

Plasma Amino Acids

Synonyms

PAA

Clinical Indication

Plasma amino acids can assist in the diagnosis of suspected inherited metabolic disorders. For example phenylketonuria, maple syrup urine disease, glycine encephalopathy, urea cycle defects and some organic acidurias.

Although not an exhaustive list, patients with a disorder of amino acid metabolism often present with non-specific symptoms such as hyperammonaemia, seizures, developmental delay, metabolic acidosis and hypoglycaemia.

Part of Profile / See Also

Request Form

Combined Pathology manual Blood form or ICE request.

Availability / Frequency of Analysis

Referred test: Analysed by Great Ormond Street Hospital (8692), if specific criteria met. Amino acid analysis by HPLC is adequate for the majority of disease types, ion exchange is reserved for the cases not reliably measured by HPLC technique.

Turnaround Time

3 weeks

Patient Preparation

Plasma amino acids fluctuate widely depending on the protein intake and whether the patient is in a fed or fasted state. Dietary restrictions may cause characteristic amino acid patterns to disappear and result in false negative results.

The most diagnostically useful sample is collected during acute illness if possible or when the patient is in crisis/metabolic decompensation.

If non-acute situation, a fasting sample is preferred (2-3 hrs post feed in infants).

Please provide clinical details including feeding regime, state if protein has been withdrawn and provide details of current medication.

Sample Requirements

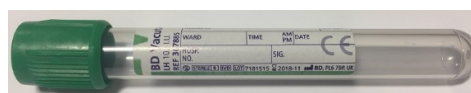
Specimen Type

Plasma (Serum is also acceptable)

Volume

1.0 ml

Container



Dark green top (lithium-heparin)



Or Paediatric lithium heparin (Orange top – Sarstedt tube)



Or Paediatric lithium heparin (Pale green top – BD Microtainer tube).

Samples should be transported to laboratory immediately.

Reference Range & Units

Report will contain an interpretive comment. Individual amino acid concentrations are only reported when clinically relevant/significant.

Interferences
Interpretation & Clinical
Decision Value (if applicable)

Provided by the referral laboratory

References
Test code

PAA

Lab Handling

Centrifuge and aliquot plasma/serum ideally within 30 minutes of collection but no longer than 2 hours post collection into 2 tubes and freeze at -20°C. Ensure the sample type is written on the aliquot. Samples sent frozen to GOSH.



8692
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 ISO 15189:2012**