Emergency Management Protocol for Glutaric Acidemia Type 1 (GA 1)
(Primary Analyte C5DC < 0.46 μmol/L; Secondary Analyte C5DC/C8 <4.60 and C5DC/C16 <0.20)
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk GA 1 Screen Results

1. Contact the family within **one hour** of notification. Inform family of newborn screen results and assess clinical status (poor feeding, vomiting, lethargy, tachypnea).
2. **Immediate** consultation with a geneticist. Pager number listed below.
3. History and Physical Exam **on same day of notification** either in the pediatrician's office or at the local Emergency department, in consultation with a geneticist.
   May appear normal at birth.
   Assess specifically for signs and symptoms of Metabolic Crises:
   - Macrocephaly
   - Muscle hypotonia
   - Low blood sugar
   - Vomiting
   - Neurological problems
   - Poor appetite
4. **If symptomatic, immediate** phone consultation with a geneticist regarding treatment and clinical management is required.
5. If not symptomatic, schedule diagnostic workup with a geneticist within 24 to 48 hours.

Feeding Precautions

Initiate **feeding precautions** by close of business by giving the parents the following instructions:

1. Wake baby and feed every 3 hours.
2. Use an alarm clock to ensure feedings occur routinely throughout the day and night.
3. Avoid fasting (defined as more than 3 to 4 hours without a feeding).
4. Contact doctor **immediately** or **go to the local emergency department** if baby is not able to feed, is vomiting and/or lethargic.
5. Continue feeding precautions until instructed to stop by a geneticist.

Home Care Precautions

Initiate **home care precautions** by close of business by giving the parents the following instructions:

1. Seek medical attention immediately if baby has concerning symptoms including excessive sleeping, poor feeding, abnormal breathing, fever, decreased urination, vomiting or any minor illness.
2. Seek medical attention immediately if baby is feeding poorly or has difficulty waking up to feed. NOTE: This may be difficult to assess with breast-feeding infants. If there is any concern of poor feeding or poor milk flow, bottle supplementation must be used. Mother should be encouraged to pump and bottle-feed (breast milk or formula) until appointment with a geneticist is achieved.
3. Seek medical attention if baby develops an illness, infection or fever. A metabolic crisis can be triggered by these symptoms.
4. Contact information for the geneticist (pager number listed below).
5. If baby is difficult to arouse or awaken call 911.

Description

This disorder is caused by a deficiency of the enzyme Glutaryl Co-A dehydrogenase. People with this inherited organic acid disorder cannot properly break down certain components of protein (glutaryl-CoA to crotonyl-CoA), causing an increase in organic acids (glutaric acid) in blood and urine when a person eats a normal amount of protein, or becomes sick.

Resources

- **ACMG Newborn Screening ACT Sheets:** https://www.ncbi.nlm.nih.gov/books/NBK55827/
- **Integris Pediatric Specialty Clinic, Inborn Error of Metabolism (IEM) Clinic**
  Geneticist pager: (405) 630-3794
- **OU Children's Physicians – Genetics Clinic**
  Page Operator: (405) 271-3636
- **Newborn Screening Follow-Up Program**
  (405) 271-6617 option 2 or (800) 766-2223; www.nsp.health.ok.gov