



**Molecular Diagnostic Laboratory**  
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## **Prothrombin Mutation Analysis**

Prothrombin (*F2*) point mutation G20210A

### **Indications for Molecular Testing**

- Family history of venous thromboembolism (VTE)
- Unprovoked thrombotic event at age of <45
- Women with multiple stillbirths or spontaneous abortions

### **Testing Methodology**

The assay utilizes the Invader Plus® chemistry manufactured by Hologic for the detection of the F2 20210G>A (c.\*97G>A) mutation in genomic DNA (AF478696.1 transcript isoform). Invader and allele-specific probes match the mutant and the wild-type alleles and have overlapping 5'-ends that are cleaved upon perfect hybridization to the amplified DNA. The cleaved 5'-end of the primary probes transiently hybridize with a corresponding fluorescence resonance energy transfer (FRET) cassettes triggering the cleavage of the fluorophore from the cassette by the cleavage enzyme and allowing signal release and detection.

### **Interpretation of DNA analysis**

Prothrombin (coagulation factor II) is the precursor of thrombin, which participates as a serine protease in the coagulation cascade. The Pro G-20210-A mutation is present in 1-2% of the general Caucasian population and has been associated with an increased risk of venous thrombosis similar to that of factor V Leiden (FVL). For individuals presenting with venous thromboembolism (VTE), Pro G-20210-A occurs in 6-8% of those of all ages. About 10% of FVL carriers with first VTE will also have the prothrombin variant. In addition, Pro G-20210-A has been associated with myocardial infarction in young women, cerebral vein thrombosis in oral contraceptive users, preeclampsia and pregnancy complications and infarctions at other sites. The relative risk for thrombosis in the Pro G-20210-A heterozygous state (AG) is increased by a factor of 2.8. Patients with both FVL and this *F2* mutation have a 20-fold increased risk for recurrent VTE.

### **Specimen Requirements**

**Peripheral blood**--1 lavender-top (EDTA) tube. Invert several times to mix blood.

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**Barnes-Jewish Hospital, Institute of Health**  
**Mail Stop 90-28-344**  
**425 South Euclid Avenue, Room 5970**  
**St. Louis, MO 63110**

Clinical information must be provided with specimen referral in order to correctly interpret test results.

### **Current Pricing**

Contact Lab Customer Service for current pricing 314 362-1470.  
CPT code: 81240

1. Heit, JA. Thrombophilia: Common Questions on Laboratory Assessment and Management. *Hematology Am Soc Hematol Educ Program*. 2007:127-35.
2. Varga EA and Kujovich JL. Management of inherited thrombophilia: guide for genetics professionals. *Clinical Genetics*. 2012 Jan. 81(1): 7-17.