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Content

CAN PHARMACOGENETIC VARIANTS IN *TPMT*, *MTHFR* AND *SLCO1B1* GENES BE USED AS POTENTIAL MARKERS OF OUTCOME PREDICTION IN SYSTEMIC SCLEROSIS PATIENTS?

Marina Jelovac,[1] Nikola Kotur,[1] Bojan Ristivojevic,[1] Djordje Pavlovic,[1] Vesna Spasovski, [1] Nemanja Damjanov, [2,3] Sonja Pavlovic, [1] Branka Zukic[1]

[1]*Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia;* [2]*Institute of Rheumatology, Belgrade, Serbia;* [3]*Medical School, University of Belgrade, Belgrade, Serbia*

Introduction: Systemic sclerosis (SSc) is a rare autoimmune disorder that affects connective tissues and has the highest morbidity and mortality among rheumatologic diseases. Clinical presentations as well as disease progression are highly heterogeneous between patients, implying a strong need for individualization of therapy.

Methods: Four pharmacogenetic variants, namely *TPMT* rs1800460, *TPMT* rs1142345, *MTHFR* rs1801133 and *SLCO1B1* rs4149056 were tested for association with severe disease outcomes in 102 patients with SSc from Serbia treated either with immunosuppressants azathioprine (AZA) and methotrexate (MTX) or with other types of medications. Genotyping was performed using PCR-RFLP and direct Sanger sequencing. R software was used for statistical analysis and development of polygenic risk score (PRS) model.

Results: Association was found between *MTHFR* rs1801133 and higher risk for elevated systolic pressure in all patients except those prescribed with MTX, and higher risk for kidney insufficiency in patients prescribed with other types of drugs. In patients treated with MTX, variant *SLCO1B1* rs4149056 was protective against kidney insufficiency. For patients receiving MTX a trend was shown for having a higher PRS rank and elevated systolic pressure.

Conclusion: Our results open a door wide for more extensive research on pharmacogenomics markers in patients with SSc. Altogether, pharmacogenomics markers could predict the outcome of patients with SSc and help in prevention of adverse drug reactions.

Key words: Systemic Sclerosis; Pharmacogenetics markers; Methotrexate; Azathioprine; Rheumatologic Dis-eases; Personalized therapy

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