MAYO CLINIC LABORATORIES

Test Definition: GPI1

Glucose Phosphate Isomerase, B

Overview

Useful For
The evaluation of individuals with Coombs-negative chronic hemolysis

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)

Specimen Volume:6 mL

Collection Instructions: Send 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.

Reject Due To

Gross Hemolysis	Reject

Specimen Minimum Volume
1 mL

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information
The glucose 6-phosphate (G6P) isomerase enzyme interconverts G6P and fructose-6-phosphate in the second step of glycolysis. Glucose phosphate isomerase (GPI) deficiency (OMIM 613470) is a cause of nonspherocytic hemolytic anemia and has been reported in patients from varied ethnic backgrounds. As investigational methods have improved, the

number of confirmed diagnoses has increased, although the disorder remains rare. Inheritance is autosomal recessive. Clinically significant GPI deficiency manifests in variable severity ranging from mild to severe anemia, with jaundice, gallstones, splenomegaly. Some cases of neonatal death/hydrops fetalis have been reported to be associated with GPI deficiency. A subset of patients shows neurologic impairment and granulocyte dysfunction. Heterozygotes are expected to have a normal phenotype.

Reference Values

> or =12 months: 40.0-58.0 U/g Hb
Reference values have not been established for patients who are <12 months of age.

Interpretation

Most clinically significant hemolytic anemias due to glucose phosphate isomerase (GPI) deficiency are associated with activity levels under 30% of mean normal; however, some clinically affected patients can have higher activity due to reticulocytosis. Heterozygotes usually show 40% to 60% of mean normal activity and are hematologically normal. Increased GPI activity is variably seen when young red blood cells are being produced in response to the anemia (reticulocytosis) or in newborns.

Cautions

Recent transfusion may mask the patient's intrinsic enzyme activity and cause unreliable results.
Reticulocytosis from any cause can mask some glucose phosphate isomerase (GPI) deficiency cases by raising the activity level. Comparison to other RBC enzyme activity levels or correction for reticulocytosis may be useful.

Clinical Reference

1. Manco L, Bento C, Victor BL, et al: Hereditary nonspherocytic hemolytic anemia caused by red cell glucose-6-phosphate isomerase (GPI) deficiency in two Portuguese patients: Clinical features and molecular study. Blood Cells Mol Dis. 2016 Sep;60:18-23
2. Mojzikova R, Koralkova P, Holub D, et al: Two novel mutations (p.(Ser160Pro) and p.(Arg472Cys)) causing glucose-6-phosphate isomerase deficiency are associated with erythroid dysplasia and inappropriately suppressed hepcidin. Blood Cells Mol Dis. 2018 Mar;69:23-29
3. Fairbanks VF, Klee GG: Biochemical aspects of hematology. In: Burtis CA, Ashwood ER, eds. Tietz Textbook of Clinical Chemistry. 3rd ed. WB Saunders Company; 1999:1642-1646
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

Glucose phosphate isomerase (GPI) interconverts glucose 6-phosphate (G6P) and fructose 6-phosphate (F6P). In this assay, the F6P is then further converted to 6-phosphogluconate (6-PG) through the G6P dehydrogenase (G6PD) reaction resulting in the reduction of nicotinamide adenine dinucleotide phosphate (NADP[+]) to NADPH. The reduction of NADP(+) is measured spectrophotometrically by the increase in absorbance 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; 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

Specimen Retention Time
7 days

Performing Laboratory Location
Rochester

Fees & Codes

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
84087