Family Screening Guidelines for Cardiomyopathies

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Evaluation of the patient with heart failure or cardiomyopathy

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Family screening — The 2009 HFSA guideline recommends screening first-degree relatives (parents, siblings, children) of patients with cardiomyopathy (including all types for which the guidelines recommend detailed family history). These guidelines are supported by evidence that cardiomyopathy is frequently familial and that affected family members are frequently asymptomatic [27]. Progressive disease may occur within a relatively short period of time in initially asymptomatic family members with abnormal electrocardiographic or echocardiographic findings [28-30]

The following screening is recommended for first-degree relatives of patients with cardiomyopathy [27]:

● Clinical screening for cardiomyopathy in asymptomatic first-degree relatives is recommended whether or not genetic testing has been undertaken, and whether or not a genetic cause has been found if genetic testing was performed. Screening should include the following:

● History (with special focus on HF symptoms, arrhythmias, pre-syncope, and syncope)
• Physical examination (with special attention to the cardiac and skeletal muscle systems)
• Electrocardiogram (ECG)
• Echocardiogram
• Creatine kinase MM fraction (at initial evaluation only)
• Signal-averaged ECG in ARVC only
• Holter monitoring in HCM and ARVC
• Exercise treadmill testing in HCM
• Cardiovascular magnetic resonance imaging in ARVC

• Asymptomatic first-degree relatives with negative clinical and/or genetic screening should be rescreened at intervals or any time that symptoms or signs appear. The frequency of recommended rescreening varies with cardiomyopathy type:
  • HCM – Every three years until 30 years old, except yearly during puberty
  • DCM – Every three to five years beginning in childhood
  • ARVC – Every three to five years after age 10
  • LVNC – Every three years beginning in childhood
  • RCM – Every three to five years beginning in adulthood

More frequent screening is recommended if a mutation is present.

• Repeat clinical screening at one year is suggested in first-degree relatives with any abnormal clinical screening tests.