Overview

Useful For
This test is the preferred test for the confirmation of a diagnosis of aminolevulinic acid dehydratase deficiency porphyria.

Genetics Test Information
Aminolevulinic acid dehydratase (ALAD) activity can be inhibited in situations including hereditary tyrosinemia type 1, lead intoxication, and exposure to styrene, trichloroethylene, or bromobenzene. These causes should be ruled out when considering a diagnosis of ALAD deficiency porphyria (ADP). This method will not exhibit a decreased ALAD enzyme activity due to lead intoxication.

Due to longer specimen stability, this test is the preferred test for analysis of ALAD.

This test will not detect lead intoxication.

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
Enzymatic End point/Spectrofluorometric

NY State Available
Yes

Specimen

Specimen Type
Whole blood

Specimen Required
All porphyrin tests on whole blood can be performed on 1 draw tube.

Container/Tube:

Preferred: Green top (sodium heparin)

Acceptable: Lavender top (EDTA) or green top (lithium heparin)
Test Definition: ALAD
ALA Dehydratase, WB

Specimen Volume: Full tube

Collection Instructions:
1. Patient should abstain from alcohol for 24 hours.
2. Immediately place specimen on wet ice.

Additional Information: Include a list of medications the patient is currently taking.

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>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
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Clinical and Interpretive

Clinical Information
Porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway. A defect in the second enzyme of this pathway causes 5-aminolevulinic acid (ALA) dehydratase (ALAD) deficiency porphyria (ADP). A marked deficiency of ALAD causes the accumulation and subsequent urinary excretion of large amounts of ALA. Urinary porphobilinogen (PBG) remains essentially normal, which rules out other forms of acute porphyria.

ADP is an autosomal recessive acute hepatic porphyria that produces neurologic symptoms similar to those seen in acute intermittent porphyria. Symptoms include acute abdominal pain, peripheral neuropathy, nausea, vomiting, constipation, and diarrhea. Respiratory impairment, seizures, and psychosis are possible during an acute period. ADP is extremely rare with only 7 cases described in the literature since 1979.

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

Reference Values
Reference ranges have not been established for patients who are <16 years of age.

> or =4.0 nmol/L/sec
3.5-3.9 nmol/L/sec (indeterminate)
<3.5 nmol/L/sec (diminished)

Interpretation
Abnormal results are reported with a detailed interpretation including an overview of the results and their significance, a correlation to available clinical information provided with the specimen, differential diagnosis, and 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 assay is not useful in assessment of lead intoxication as it reactivates aminolevulinic acid dehydratase (ALAD) that has been inhibited by lead. The preferred test for lead toxicity is PBBD / Lead with Demographics, Blood.

Abstinence from alcohol is essential for at least 24 hours prior to specimen collection as ethanol suppresses ALAD activity.

False-positive values may result from enzyme degradation due to improper specimen handling. It is essential to adhere to instructions outlined in the Specimen Required and the Specimen Stability Information fields.

Clinical Reference


Performance

Method Description

PDF Report
No
Day(s) and Time(s) Test Performed
Tuesday, Thursday; 1 p.m.

Analytic Time
3 days (not reported on Saturday or Sunday)

Maximum Laboratory Time
4 days

Specimen Retention Time
14 days

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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