Hypertrophic cardiomyopathy (HCM) is a myocardial disorder defined by ventricular hypertrophy that cannot be explained by another cardiac or systemic disease. The left ventricle is most commonly affected. HCM is a lifelong condition that can worsen over time. It is the most common inherited cardiac disorder.

Could my patient have HCM?
The symptoms of HCM vary from person to person.
- Sometimes people have few or no symptoms and/or no complaints of physical limitations.
- A person may underestimate or under-report their symptoms and the impact on their exercise capacity.
- People may have a family member who died suddenly or prematurely, or has similar symptoms or a diagnosis of HCM.
- Importantly, HCM symptoms can be non-specific, overlapping with other heart conditions and diseases (see below).

HCM is underdiagnosed and misdiagnosed
Anyone can have HCM, regardless of sex, age or race

The overall prevalence of HCM in the USA is estimated as 700,000

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Anyone can have HCM, regardless of sex, age or race

Why does diagnosis matter?
Correct diagnosis is key to ensuring the right monitoring and management arrangements are in place to reduce symptoms and the risk of adverse outcomes.

Common symptoms†
- Dyspnea
- Fatigue
- Palpitations
- Lightheadedness
- Atypical chest pain
† Some individuals do not experience symptoms

Higher mortality
- Sudden cardiac death may be the first sign, but this is rare overall
- 3x increased mortality compared with the general population

Increased risk of:
- Atrial fibrillation
- Stroke
- Heart failure

Reduced function over time
- Activity and quality of life decrease over years to decades

High burden in younger people
- High risk of adverse cardiac outcome by age 60 in those diagnosed before 40

Genetic changes are linked with disease course
- Onset of complications/death is earlier if a known gene mutation is recognized

Heart failure (HF) affects up to 45% of people with HCM. Important differences exist in the clinical profile, pathophysiology, management and outcome between HF in HCM and conventional congestive HF, so accurate diagnosis is important.

In a survey of 444 patients with HCM:*
- 43% were originally diagnosed with an innocent heart murmur
- 25% waited 3 years or longer for a diagnosis of HCM after developing symptoms
- 85% said that, since their diagnosis, their symptoms had increased or interfered with their regular activities

* Salberg et al. 2016.
Systolic anterior motion of the mitral valve leaflets can lead to mitral valve regurgitation. Asymmetric septal hypertrophy partially blocks the outflow tract.

Dilated atria
- Increased ventricular wall thickness
- Asymmetric septal hypertrophy partially blocks the outflow tract

Increased ventricular wall thickness

Stiffening and hypertrophy of the heart can lead to heart failure.

Changes within the cardiomyocyte sarcomere lead to hypercontractility and slower relaxation.

Over time, cardiac tissue stiffens and thickens as a result of the hypercontractility and impaired relaxation. Fibrosis in the heart may also contribute to the stiffening.

Typically, the clinical presentation of HCM is hypercontractility (or hyperdynamic contractions) and impaired relaxation and compliance. Histopathological findings can include myocardial disarray (enlarged, disorganized cardiomyocytes) and fibrosis.

Non-obstructive HCM (30% of people with HCM)

The heart muscle is thickened but this does not interfere with blood flow out of the left ventricle.

The hypertrophy can vary in its distribution: the septum or the apex or bottom of the heart can be affected, or the hypertrophy can be evenly distributed throughout the left ventricle.

The difficulty in relaxation of the cardiac muscle can cause symptoms.

Non-obstructive HCM can be progressive and cause symptoms, including heart failure.

Sources

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