

Beta-Ketothiolase Deficiency (BKT)

Parent Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An “abnormal” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is **very important** to determine if your baby has the disorder indicated.

Disorder Indicated: Beta-Ketothiolase Deficiency (BKT) is a condition in which the body cannot break down certain proteins or produce ketone bodies (store energy) due to an inherited enzyme deficiency. This results in an accumulation of organic acids in the body. If left untreated, BKT could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals with BKT can have healthy growth and development.

Incidence	Rare, estimated 1 in every 1,000,000 newborns.
Signs and Symptoms <i>Please note: these findings may not be present in young infants or in milder forms of the disease</i>	When a child has Beta-Ketothiolase Deficiency, you may see symptoms including: <ul style="list-style-type: none"> • Poor appetite • Sleeping longer or more often • Tiredness • Vomiting • Breathing difficulty <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i></p>
Next Steps may include:	Follow up with your child’s pediatric provider Clinical assessment Laboratory Testing
Treatment (if indicated)	Restricted diet (follow up with your child’s pediatric provider)
Additional Resources	VDH Newborn Screening http://vdhlivewell.com/newbornscreening Baby’s First Test www.babysfirsttest.org Genetics Home Reference https://ghr.nlm.nih.gov/ Organic Acidemia Association http://www.oaanews.org/

Educational content adapted from www.babysfirsttest.org

