The Role of Echocardiography in Hypertrophic Cardiomyopathy

Jing Ping Sun¹, Xing Sheng Yang¹, and Shaochun Wang²

¹Division of Cardiology, Chinese University of Hong Kong, Sha Tin, Hong Kong
²Affiliated Hospital of Jining Medical University, Jining, China

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Abstract

Hypertrophic cardiomyopathy (HCM) is a common genetic cardiovascular disease and appears in all ethnic groups. HCM is diagnosed on the basis of left ventricular hypertrophy. Echocardiography is a key technique in the diagnosis of HCM, the prognosis of patients with HCM, the management strategy for HCM, and the follow-up of patients with HCM. This review briefly describes and discusses the practical use of established echocardiography techniques and the current and emerging echocardiographic methods that can help physicians in the correct diagnostic and pathophysiological assessment of patients with HCM.

Keywords: Hypertrophic cardiomyopathy; Hypertension; Echocardiography

Introduction

The prevalence of hypertrophic cardiomyopathy (HCM) is about 0.05–0.2% of the general population [1]. The occurrence of HCM is a significant cause of sudden cardiac death in any age group and a cause of heart failure. The generally accepted definition of HCM is a disease state characterized by unexplained left ventricular (LV) hypertrophy of maximal LV wall thickness of 15 mm, with wall thickness of 13–14 mm considered borderline, particularly in the presence of other compelling information (e.g., family history of HCM), on the basis of echocardiography [1].

Diagnosis

Numerous cardiac abnormalities lead to LV hypertrophy, including primary myocardial disease, disorders of myocardial deposition, and hypertrophy due to excess afterload. These “hypertrophic cardiac syndromes” are often distinguished from one another, and from normal function, by other features, including valvular abnormalities, outflow tract obstruction, electrocardiographic patterns, diastolic LV dysfunction, and distribution of LV hypertrophy. This disease has a unique potential for clinical presentation during any phase of life from infancy to old age and may be expressed in a wide range of phenotypical forms, from severe...
symmetric LV hypertrophy to massive hypertrophy of asymmetrical distribution.

The physiopathologic mechanisms underlying HCM are complex and not clearly understood, but include dynamic LV outflow obstruction, mitral regurgitation, and diastolic dysfunction. These may all lead to shortness of breath, limited functional capacity, angina, and syncope; however, although most patients are asymptomatic throughout their lives, there is a risk of sudden cardiac death probably associated with arrhythmia and progression to advanced heart failure with LV systolic dysfunction.

Diagnosis of HCM is now mainly based on cardiac imaging methods, including two-dimensional echocardiography, MRI, and multislice CT scanning. However, echocardiography is the first choice to screen patients for this disease.

**M-Mode Echocardiography**

M-mode echocardiography was the first echocardiographic technique used for the diagnosis of HCM. Typically, M-mode scans through the left ventricle in the long parasternal view allow the detection of a thickened wall (>15 mm) and a reduced cavity of the left ventricle. The hypertrophy can be of concentric or asymmetric distribution, usually affecting the septal wall, with a septal to posterior wall ratio greater than 1.3 in normotensive patients or greater than 1.5 in hypertensive patients.

M-mode echocardiography can also determine the existence, degree, and duration of an abnormal systolic anterior movement (SAM) of the mitral valve, related to several factors, such as abnormalities in the mitral valve apparatus (i.e., posterior to anterior leaflet mismatch and abnormal disposition of the subvalvular apparatus), reduced LV outflow tract (LVOT) dimensions, and the Venturi effect of the abnormally accelerated LV outflow. The duration of the contact of the mitral valve with the septal wall allows the classification of the severity of the SAM, given the high temporal resolution of M-mode scans across the mitral valve in the parasternal long axis (Figure 1A). Additionally, M-mode interrogation of the aortic valve leaflets provides indirect hemodynamic information about the existence of LV outflow obstruction as the early closing of an otherwise normally appearing aortic valve can be detected during the middle of systole (Figure 1B). This mid-systolic closure or notching of the aortic valve must be differentiated from other causes of severe LV hypertrophy such as the presence of a subaortic fixed stenosis (subaortic membrane), where the abnormal closure of the aortic valve occurs in early systole, or a valvular aortic stenosis, where the valve does not open properly throughout the whole systole and thickening of the aortic leaflets is present.

**Two-Dimensional Echocardiography**

Two-dimensional echocardiography provides a powerful tool to evaluate patients with HCM with more anatomical information of the whole cardiac cavities. It also allows a better understanding of the distribution of the hypertrophy than M-mode echocardiography, especially in those rare presentations affecting localizations other than the septal wall or the posterior wall such as the LV inferior or lateral

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*Figure 1* M-mode images of the parasternal long-axis view from a patient with hypertrophic cardiomyopathy (A) showing the mitral anterior leaflet anterior motion (arrow) during systole and (B) showing the aortic valve closed early in the middle of systole (arrow).
The distribution of LV hypertrophy may be well determined by the use of 2D echo echocardiography in the short-axis view of the left ventricle, where the whole transverse section of the LV wall and cavity can be examined in most cases. From this view, this technique may also be able to detect right ventricular involvement (Figure 2). Also, from the apical views, 2D echocardiography allows the evaluation of the distribution of the LV hypertrophy that may not only affect the basal septum mostly but can also involve the whole septal or inferior wall (Figure 2).

