Suggested Follow-up for Elevated C16 Palmitoyl Carnitine and
Elevated C18:1 Oleyl Carnitine

Possible Causes: When both C16 and C18:1 are elevated the possible diagnosis includes **Carnitine Palmitoyltransferase II Deficiency (CPT II) and Carnitine/Acylcarnitine Translocase Deficiency (CACT).** Both are defects in fatty acid and carnitine transport.

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: CPT II—Usually none. May have hypoketotic hypoglycemia, cardiomyopathy and renal dysgenesis. Infants are at risk for metabolic decompensation/crisis.

CACT—May have cardiomyopathy, hypotonia, hypoketotic hypoglycemia, hyperammonemia, liver dysfunction. 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.

Advice for Family: Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated C16 with Elevated 18:1),* may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet Resources:

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

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

http://www.genetests.org/query?dz=cpt2

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

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