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)*

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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
http://ghr.nlm.nih.gov/ghr/locuslink/383#summary

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

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/22236?db=genetests&country=United%20States

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests

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