Diagnostic Dilemmas
(therefore, treatment dilemmas)

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• Be able to perform a thorough history and examination of the athlete with a heart murmur
• Be able to counsel the athlete with multiple concussions regarding short and long-term risks
• Be able to provide guidance for the obese athlete
16 yo AAM varsity basketball player comes for preseason exam

- He has never had any chest pain, syncope, palpitations, dyspnea on exertion
- His great-uncle on his mother’s side passed away at 45 from a heart problem
- He is 6'5" and 185 pounds (200 in the program)
- BP is 125/80 and HR is 60 regular
- His femoral and brachial pulses are equal and simultaneous
- He has a 2/6 systolic murmur at the right upper sternal border when lying down

WHAT NOW?
WHAT ARE YOU THINKING ABOUT?

- What happens to his exam when he stands?
  - Decreases- Flow murmur or Aortic stenosis
  - Increases- HCM

- Does he have Marfan’s?
  - Ghent criteria
Ghent Criteria

In the presence of family history of Marfan’s

- Ectopia lentis
- A systemic score ≥ 7 points
- Aortic Root Dilatation Z score
  - ≥ 2 above 20 yrs. old OR
  - ≥ 3 below 20 yrs. old

In the absence of family history

- Aortic Root Dilatation Z score ≥ 2 AND
  - Ectopia Lentes OR
  - FBN1 OR
  - Systemic Score ≥ 7pts
- Ectopia lentis AND FBN1 with known Aortic Root Dilatation

>7 is concerning but not diagnostic

- Wrist AND thumb sign 3
- Wrist OR thumb sign 1
- Pectus Carinatum Deformity 2
- Pectus Excavatum or Chest Asymmetry 1
- Hindfoot Deformity 2
- Plain Flat Foot 1
- Spontaneous Pneumothorax 2
- Dural Ectasia 2
• Protucio Acetabulae 2
• Scoliosis or Thoracolumbar Kyphosis 1
• Reduced Elbow Extension 1
• 3 of 5 Facial Features 1
  – Dolichocephaly
  – Downward slanting palpebral fissures
  – Enophthalmos
  – Retrognathia
  – Malar hypoplasia
• Skin Striae 1
• Severe Myopia 1
• Mitral Valve Prolapse 1
• Reduced Upper Segment / Lower Segment & Increased Arm Span / Height 1
  – Upper/Lower Segment Ratio < 0.85 in whites, < 0.78 in blacks AND Increased Arm Span/Height > 1.05

Shprintzen-Goldberg Syndrome
Physiologic
• Long arms, legs, and fingers
• Curved spine

Craniosynostosis facial features that may include
• Dolichocephaly
• Widely spaced wandering eyes with outside corners of the eyes that point downward
• High, narrow roof of the mouth with micrognathia
• Low-set ears that are rotated backward

Other features
• One or more fingers that are permanently bent
• Joint hypermobility
• Heart or brain abnormalities
• Weak muscle tone in infancy

Loeys-dietz Syndrome
Cardiovascular
• Aortic dissection
• Heart defects at birth (ASD, PDA, bicuspid aortic valve)

Eyes, Head and Neck
• Hypertelorism with blue/gray sclera
• Wide or split uvula
• Cleft palate

Skin
• Easy bruising, wide scars, soft skin texture, and translucent skin

Bones
• Club foot
• Osteoporosis
• Instability or malformation of the spine in the neck

Other
• Irritable Bowel Syndrome
• Food allergies
• Difficulty absorbing food and chronic diarrhea, abdominal pain, and/or gastrointestinal bleeding and inflammation
• Rupture of the spleen or bowel
• Rupture of the uterus during pregnancy
## Incidence of Sudden Cardiac Death in NCAA Athletes

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Athlete-Years</th>
<th>SCD Incidence</th>
<th>Incidence per Athlete-Year</th>
<th>IRR</th>
<th>95% CI</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Overall</td>
<td>4,242,519</td>
<td>79</td>
<td>1 in 53,703</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Sex</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Male</td>
<td>2,418,563</td>
<td>64</td>
<td>1 in 37,790</td>
<td>3.22</td>
<td>1.9 - 5.5</td>
<td>&gt;0.0001*</td>
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<tr>
<td>Female</td>
<td>1,823,999</td>
<td>15</td>
<td>1 in 121,593</td>
<td>1.00</td>
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<td></td>
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<tr>
<td>Division</td>
<td></td>
<td></td>
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<tr>
<td>Division 1</td>
<td>1,263,441</td>
<td>38</td>
<td>1 in 33,942</td>
<td>3.90</td>
<td>1.1 - 1.6</td>
<td>0.0131*</td>
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<tr>
<td>Division 2</td>
<td>930,434</td>
<td>22</td>
<td>1 in 42,292</td>
<td>2.05</td>
<td>1.1 - 4.0</td>
<td>0.0231*</td>
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<tr>
<td>Division 3</td>
<td>1,648,128</td>
<td>19</td>
<td>1 in 86,744</td>
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<tr>
<td>Race</td>
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<td></td>
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<tr>
<td>White</td>
<td>3,075,942</td>
<td>45</td>
<td>1 in 68,354</td>
<td>1.00</td>
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<tr>
<td>Black</td>
<td>644,715</td>
<td>30</td>
<td>1 in 21,491</td>
<td>3.18</td>
<td>1.9 - 5.2</td>
<td>&gt;0.0001*</td>
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<td>Hispanic</td>
<td>168,763</td>
<td>3</td>
<td>1 in 56,254</td>
<td>1.22</td>
<td>0.2 - 3.8</td>
<td>0.6974</td>
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<td>Other</td>
<td>353,042</td>
<td>1</td>
<td>1 in 353,042</td>
<td>0.19</td>
<td>0.005 - 1.1</td>
<td>0.0491*</td>
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**Family History of a Great Uncle who Died at 45**
• SCD patients (ischemic SCD) who died had a higher incidence of a first degree relative with ACI than those who survived

