MAYO CLINIC LABORATORIES

Test Definition: PBGD_

Porphobilinogen Deaminase, Whole Blood

Overview

Useful For
Confirmation of a diagnosis of acute intermittent porphyria

Genetics Test Information
This test is for diagnosis of acute intermittent porphyria.

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

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

Ordering Guidance
This test is for diagnosis of acute intermittent porphyria. Porphobilinogen deaminase, also known as hydroxymethylbilane synthase and uroporphyrinogen I synthase, is commonly confused with uroporphyrinogen III synthase, the enzyme deficient in congenital erythropoietic porphyria.

Necessary Information
1. Patient's age is required
2. Include a list of medications the patient is currently taking.

Specimen Required

Patient Preparation: For 24 hours before specimen collection, patient **must not** consume any alcohol. This is essential as ethanol induces porphobilinogen deaminase 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: 4 mL; full tube
Collection Instructions: Refrigerate specimen as soon as possible.

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:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

3 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	8 days	
	Ambient	7 days	

Clinical & 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 (HMBS).

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 a deficiency variant 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 PBGD enzyme activity in erythrocytes, although 5% to 10% of affected individuals exhibit normal erythrocyte PBGD activity. In addition, molecular genetic confirmation (CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify gene list ID: IEMCP-WCJKC9) is available on a clinical basis and can be particularly helpful in identifying asymptomatic family members at risk of acute symptoms.

A stepwise approach is typically most effective when evaluating patients with suspected porphyria. See Porphyria (Acute) Testing Algorithm or call 800-533-1710 to discuss testing strategies.

Reference Values

> or =7.0 nmol/L/sec
6.0-6.9 nmol/L/sec (Indeterminate)
<6.0 nmol/L/sec (Diminished)

Reference ranges have not been established for patients who are younger than 16 years.

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.

Cautions

A normal result does not rule out acute intermittent porphyria; 5% to 10% of affected individuals will have normal erythrocyte porphobilinogen deaminase 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

1. Tortorelli S, Kloke K, Raymond K. Disorders of porphyrin metabolism. In: Dietzen DJ, Bennett MJ, Wong ECC, eds. Biochemical and Molecular Basis of Pediatric Disease. 4th ed. AACC Press; 2010:307-324
2. 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
3. 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 August 3,2026. Available at https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225540906

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[8]:1182-1185; Bustad HJ, Kallio JP, Vorland M, et al. Acute intermittent porphyria: An Overview of therapy developments and future perspectives focusing on stabilisation of HMBS and proteostasis regulators. Int J Mol Sci. 2021;22[2]:675. doi:10.3390/ijms22020675)

PDF Report
No

Day(s) Performed
Tuesday

Report Available
2 to 8 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
82657

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PBGD_	PBG Deaminase, WB	12810-8

Result ID	Test Result Name	Result LOINC® Value
4022	PBG Deaminase, WB	12810-8
28400	Interpretation	59462-2
606470	Reviewed By	18771-6