Overview

Useful For
Identifying defects of red cell enzyme metabolism
Evaluating patients with hemolytic anemia

Profile Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>EEEV</td>
<td>Erythrocyte Enzyme Interpretation</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>G6PD</td>
<td>G-6-PD, QN, RBC</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>PK</td>
<td>Pyruvate Kinase, RBC</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>GPI</td>
<td>Glucose Phosphate Isomerase, B</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>HEXK</td>
<td>Hexokinase, B</td>
<td>No</td>
<td>Yes</td>
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</table>

Reflex Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>GLTI</td>
<td>Glutathione, B</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>RBCE</td>
<td>Reflexed RBC Enzymes</td>
<td>No</td>
<td>No</td>
</tr>
</tbody>
</table>

Testing Algorithm
This is a consultative evaluation in which the case will be evaluated at Mayo Clinic Laboratories, the appropriate tests performed at an additional charge, and the results interpreted.

Note: RBCE / Reflexed RBC Enzymes, Blood includes: adenylate kinase, phosphofructokinase, phosphoglycerate kinase, triosephosphate isomerase, and pyrimidine 5'nucleotidase.

See Benign Hematology Evaluation Comparison in Special Instructions.

Special Instructions
- Metabolic Hematology Patient Information
- Benign Hematology Evaluation Comparison

Method Name
EEEV: Consultative Interpretation
G6PD, GPI, PK, GLTI, HEXK, RBCE: Kinetic Spectrophotometry (KS)

NY State Available
Test Definition: EEEVP
RBC Enzyme Evaluation

Yes

Specimen

Specimen Type
Whole Blood ACD-B

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

Specimen Volume: 12 mL

Collection Instructions: Do not transfer blood to other containers.

Forms
1. Metabolic Hematology Patient Information (T810) is available in Special Instructions
2. If not ordering electronically, complete, print, and send a Benign Hematology Test Request Form (T755) with the specimen.

Specimen Minimum Volume
5 mL

Reject Due To

| Gross hemolysis | Reject |

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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<tbody>
<tr>
<td>Whole Blood ACD-B</td>
<td>Refrigerated</td>
<td>8 days</td>
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Clinical and Interpretive

Clinical Information
All enzyme defects, including erythrocyte enzyme errors, are inherited; some are sex-linked and located on the X chromosome. Some family members have no hematologic abnormalities, while others have a hemolytic anemia. For a number of RBC enzyme defects (eg, deficiencies of hexokinase, glucose phosphate isomerase, pyruvate kinase), the sole clinical manifestation is hemolytic anemia. Glucose-6-phosphate dehydrogenase deficiency is the most common metabolic error of the red cell and presents with acute hemolytic anemia in response to oxidant stress (eg, drugs, acute infections, fava bean ingestion).

This is a consultative evaluation looking at red cell enzyme defects as the cause for early red cell destruction.

Reference Values
Definitive results and an interpretive report will be provided.
Interpretation
A hematopathologist expert in these disorders evaluates the case, appropriate tests are performed, and an interpretive report is issued.

Cautions
No significant cautionary statements

Clinical Reference

Performance

Method Description
Erythrocyte Enzyme Interpretation:
A hematopathologist who is an expert in these disorders evaluates the case, appropriate tests are performed and an interpretive report is issued.

Glucose-6-Phosphate Dehydrogenase (G6PD):

Pyruvate Kinase:
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 di-nucleotide and measuring the rate of decrease in absorbance at 340 nm. (Beutler E: Red Cell Metabolism: A Manual of Biochemical Methods. Third edition. New York, Grune and Stratton, 1984, pp 68-71)

Glucose Phosphate Isomerase:
Washed erythrocytes are hemolyzed and the hemolysate is mixed with glucose, adenosine triphosphate (ATP), glucose-6-phosphate dehydrogenase, and nicotinamide adenine dinucleotide phosphate (NADP). The reduction of NADP is measured spectrophotometrically and is proportional to the enzymatic conversion of ATP and glucose to glucose-6-phosphate. (Beutler E: Red Cell Metabolism: A Manual of Biochemical Methods. Third edition. New York, Grune and Stratton, 1984, pp 40-42)

Hexokinase:
Hexokinase (in the presence of magnesium) catalyzes the reaction of ATP and glucose to G-6-P and ADP. In this assay the formation of glucose-6-phosphate (G-6-P) is measured by linking its further oxidation to 6-phosphogluconate (6-PG) to the reduction of NADP through the glucose-6-phosphate dehydrogenase (G-6-PD) reaction. The increase in absorbance which occurs as NADP+ is reduced is measured at 340 nm. (Beutler E: Red cell metabolism: A Manual of Biochemical Methods. Third edition. Grune and Stratton, New York, 1984, pp 38-40)
Glutathione:

Virtually all of the nonprotein sulfhydryl of red cells is in the form of reduced glutathione (GSH). 5,5'-dithiobis (2-nitrobenzoic acid) is a disulfide compound which is readily reduced by sulfhydryl compounds, forming a highly colored yellow anion. The absorbance of this resultant yellow substance is measured by 412 nm and compared to that of a known standard. (Beutler E: Red cell metabolism. In A Manual of Biochemical Methods, Second edition, Grune and Stratton, 1984)

Day(s) and Time(s) Test Performed
Monday through Friday; Varies

Analytic Time
2-10 days (not reported Saturday or Sunday)

Maximum Laboratory Time
13 days

Specimen Retention Time
14 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
See Individual Test IDs

CPT Code Information
82955-G-6-PD
84087-Glucose phosphate isomerase
84220-Pyruvate kinase
82657-Hexokinase
82978-Glutathione (if appropriate)
83915-RBC Enzymes (if appropriate)

LOINC® Information
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<th>Test Order Name</th>
<th>Order LOINC Value</th>
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<tr>
<td>EEEVP</td>
<td>RBC Enzyme Evaluation</td>
<td>In Process</td>
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<table>
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<td>G6PD_</td>
<td>G-6-PD, QN, RBC</td>
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<td>GPI_</td>
<td>Glucose Phosphate Isomerase, B</td>
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<td>HEXK_</td>
<td>Hexokinase, B</td>
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<td>PK_</td>
<td>Pyruvate Kinase, RBC</td>
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