

Newborn Screening ACT Sheet

[Increased Arginine]

Argininemia

Differential Diagnosis: None.

Condition Description: Argininemia results from inherited defects in arginase, an enzyme in the urea cycle that helps convert ammonia to urea. Arginase deficiency leads to elevated plasma arginine and in some cases, hyperammonemia. Presentation in the neonatal period is rare.

You Should Take the Following Actions:

- Inform family of the newborn screening result the same day.
- Ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn for signs of poor feeding, lethargy, hypotonia, tachypnea, seizures, and signs of liver disease. If any sign is present or infant is ill, immediately transport to the hospital for emergency treatment of hyperammonemia in consultation with metabolic specialist.
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide the family with basic information about argininemia and its management.
- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: [Plasma amino acids](#): Arginine is usually significantly elevated in argininemia. [Urine organic acids or quantitative orotic acid](#) may demonstrate elevated orotic acid, which is more readily detected by the quantitative test. [Plasma ammonia](#) will reveal hyperammonemia when present. Additional [enzymatic or molecular genetic testing](#) may be required in cases where plasma arginine is mildly elevated.

Clinical Considerations: Neonates with argininemia are usually asymptomatic but can develop mild to moderate hyperammonemia once receiving dietary protein. Later signs include developmental delay, seizures, and lower extremity spasticity. Rarely, argininemia may cause severe neonatal illness as seen in the other urea cycle disorders.

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](#)

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/NBK1159/>

Medline Plus

- <https://medlineplus.gov/genetics/condition/arginase-deficiency/>

Condition Information for Families- HRSA Newborn Screening Clearinghouse

- <https://newbornscreening.hrsa.gov/conditions/arginase-deficiency>

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/>