

Elevated Organic Acid: C5-OH 3-hydroxy-isovalerylcarnitine

Parent Fact Sheet

<p>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.</p>	
<p>Disorder Indicated: Hydroxymethylglutaric Aciduria (HMG), 3-Methylcrotonyl-CoA carboxylase Deficiency (3-MCC), and Beta-Ketothiolase Deficiency (BKT) are conditions in which the body cannot break down certain proteins due to an inherited enzyme deficiency. Individuals with HMG also cannot produce ketone bodies. This results in an accumulation of organic acids in the body. If left untreated, it could cause brain damage or death. However, if the disorders are detected early and treatment is begun, individuals with these conditions can have healthy growth and development.</p>	
<p>Incidence</p>	<p>3-MCC: 1 in every 35,000 to 50,000 newborns. BKT: Rare, estimated 1 in every 1,000,000 newborns HMG: Extremely rare- Less than 100 cases worldwide</p>
<p>Signs and Symptoms</p> <p><i>Please note: these findings may not be present in young infants or in milder forms of the disease</i></p>	<p>When a child has HMG or 3-MCC, you may see symptoms including:</p> <ul style="list-style-type: none"> • Poor appetite/ Vomiting/ Low blood sugar (hypoglycemia) • Sleeping longer or more often/Tiredness • Irritability/ Behavior changes • Weak muscle tone (hypotonia) • Muscle tightness (spasticity) • Fever • Diarrhea <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i></p>
<p>Next Steps may include:</p>	<p>Follow up with your child’s pediatric provider</p> <p>Clinical assessment Laboratory Testing</p>
<p>Treatment (if indicated)</p>	<p>Restricted diet (follow up with your child’s pediatric provider)</p>
<p>Additional Resources</p>	<p>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/</p>

Educational content adapted from www.babysfirsttest.org

