



## Newborn Screening FACT Sheet

### **Methylmalonic Acidemia (MMA) with Homocystinuria (Cbl C, D, E, F, G, and J)**

#### **What is Methylmalonic Acidemia (MMA) with homocystinuria (Cbl C, D, E, F, G, and J)?**

MMA is a condition in which the body is unable to process certain fats and proteins. It is considered an organic acid condition because it can lead to a harmful excess of certain toxins and organic acids. MMA with homocystinuria (Cbl C, D, E, F, G, and J) is one type of MMA. Individuals with this form of MMA have trouble producing certain cobalamin enzymes, which causes harmful levels of homocysteine and methylmalonic acid to build up in their bodies.

#### **What Causes Cbl C, D, E, F, G, or J?**

When we eat food, enzymes help break it down. Certain enzymes break down proteins into their building blocks, amino acids. Other enzymes break down these amino acids.

Some enzymes need help from vitamins like vitamin B<sub>12</sub> (also called cobalamin). The enzymes that break down the amino acids isoleucine, valine, methionine, and threonine need vitamin B<sub>12</sub> to work correctly.

If your baby has Cbl C, D, E, F, G, or J, his or her body cannot use vitamin B<sub>12</sub> correctly. Your baby's body either does not make enough or makes non-working enzymes that are supposed to turn vitamin B<sub>12</sub> from food into a form that the body can use.

Cbl C, D, E, F, G, and J is an autosomal recessive genetic condition. This means that a child must inherit two copies of the non-working gene for Cbl C, D, E, F, G, or J 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 C, D, E, F, G, or J is rare, when both parents are carriers, they can have more than one child with the condition.

#### **What Symptoms or Problems Occur with Cbl C, D, E, F, G, or J** [*Symptoms are something out of the ordinary that a parent notices.*]

Signs of Cbl C, D, E, F, G, or J could begin anywhere between the first few days of life and 14 years of age. Children with Cbl C usually show symptoms between the first few days and the first month of life.

Children with Cbl D deficiency do not show signs until later in childhood. If your baby has Cbl C, D, E, F, G, or J you might notice signs including:

- delayed growth
- small head size
- skin rash
- vomiting
- poor appetite
- diarrhea
- fever
- sleeping longer or more often
- tiredness
- weak muscle tone (called hypotonia)

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 health care provider immediately.

#### **What is the Treatment for Cbl C, D, E, F, G, or J?**

**Dietary Treatment** - Your baby will probably need to be on a restricted diet to avoid proteins that his or her body cannot break down. A dietician or nutritionist can help you plan a low-protein diet that still gives your baby the right nutrients for healthy growth.

Your baby's doctor might recommend special formulas or foods especially for children with Cbl C, D, E, F, G, or J. These formulas will likely need to continue through adulthood.

Eating often will also help prevent your baby from experiencing many of the signs mentioned in the previous section. Illnesses and infections can also trigger these signs.

**Supplements and Medications** - Supplements can also help treat Cbl C, D, E, F, G, or J. Vitamin B<sub>12</sub> can help reduce the signs and symptoms of the condition in some children. Your baby's doctor may need to try this treatment for a short period of time in order to determine if it is an effective treatment for your baby. Talk to your baby's doctor before starting vitamin B<sub>12</sub> treatment.

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L-carnitine is another substance that helps get rid of harmful waste products in the body. Some babies do not need this supplement, but your baby's body might not be making enough carnitine naturally. Your baby's doctor can tell if your baby needs these supplements and write an appropriate prescription.

Betaine supplements can help lower homocysteine levels in your baby's blood. Your baby's doctor can write a prescription for these supplements.

#### **Things to Remember**

Even minor illness can lead to a Metabolic Crisis in children with Cbl C, D, E, F, G, or J. 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 Cbl C, D, E, F, G, or J 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 dietitian who can help plan your child's specialized diet, or other medical resources in your community. Some children Cbl C, D, E, F, G, or J 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.