Overview

Useful For
Preferred screening test 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 be longer than 72 hours

Genetics Test Information
This test is preferred during symptomatic periods for acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), and variegate porphyria (VP) when specimen transport will be longer than 72 hours. If the specimen will be received at Mayo Clinic Laboratories within 72 hours of collection, PQNRU / Porphyrins, Quantitative, Random, Urine is recommended.

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
- Urine Preservatives-Collection and Transportation for 24-Hour Urine Specimens
- 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 24-hour urine test should be ordered when the specimen will not reach MCL within 72 hours. If the specimen
will reach MCL within 72 hours, order PQNRU / Porphyrins, Quantitative, Random, Urine.

**Shipping Instructions**

Ship specimen in amber container to protect from light.

**Necessary Information**

1. **24-Hour volume is required.**

2. Collection date and time should be documented upon completion of the 24-hour collection.

3. Include a list of medications the patient is currently taking.

4. See Urine Preservatives in Special Instructions for multiple collections.

**Specimen Required**

**Patient Preparation:** Patient should abstain from alcohol for 24 hours prior to, as well as during, collection.

**Supplies:**

Amber, 60-mL urine bottle (T596)

Sodium Carbonate, 5 gram (T272)

**Specimen Volume:** 20-50 mL

**Collection Instructions:**

1. Collect a 24-hour urine specimen.

2. Add 5 g of sodium carbonate (T272) as preservative at start of collection. This preservative is intended to achieve a pH of >7. **Do not substitute sodium bicarbonate for sodium carbonate.**

3. The container should be refrigerated and protected from light as much as possible during collection. An aliquot should be frozen when collection is complete.

**Forms**

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

**Urine Preservative Collection Options**

**Note:** The addition of preservative or application of temperature controls **must occur within 4 hours of completion** of the collection.

<table>
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<th>Preservative</th>
<th>Required</th>
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<tr>
<td>Ambient</td>
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<td>Refrigerate</td>
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<td>50% Acetic Acid</td>
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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 porphyrrias; 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.

Urinary porphyrin determination is helpful in the diagnosis of most porphyrias including CEP, PCT, AIP, HCP, and VP. In addition, measurement of porphobilinogen (PBG) in urine is important in establishing the diagnosis of the acute neurologic porphyrias (AIP, HCP and VP). Neither urine porphyrins nor PBG is helpful in evaluating patients suspected of having EPP or XLDPP.

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 (OCTACARBOXYL)**

< or =30 nmol/24 hours

**HEPTACARBOXYLPORPHYRINS**

< or =9 nmol/24 hours

**HEXACARBOXYLPORPHYRINS**

< or =8 nmol/24 hours

**PENTACARBOXYLPORPHYRINS**

< or =10 nmol/24 hours

**COPROPORPHYRINS (TETRACARBOXYL)**

Males: < or =230 nmol/24 hours

Females: < or =168 nmol/24 hours
PORPHOBILINOGEN

< or =2.2 mcmol/24 hours

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 one of the laboratory directors in case the referring physician has additional questions.

Cautions
Urine preservative should be used; 24-hour collections should be preserved by adding 5.0 g of sodium carbonate to a light-resistant collection container prior to beginning collection. Porphobilinogen (PBG) and porphyrins are susceptible to degradation at high temperature, at pH <5.0, and on 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


Performance

Method Description
An aliquot of urine is acidified and mesoporphyrin is added as an internal standard. Porphyrins in the acidified urine are separated by HPLC and the eluted porphyrins and internal standard 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)


PDF Report
No
Day(s) and Time(s) Test Performed
Monday through Friday; 7 a.m.

Analytic Time
2 days (not reported on 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

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