Suggested Follow-up for Carnitine Uptake/Transport Deficiency
Low Free Carnitine

Possible Causes: When free carnitine is low the possible diagnosis is **carnitine uptake/transport deficiency**. In carnitine uptake/transport deficiency, carnitine transport across the plasma membrane is inhibited. The reduction in carnitine limits the formation of acylcarnitine and subsequently limits energy production. Skeletal and heart muscle tissues are particularly affected in this process.

Next Steps if Abnormal: **Potential medical emergency when the free carnitine is low and the sum of C3 + C16 is less than 2.** 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: Tachycardia, hepatomegaly, reduced muscle tone, poor feeding

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: Carnitine supplementation. Avoid fasting. Feed every four hours through the night for first several months.

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

NOTE: In some newborns, the low free carnitine is reflective of maternal CUD.

Internet Resources:

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

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

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