NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED C5 ACYLCARNITINE

CONDITION: ISOVALERIC ACIDEMIA (IVA)

DIFFERENTIAL DIAGNOSIS: Isovaleric acidemia (IVA)

METABOLIC DESCRIPTION: IVA results from a defect in the metabolism of the branched chain amino acid, leucine (isovaleryl-CoA dehydrogenase in IVA). Specific metabolites accumulate and are potentially toxic.

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, odor of sweaty feet).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn; if 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, tachypnea, odor of sweaty feet).
- Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Plasma acylcarnitine analysis confirms the increased C5. Urine organic acid analysis will show isovalerylglycine in IVA. Urine acylglycine and acylcarnitine analysis may also be informative.

CLINICAL EXPECTATIONS: Isovaleric acidemia presents in the neonate with metabolic ketoacidosis, a “sweaty feet” odor, dehydration, hyperammonemia, ketonuria, vomiting, hypoglycemia, and failure to thrive. Milder variants without neonatal illness exist. Long term prognosis of IVA with appropriate therapy is good.

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

SPECIALISTS:

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