Overview

Useful For
Evaluation of nonspherocytic hemolytic anemia
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
Kinetic Spectrophotometry

NY State Available
Yes

Specimen

Specimen Type
Whole Blood ACD-B

Specimen Required
Collection Container/Tube:

Preferred: Yellow top (ACD solution B)
Acceptable: Lavender top (EDTA)

Specimen Volume: 6 mL

Collection Instructions: Send specimen in original tube. Do not transfer blood to other containers.

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

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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 (CNSHA). It is an autosomal recessive disorder and parents of affected patients are typically carriers. Some PK carrier states can exacerbate other RBC 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 has 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
> or =12 months of age: 5.5-12.4 U/g Hb

Reference values have not been established for patients who are less than 12 months of age.

Interpretation
Pyruvate kinase (PK) deficiency is the most easily masked of the RBC enzyme disorders and can be difficult to classify without complete information, which may require comparison to other RBC enzyme activity levels or correlation with results of PKLR gene molecular testing (PKLRG / Pyruvate Kinase Liver and Red Blood Cell [PKLR] Full Gene Sequencing and Large Deletion Detection, 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 red cells are 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 RBC 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 WBC count is very high, false-negative results may occur due to inability to adequately remove WBCs from the assay.

**Clinical Reference**


**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 Saturday

**Report Available**

1 to 4 days

**Specimen Retention Time**

7 days

**Performing Laboratory Location**

Rochester

**Fees and Codes**

**Fees**

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
Test Definition: PK1
PK Enzyme Activity, B

- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. 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. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
84220

LOINC® Information

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