Test Definition: NAACD

N-Acetylaspartic Acid, Canavan Disease, Random, Urine

Overview

Useful For

Diagnosis and monitoring of individuals with Canavan disease

Follow-up quantitation of abnormal organic acid elevations of N-acetylaspartic acid

Special Instructions

- Biochemical Genetics Patient Information

Highlights

This test provides quantitative analysis of N-acetylaspartic acid in urine for the diagnosis and monitoring of individuals with Canavan disease.

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	
	Ambient	14 days	
	Refrigerated	90 days	

Clinical & Interpretive

Clinical Information
Canavan disease is a leukodystrophy disorder characterized by delayed development beginning at age 3 to 6 months followed by regression, head lag, macrocephaly, and hypotonia. Although the phenotypic spectrum ranges in severity, death usually occurs within the first two decades of life. Affected patients receive symptomatic care and supportive management. While there is currently no treatment available for Canavan disease, clinical trials may be available.

The incidence of Canavan disease unknown, although there is a higher incidence in the Ashkenazi Jewish population. Canavan disease is inherited in an autosomal recessive manner and is caused by disease-causing variants in the *ASPA* gene. This leads to a deficiency of the enzyme aspartoacylase and increased urinary excretion of N-acetylaspartic acid (NAA). For patients with a clinical suspicion of Canavan disease, the biochemical diagnosis is established via elevated NAA in urine. Molecular genetic testing of the *ASPA* gene is available for confirmation and testing of at-risk family members to provide assessment for recurrence risks; order CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify Gene List ID: IEMCP-TF8VHL.

Reference Values
< or =50.00 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. Bley A, Denecke J, Kohlschutter A, et al. The natural history of Canavan disease: 23 new cases and comparison with patients from literature. Orphanet J Rare Dis. 2021;16(1):227. doi:10.1186/s13023-020-01659-3
2. Nagy A, Bley AE, Eichler F. Canavan disease. In: Adam MP, Feldman J, Mirzaa GM, et al., eds. GeneReviews [Internet]. University of Washington, Seattle; 1999. Updated December 21, 2023. Available at www.ncbi.nlm.nih.gov/books/NBK1234/

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.

MAYO CLINIC LABORATORIES

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CPT Code Information

83921

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
NAACD	N-acetylaspartic acid, U	76023-1

Result ID	Test Result Name	Result LOINC® Value
621425	Interpretation	59462-2
621424	N-acetylaspartic acid	76023-1
621426	Reviewed By	18771-6