

Multiple Carboxylase Deficiency (MCD, holocarboxylase synthetase)

Organic acid disorder

What is it?

Multiple Carboxylase deficiency (MCD) is an inherited organic acid disorder. People with this organic acid disorder cannot properly break down certain components of protein and sometimes fats. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down protein, certain organic acids build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

What are the symptoms?

A person with this disorder can appear normal at birth. People with this disorder need to receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

The symptoms of MCD include food refusal, vomiting, skin rashes, breathing problems, hypotonia, seizures and lethargy. Severe metabolic/lactic acidosis, organic aciduria, mild hyperammonemia and variable hypoglycemia can lead to coma and death if not treated.

Inheritance and frequency

This disorder is inherited in an autosomal recessive manner. This means that for a person to be affected with MCD he or she must have inherited two non-working copies of the gene responsible for causing MCD. 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 MCD. Typically, there is no family history of MCD in an affected person. About 1 in 87,000 babies born have MCD.

How is it detected?

These disorders can 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?

These disorders may be treated with a special diet that is low in protein and sometimes fat. A special medication may also be recommended. A special medication may also be recommended. A specifically tailored treatment regimen is typically provided by a metabolic genetics 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

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