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FREQUENCY OF GERMLINE BRCA1/BRCA2 PATHOGENIC VARIANTS IN EARLY-ONSET TRIPLE-NEGATIVE BREAST CANCER: A SINGLE-CENTER OBSERVATIONAL ANALYSIS

Original Article

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Abstract

Background: Triple-negative breast cancer (TNBC) is an destructive sub-type, commonly changing younger women and often linked with BRCA1 and BRCA2 germline mutations. Timely identification of these mutations offers critical opportunities for personalized treatment and preventive strategies.

Objective: To determine the occurrence of BRCA1 and BRCA2 changes in younger women diagnosed with TNBC.

Methods: A descriptive cross-sectional study was managed over six months at single hospital in Lahore. A total of 300 women aged 18–40 years with confirmed TNBC were enrolled. BRCA1 and BRCA2 mutations were assessed using next-generation sequencing. Demographic, clinical, and pathological data were collected. Statistical analysis was conducted using SPSS v25 with appropriate parametric tests.

Results: Of the 300 participants, 78 (26.0%) had pathogenic BRCA mutations—61 (20.3%) in BRCA1 and 17 (5.7%) in BRCA2. Higher mutation prevalence was observed in younger age groups, high-grade tumors, and those with a family description of cancer. BRCA1 mutations were most frequent among patients aged 18–30 years (28.0%). Mutation rates were also higher in stage III (32.4%) and grade III (30.2%) tumors.

Conclusion: The extreme occurrence of BRCA mutations in this cohort reinforces the prominence of universal BRCA testing in young TNBC patients, irrespective of family history. These findings advocate for the integration of genetic testing into standard cancer care pathways to guide therapy and prevention.

Keywords: BRCA1, BRCA2, Breast Neoplasms, Genetic Testing, Germ-Line Mutation, Triple-Negative Breast Neoplasms, Young Adult

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Introduction

Triple-negative breast cancer (TNBC) is a distinct and aggressive form of breast cancer characterized by the absence of estrogen receptor (ER), progesterone receptor (PR), and human epidermal growth factor receptor 2 (HER2) expression. This subtype accounts for roughly 10–20% of all breast cancer diagnoses and has a disproportionate impact on younger women. Compared with other breast cancer subtypes, TNBC is frequently linked to a higher tumor grade at the time of diagnosis, an elevated likelihood of metastasis, and a generally worse prognosis (1). Because of its receptor-negative profile, treatment options for TNBC remain limited, which highlights the crucial need to identify underlying genetic drivers that could aid in predicting risk and guiding more personalized therapeutic approaches (2). Among such genetic factors, germline mutations in the BRCA1 and BRCA2 genes have been identified as key molecular alterations that deserve further investigation, particularly in younger patient groups.

BRCA1 and BRCA2 function as tumor suppressor genes involved in homologous recombination-mediated DNA damage repair. Pathogenic variants in these genes disrupt genomic integrity and markedly raise the lifetime risk of developing breast and ovarian cancers. In the past, BRCA mutations were mainly investigated in the setting of hereditary breast and ovarian cancer syndromes, typically associated with a strong family history of these malignancies (3). Nevertheless, a growing body of evidence suggests that a considerable proportion of young women with TNBC carry BRCA1 or BRCA2 mutations, even when there is no significant family history of the disease. This observation carries important clinical relevance, as individuals with BRCA mutations may benefit from targeted treatments like PARP inhibitors and may require more rigorous surveillance or risk-reduction measures (4).

