MAYO CLINIC LABORATORIES

Test Definition: PBALP

Porphobilinogen and Aminolevulinic Acid, Plasma

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

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

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

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

If ordered for diagnostic purposes, results are most informative when the specimen is obtained while the patient is having symptoms.

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

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

Highlights

This test is an alternative for the evaluation of a suspected acute porphyria when a urine specimen cannot be obtained during a symptomatic episode.

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 **must not** consume any alcohol for at least 24 hours prior to specimen collection.
Supplies: Amber Frosted Tube, 5mL (T915)
Collection Container/Tube:
Preferred: Green top (sodium heparin)
Acceptable: Green top (lithium heparin), lavender top (EDTA), yellow top (ACD A or B)
Submission Container/Tube: Amber vial
Specimen Volume: 1 mL
Collection Instructions:
1. It is recommended that specimen collection occur during the acute phase. Porphobilinogen and aminolevulinic acid may be normal when the patient is not exhibiting symptoms.
2. Centrifuge and aliquot plasma into plastic vial.

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

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 & 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 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.

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 (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 (APGP / Acute Porphyria Gene Panel, Varies), which allows for diagnosis of at-risk family members.

The very rare (<10 cases described) autosomal recessive ALA dehydratase deficiency porphyria (ADP) is also a primary acute porphyria causing neurovisceral symptoms with variable age of onset. Biochemically, ADP is characterized by an isolated significant elevation of ALA. More commonly, however, isolated elevations of ALA are due to secondary inhibition of ALA dehydratase with acute lead intoxication, which results in the highest degree of aminolevulinic aciduria. Less significant elevations are seen in chronic lead intoxication, tyrosinemia type I, alcoholism, and pregnancy.
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

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 porphobilinogen (PBG) and aminolevulinic acid.

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

Clinical Reference

1.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
2. 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;362(2):113-121. doi:10.1016/j.amjms.2021.03.004

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.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Wednesday

Report Available

3 to 9 days

Specimen Retention Time

14 days

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
82135

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PBALP	PBG and ALA, P	96911-3

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