METHYLMALONATE

Relevant disorders

Methylmalonic acidaemia, cobalamin defects, Vitamin B12 deficiency

Related Metabolic Tests

Urinary organic acids
Homocysteine
Plasma amino acids (methionine)

Indication for Test

Methylmalonic acidaemia is the result of low activity of the mutase enzyme responsible for transforming methylmalonyl CoA into succinyl CoA. Either B12 cofactor deficiency or mutase deficiency can cause methylmalonic acid (MMA) to accumulate with a variety of presentations dependant on the site of the defect. The non-B12 responsive mutase deficiency is usually the most severe form presenting neonatally with acidosis, hyperammonaemia, hypocalcaemia, neutropenia and liver disease. Neonatal death is not uncommon thus prenatal diagnosis can be offered using MMA concentration in amniotic fluid.

MMA can also be mildly or grossly elevated in Vitamin B12 deficiency.

Methodology

Stable isotope dilution GC-MS.

Sample requirements

5 mL random urine (no preservative).

5ml amniotic fluid (for prenatal diagnosis of methylmalonic acidaemia).
Laboratory must be contacted before sending sample (see contact details below).
Centrifuge the amniotic fluid and send half of the supernatant only. Cells are not required for this assay. The sending laboratory should retain the rest of the supernatant and store at -20˚C.

Transport information/ Contact Details

Send by first class post. Normal packaging.

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Turn Around Time

2 weeks for urine analysis
Within 7 days for pre-natal diagnosis

Reference Ranges

- Urinary methylmalonic acid

1.0– 8.0 µmol/mmol creatinine

Reference range established internally from patient data

(N.B: reference range only valid with data produced by the Department of Clinical Chemistry, Sheffield Children's NHS Foundation Trust).

- Amniotic fluid methylmalonic acid

Interpretation provided with the report

References

- Holm et al J.Inher.Metab.Dis.12.Suppl2.(1989)