Suggested Follow-up for Elevated C16OH 3-OH Palmitoyl Carnitine

Possible Causes: Elevated C16OH is the primary marker for **long chain 3-OH acyl co-A dehydrogenase deficiency (LCHAD) and Trifunctional Protein Deficiency (TFP).** LCHAD and TFP are defects in long chain fatty acid oxidation.

Next Steps if Abnormal: **Potential medical emergency.** See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies include plasma total and free carnitines, plasma acylcarnitines and urine organic acids. In addition, repeat acyl carnitine profile on filter paper and send to the DHEC laboratory.

Neonatal Presentation: Hypoketotic hypoglycemia, cardiomyopathy, myopathy, hepatic encephalopathy. Infants are at risk for metabolic decompensation/crisis.

Emergency Treatment: Treatment of metabolic crisis includes provision of sufficient calories (concentrated dextrose infusion with appropriate electrolytes) to correct catabolic state and biochemical abnormalities if needed.

Standard Treatment: Avoid fasting. Feed every four hours through the night for first several months. Fat restricted diet with use of MCT oil as fat source. May need cornstarch supplementation at bedtime to maintain blood glucose levels overnight. Carnitine supplementation if helpful.

Advice for Family: Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated C16OH)*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

NOTE: HELLP syndrome (hemolysis, elevated liver enzymes, low platelets)/AFLP (acute fatty liver of pregnancy) occurs in 20% of pregnancies where the fetus is affected by LCHAD. TFP is thought to be a less common cause of HELLP syndrome.

Internet Resources:

http://oregon.gov/DHS/ph/nbs/expand.shtml

http://web1.tch.harvard.edu/newenglandconsortium/scientists_physicians2.html

http://ghr.nlm.nih.gov/condition=longchain3hydroxyacylcoenzymeadehydrogenasedeficiency

http://ghr.nlm.nih.gov/condition=mitochondrialtrifunctionalproteindeficiency

http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm