Cardiac Issues in the Adolescent Athlete

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The Heart Center
Sudden Cardiac Death

- Incidence is 0.6-6.2 % per 100,000 children in the US
- 20-25 % of the deaths occur during sports
- Hypertrophic Cardiomyopathy is the number 1 cause
Hypertrophic Cardiomyopathy

• 36% of all causes of sudden cardiac death in children
• May actually be higher because of “confusion” about postmortem findings
• Is a “deal breaker” with respect to competitive athletics.
A Little Background Info

Teare, et al (British Heart Journal, 1958)

1st pathologic report:

“asymmetrical hypertrophy of the heart”

Most studies that followed focused on LV outflow obstruction
<table>
<thead>
<tr>
<th>Name confusion</th>
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</thead>
<tbody>
<tr>
<td>Asymmetrical hypertrophic cardiomyopathy</td>
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<tr>
<td>Asymmetrical hypertrophy of the heart</td>
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<td>Asymmetrical septal hypertrophy</td>
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<td>Brock’s disease</td>
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<td>Diffuse muscular subaortic stenosis</td>
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<tr>
<td>Diffuse subvalvular aortic stenosis</td>
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<td>Dynamic hypertrophic subaortic stenosis</td>
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<td>Dynamic muscular subaortic stenosis</td>
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<td>Familial hypertrophic subaortic stenosis</td>
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<tr>
<td>Familial muscular subaortic stenosis</td>
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<td>Familial myocardial disease</td>
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<tr>
<td>Functional aortic stenosis</td>
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<tr>
<td>Functional hypertrophic subaortic stenosis</td>
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<tr>
<td>Functional obstructive cardiomyopathy</td>
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<tr>
<td>Functional obstruction of the left ventricle</td>
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<tr>
<td>Functional obstructive subvalvular aortic stenosis</td>
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<tr>
<td>Functional subaortic stenosis</td>
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<tr>
<td>Hereditary cardiovascular dysplasia</td>
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<td>HYPERTROPHIC CARDIOMYOPATHY</td>
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<tr>
<td>Hypertrophic constrictive cardiomyopathy</td>
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<td>Hypertrophic hyperkinetic cardiomyopathy</td>
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<tr>
<td>Hypertrophic infundibular aortic stenosis</td>
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<td>Hypertrophic nonobstructive cardiomyopathy</td>
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<td>Hypertrophic obstructive cardiomyopathy</td>
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<td>Hypertrophic stenosing cardiomyopathy</td>
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<tr>
<td>Hypertrophic subaortic stenosis</td>
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<tr>
<td>Idiopathic hypertrophic cardiomyopathy</td>
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<td>Idiopathic muscular hypertrophic subaortic stenosis</td>
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<td>Idiopathic muscular stenosis of the left ventricle</td>
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<td>Idiopathic myocardial hypertrophy</td>
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<td>Idiopathic stenosis of the flushing chamber of the left ventricle</td>
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<td>Idiopathic ventricular septal hypertrophy</td>
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<td>Irregular hypertrophic cardiomyopathy</td>
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<td>Left ventricular muscular stenosis</td>
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<td>Low subvalvular aortic stenosis</td>
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<td>Muscular aortic stenosis</td>
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<td>Muscular hypertrophic stenosis of the left ventricle</td>
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<tr>
<td>Muscular stenosis of the left ventricle</td>
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<td>Muscular subaortic stenosis</td>
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<tr>
<td>Muscular subvalvular aortic stenosis</td>
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<td>Non-dilated cardiomyopathy</td>
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<td>Nonobstructive hypertrophic cardiomyopathy</td>
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<td>Obstructive cardiomyopathy</td>
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<tr>
<td>Obstructive hypertrophic aortic stenosis</td>
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<td>Obstructive hypertrophic cardiomyopathy</td>
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<tr>
<td>Obstructive hypertrophic myocardiopathy</td>
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<tr>
<td>Obstructive myocardiopathy</td>
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<td>Pseudoaortic stenosis</td>
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<td>Stenosing hypertrophy of the left ventricle</td>
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<td>Stenosis of the ejection chamber of the left ventricle</td>
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<td>Subaortic hypertrophic stenosis</td>
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<tr>
<td>Subaortic idiopathic stenosis</td>
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<tr>
<td>Subaortic muscular stenosis</td>
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<tr>
<td>Subvalvular aortic stenosis of the muscular type</td>
</tr>
<tr>
<td>Teare’s disease</td>
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</tbody>
</table>
Definition

- Thickened but nondilated left ventricle
- absence of another cardiac or systemic disease
- i.e., Aortic stenosis, HTN, athlete’s heart
Definition

