MAYO CLINIC LABORATORIES

Test Definition: UPGD

Uroporphyrinogen Decarboxylase, Whole Blood

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 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 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.

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 (sporadic), the most common form of PCT, exhibits normal erythrocyte 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 **must not** consume any alcohol for 24 hours before specimen collection. This is essential 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: 4 mL

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:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

3 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

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

Clinical & 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 inhibition of hepatocyte or erythrocyte uroporphyrinogen decarboxylase (UROD; see The Heme Biosynthetic Pathway). 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 less than 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 =1.00 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 enzyme activity.

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. Nuttall KL, Klee GG. Analytes of hemoglobin metabolism-porphyrins, iron, and bilirubin. In: Burtis CA, Ashwood ER, eds. Tietz Textbook of Clinical Chemistry. 5th ed. WB Saunders Company; 2001:584-607
3. 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 April 22, 2024. Available at https://ommbid.mhmedical.com/content.aspx?sectionid=225540906&bookid=2709
4. Singal AK. Porphyria cutanea tarda: Recent update. Mol Genet Metab. 2019;128(3):271-281. doi:10.1016/j.ymgme.2019.01.004

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Performance

Method Description
This procedure is performed by incubation of lysed red blood 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

Report Available
3 to 9 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
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