

# **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.

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	Reject
hemolysis	

### Specimen Stability Information

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

## **Clinical & Interpretive**



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### **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



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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**

Mayo Clinic Laboratories - Rochester Main Campus

# Fees & Codes

### **Fees**

- Authorized users can sign in to <u>Test Prices</u> for detailed fee information.
- Clients without access to Test Prices can contact <u>Customer Service</u> 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact <u>Customer Service</u>.

# **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