Homocysteine, Total Quantitative, Plasma

Test Code: HO
Turnaround time: 7 days - 10 days
CPT Codes: 82542 x1, 83090 x1, 83789 x1

Condition Description

Homocystinuria is an autosomal recessive metabolic disorder resulting from the body's inability to process the amino acids methionine and homocystine. The majority of cases of this condition are due to deficiency of the enzyme cystathionine synthase, which causes an accumulation of methionine, homocystine and various metabolites of homocystine.

Clinical manifestations vary in degree, type and age of onset. They include diffuse thromboembolism, dislocation of the optic lens, osteoporosis, tall stature due to thin, lengthened long bones, seizures, psychiatric disturbances, and mental retardation.

Indications

This test is indicated for:

- Individuals suspected to have homocystinuria, or hyperhomocysteinemia
- Individuals with premature vascular clotting or those patients with a positive family history (elevated homocysteine should be suspected in these cases).

Methodology

- High Performance Liquid Chromatography (HPLC)
- Liquid Chromatography-Mass Spectrometry (LC-MS/MS)

Detection

This test is very sensitive for homocysteine but should be interpreted in light of clinical symptoms and feeding status.

Reference Range

Click here for reference range.

Specimen Requirements

Type: Plasma

Specimen Requirements:

In EDTA (purple top) tube: 1-5 ml

Sample should be collected while fasting or 2-4 hours postprandial.

Centrifuge to separate plasma immediately (ideally within 30 minutes of collection) and freeze.

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

Special Instructions

Please indicate on the test requisition form any medications or dietary changes.

Related Tests

- Organic Acid Analysis (OA), Plasma Amino Acids (AA), and Urine Amino Acids (UA) are used in the diagnosis and evaluation of patients with metabolic conditions.
- Cystathionine Beta-Synthase Gene Sequencing (EY)