



Newborn Screening FACT Sheet

2Methylbutyryl-CoA dehydrogenase deficiency (2MBG)

What is 2MBG?

The disorder 2 methylbutyrylglycinuria (2MBG) is a condition in which the body is unable to break down certain proteins. It is classified as an organic acid condition because 2MBG can lead to a harmful amount of organic acids and toxins in the body. Symptoms of the condition vary. If 2MBG is identified early and treatment is begun, individuals with 2MBG can often lead healthy lives.

What Causes 2MBG?

When we eat food, enzymes help break it down. Some enzymes break down proteins into their building blocks, called amino acids. Other enzymes further break down the amino acids. In 2MBG, the enzyme 2-methylbutyryl-CoA dehydrogenase is not working correctly. This enzyme's job is to break down the amino acid isoleucine for energy. If your baby has 2MBG, your baby's body does not make enough or makes non-working 2-methylbutyryl-CoA dehydrogenase. When this happens, your baby's body cannot break down isoleucine, which causes harmful organic acids to build up in your baby's body.

What Symptoms or Problems Occur with 2MBG?

[Symptoms are something out of the ordinary that a parent notices.]

There have been very few reported cases of 2MBG. But of those reported, all of the babies were healthy at birth. Signs of 2MBG often begin in infancy, sometimes as early as a few days after birth. In other cases, signs do not develop until childhood.

- poor appetite
- sleeping longer or more often
- tiredness
- irritability
- vomiting
- fever
- weak muscle tone (also called hypotonia)
- delayed growth
- tight muscles (called spasticity)
- developmental delays

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.

What is the Treatment for 2MBG?

When 2MBG is diagnosed early, treatment can help prevent signs and symptoms. In fact, some children will never need treatment.

1. Dietary Treatment - The best way to prevent the symptoms of 2MBG is to have a very restricted diet for your child. A dietician or nutritionist can help you plan a low protein diet that still gives your baby the nutrients he or she needs for healthy growth. Your baby's doctor might recommend special baby formulas and foods designed for children with 2MBG. These formulas will likely need to be continued through adulthood. In addition, your baby will need to eat often to help prevent your baby from experiencing many of the signs mentioned in the Signs and Symptoms section. Illnesses and infections can also trigger these signs.

2. Medications - Your baby's doctor might prescribe L-carnitine supplements. L-carnitine is a natural substance, but your baby's body might not produce enough of it. Taking these supplements can help get rid of harmful substances in your baby's body. Your baby's doctor will need to write you a prescription for these supplements.

Things to Remember

If 2MBG is treated early, your baby can have healthy growth and development. It is important to get screened and start treatment early. If 2MBG is not treated, children could develop breathing problems, seizures, coma, or brain damage.