MAYO CLINIC LABORATORIES

# Test Definition: PBGDW

Porphobilinogen Deaminase, Washed Erythrocytes

## Overview

### Useful For

Confirmation of a diagnosis of acute intermittent porphyria using washed erythrocyte specimens

### 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

Washed RBC

### 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. Volume of packed cells and total volume of specimen (red cells + saline) are required and must be sent with specimen.**

2. **Patient's age is required**
3. 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.
**Collection Container/Tube:**
**Preferred:** Green top (sodium heparin)
**Acceptable:** Lavender top (EDTA) or green top (lithium heparin)
**Submission Container/Tube:** Plastic vial
**Specimen Volume:** Entire washed erythrocyte suspension
**Collection Instructions:**
Collect and process whole blood specimen as follows:
1. Transfer entire specimen to a 12-mL graduated centrifuge tube.
2. Centrifuge specimen at 4 degrees C for 10 minutes at 2000 rpm.
**3. Record volume of packed cells and the total volume of the specimen.**
4. Discard supernatant plasma.
5. Wash packed erythrocytes 2 times by resuspension of at least an equal amount of cold 0.9% saline, mix, and centrifuge for 5 minutes at 2000 rpm, discarding supernatant after each washing.
6. Resuspend packed cells to the original total volume with 0.9% saline. Invert specimen gently to mix.
7. Transfer washed erythrocytes into a plastic vial and freeze.

## 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

Washed and resuspended erythrocytes: 1 mL

## Reject Due To

| | |
|---|---|
| Cell suspension not available | Reject |

## Specimen Stability Information

| Specimen Type | Temperature | Time | Special Container |
|---|---|---|---|
| Washed RBC | Frozen (preferred) | 14 days | |
| | Ambient | 48 hours | |
| | Refrigerated | 14 days | |

## Clinical & Interpretive

### Clinical Information

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 Definition: PBGDW

Porphobilinogen Deaminase, Washed Erythrocytes

## 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 |
|---|---|---|
| PBGDW | PBG Deaminase, RBC | 2812-6 |

| Result ID | Test Result Name | Result LOINC® Value |
|---|---|---|
| 31944 | PBG Deaminase, RBC | 2812-6 |
| 31945 | Interpretation | 59462-2 |
| BG575 | Total cell Suspension | 94496-7 |
| BG576 | Packed cell volume | 94497-5 |
| 606471 | Reviewed By | 18771-6 |