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Tumor necrosis factor- promoter polymorphism (308G/A) in Egyptian patients with systemic lupus erythematosus (SLE)

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Background: Tumor Necrosis Factor alpha (TNF-alpha) and lymphotoxin alpha are pivotal cytokines in the pathogenesis of systemic lupus erythmatosis.

Objectives: To investigate the possible association of the polymorphism of the TNF promoter gene at position -308 and that of the LTA gene at position 252 with susceptibility to SLE and with phenotypic disease features in Egyptians patients.

Subjects and Methods: A case control study involving 100 SLE patients and 100 unrelated healthy controls. Polymerase chain reaction and restriction fragment length (PCR-RFLP) methods were applied to detect polymorphism in TNF Promoter (-308 G>A) and LTA 252 A>G.

Results: We found TNF- 308 genotype AA was significantly increased by 26% in SLE patients compared to 10% in the control group and genotype LTA 252 GG showed a significant increase by 22% in SLE patients compared to 6% in the control group. Mutant allele A of TNF and mutant allele G of LTA were significantly associated with SLE ($p<0.001$, OR=2.29, 2.31 and CI=1.49-3.52, 1.48-3.6 respectively). Genotype (AA+GA) of TNF was significantly associated with clinical manifestations as malar rash, arthritis, oral ulcers,serositis and systemic lupus erythematosus disease activity index(SLEA1). Genotype (GG+GA) of LTA was significantly associated with arthritis.

Conclusion: These results suggest that TNF and LTA genetic polymorphisms contribute to SLE susceptibility in the Egyptian population and are associated with disease characteristics.

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Chitinase 3-like-1 and its receptors in Hermansky-Pudlak syndrome-associated lung disease

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Hermansky-Pudlak Syndrome (HPS) comprises a group of inherited disorders caused by mutations that alter the function of lysosome-related organelles. Pulmonary fibrosis is the major cause of morbidity and mortality in BLOC-3 mutant HPS-1 and HPS-4 patients. Chitinase 3-like-1 (CHI3L1), a prototypic chitinase-like protein, plays a protective role by ameliorating cell death and stimulating fibro-proliferative repair. Here we demonstrate that circulating CHI3L1 levels are higher in HPS patients with pulmonary fibrosis compared to those that remain fibrosis-free and that these levels associate with disease severity. Using murine models we also demonstrate that a defect in CHI3L1 inhibition of epithelial apoptosis and exaggerated CHI3L1-driven fibro-proliferation play important roles in HPS fibrosis. Lastly we demonstrate that these divergent responses are mediated by differences in the trafficking and effector functions of two CHI3L1 receptors. Specifically, the enhanced sensitivity to apoptosis is due to the BLOC-3 dependent and thus abnormal, trafficking of IL-13Ra2. In contrast, the fibrosis is due to interactions of CHI3L1 and CRTH2, which traffics normally in BLOC-3 HPS.

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