

Abstract:

Multiple sclerosis (MS) is a complex disease in the central nervous system (CNS) and the most common inflammatory demyelinating and degenerative disease in young adults. Most studies have shown that vitamin D may interfere with the pathophysiology of MS within the CNS. Vitamin D does not only interfere with immune responses of the periphery, but it can play a role in regulating the neurodegeneration and repair processes within the CNS. *CYP27B1* gene encodes the enzyme that catalyses the formation of the biological active metabolite 1, 25(OH) 2D form of the abundant metabolite 25(OH) D. *CYP27B1* is expressed on a variety of cells including neurons, glial cells and invaded lymphocyte. We analyzed the polymorphism profiling of *CYP27B1* gene in rs703842 SNP among 209 MS patients and 221 age_matched healthy individual controls in eastern of Iran. DNA was extracted from whole blood using the boiling method. The rs703842 was genotyped applying *RsaI* restriction enzyme. SPSS 19 for Windows (SPSS Inc., Chicago, IL) was used to analyze the data. Genotypic and allelic frequencies were compared between patients and controls using the Chi-Square Tests. We investigated the genotypic and allelic analyses of this risk SNP but did not find any association with complete MS cohort, but the stratification of gender and MS subtype analyses were showed significantly associated with genotyping in MS ($p > 0.05$).

Keywords: polymorphism, *CYP27B1*, multiple sclerosis, PCR-RFLP



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**Association study of promoter
polymorphism of the *CYP27B1* gene with
multiple sclerosis patients in East of Iran**

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