SOME HEARTS ARE EXTRAORDINARY

Learn more about **HYPERTROPHIC CARDIOMYOPATHY**



HCM Patient Foundation

SUPPORT US!

HCM Patient Foundation



DID YOU KNOW?

WHAT IS HYPERTROPHIC CARDIOMYOPATHY *(HCM)*?

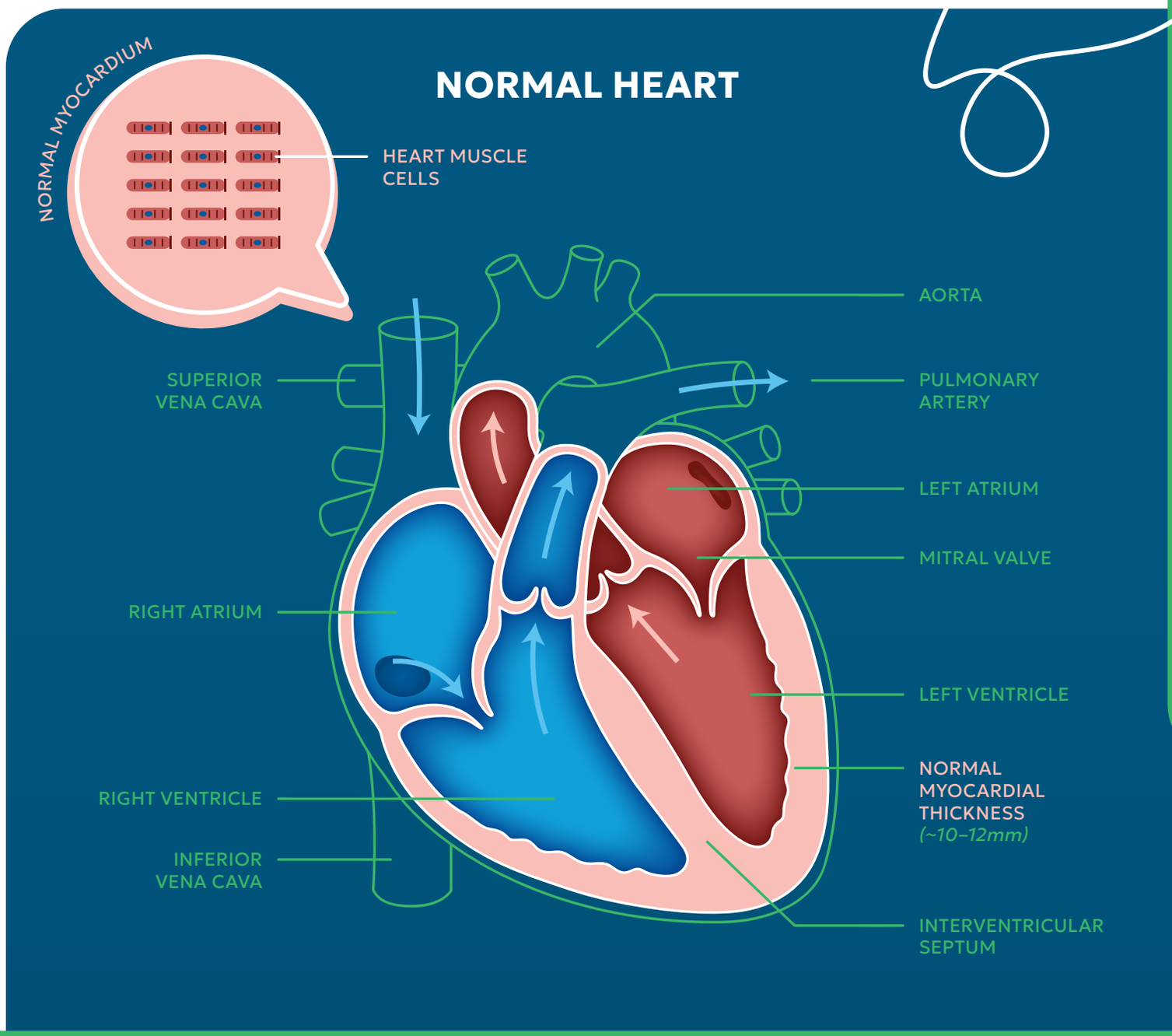
So you or someone you care about has been diagnosed with **Hypertrophic Cardiomyopathy** *(we abbreviate it as HCM because its full name is quite a mouthful)*. Keep calm and don't panic! The important thing is to understand as much as possible about the condition and to find experienced healthcare professionals to help you in managing it.

The most important thing to remember is this:
IT IS PERFECTLY POSSIBLE TO LIVE A LONG AND FULFILLING LIFE WITH HCM!

WHAT DOES HCM MEAN? Hypertrophic means abnormal thickness. Myopathy *(both words come from Greek)* is a generic term for any disease that affects a muscle, the heart one in this case. So HCM is an abnormal thickening of the heart, and mainly of the septum.



