

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

Methionine Screen
Out-of-Range (abnormal)

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

YES

Primary Analyte
Methionine \geq **100 μ mol/L & < 160 μ mol/L**
¹ For reference ranges and list of other analytes that will be reported, see Table 1.

Primary Analyte
Methionine \geq **160 μ mol/L**

Contact family within 24 hours to assess infant's clinical status.
 Repeat Filter Paper within 48 hours.

In-Range

Out-of-Range

Not consistent with.
 No further follow-up indicated.

From time of report, the contacted provider will:

1. Contact family by COB.
2. In consultation with the metabolic specialist, arrange clinical evaluation utilizing the Medical Management Protocol (initial assessment can be done by provider or metabolic specialist).
3. Refer for evaluation and diagnostic work-up by the metabolic specialist. Evaluation by the metabolic specialist must occur within 2 weeks of notification of out of range results.

Appointment with metabolic specialist for diagnostic testing (testing must be coordinated by the specialist or newborn screening program):

1. Plasma amino acids
2. Total homocysteine
3. Urine Organic Acids
4. Other lab and/or DNA may be indicated

Table 1. In-range Methionine Screen Results¹:

Primary Analyte:
 Methionine < 100 μ mol/L

Secondary Analyte²:
 Methionine/Phenylalanine ratio < 1.2

¹These values are utilized for newborns less than 60 days old.
²Elevations of the secondary analytes are reported as "not consistent with a disorder of Methionine metabolism" if methionine is in-range.

Diagnostic Testing Inconclusive:
 Monitoring and medical management as advised by metabolic specialist.

Diagnostic Testing Consistent with: Refer to metabolic specialist for medical management.

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

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.