

# Parents' Guide to MCADD

Medium Chain Acyl-CoA Dehydrogenase Deficiency



California  
Department of  
Health Services



Newborn Screening Program  
Genetic Disease Branch  
[www.ca.gov/gdb](http://www.ca.gov/gdb)

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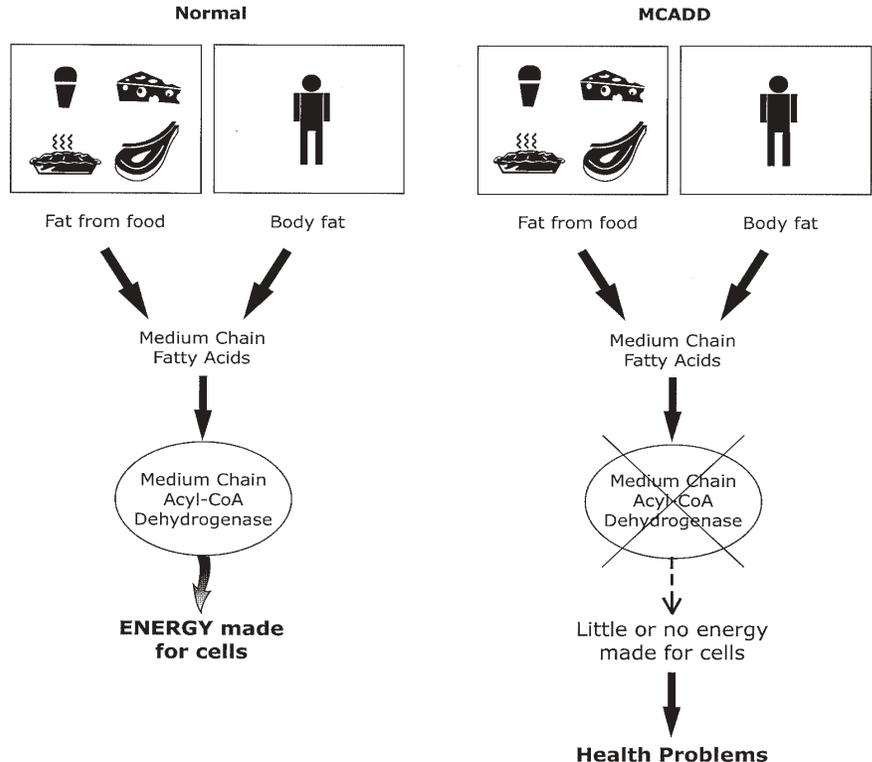
**The information in this booklet is general and is not meant to be specific to each child with MCADD. Certain treatments may be recommended for some children but not others. Children with MCADD should be followed by a physician specializing in metabolic diseases (metabolic specialist) in addition to their primary doctor. For a list of metabolic clinics, visit our website at [www.dhs.ca.gov/gdb](http://www.dhs.ca.gov/gdb).**

\*Underlined words in booklet are defined in the glossary

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## What is MCADD?

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



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## 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 while – such as when we miss a meal or when we sleep.

When the MCAD enzyme is missing or not working, the body cannot use 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, also called hypoglycemia, and to the build up of harmful substances in the blood.

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## What causes the MCAD enzyme to be missing 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.

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

MCADD can cause bouts of illness called metabolic crises. Children with MCADD usually get symptoms for the first time between 3 months and 2 years of age. Some of the first symptoms of a metabolic crisis are extreme sleepiness, behavior changes, irritable mood, and poor appetite. Crises are often set off by an infection; so illness may start with symptoms of fever, diarrhea, and vomiting. Low blood sugar then follows. If a metabolic crisis is not treated, a child with MCADD can develop breathing problems, seizures, and coma, sometimes leading to death.

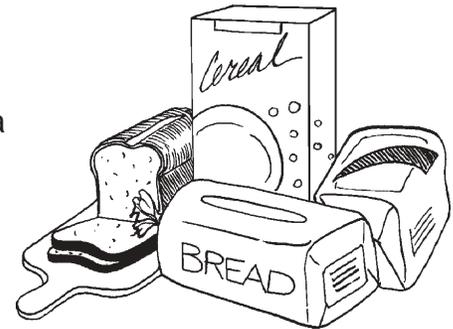
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Between bouts of metabolic crises, people with MCADD are usually healthy. However, repeated episodes can cause permanent brain damage. This may result in learning problems, mental retardation or spasticity.

Symptoms often happen after having nothing to eat for more than a few hours. During long periods without eating food, the glucose in the body is used up. The body then attempts to break down fat, which leads to the build up of harmful substances in the blood.

Low blood sugar can occur, with or without other symptoms of metabolic crisis, just by going too long without food. Low blood sugar can cause a person to feel weak, shaky or dizzy, and have clammy, cold skin. If not treated, low blood sugar can lead to coma and even death.

