What is hypertrophic cardiomyopathy (HCM)?

Cardiomyopathy is a general term describing any condition in which the heart muscle is structurally and functionally abnormal (the heart itself is, of course, a specialized type of muscle). While there are many types of cardiomyopathy, many of which are genetic and familial, we are concerned here with only ***hypertrophic cardiomyopathy (HCM)***.

HCM is a genetic disease affecting the heart muscle. The most consistent feature of HCM is excessive thickening of that portion of the heart muscle known as the left ventricle (heart muscle thickening = *hypertrophy*; diseased heart muscle = *cardiomyopathy*). In quantitative terms, hypertrophy is usually defined as a wall thickness of 15 mm or more when measured by ultrasound (echocardiogram). The consequences of HCM to patients are related, in part or solely, to the abnormally thickened left ventricular heart muscle which in turn is a consequence of the basic genetic defect. Hypertrophy may be widespread throughout the left ventricle, but may also be more limited in distribution, and there is no single pattern of muscle thickening which is "typical" of HCM. The region of the left ventricle which is usually the site of the most prominent thickening is the ventricular septum; that is, that portion of muscle which separates the left and right ventricular cavities.

The heart (specifically the left ventricle) may also thicken in other individuals who do not have HCM, either as a result of high blood pressure, obstructive heart valve disease, or even prolonged and intense athletic training in certain sports. The type of hypertrophy associated with high blood pressure is often referred to as secondary (i.e., a consequence of the increased blood pressure). In HCM, however, the muscular thickening of the heart wall is *primary* – that is, due to a genetic defect and not a reaction to other factors.

In addition, when the heart muscle of HCM is viewed under a light microscope, it usually shows several particular abnormalities, the most prominent of which is called ***myocardial cell (myocyte) disarray or disorganization*** (Figure 1), in which normal parallel alignment of heart muscle cells has been lost and many of the muscle cells are arranged in a characteristically chaotic and disorganized pattern. It is likely that this cell disarray interferes with normal electrical transmission of impulses and predisposes some patients to irregularities of heart rhythm, as well as

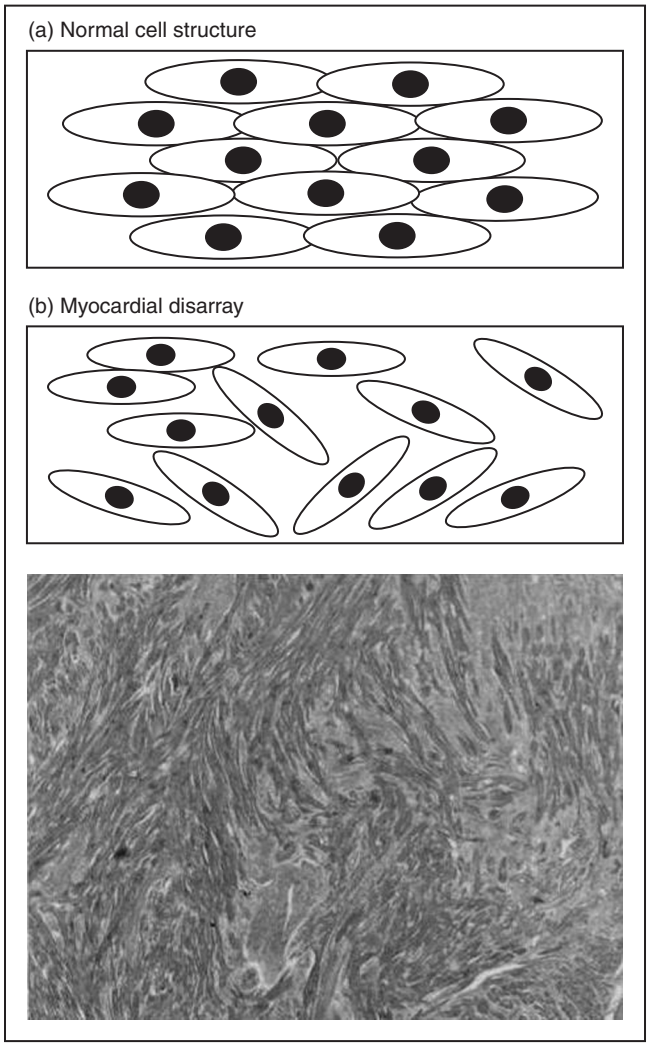

Figure 1 *The cell structure and architecture of the HCM heart.* Diagrams contrast the regular and parallel alignment of muscle cells in the normal heart (a) with the irregular, disorganized alignment of cells ("myocardial disarray") found in some areas of the HCM heart (b). At the bottom is a micrograph of an actual area of an HCM heart (from a histologic section) showing the disorganized and chaotic arrangement of cardiac muscle cells (myocytes).

altering the heart contraction. In addition, there are often scars (comprised of collagen; i.e., fibrosis of various size and extent within the wall of the left ventricle), which probably result from inadequate blood supply to the heart muscle.

Historical perspective and names

The first modern description of HCM was in 1958 by a British pathologist, Dr. Donald Teare, who likened the disease to a tumor of the heart. However, there is some evidence that HCM was initially recognized in the mid-1850s by German and French investigators. Nevertheless, over these many years the condition has been known by a vast number of names. Indeed, this issue of nomenclature *is* often confusing to patients and even some physicians (Figure 2).

