Hypertrophic cardiomyopathy: through a window of 50 years

Hipertrofik kardiyomiyopati: 50 yıllık pencereden bir bakış

Next year, 2007 will mark the 50th anniversary of the modern description of hypertrophic cardiomyopathy (HCM) by 2 seminal papers. The first was from Sir Russell Brock of Guys Hospital, London, who described functional obstruction of the left ventricle (1). The second, from Dr. Donald Teare of the Department of Pathology of St. George's Hospital, London described asymmetrical hypertrophy of the heart in young adults (2). Drs. Teare and Brock's papers make excellent reading 50 years later. In one, the surgeon encounters subaortic stenosis in 3 patients initially thought to have valvular aortic stenosis. In the second, the pathologist starkly describes 9 young patients who died suddenly with dramatically abnormal thickening of the septum, so thick it might be confused with tumor.

In Dr. Teare's case descriptions, we recognize many features of HCM encountered in clinical practice: many are discussed in detail in this supplement of the AKD. The first, sudden death, is HCM's most catastrophic complication. Those who care for HCM patients are particularly focused on this complication because young patients may be exposed to this risk for many years and because of difficulty predicting which patients are susceptible. The association of sudden death with exertion clearly described by Teare has stood the test of time. Hypertrophic cardiomyopathy is perhaps most famous because of its association with sudden cardiac death in young athletes. Although risk factors for sudden cardiac death have been identified, the positive predictive value of these risk factors is low; the presence of multiple risk factors increases risk. In patients deemed to be at high risk the implantable defibrillator has been shown to be effective in primary prevention of sudden death (3). In HCM centers in the US, device implantation approaches 20% of patients. But, our ability to prevent death from ventricular fibrillation with the defibrillator exceeds our ability to predict who is likely to benefit from its implantation. A future challenge is accurate identification of patients who are truly at high risk.

Dr. Teare noted an association with atrial fibrillation, and clinical deterioration with its onset. Similarly, he noted HCM's association with stroke when atrial fibrillation appears. Finally, the inherited nature of HCM was noted; a family with 3 affected members is described - 2 died suddenly at a young age. Advances in the molecular genetic cause of HCM have been rapid recent years. About half the patients with clear HCM may have an abnormal gene detected on one of the 10 genes identified as a cause of HCM (4,5).

Dr. Brock reported 3 patients with obstructive HCM. These patients mimicked aortic stenosis and highlight another deviling aspect of HCM care. Hypertrophic cardiomyopathy is the great masquerader of cardiology and often is misdiagnosed today as other conditions: asthma, aortic stenosis, mitral regurgitation, rheumatic heart disease, dilated cardiomyopathy, coronary artery disease and perhaps worst of all - HCM can masquerade as normal. In Dr. Brock's cases, the first was a woman aged 58 who had heart failure, angina and syncope, and hypotension and thought to have aortic valvular stenosis. At operation, after the aortic valve was found to be normal, a subvalvular gradient was documented by catheter pull-back. An expanding dilator was inserted into the ventricle but it was not possible to resuscitate the patient. A second patient, age 63, had dyspnea and hypertension. She was clinically thought to have aortic stenosis but cardiac catheterization showed subvalvular stenosis. Bougees were passed into the subvalvular area and an expanding dilator, but the patient died early post-operatively. The third patient was detected by catheterization directly into the left ventricle. The diagnosis of subvalvular obstruction was established and the patient was not operated.

Important conclusions of Brock's paper were: 1. Left ventricular obstruction could be caused by left ventricular hypertrophy. 2. Subvalvular aortic stenosis was an important differential diagnostic distinction from aortic stenosis. It would then be useful to search for calcification in all adults with a supposed aortic valvular stenosis. 3. In 1957, before the advent of cardiopulmonary bypass, subvalvular stenosis appeared inoperable. Indeed, one of the patients in this early series of 3 was not operated because of the absence of calcification in the aortic valve, and after catheterization diagnosis.

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In 1963, Cleland reported the first series of patients operated after the advent of cardiopulmonary bypass, with good results in the majority of cases (6). The operative details indicate excision of subvalvular muscle bars. The prevailing notion at that time was that obstruction was caused by a subvalvular muscular ring, a muscular sphincter comparable to that found in infundibular stenosis of the right ventricle outflow tract. It was not until the M-mode echocardiographic observations of Shah et al that systolic anterior motion (SAM) of the mitral valve was widely understood as the cause of LV outflow obstruction in most cases (7). Newer understanding of the hydrodynamic mechanism of SAM has led to novel, and perhaps better operations, that include repairs to the mitral valve and the papillary muscles (8).

It is hoped that this supplement in the AKD will serve as an introduction to progress in HCM diagnosis and therapy (9-11). Though impossible to adequately cover 50 years of discovery, it is hoped the reader will find this introduction interesting and useful.

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