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Genetically, breast cancer is categorized into sporadic and familial. \textit{BRCA1} is the most studied gene in the field of breast cancer research. Although important progress has been made in understanding the pathogenesis of breast cancer, however, our idea is nebulous. Studies to find out the role of \textit{BRCA1} promoter SNPs and promoter DNA methylation in any Indian population has not been carried out. Hence, the present study was planned to unveil the association pattern of \textit{BRCA1} promoter SNP c.-2265C>T (rs11655505) with the predisposition of breast cancer and role of promoter methylation in sporadic breast cancer in an Indian population. Screening for \textit{BRCA1} (c.-2265C/T) rs11655505 variation was performed in 352 female breast cancer patients and 380 healthy women, irrespective of their age and menopausal status. Later for the ease of statistical analysis they were grouped according to their age, menopausal status and the familial history. Twenty nine sporadic breast tumors and 26 breast biopsy materials were also used in this study. Isolation of DNA from blood and tissue samples, their mutational analysis was performed. Isolation of mRNA from tissues was done as well as gene expression and promoter methylation status was studied for sporadic cancer. Statistical analysis revealed that CT genotype of rs11655505 (c.-2265C/T) is significantly associated with menopausal breast cancer patients \((p=0.01)\). Meanwhile none of the allele found associated with breast cancer risk in premenopausal, familial
and non-familial cancer patients. Somatic BRCA1 gene mutations had not been found in sporadic breast tumors. An alternative mechanism, hypermethylation of CpG island in the promoter region of the gene is known to be strongly associated with gene silencing. Results of this study suggested that the methylation of BRCA1 promoter is found in 31.03% of the tumor samples which is higher than that of earlier reports. CT genotype was found to be associated significantly with the low expression of BRCA1 gene in tumor samples \( (p=0.04) \). Our present study suggests that CT genotype of rs11655505 (c.-2265C/T) is significantly associated with menopausal breast cancer patients and decreased expression of \textit{BRCA1} gene in sporadic breast cancer cases. Methylated status of BRCA1 gene promoter is associated with decreased expression of the gene in tumor samples but not in normal biopsy samples. In addition to this the methylated status was significantly associated with the ER negative tumor samples.