Test Definition: ALAD

ALA Dehydratase, WB

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)

Specimen Minimum Volume

3 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	7 days	
	Ambient	4 days	

Clinical and 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

1. Tortorelli S, Kloke K, Raymond K: Chapter 15: Disorders of porphyrin metabolism. In Biochemical and Molecular Basis of Pediatric Disease. Fourth edition. Edited by DJ Dietzen, MJ Bennett, ECC Wong. AACC Press 2010, pp 307-324

2. Nuttall KL, Klee GG: Analytes of hemoglobin metabolism-porphyrins, iron, and bilirubin. In Tietz Textbook of Clinical Chemistry. Fifth edition. Edited by CA Burtis, ER Ashwood. WB Saunders Company, 2001, pp 584-607

3. Anderson KE, Sassa S, Bishop DF, Desnick RJ: Disorders of Heme Biosynthesis: X-Linked Sideroblastic Anemia and the Porphyrias In The Online Metabolic and Molecular Bases of Inherited Disease. Edited by D Valle, AL Beaudet, B Vogelstein, et al. McGraw-Hill, Accessed August 9, 2017 Available at http://ommbid.mhmedical.com/content.aspx?bookid=971&Sectionid=62638866

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, Thursday

Report Available

2 to 4 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 US Food and Drug Administration.

CPT Code Information

82657

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
ALAD	ALA Dehydratase, WB	12916-3

Result ID	Test Result Name	Result LOINC Value
4021	ALA Dehydratase	12916-3
28399	Interpretation	59462-2
606468	Reviewed By	18771-6