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Novel Sequence Variants and a High Frequency of Recurrent Polymorphisms in *BRCA1* Gene in Breast Cancer Women of North Coastal Andhra Pradesh

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ABSTRACT Owing to its high incidence coupled with relatively good prognosis, breast cancer is the most prevalent cancer in the world today. Germ line mutations in the susceptibility gene *BRCA1* in hereditary breast/ovarian cancer, though low in prevalence, are highly penetrant and show geographical variations. Most cancer-associated *BRCA1* mutations identified to date result in the premature translational termination of the protein. However, the molecular and genetic effects of missense mutations remain largely unknown. There have been only a few reports from India on mutations in *BRCA1* and none from North Coastal Andhra Pradesh. We have analyzed 114 breast cancer patients with (N = 9) and without (N = 105) a family history of breast cancer, 22 at risk relatives from familial (n=11), sporadic (n=11) cases and 97control subjects for mutations in exons 2 and 11 and their intron-exon boundaries of *BRCA1 gene* by direct sequencing. Sequence alignment was carried by CLUSTAL W and PSI-BLAST. Out of eight sequence variants found, one novel deleterious frame-shift mutation (c.2717insA), one novel polymorphism (c.1400A>G), five previously reported common polymorphisms in exon 11 and one intronic (intron 1) variant (base1822C>T) were observed. All the identified polymorphisms in exon 11 fall in DNA binding domain of BRCA1 protein.