Test Definition: PUPYP

Purines and Pyrimidines Panel, Plasma

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

Aiding in the diagnosis of individuals with suspected dihydropyrimidine dehydrogenase deficiency

Genetics Test Information

There are at least 35 known inherited disorders of purine and pyrimidine metabolism, which cause a variety of neurological, immunological, hematological, and renal manifestations.

Testing Algorithm

For information see Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm.

Special Instructions

- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

Highlights

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

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Plasma

Ordering Guidance

The preferred test to rule-out inherited disorders of purine and pyrimidine metabolism is PUPYU / Purines and Pyrimidines Panel, Random, Urine.

This test **does not evaluate** succinyladenosine. If succinyladenosine analysis is needed, order PUPYU / Purines and

Pyrimidines Panel, Random, Urine.

Necessary Information

Patient's age is required.

Specimen Required

Collection Container/Tube: Lavender top (EDTA)
Submission Container/Tube: Plastic vial
Specimen Volume: 0.5 mL Plasma
Collection Instructions:
1. Centrifuge at 4 degrees C and aliquot plasma into plastic vial.
2. Send plasma frozen.

Forms

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

Specimen Minimum Volume

Plasma: 0.2 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen	90 days	

Clinical & Interpretive

Clinical Information

Purines (adenine, guanine, xanthine, hypoxanthine, uric acid) 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 play a role in signal transduction and translation. Purines and pyrimidines originate primarily from endogenous synthesis, with dietary sources contributing only a small amount. 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 clinical manifestations that may include immunodeficiency, developmental delay, nephropathy, and neurologic involvement. The most common disorder of purine metabolism is 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.

Dihydropyrimidine dehydrogenase deficiency can result in a severe disorder in infancy involving seizures, intellectual disability, microcephaly, and hypertonia. It is caused by variants in the *DPYD* gene and is associated with high levels of uracil and thymine.

Reference Values

Age range	0-1 years	>1-4 years	5-18 years	>18 years
Uracil	< or =2	< or =2	< or =2	< or =2
Thymine	< or =2	< or =2	< or =2	< or =2
Adenine	< or =3	< or =3	< or =3	< or =3
Hypoxanthine	< or =35	< or =17	< or =15	< or =15
Xanthine	< or =6	< or =6	< or =6	< or =3
Dihydroorotic	< or =2	< or =2	< or =2	< or =2
Uric Acid	100-450	150-500	150-500	150-500
Deoxythymidine	< or =2	< or =2	< or =2	< or =2
Deoxyuridine	< or =2	< or =2	< or =2	< or =2
Uridine	< or =14	< or =9	< or =9	< or =9
Deoxyinosine	< or =2	< or =2	< or =2	< or =2
Deoxyguanosine	< or =2	< or =2	< or =2	< or =2
Inosine	< or =2	< or =2	< or =2	< or =2
Guanosine	< or =2	< or =2	< or =2	< or =2
Dihydrouracil	< or =3	< or =3	< or =3	< or =3
Dihydrothymine	< or =2	< or =2	< or =2	< or =2
N-carbamoyl-beta-alanine	< or =2	< or =2	< or =2	< or =2
N-carbamoyl-beta-aminoisobutyric acid	< or =2	< or =2	< or =2	< or =2

All results reported as nmol/mL

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 is required for follow-up of abnormal results.

Clinical Reference

1. Jinnah HA, Friedmann T. Lesch-Nyhan disease and its variants. 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 February 12, 2026. Available at https://ommbid.mhmedical.com/content.aspx?sectionid=225089443
2. Nyhan WL, Hoffmann GF, Al-Aqeel AI, Barshop BA. Introduction to the disorders of purine and pyrimidine metabolism. Atlas of Inherited Metabolic Diseases. 4th ed. CRC Press; 2020:495-495
3. Balasubramaniam S, Duley JA, Christodoulou J. Inborn errors of purine metabolism: clinical update and therapies. J Inherit Metab Dis. 2014;37:669-686
4. 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

Filtered EDTA plasma is mixed with an internal standard mixture and analyzed for uracil, thymine, adenine, hypoxanthine, xanthine, dihydroorotic, uric acid, deoxythymidine, deoxyuridine, uridine, deoxyinosine, deoxyguanosine, inosine, guanosine, dihydrouracil, dihydrothymine, N-carbamoyl- beta-alanine and N-carbamoyl- beta-aminoisobutyric acid by liquid chromatography-tandem mass spectrometry. 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.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Tuesday

Report Available

3 to 9 days

Specimen Retention Time

1 month

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.
- 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

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PUPYP	Purines and Pyrimidines Panel, P	79665-6

Result ID	Test Result Name	Result LOINC® Value
92310	Interpretation (PUPYP)	79659-9
92292	Uracil	75152-9
92293	Thymine	75121-4
92294	Hypoxanthine	75135-4
92295	Xanthine	75144-6
92296	Dihydroorotic	79654-0
92297	Uric Acid	14933-6
92298	Deoxythymidine	48162-2
92299	Deoxyuridine	47957-6
92300	Uridine	75159-4
92301	Deoxyinosine	75127-1
92302	Deoxyguanosine	75134-7
92303	Inosine	75150-3
92304	Guanosine	79670-6
92305	Dihydrouracil	79682-1
92306	Dihydrothymine	79669-8
92307	N-carbamoyl-B-alanine	79656-5
92308	N-carbamoyl-B-aminoisobutyric acid	79582-3
92309	Reviewed By	18771-6
606842	Adenine	75131-3