Suggested Follow-up for Biotinidase Deficiency

Possible Causes:

Biotinidase is a key enzyme in the biotin cycle. Biotinidase releases biotin from dietary proteins and recycles biotin. Biotinidase deficiency is an autosomal recessive disorder, which results in a lack of adequate production of biotinidase.

Infants who have untreated biotinidase deficiency may develop hypotonia, seizures, ataxia, developmental delay, breathing problems, hair loss, and hearing loss. Screening may also identify infants with partial biotinidase deficiency or carrier status.

Estimated Inheritance: 1:60,000

Abnormal 1st tier screen: Deficient Biotinidase (BIO)

Method of Notification: All abnormal 1st and 2nd tier screening results are called, faxed, and/or sent to the provider of record.

Next Steps if Abnormal:

See the infant as soon as possible to ascertain health status. When quantity allows, a portion of the initial specimen will be sent to the Greenwood Genetic Center (GGC) Laboratory for secondary testing.

If biotinidase is abnormal on the 2nd tier test, consult with a pediatric metabolic specialist. Initiate treatment and diagnostic evaluation as recommended. Report all findings to the SC Newborn Screening Program.

Neonatal Presentation: Usually none. However, some affected infants may have symptoms as early as one week of age.

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.

Special Considerations: Transfusion of red blood cells prior to drawing the newborn screening specimen may affect the biotinidase result. Repeat screening for biotinidase should be done 120 days after the last transfusion for accurate assessment.

Internet Resources:

http://ghr.nlm.nih.gov/condition/biotinidase-deficiency

https://www.babysfirsttest.org/newborn-screening/conditions/biotinidase-deficiency

https://www.acmg.net/PDFLibrary/Biotinidase-Deficiency-ACT-Sheet.pdf