KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: INCREASED SUCCINYLACETONE (SUAC)
CONDITION: TYROSINEMIA (TYR I)

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

METABOLIC 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 hepatotoxic and leads to elevated tyrosine. Tyrosinemia II and III are due to other defects in the tyrosine degradation.

ACTION TO BE TAKEN **IMMEDIATELY:**

- 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.

CONFIRMATION OF DIAGNOSIS: Plasma amino acid analysis will show increased **tyrosine** in all of the Tyrosinemias. Urine organic acid analysis will reveal increased **succinylacetone** in tyrosinemia I only.

CLINICAL EXPECTATIONS: 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.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

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