

Overview

Useful For

Preferred test to begin assessment for congenital erythropoietic porphyria and porphyria cutanea tarda and during symptomatic periods for acute intermittent porphyria, hereditary coproporphyria, and variegate porphyria when specimen transport **will not exceed 72 hours**

Genetics Test Information

This test is preferred test during symptomatic periods for acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), and variegate porphyria (VP). The random urine collection for this test allows for the diagnosis to be established and treatment to be initiated quickly. However, this test should only be ordered when the specimen will be received at MCL within 72 hours of collection. If it will be longer, PQNU / Porphyrins, Quantitative, 24 Hour, Urine should be ordered.

Testing includes porphobilinogen which is useful in the evaluation of the acute porphyrias.

This is the preferred test to begin assessment for congenital erythropoietic porphyria (CEP) and porphyria cutanea tarda (PCT).

Testing Algorithm

[The following algorithms are available in Special Instructions:](#)

-Porphyria (Acute) Testing Algorithm

-Porphyria (Cutaneous) Testing Algorithm

Special Instructions

- [The Heme Biosynthetic Pathway](#)
- [Porphyria \(Acute\) Testing Algorithm](#)
- [Porphyria \(Cutaneous\) Testing Algorithm](#)

Method Name

High-Performance Liquid Chromatography (HPLC) with Fluorometric Detection

Includes quantitation of coproporphyrins, uroporphyrins, and intermediate porphyrins (heptacarboxyl, hexacarboxyl, and pentacarboxyl).

Includes liquid chromatography-tandem mass spectrometry (LC-MS/MS) determination of porphobilinogen.

NY State Available

Yes

Specimen

Specimen Type

Urine

Advisory Information

This random urine test should be ordered when the specimen will reach MCL within 72 hours. If transportation will

take longer than 72 hours, order PQNU / Porphyrins, Quantitative, 24 Hour, Urine.

Shipping Instructions

[Ship specimen in amber bottle to protect from light.](#)

Necessary Information

Include a list of medications the patient is currently taking.

Specimen Required

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

Supplies: Urine Container - Amber, 60 mL (T596)

Container/Tube: Amber, 60-mL urine bottle (T596)

Specimen Volume: 20-50 mL

Collection Instructions: Collect a random urine specimen.

Forms

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

Specimen Minimum Volume

15 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Frozen	72 hours	LIGHT PROTECTED

Clinical and Interpretive

Clinical Information

The porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway. Depending on the specific enzyme involved, various porphyrins and their precursors accumulate in different specimen types. The patterns of porphyrin accumulation in erythrocytes and plasma and excretion of the heme precursors in urine and feces allow for the detection and differentiation of the porphyrias.

The porphyrias are typically classified as erythropoietic or hepatic based upon the primary site of the enzyme defect. In addition, hepatic porphyrias can be further classified as chronic or acute, based on their clinical presentation.

The primary acute hepatic porphyrias: acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), and variegate porphyria (VP), are associated with neurovisceral symptoms that typically onset during puberty or later.

Common symptoms include severe abdominal pain, peripheral neuropathy, and psychiatric symptoms. Crises may be precipitated by a broad range of medications (including barbiturates and sulfa drugs), alcohol, infection, starvation, heavy metals, and hormonal changes. Photosensitivity is not associated with AIP, but may be present in HCP and VP.

Cutaneous photosensitivity is associated with the chronic hepatic porphyrias: porphyria cutanea tarda (PCT) and the erythropoietic porphyrias; erythropoietic protoporphyria (EPP), X-linked dominant protoporphyria (XLDPP), and congenital erythropoietic porphyria (CEP). Although genetic in nature, environmental factors may exacerbate symptoms, significantly impacting the severity and course of disease.

CEP is an erythropoietic porphyria caused by uroporphyrinogen III synthase deficiency. Symptoms typically present in early infancy with red-brown staining of diapers, severe cutaneous photosensitivity with fluid-filled bullae and vesicles. Other common symptoms may include thickening of the skin, hypo- and hyperpigmentation, hypertrichosis, cutaneous scarring, and deformities of the fingers, eyelids, lips, nose, and ears. A few milder adult-onset cases have been documented as well as cases that are secondary to myeloid malignancies.

