MAYO CLINIC LABORATORIES

Test Definition: PYR

Pyruvic Acid, Blood

Overview

Useful For

Screening for possible disorders of mitochondrial metabolism, when used in conjunction with blood lactate collected at the same time, to determine the lactate-to-pyruvate ratio

Genetics Test Information

The 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 either elevated or normal blood lactate levels. Pyruvic acid levels alone have little clinical utility.

Testing Algorithm

For information see Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

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

Whole blood

Additional Testing Requirements

This test does not calculate the lactate:pyruvate ratio. If you would like the lactate:pyruvate ratio, order LAPYP / Lactate Pyruvate Panel, Plasma.

Specimen Required

Call 800-533-1710 or 507-266-5700 to order special collection tube.

Patient Preparation:
Fasting: 4 hours, required
Supplies: Perchloric Acid-Pyruvate Tube (T012)
Container/Tube: Special collection tube containing 2.5 mL of 6% perchloric acid
Specimen Volume: Exactly 1 mL

Collection Instructions:

1. Special collection tube must be prechilled prior to collection.
2. Draw enough blood directly into syringe to add exactly 1 mL of blood to the prechilled special collection tube.
3. Taking care to not spill any of the preservative, cautiously remove the cap from the tube.
4. Immediately transfer blood, once drawn, to the prechilled, special collection tube, recap, and shake vigorously to mix.

Additional Information:

1. **Check expiration date before using.** Supplied collection tube expires 14 months after preparation.
2. If perchloric acid (preservative) spills, obtain a new, prechilled tube.

Forms

1. Biochemical Genetics Patient Information (T602)
2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

See Specimen Required

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated	15 days	PYRUVATE

Clinical & 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 isolated elevated pyruvate is not diagnostic of any inborn error of metabolism, analysis with lactate may suggest an inborn error of metabolism as some present with lactic acidosis or a high lactate-to-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 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, though further confirmatory testing will be required to establish a diagnosis.

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

0.08-0.16 mmol/L

NIH Unit
0.7-1.4 mg/dL

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 if the patient is acutely ill.

Cerebrospinal fluid (CSF) L:P ratio may assist in evaluation of patients with neurologic dysfunction and normal blood L:P ratios. Blood and CSF specimens should be collected at the same time.

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. The determination of pyruvic acid is of diagnostic value when lactic acid is measured and the L:P ratio is established in 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: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed January 14, 2025.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: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019. Accessed January 14, 2025. Available at http://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225087140
3. Shoffner JM. Oxidative phosphorylation diseases. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA, eds. Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; 2019 Accessed January 14, 2025.. Available at

http://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225088339
4. Parikh S, Goldstein A, Koenig MK, et al. Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genet Med. 2015;17(9):689-701. doi:10.1038/gim.2014.177

Performance

Method Description
Pyruvate, in the presence of excess nicotinamide adenine dinucleotide, hydrogen ions, 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.(Fleischer WR, Forman DT, Huckabee WE, Antonis A, Young K. Enzymatic methods for lactic and pyruvic acids. In: MacDonald RP, ed. Standard Methods of Clinical Chemistry. Vol 6. 1970:245-259; 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[2]:244-254; Cowan T, Pasquali M. Laboratory investigations of inborn errors of metabolism. In: Sarafoglou K, Hoffman GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. 2nd ed. McGraw-Hill; 2017:1139-1158)

PDF Report
No

Day(s) Performed
Monday, Thursday

Report Available
2 to 5 days

Specimen Retention Time
3 weeks

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & 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 account representative. 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. It has not been cleared or approved by the US Food and Drug Administration.

Test Definition: PYR

Pyruvic Acid, Blood

CPT Code Information

84210

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PYR	Pyruvic Acid, B	14121-8

Result ID	Test Result Name	Result LOINC® Value
8657	Pyruvic Acid, B	14121-8
7729	Pyruvic Acid, B	2905-8