HCM is more than a thickened heart muscle. It is a disease that begins deep within the heart muscle cells, where tiny structures responsible for each heartbeat do not use energy as efficiently as they should. By understanding these mechanisms, researchers are **working toward treatments** that address the root causes of HCM rather than simply managing its consequences.



But first of all, let us explain how the heart works. It has two sides – left and right. The right one pumps deoxygenated blood to the lungs. The left-hand side takes the blood that the lungs just oxygenated and sends it back to the body. Then the cycle is closed.

The two sides are very similar in structure. They both have an atrium and a ventricle. The **atrium** is where the blood collects when it enters the heart. The **ventricles** do the actual pumping work. To a certain extent, it is simple physics.

There are also four valves in the heart. They react to the pressure and regulate the flow. There are also two veins and two arteries going in and out of it. It is a delicately balanced but very robust system.

An average heart will pump about seven thousand liters of blood per day. It can fill three supertankers with blood in a human lifetime!

The two sides are divided by a wall called the **septum**, and **it is exactly this wall that becomes thicker in HCM** *(sometimes double the average size and more)*.

But HCM is not simply a condition in which the heart muscle becomes thicker. While the enlarged heart muscle is the most visible feature of the disease, the underlying changes begin much deeper, within the individual heart muscle cells which become disarranged and less efficient.

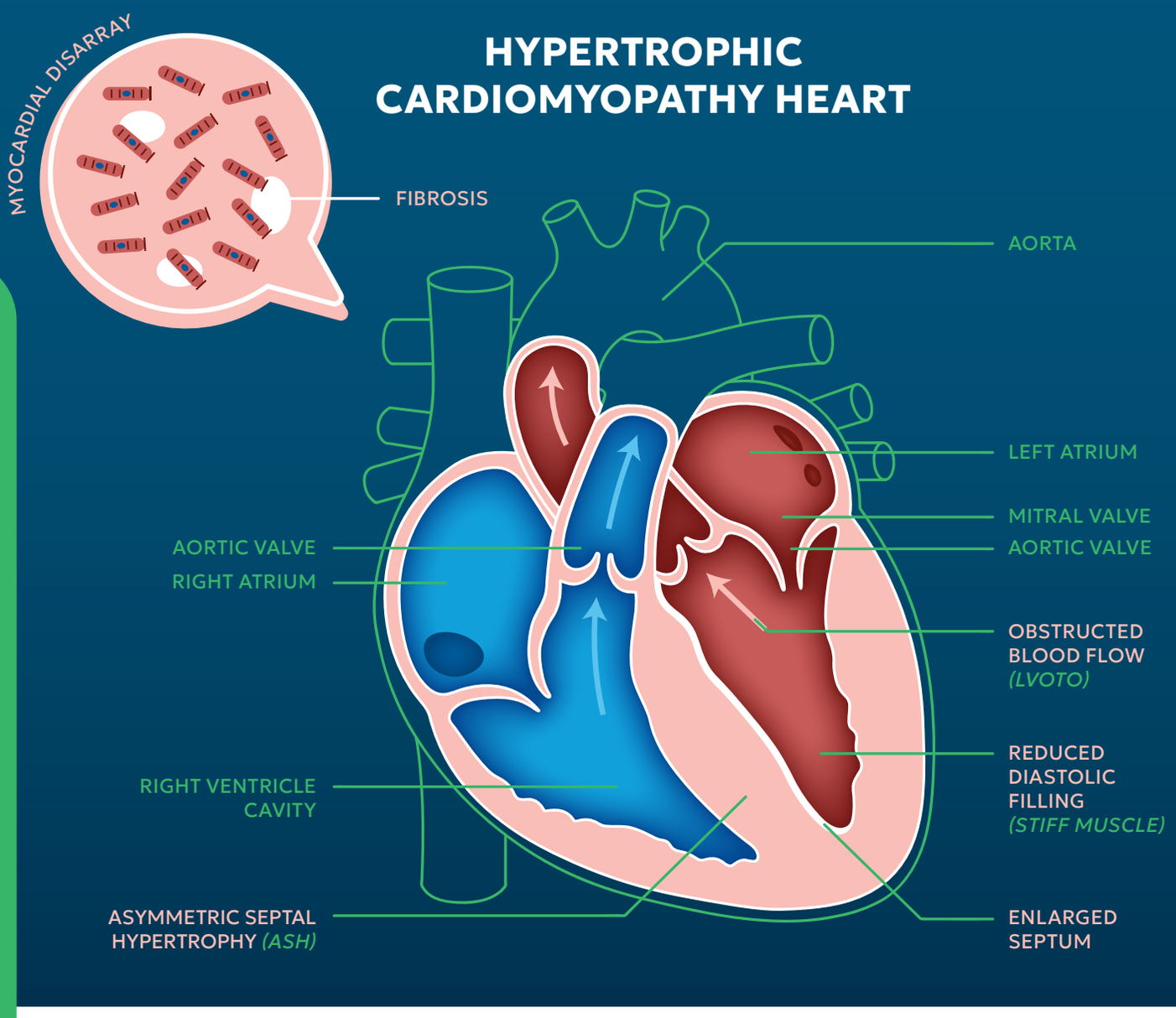
In many people with HCM, genetic defects *(mutations)*, affect structures called **sarcomeres**, the tiny molecular *"motors"* inside heart muscle cells that generate the force needed for the heart to pump blood around the body. These motors are essential for every heartbeat and in HCM they often work less efficiently. As a result, the heart requires significantly more energy to perform the same amount of work as a healthy heart. Think of an expensive sports car that consumes much more fuel to travel the same distance as a more economical vehicle. The heart can still function, but it does so at a higher energy cost.