Remarkably, HCM has been given over 75 separate names or designations by individual clinicians and scientists over the last almost 50 years (Figure 2). Literally, no other disease can make that claim. Why has this occurred? The principal reason for the proliferation of names undoubtedly has been the heterogeneity and diversity with which HCM is expressed, a major point in ultimately understanding this disease. Also, since very few cardiologists have treated large numbers of patients with HCM, they often came to regard the overall disease based solely on their personal (and sometimes limited) experiences.

Many of the alternate names for HCM emphasize obstruction to left ventricular outflow, which is a highly visible feature of the disease. Obstruction is probably present under resting conditions in just 25% of all patients; however, about 70% of all HCM patients have the capacity for obstruction, either at rest or (if not present at rest) when provoked by physiologic exercise. Therefore, names for this disease have included IHSS (or idiopathic hypertrophic subaortic *stenosis*) which was the first popular term used in the United States ("stenosis" means obstruction). The same can be said for HOCM (hypertrophic obstructive cardiomyopathy) which is still widely used in the United Kingdom. Indeed, you may well hear your disease referred to by more than the designation … HCM.

Presently, virtually all HCM experts and other cardiovascular specialists now regard *hypertrophic cardiomyopathy* (or *HCM*) as the best single name for the broad disease spectrum. This term emphasizes the *hypertrophy* which is the diagnostic marker in most patients and the fact that this disease is a *cardiomyopathy* – or heart muscle disorder – and without mentioning obstruction (which is *not* present in each patient). Therefore, the terms "HCM *with* obstruction" or "HCM *without* obstruction" are preferred.

Terms used to describe hypertrophic cardiomyopathy

Acquired aortic subvalvular stenosis	Hypertrophic apical cardiomyopathy	Low subvalvular aortic stenosis
Apical asymmetric septal hypertrophy	HYPERTROPHIC CARDIOMYOPATHY (HCM)	Mid-ventricular hypertrophic cardiomyopathy
Apical hypertrophic cardiomyopathy	Hypertrophic constrictive cardiomyopathy	Mid-ventricular hypertrophic obstructive cardiomyopathy
Apical hypertrophic nonobstructive cardiomyopathy	Hypertrophic disease	Mid-ventricular obstruction
Apical hypertrophy	Hypertrophic hyperkinetic cardiomyopathy	Muscular aortic stenosis
Asymmetric left ventricular hypertrophy	Hypertrophic infundibular aortic stenosis	Muscular hypertrophic stenosis of the left ventricle
Asymmetric septal hypertrophy	Hypertrophic nonobstructive apical cardiomyopathy	Muscular stenosis of the left ventricle
Asymmetrical apical hypertrophy	Hypertrophic nonobstructive cardiomyopathy	Muscular subaortic stenosis
Asymmetrical hypertrophic cardiomyopathy	Hypertrophic nonobstructive cardiomyopathy with giant negative T-waves	Muscular subvalvular aortic stenosis
Asymmetrical hypertrophy of the heart	Hypertrophic obstructive cardiomyopathy	Non-dilated cardiomyopathy
Asymmetrical septal hypertrophy (ASH)	Hypertrophic obstructive cardiomyopathy of the left ventricle	Nonobstructive hypertrophic cardiomyopathy
Brock's disease	Hypertrophic restrictive cardiomyopathy	Obstructive cardiomyopathy
Diffuse muscular subaortic stenosis	Hypertrophic stenosing cardiomyopathy	Obstructive hypertrophic aortic stenosis
Diffuse subvalvular aortic stenosis	Hypertrophic subaortic stenosis	Obstructive hypertrophic cardiomyopathy
Dynamic hypertrophic subaortic stenosis	Idiopathic hypertrophic cardiomyopathy	Obstructive hypertrophic myocardiopathy
Dynamic muscular subaortic stenosis	Idiopathic hypertrophic obstructive cardiomyopathy	Obstructive myocardiopathy
Familial hypertrophic subaortic stenosis	Idiopathic hypertrophic subaortic stenosis (IHSS)	Pseudoaortic stenosis
Familial hypertrophic cardiomyopathy	Idiopathic hypertrophic subvalvular stenosis	Stenosing hypertrophy of the left ventricle
Familial muscular subaortic stenosis	Idiopathic muscular hypertrophic subaortic stenosis	Stenosis of the ejection chamber of the left ventricle
Familial myocardial disease	Idiopathic muscular stenosis of the left ventricle	Subaortic hypertrophic obstructive cardiomyopathy
Functional aortic stenosis	Idiopathic myocardial hypertrophy	Subaortic hypertrophic stenosis
Functional aortic subvalvular stenosis	Idiopathic stenosis of the flushing chamber of the left ventricle	Subaortic idiopathic stenosis
Functional hypertrophic subaortic stenosis	Idiopathic ventricular septal hypertrophy	Subaortic muscular stenosis
Functional obstructive cardiomyopathy	Irregular hypertrophic cardiomyopathy	Subvalvular aortic stenosis
Functional obstruction of the left ventricle	Left ventricular muscular stenosis	Subvalvular aortic stenosis of the muscular type
Functional obstructive subvalvular aortic stenosis		Teare's disease
Functional subaortic stenosis		Typical hypertrophic obstructive cardiomyopathy
Hereditary cardiovascular dysplasia		

Figure 2 HCM has acquired many names (about 75) in four decades which reflects the diversity with which the disease is expressed. Hypertrophic cardiomyopathy is the preferred name at this time.