Hypertrophic Cardiomyopathy

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Key points

- Lintroduction
- Clinical Diagnosis and causes of hypertrophic cardiomyopathy.
- Impact on patients
- Pathophysiology
- Treatment and Management

Hypertrophic Cardiomyopathy (HCM) is a prevalent cardiac disorder resulting from the transmission of gene variants. As a hereditary condition, it is more frequently observed in men than women worldwide. In the United States, approximately one in every two hundred or five hundred young individuals may exhibit asymptomatic hypertrophy, while symptomatic hypertrophy is less prevalent at a rate of 1 in 3000. Typical symptoms of this cardiac ailment entail instances of heart attacks, abnormal 12-lead ECG readings, and heart murmur sounds. Accurate diagnosis of the disease can be made through cardiac imaging within a clinical setting.

Hypertrophic Cardiomyopathy leads to the deformation of valve cusps in the left ventricle, resulting in incomplete systolic opening. This obstruction to outflow can result in a significant increase in systolic pressures, medically known as left ventricular outflow tract obstruction (LVOTO). Among HCM patients, 66% exhibit LVOTO, while the remaining cases are non- obstructive. It is worth noting that HCM and left ventricular hypertrophy (LVH) disease share similar symptoms, making the diagnosis process challenging.1

Clinical Diagnosis

As HCM can often be confused with LVH, it is crucial to define the disease precisely. Medical professionals define HCM as "a disease state that is confined solely to the heart in terms of its morphologic expression." Various medical procedures, including 2D echocardiography imaging or cardiovascular magnetic resonance, can be utilized to confirm the increased thickness of the left ventricle to diagnose the condition. The diagnostic process differs for adults and children, with genetic testing being a viable option for adults with an HCM patient in the family, along with imaging techniques. In the case of children, size and body growth criteria should also be taken into account, along with pretest probability and screening conditions. Additionally, family members carrying the "pathogenic sarcomere variant" are at risk of eventually developing the HCM condition.2

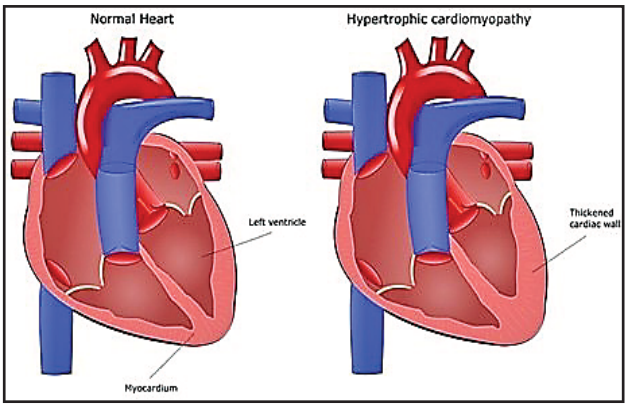

Fig 1: Comparison between normal heart and heart with HCM condition

Causes of the condition

Echocardiographic assessments utilizing DNA sequencing have revealed that Hypertrophic Cardiomyopathy (HCM) is caused by the inheritance of mutant gene coding of sarcomere proteins, making the disease genetic. The presence of abnormal ECGs and physical findings further supports this conclusion. Research suggests that a patient with HCM has a 30%-60% probability of with HCM has a 30%-60% probability of carrying a pathogenic gene variant. However, there are exceptions where no such pathogenic gene variants are found. In cases where pathogenic gene variants are present, the responsible genes are typically MYH7 and MYBPC3, which are found in 70% of HCM patients. In addition to these two genes, at least 1500 gene variants are associated with HCM. Furthermore, any family member has a fifty percent chance of developing the HCM condition if other family members carry any of the pathogenic gene variants.3

Impact on patients

As of yet, there is no cure for Hypertrophic Cardiomyopathy (HCM), but most patients with the condition can expect to reach their normal life expectancy without requiring major treatments. However, the disease can impact various processes and parts of the body in different ways.

Pathogenic gene variants in sarcomere proteins can trigger myocardial variations, hypertrophy, and fibrosis, which can reduce ventricle size and make it stiff. This can significantly impact the performance of diastolic and systolic heart functions. Additionally, HCM can lead to small vessel ischemia, elongation of the mitral valve leaflets, and cause irregularities in the sub-mitral valve apparatus. Patients with HCM have an increased risk of experiencing "heart strokes, impaired diastolic function, and ventricular arrhythmia." In severe cases, patients may experience thromboembolic stroke, and without cardiovascular therapies, fatalities may occur. However, modern therapies have been developed to reduce mortality rates, including Sudden Cardiac Death (SCD) stratification strategies.4

