MAYO CLINIC LABORATORIES

Test Definition: OAUS

Organic Acids Screen, Urine Spot

Overview

Useful For
Diagnosis of inborn errors of metabolism using dried urine specimens

Special Instructions
- Biochemical Genetics Patient Information

Highlights
This test provides a qualitative report of abnormal levels of organic acids identified via gas chromatography mass spectrometry.

Diagnostic specificity of inborn errors of metabolism via urine organic acids analysis is variable due to factors such as specimen collection when the patient is asymptomatic versus acutely ill, taking dietary supplements, or anabolic versus catabolic status of the patient.

Method Name
Gas Chromatography Mass Spectrometry (GC-MS)

NY State Available
Yes

Specimen

Specimen Type
Urine

Ordering Guidance
The preferred screening test for organic acidemias is OAU / Organic Acids Screen, Random, Urine, which should be ordered unless collecting or sending urine is difficult or not feasible.

Necessary Information
1. Patient's age is required.
2. Biochemical Genetics Patient Information (T602) is recommended, but not required, to be filled out and sent with the specimen to aid in the interpretation of test results.

Specimen Required
Supplies: Card-Blood Spot Collection (Filter Paper) (T493)
Collection Container:
Preferred: Blood Spot Collection Card
Acceptable: Whatman Protein Saver 903 paper, PerkinElmer 226 filter paper, Munktell filter paper

Specimen Volume: 1 Filter paper card soaked with urine, typically requires 2 to 3 mL of urine

Collection Instructions

1. Soak a filter paper card with urine, approximately 22 cm(2) in area (typically requires 2 to 3 mL of urine; exact measurement is not important as urine volume will be normalized to creatinine).

Note: Filter paper sample may be collected by dipping the card into a collection cup of urine. Avoid dilute urine if possible.

2. Let urine dry on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

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

2 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)	28 days	FILTER PAPER
	Ambient	14 days	FILTER PAPER
	Refrigerated	14 days	FILTER PAPER

Clinical & Interpretive

Clinical Information

Organic acids occur as physiologic intermediates in a variety of metabolic pathways. Organic acidurias are a group of disorders in which one or more of these pathways are blocked, resulting in a deficiency of normal products and an abnormal accumulation of intermediate metabolites (organic acids) in the body. These excess metabolites are excreted in the urine.

The incidence of individual inborn errors of organic acid metabolism varies from 1 in 10,000 to greater than 1 in 1,000,000 live births. Collectively, their incidence approximates 1 in 3000 live births. This estimate, however, does not include other inborn errors of metabolism (ie, amino acid disorders, urea cycle disorders, congenital lactic acidemias) for which diagnosis and monitoring may require organic acid analysis. If all possible disease entities were included, the incidence of conditions where informative organic acid profiles could be detected in urine is likely to approach 1 in 1000 live births.

Organic acidurias typically present with either an acute life-threatening illness in early infancy or unexplained developmental delay with intercurrent episodes of metabolic decompensations in later childhood. A situation of severe and persistent metabolic acidosis of unexplained origin, elevated anion gap, and severe neurologic manifestations, such as seizures, should be considered strong diagnostic indicators of one of these diseases. The presence of ketonuria, occasionally massive, provides an important clue toward the recognition of disorders, especially in the neonatal period. Hyperammonemia, hypoglycemia, and lactic acidemia are frequent findings, especially during acute episodes of metabolic decompensations.

Reference Values

An interpretive report will be provided.

Interpretation

When no significant abnormalities are detected, the organic acid analysis is reported and interpreted in qualitative terms only. 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, and recommendations for additional biochemical testing, and in vitro confirmatory studies (enzyme assay, molecular analysis).

Cautions

The diagnostic specificity of organic acid analysis under acute and asymptomatic conditions may vary considerably.

Informative profiles may not always be detected in disorders where the excretion of diagnostic metabolites reflects the residual activity of the defective enzyme, the dietary load of precursors, and the anabolic/catabolic status of a patient.

In some cases, methods of higher specificity and sensitivity, such as acylcarnitine and acylglycine analysis, can effectively overcome the limitations of standard organic acid analysis for the investigation of patients who are not acutely ill.

Urine samples with creatinine values below 10 mg/dL may be too dilute for accurate interpretation.

Toluene should not be used as a preservative as it will interfere with the test.

Clinical Reference

1. Lehotay DC, Clarke JT. Organic acidurias and related abnormalities. Crit Rev Clin Lab Sci. 1995;32(4):377-429
2. Ferreira CR, van Karnebeek CDM. Inborn errors of metabolism. In: de Vries LS, Glass HC, eds. Neonatal Neurology. Elsevier; 2019:449-481. Handbook of Clinical Neurology. Vol 162
3. Chapman KA. Practical management of organic acidemias. Trans Sc Rare Dis. 2019;1-12. doi:10.3233/TRD-190039

Performance

Method Description

Urine collected on filter paper is reconstituted in water and a volume corresponding to 0.25 mg of creatinine is acidified and then extracted with ethyl acetate. After separation and evaporation of the solvent phase, the dry residue is sialylated and analyzed by capillary gas chromatography mass spectrometry. When indicated, oximation of 2-keto acids is performed by reaction with hydroxylamine hydrate.(Sweetman L. Organic acid analysis. In: Hommes FA, ed.

Techniques in Diagnostic Human Biochemical Genetics. Wiley-Liss; 1991:143-176; Hoffman GF, Feyh P. Organic acid analysis. In: Blau N, Duran M, Blaskovics ME, Gibson KM, eds. Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases. Springer; 2003:27-44; Cowan T, Pasquali M. Laboratory investigations of inborn errors of metabolism. In: Sarafoglou K, Hoffman GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. 2nd ed. McGraw-Hill; 2017:1143)

PDF Report
No

Day(s) Performed
Monday through Saturday

Report Available
3 to 5 days

Specimen Retention Time
Filter paper urine spot: 28 days; Reconstituted urine: 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
83919

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
OAUS	Organic Acids Scrn, US	49287-6

Result ID	Test Result Name	Result LOINC® Value
610707	Organic Acids Scrn, US	49287-6