Emergency Management Protocol for the Fatty Acid Oxidation Disorders (FAOD) of:
Short-chain acyl-CoA dehydrogenase deficiency (SCAD) & Glutaric acidemia Type II (GAII)
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk SCAD & GAII Screen Results

1. Contact the family by COB (close of business) & initiate *Feeding Precautions* (listed below).
2. Initiate *Home Care Precautions* (listed below) by COB.
3. History and Physical Exam within 8 to 24 hours to assess:
   - Family history of FAOD (family history of SIDS or affected siblings, aunts, uncles etc.)
   - Assess specifically for signs and symptoms of metabolic crisis (acidosis):

SCAD
- Lethargy
- Hypoketotic hypoglycemia
- Seizures
- Vomiting

GAII (same as SCAD)
- "Sweaty feet" odor
- Facial dysmorphism
- Metabolic Acidosis
- Hyperammonemia

4. Immediate phone consultation with a geneticist regarding treatment and clinical management is required.
5. Most infants with **SCAD** are asymptomatic.
6. IV glucose therapy is indicated if infant has signs & symptoms of metabolic crisis.
7. 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 if baby is not tolerating feedings or becomes ill,
5. Failure to feed your baby every 3 hours could result in possible **coma or death**, and
6. Continue feeding precautions until instructed to stop by the 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 or any minor illness.
2. Seek medical attention immediately if baby is feeding poorly. 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 geneticst is achieved.
3. Contact information for the geneticist (pager number listed below).
4. If baby is difficult to arouse or awaken call 911.

Description

SCAD deficiency and GAII are fatty acid oxidation (FAO) disorders. FAO occurs during prolonged fasting and/or during periods of increased energy demands (fever, stress) after glycogen stores become depleted and energy production relies more on fat metabolism. Fatty acids and potentially toxic derivatives accumulate in the body. **SCAD** can have variable presentation. Most affected neonates are asymptomatic. However, an affected neonate can be extremely ill with vomiting, lethargy, seizures, and hypoketotic hypoglycemia. Treatment consists primarily of avoidance of fasting and vitamin/cofactor supplementation. **GAII** often presents in the neonate with poor feeding, marked lethargy, and facial and renal dysmorphism. Additional symptoms include "sweaty feet" odor, failure to thrive, metabolic acidosis and hyperammonemia.

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