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Impact of X-linked tumour suppressor gene (FOXP3) variants on Indian breast cancer patients

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Breast cancer (BC), a malignant breast neoplasm is one of the leading causes of female deaths worldwide. The incidence of BC is constantly rising in developing countries like India (21%) with a 3 % annual increase. Breast cancer pathogenesis depends on various epidemiological factors; genetic susceptibility also plays an important role in its aetiology and progression. Forkhead box protein 3 (FOXP3) an X-linked transcription factor gene suggested to be down regulated in the mammary cancer tissues compared to normal breast epithelial cells. Several mutations in this gene have been reported in breast and prostate cancer. However, studies pertaining to the role of FOXP3 germ line mutations in the genetic susceptibility to breast cancer in Indian population are lacking. The present study was conducted on a total of 446 subjects of Indian origin that includes both BC patients and normal healthy controls. Genomic DNA was isolated from blood samples and genotyping was carried out for three selected functional polymorphisms of FOXP3 gene (rs3761548, rs3761549, rs2294021) using PCR-RFLP method. Of the three SNPs the genotype distribution of rs3761548 differed between patients and controls (CA vs. CC+AA: OR: 1.72 (1.12-2.60); $p=0.01$). Stratified data of individual SNPs showed significant influence of rs2294021 and rs3761548 on age at onset as well as the disease progression. Further, haplotypes of these two SNPs exhibited significant impact on age at onset and genetic predisposition towards the disease. In conclusion, the selected functional polymorphisms of FOXP3 that are coupled with low gene expression sway various aspects of breast cancer in Indian population.

Biography

Parveen Jahan is presently working as Associate Professor at Maulana Azad National Urdu University, obtained PhD from Osmania University (OU), India. She has 30 years of teaching and research experience with 55 research publications and 92 research presentations to her credit. She is editorial member and reviewer for reputed journals, organized workshops and seminars, and visited several places. She is Principle investigator for Government funded (DST, ICMR, UGC, APCOST) research projects. Doctoral and Post-doctoral students are pursuing their research under her supervision in the area of human molecular genetics with a focus on women related disorders. She held responsibilities as Chairperson BOS for Biotechnology and placement coordinator at OU.

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