Novel mutations in the *BRCA2* gene in Yemeni women with breast cancer

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Abstract

Background: Breast cancer is a major cause of death among women worldwide. It is the most commonly diagnosed cancer among women in Yemen. In 2011, an average of breast incidence rate in Yemen was 20.9(1261). Among many etiological factors of BC, mutations in BRCA2 gene were found to be the primary cause in 5–10% of cases worldwide. The etiology of (BC) in Yemen is scarcely investigated. The study aims to describe the pattern of mutations including single nucleotide polymorphisms (SNP) and variants of the *BRCA2 gene* (exon11) among Yemeni women patients diagnosed with BC.

Methods: In this study, the DNA was extracted from tissue blocks of the patients who attended the Oncology Center-Sanaa City and undergone breast biopsy, from May 2015 to May 2016.One hundred and fifty suspected patients were enrolled in this study; one hundred samples were breast cancer and fifty samples were benign lesions of the breast which were used as controls. Polymerase chain Reaction (PCR) using primers that target regions from 3281 to 3731 (A) and 4967 to 5673 (B) of *BRCA 2 gene* (exon 11), and Sanger sequencing were

performed for all samples. The study was approved by Ethics Review Committee Board of Al-Neelain University.

Results and Discussion: Of the 150 suspected cases, 100 samples were cancer, while fifty were benign lesions of the breast. The age of the cancer patients ranges from 22 to 75 years, the median age was 46.2 years. Among breast cancer patients, nineteen novel mutations were detected in *BRCA2 gene* within two regions in selected positions A & B, while no variants was detected in all fifty benign lesions of the breast. Seventeen out of the nineteen novel mutations were pathological, while (2/19) were silent mutations. There is significant difference in the frequency of the detected mutations between cancer patients and controls. Mutations in *BRCA2 gene* are associated with 5-10 % of all cases of BC worldwide. The results revealed higher frequency than those reported figures.

Conclusion: the study concluded that there are novel mutations in *BRCA2* gene which are strongly associated with *BC* in Yemini patients. This association could be used as an effective tool for screening and gene therapy.

Keywords: *BRCA2 gene,* pathological mutations, novel mutation, silent mutations, Breast cancer,