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Search for new genetic mutations for breast cancer among high risk groups in Jordanian population

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Introduction: Breast cancer is the most common cancer in females. Worldwide more than 1 million new cases are diagnosed each year and nearly 600,000 women die from breast cancer each year. Breast cancer contributes to 37.4% of female cancers in Jordan and the second cause of death among women after ischemic heart disease. 22.8% of mortality from cancer among women was due to breast cancer with 211 reported deaths. Hereditary predisposition is estimated to cause about 7% of all breast cancers. Of all familial breast cancers, about 50% of cases are due to mutations in the BRCA1 and BRCA2 genes and are associated with early-onset breast cancer. The above two genes were identified in Ashkenazi Jewish. Studying other close populations could help identifications of new genes. A study conducted in Jordan showed a high consanguineous mating rate of 63.7%. Therefore, in such country with a very high consanguinity rate, genetic studies could identify new mutations or new genes related to breast cancer.

Aim & Methodology: In this cross-sectional study, we aimed to evaluate the potential contribution and the frequency of mutations in the BCRA1, BCRA2 gene in selected Jordanian patients with breast and ovarian cancers and in their first degree family members. We also aimed to search for new mutations in selected Jordanian patients with breast or/and ovarian cancer. We used the NICE guidelines for strong family history of breast/ovarian cancers in the selection criteria.

Results: 205 patients and 200 controls were included in the study. The mean age of study participants was 44. 195 patients had breast cancer and 10 cases had ovarian cancer at primary site. 15 patients had bilateral cancer. Results showed that there were 35 patients with confirmed mutations for breast cancer and 41 patients with confirmed or possible mutations with prevalence of 17% for the first group and 20% for the second group. Regarding site of mutation, 7% had confirmed mutations in BRCA1 gene and 12% had confirmed mutations in BRCA2 gene. Six patients had possible new mutations in BRCA2 gene and needs to be studied further among families of the patients.

Discussion & Recommendations: There is an urgent need for a national program for familial cancer referral and management system. This could be achieved through establishing a familial breast and ovarian cancers service in the first stage and expanding this in the future for all cancers. Patients and family members should be seen for genetic testing, counseling and management. This will lead to reducing mortality and morbidity from breast cancer and could save millions from treatment of advanced breast cancer or ovarian cancer for these high risk groups.

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Controversies in screening breast cancer

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There is established consensus in breast health community that mammography is the mainstay imaging examination for screening breast cancer. However, there are varied national recommendations among stakeholders and major institutions in the topic of breast cancer screening. Lack of consensus in screening criteria includes risk stratification, age to initiate screening and the interval of screening. The differences in practice guidelines are mainly due to variation in design and interpretation of screening trials over the past decades. As debates for and against the use of screening mammography continue to escalate, both providers and patients are often confused and wrongly perceive the recommendations as directives. The purpose of this presentation is to review the current guidelines, analyze the reason for the controversies in screening mammography and shine light on the upcoming trends of future screening guidelines.

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