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Master Thesis Defense

Entitled

PREVALENCE OF BRCA1 AND BRCA2 MUTATIONS AMONG BREAST AND OVARIAN CANCER PATIENTS IN NORTHERN EMIRATES

by

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Abstract

Breast cancer is the most common cancer and the second most cause of death among women. Mutations in BRCA1 and BRCA2 genes confer high susceptibility to both breast and ovarian cancer. However, data on the prevalence of the *BRCA1/2* mutations are scare. The genetic component of breast cancer in UAE is largely unknown and no study has evaluated the *BRCA* mutations status in breast and ovarian cancer patients in UAE population. This retrospective study aimed to establish mutation frequencies of the BRCA genes in breast and ovarian cancer patients from Northern Emirates and sought to examine potential association of *BRCA* carriers and Triple- Negative Breast Cancer (TNBC). The study population included patients who underwent *BRCA* genetic testing at Sheikh Khalifa Specialty Hospital (SKSH) to determine hereditary breast/ovarian cancer. Mutations in BRCA1 and BRCA2 were analyzed by Sanger sequencing or next generation sequencing (NGS) along with multiple ligation probe amplification (MLPA).

Of total 626 patients, 224 (85.5%) had no mutation. *BRCA* mutations were identified in 38 patients (14.5%). *BRCA1* and *BRCA2* mutations were detected in 6.8% and 7.6% of the patients, respectively. Variants of unknown significance in *BRCA1*was found in 0.4% of patients (one patient). TNBC accounted for 22% of all patients with breast cancer (BC) who underwent immunohistochemistry (28/127). Importantly, one novel *BRCA1* mutation: c. (80+1_81-1) _ (441+1_442-1) dup in exons 3,5,6,7 was observed in one patient with ovarian cancer who showed positive family history and age ≤ 45. Moreover, two novel deletion mutations were identified in the *BRCA2* gene. One deletion in exon 5 in ovarian cancer patient who showed positive family history and age ≤ 45. The second was a deletion in exons 10-13 observed in male patient with breast cancer with age ≤ 45. Our results in this study will help to establish the spectra of *BRCA* mutations and risks associated with breast and ovarian cancer in UAE patients.

Keywords: UAE, Hereditary, Breast cancer, Ovarian cancer, *BRCA1*, *BRCA2*, MLPA, NGS, TNBC.