SUMMARY

DNA analysis techniques are powerful tools in the study of the molecular basis of disease. Hemostasis is a complex homeostatic mechanism protectinγ the organism from blood loss. Its main part is the coagulation mechanism, a complex of plasma proteins creating an activation cascade aiming at blood clot formation.

The term thrombophilia refers to a group of clinical syndromes characterized by blood hypercoagulable state, which often result in thromboembolic occurences. In its hereditary form, thrombophilia is due to mutations in coagulation and anticoagulation factor genes.

In this thesis the coagulation factor IX gene was studied in thrombophilic patients showing normal coagulation lab values after in vitro addition of normal recombinant human factor IX. The promoter and the coding regions (exons) of the FIX gene were amplified and scanned for insertion/deletion as well as for point mutations.

50 patients were checked for insertion/deletion mutations by polyacrylamide gel electrophoresis of the amplified regions without any positive result. 28 of the above patients were furtherly checked for point mutations by sequencing the amplified regions. In 3 patients the polymorphism A21975T was detected in exon 6. This polymorphism causes the substitution of the residue Thr^{194} by Ala in factor IX protein (T194A), has already been described in literature and has a prevalence of 0.23 in normal individuals, showing no effect in the factor's conformation and function.