

Coagulation Factor V Leiden R506Q

This mutation (G1691A) occurs in about 8 % of Caucasians and encodes a structurally altered factor V protein (R506Q) which is cleaved less efficiently (about 10%) by activated protein C than the wild type protein. Consequently, the R506Q mutation causes a pro-thrombotic state. Clinically, it is associated with deep venous thromboembolism, recurrent miscarriages, portal vein thrombosis in cirrhotic patients, early kidney transplant loss and other forms of venous thromboembolism.

References

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