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Abstract Topic: - Statistical genetics and genetic epidemiology

Abstract Title: - Association of single nucleotide polymorphisms in IL-6 gene with tuberculosis: A case control study from population of Punjab, India.

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Aims: - Tuberculosis (TB) is an infectious disease which remains a major health problem, worldwide. Interestingly, out of total infected individuals only 5-10% develop clinical signs of this disease. The host genetic factors may contribute to this differential susceptibility to TB. Among other factors, cytokine gene polymorphism have garnered significant attention. Many pro and anti-inflammatory cytokines are involved in pathophysiology of TB. Interleukin (IL)-6 act as a pro-inflammatory as well as anti-inflammatory cytokine which plays a crucial role in immune response against this disease. It has been observed that absence of IL-6 leads to delayed expression of IFN-Gamma response in TB. Keeping in view the importance of IL-6 in TB and to fill the existing gap, present study was designed with the aim of evaluating the association of rs2069824 C/T and rs1800795 G/C SNPs of IL-6 in TB patients from population of Punjab, India.

Methods: - This is a case-control study. A total of 450 subjects including 300 TB patients along with age, sex and endogamous group matched 150 normal healthy controls (NHCs) were recruited for the study. Genotyping of selected SNPs of IL-6 was done by Amplification Refractory Mutation System-Polymerase Chain Reaction (ARMS-PCR) method. Plasma levels of IL-6 were measured by enzyme linked immunosorbent assay (ELISA). Statistical significance level was calculated at 0.05. This study was approved by the institutional ethics committee of Panjab University, Chandigarh.

Results: - It was found that statistically significant difference exists in distribution of genotypic ($p < 0.00001$) and allelic counts ($p < 0.0003$) of rs2069824 C/T SNP in TB patients and NHCs. The CT and CC genotypes were associated with decreasing the risk of TB in co-dominant model ($p < 0.0001$). No association of rs1800795 G/T SNP of IL-6 was found with TB in the studied population. Plasma levels of IL-6 were measurable in some of the TB patients and did not vary significantly with time period of ATT.

Conclusions: - The SNP rs2069824 C/T of IL-6 is associated with TB in the population of Punjab, India.

Keywords: - It was found that statistically significant difference exists in distribution of genotypic ($p < 0.00001$) and allelic counts ($p < 0.0003$) of rs2069824 C/T SNP in TB patients and NHCs. The CT and CC genotypes were associated with decreasing the risk of TB in co-dominant model ($p < 0.0001$). No association of rs1800795 G/T SNP of IL-6 was found with TB in the studied population. Plasma levels of IL-6 were measurable in some of the TB patients and did not vary significantly with time period of ATT.