

Overview

Useful For

Assisting in the differential diagnosis of the acute hepatic porphyrias

Testing Algorithm

For more information see:

[-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; order either 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 not consume any alcohol for 24 hours before specimen collection.

Supplies: Urine Tubes, 10 mL (T068)

Collection Container/Tube: Clean, plastic urine collection container

Submission Container/Tube: Plastic vial

Specimen Volume: 2 mL

Collection Instructions: Collect a random urine specimen.

Forms

[If not ordering electronically, complete, print, and send a Biochemical Genetics 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 & 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. For more information see [The Heme Biosynthetic Pathway](#).

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 it 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. Once the biochemical diagnosis of an acute porphyria is established, molecular genetic testing is available (APGP / Acute Porphyria Gene Panel, Varies), which allows for diagnosis of at-risk family members.

For more information, see the following or call 800-533-1710 to discuss testing strategies:

- [The Heme Biosynthetic Pathway](#)
- [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, White A, Raymond K. Disorders of porphyrin metabolism. In: Dietzen DJ, Bennett MJ, Wong ECC, Haymond S, eds. Biochemical and Molecular Basis of Pediatric Disease. 5th ed. Academic Press; 2021:503-528
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 July 5, 2024. Available at <https://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=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
4. Anderson KE, Lobo R, Salazar D, et al. Biochemical diagnosis of acute hepatic porphyria: Updated expert recommendations for primary care physicians. Am J Med Sci. 2021;362(2):113-121. doi:10.1016/j.amjms.2021.03.004

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 ([13]C5, [15]N ALA-IS) and filtered using a 0.2 µm 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

Mayo Clinic Laboratories - Rochester Main Campus

Fees & 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 account representative. 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. It has not been cleared or approved by the US 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