

# 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

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource 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. 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 practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.



## ACT Sheet

## **Local Resources** (Insert Local Website Links)

State Resource Site (Insert Website Information)

	Name			
	URL			
	Comments			
Local Ro	esource Site (Insert V	Vebsite 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/

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