VDH VIRGINIA DEPARTMENT OF HEALTH

Division of Child and Family Health- Newborn Screening Follow-Up Program

Elevated Fatty Acid: C16-OH *3-hydroxypalmitoylcarnitine*

Health Care Professional Fact Sheet

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.

Disorder(s) 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
Analyte Measured	**Primary Marker**: C16-OH *3-hydroxypalmitoylcarnitine* **Secondary Markers***: C16 *palmitoylcarnitine,* C18:1OH *3-hydroxyoleoylcarnitine* ** an abnormal secondary marker paired with an abnormal primary marker will result in critical group and require referral and diagnostic testing*
Normal Test Results	C16-OH < 0.10 µmol/L
Abnormal Test Results	C16-OH ≥ 0.10 µmol/L to < 0.19 µmol/L
Critical Test Results	C16-OH ≥ 0.19 µmol/L ***(Critical results require immediate evaluation and follow-up)***
Signs and Symptoms *Please note: these findings may not be present in young infants or in milder forms of the disease*	When a child has a fatty acid oxidation disorder, you may see symptoms including: • Poor appetite • Tiredness/Sleeping longer or more often • Vomiting/Diarrhea • Hypoglycemia (low blood sugar) • Behavior changes/irritability • Breathing difficulty • Seizures *Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections*
Next Steps ***may*** include:	**Discuss the next steps of evaluation and *possible* treatment with the regional Geneticist** Provide parental education (see accompanying sheet) Clinical Assessment Assay: Plasma Acylcarnitines, Urine Organic Acids
Treatment **(*if indicated*)**	Restricted diet/supplements (Discuss with the regional Geneticist)
Additional Resources	VDH Newborn Screening *http://vdhlivewell.com/newbornscreening* Baby's First Test *www.babysfirsttest.org* American College of Medical Genetics (ACMG) ACT Sheets *www.ACMG.net* Genetics Home Reference *https://ghr.nlm.nih.gov/* Fatty Oxidation Disorder Family Support Group *http://www.fodsupport.org/*

Educational content adapted from www.babysfirsttest.org

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