

Reporting Title: PBG Deaminase, RBC**Performing Location:** Mayo Clinic Laboratories - Rochester Main Campus**Ordering Guidance:**

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.

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 Requirements:

Patient Preparation: Patient **must not** consume any alcohol for 24 hours before specimen collection. 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.

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

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
PBGDW	BG575	Total cell Suspension	Plain Text	Yes
PBGDW	BG576	Packed cell volume	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
31944	PBG Deaminase, RBC	Alphanumeric	nmol/L/sec	2812-6
31945	Interpretation	Alphanumeric		59462-2
BG575	Total cell Suspension	Alphanumeric		94496-7
BG576	Packed cell volume	Alphanumeric		94497-5
606471	Reviewed By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82657

Reference Values:

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

> or =7.0 nmol/L/sec

6.0-6.9 nmol/L/sec (indeterminate)

<6.0 nmol/L/sec (diminished)