MAYO CLINIC LABORATORIES

Test Definition: PBGD_

Porphobilinogen Deaminase, Whole Blood

Reporting Title: PBG Deaminase, WB
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. Patient's age is required
2. 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.
Container/Tube:
Preferred: Green top (sodium heparin)
Acceptable: Lavender top (EDTA) or green top (lithium heparin)
Specimen Volume: 4 mL
Collection Instructions: Refrigerate specimen as soon as possible.

Specimen Minimum Volume:

3 mL

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
Whole blood	Refrigerated (preferred)	8 days	
	Ambient	7 days	

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Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
4022	PBG Deaminase, WB	Alphanumeric	nmol/L/sec	12810-8
28400	Interpretation	Alphanumeric		59462-2
606470	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)