MCAD *At a Glance*

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 go for a long time without food or calories (fasting).

More about fasting:

- During fasting, fatty acids are an important source of energy for liver and other tissues. If they are not converted to energy, it results in lethargy (lack of energy) and hypoglycemia (low blood sugar).
- Energy from fat keeps us going whenever our bodies run low of our main source of energy (glucose.) This might happen if we miss a meal or when we sleep.
- Once sugar or glucose is gone, the body tries to use fat without success. This leads to low blood sugar (known as hypoglycemia) and the buildup of harmful substances in the blood.

About one in every 5-17,000 babies is born with MCAD. MCAD is most prevalent in Caucasians, especially those of Northern European descent, although it can occur in any racial or ethnic group.

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 (low blood sugar), vomiting, and lethargy triggered by a common illness.

Signs of hypoglycemia:

- Weak
- Shaky
- dizzy with clammy cold skin

If untreated, seizures could occur and such episodes could quickly progress to coma and death. Enlarged liver and acute liver disease may also be present.

With newborn screening MCAD is usually diagnosed at birth in the United States.

Children with MCAD do not have any distinct physical features. Although MCAD cannot be seen, it is a serious condition. Good communication between parents, doctors, and school personnel is very important.

Note: There is wide variability within individuals who have this condition.

*Fatty Acid Oxidation Disorders *At a Glance*

We have enzymes that break down fats in our bodies. This process is known as fatty acid oxidation. 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

Fatty acids are a major source of energy for the heart and muscles. Energy from fat keeps us going when our bodies run low on our main source of energy (glucose). This might happen when we miss a meal or sleep. Once sugar or glucose is gone, the body tries to use fat without success. This leads to low blood sugar (known as hypoglycemia) and the buildup of harmful substances in the blood.

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 low blood sugar (hypoglycemia) leading to many hospitalizations within the first years of life

- Lack of energy (lethargy)

- Muscle weakness

- 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.

MCAD - Things to Think About

1. Medical / Dietary Needs

What you need to know

Some children may have an 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. This is especially important during times of high metabolic stress, such as during an illness. This means frequent feeds as infants. Check with parents on specific duration. As the child ages, they may need a cornstarch supplement at bedtime if they have the severe form of MCAD.

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.

Low blood sugar (hypoglycemia) must be avoided. It can come and go and may become more of an issue in teen years with hormonal changes.

Sick day plans need to be in place for illness or other times when a child cannot eat.

What you can do

During the school day, a student may require low fat/high carbohydrate foods throughout the day. Parents may leave cereal, popsicles, or other treats/food with the teacher or nurse to use as needed (i.e. school parties).

When a child is sick they may require more fluids and extra starchy food. Parents may leave these drinks with the nurse to use as needed.

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.

2. Education Supports

What you need to know

It is important to have HIGH EXPECTATIONS for learning for children who have VLCAD.

Good communication between parents, doctors, and school personnel is very important.

A 504 plan for diet and/or gym may be required.

If an individual with classic MCAD or any other FAOD has an acute metabolic event that leads to a brain injury, they may have secondary academic consequences.

Secondary academic consequences due to brain injury may include:

- Loss of developmental milestones
- Acquiring aphasia (an impairment in language ability)
- Attention deficit disorder

3. Behavior & Sensory Support

What you need to know

In a 2011 study at Children's Hospital Boston in *which An Educator's Guide to MCADD* was developed (see Resources below), over half of the children tended to internalize their behaviors more than their peers. Children who internalize behaviors may:

- Monitor their own actions to excess
- Be more compliant, so their problems may go unnoticed
- Worry excessively (e.g. Say “I’m not good at this”)
- Be very sensitive
- Have high anxiety
- Be more likely to withdraw from social situations

What you can do

Results from this study indicated that children with MCAD need a little extra encouragement to join activities, speak up in class, or tell the teacher if they have a problem.

4. Physical Activity, Trips, Events

What you need to know

It is very important that individuals with MCAD have access to fluids and snacks. They need to bring snacks or drinks on field trips that last more than a few hours.

Students may have challenges with executive functioning, distractibility, anxiety, hearing and speech.

What you can do

If the student has challenges with executive functioning, distractibility, anxiety, hearing and/or speech, consider the need for extra supports.

