Cardiac Disease in Fatty Acid Oxidation Disorders

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Introduction to the Heart

• Heart uses fatty acids as a preferred fuel
• Heart needs fuel for 2 major functions:
  • Muscle contraction (the pump & plumbing)
  • Conduction system (the electrical wiring)

• How to treat muscle and conduction problems
Keep in mind…

• This is a general discussion about problems known to occur in most types of FAODs
• Your child’s cardiologist knows the most about your child’s heart
FAODs and the Heart

1) Risk for developing cardiomyopathy (CM)
   • CM is disease of the heart muscle- abnormal muscle contraction can mean the heart cannot generate enough force to deliver oxygen-containing blood to the body and the brain

2) Risk for developing cardiac arrythmias
   • Abnormality of the conduction system- the electrical system of the heart- these electrical signals tell the heart when to beat and allow this to happen in an organized way

   • *The reason why individuals with FAOD develop heart muscle and conduction system abnormalities is not known exactly, but we have some ideas…*
Cardiomyopathy: 2 primary types

- Dilated Cardiomyopathy
- Hypertrophic Cardiomyopathy
- Both can occur simultaneously
- One can morph into the other (usually HCM turns into dilated HCM)
Dilated Cardiomyopathy (DCM)
What is Cardiomyopathy?

Hypertrophic Cardiomyopathy - HCM
How do we diagnose cardiomyopathy?

• Echocardiography (ECHO) is the single best test to diagnose and follow CM

• Electrocardiography (ECG or EKG) can be helpful to screen for CM, but cannot diagnose CM

• Other tests can also be used (ex.- cardiac MRI) but are usually not necessary or practical

• Simple tests are also very useful- chest x-ray, some basic blood tests (if organs do not receive enough oxygenated blood they get sick)
CM can be detected by an x-ray:

Normal

Enlarged heart
Echocardiogram: Gold Standard

- Used to make diagnosis
- Follow for changes over time
Basics of the heart: the plumbing

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How does EHCO help the cardiologist? How does it help a patient?

• ECHO is very good for:
  Making a diagnosis
  Detecting subtle abnormalities early in disease
  Following changes over time
  Making objective measurements of heart function (ejection fraction, shortening fraction)
• ECHO is not good for:
  Day-to-day management
  Detecting arrhythmias
What does an ECHO see?

- What a normal heart looks like:
What does an ECHO see?

• What a normal heart looks like:
Dilated Cardiomyopathy: by ECHO
Dilated Cardiomyopathy: by ECHO
Hypertrophic cardiomyopathy: by ECHO
Other ways that CM can be detected:

- **SYMPTOMS**: what parents may notice
  - Fatigue- inability to do the same activities, or tiring more quickly
  - Shortness of breath with activity
  - Weight loss
  - Nausea/vomiting, or early satiety
  - Swelling in face, abdomen, legs
  - Other symptoms may occur but are not typical: chest pain, fainting
How is CM detected?:

- SIGNS: what doctors will notice
  - Increase in heart rate
  - Extra heart sound (gallop, murmur)
  - Pulmonary edema: extra fluid in the lungs
  - Ascites/ Edema (fluid build-up in abdomen or limbs)
  - Liver enlargement
Children and compensated heart failure

• Sometimes signs/symptoms are subtle
• Slow changes are hard to recognize
• Kids compensate, until then don’t…
• Surveillance is intended to catch early signs/changes
What do these signs indicate:

• HEART FAILURE= pump failure, evidence that heart fails to deliver adequate blood/oxygen to tissues

• Congestive heart failure= pump failure resulting in back-up of blood
  • Left side- backs up into lungs
  • Right side- backs up into body
OTHER TESTS:
• BNP- hormone released by a “stretched” heart
• Blood tests that measure oxygen delivery to organs:
  • Kidney function
  • Lactate
  • Oxygen content of blood going back to heart
  • Liver function
  • Cognitive function
Health Supervision Guidelines: example

- Echocardiogram at presentation and every year to evaluate for CM
- Echo as needed to evaluate shortness of breath, tachycardia, or other signs/symptoms of heart failure
- ECG annually to screen for abnormalities
- Holter monitor annually (24 hour monitor)
- ECG and Holter if any syncope (fainting) or other symptoms concerning for arrhythmia

- Individualized care for each patient
Why should a FAOD patient see a cardiologist?