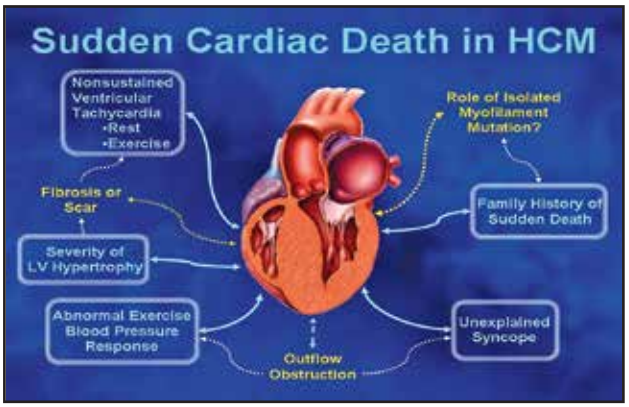

Fig 2: Consequences of HCM on patients

Pathophysiology

The disordered physiological processes associated with HCM are based on left ventricular outflow tract obstruction (LVOTO), impaired diastolic function, myocardial ischemia, and arrhythmias. HCM can be caused by one or a combination of these outcomes. Let us discuss each component in detail:

LVOTO is the major component of HCM, present in 75% of all cases. LVOTO is caused by anatomic variations in the mitral valve or septal hypertrophy, which narrow the LVOT. This increases the risk of heart failure and systolic pressure. CMR imaging is recommended for diagnosing LVOTO to locate the obstruction.

Impaired diastolic pressure is another abnormality observed in HCM. Delayed inactivation between cells for calcium reuptake and irregular contracting and relaxing in the ventricle are common symptoms. CMR imaging can also identify this condition.

Mitral regurgitation is another component of HCM that may or may not be caused directly by LVOTO. It results in the loss of leaflet coaptation, increased leaflet length, and abnormal insertion of papillary muscles. A close assessment of the mitral valve using an invasive approach is required for diagnosis.

HCM may cause a mismatch between the supply and demand of myocardial oxygen, resulting in myocardial ischemia. This condition can trigger infarction, LV aneurysms, and impairment of blood flow. Autonomic malfunction is yet another characteristic of HCM that results in dysfunctional vasodilatation. This can cause abnormal blood pressure, and during workouts, a significant drop in systolic pressure can be experienced. A particular surgical therapy can be beneficial in normalizing the blood pressure response.5

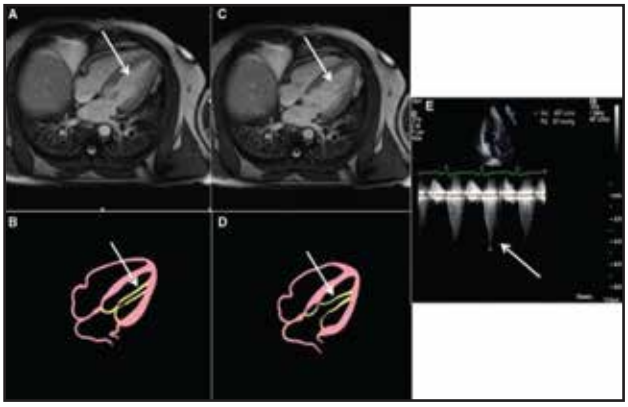

Fig 3: "Left Ventricular Outflow Tract Obstruction in Hypertrophic Cardiomyopathy Patients Without Severe Septal Hypertrophy"

Treatment and Management

If one is diagnosed with Hypertrophic Cardiomyopathy (HCM) and emphasizes the importance of shared decision-making in the treatment plan. There is need for a multidisciplinary HCM center that specializes in treating HCM, providing genetic counseling, and offering advanced HF therapies and transplants. The diagnostic tests for HCM including "clinical diagnosis, echocardiography, cardiovascular magnetic resonance imaging, cardiac computed tomography, heart rhythm assessment, angiography and invasive hemodynamic assessment, exercise stress testing, genetics and family screening, and genotype-positive, phenotype-negative assessment" and these tests can be used to determine the possible management option for the patient. Doctors also stress about the importance of yearly assessments and the need for periodic screening for individuals who are genotype positive and phenotype-negative.6

Recommendation of HCM patients

Hypertrophic Cardiomyopathy (HCM) has no cure but can be managed with proper care and surveillance. Recommendations for symptomatic HCM patients include medication, Shared decision-making for surgical relief of LVOTO, and experienced HCM centers. Recommendations for patients with nonobtrusive HCM include beta-blockers or non-dihydropyridine calcium channel blockers, and for patients with AF or Ventricular Arrhythmias, anticoagulants and catheter ablation can be considered. Lifestyle recommendations include low-intensity exercise, occupation guidelines, pregnancy guidelines, and management of comorbidities. Genotype

positive and phenotype-negative individuals have no restrictions on sports.

Conclusion

Hypertrophic Cardiomyopathy (HCM) is a complex and potentially life-threatening disease that requires proper surveillance, care, and management. Although there is no known cure for HCM, with the help of modern diagnostic techniques and advanced therapies, the condition can be effectively managed, allowing patients to live a full and healthy life. Effective management strategies for HCM include medication, shared decision-making, surgical procedures, and lifestyle modifications. It is crucial that patients with HCM work closely with their healthcare providers to develop a personalized care plan that is tailored to their unique needs and medical history. By following the guidelines and recommendations outlined in this thread, HCM patients can minimize their risk of complications and lead a healthy, fulfilling life.7

References

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2019 Feb 1;37(1):45-54.