



## **MCADD** **(Medium Chain Acyl-CoA Dehydrogenase Deficiency)**

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### **What is it?**

MCADD stands for medium chain acyl-CoA dehydrogenase deficiency. It is one type of fatty acid oxidation disorder. People with MCADD have problems breaking down fat into energy for the body.

### **What causes it?**

MCADD occurs when an enzyme called medium chain acyl-CoA dehydrogenase (MCAD) is either missing or not working properly. This enzyme's job is to break down certain fats in the food we eat into energy. It also breaks down fat already stored in the body.

Energy from fat keeps us going whenever our bodies run low of their main source of energy, a type of sugar called glucose. Our bodies rely solely on glucose. Although glucose is a good source of energy, there is a limited amount available. Once the glucose has been used up, the body tries to use fat without success. This leads to low blood sugar, called hypoglycemia, and to the buildup of harmful substances in the blood.

### **If MCADD is not treated, what problems occur?**

MCADD can cause bouts of illness called metabolic crises. Children with MCADD often show effects for the first time between 3 months and 2 years of age. Some of the first symptoms of a metabolic crisis are:

- 1) Extreme sleepiness.
- 2) Behavior changes.
- 3) Irritable mood.
- 4) Poor appetite.

Other symptoms then follow:

- 1) Fever
- 2) Diarrhea
- 3) Vomiting
- 4) Hypoglycemia (low blood sugar)

If a metabolic crisis is not treated, a child with MCADD can develop:

- 1) Breathing problems.
- 2) Seizures.
- 3) Coma, sometimes leading to death.

Between episodes of metabolic crisis, people with MCADD are usually healthy. However, repeated episodes can cause permanent brain damage. This may result in learning problems, mental retardation or spasticity.

Symptoms often happen after having nothing to eat for more than a few hours. Hypoglycemia can occur, with or without the other symptoms of metabolic crisis, just by going too long without food. Hypoglycemia can cause a person to feel weak, shaky or dizzy and have clammy, cold skin. If not treated, hypoglycemia can lead to coma and possibly death.

Hypoglycemia and metabolic crises also are more likely to occur when a person with MCADD gets sick or has an infection. Some children with MCADD never have symptoms and are found to be affected only after a brother or sister is diagnosed.

### **What is the treatment for MCADD?**

Your baby's primary doctor will work with a metabolic doctor to care for your child. Your

doctor may also suggest that you meet with a dietician familiar with MCADD.

Certain treatments may be advised for some children but not others. When necessary, treatment usually is needed throughout life. The following are treatments often recommended for children with MCADD:

### ***Avoid Going a Long Time Without Food***

Babies and young children with MCADD need to eat often to avoid hypoglycemia or a metabolic crisis. These children should not go without food for more than four to six hours. Some babies may need to eat even more often than this. It is important that babies be fed during the night. They need to be awakened to eat if they do not wake up on their own.

Young children with MCADD may need to have a starchy snack before bed and another during the night. They need another snack first thing in the morning. Raw cornstarch mixed with water, milk or other drink is a good source of long-lasting energy that is sometimes suggested for children older than age 1. Your dietician can give you ideas for good nighttime snacks.

When they are well, most teens and adults with MCADD can go without food for up to 12 hours without problems. They do need to continue other treatments throughout life.

### ***Diet***

Sometimes a low-fat, high-carbohydrate diet is recommended. Carbohydrates give the body many types of sugar that can be used as energy. In fact, for children needing this treatment, most food in the diet should be carbohydrates (bread, pasta, fruit, vegetables, etc.) and protein (lean meat and low-fat dairy foods). Your dietician can create a food plan with the correct type and amount of fat your child needs. Any diet changes should be made under the guidance of a dietician.

Ask your doctor whether your child needs to have any changes in his or her diet.

### ***L-Carnitine***

Some children may be helped by taking L-carnitine. This is a safe and natural substance that helps the body make energy. It also helps the body get rid of harmful wastes. Your doctor will decide whether your child needs L-carnitine. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor. Do not use any medication without checking with your doctor.

### ***Call Your Doctor at the Start of Illness***

Always call your health-care provider when your child has any of the following:

- 1) Poor appetite
- 2) Low energy or excessive sleepiness
- 3) Vomiting
- 4) Diarrhea
- 5) Infection
- 6) Fever

People with MCADD need to eat extra starchy food and drink more fluids during an illness – even if they may not feel hungry – or they could develop hypoglycemia or a metabolic crisis. Children who are sick often don't want to eat. If they won't or can't eat, they may need to be treated in the hospital to prevent serious health problems.

Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.

### ***What happens when MCADD is treated?***

With prompt and careful treatment, children MCADD usually live healthy lives with typical growth and development. The goal of treatment is to prevent long-term problems. However, children who have repeated metabolic crises may have life-long learning disabilities, spasticity, or other effects.

