



Newborn Screening ACT Sheet [Increased Valine] Maple Syrup (Urine) Disease

Differential Diagnosis:

- Maple syrup urine disease (MSUD);

Condition 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: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- 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.

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:

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 can not be differentiated, so if the baby has hydroxyprolinemia confirmatory amino acid analysis will show only increased hydroxyproline.

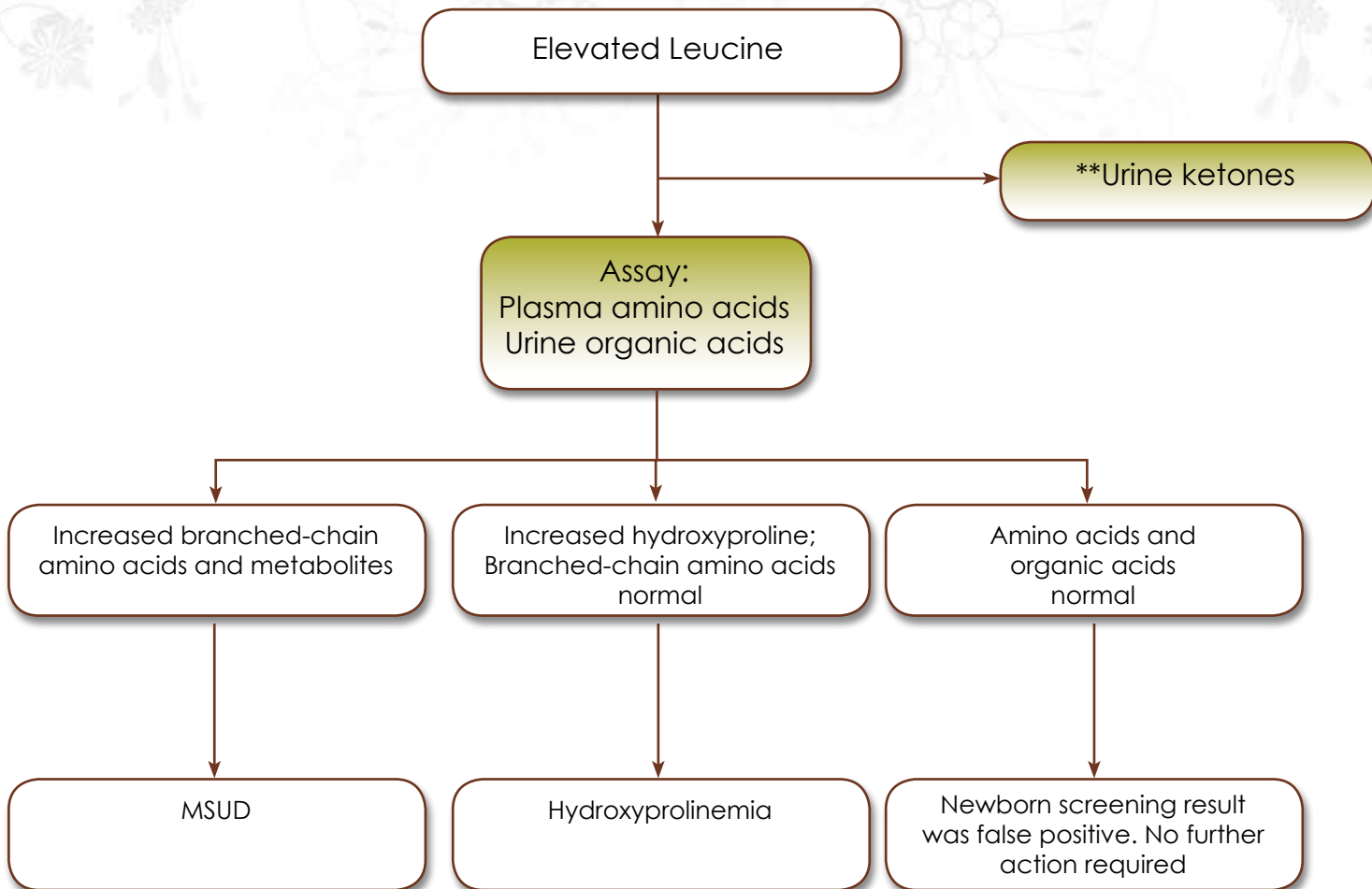
Clinical Expectations:

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

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



Leucine Elevated



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

Abbreviations / Key:

MSUD = Maple (syrup) urine disease

** = When the positive predictive values of screening are sufficiently high and the risk to the baby is high, some initiate diagnostic studies at the same time as confirmation of the screening result is done.

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality clinical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. 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. It may be prudent, however, to document in the patient record the rationale for any significant deviation from these standards and guidelines.