Biotinidase Deficiency

Biotinidase absent/reduced

Serum Biotinidase Assay

- Biotinidase < 10% of normal
  - Biotinidase Deficiency Profound
    - Optional confirmatory testing
      - BTD Gene Sequencing

- Biotinidase 10–30% of normal
  - Biotinidase Deficiency Partial
    - Optional confirmatory testing
      - BTD Gene Sequencing

- Biotinidase Normal
  - Newborn screening result was false positive.
    - No further action required

‡ If symptomatic, obtain: Glucose, electrolytes, blood gas, lactate, ammonia

Abbreviations/Key
BTD = Biotinidase
‡ = 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 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.