AMINO ACIDS – EYE FLUID (quantitative)

Relevant disorders

Phenylketonuria, maple syrup urine disease, urea cycle disorders, organic acidaemias, non ketotic hyperglycinaemia, Disorders of amino acid transport including cystinuria, Hartnup disease, Lysinuric protein intolerance

Related Metabolic Tests

Quantitative orotic acid, qualitative organic acids

Indication for Test

Amino acids are the individual building blocks of peptides and proteins. A number of inborn errors of metabolism affect the catabolic or synthetic pathways of these amino acids and therefore analysis of the amino acids in a sample of plasma, urine or CSF can be used to diagnose disorders such as PKU, maple syrup urine disease, cystinuria, non-ketotic hyperglycinaemia and the urea cycle disorders. Amino acid analysis is also used for monitoring the efficacy of treatment in patients with these disorders, as well as patients on low protein diets for other reasons (e.g. those with organic acidaemias).

Methodology

Quantitative amino acid analysis is performed on a Biochrom amino acid analyser which uses a cation exchange resin and Ninhydrin detection.

Sample requirements

Plain sample (post mortem)

Turn Around Time

5 – 14 days (but if requested urgently can be performed more quickly).

Transport information/Contact details

Send all samples by first class post or courier if urgent to:

Department of Clinical Chemistry
Sheffield Children's NHS Foundation Trust
Western Bank, Sheffield
S10 2TH, UK

Simon Olpin (Consultant Clinical Scientist)
0114 2717267

Reference Ranges

Interpretation will be provided with the report.

References

- A Clinical Guide to Inherited Metabolic Diseases. JTR Clarke
- Vadecum Metabolicum Manual of Metabolic Paediatrics Zschoche/Hoffman
- The Molecular and Metabolic Basis of Inherited Disease. Eighth Edition 2001. Scriver et al.