Use of Echocardiography to Diagnose Obstructive Hypertrophic Cardiomyopathy

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Interview Summary

Fabian Knebel, Sana Klinikum, Berlin, Germany, and Department of Cardiology and Angiology, Charité Campus Mitte, Berlin, Germany, opened the podcast with a definition of hypertrophic cardiomyopathy (HCM), which is characterised by left ventricular hypertrophy. Knebel then described the symptoms of the condition, such as shortness of breath and syncope after physical exertion. Up to one in 400 persons carry the genetic mutations that can lead to HCM, which is a dangerous condition if left undiagnosed, potentially leading to ventricular tachyarrhythmias and sudden cardiac death, or end-stage heart failure. HCM is a genetic condition, and genetic testing should be performed in cases of unexplained death so that families can be tested if HCM is diagnosed. Echocardiography is the first imaging method of choice for patients with HCM due to its relatively low cost and wide availability, but cardiac MRI may also be performed to measure left ventricular wall thickness, fibrosis, and left ventricular outflow tract (LVOT) obstruction, and to evaluate the success of therapies such as septal myectomy. When using echocardiography, the first obvious finding to indicate HCM is a thickened left ventricular wall, usually in the interventricular septum. European and American guidelines agree that an end-diastolic left ventricular wall thickness of ≥15 mm should be considered HCM. Knebel provided advice on how to effectively perform echocardiography, including using apical cut planes, and measuring the pressure gradient in the LVOT during the Valsalva manoeuvre. A case study was presented of a patient presenting with signs and symptoms suggestive of myocardial infarction, demonstrating the potential complexity of diagnosing HCM. Knebel concluded with a summary of current therapeutic options, such as septal reduction surgery and septal branch ablation, and pointed out that in the near future there will be medications to reduce LVOT obstruction and alleviate symptoms.

What is Hypertrophic Cardiomyopathy?

HCM is a rare disease that is characterised by increased left ventricular wall thickness, referred to as left ventricular hypertrophy.[1] The hypertrophy can be mild or extensive. HCM is classified as obstructive and non-obstructive. The disease is progressive, meaning that a patient without obstruction can develop obstruction over time as left ventricular hypertrophy increases.[2]

Symptoms of Hypertrophic Cardiomyopathy

HCM has a complex pathophysiology that dictates the symptoms patients experience.[1] First of all, the symptoms of HCM depend on the degree of left ventricular hypertrophy. Hypertrophy leads to stiffness of the left ventricle, which causes shortness of breath during physical exercise. As left ventricular hypertrophy increases over time, the patient can develop LVOT obstruction, which exacerbates the shortness of breath, and may cause syncope after physical exertion. In some patients with HCM, the mitral valve is affected, causing mitral regurgitation, which can also lead to shortness of breath.

Unfortunately, there are no symptoms specific to HCM that could differentiate it from other cardiac conditions.[3] The typical symptoms can also be present in patients with heart failure due to other causes, after myocardial infarction, in dilated cardiomyopathy, and in other diseases. As a result, it is difficult to diagnose HCM using the patient history alone.

How Common is Hypertrophic Cardiomyopathy?

It was previously thought that HCM was a very rare disease affecting just one in 100,000 individuals. However, contemporary genetic analyses have revealed that up to one in 400 persons carry genetic mutations that can lead to HCM.[4] To put that into perspective, in a large aeroplane at least one passenger would be at risk of developing the condition. In most cases, patients do not know that they are harbouring genetic mutations that can cause HCM.

Is Hypertrophic Cardiomyopathy Dangerous?

HCM is a dangerous condition if left undiagnosed. Patients with the condition may develop heart rhythm disturbances, called ventricular tachyarrhythmias, that can lead to sudden cardiac death.[2] Another common cause of death is end-stage heart failure. The potentially serious consequences of HCM make it important to diagnose. Because the disease is genetic, when families have an unexplained death that could potentially be HCM, it is important to perform genetic testing to check for HCM so that if the result is positive, relatives can also be tested and treated.[5]

How is Hypertrophic Cardiomyopathy Typically Diagnosed?

The first indication that HCM might be present usually occurs in the echocardiography laboratory. Patients with HCM often have a long history of unexplained symptoms before the disease is diagnosed when a physician examines the ECG and echocardiography findings.[3]

While echocardiography is the first imaging method of choice for patients with HCM due to its relatively low cost and wide availability, in certain scenarios, cardiac MRI should also be performed. Reasons to conduct a cardiac MRI scan include to measure the wall thickness, to detect the extent of fibrosis in the heart, to measure certain aspects of LVOT obstruction, and to determine the success of therapies such as septal myectomy.[6,7]

Use of Echocardiography to Diagnose Hypertrophic Cardiomyopathy

When using echocardiography to diagnose HCM, the first obvious finding to indicate a diagnosis is a thickened left ventricular wall.[6] In cases where there is borderline hypertrophy of the left

ventricle, it can be challenging to distinguish normal values from pathological values, and more detailed investigation is needed.

