MAYO CLINIC LABORATORIES

Test Definition: AAPD

Amino Acids, Quantitative, Random, Urine

Overview

Useful For

Evaluating patients with possible inborn errors of metabolism using random urine specimens

May aid in evaluation of endocrine disorders, liver diseases, muscle diseases, neoplastic diseases, neurological disorders, nutritional disturbances, renal failure, and burns

Testing Algorithm

Testing includes quantitation of the following amino acids: phosphoserine, phosphoethanolamine, taurine, threonine, serine, asparagine, hydroxyproline, glutamic acid, glutamine, aspartic acid, ethanolamine, sarcosine, proline, glycine, alanine, citrulline, alpha-aminoadipic acid, alpha-amino-n-butyric acid, valine, cystine, cystathionine, methionine, isoleucine, leucine, tyrosine, phenylalanine, beta-alanine, beta-aminoisobutyric acid, ornithine, lysine, 1-methylhistidine, histidine, 3-methylhistidine, carnosine, anserine, argininosuccinic acid, allo-isoleucine, homocitrulline, gamma-amino-n-butyric acid, hydroxylysine, tryptophan, and arginine.

Method Name

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)
Portions of this test are covered by patents held by Quest Diagnostics

NY State Available

Yes

Specimen

Specimen Type

Urine

Additional Testing Requirements

Not all patients with homocystinuria will be detected by this assay. If homocystinuria is a concern, order HCYSP / Homocysteine, Total, Plasma or HCYSS / Homocysteine, Total, Serum in tandem with this test.

Necessary Information

1. Patient's age is required.
2. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information.
3. If prolidase deficiency is a concern, indicate on the amino acid order "Pretreat with acid hydrolysis prior to analysis". The acid hydrolysis will break up in vitro proline and hydroxyproline containing dipeptides, which are cleaved *in vivo* by prolidase.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)
Specimen Volume: 2 mL

Collection Instructions: Collect a random urine specimen.

Forms

If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

1 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen (preferred)	70 days	
	Refrigerated	14 days	

Clinical & Interpretive

Clinical Information

Amino acids are the basic structural units that comprise proteins and are found throughout the body. Many inborn errors of amino acid metabolism that affect amino acid transport or metabolism have been identified, such as phenylketonuria and tyrosinemia. Amino acid disorders can manifest at any age, but most become evident in infancy or early childhood. These disorders result in the accumulation or the deficiency of 1 or more amino acids in biological fluids, which leads to the clinical signs and symptoms of the specfic amino acid disorder.

The clinical presentation is dependent upon the specific amino acid disorder. In general, affected patients may experience failure to thrive, neurologic symptoms, digestive problems, dermatologic findings, and physical and cognitive delays. If not diagnosed and treated promptly, amino acid disorders can result in intellectual disabilities and, possibly, death.

In addition, amino acid analysis may have clinical importance in the evaluation of several acquired conditions including endocrine disorders, liver diseases, muscle diseases, neoplastic diseases, neurological disorders, nutritional disturbances, kidney failure, and burns. General elevations in urine amino acid levels, called aminoaciduria, can be seen in disorders with amino acid transport defects, such as lysinuric protein intolerance and Hartnup disease, as well as in conditions with renal tubular dysfunction including Lowe syndrome and Dent disease.

Reference Values

Amino Acid		Age groups					
		< or =12 months	13-35 months	3-6 years	7-8 years	9-17 years	> or =18 years
Phosphoserine	PSer	<1	<1	<1	<1	<1	<1

Phosphoethanolamine	PEtN	15-341	33-342	19-164	12-118	<88	<48
Taurine	Tau	37-8300	64-3255	76-3519	50-2051	57-2235	24-1531
Asparagine	Asn	25-1000	62-884	28-412	38-396	22-283	25-238
Serine	Ser	18-4483	284-1959	179-1285	153-765	105-846	97-540
Hydroxyproline	Hyp	<2536	<89	<46	<19	<22	<15
Glycine	Gly	362-18614	627-6914	412-5705	449-4492	316-4249	229-2989
Glutamine	Gln	139-2985	263-2979	152-1325	164-1125	188-1365	93-686
Aspartic Acid	Asp	<64	<56	<30	<9	<11	<10
Ethanolamine	EtN	282-3782	256-947	193-643	137-564	158-596	95-471
Histidine	His	145-3833	427-3398	230-2635	268-2147	134-1983	81-1128
Threonine	Thr	25-1217	55-763	30-554	25-456	37-418	31-278
Citrulline	Cit	<72	<57	<14	<9	<14	<12
Sarcosine	Sar	<75	<12	<9	<2	<3	<3
Beta-Alanine	bAla	<219	<92	<25	<25	<49	<52
Alanine	Ala	93-3007	101-1500	64-1299	44-814	51-696	56-518
Glutamic Acid	Glu	<243	12-128	<76	<39	<62	<34
1-Methylhistidine	1MHis	17-419	18-1629	10-1476	19-1435	12-1549	23-1339
3-Methylhistidine	3MHis	88-350	86-330	56-316	77-260	47-262	70-246
Argininosuccinic Acid	Asa	<77	<48	<37	<24	<69	<15
Carnosine	Car	27-1021	16-616	18-319	<161	<109	<35
Anserine	Ans	<277	<820	<398	<141	<369	<38
Homocitrulline	Hcit	<295	11-158	<71	<62	<33	<30
Arginine	Arg	10-560	20-395	14-240	<134	<153	<114
Alpha-aminoadipic Acid	Aad	10-275	15-324	10-135	<84	<76	<47
Gamma Amino-n-butyric Acid	GABA	<25	<13	<11	<6	<5	<5
Beta-aminoisobutyric Acid	bAib	18-3137	<980	15-1039	24-511	11-286	<301
Alpha-amino-n-butyric Acid	Abu	<63	<56	<38	<30	<31	<19
Hydroxylysine	Hyl	<150	<57	<34	<26	<31	<12
Proline	Pro	28-2029	<119	<78	<20	<28	<26
Ornithine	Orn	<265	<70	<44	<17	<18	<25
Cystathionine	Cth	<302	<56	<26	<18	<44	<30
Cystine	Cys	12-504	11-133	<130	<56	<104	10-98
Lysine	Lys	19-1988	25-743	14-307	17-276	10-240	15-271
Methionine	Met	<41	<41	<25	<23	<20	<16
Valine	Val	11-211	11-211	<139	16-91	<75	11-61
Tyrosine	Tyr	39-685	38-479	23-254	22-245	12-208	15-115
Isoleucine	Ile	<86	<78	<62	<34	<28	<22

