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, kidney failure, and burns

Testing Algorithm

Testing includes quantitation of the following amino acids: 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, 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 CMMPP / Cobalamin, Methionine, and Methylmalonic Acid Pathways, Plasma or 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 specific 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	Abbr	<2 months	2-35 months	3-6 years	7-17 years	> or =18 years
Taurine	Tau	<9026	<5604	<3680	<3954	<2134
Asparagine	Asn	<687	<1159	<238	<322	<204

Serine	Ser	<3202	<2741	<1386	<1116	<658
Hydroxyproline	Hyp	<2254	<1775	<89	<42	<48
Glycine	Gly	<13336	<9872	<4997	<4467	<4068
Glutamine	Gln	<2931	<3681	<1124	<1038	<764
Aspartic Acid	Asp	<290	<72	<46	<13	<12
Ethanolamine	EtN	<1589	<1765	<776	<536	<500
Histidine	His	<3806	<4578	<2596	<2294	<1508
Threonine	Thr	<1930	<1769	<496	<575	<384
Citrulline	Cit	<61	<67	<23	<17	<34
Sarcosine	Sar	<177	<52	<6	<10	<6
Beta-Alanine	bAla	<174	<79	<99	<139	<58
Alanine	Ala	<2856	<2360	<961	<794	<472
Glutamic Acid	Glu	<281	<210	<127	<63	<62
1-Methylhistidine	1MHis	<493	<470	<517	<453	<265
3-Methylhistidine	3MHis	<196	<662	<3346	<2027	<1920
Argininosuccinic Acid	Asa	<96	<103	<65	<57	<37
Homocitrulline	Hcit	<164	<169	<110	<107	<87
Arginine	Arg	<250	<147	<81	<42	<65
Alpha-aminoadipic Acid	Aad	<264	<397	<349	<138	<73
Gamma Amino-n-butyric Acid	GABA	<8	<14	<4	<3	<4
Beta-aminoisobutyric Acid	bAib	<2214	<2488	<840	<251	<265
Alpha-amino-n-butyric Acid	Abu	<58	<44	<37	<29	<19
Hydroxylysine	Hyl	<101	<87	<44	<22	<16
Proline	Pro	<1776	<1028	<29	<42	<98
Ornithine	Orn	<344	<238	<228	<291	<193
Cystathionine	Cth	<118	<53	<23	<24	<33
Cystine	Cys	<486	<285	<111	<84	<142
Lysine	Lys	<2217	<1321	<814	<463	<295
Methionine	Met	<76	<45	<24	<22	<15
Valine	Val	<308	<258	<119	<91	<73
Tyrosine	Tyr	<453	<603	<289	<371	<155
Isoleucine	Ile	<99	<131	<52	<29	<26
Leucine	Leu	<286	<215	<87	<70	<64
Phenylalanine	Phe	<342	<306	<178	<123	<98
Tryptophan	Trp	<241	<329	<222	<218	<140
Allo-isoleucine	AlloIle	<3	<6	<1	<2	<4

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

This assay does not measure total homocysteine, phosphoethanolamine, or imidodipeptides. Therefore, this assay should not be used as a test for homocystinuria, hypophosphatasia, and prolidase deficiency. See Additional Testing Requirements and Necessary Information for more information or contact a biochemical genetics counselor at 800-533-1710.

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 October 24, 2024. Available at https://ommbid.mhmedical.com/book.aspx?bookID=2709#225069340

2. Pasquali M, Longo N. Amino acids. In: Blau N, Dionisi Vici C, Ferreira CR, Vianey-Saban C, van Karnebeek CDM, eds. Physician's Guide to the Diagnosis, Treatment and Follow-up of Inherited Metabolic Diseases. 2nd ed. Springer-Verlag; 2022:41-50

Performance

Method Description

Quantitative analysis of amino acids is performed by liquid chromatography-tandem mass spectrometry. Patient samples are combined with isotopically labeled internal standard. Following protein precipitation, the supernatant is subjected to hydrophilic-interaction liquid chromatography for the separation of isomers with MS/MS detection of the underivatized 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

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
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
34477	Hydroxyproline	28601-3
34478	Aspartic Acid	30061-6
34479	Ethanolamine	28605-4
34480	Sarcosine	28610-4
34481	Argininosuccinic Acid	32229-7
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