ABSTRACT: GATA3 is an enriched transcription factor in mammary epithelium. To date, there has been no study on the relationship between microsatellites in the GATA3 gene and breast cancer risk. In this study, we investigated the existence of polymorphisms in the cytosine-thymine (CT) dinucleotide repeat in intron 3 of the GATA3 gene and its association with breast cancer risk. A case-control study of 206 breast cancer patients and 262 controls was conducted in Iranian women. Several different CT repeat alleles of GATA3 were detected in both the patients and controls. The frequencies of 17 and 18 alleles in patients were significantly lower than controls. Our findings demonstrate that women who carry 17-CT (OR = 0.5; p = 0.003) or 18-CT (OR = 0.41, p = 0.02) alleles of GATA3 gene are at lower risk of developing breast cancer. The highest protection against breast cancer was observed with heterozygotes of 16/17 repeats (OR = 0.12, p = 0.02). Also, the presence of the 17-CT allele has a positive relation with estrogen receptor expression. However, we found that the allelic length of GATA3 polymorphisms had no significant effect on the age onset or grade of the disease, as well as the expression of progesterone receptors and HER2.

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