GECKO
Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

Hypertrophic Cardiomyopathy (HCM)

How does HCM present?

The **onset** (from infancy to adulthood) and presentation of HCM is highly variable, even within families.

Symptoms can include dyspnea, chest pain, palpitations, syncope and, in some cases, sudden cardiac death even in the absence of marked hypertrophy.

Evaluation can be prompted by:

- a positive family history of HCM
- the presence of symptoms such as a cardiac event
- findings on physical examination, such as
  - the detection of a heart murmur, typically a systolic murmur
  - a pronounced apical point of maximal impulse
  - an abnormal carotid pulse
  - the presence of a fourth heart sound
- abnormal findings on an echocardiogram conducted for other reasons
- irregularities on a 12-lead ECG e.g. repolarization abnormalities, arrhythmias

Who should be offered genetic testing?

Genetic testing is recommended for:

- **All** patients with a confirmed or suspected clinical diagnosis of HCM.
- Individuals with a known pathogenic/likely pathogenic HCM-genetic variant in their family, beginning with first-degree relatives.
  - More distant relatives can be considered if intervening relatives are unable or unwilling.
- Deceased individuals with a suspected inherited cardiac condition as part of an autopsy.

Up to 60% of affected individuals will have a positive genetic test result.

HCM is most often an autosomal dominant condition with a 50% risk to all first-degree relatives

Genetic testing is currently offered in a specialty clinic (genetics or cardiology).

Why offer genetic testing?

Genetic testing for HCM can help with:

- Clarification of HCM status for individuals with borderline clinical investigations.
- Initiation of cascade testing of at-risk relatives. This can identify those at risk for HCM and those who are not at risk and would not need ongoing surveillance.
- Positive results can indicate earlier onset of disease and worse outcomes.
- Assistance with life planning (e.g., decisions about careers, participation in competitive sports).
- Provision of relief, to those who test negative for a known familial genetic variant, from worry that they are at greater risk of developing the disease in the future, and knowledge that their children are not at risk of inheriting the predisposition to HCM.

How is HCM managed?

Clinical screening should be individualized.

Echography screening every 1-2 years until the age of 20 and every 3-5 afterwards is reasonable, given the current limited available data.

Referral to an HCM specialist is recommended for treatment and risk stratification (identifying those at greatest risk for sudden cardiac death).

All first-degree relatives should be offered baseline clinical screening by echocardiogram and ECG, regardless of genetic test results. Continued follow-up will be based on genetic test results of the index case and relatives.

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