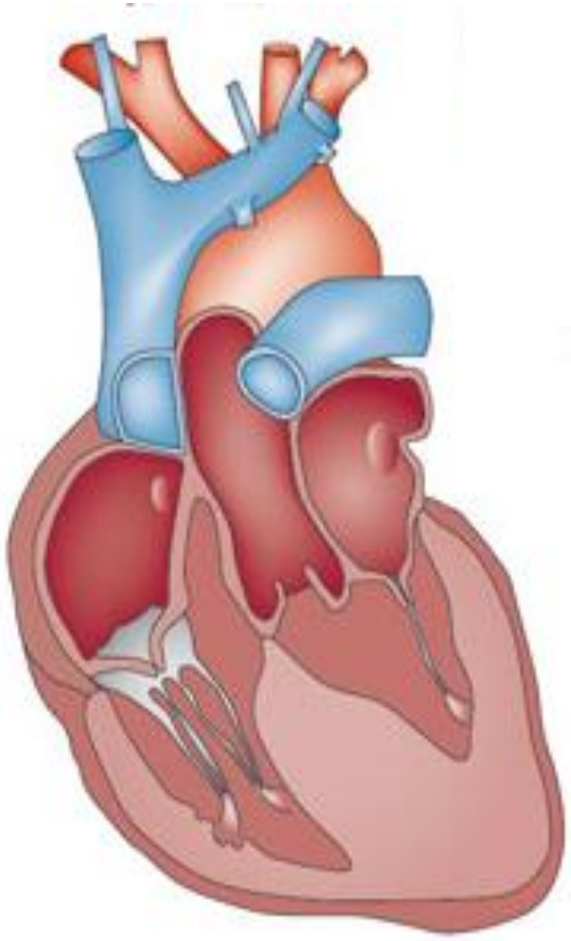
HYPERTROPHIC CARDIOMYOPATHY: MOST COMMON GENETIC HEART CONDITION

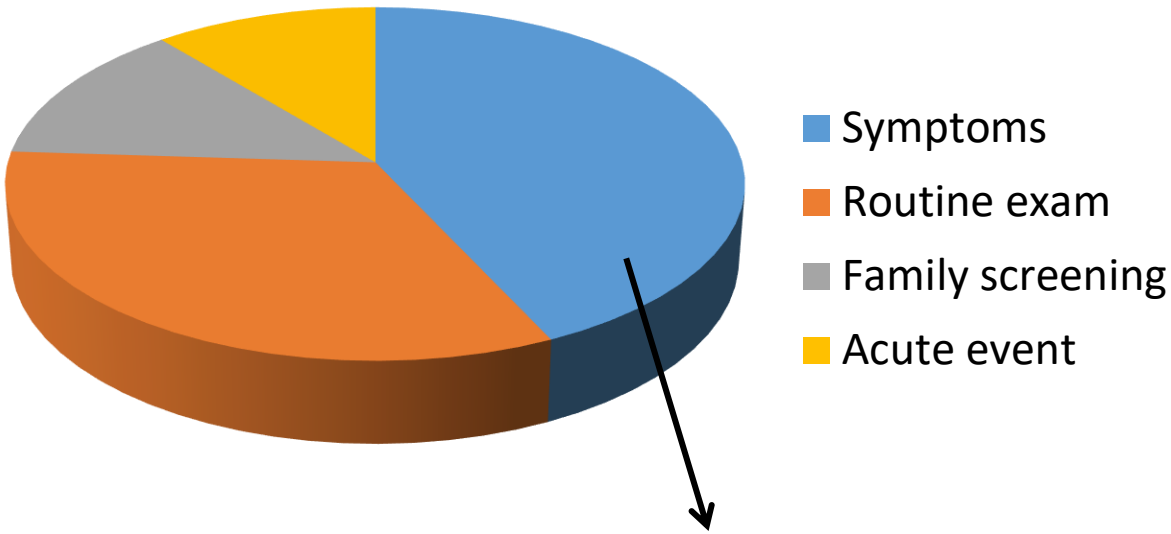


- Unexplained hypertrophy ≥ 15 mm in any segment
- Age of onset and symptoms are highly variable
- Substantial number of patients experience major adverse complications are heart failure, atrial fibrillation and ventricular arrhythmias that can lead to SCD
- However, a subset of patients have no or mild symptoms and normal longevity

CLINICAL PRESENTATION OF HCM

46% are asymptomatic at time of diagnosis

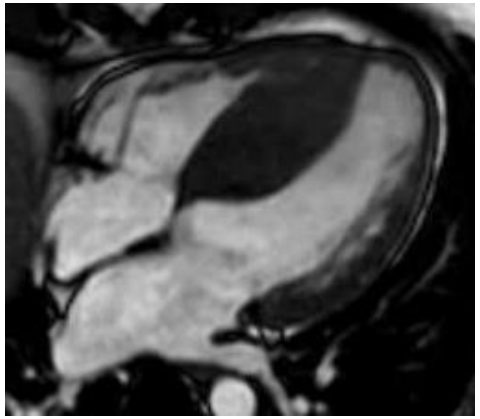
(e.g. murmur, abnormal ECG)



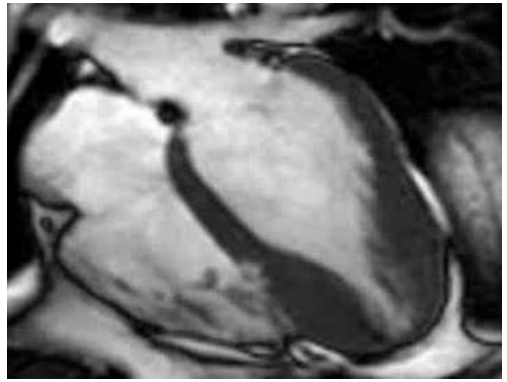
Dyspnea
Chest pain
Syncope/Presyncope
Fatigue
Palpitations

