Overview

Useful For
Diagnosis of inborn errors of metabolism

Highlights
The urine organic acids (OAU) 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 is variable due to factors such as specimen collection when patient is asymptomatic versus acutely ill, taking dietary supplements, or anabolic versus catabolic.

Testing Algorithm
The following algorithms are available in Special Instructions:

- Newborn Screening Follow-up for Elevations of C8, C6, and C10 Acylcarnitines (also applies to any plasma or serum C8, C6, and C10 acylcarnitine elevations)
- Newborn Screening Follow-up for Isolated C4 Acylcarnitine Elevations (also applies to any plasma or serum C4 acylcarnitine elevation)
- Newborn Screening Follow-up for Isolated C5 Acylcarnitine Elevations (also applies to any plasma or serum C5 acylcarnitine elevation)
- Porphyria (Acute) Testing Algorithm
- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

Special Instructions
- Porphyria (Acute) Testing Algorithm
- Newborn Screening Follow-up for Isolated C4 Acylcarnitine Elevations (also applies to any plasma or serum C4 acylcarnitine elevation)
- Newborn Screening Follow-up for Elevations of C8, C6, and C10 Acylcarnitine Elevations (also applies to any plasma or serum C8, C6, and C10 acylcarnitine elevations)
- Newborn Screening Follow-up for Isolated C5 Acylcarnitine Elevations (also applies to any plasma or serum C5 acylcarnitine elevations)
- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

Method Name
Gas Chromatography-Mass Spectrometry (GC-MS)

NY State Available
Yes

Specimen

Specimen Type
Urine
Necessary Information
1. Patient’s age is required.
2. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information.

Specimen Required
Supplies: Urine Tubes, 10 mL (T068)

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.
2. No preservative.

Forms
If not ordering electronically, complete, print, and send an Inborn Errors of Metabolism Test Request (T798) with the specimen.

Specimen Minimum Volume
4 mL

Reject Due To
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
</tr>
</thead>
<tbody>
<tr>
<td>Urine</td>
<td>Frozen (preferred)</td>
<td>416 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Refrigerated</td>
<td>14 days</td>
<td></td>
</tr>
</tbody>
</table>

Clinical and 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 >1 in 1,000,000 live births. Collectively, their incidence approximates 1 in 3,000 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 also require organic acid analysis. All possible disease entities included, the incidence of conditions where informative organic acid profiles could be detected in urine is likely to approach 1 in 1,000 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 is a reflection of 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 non-acutely ill patients.

Clinical Reference


Performance

Method Description
Test Definition: OAU
Organic Acids Scrn, U

Day(s) and Time(s) Test Performed
Monday through Saturday; 7 a.m.

Analytic Time
3 days (not reported on Sunday)

Maximum Laboratory Time
6 days

Specimen Retention Time
2 months

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 U.S. Food and Drug Administration.

CPT Code Information
83919

LOINC® Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>OAU</td>
<td>Organic Acids Scrn, U</td>
<td>2676-5</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>80619</td>
<td>Organic Acids Scrn, U</td>
<td>2676-5</td>
</tr>
</tbody>
</table>