

## Suggested Follow-up for Elevated Free Carnitine/C16 Palmitoyl Carnitine + C18 Steraroyl Carnitine

**Possible Causes:** When the ratio of free carnitine to the sum of C16 and C18 is elevated the possible diagnosis includes **Carnitine Palmitoyltransferase I Deficiency (CPT I)**. This is a defect in conversion of long chain fatty acyl co-A molecules to their corresponding acylcarnitine molecules. Deficiency of this enzyme reduces the availability of acylcarnitines for transport into the mitochondrial matrix for 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, and plasma acylcarnitines. In addition, repeat acyl carnitine profile on filter paper and send to the DHEC laboratory.

**Neonatal Presentation:** Usually none. May have hypoketotic hypoglycemia, seizures and hepatomegaly. 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 once diagnosis is clearly established. **Carnitine is contraindicated in treatment of CPT I.**

**Advice for Family:** Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated Free Carnitine to C16 + C18)*, 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) can occur in pregnancies where the fetus is affected by CPT I.

**Internet Resources:**

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

[http://web1.tch.harvard.edu/newenglandconsortium/scientists\\_physicians2.html](http://web1.tch.harvard.edu/newenglandconsortium/scientists_physicians2.html)

<http://www.genetests.org/query?dz=cpt1a>

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