HCM MANIFESTS WITH A WIDE RANGE OF MORPHOLOGIES

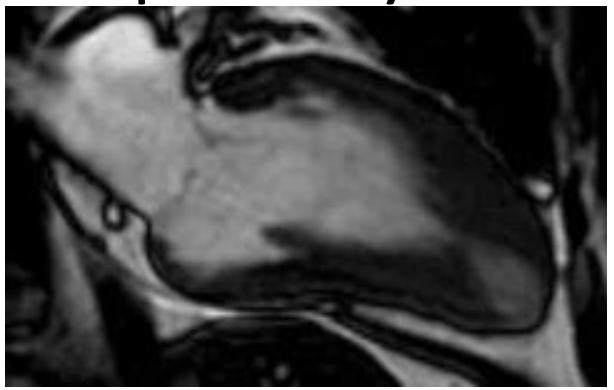
Asymmetric septal



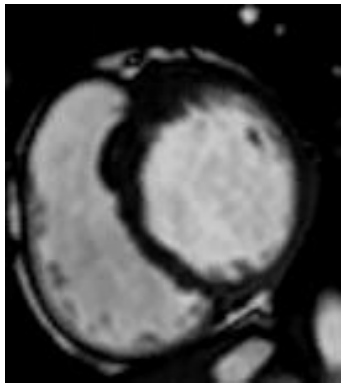
Apical



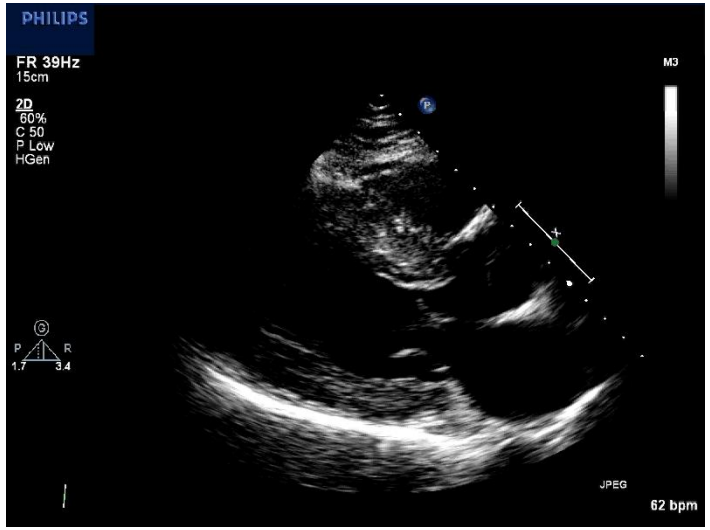
Mid ventricular with apical aneurysm

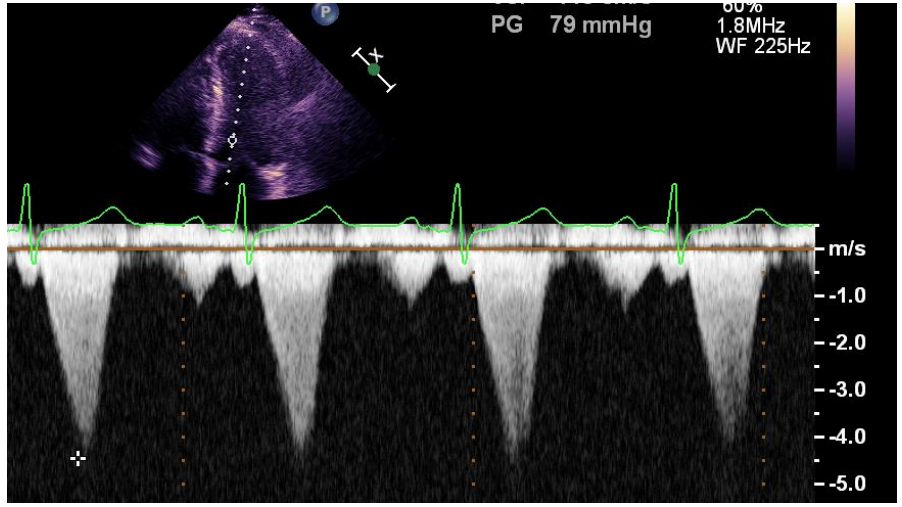


Focal anterior wall



LVOT obstruction
~1/2-2/3 of patients



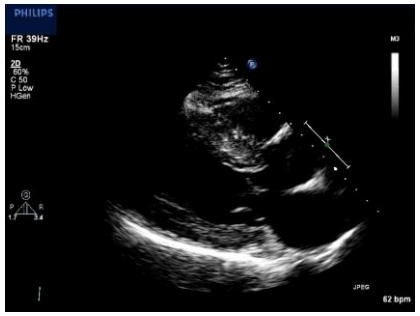


LVOTG = 4v2

IMAGING AND FUNCTIONAL MODALITIES

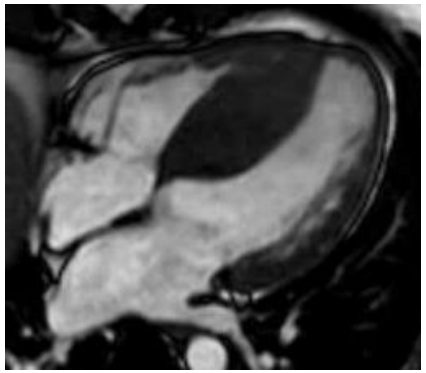
Echocardiography

- Mainstay of diagnosis and serial surveillance
- Function, LVOT gradients, mitral valve SAM and intrinsic disease



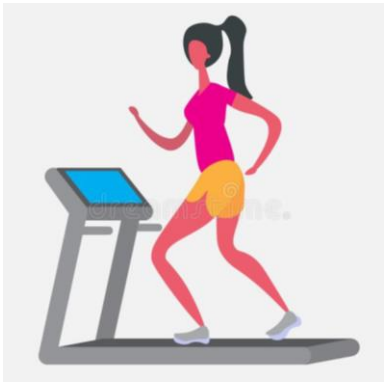
CMR

- Helpful for most patients, at least as one time imaging
- Diagnosis, prognosis, treatment

