Hypertrophic Cardiomyopathy: Standardization of Echocardiographic Assessment in an Era of New Therapies

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Hypertrophic cardiomyopathy (HCM) has become the most common inherited myocardial disease, with an estimated global prevalence between 1:200 and 1:500. Despite its relatively high frequency in the general population, the disease remains significantly underdiagnosed. Only about 15% of affected individuals are clinically identified, due mainly to the wide variability of phenotypes and clinical manifestations.[1,2] Slightly more than half of patients may develop progressive symptoms or experience adverse events throughout their lifetime. Early identification, risk stratification, and cardiovascular therapies and interventions have reduced mortality rates to < 1.0% per year.[3]

The pathophysiology of HCM is based on myocardial hypertrophy in the absence of secondary causes, associated with hypercontractility and diastolic dysfunction, resulting from abnormal myosin activation. Approximately 75% of patients present with left ventricular outflow tract (LVOT) obstruction at rest or after provocative maneuvers. In the absence of an obstructive pattern, the disease course is usually favorable, oligosymptomatic, or asymptomatic, with a minority progressing to advanced stages.[3–5]

Transthoracic echocardiography (TTE) is essential for diagnosing HCM. Suspicion should arise in the presence of diastolic myocardial thickness ≥ 15 mm in the absence of any conditions that justify hypertrophy in a non-dilated ventricle. In patients with a family history of HCM or genetic mutation, diastolic myocardial thickness ≥ 13 mm is considered sufficient. Other indications for performing TTE include systolic murmur suggestive of dynamic obstruction in the LVOT and suggestive symptoms, such as dyspnea, chest pain, and syncope, related to dehydration, exercise, and the postprandial period.[1,2,6]

When HCM is suspected, TTE should contain the relevant information for case management, including the following: indexed left atrial volume; myocardial thickness of the septum and posterior wall; location of the segment with the greatest increase in thickness; left ventricular ejection fraction; global longitudinal strain; analysis of diastolic function; description of apical aneurysm when present; description of the presence and location of intraventricular gradient; description of systolic anterior motion of the mitral valve; mitral valve apparatus abnormalities; and mitral regurgitation.[2,3]

Given the labile nature of LVOT gradients, the absence of obstruction at rest does not exclude latent obstructive HCM. Provocative maneuvers such as the Valsalva maneuver or rapid squat-to-stand maneuver should be routinely used to unmask gradients. Furthermore, postprandial echocardiography has emerged as a powerful tool, as mesenteric vasodilation and the adrenergic response after a meal can significantly elevate gradients in more than a third of patients who would be erroneously classified as non-obstructive when fasting. Whenever resting maneuvers are inconclusive, exercise stress echocardiography (ESE) remains the gold standard for assessing the functional relevance of the obstruction and should be performed after a meal.[2,7]

Recent studies have demonstrated the relevance of hemodynamic assessment in hypertrophic cardiomyopathy under different physiological conditions, including rest, physical exertion, fasting, and the postprandial period. After food intake, even at rest, the presence of systolic anterior motion of the mitral valve and an increased left ventricular outflow tract gradient were observed, findings that became more pronounced during postprandial exertion. This case highlights the importance of a comprehensive use of the available diagnostic tools for characterizing obstruction, as such an approach contributes to therapeutic optimization and to guiding lifestyle measures.[8]

Using TTE and postprandial ESE to study 252 patients with HCM, Massera et al. identified a LVOT gradient ≥ 50 mmHg in 35.7% of patients without obstruction under baseline conditions, including 15.1% on the postprandial physical stress phase alone. More than 50% of patients undergoing invasive treatment or myosin inhibitors had a LVOT gradient ≥ 50 mmHg only on postprandial assessment (TTE and ESE).[7]

Accordingly, clinical routines should incorporate echocardiography with specific protocols for HCM, including provocative maneuvers and postprandial assessment. However, there is still a lack of standardization regarding the type of diet and the interval between the meal and echocardiographic assessment.

Keywords

Hypertrophic Cardiomyopathy; Echocardiography; Left Ventricular Outflow Obstruction.

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