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Factor II (Prothrombin) Mutation Analysis

Test Ordering Code:	1062
CPT Codes:	83891, 83896 x 2, 83898, 83903
Specimen:	Whole blood anticoagulated with EDTA, 1mL minimum. Maintain specimen at 4° C after collection and ship at room temperature.
Turnaround Time:	5-7 business days
Clinical Significance:	A mutation in the factor II (prothrombin) gene (G20210A) has been found to be associated with thrombosis and is the second most common genetic cause of thrombosis. Patients heterozygous for this mutation have an increased risk of thrombotic disorders. The overall frequency of the mutant allele is approximately 1-6% in Caucasian populations, but rare in others. If this allele is co-inherited with the Factor V Leiden mutation, the severity of thrombotic problems may be more severe and the age of onset may be earlier.
Indications for Testing:	History of recurrent venous thromboembolism (VTE) First VTE at younger than 50 years of age First Unprovoked VTE at any age First VTE at an unusual anatomic site: (cerebral, mesenteric, portal, or hepatic veins) First VTE related to pregnancy, the puerperium, or oral contraceptive use Women with unexplained pregnancy loss First VTE at any age in patients with first-degree family members with VTE
Methodology:	Real - Time PCR
Reporting of Results:	Negative: The patient is negative for Factor II, G20210A allele. Heterozygous: The patient is heterozygous for Factor II, G20210A allele. Homozygous: The patient is homozygous for Factor II, G20210A allele.
Intended Use:	This test only detects the factor V Leiden mutation (G1691A), and not mutations in other genes associated with thrombophilia.
References:	McGlennenRC, Key NS. Clinical and laboratory management of the prothrombin G20210A mutation. Arch Pathol Lab Med. 2002; 126(11):1319-25.