

**Emergency Management Protocol for the Fatty Acid Oxidation Disorders (FAOD) of:
Long-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) &
Trifunctional protein deficiency (TFP) Screening**

Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk LCHAD & TFP 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
 - Cardiomyopathy
 - Lactic acidosis
 - Cardiac insufficiency
 - Elevated liver transaminases
 - Failure to thrive
 - Maternal liver disease during pregnancy
4. If symptomatic, immediate phone consultation with a metabolic specialist 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 metabolic specialist 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 metabolic specialist.

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 metabolic specialist is achieved.
3. Contact information for the metabolic specialist (phone numbers listed below).
4. If baby is difficult to arouse or awaken call 911.

Description

LCHAD and TFP deficiencies 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 FAO disorders, which are caused by a deficiency in one of the enzymes involved in FAO. LCHAD and TFP deficiencies usually present acutely and are associated with high mortality unless treated promptly. Milder variants do exist. Features include hepatomegaly, cardiomyopathy, lethargy, hypoketotic hypoglycemia, elevated liver transaminases, lactic acidosis, and failure to thrive.

Resources

Metabolic Nurse Specialist – OKC and Tulsa Mary Monks, RN

Phone (405) 271-8685 Pager (405) 559-1378

Newborn Screening Follow-up Program (405) 271-6617 or 1-800-766-2223

www.nsp.health.ok.gov

Metabolic Specialists

Klaas Wierenga, MD

Phone: (405) 271-8685 Pager: (405) 218-1422

Susan Palmer, MD

Phone: (405) 271-8685 Pager: (405) 502-8973