

## **3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)**

Organic acid disorders

### ***What is it?***

3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC) is an inherited organic acid disorder. People with 3-MCC 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 one of these disorders can appear normal at birth. People with these disorders need to receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

The symptoms of 3-MCC can be very variable between people. Newborns may present with vomiting, diarrhea, failure to thrive, and seizures. Some people with 3-MCC may have low blood sugar and liver problems while others may have no symptoms at all. Many symptoms of 3-MCC can be prevented by immediate treatment and lifelong management.

### ***Inheritance and frequency***

These disorders are inherited in an autosomal recessive manner. This means that for a person to be affected with 3-MCC he or she must have inherited two non-working copies of the gene responsible for causing 3-MCC. 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 3-MCC. Typically, there is no family history of 3-MCC in an affected person. About 1 in 50,000 babies born have 3-MCC. The mother may also be tested as well because several cases of maternal 3-MCC deficiency have been identified following an abnormal newborn screening result in their offspring.

### ***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>

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