عنوان مقاله:

Contribution of BRCA1 and BRCA2 Germ Line Mutations to Triple Negative Breast Cancer Patients

محل انتشار:

اولین سمپوزیوم بین المللی سرطان نسترن (سال: 1394)

تعداد صفحات اصل مقاله: 1

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خلاصه مقاله:

Objectives: Women harboring BRCA1/2 mutations have high lifetime risks of developing breast and ovarian cancer. Current criteria for BRCA1/2 testing to identify individuals at-risk rely predominantly on family history of breast/ovarian cancer and early age of disease onset. Here we investigated if the triple-negative breast cancer (TNBC) phenotype is pertinent in identifying candidates for BRCA1/2 testing in Pakistan. Methods: Five hundred and twenty-three breast cancer patients including 237 diagnosed with early-onset disease (<30 years of age) and 286 with a positive family history of breast/ovarian cancer were enrolled at the SKMCH & RC from June 2001 to February 2014. All patients underwent comprehensive BRCA1/2 testing for small-range mutations and large genomic rearrangements. Results: Of the breast cancer patients, 36.7% (192/523) presented with TNBC. BRCA1/2 mutation screening revealed 73 mutations in this group: 71 (97.3%) in BRCA1 and two (2.7%) in BRCA2. The prevalence of BRCA1 mutations was 37.0% (71/192), which was significantly higher compared to that of non-TNBC cases (10.3% (34/331)) (P < 0.0001). In the subgroups of early-onset and familial breast/ovarian cancer patients BRCA1 mutations were also more common in TNBC than non-TNBC patients (14.5% (13/90) vs. 5.5% (8/147), P=0.03) and (56.9% (58/102) vs.14.1% (26/184), P < 0.0001), respectively. TNBC patients harboring BRCA1 mutations (n=71) were diagnosed at later age than non-carriers (n=119) (p=0.002). Conclusion: The high prevalence of BRCA1 mutations in Pakistani TNBC patients with early- onset disease regardless of a family history of breast/ovarian cancer and the predominance of .BRCA1 mutations suggest TNBC status to be included as a criterion for BRCA1 testing in Pakistan

کلمات کلیدی: