

## Genetics of Atopic Dermatitis: Role of Cytokines Genes Polymorphisms

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**Abstract :** Atopic dermatitis (AD), also known as atopic eczema, is a chronic inflammatory skin disease characterized by severe itching and recurrent relapsing eczema-like skin lesions, affecting up to 20% of children and 10% of adults in industrialized countries. AD is a complex multifactorial disease, and its exact etiology and pathogenesis have not been fully elucidated. The aim of this study was to investigate the impact of gene polymorphisms of T helper cell subtype Th1 and Th2 cytokines, interferon-gamma (IFN- $\gamma$ ), interleukin-6 (IL-6) and transforming growth factor (TGF)- $\beta$ 1 on AD susceptibility in a Saudi cohort. One hundred four unrelated patients with AD and 195 healthy controls were genotyped for IFN- $\gamma$  (874A/T), IL-6 (174G/C) and TGF- $\beta$ 1 (509C/T) polymorphisms using ARMS-PCR and PCR-RFLP technique. The frequency of genotypes AA and AT of IFN- $\gamma$  (874A/T) differed significantly among patients and controls ( $P \leq 0.001$ ). The genotype AT was increased while genotype AA was decreased in AD patients as compared to controls. AD patients also had higher frequency of T containing genotypes (AT+TT) than controls ( $P = 0.001$ ). The frequencies of allele T and A were statistically different in patients and controls ( $P = 0.04$ ). The frequencies of genotype GG and allele G of IL-6 (174G/C) were significantly higher while genotype GC and allele C were lower in AD patients than controls. There was no significant difference in the frequencies of alleles and genotypes of TGF- $\beta$ 1 (509C/T) polymorphism between patient and control groups. These results showed that susceptibility to AD is influenced by presence or absence of genotypes of IFN- $\gamma$  (874A/T) and IL-6 (174G/C) polymorphisms. It is concluded that T-allele and T-containing genotypes (AT+TT) of IFN- $\gamma$  (874A/T) and G-allele and GG genotype of IL-6 (174G/C) polymorphisms are susceptible to AD in Saudis. On the other hand, the TGF- $\beta$ 1 (509C/T) polymorphism may not be associated with AD risk in Saudi population however further studies with large sample size are required to confirm these findings.

**Keywords :** atopic dermatitis, interferon- $\gamma$ , interleukin-6, transforming growth factor- $\beta$ 1, polymorphism

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