TEXAS Department of State Health Services

Newborn Screening ACT Sheet

Increased Leucine
Maple Syrup (Urine) Disease

Differential Diagnosis: Maple syrup urine disease (MSUD); hydroxyprolinemia.

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

Conditions associated with this analyte have been identified by the Society of Inherited Metabolic Disorders (SIMD) as critical, and require immediate action.

Medical Emergency: Take the Following IMMEDIATE Actions

- **Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea).**
- **Consult with pediatric metabolic specialist. (See attached list.)**
- **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.**
- **Initial testing: plasma quantitative amino acids and urine organic acids.**
- **Repeat newborn screen if second screen has not yet been done.**
- **Provide the family with basic information about MSUD and dietary management.**
- **Report findings to newborn screening program.**

Diagnostic Evaluation: In MSUD, plasma amino acid analysis reveals elevations of leucine, isoleucine, alloleucine, 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 mental retardation, hyperactivity, failure to thrive, seizures, coma, cerebral edema, and possibly death. Hydroxyprolinemia is probably benign.

Additional Information:

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

STAR G FELSI
http://www.newbornscreening.info/Parents/aminoaciddisorders/MSUD.html
http://www.newbornscreening.info/Pro/aminoaciddisorders/MSUD.html