The Role of Genetic Testing in Hypertrophic Cardiomyopathy[1]

Hypertrophic cardiomyopathy (HCM) is:

- characterized by cardiac hypertrophy in the absence of another cardiac or systemic disease
- said to affect 1 in 500 people
- the most common cause of sudden cardiac death in the young

The evaluation and management of HCM is outlined in the table on the right. The principal role of genetic testing is not to confirm a diagnosis but rather to identify the causative gene in the affected individual and to provide a clinical tool for screening family members at risk of developing the disease. In general, affected individuals and their first degree relatives should be referred to both cardiology and genetics specialists.

For more information on HCM see the GECKO *on the run* in Educational Resources at www.geneticseducation.ca .

Table: Evaluation and management of HCM

Evaluation	• Personal and family history • Physical examination • ECG • 2-dimensional echocardiography
Diagnosis	• Generally established by echocardiography • ECG abnormalities may occasionally precede the onset of left ventricular hypertrophy on the echocardiogram • Distinguish patients with HCM from patients with physiological causes of hypertrophy (e.g., athlete's heart) or infiltrative disorders

Management	• Risk stratification of patients is recommended to determine the risk for Sudden Cardiac Death (SCD)
	• Major risk factors are:
	  • family history of premature SCD
	  • unexplained syncope
	  • non-sustained ventricular tachycardia (VT)
	  • abnormal blood pressure response to exercise
	  • massive left ventricular hypertrophy (maximum left ventricular wall thickness >30 mm)
	• *In patients considered high risk for SCD, an implantable cardioverter-defibrillator (ICD) is indicated*
	• *Exercise restriction is recommended to minimize arrhythmia provocation in high-risk individuals*

Surveillance	• Regular echocardiographic and ECG monitoring • All first-degree relatives (including children) of an affected person should have regular cardiac exams, echocardiograms and ECGs, unless they test negative for a known disease-causing familial mutation

[1] This GECKO POC Tool was adapted from Gollob *et al.*, Recommendations for the Use of Genetic Testing in the Clinical Evaluation of Inherited Cardiac Arrhythmias Associated with Sudden Cardiac Death: Canadian Cardiovascular Society/Canadian Heart Rhythm Society Joint Position Paper. *Canadian Journal of Cardiology* 2011; 27: 232–245.

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