ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

ACT Sheet

Newborn Screening ACT Sheet

[Absent/ Reduced Biotinidase Activity]

Biotinidase Deficiency

Differential Diagnosis: Biotinidase deficiency (profound or partial).

Condition Description: Biotinidase deficiency is caused by decreased levels of the enzyme biotinidase, which is responsible for recycling the vitamin biotin. This deficiency leads to decreased biotin levels and disrupts the activity of several biotin-requiring enzymes (carboxylases). Presentation in the neonatal period is rare.

You Should Take the Following Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (typically asymptomatic; rarely poor feeding, lethargy, hypotonia).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, lethargy, hypotonia).
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide family with basic information about biotinidase deficiency and its management.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Serum biotinidase activity: Biotinidase activity is markedly reduced or absent in profound biotinindase deficiency, and partially reduced in partial biotinidase deficiency. Urine organic acids: may demonstrate increased 3-hydroxyisovaleric acid and 3-methylcrotonylglycine with profound deficiency. Molecular genetic testing may be required to confirm the diagnosis.

Clinical Considerations: The neonate is usually asymptomatic but episodic hypoglycemia, lethargy, hypotonia, can occur at any time from the neonatal period through childhood. Untreated profound biotinidase deficiency leads to developmental delay, seizures, alopecia, and hearing deficits. Patients with partial biotinidase deficiency are typically asymptomatic. Biotin treatment is highly effective.

Additional Information:

How to Communicate Newborn Screening Results
Gene Reviews
Medline Plus
Condition Information for Families- HRSA Newborn Screening Clearinghouse
ClinGen Actionability Report

Referral (local, state, regional, and national):

Find a Genetics Clinic Directory
Genetic Testing Registry

State and Other Resources

State Newborn Screening Program

Metabolic (Bloodspot) Screening, Montana's Newborn Screening Programs
406-444-0984, dphhs.mt.gov/ecfsd/cshs/newbornscreeningprograms/

Short-term Follow-up Coordinator - Crystal Fortune, MT Laboratory Services Bureau
406-444-0930 or 800-821-7284 cfortune@mt.gov

Genetics/Metabolics Consultants

Shodair Children's Hospital Medical Genetics Staff will contact you with recommendations.
Metabolic Clinic: 406-444-1099; Clinics held across Montana: 406-444-1016
shodair.org/about-shodair/contact/

If emergency consultation with a metabolic specialist is necessary, contact One Call (24/7) at Children's Hospital Colorado (800-525-4871) and ask to speak to the metabolic specialist on call.

Information for Clinicians and Families

Montana Medical Home Portal (see Newborn Disorders and Parents & Families sections)
mt.medicalhomeportal.org/newborn/biotinidase-deficiency

Parent/Family Support

Metabolic Support UK
www.metabolicsupportuk.org

National Resources (with web addresses)

Additional Information

How to Communicate Newborn Screening Results
www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf

Gene Reviews
www.ncbi.nlm.nih.gov/books/NBK1322/

Medline Plus
medlineplus.gov/genetics/condition/biotinidase-deficiency/

Condition Information for Families-HRSA Newborn Screening Clearinghouse
newbornscreening.hrsa.gov/conditions/biotinidase-deficiency

ClinGen Actionability Report

actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC098

Referral (local, state, regional and national)

Find a Genetics Clinic Directory
clinics.acmg.net

Genetic Testing Registry
www.ncbi.nlm.nih.gov/gtr/