Emergency Management Protocol for 3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency (HMG)/3 methylcrotonyl-coenzyme A carboxylase deficiency (3MCC)/Multiple carboxylase deficiency (MCD)
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk HMG/3MCC/MCD/3MGA/2M3HBA Screen Results

1. Contact the family by COB (close of business) & initiate *Feeding Precautions* (listed below).
2. **Immediate consultation with a geneticist**– pager number listed below.
3. Initiate *Home Care Precautions* (listed below) by COB.
4. History and Physical Exam on same day to assess:
   Symptoms may appear within a few days of birth
   - Assess specifically for signs and symptoms of Metabolic Crises:
     - Poor Feeding/ loss of appetite
     - Hypoglycemia
     - Lethargy
     - Metabolic Ketoacidosis
     - Vomiting /Dehydration
     - Seizures
     - Heart abnormalities
     - Failure to thrive
     - Hypotonia
     - Coma
5. If symptomatic, immediate phone consultation with a geneticist regarding treatment and clinical management is required.
6. 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, does not tolerating feedings or becomes ill.
5. Failure to feed baby every 3 hours could result in possible **coma or death**.
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 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 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

These disorders are caused by the body's inability to process certain proteins and fats properly. Abnormal levels of organic acids in the blood, urine and tissue can be toxic and lead to serious health problems.

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