In most cases, left ventricular wall thickening is located in the interventricular septum. However, there are also forms of HCM in which increased wall thickness occurs in the apex of the left heart or in the mid-ventricular segments, either symmetrically or asymmetrically.[8]

In terms of cut-off values, there is consensus in the guidelines of both the European and American cardiology societies that an end-diastolic left ventricular wall thickness of ≥15 mm should be considered HCM.[1,9] The reference value for healthy cohorts is 12 mm, meaning that a measurement of 13–14 mm falls into a grey zone and further evaluation is needed, including family history, non-cardiac symptoms and signs, ECG abnormalities, laboratory tests, and multimodality cardiac imaging.[1]

Measuring wall thickness can be technically challenging because the heart is constantly moving, and because the thickness may be asymmetrical. A rule of thumb is to measure wall thickness at the point with the largest extent of hypertrophy.[10] Subtle signs or artefacts on echocardiography may also complicate the diagnosis. Apical cut planes should be used to avoid overlooking asymmetrical hypertrophy. In addition, LVOT obstruction should be ruled out; techniques for diagnosis of LVOT obstruction are described below.

In most cases, echocardiography is performed with the patient lying down at rest. For patients whose symptoms manifest during physical exertion, there are two options. One is to perform stress echocardiography with the patient walking on a treadmill; however, image quality can be poor with this technique due to the patient's movement. Another challenge that physicians may encounter when using exercise stress echocardiography is that patients may develop rhythm disturbances, such as atrial fibrillation or a ventricular tachyarrhythmia, while on the treadmill, which could lead to misdiagnosis or underdiagnosis of HCM.

An elegant method to overcome the limitations of stress echocardiography is the Valsalva manoeuvre, whereby the patient closes their mouth and nose, and attempts to exhale forcefully for approximately 10 seconds, during which the pressure gradient in the LVOT is measured. A sharp increase in the pressure gradient in the LVOT indicates a diagnosis of hypertrophic obstructive cardiomyopathy (HOCM). Guidelines recommend starting with the Valsalva manoeuvre instead of exercise stress echocardiography[1,9] due to the low cost, ease of use, and diagnostic accuracy.

Using Echocardiography to Diagnose Left Ventricular Outflow Tract Obstruction

There are three ways to use echocardiography to diagnose LVOT obstruction. The first is visual assessment of the LVOT, which can be seen clearly in the parasternal long axis. The outflow tract is typically 20 mm wide, but is reduced to, for example, 5–6 mm when obstruction is present. The second tool is Doppler echocardiography. Here, the physician will see a turbulent flow in the LVOT, which shows up as green or yellow on the image depending on the device used.[11] The third technique is provocation with the Valsalva manoeuvre or exercise stress echocardiography, which can quantify the extent of LVOT obstruction based on the pressure gradient.[11]

An effective method for diagnosing HOCM is the Brockenbrough-Braunwald-Morrow sign, which can be assessed in patients with a ventricular ectopic beat. HOCM is diagnosed when there is a post-extrasystolic potentiation of the outflow tract gradient.[12]

Case Study

The case of a patient with a rare manifestation of HCM illustrates the potential complexity of diagnosis. The patient presented with signs and symptoms suggestive of myocardial infarction, including chest pain, ECG changes, and elevated troponin. However, on further examination with imaging, the coronary vessels looked completely normal.

Echocardiography revealed that the patient had severe HCM, especially affecting the mid-ventricular segments of the myocardium. Due to the thick mid-ventricular myocardium, contraction of the left ventricle led to the development of a left ventricular aneurysm in the apical segments. An MRI showed that the patient had already developed a scar in the apical myocardial segments despite the absence of coronary artery disease. A literature search showed that up to 5% of patients with HCM have this combination of mid-ventricular obstruction, apical aneurysm, and a scar.[13]

Conclusion

Diagnosis of HCM can be challenging. The most obvious cause of left ventricular hypertrophy in Western countries is usually arterial hypertension, but physicians should keep in mind that the presence of hypertension does not exclude HCM.[14] Left ventricular hypertrophy should always be taken seriously and investigated, even in cases where the degree of thickening is borderline. A Valsalva manoeuvre should always be performed at the first examination to rule out HOCM. HCM is not extremely rare, and should be in the physician's mind as a possible diagnosis. Effective therapies are available, including septal reduction surgery and septal branch ablation, and in the near future there will be medications to reduce LVOT obstruction and alleviate symptoms.[7,15]

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