Overview

Useful For
Evaluation of individuals with Coombs-negative nonspherocytic hemolytic anemia
Evaluation of individuals with exercise intolerance or myopathy
Genetic studies in families with phosphofructokinase deficiency

Method Name
Kinetic Spectrophotometry

NY State Available
Yes

Specimen

Specimen Type
Whole Blood ACD-B

Specimen Required
Collection Container/Tube:

Preferred: Yellow top (ACD solution B)
Acceptable: Lavender top (EDTA)

Specimen Volume: 6 mL

Collection Instructions: Send specimen in original tube. Do not transfer blood to other containers.

Forms
If not ordering electronically, complete, print, and send a Benign Hematology Test Request (T755) with the specimen.

Specimen Minimum Volume
1 mL

Reject Due To

| Gross hemolysis | Reject |

Specimen Stability Information

<table>
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<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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<tbody>
<tr>
<td>Whole Blood ACD-B</td>
<td>Refrigerated</td>
<td>11 days</td>
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Clinical and Interpretive

Clinical Information
Phosphofructokinase (PFK) is the third enzyme in glycolysis. It converts fructose-6-phosphate to fructose 1,6-diphosphate. PFK deficiency, also called glycogen storage disease, type VII or Tarui disease (OMIM 232800), is a rare hereditary autosomal recessive disorder that is typically noticed in childhood. Different clinical subtypes (classical, late-onset, infantile and hemolytic) have been described. Manifestations can vary including exercise intolerance, exertional myopathy, nausea, stiffness, and myoglobinuria. Although not classically described, a second-wind effect is noticed by some patients (1). A subset of individuals have compensated (high normal hemoglobin values) or mild hemolytic anemia, episodic jaundice, hyperuricemia, or gout-like symptoms. No distinctive morphologic abnormalities are seen on the peripheral blood smear. Red blood cell PFK activity is typically partially decreased (30-50% mean normal) and muscle biopsy PFK activity is markedly decreased.

Reference Values
> or =12 months of age: 5.8-10.9 U/g Hb

Reference values have not been established for patients who are <12 months of age.

Interpretation
Clinically significant disorders due to phosphofructokinase (PFK) deficiency are associated with red blood cell activity levels less than 50% of mean normal. Unaffected heterozygotes have been reported with levels of 63% of normal. Therefore genetic correlation will often be important in ambiguous cases.

Cautions
Recent transfusion may mask the patient's intrinsic erythrocyte (RBC) enzyme activity and cause unreliable results.

Some enzyme deficiency disorders can be masked by reticulocytosis and comparison of activities of other RBC enzyme activities in this panel can be useful.

Some enzyme deficiency disorders can be slightly decreased in normal neonates or conversely masked in the neonatal period. Repeat testing after 1 year of age can be useful if features of myopathy are present.

Clinical Reference
Test Definition: PFK1
Phosphofructokinase, B

10.1002/humu.1380060102


Performance

Method Description
Phosphofructokinase (PFK) catalyzes the phosphorylation of fructose-6-phosphate (F6P) by adenosine triphosphate (ATP) to fructose-1,6-diphosphate (F1,6-diP). F1,6-diP is then converted to dihydroxyacetone phosphate (DHAP) through subsequent aldolase and triosephosphate isomerase (TPI) catalyzed reactions. The rate of formation of DHAP is measured by linking its reduction to alpha-glycerophosphate by alpha-glycerophosphate dehydrogenase which results in the oxidation of 1,4-dihydrionicotinamide adenine dinucleotide NADH to NAD(+). The decrease in absorbance at 340 nm is measured spectrophotometrically as the NADH is oxidized on an automated chemistry analyzer.(Beutler E: Red cell metabolism. A Manual of Biochemical Methods. 3rd ed. Grune and Stratton; 1984: 68-71; van Solinge WW, van Wijk: Enzymes of the red blood cell. In: Rifai N, Horvath AR, Wittwer CT, eds. Tietz Textbook of Clinical Chemistry and Molecular Diagnostics. 6th ed. Elsevier; 2018:chap 30)

PDF Report
No

Day(s) Performed
Performed weekly, varies

Report Available
5 days

Specimen Retention Time
7 days

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 US Food and Drug Administration.

CPT Code Information
82657

LOINC® Information
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<td>Phosphofructokinase, B</td>
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