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Genetic Analysis of PTPN22 C1858T and GAD65 Antibodies in Iranian Children and Adolescents with Type 1 Diabetes

Pourya Fathollahzadeh[1], Farshad Foroughi[2], Shamim Nonejad[3], Fatemeh Saffari[4], Amir Javadi[5], Sanaz Keshavarz Shahbaz[6*]

[1]Student Research Committee, Qazvin University of Medical Sciences, Qazvin, Iran USERN Office, Qazvin University of Medical Sciences, Qazvin, Iran
[2]Cellular and Molecular Research Center, Research Institute for Prevention of Non-Communicable Disease, Qazvin University of Medical Sciences, Qazvin, Iran
[3]Department of Biochemistry & Genetics, School of Medicine, Qazvin University of Medical Sciences, Qazvin, Iran
[4]Departmentt of Pediatric Endocrinology, Children Growth Research Center, Research Institute for Prevention of Non-Communicable Diseases, Qazvin University of Medical Sciences, Qazvin, Iran
[5]Department of Community Medicine, School of Medicine, Qazvin University of Medical Sciences, Qazvin, Iran
[6]Research Institute for Prevention of Non-Communicable Disease, Qazvin University of Medical Sciences, Qazvin, Iran

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***Corresponding Author:**
Research Institute for Prevention of Non-Communicable Disease, Qazvin University of Medical Sciences, Qazvin, Iran

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ABSTRACT

Introduction: Type 1 diabetes (T1D) is an autoimmune disorder that destroys pancreatic beta cells, leading to chronic hyperglycemia. Genetic factors, particularly the *PTPN22* gene C1858T polymorphism, are thought to increase susceptibility to T1D. This study investigated the association of the *PTPN22* gene C1858T polymorphism with serum GAD65 autoantibody levels in Iranian children and adults with T1D to enhance the understanding of genetic predispositions in this population.
Methods and Materials: This case-control study included 98 patients with T1D and 94 healthy controls, matched by age and gender, from Qods Hospital in Qazvin, Iran. Blood samples were collected, and serum levels of GAD65 autoantibodies were measured using the ELISA method. Genomic DNA was extracted from peripheral blood mononuclear cells and amplified by PCR to identify the PTPN22 C1858T polymorphism. PCR products were analyzed via RFLP and agarose gel electrophoresis. Statistical analyses were performed using IBM-SPSS software version 24, comparing the prevalence of the C1858T polymorphism and GAD65 autoantibody levels between patients and the control group with chi-square and student's t-tests.
Results: The study involved 98 T1D patients (age range 3-18 years; 54% male) and 94 healthy controls (age range 4-18 years; 50% male). Serum GAD65 antibody levels significantly elevated in T1D patients (mean: 85.6 U/mL) compared to the control group (mean: 12.4 U/mL; $p = 0.001$). The CT genotype was present in 16.3% of T1D patients versus 9.6% of controls, and the T allele frequency was 8.2% in patients compared to 4.8% in controls. However, these differences were insignificant ($p = 0.10$ and $p = 0.09$, respectively). Among T1D patients, those with the CT genotype had a mean GAD65 level of 92.1 U/mL, whereas those with the CC genotype had a mean level of 83.2 U/mL, indicating no significant difference ($p = 0.45$). The analysis also revealed that 53% of the patient group were male, with an average age of 10.2 years, compared to 51% male and an average age of 9.8 years in the control group. The CT genotype was associated with a 1.8-fold increased risk of T1D, but this association was not statistically significant ($p = 0.08$).
Conclusion and Discussion: Our findings suggest a notable, albeit statistically non-significant, increase in the 1858T allele frequency among T1D patients. The association between the PTPN22 C1858T polymorphism and GAD65 autoantibody levels warrants further investigation with larger sample sizes to establish a definitive genetic link. These findings highlight the potential importance of genetic screening in predicting the risk of T1D, which could lead to earlier diagnosis and improved management strategies for this autoimmune disease.

Keywords: Anti-GAD65 autoantibody, Diabetes mellitus, Polymorphism

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