

## 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

### Ordering Guidance

Body fluids are not acceptable specimens for this test.

For testing urine specimens, order TRYPU / Tryptophan, Random, Urine.

For testing spinal fluid specimens, order AACSF / Amino Acids, Quantitative, Spinal Fluid.

### 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: 4 hours, required;** 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 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**

&lt;24 months: 12-103 nmol/mL

2 years-17 years: 21-114 nmol/mL

&gt; or =18 years: 21-108 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**

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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. Klaessens S, Stroobant V, De Plaen E, Van den Eynde BJ. Systemic tryptophan homeostasis. *Front Mol Biosci.* 2022;9:897929
2. Larson A, Goodman S. Glutaric acidemia type 1. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. *GeneReviews* [Internet]. University of Washington, Seattle; 2019. Accessed April 22, 2024. Available at [www.ncbi.nlm.nih.gov/books/NBK546575/](http://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 April 22, 2024. <https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225086303>

**Performance****Method Description**

Quantitative analysis of amino acids is performed by liquid chromatography tandem mass spectrometry. 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.

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- 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