



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.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE 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.

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:

Diagnostic Evaluation:

Plasma amino acid analysis will show increased tyrosine in all of the tyrosinemias. Urine organic acid analysis will 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. Nitisnone (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.

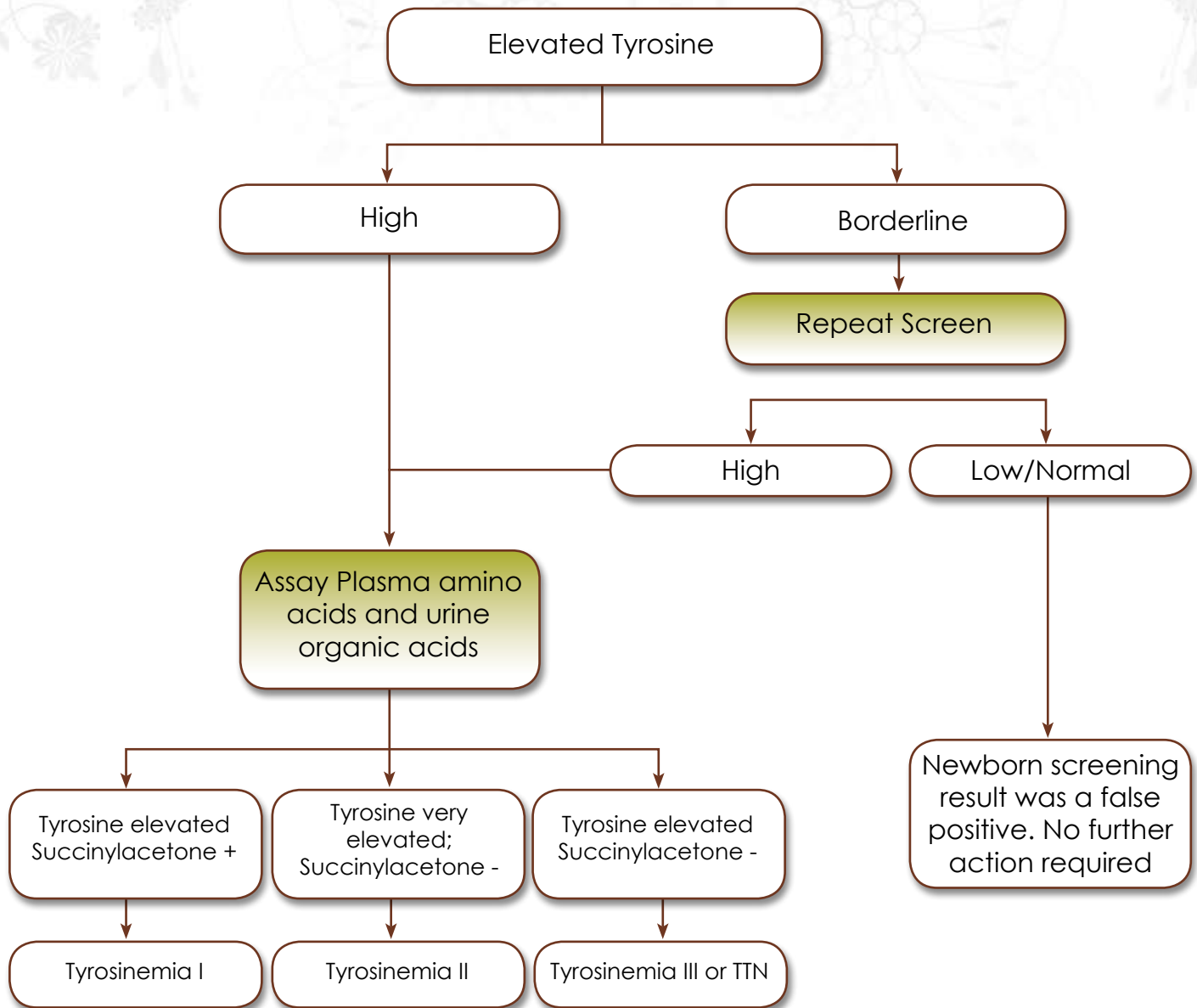
Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen.



NEWBORN SCREENING

Screening Today for a Safer Tomorrow

Tyrosine Elevated



Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations / Key:

TTN = Transient tyrosinemia of the neonate

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