Association of the dopamine transporter gene (DAT1) core promoter polymorphism -67 T allele with schizophrenia

Abstract

Background: Dysfunction of the central dopaminergic neurotransmission has been suggested to play an important role in the etiology of schizophrenia. The dopamine transporter (DAT1) mediates the active reuptake of dopamine from the synapses and thereby plays a key role in the regulation of the dopaminergic neurotransmission. In this study, we sought to determine the possible association of the DAT1 gene core promoter polymorphism -67A/T with schizophrenia in a case/control study.

Methods: The allele and genotype frequencies of the polymorphism were studied in 100 patients and 100 controls, which were matched on the basis of sex, age and ethnicity.

Results: The genotype frequencies in the patients group were as follows: AA 29%, AT 59%, TT 12% vs. the genotype frequencies in the control group: AA 57%, AT 38%, TT 5%, \( \chi^2 = 16.54 \), df = 2, OR = 2.25 (95% CI 1.46-3.45, \( P = 0.0003 \)).

Conclusion: For the first time, these findings provide tentative evidence for the contribution of the DAT1 gene core promoter polymorphism to the etiopathology of schizophrenia at least in the Iranian male population that we studied. Replication studies of independent samples and family-based association studies are necessary to further evaluate the significance of our findings.

Keywords: DAT1 / Promoter / Schizophrenia / Association / Dopamine Polymorphism