Test Definition: PBALP

PBG and ALA, P

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 (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.
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

The following algorithms are available:
-Porphyria (Acute) Testing Algorithm
-Porphyria (Cutaneous) Testing Algorithm

Special Instructions

- 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 frozenand 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
Specimen Volume: 1 mL
Collection Instructions: 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.

Forms

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

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Minimum Volume

0.3 mL

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 aminolevulinic acid 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 aminolevulinic acid (ALA). More commonly, however, isolated elevations of ALA are due to secondary inhibition of ALA dehydratase with 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 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 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

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).(Unpublished Mayo method)

PDF Report
No

Specimen Retention Time
14 Days

Performing Laboratory Location

Rochester

Fees & Codes

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 US Food and Drug Administration.

CPT Code Information

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