

# Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

A fatty acid oxidation disorder

## *What is it?*

Medium Chain Acyl-CoA Dehydrogenase Deficiency (also known as MCAD) is an inherited fatty acid oxidation disorder. People with fatty acid oxidation disorders, like MCAD, cannot breakdown fats for energy. Once the body uses up its primary source of energy (glucose, or blood sugar), the body begins to fail because it cannot then make energy from fats. Therefore, people with MCAD must eat on a very regular basis and cannot go long without food.

## *What are the symptoms?*

A person with MCAD can appear normal at birth. Symptoms usually appear sometime in infancy. Some individuals with MCAD have recurrent episodes of hypoglycemia, lack of energy, vomiting, or seizures. These symptoms can progress very quickly to coma, cardiac arrest, brain damage, or even death in children who are not eating well. Many symptoms of MCAD can be prevented by immediate treatment and lifelong management. People with MCAD typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

## *Inheritance and Frequency*

MCAD is inherited in an autosomal recessive manner. This means that for a person to be affected with MCAD, he or she must have inherited two non-working copies of the gene responsible for causing MCAD. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have MCAD. Typically, there is no family history of MCAD in an affected person. MCAD is the most common fatty acid oxidation disorder, affecting about 1 in 10,000 individuals. It is most common among individuals of Northern European descent.

## *How is it detected?*

MCAD may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

## *How is it treated?*

MCAD is treated by eating frequently and avoiding fasting, and sometimes medication, as recommended by a genetic metabolic medical professional.

**DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.**

## **For more information:**

### **Genetics Home Reference**

Website: <http://www.ghr.nlm.nih.gov>

### **Save Babies Through Screening Foundation**

4 Manor View Circle

Malvern, PA 19355-1622

Toll Free Phone: 1-888-454-3383

Fax: (610) 993-0545

Email: [email@savebabies.org](mailto:email@savebabies.org)

Website: <http://www.savebabies.org>

### **FOD (Fatty Oxidation Disorder) Family Support Group**

1559 New Garden Rd, 2E

Greensboro, NC 27410

Phone: (336) 547-8682 [8am - 8pm EST every day]

Fax: (336) 292-0536 [email/call ahead between 8am and 8pm before faxing]

Email: [deb@fodsupport.org](mailto:deb@fodsupport.org)

Website: <http://www.fodsupport.org>

### **United Mitochondrial Disease Foundation**

8085 Saltsburg Road, Suite 201

Pittsburgh, PA 15239

Phone: (412) 793-8077

FAX: (412) 793-6477

email: [info@umdf.org](mailto:info@umdf.org)

website: <http://www.umdf.org>

### **American College of Medical Genetics**

Newborn Screening ACT Sheets and Confirmatory Algorithms

<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>

### **Cardinal Glennon Children's Hospital**

St. Louis, Missouri 314-577-5639

Website: <http://pediatrics.slu.edu/index.phtml?page=geneticsdiv>

### **Children's Hospital at University Hospital and Clinics**

Columbia, Missouri 573-882-6991

Website: <http://www.genetics.missouri.edu/>

### **Children's Mercy Hospital**

Kansas City, Missouri 816-234-3290

Website: <http://www.childrens-mercy.org/content/view.aspx?id=155>

### **St. Louis Children's Hospital**

St. Louis, Missouri 314-454-6093

Website: <http://www.peds.wustl.edu/genetics/>