Amino Acid Profile, Quantitative, Plasma

Test Code: AA
Turnaround time: 7 days (STAT results: 1 day.)
CPT Codes: 82139 x1

Condition Description

Plasma amino acids analysis will detect specific amino acid disorders such as phenylketonuria (PKU), maple syrup urine disease (MSUD), urea cycle defects, non-ketotic hyperglycinemia and homocystinuria, etc. Plasma amino acids values are compared to age-matched normal values. Quantitative analysis of amino acids can also be performed to monitor established patients diagnosed with metabolic disorders.

Indications

This test is indicated in the case of:

- Infants with a positive newborn screening result indicative of a metabolic disorder.
- Evaluation of patients with signs of a possible metabolic condition, such as lethargy, vomiting, and failure to thrive.
- Monitoring for individuals diagnosed with a metabolic condition.

Methodology

Quantitative ion exchange chromatography, reported as micromoles/L.

Detection

This test is very sensitive for specific amino acid disorders, but detection can be sensitive to the clinical and nutritional status of the patient.

Specimen Requirements

Type: Plasma

Specimen Requirements:

In sodium heparin (green top) tube: 1-2 ml

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

Centrifuge to separate plasma and freeze.

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

Special Instructions

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

Related Tests

- [Organic Acids Quantitative Analysis (OA)](https://www.eglgenetics.com) is used in the diagnosis and evaluation of patients with metabolic conditions.