



## Genetic Fact Sheets for Parents

# Fatty Acid Oxidation Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – <http://www.newbornscreening.info>

**Disorder Name: Medium chain acyl-CoA dehydrogenase deficiency**

**Acronym: MCADD**

- What is MCADD?
- What causes MCADD?
- If MCADD is not treated, what problems occur?
- What is the treatment for MCADD?
- What happens when MCADD is treated?
- What causes the MCAD enzyme to be absent or not working correctly?
- How is MCADD inherited?
- Is genetic testing available?
- What other testing is available?
- Can you test during a future pregnancy?
- Can other members of the family have MCADD or be carriers?
- Can other family members be tested?
- How many people have MCADD?
- Does MCADD happen more often in a certain ethnic group?
- Does MCADD go by any other names?
- Where can I find more information?

This fact sheet contains general information about MCADD. Every child is different and some of this information may not apply to your child specifically. Certain treatments may be recommended for some children but not others. Children with MCADD should be followed by a metabolic doctor in addition to their primary doctor.

## What is MCADD?

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.

## Fatty Acid Oxidation Disorders:

Fatty acid oxidation disorders (FAODs) are a group of rare inherited conditions. They are caused by enzymes that do not work properly.

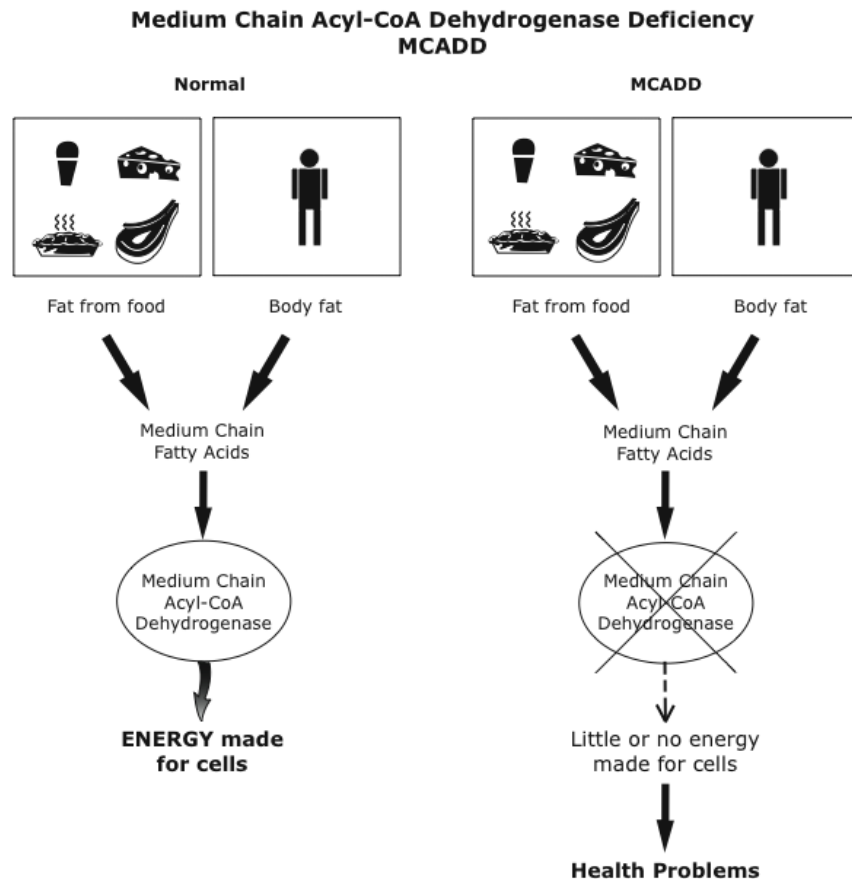
A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

The symptoms and treatment vary between different FAODs. They can also vary from person to person with the same FAOD. See the fact sheets for each specific FAOD.

FAODs are inherited in an autosomal recessive manner and affect both males and females.

## What causes MCADD?

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 on fat when we don't eat for a stretch of time – like when we miss a meal or when we sleep.

When the MCADD enzyme is missing or not working well, the body cannot use certain types of fat for energy and must 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 build up 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 three months and three years of age. Some of the first symptoms of a metabolic crisis are:

- extreme sleepiness
- behavior changes
- irritable mood
- poor appetite

Some of these other symptoms may also follow:

- fever
- diarrhea
- vomiting
- hypoglycemia

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

- breathing problems
- seizures
- 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, intellectual disabilities, or spasticity.

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

Hypoglycemia and metabolic crises are also more likely to occur when a person with MCADD gets sick or has an infection.

Some children with MCADD have very mild symptoms or no symptoms at all.

