MAYO CLINIC LABORATORIES

Test Definition: AAQP

Amino Acids, Quantitative, Plasma

Overview

Useful For

Evaluating patients with possible inborn errors of metabolism using plasma 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, glutamic acid, glutamine, proline, alanine, citrulline, alpha-amino-n-butyric acid, valine, cystine, methionine, isoleucine, leucine, tyrosine, phenylalanine, beta-alanine, ornithine, lysine, histidine, argininosuccinic acid, allo-isoleucine, arginine, phosphoserine, phosphoethanolamine, hydroxyproline, glycine, aspartic acid, ethanolamine, sarcosine, 1-methylhistidine, 3-methylhistidine, carnosine, anserine, homocitrulline, alpha-aminoadipic acid, gamma-amino-n-butyric acid, beta-aminoisobutyric acid, hydroxylysine, cystathionine, and tryptophan.

For more information see Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm.

Special Instructions

- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

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

Plasma

Additional Testing Requirements

Not all patients with homocystinuria/homocystinemia will be detected by this assay. If there is a concern for homocystinemia, order HCYSP / Homocysteine, Total, Plasma in conjunction with this amino acids profile.

Shipping Instructions

Send plasma frozen.

Necessary Information

1. Patient's age is required.

2. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information.

Specimen Required

Patient Preparation: Fasting (overnight preferred, 4 hours minimum). Infants should be drawn just before next feeding (2-3 hours without total parenteral nutrition, if possible).

Collection Container/Tube:

Preferred: Green top (sodium heparin)

Acceptable: Lavender top (EDTA), plasma gel tube, green top (lithium heparin)

Submission Container/Tube: Plastic vial

Specimen Volume: 0.5 mL

Collection Instructions:

1. Collect specimen and place on wet ice. Note: Thrombin-activated tubes **should not be used** for collection.
2. Centrifuge immediately or within 4 hours of collection if the specimen is kept at refrigerated temperature.
3. Being careful to ensure that no buffy coat is transferred, aliquot plasma into a plastic vial and freeze.

Forms

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

Specimen Minimum Volume

0.3 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen	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, such as phenylketonuria and tyrosinemia, have been identified. 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.

Treatment for amino acid disorders includes very specific dietary modifications. Nonessential amino acids are synthesized by the body, while essential amino acids are not and must be obtained through an individual's diet. Therapeutic diets are coordinated and closely monitored by a dietician or physician. They are structured to provide the necessary balance of amino acids with particular attention to essential amino acids and those that accumulate in a particular disorder. Patients must pay close attention to the protein content in their diet and generally need to supplement with medical formulas and foods. Dietary compliance is monitored by periodic analysis of plasma amino acids.

In addition, plasma 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.

Reference Values

Amino acids	Age groups		
	<24 months	2-17 years	> or =18 years
Phosphoserine (PSer)	<109	<95	<18
Phosphoethanolamine (PEtN)	<6	<5	<12
Taurine (Tau)	37-177	38-153	42-156
Asparagine (Asn)	25-91	29-87	37-92
Serine (Ser)	69-271	71-208	63-187
Hydroxyproline (Hyp)	8-61	7-35	4-29
Glycine (Gly)	111-426	149-417	126-490
Glutamine (Gln)	316-1,020	329-976	371-957
Aspartic Acid (Asp)	2-20	<11	<7
Ethanolamine (EtN)	<70	<64	<67
Histidine (His)	10-116	12-132	39-123
Threonine (Thr)	47-237	58-195	85-231
Citrulline (Cit)	9-38	11-45	17-46
Sarcosine (Sar)	<5	<5	<5
b-Alanine (bAla)	<28	<27	<29
Alanine (Ala)	139-474	144-557	200-579
Glutamic Acid (Glu)	31-202	22-131	13-113
1-Methylhistidine (1MHis)	<11	<20	<28
3-Methylhistidine (3MHis)	<1	<1	2-9
Argininosuccinic Acid (Asa)	<2	<2	<2
Carnosine (Car)	<13	<1	<1
Anserine (Ans)	<1	<1	<1
Homocitruline (Hcit)	<5	<2	<2
Arginine (Arg)	29-134	31-132	32-120
a-Aminoadipic Acid (Aad)	<4	<3	<3
g-Amino-n-butyric Acid (GABA)	<4	<3	<2

b-Aminoisobutyric Acid (bAib)	<9	<5	<5
a-Amino-n-butyric Acid (Abu)	7-28	7-31	9-37
Hydroxylysine (Hyl)	<4	<3	<2
Proline (Pro)	85-303	80-357	97-368
Ornithine (Orn)	20-130	22-97	38-130
Cystathionine (Cth)	<2	<2	<5
Cystine (Cys)	2-32	2-36	3-95
Lysine (Lys)	49-204	59-240	103-255
Methionine (Met)	11-35	11-37	4-44
Valine (Val)	83-300	106-320	136-309
Tyrosine (Tyr)	26-115	31-106	31-90
Isoleucine (IIe)	31-105	30-111	36-107
Leucine (Leu)	48-175	51-196	68-183
Phenylalanine (Phe)	28-80	30-95	35-80
Tryptophan (Trp)	17-75	23-80	29-77
Alloisoleucine (Allolle)	<2	<3	<5

All results reported in nmol/mL

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 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

Reference values are for fasting patients.

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 Sep 9, 2022 Available at https://ommbid.mhmedical.com/book.aspx?bookID=2709#225069340

2. 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 spinal 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-MSMS. 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
AAQP	Amino Acids, QN, P	35083-5

Result ID	Test Result Name	Result LOINC® Value
3512	Taurine	20657-3
3517	Asparagine	20638-3
3516	Serine	20656-5
3522	Glycine	20644-1
3518	Glutamine	20643-3
3535	Histidine	20645-8

3515	Threonine	20658-1
3521	Citrulline	20640-9
3532	Beta-Alanine	26604-9
3523	Alanine	20636-7
3520	Glutamic Acid	20642-5
32341	Argininosuccinic Acid	32227-1
3536	Arginine	20637-5
3524	Alpha-amino-n-butyric Acid	20634-2
3519	Proline	20655-7
3533	Ornithine	20652-4
3526	Cystine	22672-0
3534	Lysine	20650-8
3527	Methionine	20651-6
3525	Valine	20661-5
3530	Tyrosine	20660-7
3528	Isoleucine	20648-2
3529	Leucine	20649-0
3531	Phenylalanine	14875-9
3570	Interpretation (AAQP)	49247-0
32347	Allo-isoleucine	22670-4
34449	Phosphoserine	20654-0
34450	Alpha-aminoadipic Acid	26600-7
34451	Cystathionine	26607-2
34452	Beta-aminoisobutyric Acid	26605-6
34453	1-Methylhistidine	20633-4
34454	3-Methylhistidine	20635-9
34455	Carnosine	26606-4
34456	Homocitrulline	55876-7
34457	Phosphoethanolamine	26612-2
34458	Hydroxyproline	20647-4
34459	Aspartic Acid	20639-1
34460	Ethanolamine	26608-0
34461	Sarcosine	26613-0
34462	Anserine	26599-1
34463	Gamma-amino-n-butyric Acid	26609-8
34464	Hydroxylysine	26610-6
34465	Tryptophan	20659-9