



Newborn Screening FACT Sheet

Beta Ketothiolase Deficiency (BKT/BKD)

What is BKT?

Beta-ketothiolase deficiency (BKT) is a condition in which the body is unable to break down certain proteins. People with the condition are also unable to produce ketone bodies, substances that help the body store energy. BKT is considered an organic acid condition because it can lead to a dangerous amount of organic acids and toxins in the body. Early detection and treatment can often prevent the severe outcomes of this condition.

What Causes BKT?

Enzymes help start chemical reactions in the body. BKT happens when an enzyme called “mitochondrial acetoacetyl-CoA thiolase” is missing or not working. This enzyme helps break down the amino acid isoleucine. All foods with protein contain isoleucine. Harmful matter can build up in the blood and cause problems when someone with BKT eats protein.

What Symptoms or Problems Occur with BKT?

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

BKT can cause periods of illness called Metabolic Crises. Some of the first symptoms of a Metabolic Crisis are:

- too much sleepiness or lack of energy
- fever / vomiting / diarrhea
- poor appetite
- ketones in the urine

Later problems can include:

- intellectual disabilities
- dystonia (muscle spasms)
- enlarged heart
- low blood platelets
- higher risk for infections
- not growing well
- kidney problems

What is the Treatment for BKT?

The treatments often recommended for babies and children with BKT are:

1. Medication – The doctor may prescribe L-carnitine for your child. This is safe and natural and helps the body make energy. It also helps the body get rid of harmful wastes.

2. Do not go a long time without food – Some babies and young children can have a Metabolic Crisis if they don’t eat often enough. Your doctor will tell you how to space your child’s meals. Most healthy children over the age of 10 can go without food for up to 12 hours without problems.

3. Low-protein diet – Some children will need to eat a low-protein diet. High-protein foods to be limited include:

- milk and milk products
- meat and poultry
- fish
- eggs
- dried beans
- nuts/peanut butter

Children with BKT need some protein to grow properly. A dietitian can make a food plan for your child. Dietitians know the right foods for your child to eat.

Things to Remember

Even minor illness such as a cold or flu can lead to a Metabolic Crisis. Call your doctor right away when your child has any of the following:

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

An ill child with BKT needs more liquids, starchy foods (such as rice, cereal, bread), and sugars to avoid a Metabolic Crisis. When your child is ill, you will also need to limit protein.

Children with symptoms of a Metabolic Crisis need to be treated in a hospital.