MAYO CLINIC LABORATORIES

Test Definition: EEEVI

Red Blood Cell (RBC) Enzyme Interpretation

Overview

Useful For

Interpretation of results for the red blood cell enzyme evaluation

Identifying defects of red cell enzyme metabolism

Evaluating patients with Coombs-negative hemolytic anemia

Method Name

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

Medical Interpretation

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD-B	Refrigerated	11 days	

Clinical & Interpretive

Clinical Information

Erythrocyte enzyme deficiencies are inherited causes of hemolytic anemia. Some are very common, such as glucose 6-phosphate dehydrogenase (G6PD) deficiency, and others are very rare, found in only a few families around the world. Most are autosomal in inheritance, but some are sex-linked and located on the X chromosome. Most enzyme deficiencies result in chronic nonspherocytic hemolytic anemia of variable severity; however, some, such as G6PD, can be hematologically normal with episodic acute hemolysis due to a trigger event such as medications, toxins, or some foods. The red blood cell (RBC) enzymopathies do not typically show recurrent pathognomonic changes on the peripheral blood smear other than generic features of hemolytic anemia, although some such as pyruvate kinase deficiency can have echinocytes and pyrimidine 5' nucleotidase (P5NT) deficiency is associated with basophilic stippling. RBC enzyme activity levels are best evaluated as a panel as reticulocytosis can mask some deficient states and comparison to the background enzyme activity is useful.

This is a consultative evaluation of red cell enzyme activity as a potential cause of early red cell destruction.

Reference Values

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

Definitive results and an interpretive report will be provided.

Interpretation

A hematopathologist expert in these disorders evaluates the case and an interpretive report is issued.

Cautions

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

A very high white blood cell count can contribute to interference and falsely raise the activity for some enzymes.

Some enzyme deficiency disorders can be masked by reticulocytosis and comparison of activities of other red blood cell enzyme activities in this panel can be useful.

Clinical Reference

1. 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(3):388-397
2. Beutler E. Glucose-6-phosphate dehydrogenase deficiency and other enzyme abnormalities. In: Beutler E, Lichtmann MA, Coller BS, Kipps TJ, eds. Hematology. 5th ed. McGraw-Hill Book Company; 1995:564-581

Performance

Method Description

A hematopathologist who is an expert in these disorders evaluates the case and an interpretive report is issued.

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

2 to 13 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

Not Applicable

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
EEEVI	Erythrocyte Enzyme Interpretation	59466-3

Result ID	Test Result Name	Result LOINC® Value
608087	Erythrocyte Enzyme Interpretation	59466-3
608109	Reviewed By	18771-6