

Parent Fact Sheet

Methylmalonic Aciduria (MMA) (meth-ill-mal-ON-ic a-sid-EE-mia) **Mutase Deficiency (MUT)** (Mu-tase De-fi-shen-see)

What is MMA and MUT ?

Methylmalonic aciduria (MMA) is a “parent” metabolic disorder that can be diagnosed and named differently depending on which enzyme imbalance your child has. These conditions occur when the body is not able to completely break down protein. When the body cannot break down protein, a build-up of acids occurs in the blood. This build up of acids causes the child to become sick. Your child’s diagnosis will depend on the enzyme affected. The most common types of MMA are **MUT^o**, **MUT⁻**, **cbIA**, and **cbIB**.

What type of problems occurs with MMA?

A baby who has this condition may have a poor appetite, muscle tone weakness, vomiting, larger than normal size liver, and problems breathing. Common colds or eating too much protein can make your child sicker. If untreated, this condition can lead to delays in growth and a delay in reaching normal milestones. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have MMA?

This condition occurs in less than 1 in every 100,000 births. MMA occurs more often in certain populations, such as the Amish and people of Swedish descent. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

What is the treatment of MMA?

The treatment of this condition is usually a low protein diet. A special medical formula and supplements of vitamin B12 and carnitine may also be used. Your baby’s metabolic doctor will help you make sure that your baby gets the right diet and medical care.

Where in Virginia can I take my baby for care?

Please speak to your baby’s pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is <http://www.vahealth.org/gns>.



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