

**Emergency Management Protocol for  
Maple Syrup Urine Disease (MSUD) Screening**  
Newborn Screening Program of the Oklahoma State Department of Health

**Differential Diagnosis:** MSUD, Hydroxyprolinemia

**Evaluation & Initial Management Guidelines for Significantly Out-of-Range MSUD Screen**

1. Contact the family within one hour of notification. Inform family of newborn screen result and assess clinical status (poor feeding, vomiting, lethargy, tachypnea).
2. Immediately consult with the metabolic specialist.
3. History and Physical Exam by COB in consultation with the metabolic specialist:
  - **Assess specifically for signs and symptoms of Maple Syrup Urine Disease**
    - Poor Feeding
    - Vomiting
    - Lethargy
    - Tachypnea
    - Alternating Hypotonia/Hypertonia
    - Opisthotonus
    - Ketonuria
    - Possible high-pitched cry
  - Family history of MSUD (family history of SIDS or affected siblings, aunts, uncles etc.)
  - **Immediate** phone consultation with a metabolic specialist regarding treatment and emergency clinical management is required if infant is symptomatic.
4. In coordination with the metabolic specialist, arrange immediate transportation to metabolic center for diagnostic work-up and medical management.

**Description**

Maple Syrup Urine Disease (MSUD) results when branch chain amino acids, Valine, Isoleucine and Leucine cannot be metabolized. Vomiting and lethargy may be present. Untreated in the newborn period, MSUD will quickly progress to cerebral edema, seizures, coma, irreversible mental retardation and possibly death. **MSUD is life threatening.** Coma and central respiratory failure can occur in the first 4 days of life. Immediate intervention is warranted to prevent profound mental retardation, and death.

**Resources**

Mary Monks, RN, Metabolic Nurse Specialist, pager (405) 559-1378, phone (405) 271-8685.  
Newborn Screening Follow-up Program (405) 271-6617 or 1-800-766-2223.

**Metabolic Specialist in Oklahoma**

**Klaas Wierenga, M.D.**  
Phone: (405) 271-8685  
Pager: (405) 218-1422

**Susan Palmer, M.D.**  
Phone: (405) 271-8685  
Pager: (405) 502-8973