Leucine	Leu	<200	15-167	12-100	13-73	<62	<51
Phenylalanine	Phe	14-280	34-254	20-150	21-106	11-111	13-70
Tryptophan	Trp	14-315	14-315	10-303	10-303	15-229	18-114
Allo-isoleucine	AlloIle	<29	<10	<8	<8	<8	<7

All results reported as nmol/mg creatinine.

Interpretation

When no significant abnormalities are detected, a simple descriptive interpretation is provided. When abnormal results are detected, a detailed interpretation is given, including an overview of the results and of their significance, a correlation to available clinical information, elements of differential diagnosis, recommendations for additional biochemical testing and in vitro confirmatory studies (enzyme assay, molecular analysis), name and phone number of key contacts who may provide these studies, and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

Not all patients with homocystinuria and prolidase deficiency will be detected by this assay. See Additional Testing Requirements and Necessary Information for more information.

Clinical Reference

1. Part 8: Amino Acids. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill, 2019. Accessed September 9, 2022. Available at https://ommbid.mhmedical.com/book.aspx?bookID=2709#225069340
2. Camargo SMR, Bockenhauer D, Kleta R: Aminoacidurias: Clinical and molecular aspects. Kidney Int. 2008 Apr;73(8):918-925. doi: 10.1038/sj.ki.5002790
3. Duran M: Amino acids. In: Blau N, Duran M, Gibson KM, eds. Laboratory Guide to the Methods in Biochemical Genetics. Springer-Verlag; 2008:53-89

Performance

Method Description

Quantitative analysis of amino acids is performed by liquid chromatography tandem mass spectrometry (LC-MS/MS) by labeling amino acids present in plasma, urine, and cerebrospinal fluid with aTRAQ Reagent 121. Samples are dried and reconstituted with aTRAQ Reagent 113-labeled standard mix. Amino acids are separated and detected by LC-MS/MS. The concentrations of amino acids are established by comparison of their ion intensity (121-labeled amino acids) to that of their respective internal standards (113-labeled amino acids).(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available
3 to 5 days

Specimen Retention Time
2 weeks

Performing Laboratory Location
Rochester

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
82139

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
AAPD	Amino Acids, QN, Random, U	35087-6

Result ID	Test Result Name	Result LOINC® Value
3452	Taurine	28595-7
3456	Asparagine	28603-9
3455	Serine	30058-2
3460	Glycine	30066-5
3457	Glutamine	30056-6
3477	Histidine	30047-5
3454	Threonine	30057-4
3459	Citrulline	30161-4
3472	Beta-alanine	28588-2
3461	Alanine	30068-1
3458	Glutamic Acid	30059-0
3476	1-Methylhistidine	28606-2
3478	3-Methylhistidine	28594-0
3479	Carnosine	28597-3
3480	Arginine	30062-4

3462	Alpha-aminoadipic Acid	28598-1
3473	Beta-aminoisobutyric Acid	28602-1
3463	Alpha-amino-n-butyric Acid	28590-8
3483	Proline	30067-3
3474	Ornithine	30049-1
3466	Cystathionine	28599-9
3465	Cystine	30065-7
3475	Lysine	30048-3
3467	Methionine	30063-2
3464	Valine	30064-0
3470	Tyrosine	30054-1
3468	Isoleucine	30052-5
3469	Leucine	30053-3
3471	Phenylalanine	30055-8
3481	Interpretation (AAPD)	49248-8
34466	Phosphoserine	28600-5
34467	Phosphoethanolamine	28604-7
34477	Hydroxyproline	28601-3
34478	Aspartic Acid	30061-6
34479	Ethanolamine	28605-4
34480	Sarcosine	28610-4
34481	Argininosuccinic Acid	32229-7
34482	Anserine	28596-5
34483	Homocitrulline	32248-7
34484	Gamma-amino-n-butyric Acid	28593-2
34485	Hydroxylysine	30050-9
34486	Tryptophan	28608-8
34487	Allo-isoleucine	73908-6
113130	Reviewed By	18771-6