# Test Definition: PGK1

Phosphoglycerate Kinase Enzyme Activity, Blood

## 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) or yellow top (ACD solution A)

**Specimen Volume:** 6 mL

**Collection Instructions:**

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot**.

### Forms

If not ordering electronically, complete, print, and send a Benign Hematology Test Request (T755) with the specimen.

### Specimen Minimum Volume

1 mL

### 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 | |

## Clinical & Interpretive

### Clinical Information

Phosphoglycerate kinase (PGK) is an enzyme that converts 1,3-diphosphoglycerate to 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 be mildly affected.

### Reference Values

> 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 below 20% of normal mean.(1)

### Cautions

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

Some enzyme deficiency disorders can be masked by reticulocytosis, and comparison of activities of other red blood cell enzyme activities in this panel may be useful.

### 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(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(3):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 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 reduced nicotinamide 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; 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

1 to 6 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 |
|---|---|---|

| | | |
|---|---|---|
| PGK1 | Phosphoglycerate Kinase, B | 44053-7 |

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