

Methylmalonic Acidemia (Cbl A,B; vitamin B12 disorders)

An organic acid disorder

What is it?

Methylmalonic acidemia (Cbl A,B; vitamin B12 disorders) represents a family of organic disorders of the metabolism of branched-chain amino acids. Methylmalonic Acidemia (Cbl A,B) is an inherited organic acid disorder. People with organic acid disorders CblA,B, cannot properly break down certain components of protein and fats. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the proteins and fats, 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 Cbl A,B can appear normal at birth. The symptoms of Cbl A,B can be very variable between people. Some people with CblA,B will have the following symptoms after a few days of life: poor feeding, lack of energy, vomiting, low muscle tone, seizures, and trouble breathing. Kidney failure may develop. People with Cbl A,B may also present with the following symptoms later in infancy: failure to thrive, developmental delay and seizures. People with CblA,B may have no symptoms at all. Many symptoms of CblA,B can be prevented by immediate treatment and lifelong management. People with CblA,B typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

CblA,B is inherited in an autosomal recessive manner. This means that for a person to be affected with CblA,B, he or she must have inherited two non-working copies of the gene responsible for causing CblA,B. 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 CblA,B. Typically, there is no family history of CblA,B in an affected person. About 1 in 50,000 babies born have CblA,B.

How is it detected?

CblA,B 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?

CblA,B is treated by eating a diet low in protein and drinking a special formula, 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>

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