Supplemental Information

GUIDANCE DOCUMENT FOR CLINICAL EVALUATION OF FAMILY MEMBERS OF SUDDEN UNEXPLAINED DEATH

Instructions

- You may be caring for a patient who is a family member of a sudden unexplained death victim. The purpose of this document is to explain to medical providers the current recommendations for the clinical evaluation of family members of sudden unexplained death.
- This document is associated with an observational registry called the Sudden Death in the Young (SDY) Family Sub-study, but the tests listed are not required for research purposes. Tests should be ordered at your discretion for the medical evaluation of your patient.
- Recommendations in this document are based on the 2013 HRS/EHRA/APHRS Expert Consensus Statement on the Diagnosis and Management of Patients with Inherited Primary Arrhythmia Syndromes. (Priori S et al. *Heart Rhythm*, 2013). An algorithm from those recommendations is included on page 2.

Frequently Asked Questions

Q1. Who Should Be Evaluated?

A1. All first-degree relatives should be evaluated along with other more distantly related individuals who have a history of suspicious symptoms. Examples of suspicious symptoms include syncope, near syncope, palpitations, prior cardiac arrest, and seizures. Also, any obligate carriers should be evaluated. An obligate carrier is a family member who must be a mutation carrier based on their relationship to other family members that are known to be affected.

Q2. What Tests Are Recommended for All Patients Being Evaluated?

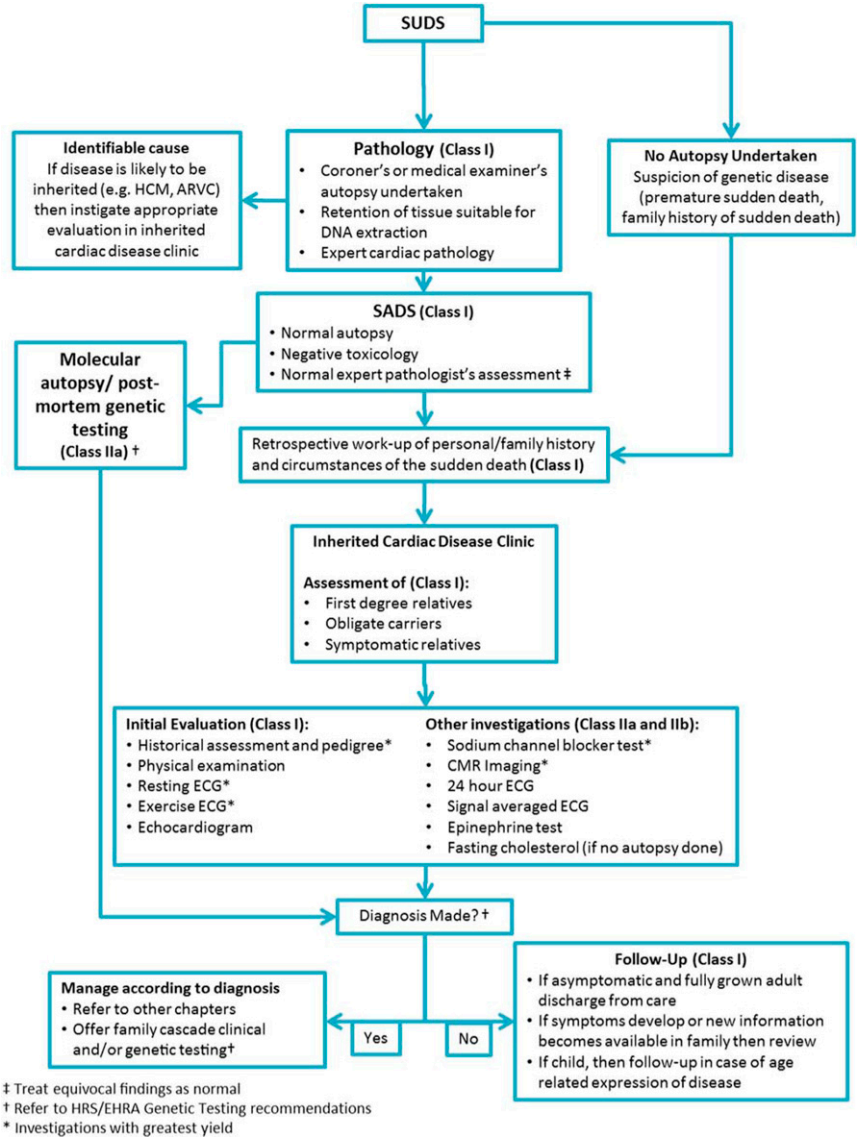
A2. A Class I recommendation is for all patients to have a history and physical exam, resting electrocardiogram, exercise electrocardiogram, and transthoracic echocardiogram.

Q3. How Do I Decide on Additional Testing?

A3. Additional tests include a sodium channel blocker (i.e. procainamide) challenge, cardiac MRI, 24–48 hour ambulatory ECG (i.e. Holter) monitor, signal averaged ECG (SAECG), catecholamine (i.e. epinephrine) challenge, and fasting cholesterol panel. Some additional tests may become unnecessary if the results of prior testing point to a specific diagnosis. For example, if a sodium channel blocker challenge reveals a diagnosis of Brugada Syndrome, additional testing with a cardiac MRI to look for Arrhythmogenic Right Ventricular Cardiomyopathy may be unnecessary.

Q4. When Should Genetic Testing Be Ordered?

A4. Recommendations for genetic testing can be found in the 2011 HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for Channelopathies and Cardiomyopathies (Ackerman MJ et al. Heart Rhythm, 2011). If a disease-causing mutation has been previously found in the family, targeted genotyping for that mutation should be performed in all family members. If no prior mutation has been found, sequencing of a panel of genes may be indicated if evidence exists to suggest a specific diagnosis. Commercial genetic testing companies may be used to perform targeted genotyping and assist in selection of specific gene panels for sequencing.



SUPPLEMENTAL FIGURE 1

Algorithm for evaluation of relatives of sudden unexplained death victims. (Adapted from Priori SG, Wilde AA, Horie M, et al. 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;10[12]:1932–1963).