PCT is the most common form of porphyria and is most commonly sporadic (acquired) but in about 25% of cases it is inherited in an autosomal dominant manner. 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 and with 30% to 40% of patients developing cirrhosis. In addition, there is an increased risk of hepatocellular carcinoma.

Hepatoerythropoietic porphyria (HEP) is observed when an individual inherits PCT from both parents. Patients exhibit a similar clinical presentation to what is seen in CEP.

In addition, porphyrinuria may result from exposure to certain drugs and toxins or other medical conditions (ie, hereditary tyrosinemia type I). Heavy metals, halogenated solvents, various drugs, insecticides, and herbicides can interfere with heme production and cause "intoxication porphyria." Chemically, the intoxication porphyrias are characterized by increased excretion of, uroporphyrin and/or coproporphyrin in urine.

The workup of patients with a suspected porphyria is most effective when following a stepwise approach. See Porphyria (Acute) Testing Algorithm and Porphyria (Cutaneous) Testing Algorithm in Special Instructions or call 800-533-1710 to discuss testing strategies.

Reference Values

UROPORPHYRINS, OCTA

< or =30 nmol/L

HEPTACARBOXYLPORPHYRINS

< or =7 nmol/L

HEXACARBOXYLPORPHYRINS

< or =2 nmol/L

PENTACARBOXYLPORPHYRINS

< or =5 nmol/L

COPROPORPHYRINS, TETRA

< or =110 nmol/L

PORPHOBILINOGEN

< or =1.3 mcmmol/L

Interpretation

Abnormal results are reported with a detailed interpretation which 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 1 of the laboratory directors in case the referring physician has additional questions.

Cautions

Porphobilinogen (PBG) and porphyrins are susceptible to degradation at high temperature, at pH <5.0, and exposure to light.

Neither erythropoietic protoporphyria nor X-linked dominant protoporphyria are detected utilizing urine porphyrins and PBG measurements.

Ethanol and a variety of medications are known to interfere with heme synthesis leading to elevations in urine porphyrins, particularly coproporphyrin. Coproporphyrin elevation without concomitant PBG elevation should not be used as the basis for the diagnosis of porphyria, but may warrant follow-up testing with fecal porphyrin analysis.

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. AACCC 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>

Performance

Method Description

An aliquot of urine is acidified and mesoporphyrin is added as an injection marker. Porphyrins in the acidified urine are separated by HPLC and the eluted porphyrins are quantified by comparison of their fluorescence intensity to that of known porphyrin standards.(Ford RE, Ou C-N, Ellefson RD: Liquid chromatographic analysis for urinary porphyrins. *Clin Chem* 1981;27:397)

Porphobilinogen (PBG) in urine is quantified by liquid chromatography-tandem mass spectrometry after addition of stable isotope-labeled PBG internal standard and solid-phase extraction.(Ford RE, Magera MJ, Kloke KM, et al: Quantitative measurement of porphobilinogen in urine by stable-isotope dilution liquid chromatography-tandem mass

spectrometry. Clin Chem 2001 September;47[9]:1627-1632)

PDF Report

No

Day(s) and Time(s) Test Performed

Monday through Friday; 9 a.m.

Analytic Time

2 days (not reported Saturday or Sunday)

Maximum Laboratory Time

4 days

Specimen Retention Time

1 week

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

84110-Porphobilinogen, quantitative

84120-Porphyrins, quantitation and fractionation

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
PQNRU	Porphyrins, QN, Random, U	93707-8

Result ID	Test Result Name	Result LOINC Value
32332	Uroporphyrin, Octa	25166-0
32333	Heptacarboxylporphyrins	34314-5
32334	Hexacarboxylporphyrins	In Process
32335	Pentacarboxylporphyrins	34352-5
32336	Coproporphyrin, Tetra	25167-8



Result ID	Test Result Name	Result LOINC Value
32337	Porphobilinogen	2811-8
32338	Interpretation	49291-8