

## Suggested Follow-up for Elevated C4OH: 3-OH Butyryl Carnitine

**Possible Causes:** Elevated C4OH is the primary marker for **medium and short chain acyl co-A dehydrogenase deficiency (M/SCHAD)**. M/SCHAD is caused by deficiency of 3-OH acyl coA dehydrogenase and is a defect in fatty acid oxidation.

**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. Infants are at risk for metabolic decompensation/crisis, hypoglycemia. Plasma insulin may be elevated.

**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. May need cornstarch supplementation at bedtime to maintain blood glucose levels overnight. Carnitine supplementation if helpful. Consider medication for infants with documented hyperinsulinism.

**Advice for Family:** Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated C4OH)*, 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://ghr.nlm.nih.gov/condition=3hydroxyacylcoenzymeadehydrogenasedeficiency>

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