

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 (KS)

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
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Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD-B	Refrigerated	20 days	

Clinical & Interpretive

### Clinical Information

Phosphoglycerate kinase (PGK) is an enzyme that converts 1,3-diphosphoglycerate to 3-phosphoglyceric acid) 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 younger than 12 months.

### Interpretation

In phosphoglycerate kinase 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):e32065. doi:10.1371/journal.pone.0032065
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(10):528-534
3. Beutler E: PGK deficiency. Br J Haematol. 2007;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. 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 reaction as 1,3-DPG is converted to glyceraldehyde-3-phosphate 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

Day(s) Performed

Weekly

Report Available

5 days

Specimen Retention Time

7 days

Performing Laboratory Location

Rochester

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.

CPT Code Information

82657

LOINC® Information

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

Result ID	Test Result Name	Result LOINC® Value
PGKCL	Phosphoglycerate Kinase, B	44053-7