TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening ACT Sheet

Increased Arginine

Amino Aciduria/Urea Cycle Disorder

Differential Diagnosis

Argininemia (ARG)

Condition Description

The urea cycle is the enzyme cycle whereby ammonia is converted to urea. In argininemia, defects in arginase, a urea cycle enzyme, may result in hyperammonemia.

Take the Following IMMEDIATE Actions

- **Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea);**
- **Immediate telephone consultation with pediatric metabolic specialist (See attached list.);**
- **Evaluate the newborn (poor feeding, vomiting, lethargy, hypotonia, tachypnea, seizures and signs of liver disease);**
- **If any sign is present or infant is ill, IMMEDIATELY initiate emergency treatment for hyperammonemia in consultation with metabolic specialist;**
- **Transport to hospital for further treatment in consultation with metabolic specialist;**
- **Initiate timely confirmatory/diagnostic testing and management, as recommended by specialist;**
- **Initial testing: immediate plasma ammonia, plasma quantitative amino acids, and urine orotic acid;**
- **Repeat newborn screen if second screen has not been done;**
- **Provide family with basic information about hyperammonemia; and**
- **Report findings to newborn screening program.**

Diagnostic Evaluation

Specific diagnosis is made by plasma quantitative amino acid analysis revealing increased arginine and urine orotic acid analysis revealing increased orotic acid, respectively. Blood ammonia determination may also reveal hyperammonemia.

Clinical Considerations

Argininemia is usually asymptomatic in the neonate although it can present with a mild-moderate hyperammonemia once the baby receives dietary protein. Later signs include intellectual disability, seizures and spastic diplegia if untreated. Rarely, argininemia may cause severe neonatal illness as seen in the other urea cycle disorders.

Additional Information

American Medical Genetics and Genomics – Argininemia
U.S. National Library of Medicine, Medline Plus – Arginase deficiency
STAR G FELSI – Argininemia
STAR G FELSI – Argininemia/ arginase deficiency
STAR G FELSI – Argininosuccinic Acid lyase Deficiency
STAR G FELSI – Argininosuccinyl-CoA lyase deficiency

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