Hypertrophic cardiomyopathy

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).

Be aware that, before a definitive diagnosis is made, symptoms can be mistakenly attributed to:

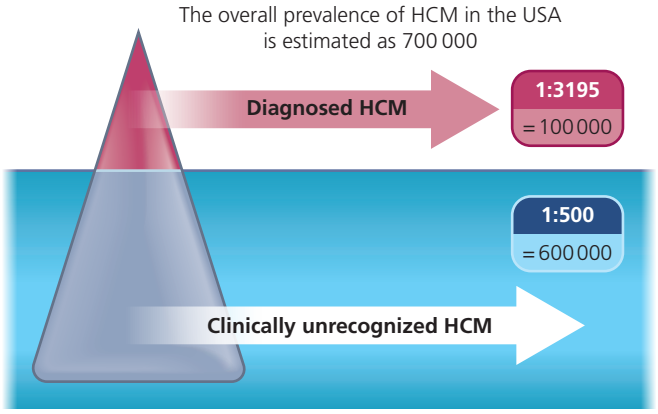
- Asthma
- Coronary artery disease
- Sleep apnea
- Deconditioning
- Chronic obstructive pulmonary disease
- Mitral valve prolapse
- Athlete's heart
- Anxiety

In older people, symptoms may be misattributed to valvular or hypertensive heart disease or heart failure.

HCM is underdiagnosed and misdiagnosed

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

It is likely that only a fraction of people with HCM are correctly identified



HCM is caused by dysfunction of the cardiac sarcomere (the structural unit of the cardiac muscle), mostly as a result of genetic mutations.

Just under half of people with HCM have a pathogenic or likely pathogenic sarcomere mutation. Over 1400 different mutations in 11 genes have been associated with heritable forms of HCM. About 70% of known pathogenic HCM mutations affect the *MYH7* and *MYBPC3* genes, which encode the β-myosin heavy chain and cardiac myosin-binding protein C, respectively.

Mutations that cause HCM lock myosin in an 'on' state, causing hypercontractility and decreased sarcomere relaxation.

In non-familial HCM, a genetic change cannot be identified and there is no family history of HCM. These individuals tend to be older; many also have hypertension and/or a combination of other comorbid factors such as obesity, diabetes and obstructive sleep apnea.

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
- 3× 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.