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 mental retardation, 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

Additional Information:
Emergency Protocols (New England Consortium of Metabolic Programs)
http://newenglandconsortium.org/for-professionals/acute-illness-protocols/urea-cycle-disorders/arginase-deficiency/

Gene Reviews
http://www.ncbi.nlm.nih.gov/books/NBK1159/

Genetics Home Reference

Referral (local, state, regional and national):
Testing

Clinical Services

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx