TEXAS Department of State Health Services

Newborn Screening FACT Sheet

Methylmalonic Acidemia (MMA) Cobalamin A, B Type (Cbl A, B)

What is Methylmalonic Acidemia Cobalamin A, B Type (Cbl A, B)?
Methylmalonic acidemia (MMA) is a condition with many different forms, which all have different causes and treatments. Methylmalonic acidemia caused by cobalamin disorders A and B (Cbl A, B) is just one type of MMA.

MMA is a condition in which the body is unable to break down certain fats and proteins. It is considered an organic acid condition because it can lead to a harmful amount of organic acids and toxins in the body. MMA caused by cobalamin A or cobalamin B deficiencies is one type of MMA. Children with this form of the condition have trouble producing cobalamin enzymes A and B. Cobalamin enzymes are necessary for the body to break down certain foods.

What Causes MMA Cbl A, B?
When we eat food, enzymes help to break it down. Some enzymes break down proteins into their building blocks, amino acids. Other enzymes break down these amino acids. In Cbl A, B, the enzymes "cobalamin A" and "cobalamin B" are not working correctly. Cbl A, B enzymes help break down amino acids. Children with Cbl A, B either do not make enough or make non-working Cbl A, B enzymes. When these enzymes do not work, their bodies cannot break down the amino acids isoleucine, valine, methionine, and threonine. This causes a build-up of harmful substances in the body.

Cbl A, B is an autosomal recessive genetic condition. This means that a child must inherit two copies of the non-working gene for Cbl A, B, 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 Cbl A, B is rare, when both parents are carriers, they can have more than one child with the condition.

What Symptoms or Problems Occur with MMA Cbl A, B Type?
[Symptoms are something out of the ordinary that a parent notices.]

Early Signs
The signs of MMA Cbl A, B can begin at any time from birth to adulthood. In most cases, signs first appear during infancy (as early as the first few days after birth). For babies, signs of Cbl A, B can include:

- sleeping longer or more often
- 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 Cbl A, B Type?
Supplements and Medications - There are two types of MMA: cobalamin disorders and MUT deficiencies. One of the ways these two types of MMA differ is their response to vitamin B_{12}. MUT deficiencies are considered non-vitamin B_{12} responsive.

Cobalamin deficiencies are vitamin B_{12} responsive. In these cases, vitamin B_{12} injections can prevent symptoms. This type of treatment is more successful for cobalamin A disorders than for cobalamin B disorders, but it is helpful for both.

Your baby's doctor might also 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 Treatment – Some forms of Cobalamin A and B may not require dietary management. Other children may need a very carefully monitored diet. Children with MMA need to avoid certain fats and proteins because their bodies cannot break down these substances. Your baby's doctor can recommend special formulas made for babies 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 without food, illnesses, and infection may trigger many of the signs mentioned in the Early Signs section.

Things to Remember
Even minor illness can lead to a Metabolic Crisis in children with MMA Cbl A, B Type. 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 Cbl A, B 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 will 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 MMA have developmental delays. If you think that your baby is not meeting his or her developmental milestones, ask your baby's doctor about the next steps in requesting a developmental evaluation and care.