Plasminogen Activator Inhibitor-1, PAI-1 (SERPINE1) Genotyping
ARUP test code 2004980

PAI-1 Specimen: Whole Blood

PAI-1 Interpretation: 4G/5G

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Indication for testing: Assess genetic risk for thrombosis.

Result: PAI-1 Genotype 4G/5G

Interpretation: One copy each of the PAI-1 4G and 5G alleles (4G/5G) were detected. The 4G allele is associated with increased PAI-1 plasma activity levels and confers a mild risk for venous thromboembolism in individuals having no other thrombophilic impairment. In individuals with other thrombotic risk factors, the identified 4G/5G genotype confers a significant risk for venous thromboembolism as well as an increased risk for myocardial infarction.

Recommendations: If clinically indicated, testing for other inherited or acquired thrombophilic disorders is recommended. For additional testing information, refer to the Antiphospholipid Syndrome, Hypercoagulable States, Thrombophilia, or Venous Thromboembolism topics at arupconsult.com.

This result has been reviewed and approved by Yuan Ji, Ph.D.
Background Information: Plasminogen Activator Inhibitor-1, PAI-1 (SERPINE1) Genotyping:

Characteristics: The 4G allele within in the promoter region of the PAI-1 (SERPINE1) gene is associated with higher plasma PAI-1 activity when compared with the 5G allele. Heterozygosity or homozygosity for the 4G allele confers a risk for venous thromboembolism (VTE), especially in individuals with other thrombophilic risk factors, as well as a risk for myocardial infarction. Frequency of the 4G Allele: Caucasian 0.52, Hispanic 0.38, African-American 0.13-0.28.

Variant Tested: The PAI-1 promoter 4G/5G polymorphism located in the promoter region of the SERPINE1 gene. NM_000602.3(SERPINE1) c.-817dupG (from start of translation).

Inheritance: Autosomal dominant.

Clinical sensitivity: Unknown.

Methodology: Polymerase chain reaction and fluorescence monitoring.

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Variants in the PAI-1 (SERPINE1) gene, other than the 4G/5G polymorphism, are not evaluated. Diagnostic errors can occur due to rare sequence variations.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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H=High, L=Low, *=Abnormal, C=Critical