

**Follow-Up Management Protocol for
Newborns with Elevated Arginine Screening**
Newborn Screening Program of the Oklahoma State Department of Health

Differential Diagnosis: Argininemia

Evaluation & Initial Management Guidelines for Significantly Elevated Arginine Screen

1. Contact the family by close of business. Inform family of newborn screen result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea).
2. Immediately consult with the geneticist.
3. History and Physical Exam within 24 hours in consultation with the geneticist:
 - **Assess specifically for signs and symptoms of hyperammonemia:**
 - Poor Feeding
 - Vomiting
 - Lethargy
 - Tachypnea
 - Hypotonia
 - Seizures
 - Hyperammonemia
 - Signs of Liver Disease
 - Family history of urea cycle disorder (family history of SIDS or affected siblings, aunts, uncles etc.)
4. If symptomatic, immediate phone consultation with a geneticist regarding treatment and emergency clinical management of hyperammonemia is required.
5. If not symptomatic, consult with a geneticist for medical management, and schedule diagnostic work-up within 24 hours.

Description

The urea cycle disorders result from defects in the metabolism of the extra nitrogen produced by the breakdown of protein and other nitrogen-containing molecules. Infants with Argininemia appear normal at birth. Argininemia is usually asymptomatic in the neonatal period, although it can present with mild to moderate hyperammonemia. Untreated, argininemia usually progresses to severe spasticity, loss of ambulation, complete loss of bowel and bladder control, severe cognitive and intellectual disabilities and seizures.

Resources

- **ACMG Newborn Screening ACT Sheets:**
<https://www.ncbi.nlm.nih.gov/books/NBK55827/>
- **Integris Pediatric Specialty Clinic, Inborn Error of Metabolism (IEM) Clinic**
Geneticist pager: (405) 630-3794
- **OU Children's Physicians – Genetics Clinic**
Page Operator: (405) 271-3636
- **Newborn Screening Follow-Up Program**
(405) 271-6617 option 2 or (800) 766-2223; www.nsp.health.ok.gov