Emergency Management Protocol for
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) Screening
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for Significantly Elevated MCAD 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 MCAD (family history of SIDS or affected siblings, aunts, uncles etc.)
   - Assess specifically for signs and symptoms of metabolic crisis (acidosis):
     - Lethargy
     - Nausea or vomiting
     - Hypoglycemia with lack of or only 'trace' amounts of urinary ketones
     - Hepatomegaly
     - *'Reye'* like syndrome
     - Seizures
     - Coma
4. If symptomatic, immediate phone consultation with a geneticist regarding treatment and clinical management is required (IV glucose therapy is indicated if infant has signs & symptoms of metabolic crisis).
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 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, jittery movements, 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

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) is the most frequently occurring fatty acid oxidation disorder (FAOD) and one of the most frequently identified inborn errors of metabolism. The incidence of MCAD ranges from 1/15,000 to 1/20,000 with mortality rates as high as 43% at the initial crisis. It is caused by an intramitochondrial defect in the beta-oxidation of fatty acids and is a major cause of hypoketotic hypoglycemia. MCAD is also a cause for lethargy, liver dysfunction with hepatomegaly, metabolic acidosis, hyperammonemia and sudden death. MCAD **is life threatening**. Immediate intervention is warranted to prevent metabolic crisis.

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