

Beta-Ketothiolase Deficiency (BKT)

Health Care Professional Fact Sheet

<p>A newborn screening test is a screen and not diagnostic testing. An “abnormal” or “critical” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is vital to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.</p>	
<p>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.</p>	
Incidence	Rare, estimated 1 in every 1,000,000 newborns.
Analyte Measured	<p>Primary Marker: C5:1 (<i>tiglylcarnitine</i>)</p> <p>Secondary Marker*: C5-OH (<i>3-hydroxy-isovaleryl carnitine</i>)</p> <p><i>*an abnormal secondary marker paired with an abnormal primary marker will result in critical group and require referral and diagnostic testing</i></p>
Normal Test Results	C5:1 < 0.10 µmol/L
Abnormal Test Results	C5:1 ≥ 0.10 µmol/L to < 1.0 µmol/L
Critical Test Results	<p>C5:1 ≥ 1.0 µmol/L</p> <p>(Critical results require immediate evaluation and follow-up)</p>
Signs and Symptoms	<p>When a child has Beta-Ketothiolase Deficiency, you may see symptoms including:</p> <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>
Signs and Symptoms	<p><i>Please note: these findings may not be present in young infants or in milder forms of the disease</i></p>
Next Steps may include:	<p>Discuss the next steps of evaluation and possible treatment with the regional Geneticist</p> <p>Provide parental education (see accompanying sheet)</p> <p>Clinical Assessment</p> <p>Assay: Urine Organic Acids, Plasma Acylcarnitine</p>
Treatment (if indicated)	Restricted diet (Discuss with the regional Geneticist)
Additional Resources	<p>VDH Newborn Screening http://vdhlivewell.com/newbornscreening</p> <p>Baby’s First Test www.babysfirsttest.org</p> <p>American College of Medical Genetics (ACMG) ACT Sheets www.ACMG.net</p> <p>Genetics Home Reference https://ghr.nlm.nih.gov/</p> <p>Organic Acidemia Association http://www.oaanews.org/</p>

Educational content adapted from www.babysfirsttest.org

