

## 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](#)

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.

## State and Other Resources

### State Newborn Screening Program

Metabolic (Bloodspot) Screening, Montana's Newborn Screening Programs  
406-444-0984, [dphhs.mt.gov/ecfsd/cshs/newbornscreeningprograms/](http://dphhs.mt.gov/ecfsd/cshs/newbornscreeningprograms/)

Short-term Follow-up Coordinator - Crystal Fortune, MT Laboratory Services Bureau  
406-444-0930 or 800-821-7284 [cfortune@mt.gov](mailto:cfortune@mt.gov)

### Genetics/Metabolics Consultants

Shodair Children's Hospital Medical Genetics Staff will contact you with recommendations.  
Metabolic Clinic: 406-444-1099; Clinics held across Montana: 406-444-1016  
[shodair.org/about-shodair/contact/](http://shodair.org/about-shodair/contact/)

**If emergency consultation with a metabolic specialist is necessary**, contact One Call (24/7) at Children's Hospital Colorado (800-525-4871) and ask to speak to the metabolic specialist on call.

### Information for Clinicians and Families

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

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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/)