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## Test Update

From Your Laboratory Service Provider, PathGroup Labs

### Plasminogen Activator Inhibitor-1 (PAI-1) 4G/5G Polymorphism

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#### Overview

PathGroup is now offering a PAI-1 genotyping assay for 4G/5G polymorphism. This assay is used as an aid to diagnosis venous thromboembolism and related thrombotic disorders. Plasminogen activator inhibitor-1 (PAI-1) is a primary regulator of the fibrinolytic system *in vivo*<sup>1</sup>. PAI-1 is an important inhibitor of the fibrinolytic system; therefore, elevated levels of PAI-1 could suppress fibrinolysis and increase risk of thrombosis. A common single nucleotide polymorphism (4G/5G) at position - 675 of the promoter region leads to differences in PAI-1 production. In vitro studies suggest that the 4G allele has higher activity than the 5G allele because the 5G allele contains an additional binding site for a DNA-binding protein that can be a transcriptional repressor<sup>1</sup>. In two out of three studies, the lowest levels of PAI-1 occur in those subjects with the 5G/5G genotype, intermediate levels in 4G/5G subjects, and highest levels in 4G/4G subjects<sup>2</sup>. Most recent publications have shown that, homozygosity or heterozygosity for the 4G PAI-1 allele is associated with thrombotic risk, myocardial infarction, pregnancy complications, and risk of meningococcal sepsis in children. No association of 5G allele with these conditions has been shown. Prevalence of 4G/4G, 4G/5G, and 5G/5G genotypes are 31.8%, 46.6%, and 21.6% respectively.

#### Clinical Utility

- ✍ Venous thromboembolism
- ✍ Evaluation of thrombotic risk
- ✍ Myocardial infarction
- ✍ Coronary artery disease
- ✍ Pregnancy complications
- ✍ Risk meningococcal sepsis

**Method:** Genotyping by Invader™ technology

**Specimen Collection and Storage:** 3-5 ml EDTA whole blood (lavender-top tube). Store and transport at room temperature. If delayed more than 72 hours, store and transport refrigerated. Do not freeze specimen.



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