HEXANOYLGLYCINE

Relevant disorders

Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)
Multiple Acyl CoA Dehydrogenase deficiency (MADD; GA2)

Related Metabolic Tests

Urine organic acids
Acylcarnitines

Indication for Test

Hexanoylglycine is a diagnostic metabolite for Medium Chain Acyl CoA Dehydrogenase deficiency (MCAD) and Multiple AcylCoA dehydrogenase deficiency (MAD or GAII). Diagnosis of patients in non-crisis periods can depend on detection of often low concentrations of acylglycines. Quantitation with a stable isotope internal standard can clarify a diagnosis, especially useful for newborn screening follow-up testing, where an initial increased octanoylcarnitine dried blood spot result was not confirmed by a detectable peak of hexanoylglycine by organic acid profiling.

Methodology

LC-MS/MS using a deuterated internal standard

Sample requirements

2ml random urine, no preservative

Transport information/ Contact Details

Send by first class post.

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Turn Around Time

ASAP. Metabolic laboratory MUST be contacted (see contact information below).

Reference Ranges

Urinary Hexgly 0.1 - 1.1 µmol/mmol creatinine [MCADD > 2.1]

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

There have been rare cases of MCADD where hexanoylglycine excretion was within the normal range. Therefore normal excretion of hexanoylglycine does not confirm exclusion of MCADD.

References

- Ramsdell HS et al Mass spectrometric studies of twenty one metabolically important acyglycines. Biomed.Mass.Spectrom. Vol 4 No 4 1977

- Rinaldo P et al Medium-chain acyl-CoA dehydrogenase deficiency. Diagnosis by stable-isotope dilution measurement of urinary n-hexanoylglycine and 3-phenylpropionylglycine.
N Engl J Med. 1988 Nov 17;319(20):1308-13. Erratum in: N Engl J Med 1989 May 4;320(18):1227.