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. Urine amino acids analysis can evaluate renal function, some specific amino acid disorders, and renal transport disorders. Plasma and urine amino acids values are compared to age-matched normal values. Quantitative analysis of amino acids can also be performed on spinal fluid. Filter paper analysis is used to monitor established patients diagnosed with PKU and MSUD.

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 creatinine.

Detection

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

Specimen Requirements

Additional Specimen Collection/Handling Instructions Required for this Test
Samples submitted for the evaluation of glycine encephalopathy should be accompanied by a simultaneously obtained plasma sample for calculation of CFS/Plasma ratio.

Type: Spinal Fluid (CSF)

Specimen Requirements:

In a sterile container without additives: 1 ml
Freeze.

Note: Blood contamination can interfere with test results.

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 (QA)** is used in the diagnosis and evaluation of patients with metabolic conditions.