CLINICAL VIGNETTE

A Case of Hypertrophic Cardiomyopathy

Sahar Sohrabian, MD

A 50-year-old male with a past medical history of hypertension, presented to Cardiology with dyspnea on exertion. Over the last few months, he noted unusual fatigue and palpitations during his tennis lessons. He also noticed dizziness when going up a few flights of stairs at work. He had no syncopal episodes and also denied chest pain, orthopnea, paroxysmal nocturnal dyspnea, or lower extremity edema. He had no other cardiac risk factors, no history of smoking, and no family history of cardiac disease.

On exam, patient's vitals were within normal limits, his heart rate was mildly elevated at 99. His physical exam was otherwise unremarkable. Cardiac exam had normal rate and rhythm, with no murmurs, rubs, or gallops. Patient's EKG was consistent with normal sinus rhythm, with T wave inversions in V3/V4. His labs were only remarkable for an elevated triglycerides at 213. An echocardiogram, treadmill stress echocardiogram, and event monitor were ordered for further risk stratification. Patient's event monitor was consistent with normal sinus rhythm with episodes of sinus tachycardia, but without other significant arrhythmias or pauses. Echocardiogram showed normal cardiac function with ejection fraction estimated at 60-65%.There was moderate septal left ventricular hypertrophy with grade I diastolic dysfunction. There was also mild chordal systolic anterior motion of the mitral valve with mild mitral regurgitation. His treadmill echocardiogram was consistent with fair exercise tolerance, patient achieved 7.0 METS. His ECG had significant ST depressions in the inferior-lateral leads during peak exercise, with a drop in blood pressure. Patient was then referred for cardiac catheterization.

Cardiac catheterization showed normal coronary arteries. He had a resting left ventricular outflow tract mean gradient of 18 mmHg. He also had a mean gradient of 101 mmHg and peak gradient of 134 mmHg across the left ventricular outflow tract after following a premature ventricular contraction. He was given the diagnosis of hypertrophic cardiomyopathy and referred for a Cardiac MRI. MRI was also consistant with hypertrophic cardiomyopathy. It demonstrated hypertrophy of the basal interventricular septum with narrowing of the left ventricular outflow tract and minimal systolic anterior motion of the mitral valve. The patient was started on metoprolol and instructed to keep well hydrated throughout the day and strongly advised to refrain from competitive sports such as single's tennis. His symptoms have improved and he remains stable.

Hypertrophic cardiomyopathy (HCM) is an autosomal dominant, genetically determined cardiomyopathy. The disease arises after a mutation occurs in a sarcomere gene. HCM is characterized by left ventricular hypertrophy in the absence of aortic stenosis or hypertension. The prevalence of hypertrophic cardiomyopathy is 0.2% worldwide.[1] The diagnosis of HCM can be compatible with a normal lifespan. Many patients do not have symptoms or only mild limitations. Patients with a diagnosis of HCM can have outflow gradients and heart failure symptoms, however many patients may also be asymptomatic with a minimal outflow tract gradient at rest. Patients with progressive disease usually present with dyspnea, chest pain, palpitations, or syncope. The most common presentation of HCM is dyspnea. The dyspnea can be a result of 1) Diastolic dysfunction, 2) LVOT obstruction 3) Mitral regurgitation or 4) Systolic dysfunction.[3] The LVOT pressure gradient in HCM is dynamic and is influenced by myocardial contractility and loading conditions. Patients that do not have LVOT gradients on resting echocardiogram should have an exercise stress echo test to further evaluate for the presence of LVOT obstruction with exertion.[4]

The physical exam in a patient with HCM may be variable. The classic physical findings include systolic murmur. The systolic murmur is usually due to left ventricle upper septal hypertrophy and systolic anterior motion of the mitral valve. The murmur is a harsh crescendo-decrescendo systolic murmur that is heard best at the apex and left lower sternal border. A systolic thrill may also be felt at the apex or lower left sternal border. A parasternal lift suggests significant mitral regurgitation or pulmonary hypertension.[5-7]

In addition to the clinical cardiac history and physical exam, an electrocardiogram and cardiac imaging should be done in the initial diagnosis of HCM. Patients with evidence of HOCM on ECG and echo, should have ambulatory ECG monitoring and exercise stress testing for further risk stratification. The ECG in a patient with HCM is usually abnormal. ECG findings may include repolarization changes, prominent abnormal Q waves in the inferior (II, III, and AVF) and lateral leads (I, aVL, and V4-V6), P wave abnormalities due to left atrial or bi-atrial enlargement, left axis deviation, or deeply inverted T waves in the mid-precordial leads (V2-V4). Echocardiogram can confirm a clinical diagnosis of HCM when there is unexplained increased LV wall thickness > 15mm anywhere in the LV wall. Typically LV wall thickening is asymmetric, however any pattern of LV wall thickening may be present. Systolic anterior motion (SAM) of the mitral valve is not required for a diagnosis of HCM. Patients that do have SAM can still have LVOT obstruction. If patients do not have a definitive diagnosis after

an echocardiogram, a cardiovascular magnetic resonance can be performed for diagnosis and further risk stratification. Cardiac catheterization is also an option for diagnosis when a patient also needs to be evaluated for obstructive coronary artery disease or in patients without a definitive diagnosis by imaging.[8]

Screening is recommended for first degree relatives of patients with definitive HCM. Family screening should include a history and physical, ECG, and echocardiography. The clinical evaluation is recommended annually, from 12-18 years of age. There is also a possibility of delayed onset hypertrophy, therefore family members with normal studies should get re-evaluated every five years after the age of 18.[9]

Medical treatment for HCM is initiated for patients with symptoms of heart failure and a LVOT obstruction. First line therapy includes initiation of a beta blocker. Patients who continue to be symptomatic despite beta blocker therapy can be switched to a non-dihydropyridine calcium channel blocker such as verapamil. For patients who continue to be symptomatic, disopyramide can be added to the beta blocker or calcium channel blocker therapy. Caution should be advised with the use of diuretics and vasodilators in patients with HCM.[10-13] Patients that have advanced heart failure symptoms despite optimal medical therapy along with resting or provocable gradients > 50 mmHg can be candidates for invasive septal reduction therapy. The options for septal reduction include, surgical septal myectomy or alcohol septal ablation. Generally surgical myectomy is considered first line therapy, unless the patient has too many co-morbidities and is considered high risk for surgery.[14]

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