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
CblE, CblG, or MTHFR def.
Optional Confirmatory Testing:
Cbl Complement Studies (fibroblasts); Molecular

Decreased methionine; Increased total homocysteine; Elevated MMA
CblC, CblD, or CblF deficiency

Normal
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.