

## 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.
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- 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

Newborn Screening Program, Utah Department of Health and Human Services  
801-584-8256, [newbornscreening.health.utah.gov/](http://newbornscreening.health.utah.gov/)

### Genetics/Metabolic Consultants

Pediatric Medical Genetics, University of Utah Department of Pediatrics  
801-213-3599, [healthcare.utah.edu/pediatrics/programs-services/genetics.php](http://healthcare.utah.edu/pediatrics/programs-services/genetics.php)

### Information for Clinicians and Families

Utah Medical Home Portal (see Newborn Disorders and Parents & Families sections)  
[ut.medicalhomeportal.org/newborn/argininemia](http://ut.medicalhomeportal.org/newborn/argininemia)

### Parent/Family Support

National Urea Cycle Disorders Foundation  
[www.nucdf.org/](http://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](http://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/](http://www.ncbi.nlm.nih.gov/books/NBK1159/)

Medline Plus  
[medlineplus.gov/genetics/condition/arginase-deficiency/](http://medlineplus.gov/genetics/condition/arginase-deficiency/)

Condition Information for Families-HRSA Newborn Screening Clearinghouse  
[newbornscreening.hrsa.gov/conditions/arginase-deficiency](http://newbornscreening.hrsa.gov/conditions/arginase-deficiency)

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

Find a Genetics Clinic Directory  
[clinics.acmg.net](http://clinics.acmg.net)

Genetic Testing Registry  
[www.ncbi.nlm.nih.gov/gtr/](http://www.ncbi.nlm.nih.gov/gtr/)