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 geneticist.
3. History and Physical Exam within 24 hours in consultation with the geneticist:
   - **Assess specifically for signs and symptoms of liver disease.**
4. If symptomatic, immediate phone consultation with a geneticist regarding treatment is required.
5. If not symptomatic, consult with geneticist 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 cognitive and intellectual disabilities, ectopia lentis, a marfanoid appearance including arachnodactyly, osteoporosis, other skeletal deformities and thromboembolism. Hypermethioninemeia may be benign.

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