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Echocardiography for Hypertrophic Cardiomyopathy

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ABSTRACT

Hypertrophic cardiomyopathy (HCM) is a genetic cardiomyopathy. The prevalence of phenotypic expression, in the absence of another systemic or cardiac disease causing increased left ventricular (LV) wall thickness, is estimated to be 1:500. The frequency of clinical presentation is far less, highlighting the need for a non-invasive diagnostic imaging tool. Echocardiography is readily available and allows for structural characterization and hemodynamic assessment of the hypertrophic heart and to screen patients at-risk for HCM, such as first degree relatives of affected individuals, and differentiate HCM from the athletic heart. Echocardiography can also be used to assess for anatomic abnormalities of the mitral valve apparatus that may exacerbate LV outflow track obstruction and to further risk stratify patients during exercise. Finally, echocardiography plays an integral role in guiding alcohol septal ablation procedures.

Hypertrophic cardiomyopathy (HCM) is characterized morphologically by a hypertrophied, non-dilated left ventricle (LV) in the absence of another systemic or cardiac disease that is capable of producing the magnitude of wall thickening evident (e.g. systemic hypertension, aortic stenosis/AS).[1] HCM, with classical asymmetric septal hypertrophy (ASH) and typical pathologic findings was reported by Teare in 1958.[2] He described eight cases of ASH, seven of which were the result of sudden cardiac death (SCD) in young adults and an additional case of a patient's brother with SCD with identical pathology. He described bizarre and disorganized arrangement of muscle bundles associated with hypertrophy of individual muscle fibers and their nuclei. HCM is clearly a genetic, inherited cardiomyopathy with characteristic cardiac morphology, structure and function.

Maron and colleagues[3] estimate the prevalence of the hypertrophic phenotype in HCM to be 0.2% or 1:500 in the general population. However, the frequency of clinical presentation in the cardiology office and/or hospital is far less. This suggests that many affected individuals remain unidentified.[4] The clinical course can often be complicated by heart failure (HF) symptoms, atrial fibrillation associated with HF or SCD due to unpredictable ventricular arrhythmias.[4]

Transthoracic echocardiography (TTE) is the non-invasive imaging modality of choice in making the initial diagnosis of HCM. Clinical recommendations on the multimodality imaging of HCM from 2011 recommend that an echocardiographic evaluation of a patient with suspected HCM includes evaluation for the presence and distribution of LV and/or right ventricular (RV) hypertrophy, estimation of systolic and diastolic function, estimation of left atrial (LA) size and pulmonary systolic pressure, identification of resting or provocable dynamic obstruction, and characterization of the mitral valve apparatus. Echocardiography has also been recommended as an adjunctive imaging modality to guide either surgical myectomy or alcohol septal ablation procedures.[6]

Echocardiography

TTE allows accurate assessment of ventricular volumes in HCM, which are typically normal or reduced. The presence

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Abbreviations and Acronyms

AS = Aortic stenosis

ASH = Asymmetric septal hypertrophy

HCM = Hypertrophic cardiomyopathy

HF = Heart failure

IVS = Inter-ventricular septum

LA = Left atrial

LV = Left ventricle or left ventricular

LVH = Left ventricular hypertrophy

LVOT = Left ventricular outflow tract

MR = Mitral regurgitation

MV = Mitral valve

RV = Right ventricular

SAM = Systolic anterior motion

SCD = Sudden cardiac death

TTE = Transthoracic echocardiography

and distribution of LV hypertrophy (LVH) can also be determined, with septal wall thickness ≥ 15 mm or inter-ventricular septum dimension (IVSd)/LV posterior wall dimension (LVPWd) ratio ≥ 1.3 supporting HCM in the absence of another explanation for LVH, such as hypertension or AS.

In addition to ASH, there are a number of morphologic subtypes. ASH is most commonly seen, in approximately 75% of cases (Fig 1). Basal hypertrophy accounts for an additional 15% of cases, then concentric and apical or lateral wall variants.[5] In cases of unusual distribution, particularly apical-variant HCM, commercial ultrasound contrast agents may be useful (Fig 2).

