Overview

Useful For
Diagnosis of congenital erythropoietic porphyria

Genetics Test Information
This test is not appropriate for assessment of acute abdominal pain.

Highlights
Congenital erythropoietic porphyria (CEP) is a disease usually seen in pediatric patients.

In our testing experience over the last 10 years, fewer than 5 adult patients have been diagnosed with CEP associated with a myelodysplastic syndrome.

Testing Algorithm
The following algorithms are available in Special Instructions:

- Porphyria (Acute) Testing Algorithm
- Porphyria (Cutaneous) Testing Algorithm

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)

NY State Available
Yes

Specimen

Specimen Type
WB Heparin

Advisory Information
This test is most appropriately used for pediatric patients.

This test measures uroporphyrinogen (UPG) III synthase to confirm congenital erythropoietic porphyria, which is typically seen in early infancy. It does not measure UPG I synthase (also known as porphobilinogen deaminase), the enzyme deficient in acute intermittent porphyria (AIP). For AIP (and UPG I synthase), order PBGD_ / Porphobilinogen Deaminase, Whole Blood.

Necessary Information
Include a list of medications the patient is currently taking.
Specimen Required
All porphyrin tests on erythrocytes can be performed on 1 draw tube.

Patient Preparation: Patient should abstain from alcohol for 24 hours.

Container/Tube: Green top (heparin)

Specimen Volume: Full tube

Collection Instructions: Immediately place specimen on wet ice.

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 in Special Instructions:
   - Informed Consent for Genetic Testing (T576)
   - Informed Consent for Genetic Testing-Spanish (T826)

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

Specimen Minimum Volume
3 mL

Reject Due To
| Gross hemolysis | Reject |

Specimen Stability Information

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<th>Temperature</th>
<th>Time</th>
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<tr>
<td>WB Heparin</td>
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Clinical and Interpretive

Clinical Information
The porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway. Congenital erythropoietic porphyria (CEP) is an extremely rare, autosomal recessive porphyria that typically presents in early infancy. Also known as Gunther disease, CEP results from a deficiency of uroporphyrinogen III (co-) synthase (UROIIIS). In most cases, the disorder is suggested during the first few days or weeks of life by pink, violet, or brown urinary staining of diapers. Clinical symptoms include hemolytic anemia, hepatosplenomegaly, skin photosensitivity, scarring and blistering, red or brown dental discoloration (erythrodontia), and hypertrichosis (excess body hair). Growth and cognitive developmental delays are commonly observed in individuals with CEP. A few cases of adult-onset CEP have been reported, typically associated with a myelodysplastic syndrome.

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

Reference Values
> or =75 Relative Units (normal)

See The Heme Biosynthetic Pathway in Special Instructions.

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
This test is **not** useful for ruling out acute intermittent porphyria (AIP) a disorder caused by decreased uroporphyrinogen I synthase (also known as porphobilinogen deaminase). For AIP, order PBGD_ / Porphobilinogen Deaminase, Whole Blood.

This test does not reliably distinguish between individuals who are carriers for congenital erythropoietic porphyria (CEP), and are at risk for having an affected child.

If possible, specimens from patients suspected of having CEP should be drawn prior to blood transfusions; uroporphyrinogen (UPG) III synthase activity in transfused erythrocytes can cause false-negative results.

Abstinence from alcohol for at least 24 hours is essential for accurate results. While the effects of alcohol on this enzyme have not yet been determined, alcohol is known to suppress or induce other enzymes in the heme biosynthetic pathway.

Clinical Reference

Performance

Method Description
Washed cells are incubated with aminolevulinic acid as substrate and the series I and III porphyrin isomers formed are measured. The proportion of series III isomers formed in relation to total porphyrins (I + III isomers) represents the uroporphyrinogen III synthase activity. The values are reported as Relative Units.(Unpublished Mayo method)
PDF Report
No

Day(s) and Time(s) Test Performed
Varies

Analytic Time
7 days (Not reported on Saturday or Sunday)

Maximum Laboratory Time
14 days

Specimen Retention Time
2 weeks

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
82657

LOINC® Information

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<td>Uroporphyrinogen III Synthase, RBC</td>
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