HYPERTROPHIED NON-DILATED LEFT VENTRICLE
Hypertrophic Cardiomyopathy

- Primary and (usually) familial disorder
- Genetic disease of the cardiac sarcomere, caused by mutations in one of a number of genes
- Markedly variable clinical, morphologic and hemodynamic abnormalities
Autosomal Dominant
- mutation in any 1 of 8 genes that encode proteins of the cardiac sarcomere
- prognosis varies with mutation
- genotyping is complex and not in routine use
Epidemiology

Hypertrophic Cardiomyopathy (HCM)

- Complex, relatively common genetic cardiac disorder
- 1:500 in the general adult population
- Most common genetically transmitted CV disease
- Men = women
- Prevalent in many races and countries
Epidemiology

Common cause of sudden cardiac death in young people
- including trained athletes
- cause of death/disability in patients of all ages
Epidemiology

Frequently compatible with NORMAL longevity
Natural history

- Clinical presentation during any phase of life
  - infancy to old age (day one to over 90 years)

- May remain stable over long periods of time
  - up to 25% of a HCM cohort achieving normal longevity

- 75 years of age or older
Natural history

Regional cohort and free of referral bias

Approximately 90 percent were asymptomatic at presentation

Eight year follow-up

- 69 percent remained asymptomatic or had only mild symptoms
- 25 percent had incapacitating symptoms or died
Annual mortality compared to the general population was substantially increased in the 43 patients identified during childhood (1.3 versus 0.08 percent).

Annual mortality was not increased in those identified in adulthood (2.2 versus 1.9 percent).
Hypertrophic Cardiomyopathy

Normal heart vs. Hypertrophic cardiomyopathy

- **Right ventricle**
- **Left ventricle**

Enlargement of the heart muscle
Morphology

- Left ventricular cavity small (or normal) in size
- Increase LV mass due entirely to increased wall thickness
Pathology
LVH

- rarely can occur as a congenital heart defect in infancy
- more commonly develops during adolescence after a latency period
- progression likely predetermined morphologic expression
- not associated with symptoms or sudden death
Mitral Valve

Abnormalities are a primary feature

Systolic Anterior Motion (SAM)
Histology

Affected muscle cells:
- increased diameter
- bizarre shapes
- arranged in chaotic disorganized fashion
Histology - why we care

- Substrate for dysrhythmias --> SCD
- Systolic/diastolic dysfunction
- Intramural coronary arteries
Intramural Coronaries

Decrease size of lumen

Thickening of vessel wall

Suggestive of “small vessel disease”
Pathophysiology

Left Ventricular Outflow Tract Obstruction (LVOTO)

- ? clinical significance
- SAM
  - results in dynamic subaortic obstruction -->
  - increased pressures in LV -->
  - increase wall stress and oxygen demand
Pathophysiology

LVOTO gradient:
- decreased by decreasing contractility
- B-blockers
- Increased LV volume/arterial pressure
Pathophysiology

- LVOTO gradient
  - increased by:
    - decreased arterial pressure/LV volume
    - valsalva
    - maneuvers that increase contractility
    - PVC, certain meds
Pathophysiology

Myocardial Ischemia

- regional
- decreased capillary to muscle mass ratio
- small vessel disease
  - difficult to measure/correlate
Pathophysiology

Diastolic Dysfunction

- 80% have abnormal relaxation and filling
- independent of symptoms, LVOTO, LVH
Clinical Features

Physical Examination

- findings directly related to hemodynamic state
Physical Exam

- double/triple apical impulse
- ventricular contraction
- increased atrial contraction
- early diastolic filling
Physical Exam

- Murmur varies directly with subaortic gradient
- best at lower left sternal border and apex
- generally systolic ejection murmur
Physical Exam

- Maneuvers that increase the murmur
  - standing
  - Valsalva

- Maneuvers that decrease the murmur
  - squatting
  - isometric hand grip
Second heart sound

- may be paradoxically split
Physical Exam

- If no LVOTO
- PE findings may be essentially normal
Symptoms

- Onset generally between 20 and 40 years of age
- decreased exercise capacity
- dyspnea on exertion/fatigue
- chest pain
- dizziness with position changes
- palpitations
- syncope
Symptoms

- severity/character NOT related to LVOTO (presence or gradient)
- infants and children with HCM most often identified due to murmur
  - leading to evaluation
ECG

- 12 lead surface ECG may be 1st manifestation
- precedes symptoms and echo findings
- abnormal in >90% (no particular pattern)
- LVH
- ST-T changes (inverted T’s)
- deep Q-waves
ECG

