

Methylmalonic Acid (MMA) Serum Test Information



Methylmalonic acid (MMA) is a dicarboxylic acid that is a C-methylated derivative of malonate. It is usually secreted in very small quantities from the human body; however, MMA levels can be elevated markedly in patients with rare inherited metabolic diseases collectively known as the methylmalonic acidurias. Also, since vitamin B12 is involved in the conversion of methylmalonyl CoA (a form of MMA) to succinyl CoA, MMA concentration in serum or urine increases when vitamin B12 deficiency is present in patients. MMA testing is very sensitive when used as the confirmatory test for early to mild vitamin B12 deficiency, and it is more specific than homocysteine for such diagnosis. It also offers a sensitive marker for vitamin B12 deficiency.

Gas chromatography with mass spectrometry (GC-MS) has been used extensively for urinary organic acid profiling, including MMA. Recently, liquid chromatography-tandem mass spectrometry (LC-MS/MS) has been used in MMA analysis, which shows a high degree of correlation with GC-MS assay results while providing additional advantages of sensitivity, speed, and reproducibility. Our lab developed a rapid LC-MS/MS assay combined with a stable-isotope labeled internal standard for serum MMA level quantification. For more information, call the lab at 513-636-4203.

Sample Type:

Serum (Red, No Gel)

Volume:

1.5 mL

0.5 mL (minimum)

Specimen Preparation:

Spin/pour off within 2 hours of collection. Transfer serum to a standard transport tube and refrigerate or freeze immediately.

Unacceptable Specimens:

Ambient temperature, grossly-hemolyzed, or lipemic specimens.

Stability:

Ambient: Unacceptable

Refrigerated: 4 days

Frozen: 1 month

Methodology:

Liquid chromatography-tandem mass spectrometry (LC-MS/MS)

Reporting Units:

Quantitative: mcmol/L

LC-MS/MS Calibration Range:

0.04 mcmol/L - 4.24 mcmol/L

Reference Interval:

0.04 – 0.40 mcmol/L

Shipping Conditions:

Frozen (dry ice), next day

Testing Schedule:

Fri, 1st Shift (for testing outside this schedule, please call 513-636-4203). **Turnaround time:** 3 – 7 days.

CPT Code:

83921

Contact Information:

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Shipping Address:

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Cincinnati Children's Hospital Medical Center
240 Albert Sabin Way
Cincinnati, Ohio 45229-3039

References:

1. Herrmann W. and Obeid R. Causes and early diagnosis of vitamin B12 deficiency. Dtsch Arztebl Int 2008 (105)680-685.
2. Obeid R. et al. Comparison of two methods for measuring methylmalonic acid as a marker for vitamin B12 deficiency. Diagnosis 2015 (2) 67-72.