



Newborn Screening FACT Sheet

Elevated C4 Isobutyrylglycinuria (IBG)

What is IBG?

Isobutyrylglycinuria (IBG) is a condition in which the body is unable to break down certain proteins. It is classified as an organic acid condition because IBG can lead to harmful amounts of organic acids and other toxins in the body. Symptoms of the condition vary. If IBG is identified early and treatment is begun, individuals with the condition may be able to lead healthy lives.

What Causes IBG?

When we eat food, enzymes help break it down. Some enzymes break down proteins into their building blocks, called amino acids. Other enzymes help break down those amino acids. In IBG, the enzyme isobutyryl-CoA dehydrogenase is not working correctly. This enzyme's job is to help break down the amino acid valine. Babies with IBG either do not make enough or make non-working isobutyryl-CoA dehydrogenase. When this enzyme does not work correctly, the body cannot break down valine. If your baby's body cannot break down valine, high acid levels build up in the body. Everyone has some acid in his or her blood, but high levels can be toxic.

What Symptoms or Problems Occur with IBG?

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

IBG is a very rare condition. A child with IBG had signs that began at age one. Signs of IBG include:

- Delayed growth
- Sleeping longer or more often
- Tiredness
- Pale skin
- Trouble regulating body temperature (your baby may get cold easily)
- Difficulty breathing

Many of these signs may occur when your baby eats food 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 IBG?

1. Dietary Treatment - Your baby will probably need to follow a restricted diet in order to avoid eating proteins that his or her body cannot break down. A nutritionist or a dietician can help plan your baby's diet. Your baby's doctor might also recommend special formulas and foods for children with IBG. These formulas will likely need to continue through adulthood.

Your baby will also need to eat often in order to avoid many of the signs mentioned in the Symptoms and Problems section.

2. Supplements and Medications - Your baby may need to take prescription L-carnitine supplements. L-carnitine is a substance that is naturally produced in the body, but your baby's body might not make enough of it. These supplements help to break down fats and get rid of harmful waste substances that build up in the body. Your baby's doctor will need to write a prescription for these supplements.

L-carnitine supplements can also help prevent or reverse some of the more severe health complications associated with IBG mentioned above.

Things to remember:

We do not yet know how effective treatments for isobutyrylglycinuria (IBG) are. With early and ongoing treatment, your child will likely have healthy growth and development. This is why newborn screening is so important.

Children who do not receive treatment for IBG are at risk for more severe health complications, including a low red blood cell count (called anemia) and an enlarged, weak heart (known as cardiomyopathy).

L-carnitine treatments can help prevent, and even reverse, anemia and heart trouble that may develop.