Association between single nucleptide polymorphism (rs1800468) in the transforming growth factor beta 1 gene (TGFB1) and multiple sclerosis

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Abstract

Introduction: Multiple sclerosis (MS) is a neurodegenerative autoimmune disease characterized by recurrent episodes of demyelination and axonal injury mediated primarily by CD4+ T-helper cells with a pro inflammatory Th1 phenotype, macrophages, and soluble mediators of inflammation. Cytokines released by Immune cells play an important role in the pathogenesis of the disease characterized by periods of exacerbations and remissions. Among them, there are pro-inflammatory cytokines like interleukin-2 (IL-2), tumor necrosis factor-a (TNF-a) or interferon-g (IFN-g) produced by Th1 cells and cytokines with immunosuppressive properties like IL-4, transforming growth factor-b1 (TGF-b1) and/or IL-10. Among them Transforming growth factor-b1 (TGF-b1) has an important role in suppression of the immune system in autoimmune diseases. TGF-b is a potent regulatory cytokine with diverse effects on hemopoietic cells. The pivotal function of TGF-b in the immune system is to maintain tolerance via the regulation of lymphocyte proliferation, differentiation, and survival. There are some reports about variation in -800 nucleotide in autoimmune diseases uch as Parkinson, Alzimer, Rheumatoid arthritis. Defects in TGF-b1such as in its expression correlate with the onset of several autoimmune diseases. Paradoxically, TGF-b1 also acts as a pro-inflammatory cytokine and induces interleukin 17-producing pathogenic T helper cells (Th IL-17 cells) synergistically during an inflammatory response in which interleukin 6 is produced. TGF-b1 gene located on the complement strand of chromosome 19 an the location is 19q13.1 intreset was to determine wether the *TGFB1* promoter region (rs1800468) polymorphism is related to the molecular pathogenesis of multiple sclerosis in Iranian population.

Materials and Methods: The blood samples were collected from 50 multiple sclerosis patients and PBMCs gathered. Genomic DNA from PBMCs extracted, PCR primers designed for promoter region of TGF-b1 and the length of amplicon is 680 bp, then PCR amplification exacuted. Digestion of PCR products were done by Tai1 restriction enzyme and agarose gel electrophoresis runned to separate digested fragments. Our data analysed by

Results: single nucleotide polymorphism (rs1800468) exists significantly in multiple sclerosis patients.

Conclusion: -800 SNP is significant in Iranian patients with multiple sclerosis and cause of its position, which is in promoter region, we clime that -800 SNP can have a significant track on the TGF-b1 expression.

Key words: *SNP, TGF-b1, Multiple Sclerosis.*