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Status and importance of decoding breast cancer genetics in khyber pakhtunkhwa population, Pakistan

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Breast cancer is the result of cumulative mutations within oncogenes and tumor suppressor genes that result in the clonal development of progressively malignant cells. Traditionally, breast cancer was categorized by the presence or absence of estrogen receptor-alpha (ER□), progesterone receptor (PR), and human epidermal growth factor receptor (EGFR/HER2). Unfortunately, mutations in the DNA repair genes responsible for maintaining genomic stability such as BRCA1, BRCA2 and TP53 have not been included in such molecular categorization. Although it is widely accepted that mutations in DNA repair genes are causally linked to the onset of breast cancer development, and are more prevalent in triple negative breast cancer, as defined by the absence of ER□, PR, and HER2, and are thus more difficult to treat clinically.

Large scale population studies including GWAS, have primarily focused on the genetic predispositions associated with chronic disease amongst the Caucasian population. However, amongst all developing countries Pakistan has one of the highest rates of breast cancer, accounting for approximately 38% of total cancer diagnoses within the country. With the advent of more effective sequencing procedures, the ability to assess cancer risk and genetic diversity at a subpopulation level has allowed clinical researchers to develop innovative and personalized treatment protocols specific to their communities. With respect to breast cancer, a continuing need in developing countries is to elucidate the genetic predispositions that associate with cancer susceptibility, onset, development, progression, relapse, and drug response. Subsequently, in Pakistan there has been an enormous effort to collect genomic data from various tribal populations within the country in order to develop epidemiologic risk factors linked to breast cancer development. In this study, identification of known genetic risk alleles for breast cancer such as BRCA1, BRCA2, and TP53 was performed solely in patients located in the Khyber Pakhtunkhwa region of Pakistan, so as to better understand the prevalence and genetic risk factors associated with breast cancer development in those of Pathan or Pashtun ancestry.

Biography

This is Najeeb from Pakistan. I have a Ph.D. in Biochemistry and Molecular Biology. Recently, I am working on human genetic variability and susceptibility to cancer. My focus is to find the most associated gene polymorphism with cancer risk in our region and check the potential of that particular gene polymorphism as a biomarker for disease monitoring especially early diagnosis.