5. School Absences & Fatigue

What you need to know

There is wide variability in this condition. Some individuals with MCAD or any FAOD may have increased absences due to their condition. Most do not.

They should not require accommodations for fatigability as long as they are provided adequate nutrition and fluids. Absences may be due to viruses, stress, episodes of low blood sugar, etc.

6. Emergency Planning

What you need to know

All individuals who have MCAD need an emergency letter to be given to health care providers in the event of a crisis. The letter should address the following:

- When a child with MCAD has a viral illness or otherwise has decreased intake, they may become lethargic or hypoglycemic.
- Glucose through an IV is essential to prevent hypoglycemia and other complications.
- Symptoms that signal that a child with MCAD may be in trouble may include:
    - Lack of energy
    - Lethargy
    - Muscle weakness
    - Muscle cramps
    - Any vomiting or diarrhea

What you can do

Obtain a copy of the emergency letter so treatment can begin as soon as possible.

7. Resources

New England Consortium of Metabolic Programs

http://newenglandconsortium.org/

The New England Consortium of Metabolic Programs brings together healthcare professionals at all levels involved in identifying and treating individuals with metabolic disorders.

The goals of the Consortium are to disseminate information, collaborate in the development of social support programs and educational materials, support organizations for parents and adults with metabolic disorders, provide training for students in medicine and related fields, jointly develop and conduct research projects, and establish uniform treatment protocols for individuals with metabolic disorders.

An Educator's Guide to MCADD

http://www.gemssforschools.org/Libraries/MCAD_documents/An_Educators_Guide_to_MCADD.sflb.ashx

This project was supported by the NEGC and HRSA, with help from physicians at Children's Hospital Boston, Elementary school teachers, Parents of children with MCADD, The FOD Family Support Group.

FOD Family Support Group

http://www.fodsupport.org/

"All in this together"

Genetic Fact Sheets for Parents

http://www.newbornscreening.info/Parents/fattyaciddisorders/MCADD.html

Fatty Acid Oxidation Disorders - MCAD
by Screening, Technology & Research in Genetics/Expanded Newborn Screening

Genetics Home Reference

http://ghr.nlm.nih.gov/condition/medium-chain-acyl-coa-dehydrogenase-deficiency

Your Guide to Understanding Genetic Conditions

National Center for Biotechnology Information (NCBI) Bookshelf - VLCAD

http://www.ncbi.nlm.nih.gov/books/NBK1424/

Learn more about the genetics of MCAD

***Note:** This printable version does not include the information found under the green button marked "Transitions" on the website. Those general pages may be printed separately.*

8. Meet a Child with MCAD

Meet Shawna!

GEMSS would like to thank Shawna and her mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!

Shawna is in 4th grade and is a very active and generous little girl. Her favorite things to do are basketball, Tae Kwon Do, horseback riding, and swimming. She loves being with her friends and going to school every day and she is very outgoing. When she was only 2 weeks old, Shawna was diagnosed with MCADD through New Born Screening.

School can be a little difficult for Shawna at times. She struggles with her fluency in reading and retaining math facts. She goes to Intensive Title Reading and Title Math and she has improved greatly in just a matter of months.

Shawna is her own advocate at school. She understands her MCADD and does not hesitate to explain it to friends, teachers, nurses, doctors, and her friend's parents. We have an emergency plan in place at the school for teachers and nurses. At home, Shawna is working on being more independent with her homework although it is checked when it is completed.

Shawna is very generous toward others. Although she is has spent a lot of time in the hospital herself, she always is worried about how other children are doing. After receiving a wish from the Make-A-Wish Foundation, she wanted to raise money to donate to other children who are ill. With the help of the Center for Civic Engagement at St Lawrence University Shawna helped organize Shawna's Walk For Wishes. Shawna is proud to announce that after completing this year's, 3rd Annual Shawna's Walk For Wishes, we have raised a grand total of just over $21,000. If you would like to check out Shawna and her mom talking about MCADD and her wish, click here to see their YouTube video.

Her mother has some advice for teachers and that is "to not judge a book by its cover." "I always hear that she doesn't look sick, and many times it means that they are missing the subtle signs," says her mother Melissa. Her mother's advice to other parents is to "be an active advocate for your child, be an open book about the disorder, and teach your children to advocate for themselves."