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Factor V (Leiden) Mutation Analysis

Test Ordering Code:	1061
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:	The factor V Leiden mutation (G1691A; R506Q) predisposes patients to thrombophilia and is the most common genetic cause of venous thrombosis. Patients heterozygous for this mutation have an increased risk of thrombosis and it is estimated to be involved in approximately 20 - 40% of cases of venous thrombosis. The overall frequency of the mutant allele is approximately 3% in Caucasian populations.
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 V Leiden, R506Q allele. Heterozygous: The patient is heterozygous for Factor V Leiden, R506Q allele. Homozygous: The patient is homozygous for Factor V Leiden, R506Q allele.
Intended Use:	This test only detects the factor V Leiden mutation (G1691A), and not mutations in other genes associated with thrombophilia.
References:	Bertina RM, et al. Mutation in blood coagulation factor V associated with resistance to activated protein C. Nature 1994; 369: 64-67 Voelkerding KV. Resistance to activated protein C and a novel factor V gene mutation. Clin Lab Med, 1996, 16(1): 169-86 (review) Press RD, et al. Clinical utility of factor V Leiden (R506Q) testing for the diagnosis and management of thromboembolic disorders. Arch Pathol Lab Med. 2002 Nov; 126(11): 1304-18