Emergency Management Protocol for
Carnitine palmitoyltransferase I deficiency (CPT I)
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk CPT1 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 CPT1 (family history of SIDS, fatty acid oxidation disorder, or affected siblings, aunts, uncles etc.)
   - Assess specifically for signs and symptoms of metabolic crisis (acidosis):
     - Cardiac insufficiency
     - Cardiac arrhythmias
     - Marked hypoglycemia
     - Metabolic acidosis
     - Facial dysmorphism
4. If symptomatic, immediate phone consultation with a geneticist regarding treatment and clinical management is required.
5. IV glucose therapy is indicated if infant has signs & symptoms of metabolic crisis.
6. If not symptomatic, schedule diagnostic workup with the 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

This disorder is caused by a deficiency of the enzyme CPT1, preventing the fatty acid carnitine-acylcarnitine linkage required to transport fatty acids into the mitochondria. This results in accumulation of free carnitine (C0) and prevents the fatty acid oxidation response necessary to generate energy during fasting and increased energy needs (fever, stress). CPT1 can have a variable presentation. Critical hypoketotic hypoglycemia is a common presenting feature. Newborns may appear asymptomatic but can progress to fasting hypoketotic hypoglycemia, lethargy, hepatomegaly, and seizures, usually precipitated by fasting or acute illness.

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