Low blood sugar and metabolic crises are also more likely to occur when a person with MCADD gets sick or has an infection. When we are sick, our bodies need extra energy to fight the illness. People with MCADD need to eat extra starchy food and



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drink more fluids during any illness - even if they may not feel hungry - or they could develop low blood sugar or a metabolic crisis. Children who are sick often don't want to eat or they vomit and can not keep food down. If they won't or can't eat, they need to be treated in the hospital with intravenous (IV) glucose to prevent problems.

Some children with MCADD never have symptoms and are only found after a brother or sister is diagnosed.

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

Your baby's primary doctor will work with a metabolic specialist 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 pages describe the treatments often recommended for children with MCADD.



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## 1. Avoid going a long time without food

Babies and young children with MCADD need to eat often to avoid low blood sugar or a metabolic crisis. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often than this. It is important that babies be fed during the night. You need to wake them up 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 may 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. This is sometimes suggested for children older than one year of age. Your dietician can give you other ideas for good 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 the other treatments throughout life.

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## 2. 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 food).

Ask your doctor whether or not your child needs to have any changes in his or her diet. Any diet changes should be made under the guidance of a dietician.



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### 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. Do not use any medications without checking with your doctor.

### 4. Call your doctor immediately at the sign of any of these symptoms in your child:

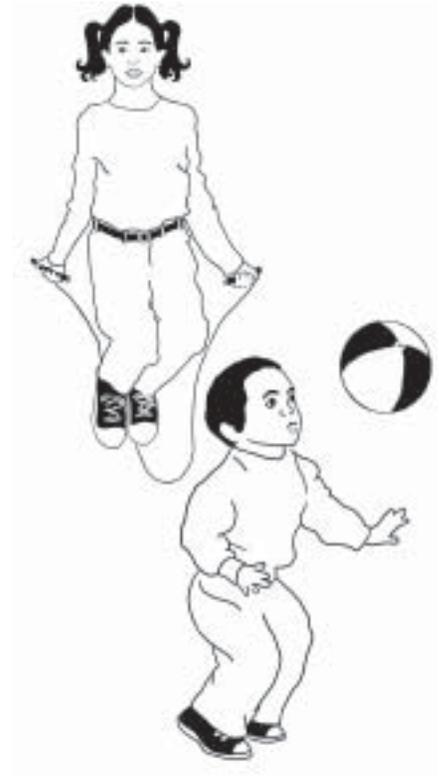
- poor appetite
- low energy or excessive sleepiness
- vomiting
- diarrhea
- an infection
- a fever



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## 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, or other effects.



## How is MCADD inherited?

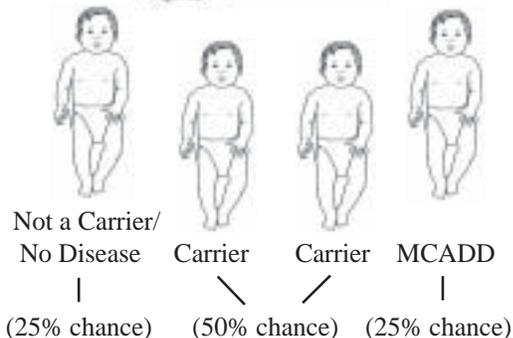
MCADD affects both boys and girls equally.

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 non-working gene for the condition from each parent. This is called autosomal recessive inheritance.

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

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have two working genes. This means the child is not a carrier and does not have the disease. There is a 50% chance for the child to be a carrier, just like the parents. There is a 25% chance for the child to have MCADD.

MCADD Carrier MCADD Carrier



*Chances apply to each pregnancy*

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

### 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 the pair of genes that cause MCADD. Up to 80% of children with MCADD have one particular gene change in both genes of this pair. About 2% of affected children have gene changes that cannot be found through current tests. About 18% have only one gene change show up through testing. The other cannot be easily found, even though we know it is present. These percentages may differ in non-Caucasian families.

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



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## What other testing is available?

MCADD can also be confirmed by either 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 pregnancy?

If both gene changes are known in your child with MCADD, DNA testing can be done during future pregnancies to determine if the sibling also has MCADD. The sample needed for this test is obtained by either CVS or amniocentesis.

If the gene changes are not known in the child with MCADD, 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.



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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?**

The brothers and sisters of a baby with MCADD have a chance of also having MCADD even if they have had no symptoms. Finding out if 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.

Brothers and sisters who do not have MCADD still have a chance to be carriers like their parents. Carriers do not have the disorder and will never develop it.

Each of the parents' brothers and sisters has 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.

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When both parents are known MCADD carriers or have had a baby with MCADD, subsequent newborns should have special diagnostic testing in addition to the newborn screen to test for MCADD.

