Suggested Follow-up for Elevated C3DC: Malonyl Carnitine

Possible Causes: Elevated C3DC is the primary marker for **malonic acidemia (MA).** Malonic acidemia is a disorder of ketone metabolism arising from a deficiency of the enzyme malonyl coA decarboxylase.

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: May have hypotonia, hypoglycemia, hypertrophic cardiomyopathy, diarrhea, vomiting, ketosis and/or seizures. Infants are at risk for metabolic decompensation/crisis.

Emergency Treatment: Treatment of metabolic crisis includes provision of sufficient nonprotein calories (concentrated dextrose infusion with appropriate electrolytes) to correct catabolic state and biochemical abnormalities if needed.

Standard Treatment: Carnitine supplementation. May be prescribed fat controlled diet with MCT as major fat source.

Advice for Family: Provide basic information about organic acid disorders. The handout, *When Baby Needs a Second Test for an Organic Acid Disorder (Elevated C3DC)*, 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://ghr.nlm.nih.gov/condition=malonylcoenzymeadecarboxylasedeficiency

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