

Methylmalonic Acid (MMA) Urine 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.

Gas chromatography with mass spectrometry (GC-MS) has been used extensively for urinary organic acid profiling, including MMA. Our lab offers a rapid GC-MS assay combined with a stable-isotope labeled internal standard for urinary MMA level quantification and interpretation. For more information, call the lab at 513-636-4203.

Sample Type:

Urine, random collection

Volume:

5 mL (minimum)

Specimen Preparation:

Random urine collection, freeze ASAP.

Unacceptable Specimens:

Ambient temperature

Stability:

Ambient: Unacceptable

Refrigerated: 4 days

Frozen: 1 month

Methodology:

Gas chromatography-mass spectrometry (GC-MS)

Reporting Units:

Quantitative: mmol/mol creatinine

Reference Interval:

≤ 3.6 mmol/mol creatinine

Shipping Conditions:

Frozen (dry ice), next day.

Testing Schedule:

Biweekly, depending on batch volumes (for urgent testing, please call 513-636-4203). **Turnaround time:** 7-10 days.

CPT Code:

83921

Contact Information:

Clinical Mass Spectrometry

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Fax: 513-803-5014

Email: pathology@cchmc.org

Website: www.cincinnatichildrens.org/mass-spec

Shipping Address:

Clinical Mass Spectrometry Facility, MLC 7019
Division of Pathology and Laboratory Medicine
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. Lindenbaum J. et. al. Diagnosis of cobalamin deficiency: II. Relative sensitivities of serum cobalamin, methylmalonic acid, and total homocysteine concentrations. *Am. J Hematol* 1990 (34) 99-107.