
Reporting Title: PBG Deaminase, RBC**Performing Location:** Rochester**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 erythrocyte suspension (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: Abstinence from alcohol for at least 24 hours prior to specimen collection 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: 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.

Specimen Minimum Volume:

1 mL of washed and resuspended erythrocytes

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	
	Refrigerated	14 days	
	Ambient	48 hours	

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)