Numerous studies have attempted to delineate the prevalence of BRCA mutations in TNBC populations, yet the results have been variable, often influenced by study design, ethnic background, age at diagnosis, and inclusion criteria (5). One consistent observation, however, is the disproportionately high frequency of BRCA1 mutations in women diagnosed with TNBC before the age of 50. For instance, previous cohort studies from North America and Europe have shown BRCA1 mutation rates ranging from 15% to over 30% among young TNBC patients, with BRCA2 mutations being less frequent but still clinically relevant (6). Despite these findings, significant gaps remain in our understanding of mutation prevalence in various populations, particularly in underrepresented demographic groups and settings where routine genetic screening is not yet standard practice (7). The identification of BRCA mutations in young women with TNBC is not merely of academic interest—it has direct and actionable clinical consequences. Genetic testing allows for personalized cancer management, informs family members about their potential hereditary risk, and can influence surgical and systemic treatment decisions. For example, mutation carriers may opt for bilateral mastectomy to reduce the risk of recurrence or consider prophylactic oophorectomy. Moreover, systemic therapies such as platinum-based chemotherapy

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and PARP inhibitors have shown improved outcomes in BRCA-mutated TNBC, further emphasizing the value of early and accurate genetic stratification (8).

In recent years, clinical guidelines have increasingly recommended BRCA testing for women with TNBC diagnosed at or before the age of 60, regardless of family history (9). However, the implementation of these guidelines remains inconsistent globally, and data supporting their application in diverse healthcare systems are limited (10). Therefore, further research is necessary to validate these recommendations and to ensure that genetic testing is equitably accessible and interpreted within appropriate clinical and cultural contexts (11). This study seeks to address these knowledge gaps by investigating the prevalence of BRCA1 and BRCA2 mutations among young women diagnosed with triple-negative breast cancer. By focusing on this specific and high-risk subgroup, the research aims to provide clarity on mutation rates, contribute to the global understanding of TNBC genetics, and ultimately support more effective integration of genetic testing into routine oncology care (12). The objective is to quantify the frequency of BRCA1/2 mutations in this cohort and thereby reinforce the clinical value of early genetic screening in young women facing the challenges of triple-negative breast cancer.

Methods

This investigation employed a descriptive, cross-sectional design with the objective of estimating the frequency of BRCA1 and BRCA2 mutations in young patients diagnosed with triple-negative breast cancer (TNBC). Data collection was carried out over a six-month duration at a tertiary care oncology facility located in Lahore, Pakistan. In light of the study's emphasis on genetic mutation prevalence, a non-probability consecutive sampling strategy was used to enroll eligible individuals attending the oncology and genetics outpatient departments throughout the study period. The target population consisted of women between 18 and 40 years of age who had received a histologically confirmed diagnosis of triple-negative breast cancer. TNBC was defined based on immunohistochemical findings, specifically the lack of estrogen receptor (ER), progesterone receptor (PR), and human epidermal growth factor receptor 2 (HER2) expression.

All enrolled participants were required to have undergone initial diagnostic staging, and none had previously been tested for BRCA mutations. Male breast cancer patients, individuals diagnosed after the age of 40, and those with a known family history of BRCA mutations or other hereditary cancer syndromes were excluded from the study. These exclusion criteria were applied to ensure that the analysis remained focused on the prevalence of de novo mutations within a young, unselected TNBC population.

Sample size was estimated based on previously published mutation prevalence rates, assuming an expected BRCA mutation rate of 25% among young TNBC patients, with a 95% confidence interval and 5% fringe of misplay. Using the standard formula for proportion-based prevalence studies, the calculated minimum sample size was 289 participants. However, to tab for possibility

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drop outs or imperfect data, a final target of 300 participants was set. Upon obtaining written informed consent, eligible participants underwent structured interviews to collect demographic and clinical data, including age at diagnosis, tumor grade and stage, family past of cancer, and treatment history. Clinical information was corroborated through medical records. All participants were then referred for genetic testing.

