Echocardiography in hypertrophic cardiomyopathy

Cardiac imaging, most commonly echocardiography, is used to aid the diagnosis of HCM and to monitor patients after diagnosis. It enables the visualization of cardiac function and structure, including hypertrophy, LVOTO, and SAM. It provides information essential in the assessment of risk for SCD and stroke.

What to order?

1. Suspected HCM → **order TTE**
2. HCM with resting LVOT < 50 mmHg → **order TTE with provocative maneuvers**
3. HCM with no resting LVOT gradient or provocative LVOT gradient < 50 mmHg → **order exercise TTE**
4. First-degree HCM relatives → **order TTE at initial screening and periodic follow-up**

Key considerations and what to look for

LV hypertrophy

Imaging

- Increased wall thickness can occur anywhere; record presence, severity, and distribution of hypertrophy using cross-sectional imaging from several projections.

Diagnosis

- LV wall thickness ≥ 15 mm, or ≥ 13 mm with positive family history of HCM.
- Concentric LV hypertrophy: more common in phenocopies such as amyloidosis and storage disease, particularly when associated with systolic impairment and RV hypertrophy.

Prognosis: Maximum LV wall thickness is important risk factor for SCD.

SAM, LVOT, and MR

Imaging

- SAM of the mitral valve can contribute to LVOTO and cause MR.
  - Most patients with HCM have papillary muscle abnormalities that contribute to the severity of MR.
- Maneuvers such as Valsalva and standing from a squat decrease LV filling, thus increasing LVOTO. At-rest or standing echo may miss important HCM pathophysiology given the dynamic nature of LVOT gradient.
  - Valsalva: increases HCM murmur intensity, particularly of dynamic subvalvular LVOTO.
  - Exercise: increases cardiac contractility and LVOTO; Doppler echo imaging during these maneuvers can detect significant LVOTO not present at rest.

Diagnosis

- LVOT gradient ≥ 30 mmHg at rest or with provocation defines obstructive HCM.
- LVOT gradient ≥ 50 mmHg: threshold for considering septal reduction therapy and indicates severe obstruction.
- Differentiate other causes of LVOTO, such as the subaortic membrane.
- Determine severity/mechanisms of SAM MR when assessing symptoms and planning septal reduction strategies.

Prognosis: LVOTO is associated with risk of SCD in adults with HCM.

Diastolic function

Imaging

- Key parameters to record: LA volume, transmitral flow velocities, tissue Doppler imaging, pulmonary vein flows, and estimated pulmonary artery systolic pressure; e' velocities decreased, E/e' and LA volume increased.
- Considerations for LA volume:
  - Left atrium: often enlarged (SAM-related MR and elevated LV filling pressures are most common mechanisms).

Prognosis: Increased LA volume is associated with increased risk of AF, stroke, and SCD.

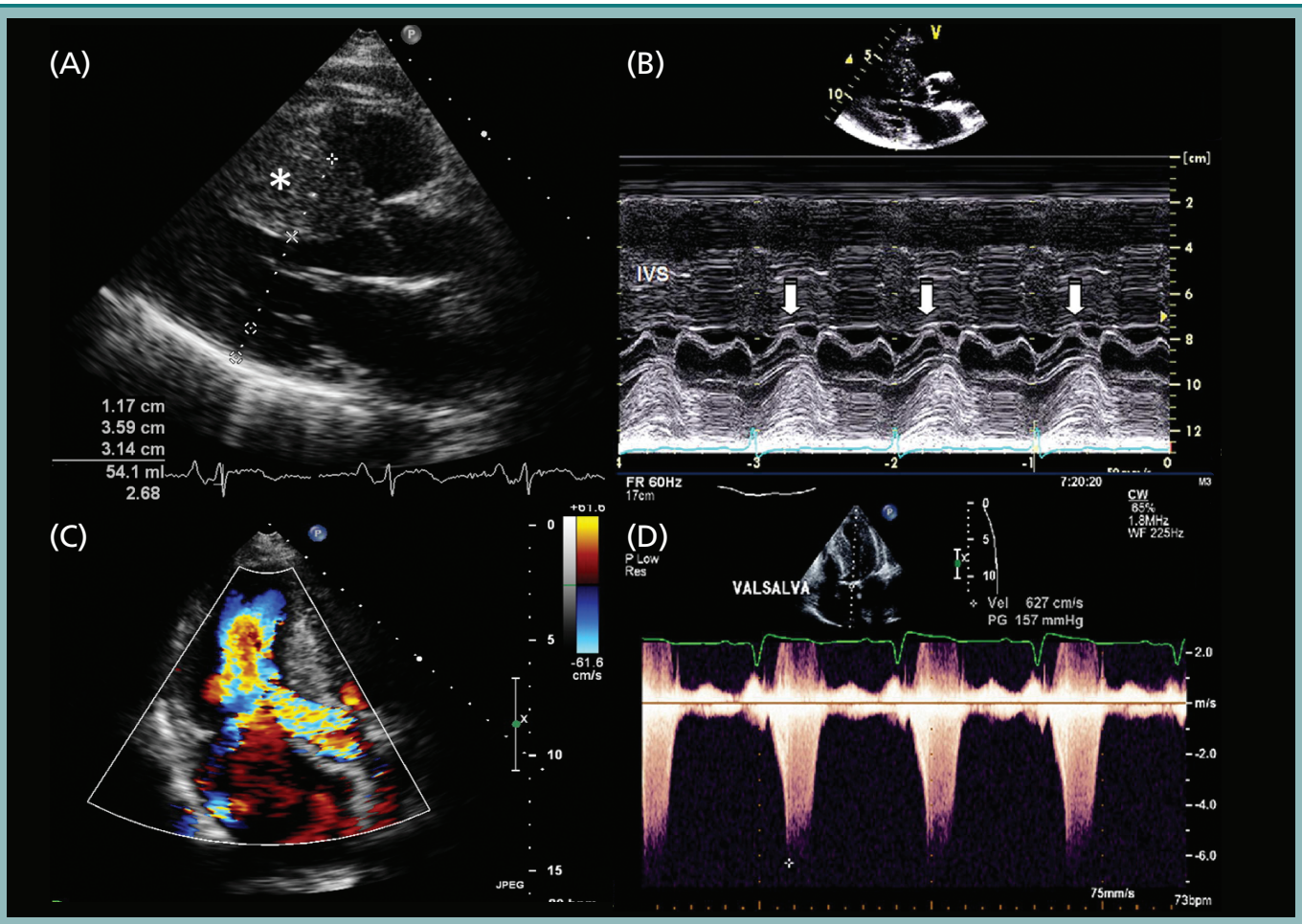
Systolic function

Imaging

- LV systolic function: LVEF 55–70% is considered normal; in HCM, typically normal to hyperdynamic (LVEF > 65–75%) and > 75% in some patients; stroke volume typically normal or reduced.
- Systolic dysfunction (LVEF < 50%): develops in 4–9% of patients with HCM; reported prognosis is poor.
- LV global longitudinal strain (from tissue Doppler or strain imaging): often reduced (< –16%; normal values –16% to –22%) and an early marker of phenotypic expression.

Prognosis: LVEF ≤ 50% is risk factor for SCD.

Additional considerations: Check for LV apical aneurysm (risk factor for SCD) best seen with CMR, which can also demonstrate LV thrombus; contrast echo is useful when CMR is contraindicated/unavailable.



(A) Asymmetric septal hypertrophy. (B) M-mode across the mitral leaflets showing prominent SAM of the anterior mitral leaflet. (C) Turbulence of blood flow due to LVOTO from SAM. (D) Late-peaking dynamic LVOTO accentuated after Valsalva. Reproduced, with permission, from Afonso et al. 2008 and Jamil et al. 2013.

Most common misdiagnoses

Common cardiovascular and pulmonary diseases	HCM phenocopies	Other conditions
Hypertension, hypertensive heart disease (including coronary artery disease), aortic stenosis, mitral prolapse, asthma, COPD	Inborn errors of metabolism or metabolic storage disorders including amyloidosis, Danon disease, Fabry disease, Noonan syndrome	Panic attacks, anxiety, depression, sedentary lifestyle ('out of shape'), athlete's heart

Next steps

- If echo is inconclusive or of inadequate quality, CMR imaging is indicated for diagnostic clarification and can also be used for risk stratification. Cardiac computed tomography may also be considered.
- When LV areas are poorly visualized, LV opacification with echo contrast agents or CMR should be considered.

Glossary
AF, atrial fibrillation;
CMR, cardiac magnetic resonance;
COPD, chronic obstructive pulmonary disease;
HCM, hypertrophic cardiomyopathy;
LA, left atrial;
LV, left ventricular;
LVEF, left ventricular ejection fraction;
LVOT, left ventricular outflow tract;
LVOTO, left ventricular outflow tract obstruction;
MR, mitral regurgitation (mitral insufficiency or incompetence defined as an abnormal reversal of blood flow from the LV to the left atrium);
SAM, systolic anterior motion (the dynamic movement of the mitral valve during systole anteriorly towards the LVOT);
SCD, sudden cardiac death;
TTE, transthoracic echocardiogram.

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