Left atrial dimension, an index of chronic diastolic dysfunction and mitral regurgitation, which are both usually observed in HCM patients, can also be assessed with 2D echocardiography, mainly from the apical views. Finally, 2D echocardiography permits the estimation of LV systolic function as in any cardiac disease.

**Color Doppler Echocardiography**

Color Doppler 2D echocardiography detects the presence of a turbulent flow in the LVOT in the case of LV obstruction. The most frequent obstruction position is at the outflow tract, but may occur at three levels: LVOT, mid ventricular, and apical in patients with obstructive HCM. The presence of a flow convergence area in the LVOT can point out the existence of obstruction at this level probably due to an associated SAM, while visualization of a turbulent flow in the mid cavity may help in the diagnosis of these less frequent forms of HCM (Figure 3). Mid-ventricular obstruction may also

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**Figure 2**  Two-dimensional images from a patient with hypertrophic cardiomyopathy showing the distribution of hypertrophic walls: (A) thickened lateral wall with a left ventricular chamber of normal size; (B) apical four-chamber view showing thickened left ventricular anterior and inferior walls; (C) parasternal short-axis view showing only a normal anterior septal wall but thickened remaining segments, including the right ventricular free wall; (D) right ventricular inflow tract view showing thickened right ventricular free wall and septal wall. LA, left atrium; LV, left ventricle; RA, right atrium; RV right ventricle.
develop secondary to apical myocardial infarction in a patient with angiographically normal coronary arteries or in patients with coronary artery disease and diffuse involvement of ventricular hypertrophy [4, 5]. Also, color Doppler echocardiography diagnoses the presence of mitral regurgitation as a consequence of SAM and/or the coexistence of organic valve disease [6]. The mitral regurgitant jet is usually eccentric and directed posterolaterally to the left atrium.

**Pulsed Wave and Continuous Wave Doppler Echocardiography**

Pulsed wave Doppler echocardiography is a reliably tool to detect the level at which the obstruction is produced by mapping flow velocity from the apex to the outflow tract in the presence of LV or mid-cavity obstruction. Especially, pulsed wave Doppler echocardiography was used with color Doppler echocardiography (Figure 3).

Additionally, LV diastolic dysfunction often occurs in patients with HCM, mostly indicating impaired myocardial relaxation regardless of symptoms or the presence of LVOT obstruction. Assessment of diastolic function is helpful in the evaluation of symptoms and disease staging. Doppler echocardiographic parameters are sensitive measures of diastolic function. Therefore a comprehensive evaluation of diastolic function, including Doppler imaging of mitral valve inflow, tissue Doppler velocities at the mitral annulus, pulmonary vein flow velocities, pulmonary artery systolic pressure, and the size and volume of the left atrium, is recommended as part of the routine assessment of HCM.

Pseudonormalization is also often seen in patients with LVOT obstruction and secondary mitral regurgitation with increased atrial pressure. Also, a restrictive pattern may be seen as an effect of increased chamber stiffness causing both rapid atrial-ventricular pressure equilibration (rapid deceleration time) and compensatory increases in left atrial pressure (increased E-wave velocity) [7, 8]. Finally, pulsed wave Doppler imaging of the LV inflow combined with velocities of the mitral annulus determined with pulsed tissue Doppler imaging
may also provide accurate estimates of LV filling pressures [9].

Continuous wave Doppler echocardiography is an essential tool for the estimation of the severity of intraventricular obstruction. Excellent correlation has been demonstrated between pressure gradients determined from continuous wave Doppler echocardiography and cardiac catheterization in different subsets of patients with HCM [10]. Typically, the spectral continuous wave Doppler echocardiogram shows a dagger-shaped gradient across the LVOT [11].

Two-dimensional and Doppler techniques are useful tools to estimate the effect of therapy in patients with HCM. Figure 4 showed continuous wave Doppler images of a patient with HCM before and after percutaneous transluminal septal myocardial ablation; the LVOT obstruction was relieved.

The clinical uses of various echocardiographic techniques for screening and diagnosis of HCM are summarized in Table 1.

Two-Dimensional Speckle Tracking Echocardiography

Two-dimensional strain is a unique imaging mode that permits objective analysis of myocardial motion throughout the entire cardiac cycle.

