Methionine Screening - Homocystinuria

**Differential Diagnosis:** Classical Homocystinuria (cystathionine beta synthase (CBS) deficiency; Hypermethioninemia; GNMT deficiency; Adenosylhomocysteine hydrolase deficiency; Liver disease; Hyperalimentation
Oklahoma State Department of Health Newborn Screening Program

### Methionine Screen NICU on TPN

**Out-of-Range (abnormal)**

**Primary Analyte**

**Methionine** > 160 µmol/L

**YES**

1. Assess infant’s clinical status - Repeat filter paper after TPN is discontinued or as scheduled time to recollect per NICU policy.
2. If repeat is elevated – Consult/Refer for evaluation and diagnostic work-up by the metabolic specialist.
   - Evaluation by the metabolic specialist must occur within 2 weeks of notification of two out of range results.

**Primary Analyte**

**Methionine** > 160 µmol/L

**NO**

- Not consistent with disorder of methionine metabolism.
- No further action needed.

### Table 1. In-range Methionine Screen Results:

<table>
<thead>
<tr>
<th>Primary Analyte: Methionine &lt; 100 µmol/L</th>
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<tbody>
<tr>
<td>Secondary Analyte: Methionine/Phenylalanine ratio &lt; 1.2</td>
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1. For reference ranges and list of other analytes that will be reported, see Table 1.

### Diagnostic Testing

**Consistent with:**
- Refer to metabolic specialist for medical management.

**Within Normal Limits:** Not consistent with disorder of methionine metabolism. No further follow-up indicated.

**Inconclusive:**
- Monitoring and medical management as advised by metabolic specialist.

#### From the time the screen is reported to the provider, the Metabolic Nurse Specialist will monitor follow-up by:

1. Confirming the provider contacts family by COB.
2. Facilitating and confirming a clinical evaluation by a provider or metabolic specialist is achieved before COB.
3. Facilitating and confirming infant presents for a diagnostic workup with a metabolic specialist within 24 hours.
4. Coordinating collection and processing of diagnostic tests and communicating test results to provider and short-term follow-up program (STFU).
5. Communicating with STFU if the above timelines are not met.