## **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 is usually needed throughout life. The following are treatments often recommended for children with MCADD:

### **1. Avoid going a long time without food**

Infants and young children with MCADD need to eat frequently to prevent hypoglycemia or a metabolic crisis. Your metabolic doctor will tell you how often your child needs to be fed. In general, it is often suggested that infants be fed every four to six hours. Some babies need to eat even more frequently than this. It is important that infants be fed during the night. They may need to be woken up to eat if they do not wake up on their own. Your metabolic doctor and dietician will give you an appropriate feeding plan for your infant. Your doctor will also give you a 'sick day' plan, tailored to your child's needs, for you to follow during illnesses or other times when your child will not eat.

Your metabolic doctor will continue to advise you on how often your child should eat as he or she gets older. When they are well, many teens and adults with MCADD can go without food for up to 12 hours without problems. Most children do not have metabolic crises past the age of ten. However, some may need to continue treatment throughout life.

### **2. Diet**

Sometimes a diet that is low in fat and high in carbohydrates is recommended. Carbohydrates give the body many types of sugar that can be used as energy. In fact, for children needing this treatment, most foods 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 an experienced dietician.

Ask your doctor whether or not your child needs to have any changes in his or her diet.

### **3. L-carnitine**

Some children may be helped by taking L-carnitine. This is a safe and natural substance that helps body cells make energy. It also helps the body get rid of harmful wastes.

Your doctor will decide whether or not your child needs L-carnitine. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor. Do not use any medications without checking with your doctor.

#### **4. Call your doctor at the start of any illness**

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

- poor appetite
- low energy or excessive sleepiness
- vomiting
- diarrhea
- an infection
- a fever

Children with MCADD need to eat extra starchy food and drink more fluids during any 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 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 with 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, chronic muscle weakness, or other effects.

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

Genes tell the body to make various enzymes. The ACADM gene instructs the body to make the medium chain acyl-CoA dehydrogenase (MCAD) enzyme. Everyone has two copies of the ACADM gene. People with MCADD have changes, also called variants in both copies of their ACADM genes that cause them to not work correctly. Because of the variants in the ACADM genes, the MCAD enzyme either does not work properly or is not made at all.

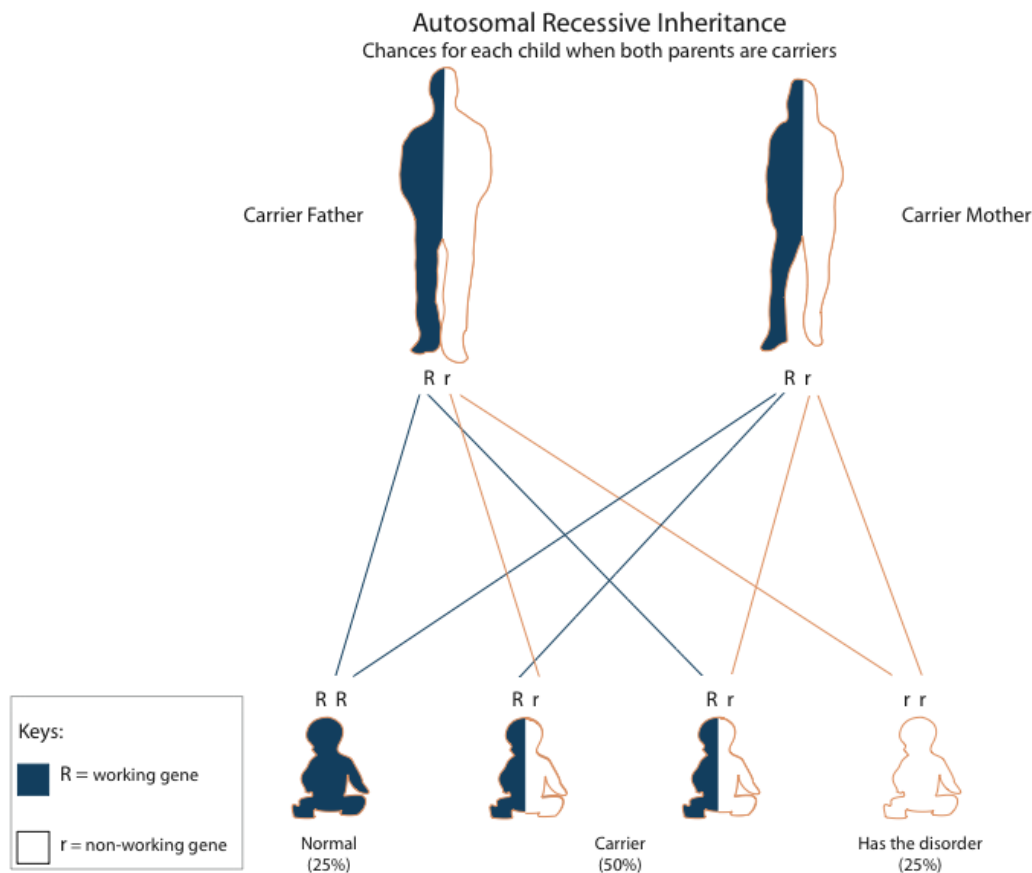
#### **How is MCADD inherited?**

MCADD is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has two copies of the ACADM gene that make the MCAD enzyme. In children with MCADD, neither of these genes works correctly. These children inherit one non-working ACADM gene from each parent.

