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## Polymorphisms in the MTHFR gene are associated with breast cancer.

[Ergul E](#), [Sazci A](#), [Utkan Z](#), [Canturk NZ](#).

### Source

Department of Medical Biology and Genetics, Faculty of Medicine, University of Kocaeli, Kocaeli, Turkey.

### Abstract

The methylenetetrahydrofolate reductase (MTHFR) gene is a polymorphic gene involved in folate metabolism, DNA biosynthesis, methylation and genomic integrity in actively dividing cells. The MTHFR C677T and A1298C polymorphisms are likely to play an important role in the susceptibility to breast cancer. In this case-control study, we examined the role of MTHFR C677T and A1298C polymorphisms in breast cancer patients. We genotyped 118 premenopausal women with sporadic breast cancer and 193 controls, using a PCR-RFLP method. The allele frequencies of the MTHFR 677T were 31.36% in the breast cancer cases and 28.76% in the controls. The allele frequencies of the MTHFR 1298C were 37.29% in the breast cancer subjects and 31.35% in the controls. Frequencies of MTHFR C677C, C677T and T677T were 50.8, 33.9 and 14.4% in the breast cancer patients and 48.7, 45.1 and 6.2% in the controls, respectively. The results of a chi(2) analysis indicated that the MTHFR 677T allele was significantly distributed (chi(2) = 7.234; p = 0.027). Likewise, the MTHFR T677T genotype showed a 2.5-fold increased risk for breast cancer and the C1298C genotype showed a 1.9-fold increased risk for breast cancer. In the compound genotypes, T677T/A1298A and C677C/C1298C showed a 4.472- and a 2.301-fold increased risk for breast cancer (OR = 4.472, p = 0.001, and OR = 2.301, p = 0.024), respectively. In conclusion, our data suggest that the MTHFR 677T, 1298C alleles, T677T, C1298C genotypes, and C677C/C1298C and T677T/A1298A compound genotypes are genetic risk factors for premenopausal women with sporadic breast cancer.

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