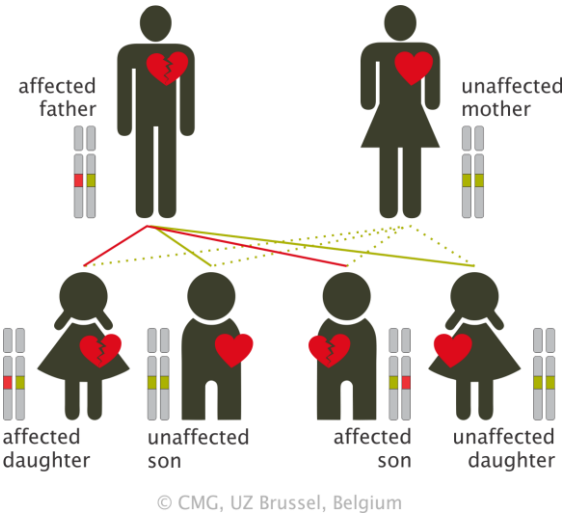
3. Prevalence & Inheritance

Around 1 in every 500 persons has HCM; this is the prevalence of the disease. HCM often has a genetic cause. This means that it is caused by a defect (a mutation) in one or more genes that can be passed on through families.

A gene is part of our DNA which contains a code for making a molecule (a protein). HCM is caused by a mutation in the genes that contain codes for specific molecules (proteins) in the heart. Every person has two copies of each gene that can be linked to HCM. A mutation in only one of the two copies of one of these genes (from the father or from the mother) is enough to develop HCM. This is called an autosomal dominant disorder and a parent who carries it has a 50% (1 in 2) chance of passing the mutation to each child.

A person affected with an autosomal dominant disorder has a 50 percent (1 in 2) chance of passing the mutated gene to each child. The chance that a child will not inherit the mutated gene is also 50 percent. In some cases, a new (de novo) mutation can occur in the egg or sperm cells or in an embryo. In these cases, the child's parents do not have the mutation and HCM, but the child does

have HCM and can pass the mutated gene to his or her own children.



Autosomal dominant inheritance

4. Symptoms

Most people with HCM have no symptoms, or have a stable condition throughout adult life. A few individuals develop serious symptoms and some persons find that their symptoms become worse over time. This may be because they develop arrhythmias (abnormal heart rhythm) or because their heart muscle becomes progressively stiffer making it more difficult for the heart to pump. The most common symptoms are shortness of breath (dyspnoea), chest pain, palpitations (due to arrhythmias), light-headedness and blackouts.

5. Diagnosis

The most common tools to make the diagnosis of HCM are the medical and family history, physical examination, a heart electrical tracing (the electrocardiogram or ECG), a heart ultrasound scan (echocardiogram), exercise testing, heart rhythm monitor (Holter), and a heart magnetic scan (magnetic resonance imaging or MRI) (*see below*).

5.1 ECG (electrocardiogram)

This is the most basic test. Small sticky patches (electrodes) are put onto the chest and sometimes to arms and legs. These are connected by wires to an ECG recording machine, which picks up the electrical activity for a few seconds that makes the heartbeat. Sometimes additional or repeated ECG-tests are necessary.

5.2. Echocardiogram (echo)

Echocardiogram uses ultrasound waves to look at the structures of the heart. An echocardiogram can detect different types of structural changes in the heart, for example heart muscle diseases like ARVC and heart valve abnormalities. Areas of thinning of the heart muscle can be identified.

5.3. Exercise test (stress test)

Exercise test is the same as the ECG described above, but is recorded before, during and after exercising on a treadmill or an exercise bike. This records any changes in the electrical patterns that occur with exercise.

5.4. Holter monitoring

Holter monitoring involves a small digital machine, which can be worn on a belt round the waist. Four or six ECG electrodes from the machine are taped to the chest. It then records the electrical activity of the heart for 24-48 hours, or for up to seven days. During the monitoring all activities are listed in a 'diary'.

5.5. MRI

An MRI scan uses a magnetic field to create images of the heart. The scanner itself is a large tube with a table in the middle, allowing the patient to slide into the tunnel. The test takes about one hour. An MRI is very good at showing the structure of the heart and blood vessels, showing the condition of the heart muscle, and identifying any scarring (fibrosis) within the heart.

5.6. Genetic testing

In over half of the HCM families, a mutation can be found in one of the genes that can cause HCM. Because not all genes that can cause HCM are known, a negative result of the genetic testing (i.e. when no mutation is found) does not rule out an inherited cause of the HCM.

6. Therapy

Although there is no cure for HCM, treatments help to control symptoms, and to decrease the long-term risk. Most symptoms can be controlled by using medications, such as beta blockers, calcium channel blockers, anti-arrhythmic medicines and anticoagulants. If patients are at high risk of sudden death (for example after a previous cardiac arrest) or if symptoms cannot be controlled by medication, the implantation of an internal cardiac defibrillator (ICD) can be considered. The ICD constantly monitors the

electrical activity of the heart and can recognize severe arrhythmias. The ICD can be programmed specifically to each individual patient. It can treat severe and rapid arrhythmias by sending electrical impulses or give an electric shock, and return a normal heartbeat. An ICD consists of 2 parts: the battery (the device) and the electrical lead that monitors the electrical activity of the heart and provides electrical impulses or an electrical shock to the heart. The lead of an ICD can be placed in the right heart chamber (via blood vessels) or under the skin in an area of the chest overlying the heart.

A small number of individuals who have HCM with obstruction will still have symptoms despite the medications. These individuals may need further treatment to help reduce their symptoms and improve their heart muscle function. There are two main types of treatment: myectomy or alcohol septal ablation. Myectomy is an open-heart surgical procedure to remove part of the thickened part of the heart muscle that is causing obstruction (i.e. the interventricular septum). Alcohol septal ablation is the injection of a small amount of alcohol by a catheter in the blood vessel that supplies blood to the thickened part of the heart (i.e. the interventricular septum). Alcohol destroys a part of the thickened heart muscle that is causing the obstruction. Both myectomy and alcohol septal ablation will allow blood to flow out of the heart more easily.

7. Lifestyle & Sports

There are key recommendations for patients (and families) who are diagnosed with HCM, to prevent them from arrhythmias:

- avoidance of strenuous exercise – especially intense, competitive sport and heavy weight lifting;
- regular check-ups to monitor any changes in the disease;
- encouragement of relatives to be screened.

The diagnosis of HCM and the ability to pass on the condition can lead to anxiety and many other questions. Medical social workers or psychologists have experience with this and may be helpful for the patient and the family members.

8. Follow-up

The heart doctor (cardiologist) will advise on how often follow-up is needed depending on symptoms, age, and treatment.

9. Family Screening

If a mutation in a gene is found in a patient with HCM (*see genetic testing*), family members of this patient (to start with the first-degree family members: mother, father, brothers, sisters, and children) can have genetic testing at a specialist genetic heart clinic. Family members in whom the same mutation (familial) is found, are called mutation carriers and will be followed by a cardiologist. Family members in whom the familial mutation is not found can be reassured. If there is not a mutation identified in a patient with HCM, family members of this patient (to start with the first-degree family members) are advised to visit a cardiologist for heart tests. HCM usually develops after puberty, as children grow into adults. However, some children have symptoms of the disease at an earlier age. Therefore the recommendation for children to be tested is from the age of ten years and earlier when the disease has manifested earlier in life in family members.

10. HCM and Pregnancy

Before becoming pregnant it is important to discuss any potential risks, any medication changes and care during the pregnancy.

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