Suggested Follow-up for Elevated C4 (Butyryl Carnitine)
with Elevated C5 (Isovaleryl Carnitine)

Possible Causes: When both the C4 and the C5 are elevated, the possible cause is **glutaric aciduria type II (GA II).** This disorder is also known as multiple acyl co-A dehydrogenase deficiency. This is a defect in multiple dehydrogenases involved in amino acid catabolism and fatty acid oxidation. Other acyl carnitines may also be elevated.

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: Poor feeding, vomiting, lethargy, hypotonia, seizures, acidosis, hypoglycemia. Some have dysmorphic features and congenital anomalies, particularly cystic kidneys. 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. Riboflavin may be helpful. May be prescribed fat restricted, protein controlled diet. Carnitine supplementation.

Advice for Family: Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated C4 and C5)*, 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://ghr.nlm.nih.gov/condition=glutaricacidemiatypeii

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