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

ACT Sheet

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

State and Other Resources

State Newborn Screening Program

Nevada Newborn Screening Program, Nevada State Public Health Laboratory
775-682-6238, med.unr.edu/nsphl/newborn-screening

Genetics/Metabolic Consultants

Nicola Longo, MD, PhD, University of Utah Genetics/Pediatrics
801-585-2457, healthcare.utah.edu/fad/mddetail.php?physicianID=u0305101

Information for Clinicians and Families

Nevada Medical Home Portal (see Newborn Disorders and Parents & Families sections)
nv.medicalhomeportal.org/newborn/argininemia

Parent/Family Support

National Urea Cycle Disorders Foundation
www.nucdf.org/

National Resources (with web addresses)

Additional Information

How to Communicate Newborn Screening Results
www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf

Gene Reviews
www.ncbi.nlm.nih.gov/books/NBK1159/

Medline Plus
medlineplus.gov/genetics/condition/arginase-deficiency/

Condition Information for Families-HRSA Newborn Screening Clearinghouse
newbornscreening.hrsa.gov/conditions/arginase-deficiency

Referral (local, state, regional and national)

Find a Genetics Clinic Directory
clinics.acmg.net

Genetic Testing Registry
www.ncbi.nlm.nih.gov/gtr/