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Thanks!

• Families and patients
• Kathi
  – “Steal shamelessly, share seamlessly”
    • None of us can do this alone
Why are cardiologists interested in patients with Duchenne muscular dystrophy?
The Heart is a Muscle Too!!!

Types of Muscle

- Cardiac muscle
- Skeletal muscle
- Smooth muscle

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changing the outcome together
What does cardiomyopathy mean?

- Cardiomyopathy = disease that affects the heart muscle
- Patients with Duchenne get DILATED CARDIOMYOPATHY
How does thinning and enlargement happen?

- In DMD, scar replaces muscle in the heart.
The progression of cardiomyopathy in DMD

Scar forms → Heart function ↓ as there is less muscle → Eventually, not enough blood is pumped to the body, which we call “Heart Failure”
What is “Heart Failure”? 

- **DOES NOT** mean the heart has stopped working

- **DOES** mean that the heart is not able to pump enough blood to give the body the blood and oxygen it needs

- People can live for a long time with heart failure **if treated**
Who should care for my son’s heart?

- **Cardiologist** is a “heart doctor”
  - Not all cardiologists are the same
  - **Pediatric cardiologists**
    - Train in pediatrics and cardiology
  - **Adult cardiologists**
    - Train in adult medicine and cardiology
- **Some cardiologists** have special interests
  - Heart failure/transplantation
  - Neuromuscular disorders
  - Talk to your son’s doctor about finding an expert who is comfortable treating “heart failure”
When should cardiac care begin?

• Begin at the beginning!!
• Repeat visits:
  • At least annually until age 10
    • Or with the onset of cardiac signs and symptoms
  • Biannually after the age of 10
    • Or more frequently based on cardiac signs and symptoms
  • Prior to any major surgery or during periods of significant illness
Why do we need to follow so frequently?

• We often speak in averages when we talk about the expected course....every patient is different!
Types of heart testing

• Electrical
• Heart Function
• Heart scar
Heart testing: Electrical signals and heart rhythm

- Electrocardiogram (ECG or EKG)
  - Evaluates heart rate and rhythm
The ECG in DMD

• In DMD the ECG is abnormal at an early age
• Abnormality **NOT** predictive of clinical course
• Boys with DMD have a heart rate often elevated 10-15 beats per minute above “normal”
• Important to obtain a baseline ECG and to watch for changes over time and for the development of abnormal heart rhythms
Heart testing: Monitoring for abnormal rhythms

• Holter Monitor
  – Evaluates heart rate and rhythm for 24 hours to see if abnormal rhythms are happening
  – More likely to see changes if patients feel symptoms or have lower heart function
How will the heart function be checked?
How will the heart be checked?

• Echocardiogram (aka ECHO)-ultrasound of the heart
  • Evaluate anatomy and function
  • Evaluate valve function
  • Advantages:
    • Readily available/portable
    • Quick
  • Disadvantages:
    • Image quality can unreliable (especially in the DMD patient)
      • Scoliosis, body size
      • Not as accurate for function of the right ventricle
How will the heart be checked?

• Cardiac MRI
  • Advantages
    • No radiation exposure
    • Detailed and accurate cardiac information is obtained
      • Additional information regarding fibrosis
  • Disadvantages
    • May require IV placement
    • Some patients can be claustrophobic
    • Expensive (though limited studies can be close in price)
    • Sedation is often required in younger children or patients who are claustrophobic
What signs and symptoms should I watch for?

- Heart failure symptoms can be difficult to identify in DMD patient
  - Rapid weight gain (or loss)
  - Swelling/puffiness of feet
  - Heart racing/skipping beats or fainting
  - Chest pain (common)
    - Usually musculoskeletal
    - Myocarditis
- Nausea, vomiting, diarrhea, abdominal pain
  - Can result from lack of adequate blood flow to the gut
What treatments are available?

- Standard heart failure drugs include:
  - ACE inhibitors
    - enalapril, lisinopril, perindopril
  - Angiotensin- receptor blockers
    - Losartan
  - β-blockers
    - metoprolol, carvedilol
  - Diuretics
    - furosemide, hydrochlorothiazide
  - Aldosterone receptor antagonists
    - Spironolactone, eplerenone
  - Anti-coagulation
    - Coumadin, Aspirin
When to start treatments

• We know patient will develop cardiac dysfunction at some point

• Should cardiac meds be started at diagnosis?
  • No data exists to suggest benefit starting at diagnosis

• Start ACE inhibitors when evidence of
  • Left ventricular enlargement
  • Ventricular dysfunction
  • Myocardial fibrosis
Other treatments for advanced disease

- Implantable defibrillator (ICD) for heart rhythm problems
- Ventricular assist device (VAD) for heart function problems
- We are still learning the benefits of these devices and how to use them
Should carriers have their hearts checked?

- Little natural history data (into the 40s and 50s…we are just learning)
- Often cardiac disease is the only manifestation
- Cardiomyopathy risk increases with age
  - Approximately 350 DMD/BMD carriers
    - age < 16 yrs: all normal
    - age 16-30 yrs: 6%; 31-50 yrs: 9%; > 50 yrs: 16% DCM
- Current recommendations
  - Baseline evaluation as young adult
  - Frequency of follow-up unclear (? Every 3 to 5 years)
  - Be aware of symptoms
  - Take care of yourself
    - minimize other CV risks
      - smoking, HTN, cholesterol
Conclusions

- Cardiac evaluation should begin at diagnosis
- Ongoing cardiac follow-up is important and the best way to insure long term cardiac health
- When there is evidence of fibrosis, abnormal function or by age 10yo, treatment is recommended
THANK YOU

Questions?