Case # 4: Pre-participation Screening Detects Cardiac Disease in an Asymptomatic Preadolescent Athlete

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Patient presentation

An asymptomatic 13-year-old African-American male was evaluated during a large, station-based Pre-participation Physical Evaluation (PPE).

History

He is a talented, high-intensity athlete and otherwise healthy. He was asymptomatic and specifically denied chest pain (CP), shortness of breath (SOB), palpitations, lightheadedness, or syncope. The patient reported a family history of cardiovascular disease (grandparents) and hypertension (both parents), but denied a family history of structural cardiac abnormalities or premature sudden cardiac death. He also mentioned a personal history of occasional high blood pressure readings. During a later chart review, BPs ranging from 146-166/62-76 were noted over the preceding 2 years. Despite this, he denied all other medical conditions, medicine use aside from an occasional over-the-counter medication (NSAID/Tylenol), the use of alcohol, illicit drugs, steroids, or tobacco. He also denied prior restrictions on sports participation.

Physical Examination

The patient was afebrile (T 98). Although he was large for his age, with both height (6 feet 0 inch) and weight (172 lbs) being greater than the 99th percentiles for age, he was a healthy weight per BMI (22). Pulse (60) and RR (14) were WNL, but blood pressure (140/76) was noticeably elevated especially when compared to age-related normative values.
Generally, the patient was well-appearing and devoid of any obvious dysmorphic features. His cardiac exam did reveal a grade 3/6 crescendo-decrescendo systolic murmur heard best at the left upper sternal border. Murmur increased with Valsalva maneuver. Lungs, Abdomen, Genitourinary, and extremity exams were unremarkable.

**Differential Diagnosis**

- Pediatric Hypertrophic Cardiomyopathy (HCM) due to a sarcomeric defect
- Athlete’s Heart (physiologic adaptation to exercise)
- Primary hypertensive with left ventricular hypertrophy (LVH)
- Secondary hypertension (e.g. renal disease, anabolic steroid use, coarctation of the aorta)
- Aortic stenosis

**Challenge Question #1** - What is the best explanation for why the murmur of hypertrophic cardiomyopathy (HCM) increases with the Valsalva maneuver?

A) Cardiac output increases, left ventricular (LV) volume increases – increasing outflow obstruction
B) Cardiac output decreases, LV volume increases – increasing outflow obstruction
C) Cardiac output increases, LV volume decreases – increasing outflow obstruction
D) Cardiac output decreases, LV volume decreases – increasing outflow obstruction

**Lab Studies**

CBC, CMP, Thyroid Function panel, and urinalysis were WNL.
GeneDx Hypertrophic Cardiomyopathy (HCM) Panel:
No disease-causing gene mutation was detected

**Other Studies**

EKG: sinus rhythm, prominent precordial voltage, “dome-shaped” convex ST segment elevations in the anterior precordial leads (V1-V2), and diffuse T wave inversions (most notably in the inferolateral leads).
Challenge Question #2 - Which of the following best describes the sensitivity of EKG in detecting (HCM)?

A) <30%
B) 50% - 75%
C) 75% - 90%
D) >90%

2D Echo: Concentric LVH (maximal wall thickness of 1.6 cm) with speckled appearance of myocardium. Hyperdynamic global systolic function (EF > 75%). No LV outflow obstruction or systolic anterior motion of the mitral valve.

Cardiac MRI: Abnormal LV myocardial mass at 125 g/m² (47-87 g/m²). Concentric LV thickening with slightly more prominent apical involvement (1.6 cm in anterior basilar septum and basilar inferolateral wall). No abnormal uptake of gadolinium was noted.
Stress ECHO: Standard Bruce protocol: Achieved predicted maximal HR without CP or SOB. Hypertensive response to exercise (baseline BP 152/73, HR 71; peak BP 220/60, HR 84), widespread repolarization abnormalities, and dynamic LV outflow tract obstruction
Working Diagnosis

Hypertrophic Cardiomyopathy

Treatment

Internal cardiac defibrillator (ICD) placement was not deemed necessary, but the patient was given a home automatic external defibrillator (AED) and instructed to avoid competitive sports. He was also offered counseling to help cope with the suspected diagnosis of HCM and exclusion from competitive sports. After 3 months of rest, the repeat EKG and ECHO were unchanged and resting blood pressures remained markedly elevated (146-168/62-74). After this observation, he was started on Atenolol 25 mg twice a day for improved blood pressure control and to see if cardiac mass resolution would be achieved once his target BP (<125/75) was maintained. At a subsequent visit, BP remained markedly elevated and the Atenolol dosage was increased to 50 mg twice a day and a daily dose of Hydrochlorothiazide 12.5 mg was added.

