

# TEST: <u>DETECTION OF THE PLASMINOGEN ACTIVATOR INHIBITOR-1 (PAI-1)</u> 4G/5G GENE POLYMORPHISM BY PCR

### PRINCIPLE:

Plasminogen activator inhibitor-1 (PAI-1) is an essential regulatory component of the fibrinolytic pathway. It binds to tissue plasminogen activator and inhibits conversion of plasminogen to plasmin, which leads to decreased fibrinolysis. A single base deletion at nucleotide 675 (4G/5G) from the start of the promoter region of *PAI-1* gene is responsible for higher levels of PAI-1 protein, due to high levels of transcription. Individuals with 4G/5G (heterozygous) or 4G/4G (homozygous) genotypes have elevated levels of PAI-1, which can suppress fibrinolysis and cause an increased risk of thrombosis. PAI-1 levels may be associated with pathogenetic mechanisms of myocardial infarction, coronary artery disease and ischemic stroke. Additionally, women with pregnancy complications, such as pre-eclampsia, stillbirth and recurrent spontaneous miscarriages, were shown to be homozygous (4G/4G) for the *PAI-1* gene.

## SPECIMEN COLLECTION AND PREPARATION:

10mL whole blood collected in lavender top EDTA tubes (two 5ml tubes). Specimen should be delivered to the laboratory within 72 hours at room temperature. Peripheral blood specimens that are clotted, have not been collected in EDTA, or frozen are not acceptable.

#### **METHOD:**

Polymerase chain reaction (PCR) and reverse hybridization.

#### REFERENCES:

- 1. Doggen CJ, Bertina R, Manger C et al. *Thromb Haemost* 1999;82:115-120
- 2. Buchholz T, Lohse P, Rogenhofer N et al. Hum Reprod 2003;18:2473-77
- 3. Dossenbach-Glaninger A et al. *Clinical Chemistry* 2003;49:1081-86

**Normal Range:** Reported as Normal (5G/5G), Heterozygous Mutated (4G/5G) or Homozygous Mutated (4G/4G)

Turnaround time: 10 business days