HRS/EHRA Recommendations for Cardiovascular Genetic Testing

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<th>IS RECOMMENDED*</th>
<th>CAN BE USEFUL**</th>
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| **Hypertrophic Cardiomyopathy (HCM)**  
Clinical Diagnosis |  |
| **Dilated Cardiomyopathy (DCM)**  
Clinical diagnosis with cardiac conduction disease and/or family history of premature sudden death |  |
| **Long QT Syndrome (LQTS)**  
Clinical suspicion, or patients with QT interval >480ms (adolescents) or >500ms (adults) |  |
| **Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)**  
Clinical suspicion |  |

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| **Brugada Syndrome (BrS), Type I**  
Clinical suspicion |  |
| **Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)**  
Clinical diagnosis |  |

**Class Ia recommendation**

**American Heart Association**
Scientific Statement on the Genetic Testing for Inherited Cardiovascular Diseases

Genetic testing is informative and useful for the clinical management of various inherited cardiovascular diseases such as:
- Cardiomyopathies
- Arrhythmic disorders
- Thoracic aortic aneurysms and dissections
- Familial hypercholesterolemia

HFSA Guideline #4
Genetic Testing is Recommended for Patients with Cardiomyopathy

WHY
Genetic testing is recommended to determine if a pathogenic variant can be identified to facilitate patient management and family screening. The identification of at-risk family members is critical because the first presentation may be sudden death. Without genetic testing, all family members of a patient with cardiomyopathy need lifetime clinical screening.

WHO
- Test all patients with idiopathic forms of HCM, DCM, ARVC, and RCM, as diagnosed according to the guidelines in GeneReviews1,2,3
- Most severely affected individual in the family with earliest onset disease should be tested first
- Testing “should be considered” for cases of peripartum cardiomyopathy, sudden death

WHEN
- Genetic testing should be initiated when a new cardiomyopathy diagnosis is made
- Probands who test negative should be re-tested when panel sensitivity has increased 5-10% or more

HOW
- Authors advocate for the use of multi-gene panel testing in proband
- Targeted cascade genetic testing should be performed for first-degree relatives when proband is positive

References: