Overview

Useful For
Direct mutation analysis for the prothrombin (PT) G20210A allele should be reserved for patients with clinically suspected thrombophilia. There may be additional indications for direct PT G20210A mutation testing, such as in determining the duration of anticoagulation therapy of venous thromboembolism patients and screening for women contemplating hormone therapy.

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

Method Name
DirectMutationAnalysis

NY State Available
Yes

Specimen

Specimen Type
Whole blood

Specimen Required

Container/Tube:

Preferred: Yellow top (ACD solution A or B)

Acceptable: EDTA or sodium citrate

Specimen Volume: Full tube

Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.

Additional Information: Can be combined with other molecular coagulation tests:

- MTHAC / 5,10-Methylenetetrahydrofolate Reductase A1298C, Mutation, Blood
- F5DNA / Factor V Leiden (R506Q) Mutation, Blood
- MTHFR / 5,10-Methylenetetrahydrofolate Reductase C677T, Mutation, Blood
- MTHP / 5,10-Methylenetetrahydrofolate Reductase C677T and A1298C Mutations, Blood
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 in Special Instructions:

   - Informed Consent for Genetic Testing (T576)
   - Informed Consent for Genetic Testing-Spanish (T826)

2. Coagulation Patient Information (T675) in Special Instructions

3. If not ordering electronically, complete, print, and send a **Coagulation Test Request** (T753) with the specimen.

**Specimen Minimum Volume**

1 mL in a 3-mL ACD tube

**Reject Due To**

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<thead>
<tr>
<th>Condition</th>
<th>Acceptance</th>
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<tr>
<td>Hemolysis</td>
<td>Mild OK; Gross OK</td>
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<tr>
<td>Lipemia</td>
<td>Mild OK; Gross OK</td>
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<tr>
<td>Icterus</td>
<td>NA</td>
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<tr>
<td>Other</td>
<td>Green-top (heparin) tube or extracted DNA</td>
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**Specimen Stability Information**

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<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
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<tr>
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<tr>
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**Clinical and Interpretive**

**Clinical Information**

Venous thromboembolism (VTE) is a syndrome of deep vein thrombosis and its complication, pulmonary embolism. The prothrombin (PT) G20210A mutation (F2 rs1799963) is a common polymorphism within the 3' untranslated region of the prothrombin gene, affecting 1.5% to 3% of Caucasian Americans, especially persons of southern European ancestry. The PT G20210A allele is uncommon among African Americans (carrier frequency of 0.4%). The PT G20210A mutation is associated with a 3-fold increased risk of venous thromboembolism due to increased plasma prothrombin activity among carriers.

The PT G20210A gene mutation test is a direct mutation analysis of patient blood leukocyte genomic DNA. At present, there are no other methods of detecting this VTE risk factor except for direct mutation testing.

**Reference Values**

Negative

**Interpretation**
The interpretive report will include sample information, assay information, background information, and conclusions drawn from the test results (normal, heterozygous prothrombin [PT] G20210A, homozygous PT G20210A).

**Cautions**

This direct mutation analysis will not detect individuals with thrombophilia caused by mechanisms other than the prothrombin (PT) G20210A mutation. Special Coagulation Clinic, Thrombophilia Center, and/or Medical Genetics consultations are available for Mayo Clinic patients 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**


**Performance**

**Method Description**

Direct mutation analysis using PCR amplification, signal generation, and release by cleavage of sequence specific alleles.(Invader Factor II, Invader Plus Chemistry, Hologic, Madison, WI)

**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Monday through Friday; 12 p.m.

**Analytic Time**

3 days

**Maximum Laboratory Time**

5 days

**Specimen Retention Time**

Whole blood stored 2 weeks

**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 has been modified from the manufacturer's instructions. Its performance characteristics were 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
81240-F2 (prothrombin, coagulation factor II) (eg, hereditary hypercoagulability) gene analysis, 20210G->A variant

LOINC® Information

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<td>Prothrombin G20210A Mutation, B</td>
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<td>21804</td>
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