DEFINITIONS

Hypertrophic cardiomyopathy (HCM) is a genetic disorder characterized by ventricular hypertrophy in the absence of an identifiable cause for the hypertrophy (1–4). The hypertrophy is usually asymmetric and involves the interventricular septum in 90% of patients. Most commonly, the outflow septum (i.e., subaortic area) is the major focus.
of hypertrophy, but the midventricular or apical septum may occur in isolation or concomitantly. Rarely (5%) the right ventricle is involved. The extent of the hypertrophy varies tremendously and accounts for different manifestations of the disease (1-5). The hypertrophy is associated with myocardial fiber disarray on microscopy.

The disorder is an inherited autosomal dominant mutation or arises by spontaneous mutation. HCM is a heterogeneous disorder of the sarcomere, and to date 10 genes and more than 200 mutations have been described affecting β-myosin heavy chain, cardiac myosin-binding protein C, cardiac troponin T, troponin I, α-tropomyosin, regulatory myosin light chains, actin, and titin (4,6,7). Although not practical as yet, genetic screening may provide better detection and outcome prediction. The troponin T and some β-myosin heavy-chain mutations have been associated with a poor outcome.

PREVALENCE

Within the general population, HCM may affect as many as 1 per 500 individuals and represents about one-quarter the prevalence of all forms of congenital heart disease (3,4,7). However, many individuals with HCM are asymptomatic and may remain so indefinitely.

CLASSIFICATION OF HCM

The clinical manifestations of HCM are varied and dependent on the extent and location of the hypertrophy and the secondary effects of the hypertrophy (8). Based on hemodynamic criteria, patients may have either an obstructive or nonobstructive form of the disease (Table 1). The obstruction may occur at rest, with provocation (latent), or intermittently (labile). The degree of obstruction varies directly with the inotropic state of the heart and inversely with the systemic vascular resistance and preload.

CLINICAL PRESENTATION

The clinical manifestations of HCM are extremely varied. Many patients are asymptomatic (3-5,9). Symptoms in children are very uncommon, although they are not immune to sudden death (10,11). Symptoms may first develop during adolescence, possibly because of an accelerated growth of myocardial hypertrophy. Most patients become symptomatic in their fourth or fifth decade. Because of the dynamic nature of the obstructive form of HCM, symptoms will vary in intensity from day to day, depending on normal variation in periph-