This increased energy demand is believed to be **one of the earliest abnormalities in HCM** and may contribute to many of the problems that develop later in the disease. Importantly, not all cases of HCM are caused by a single identifiable genetic mutation.

In some individuals, a combination of genetic factors and other influences may increase the likelihood of developing heart muscle thickening. Factors such as obesity may also contribute and act as additional triggers in susceptible individuals.

Understanding these early cellular changes is helping researchers develop new treatments. Rather than only treating symptoms, scientists are increasingly focusing on therapies that improve the heart's energy efficiency and target the underlying disease mechanisms.

This research is paving the way for more personalized treatments tailored to the specific biological changes present in each patient.



OBSTRUCTIVE *VS* NON-OBSTRUCTIVE HCM

Hypertrophic cardiomyopathy *(HCM)* is **one disease**, but it can present in two different forms: **obstructive** and **non-obstructive** HCM. Both are caused by the same underlying changes in the heart muscle and often have the same genetic origin. The difference lies in how the thickened heart muscle affects blood flow.

Think of your heart as a pump, connected to a large pipe that carries blood to the rest of your body.

In obstructive HCM, the exit from the pump becomes partially narrowed. As blood is pushed through this narrow opening at high speed, it creates powerful forces — similar to the airflow that lifts an airplane's wing. These forces pull the heart's mitral valve towards the thickened wall of the heart, creating a temporary blockage that makes it harder for blood to leave the heart. This blockage is called **obstruction**.

It can cause symptoms such as breathlessness, chest discomfort, tiredness, dizziness, or reduced ability to exercise.

In non-obstructive HCM, there is **no blockage** to the blood leaving the heart. However, the heart muscle is often stiffer than normal and cannot relax properly between heartbeats. Imagine trying to fill a balloon that has become thick and rigid — it can still hold air, but it takes more effort to fill it. In the same way, a stiff heart fills less efficiently with blood, especially during exercise. This can also lead to breathlessness, fatigue, and reduced exercise capacity.

Some people with HCM have **no obstruction when they are resting** but develop a significant obstruction when the heart has to work harder — for example during exercise or when certain physiological changes occur. This is known as **provoked *(or latent)* obstruction**.

Current estimates are that about two thirds of people with HCM have the obstructive form of the disease, although this ratio may actually be closer to fifty-fifty. Also note that **the value of obstruction is dynamic** — it changes from moment to moment. The measured degree of obstruction depends on factors such as:



