The role of PTPN22 gene polymorphism in childhood immune thrombocytopenic purpura.

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Abstract

Immune thrombocytopenia is an autoimmune disorder characterized by antibody-mediated platelet destruction. A protein tyrosine phosphatase (PTPN22) present in lymphocytes is an important negative regulator of signal transduction for the T-cell receptor-MHC complex and has been associated with autoimmune disorders that produce autoantibodies. The present study investigated the frequency of the 1858C>T single-nucleotide polymorphism (SNP) in the PTPN22 gene in idiopathic thrombocytopenic purpura (ITP) patients. This case series study included 50 children with ITP, 24 acute and 26 chronic cases, and 50 normal children as a control group. All were subjected to clinical history and laboratory investigations including complete blood count, genotyping of PTPN22 1858C/T SNP by polymerase chain reaction-restriction fragment length polymorphism and platelet antibodies using platelets suspension immunofluorescence test for the cases. Thirteen patients (26%) were positive for the PTPN22 1858C>T SNP. Three patients were homozygous for the mutation and 10 were heterozygous. Comparison of the 26% of the ITP patients who were positive for the PTPN22 1858C>T mutation with the 6% positive in the control group yielded a P value of 0.006. Antiplatelet antibodies were detected in five patients (20.8%) with acute ITP and in three patients (11.5%) with chronic ITP; no significant association between the presence of PTPN22 1858C>T mutation and the presence of antiplatelet antibodies was detected. The prevalence of PTPN22 gene mutation was higher in ITP patients, thus it may be considered as a genetic risk factor in the development of ITP in Egyptian children.

Keywords:

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