SCHOOL OF MEDICINE
INDIANA UNIVERSITY

Department of Medical and Molecular Genetics
Division of Diagnostic Genomics

Laboratory Test Directory

Qualitative Urine Organic Acid Analysis

CPT Code(s): 83919, 82570

Service Code (IU Health): 53065512, 53065488

Ordering Recommendation: This test is intended to be used to screen for inborn errors of metabolism associated with the urinary excretion of high levels of organic acids. See below section "Analytes Tested" for a list of clinically relevant urine metabolites that are screened by this assay.

Synonyms: UOA

Methodology: Gas chromatography mass spectrometry (GC-MS).

Performed: multiple times a week (Monday-Friday)

Turn Around Time: 2-10 days

Specimen Requirements

Patient Preparation: None required for urine collection.

Collect: Can be collected at random, in a tube free of preservatives or other additives.

Specimen Volume: 10 mL (4 mL minimum)

Storage: Must be stored at -20°C.

Shipment: Ship frozen samples on 3-5 lbs of dry ice in an insulated container using an overnight courier.

Unacceptable Conditions:

- Specimen requirements listed above not met.
- Sample fully thawed during shipment.
- Sample has evidence of lipemia or contamination with lotions, powders or other foreign substances.

Stability: 30 days freezer (-20°C)

Reference Interval: Qualitative interpretation only

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Interpretive Data

Interpretation:

An interpretation of results is provided with each test. In this interpretation, abnormal organic acid elevations are reported qualitatively.

Limitations:

The ability of this test to detect diagnostic perturbations is affected by a number of factors including the physiologic status of the patient at time of sampling, the genetic basis of the patient's disease, diet, medications, and urine creatinine concentration. For some disorders, urine organic acid analysis may appear normal if the patient is asymptomatic during sampling.

Succinylacetone is NOT detected by this analysis. In addition, clinically relevant orotic acid elevations may be missed by this assay. Targeted quantitative assays should be ordered for these compounds.

Results should be viewed in the context of clinical presentation and concurrent laboratory studies. In the case of a new diagnosis, results should be confirmed with additional biochemical or molecular genetic testing.

This test was developed and its performance characteristics determined by Indiana University Biochemical Genetics Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA '88) as qualified to perform high complexity clinical laboratory testing. CLIA #15D0647198 ▪ CAP #1678930

References:

Rinaldo, P., Ch 3.1 Organic acids, Laboratory Guide to the Methods in Biochemical Genetics, Springer 2009, pg 137-169.

Jones, P.M., Bennett, M.J., Urine organic acid analysis for inherited metabolic disease using gas chromatography-mass spectrometry, Methods Mol Biol, 2010, pg 423-31.

Analytes Tested*

*Note: Analytes shown here are only a representative subset of organic acids that are detectable by this assay. The below listed analytes are NOT routinely detected in all specimens. For questions about the detection of additional organic acids not listed here, contact the lab.

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Organic acids	
2-hydroxy-3-methylbutyric acid	Fumaric acid
2-hydroxyglutaratic acid	Glutaric Acid
2-methylcitric acid	Glycolic Acid
2-oxoglutaratic acid	Hexanoylglycine
3-hydroxy-2-methylbutanoic acid	Hippuric acid
3-hydroxy-3-methylglutaric acid	Homogentisic acid
3-hydroxybutyric acid	Homovanillic acid
3-hydroxyglutaric acid	hydantoin-5-propionic acid
3-hydroxyisovaleric acid	Indole-3-acetic acid
3-hydroxyphenylacetic acid	Isovalerylglycine
3-hydroxypropionic	Lactic acid
3-ketovalproic acid	Malonic acid
3-methylcrotonylglycine	Methylmalonic acid
3-methylglutaconic	Methylsuccinic acid
3-methylglutaric acid	mevalonyllactone
4-hydroxybutyric acid	N-acetyl glycine
4-hydroxyphenylacetic acid	N-acetyl L-alanine
4-Hydroxyphenyllactic acid	N-acetyl L-tyrosine
5-hydroxyindoleacetic acid	N-acetylaspartic acid
5-oxoproline	Propionylglycine
Acetoacetic acid	Suberic Acid
Aconitic acid	suberylglycine
Adipic acid	Succinic Acid
alpha- Hydroxyisobutyric acid	Tiglylglycine
Azelaic acid	Valproic acid
Citric acid	Vanillyllactic acid
Ethylmalonic acid	Vanillylmandelic acid