



Homocysteine (Total), Methylmalonic Acid,
and Methylcitric Acid, Blood Spots

Patient ID M0002122	Patient Name SAMPLEREPORT, HCMM A	Birth Date 1971-09-28	Gender F	Age 40
Order Number M0002122	Client Order Number M0002122	Ordering Physician ,	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 27 Jun 2012 00:45		


HCMM, BS

Reason For Referral

Reason for referral not provided.

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Homocysteine

 **27.9 nmol/mL**
High

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Reference Value
<15.0

Methylmalonic Acid

3.8 nmol/mL

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Reference Value
<5.0

Methylcitric Acid

0.5 nmol/mL

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Reference Value
<1.0

Interpretation

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These results are suggestive of a diagnosis of either homocystinuria (cystathionine beta synthase deficiency), methylene tetrahydrofolate reductase deficiency (MTHFR) or another defect in cobalamin metabolism. We recommend laboratory evaluation to include a quantitative plasma amino acid, total homocysteine, methylmalonic acid, and urine organic acid analyses.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Reviewed By

MCR

Jennifer Peterson

Received: 27 Jun 2012 00:45

Reported: 20 Feb 2013 07:57

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905