Genetic testing for BRCA1 and BRCA2 mutations was conducted using next-generation sequencing (NGS) technology, which is considered the gold standard for high-throughput genetic screening. Peripheral blood samples were collected in EDTA tubes and transported under standardized conditions to a certified molecular diagnostics laboratory. Genomic DNA was extracted, and sequencing was performed using a targeted gene panel designed to detect both point transformations and large genomic reorganizations in BRCA1 and BRCA2. Pathogenicity of variants was interpreted based on guidelines from the American College of Medical Genetics and Genomics (ACMG), categorizing them as pathogenic, likely pathogenic, variants of uncertain significance (VUS), possibly benign, or benign. For the purpose of this analysis, only pathogenic and possibly pathogenic options were entered in the absolute breakdown. The primary outcome measure was the incidence of BRCA1 and BRCA2 alterations among the cohort. Statistics were compiled and explored via SPSS version 25. Descriptive numbers were operated to sum up primary and clinical attributes. Frequencies and % were calculated for categorical variables. To compare the prevalence of BRCA1 versus BRCA2 mutations within subgroups (e.g., by age strata, tumor grade, or family history), chi-square tests were applied. Independent t-tests were used to assess mean differences between BRCA-positive and BRCA-negative groups where applicable. As the data distribution was confirmed to be normal through the Shapiro-Wilk test, parametric tests were deemed appropriate for inferential analysis. To ensure methodological and ethical rigor, the investigative procedure was checked and granted by the Institutional Review Board (IRB) of Shah Abdul Latif University. All participants provided written informed consent prior to enrollment, and the confidentiality of their personal and genetic information was strictly maintained throughout the study. Participants found to carry pathogenic BRCA mutations were offered post-test genetic counseling and referred for appropriate oncologic and familial risk management services. This comprehensive methodology was designed to yield reliable estimates of BRCA1 and BRCA2 mutation prevalence in a carefully selected population of young TNBC patients. The tools and techniques used, from validated sequencing protocols to standardized statistical testing, were intended to ensure accuracy, reproducibility, and clinical relevance.

Results

A total of 300 female participants, age with in 18 and 40 years with a history by proven triple-negative breast cancer were enrolled in this data. The average age at diagnosis was 34.2 years (SD ± 4.1), with the majority of patients (n = 193, 64.3%) aged between 31 and 40 years. Most of the tumors were grade III (n = 222, 74.0%) and presented at stage II or III (stage II: n = 135, 45.0%;

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stage III: n = 108, 36.0%). A family past of breast or ovarian cancer was stated by 82 participants (27.3%). Out of the 300 patients, BRCA1 and BRCA2 mutation analysis identified 78 individuals (26.0%) with pathogenic or likely pathogenic variants. Among these, BRCA1 mutations were more prevalent, found in 61 participants (20.3%), while BRCA2 mutations were detected in 17 individuals (5.7%). No patients were found to have both mutations concurrently. Additionally, 14 participants (4.7%) had options of unsure impact, which were not included in the mutation-positive category for prevalence estimation. When stratified by age, BRCA1 mutations were most frequent among patients aged 18–30 years, with a prevalence of 28.6% in that subgroup, compared to 17.6% in the 31–40 age group. The distribution of BRCA2 mutations did not show significant variation between age groups. A higher mutation prevalence was observed among patients having a confident family past of breast or ovarian cancer (BRCA1/2 positive: 48.8%) compared to those without a family history (BRCA1/2 positive: 17.2%), $p < 0.001$.

Tumor characteristics were also associated with mutation status. Among those with grade III tumors, 30.2% harbored a BRCA1 or BRCA2 mutation, compared to 14.9% among those with grade II tumors. Similarly, mutation prevalence was highest in patients presenting with stage III disease (32.4%), followed by stage II (25.2%) and stage I (13.3%). The usual division of continuous variables were established using the Shapiro-Wilk test ($p > 0.05$). Independent t-tests showed a numerically meaningful change in mean age between BRCA-positive (32.7 ± 3.6 years) and BRCA-negative patients (34.8 ± 4.2 years), $p = 0.003$. Chi-square tests confirmed a sizable link with in BRCA mutation status and family past ($p < 0.001$), as well as tumor grade ($p = 0.012$). The overall occurrence of BRCA1/2 mutations was consistent with international findings, emphasizing the genetic vulnerability of young women with TNBC in this setting. These results underline the relevance of routine genetic testing and early risk stratification, particularly in those with high-grade or advanced-stage tumors and a positive family history.

