

Physician Action Sheet for Abnormal Newborn Screen – Biotinidase Deficiency

Newborn Screen Findings: Deficient or decreased levels of biotinidase on newborn screen. *Because environmental factors (such as heat) can affect screening results, the Indiana University Newborn Screening (NBS) Lab may request a repeat newborn screen before requesting the next step in confirmatory testing.*

The Indiana University NBS Lab should have already contacted your office & the infant's birthing facility about the abnormal results & recommended confirmatory testing. The IU NBS Lab has also notified the Metabolic Specialist. *Please note that the IU NBS Lab does not contact the patient's family.*

Required Actions

1. **Contact the Metabolic Specialist (317) 274-3966 (ask for Newborn Screening Nurse or Genetic Counselor) to discuss abnormal results, follow-up recommendations & confirmatory testing.**
2. **Contact the family as soon as possible.**
 - a. Inform the family of the abnormal NBS result & the need for additional confirmatory testing.
 - b. Determine the condition of the infant. *See the infant in your office immediately if there are concerns of poor feeding, hypotonia or lethargy. These symptoms require immediate evaluation & treatment.*
 - c. Review the list of symptoms that require immediate contact to your office (see above).
3. **Arrange for confirmatory laboratory testing at the infant's birthing facility.**
4. **Plan for follow-up with the Metabolic Specialist if biotinidase deficiency is confirmed.**

Clinical Summary: Autosomal recessive disorder caused by reduced activity of the biotinidase enzyme. Deficiency of biotinidase results in abnormal biotin recycling & subsequent multiple carboxylase deficiencies. If untreated, symptoms of the disorder typically appear later in infancy & can include skin rashes, episodic hypoglycemia, metabolic acidosis, hypotonia, seizures, developmental delay, ataxia & progressive vision & hearing loss. Symptoms are irreversible if treatment is not initiated in a timely manner.

Treatment: Individuals with biotinidase deficiency are treated with oral biotin supplements in larger doses than the Recommended Dietary Allowance, under the direction of a Metabolic Specialist. Treatment is life-long & will require continued follow-up with Metabolic Specialist. Early diagnosis and prompt treatment will help ensure the best outcome for individuals with biotinidase deficiency.

Incidence: 1/76,000 in Indiana (1/60,000 nationally). Genetic testing available.

Contact Information:

- Regular business hours: (317) 274 – 3966; ask for the Newborn Screening Nurse or Genetic Counselor.
- Emergency night/weekend contact: (317) 944 – 5000; have the on-call Metabolism physician paged.

Resources:

- Newborn Screening ACT sheets – www.acmg.net (select "Resources," click on "ACT Sheets")
- Information for clinicians – www.ncbi.nlm.nih.gov/books/NBK1322