

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency

What is Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency?

Medium chain acyl-CoA dehydrogenase (also known as MCAD) deficiency is an inherited condition that affects the way a person's body breaks down certain fats (medium chain fatty acids). A person with MCAD deficiency cannot breakdown medium chain fatty acids for energy. Consequently, the person's body begins to fail once the food eaten has been exhausted. In addition, the medium chain fatty acids then build up in the liver and brain. In the case of MCAD deficiency the inability to break down fats for energy and the build up of fats leads to mental retardation, other serious health problems and even death.

About one baby in 12,000 in the United States is born with MCAD deficiency. The condition occurs in all ethnic groups, but it is most common in individuals of Northern European ancestry.

How Does MCAD Deficiency Affect a Child?

Without treatment, children with MCAD deficiency experience episodes of low blood sugar, lack of energy, vomiting, and seizures. The episodes can become serious enough to lead to permanent mental retardation, liver and heart damage, coma, and even death.

What Causes MCAD Deficiency?

MCAD deficiency is a genetic condition caused by a change in the ACADM (Acyl-Coenzyme A Dehydrogenase) gene. The ACADM gene is responsible for making an enzyme called medium chain acyl CoA dehydrogenase, which is responsible for breaking down medium chain fatty acids into energy. When there is an alteration in the ACADM gene, medium chain acyl CoA dehydrogenase levels go down and medium chain fatty acids build up in the liver and brain.

MCAD deficiency is inherited in an autosomal recessive pattern, which means two copies of the ACADM gene must be changed for a person to be affected with MCAD deficiency. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "carriers," with one copy of the changed gene and one copy of the normal gene.

When both parents are carriers, there is a one-in-four (or 25%) chance that both will pass the changed ACADM gene on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal ACADM gene, and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed ACADM gene from one parent and a normal ACADM gene from the other, making that child a carrier like the parents. These chances are the same in each pregnancy with the same parents.

Is There a Test for MCAD Deficiency?

Yes. Babies are tested through newborn screening for MCAD deficiency before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has more than a normal amount of C8 (octanoylcarnitine, an acylcarnitine that helps with the break down of medium chain fatty acids). Some states screen newborns for MCAD deficiency, and Tennessee is one of them.

Can MCAD Deficiency Symptoms Be Prevented?

Yes. In most cases, not skipping any meals and eating frequently can prevent the symptoms of MCAD deficiency. If food cannot be tolerated (for instance when someone has a virus, cold, flu, etc.), the use of IV glucose (sugar) may be needed. Also, a specialized diet with replacement carnitine may be prescribed. Children and adults with MCAD deficiency require follow-up care at a medical center or clinic with an experienced dietician or nutritionist who specializes in this condition.

DISCLAIMER: The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

Resources:



FOD (Fatty Oxidation Disorder) Family Support Group
805 Montrose Drive
Greensboro, NC 27410
Phone: 336-547-8682
Email: deb@fodsupport.org
www.fodsupport.org



MUMS National Parent-to-Parent Network
Julie J. Gordon
150 Custer Court
Green Bay, Wisconsin 54301-1243
Phone: 1-877-336-5333 (Parents only please)
Phone: 1-920-336-5333
Fax: 1-920-339-0995
E-mail: mums@netnet.net
www.netnet.net/mums/



United Mitochondrial Disease Foundation
8085 Saltsburg Road, Suite 201
Pittsburgh, PA 15239
Phone: 412-793-8077
Fax: 412-793-6477
Email: info@umdf.org
www.umdf.org

References:

- American Academy of Pediatrics (1996): Newborn Screening Fact Sheets (RE9632). Pediatrics 98:473-501.
<http://aappolicy.aappublications.org/cgi/reprint/pediatrics;98/3/473.pdf>
- GeneTests (Medium Chain Acyl-Coenzyme A Dehydrogenase Deficiency)
<http://www.genetests.org>
- Online Mendelian Inheritance in Man (OMIM topic 607008)
<http://www.ncbi.nlm.nih.gov/Omim>

- Scriver, C.R. and Kaufman, S (2001) Mitochondrial Fatty Acid Oxidation Disorders. In: Scriver, C.R., Kaufman, S., Eisensmith, E., Woo S.L.C., Vogelstein, B. Childs, B. (eds) The Metabolic and Molecular Bases of Inherited Disease, 8th ed. McGraw-Hill, New York, Ch.101 pg. 2311-2315.