The American College of Cardiology Foundation (ACCF) and the American Heart Association (AHA) have updated guidelines on the diagnosis and treatment of hypertrophic cardiomyopathy (HCM). This summary focuses on portions of the recommendations that pertain to genetic testing strategies, screening of patients and family members, and monitoring of asymptomatic patients.

HCM is a common genetic cardiovascular disease that affects approximately one out of every 500 persons. Generally, the condition is characterized by unexplained left ventricular (LV) hypertrophy associated with nondilated ventricular chambers, without the presence of another cardiac or systemic disease that, by itself, would be capable of producing the extent of hypertrophy seen in a given patient (with the caveat that patients who are positive for HCM on genetic testing may have no observable characteristics of hypertrophy).

Clinically, HCM is usually recognized in a patient by a maximum LV wall thickness of 15 mm or greater. In children, increased LV wall thickness is defined as being two or more standard deviations above the mean wall thickness for the child’s age, sex, or body size. In principle, however, any degree of wall thickness is compatible with the presence of genetic HCM. Although there are a variety of patterns and distributions of LV hypertrophy associated with HCM, about one-third of patients have largely segmental wall thickening that involves only a small portion of the left ventricle; such patients with HCM usually have a normal calculated LV mass.

Differential diagnosis of HCM and other cardiac conditions with LV hypertrophy may arise, most often with hypertensive heart disease, or with the physiologic remodeling that can result from intense athletic training, usually when maximum wall thickness is between 13 and 15 mm. Noninvasive markers, such as sarcomeric mutations, family history of HCM, LV cavity dimension, diastolic function, a pattern of LV hypertrophy, or short deconditioning periods, often help resolve these distinctions.

Certain disorders can mimic clinically diagnosed HCM. These include metabolic or infiltrative storage disorders with LV hypertrophy in infants, older children, and young adults, such as mitochondrial disease, Fabry disease, or storage diseases caused by genetic mutations. HCM should not be used to describe the diagnosis in these patients, or in those with LV hypertrophy that occurs in the context of a multisystem disorder. When a patient with HCM presents in the end stages of the disease, the differential diagnosis may require distinction from dilated cardiomyopathy.

Genetic Testing Strategies and Family Screening Recommendations

The assessment of patients with HCM should include genetic counseling and an evaluation of familial inheritance. Patients who undergo genetic testing should receive counseling from someone knowledgeable in the...
genetics of cardiovascular disease, so that patients can fully comprehend the results and clinical significance of such testing. Testing for HCM and other genetic causes of unexplained cardiac hypertrophy should be performed in patients with an atypical clinical presentation of HCM, or when another genetic condition is suspected of causing it.

Genetic testing of the index patient may help identify first-degree family members who are at risk of developing HCM. Clinical screening, with or without genetic testing, is recommended in first-degree relatives of patients who have HCM. Genetic testing is not indicated in relatives of index patients who do not have a definite pathogenic mutation. Relatives who are members of families with HCM, but who have negative genetic test results, do not require ongoing clinical screening. The usefulness of genetic evaluation in assessing the risk of sudden cardiac death in HCM has not been determined.

**Electrocardiography and Echocardiography Recommendations**

Twelve-lead electrocardiography is recommended in the initial evaluation of patients with HCM, and as a screening component for first-degree relatives of those with HCM. To detect ventricular tachycardia and identify patients who may be candidates for implantable cardioverter-defibrillator therapy, the initial evaluation of patients with HCM should include 24-hour ambulatory electrocardiographic monitoring. Screening with 12-lead electrocardiography is also recommended for adolescent first-degree relatives of patients with HCM who have no evidence of hypertrophy on echocardiography. Twenty-four–hour monitoring or event recording is recommended in patients with HCM who develop palpitations or lightheadedness. Repeat electrocardiography is recommended for patients with HCM when there is worsening of symptoms.

Transthoracic echocardiography is recommended in the initial evaluation of all patients with suspected HCM, and as a screening component for family members of patients with HCM (unless the family member is genotype negative in a family that has known definitive genetic mutations for HCM). Patients with HCM who have a change in clinical status or a new cardiovascular event should undergo repeat transthoracic echocardiography. Periodic screening (every 12 to 18 months) is recommended for children of patients with HCM, starting at 12 years of age. Screening may be initiated before 12 years of age if the child experiences a growth spurt; if signs of puberty are evident; if the child is planning to engage in intense competitive sports; or if there is a family history of sudden cardiac death.

**Stress Testing Recommendations**

Treadmill exercise testing is a reasonable method of determining functional capacity and response to therapy.

**Guidelines for Asymptomatic Patients**

For patients with HCM, comorbidities that may contribute to cardiovascular disease, such as diabetes mellitus, hyperlipidemia, hypertension, and obesity, should be treated in compliance with relevant existing guidelines.

Low-intensity aerobic exercise is a reasonable component of a healthy lifestyle for patients with HCM.

The usefulness of beta blockers and calcium channel blockers in managing HCM in asymptomatic patients with or without obstruction has not been clearly established.

Regardless of the severity of obstruction, septal reduction therapy should not be performed for asymptomatic patients who have HCM with normal effort tolerance. In patients who have HCM at rest or with provokable outflow tract obstruction, pure vasodilators and high-dose diuretics are potentially harmful, regardless of symptom status. ■

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**Answers to This Issue’s CME Quiz**

| Q2. B | Q6. C | Q10. A, D |