HOW WELL HYDRATED A PERSON IS



HEART RATE



HOW FORCEFULLY THE HEART IS CONTRACTING



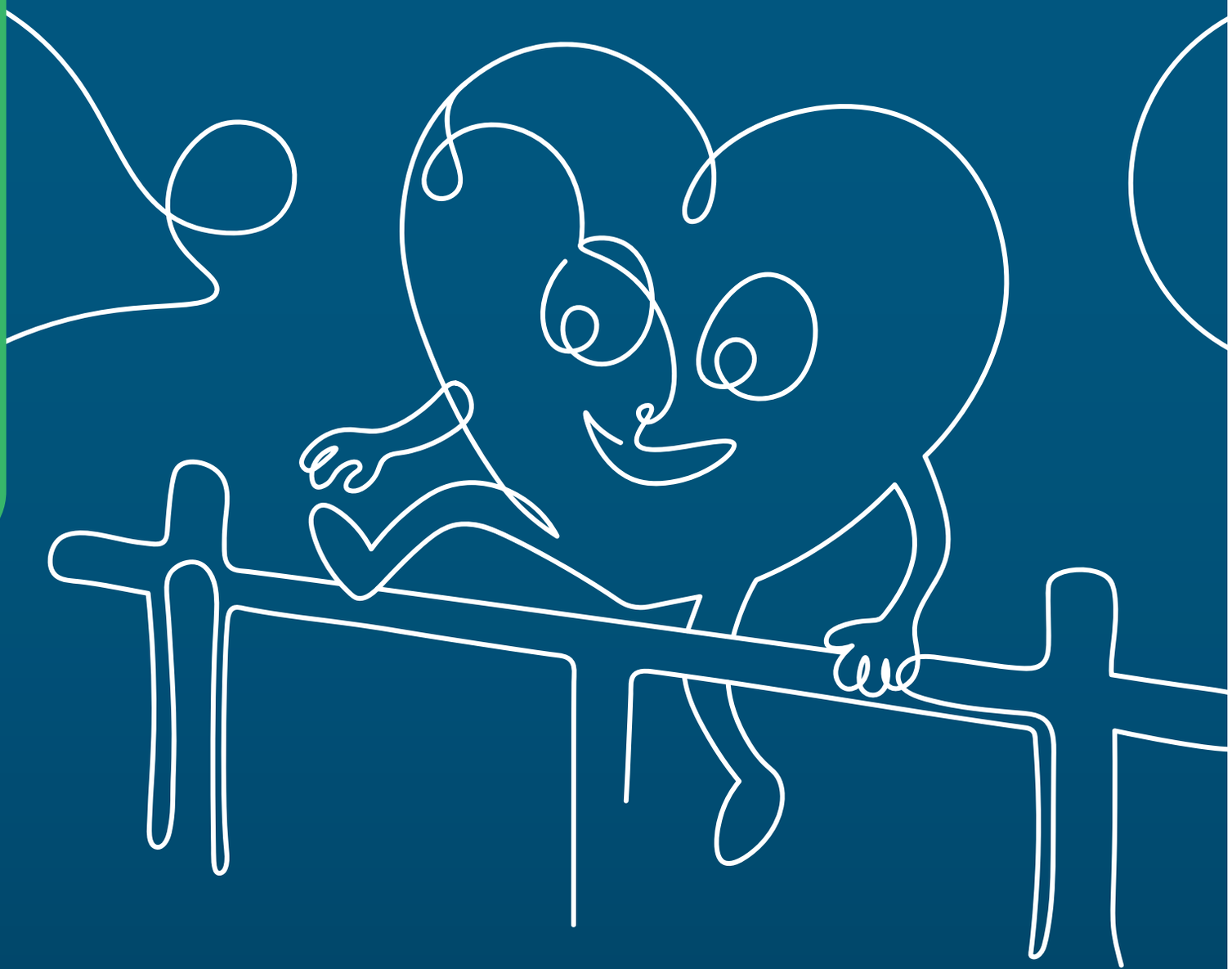
OTHER PHYSIOLOGICAL CONDITIONS



As a result, the pressure gradient measured on one echocardiogram may be different from the gradient measured a few months later, even though the disease itself has not changed.

It is important to know **which type of HCM** you have because **different treatments** are available. Doctors may perform an echocardiogram during exercise or while asking you to perform simple manoeuvres, such as leaning down or changing position, to determine whether obstruction is present.

Treatments that remove or reduce the obstruction — whether through medication or a surgical procedure — can dramatically improve symptoms and quality of life. However, they **do not cure HCM**, because the underlying heart muscle disease remains.





Obstructive and **non-obstructive HCM** are two forms of the same condition. The main difference is whether blood flow out of the heart is partially blocked.
Obstruction can also be provoked and is dynamic. Identifying the correct form helps your healthcare team choose the treatment that is best for you.



ELECTROCARDIOGRAM (ECG)



ECHOCARDIOGRAM



EXERCISE TESTING



GENETIC TESTING



HEART RHYTHM MONITORING



CARDIAC MRI



HOW IS HCM DIAGNOSED

Hypertrophic cardiomyopathy *(HCM)* can be diagnosed in several ways. Many people first come to medical attention after an abnormal heart test, such as an **electrocardiogram *(ECG)*** or an **echocardiogram *(heart ultrasound)***, performed because of symptoms, a sports screening, a routine health check, or a pre-operative evaluation. In other cases, **HCM is discovered** after a heart-related event such as an abnormal heart rhythm, fainting, heart failure symptoms, or, more rarely, a cardiac arrest. Increasingly, HCM is also identified through family screening with **genetic testing** when a relative has already been diagnosed with the condition.

The most important tests for diagnosing HCM are **cardiac imaging tests**. An **echocardiogram** is the cornerstone of diagnosis and can identify the characteristic thickening of the heart muscle. In some cases, a **cardiac magnetic resonance *(CMR or cardiac MRI)*** scan is performed to provide additional detail. While MRI can offer valuable information, most patients can be diagnosed accurately with a **high-quality echocardiogram**.

An **ECG** is also an important tool. Although it cannot confirm HCM on its own, it can raise suspicion and help identify individuals who need further investigation. This is particularly useful when screening family members of people with HCM, especially younger relatives.

Once HCM is suspected or confirmed, additional tests may be needed to determine the exact cause of the heart muscle thickening. Some rare conditions, such as **Fabry disease** or **Danon disease**, can mimic HCM and require different management. These investigations may include genetic testing and other specialized examinations.

The reassuring message for most patients is that HCM can usually be diagnosed using tools that are widely available and non-invasive: careful description of medical history, a standard ECG, and a good-quality echocardiogram. These tests allow cardiologists to identify the condition and begin planning the most appropriate care.

DIFFERENCES BETWEEN CARDIOMYOPATHIES

Cardiomyopathies are diseases of the heart muscle. While they can share some symptoms, such as shortness of breath, palpitations, or fatigue, they affect the heart in different ways and often require different treatments.