In the presence of increased wall thickness, it remains important to exclude the infiltrative causes of cardiomyopathy. These pathologies include cardiac amyloidosis, Fabry's disease, Freidrich's ataxia and glycogen storage diseases. There are often systemic signs of these disease processes that help point toward diagnosis. Cardiac sarcoidosis, Wegener's granulomatosis and hemochromatosis represent infiltrative cardiac diseases that result in dilated cardiomyopathies.[1]In these cases, a multi-modality imaging approach with cardiac magnetic resonance can often be useful in tissue characterization[5,6] (Fig 3).

Although the 2006 American Heart Association recommended classification of cardiomyopathies bundles several of the above cardiomyopathy types together as genetic cardiomyopathies, many suggest that a more useful classification would be at the molecular level, where cardiomyopathies are stratified as diseases of molecular components. For example, cytoskeletal abnormalities in dilated cardiomyopathies, desmosomal disease in the case of arrhythmogenic RV cardiomyopathy or primary sarcomere disorder as seen in HCM.[7,8]

LV ejection fraction (LVEF), whether by visual estimation or biplane Simpson's method, is typically normal or increased in HCM, despite low stroke volumes. The assessment of LV diastolic function is equally important in HCM as it can be the earliest sign of significant myocardial disease. Interpretation of the diastology requires comprehensive review of spectral Doppler signal of the mitral inflow, pulmonary vein Doppler, tissue Doppler of the mitral annulus and measurement of LA volume.[6] In patients with HCM, mitral annular velocities are reduced and correlate with New York Heart Association functional class and exercise capacity[9,10] (Fig 4). Nagueh and colleagues demonstrated that tissue Doppler imaging is an accurate and sensitive method for identifying subjection with familial HCM, regardless of phenotypic expression. In a study of 30 subjects with familial HCM, those with the gene mutation had abnormally low tissue velocities regardless of whether LVH had yet to develop compared to age and sex matched controls.[10]

Assessment of the mitral valve apparatus is also important, as abnormalities of the apparatus may predispose the leaflets to systolic anterior motion (SAM) and LV outflow tract (LVOT) obstruction. It is important to use echocardiography to distinguish between the obstructive and non-obstructive forms of HCM. Echocardiography is essential in differentiating the dynamic obstruction seen in HCM from fixed obstructions such as AS or subaortic membranes. Up to one-third of HCM patients will have obstruction under resting conditions (> 30 mmHg). Another third have no evidence of obstruction at rest (< 30 mmHg) but develop obstruction with provocation (> 30 mmHg).[4]The presence of a resting LVOT obstruction > 30 mmHg is useful in risk stratification for SCD, associated with a relative risk of 2 to 1.[11]

It is necessary to implement all echocardiographic modes (m-Mode, 2D, pulsed, continuous and color flow Doppler) to characterize the level of obstruction and assess the secondary effects. Resting LVOT gradients should be assessed at baseline and also with provocation, by Valsalva maneuver or amyl nitrite. Exercise can also be useful for provocation and is discussed later.

The hallmark of LVOT obstruction is SAM of the anterior mitral valve leaflet. This has been functionally described as secondary to Venturi phenomenon or drag effect.[11–13] Hypertrophy of the papillary muscles may result in anterior displacement and also contribute to LVOT obstruction. Although highly specific to HCM, there are other conditions associated with SAM including: hypercontractile states, anomalous papillary muscle configuration, anteroapical infarction with hyperkinesis of the base, post mitral valve (MV) replacement and patients with sigmoid shaped septa and LVH. As a result of SAM and subsequent outflow obstruction, there may be significant eccentric mitral regurgitation (MR) (Figs 5 and 6). The evaluation of MR in patients with HCM should follow the American Society of Echocardiography's recommended guidelines of severity.[14] However, particular attention should be made to determine whether there is intrinsic MV disease present.

Echocardiography clearly plays an important clinical role in HCM diagnosis and management. Beyond evaluation of structure and morphology, it allows us to screen at-risk patients for HCM, evaluate for structural abnormalities of the mitral apparatus and configuration of the papillary muscles, differentiate HCM from the athletic heart, risk stratify with exercise echocardiography and guide alcohol septal ablation procedures.

Genetic testing and surveillance echocardiography

HCM is inherited as an autosomal dominant trait and has been linked to several different gene mutations. Specifically, certain