Methionine Elevated or Decreased

Elevated or decreased Methionine

Assay
- Plasma amino acids
- Plasma homocysteine
- Plasma MMA

Increased methionine; Increased total homocysteine
- Homocystinuria
  (CBS deficiency)

Increased methionine; Normal/slightly increased total homocysteine
- Hypermethioninemia requiring further work up for:
  - Mat I/III deficiency
  - GNMT deficiency
  - AdoHcy hydrolase deficiency

Decreased methionine; Increased total homocysteine; Normal MMA
- CbiE, CbiG, or MTHFR def.

Decreased methionine; Increased total homocysteine; Elevated MMA
- CbiC, CbiD, or CbiF deficiency

Optional Confirmatory Testing:
- Cbi Complement Studies (fibroblasts); Molecular
- Newborn screening result was false positive. No further action required

Action steps are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key
- AdoHcy hydrolase = Adenosylhomocysteine hydrolase
- Cbi = cobalamin
- CBS = Cystathionine β-synthase
- GNMT = glycine N-methyltransferase
- Mat = Methyladenosyltransferase
- MMA = Methylmalonic Acidemia
- MTHFR def = Methylene Tetrahydrofolate Reductase Deficiency

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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