Suggested Follow-up for Elevated C8: Octanoyl Carnitine

Possible Causes: Elevated C8 is the primary marker for **medium chain acyl co-A dehydrogenase deficiency (MCAD)** and **medium chain ketoacyl co-A thiolase deficiency (MCKAT).** Both are disorders of fatty acid oxidation. MCKAT has only been described in one infant worldwide.

Next Steps if Abnormal: 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: MCAD—Usually none. MCKAT—Vomiting, dehydration, metabolic acidosis, liver dysfunction

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. Carnitine supplementation if helpful. **Infants with MCAD should not be fed formulas that have medium chain triglycerides (MCT) as a fat source.**

Advice for Family: Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated C8),* 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=mcad

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