KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ABSENT BIOTINIDASE ACTIVITY

CONDITION: BIOTINIDASE DEFICIENCY (BIOT)

DIFFERENTIAL DIAGNOSIS: Biotinidase deficiency

METABOLIC DESCRIPTION: *Biotinidase deficiency results from defective activity of the biotinidase enzyme.*

ACTION TO BE TAKEN **IMMEDIATELY:**

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, lethargy, hypotonia).
- See and evaluate infant.
- Consultation/referral to a metabolic specialist to determine appropriate follow-up.
- If infant can not be seen immediately at metabolic specialist, undertake confirmatory testing in consultation with metabolic specialist.
- Emergency treatment if symptomatic.
- Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Enzyme assay for biotinidase reveals low or absent activity. Plasma acylcarnitine analysis may show normal or increased 3-hydroxyisovaleric acid and 3-methylcrotonyl-glycine. C5-OH acylcarnitine may be high but lack of an abnormal acylcarnitine profile does not rule out biotinidase deficiency.

CLINICAL EXPECTATIONS: The neonate is usually asymptomatic but episodic hypoglycemia, lethargy, hypotonia and mild developmental delay can occur at any time from the neonatal period through childhood. Untreated biotinidase deficiency leads to developmental delay, seizures, alopecia, and hearing deficits. Biotin treatment is available and highly effective.

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

SPECIALISTS:

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DISCLAIMER: These standards and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical 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 reasonable 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's record the rationale for any significant deviation from these standards and guidelines