

Overview

Useful For

Diagnosis of congenital erythropoietic porphyria

This test is **not useful for** diagnosis of acute intermittent porphyria (AIP).

Genetics Test Information

This test is **not appropriate** for assessment of acute abdominal pain.

Testing Algorithm

The following algorithms are available:

- [Porphyria \(Acute\) Testing Algorithm](#)
- [Porphyria \(Cutaneous\) Testing Algorithm](#)

Special Instructions

- [The Heme Biosynthetic Pathway](#)
- [Informed Consent for Genetic Testing](#)
- [Porphyria \(Acute\) Testing Algorithm](#)
- [Porphyria \(Cutaneous\) Testing Algorithm](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)

Highlights

Congenital erythropoietic porphyria (CEP) is a disease usually seen in pediatric patients.

In our testing experience over the last 10 years, fewer than 5 adult patients have been diagnosed with CEP associated with a myelodysplastic syndrome.

Method Name

High-Performance Liquid Chromatography (HPLC)

NY State Available

Yes

Specimen

Specimen Type

WB Heparin

Ordering Guidance

This test is most appropriately used for pediatric patients.

This test measures uroporphyrinogen (UPG) III synthase to confirm congenital erythropoietic porphyria, which is typically seen in early infancy. It does not measure UPG I synthase (also known as porphobilinogen deaminase), the enzyme deficient in acute intermittent porphyria (AIP). For AIP (and UPG I synthase), order PBGD_ / Porphobilinogen Deaminase, Whole Blood.

Necessary Information

- 1. Include a list of medications the patient is currently taking.
- 2. Date of transfusion, if performed

Specimen Required

All porphyrin tests on erythrocytes can be performed on one collection tube.

Patient Preparation: Patient should abstain from alcohol for 24 hours prior to specimen collection.

Container/Tube: Green top (sodium or lithium heparin)

Specimen Volume: 4 mL

Collection Instructions: Immediately place specimen on wet ice.

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

3 mL

Reject Due To

Gross hemolysis	Reject
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Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
WB Heparin	Refrigerated	7 days	

Clinical & Interpretive

Clinical Information

The porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway.

Congenital erythropoietic porphyria (CEP) is an extremely rare, autosomal recessive porphyria that typically presents in early infancy. Also known as Gunther disease, CEP results from a deficiency of uroporphyrinogen III (co-) synthase (UROIII). In most cases, the disorder is suggested during the first few days or weeks of life by pink, violet, or brown urinary staining of diapers. Clinical symptoms include hemolytic anemia, hepatosplenomegaly, skin photosensitivity, scarring and blistering, red or brown dental discoloration (erythrodontia), and hypertrichosis (excess body hair). Growth and cognitive developmental delays are commonly observed in individuals with CEP. A few cases of adult-onset CEP have been reported, typically associated with a myelodysplastic syndrome.

The workup of patients with a suspected porphyria is most effective when following a stepwise approach. Molecular confirmatory testing is available on a clinical basis; order CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify UROS Gene List ID: IEMCP-8W4945. See [Porphyria \(Cutaneous\) Testing Algorithm](#) or call 800-533-1710 to discuss testing strategies.

Reference Values

> or =75 Relative Units (normal)

See [The Heme Biosynthetic Pathway](#)

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, and a phone number to reach a laboratory director in case the referring physician has additional questions.

Cautions

This test is **not** useful for ruling out acute intermittent porphyria (AIP), a disorder caused by decreased uroporphyrinogen I synthase (also known as porphobilinogen deaminase). For AIP, order PBGD_ / Porphobilinogen Deaminase, Whole Blood.

This test does not reliably distinguish between individuals who are carriers for congenital erythropoietic porphyria (CEP) and are at risk for having an affected child.

If possible, specimens from patients suspected of having CEP should be collected prior to blood transfusions; uroporphyrinogen (UPG) III synthase activity in transfused erythrocytes can cause false-negative results.

Abstinence from alcohol for at least 24 hours is essential for accurate results. While the effects of alcohol on this enzyme have not yet been determined, alcohol is known to suppress or induce other enzymes in the heme biosynthetic pathway.

Clinical Reference

1. Tortorelli S, Kloke K, Raymond K: Disorders of porphyrin metabolism. In: Dietzen DJ, Bennett MJ, Wong EDD, eds. Biochemical and Molecular Basis of Pediatric Disease. 4th ed. AACCPress; 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 May 6, 2022. Available at <https://ommbid.mhmedical.com/content.aspx?sectionid=225540906&bookid=2709>

4. Erwin AL, Desnick RJ: Congenital erythropoietic porphyria: Recent advances. Mol Genet Metab. 2019 Nov;128(3):288-297. doi: 10.1016/j.ymgme.2018.12.008

Performance

Method Description

Washed cells are incubated with aminolevulinic acid as substrate and the series I and III porphyrin isomers formed are measured. The proportion of series III isomers formed in relation to total porphyrins (I + III isomers) represents the uroporphyrinogen III synthase activity. The values are reported as Relative Units.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Varies

Report Available

3 to 9 days

Specimen Retention Time

Residual whole blood, processed specimen: 2 weeks

Performing Laboratory Location

Rochester

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 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
UPGC	Uroporphyrinogen III Synthase, RBC	11066-8

Result ID	Test Result Name	Result LOINC® Value
80288	Uroporphyrinogen III Synthase, RBC	11066-8