Biotinidase Deficiency (BIOT)

Biotinidase (BIOT) is a key enzyme in the biotin cycle. Biotinidase releases biotin from dietary proteins and recycles biotin. Free biotin is necessary for activation of four carboxylase enzymes. Carboxylases are important enzymes in the metabolism of amino acids, gluconeogenesis, and in the synthesis of fatty acids.

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

Inheritance:	Autosomal recessive
Estimated Inheritance:	1:60,000
Abnormal Screen Result:	**Deficient Biotinidase**
Method of Notification:	All abnormal test results are called and faxed to provider of record.
Next Steps if Abnormal:	**See infant as soon as possible to ascertain health status and repeat biotinidase on filter paper. If biotinidase is still deficient on repeat testing, contact a pediatric metabolic specialist for further instructions**. Initiate treatment and diagnostic evaluation as recommended by specialist. **Report all findings to state newborn screening program**.
Neonatal Presentation:	Usually none. However, some affected infants have symptoms as early as one week of age.
Treatment:	Daily biotin supplements for life.

Special Considerations

Transfusion - 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. If the date of the last transfusion is unknown, put the date of hospital discharge on the collection form next to "**Transfused**".