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

Monitoring of patients treated for various inborn errors of metabolism or other causes of amino acid imbalances

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, hydroxyproline, glycine, aspartic acid, ethanolamine, sarcosine, 1-methylhistidine, 3-methylhistidine, 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

Ordering Guidance

Body fluids are not acceptable specimens for this test.
For testing urine specimens, order AAPD / Amino Acids, Quantitative, Random, Urine.
For testing spinal fluid specimens, order AACSF / Amino Acids, Quantitative, Spinal Fluid.

Additional Testing Requirements

Not all patients with homocystinuria/homocystinemia will be detected by this assay. For quantitation of total homocysteine, order CMMPP / Cobalamin, Methionine, and Methylmalonic Acid Pathways, Plasma or HCYSP / Homocysteine, Total, Plasma in conjunction with this amino acids profile.

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: Patient should fast a minimum of 4 hours; infants should have specimen collected before next feeding (2-3 hours without total parenteral nutrition if possible).

Supplies: Sarstedt Aliquot Tube, 5 mL (T914)

Collection Container/Tube: Green top (sodium 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 are abnormal 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
Taurine (Tau)	31-354	32-181	21-123
Asparagine (Asn)	18-94	25-80	23-94
Serine (Ser)	59-224	53-166	55-146
Hydroxyproline (Hyp)	<121	<73	<38
Glycine (Gly)	80-500	80-500	80-500
Glutamine (Gln)	356-857	353-790	447-774
Aspartic Acid (Asp)	<48	<17	<13
Ethanolamine (EtN)	<70	<30	<20
Histidine (His)	46-147	56-119	61-120
Threonine (Thr)	49-358	48-205	73-325
Citrulline (Cit)	8-42	12-44	18-57
Sarcosine (Sar)	<20	<20	<20
b-Alanine (bAla)	<36	<36	<36
Alanine (Ala)	139-474	144-557	200-579
Glutamic Acid (Glu)	28-376	16-182	13-148
1-Methylhistidine (1MHis)	<12	<12	<12
3-Methylhistidine (3MHis)	<11	<30	<35
Argininosuccinic Acid (Asa)	<5	<5	<5
Homocitruline (Hcit)	<5	<2	<2
Arginine (Arg)	28-164	28-156	45-144
a-Aminoadipic Acid (Aad)	<4	<4	<4
g-Amino-n-butyric Acid (GABA)	<4	<4	<4

b-Aminoisobutyric Acid (bAib)	<9	<5	<5
a-Amino-n-butyric Acid (Abu)	<40	<40	<40
Hydroxylysine (Hyl)	<4	<4	<4
Proline (Pro)	102-342	99-389	107-383
Ornithine (Orn)	32-171	32-148	39-154
Cystathionine (Cth)	<4	<4	<4
Cystine (Cys)	6-131	3-151	8-310
Lysine (Lys)	83-304	61-291	105-335
Methionine (Met)	12-57	13-41	13-40
Valine (Val)	94-382	111-367	134-357
Tyrosine (Tyr)	27-188	36-133	36-113
Isoleucine (IIe)	23-149	26-150	29-153
Leucine (Leu)	59-213	51-216	79-217
Phenylalanine (Phe)	36-105	38-116	45-106
Tryptophan (Trp)	12-103	21-114	21-108
Alloisoleucine (Allolle)	<5	<5	<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.

Patients with phenylketonuria (PKU) who are treated with pegvaliase-pqpz (Palynziq) may have false low phenylalanine concentrations due to persistent activity in collected blood specimens.

This assay does not measure total homocysteine. If relevant disorders are considered, also order CMMPP / Cobalamin, Methionine, and Methylmalonic Acid Pathways, Plasma or HCYSP / Homocysteine, Total, Plasma analysis.

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 (LC-MS/MS). 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
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
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
34456	Homocitrulline	55876-7
34458	Hydroxyproline	20647-4
34459	Aspartic Acid	20639-1
34460	Ethanolamine	26608-0
34461	Sarcosine	26613-0
34463	Gamma-amino-n-butyric Acid	26609-8
34464	Hydroxylysine	26610-6
34465	Tryptophan	20659-9