Suggested Follow-up for Biotinidase Deficiency

Possible Causes: Autosomal recessive disorder resulting in lack of adequate production of the enzyme biotinidase. Infants who have untreated biotinidase deficiency may develop hypotonia, seizures, developmental delay, ataxia, breathing problems, hair loss and hearing loss.

Next Steps if Abnormal: Repeat biotinidase screening on filter paper and send to the DHEC laboratory. No treatment necessary until result of repeat testing known. If biotinidase is still deficient in repeat specimen, refer to metabolic specialist. Further evaluation/testing is necessary to clarify diagnosis.

Neonatal Presentation: Usually none.

Emergency Treatment: None.

Standard Treatment: Daily biotin supplements for life.

Advice for Family: Provide basic information about biotinidase deficiency. The handout, *When Baby Needs a Second Test for Biotinidase Deficiency*, may be used for this purpose.

Internet Resources:

http://www.biotinidasedeficiency.20m.com/

http://www.genetests.org/query?dz=biotin

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