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

Gross hemolysis	Reject

Specimen Stability Information

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

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 P5Ã¢â‚¬â„¢NT is caused by homozyogous 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 P5Ã¢â‚¬â„¢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

1. Rees DC, Duley JA, Marinaki AM: Pyrimidine 5' nucleotidase deficiency. Br J Haematol 2003 Feb;120(3):375-383

2. Fairbanks VF, Klee GG: Biochemical aspects of hematology. In Tietz Textbook of Clinical Chemistry. Third edition. Edited by CA Burtis, ER Ashwood, Philadelphia, WB Saunders Co., 1999, pp 1642-1647

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

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

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

Test ID	Test Order Name	Order LOINC Value
P5NT	Pyrimidine 5' Nucleotidase, B	2902-5

Result ID	Test Result Name	Result LOINC Value
2734	Pyrimidine 5' Nucleotidase, B	2902-5