**Plasminogen Activator Inhibitor-1, PAI-1 (SERPINE1) Genotyping**

ARUP test code 2004980

**PAI-1 Specimen**  
Whole Blood

**PAI-1 Interpretation**  
5G/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 5G/5G

**Interpretation:** Two copies of the PAI-1 5G allele (5G/5G) were detected. The 4G allele, which is associated with increased PAI-1 plasma activity levels, was not detected. This result does not exclude other genetic, or non-genetic, causes for thrombosis.

**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