Hypertrophic cardiomyopathy *(HCM)* is characterized by an abnormal thickening of the heart muscle, most commonly the wall separating the heart's two lower chambers *(the septum)*. Despite the increased muscle thickness, the heart's pumping function is often normal or even stronger than normal. In some patients, the thickened muscle can partially block blood flow out of the heart, a condition known as obstructive HCM.

Dilated cardiomyopathy *(DCM)* is almost the opposite. In DCM, the heart chambers become enlarged and the heart muscle becomes weakened, reducing the heart's ability to pump blood effectively. This can lead to heart failure symptoms such as fatigue, breathlessness, and fluid retention.

Restrictive cardiomyopathy *(RCM)* occurs when the heart muscle becomes stiff and less able to relax between heartbeats. The heart chambers may remain normal in size, but filling with blood becomes difficult. Patients often experience shortness of breath, exercise intolerance, and swelling of the legs or abdomen.

Arrhythmogenic cardiomyopathy *(ACM)* primarily affects the heart muscle by replacing normal heart tissue with scar tissue and fat. This process increases the risk of abnormal heart rhythms *(arrhythmias)* and can sometimes lead to sudden cardiac arrest, particularly in younger individuals.

TYPE OF CARDIOMYOPATHY	MAIN PROBLEM	EFFECT ON THE HEART
Hypertrophic *(HCM)*	Heart muscle becomes abnormally thick	Heart may pump normally but fill less efficiently; obstruction may occur
Dilated *(DCM)*	Heart chambers enlarge and weaken	Reduced pumping ability
Restrictive *(RCM)*	Heart muscle becomes stiff	Reduced filling of the heart
Arrhythmogenic *(ACM)*	Heart muscle replaced by scar and fatty tissue	Increased risk of arrhythmias

UNDERSTANDING THE GENETICS OF HCM

Hypertrophic cardiomyopathy *(HCM)* is the most common inherited heart muscle disease. In about half of people with HCM, the condition is caused by a genetic change *(mutation)* that can be identified through genetic testing. These mutations are usually found in genes responsible for producing proteins that form the sarcomere — the microscopic *"engine"* that enables heart muscle cells to contract and pump blood.

HOW IS HCM INHERITED? HCM is most often inherited in an **autosomal dominant pattern**. This means that a person with a disease-causing mutation has a 50% chance of passing it on to each child. Because of this, HCM often runs in families and may affect several generations.

MUTATIONS IN WHICH GENES CAUSE HCM? More than 20 genes have been associated with HCM, but only a handful account for most cases. MYBPC3 and MYH7 together account for approximately 70–80% of all disease-causing variants identified in HCM patients with a positive genetic test. Other more prominent variants are TNNT2 *(3-7 %)* and TNNI3 *(2 to 5 %)*.

GENETIC TESTING SHOWED THAT I HAVE AN HCM-CAUSING GENE. WILL I DEVELOP THE DISEASE? You may carry the gene and still not express the disease. You can be one of the two:

- **Genotype-positive, phenotype-negative:** You carry the mutation but have no evidence of HCM on testing.

- **Genotype-positive, phenotype-positive:** You carry the mutation and have developed measurable signs of HCM.

If you are **genotype - positive** but **phenotype - negative**, current guidelines generally recommend periodic follow-up with a cardiologist experienced in inherited heart diseases. The interval depends on your age, the specific mutation, and family history.





CAN A GENETIC RESULT PREDICT HOW MY DISEASE WILL DEVELOP? No, it cannot. A genetic result does not predict the future with certainty. Even if the disease is expressed, its course cannot be predicted just from the gene type. Researchers have identified trends suggesting that some genetic variants may be associated with more severe disease than others. However, there is considerable variation even among relatives who carry exactly the same mutation. Some people develop significant heart muscle thickening and symptoms, while others experience only mild disease or remain symptom-free for many years.

This variability is one of the major challenges in HCM research. Scientists are working to better understand why the same genetic change can affect individuals so differently. The long-term goal is to use genetic information to provide more personalized care, improve risk prediction, and eventually develop treatments that can prevent disease from developing in people who carry HCM-causing mutations.

WHY SHOULD I DO GENETIC TESTING? Genetic testing can provide important benefits. First, it can help confirm that a patient's heart muscle thickening is truly caused by HCM and not by another condition, such as high blood pressure. Second, once a disease-causing mutation has been identified in one family member, other relatives can be tested for the same mutation. Family members who do not carry the mutation can usually be reassured that their risk is similar to that of the general population. Those who do carry the mutation can be monitored regularly so that any signs of disease can be detected early. Finally, new treatment options are appearing all the time and knowing your HCM genetic status can help you in getting quicker access to them.

Genetics plays a central role in HCM.
Genetic testing can help confirm the diagnosis, guide family screening, and support earlier and more personalized care for patients and their relatives.

