Phenylketonuria (Monitoring): Phenylalanine and Tyrosine, Quantitative, Dried Blood Spot

Test Code: FP
Turnaround time: 7 days
CPT Codes: 82136 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. 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.

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.

Reference Range

Call lab for reference ranges if needed.

Specimen Requirements

Type: Dried Blood Spot

Specimen Requirements:

Peripheral blood from finger prick or heel stick spotted on filter paper, completely saturating the circle. Air dry sample.

Specimen Collection and Shipping: Do not expose specimen to heat or direct sunlight. Keep the specimen dry. Ship sample at room temperature with overnight delivery.

Special Instructions

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

Related Tests

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