



# Test Definition: PGKC

Phosphoglycerate Kinase Enzyme Activity,  
Blood

## Overview

### Useful For

Evaluation of individuals with Coombs-negative nonspherocytic hemolytic anemia, especially if X-linked inheritance pattern, as a part of a profile

Evaluation of individuals with myopathic or neurologic symptoms

### Method Name

Only orderable 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 orderable 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
Fully clotted	Reject

### Specimen Stability Information

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

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**Clinical & Interpretive****Clinical Information**

Phosphoglycerate kinase (PGK) is an enzyme that converts 1,3-diphosphoglycerate into 3-phosphoglyceric acid), during glycolysis, representing one of the adenosine triphosphate-generating steps. 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 heterozygous individuals may possibly be mildly affected.

**Reference Values**

Only orderable 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 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
5. Echaniz-Laguna A, Nadjar Y, Behin A, et al. Phosphoglycerate kinase deficiency: A nationwide multicenter retrospective study. *J Inherit Metab Dis*. 2019;42(5):803-808

**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-dihyronicotinamide 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; Rab MAE, van Wijk R. Enzymes of the red blood cell. In: Rifai N, Chiu RWK, Young I, Burnham CAD, Wittwer CT, eds. Tietz Textbook of Laboratory Medicine. 7th ed. Elsevier; 2023:chap 78)

**PDF Report**

No

**Day(s) Performed**

Tuesday, Thursday

**Report Available**

5 days

**Specimen Retention Time**

7 days

**Performing Laboratory Location**

Mayo Clinic Laboratories - Rochester Main Campus

**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
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## Test Definition: PGKC

Phosphoglycerate Kinase Enzyme Activity,  
Blood

PGKCL	Phosphoglycerate Kinase, B	44053-7
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