



NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED C5-OH ACYLCARNITINE

CONDITION: ORGANIC ACIDEMIAS (3MCC, HMG, BKT & MCD)

DIFFERENTIAL DIAGNOSIS: Most likely 3-methylcrotonyl-CoA carboxylase (3MMC) deficiency (**infant or mother**); may be 3-hydroxy-3-methylglutaryl (HMG)-CoA lyase deficiency; β -ketothiolase (BKT) deficiency; or multiple carboxylase (MCD) deficiency including biotinidase deficiency.

METABOLIC DESCRIPTION: Each of the disorders is caused by a deficiency of the relevant enzyme. In most of the disorders, the substrate, for which the enzyme is named, accumulates along with its potentially toxic metabolites.

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

- ◆ Contact family to inform them of the newborn screening results and ascertain clinical status (poor feeding, vomiting, lethargy).
- ◆ Consult with pediatric metabolic specialist.
- ◆ Evaluate the newborn (hypoglycemia, ketonuria, metabolic acidosis). If any of these parameters are abnormal or the infant is ill, initiate emergency treatment as indicated by metabolic specialist and transport IMMEDIATELY to tertiary center with metabolic specialist.
- ◆ Initiate timely confirmatory/diagnostic testing as recommended by specialist.
- ◆ Educate family about signs, symptoms and need for urgent treatment of metabolic acidosis (poor feeding, vomiting, lethargy).
- ◆ Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Confirmatory tests include urine organic acids on infant and mother, plasma acylcarnitine analysis, and serum biotinidase assay. The organic acids analysis on infant and mother should clarify the differential except for biotinidase deficiency (clarified by biotinidase assay).

CLINICAL EXPECTATIONS: The neonate is usually asymptomatic in 3MMC deficiency. However, episodic hypoglycemia, lethargy, hypotonia and mild developmental delay can occur at any time from the neonatal period through childhood for any of these disorders. There is beneficial treatment that is specialized to each condition.

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

SPECIALISTS:

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