**Overview**

**Method Name**
Fast Atom Bombardment-Mass Spectrometry (FAB-MS)

**NY State Available**
Yes

**Specimen**

**Specimen Type**
Urine

**Specimen Required**

**Collection Container:** Plastic urine container

**Specimen Volume:** 5-25 mL

**Collection Instructions:**
1. Collect 5-25 mL random urine without preservative.
2. Ship frozen in a plastic container.

**NOTE:** Submit with specimen:
1. Clinical history/Preliminary diagnosis
   - Because URSO can mask detection of bile acid synthetic defects it is preferable for patients to be off Urso or Actigall for 5 days before sample collection.
   - If possible, send Urine & Serum (ZW166 - Bile Acids Serum, referral lab code 9001004). Urine is analyzed for all patients - if Urine shows evidence of a metabolic abnormality, Serum will be tested. Urine and serum must be ordered separately as they are 2 separate tests with separate charges.

**Reject Due To**

- Hemolysis   | NA
- Lipemia     | NA
- Icterus     | NA
- Other       | Collection in or on diaper or cotton balls

**Specimen Minimum Volume**
1 mL

**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>
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<tr>
<td></td>
<td>Ambient</td>
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<td>Refrigerated</td>
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Clinical & Interpretive

Clinical Information
Diagnostic testing in pediatric and adult patients presenting with conditions of cholestatic liver disease, neurological disease, or fat-soluble vitamin malabsorption of unknown etiology. Urine FAB-MS analysis provides a rapid and cost-effective means of diagnosing the most common of the genetic defects in the metabolism of cholesterol to the primary bile acids. Mass spectrometry testing may be used to monitor the biochemical response to primary bile acid therapy and to help in decisions on dose adjustments, where compliance should lead to a reduction in levels of atypical bile acids.

Performance

Method Description
Application of liquid secondary ionization mass spectrometry using fast atom bombardment (FAB-MS) ionization to detect the negative ions associated with the presence of increased concentrations of atypical bile acids in urine resulting from the loss of activity of one of the key enzymes that catalyze the production of normal primary bile acids by the liver. Each enzyme defect yields a distinct and specific mass spectrum that permits the diagnosis of the genetic defect.

PDF Report
Referral

Performing Laboratory Location
Child Hospital Medical Center Division of Clinical Mass Spectrometry

Fees & Codes

Test Classification
This test was developed and its performance characteristics were determined and validated by the Clinical Mass Spectrometry Laboratory at Cincinnati Childrens Hospital Medical Center. It has not been cleared or approved by the U. S. Food and Drug Administration. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA 88) as qualified to perform high-complexity laboratory testing.

CPT Code Information
83789