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.

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

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

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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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