Test Definition: PKC

Pyruvate Kinase Enzyme Activity, Blood
(Non-Orderable)

Overview

Useful For

Only orderable as part of a profile. For more information see:
HAEV1 / Hemolytic Anemia Evaluation, Blood
EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

Evaluation of nonspherocytic hemolytic anemia as a part of a profile

Evaluation of neonatal anemia or jaundice

Evaluation of unexplained noninfectious hepatic failure

Evaluation of unexplained iron overload

Evaluation of unusually severe hemoglobin S trait

Evaluation of unusually severe glucose-6-phosphate dehydrogenase deficiency

Investigating families with pyruvate kinase deficiency to determine inheritance pattern and for genetic counseling

Method Name

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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

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

Deficiencies of most of the enzymes of the Embden-Meyerhof (glycolytic) pathway, including pyruvate kinase (PK), have been reported. PK deficiency (OMIM 266200) is the erythrocyte enzyme deficiency most frequently found to be a cause of chronic nonspherocytic hemolytic anemia. It is an autosomal recessive disorder and parents of affected patients are typically carriers. Some PK carrier states can exacerbate other red blood cell disorders (ie, coincident glucose 6-phosphate dehydrogenase deficiency or hemoglobin S trait).

Clinically significant PK deficiency manifests in widely variable severity ranging from incidental compensated mild normocytic anemia to severe anemia. Neonatal jaundice is very common, and a significant subset of neonates have perinatal complications. Other symptoms include early gallstones and splenomegaly. Iron overload, even in the absence of frequent transfusions, is very common. Rare severe PK deficiency is associated with hydrops fetalis/fetal demise or unexplained noninfectious hepatic failure. Acquired PK deficiency can arise secondary to myeloid neoplasms.

Reference Values

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HAEV1 / Hemolytic Anemia Evaluation, Blood
EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

> or =12 months of age: 5.5-12.4 U/g Hb
Reference values have not been established for patients younger than 12 months.

Interpretation

Pyruvate kinase (PK) deficiency is the most easily masked of the red blood cell (RBC) enzyme disorders and can be difficult to classify without complete information, which may require comparison to other RBC enzyme activity levels and/or correlation with results of *PKLR* gene molecular testing (PKLRZ / *PKLR* Full Gene Analysis, Varies). Most hemolytic anemias due to PK deficiency are associated with activity levels less than 40% of mean normal. However, some patients with clinically significant hemolysis can have normal or only mildly decreased PK enzyme activity, which, paradoxically, may occur in individuals with the most severe symptoms. Isolated carriers (heterozygotes) may show mildly decreased activity and are typically hematologically normal, although the carrier state may exacerbate other RBC disorders such as glucose 6-phosphate dehydrogenase deficiency, RBC membrane disorders, or hemoglobinopathies. Some alterations in

other genes (ie, *KLF1*) can be associated with decreased PK levels.

Elevated PK concentrations can be found in those patients with younger erythrocyte population. This may be due to the patient being a newborn or young RBCs being produced in response to the anemia (reticulocytosis). Rare PK deficient cases have been associated with minimally increased PK levels; however, comparison to other RBC enzyme activity would be critical in these cases for accurate interpretation.

Cautions

Pyruvate kinase (PK) activity level can vary from markedly decreased to normal levels in affected individuals due to a compensated increase in enzyme by reticulocytes. Comparison of PK activity levels to other red blood cell enzyme activity can be very useful.

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

Because leukocytes also contain PK, if the white blood cell (WBC) count is very high, false-negative results may occur due to inability to adequately remove WBCs from the assay.

Clinical Reference

1. Grace RF, Bianchi P, van Beers EJ, et al. The clinical spectrum of pyruvate kinase deficiency: data from the Pyruvate Kinase Deficiency Natural History Study. Blood. 2018 ;131(20):2183-2192
2. Gallagher PG, Glader B. Diagnosis of pyruvate kinase deficiency. Pediatr Blood Cancer. 2016;63(5):771-772
3. Grace RF, Zanella A, Neufeld EJ, et al. Erythrocyte pyruvate kinase deficiency: 2015 status report. Am J Hematol. 2015;90(9):825-830
4. Zanella A, Fermo E, Bianchi P, Chiarelli LR, Valentini G. Pyruvate kinase deficiency: the genotype-phenotype association. Blood Rev. 2007;21(4):217-231

Performance

Method Description

Pyruvate kinase catalyzes the phosphorylation of adenine diphosphate to adenine triphosphate by converting phosphoenolpyruvate to pyruvate. The amount of pyruvate formed is quantitated by adding lactate dehydrogenase and reduced nicotinamide adenine dinucleotide (NADH) and measuring the rate of decrease in absorbance spectrophotometrically at 340 nm as the NADH is oxidized to NAD(+) on an automated chemistry analyzer.(Beutler E: Red Cell Metabolism. In: A Manual of Biochemical Methods. 3rd ed. Grune and Stratton; 1984:68-71; 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

Monday through Friday

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Pyruvate Kinase Enzyme Activity, Blood
(Non-Orderable)

Report Available
5 days

Specimen Retention Time
28 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
84220

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PKC	PK Enzyme Activity, B	32552-2

Result ID	Test Result Name	Result LOINC® Value
PKCL	PK Enzyme Activity, B	32552-2