

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

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	Frozen (preferred)	70 days	
	Refrigerated	14 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

- < or =35 months: 14-315 nmol/mg creatinine
- 3-8 years: 10-303 nmol/mg creatinine
- 9-17 years: 15-229 nmol/mg creatinine
- > or =18 years: 18-114 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. Roth KS: Disorders of membrane transport. In: Sarafoglou K, Hoffmann GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. McGraw-Hill Medical Division; 2009:108-112

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 March 08, 2023. Available at <https://ommbid.mhmedical.com/content.aspx?sectionid=225555835&bookid=2709>

Performance

Method Description

Quantitative analysis of amino acids is performed by liquid chromatography tandem mass spectrometry (LC-MS/MS) by labeling amino acids present in plasma, cerebrospinal 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 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).(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