MAYO CLINIC LABORATORIES

Test Definition: PGK1

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

Kinetic Spectrophotometry (KS)

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Specimen Required

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.

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

> 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, RBC activity levels have been reported ranging from 1% to 49% of mean normal; however, affected patients more typically have values below 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): e32065

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. N Engl 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; 1984:40-42; 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

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
PGK1	Phosphoglycerate Kinase, B	44053-7

Result ID	Reporting Name	LOINC®

PGKCL	Phosphoglycerate Kinase, B	44053-7