Methylmalonic Acid

CPT Code: 83921
Order Code: 34879
ABN Requirement: No
Synonym: MMA
Specimen:
Preferred: Serum
Acceptable: Plasma
Volume: 2.0 mL
Minimum Volume: 1.0 mL
Container:
Preferred: R, red-top tube (no gel)
Acceptable: SS, serum separator tube, or RB, royal blue-top tube, no additive , or Gn, green-top tube, sodium heparin or RB, royal blue-top tube, sodium heparin

Collection:

1. Allow blood to clot at room temperature for 30 minutes. Centrifuge and separate the serum from cells immediately.
2. Aliquot serum into transport tube labeled as “Red Top Serum” and cap tightly. Discard original tube.

Note: If a Serum Separator Tube (SST) is used for specimen collection, serum must be separated from the original SST tube and aliquoted into transport tube labeled “SST serum.”

Transport: Store serum at 2°C to 8°C after collection and ship the same day per packaging instructions included with the provided shipping box.

Stability:

Ambient (15-25°C): 4 days
Refrigerated (2-8°C): 7 days
Frozen (-20°C): 10 months

Methodology: GAS CHROMATOGRAPHY/MASS SPECTROMETRY (GC/MS)

Turn Around Time: 3 to 4 days

Reference Range:

Age	nmol/L
All Ages	87-318

Clinical Significance: The Methylmalonic Acid (MMA) test is used in the diagnosis of acquired cobalamin (vitamin B12) deficiency in adults and to screen for inherited organic acidemia in neonates and infants. Elevated MMA in either blood or urine indicates vitamin B12 deficiency in adults, with MMA acting as a functional biomarker for vitamin B12 status. In neonates and infants, elevated MMA is associated with inborn errors of metabolism [1].

Adults with signs and symptoms of cobalamin deficiency, including peripheral neuropathy, ataxia, memory impairment, depression, behavioral changes, and anemia, should be tested for MMA, especially if they are elderly or have experienced intestinal malabsorption or digestive disorders [1]. In the United States all newborns should be screened for MMA as part of the Department of Health and Human Services (HHS) Recommended Universal Newborn Screening Panel [2].

MMA can be acquired due to underlying medical conditions that lead to B-vitamin deficiencies or inherited as an autosomal recessive inborn error of metabolism. If a neonate or infant has elevated MMA suggestive of an organic acidemia, the parents may elect to undergo carrier testing, or have their other children undergo genetic testing. Siblings of a child with MMA-related mutation have a 25% chance of being affected and a 50% chance of being a carrier [3].

References:

1. AACC. Methylmalonic acid. Updated May 10, 2019. https://labtestsonline.org/tests/methylmalonic-acid. Accessed July 20, 2019.
2. Department of Health and Human Services. Recommended Uniform Screening Panel. https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html. Updated February 2019. Accessed July 2019.
3. Manoli, et al. Isolated methylmalonic academia. Updated: December 1, 2016. In: Adam MP, Ardinger HH, Pagon RA, et al. editors. GeneReviews [Internet]. Seattle (WA)L University of Washington

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