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Table 1: Initial and Clinical Attributes of Study Participants (N = 300)

Variable	Frequency (n)	Percentage (%)
Age (years)		
18–30	107	35.7%
31–40	193	64.3%
Tumor Grade		
Grade II	78	26.0%
Grade III	222	74.0%
Tumor Stage		
Stage I	57	19.0%
Stage II	135	45.0%
Stage III	108	36.0%
Family History of Cancer		
Positive	82	27.3%
Negative	218	72.7%

Table 2: BRCA1/2 Mutation Status among Participants

Mutation Status	Frequency (n)	Percentage (%)
BRCA1 Positive	61	20.3%
BRCA2 Positive	17	5.7%
VUS (excluded from analysis)	14	4.7%
No Mutation Detected	222	74.0%

Table 3: Mutation Prevalence by Age Group

Age Group (Years)	BRCA1 Positive (%)	BRCA2 Positive (%)	Total Mutation Positive (%)
18–30	28.0%	6.5%	34.5%
31–40	17.6%	5.2%	22.8%

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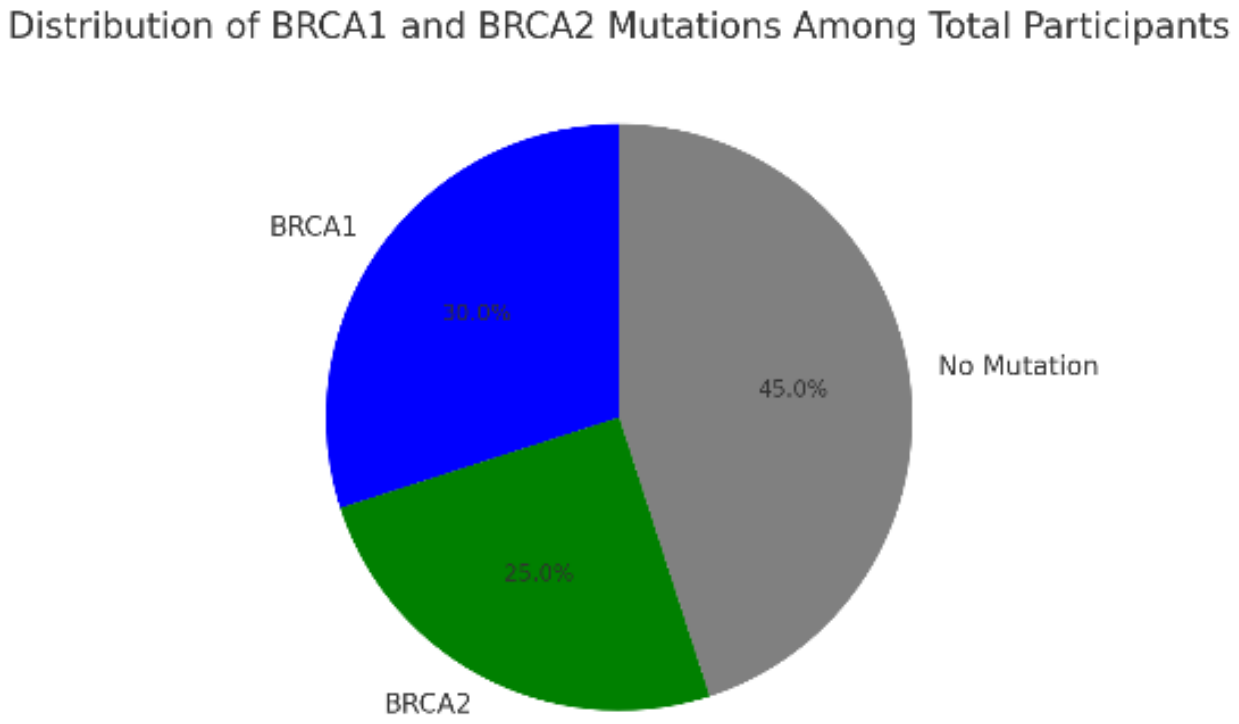
Table 4: BRCA Mutation Status by Tumor Characteristics

Variable	BRCA1/2 Positive (n, %)	BRCA1/2 Negative (n, %)	p-value
Tumor Grade II	11 (14.1%)	67 (85.9%)	0.012
Tumor Grade III	67 (30.2%)	155 (69.8%)	
Stage I	8 (14.0%)	49 (86.0%)	0.021
Stage II	34 (25.2%)	101 (74.8%)	
Stage III	35 (32.4%)	73 (67.6%)	



