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

ACT Sheet

Newborn Screening ACT Sheet

[Decreased acid sphingomyelinase]

Acid Sphingomyelinase Deficiency (ASMD)

Differential Diagnosis: None.

Condition Description: Acid sphingomyelinase deficiency (ASMD), also known as Niemann-Pick Disease Types A/B, are lysosomal disorders caused by deficiency of acid sphingomyelinase (ASM), resulting in the accumulation of sphingomyelin. The infantile neurovisceral form (previously referred to as "Type A") presents in infancy with hepatosplenomegaly, interstitial lung damage, and psychomotor regression. The chronic visceral form (previously referred to as "Type B") most often presents in childhood with similar symptoms which are usually less severe.

You Should Take the Following Actions:

- Inform the family of newborn screening result.
- Ascertain clinical status (difficulty breathing, neurologic changes).
- Consult with pediatric metabolic specialist the same day.
- Evaluate the newborn (hepatosplenomegaly, pulmonary compromise, neurologic changes).
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide the family with basic information about ASMD, and its management.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Leukocyte acid sphingomyelinase enzyme assay: decreased ASM enzyme activity is suggestive of ASMD, but this result alone does not exclude pseudodeficiency, which causes decreased enzyme levels, but no disease. Plasma or dried blood spot lysosphingomyelin concentration: elevated lysosphingomyelin can confirm the diagnosis, but does not differentiate the neurovisceral form from the chronic visceral form. Molecular genetic testing can confirm the diagnosis and distinguish the two subtypes of ASMD.

Clinical Considerations: The infantile neurovisceral form of ASMD is characterized by neonatal onset with progressive hepatosplenomegaly, pulmonary disease, neurodegeneration, cherry-red spot of the retina, and death by 3 years of age. The chronic visceral form of ASMD is associated with variable age of onset, similar manifestations, and a less severe course. Treatment for both types is supportive.

Additional Information:

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

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

Find a Genetics Clinic Directory
Genetic Testing Registry

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Local Resources *(Insert Local Website Links)*

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

Additional Information

How to Communicate Newborn Screening Results

- https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf

Gene Reviews

- https://www.ncbi.nlm.nih.gov/books/NBK1370/

Medline Plus

- https://medlineplus.gov/genetics/condition/niemann-pick-disease/

Condition Information for Families-HRSA Newborn Screening Clearinghouse

- https://newbornscreening.hrsa.gov/conditions/niemann-pick-disease

Referral (local, state, regional and national)

Find a Genetics Clinic Directory

- https://clinics.acmg.net

Genetic Testing Registry

- https://www.ncbi.nlm.nih.gov/gtr/