

## Overview

### Useful For

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

### Testing Algorithm

For more information see:

- [-The Heme Biosynthetic Pathway](#)
- [-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)

### NY State Available

Yes

## Specimen

### Specimen Type

Urine

### Shipping Instructions

[Ship specimen protected from light.](#)

### Necessary Information

Include a list of medications the patient is currently taking.

### Specimen Required

**Patient Preparation:** Patient **must not** consume any alcohol for at least 24 hours prior to collection.

**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 above 5.0.

3. Specimens should be protected from light and frozen immediately following collection.

**Forms**

[If not ordering electronically, complete, print, and send a Biochemical Genetics 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 & 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 (PBGD<sub>2</sub> / 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 (APGP / Acute Porphyria Gene Panel, Varies), 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.

For more information, see the following or call 800-533-1710 to discuss testing strategies:

- [-The Heme Biosynthetic Pathway](#)
- [-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**

- Tortorelli S, White A, Raymond K: Disorders of porphyrin metabolism. In: Dietzen DJ, Bennett MJ, Wong ECC, Haymond S, eds. *Biochemical and Molecular Basis of Pediatric Disease*. 5th ed. Academic Press; 2021:503-528
- 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
- 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 July 5, 2024. Available at <https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225540906>
- Anderson KE, Lobo R, Salazar D, et al: Biochemical diagnosis of acute hepatic porphyria: Updated expert recommendations for primary care physicians. *Am J Med Sci*. 2021 Aug;362(2):113-121. doi: 10.1016/j.amjms.2021.03.004

**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;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 4 days

**Specimen Retention Time**

1 week

**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**

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