Synergy
in Hemophilia

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Frequency of FV Leiden and FII G20210A Mutations in Patients with Inherited Antithrombin Deficiency from Serbia

Branko Tomic[1], Mirjana Kovac[2], Iva Pruner[1], Maja Gvozdenov[1], Sofija Dunjic[1], Marija Cumbo[1], Dragica Radojkovic[1], Valentina Djordjevic[1]

[1]Institute of Molecular Genetics and Genetic Engineering, University of Belgrade

[2]Faculty of Medicine, University of Belgrade; Blood Transfusion Institute of Serbia, Hemostasis Department

PURPOSE: Thrombosis is multicausal disease in which both acquired and genetic risk factors play important roles. The most frequent genetic risk factors known to date are the Factor V G1691A (FV Leiden) and FII G20210A mutations. On the other hand, inherited antithrombin (AT) deficiency, caused by mutations in the AT gene (SERPINC1) is a very rare disorder, but it is associated with significant risk for thrombotic complications. AT deficiency is classified into two types: type-I is a quantitative disorder characterized by decreased amount and activity of AT, while type-II is a qualitative - functional disorder. Aim of our study was to analyze the frequency of FV Leiden and FII G20210A mutations in patients with inherited AT deficiency from Serbia.

METHODOLOGY: A study was carried out in large group of AT deficiency patients from Serbia. Cohort of 42 subjects (15m/27f; 36.7±18.7y) from 18 Serbian families included 24 symptomatic and 18 asymptomatic first-degree relatives. Among them, type-I AT deficiency were detected in 9 families (19 members: 6m/13f; 37.1±19.0y) and type-II in 9 families (23 members: 9m/14f; 36.5±18.8y). FV Leiden and FII G2010A mutations were detected by PCR, followed by digestion with specific restriction enzymes (PCR-RFLP).

RESULTS: We have detected 3 FV Leiden heterozygous carriers in 3 different families (1 with type-I and 2 with type-II AT deficiency). All 3 carriers were symptomatic. Regarding FII G20210A mutation, 2 heterozygous carriers, both asymptomatic and from same family with type-I deficiency, were identified. According to our findings in families with AT deficiency from Serbia frequency of FV Leiden and FII G20210A mutation are 16.7% and 5.6%, respectively.

CONCLUSION: This is the first study in which frequency of FV Leiden and FII G20210A mutations in patients with inherited AT deficiency from Serbia were examined. Results of our study suggest that these mutations can be relevant for AT deficiency patients' phenotype, but further studies are required.

Keywords: Antithrombin deficiency, FV Leiden, FIIG20210A