Leucine Elevated

Assay:
Plasma amino acids
Urine organic acids

Increased branched-chain amino acids and metabolites
MSUD

Increased hydroxyproline;
Branched-chain amino acids normal
Hydroxyprolinemia benign

Amino acids and organic acids normal
Newborn screening result was false positive. No further action required

‡ Urine ketones

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: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.