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Analysis of ERBB2 rs2517956 in breast cancer susceptibility-A study from India

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Breast cancer (BC) is one of the most common cancers affecting the morbidity and mortality of females worldwide. The incidence of breast cancer is constantly rising in India with 3% annual increase. Numerous risk factors have been identified for breast cancer, however, the genetic predisposition may play an important role inbreast carcinogenesis. ErbB2, a receptor tyrosine kinase with intrinsic tyrosine kinase activity was known to play a significant role in human malignancies. Several studies indicated that amplification of ErbB2 disrupts normal cell-control mechanisms and gives rise of aggressive tumor cells. ERBB2 was found to overexpress in approximately 30% of human breast cancers. Anumber of case-control studies had been conducted to explore the association between the genetic polymorphisms of the ERBB2 gene in various diseases including breast cancer. However, the results remained inconsistent and this could be due to the difference in frequencies of polymorphic variants that differsby ethnicity. The present case-control study was conducted in a total of 468 subjects to better understand the role of promoter polymorphism (rs2517956) in predisposition to BC for the first time in Indian population. Genomic DNA was isolated from blood samples and PCR-RFLP was performed using specific primers. The results revealed a significant association of this polymorphism with breast cancer risk ($p<0.05$). However, there was no influence of this SNP on various demographic andclinical parameters such as menopausal status, tumor stage, ER, PR and HER2 receptor status. In conclusion, it appears that rs2517956 of ERBB2 gene play a significant role in breast cancer susceptibility in Indian women.

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