- ECHO can detect subtle abnormalities before symptoms are present
- There are medications that help slow the progression CM in children and help them feel better
- New therapies may reverse cardiomyopathy
- Some ECG abnormalities can be treated (medications, devices)
- There are many medications that should be avoided in someone with an abnormal ECG
Arrhythmias in FAODs

• Many types of electrical malfunctions (arrhythmias)
• Not one specific type of arrhythmia that is seen in FAODs, several have been described
• A sick heat is prone to arrhythmia
• Metabolic derangement can cause arrhythmia
Conduction system: the wiring/electrical

- Sinus node
- AV node
What an ECG looks like:
What an ECG tells you:

- **Sinus Node**: Electrical impulse spreads from the Sinus Node throughout the Left and Right Atria.
- **Atrioventricular Node**: Electrical impulse spreads from Bundle Branches throughout the Left and Right Ventricles.
- **His Bundle**: 
- **Left Bundle Branch**: 
- **Right Bundle Branch**: 

The ECG waveform:
- **P Wave**: PR Segment
- **QRS Complex**: 
  - **PR Interval**: 
  - **QT Interval**: 
  - **ST Segment**: 
  - **T Wave**: 
Compare normal to prolonged-QT

[heart institute logo]

[diagram showing normal and long-QT ECGs]
Arrhythmias in FAODs

• Prolonged QT, conduction delays, atrial flutter, ventricular arrhythmias, abnormal repolarization

• Most likely to occur in decompensated patient, very unpredictable

• Treated the same as they are for any child (or adult)

• Correction of metabolic derangement is #1
How to detect arrhythmia

• ECG
• Telemetry (monitor)
• Holter Monitor
• Event Monitor
• Loop recorder
Loop recorder
Treatment of a arrythmias

- Medications *may* be protective:
- Avoidance of acidosis, electrolyte supplementation
- No treatment specific to FAOD
Antiarrhythmic medications:

- **Class I:** affect sodium channels
  - disopyramide is a class Ia antiarrhythmic
  - flecainide is a class Ic

- **Class II:** b-blockers (propranolol)
- **Class III:** potassium channel blockers
  - amiodarone - has class I, II, III and IV properties
  - sotalol - also has b-blocker activity

- **Class IV:** calcium channel blockers (verapamil)
This is only a sampling of the medications that can be used for atrial arrhythmias.

Treatment of childhood arrhythmias is sometimes more of an art than a science.

Antiarrhythmics have toxicities and can also cause arrhythmias.

A cardiologist who specializes in cardiomyopathy and one who specializes in rhythm problems make a good team when it comes to the care of a patient with FAOD.
Implantable Cardioverter Defibrillator (ICD)
## Signs and Symptoms in 50 LCHAD patients

<table>
<thead>
<tr>
<th>Signs and Symptoms</th>
<th>11 Patients Presenting Without Acute Metabolic Derangement</th>
<th>39 Patients Presenting With Acute Metabolic Derangement</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Number</td>
<td>Percentage</td>
</tr>
<tr>
<td>Hepatomegaly</td>
<td>6/10</td>
<td>60%</td>
</tr>
<tr>
<td>Hepatic dysfunction</td>
<td>8/10</td>
<td>80%</td>
</tr>
<tr>
<td>Cholestasis</td>
<td>3/10</td>
<td>30%</td>
</tr>
<tr>
<td>Cardiomyopathy</td>
<td>4/11</td>
<td>36%</td>
</tr>
<tr>
<td>Failure to thrive</td>
<td>8/11</td>
<td>73%</td>
</tr>
<tr>
<td>Feeding difficulties</td>
<td>6/11</td>
<td>55%</td>
</tr>
<tr>
<td>Vomiting</td>
<td>5/11</td>
<td>45%</td>
</tr>
<tr>
<td>Hypotonia</td>
<td>7/11</td>
<td>64%</td>
</tr>
<tr>
<td>Lethargy</td>
<td>3/10</td>
<td>30%</td>
</tr>
<tr>
<td>Psychomotor retardation</td>
<td>3/11</td>
<td>27%</td>
</tr>
<tr>
<td>Peripheral neuropathy</td>
<td>1/11</td>
<td>9%</td>
</tr>
<tr>
<td>Microcephaly</td>
<td>3/11</td>
<td>27%</td>
</tr>
</tbody>
</table>
Summary: the heart of FAOD

- Cardiology follow-up every year is important
- Early detection of heart problems may help avoid serious illness and early death
- Most pediatric cardiologists will not know much about FAODs; find a cardiomyopathy specialist in your area
- Educate your cardiologist about what your child or family member needs every year or every few years?:
  - Screening for CM
  - Screening for abnormal conduction

• Pediatric cardiologist understand how to take care of these problems, find a doctor who is willing to work with your family!
Thank you

Questions?