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Prevalence of BRCA 1 and BRCA2 in breast cancer patients in India

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Background: Breast cancer is the most common cancer affecting women all over the world. In India, it is the second most common solid-organ malignancy after carcinoma cervix. Worldwide, approximately 5%–10% of the cases have been associated with various germline mutations. However, in India, studies correlating germline BRCA mutation with various clinicopathological variables are rare. In this study, we have tried to find out the difference in BRCA-mutated and BRCA-nonmutated patients.

Materials and Methods: From May 2015 to May 2017, 50 patients with carcinoma breast were subjected to BRCA mutational studies by next-generation sequencing and further subdivided into BRCA-mutated and BRCA-nonmutated subgroups. Their clinical, pathological, and response to primary treatment were recorded and compared between two subgroups. They were followed up for a minimum of 9 months, and response to treatment was also recorded.

Results: Out of 50 patients with carcinoma of the breast, only six patients were detected to be mutated and pathological mutations were detected in two (4%) patients only. All the BRCA-positive patients were female only. The most common age of presentation was >50 years while BRCA-positive patients presented earlier. Triple-negative breast cancer (TNBC) was the most common presentation and most patients presented in Stage III.

Conclusion: Germline mutations in carcinoma breast can account for around 5%–10% of total breast cancers all over the world, but in our study, we have reported that 4% of the patients had BRCA mutations. BRCA-mutated carcinoma breast presents at a younger age and more frequently with the bilateral presentation as compared to BRCA-negative disease. BRCA-mutated carcinoma breast presents with more advanced disease and usually has a significant family history of either first- or second-degree relatives being affected. Overall TNBC status was more commonly found in both subsets of the patients. Overall BRCA-positive disease had a more aggressive course of the illness as compared to BRCA-negative patients

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