Test Definition: AACSF

Amino Acids, Quantitative, Spinal Fluid

Overview

Useful For

Evaluating patients with possible inborn errors of amino acid metabolism, particularly nonketotic hyperglycinemia (glycine encephalopathy) and serine biosynthesis defects, especially when used in conjunction with concomitantly collected plasma specimens

Testing Algorithm

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

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

CSF

Additional Testing Requirements

This test should be ordered in conjunction with AAQP / Amino Acids, Quantitative, Plasma. The specimens for both tests (AAQP / Amino Acids, Quantitative, Plasma and this test) should be collected at the same time.

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

Container/Tube: Sterile vial
Specimen Volume: 0.2 mL
Collection Instructions: Collect specimen from second collection vial.

Forms

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

Specimen Minimum Volume

0.1 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
CSF	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 that affect amino acid transport and metabolism 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 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.

Cerebrospinal fluid (CSF) specimens are highly informative for a subset of these conditions, such as nonketotic hyperglycinemia and serine biosynthesis defects. CSF specimens are most informative when a plasma specimen is collected at the same time, and the ratio of the amino acid concentrations in CSF to those in plasma is calculated.

Reference Values

Amino Acid	< or =31 days	32 days-23 months	2-18 years	> or =19 years
Taurine (Tau)	8-28	4-13	4-10	3-9
Asparagine (Asn)	7-25	5-17	4-12	5-14
Serine (Ser)	43-127	37-87	22-57	18-58
Hydroxyproline (Hyp)	<12	<8	<3	<3

Glycine (Gly)	<60	<27	<20	<28
Glutamine (Gln)	447-1547	384-716	375-770	452-1283
Aspartic Acid (Asp)	<12	<12	<12	<12
Ethanolamine (EtN)	11-152	6-41	7-25	6-24
Histidine (His)	19-63	12-32	9-26	12-36
Threonine (Thr)	35-212	19-89	13-51	13-57
Citrulline (Cit)	<6	<5	<4	<4
Sarcosine (Sar)	<21	<21	<21	<21
Beta-alanine (bAla)	<17	<17	<17	<17
Alanine (Ala)	20-92	18-69	16-54	22-80
Glutamic Acid (Glu)	<12	<4	<3	<2
1-Methylhistidine (1MHis)	<5	<1	<1	<3
3-Methylhistidine (3MHis)	<4	<1	<3	<5
Argininosuccinic Acid (Asa)	<4	<4	<4	<4
Homocitrulline (Hcit)	<1	<1	<1	<1
Arginine (Arg)	7-37	11-36	13-30	14-32
Alpha-aminoadipic Acid (Aad)	<2	<2	<2	<2
Gamma-amino-n-butyric Acid (GABA)	<10	<10	<10	<10
Beta-aminoisobutyric Acid (bAib)	<1	<1	<1	<1
Alpha-amino-n-butyric Acid (Abu)	<21	<7	<5	<10
Hydroxylysine (Hyl)	<1	<1	<1	<1
Proline (Pro)	<14	<4	<4	<2
Ornithine (Orn)	<32	<15	<12	<15
Cystathionine (Cth)	<4	<1	<1	<2
Cystine (Cys)	<3	<3	<3	<3
Lysine (Lys)	16-67	17-41	13-45	23-54
Methionine (Met)	<19	<7	<5	<12
Valine (Val)	16-83	10-36	10-27	13-52
Tyrosine (Tyr)	<70	<22	<16	<65
Isoleucine (Ile)	2-30	2-14	3-11	3-17
Leucine (Leu)	14-72	9-25	8-23	10-53
Phenylalanine (Phe)	9-49	7-18	5-18	8-23
Tryptophan (Trp)	<14	<14	<14	<14
Allo-isoleucine (AlloIle)	<2	<2	<2	<2

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 provided. This interpretation includes 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 the telephone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

Proper specimen collection and handling are crucial to achieve reliable results.

Blood contamination can interfere with test results.

Clinical Reference

1. Rinaldo P, Hahn S, Matern D. Inborn errors of amino acid, organic acid, and fatty acid metabolism. In: Burtis CA, Ashwood ER, Bruns DE. Tietz Textbook of Clinical Chemistry and Molecular Diagnosis. 4th ed. WB Saunders Company; 2005:2207-2247

2. Van Hove JLK, Coughlin C II, Swanson M, et al. Nonketotic hyperglycinemia. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2002. Updated May 23, 2019. Accessed October 24, 2024. Available at www.ncbi.nlm.nih.gov/books/NBK1357/

3. El-Hattab AW. Serine biosynthesis and transport defects. Mol Genet Metab. 2016;118(3):153-159. doi:10.1016/j.ymgme.2016.04.010

4. 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 (AA) 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

Mayo Clinic Laboratories - Rochester Main Campus

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
AACSF	Amino Acids, QN, CSF	35507-3

Result ID	Test Result Name	Result LOINC® Value
81934	Taurine	26614-8
30107	Threonine	22643-1
30108	Serine	22644-9
30109	Asparagine	26603-1
30110	Glutamic Acid	22652-2
30111	Glutamine	22641-5
30112	Proline	22645-6
30115	Glycine	22650-6
30116	Alanine	22657-1
30117	Citrulline	22654-8
30118	Alpha-amino-n-butyric Acid	26586-8
30119	Valine	22649-8
30120	Cystine	22653-0
30121	Methionine	22648-0
30122	Isoleucine	22659-7
30123	Leucine	9412-8
30124	Tyrosine	22642-3

30125	Phenylalanine	22646-4
30126	Ornithine	22647-2
30127	Lysine	22651-4
30128	Histidine	9453-2
30129	Arginine	22656-3
50435	Interpretation	49303-1
34566	Hydroxyproline	26596-7
34567	Aspartic Acid	22655-5
34568	Ethanolamine	26593-4
34569	Sarcosine	26598-3
34570	Beta-alanine	26589-2
34571	1-Methylhistidine	26584-3
34572	3-Methylhistidine	26585-0
34573	Argininosuccinic Acid	40838-5
34576	Homocitrulline	55875-9
34577	Alpha-aminoadipic Acid	26587-6
34578	Gamma-amino-n-butyric Acid	26594-2
34579	Beta-aminoisobutyric Acid	26590-0
34580	Hydroxylysine	26595-9
34581	Cystathionine	26592-6
34582	Tryptophan	26602-3
34583	Allo-isoleucine	22658-9