Outcome

The cardiac murmur, screening EKG, ECHO, and cardiac MRI were suggestive of Hypertrophic Cardiomyopathy; however, genetic testing was negative. Disqualification was made from participation in competitive sports due to suspicion of HCM. Deconditioning for 3 months did not result in resolution of his cardiac mass. Patient was diagnosed with Stage I/II hypertension and started on two antihypertensive agents. He has remained asymptomatic since his initial evaluation and disqualification.
Challenge Question #3 - According to the 36th Bethesda Conference, what is the recommendation for sports participation in athletes with (HCM)?

A) No sports participation
B) Class IA exercise (Low static/low dynamic components) i.e. bowling
C) Class IIB exercise (moderate static/moderated dynamic components) i.e. football
D) Unlimited participation if the athlete is asymptomatic.

Author’s Comments

HCM is the most common cause of sudden cardiac death (SCD) in sports [1], largely affects males (9:1, M: F) and results in a disproportionately higher rate of death among African-Americans. [2] While the prevalence of HCM is > 1:500 in adults [3], it is much less common in children (0.3 to 0.5 per 100,000). [4] Athlete’s Heart results in increased LV wall thickness due to systematic training, but usually resolves with a period of deconditioning. [5] Systemic hypertension-induced LVH can also present similarly to HCM, but regression of LV wall thickening often occurs with adequate blood pressure control. [6] Genetic testing of sarcomere protein genes identifies a disease-causing mutation in up to 60% of HCM cases [4]; therefore, a negative test does not exclude diagnosis. HCM patients with sarcomere mutations usually present earlier and have a higher prevalence of SCD [7] To reduce risk of SCD in HCM: (1) Exercise restriction and (2) ICD placement if patient experiences life threatening ventricular arrhythmias or ≥2 major risk factors (severe LVH, syncope, NSVT, or family history of SCD) [4]

Editor’s Comments

This case underscores the importance of cardiac auscultation in a quiet environment during the pre-participation physical examination and the need for appropriate work-up when a possibly pathologic murmur is noted.

References

Challenge question answers:

1) Answer D – “Intraventricular obstruction usually occurs at the left ventricular outflow tract, but can be midventricular. The obstruction at the left ventricular outflow tract is caused by systolic anterior motion of the mitral apparatus towards the hypertrophied septum. The drag forces across the mitral valve pull the leaflets anteriorly to the basal septum and lead to both LV outflow tract obstruction and malcoaptation of the mitral leaflets, resulting in mitral regurgitation. Midventricular obstruction is caused by hypertrophied papillary muscles and midventricular hypertrophy. The left ventricular obstruction is dynamic and the degree of obstruction depends largely on loading conditions and cardiac contractility. Therefore, physiological and pharmacological manoeuvres that decrease left ventricular volume (Valsalva manoeuvre, vasodilatation caused by nitrates, or standing from squatting to an upright position) or increase contractility (dobutamine) are associated with augmentation of obstruction” (Veselka J, et al. Lancet 2017; 389: 1253–67)

2) Answer D - Over 90% of patients with HCM will have an abnormal ECG. Abnormalities include T wave inversion, ST segment depression, pathological Q waves, conduction delay, left-axis deviation and left atrial enlargement. (Drezner JA, et al. Br J Sports Med 2013;47:137–152)

3) Answer B – The actual risks to an individual athlete are difficult to evaluate. The Bethesda Conference working group took a conservative approach and recommended low intensity exercise. (Maron B, et al. JACC Vol. 45, No. 8, 2005 Maron and Zipes 1321 April 19, 2005:1318 –21)