

PHYSICIAN ACT SHEET for POSITIVE Newborn Screening Result for BIOTINIDASE DEFICIENCY

Disease Category: Inborn error of metabolism of biotin/holocarboxylase

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- **Contact family to inform them of the newborn screening result and to schedule an immediate office visit.**
- **Schedule a lab visit for the newborn within 24-48 hours for confirmatory testing.**
- **Consult a pediatric metabolic specialist or geneticist.** Refer to a metabolic center if considered appropriate.
- **Report findings** back to state newborn screening program.

Meaning of the Screening Result: Decreased biotinidase enzyme activity suggests biotinidase deficiency. This could mean the newborn has complete deficiency, partial deficiency, is a carrier or that the specimen was denatured due to heat exposure at some point during specimen handling.

- Enzyme activity of ≤ 8.0 ERU strongly suspected of complete deficiency.
- Enzyme activity $> 8.1 - 16$ ERU may be complete deficiency but are more often suggestive of partial deficiency or carrier status.

Confirmation of Diagnosis: For screening results < 8.0 ERU, collect by venipuncture 2-3 cc. of blood in a red top tube. Serum must be separated as soon as possible, and should be frozen and shipped overnight on dry ice in a sealed, leak-proof container labeled as biohazardous material. The specimen **MUST BE KEPT FROZEN**. A control specimen from a **NON-RELATED** person should also be collected and handled in the same way. The Pediatric Metabolic Specialists prefer confirmatory testing be completed at the Biochemical Genetics Laboratory in Baltimore, MD. (see following instructions for more information)

Condition Description: This autosomal recessive genetic condition is caused by an enzyme defect that impairs the breakdown of biotin to be used and recycled for normal metabolism.

Clinical Expectations: Biotinidase deficiency has variable presentation. The newborn may be asymptomatic. However, the clinical phenotype can progress to include metabolic acidosis, which can result in coma and death. Other symptoms may include ataxia, seizures, hearing loss, alopecia, developmental delay and skin rash.