Overview

Useful For
Assistance in the differential diagnosis of the various acute hepatic porphyrias

Testing Algorithm
The following algorithms are available in Special Instructions:

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

Special Instructions

The Heme Biosynthetic Pathway
- Porphyria (Acute) Testing Algorithm
- Porphyria (Cutaneous) Testing Algorithm

Method Name
Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

NY State Available
Yes

Specimen

Specimen Type
Urine

Advisory Information
The preferred test for lead toxicity in children is blood lead (see PBDV / Lead, Venous, with Demographics, Blood or PBDC / Lead, Capillary, with Demographics, Blood).

Necessary Information
Patient's age is required.

Specimen Required

Patient Preparation: Patient should abstain from alcohol for 24 hours prior to and during testing.

Supplies: Urine Tubes, 10 mL (T068)

Specimen Volume: 2 mL

Collection Instructions: Collect a random urine specimen.

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

Specimen Minimum Volume
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>Urine</td>
<td>Refrigerated (preferred)</td>
<td>28 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Frozen</td>
<td>45 days</td>
<td></td>
</tr>
</tbody>
</table>

Clinical and Interpretive

Clinical Information

The porphyrias are a group of inherited disorders resulting from enzyme defects in the heme biosynthetic pathway. Depending on the specific enzyme involved, various porphyrins and their precursors 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 in Special Instruction for more information.

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: aminolevulinic acid dehydratase deficiency porphyria (ADP), 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. A broad range of medications (including barbiturates and sulfa drugs), alcohol, infection, starvation, heavy metals, and hormonal changes may precipitate crises. Photosensitivity is not associated with AIP, but may be present in HCP and VP.

The excretion of aminolevulinic acid (ALA) can be increased due to one of the inherited acute porphyrias or due to secondary inhibition of ALA dehydratase. Among the secondary causes, acute lead intoxication results in the greatest increases of aminolevulinic aciduria. Less significant elevations are seen in chronic lead intoxication, tyrosinemia type I, alcoholism, and pregnancy.

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

Reference Values

<1 year: < or =10 nmol/mL

1-17 years: < or =20 nmol/mL

> or =18 years: < or =15 nmol/mL

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**

No significant cautionary statements

**Clinical Reference**


**Performance**

**Method Description**

Aminolevulinic acid (ALA) is determined by liquid chromatography-tandem mass spectrometry (LC-MS/MS) stable isotope dilution analysis. The urine is mixed with an internal standard (5-aminolevulinic acid, 13C5, 15N, ALA-IS) and filtered using a 0.2 mcM nylon filter vial. The ratios of the extracted peak areas of ALA to ALA-IS determined by LC-MS/MS are used to calculate the concentration of ALA present in the sample. (Lacey, JM, Magera MJ, and Tortorelli S: Delta Aminolevulinic Acid Quantitation in Urine by LC-MS/MS. J Am Soc Mass Spectrom 2011;22, S1:pp 69)

**PDF Report**

No

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

Tuesday; 3 p.m.

**Analytic Time**

3 days (not reported Saturday or Sunday)

**Maximum Laboratory Time**

6 days

**Specimen Retention Time**

14 days

**Performing Laboratory Location**

Rochester

**Fees and Codes**

Fees
Test Definition: ALAUR
Aminolevulinic Acid, U

- 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
82135

LOINC® Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>ALAUR</td>
<td>Aminolevulinic Acid, U</td>
<td>34284-0</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>61547</td>
<td>Aminolevulinic Acid, U</td>
<td>34284-0</td>
</tr>
<tr>
<td>34347</td>
<td>Interpretation (ALA), U</td>
<td>59462-2</td>
</tr>
<tr>
<td>34348</td>
<td>Reviewed By</td>
<td>18771-6</td>
</tr>
</tbody>
</table>