Hypertrophic Cardiomyopathy

Diagnosis, genetic and family screening, sudden death risk stratification with ICD decision-making, and medical/invasive management of obstructive and nonobstructive HCM.

2024 AHA/ACC/AMSSM/HRS/PACES/SCMR
Hypertrophic Cardiomyopathy Guideline
Ommen et al. · JACC 2024;83(23):2324–2405

COR — I · Recommended | IIa · Reasonable | IIb · May Consider | III · No Benefit / Harm

1 Clinical Diagnosis & Imaging

Initial workup for suspected HCM

COR / LOE	
I B-NR	Comprehensive physical exam (incl. Valsalva, squat-to-stand, passive leg raise) plus complete **3-generation family history** recommended as part of initial diagnostic assessment, followed by ECG and imaging to identify LVH.
I B-NR	**TTE** recommended for initial evaluation of suspected HCM. Repeat every 1–2y (children) or per clinical status; repeat immediately with any new symptom or clinical event.
I B-NR	Resting LVOT gradient **<50 mmHg**: TTE with provocative maneuvers recommended. If still <50 mmHg and symptomatic, **exercise TTE** to detect dynamic LVOTO.
I B-NR	**CMR** reasonable when echo is inconclusive (apical HCM/aneurysm, atypical hypertrophy) and for LGE quantification informing SCD risk.

FIRST-DEGREE RELATIVE	START SCREENING	REPEAT ECG/ECHO
Child/adolescent, genotype+ or early-onset family	At diagnosis in relative	Every 1–2y
Other children/adolescents	Any time after diagnosis, by puberty	Every 2–3y
Adults	At diagnosis in relative	Every 3–5y

2 Genetics & Family Screening

PROBAND & CASCADE TESTING

COR / LOE	
I B-NR	Genetic testing recommended in the **proband** to enable cascade testing of at-risk relatives. First-tier panel: *MYH7, MYBPC3, TNNI3, TNNT2, TPM1, MYL2, MYL3, ACTC1*.
I B-NR	First-degree relatives: offer **both** clinical screening (ECG + 2D echo) **and** cascade genetic testing when a pathogenic/likely pathogenic variant is identified in the proband.
III NB · B-NR	If proband has **no pathogenic variant** (benign/likely-benign only), cascade genetic testing of family is not useful. Ongoing clinical screening not indicated in genotype-negative relatives of genotype+ families.

COUNSELING & LIMITS OF TESTING

COR / LOE	
I B-NR	Genetic counseling by an expert in **cardiovascular genetics** recommended so risks, benefits, and results can be reviewed in shared decision-making.
IIb B-NR	Usefulness of genetic testing for SCD risk assessment in adults is **uncertain**; usefulness of testing VUS-carrying phenotype-negative relatives for reclassification is uncertain.

3 SCD Risk Stratification & ICD Decision-Making

Major clinical risk factors — assess at initial evaluation and every 1–2y (I, B-NR)

MAJOR RISK FACTOR	CRITERIA
Family history of SCD	≥1 first-degree/close relative, HCM-attributable, ≤50y
Massive LVH	≥30 mm any LV segment (borderline ≥28 mm)
Unexplained syncope	Recent, suspected arrhythmic (not vasovagal or LVOTO-related)
LV systolic dysfunction	EF <50%
LV apical aneurysm	With transmural scar/LGE
Extensive LGE on CMR	≥15% of LV mass
NSVT	On ambulatory monitoring

COR / LOE	
IIa B-NR	Echo-derived LA diameter + max LVOT gradient inform the validated **5-year SCD risk calculator** (separate adult and pediatric versions) to aid shared decision-making.
I C-EO	Individual clinical judgment applied to conventional risk markers, with thorough shared decision-making, is recommended for every ICD discussion.
I B-NR	**Secondary prevention:** prior documented cardiac arrest or sustained VT — ICD placement recommended.
IIa B-NR	**Primary prevention:** ≥1 major SCD risk factor above — reasonable to offer ICD, after estimating 5-year risk to inform shared decision-making.
IIb B-NR	No major risk factors, or decision uncertain — ICD **may be considered** if extensive LGE or NSVT present (adults); pediatric: consider extensive LGE + systolic dysfunction.
III Harm · B-NR	No SCD risk factors — ICD should **not** be placed. ICD for the sole purpose of sports eligibility should **not** be placed.

4 Obstructive HCM — Medical Therapy Ladder

For symptoms attributable to LVOTO

COR / LOE	
I B-NR	**Step 1 —** Nonvasodilating beta blocker, titrated to symptomatic effect or maximally tolerated dose.

↓ symptoms persist

COR / LOE	
I B-NR/C-LD	**Step 2 —** Beta blocker ineffective/not tolerated → substitute nondihydropyridine CCB (**verapamil** or **diltiazem**).

↓ symptoms persist

COR / LOE	
I B-R	**Step 3 —** Add a **cardiac myosin inhibitor** (adults only), or **disopyramide** (+ AV-nodal blocker), or refer for SRT at an experienced center.

↓ acute hypotension

COR / LOE	
I C-LD	Unresponsive to fluids: IV **phenylephrine** (or other pure vasoconstrictor), alone or with a beta blocker.

Caveats: Persistent dyspnea with volume overload despite GDMT — cautious low-dose oral diuretics may be considered (IIb, C-EO); discontinuing vasodilators (ACEi/ARB, dihydropyridine CCB) or digoxin may also be reasonable, as these worsen LVOTO (IIb, C-EO). Severe rest dyspnea, hypotension, gradient >100 mmHg, or age <6 weeks — **verapamil is potentially harmful** (III: Harm, C-LD).

5 Obstructive HCM — Invasive Treatment (SRT)

INDICATIONS

COR / LOE	
I B-NR	Symptomatic despite GDMT, eligible anatomy — **SRT** (surgical myectomy or alcohol septal ablation) at an experienced HCM center recommended for LVOTO relief.
I B-NR/C-LD	Associated structural disease needing surgery (papillary muscle anomaly, elongated leaflet, multivessel CAD, AS) → **surgical myectomy**. Surgery contraindicated/high-risk in adults → **alcohol septal ablation**.

PATIENT SELECTION

COR / LOE	
IIb B-NR	Earlier (NYHA II) myectomy at comprehensive centers reasonable with: severe/progressive pulmonary hypertension, LA enlargement + symptomatic AF, poor functional capacity, or gradient >100 mmHg in children/young adults.
III Harm	Asymptomatic with normal exercise capacity — SRT **not recommended**. Mitral valve replacement should **not** be performed solely for LVOTO relief when SRT is an option.

6 Nonobstructive HCM, Preserved EF

COR / LOE	
I C-LD	Exertional angina or dyspnea — **beta blocker or nondihydropyridine CCB** recommended; add oral diuretics if dyspnea persists (IIa, C-EO).
IIb C-LD/B-R	ACEi/ARB benefit for symptoms not well established. Apical HCM with severe symptoms + small LV cavity — apical myectomy at experienced centers may be considered. Age ≤45y with pathogenic variant + mild phenotype — **valsartan** may slow remodeling.

Ommen SR, Ho CY, Asif IM, et al. 2024 AHA/ACC/AMSSM/HRS/PACES/SCMR Guideline for the Management of Hypertrophic Cardiomyopathy. *J Am Coll Cardiol*. 2024;83(23):2324–2405.

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