



GENETICS AND GENODERMATOSES

XRCC1 RS1799782 ARG194TRP AND RS25487 ARG399GLN POLYMORPHISMS INFLUENCE ON DERMATOMYOSITIS AND SYSTEMIC LUPUS ERYTHEMATOSUS IN BULGARIAN PATIENTS

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Introduction: It is well known that genetic, hormonal, and environmental factors are potential triggers of the autoimmune process in systemic lupus erythematosus (SLE) and dermatomyositis (DM). Evidences have suggested that the impaired DNA repair efficiency is implicated in the development of autoimmune diseases as well. XRCC1 and XRCC3 play central role in mammalian DNA repair processes and might influence susceptibility to such conditions.

Objective: The aim of this case-control study was to investigate the influence of the XRCC1 Arg194Trp (C>T) and Arg399Gln (G>A) polymorphisms over DM and SLE susceptibility in Bulgarian patients.

Materials and methods: A cohort of 88 patients, subdivided as group of 55 with SLE and 33 with DM, as well as 94 unrelated healthy controls, were included in this study.

Results: None of the polymorphisms showed association with SLE, DM or their clinical parameters. The allele and genotype frequency of the two SNPs was similar to those found in the other Caucasian healthy population.

Conclusion: Our results indicate that the XRCC1 rs1799782 Arg194Trp and rs25487 Arg399Gln polymorphisms are not independent factors in the development of SLE and DM.

Keywords: XRCC1, XRCC3, dermatomyositis, systemic lupus erythematosus

