Test Definition: PUPYU

Purines and Pyrimidines Panel, U

Overview

Useful For

Evaluating patients with symptoms suspicious for disorders of purine and pyrimidine metabolism

Monitoring patients with disorders of purine and pyrimidine metabolism

Laboratory evaluation of primary and secondary hyperuricemias

Genetics Test Information

At least 35 known inherited disorders of purine and pyrimidine metabolism exist representing a diversity of neurological, immunological, hematological, and renal manifestations.

Highlights

This test provides a quantitative report of abnormal levels of purines and pyrimidines identified via liquid chromatography-mass spectrometry.

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Urine

Necessary Information

Patient's age is required.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 3 mL

Collection Instructions: Collect a random urine specimen.

Forms

If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume

2 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	90 days	

Clinical and Interpretive

Clinical Information

Purines (adenine, guanine, xanthine, hypoxanthine) and pyrimidines (uracil, thymine, cytosine, orotic acid) are involved in all biological processes, providing the basis for storage, transcription, and translation of genetic information as RNA and DNA. Purines are required by all cells for growth and survival and also play a role in signal transduction and translation. Purines and pyrimidines originate primarily from endogenous synthesis, with dietary sources playing only a minor role. The end product of purine metabolism is uric acid (2,6,8-trioxypurine), which must be excreted continuously to avoid toxic accumulation.

Disorders of purine and pyrimidine metabolism can involve all organ systems at any age. The diagnosis of the specific disorders of purine and pyrimidine metabolism is based upon the clinical presentation of the patient, determination of specific concentration patterns of purine and pyrimidine metabolites, and confirmatory enzyme assays and molecular genetic testing.

Numerous inborn errors of purine and pyrimidine metabolism have been documented. Clinical features are dependent upon the specific disorder but represent a broad spectrum of manifestations that may include immunodeficiency, developmental delay, nephropathy, and neurologic involvement. The most commonly described disorder of purine metabolism involves a deficiency of hypoxanthine-guanine phosphoribosyl transferase (HPRT) which causes 3 overlapping clinical syndromes depending on the amount of residual enzyme activity. The majority of patients with HPRT deficiency have classic Lesch-Nyhan syndrome, a severe -X-linked disorder characterized by crystals in urine, neurologic impairment, mild to severe intellectual disability, development of self-injurious behavior, and uric acid nephropathy.

Treatments for Lesch-Nyhan syndrome include allopurinol, urine alkalinization and hydration for nephropathy, and supportive management of neurologic symptoms. For milder forms of HPRT deficiency, treatment that can mitigate the potentially devastating effects of these diseases are disorder dependent; therefore, early recognition through screening and subsequent confirmatory testing is highly desirable.

Reference Values

Purines and Pyrimidines Panel, Urine

Reference Values

(all results reported as mmol/mol creatinine)

Age range	0-3 years	4-6 years	7-12 years	13-18 years	>18 years
Uracil	< or =50	< or =30	< or =25	< or =20	< or =20
Thymine	< or =3	< or =3	< or =3	< or =3	< or =3

Adenine	< or =3	< or =3	< or =3	< or =3	< or =3
Hypoxanthine	< or =65	< or =30	< or =30	< or =30	< or =30
Xanthine	< or =54	< or =21	< or =35	< or =15	< or =20
Orotic	< or =4	< or =4	< or =3	< or =3	< or =5
Dihydroorotic Acid	< or =3	< or =3	< or =3	< or =3	< or =3
Uric Acid	350-2500	200-2000	200-1400	150-700	70-700
Deoxythymidine	< or =3	< or =3	< or =3	< or =3	< or =3
Deoxyuridine	< or =3	< or =3	< or =3	< or =3	< or =3
Thymidine	< or =3	< or =3	< or =3	< or =3	< or =3
Uridine	< or =10	< or =3	< or =3	< or =3	< or =3
Deoxyadenosine	< or =3	< or =3	< or =3	< or =3	< or =3
Deoxyinosine	< or =3	< or =3	< or =3	< or =3	< or =3
Deoxyguanosine	< or =3	< or =3	< or =3	< or =3	< or =3
Adenosine	< or =3	< or =3	< or =3	< or =3	< or =3
Inosine	< or =6	< or =3	< or =3	< or =3	< or =3
Guanosine	< or =4	< or =3	< or =3	< or =3	< or =3
5-Aminoimidazole-4-carboxamide 1-beta-D-ribofuranoside (AICAR)	< or =3	< or =3	< or =3	< or =3	< or =3
Succinyladenosine	< or =16	< or =3	< or =3	< or =3	< or =3
S-Sulfocysteine	< or =11	< or =5	< or =5	< or =5	< or =5
Dihydrouracil	< or =15	< or =6	< or =6	< or =6	< or =6
Dihydrothymine	< or =11	< or =3	< or =3	< or =3	< or =3
N-carbamoyl-B-alanine	< or =30	< or =10	< or =10	< or =10	< or =10
N-carbamoyl-B-aminoisobutyric Acid	< or =20	< or =3	< or =3	< or =3	< or =3

