

Overview

Useful For

Investigating inadequate tryptophan intake and monitoring dietary treatment

Method Name

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Plasma

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	OK
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hemolysis	
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 have been identified, including glutaric acidemia type 1, which affect other metabolic activities. Amino acid disorders can manifest at any time in a person's life, 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 particular amino acid disorder.

Tryptophan is an essential amino acid necessary for the synthesis of serotonin, melatonin, and niacin. Low plasma concentrations of tryptophan have been associated with clinical observations of insomnia, anxiety, and depression.

Glutaric acidemia type 1 is an autosomal recessive disorder of tryptophan and lysine metabolism caused by a deficiency of glutaryl-CoA dehydrogenase. Early diagnosis and treatment are essential to help prevent encephalopathic crises leading to brain degeneration. These can be provoked by infections, trauma, fever, and fasting. Treatment consists of preventing neurodegeneration through L-carnitine supplementation and strict adherence to an emergency protocol. Dietary protein, particularly lysine and tryptophan, is restricted during the vulnerable period of brain development from 0 to 5 years of age. In addition to other indices of malnutrition, the measurement of plasma concentration of tryptophan is used as an indicator of appropriate dietary therapy.

Reference Values

- < or =23 months: 17-75 nmol/mL
- 2 years-17 years: 23-80 nmol/mL
- > or =18 years: 29-77 nmol/mL

Interpretation

If the result is within the respective age-matched reference range, no interpretation is provided. When an abnormal result is reported, an interpretation may be added including a correlation to available clinical information, elements of differential diagnosis, and recommendations for additional biochemical testing, if applicable.

Cautions

Abnormal plasma concentrations of tryptophan are not diagnostic for a specific disorder and must be interpreted in the context of a patient's clinical presentation and other laboratory results.

Clinical Reference

1. Hoffmann GF, Schulze A: Organic acidurias. In: Sarafoglou K, Hoffmann GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. McGraw-Hill Medical Division; 2009:108-112
2. Larson A, Goodman S. Glutaric acidemia type 1. In: Adam MP, Everman DB, Mirzaa GM, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2019. Accessed March 08, 2023. Available at www.ncbi.nlm.nih.gov/books/NBK546575/
3. Goodman SI, Frerman FE. Organic acidemias due to defects in lysine oxidation: 2-ketoadipic acidemia and glutaric acidemia. 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 March 08, 2023. <https://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=225086303>

Performance

Method Description

Quantitative analysis of amino acids (AA) is performed by liquid chromatography tandem mass spectrometry (LC-MS/MS) by labeling amino acids present in plasma, spinal fluid, and urine 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 AA 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

82131

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
TRYPP	Tryptophan, P	20659-9

Result ID	Test Result Name	Result LOINC® Value
82955	Tryptophan	20659-9
34619	Interpretation (TRYPP)	59462-2
38056	Reviewed By	18771-6