MAYO CLINIC LABORATORIES

Test Definition: PGKC

Phosphoglycerate Kinase, B

Overview

Useful For

Evaluation of individuals with Coombs-negative nonspherocytic hemolytic anemia, especially if X-linked inheritance pattern.
Evaluation of individuals with myopathic or neurologic symptoms

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)	20 days	

Clinical & Interpretive

Clinical Information

Phosphoglycerate kinase (PGK) is an enzyme that converts 1,3-diphosphoglycerate (1,3-DPG) to 3-phosphoglyceric acid (3-PGA) in one of the adenosine triphosphate (ATP) generating steps in glycolysis. PGK deficiency (OMIM 300653) is an

X-linked disorder with a variable clinical phenotype. Manifestations include hemolytic anemia, myopathy/rhabdomyolysis, or neurologic impairment. Patients can have 1 or 2 systems affected but rarely have all 3. Clinical severity may not correlate with enzyme activity and female heterozygotes may possibly be mildly affected.

Reference Values

Only available as part of a profile. For more information see:
-HAEV1 / Hemolytic Anemia Evaluation
-EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation
> or =12 months: 142-232 U/g Hb
Reference values have not been established for patients who are less than12 months of age.

Interpretation

In phosphoglycerate kinase (PGK) deficiency, red blood cell activity levels have been reported ranging from 1% to 49% of mean normal; however, affected patients more typically have values less than 20% of normal mean.(1)

Cautions

Recent transfusion may mask the patient's intrinsic enzyme activity and cause unreliable results.

Clinical Reference

1. Chiarelli LR, Morera SM, Bianchi P, et al: Molecular insights on pathogenic effects of mutations causing phosphoglycerate kinase deficiency. PLoS One. 2012;7(2)
2. Valentine WN, Hsieh HS, Paglia DE, et al: Hereditary hemolytic anemia associated with phosphoglycerate kinase deficiency in erythrocytes and leukocytes: a probable X-chromosome-linked syndrome. New Eng J Med. 1969;280:528-534
3. Beutler E: PGK deficiency. Br J Haematol. 2007 Jan;136(1):3-11
4. 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

Phosphoglycerate kinase (PGK) catalyzes the phosphorylation of adenosine diphosphate (ADP) to adenosine triphosphate (ATP) by conversion of 1,3-diphosphoglycerate (1,3-DPG) to 3,phosphoglyceric acid (3-PGA). In this assay, the reaction is driven in the reverse direction. The formation of 1,3-DPG is then measured through the glyceraldehyde phosphate dehydrogenase (GAPD) reaction as 1,3-DPG is converted to glyceraldehyde-3-phosphate (GAP) resulting in the oxidation of 1,4-dihydronicotinamide adenine dinucleotide (NADH) to NAD(+). The decrease in absorbance, which occurs as NADH is oxidized, is measured spectrophotometrically at 340 nm on an automated chemistry analyzer.(Beutler E: Red Cell Metabolism. A Manual of Biochemical Methods. 3rd ed. Grune and Stratton; 1984:53-55; 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