Overview

Useful For
Confirmation of a diagnosis of acute intermittent porphyria (AIP)

Testing Algorithm
The following algorithms are available in Special Instructions:

- Porphyria (Acute) Testing Algorithm
- Porphyria (Cutaneous) Testing Algorithm

Special Instructions

The Heme Biosynthetic Pathway
Informed Consent for Genetic Testing
Porphyria (Acute) Testing Algorithm
Porphyria (Cutaneous) Testing Algorithm
Informed Consent for Genetic Testing (Spanish)

Method Name
Enzymatic End point/Spectrofluorometric

NY State Available
Yes

Specimen

Specimen Type
Whole blood

Advisory Information
This test is for diagnosis of acute intermittent porphyria. Porphobilinogen deaminase, also known as uroporphyrinogen I synthase, is commonly confused with uroporphyrinogen III synthase, the enzyme deficient in congenital erythropoietic porphyria (CEP). For CEP cases, order UPGC / Uroporphyrinogen III Synthase (Co-Synthase) (UPG III S), Erythrocytes.

Necessary Information
Include a list of medications the patient is currently taking.

Specimen Required
All porphyrin tests on whole blood can be performed on 1 draw tube.

Patient Preparation: Abstinence from alcohol for at least 24 hours prior to specimen collection is essential as ethanol induces porphobilinogen deaminase (PBGD) activity, which may lead to a false-normal result.

Container/Tube:
Preferred: Green top (sodium heparin)
Acceptable: Lavender top (EDTA) or green top (lithium heparin)

Specimen Volume: Full tube

Collection Instructions:
1. Patient should abstain from alcohol for 24 hours.
2. Immediately place specimen on wet ice.

Forms
1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

   - Informed Consent for Genetic Testing (T576)
   - Informed Consent for Genetic Testing-Spanish (T826)

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

Specimen Minimum Volume
3 mL

Reject Due To

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<th>Gross hemolysis</th>
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Specimen Stability Information

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<tr>
<th>Specimen Type</th>
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<tr>
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Clinical and Interpretive

Clinical Information

The porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway. Acute intermittent porphyria (AIP) is caused by diminished erythrocyte activity of porphobilinogen deaminase (PBGD), also known as uroporphyrinogen I synthase or hydroxymethylbilane synthase. Onset of AIP typically occurs during puberty or later. Individuals may experience acute episodes of neuropathic symptoms. Common symptoms include severe abdominal pain, peripheral neuropathy, and psychiatric symptoms. Crises may be precipitated by a broad range of medications (including barbiturates and sulfa drugs), alcohol, infection, starvation, heavy metals, and hormonal changes. AIP is inherited in an autosomal dominant manner. At-risk family members of patients with a biochemical diagnosis of AIP should undergo appropriate testing. Timely diagnosis is important as acute episodes of AIP can be fatal. Treatment of AIP includes the prevention of symptoms through avoidance of precipitating factors. More than 80% of individuals with deficiency mutation in the HMBS gene remain asymptomatic throughout their lives.

The biochemical diagnosis of AIP is made by demonstrating increased urinary excretion of porphobilinogen (PBG).
and is most accurate during an acute episode. In addition, the diagnosis of AIP can be confirmed through the measurement of porphobilinogen deaminase (PBGD) enzyme activity in erythrocytes, although 5% to 10% of affected individuals exhibit normal erythrocyte PBGD activity. In addition, molecular genetic confirmation (HMBSZ / HMBS Gene, Full Gene Analysis) is available on a clinical basis and can be particularly helpful in identifying asymptomatic family members at risk of acute symptoms.

The workup of patients with a suspected porphyria is most effective when following a stepwise approach. See Porphyria (Acute) Testing Algorithm in Special Instructions or call 800-533-1710 to discuss testing strategies.

Reference Values
Reference ranges have not been established for patients who are <16 years of age.

- > or = 7.0 nmol/L/sec
- 6.0-6.9 nmol/L/sec (indeterminate)
- < 6.0 nmol/L/sec (diminished)

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 a laboratory director in case the referring physician has additional questions.

Cautions
A normal result does not rule-out acute intermittent porphyria; 5% to 10% of affected individuals will have normal erythrocyte PBGD activity. Additionally, enzyme activity may be increased during an acute attack, therefore, the enzyme level should be assessed when the patient is asymptomatic.

Clinical Reference

Performance
Method Description
Measurement of porphobilinogen deaminase (PBGD) activity is based on the measurement of the rate of synthesis of uroporphyrin from porphobilinogen (PBG) in incubated, lysed erythrocytes. Low yield of uroporphyrin from PBG indicates a deficiency of PBGD. (Ford RE, Ou CN, Ellefson RD: Assay for erythrocyte uroporphyrinogen I synthase activity, with porphobilinogen as substrate. Clin Chem 1980;26:1182-1185)
PDF Report
No

Day(s) and Time(s) Test Performed
Tuesday, Thursday; 1 p.m.

Analytic Time
2 days (not reported on Saturday or Sunday)

Maximum Laboratory Time
4 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
82657

LOINC® Information

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