Importance of ATM gene as a susceptible trait: predisposition role of polymorphism in breast cancer.

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Abstract

The involvement of ATM gene and specifically, the important role of D1853N polymorphism, as a three-hit hypothesis has been previously reported in an Iranian proband affected with brain tumor and this polymorphism could be screened in her relatives as well. The aim of present study was to investigate the involvement of D1853N polymorphism as a predisposition factor in 129 Iranian patients affected with primary breast cancer and 248 sex- and age-matched healthy controls. Mutant allele-specific PCR amplification (MASA) assay was performed to analyze the D1853N polymorphism in the ATM gene. The frequency of D1853N polymorphism in cases, internal and external controls was 31.0% (40/129), 26.9% (28/104) and 12.5% (18/144), respectively. The frequency of D1853N in total control groups, including normal external control and pedigree internal control, was 18.6% (46/248). The odds ratio was calculated with the logistic regression test, with an estimated relative risk of 2.579 (P = 0.005). The significant difference was observed between the patient-carriers of this alteration and external controls (P = 0.001). The number of controls harboring D1853N polymorphism was higher in internal control compared to external controls, and the difference was statistically significant (P = 0.004). The significant difference was observed between the patient-carriers and external controls and could be considered as a predisposing and diagnostic marker in the population and specifically in the cancer-prone pedigrees.

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