MAYO CLINIC LABORATORIES

Test Definition: AACSF

Amino Acids, QN, CSF

Overview

Useful For

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

Testing Algorithm

Includes quantitation of the following amino acids: phosphoserine, phosphoethanolamine, taurine, threonine, serine, hydroxyproline, asparagine, glutamic acid, 1-methylhistidine, 3-methylhistidine, argininosuccinic acid, carnosine, anserine, 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.
See Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm in Special Instructions.

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 AACSF / Amino Acids, Quantitative, Spinal Fluid) 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.

Reject Due To

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

Specimen Minimum Volume

0.1 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
CSF	Frozen (preferred)	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 particular 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 mental retardation and 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 drawn at the same time and the ratio of the amino acid concentrations in CSF to plasma is calculated.

Reference Values

CSF Amino Acid Reference Values (nmol/mL)	Age Groups	< or =31 days (n=73)	32 days-23 months (n=88)
2-18 years (n=189)	> or =19 years (n=32)	Phosphoserine (PSer)	<1
<1	<1	<1	Phosphoethanolamine (PEtN)
<15	<10	<8	<7
Taurine (Tau)	8-48	<28	<13
<20	Asparagine (Asn)	8-34	5-16
<10	5-20	Serine (Ser)	44-136
26-71	21-51	19-40	Hydroxyproline (Hyp)
<7	<3	<1	<2
Glycine (Gly)	5-115	<33	<11
<35	Glutamine (Gln)	467-1832	301-1128

326-1092	380-1348	Aspartic Acid (Asp)	<1
<1	<1	<2	Ethanolamine (EtN)
11-193	7-155	7-153	7-153
Histidine (His)	11-70	9-28	9-21
9-28	Threonine (Thr)	32-143	11-77
14-38	23-57	Citrulline (Cit)	<11
<6	<3	<9	Sarcosine (Sar)
<1	<1	<1	<1
Beta-alanine (bAla)	<26	<25	<25
<25	Alanine (Ala)	24-124	16-53
12-34	19-60	Glutamic Acid (Glu)	<12
<3	<1	<4	1-Methylhistidine (1MHis)
<3	<1	<2	<3
3-Methylhistidine (3MHis)	<4	<1	<1
<2	Argininosuccinic Acid (Asa)	<1	<2
<1	<1	Carnosine (Car)	<1
<1	<1	<1	Anserine (Ans)
<9	<9	<7	<3
Homocitrulline (Hcit)	<3	<1	<1
<1	Arginine (Arg)	5-39	11-35
11-27	11-32	Alpha-aminoadipic Acid (Aad)	<1
<1	<1	<1	Gamma-amino-n-butyric Acid (GABA)
<1	<1	<1	<1
Beta-aminoisobutyric Acid (bAib)	<1	<1	<1
<1	Alpha-amino-n-butyric Acid (Abu)	<15	<6
<5	<14	Hydroxylysine (Hyl)	<1
<1	<1	<1	Proline (Pro)
<17	<6	<2	<6
Ornithine (Orn)	<24	<12	<6
<11	Cystathionine (Cth)	<1	<2
<1	<1	Cystine (Cys)	<2
<2	<1	<1	Lysine (Lys)
11-63	9-33	10-25	13-42

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, and 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 Tietz Textbook of Clinical Chemistry and Molecular Diagnosis. Fourth edition. Edited by CA Burtis, ER Ashwood, DE Bruns. St. Louis, WB Saunders Company, 2005, pp 2207-2247
2. Van Hove J, Coughlin C II, Scharer G: Glycine Encephalopathy. In GeneReviews. 2002 Nov 14. Edited by MP Adam, HH Ardinger, RA Pagon, et al. Retrieved 2019 May 10. Available at https://www.ncbi.nlm.nih.gov/books/NBK1357/
3. El-Hattab AW: Serine biosynthesis and transport defects. Mol Genet Metab 2016;118:153-159
4. Duran M: Amino acids. In Laboratory Guide to the Methods in Biochemical Genetics. Edited by N Blau, M Duran, KM Gibson. Springer-Verlag, Berlin Heidelberg, 2008, pp 53-89

Performance

Method Description

Quantitative analysis of the amino acids is performed by liquid chromatography-tandem mass spectrometry (LC-MS/MS) by labeling amino acids present in cerebrospinal fluid (CSF) 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).(Lacey JM, Casetta B, Daniels SB, et al: Quantitation in plasma, urine and CSF by iTRAQ reagent amino acid analysis kit and MS-MS. J Am Soc Mass Spectrom [2008] 19[5] S97)

PDF Report

No

Specimen Retention Time

2 weeks

Performing Laboratory Location

Rochester

Fees & Codes

Test Classification

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

CPT Code Information

82139