Newborn Screening ACT Sheet
[Increased Arginine]
Amino Aciduria/Urea Cycle Disorder

Differential Diagnosis: Argininemia

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.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea).
- Immediate consult with pediatric metabolic specialists.
- Evaluate the newborn (poor feeding, 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.
- Provide family with basic information about hyperammonemia.
- Report findings to newborn screening program.

Diagnostic Evaluation: Specific diagnosis is made by plasma 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 developmental delay, seizures and spastic diplegia if untreated. Rarely, argininemia may cause severe neonatal illness as seen in the other urea cycle disorders.

Additional Information:
- Emergency Protocols (New England Consortium of Metabolic Programs)
- Gene Reviews
- Genetics Home Reference

Referral (local, state, regional and national):
- Testing
- Clinical Services
- Find Genetic Services
LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site  (insert state newborn screening program website information)

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Local Resource Site  (insert local and regional newborn screening website information)

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APPENDIX: Resources with Full URL Addresses

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Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. 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 guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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