

**Follow-Up Management Protocol for  
Newborns with Elevated Methionine Screening**  
Newborn Screening Program of the Oklahoma State Department of Health

**Differential Diagnosis:** Classical homocystinuria (cystathionine beta synthase deficiency);  
Hypermethioninemia (due to MAT I/III deficiency); liver disease; hyperalimentation

**Evaluation & Initial Management Guidelines for Significantly Elevated Methionine Screen**

1. Contact the family by close of business. Inform family of newborn screen result and ascertain clinical status.
2. Immediately consult with the metabolic specialist.
3. History and Physical Exam within 24 hours in consultation with the metabolic specialist:
  - **Assess specifically for signs and symptoms of liver disease.**
4. If symptomatic, immediate phone consultation with a metabolic specialist regarding treatment is required.
5. If not symptomatic, consult with metabolic specialist for medical management, and to schedule diagnostic work-up to occur within 24-48 hours.

**Description**

Methionine from ingested food is usually converted to homocysteine. In classical homocystinuria, homocysteine cannot be converted to cystathionine. As a result, the concentration of homocysteine and its precursor, methionine, will become elevated.

Homocystinuria is usually asymptomatic in the neonate. If untreated, these children eventually develop mental retardation, ectopia lentis, a marfanoid appearance including arachnodactyly, osteoporosis, other skeletal deformities and thromboembolism. Hypermethioninemia may be benign.

**Resources**

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

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**Susan Palmer, M.D.**

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