TEXAS Department of State Health Services

Newborn Screening FACT Sheet

Methylmalonic Acidemia Mutase Deficiency (methylmalonyl-CoA mutase deficiency) (MUT)

What is Methylmalonic Acidemia Mutase Deficiency?
Methylmalonic acidemia (MMA) is a condition with many different forms, all of which have different causes and treatments. MMA caused by methylmalonyl-CoA mutase deficiency is just one type of MMA.

What Causes MMA Mutase Deficiency?
When we eat food, enzymes help break it down. Some enzymes break down proteins into their building blocks, amino acids. Other enzymes break down fats into their building blocks, fatty acids. More enzymes break down these amino acids and fatty acids.

In MMA, the enzyme methylmalonyl-CoA mutase is not working correctly. This enzyme helps break down odd-chain fatty acids and the amino acids isoleucine, valine, methionine, and threonine. If your baby is affected with MMA mutase deficiency, then his or her body is either not making enough or making non-working methylmalonyl-CoA mutase enzymes.

In "MMA mutase deficiency 0" forms of MMA mutase deficiency, this enzyme is completely deficient. That means that there are no working methylmalonyl-CoA mutase enzymes in the body. In the "MMA mutase deficiency+" forms, some methylmalonyl-CoA mutase enzymes work correctly, but there are not enough. Without enough working enzymes, your baby's body has trouble using fats and proteins for energy.

MMA mutase deficiency is an autosomal recessive genetic condition. This means that a child must inherit two copies of the non-working gene for MMA mutase deficiency, one from each parent, in order to have the condition. The parents of a child with an autosomal recessive condition each carry one copy of the non-working gene, but they typically do not show signs and symptoms of the condition. While having a child with MMA mutase deficiency is rare, when both parents are carriers, they can have more than one child with the condition.

What Symptoms or Problems Occur with MMA Mutase Deficiency?
[Symptoms are something out of the ordinary that a parent notices.]

Early Signs -Signs of MMA mutase deficiency can start at any time from birth to adulthood. In most cases, the signs begin during infancy (either in the first few days or in the first few months of life). For babies, signs of MMA mutase deficiency can include:

- sleeping longer or more often
- tiredness
- vomiting
- weak muscle tone (also called hypotonia)
- fever
- breathing trouble
- frequent illnesses and infections
- increased bleeding and bruising

Many of these signs may occur when your baby eats foods that his or her body cannot break down. They can be triggered by long periods of time without eating, illnesses, and infections.

If your baby shows any of these signs, be sure to contact your baby's doctor immediately.

What is the Treatment for MMA Mutase Deficiency?
There are two types of MMA: cobalamin disorders and MMA mutase deficiency. Cobalamin disorders are considered vitamin B_{12} responsive. MMA mutase deficiency deficiencies are non-vitamin B_{12} responsive. You may hear about other babies with MMA receiving vitamin B_{12} injections. This treatment will not help a baby with MMA mutase deficiency.

Supplements and Medication - Your baby's doctor might recommend L-carnitine supplements. These supplements help the body break down fats, and they can remove harmful substances from the body. Your baby's doctor will need to write a prescription for these supplements.

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Dietary Treatments - Your baby will need a very carefully monitored diet. Children with MMA need to avoid certain fats and proteins because their bodies cannot break down these substances, causing a buildup of toxic substances. Your doctor can recommend special formulas and foods made for children with organic acid conditions. These formulas will likely need to be continued through adulthood.

It is also important for your baby to eat frequently. Long periods of time without food, illness, or infections may trigger many of the signs mentioned in the previous section.

Things to Remember

Even minor illness can lead to a Metabolic Crisis in children with MMA mutase deficiency**.** Call your doctor right away when your child has any of the following:

- loss of appetite
- vomiting
- diarrhea
- infection or illness
- fever

Children need extra fluids and starchy food (such as bread, rice, cereal, noodles) when they're sick in order to prevent a Metabolic Crisis. During illness, you should limit protein and give your child starchy foods and fluids. Sick children with MMA mutase deficiency may need to be treated in the hospital to avoid serious health problems.

Work with your baby's doctor to determine the next steps for your baby's care. Your baby's doctor may help you coordinate care with a physician who specializes in metabolism, a dietician who can help plan your child's specialized diet, or other medical resources in your community. Some children with methylmalonic acidemia have developmental delays. If you think that your baby is not meeting his or her developmental milestones, ask your baby's health care provider about the next steps in requesting a developmental evaluation and care.