INTRODUCTION

Phenomena of thrombophilia are commonly defined as blockage of blood circulation by clots, which originate in the veins or are stem from a thrombus in another area of the body. Thrombosis can be the result of events that activate the coagulation system, which include injury, surgery, immobilization, pregnancy and the use of estrogens and oral contraceptives. In addition, the genetic background of a person influences the individual risk of thrombosis. Mutations in genes coding for factors of blood homeostasis and fibrinolysis may lead to a lifelong increased risk of thrombosis. Today, several of such genetic alterations are known.

Factor V is an inactive pro-cofactor of the coagulation system in the blood. The active form (Factor Va) can be inactivated by proteolysis at specific amino acids. According to the current hypothesis, Factor-V-related thrombosis can be caused by a multitude of genetic mutations affecting sites of the Factor V protein that are crucial for inactivation by proteolysis.

A common mutation is a G→A transition in exon 10 of the Factor V gene, which leads to the substitution of the amino acid arginine by a glutamine in position 506 of the Factor V protein. This mutated form of Factor V is known as Factor V Leiden and is resistant to inactivation by the active protein C (APC). The Factor V Leiden mutation is found in over 90% of patients with APC-resistance. This mutation is relatively frequent in Caucasian Europeans and North Americans and very rare in non-Caucasians. In Europe, the frequency of Factor V Leiden carriers is significantly higher in Southern Europeans populations (7%) compared to Northern European populations (2% to 3%).

Individuals that are heterozygous for Factor V Leiden have a seven (7) times higher risk to develop deep vein thrombosis, compared to the normal population, whereas homozygous carriers of the mutation have an eighty (80) times higher risk.

Using Real-Time PCR this mutation can be detected quickly and with high specificity and sensitivity.