



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: INCREASED LEUCINE

CONDITION: MAPLE SYRUP URINE DISEASE (MSUD)

DIFFERENTIAL DIAGNOSIS: Maple syrup urine disease (MSUD); hydroxyprolinemia.

METABOLIC DESCRIPTION: In MSUD, leucine, isoleucine, and valine (branched chain amino acids) can not be metabolized further than their α -ketoacid derivatives. The amino acids and organic acids accumulate and produce severe toxicity.

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

- ◆ Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea).
- ◆ Consult with pediatric metabolic specialist.
- ◆ Evaluate the newborn (poor feeding, lethargy, tachypnea, alternating hypertonia/hypotonia, seizures). If any sign is present or infant is ill, transport to hospital for further treatment in consultation with metabolic specialist.
- ◆ Initiate timely confirmatory/diagnostic testing and management, as recommended by specialist.
- ◆ Provide the family with basic information about MSUD and dietary management.
- ◆ Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: In MSUD, plasma amino acid analysis reveals elevations of **leucine, isoleucine, alloisoleucine,** and **valine** (the branched chain amino acids) and urine organic acid analysis reveals abnormal branched-chain **hydroxyl-** and **ketoacids**. In expanded screening, leucine/isoleucine and hydroxyproline cannot be differentiated, so if the baby has hydroxyprolinemia confirmatory amino acid analysis will show only increased **hydroxyproline**.

CLINICAL EXPECTATIONS: MSUD presents in the neonate with feeding intolerance, failure to thrive, vomiting, lethargy and maple syrup odor to urine and cerumen. If untreated, it will progress to irreversible intellectual disabilities, hyperactivity, failure to thrive, seizures, coma, cerebral edema, and possibly death. Hydroxyprolinemia is probably benign.

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

SPECIALISTS:

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