  Circulation, October 3, 2006, Vol 114,(14)
  Circulation: Arrhythmia and Electrophysiology, August 2012, Vol 5, (4)

• If a first degree relative has been diagnosed with a cardiogenetic disease, the other first degree relatives should be screened
  – Brugada’s (ECG, flecainide challenge)
  – LQTS (ECG, genetic testing)
  – ARVD (ECG, echo)
  – Marfan’s (serial echo)
  – Dilated cardiomyopathy (serial echo)
  – HCM (ECG, echo, serial echo)

17 yo football player is here for preseason exam

• Healthy
• Has had his second concussion of his high school football career last fall
• Plays as defensive back and on the varsity team but not starting
• His dad is a lawyer and his mom is a CPA and they are concerned that he will not get into Harvard if he has another concussion

WHAT NOW?
WHAT ARE YOU THINKING ABOUT?
Need More Information

• Was he going to get into Harvard before?
• Does he want to go to Harvard?
• What is the history of the 2 previous concussions?
  – How long did he take to get back to himself?
  – What was the nature of the injury?
• Risk factors for prolonged return to sports

Recommendations

• Neuropsychological testing
  – Baseline, post-injury
  – Computer based vs SCAT vs testing in neuropsychology
• Special teams
• Special helmet
• Special mouthguard
• DHA supplementation
14 yo freshman is coming for his health maintenance visit

- Mentions that he wants to play football because he was told that he is big and so he is going to be good
- He has not played sports before and has always had problems with his weight
- He has seen you in the past for bilateral patellofemoral dysfunction and some achy back pain
- His preseason evaluation form is otherwise normal. His height is 5'8” weight is 280 pounds. HR is 72 and Blood pressure is 140/85
  - Cardiac exam – no murmur and normal S1, S2
  - No organomegaly but hard to be sure
  - Difficulty with 5 hop and duck walk

Thoughts

- Height is 75% weight is >>99%
- Calculate BMI but athletes need body fat analysis
- Be sure that you have the right size blood pressure cuff (bladder should cover 80% of upper arm circumference)
- Ask
  - Why he wants to play football now
  - How competitive is the team?
  - What has the coaching staff said?
  - Any psychological issues that need to be addressed?
  - Dietician vs nutritionist vs weight loss program vs community program
Beginner Exercise Program

Start slowly, with moderate-intensity, non-impact workouts

- Take walks, ride a bike, swim, skate or do calisthenics such as push-ups, sit-ups, crunches or walking stairs
  - Start doing push-ups from a kneeling position to make them easier
  - Crunches may be easier because they do not require a child with weak core muscles to go all the way down
  - Walk and cycle up and down hills to raise and lower the heart rate and use different muscles (difficult in Houston)
  - Change strokes every few laps when swimming to vary muscle use
- Try to add strength, flexibility and endurance exercises to each workout. For example, children can use dumbbells while walking or using an exercise bike

Intermediate Workout

Raise the intensity level of workouts after the child improves his cardio stamina and muscular endurance

- Pick up the pace of walks or rides, trying to finish the course a few minutes early, or adding more minutes
- Add a step aerobics workout, which is low impact
- Add a few minutes of jumping rope and jumping jacks
- Use a treadmill, elliptical machine, rowing machine or exercise bike
  - Do not need full aerobic workouts using these machines; just raise his heart rate each week as he improves his condition
- Create a circuit training workout that includes a variety of exercise
  - 30 seconds of jumping jacks, 30 seconds of jump rope, 30 seconds of crunches and 30 seconds of stairs. Take a two- or three-minute break, then start another circuit, which might include push-ups, jogging in place, squats and lunges.
  - Keep the circuit going for 30 minutes or more.
Conclusion

• There is almost never a completely simple problem that comes into your office
• Think about the short and long term issues of each patient
• Have open discussion and understand where your patients and parents are coming from