

Breast Cancer and BRCA Gene: A Study on Genetic and Environmental Interaction

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Abstract : Breast cancer is the most common malignancy among women globally, including India. Human breast cancer results from the genetic and environmental interaction. The present study attempts to understand the molecular heterogeneity of BRCA1 and BRCA2 genes, as well as to understand the association of various lifestyle and reproductive variables for the Breast Cancer risk. The study was conducted amongst 110 patients and 128 controls with total DNA sequencing of flanking and coding regions of BRCA1 BRCA2 genes that revealed ten Single Nucleotide Polymorphisms (SNPs) (6 novels). The controls selected for the study were age, sex and ethnic group matched. After written and informed consent biological samples were collected from the subjects. After detailed molecular analysis, significant ($p < 0.005$) molecular heterogeneity is revealed in terms of SNPs in BRCA1 (4 Exonic & 1 Intronic) and BRCA2 (2exonic and 3 Intronic) genes. The augmentation study investigated significant ($p < 0.05$) association with positive family history, early age at menarche, irregular menstrual periods, menopause, prolong contraceptive use, nulliparity, history of abortions, consumption of alcohol and smoking for breast cancer risk. To the best of authors knowledge, this study is the first of its kind, envisaged that the identification of the SNPs and modification of the lifestyle factors might aid to minimize the risk among the Bengalee Hindu females.

Keywords : breast cancer, BRCA, lifestyle, India

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