

Newborn Screening ACT Sheet

Niemann-Pick Disease Type A and B

Condition Description: Niemann-Pick disease types A and B are lysosomal storage disorders (LSD) caused by a defect in acid sphingomyelinase (ASM), resulting in accumulation of sphingomyelin. These are autosomal recessive disorders.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Consult with genetic metabolic specialist.
 - Contact family to inform them of the newborn screening result.
 - Examine the patient with particular attention to hepatosplenomegaly and neurologic findings.
 - Provide the family with basic information about Niemann-Pick disease.
 - Report confirmatory findings to newborn screening program.
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Diagnostic Evaluation: Confirmatory sphingomyelinase enzyme assay. If low, the patient should have sphingomyelin phosphodiesterase 1 (*SMPD1*) gene analysis. Gene analysis may allow for separation of Type A from Type B.

Clinical Considerations: Type A disease is characterized by neonatal onset, massive hepatosplenomegaly, pulmonary infiltration, neurodegeneration and early death. Type B is associated with variable age of onset, similar visceral manifestations, but no central nervous system involvement. Treatment in both types is supportive. Liver transplantation, hematopoietic stem cell transplantation, or enzyme replacement therapy (ERT) may be considered. ERT is highly complicated and should be given only under the guidance of a specialist with expertise in lysosomal storage disorders.

Additional Information:

[Genetics Home Reference](#)

OMIM

[Niemann-Pick Type A](#)

[Niemann-Pick Type B](#)

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.

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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:

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition/niemann-pick-disease>

OMIM

Niemann-Pick Type A

<http://www.ncbi.nlm.nih.gov/omim/257200>

Niemann-Pick Type B

<http://www.ncbi.nlm.nih.gov/omim/607616>

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/26084?db=genetests&country=United%20States

Clinical Services

<http://www.genetests.org>

Find Genetic Services

<http://www.acmg.net/gis>

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