MAYO CLINIC LABORATORIES

Test Definition: PTP

Porphyrins, Total, Plasma

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: 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: For 24 hours before specimen collection, patient **must not** consume any alcohol.
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 Plasma
Collection Instructions: Centrifuge specimen and aliquot plasma into an amber vial.

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

Specimen Minimum Volume
Plasma: 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 (preferred)	14 days	LIGHT PROTECTED
	Refrigerated	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 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. See the Heme Biosynthetic Pathway

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 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 protoporphyria (XLP), 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 genetic variant in *UROD* increases susceptibility to the disorder. 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 alopecia may develop 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.

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 XLP 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 with chronic kidney failure who develop bullous dermatosis similar to that associated with PCT may have increased plasma porphyrins.

A stepwise approach is typically most effective when evaluating patients with suspected porphyria. See Porphyria (Acute) Testing Algorithm and Porphyria (Cutaneous) Testing Algorithm or call 800-533-1710 to discuss testing strategies.

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. AACC 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 May 28, 2026. 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