Interpretation

Abnormal concentrations of measurable compounds will be reported along with an interpretation. The interpretation of an abnormal metabolite pattern includes an overview of the results and of their significance, a correlation to available clinical information, possible differential diagnosis, recommendations for additional biochemical testing and confirmatory studies (enzyme assay, molecular analysis), name, and phone number of contacts who may provide these studies, and a phone number of the laboratory directors in case the referring physician has additional questions.

Cautions

Additional confirmatory testing via enzyme assays and molecular genetic testing is required for follow-up of abnormal results.

Clinical Reference

1. Jinnah HA, Friedmann T: Lesch-Nyhan Disease and Its Variants. In The Online Metabolic and Molecular Bases of Inherited Disease. Edited by D Valle, AL Beaudet, B Vogelstein, et al. New York, NY. McGraw-Hill 2014. Accessed April 23, 2019. Available at http://ommbid.mhmedical.com/content.aspx?bookid=971&sectionid=62635320

2. Balasubramaniam S, Duley JA, Christodoulou J: Inborn errors of purine metabolism: clinical update and therapies. J Inherit Metab Dis 2014;37:669-686

3. Balasubramaniam S, Duley JA, Christodoulou J: Inborn errors of pyrimidine metabolism: clinical update and therapy. J Inherit Metab Dis 2014;37:687-698

Performance

Method Description

Diluted, filtered urine is mixed with an internal standard mixture and analyzed for uracil, thymine, adenine, hypoxanthine, xanthine, orotic, dihydroorotic, deoxythymidine, deoxyuridine, thymidine, uridine deoxyadenosine, deoxyinosine, deoxyguanosine, adenosine, inosine, guanosine, 5-aminoimidazole-4-carboxamide 1-beta-D-ribofuranoside (AICAR), succinyladensoine, S-sulfocysteine, dihydrouracil, dihydrothymine, n-carbamoyl-beta-alanine, and N-carbamoyl-beta-aminoisobutyric acid by liquid chromatography-tandem mass spectrometry (LC-MS/MS). The ratios of the extracted peak areas of the purine and pyrimidine analytes to the added internal standards are used to calculate the concentration of purines and pyrimidines present in the sample.(la Marca G, Casetta B, Malvagia S, Pasquini E, et al: Implementing tandem mass spectrometry as a routine tool for characterizing the complete purine and pyrimidine metabolic profile in urine samples. J Mass Spectrom 2006;41:1442-1452; Monostori P, Kilinke G, Hauke J, et al: Extended diagnosis of purine and pyrimidine disorders from urine: LC MS/MS assay development and clinical validation. PLoS One.2019 Feb 28;14(2):e0212458. doi: 10.1371/journal.pone.0212458)

PDF Report

No

Day(s) and Time(s) Test Performed

Tuesday; 8 a.m.

Analytic Time

7 days (Not reported on Saturday or Sunday)

Maximum Laboratory Time

16 days

Specimen Retention Time

6 months

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
PUPYU	Purines and Pyrimidines Panel, U	79673-0

Result ID	Test Result Name	Result LOINC Value
42201	Interpretation (PUPYU)	79677-1
41978	Uracil	25140-5
38249	Thymine	48157-2
38239	Adenine	59203-0
41980	Hypoxanthine	38366-1
41981	Xanthine	38371-1
38246	Orotic Acid	17869-9
38251	Dihydroorotic	78694-7
41979	Uric Acid	34385-5
38252	Deoxythymidine	59215-4
38253	Deoxyuridine	59193-3
38248	Thymidine	59215-4
38250	Uridine	59216-2
38241	Deoxyadenosine	59199-0
38243	Deoxyinosine	59202-2
38242	Deoxyguanosine	59201-4
38240	Adenosine	75160-2
38245	Inosine	59210-5
38244	Guanosine	78691-3
38254	AICAR	75151-1
38247	Succinyladenosine	59214-7
606745	S-Sulfocysteine	33876-4
38255	Dihydrouracil	79685-4
38256	Dihydrothymine	78693-9
38257	N-carbamoyl-beta-alanine	59251-9

Result ID	Test Result Name	Result LOINC Value
38258	N-carbamoyl-beta-aminoisobutyric Acid	79647-4
42200	Reviewed By	18771-6