AMINO ACIDS - URINE (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
Amino acids -plasma

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

5ml Urine in a plain universal container (either a random sample or an aliquot of a 24 hour collection).

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

Joanne Croft (Clinical Scientist)
0114 2717307

Reference ranges

Urine amino acid ref ranges µmol/mmol creatinine (N.B: these reference ranges are only valid with data produced by the Department of Clinical Chemistry, Sheffield Children's NHS Foundation Trust).

Age	0-1m	1-6m	6m-2yr	2-10yr	10-13yr	>13yr
TAU	8-226	6-89	9-159	13-230	18-176	16-160
ASP	2-12	2-16	3-12	1-9	1-9	2-7
THR	20-138	17-92	14-62	9-48	8-28	7-29
SER	50-282	42-194	45-137	23-94	23-67	21-50
ASN	0-84	0-58	0-36	0-30	0-18	0-23
GLU	0-30	0-29	0-18	0-10	0-9	0-12
GLN	52-205	63-229	62-197	31-236	20-112	20-76
PRO	9-213	0-130	0-14	0-9	0-9	0-9
GLY	165-1097	210-743	110-445	84-362	64-165	43-173
CIT	0-11	0-10	0-8	0-6	0-5	0-4
ALA	62-244	62-206	36-162	17-115	21-62	16-68
BAIB	0-87	0-216	0-266	0-175	0-58	0-91
CYS	23-78	13-48	10-29	8-30	7-23	6-34
VAL	3-26	4-19	6-21	3-20	3-17	3-13
MET	7-27	6-22	7-29	3-21	3-10	2-16
ISO	0-6	0-5	0-6	0-6	0-6	0-4
LEU	3-25	4-12	3-17	3-18	3-14	2-11
TYR	6-55	12-52	11-54	7-35	6-25	2-23
PHE	4-32	7-28	10-31	5-26	5-17	2-19
HIS	80-295	92-278	87-287	45-255	43-159	26-153
3MHIS	20-39	19-40	20-57	18-61	20-56	19-47
ORN	0-19	0-13	0-8	0-7	0-6	0-5
LYS	22-171	15-199	13-79	10-68	10-56	7-58
ARG	0-14	0-11	0-11	0-9	0-6	0-5

References

- A Clinical Guide to Inherited Metabolic Diseases. JTR Clarke
- Vademecum Metabolicum, Diagnosis and Treatment of Inborn Errors of Metabolism.
- The Molecular and Metabolic Basis of Inherited Disease. Eighth Edition 2001. Scriver et al.