

Overview

Useful For

Preferred test for the confirmation of a diagnosis of porphyria cutanea tarda type II and hepatoerythropoietic porphyria

Genetics Test Information

Uroporphyrinogen decarboxylase (UROD) is deficient in cases of type II porphyria cutanea tarda (PCT). Enzyme analysis is uninformative in sporadic PCT cases.

Testing Algorithm

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. If guidance is needed for an acute form of porphyria, the [Porphyria \(Acute\) Testing Algorithm](#) is also available in Special Instructions.

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)/Incubation of Lysed Erythrocytes

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Ordering Guidance

Porphyria cutanea tarda (PCT) type I, the most common form of PCT, exhibits normal RBC enzyme activity. The preferred test for diagnosis of type I is PQNU / Porphyrins, Quantitative, 24 Hour, Urine or PQNRU / Porphyrins, Quantitative, Random, Urine.

Necessary Information

Include a list of medications the patient is currently taking.

Specimen Required

Patient Preparation: Patient should abstain from alcohol for 24 hours. Abstinence from alcohol is essential for at least 24 hours as alcohol suppresses enzyme activity for 24 hours after ingestion.

Container/Tube:

Preferred: Green top (sodium heparin)

Acceptable: Lavender top (EDTA) or green top (lithium heparin)

Specimen Volume: Full tube

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
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Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	14 days	
	Ambient	7 days	

Clinical and Interpretive

Clinical Information

The porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway. Porphyria cutanea tarda (PCT) is the most common porphyria resulting from a partial deficiency of hepatocyte or erythrocyte uroporphyrinogen decarboxylase (UROD; see [The Heme Biosynthetic Pathway](#) in Special Instructions). PCT is classified into 3 subtypes. The most frequently encountered is type I, a sporadic or acquired form, typically associated with concomitant disease or other precipitating factors. Patients exhibit normal UROD activity in erythrocytes but decreased hepatic activity. This differs from type II PCT in which patients exhibit approximately 50% activity in both erythrocytes and hepatocytes. Type II accounts for about 20% of cases and is inherited in an autosomal dominant manner with low penetrance. Type III is a rare familial form seen in <5% of PCT cases. As in type I, patients with type III PCT have normal UROD activity in erythrocytes with decreased hepatic activity. Type III cases are distinguished from type I by the history of other affected family members.

Hepatoerythropoietic porphyria (HEP) is a rare autosomal recessive form of porphyria that typically presents in early childhood. Patients have a severe deficiency of UROD, with activity levels 10% of normal in both hepatocytes and erythrocytes.

All forms of PCT and HEP result in accumulation of uroporphyrin and intermediary carboxyl porphyrins in skin, subcutaneous tissues, and the liver. The most prominent clinical characteristics are cutaneous photosensitivity and

scarring on sun-exposed surfaces. Patients experience chronic blistering lesions resulting from mild trauma to sun-exposed areas. These fluid-filled vesicles rupture easily, become crusted, and heal slowly. Secondary infections can cause areas of hypo- or hyperpigmentation or sclerodermatous changes and may result in the development of alopecia at sites of repeated skin damage. Liver disease is common in patients with PCT as evidenced by abnormal liver function tests, with 30% to 40% of patients developing cirrhosis. In addition, there is an increased risk of hepatocellular carcinoma.

Reference Values

> or =1.0 RU (normal)

0.80-0.99 RU (indeterminate)

<0.80 RU (indicative of PCT type II)

RU = Relative Units

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

Alcohol ingestion within 24 hours of specimen collection may lead to a false-positive result.

Exposure of specimens to significant heat during the summer months may cause a decrease in the uroporphyrinogen decarboxylase (UROD) enzyme activity.

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. New York, McGraw-Hill, 2014. Accessed August 9, 2017. Available at <http://ommbid.mhmedical.com/content.aspx?bookid=971&Sectionid=62638866>
4. Doss MO, Kuhnel A, Gross U: Alcohol and porphyrin metabolism. *Alcohol* 2000;35(2):109-125

Performance

Method Description

This procedure is performed by incubation of lysed red cells with delta-aminolevulinic acid as the substrate followed by analysis of the porphyrins formed.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Tuesday, Thursday

Report Available

3 to 6 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
UPGD	UPG Decarboxylase, WB	49596-0

Result ID	Test Result Name	Result LOINC Value
8599	UPG Decarboxylase, WB	49596-0
606379	Interpretation (UPGD)	59462-2
606380	Reviewed By	18771-6