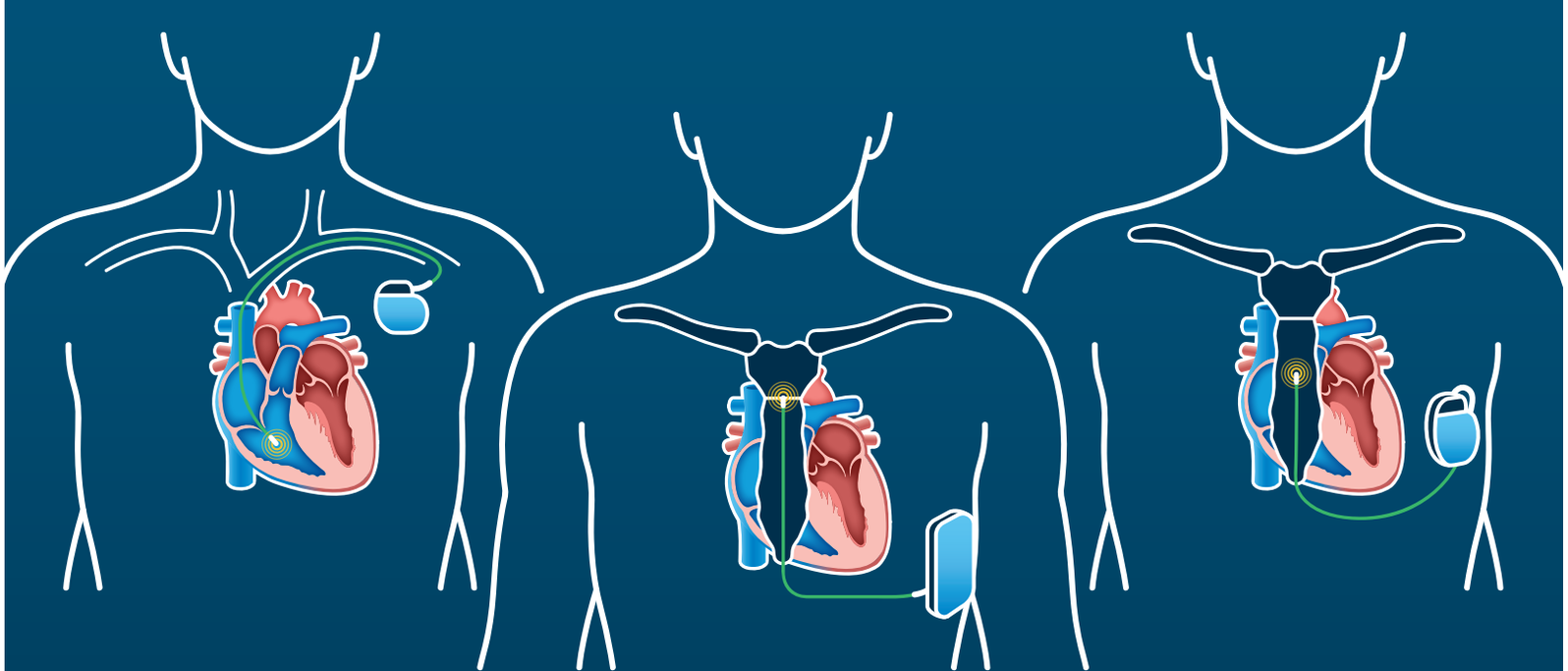
UNDERSTANDING THE RISK OF SUDDEN CARDIAC ARREST AND MAKING DECISIONS ABOUT AN IMPLANTABLE CARDIOVERTER DEVICE *(ICD)*

One of the greatest concerns for people diagnosed with HCM is the possibility of **sudden cardiac arrest *(SCA)***. Although this risk is real, it is important to remember that **the vast majority of people with HCM will never experience sudden cardiac arrest**. The challenge for doctors is to identify the smaller group of patients who are at increased risk and who may benefit from additional protection.

Risk assessment is based on several features of the disease, including the thickness of the heart muscle, heart rhythm abnormalities, family history, and other clinical findings. Rather than looking at each factor in isolation, doctors combine them using a validated risk calculator developed from thousands of patients. This provides an estimate of the likelihood of a serious heart rhythm event over the next five years.

It is important to understand the numbers behind this likelihood. If you are told *"Your risk is 5% for 5 years,"* you can think about it in this way *"Imagine 100 people who have the same characteristics as you. Over the next five years, about five of them may experience a serious heart rhythm event, while 95 will not."* A risk is a probability — not a certainty — and there are many other risks in life which are comparable in magnitude.

Importantly, there is **no single definition of an *"acceptable"* risk**. A young adult planning a family may feel differently about risk than someone in later life whose priorities are focused on maintaining quality of life. The *"right"* decision therefore depends not only on medical evidence but also on the patient's personal values, lifestyle, and goals.



TRANSVENOUS ICD
(TV-ICD)

A lead is inserted through a vein into the heart. The ICD device is implanted beneath the collarbone, the bone between the shoulder blade and the breastbone.

SUBCUTANEOUS ICD
(S-ICD)

A lead is placed under the skin alongside the breastbone.
The ICD device is implanted under the skin on the left side of the chest.

EXTRAVASCULAR ICD
(EV-ICD)

A lead is placed beneath the breastbone, outside the heart and blood vessels. The ICD device is implanted under the skin on the left side of the chest.

