Overview

Useful For
Evaluation of nonspherocytic hemolytic anemia
Evaluation of neonatal anemia
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
KineticSpectrophotometry (KS)

NY State Available
Yes

Specimen

Specimen Type
Whole Blood ACD-B

Specimen Required
Container/Tube:
Preferred: Yellow top (ACD solution B)
Acceptable: EDTA

Specimen Volume: 6 mL

Collection Instructions: Do not transfer blood to other containers.

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

Specimen Minimum Volume
1 mL

Reject Due To

| Gross hemolysis | Reject |

Specimen Stability Information
**Clinical and Interpretive**

**Clinical Information**

Deficiencies of most of the enzymes of the Embden-Meyerhof (glycolytic) pathway, including pyruvate kinase (PK), have been reported. PK deficiency, although relatively rare, 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. It is possible the mutation arose as partial protection against malaria. PK deficiency is the most easily masked of the RBC enzyme disorders and is therefore difficult to classify without complete information which may require correlation with results of *PKLR* gene molecular testing (*PKLRG* / Pyruvate Kinase Liver and Red Blood Cell (*PKLR*) Full Gene Sequencing and Large Deletion Detection). 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. Carriers (heterozygotes) may show mildly decreased activity and are hematologically normal. Some PK carrier states can exacerbate other RBC disorders (ie coincident glucose-6-phosphate dehydrogenase: G6PD deficiency or hemoglobin S trait).

Clinically significant PK deficiency manifests in widely variable severity ranging from incidental compensated mild normocytic anemia to severe neonatal anemia. Other symptoms include jaundice, early gallstones, splenomegaly and iron overload even in the absence of frequent transfusions. Rare severe forms are associated with hydrops fetalis/fetal demise. Rarely, acquired PK deficiency can arise secondary to myeloid neoplasms.

**Reference Values**

> or = 12 months: 6.7-14.3 U/g Hb

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

**Interpretation**

Most hemolytic anemias due to pyruvate kinase (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. Carriers (heterozygotes) may show mildly decreased activity and are hematologically normal.

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).

**Cautions**

Because leukocytes also contain pyruvate kinase (PK) that is not diminished in hereditary erythrocytic PK deficiency, freeing the blood of white blood cells is always critical to this test. 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
A red cell hemolysate is incubated with adenosine diphosphate and phosphoenolpyruvate. The amount of pyruvate formed is quantitated by adding lactic dehydrogenase and reduced nicotinamide adenine dinucleotide and measuring the rate of decrease in absorbance at 340 nm. (Beutler E: Red Cell Metabolism. In A Manual of Biochemical Methods. Third edition. New York, Grune and Stratton, 1984, pp 68-71)

PDF Report
No

Day(s) and Time(s) Test Performed
Monday through Saturday

Analytic Time
1 day

Maximum Laboratory Time
4 days

Specimen Retention Time
28 days

Performing Laboratory Location
Rochester

Fees and 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 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 U.S. Food and Drug Administration.

CPT Code Information
84220

LOINC® Information
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