

Overview

Useful For

Aiding in the screening and monitoring of Hartnup disease

Highlights

Determination of tryptophan by conventional amino acid profiling methods (ninhydrin based, high-performance liquid chromatography) is hampered by coelution with other compounds. This test utilizes liquid chromatography tandem mass spectrometry to quantify tryptophan and is interference free.

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Urine

Ordering Guidance

Other body fluids are not acceptable specimens for this test. For testing spinal fluid specimens, order AACSF / Amino Acids, Quantitative, Spinal Fluid.

Testing for tryptophan using plasma specimens is available; order TRYPP / Tryptophan, 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

Supplies: Urine Tubes, 10 mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 2 mL

Collection Instructions: Collect a random urine specimen.

Forms

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

Specimen Minimum Volume

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 |
|---------------|--------------------|---------|-------------------|
| Urine | Refrigerated | 14 days | |
| | Frozen (preferred) | 70 days | |

Clinical & Interpretive

Clinical Information

Amino acids are the basic units that make up proteins and are crucial to virtually all metabolic processes in the body. Tryptophan is an essential amino acid necessary for the synthesis of serotonin, melatonin, and niacin.

Hartnup disease is a rare, usually benign, autosomal recessive disorder of renal and intestinal neutral amino acid transport. Reduced intestinal absorption of tryptophan and subsequent loss in the urine lead to a reduction of available tryptophan for the synthesis of niacin. The clinical features associated with Hartnup disease include an erythematous skin rash on exposed surfaces that is identical to the rash seen in pellagra (niacin deficiency) and cerebral ataxia. Biochemically, it is characterized by increased renal excretion of tryptophan and other neutral amino acids. Newborn screening studies reveal that most affected individuals remain asymptomatic, suggesting that clinical expression of symptoms is dependent on additional genetic or environmental factors (ie, multifactorial disease).

Reference Values

- <2 months: <241 nmol/mg creatinine
- 2-35 months: <329 nmol/mg creatinine
- 3-6 years: <222 nmol/mg creatinine
- 7-17 years: <218 nmol/mg creatinine
- > or =18 years: <140 nmol/mg creatinine

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 and recommendations for additional biochemical testing, if applicable.

Cautions

Abnormal urine 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. Levy HL. Hartnup disorder. In: Valle D, 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. Available at <https://ommbid.mhmedical.com/content.aspx?bookId=2709§ionid=225555835>

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

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 |
|---------|-----------------|--------------------|
|---------|-----------------|--------------------|

| | | |
|-----------|------------------------|---------------------|
| TRYPU | Tryptophan, U | 28608-8 |
| Result ID | Test Result Name | Result LOINC® Value |
| 83823 | Tryptophan, U | 28608-8 |
| 34618 | Interpretation (TRYPU) | 59462-2 |
| 113131 | Reviewed By | 18771-6 |