Test Definition: P5NT
Pyrimidine 5’ Nucleotidase, B

Overview

Useful For
Evaluation of persistent reticulocytosis and marked basophilic stippling
Evaluation of hemolytic anemia

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: 5 mL

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

Specimen Minimum Volume
3 mL

Reject Due To

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<th>Gross hemolysis</th>
<th>Reject</th>
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Specimen Stability Information

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<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>20 days</td>
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Clinical and Interpretive

Clinical Information
Pyrimidine 5’ nucleotidase (P5’NT) is involved in the catabolism of RNA, which is a normal constituent of reticulocytes but not of mature erythrocytes. A deficiency of P5’NT (also called uridine 5’ monophosphate hydrolase) is a cause of congenital non-spherocytic hemolytic anemia (OMIM #266120) and is associated with a persistent reticulocytosis. Deficiency of P5’NT (also called uridine 5’ monophosphate hydrolase) is caused by homozygous or compound heterozygous mutations of the NT5C3A gene at chromosome 7p14 and results in the abnormal accumulation of pyrimidine nucleotides. The disorder is classically associated with basophilic stippling of the red blood cells. Assaying for the presence of pyrimidine nucleotides serves as a surrogate marker for P5’NT deficiency, as the enzymatic assay is difficult.

Reference Values

Normal

Interpretation

A normal result indicates the absence of pyrimidine nucleotides and indicates normal P5’NT function. An abnormal result (abnormal spectral scan) indicates the presence of pyrimidine nucleotides and likely P5’NT deficiency. Lead poisoning can also inhibit P5NT function. If this is suspected, correlation with blood lead levels is recommended.

Cautions

Lead inhibits P5NT activity; therefore, blood lead levels should be performed to exclude a reversible cause.

Clinical Reference


Performance

Method Description

Pyrimidine nucleotides have a spectral absorption curve that is markedly different from that exhibited by (normally present) adenine nucleotides, eg, adenosine triphosphate. The former have a peak at about 270 nm; the latter at about 257 nm. Thus, pyrimidine 5’ nucleotidase deficiency may be ascertained by demonstrating a very high spectral absorption maximum of 270 nm in erythrocyte extracts. (Beutler E: Red Cell Metabolism. A Manual of Biochemical Methods. Third edition. Grune and Stratton. 1984, pp 100-102)

PDF Report

No

Day(s) and Time(s) Test Performed

Monday through Friday

Analytic Time

10 days

Maximum Laboratory Time

10 days

Specimen Retention Time

1 week

Performing Laboratory Location
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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
83915

LOINC® Information

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