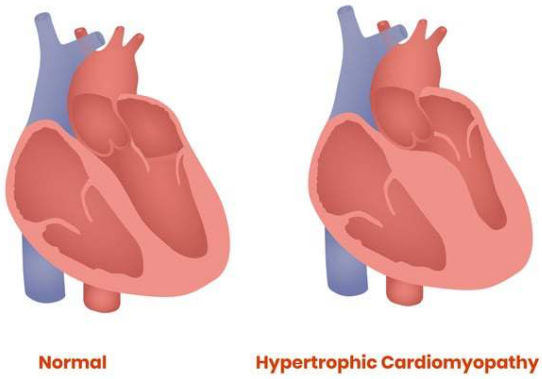
THUMBNAIL GUIDE TO HYPERTROPHIC CARDIOMYOPATHY (HCM)



What is it?

- Most common inherited monogenic cardiac disorder, though all cases are not genetic
- Results from hypertrophy of one or more ventricular segments, most often and predominantly asymmetric septal hypertrophy, which causing outflow obstruction of the left ventricle and therefore slowly exacerbates LV hypertrophy

How common is it?

- Affects 0.2-0.5% of the population, but majority of cases are underdiagnosed.
- It is more common in males than in females
- When diagnosed, prevalence is highest in patients 55–64 years of age

Important prompts to consider diagnosis

- Unexplained syncope
- Presyncope
- Dyspnea, fatigue
- Heart failure
- Lightheadedness or dizziness after exertion
- Chest pain, particualrly on exertion
- Progressive exertional intolerance
- Palpitations/atrial fibrillation
- Family history of HCM and/or sudden death
- May be largely or even completely asymptomatic

Initial evaluation: key components

- Electrocardiogram
- Echocardiogram (exercise echo to provoke outflow gradient if symptom burden is low)
- Cardiac MRI
- Genetic testing
- Ambulatory ECG monitoring
- Cardiac auscultation may reveal
    - Harsh crescendo-decrescendo systolic murmur heard best at left lower sternal border
    - Mid-to-late systolic or holosystolic apical murmur
    - Paradoxically split second heart sound

Differential diagnosis includes

- Hypertensive cardiomyopathy
- Aortic valvular stenosis
- "athlete's heart"
- Genetic "phenocopies" of HCM include
    - Anderson-Fabry disease
    - Pompe disease
    - Danon disease
    - Myocardial amyloidosis
    - AMPK-mediated glycogen storage disease

Confirming the diagnosis

- Demonstration of LV wall thickness $\geq$ 15mm not otherwise explained by abnormal LV loading conditions such as valvular disease, congenital abnormalities, or persistent hypertension
- Unexplained thickness of $\geq$ 13mm is diagnostic in genotype-positive individuals or in relatives of other patients with HCM

Risk stratification: most important to determine indication for ICD implantation

- ESC risk prediction model: https://doc2do.com/hcm/webHCM.html

Symptomatic treatment

- Medical treatment is indicated for NYHA Class > II
    - **beta-blockade** and **verapamil** alone or in combination
        - caution: verapamil may increase LVOTO-associated symptoms in some individuals due to its vasodilatory effect.
    - **Disopyramide** may be added for patients with symptoms refractory to the use of beta-blocker or calcium channel blocker therapy

Disease-targeted therapy: Medical

- Mavacamten: cardiac myosin inhibitor

Disease-targeted therapy: Interventional

- Indicated when medical therapy fails to control NYHA Class III symptoms or following LVOTO-associated syncope or near syncope refractory to medical therapy
- Septal reduction therapy by
    - Alcohol septal ablation
    - Surgical myomectomy
- ICD placement (primary prevention of sudden cardiac death [SCD]) for
    - severe hypertrophy (>30mm)
    - family history of sudden death in a first degree relative
    - recent unexplained syncope
    - "end stage" HCM (LVEF < 50%)
    - Consider strongly in context of other risks if
        - Holter shows nonsustained ventricular tachycardia (NSVT)
        - > 15% late gadolinium enhancement on cardiac MRI
        - apical aneurysm
        - genetic mutations associated with high prevalence of SCD
- ICD placement (secondary prevention) for
    - SCD survivors
    - Sustained VT

Ready references:

- https://www.acc.org/Latest-in-Cardiology/Articles/2020/02/25/06/34/Diagnosis-of-Hypertrophic-Cardiomyopathy#sort=%40commonsortdate%20descending
- https://www.acc.org/Latest-in-Cardiology/ten-points-to-remember/2022/01/24/20/39/Diagnosis-and-Evaluation-of-HCM
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- Earn free CE/CME at https://cardiometabolic-ce.com/category/hcm/