

Beta Ketothiolase (BKT, mitochondrial acetoacetyl-CoA thiolase, short-chain ketoacylthiolase)

Organic acid disorder

What is it?

Beta Ketothiolase (BKT) is an inherited organic acid disorder. People with an organic acid disorder cannot properly break down certain components of protein and sometimes fats. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down protein, certain organic acids build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

What are the symptoms?

A person with this disorder can appear normal at birth. People with this disorder need to receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

The symptoms of BKT include intermittent episodes of severe metabolic acidosis and ketosis accompanied by vomiting, diarrhea and coma that may progress to death. Death or neurological complications can occur. Other symptoms include cardiomyopathy, poor weight gain, renal failure and short stature.

Inheritance and frequency

This disorder is inherited in an autosomal recessive manner. This means that for a person to be affected with BKT he or she must have inherited two non-working copies of the gene responsible for causing BKT. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non-working copies of their gene, causing the baby to have BKT. Typically, there is no family history of BKT in an affected person. BKT is a rare organic acid disorder; the frequency of BKT in the general population is unknown.

How is it detected?

These disorders can be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

These disorders may be treated with a special diet that is low in protein and sometimes fat. A special medication may also be recommended. A special medication may also be recommended. A specifically tailored treatment regimen is typically provided by a metabolic genetics professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: <http://www.ghr.nlm.nih.gov>

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