Does NOT predict/identify patients at risk for sudden death
Echo
Stress testing

- Microvascular ischemia
- Vascular responses
  - Syncope or presyncope
- Arrhythmias
- Muscle fatigue
Conundrum: Athlete’s Heart

- Long term athletic training increases LV
  - Diastolic dimension
  - Wall thickness
  - Myocardial mass
  - More pronounced in runners/cyclist
Athlete’s Heart

Difficult to distinguish between true HCM and athletic heart
"Gray Zone" of LV Wall Thickness

HCM*  \( \cap \) Athlete's Heart

+ Unusual Patterns of LVH\(\uparrow\)
+ LV Cavity < 45mm
- LV Cavity > 55mm
+ LA Enlargement
+ Bizarre ECG Patterns
+ Abnormal LV Filling
+ Female Gender
- Thickness with Deconditioning
+ Family History of HCM
Athlete’s Heart: what to do?

Period of deconditioning

- 4-8 weeks

- LVH should regress if no HCM is present
Sudden Cardiac Death

<table>
<thead>
<tr>
<th>Cause of death</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypertrophic cardiomyopathy</td>
<td>42</td>
</tr>
<tr>
<td>Increased LV Mass/Possible HCM</td>
<td>9</td>
</tr>
<tr>
<td>Anomalous coronary artery</td>
<td>17</td>
</tr>
<tr>
<td>Other CAD</td>
<td>9</td>
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<tr>
<td>Myocarditis</td>
<td>5</td>
</tr>
<tr>
<td>Dilated cardiomyopathy</td>
<td>4</td>
</tr>
<tr>
<td>Ruptured aortic aneurysm</td>
<td>3</td>
</tr>
<tr>
<td>Aortic valve stenosis</td>
<td>3</td>
</tr>
<tr>
<td>ARVD</td>
<td>2</td>
</tr>
<tr>
<td>MVP</td>
<td>2</td>
</tr>
<tr>
<td>Unknown/other</td>
<td>4</td>
</tr>
</tbody>
</table>

Data derived from Burke et al., [2] Maron et al., [35, 43] and Van Camp et al. [64]. LV, left ventricular; HCM, hypertrophic cardiomyopathy; CAD, coronary artery disease; ARVD, arrhythmogenic right ventricular dysplasia; MVP, mitral valve prolapse.
What options do we have with respect to the high school/college athlete?
Prevention of Sudden Death

- Those identified as high risk are treated aggressively
- Occurs at any time
  - during mild exertion
  - sedentary activities (or during sleep)
Sudden Cardiac Death

### Table 2. Risk Factors for Sudden Death in HCM*

<table>
<thead>
<tr>
<th>Major</th>
<th>Possible in Individual Patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cardiac arrest (ventricular fibrillation)</td>
<td>Atrial fibrillation</td>
</tr>
<tr>
<td>Spontaneous sustained ventricular tachycardia</td>
<td>Myocardial ischemia</td>
</tr>
<tr>
<td>Family history of premature sudden death</td>
<td>LV outflow obstruction</td>
</tr>
<tr>
<td>Unexplained syncope</td>
<td>High-risk mutation</td>
</tr>
<tr>
<td>LV thickness greater than or equal to 30 mm</td>
<td>Intense (competitive) physical exertion</td>
</tr>
<tr>
<td>Abnormal exercise blood pressure</td>
<td></td>
</tr>
<tr>
<td>Nonsustained ventricular tachycardia (Holter)</td>
<td></td>
</tr>
</tbody>
</table>

*See text for details.

HCM = hypertrophic cardiomyopathy; LV = left ventricular.
Preparticipation Screening

Not generally recommended beyond physical exam

– unless significant family history exists
Risk Stratification

Sudden death
- Rare before 10 years of age
- Generally due to VT/VF in early morning hours
- Occurs with minimal (if any) physical exertion
Risk Stratification

Risk factors

- Prior cardiac arrest/sustained VT
- Family history of HCM-related death
- Recurrent syncope (especially exertional)
- LVH > 30mm
- Intramural course of proximal LAD
Risk Stratification

- Electrophysiology testing can trigger VT/VF even in low-risk patients
  - does not justify aggressive treatment
Risk Stratification

- Infantile presentation (SIDS)
  - Few patients have been identified
  - Rarely due to genetic transmission
  - Usually have other conditions
    - Noonan Syndrome
    - Metabolic diseases (Pompe’s)
    - IDM
Risk Stratification

Infantile presentation

- Course likely biased by how identified
  - If incidental finding --> do very well
  - If sick --> poor prognosis
- 9/11 died in 1st year of life
Risk Stratification

- Childhood presentation
  - Usually asymptomatic
  - Mortality generally higher than if diagnosed as adult
  - Sudden/spontaneous death
Evaluation

