

2 HYDROXY GLUTARIC ACID - DETERMINATION OF CHIRALITY

Relevant disorders

D- or L- 2-hydroxy glutaric aciduria

Related Metabolic Tests

Urine organic acids

Indication for Test

2-Hydroxyglutaric acid (2HG) is thought to be mainly formed from the reduction of 2-ketoglutarate of the Krebs cycle. Normally detected in small amounts in urine this compound is notably elevated in Glutaric aciduria type 2 and 2-hydroxyglutaric aciduria. The latter disorder can be divided into 2 depending on the chirality of 2HG - ie whether the 2HG is the L- (laevorotatory) or the D-(dextrorotatory) stereoisomer (enantiomer).

D-2-Hydroxyglutaric aciduria: only a few cases described with varied presentation from severe neuropathy, seizures, blindness and alopecia to protein losing enteropathy, vomiting, cardiomyopathy and normal mental development. In some infants the 2HG has normalised without intervention. The 'normal' form of 2HG (from 2KG) a raised level of D-2HG is seen as the less severe disorder.

L-2-Hydroxyglutaric aciduria: more than 20 cases have been described with symptoms characterised in many as progressive ataxia, peripheral leukodystrophy (U-fibres), cerebellar atrophy, changes in basal ganglia, extrapyramidal signs, mental regression and eventual development of seizures. The lesion responsible for this disorder has not been characterised. Seizures can be controlled with anti-epileptics, but overall the prognosis is poor.

Methodology

Gas Chromatography Mass Spectrometry

Sample requirements

5ml urine, no preservative. Please send evidence of increased excretion of 2-hydroxy glutarate (i.e. an organic acid chromatogram of the same sample).

Transport information/ Contact Details

Send by first class post.

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Turn Around Time

4 – 6 weeks

Reference Ranges

Not applicable

References

- Duran M et al. L-2-Hydroxyglutaric aciduria: an inborn error of metabolism? J Inherit Metab Dis. 1980;3(4):109-12.
- Watanabe H et al . Identification of the D-enantiomer of 2-hydroxyglutaric acid in glutaric aciduria type II. Clin Chim Acta. 1995 Jul 14;238(2):115-24.