

Biotinidase Deficiency

Action required

Contact metabolic specialist immediately.

Issues to discuss with specialist

- Laboratory evaluation of infant
 - § Set up appointment in metabolic clinic
 - § Enzyme assay for biotinidase activity
- Oral biotin
 - § Before or after laboratory evaluation
 - § Before or after metabolic clinic visit

False positives

- Common
- Variants seen with partial biotinidase deficiency
- Screening result can be impacted by environmental factors, such as heat

Review with family

Family has **not** been notified of result by MDH.

After discussion with metabolic specialist, contact family to coordinate clinic visit, and referral to metabolic clinic. Expect infant to be stable when family is contacted and at clinic visit.

Prompt follow-up is important.

NICU issues

Positive newborn screens are seen more commonly in babies who spend time in the NICU than in healthy term infants.

Newborn screens cannot be accurately interpreted after transfusion.

Clinical summary

Biotinidase deficiency is an autosomal recessive disorder that results from the defective activity of the biotinidase enzyme. Newborns are asymptomatic. If an infant is not screened and/or left untreated, symptoms begin to appear later in infancy and can include seizures, developmental delay, facial rash, ataxia, and progressive vision and hearing loss.

Affected children require life-long treatment with oral biotin and monitoring by both primary care and specialty providers.

Incidence: ~ 1/50,000; affects all ethnic groups. High incidence in Minnesota Somali population.

Clinical expectations

If treated promptly, children with biotinidase deficiency can be asymptomatic and are expected to develop normally. Typically, neurologic problems only occur in children with biotinidase deficiency who had symptoms prior to biotin treatment.

Episodic hypoglycemia, lethargy, hypotonia, and mild developmental delay can occur even with treatment through childhood

Resources

GeneTests www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

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

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening