METHIONINE IN HOMOCYSTINURA

Guideline Recommendations

Monitoring methionine <u>in addition</u> to homocysteine is crucial to assess the effectiveness of treatment and risk of complications

Diagnosis	Recommended Plasma Methionine Level	Reason
CBS Deficiency (Classical Homocystinuria)	Less than 1000umol/L	Decreases risk of cerebral edema and encephalopathy
Remethylation Disorders (Cobalamin and Severe MTHFR)	10-40 µmol/L Within Normal Limits	Ensure availability of methyl donor reactions

Important Considerations

Betaine treatment must be managed appropriately to ensure plasma methionine levels remain in the recommended range

Regular labs are recommended for close monitoring of plasma methionine along with plasma homocysteine levels
Childhood (Every 3-4 months), Adolescence & Adulthood (Every 6 months)

Low methionine in patients with remethylation disorders increases risk of developmental delay, seizures and intellectual disability.

Suggested Laboratory Findings-CBS Deficiency

Following a high homocysteine lab result, plasma methionine levels within the following ranges, could suggest a diagnosis of inherited homocystinuria.

	Childhood-Onset Multisystem Disease	Adult-Onset Thromboembolic Disease	Normal Range
Plasma Methionine Concentration	**200-1,500 µmol/L**	**>50 µmol/L**	**10-40 µmol/L**

This is for educational purposes only. Please consult your medical team before making any changes to your care.

https://www.ncbi.nlm.nih.gov/books/NBK1524/ https://jcp.bmj.com/content/75/11/744.long

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