Newborn Screening ACT Sheet
[Increased Tyrosine]
Tyrosinemia

Differential Diagnosis: Tyrosinemia I (hepatorenal); tyrosinemia II (oculocutaneous); tyrosinemia III; transient hypertyrosinemia; liver disease.

Condition Description: In the hepatorenal form, tyrosine (from ingested protein and phenylalanine metabolism) cannot be metabolized by fumarylacetoacetate hydrolase to fumaric acid and acetoacetic acid. The resulting fumarylacetoacetate accumulates and is converted to succinylacetone, the diagnostic metabolite, which is liver toxic and leads to elevated tyrosine. Tyrosinemias II and III are due to other defects in tyrosine degradation.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result.
- Consult with pediatric metabolic specialist.
- Evaluate the newborn and refer as appropriate.
- Initiate confirmatory/diagnostic tests in consultation with metabolic specialist.
- Provide family with basic information about tyrosinemia.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma amino acid analysis will show increased tyrosine in all of the tyrosinemias. Urine organic acid analysis may reveal increased succinylacetone in tyrosinemia I.

Clinical Considerations: Tyrosinemia I is usually asymptomatic in the neonate. If untreated, it will cause liver disease and cirrhosis early in infancy. Nitisinone (NTBC) treatment will usually prevent these features. Tyrosinemia II is asymptomatic in the neonate but will cause hyperkeratosis of the skin, corneal ulcers, and in some cases, mental retardation unless treated with a tyrosine restricted diet. Tyrosinemia III may be benign.

Additional Information:
Gene Reviews (Tyrosinemia I)
Genetics Home Reference

Referral (local, state, regional and national):
Testing
- Tyrosinemia I
- Tyrosinemia II
- Tyrosinemia III

Clinical Services
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

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:

Gene Reviews (Tyrosinemia I)
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=tyrosinemia

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=tyrosinemia

Referral (local, state, regional and national):

Testing

Tyrosinemia I
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2286?db=genetests

Tyrosinemia II
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/68759?db=genetests

Tyrosinemia III
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/56600?db=genetests

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

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

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