

**Harvard Medical School Department of
Continuing Education and the Cardiovascular
Division of the Department of Medicine,
Brigham and Women's Hospital**



Cardiology Rounds
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Contemporary Paradigms of Hypertrophic Cardiomyopathy
Carolyn Ho, MD

Objectives:

This issue of *Cardiology Rounds* will help readers to:

- review the current knowledge regarding the genetic etiology of hypertrophic cardiomyopathy (HCM)
- describe newly recognized causes of inherited HCM that are not related to sarcomere gene mutations
- discuss the implications of genetic-based diagnosis and integration into contemporary patient management
- discuss the potential of genetic advances in furthering our understanding of the early pathophysiology of HCM and their future impact on patient care

Questions: (Only one response is correct)

1. Pathologic hallmarks of hypertrophic cardiomyopathy include:
 - a. myocyte hypertrophy, hemosiderin deposition, scant interstitial fibrosis
 - b. myocyte hypertrophy, pronounced interstitial fibrosis, vacuolization
 - c. myocyte hypertrophy, myocardial disarray, interstitial fibrosis
 - d. myocyte hypertrophy, increased interstitial space, inflammatory cell infiltration

2. The genetic etiology of HCM is:
 - a. x-linked inheritance of sarcomere gene mutations
 - b. autosomal recessive inheritance of genes responsible for glucose energetics
 - c. autosomal dominant or sporadic mutations in genes that encode elements of contractive apparatus (sarcomere).
 - d. autosomal dominant inheritance of mutations in genes responsible for myocyte growth and hypertrophic response to pressure overload

3. Gene mutations associated with hypertrophic cardiomyopathy are:
 - a. small in number and associated with specific clinical characteristics
 - b. small in number and not highly predictive of clinical course
 - c. large in number and not highly predictive of clinical course
 - d. large in number and associated with specific clinical characteristics

4. Development of left ventricular hypertrophy (LVH) in hypertrophic cardiomyopathy is:
- a. invariable and detectable early in life allowing for accurate diagnosis of children in families with HCM
 - b. invariable in adolescents but not infants or young children
 - c. variable in magnitude, but ubiquitously present
 - d. variable in magnitude and in the age, when detectable with abnormalities in diastolic function often preceding LVH.
5. A single comprehensive clinical screening in children or adolescents in families with HCM is adequate to determine the risk of developing the disease
- True False
6. Only members of families with typical autosomal dominant HCM require clinical screening; relatives of individuals with apparently sporadic disease are not at risk for disease development.
- True False
7. The benefits of genetic screening in hypertrophic cardiomyopathy include all except:
- a. determination of the precise genetic etiology and establishing the definitive diagnosis of HCM
 - b. enabling definitive determination of the risk of disease development in family members, irrespective of age or the presence of LVH
 - c. allowing for therapy to be tailored based on the identification of the specific mutation present
 - d. identification of family members at risk for developing overt HCM early in life, in the preclinical/prehypertrophic phase of disease

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