Suggested Follow-up for Elevated C5: Isovaleryl/2-Methybutyryl Carnitine

Possible Causes: Elevated C5 is the primary marker for **isovaleric acidemia (IVA)**, a disorder of leucine (LEU) metabolism. It is also elevated in **2-methylbutyryl co-A dehydrogenase deficiency (2-MBCD)**, a very rare disorder of isoleucine (ILE) metabolism.

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, tachypnea, lethargy, hypoglycemia, metabolic acidosis, ketosis, hyperammonemia. 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: IVA—Protein restricted diet with use of metabolic formula without LEU. Glycine (GLY) supplementation. Carnitine supplementation. Avoid fasting.

2-MBCD—Carnitine supplementation. Moderate protein restriction. Avoid fasting.

Advice for Family: Provide basic information about organic acid disorders. The handout, *When Baby Needs a Second Test for an Organic Acid Disorder (Elevated 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=isovalericacidemia

http://ghr.nlm.nih.gov/condition=2methylbutyrylcoenzymeadehydrogenasedeficiency

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