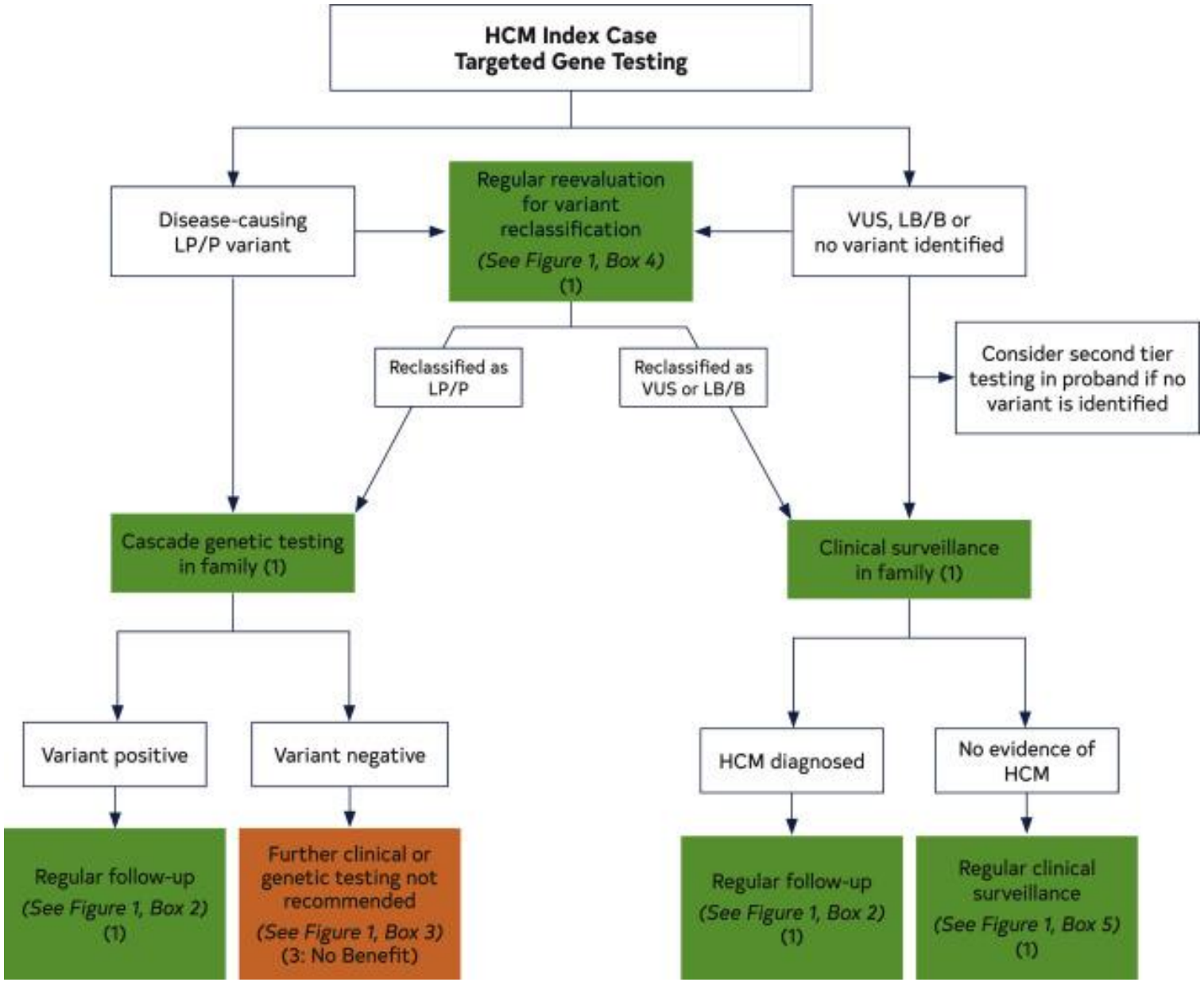


Stress testing – CPET ideal

- Assess cardiorespiratory fitness, symptom progression particularly for patients with advanced HF
- Provocable LVOTO, hemodynamic response, arrhythmias
- Peak VO2 strong prognostic value



GENETIC EVALUATION AND TESTING



- **Genetic evaluation and counseling, comprehensive FH, and offering genetic testing is standard of care.**
- **Diagnostic utility to distinguish HCM from other conditions (i.e storage diseases)**

Ommen et al., Circ and JACC 2020