



# HYPERTROPHIC CARDIOMYOPATHY

## Hypertrophic Cardiomyopathy HCM

Hypertrophic cardiomyopathy (HCM) is an uncommon heart muscle disease, characterized by an abnormal increase in the thickness of the left ventricle, the major pumping chamber of the heart. This increase in mass or "hypertrophy" occurs with an external cause such as high blood pressure or valvular heart disease. Although the pumping function of the thickened heart is usually normal, the ability of the heart muscles themselves is abnormal, and the hypertrophy may cause "obstruction" or blockage of the blood flow out of the heart, so called hypertrophic obstructive cardiomyopathy.

Occurring with a prevalence of between 0.1 and 1.0 people per 1000 population, HCM presents with a wide clinical spectrum, ranging from a completely benign course with no symptoms to severe heart failure and sudden cardiac death, HCM is the most common cause of sudden cardiac death in otherwise healthy young individuals such as athletes. Arrhythmias are common, and range from occasional extra beats to more serious and life threatening forms.

Diagnosis of HCM usually occurs during evaluation of symptoms such as shortness of breath, palpitations, chest pain or fainting episodes. The diagnosis is usually suspected by the presence of a heart murmur or abnormal cardiogram (EKG), and is usually confirmed by an ultrasound examination of the heart (echocardiogram.) The identification of the genetic basis of the disease means that more asymptomatic people with HCM are being diagnosed by echocardiographic screening of family members of affected individuals. Treatment is currently based on symptoms and the severity of family history, i.e. incidence of sudden death. There is no strict correlation between the degree of thickening of the left ventricle and risk for sudden death. Medical therapy is still the primary treatment, although some patients may require permanent pacemakers or implantable defibrillators.

Advances in molecular genetics have enabled researchers to establish that HCM is a genetic disease, but the pattern of inheritance and clinical expression of the disease are complex. It is inherited as an "autosomal dominant" trait, meaning that 50% of children from an affected parent will inherit the abnormal gene. It is very important to note, however, that the disease has variable penetrance." This means that the abnormal gene does not express itself as HCM in every person who inherits the abnormal gene. Therefore, many people will inherit the gene for HCM but will not develop the disease. Further complicating matters, HCM is not a single genetic disease. To date, three genes have been linked to HCM and additional linked sites on chromosomes have been identified. Within each associated gene, multiple distinct mutations can cause the disease, and may cause differing severity of the disease. Even within family members sharing the same mutation, the disease may present differently, indicating a role for environmental or other genetic factors. Finally, not all patients with HCM inherit the abnormal gene. Spontaneous gene mutations clearly occur, but the true frequency of sporadic (not inherited) cases is unknown.

Unfortunately, the genetic defects identified to date likely account for only a minority of cases of HCM, and large scale genetic testing is not yet available. Further genetic research will lead to better and earlier diagnosis of HCM, increase our understanding of the various clinical manifestations of the disease, and possibly lead to effective prevention and therapy for those with abnormal HCM genes.

For more information about Hypertrophic cardiomyopathy, ask your physician, or contact the C.A.R.E. Foundation at (800) 404-9500