Test Definition: PYRC

Pyruvic Acid, CSF

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

- Biochemical Genetics Patient Information
- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

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

Container/Tube: Sterile vial

Specimen Volume: 0.6 mL

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

0.3 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
CSF	Frozen (preferred)	7 days	
	Ambient	7 days	
	Refrigerated	7 days	

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 artifactually 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

1. Munnich A, Rotig A, Cormier-Daire V, Rustin P: Clinical Presentation of Respiratory Chain Deficiency. In The Online Metabolic and Molecular Bases of Inherited Disease. Edited by D Valle, S Antonarakis, A Ballabio, et al. McGraw-Hill. Accessed 1/13/2020. Available at http://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225086827

2. Robinson BH..Lactic Acidemia: Disorders of Pyruvate Carboxylase and Pyruvate Dehydrogenase. In *The Online Metabolic and Molecular Bases of Inherited Disease. Edited by D* Valle, S Antonarakis, A Beaudet, et al. McGraw-Hill. Accessed 1/13/2020. Available at http://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225087140

3. Shoffner JM: Oxidative Phosphorylation Diseases. In The Online Metabolic and Molecular Bases of Inherited Disease. Edited by D Valle, S Antonarakis, A Ballabio, et al. McGraw-Hill. Accessed 1/13/2020. Available at http://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225088339

Performance

Method Description

Pyruvate, in the presence of excess NADH, H+, and lactic dehydrogenase, is reduced to lactate. The reaction is stoichiometric; the decrease in absorbance at 340 nm is directly proportional to the concentration of pyruvate.(Huckabee WE: Relationships of pyruvate and lactate during anaerobic metabolism. I. Effects of infusion of pyruvate or glucose and of hyperventilation. J Clin Invest 1958;37:244-254; Benoist J, Alberti C, Leclercq S, et al: Cerebrospinal fluid lactate and pyruvate concentrations and their ratio in children: age-related reference intervals. Clin Chem 2003;49[3]:487-494; Cowan T, Pasquali M: Laboratory Investigations of Inborn Errors of Metabolism. In Pediatric Endocrinology and Inborn Errors of Metabolism. Second Edition. Edited by K Sarafoglou, GF Hoffman, KS Roth. 2017, pp 1139-1158)

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

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

Test ID	Test Order Name	Order LOINC Value
PYRC	Pyruvic Acid, CSF	14122-6

Result ID	Test Result Name	Result LOINC Value
83356	Pyruvic Acid, CSF	14122-6