MAYO CLINIC LABORATORIES

Test Definition: GPIC

Glucose Phosphate Isomerase Enzyme Activity, Blood

Overview

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

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 hemolysis	Reject

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

The glucose 6-phosphate isomerase (GPI) enzyme interconverts glucose 6-phosphate and fructose 6-phosphate in the second step of glycolysis. 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 and 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

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

> or =12 months of age: 40.0-58.0 U/g Hb
Reference values have not been established for patients who are younger than12 months of age.

Interpretation

Most clinically significant hemolytic anemias due to glucose phosphate isomerase (GPI) deficiency are associated with activity levels less than 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 red blood cell (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;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;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 (6PG) 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

Day(s) Performed

Weekly

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

84087

LOINC® Information

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Test Definition: GPIC

Glucose Phosphate Isomerase Enzyme Activity, Blood

Test ID	Test Order Name	Order LOINC® Value
GPIC	Glucose Phosphate Isomerase, B	44050-3

Result ID	Test Result Name	Result LOINC® Value
GPICL	Glucose Phosphate Isomerase, B	44050-3