MAYO CLINIC LABORATORIES

Test Definition: PFKC

Phosphofructokinase, B

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

Only available as part of a profile. For more information see:
-HAEV1 / Hemolytic Anemia Evaluation, Blood
-EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood
Kinetic Spectrophotometry

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Specimen Required

Only available as part of a profile. For more information see:
-HAEV1 / Hemolytic Anemia Evaluation, Blood
-EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

Reject Due To

Gross hemolysis Reject

Specimen Minimum Volume

1 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD-B	Refrigerated (preferred)	11 days	

Clinical & 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 and include 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 has 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

Only available as part of a profile. For more information see:
-HAEV1 / Hemolytic Anemia Evaluation, Blood
-EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood
> or =12 months of age: 5.8-10.9 U/g Hb
Reference values have not been established for patients who are less than12 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 and repeat testing after 1 year of age can be useful if features of myopathy are present.

Clinical Reference

1. Sherman JB, Raben N, Nicastri C, et al: Common mutations in the phosphofructokinase-M gene in Ashkenazi Jewish patients with glycogenesis VII--and their population frequency. Am J Hum Genet. 1994 Aug;55(2):305-313
2. Tarui S, Okuno G, Ikura Y, Tanaka T, Suda M, Nishikawa M: Phosphofructokinase deficiency in a skeletal muscle. A new type of glycogenosis. Biochem Biophys Res Commun. 1965 May 3;19:517-523
3. Musumeci O, Bruno C, Mongini T, et al: Clinical features and new molecular findings in muscle phosphofructokinase deficiency (GSD type VII). Neuromuscul Disord. 2012 Apr;22(4):325-330
4. Nakajima H, Raben N, Hamaguchi T, Yamasaki T: Phosphofructokinase deficiency; past, present and future. Curr Mol Med. 2002 Mar;2(2):197-212
5. Auranen M, Palmio J, Ylikallio E, et al: PFKM gene defect and glycogen storage disease GSDVII with misleading enzyme histochemistry. Neurol Genet. 2015 Jun 4;1(1)
6. Raben N, Sherman JB. Mutations in muscle phosphofructokinase gene. Hum Mutat. 1995;6(1):1-6
7. Koralkova P, van Solinge WW, van Wijk R: Rare hereditary red blood cell enzymopathies associated with hemolytic anemia-pathophysiology, clinical aspects and laboratory diagnosis. Int J Lab Hematol. 2014;36:388-397

Performance

Method Description

Phosphofructokinase (PFK) catalyzes the phosphorylation of fructose 6-phosphate (F6P) by adenosine triphosphate (ATP) to fructose 1,6-diphosphate (FDP). FDP 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-dihydronicotinamide 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

Specimen Retention Time

7 days

Performing Laboratory Location

Rochester

Fees & Codes

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