Suggested Follow-up for Elevated C10:2

Possible Causes: Elevated 10:2 is the primary marker for dienoyl co-A reductase deficiency**.** This disorder is a very rare 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 could 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: Very little is known about this disorder. One case in an African-American infant has been described. Infant had hypotonia, small ventricular septal defect, short extremities and microcephaly; however, the relationship of phenotype to this disorder is not known.

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

Standard Treatment: Proposed treatment includes avoidance of fasting, reduction of long chain fat in the diet and supplementation with MCT and perhaps carnitine.

Advice for Family: Provide basic information about fatty acid disorders. The handout, *When Baby Needs a Second Test for a Fatty Acid Disorder (Elevated C10:2)*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet Resources:

No internet resources were found. This information sheet was based upon information obtained from *The Metabolic and Molecular Bases of Inherited Disease, 8th Edition.*