AMINO ACIDS - PLASMA (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

Amino acids – CSF and Urine
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

1 ml lithium heparin plasma (fluoride and serum are acceptable)

NB samples for free homocystine and cystine must be separated and deproteinised within 30 minutes of venepuncture.

Transport information/Contact details

Send by first class post Monday – Thursday with normal packaging, or courier if urgent.

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

Joanne Croft (Clinical Scientist)
0114 2717307

Turn Around Time

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

Reference Ranges (age dependent)

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

Age	<7d	7d-6m	6m-2yr	2-10yr	10-17yr	adult
TAU	40-420	0-360	0-250	20-260	57-242	75-225
ASP	20-70	0-120	0-110	0-80	5-85	10-90
THR	10-400	60-360	20-280	40-180	45-240	50-300
SER	50-350	60-300	40-280	60-240	68-220	75-200
ASN	0-175	15-95	0-110	0-87	15-84	30-80
GLU	0-250	20-280	0-300	0-200	5-150	10-100
GLN	487-1031	392-988	550-830	550-830	550-830	413-690
PRO	50-450	125-375	75-450	66-333	83-341	10-350
GLY	200-600	140-420	100-425	120-480	110-465	100-450
CIT	0-40	0-55	10-60	0-54	0-57	0-60
ALA	100-800	200-700	100-800	150-650	175-625	200-600
BAIB	-	0-45	3-41	0-36	0-33	0-30
CYS	36-100	24-100	34-105	45-80	46-85	48-90
VAL	50-400	100-400	75-387	50-375	95-338	140-300
MET	0-80	15-65	0-50	0-54	8-47	15-40
ISO	0-150	23-143	20-150	0-135	20-123	40-110
LEU	20-280	72-240	40-260	60-260	70-170	80-180
TYR	30-135	13-200	20-160	30-130	25-125	20-110
PHE	40-110	32-128	40-140	20-130	30-115	40-100
HIS	45-150	32-144	35-130	50-130	30-130	10-130
TRY	0-50	10-95	7-86	15-85	12-83	10-80
ORN	25-225	30-285	20-200	40-160	35-155	30-150
LYS	105-315	100-360	40-280	50-233	85-237	120-240
ARG	10-70	0-135	0-120	0-160	0-150	0-140

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.