When the estimated risk is considered high enough, doctors may recommend an **implantable cardioverter-defibrillator *(ICD)***. An ICD is a small device placed under the skin that continuously monitors the heart rhythm and can deliver a life-saving shock if a dangerous rhythm occurs. Evidence shows that ICDs are highly effective at preventing death from life-threatening ventricular arrhythmias in people at increased risk. Of course, the implant of a foreign object in your body also comes with its own sets of challenges.

Deciding whether to have an ICD should always be a **shared decision**. Shared decision-making is more than simply having a conversation. It means that patients receive clear information which they have enough time to process, understand both the benefits and limitations of treatment, and have the opportunity to express what matters most to them. Only then can the patient and healthcare team make a decision together that is both medically appropriate and personally right.

ICD TYPE	NOW IT WORKS	MAIN ADVANTAGES
Transvenous ICD *(TV-ICD)*	Leads are inserted through a vein into the heart and connected to a device implanted under the skin below the collarbone.	Can deliver shocks and provide pacing for slow heart rhythms, anti-tachycardia pacing *(ATP)*, and cardiac resynchronization if needed. Longest clinical experience.
Subcutaneous ICD *(S-ICD)*	The device is implanted under the skin on the side of the chest, with the lead placed under the skin alongside the breastbone rather than inside the heart.	No leads inside the heart or blood vessels, reducing the risk of lead - related complications and bloodstream infections. Particularly attractive for younger patients.
Extravascular ICD *(EV-ICD)*	A newer system in which the lead is placed beneath the breastbone but remains outside the heart and blood vessels.	Combines the advantage of avoiding intravascular leads with the ability to provide limited pacing, including anti-tachycardia pacing.



Risk prediction is a tool to guide discussion — not to predict the future with certainty.
The decision to receive an **ICD** should combine the best available medical evidence with your own preferences, priorities, and life goals.

EXISTING AND EMERGING TREATMENTS

Until very recently it was possible to treat only symptoms of patients, treatment of HCM itself was nothing but a dream. Oh, how things have changed! **New classes of drugs** that target the very mechanism of HCM appeared and are being **tested regularly**.

These are the currently most used drugs in the treatment of HCM:

DRUG CLASS	EXAMPLES	MAIN PURPOSE IN HCM	TYPICAL USE
Beta-blockers	Atenolol, Bisoprolol, Metoprolol, Propranolol	Slow the heart rate, improve filling of the heart, reduce chest pain and shortness of breath	Usually first-line treatment for symptomatic HCM, particularly obstructive HCM
Calcium channel blockers	Diltiazem, Verapamil	Improve relaxation of the heart muscle and reduce symptoms	Alternative when beta-blockers are ineffective or not tolerated
Cardiac myosin inhibitors	Aficamten, Mavacamten	Reduce excessive contraction of the heart muscle and relieve obstruction	Approved for symptomatic obstructive HCM in appropriate patients
Antiarrhythmic drugs	Amiodarone, Disopyramide, Sotalol	Control arrhythmias and, in the case of disopyramide, reduce obstruction	Used in selected patients with rhythm disturbances or persistent obstruction
Anticoagulants *(blood thinners)*	Apixaban, Dabigatran, Rivaroxaban, Warfarin	Prevent stroke caused by atrial fibrillation	Recommended for HCM patients who develop atrial fibrillation
Diuretics	Furosemide, Torasemide	Reduce fluid retention and breathlessness	Used cautiously in patients with heart failure symptoms, especially non-obstructive HCM
Heart failure therapies	Dapagliflozin, Empagliflozin and others	Treat advanced heart failure when systolic function becomes impaired	Primarily for the small minority of patients who progress to end-stage HCM

As of July 2026 there are 13 active **Phase II** and **Phase III** trials for HCM medications. On top of the existing classes of drugs the following new treatments are being tested:

EDG-7500	Edgewise Therapeutics	Next-generation sarcomere modulator	Obstructive & non-obstructive	**Phase II** *(CIRRUS-HCM)*
Ninerafaxstat	Imbria Pharmaceuticals	Improves cardiac energetics	Non-obstructive	**Phase III** *(FORTITUDE-HCM)*
TN-201	Tenaya Therapeutics	MYBPC3 gene replacement	Genetic HCM	**Phase I/II**

Sarcomere Modulators: Fine-tune heart muscle contraction only when needed, preserving normal pumping while reducing hypercontractility.

Medicines for Improved Cardiac Energetics: Improves the heart's energy production by providing a precursor for NAD^+, an essential molecule involved in mitochondrial energy metabolism

Gene Replacement Therapies: Replace a faulty gene to restore normal heart muscle function at its source.



THE PSYCHOLOGICAL IMPACT OF LIVING WITH HYPERTROPHIC CARDIOMYOPATHY

Being diagnosed with HCM affects more than just the heart — it can also have a profound emotional impact. For many people, the diagnosis brings uncertainty about the future, concerns about symptoms *(e.g. sudden cardiac arrest)*, and worries about how the condition may affect their family, work, and everyday life. Unlike some illnesses, HCM is often an **invisible disease**: many patients look healthy on the outside but struggle with fatigue, breathlessness, or reduced physical ability that others cannot see or easily understand.

