

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

### Health Care Professional Fact Sheet

<p>A newborn screening test is a <b>screen</b> 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 <b>vital</b> 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><b>Disorder Indicated:</b> 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>	
Incidence	<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>
Analyte Measured	Organic Acid: C5-OH (3-hydroxy-isovalerylcarnitine)
Normal Test Results	C5-OH < 1.0 µmol/L
Abnormal Test Results	C5-OH ≥ 1.0 µmol/L to < 2.0 µmol/L
Critical Test Results	<p>C5-OH ≥ 2.0 µmol/L  <b>(Critical results require immediate evaluation and follow-up)</b></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 elevated C5-OH, you may see symptoms including:</p> <ul style="list-style-type: none"> <li>• Poor appetite/ Vomiting/ Low blood sugar (hypoglycemia)</li> <li>• Sleeping longer or more often/Tiredness</li> <li>• Irritability/ Behavior changes</li> <li>• Weak muscle tone (hypotonia)</li> <li>• Muscle tightness (spasticity)</li> <li>• Fever</li> <li>• Diarrhea</li> </ul> <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i></p>
Next Steps <i>may</i> include:	<p><b>Discuss the next steps of evaluation and possible treatment with the regional metabolic consultant</b></p> <p>Provide parental education (see accompanying sheet)</p> <p>Clinical Assessment</p> <p>Assay: Urine Organic Acids, Plasma Acylcarnitine</p>
Treatment <i>(if indicated)</i>	Restricted diet (Discuss with the regional metabolic consultant)
Additional Resources	<p>VDH Newborn Screening <a href="http://vdhlivewell.com/newbornscreening">http://vdhlivewell.com/newbornscreening</a></p> <p>Baby’s First Test <a href="http://www.babysfirsttest.org">www.babysfirsttest.org</a></p> <p>American College of Medical Genetics (ACMG) ACT Sheets <a href="http://www.ACMG.net">www.ACMG.net</a></p> <p>Genetics Home Reference <a href="https://ghr.nlm.nih.gov/">https://ghr.nlm.nih.gov/</a></p> <p>Organic Acid Association <a href="http://www.oaanews.org/">http://www.oaanews.org/</a></p>

Educational content adapted from [www.babysfirsttest.org](http://www.babysfirsttest.org)

