KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED C5-DC ACYLCARNITINE

CONDITION: GLUTARYL-COA DEHYDROGENASE DEFICIENCY (GA-I)

DIFFERENTIAL DIAGNOSIS: Glutaric aciduria (GA-I)

METABOLIC DESCRIPTION: GA-I is caused by a defect of glutaryl-CoA dehydrogenase which limits the metabolism of glutaryl-CoA to crotonyl-CoA, resulting in increased glutaric acid (toxic) and its metabolites.

ACTION TO BE TAKEN **IMMEDIATELY:**

- Contact family to inform them of the newborn screening result.
- Consult with pediatric metabolic specialist.
- Evaluate the newborn for macrocephaly and muscle hypotonia. Initiate confirmatory/diagnostic testing as recommended by metabolic specialist.
- Refer to metabolic specialist to be seen as soon as possible but not later than three weeks.
- Educate family about diagnostic possibilities, complexity of diagnostic work-up and the possibility of neurodegenerative crisis with an intercurrent infectious illness.
- IMMEDIATE treatment with IV glucose is needed for intercurrent infectious illness.
- Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Urine organic acid analysis will reveal **elevated glutaric acid** and **3-hydroxyglutaric acid**. Testing should be ordered promptly and is often diagnostic. If urine organic acids don't confirm the diagnosis, the metabolic specialist will consider analyzing glutarylcarnitine in urine and 3-hydroxyglutaric acid in blood and CSF, enzyme assay in fibroblasts, and molecular analysis of the GCDH gene.

CLINICAL EXPECTATIONS: The neonate with glutaric aciduria type I is usually macrocephalic but otherwise asymptomatic. Later signs include metabolic ketoacidosis, failure to thrive and sudden onset of dystonia and athetosis due to irreversible striatal damage. With appropriate treatment, 60-70% of patients will not suffer neurodegenerative disease.

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

SPECIALISTS:

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