### **What causes the MCAD enzyme to be absent or not working correctly?**

Genes tell the body to make various enzymes. People with MCADD have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the MCAD enzyme either does not work properly or is not made at all.

### **Is MCADD inherited?**

MCADD is inherited and affects both boys and girls equally.

Everyone has a pair of genes that make the MCAD enzyme. In children with MCADD, neither of these genes works correctly. These children inherit one nonworking gene for the condition from each parent.

Parents of children with MCADD rarely have the disorder. Instead, each parent has a single nonworking gene for MCADD. They are called carriers. Carriers do not have MCADD because the other gene of this pair is working correctly. When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have MCADD. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

### **Can other members of the family have MCADD or be carriers?**

#### ***Having MCADD***

The brothers and sisters of a baby with MCADD have a chance of being affected, even if they haven't had symptoms. Finding out whether other children in the family have MCADD is important because early treatment may prevent serious health problems. Talk to your doctor or genetic counselor about testing your other children for MCADD.

#### ***MCADD Carriers***

Brothers and sisters who do not have MCADD still have a chance to be carriers like their

parents. Except in special cases, carrier testing should be done only in people older than 18.

Each of the parents' brothers and sisters has a 50 percent chance to be a MCADD carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with MCADD.

When both parents are carriers, newborn screening results are not sufficient to rule out MCADD in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

### **Can other family members be tested?**

#### ***Diagnostic Testing***

Brothers and sisters can be tested for MCADD using DNA testing or other special tests.

#### ***Carrier Testing***

If both gene changes have been found in your child with MCADD, other family members can have DNA testing to see if they are carriers.

If DNA testing would not be helpful, carrier testing can also be done by an enzyme test on a skin sample.

### **How many people have MCADD?**

About one in every 15,000 babies in the United States is born with MCADD.

### **Does MCADD happen more frequently in a certain ethnic group?**

MCADD happens more often in white people from Northern Europe and the United States. About one in every 70 Caucasians is a carrier for MCADD.

### **Does MCADD go by any other names?**

MCADD is also sometimes called:

- 1) MCAD deficiency.
- 2) ACADM deficiency.
- 3) MCADH deficiency.

### **Where can I find more information?**

Fatty Oxidation Disorders (FOD) Family Support Group

[www.fodsupport.org](http://www.fodsupport.org)

Organic Acidemia Association

[www.oaanews.org](http://www.oaanews.org)

United Mitochondrial Disease Foundation

[www.umdf.org](http://www.umdf.org)

Children Living with Inherited Metabolic Diseases (CLIMB)

[www.climb.org.uk](http://www.climb.org.uk)

Genetic Alliance

[www.geneticalliance.org](http://www.geneticalliance.org)

### **Children's Special Health Services (CSHS)**

State Capitol Judicial Wing

600 E. Boulevard Ave., Department 301

Bismarck, ND 58505-0269

Toll Free: 800.755.2714

701.328.2436

Relay TDD: 701.328.3975

CSHS website: [www.ndhealth.gov/CSHS](http://www.ndhealth.gov/CSHS)

North Dakota Department of Health website:

[www.ndhealth.gov](http://www.ndhealth.gov)

### **Family support resources available from CSHS:**

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

### ***Family Resources***

Family to Family Network

Center for Rural Health

University of North Dakota

School of Medicine and Health

PO Box 9037

Grand Forks, ND 58202-9037

Toll Free: 888 434.7436

701.777.2359

Fax: 701.777.2353

Email: [NDF2F@medicine.nodak.edu](mailto:NDF2F@medicine.nodak.edu)

[www.medicine.nodak.edu/crh](http://www.medicine.nodak.edu/crh)

Pathfinder Services of ND

Pathfinder Family Center

1600 2<sup>nd</sup> Ave. SW, Ste. 19

Minot, ND 58701

Toll Free: 800.245.5840

701.837.7500

Relay TDD: 701.837.7501

Email: [ndpath01@ndak.net](mailto:ndpath01@ndak.net)

[www.pathfinder.minot.com](http://www.pathfinder.minot.com)

Family Voices of North Dakota, Inc.

P.O. Box 163

Edgeley, ND 58433

Toll Free: 888.522.9654

701.493.2634

Fax: 701.493.2635

[www.geocities.com/ndfv](http://www.geocities.com/ndfv)

\*\*This fact sheet has general information.

Every child is different and some of these facts may not apply to your child specifically.

Certain treatments may be recommended for some children but not others. All children should be followed by a metabolic doctor in addition to their primary-care provider.

### **Acknowledgement**

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### **Disclaimer**

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North Dakota Department of Health  
Newborn Screening Program  
600 E. Boulevard Ave., Dept. 301  
Bismarck, ND 58505.0200  
800.472.2286 or 701.328.2496  
[www.ndhealth.gov/familyhealth](http://www.ndhealth.gov/familyhealth)