Test Definition: P5NT

Pyrimidine 5' Nucleotidase, Blood

Overview

Useful For

Evaluation of marked basophilic stippling

Evaluation of hemolytic anemia

Method Name

Kinetic Spectrophotometry

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Specimen Required

Specimen Type: Whole blood
Container/Tube:
Preferred: Yellow top (ACD solution B)
Acceptable: Lavender top (EDTA)
Specimen Volume: 5 mL
Collection Instructions: Send whole blood specimen in original tube. **Do not aliquot**.

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 & Interpretive

Clinical Information

Pyrimidine 5' nucleotidases (P5'Ns) are catabolic enzymes that regulate cellular nucleotide and nucleoside levels through the dephosphorylation of noncyclic nucleoside 5'-monophosphates. P5'N activity is much higher in reticulocytes than in aged red blood cells due to increased demand during erythroid maturation. Reticulocyte ribosomal RNA degradation results in pyrimidine nucleotide residues that require conversion to nucleosides to allow diffusion outside the cell. Disruption of this process results in intracellular pyrimidine nucleotide accumulation visible as coarse basophilic stippling.

Several different 5'-nucleotidase enzymes have been identified with distinctive substrate specificity, cellular localization, and tissue distribution. Only P5'N type 1 is known to be associated with P5'N deficiency (also called uridine 5' monophosphate hydrolase deficiency), a cause of congenital nonspherocytic hemolytic anemia (OMIM 266120, autosomal recessive). The disorder manifests as mild/compensated to moderate hemolytic anemia with persistent reticulocytosis. Additional features include jaundice/neonatal hyperbilirubinemia, splenomegaly, and characteristic marked basophilic stippling on the peripheral blood smear. Coincident hemoglobin E may lead to a more severe hemolytic anemia.

Pyrimidine 5' nucleotidase deficiency is caused by homozygous or compound heterozygous alterations in the *NT5C3A* gene, mapped to chromosome 7p14. Assaying for the presence of pyrimidine nucleotides serves as a surrogate marker for P5'N deficiency and is not specific for a diagnosis of hereditary P5'N deficiency. Enzyme function is magnesium ion-dependent and is inhibited by metal chelating reagents, such as EDTA. Activity is inhibited by heavy metal ions including lead, mercury, copper, nickel, and cadmium, and toxic levels can cause accumulation of intracellular pyrimidine nucleotides.

Reference Values

Normal

Interpretation

A normal result indicates the absence of pyrimidine nucleotides and indicates normal pyrimidine 5' nucleotidase (P5'N) function.

An abnormal result (abnormal spectral scan) indicates the presence of pyrimidine nucleotides and possible P5'N deficiency. Enzyme activity is inhibited by heavy metal ions, including lead, mercury, copper, nickel, and cadmium. Toxic levels can cause accumulation of intracellular pyrimidine nucleotides. If results are abnormal clinical correlation is recommended to exclude heavy metal poisoning.

Cautions

This assay serves as a surrogate marker for decreased pyrimidine 5' nucleotidase (P5'N) activity from any cause and is not specific for a diagnosis of hereditary P5'N deficiency.

Enzyme activity is inhibited by metal chelating reagents, such as EDTA.

Since enzyme activity is inhibited by heavy metal ions (including lead, mercury, copper, nickel, and cadmium) and toxic levels can cause accumulation of intracellular pyrimidine nucleotides, clinical correlation to exclude heavy metal poisoning is indicated for abnormal results.

Recent transfusion can result in false negative results.

Clinical Reference

1. Rees DC, Duley JA, Marinaki AM. Pyrimidine 5' nucleotidase deficiency. Br J Haematol. 2003;120(3):375-383
2. Zanella A, Bianchi P, Fermo E, Valentini G. Hereditary pyrimidine 5'-nucleotidase deficiency: from genetics to clinical manifestations. Br J Haematol. 2006;133(2):113-123
3. Fairbanks VF, Klee GG. Biochemical aspects of hematology. In: Burtis CA, Ashwood ER, eds. Tietz Textbook of Clinical Chemistry. 3rd ed. WB Saunders; 1999:1642-1647
4. Gregg XT, Prchal JT. Red blood cell enzymopathies. In: Hoffman R, Benz, Jr EJ, et al, eds. Hematology : Basic Principles and Practice, 7th ed. Elsevier; 2018:616-625
5. Warang P, Colah R, Kedar P. Lead poisoning induced severe hemolytic anemia, basophilic stippling, mimicking erythrocyte pyrimidine 5'-nucleotidase deficiency in beta thalassemia minor. J Clin Toxicol. 2017;7(2):1000346. doi:10.4172/2161-0495.1000346

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. 3rd ed. Grune and Stratton; 1984:100-102; 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

Monday through Friday

Report Available

10 days

Specimen Retention Time

7 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

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