

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 (CEP). For CEP cases, order UPGC / Uroporphyrinogen III Synthase (Co-Synthase), Erythrocytes.

- 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)