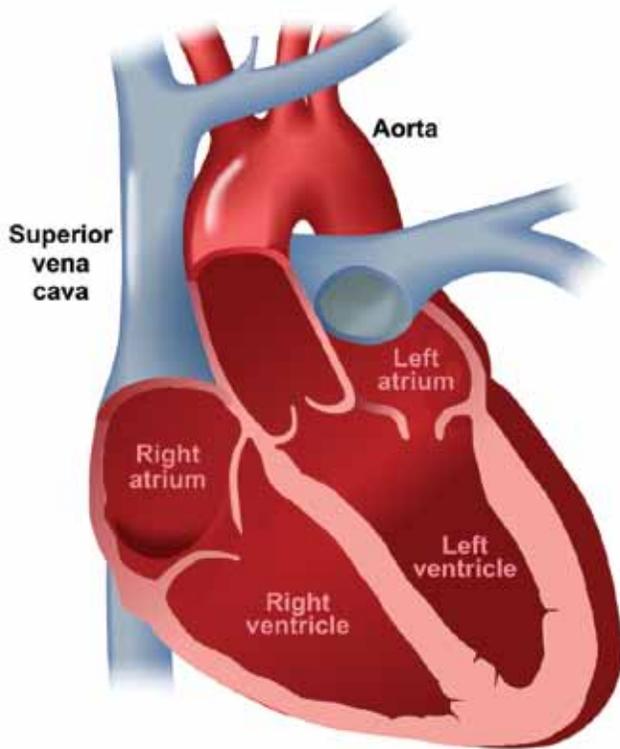
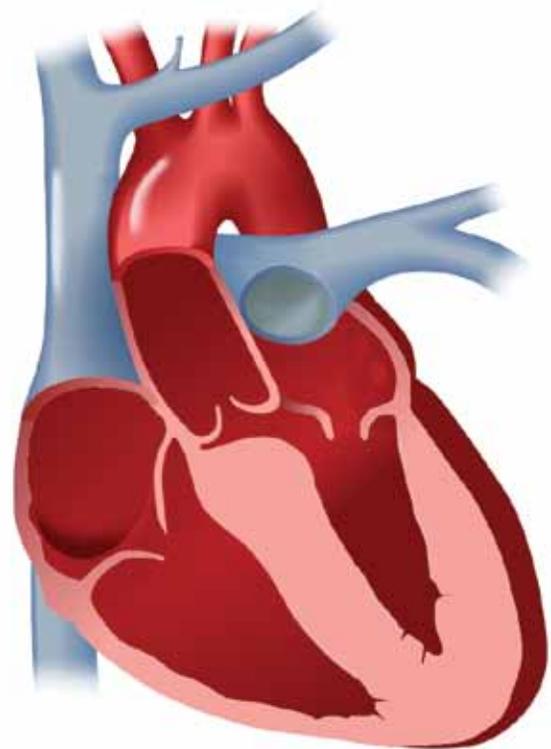


Hypertrophic Cardiomyopathy

By Gordon A. Ewy, MD



Normal Heart



Hypertrophic Heart

In this issue, we announce the appointment of Jil C. Tardiff, MD, PhD, as the *Steven M. Gootter Endowed Chair for the Prevention of Sudden Cardiac Death*. Steven was a young, vibrant, witty, athletic husband, father, son and businessman who left his house one morning for a jog. He was struck by out-of-hospital cardiac arrest (OHCA). No one witnessed this, so he was a victim of sudden cardiac death.

The average age of adults in the United States with cardiac arrest is the mid-60s, so many like Steven, who either are not witnessed or not promptly treated,

succumb at a relatively young age. In individuals over age 40, the most common cause of out-of-hospital cardiac arrest is coronary artery blockage, leading to a heart attack (myocardial infarction). So to prevent this tragedy, one addresses the classic risk factors for coronary artery disease: high cholesterol, high blood pressure, smoking, diabetes and stress. In contrast, there are many genetic causes of sudden death in younger individuals (under age 40). The most common in the United States is a disease called hypertrophic cardiomyopathy (HCM).

Jil is one of the rare, but

important breed of academic physicians who have expertise in both basic research (thus the PhD) and clinical medicine (thus the MD). Her genetic expertise in HCM is critically important as this is an inherited condition of the heart that occurs in approximately one in 500 people.

Diagnosing HCM

In HCM, the muscle of the pumping chambers of the heart becomes abnormally thick (Figure, right). It may or may not cause symptoms. It is equally distributed among males and females. It can develop in susceptible individuals

at any age (which may make the diagnosis difficult). So even if one has the genetic predisposition to the disease, their heart muscle thickness may be normal for years before it begins to thicken or enlarge. It predisposes the patient to sudden cardiac death and is the leading cause of sudden death in the young in the United States.

So why don't we just do genetic testing on everyone for HCM? More than 700 different genetic abnormalities have been identified in patients with HCM, so which ones do we test for? However, if someone in the family has this condition, the specific genetic abnormality of that individual can be identified, and others in the family can and should be tested to see if they are at risk of developing the disease.

The other problem is that individuals with hypertrophic cardiomyopathy may not have any symptoms, or symptoms may be non-specific. However symptoms, such as shortness of breath, chest pain or discomfort with exertion, dizziness or lightheadedness, fainting and palpitations, while not specific for HCM, do bring up the possibility of a cardiac condition. Unfortunately the first recognized symptom of HCM may be cardiac arrest or sudden cardiac death.

The genetics of HCM are caused by an inherited gene from parent to child. If a parent has HCM, his or her child will have a 50-percent chance of inheriting one of the HCM genes. However, the presence of the gene does not mean that the individual will have the disease. One family member may develop severe symptoms,

while another family member with the same gene may never develop any or only mild clinical signs of HCM. Conventional non-genetic family screening includes an annual electrocardiogram and echocardiogram for children under 18 years of age and every five years for adults over 18 years of age.

Treating HCM

Once discovered, the treatment of HCM depends upon its severity and the associated symptoms. In many patients the first line of treatment usually consists of medical therapy.

Patients at high risk are those who have a family history of sudden death, episodes of fainting, abnormal blood pressure in response to exercise, abnormal heart rhythms and excessive thickness of the heart muscle. Such patients often are treated with implanted cardioverter defibrillators (ICDs), so they can be rescued promptly from a life-threatening arrhythmia.

One of the characteristics of HCM is that part of the heart muscle (the part between the right and left ventricles) may become abnormally thick, causing a resistance of the blood being pumped from the left ventricle and leading to symptoms that cannot be managed by medical therapy. In these cases, the condition can be treated by either open heart surgery (removing part of the thickened septum) or in the cardiac cath lab (injecting alcohol into the septum to reduce its size).

Patients with HCM who wish to engage in sports should first closely work with their cardiologist

to develop a regimen that is tailored to their risk profile, especially in regards to any potentially strenuous activities such as football, basketball, soccer, running, bodybuilding, singles tennis, ice hockey, scuba diving, windsurfing or racquetball. They may engage in milder forms of exercise, such as walking or bowling.

An HCM Clinic

Although rare, if HCM runs in your family, it becomes a very important condition. Dr. Tardiff plans to begin a hypertrophic cardiomyopathy (HCM) clinic in conjunction with our excellent pediatric cardiologists at The University of Arizona Health Network to offer specialized care for such patients and their families. ♥

Is Your Family at Risk for HCM?

Not all sudden cardiac deaths are documented as such. If you have someone in your family who died suddenly, maybe from a car accident or even a drowning, ask people about the circumstances. Researchers suspect that some accidental deaths may be attributable to primary cardiac arrest that led to death during these types of activities.