



Hypertrophic Cardiomyopathy

A Paradigm of Cardiovascular Molecular Genetics

1990 First gene identified

2013 20 genes; 1200 mutations

Implications for screening and risk stratification

Evaluation of HCM by Echocardiography

Echo is GOLD standard for Morphologic/hemodynamic assessment

Guide Management

Risk Assessment/Prognosis

Screening of Family members

Hypertrophic Cardiomyopathy Echocardiographic Diagnosis

Left Ventricular Hypertrophy ≥ 15 mm
(Asymmetric \gg Symmetric)



In the absence of another cardiovascular or systemic disease associated with LVH or myocardial wall thickening

Gersh BJ, et al. JACC. 2011;58:e212 ACC/AHA Guidelines

LV wall thickness

(Extent and Distribution)

- Most common location of hypertrophy is the basal anterior septum.
- Imperative to determine the maximum LV wall thickness irrespective of views.
- If LV borders cannot be accurately delineated, echo contrast is useful.

LV Systolic Function

- Typically have hyper-dynamic LVEF (>65%) and small LV cavities.
- LVEF <50% : “burnt out HCM” (2 – 5%).
- Longitudinal Strain imaging can aid in detection of sub-clinical LV systolic dysfunction.
- Longitudinal strain is useful in early detection of disease.
