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Title: Thrombophilias in 60 Bulgarian patients with “idiopathic” venous thromboembolism

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Body: In the last two decades, many inherited and acquired thrombophilias predisposing venous thromboembolism (VTE) became known. The aim of this real life study is to analyze the frequencies of some thrombophilias in patients with acute VTE, in whom no underlying diseases or triggering-VTE factors are present (“idiopathic”). 60 ethnic Bulgarians (34 men and 26 women, mean age 34.47) with VTE (39 with deep vein thrombosis (DVT) only, 12 with pulmonary embolism (PE) only and 9 with both) were examined for: Leiden mutation (FVL), prothrombin factor II mutation (G20210A), plasminogen activator inhibitor-1 mutation (PAI-1), C677T and A1298C of methylenetetrahydrofolate reductase (MTHFR) mutations; deficiency of protein C (pr.C), deficiency of protein S (pr.S), deficiency of antithrombin III (AT III) and antiphospholipid antibodies syndrome (APLS). In 48 (80%) of patients (79.48% in DVT, 80.95% in PE patients) were found in total 79 abnormalities. FVL heterogeneous mutation was found in 15 (25% of all patients) and homogeneous in 2 (3.3%) patients. FVL represents 21.51% of all found abnormalities. PAI-1 was found in 17 patients and represents 21.51% of all abnormalities. 11 (18.3%) were homogeneous and 6 (10%) were heterogeneous in combination with other abnormalities. 6 (10%) had G20210A mutation and 14 (23%)- deficiency of AT III. We also found deficiency of Pr.S in 6, Pr.C in 3; mutations C677T in 4, A1298 in 7 patients. APLS was found in 5 patients. We conclude that most of the patients with “idiopathic” VTE have one or more thrombophilias. In addition to the known role of FVL as a thrombophilic factor, the PAI-1 mutation has a prominent frequency in our group.