Test Definition: PTNT

Prothrombin G20210A Mutation, Blood

Overview

Useful For

Patients with clinically suspected thrombophilia

Determination of the duration of anticoagulation therapy of venous thromboembolism patients

Screening for women contemplating hormone therapy

Genetics Test Information

This test detects the *F2* c.*97G>A variant (legacy G20210A).

Special Instructions

- Informed Consent for Genetic Testing
- Coagulation Patient Information
- Informed Consent for Genetic Testing (Spanish)

Method Name

Direct Variant Analysis

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Ordering Guidance

This assay will only detect the *F2* c.*97G>A (rs1799963) variant associated with thrombophilia. To detect other pathogenic alterations in the *F2* gene of a patient with a laboratory diagnosis of thrombophilia, order F2NGS / F2 Gene Next Generation Sequencing, Varies.

This assay will not detect alterations in individuals with thrombophilia caused by mechanisms other than the *F2* c.*97G>A variant. For those situations consider ordering AATHR / Thrombophilia Profile, Plasma and Whole Blood.

Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
Container/Tube:
Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD solution B), light-blue top (sodium citrate)
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube. **Do not** aliquot.

Forms
1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Coagulation Patient Information (T675)

Specimen Minimum Volume
1 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Extracted DNA	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	14 days	
	Refrigerated	14 days	
	Frozen	14 days	

Clinical & Interpretive

Clinical Information
The prothrombin (PT) *F2* c.*97G>A (legacy G20210A) variant is a common variant within the 3' untranslated region of the prothrombin gene, affecting 1.5% to 3% of white Americans, especially persons of southern European ancestry. The *F2* c.*97G>A variant is less common among African Americans (carrier frequency of 0.4%). The *F2* c.*97G>A variant is associated with a 3-fold increased risk of venous thromboembolism due to increased plasma prothrombin activity among carriers.

Reference Values
Negative

Interpretation
The results will be reported as:

-Negative for the c.*97G>A variant
-Heterozygous for the c.*97G>A variant
-Homozygous for the c.*97G>A variant

Cautions

This assay will not detect alterations in individuals with thrombophilia caused by mechanisms other than the prothrombin *F2* c.*97G>A variant.

Rare single nucleotide variants under the primers can cause preferential amplification of one allele. In many cases, there is no indication that this interference has occurred. Consequently, the analysis could be done on data from only one allele, which may cause a false-negative result or an incorrect allele frequency (homozygous instead of heterozygous).

Patients receiving allogenic stem cell transplants prior to having blood drawn for DNA based testing may have false-normal or abnormal results depending on the genotype of the stem cell donor.

Consultations with the Mayo Clinic Special Coagulation Clinic Thrombophilia Center, and/or Medical Genetics are available and may be especially helpful in complex cases or in situations in which the diagnosis is atypical or uncertain. Genetic counseling is recommended before testing asymptomatic family members.

Clinical Reference

1. Poort SR, Rosendaal FR, Reitsma PH, Bertina RM: A common genetic variation in the 3'untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increase in venous thrombosis. Blood. 1996;10:3698-3703
2. Makris M, Preston FE, Beauchamp NJ, et al: Co-inheritance of the 20210 A allele of the prothrombin gene increases the risk of thrombosis in subjects with familial thrombophilia. Thromb Haemost. 1997;78:1426-1429
3. De Stefano V, Martinelli I, Mannucci PM, et al: The risk of recurrent venous thrombosis among heterozygous carriers of both factor V Leiden and the G20210A prothrombin mutation. N Engl J Med. 1999;341:801-806
4. Freed J, Bauer KA: Thrombophilia: clinical and laboratory assessment and management. In: Kitchens CS, Kessler CM, Konkle BA, Streiff MB, Garcia DA, eds. Consultative Hemostasis and Thrombosis. 4th ed. Elsevier; 2019:242-265

Performance

Method Description

An allelic discrimination assay is set up using TaqMan chemistry. End-products are analyzed using a real-time polymerase chain reaction instrument for genotype detection.(Package insert: TaqMan SNP Genotyping Assays. Applied Biosystems; 2014)

PDF Report

No

Day(s) Performed

Weekly

Report Available

3 to 5 days

Specimen Retention Time

Whole blood: 2 weeks

Performing Laboratory Location

Rochester

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

81240-F2 (prothrombin, coagulation factor II) (eg, hereditary hypercoagulability) gene analysis, 20210G>A variant

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PTNT	Prothrombin G20210A Mutation, B	24475-6

Result ID	Test Result Name	Result LOINC® Value
21803	Prothrombin G20210A Mutation, B	24475-6
21804	PTNT Interpretation	69049-5
21806	PTNT Reviewed By	18771-6