
Overview

Useful For

An equivalent option to urine for first-line test for evaluation of a suspected acute porphyria

Monitoring patients undergoing treatment for an acute intermittent porphyria or other acute porphyria

Genetics Test Information

This test is used as part of the diagnostic assessment and monitoring of patients with acute intermittent porphyria (AIP) and other acute porphyrias.

Highlights

Plasma porphobilinogen (PBG) and aminolevulinic acid (ALA) are elevated during the symptomatic phase of the acute porphyrias: acute intermittent porphyria, hereditary coproporphyria, and variegate porphyria

An isolated elevation of ALA may be due to the very rare aminolevulinic acid dehydratase deficiency porphyria (ADP) or more commonly, a secondary inhibition of ALA.

This test can be used as part of the diagnostic assessment and monitoring of patients with acute intermittent porphyria (AIP) and other acute porphyrias.

Specimen is best obtained while the patient is having symptoms.

Additional testing must be performed to distinguish among the acute porphyrias

Testing Algorithm

The following algorithms are available in Special Instructions:

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

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

Special Instructions

- [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

Plasma

Shipping Instructions

Ship specimens refrigerated or frozen [and in amber vial to protect from light.](#)

Necessary Information

Include a list of medications the patient is currently taking.

Specimen Required

Patient Preparation: Patient should abstain from alcohol for at least 24 hours prior to specimen collection.

Supplies: Amber Frosted Tube, 5 mL (T192)

Collection Container/Tube:

Preferred: Green top (heparin)

Acceptable: Green top (lithium heparin), Lavender top (EDTA), Yellow top (ACD A or B)

Submission Container/Tube: Amber vial (T192)

Specimen Volume: 1 mL

Collection Instructions: It is recommended that specimen collection occur during the acute phase. Porphobilinogen (PBG) and aminolevulinic acid (ALA) may be normal when the patient is not exhibiting symptoms.

Specimen Minimum Volume

0.3 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen (preferred)	21 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 the 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, of the 5 hepatic porphyrias, 4 typically present with acute neurological manifestations and are designated

the acute porphyrias. Clinically, however, these attacks can be prolonged and chronic.

Three primary acute hepatic porphyrias: acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), and variegate porphyria (VP), are associated with neurovisceral symptoms which 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.

Plasma porphobilinogen (PBG) and aminolevulinic acid (ALA) are elevated during the acute phase of these neurologic porphyrias. Urine and fecal porphyrin analysis should be performed to confirm the diagnosis and to distinguish among AIP, HCP, and VP. A biochemical diagnosis of AIP can be confirmed by measurement of PBG deaminase activity (PBGD₂ / 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 for AIP (HMBSZ / *HMBS* Gene, Full Gene Analysis), HCP (CPOXZ / *CPOX* Gene, Full Gene Analysis), or VP (PPOXZ / *PPOX* Gene, Full Gene Analysis) which allows for diagnosis of at-risk family members.

The very rare (<10 cases described) autosomal recessive aminolevulinic acid dehydratase deficiency porphyria (ADP) is also a primary acute porphyria causing neurovisceral symptoms. It presents in childhood and biochemically, it is characterized by an isolated significant elevation of aminolevulinic acid (ALA). More commonly, however, isolated elevations of ALA are due to secondary inhibition of ALA dehydratase. Among the secondary causes, acute lead intoxication results in the highest degree of aminolevulinic aciduria. Less significant elevations are seen in chronic lead intoxication and tyrosinemia type I.

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:

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

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

Reference Values

Porphobilinogen: < or =0.5 nmol/mL

Aminolevulinic Acid: < or =0.5 nmol/mL

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, 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

Additional testing must be performed to distinguish among the acute porphyrias.

The specimen should be collected prior to treatment as therapy may decrease the amount of PBG and ALA excreted.

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

Clinical Reference

1. Tortorelli S, Kloke K, Raymond K: Chapter 15: Disorders of porphyrin metabolism. In Biochemical and Molecular Basis of Pediatric Disease. Fourth edition. Edited by DJ Dietzen, MJ Bennett, ECC Wong. AACCC Press 2010; pp 307-324
2. Sardh E, Harper P, Andersson D, Floderus: Plasma porphobilinogen as a sensitive biomarker to monitor the clinical and therapeutic course of acute intermittent porphyria attacks. Eur J Intern Med 2009 Mar;20(2):201-207
3. Floderus Y, Sardh E, Moller C, et al: Variations in porphobilinogen and 5-aminolevulinic acid concentrations in plasma and urine from asymptomatic carriers of acute intermittent porphyria gene with increased porphyrin precursor excretion. Clin Chem 2006 Apr;52(4):701-707
4. Nuttall KL, Klee GG: Analytes of hemoglobin metabolism- porphyrins, iron, and bilirubin. In Tietz Textbook of Clinical Chemistry. Fifth edition. Edited by CA Burtis, ER Ashwood. Philadelphia, PA, WB Saunders Company, 2001, pp 584-607
5. Anderson KE, Sassa S, Bishop DF, Desnick RJ: Disorders of Heme Biosynthesis: X-Linked Sideroblastic Anemia and the Porphyrins. 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 July 13, 2015. Available at ommbid.mhmedical.com/content.aspx?bookid=971&Sectionid=62638866

Performance**Method Description**

In a microcentrifuge tube internal standard and plasma are combined, centrifuged and then subjected to solid phase extraction (SPE). The SPE eluate is evaporated and the residue is then reconstituted and subjected to liquid chromatography-tandem mass spectrometry analysis (LC-MS/MS). Total run time is 3 minutes.(Unpublished Mayo method)

PDF Report

No

Day(s) and Time(s) Test Performed

Wednesday; 2 p.m.

Analytic Time

2 days (not reported on Saturday or Sunday)

Maximum Laboratory Time

6 days

Specimen Retention Time

14 Days

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-- Porphobilinogen, P

82135-- Aminolevulinic Acid, P

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
PBALP	PBG and ALA, P	In Process

Result ID	Test Result Name	Result LOINC Value
38029	Porphobilinogen, P	17474-8
38028	Aminolevulinic Acid, P	79646-6
38030	Interpretation (PBALP)	59462-2
38031	Reviewed By	18771-6