Emergency Management Protocol for
Newborns with Elevated Citrulline Screening
Urea Cycle Disorder
Newborn Screening Program of the Oklahoma State Department of Health

Differential Diagnosis: Citrullinemia type I, Argininosuccinic Aciduria; Citrullinemia Type II (citrin deficiency)

Evaluation & Initial Management Guidelines for Significantly Elevated Citrulline Screen

1. Contact the family within **one hour** of notification. 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 by COB 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.)
   - **Immediate** phone consultation with a geneticist regarding treatment and emergency clinical management of hyperammonemia is required.
4. Arrange immediate transportation to metabolic center for diagnostic work-up and medical management.

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. In citrullinemia and in argininoscuccinc academia (ASA), the accumulation of ammonia and other toxic metabolites occurs during the first few days of life. Infants with these disorders often appear normal initially but rapidly develop cerebral edema and the related signs of lethargy, anorexia, hyperventilation or hypoventilation, hypothermia, seizures, neurologic posturing, and coma. Urea cycle disorders **are life threatening**. Immediate intervention is warranted to prevent hyperammonemia and death.

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