

Overview

Useful For

First-order test for evaluation of a suspected acute porphyria: acute intermittent porphyria, hereditary coproporphyria, and variegate porphyria

Testing Algorithm

The following algorithms are available in Special Instructions:

[-Porphyria \(Acute\) Testing Algorithm](#)

[-Porphyria \(Cutaneous\) Testing Algorithm](#)

Special Instructions

- [The Heme Biosynthetic Pathway](#)
- [Porphyria \(Acute\) Testing Algorithm](#)
- [Porphyria \(Cutaneous\) Testing Algorithm](#)

Method Name

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS) Stable Isotope Dilution Analysis

NY State Available

Yes

Specimen

Specimen Type

Urine

Shipping Instructions

[Ship specimen protected from light.](#)

Specimen Required

Supplies: Urine Container-Amber, 60 mL (T596)

Specimen Volume: 20 mL

Collection Instructions:

1. Collect a random urine specimen.
2. No preservative necessary but pH must be >5.0.
3. Specimens should be frozen immediately following collection.

Forms

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

Specimen Minimum Volume

15 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)	7 days	LIGHT PROTECTED
	Refrigerated	7 days	LIGHT PROTECTED

Clinical and Interpretive

Clinical Information

The porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway. Depending on the specific enzyme involved, various porphyrins and their precursors accumulate in different specimen types. The patterns of porphyrin accumulation in erythrocytes and plasma and excretion of the heme precursors in urine and feces allow for the detection and differentiation of the porphyrias.

The porphyrias are typically classified as erythropoietic or hepatic based upon the primary site of the enzyme defect. In addition, hepatic porphyrias can be further classified as chronic or acute, based on their clinical presentation.

The primary acute hepatic porphyrias: acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), and variegate porphyria (VP), are associated with neurovisceral symptoms that typically onset during puberty or later. Common symptoms include severe abdominal pain, peripheral neuropathy, and psychiatric symptoms. A broad range of medications (including barbiturates and sulfa drugs), alcohol, infection, starvation, heavy metals, and hormonal changes may precipitate crises. Photosensitivity is not associated with AIP but may be present in HCP and VP.

Urinary porphobilinogen (PBG) is elevated during the acute phase of the neurologic porphyrias. Urine and fecal porphyrin analysis should be performed to confirm the diagnosis and to distinguish between AIP, HCP and VP. A biochemical diagnosis of AIP can be confirmed by measurement of PBG deaminase activity using PBGD_ / Porphobilinogen Deaminase, Whole Blood. VP and HCP can be confirmed by measurement of fecal porphyrins (FQPPS / Porphyrins, Feces). Once the biochemical diagnosis of an acute porphyria is established, molecular genetic testing is available, which allows for diagnosis of at-risk family members.

The workup of patients with a suspected porphyria is most effective when following a stepwise approach. The following algorithms are available in Special Instructions or call 800-533-1710 to discuss testing strategies:

The following algorithms are available in Special Instructions or call 800-533-1710 to discuss testing strategies:

[-Porphyria \(Acute\) Testing Algorithm](#)

[-Porphyria \(Cutaneous\) Testing Algorithm](#)

Reference Values

< or =1.3 mcmmol/L

Interpretation

Abnormal results are reported with a detailed interpretation that may include an overview of the results and their significance, a correlation to available clinical information provided with the specimen, differential diagnosis, and recommendations for additional testing when indicated and available, and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

It is recommended that specimen collection occur during the acute phase. Porphobilinogen (PBG) may be normal when the patient is not exhibiting symptoms.

If the specimen is collected after treatment is initiated, the level of PBG excreted may be decreased.

Specimens should be protected from light and frozen immediately following collection. PBG is susceptible to degradation at high temperatures, at pH below 5.0, and on prolonged exposure to light.

Clinical Reference

1. Tortorelli S, Kloke K, Raymond K: Disorders of porphyrin metabolism. In: Dietzen DJ, Bennett MJ, Wong ECC, eds. *Biochemical and Molecular Basis of Pediatric Disease*. 4th ed. AACCC Press; 2010:chap 15.
2. Nuttall KL, Klee GG: Analytes of hemoglobin metabolism-porphyrins, iron, and bilirubin. In: Burtis CA, Ashwood ER, eds. *Tietz Textbook of Clinical Chemistry*. 5th ed. WB Saunders Company; 2001:584-607
3. Anderson KE, Sassa S, Bishop DF, Desnick RJ: Disorders of heme biosynthesis: X-linked sideroblastic anemia and the porphyrias. 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 September 04, 2020. Available at <https://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=225540906>

Performance

Method Description

Porphobilinogen (PBG) in urine is quantified by liquid chromatography-tandem mass spectrometry after addition of stable isotope-labeled PBG internal standard and solid phase extraction.(Ford RE, Magera MJ, Kloke KM, et al: Quantitative measurement of porphobilinogen in urine by stable-isotope dilution liquid chromatography-tandem mass spectrometry. *Clin Chem*. 2001 September;47[9]:1627-1632; Stolzel U, Doss MO, Schuppan D: Clinical guide and update on porphyrias. *Gastroenterology*. 2019 Aug;157(2):365-381.e4. doi: 10.1053/j.gastro.2019.04.050)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

2 to 6 days

Specimen Retention Time

1 week

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

84110

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
PBGU	Porphobilinogen, QN, Random, U	2811-8

Result ID	Test Result Name	Result LOINC Value
29365	Porphobilinogen, U	2811-8
29366	Interpretation (PBGU)	59462-2
35032	Reviewed By	18771-6