Overview

Useful For
Preferred confirmation test for the diagnosis of aminolevulinic acid dehydratase deficiency porphyria
This test is not useful for detecting lead intoxication.

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.

Testing Algorithm
The following algorithms are available in Special Instructions:
- Porphyria (Acute) Testing Algorithm
- Porphyria (Cutaneous) Testing Algorithm
- The Heme Biosynthetic Pathway

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

Ordering Guidance
This assay is not useful in assessment of lead intoxication as it reactivates aminolevulinic acid dehydratase that has been inhibited by lead. The preferred test for lead toxicity is PBDB / Lead with Demographics, Blood.

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

Specimen Required
Patient Preparation: Abstinence from alcohol is essential for at least 24 hours prior to specimen collection as ethanol
suppresses aminolevulinic acid dehydratase (ALAD) activity, leading to false-positive results.

Container/Tube:
Preferred: Green top (sodium heparin)
Acceptable: Lavender top (EDTA) or green top (lithium heparin)
Specimen Volume: Full tube 4 mL
Collection Instructions: Immediately place specimen on wet ice.

Forms
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)

Reject Due To
Gross hemolysis  Reject

Specimen Minimum Volume
3 mL

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
</tr>
</thead>
<tbody>
<tr>
<td>Whole blood</td>
<td>Refrigerated (preferred)</td>
<td>7 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Ambient</td>
<td>4 days</td>
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</tbody>
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Clinical & 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.

**Cautions**
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**
Measurement of aminolevulinic acid (ALA) dehydratase (ALAD) activity is based on the rate of synthesis of uroporphyrin from ALA in incubated, lysed erythrocytes. Low yield of uroporphyrin from ALA indicates a deficiency of ALAD. (Unpublished Mayo method)

**PDF Report**
No

**Specimen Retention Time**
14 days

**Performing Laboratory Location**
Rochester

**Fees & Codes**

**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 US Food and Drug Administration.

**CPT Code Information**
82657