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

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.

Take the Following IMMEDIATE Actions

- **Contact family to inform them of the newborn screening result;**
- **Consult with pediatric metabolic specialist (See attached list.);**
- **Evaluate the newborn and refer as appropriate;**
- **Initiate confirmatory/diagnostic tests in consultation with metabolic specialist;**
- ***Initial testing: plasma quantitative amino acids, urine organic acids, urine succinylacetone, and liver function tests;***
- **Repeat newborn screen if the second screen has not been done;**
- **Provide family with basic information about tyrosinemia; and**
- **Report findings to newborn screening program.**

Diagnostic Evaluation

Plasma quantitative 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, intellectual disability unless treated with a tyrosine restricted diet. Tyrosinemia III may be benign.

Additional Information

American College of Genetics and Genomics – Tyrosinemia ACT Sheet
U.S. National Library of Medicine, Medline Plus – Tyrosinemia
STAR G FELSI – Tyrosinemia
STAR G FELSI – Tyrosinemia, type 1

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