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Cardiology Rounds
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Contemporary Paradigms of Hypertrophic Cardiomyopathy

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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 <u>except</u>:
    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

To receive AMA category 1 credit, you must correctly answer 60% of the test questions.

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