KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED C3 ACYLCARNITINE

CONDITION: PROPIONIC ACIDEMIA (PROP) AND METHYLMALONIC ACIDEMIA (MUT, Cbl A & B)

DIFFERENTIAL DIAGNOSIS: Propionic acidemia (PROP); Methylmalonic acidemias (MUT) including defects in B12 synthesis and transport; severe maternal B12 deficiency.

METABOLIC DESCRIPTION: PROP is caused by a defect in Propionyl-CoA carboxylase which converts propionyl-CoA to methylmalonyl-CoA; MUT results from a defect in methylmalonyl-CoA mutase which coverts methylmalonyl-CoA to succinyl-CoA or from lack of the required B_{12} cofactor for methylmalonyl-CoA mutase (cobalamin A,B,C,D and F).

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy, tachypnea.)
- Consult with pediatric metabolic specialist.
- Evaluate the newborn; check urine for ketones and, if elevated or 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 hyperammonemia and metabolic acidosis (poor feeding, vomiting, lethargy, tachypnea).
- Report findings to the newborn screening program.

CONFIRMATION OF DIAGNOSIS: Plasma acylcarnitine confirms the increased C3. Blood amino acid analysis may show increased glycine. Urine organic acid analysis will demonstrate increased metabolites characteristic of propionic acidemia or increased methylmalonic acid characteristic of methylmalonic acidemia.

CLINICAL EXPECTATIONS: Patients with PROP and severe cases of MUT typically present in the neonatal period with metabolic ketoacidosis, dehydration, hyperammonemia, ketonuria, vomiting, hypoglycemia, and failure to thrive. Long-term complications are common; early treatment may be lifesaving and continued treatment may be beneficial.

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

SPECIALISTS:

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