Test Definition: ALAUR

Aminolevulinic Acid, U

Overview

Useful For

Assistance in the differential diagnosis of the 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

Ordering Guidance

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

1 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Refrigerated (preferred)	28 days	
	Frozen	45 days	

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 following algorithms are available in Special Instructions or call 800-533-1710 to discuss testing strategies:

-Porphyria (Acute) Testing Algorithm

-Porphyria (Cutaneous) Testing Algorithm

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

1. Tortorelli S, Kloke K, Raymond K: Disorders of porphyrin metabolism. In: Dietzen DJ, Bennett MJ, Wong ECC, eds. Biochemical and Molecular Basis of Pediatric Disease. 4th ed. AACC Press; 2010:chap 15.

2. 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 September 04, 2020. Available at https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225540906

3. 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

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, 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) Performed

Tuesday, Thursday

Report Available

3 to 7 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

82135

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
ALAUR	Aminolevulinic Acid, U	34284-0

Result ID	Test Result Name	Result LOINC Value
61547	Aminolevulinic Acid, U	34284-0
34347	Interpretation (ALA), U	59462-2
34348	Reviewed By	18771-6