Hypertrophic cardiomyopathy and the Hypertrophic Cardiomyopathy Association

Hipertrofik kardiyomiyopati ve Hipertrofik Kardiyomiyopati Derneği

Hypertrophic cardiomyopathy (HCM) was once thought to be rare and inevitably tied to a very poor prognosis. Today we know it to be the most common genetic cardiac condition affecting 1 in 500 people (1-3) and one that is compatible with a normal life span in most patients. Based on the 2000 census, there are an estimate 135,000 affected individuals in Turkey.

Hypertrophic cardiomyopathy can cause significant symptoms, impaired quality of life, and disability in some patients. Hypertrophic cardiomyopathy is also unfortunately the leading cause of sudden cardiac death on the athletic playing field and for this reason, and others discussed in this article, misconceptions have invaded the popular media about the true face of this complex disease. While it is true that nearly 40% of the cardiac deaths on the playing field are from HCM (4) it is not only athletes, who die young from this disease, they simply capture the headlines in a striking fashion. Hypertrophic cardiomyopathy may cause sudden death and disability in all age groups.

The advances in public access automatic defibrillation (AED) have increased the occurrences of out of hospital resuscitation and the opportunity to bring those with sudden cardiac arrest from all causes, including HCM, to medical attention and treatment and subsequent treatment with implantable defibrillators. The efforts of AED advocates and public access defibrillation programs (PAD) should be commended. However, AED placement and appropriate utilization in the HCM population should not be viewed as the best manner in which we identify young patients.

The Hypertrophic Cardiomyopathy Association (HCMA) works with families, patients and the medical community in providing advocacy, education, and support. The HCMA was created in 1996 in response to the death the fourth member of the Founder's family from HCM. Since then the HCMA has grown to serve thousands of HCM families and thousands of medical professionals. The HCMA's services to patients and families include an interactive website (www.4hcm.org) with accurate and balanced information about the disease, lifestyle issues, family screening, and general support. The HCMA facilitates an annual meeting where patients can interact with others with HCM, and gather one-on-one support. At these meetings patients hear from, and meet top researchers and physicians in the field to address questions.

As much of the work of the HCMA is done via the internet, we have participants from around the world, including Turkey. The HCMA also provides educational materials and runs programs for the medical community in coordination with our world-renowned medical advisors. Greater general public awareness of HCM will clearly lead more patients to diagnosis and treatment. Thus the HCMA is working to gain recognition for this not uncommon disease, which is present in approximately 500,000 Americans, most of whom have no idea they are affected.

In the past 10 years the HCMA has had the opportunity to work with thousands of families and make some rather interesting observations about challenges in diagnosis, access to family screening, varied treatment options based on socioeconomic and geographic factors, and overall understanding of HCM within the medical community. Of our patient/family membership, the average HCM patient has struggled with misdiagnosis for an average of 4 years, and up to 35 years. Misdiagnoses include mitral valve prolapse, exercise induced asthma, depression, panic attack, innocent murmur, and hypochondria. Once patients have been appropriately identified with HCM and treatment begun, patients experience a significant improvement in overall quality of life. Patients who have been evaluated by a HCM center of excellence (a list of American centers is available on the HCMA's website - www.4hcm.org) have a much greater likelihood of receiving care consistent with the American College of Cardiology/European Society of Cardiology (ACC/ESC) consensus document on the treatment and management of HCM (5). When these patients follow up with community based cardiologists/electrophysiologists and maintain regular communications with the HCM center of excellence visits to emergency departments are dramatically

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decreased and patients return to work and normal activities in many cases with minimal restrictions. Those patients who have not consulted with an HCM center of excellence are often on pharmacologic treatments that are inconsistent with the ACC/ESC consensus document (5), lack a full scope understanding of their disease, and need for family screenings. They are more likely to miss time from work, participation in family activities and feel isolated when compared to their patient counterparts who have been evaluated by a HCM specialist.

Of significant concern to the HCMA is the high number of cases of young people (age 8-35 years) diagnosed originally with athletically/exercise induced asthma. This is the single most common complaint we hear about from parents of children who have died on the playing field. When questioned if the child had ever received pulmonary function testing to prove conclusively that asthma was the cause of the shortness of breath less than 5% report any pulmonary testing at all, and those that were tested report "inconclusive" findings. This is a particularly troubling issue as many of these children have been prescribed inhalers containing stimulants, which may have catastrophic consequences in HCM. It appears that if electrocardiograms were done in these children who report shortness of breath with exercise we would likely identify those with underlying heart disease.

Along a similar theme, the recent controversy over sudden deaths in children taking medications to treat Attention Deficit Disorder (ADD) and Attention Deficit Hyperactivity Disorder (ADHD) including Adderal (dextroamphetamine), Concerta and Ritalin (both methylphenidate) was brought to the HCMA's attention. Of 12 deaths in children (age 7-16 mean 12.5 years) reported, eight had underlying structural heart disease. Of these deaths, one was autopsy confirmed HCM, five are suggestive of possible HCM with poor or limited post mortem data available, or other structural heart disease. Cardiac risk factors listed in this report include 3 cardiac hypertrophy, 3 history of heart murmur, 1 history of mother with a ventricular arrhythmia, 1 bicuspid aortic valve, and 1 aberrant origin of coronary artery (6). This raises the question whether there should be an effort to require electrocardiograms in all children prior to beginning a regimen of these powerful stimulants.

There continues to be gender disparities in the diagnosis of HCM. As HCM is an autosomal dominant genetically transmitted disease (7), the occurrence of the disease is equal. The average age of diagnosis for patients with HCM is dependent upon gender with men averaging 34 years of age at diagnosis and women averaging 38 years of age. Interestingly women have reported to the HCMA that they feel symptoms on average at the same time or even slightly before men do but are often dismissed by the healthcare community as having non-cardiac causes for their complaints.

There is a lack of appreciation for HCM among health care providers. We recently documented this by a recent polling of international cardiac professionals at 3 separate scientific sessions in the United States. The respondents underestimated the prevalence of HCM 64% of the time, and were unaware of when to screen a child in an HCM family 45% of the time. Other findings included ability to recognize six risk factors of sudden death in HCM in only 16% of respondents, yet 75% are caring for HCM patients.

This is a heterogeneous disease, which has been appropriately nicknamed the "great masquerader", and has been fraught with nomenclature issues, with no fewer than 70 names being attributed to HCM in the past 50 years. The daunting task for the medical community and the HCMA is to work toward better means if identifying the millions of patients' worldwide who are currently living with HCM and guiding them to quality care.

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