



آغا خان یونیورسٹی ہسپتال، کراچی

The Aga Khan University Hospital, Karachi

Stadium Road,
P.O. Box 3500, Karachi 74800, Pakistan
Fax: (92) 21 34934294, Telephone: 34930051

THE AGA KHAN UNIVERSITY HOSPITAL CLINICAL LABORATORIES

UPDATE PLASMA AMINO ACID (QUANTITATIVE)

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INTRODUCTION:

Quantitative amino acid analysis in plasma is critical for the diagnosis and monitoring of individuals with inborn error of amino acid metabolism. This test is appropriate for diagnostic confirmation following a positive newborn screen result or in individuals who present symptomatically with developmental delay, seizures, lethargy, vomiting, or failure to thrive. The test is used to confirm the diagnosis of aminoacidopathies such as urea cycle disorders, maple syrup urine disease, phenylketonuria, tyrosinemia types I & II, vitamin B12 (cobalamin) processing disorders, non-ketotic-hyperglycinemia, argininemia, hyperprolinemia, etc. Depending on the natural history of the disorder, symptoms may be minimized or prevented by early diagnosis and treatment.

PRINCIPLE:

The test is performed by Cation-Exchange HPLC (Biochrom 30+).

SPECIMEN COLLECTION:

- Fasting sample is required, at least 2 hours in infants, 4 hours in children and 8 hours in adults.
- Adult 3-4 ml of blood, (Lithium heparin containing tube, Green top) or 1-2 ml of separated plasma is required
- Infant or child 1-2 ml (Lithium heparin containing tube, Green top) or 0.5-1 ml of separated plasma is required.
- Separate the plasma immediately within 4 hours.
- **Minimum acceptable volume of separated plasma is 0.2 ml (200 µl).**
- Transport sample frozen in dry ice to the laboratory.

UNACCEPTABLE CONDITIONS: Severely hemolyzed specimens

SPECIAL PRECAUTION: Biohazard specimen; to be handled with care.

SCHEDULE:

Reporting will be 10 days after receiving the sample.

NOTE:

- It is essential to fill in the request form related to inborn error of metabolism (inherited metabolic disease) provided at the reception of AKU Clinical Laboratory, Collection Points and Consulting Clinics.
- Instruct patient's attendant (parents or guardians) to provide previous reports related to inherited metabolic disease if available.
- Encourage the patients to get their test charged against the initial laboratory number each time so that a laboratory record related to patient can be readily available.

PLEASE FILE FOR QUICK REFERENCE