

**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 metabolic specialist.
3. History and Physical Exam by COB in consultation with the metabolic specialist:
 - **Assess specifically for signs and symptoms of hyperammonemia:**

<ul style="list-style-type: none">▪ Poor Feeding▪ Vomiting▪ Lethargy▪ Tachypnea	<ul style="list-style-type: none">▪ Hypotonia▪ Seizures▪ Hyperammonemia▪ Signs of Liver Disease
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 - Family history of urea cycle disorder (family history of SIDS or affected siblings, aunts, uncles etc.)
 - **Immediate** phone consultation with a metabolic specialist 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 argininosuccinic 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

Mary Monks, RN, Metabolic Nurse Specialist, pager (405) 559-1378, phone (405) 271-8685.
Newborn Screening Follow-up Program (405) 271-6617 or 1-800-766-2223.

Metabolic Specialist in Oklahoma

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