Emergency Management Protocol for the Fatty Acid Oxidation Disorder (FAOD) of:
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk VLCAD 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):
     - Lethargy
     - Hypoketotic hypoglycemia
     - Hepatomegaly
     - Hypotonia
     - Cardiomyopathy
     - Evidence of cardiac decompensation
     - Arrhythmias
     - Failure to thrive
4. Immediate phone consultation with a geneticist regarding treatment and clinical management is required.
5. Therapy with IV glucose and oxygen is indicated if infant has signs & symptoms.
6. Even if infant is only mildly ill treatment with IV glucose is indicated.
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 geneticist is achieved.
3. Contact information for the geneticist (pager number listed below).
4. If baby is difficult to arouse or awaken call 911.

Description

VLCAD deficiency is a fatty acid oxidation (FAO) disorders. Fatty acid oxidation occurs during periods of prolonged fasting and/or during periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In a FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO enzymes. VLCAD deficiency may present acutely in the neonate and is associated with high mortality unless treated promptly; milder variants exist. Features of severe VLCAD deficiency in infancy include hepatomegaly, cardiomyopathy and arrhythmias, lethargy, hypoketotic hypoglycemia, and failure to thrive.

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