

Elevated Fatty Acid: C16-OH 3-hydroxypalmitoylcarnitine

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: Long-chain L-3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) and Trifunctional Protein Deficiency (TFP) are fatty acid oxidation disorders that could be identified through the elevation of the fatty acid C16-OH. These disorders are conditions in which the body does not have sufficient enzymes to break down long chain fatty acids due to an inherited enzyme deficiency. Fatty acids are an important energy source for the body. If left untreated, these disorders could cause developmental delays, cardiac and liver issues, coma, or even death. However, if the conditions are detected early and treatment is begun, individuals can have healthy growth and development.

Incidence	Unknown
<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 a fatty acid oxidation disorder, you may see symptoms including:</p> <ul style="list-style-type: none"> • Poor appetite • Tiredness/Sleeping longer or more often • Vomiting/Diarrhea • Hypoglycemia (low blood sugar) • Behavior changes/irritability • Breathing difficulty • Seizures <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/ Fatty Oxidation Disorder Family Support Group http://www.fodsupport.org/</p>

Educational content adapted from www.babysfirsttest.org

