Hypertrophic Cardiomyopathy

HCM · Relieve the obstruction, stratify the sudden-death risk, screen the family

PRELOAD IS PRECIOUS
avoid vasodilators

DIAGNOSE *unexplained hypertrophy, not hypertrophy with a reason*

THE THRESHOLD

LV wall thickness ≥ 15 mm unexplained by loading conditions. **≥ 13 mm** is sufficient with a **family history** of HCM or a positive genotype. Exclude hypertension, aortic stenosis, and athlete's heart.

OBSTRUCTIVE OR NOT

LVOT gradient ≥ 30 mmHg at rest or with provocation defines obstruction; **≥ 50 mmHg** is the threshold for invasive relief. **Always provoke** — Valsalva, standing, or exercise echo — a normal resting gradient misses it.

THE MURMUR

Systolic, **louder with Valsalva or standing**, softer with squatting or handgrip. This is the opposite of aortic stenosis and is the bedside discriminator. Look for a **brisk, bifid carotid upstroke**.

SUDDEN-DEATH RISK *major risk factors from the 2024 guideline*

Major risk factor	Detail
Family history of sudden death	Death definitively or likely attributable to HCM in a **first-degree or close relative aged ≤ 50**.
Massive hypertrophy	**≥ 30 mm** in any LV segment.
Unexplained syncope	**One or more recent episodes** suspected to be **arrhythmic rather than neurocardiogenic**. The distinction is the whole point of the history.
Apical aneurysm	Independent of size — an arrhythmic substrate.
Systolic dysfunction	**LVEF < 50%** — the end-stage burnt-out phenotype.
Extensive fibrosis	Extensive **late gadolinium enhancement** on CMR.

HOW THE ICD DECISION IS MADE

With **one or more major risk factor**, it is useful to discuss the **estimated 5-year sudden-death risk and mortality** as part of **shared decision making** about an ICD. There is no single number that mandates a device — it is a conversation informed by the risk estimate, the patient's values, and device-related risk. **Secondary prevention after cardiac arrest or sustained VT is a clear indication.**

TREAT THE OBSTRUCTION *a stepwise escalation*

1. **Beta-blocker first**, titrated to symptoms — slows the rate, lengthens diastole, and reduces the dynamic gradient.
2. **Non-dihydropyridine calcium channel blocker** (verapamil or diltiazem) if beta-blockers are not tolerated or ineffective. **Avoid in severe obstruction with hypotension.**
3. **Persistent severe symptoms despite a beta-blocker or non-DHP CCB?** Either **add disopyramide** in combination with one of those drugs, or proceed to **septal reduction therapy at an experienced center**.
4. **Cardiac myosin inhibitors** (mavacamten) are now available for **symptomatic obstructive HCM**. They reduce the gradient and improve symptoms; require **EF monitoring and a REMS program**, and interact via CYP metabolism.
5. **Septal reduction** — surgical myectomy or alcohol septal ablation — for refractory symptoms with a gradient ≥ 50 mmHg. Outcomes are strongly operator- and volume-dependent.

WHAT TO AVOID, AND WHAT ELSE TO DO *preload and afterload are the enemy*

AVOID IN OBSTRUCTIVE DISEASE

Vasodilators (nitrates, ACE inhibitors, dihydropyridines, PDE5 inhibitors) — dropping afterload worsens the gradient. **Aggressive diuresis** and dehydration. **Digoxin** and other inotropes. **Treat hypotension with fluids and phenylephrine**, not with an inotrope.

ALSO ADDRESS

Atrial fibrillation is poorly tolerated — loss of atrial filling in a stiff ventricle. Anticoagulate for AF **regardless of CHA_2DS_2-VASc**. **Screen first-degree relatives** with echo and ECG, and offer genetic testing and counseling. **Exercise:** the 2024 guideline is **more permissive** — mild-to-moderate recreational activity is beneficial, and competitive sport is a shared decision at an expert center rather than a blanket ban.

THE OTHER PHENOTYPES *not everyone has obstruction*

Phenotype	Management focus
Non-obstructive HCM roughly a third of patients	No gradient to relieve, so **septal reduction and myosin inhibitors do not apply**. Treat **diastolic dysfunction**, manage AF aggressively, and stratify sudden-death risk identically.
Apical HCM	Deep precordial T-wave inversions. Look for an **apical aneurysm** — a major sudden-death risk factor and a thrombus source.
End-stage / burnt-out	**LVEF < 50%** with wall thinning. Switch to **standard HFrEF GDMT**, which is the one situation where ACE inhibitors and beta-blockers are used conventionally. Refer for **advanced heart failure and transplant assessment**.
Atrial fibrillation	Poorly tolerated at any stage. **Anticoagulate regardless of CHA_2DS_2-VASc.** Prioritize rhythm control; consider early ablation, since rate control alone often fails to restore filling.

FAMILY SCREENING & GENETICS *one diagnosis, a whole pedigree at risk*

SCREEN THE RELATIVES

First-degree relatives need **ECG and echo**. If the proband's genetic variant is known and the relative is **genotype-negative**, they can usually be released from ongoing imaging surveillance. **Genotype-positive but phenotype-negative** relatives need continued periodic screening, since penetrance is age-related.

GENETIC TESTING

Test the **proband first, with genetic counseling**. Most pathogenic variants are in **sarcomere genes**, and a negative panel does not exclude HCM. Testing changes family screening more than it changes the patient's own treatment. Consider **phenocopies** — amyloidosis, Fabry — where specific therapy exists.

PEARLS & PITFALLS

- **A normal resting echo gradient does not exclude obstruction.** If you did not provoke, you did not look.
- **Murmur louder on standing or Valsalva = HCM; louder on squatting = AS.** Reduced preload worsens dynamic obstruction.
- **Never give nitrates for chest pain in known HCM** — you can precipitate profound hypotension.
- **Syncope in HCM is a major risk factor** when arrhythmic. Pin down the circumstances before you call it vasovagal.
- **Anticoagulate HCM with AF regardless of CHA_2DS_2-VASc** — the usual threshold does not apply.

References: 2024 AHA/ACC/AMSSM/HRS/PACES/SCMR Guideline for the Management of Hypertrophic Cardiomyopathy (Circulation / JACC, 2024) · EXPLORER-HCM and VALOR-HCM trial programs.