HRS/EHRA Genetic Testing Guidelines and HRS/EHRA/ APHRS

Guidelines for Management of Arrhythmia Syndromes (HRS/EHRA/APHRS expert consensus

Silvia G Priori 1, Arthur A Wilde, Minoru Horie, Yongkeun Cho, Elijah R Behr, Charles Berul, Nico Blom, Josep Brugada, Chern-En Chiang, Heikki Huikuri, Prince Kannankeril, Andrew Krahn, Antoine Leenhardt, Arthur Moss, Peter J Schwartz, Wataru Shimizu, Gordon Tomaselli, Cynthia Tracy; ***Management of Arrhythmia Syndromes (HRS/EHRA/APHRS expert consensus statement on the diagnosis and management of patients with inherited primary arrhythmia syndromes: document endorsed by HRS, EHRA, and APHRS in May 2013 and by ACCF, AHA, PACES, and AEPC in June 2013 Heart Rhythm. 2013 Dec; 10(12):1932-63. doi: 10.1016/j.hrthm.2013.05.014.***)

LQTS

1. Comprehensive or targeted LQT1-3 testing

- Class I: •Patients with a strong clinical suspicion of LQTS by means of clinical history, family history, ECG and stress test(s); •Asymptomatic QTc prolongation (>500ms in adults) in the absence of other conditions that may prolong QTc; • Family mutation-specific testing after identification in an index case
- Class IIb: •Asymptomatic QTc prolongation (>480ms in adults)

BrS

- Class I: •Family mutation-specific testing after identification of a BrS-causative mutation in an index case
- Class IIa: •Comprehensive or targeted testing in patients with a strong clinical suspicion of BrS by means of clinical history, family history, ECG and stress test(s)
- Class III: •Not indicated in the setting of an isolated type 2 or type 3 BrS ECG

CPVT

- Class I: • Comprehensive or targeted testing in patients with a strong clinical suspicion of CPVT by means of clinical history, family history, ECG and stress test(s) • Family mutation-specific testing after identification of the CPVT-causative mutation in an index case

IVF

- Class IIa: • Genetic testing can be useful when there is a suspicion of a specific genetic disease following clinical evaluation of the IVF patient and/or family members.
- Class III: • Genetic screening of a large panel of genes in IVF patients in whom there is no suspicion of an inherited arrhythmogenic disease after clinical evaluation should not be performed.

Molecular Autopsy

- Class I • Collection of a tissue sample is recommended in all SADS cases
  •Mutation-specific genetic testing is recommended for family members following identification of a pathogenic mutation in the decedent (proband)
  •Targeted post-mortem genetic testing is recommended if circumstantial evidence suggests LQTS or CPVT Class IIa: Comprehensive post-mortem genetic testing of an arrhythmia syndrome panel can be useful.