Methylmalonic Acid, Quantitative, Urine

Test Code: BMMAU
Turnaround time: 7 days
CPT Codes: 83921 x1

Condition Description

Methylmalonic acid is the hallmark for the group of disorders called methylmalonic acidemias, which are inborn errors of metabolism in the catabolism of branch chain amino acids. Methylmalonic acidemias (MMA) are divided into those that are deficient in mutase apoenzyme and those deficient in the synthesis of adenosyl cobalamin, an essential co-factor for mutase.

Among the defects in cobalamin metabolism, two complimentary groups, Cbl A and B, affect synthesis of adenosyl cobalamin. Two other groups, Cbl E and G, affect the synthesis of methyl cobalamin, which is an essential co-factor for methionine synthase, and defects result in hyperhomocysteinemia. Also, defects in Cbl C, D, and F affect synthesis of both adenosyl cobalamin and methyl cobalamin, and will therefore result in both methylmalonic acidemia and hyperhomocysteinemia.

Mildly increased MMA in blood and urine may be found in patients with cobalamin deficiency, impaired digestion or poor feeding, and bacterial overgrowth of the small intestine. Elderly patients with cobalamin deficiency may present with peripheral neuropathy, loss of position and vibration senses, memory impairment and depression.

Methylmalonic acid quantitation can lead to early detection and follow up of inborn errors of the propionate and methylmalonic pathway, as well as defects of cobalamin synthesis; it can also detect acquired cobalamin, and/or folate deficiency as well as certain associations with increased risk of cardiovascular diseases.

GeneReviews Summaries:
- Methylmalonic aciduria (MMA)
- Propionic acidemia (PA)

Indications

This test is indicated for individuals with:
- Clinical symptoms of methylmalonic acidemia (MMA) or propionic acidemia (PA)
- Follow up to abnormal newborn screening results

Methodology

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

Detection

This test is very sensitive for methylmalonic acidemia, but detection can be sensitive to age and dietary status of the patient.

Reference Range

0.1-3.6 mmol/mol creat. ¹


Specimen Requirements

Additional Specimen Collection/Handling Instructions Required for this Test
For diagnostic purposes, specimens should ideally be collected during time of acute illness as abnormal metabolite levels may decrease, sometimes to near normal concentrations, when patient is well.

Type: Urine

Specimen Requirements:

In a clean container without preservatives: 1-5 ml. Freeze.

Fasting or first void sample is preferable.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

Related Tests

Biochemical
- Organic Acids Quantitative Analysis (OA)
- Acylcarnitine Profile (AR)
- Homocysteine, Total, Plasma (HO)
- Homocysteine, Total, Dried Blood Spot (BHOS)
- Methylmalonic Acid Quantitation, Plasma (MQ)
- Methylmalonic Acid and Methylcitric Acid, Dried Blood Spot (BMMAD)
- NBS Follow-up: Elevated C3 (BNBSF)

Molecular

- Methylmalonic Aciduria (MMA): Methylmalonyl CoA Mutase (MUT) Full Gene Sequencing (DH)
- Methylmalonic Aciduria (MMA): Methylmalonyl CoA Mutase (MUT) Gene Deletion/Duplication (NK)
- Methylmalonic Aciduria (Cbl A/Cbl B): MMAA and MMAB Full Genes Sequencing (MU)
- Methylmalonic Aciduria (Cbl A/Cbl B): MMAB and MMAB Genes Deletion/Duplication (NJ)
- Methylmalonic Aciduria and Homocystinuria, cblC Type: MMACHC Full Gene Sequencing (XU)
- Methylmalonic Aciduria and Homocystinuria, cblC Type: MMACHC Gene Deletion/Duplication (XW)
- Propionic Acidemia (PA): PCCA and PCCB Full Gene Sequencing (KK)
- Propionic Acidemia (PA): PCCA and PCCB Gene Deletion/Duplication (KI)