



THE AGA KHAN UNIVERSITY HOSPITAL CLINICAL LABORATORIES

UPDATE

Plasma Methylmalonic Acid Quantification

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Introduction

Methylmalonic acid (MMA) is an important intermediate product of amino acid decomposition in a vitamin B12-dependent reaction. Elevated levels of methylmalonic acid (MMA) result from inherited defects of enzymes involved in MMA metabolism or inherited or acquired deficiencies of vitamin B12 or its downstream metabolites. MMA is also a specific marker for the group of disorders collectively called methylmalonic acidemia, which include at least 7 different complementation groups.

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

Specimen Collection:

Essential clinical information:

- It is essential to fill the request form related to inborn error of metabolism (Inherited metabolic disease) provided at the reception of AKU Clinical laboratory, Collection Points and Consulting Clinics.
- Instruct patient's attendant to provide previous reports related to inherited metabolic disease if available
- Encourage the patients to get their test charged against the initial laboratory number each time so that the laboratory record related to patient can be readily available.

Specimen Type: Blood in heparinized tube or separated plasma.

Specimen Volume:

- Adult: 3-4 ml of blood or 0.5-1 ml of separated plasma
- Infant or child: 1-2 ml of blood or 0.5-1 ml of separated plasma.
- Minimum acceptable volume of separated plasma is 0.3 ml (300 ul).

Note: If the blood specimens cannot be sent to the laboratory immediately then centrifuge and separate the plasma into clean tube labelled with patient's identification. Separated plasma (preferred) should be transported at 2 - 8 °C.

REFERENCE RANGE: 73-271 nmol/L

SCHEDULE: Test will be performed twice a month;

- i) First Monday of the month and reported on following Wednesday.
 - ii) Third Monday of the month and reported on following Wednesday.
- (Cut-off: Monday morning 8:00 am)

PLEASE FILE FOR QUICK REFERENCE

