TEST: DETECTION OF THE FACTOR V H1299R (HR2) GENE POLYMORPHISM BY PCR

PRINCIPLE:
Coagulation Factor V is an enzyme cofactor that participates in the coagulation cascade and contributes to a normal haemostatic balance. Mutations in the *Factor V* gene are among the causes for venous thrombosis. Other conditions that are associated with mutations in the *Factor V* gene are pregnancy complications, such as recurrent pregnancy losses. The most common mutation in the *Factor V* gene is the Factor V Leiden mutation. Recently, another polymorphism in the *Factor V* gene, the His1299Arg polymorphism, has been identified and linked to hereditary thrombophilia.

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. de Visser M et al. *Thromb Haemost* 2000;83:577-82
2. Castaman G et al. *Hematologica* 2003;88:1182-89

Normal Range: Reported as Normal (H/H), Heterozygous Mutated (H/R) or Homozygous Mutated (R/R)

Turnaround time: 10 business days