

Beta-Ketothiolase Deficiency (BKT) (Bay-ta-key-toe-thigh-o-lace De-fi-shen-see)

What is BKT?

Beta-Ketothiolase Deficiency (BKT) is a metabolic disorder. This means the body has a chemical imbalance. BKT is a condition in which the body cannot properly process a particular amino acid (a building block of proteins) or the products of fat breakdown. When this happens, toxic chemicals build up in the blood. BKT is inherited. This means it is present at birth.

What type of problems occurs with BKT?

A baby who has this condition may develop vomiting, trouble breathing, and extreme tiredness. If untreated, this condition can lead to coma. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have BKT?

This condition occurs in less than 1 in every 100,000 births. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition.
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

What is the treatment of BKT?

The treatment of this condition might be a low protein diet and to avoid long periods without food. If your child becomes ill, your child might need to get medical care quickly. Intravenous (IV) fluids and medication might be needed. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care.

Where in Virginia can I take my baby for care?

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The web site is <http://www.vahealth.org/gns>.

