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HRS/EHRA Recommendations for Cardiovascular Genetic Testing[1]

IS RECOMMENDED*

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

** Class I recommendation*

CAN BE USEFUL**

Brugada Syndrome (BrS), Type I
Clinical suspicion

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)
Clinical diagnosis

*** Class IIa recommendation*

American Heart Association
Scientific Statement on the Genetic Testing for Inherited Cardiovascular Diseases[2]

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

HRS℠ Recognized as a quality educational tool by the Heart Rhythm Society.

1. Adapted from Ackerman MJ, *et al*. Heart Rhythm Society/European Heart Rhythm Association expert consensus statement on the state of genetic testing for the channelopathies and cardiomyopathies. Heart Rhythm. 2011 Aug;8(8):1308-39.
2. Musunuru K, *et al.* Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association. Circ Genom Precis Med. 2020 Aug;13(4):e000067.

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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 GeneReviews[1,2,3]**
- 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

Hershberger RE, Givertz M, Ho CY *et al.* Genetic Evaluation of Cardiomyopathy - a Heart Failure Society of America Practice Guideline, Journal of Cardiac Failure (2018), https://doi.org/10.1016/j.cardfail.2018.03.004.

1. Cirino AL, Ho C. Hypertrophic Cardiomyopathy Overview. 2008 Aug 5 [Updated 2014 Jan 16]. In: Adam MP, Ardinger HH, Pagon RA, *et al.*, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018. Available from: ncbi.nlm.nih.gov/books/NBK1768/
2. Hershberger RE, Morales A. Dilated Cardiomyopathy Overview. 2007 Jul 27 [Updated 2015 Sep 24]. In: Adam MP, Ardinger HH, Pagon RA, *et al.*, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018. Available from: ncbi.nlm.nih.gov/books/NBK1309/
3. McNally E, MacLeod H, Dellefave-Castillo L. Arrhythmogenic Right Ventricular Cardiomyopathy. 2005 Apr 18 [Updated 2017 May 25]. In: Adam MP, Ardinger HH, Pagon RA, *et al.*, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018. Available from: ncbi.nlm.nih.gov/books/NBK1131/

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