

Emergency Management Protocol for Malonic Acidemia (MAL)
(Primary Analyte - C3-DC < 0.44 μmol/L)
Newborn Screening Program of the Oklahoma State Department of Health

Evaluation & Initial Management Guidelines for High Risk Malonic Acidemia screening results

1. Contact the family within **one hour** of notification. Inform family of newborn screen results and assess clinical status.
2. **Immediate consultation with the metabolic specialist** – pager and phone numbers listed below.
3. History and Physical Exam **on same day of notification** either in the pediatrician's office or in the local Emergency department if after hours, in consultation with metabolic specialist
May appear normal at birth
 - Assess specifically for signs and symptoms of Metabolic Crises:
 - Muscle hypotonia
 - Vomiting/Diarrhea
 - Lactic Acidosis
 - Hypoglycemia
 - Seizures
 - Cardiomyopathy
 - Failure to thrive
 - Developmental Delay (childhood)
4. **If symptomatic, immediate** phone consultation with a metabolic specialist regarding treatment and emergency clinical management is required.
5. If not symptomatic, in consult with metabolic specialist for medical management, consider admission until follow-up with metabolic specialist and schedule diagnostic work-up within 24 hours.
6. Ensure that baby is awoken **every three hours** for feedings

Description

This disorder is caused by a deficiency of the enzyme Malonyl-coenzyme A (CoA) decarboxylase. People with this inherited organic acid disorder cannot convert fats to energy, which can lead to characteristic features of this disorder such as low blood sugar and cardiomyopathy.

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