

Overview

Useful For
Monitoring treatment of patients with porphyria cutanea tarda

Reflex Tests

| Test Id | Reporting Name | Available Separately | Always Performed |
|---------|------------------------------|----------------------|------------------|
| PFP | Porphyrins, Fractionation, P | No | No |

Genetics Test Information
Plasma specimens from patients with active porphyria cutanea tarda, congenital erythropoietic porphyria, and erythropoietic protoporphyria may exhibit increased plasma porphyrin levels. However, a definitive diagnosis cannot be made by plasma analysis alone.

Testing Algorithm
If total porphyrins are above 1.0 mcg/dL, then porphyrin fractionation will be performed at an additional charge.

The following algorithms are available:
[-Porphyria \(Acute\) Testing Algorithm](#)
[-Porphyria \(Cutaneous\) Testing Algorithm](#)

Special Instructions

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

Highlights
Plasma porphyrins are most appropriate for monitoring patients being treated for porphyria cutanea tarda.

Analysis of plasma porphyrins is suitable for individuals with bullous dermatosis who are in kidney failure and unable to provide a urine specimen.

Method Name
PTP: Extraction and Scanning Spectrofluorometry
PFP: High-Performance Liquid Chromatography (HPLC)

NY State Available
Yes

Specimen

Specimen Type

Plasma

Shipping Instructions

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

Necessary Information

Include a list of medications the patient is currently taking.

Specimen Required

Patient Preparation: Patient **must not** consume any alcohol for 24 hours before specimen collection.

Supplies: Amber Frosted Tube, 5 mL (T915)

Collection Container/Tube:

Preferred: Green top (sodium or lithium heparin)

Acceptable: Lavender top (EDTA)

Submission Container/Tube: Amber vial

Specimen Volume: 3 mL

Collection Instructions:

1. Centrifuge specimen and aliquot plasma into amber vial.
2. Send plasma frozen.

Forms

[If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request](#) (T798) with the specimen.

Specimen Minimum Volume

1 mL

Reject Due To

| | |
|-----------------|----|
| Gross hemolysis | OK |
| Gross lipemia | OK |
| Gross icterus | OK |

Specimen Stability Information

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|---------|-------------------|
| Plasma | Frozen | 14 days | LIGHT PROTECTED |

Clinical & Interpretive

Clinical Information

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

These enzyme defects cause various porphyrins and their precursors to accumulate in different specimen types. The detection and differentiation of the porphyrias is through evaluation of the patterns of porphyrin accumulation observed in erythrocytes and plasma and of the heme precursors excreted in urine and feces.

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. A broad range of medications (including barbiturates and sulfa drugs), alcohol, infection, starvation, heavy metals, and hormonal changes may precipitate crises. Photosensitivity is not associated with AIP but may occur in HCP and VP.

Cutaneous photosensitivity is associated with the chronic hepatic porphyria, porphyria cutanea tarda (PCT), and the erythropoietic porphyrias including 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.

Congenital erythropoietic porphyria 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.

Porphyria cutanea tarda is the most common form of porphyria and caused by hepatic inhibition of the enzyme uroporphyrinogen decarboxylase (UROD). It is most often sporadic (acquired), but in about 20% of cases, a heterozygous variant in *UROD* increases the susceptibility to disease. The most prominent clinical characteristics are cutaneous photosensitivity and scarring on sun-exposed surfaces. Patients experience chronic blistering lesions; fluid filled vesicles that rupture easily become crusted and heal slowly, which result from mild trauma to sun-exposed areas. Secondary infections can cause areas of hypo- or hyperpigmentation or sclerodermatous changes and alopecia following repeated skin damage. Liver disease is common as evidenced by abnormal liver function tests, and 30% to 40% of patients with PCT develop cirrhosis. In addition, there is an increased risk of hepatocellular carcinoma.

Hepatoerythropoietic porphyria is a rare autosomal recessive form of porphyria caused by homozygous or compound heterozygous variants in *UROD*. It typically presents in early childhood with both erythropoietic and cutaneous manifestations and is similar to what is seen in CEP.

Clinical presentation of EPP and XLDPP is identical with onset of symptoms typically occurring in childhood. Cutaneous photosensitivity in sun-exposed areas of the skin generally worsens in the spring and summer months. Common symptoms may include itching, edema, erythema, stinging or burning sensations, and occasionally scarring of the skin in sun-exposed areas.

Plasma porphyrins are most appropriate for monitoring treatment of PCT. Although analysis in plasma is not recommended for diagnosis, increases in plasma porphyrin concentrations are observed in the cutaneous porphyrias and may be elevated during acute episodes of AIP, VP, and HCP. In addition, persons in chronic kidney failure who

develop bullous dermatosis similar to that associated with PCT may have increased plasma porphyrins.

The workup of patients with a suspected porphyria is most effective when following a stepwise approach.

The following algorithms are available or call 800-533-1710 to discuss testing strategies:

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

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

Reference Values

< or =1.0 mcg/dL

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

Cautions

Plasma porphyrins, especially protoporphyrin, are extremely sensitive to light and may degrade to normal levels if not handled properly.

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. AACCC Press; 2010:307-324
2. 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 September 6, 2024. Available at <https://ommbid.mhmedical.com/content.aspx?sectionid=225540906&bookid=2709>
3. Weiss Y, Chen B, Yasuda M, Nazarenko I, Anderson KE, Desnick RJ. Porphyria cutanea tarda and hepatoerythropoietic porphyria: Identification of 19 novel uroporphyrinogen III decarboxylase mutations. Mol Genet Metab. 2019;128(3):363-366. doi:10.1016/j.ymgme.2018.11.013
4. Phillips JD. Heme biosynthesis and the porphyrias. Mol Genet Metab. 2019;128(3):164-177. doi:10.1016/j.ymgme.2019.04.008

Performance

Method Description

The plasma porphyrins profile is performed as a 2-step analysis. First, the total plasma porphyrins concentration is determined by extracting the porphyrins from plasma with a mixture of ethyl acetate and acetic acid. The porphyrins are then back extracted into dilute hydrochloric acid. Total porphyrins are quantified on this extract via scanning spectrofluorometry. If the total plasma porphyrin concentration is elevated, the extract is submitted for high-performance liquid chromatography analysis, whereby the individual porphyrin analytes are separated by differential partitioning between a stationary aliphatic surface and a moving aqueous buffer solution. As the porphyrins emerge separately from the column, the quantity of each is monitored with a fluorescence detector.(Unpublished Mayo

method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

2 to 4 days

Specimen Retention Time

14 days

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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

84311-Porphyrins, total
82542-Porphyrins, fractionation (if appropriate)

LOINC® Information

| Test ID | Test Order Name | Order LOINC® Value |
|---------|----------------------|--------------------|
| PTP | Porphyrins, Total, P | 2815-9 |

| Result ID | Test Result Name | Result LOINC® Value |
|-----------|----------------------|---------------------|
| 8731 | Porphyrins, Total, P | 2815-9 |
| 34252 | Reviewed By | 18771-6 |
| 33869 | Interpretation | 59462-2 |