For people with inherited HCM, the emotional burden may be even greater. Many worry about passing the condition on to their children or feel guilty that other family members may also be affected. Learning about genetic testing, family screening, medications, or the possible need for an implantable cardioverter-defibrillator *(ICD)* can feel overwhelming, especially in the first months after diagnosis.

These feelings are normal, yet the psychological aspects of HCM often receive less attention than the medical ones. Many patients hesitate to seek support because they believe they *"just have a heart disease"* or feel uncomfortable talking about their emotions. However, asking for psychological help is not a sign of weakness — it is an important part of learning to live well with a chronic condition.

One thing to emphasize is that **support makes a real difference**. Family, friends, and patient communities can help people feel understood and less isolated. Meeting others who are living with HCM — whether through patient organizations, support groups, rehabilitation programmes, or even conversations in a clinic waiting room — can provide practical advice, reassurance, and hope.

Staying engaged in everyday life is equally important. Rather than seeing themselves as *"patients"* all the time, people with HCM should be encouraged to remain active members of their family, workplace, and community. With guidance from their healthcare team, most patients can enjoy regular physical activity appropriate for their condition, helping to improve both physical health and emotional well-being.

Professional support can also play an important role. Psychologists experienced in caring for people with heart disease, together with comprehensive cardiac rehabilitation programmes that address both physical and emotional recovery, can help patients develop effective coping strategies, rebuild confidence, and adapt to life with HCM.



Living with HCM involves caring for both your heart and your emotional well-being.
With the support of healthcare professionals, family, friends, and other patients, most people adapt successfully and continue to lead full, meaningful lives.

PHYSICAL ACTIVITIES WITH HCM

Regular physical activity is important for everyone, including HCM patients. Exercise benefits not only the heart but also the muscles, bones, blood vessels, brain, and overall wellbeing. Rather than avoiding activity altogether, patients should be encouraged to find a form and intensity of exercise that is appropriate for their individual situation.

Historically, many patients with HCM were advised to avoid sports because of concerns about dangerous heart rhythms and sudden cardiac arrest. However, more recent research suggests that these risks may have been overestimated in some patients. Advances in cardiac imaging, exercise testing, risk assessment, and preventive therapies now allow clinicians to make more **personalized recommendations**. Today there is a shift from focusing on what patients cannot do towards identifying what they can do safely. Exercise should be viewed as an important part of a healthy lifestyle and, in many cases, can act almost like a form of medicine.

One thing to remember is that **there is no *"one-size-fits-all"* approach**. HCM affects people differently: some have significant heart muscle thickening, others experience arrhythmias, and many have few or no symptoms. Decisions about sport participation should therefore be individualized, taking into account the patient's condition, symptoms, and risk profile.

Today we also have a great variety of wearable devices and fitness trackers. These tools can help patients monitor training intensity and understand their exercise habits, but they also have some important limitations. Wearables should not be relied upon to diagnose arrhythmias, nor do they provide guarantees about safety. They should be used as supportive tools rather than substitutes for medical evaluation.



For most people with HCM, appropriately **tailored physical activity should be encouraged.**
Exercise decisions should be made jointly with healthcare professionals, based on individual risk assessment, with the goal of helping patients live full, active, and healthy lives.



SUPPORT US!

ABOUT THE HCM PATIENT FOUNDATION

In 2023 we, a group of European HCM patients, decided to found the **HCM Patient Foundation** as we realized that there were serious gaps in the diagnostics and treatment of this condition on European level. We therefore made it **our mission** to unite and support in all possible ways European HCM patients and their caretakers by helping to raise public awareness, working together with competent European and local authorities to adopt better legislation for the treatment of the condition, and supporting research in the field.

Our vision is to achieve a much better outcome of HCM in patients in the European Union, including prolonged life expectancy, better quality of life, improved understanding of genetic factors and access to the most modern medicines and treatments, including psychological care.

We dream of the day when HCM will be routinely treatable or even completely eradicated.

Our work is based on projects.
Some of them are:

• Website with rich information on the subject in 12 European languages.

• Research with 338 patients from 18 countries *(2025)*. We published the results of the research in the International Journal of Cardiology Cardiovascular Risk and Prevention.

• Free psychological support for HCM patients *(Project AURA)*.

• Online quiz to test your knowledge on HCM.

• The HCM Pills webcast in which top experts in the field explain different aspects of HCM in simple and down-to-earth terms.

• Yearly meeting of European HCM patients in Vienna *(First meeting was held in 2025, second to come in October 2026)*.

We are currently working on all these projects and developing several new ones. You can support our work as a whole or on project basis.



Learn more at:
hcmpatient.eu

2026

THIS BROCHURE WAS PRINTED WITH THE KIND SUPPORT OF

Cytokinetics®

Bristol Myers Squibb