

Newborn Screening ACT Sheet [Absent/ Reduced Biotinidase Activity] Biotinidase Deficiency

Differential Diagnosis: Biotinidase deficiency (complete and partial); see C5-OH acylcarnitine for non-biotinidase associated conditions.

Condition Description: A multiple carboxylase deficiency resulting from a reduction in available biotin secondary to deficient activity of the biotinidase enzyme.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status.
 - Evaluate infant if poor feeding, lethargy, or hypotonia are present.
 - Consultation/referral to a metabolic specialist to determine appropriate follow-up.
 - Undertake confirmatory testing in consultation with a metabolic specialist.
 - Emergency treatment if symptomatic.
 - Report findings to newborn screening program
-

Diagnostic Evaluation: Enzyme assay for biotinidase in serum or plasma reveals low activity. False positive findings are usually a processing/shipping problem. Urine organic acid analysis may show normal or increased 3-hydroxyisovaleric acid and 3-methylcrotonylglycine. Plasma acylcarnitine analysis may show normal or increased C5-OH acylcarnitine.

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

Additional Information:

[Gene Reviews](#)

[OMIM](#)

[Genetics Home Reference](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Reviews

<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=biotin>

OMIM

<http://www.ncbi.nlm.nih.gov/entrez/dispmim.cgi?id=253260>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=biotinidasedeficiency>

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/22179?db=genetests&country=United%20States

Clinical Services

<http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests>

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)