DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

HOMOCYSTEINE

Relevant disorders

Classical Homocystinuria (Cystationine β-synthase deficiency); MTHFR deficiency; Cobalamin defects; Hyperhomocystinaemia;

Related Metabolic Tests

Plasma amino acids
Urinary methylmalonic acid

Indication for Test

Determination of total homocysteine is used for the detection and monitoring of patients with inborn errors of homocysteine and/or cobalamin metabolism. These disorders include: classic homocystinuria due to the deficiency of cystathionine synthase; remethylation defects due to methylene tetrahydrofolate reductase deficiency (MTHFR) and cobalamin defects that lead to increased homocysteine (such as cblE and cblG). Clinically, classical homocystinuric patients present with Marfan-like appearance, epilepsy, mental retardation, lens dislocation and thromboembolism. Other milder forms of homocystinuria have a less severe clinical picture. All are treatable and therefore early detection is important.

In adults, mild hyperhomocysteinaemia has been recognised as a risk factor for the development of vascular disease, particularly strokes and other thromboembolitic events, but more controversially is also considered a risk factor for myocardial infarction. Mild hyperhomocysteinaemia may arise from deficiency of folate and/or vitamin B12 deficiency (co-factors in the pathway) which is relatively common in the adult population. Hyperhomocysteinaemia is also due to homozygosity for a polymorphism in the MTHFR gene which produces an enzyme that has reduced stability and therefore reduced activity. 5% of Europeans are homozygous for this polymorphism.

Sample requirements

2ml Lithium heparin sample (EDTA acceptable) separated within 30 minutes of collection. Ideally should be a fasting sample.

5ml urine (random or aliquot of 24 hours collection)

Please note: plasma is the preferred sample for total homocysteine.

Methodology

HPLC

Turn Around Time

2 weeks

Transport information/Contact details

Send all samples by first class post or courier if urgent to:

Department of Clinical Chemistry
Sheffield Children's NHS Foundation Trust
Western Bank, Sheffield
S10 2TH, UK

Jennifer Watkinson (Senior Biomedical Scientist)
0114 2717445

Reference Ranges

0 – 18 µmol/L male
0 – 16 µmol/L female

Reference from lab data and Personal Communication (L Spaapen, Stichting Klinische Genetica Limburg, Netherlands) and in house reference range. (N.B: these reference ranges are only valid with data produced by the Department of Clinical Chemistry, Sheffield Children's NHS Foundation Trust).

References

- Recommended approaches for the laboratory measurement of homocysteine in the diagnosis and monitoring of patients with hyperhomocysteinaemia. SJ Moat, JR Bonham, MS Tanner, JC Allen, HJ Powers. Ann Clin Biochem 1999; 36: 372-379.

- Vademecum Metabolicum. Diagnosis and Treatment of Inborn Errors of Metabolism. Zschoke/Hoffman 3rd Edition 2011