

Multiple acyl-CoA Dehydrogenase Deficiency (MAD, Glutaric acidemia type 2)

A fatty acid oxidation disorder

What is it?

Multiple acyl-CoA Dehydrogenase Deficiency (also known as MAD and GA-2) is an inherited fatty acid oxidation disorder. Patients with MAD cannot efficiently breakdown fats and protein into 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 MAD must eat on a very regular basis and should not go long without food.

What are the symptoms?

People with MAD may present a few different ways. Newborns may have abnormal features, such as wide spaced eyes and low set ears, genital abnormalities, rocker-bottom feet. They may also have low muscle tone, low blood sugar, large livers, and have the smell of sweaty feet. These babies have a high risk of death. On the other hand, people with MAD may not have symptoms until later in life. These symptoms may be low blood sugar, liver disease, and neurological problems. Many symptoms of late onset MAD can be prevented by immediate treatment and lifelong management. People with MAD typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

MAD is inherited in an autosomal recessive manner. This means that for a person to be affected with MAD, he or she must have inherited two non-working copies of the gene responsible for causing MAD. 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 MAD. Typically, there is no family history of MAD in an affected person. Although MAD is not a rare fatty acid oxidation disorder, the total number of people affected with MAD is not known.

How is it detected?

MAD 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?

MAD is treated by eating a diet low in fat and protein, frequent eating 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://ghr.nlm.nih.gov/ghr/page/Home>

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

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

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

STAR-G Hawaii Department of Health

<http://www.newbornscreening.info/Parents/fattyaciddisorders/GA2.html>