Parents of children with MCADD rarely have the disorder. Instead, each parent has a single non-working ACADM gene. They are called carriers. Carriers do not have MCADD because their other ACADM gene is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have MCADD. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



Genetic counseling is available to families who have children with MCADD. Genetic counselors can answer your questions about how MCADD is inherited, choices during future pregnancies, and how to test other family members. Ask your doctor about a referral to a genetic counselor.

## Is genetic testing available?

Genetic testing for MCADD can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes in both copies of the ACADM genes. In many children with MCADD, both gene changes can be found. However, in other children, only one or neither of the two gene changes can be found, even though we know they are present.

DNA testing is not necessary to diagnose your child. It can be helpful for carrier testing or prenatal diagnosis, discussed below.

## What other testing is available?

MCADD can also be confirmed either by a blood test called an acylcarnitine profile or an enzyme test on a skin sample. Talk to your doctor or your genetic counselor if you have questions about testing for MCADD.

## Can you test during a future pregnancy?

If both gene changes have been found in your child with MCADD, DNA testing can be done during future pregnancies. The sample needed for this test is obtained by either CVS or amniocentesis.

If DNA testing would not be helpful, an enzyme test can be done during pregnancy on cells from the fetus. Again, the sample needed for this test is obtained by either CVS or amniocentesis.

Parents may either choose to have testing during pregnancy or wait until birth to have the baby tested. Parents may also choose to use assisted reproductive techniques to decrease the chance that their future children would have MCADD. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

## 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 have had no 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 only be done on people over 18 years of age.

If you are a parent of a child with MCADD, your brothers and sisters have a 50% chance to be an 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.

All states offer newborn screening for MCADD. However, 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.

During pregnancy, women carrying fetuses with MCADD may be at increased risk to develop serious medical problems. Some women carrying fetuses with Fatty Acid Oxidation Disorders have developed:

- excessive vomiting
- abdominal pain
- high blood pressure
- jaundice
- abnormal fat storage in the liver
- severe bleeding

All women with a family history of MCADD should share this information with their obstetricians and other health care providers before and during any future pregnancies. Knowing about these risks allows better medical care and early treatment if needed.

## **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. MCADD occurs less frequently in some other populations with 1 in 51,000 babies in Japan and 1 in 263,500 babies in Taiwan born with MCADD.

## Does MCADD happen more often in a certain ethnic group?

MCADD happens more often in white people from Northern Europe and the United States. About 1 in every 70 Caucasians is a carrier for MCADD.

## Does MCADD go by any other names?

MCADD is also sometimes called:

- MCAD deficiency
- ACADM deficiency
- MCADH deficiency

## Where can I find more information?

Fatty Oxidation Disorders (FOD) Family Support Group

<http://www.fodsupport.org>

Organic Acidemia Association

<http://www.oaanews.org>

United Mitochondrial Disease Foundation

<http://www.umdf.org>

Metabolic Support UK

<https://www.metabolicsupportuk.org>

Baby's First Test

<http://www.babysfirsttest.org>

MedlinePlus

<https://medlineplus.gov/genetics/condition/medium-chain-acyl-coa-dehydrogenase-deficiency/>

**Document Info**

**Created by:** www.newbornscreening.info

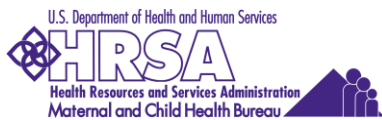
**Reviewed by:** HI, CA, OR, and WA metabolic specialists

**Review date:** September 31, 2007  
April 2, 2011  
July 13, 2013  
February 28, 2016  
April 29, 2020  
March 1, 2023

**Updated on:** March 1, 2023

**DISCLAIMER:**

THIS INFORMATION DOES NOT PROVIDE MEDICAL ADVICE. All content ("Content"), including text, graphics, images and information are for general informational purposes only. You are encouraged to confer with your doctor or other health care professional with regard to information contained on this information sheet. After reading this information sheet, you are encouraged to review the information carefully with your doctor or other healthcare provider. The Content is not intended to be a substitute for professional medical advice, diagnosis or treatment. NEVER DISREGARD PROFESSIONAL MEDICAL ADVICE, OR DELAY IN SEEKING IT, BECAUSE OF SOMETHING YOU HAVE READ ON THIS INFORMATION SHEET.



This project is supported by a grant from the Maternal and Child Health Bureau, Health Resources and Service Administration, Genetic Services Branch, MCH Project #: UH7MC30774-01-00 <http://mchb.hrsa.gov>