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ASSOCIATION OF FUNCTIONAL POLYMORPHISM IN PROTEIN TYROSINE PHOSPHATASE NON-RECEPTOR 22 (PTPN22) GENE WITH VITILIGO SUSCEPTIBILITY

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Vitiligo is an acquired, autoimmune skin disorder characterized by melanocyte loss resulting into progressive depigmentation of skin and hair. Several genes have been implicated in the pathogenesis of vitiligo. The protein tyrosine phosphatase non-receptor 22 (PTPN22) gene, which encodes the lymphoid tyrosine phosphatase (LYP) protein, is a non-HLA gene associated with autoimmune diseases. The functional PTPN22 C1858T (R620W) polymorphism is widely associated with an increased risk for vitiligo in Europeans however controversy exists in other populations. The aim of this study was to determine if the PTPN22 C1857T polymorphism confers susceptibility to vitiligo in Saudi population. Genomic DNA was extracted and amplified using tetra primer ARMS-PCR method for detection of PTPN22 C1857T polymorphism. We genotyped 125 Saudi vitiligo patients and 200 healthy controls. The frequencies of alleles and genotypes in patients and healthy controls were compared. The frequency of T-allele and CT genotype of PTPN22 C1858T polymorphism was significantly higher while the frequency of C-allele and CC genotype was lower in patients as compared to controls (P 0.0001). The homozygous genotype TT was absent in both the patients and controls. These results indicated that individuals containing allele T are susceptible to vitiligo. We conclude that PTPN22 C1858T polymorphism is strongly associated with susceptibility to vitiligo with a relative risk of >97 and can be developed as biomarker for evaluating vitiligo risk. However, further studies on PTPN22 C1857T polymorphism in larger samples from different geographical areas and ethnicity are warranted.

BIOGRAPHY

Ghaleb Bin Huraib has completed his PhD in dermatological and venereal diseases from Fribourge University, Germany. Earlier he did MBBS from faculty of medicine / King Saud University, Riyadh. He is the deputy director of Medical Services Department (MSD) for armed forces, Saudi Arabia. He has published several papers on genetic basis of dermatological diseases.

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