In families with premature death

- Genetic or echo evaluation prior to intense athletic training
  - May not fully express until 18 years old (or later)
  - Screening < 12 y.o. generally unproductive
History Screening

- History and physical examination
- 12-lead ECG
- Two-dimensional echocardiography annually in adolescence
  - 12 to 18 years of age
  - Subsequent clinical studies performed about every five years
- Younger than age 12 is not usually pursued systematically
  - Unless high-risk family history
  - Involved in particularly intense competitive sports programs
Evaluation

Flip side

- Asymptomatic young children with + DNA mutation
- No phenotypic features
- ? clinical significance
- Generally allowed to play sports (unless FH of SCD)
Clinical Course

Difficult to predict

Annual mortality rates 3-6% quoted in some studies

- Some bias in these figures (referrals of high risk pts)
- Likely more in the 1-2% per year range
Clinical Course

Pediatric patients represent a unique clinical dilemma:

- How to predict outcomes for such a diverse disease over such a long period of time?
- Most kids are asymptomatic prior to sudden death
- Yet many live without any disability (and no treatment)
Treatment

- Beta Blockers
  - Propranolol
  - Atenolol, Metoprolol (cardioselective)
Beta Blockers

- Inhibit sympathetic stimulation of the heart -->
- Reduce LVOTO
- Diminish myocardial oxygen requirement via reduction in LV contractility, wall stress or heart rate
- May improve chest pain or CHF
Verapamil

- In adults with HCM has been shown to improve
  - Symptoms
  - Exercise capacity
Treatment

Verapamil

- Generally contraindicated in children
- Absolute in infants
Diuretics

- May be used alone or with B Blockers
- Help symptoms of pulmonary congestion
Treatment

- ACE-inhibitors/digitalis
  - Generally contraindicated
Treatment - surgical

- Reserved for those with severe, drug refractory symptoms
- Functional disability
- ? in those with LVOTO gradient >50mmHg
- Morrow Procedure (myotomy-myectomy)
Morrow Procedure
Treatment - surgical

- Symptoms generally improved following procedure
- Has not been proven to prolong life
Treatment

- Dual chamber pacing
  - Sketchy evidence
  - Decrease LVOTO
  - Improve symptoms
Treatment

- Alcohol septal ablation
  - Inject ethanol into major septal perforator
  - Induce a myocardial infarction
  - Reduces LVOTO
Treatment

Alcohol septal ablation - problems

- By design you induce an MI
- Scar is nidus for dysrhythmia
Prevention of Sudden Death

- AICD (automatic implantable cardioverter defibrillators)
  - sense VT/VF --> shock --> NSR
- Life-saving
  - secondary prevention after cardiac arrest (11%/yr)
  - primary prevention in those with >1 risk factor (5%/yr)
Figure 1. Clinical presentation and treatment strategies for patient subgroups within the broad clinical spectrum of hypertrophic cardiomyopathy (HCM). See text for details. AF = atrial fibrillation; DDD = dual-chamber; ICD = implantable cardioverter-defibrillator; SD = sudden death. Adapted with permission (11). *No specific treatment or intervention indicated, except under exceptional circumstances.
So what can they do?

36th Bethesda Conference (ACC 2005)

Eligibility Recommendations for Competitive Athletes with Cardiovascular Abnormalities
Bethesda Recommendations

Those with unequivocal diagnosis of HCM

- excluded from most competitive sports with the possible exception of those of low intensity (IA)

- independent of age, gender, phenotype, symptoms, LVOTO, prior interventions

Not enough compelling data to exclude genotype +/-phenotype - from participation
The diagram illustrates the relationship between different sports and their requirements for static and dynamic components, as well as the percentage of maximum oxygen uptake (Max O2) involved. The categories are:

- **A. Low (≤40% Max O2)**: Sports requiring low levels of oxygen uptake include Billiards, Bowling, Cricket, Curling, Golf, and Riflery.
- **B. Moderate (40-70% Max O2)**: Sports requiring moderate oxygen uptake include Baseball/Softball, Fencing, Table tennis, and Volleyball.
- **C. High (>70% Max O2)**: Sports requiring high levels of oxygen uptake include Bobsledding/Luge, Body building, Boxing, Canoeing/Kayaking, Cycling, Diving, and Archery.

The diagram also highlights the increasing dynamic component from left to right, indicating a shift from low to high oxygen demands.
What we know

- Heterogeneous, relatively common disease
- Most common cause of sudden death in young athletes
- Primary care providers are 1st line
  - family h/o unexplained sudden death → refer
  - ↑murmur with ↑positioning → refer
“That's all Folks!”