MCAD
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with MCAD will have all of these features.

MCAD is one of several *Fatty Acid Oxidation Disorders (FAOD) in which there is an inability to break down certain fats, caused by an enzyme deficiency. This results in a decreased ability to fast without food or calories. MCAD can be detected through newborn screening. With treatment from infancy, children with MCAD are expected to live a full life. However, they are still at risk for complications in times of illness. Prior to newborn screening, a previously healthy child with MCAD deficiency would present with hypoglycemia , vomiting, and lethargy triggered by a common illness. If hypoglycemia is untreated, seizures could occur and such episodes could quickly progress to coma and death. Enlarged liver and acute liver disease may also be present.

Children with MCAD do not have any distinct physical features.

There is wide variability within individuals who have this condition.

*Fatty Acid Oxidation Disorders *At a Glance*

When the body can't break down fatty acids to make energy, Fatty Acid Oxidation Disorders (FAOD) occur.

MCAD and VLCAD are the most common of the FAOD, but many different disorders are included in this group.

- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
  http://www.ncbi.nlm.nih.gov/books/NBK1424/

- Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency
  http://www.ncbi.nlm.nih.gov/books/NBK6816/

- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency
  http://ghr.nlm.nih.gov/condition/long-chain-3-hydroxyacyl-coa-dehydrogenase-deficiency

- Trifunctional Protein (TFP) Deficiency
  http://www.newbornscreening.info/Parents/fattyaciddisorders/TFP.html

- Carnitine Transport Defect (Primary Carnitine Deficiency)
  http://www.newbornscreening.info/Parents/fattyaciddisorders/Carnitine.html

- Carnitine-Acylcarnitine Translocase Deficiency (Translocase)
  http://ghr.nlm.nih.gov/condition/carnitine-acylcarnitine-translocase-deficiency

- Carnitine Palmitoyl Transferase I & II (CPT I & II) Deficiency
  http://ghr.nlm.nih.gov/condition/carnitine-palmitoyltransferase-i-deficiency

- 2,4 Dienoyl-CoA Reductase Deficiency
  http://www.fodsupport.org/list.htm

- Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
  http://ghr.nlm.nih.gov/condition/short-chain-acyl-coa-dehydrogenase-deficiency

- Short Chain L-3-Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency (now called 3-Hydroxy Acyl CoA Dehydrogenase Deficiency (HADH)
  http://www.fodsupport.org/list.htm
  Electron Transfer Flavoprotein (ETF) Dehydrogenase Deficiency (GAII & MADD)
  http://www.fodsupport.org/list.htm

- 3-Hydroxy-3 Methylglutaryl-CoA Lyase Deficiency (HMG)
  http://ghr.nlm.nih.gov/condition/3-hydroxy-3-methylglutaryl-coa-lyase-deficiency

- Unclassified FAODs

When a FAOD is diagnosed and treated at birth, the prognosis for most FAOD is excellent. Symptoms and treatments can vary between different FAOD and even can vary within the same FAOD. If undiagnosed and/or untreated, disorders can lead to serious complications.

Complications include problems with the liver, heart, eyes, general muscle development, and possible death.

Symptoms vary with FAOD from complex to no symptoms:

- Chronic bouts of hypoglycemia leading to many hospitalizations within the first years of life

- Lethargy
- Hyotonia
- Liver abnormalities
- Life threatening heart problems

Treatment consists of dietary interventions specific to the particular FAOD.

Children with FAOD do not have any distinct physical features.

What you need to know

- Some children may have intolerance to heat and may need to avoid being outside when it is warm (example: above 80 degrees). This should be determined based on individual needs and decided with the family.

- It is important for an individual with MCAD or any FAOD to eat regularly and **not** go without food or calories, sometimes for as little as 3 hours; this time varies between individuals and depends on circumstances.

- A low fat (<30% of total energy from fat) diet can be beneficial. Coconut, avocado, or high fat foods are not allowed. A high carbohydrate diet may also be recommended. It is important to be sensitive to cultural differences in diet. Carnitine supplements may benefit some children.

- Hypoglycemia must be avoided.

NOTE: A child may appear hydrated and still be heading to crisis. They still require calories to prevent or help or help them through the metabolic crisis/stress.

Recommended Routine Surveillance

- Treatment of symptoms by giving simple carbohydrates orally or by IV if needed to reduce catabolism and sustain anabolism
- Avoid fasting
- Monitor weight with proper nutrition and exercise to prevent secondary complications.
- Avoid infant formulas with medium-chain triglycerides as primary source of fat

Emergency Protocols

- Individuals should have an emergency letter for health care providers, including emergency department staff, who may not be familiar with MCAD/FAOD. This letter

should detail a detailed explanation of management of acute metabolic decompensation, the importance of preventative measures, and numbers of the individual's metabolic specialist
- Sample emergency protocol http://newenglandconsortium.org/protocols/acute_illness/fatty-acid-oxidation-disorders/MCADD.pdf

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Dietician
- Geneticist / Genetic Counselor:
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
- Metabolic specialist initially and then follow-up based on individual needs Clinical trials

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed with MCAD, a form of a Fatty Acid Oxidation defect. MCAD is characterized by a lack of an enzyme involved in the metabolism of fats, which are a major source of energy between meals. Individuals with MCAD do not tolerate prolonged fasting and periods of higher energy demands. Prolonged fasting or illness can cause a person with MCAD to become hypoglycemic and can result in lethargy and metabolic decompensation. Without treatment, the decompensation can progress to coma and death. Because of these, _____ needs the following accommodations.

- Sample emergency protocol http://newenglandconsortium.org/protocols/acute_illness/fatty-acid-oxidation-disorders/MCADD.pdf

Seven Important Aspects of School Life

"MCAD at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

GeneReviews
https://www.ncbi.nlm.nih.gov/books/NBK1424/

New England Consortium of Metabolic Programs
http://newenglandconsortium.org/for-professionals/

An Educator's Guide to MCADD
http://www.gemssforschools.org/Libraries/MCAD_documents/An_Educators_Guide_to_MCADD.sflb.ashx .

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/medium-chain-acyl-coa-dehydrogenase-deficiency

Additional resources including support group information can be found on the main website.