MAYO CLINIC LABORATORIES

Test Definition: ALADW

Aminolevulinic Acid Dehydratase, Washed Erythrocytes

Overview

Useful For

Confirmation of a diagnosis of aminolevulinic acid dehydratase deficiency porphyria using washed erythrocyte specimens

This test is **not useful for** detecting lead intoxication.

Genetics Test Information

Aminolevulinic acid dehydratase (ALAD) activity is inhibited in other 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. This method will not detect a decreased ALAD enzyme activity due to lead intoxication.

This test will not detect lead intoxication.

Testing Algorithm

The following algorithms are available:
-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

Washed RBC

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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 PBDV / Lead, Venous, with Demographics, Blood.

Necessary Information

1. Volume of packed cells and total volume of specimen (red cells + saline) are required and must be sent with specimen.

2. **Patient's age is required**

3. Include a list of medications the patient is currently taking.

Specimen Required

Patient Preparation: Patient **must not** consume any alcohol for 24 hours before specimen collection. This is essential as ethanol suppresses aminolevulinic acid dehydratase activity, leading to false-positive results.

Collection Container/Tube:

Preferred: Green top (sodium heparin)

Acceptable: Lavender top (EDTA) or green top (lithium heparin)

Submission Container/Tube: Plastic vial

Specimen Volume: Entire washed erythrocyte suspension

Collection Instructions: Collect and process whole blood specimen as follows:

1. Transfer entire specimen to a 12-mL graduated centrifuge tube.
2. Centrifuge specimen at 4 degrees C for 10 minutes at 2000 rpm.
3. **Record volume of packed cells and the total volume of the specimen.**
4. Discard supernatant plasma.
5. Wash packed erythrocytes 2 times by resuspension of at least an equal amount of cold 0.9% saline, mix, and centrifuge for 5 minutes at 2000 rpm, discarding supernatant after each washing.
6. Resuspend packed cells to the original total volume with 0.9% saline. Invert specimen gently to mix.

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:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

Specimen Minimum Volume

1 mL of washed and resuspended erythrocytes

Reject Due To

Cell suspension not available	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Washed RBC	Frozen (preferred)	7 days	

	Ambient	4 days	
	Refrigerated	14 days	

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 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 8 cases described in the literature since 1979.

The workup of patients with a suspected porphyria is most effective when following a stepwise approach. Molecular confirmation is available; order CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify ALAD Gene List ID: IEMCP-D81317. See Porphyria (Acute) Testing Algorithm or call 800-533-1710 to discuss testing strategies.

Reference Values

Reference ranges have not been established for patients who are younger than 16 years.

> 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

1. Tortorelli S, Kloke K, Raymond K. Disorders of porphyrin metabolism. In: Dietzen DG, Bennett MJ, Wong ECC, eds. Biochemical and Molecular Basis of Pediatric Disease. 4th ed. AACC Press; 2010:307-324
2. 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
3. Anderson KE, Sassa S, Bishop DF, Desnick RJ. Disorders of heme biosynthesis: X-linked sideroblastic anemia and the

porphyrias. In: Valle D, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill, 2019. Accessed April 19, 2024. Available at https://ommbid.mhmedical.com/content.aspx?sectionid=225540906&bookid=2709
4. Lahiji AP, Anderson KE, Chan A. 5-Aminolevulinate dehydratase porphyria: Update on hepatic 5-aminolevulinic acid synthase induction and long-term response to hemin. Mol Genet Metab. 2020;131(4):418-423. doi:10.1016/j.ymgme.2020.10.011

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

Day(s) Performed
Tuesday

Report Available
2 to 8 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

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82657

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
ALADW	ALA Dehydratase, RBC	2813-4

Result ID	Test Result Name	Result LOINC® Value
31946	ALA Dehydratase	2813-4
31948	Interpretation	59462-2
BG573	Total Cell Suspension	94496-7
BG574	Packed Cell Volume	94497-5
606469	Reviewed By	18771-6