Test Definition: UPGC

Uroporphyrinogen III Synthase, RBC

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.

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.

Testing Algorithm

The following algorithms are available in Special Instructions:

-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)

Method Name

High-Performance Liquid Chromatography (HPLC)

NY State Available

Yes

Specimen

Specimen Type

WB Heparin

Advisory Information

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

Include a list of medications the patient is currently taking.

Specimen Required

All porphyrin tests on erythrocytes can be performed on 1 collection tube.

Patient Preparation: Patient should abstain from alcohol for 24 hours.

Container/Tube: Green top (heparin)

Specimen Volume: Full tube

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 in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume

3 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
WB Heparin	Refrigerated	7 days	

Clinical and 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 (UROIIIS). 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. See Porphyria (Cutaneous) Testing Algorithm in Special Instructions or call 800-533-1710 to discuss testing strategies.

Reference Values

> or =75 Relative Units (normal)

See The Heme Biosynthetic Pathway in Special Instructions.

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: Chapter 15: Disorders of porphyrin metabolism. In Biochemical and Molecular Basis of Pediatric Disease. Fourth Edition. Edited by DJ Dietzen, MJ Bennett, ECC Wong. AACC Press 2010, pp 307-324

2. Nuttall KL, Klee GG: Analytes of hemoglobin metabolism-porphyrins, iron, and bilirubin. In Tietz Textbook of Clinical Chemistry. Fifth edition. Edited by CA Burtis, ER Ashwood. Philadelphia, WB Saunders Company, 2001, pp 584-607

3. Anderson KE, Sassa S, Bishop DF, Desnick RJ: X-Linked sideroblastic anemia and the porphyrias. In Disorders of Heme Biosynthesis. Edited by D Valle, AL Beaudet, B Vogelstein, et al. McGraw-Hill, Accessed August 9, 2017. Available at Â https://ommbid.mhmedical.com/content.aspx?sectionid=225540906&bookid=2709&Resultclick=2

4. Desnick RJ, Astrin KH: Congenital erythropoietic porphyria: advances in pathogenesis and treatment. Br J Haematol 2002;117(4):779-795

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) and Time(s) Test Performed

Varies

Analytic Time

7 days (Not reported on Saturday or Sunday)

Maximum Laboratory Time

14 days

Specimen Retention Time

2 weeks

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. 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. This test has not been cleared or approved by the U.S. 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