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

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

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

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

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## RESOURCES

Fatty Oxidation Disorders (FOD)  
Family Support Group  
1559 New Garden Road, 2E  
Greensboro, NC 27410  
(336) 547-8682  
[www.fodsupport.org](http://www.fodsupport.org)

United Mitochondrial Disease Foundation  
8085 Saltsburg Road, Suite 201  
Pittsburgh, PA 15239  
(412) 793-8077  
[www.umdf.org](http://www.umdf.org)

Children Living with Inherited Metabolic Diseases  
CLIMB Building  
176 Nantwich Road  
Crewe, CW2 6BG  
United Kingdom  
[www.climb.org.uk](http://www.climb.org.uk)

Genetic Alliance  
4301 Connecticut Ave. NW, Suite 404  
Washington, DC 20008-2369  
(202) 966-5557  
[www.geneticalliance.org](http://www.geneticalliance.org)

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## GLOSSARY

**Acylcarnitine profile** - This is a blood test that can be used to diagnose certain fatty acid oxidation and organic acid disorders. Fatty acids get broken down into substances called acylcarnitines. These substances are then used to create energy for the body. Certain fatty acid oxidation disorders cause changes in the usual pattern of acylcarnitines.

**Amniocentesis** - Test done during pregnancy (usually between 13 and 20 weeks). A needle is used to remove a small sample of fluid from the sac around the fetus. The sample can be used to test for certain genetic disorders in the fetus.

**Autosomal recessive** - Most of the metabolic disorders that can be detected by newborn screening are inherited in an "autosomal recessive" pattern. Autosomal recessive conditions affect both boys and girls equally. How autosomal recessive inheritance works: Everyone has a pair of genes for each enzyme in the body. A separate pair of genes is responsible for making each enzyme. A person with a metabolic disorder has one enzyme that is either missing or not working properly. The problem is caused by a pair of "recessive" genes that are not working correctly. They do not make the needed enzyme. A person has to have two

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non-working "recessive" genes in order to have an autosomal recessive metabolic disorder. A person with an autosomal recessive disorder inherits one non-working gene from his or her mother and the other from his or her father. The parents are called carriers for that condition. Parents of children with a metabolic disorder rarely have the disorder themselves. Instead, for that pair of genes, each parent has one that is working correctly and one that is not working (called the "recessive gene.") People with a single non-working gene are called carriers. If one gene of the pair is working correctly, it makes up for the recessive non-working gene. Therefore, carriers usually will not have the condition.

**Carrier** - A person who has a gene mutation in one of their genes that cause a disease, but does not have any symptoms of the disease themselves. The mutation is often recessive, which means that both copies of the gene have to be mutated in order for disease symptoms to develop. Carriers are able to pass the mutation onto their children and therefore have an increased chance of having a child with the disease.

**CVS** - Chorionic Villus Sampling (CVS) is a special test done during early pregnancy (usually between 10 and 12 weeks). A small sample of the placenta is removed for testing. This sample can be used to test for certain genetic disorders in the fetus.

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**DNA** - Deoxyribonucleic acid (DNA) is a molecule that makes up chromosomes. It is composed of four units (called bases) that are designated A, T, G, and C. The sequence of the bases spell out instructions for making all of the proteins in an organism. The instructions set for each individual protein is a gene. A change in one of the DNA letters making up a gene is a mutation. In some cases, these mutations can alter the protein instructions and lead to disease. Each individual passes their chromosomes on to their children, and therefore pass down the DNA instructions. It is these instructions that cause certain traits, such as eye or hair color, to be inherited.

**Enzyme** - A molecule that helps chemical reactions take place. For example, enzymes in the stomach speed up the process of breaking down food. Each enzyme can participate in many chemical reactions without changing or being used up.

**Gene** - A segment of DNA that contains the instructions to make a specific protein (or part of a protein). Genes are contained on chromosomes. Chromosomes, and the genes on those chromosomes, are passed on from parent to child. Errors in the DNA that make up a gene are called mutations and can lead to diseases.

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**Genetic Counseling** - Genetic counseling gives patients and their families education and information about genetic-related conditions and helps them make informed decisions. It is often provided by Genetic Counselors or Medical Geneticists who have special training in inherited disorders.

**Metabolic Crisis** - This is a serious health condition caused by low blood sugar and the build-up of toxic substances in the blood. Symptoms of a metabolic crisis are: poor appetite, nausea, vomiting, diarrhea, extreme sleepiness, irritable mood and behavior changes. If not treated, breathing problems, seizures, coma, and sometimes even death can occur. Metabolic crises happen more often in people with certain metabolic disorders (some fatty acid oxidation disorders, amino acid disorders, and organic acid disorders). They are often triggered by things like illness or infection, going without food for a long time, and, in some cases, heavy exercise.

**Seizures** - These are also called “convulsions” or “fits”. During a seizure a person loses consciousness and control of his or her muscles. It may also cause involuntary movements. Seizures can happen for many reasons. Some causes are metabolic disorders, a metabolic crisis, brain injury, or infections.

**Spasticity** - This is rigidity of the muscles and increased reflexes. It is caused by increased muscle tone. It results in abnormal tightness or stiffness of the muscles.

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