Hypertrophic cardiomyopathy (HCM) is a complex type of heart disease. This condition can include:
- Abnormal thickening of the heart muscle (especially the ventricles – the lower chambers of the heart)
- Stiffness of the heart muscle
- Mitral valve changes
- Heart muscle cells that are disorganized and irregular (myocardial disarray)

These changes in the heart make it harder to pump blood and can cause irregular heart beats. About 1 in 500 people have HCM.

There are many different health risks that can lead to enlargement of the heart muscle (hypertrophy), such as high blood pressure, aging, and heavy alcohol use. However, sometimes the heart enlarges without an apparent cause. When heart enlargement happens without any underlying medical risk, it is called hypertrophic cardiomyopathy (HCM).

How is hypertrophic cardiomyopathy diagnosed?
HCM is diagnosed by a doctor who reviews your medical history, asks about your family history, performs a physical exam, and orders ultrasound testing on your heart (an echocardiogram) and heart rhythm testing (electrocardiogram). The characteristic thickening of the heart walls is usually visible on the echocardiogram.

What causes hypertrophic cardiomyopathy?
HCM is often due to a gene mutation (change in the genetic instructions) that causes the heart muscle to gradually thicken over time. Most of the known HCM genes play a role in how the heart muscle cells are formed, which can affect how the heart works. More than 20 different genes are known to cause HCM; however, not everyone who has hypertrophic cardiomyopathy has a mutation in one of the known HCM genes. It is very likely that there are other HCM genes that have not been found yet.

How is hypertrophic cardiomyopathy inherited?
The genes for hypertrophic cardiomyopathy can be passed down through the family. If a person has a mutation in one of the HCM genes, others in the family may inherit the same mutation. HCM is usually an autosomal dominant disorder. This means that, in most families, both men and women can have this condition. It also means that a person with a mutation in an HCM gene has a 50% chance to pass the mutation to each child. Rarely, HCM is caused by a mutation in a gene with a different pattern of inheritance.

GATHERING YOUR FAMILY HISTORY: WHAT TO ASK
Many families with hypertrophic cardiomyopathy are not aware of the condition. When you gather family history, it is helpful to ask about medical problems that might be related to hypertrophic cardiomyopathy.

Ask relatives whether any of the following medical problems have happened in your family:
- Fainting spells (syncope)
- Heart arrhythmias/palpitations (irregular heart beats), especially fast heart beats
- Chest pain or pressure after exercise or physical activity
- Enlarged or thickened heart
- Heart transplant
- Sudden cardiac death
- Shortness of breath with little exertion
When is inherited hypertrophic cardiomyopathy suspected?
The diagnosis of inherited HCM may be suspected based on your personal and family history. However, it is not always easy to recognize inherited hypertrophic cardiomyopathy. In some families, there may be only one or two people who develop heart symptoms that need medical attention.

How is a gene mutation identified?
A blood test can be done to look for mutations (genetic changes) in a select group of HCM genes. Hundreds of mutations in more than 20 different genes have been reported, but genetic testing is available for only some of these genes. Current genetic testing is able to find a mutation in about 60-70% of families suspected to have inherited HCM. Genetic testing is most likely to be informative when your medical history and family history shows a clear pattern of inherited HCM. Your genetics consultation will help determine whether or not testing is appropriate.

What are the benefits of genetic testing?
If a specific HCM mutation is identified in a person with HCM, genetic testing can be offered to family members. Testing can determine if family members without cardiac symptoms are at increased risk to have this condition. HCM often has no noticeable symptoms until later stages of the disease. Healthy individuals who carry a gene mutation can potentially lower the chance for serious heart problems by having regular cardiac screening. Relatives without the family mutation can be given reassurance.

What if genetic testing doesn’t find a mutation?
Genetic testing is not always able to find a mutation. This may be due to limitations in the current testing methods or the mutation may not be on any of the genes included in the current testing panel. If the family history is suspicious for inherited HCM, cardiac screening may still be recommended for family members, even when genetic testing is negative (no mutation is found).

SCREENING FOR HYPERTROPHIC CARDIOMYOPATHY
Screening recommendations for those at risk for HCM include echocardiogram (heart ultrasound) and EKG (electrocardiogram). The frequency of screening depends on your age and medical history.

Echocardiogram is a heart ultrasound. It is the most common test used to diagnose hypertrophic cardiomyopathy. This test uses sound waves to produce images of the heart. Your doctor can see the thickness of your heart muscle, how the blood flows through your heart and how your heart valves move.

EKG (Electrocardiogram) is a heart beat rhythm tracing. It records the electrical activity of your heart as it beats. An EKG can detect abnormal electrical signals that may result from thickened heart muscle. A Holter monitor is a portable form of EKG that records your heart beat for a much longer period of time. A Holter monitor is usually worn on the body for 24 to 48 hours.

Will I have heart problems if I inherit a hypertrophic cardiomyopathy mutation?
A person who inherits a HCM mutation has a much higher chance than usual to develop hypertrophic cardiomyopathy during his or her lifetime. However, some people who inherit a mutation never develop HCM or any of the related heart symptoms. Regular cardiac screening is recommended for mutation carriers.

What happens if I develop hypertrophic cardiomyopathy?
Most people with hypertrophic cardiomyopathy do not have severe heart problems and are able to live a normal life. However, a small number of people with hypertrophic cardiomyopathy have symptoms from the thickened heart muscle, such as shortness of breath, chest pain, fainting, dizziness, fatigue, and heart palpitations or arrhythmias. Less often, problems with blood flow through the heart or irregular heart rhythms can lead to sudden cardiac death. Treatment is available to minimize or prevent symptoms and reduce the risk of complications. Treatment includes regular follow-up, lifestyle changes, medications, and procedures, as needed.