

## A C/T Polymorphism at the 5' Untranslated Region of CD40 Gene in Patients Associated with Graves' Disease in Kumaon Region

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**Abstract :** Background: Graves' disease is an autoimmune disorder with a genetic predisposition, and CD40 plays a pathogenic role in various autoimmune diseases. A single nucleotide polymorphism at position -1 of the Kozak sequence of the 5' untranslated regions of the CD40 gene of exon 1 has been reported to be associated with the development of Graves' Disease. Objective: The aim of the present study was to investigate whether CD40 gene polymorphism confers susceptibility to Graves' disease in the Kumaon region. CD40 gene polymorphisms were studied in Graves' Disease patients (n=50) and healthy control subjects without anti-thyroid autoantibodies or a family history of autoimmune disorders (n=50). Material and Method: CD40 gene polymorphisms were studied in fifty Graves' Disease patients and fifty healthy control subjects. All samples were collected from STG Hospital, Haldwani, Nainital. A C/T polymorphism at position -1 of the CD40 gene was measured using the polymerase chain reaction-restriction fragment length polymorphism. Results: There was no significant difference in allele or genotype frequency of the CD40 SNP between Graves' Disease and control subjects. There was a significant decrease in the TT genotype frequency in the Graves' Disease patients who developed Graves' Disease after 40 years old than those under 40 years of age. These data suggest that the SNP of the CD40 gene is associated with susceptibility to the later onset of Graves' Disease. Conclusion: The CD40 gene was a different susceptibility gene for Graves' Disease within certain families because it was both linked and associated with Graves' Disease.

**Keywords :** autoimmune diseases, pathogenesis, diagnosis, therapy

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