Association of P2X7 gene polymorphisms with susceptibility to pulmonary tuberculosis in Zahedan, Southeast Iran

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ABSTRACT. Susceptibility to tuberculosis may be influenced by variations in human genes. The P2X7 receptor is an ATP-gated cation channel expressed in immune cells, and it influences the release of proinflammatory cytokines from monocytes and macrophages. In the present study, we aimed to evaluate the impact of P2X7 gene rs2393799 (-762T/C) and rs1718119 (Thr348Ala) polymorphisms on patient susceptibility to pulmonary tuberculosis (PTB) in a sample of the Iranian population. This case-control study was performed using 150 PTB cases and 150 controls. P2X7 receptor polymorphisms were determined using tetra-amplification refractory mutation system-polymerase chain reaction. Genotype and allelic frequencies of the rs2393799 variant
within the \textit{P2X7} gene were significantly higher in the PTB patients than in the healthy controls. The genotypes were CC in 71, CT in 54, and TT in 25 PTB patients. The genotypes were CC in 104, CT in 40, and TT in 6 healthy controls. The results indicate a significant association between rs2393799 polymorphism of the \textit{P2X7} gene and susceptibility to PTB (CT vs CC: OR = 6.5, 95\%CI = 2.5-16.9, P < 0.0001; TT vs CC: OR = 3.3, 95\%CI = 1.2-8.9, P = 0.018; TC+TT vs CC: OR = 2.56, 95\%CI = 1.59-4.12, P < 0.0001). The rs2393799 T allele is a risk factor for predisposition to PTB (OR = 2.53, 95\%CI = 1.73-3.71, P < 0.0001). No association between the rs1718119 polymorphism and PTB was found. In conclusion, the rs2393799 polymorphism in the \textit{P2X7} gene may contribute to patient susceptibility to PTB in our study population.

\textbf{Key words:} Tuberculosis; P2X7; Polymorphism