Overview

Useful For
Investigating possible disorders of mitochondrial metabolism, when used in conjunction with cerebrospinal fluid lactate, collected at the same time, to determine the lactate-to-pyruvate (L:P) ratio

Evaluating patients with neurologic dysfunction and normal blood L:P ratios

Genetics Test Information
The cerebrospinal fluid (CSF) lactate:pyruvate (L:P) ratio is considered a helpful (not diagnostic) tool in the evaluation of patients with possible disorders of mitochondrial metabolism, especially in patients with neurologic dysfunction and normal blood L:P ratios. Pyruvic acid levels alone have little clinical utility.

Testing Algorithm
See Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm in Special Instruction.

Special Instructions

Method Name
Spectrophotometry (SP)

NY State Available
Yes

Specimen

Specimen Type
CSF

Additional Testing Requirements
This test does not calculate the lactate:pyruvate ratio. To obtain this information, both this test and LABF / Lactate, Body Fluid must be ordered. The ratio can be calculated from the results obtained from these tests.

Specimen Required

Collection Instructions: Send specimen from vial 2.

Forms
1. Biochemical Genetics Patient Information (T602) in Special Instructions.

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

Specimen Minimum Volume
Test Definition: PYRC
Pyruvic Acid, CSF

0.3 mL

Reject Due To

| Gross hemolysis | Reject |

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>CSF</td>
<td>Frozen (preferred)</td>
<td>7 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Ambient</td>
<td>7 days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Refrigerated</td>
<td>7 days</td>
<td></td>
</tr>
</tbody>
</table>

Clinical and Interpretive

Clinical Information

Pyruvic acid, an intermediate metabolite, plays an important role in linking carbohydrate and amino acid metabolism to the tricarboxylic acid cycle, the fatty acid beta-oxidation pathway, and the mitochondrial respiratory chain complex. Though pyruvate is not diagnostic in itself, analysis with lactate has diagnostic value as many inborn errors of metabolism present with laboratory findings that include lactic acidosis and/or a high lactate:pyruvate (L:P) ratio.

The L:P ratio is elevated in several, but not all, mitochondrial respiratory chain disorders. Mitochondrial disorders vary widely in presentation and age of onset. Many mitochondrial disorders have neurologic and myopathic features and may involve multiple organ systems. Determination of lactate, pyruvate, and the L:P ratio in cerebrospinal fluid is helpful in directing attention toward a possible mitochondrial disorder in cases with predominantly neurologic dysfunction and normal blood lactate levels.

A low L:P ratio is observed in inherited disorders of pyruvate metabolism including pyruvate dehydrogenase complex (PDHC) deficiency. Clinical presentation of PDHC deficiency can range from fatal congenital lactic acidosis to relatively mild ataxia or neuropathy. The most common features in infants and children with PDHC deficiency are delayed development and hypotonia. Seizures and ataxia are also frequent features. Other manifestations can include congenital brain malformations, degenerative changes including Leigh disease, and facial dysmorphism.

Reference Values

0.06-0.19 mmol/L

Interpretation

An elevated lactate-to-pyruvate (L:P) ratio may indicate inherited disorders of the respiratory chain complex, tricarboxylic acid cycle disorders and pyruvate carboxylase deficiency. Respiratory chain defects usually result in L:P ratios above 20.

A low L:P ratio (disproportionately elevated pyruvic acid) may indicate an inherited disorder of pyruvate metabolism. Defects of the pyruvate dehydrogenase complex result in L:P ratios below 10.

The L:P ratio is characteristically normal in other patients. An artificially high ratio can be found in acutely ill patients.

Cautions
Correct specimen collection and handling is crucial to achieve reliable results.

Pyruvic acid levels alone have little clinical utility. Abnormal concentrations of pyruvic acid and lactate-to-pyruvate (L:P) ratios are not diagnostic for a particular disorder but must be interpreted in the context of the patient's clinical presentation and other laboratory studies.

For the L:P ratio, both analytes should be determined using the same specimen.

When comparing blood and cerebrospinal fluid (CSF) L:P ratios, blood and CSF specimens should be collected at the same time.

Clinical Reference


Performance

Method Description


PDF Report

No

Day(s) and Time(s) Test Performed

Monday, Thursday; Varies

Analytic Time

6 days

Maximum Laboratory Time

8 days

Specimen Retention Time
Test Definition: PYRC
Pyruvic Acid, CSF

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
84210

LOINC® Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>PYRC</td>
<td>Pyruvic Acid, CSF</td>
<td>14122-6</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>83356</td>
<td>Pyruvic Acid, CSF</td>
<td>14122-6</td>
</tr>
</tbody>
</table>