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Germ-line mutanome profiling of the Breast cancers in Pakistani population

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Statement of the Problem: Cancers are complex disorders. Consanguineous populations, by virtue of autozygosity, provide a unique model for the investigations of the underlying genetic component(s). A prototype example in the breast cancers is the identification of BRCA1/2 gene defects in Ashkenazi Jews. In Pakistan, the age-standardized rate (ASR) of the incidence of breast cancers in females is among the highest in Asia, whereas the mortality rate is one of the highest in the world. With the consanguinity rate of 56.4% and inbreeding coefficient (F) of 0.0331, it is extremely important to investigate the role of inherited mutations in breast cancers in Pakistani population.

Methodology and Theoretical Orientation: BROCA analysis for breast cancers consists of twenty-seven (27) established and candidate breast cancer genes involved in molecular carcinogenesis. A pilot hospital-based cohort study was designed. Eighty-five breast cancer patients and three controls with no medical history of any cancer participated in the study. The BROCA investigations were carried out by a genomic capture, massively parallel next-generation sequencing assay on Illumina HiSeq2000 assay with 100bp read lengths. Copy number variations were determined by partially-mapped read algorithm. Once the mutation was identified, it was validated by Sanger sequencing. After informed consent, the mutations were screened in the familial samples.

Findings: The analysis revealed germ-line mutations in 12% of the patients. These mutations were restricted to three genes (BRCA 1, BRCA 2, and TP53). The identified mutations consist of both novel and previously reported alterations and result in protein truncation. No mutations were identified in the remaining twenty-four (24) genes. Mutation screening in the familial samples identified carriers in four out of five families.

Conclusions and Significance: The study provides a framework for the development of preventive and treatment strategies against breast cancers in Pakistani population.

Biography

Sadia Ajaz is currently involved in molecular profiling of breast cancers in local population. The translational medicine approach integrates molecular epidemiology of breast cancers with molecular pathology and molecular cancer therapeutics. The research studies are designed according to epidemiological principles and investigate the applications of molecular discoveries in prevention and management of cancers in general and breast cancers in particular. She has eleven publications so far, including seven publications in the field of molecular oncology in peer-reviewed international journals. Her research experience includes molecular oncology projects at prestigious national and international institutions. Area(s) of Research: Biochemistry, Human Genetics and Molecular Medicine.

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