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HRS/EHRA Recommendations for Cardiovascular Genetic Testing

Clinical diagnosis of hypertrophic cardiomyopathy (HCM)	IS RECOMMENDED*
Clinical diagnosis of dilated cardiomyopathy (DCM) with cardiac conduction disease and/or family history of premature sudden death	IS RECOMMENDED*
Clinical suspicion of long QT syndrome (LQTS), or patients with QT interval >480ms (adolescents) or >500ms (adults)	IS RECOMMENDED*
Clinical suspicion of catecholaminergic polymorphic ventricular tachycardia (CPVT)	IS RECOMMENDED*
Clinical suspicion of Brugada syndrome (BrS), type I	CAN BE USEFUL**
Clinical diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC)	CAN BE USEFUL**
Clinical diagnosis of left ventricular non-compaction (LVNC)	CAN BE USEFUL**

** Class I recommendation* *** Class IIa recommendation*

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

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.

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HFSA Guideline #4

GENETIC TESTING IS RECOMMENDED FOR PATIENTS WITH CARDIOMYOPATHY

WHY TO TEST

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 TO TEST

- 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 TO TEST

- 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 TO TEST

- 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.

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