MAYO CLINIC LABORATORIES

Test Definition: AACYL

Aminoacylase-1 Deficiency, Urine

Overview

Useful For

Follow-up quantitation of abnormal organic acid elevations of N-acetylated amino acids, in particular N-acetylalanine, N-acetylglycine, N-acetylmethionine, and N-acetylglutamic acid

Diagnosis of individuals with aminoacylase-1 deficiency

Evaluating patients with neurologic and psychiatric symptoms of unknown etiology

Special Instructions

- Biochemical Genetics Patient Information

Highlights

This test provides a quantitative analysis of N-acetyl alanine, N-acetylglycine, N-acetylmethionine, and N-acetylglutamic acid for the diagnosis of aminoacylase-1 deficiency.

Method Name

Gas Chromatography Mass Spectrometry (GC-MS)

NY State Available

Yes

Specimen

Specimen Type

Urine

Necessary Information

1. Patient's age and sex are required.

2. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 10 mL

Pediatric: If insufficient collection volume, submit as much specimen as possible in a single container; the laboratory will determine if volume is sufficient for testing.

Collection Instructions:

1. Collect a random urine specimen (first morning void preferred).
2. No preservative.

Forms

1. Biochemical Genetics Patient Information (T602)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

3 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	Frozen (preferred)	90 days	
	Refrigerated	60 days	

Clinical & Interpretive

Clinical Information

Aminoacylase-1 deficiency (ACY1D) is a very rare autosomal recessive metabolic disorder caused by disease-causing variants in the *ACY1* gene and characterized by increased urinary excretion of N-acetylated amino acids, including the derivatives of serine, glutamine, alanine, methionine, glycine, leucine, and valine.(1) The phenotype is variable with less than 20 patients described in the literature. Clinical findings range from asymptomatic to significant neurologic impairments including intellectual disability, seizures, sensorineural hearing loss, and behavioral features such as attention deficit hyperactivity disorder and autism.(2) According to a recent paper, the symptoms described in the literature vary widely, thus making the relationship between clinical symptomatology and ACY1D yet unclear.(3)

For patients with a clinical suspicion of ACY1D, the biochemical diagnosis is established via elevated N-acetylated amino acids in urine. For confirmation, molecular genetic testing of the *ACY1* gene is available; order CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify Gene List ID: IEMCP-M9821N.

Reference Values

N-acetylglycine: < or =15.00 mmol/mol creatinine
N-acetyl alanine: < or =10.00 mmol/mol creatinine
N-acetylglutamic acid: < or =20.00 mmol/mol creatinine
N-acetylmethionine: < or =5.00 mmol/mol creatinine

Interpretation

When abnormal results are detected, a detailed interpretation is given including an overview of the results and of their significance; a correlation to available clinical information; elements of differential diagnosis; recommendations for additional biochemical testing and in vitro confirmatory studies (enzyme assay, molecular analysis).

Cautions

No significant cautionary statements

Clinical Reference

1. van Coster R. Aminoacylase I deficiency. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill Education; 2019. Accessed October 17, 2024. Available at https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225896774
2. Alessandri MG, Milone R, Casalini C, et al. Four years follow up of ACY1 deficient patient and pedigree study. Brain and Dev. 2018;40(7):570-575
3. Smolka V, Friedecky D, Kolarova J, et al. Aminoacylase 1 deficiency: case report on three affected siblings. AME Case Rep. 2023;8:18. doi:10.21037/acr-23-46
4. Sass JO, Mohr V, Olbrich H, et al. Mutations in ACY1, the gene encoding aminoacylase 1, cause a novel inborn error of metabolism. Am J Hum Genet. 2006;78(3):401-409. doi:10.1086/500563

Performance

Method Description

Urine volumes equivalent to 0.25 to 0.50 mg of creatinine are spiked with the mixture of labeled internal standards, allowed to equilibrate, acidified, and then extracted. After evaporation, the dry residue is derivatized to butyl esters. Specimens are analyzed by capillary gas chromatography mass spectrometry selected ion monitoring using ammonia chemical ionization and a stable isotope dilution method.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday

Report Available

3 to 9 days

Specimen Retention Time

2 months

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

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
AACYL	Aminoacylase-1 Deficiency, U	104695-2

Result ID	Test Result Name	Result LOINC® Value
621432	Interpretation	59462-2
621428	N-acetylglycine	104696-0
621429	N-acetylalanine	104697-8
621430	N-acetylglutamic acid	104698-6
621431	N-acetylmethionine	104699-4
621433	Reviewed By	18771-6