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Risk factors for Venous Thromboembolism

Venous thromboembolism (VTE), with its main clinical presentations of deep-vein thrombosis (DVT) and pulmonary embolism (PE), represents a common and serious disorder which is still a major cause of morbidity and mortality in developed countries (1). Among Europeans, first-time occurrence of VTE is age-dependent, sex-independent and has an incidence rate of 1–3 per 1,000 individuals per year (2).

It is generally known that the interplay of inherited and environmental factors and life events underlies the pathogenesis of VTE. Within environmental factors, provoking and non-provoking are known, whereby the relative impact on thrombogenic diathesis is different and for some still controversial or of emerging interest.

Genetic risk factors have been identified by studying families of thrombophilia patients including the relatively common factor (F) V Leiden and prothrombin 20210 polymorphisms together with blood group non-0. In addition, mutations in the natural anticoagulant proteins antithrombin (AT), protein C (PC) and protein S (PS) are considered as significant causes for VTE.

New techniques like high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders (BPDs) offers knowledge about novel pathogenic variants but turns also the spotlight to the question of genotype-phenotype correlations in BPDs (3).

With this talk, we will discuss a selection of environmental and genetic risk factors for VTE and their respective influence on a cumulative thrombogenic diathesis. Furthermore, the discussion will place a special emphasis on diagnostic procedures that are at the moment recommended for clinical settings and which are more related to scientific purposes.

1. Bafunno V, Margaglionex M. Genetic basis of thrombosis. Vol. 48, Clinical Chemistry and Laboratory Medicine. 2010.

2. Heit JA, Spencer FA, White RH. The epidemiology of venous thromboembolism. J Thromb Thrombolysis. 2016;41(1):3–14.

3. Simeoni I, Stephens JC, Hu F, Deevi SV V, Megy K, Bariana TK, et al. A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood. 2016;127(23):2791–803.