Experimental measurements of septal wall mechanics in transgenic mice with HCM have shown that regions of myocyte disarray have reduced systolic shortening, torsional systolic shear, and sarcomere length [12]. The focal myofiber disarray and hypertrophy in HCM is present mainly in the hypertrophic wall. In addition, systolic and diastolic sarcomere lengths are significantly shorter in areas of myofiber disarray [13]. Shorter sarcomere lengths suggest that myocytes in areas of disarray may operate at a lower point on their length-tension curve, generating less systolic tension and shortening. Sun et al. [14] found that, compared with healthy volunteers, in patients with HCM the segmental abnormalities circumferential, radial, and longitudinal strain were significantly reduced in the septal, anterior septal, and anterior walls but not in other regions. This heterogeneity of regional dysfunction might reflect the regional distributions of myocardial disarray and fibrosis. Their results demonstrate that the extent of regional abnormalities on 2D strain images is directly related to the degree of regional LV hypertrophy. These results are consistent with results of studies using tissue Doppler imaging [15]. The standard deviation of the time to peak strain was significantly higher in the groups with HCM and amyloidosis compared with the control group, indicating the asynchronized contraction.

Figure 4  Continues wave Doppler images of the left ventricular outflow tract (LVOT) from a patient with hypertrophic cardiomyopathy before (left with high velocity) and after (right with normal velocity) percutaneous transluminal septal myocardial ablation showed the LVOT obstruction was relieved.
pattern that underlies the disease in these patients. This may be one of the mechanisms of systolic dysfunction. Secondary LV hypertrophy is caused by the remodeling of myocardium in response to long-term LV pressure overload (e.g., hypertension and aortic valvular stenosis); there is myofiber compensational hypertrophy, but myofiber array is still normal in the early stage. Systolic function is normal, but the diastolic relaxation may be impaired because of LV hypertrophy.

The assessment of myocardial deformation either by Doppler myocardial imaging or 2D speckle tracking echocardiography may be used to differentiate hypertrophic myopathies with similar phenotype appearance (i.e., myocardial hypertrophy) but with very different genotype and underlying cause such as HCM, amyloidosis, hypertensive cardiomyopathy, or Fabry disease.

The differentiation of such diseases is important as they have different prognostic and therapeutic implications. The most typical pattern of deformation in HCM is segments where no myocardial deformation is present, surrounded by regions of only slightly reduced deformation (Figure 5). Conversely, in amyloidotic disease, all ventricular segments show low or absent systolic longitudinal deformation. Additionally, a study [16] demonstrated LV myocardial longitudinal strain estimated by 2D layer-specific speckle-tracking echocardiography was reduced but LV circumferential strain and LV twist was enhanced with increasing blood pressure in hypertensive patients with normal LV ejection fraction; these early changes may represent early subclinical LV systolic dysfunction and the mechanism of reserved LV gross systolic function.

Finally, longitudinal deformation is most affected in inferolateral segments of the left ventricle in patients with Fabry disease [17]. Three-dimensional strain echocardiography could provide a more comprehensive assessment of hypertrophic cardiomyopathies and also easier interpretation of deformation data by simple visualization of all ventricular segments [18]. Another potential application of myocardial deformation imaging is the prediction of the risk of ventricular arrhythmias in patients with HCM. It has been demonstrated that the presence of more than three LV segments with a reduction in the longitudinal systolic strain of 10% or more is an independent predictor of nonsustained ventricular tachycardia, which is, in turn, associated with the presence of myocardial fibrosis [19]. Finally, deformation imaging has also been used to evaluate left atrial dysfunction; a low peak systolic strain rate of the left atrium has been reported in patients with HCM and correlated with heart failure symptoms [20].

**Conclusions**

Echocardiography is an invaluable tool in the screening and diagnosis of HCM. It is also the first noninvasive imaging method for risk stratification, treatment selection and follow-up of patients. Two-dimensional and Doppler echocardiography

<table>
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<tr>
<th>Table 1</th>
<th>Echocardiographic Evaluation of Patients with Hypertrophic Cardiomyopathy.</th>
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<tr>
<td>1. Presence of hypertrophy and its distribution; report should include measurements of LV dimensions and wall thickness (septal, posterior, and maximum)</td>
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<td>2. LV EF</td>
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<td>3. RV hypertrophy and whether RV dynamic obstruction is present</td>
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<td>4. LA volume indexed to body surface area</td>
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<td>5. LV diastolic function (comments on LV relaxation and filling pressures)</td>
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<td>6. Pulmonary artery systolic pressure</td>
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<td>7. Dynamic obstruction at rest and with Valsalva maneuver; report should identify the site of obstruction and the gradient</td>
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<td>8. Mitral valve and papillary muscle evaluation, including the direction, mechanism, and severity of mitral regurgitation; if needed, TEE should be performed to satisfactorily answer these questions</td>
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<td>9. TEE is recommended to guide surgical myectomy, and TTE or TEE for alcohol septal ablation</td>
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<td>10. Screening</td>
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From [1].
are useful for evaluation of LV diastolic and systolic function, which is important for clinical practice. Two-dimensional strain is a simple, rapid, and reproducible method for early detection of early abnormalities in HCM patients with normal LV systolic function. The diagnosis of HCM is still challenging, and diagnosis can only be 100% reliable when a gene mutation is identified. No single echocardiographic parameter is ideal, and history and clinical examination play a vital role.

REFERENCES

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