

Beta-Ketothiolase (BKT) Deficiency

General Overview

Q. What is BKT deficiency?

A. BKT deficiency is a treatable disorder that affects the way the body processes protein and fats. It is treatable, but can cause life-threatening illness.

Q. What happens to protein and fats in a child with BKT deficiency?

A. Children with BKT deficiency cannot properly use the amino acid isoleucine found in the protein they eat, and they cannot properly break down fats. If left untreated, byproducts of isoleucine and fat metabolism, called ketones, build up in the bloodstream, urine, and body tissue, and cause life-threatening illness and brain damage.

Q. What is the treatment for BKT deficiency?

A. To minimize the risk of brain damage, treatment must begin shortly after birth, and is life-long. BKT deficiency is treated with a special low-protein diet that limits isoleucine intake and avoids fasting. Some doctors may also prescribe dietary supplements. Emergency care must be taken if a person with BKT deficiency becomes ill and has difficulty keeping food down. This is usually treated in the hospital with an IV. People with BKT deficiency require specialized treatment through a specialty clinic with experience in treating this disorder.

Q. What are the effects of having BKT deficiency if it is not treated?

A. Untreated BKT deficiency can result in brain damage, loss of muscle function, coma or death.

Q. Why would a child have BKT deficiency?

A. BKT deficiency is an inherited disorder. It results when a baby receives a double-dose of a specific non-working gene involved in breaking down isoleucine (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. How common is BKT deficiency?

A. The prevalence of BKT deficiency is unknown, but very rare. However, parents who have a child with BKT deficiency each carry one copy of the non-working gene. That means, with each pregnancy, there is a one in four chance of the child having BKT deficiency.

For more information about BKT deficiency, please see the Disorders section of our website:
www.doh.wa.gov/nbs.

