



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:

Bryce Heese, MD
Biochemical Genetics
Children's Mercy Hospital- Kansas City, MO

Clinic phone: 816-234-3771
Hospital Operator: 816-234-3000
Office Fax: 816-302-9963

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