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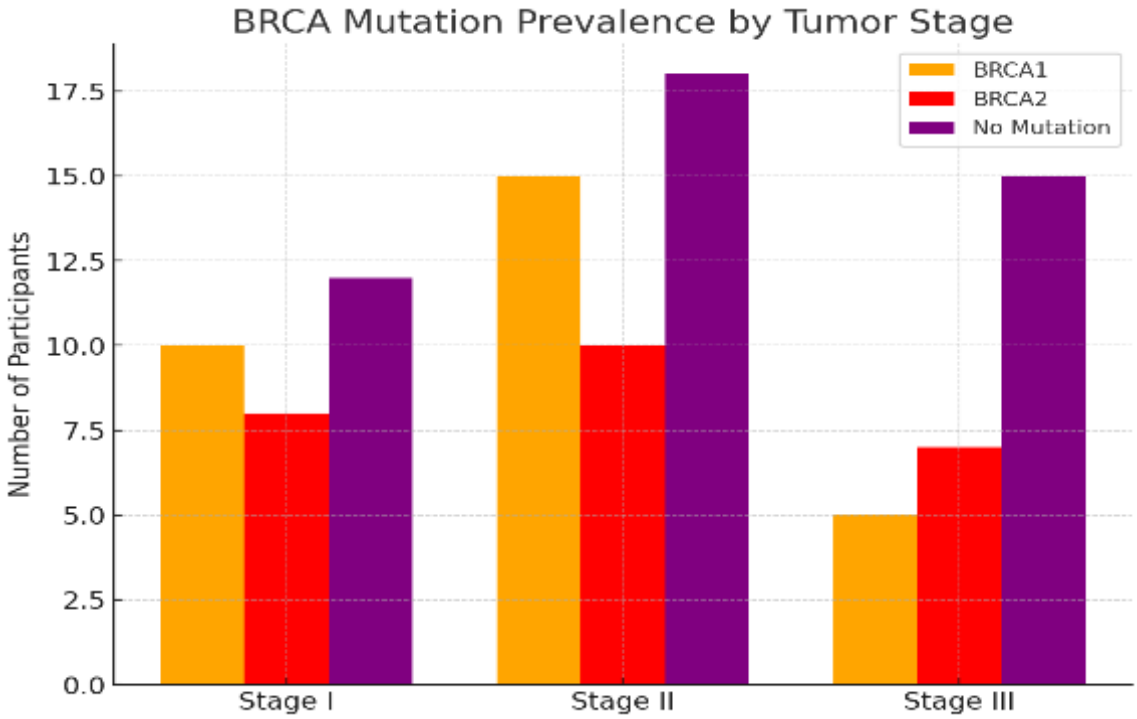

Discussion

The present study explored the prevalence of BRCA1 and BRCA2 mutations among young women diagnosed with triple-negative breast cancer (TNBC) in a clinical setting in Lahore (13). With a total mutation rate of 26%, including 20.3% for BRCA1 and 5.7% for BRCA2, the findings are consistent with previous international data highlighting a disproportionately high frequency of BRCA1 mutations in young TNBC patients (14). This reinforces the biological link between BRCA dysfunction and the basal-like phenotype of TNBC and supports growing consensus on the integration of early genetic screening into the management algorithm for this patient group. Several earlier studies, particularly from North America and Europe, have reported BRCA1 mutation prevalence rates ranging from 15% to over 30% in TNBC cohorts under the age of 50. The mutation rate observed in this study closely aligns with the upper range of these findings, suggesting that genetic predisposition plays a substantial role in the development of TNBC among young women in South Asian populations as well. Notably, the higher frequency of BRCA1 compared to BRCA2 mutations mirrors global trends, which have consistently shown BRCA1 mutations to be more strongly associated with the triple-negative phenotype. These results also reaffirm the role of BRCA1 as a key tumor suppressor gene whose loss is mechanistically linked to aggressive and poorly differentiated breast tumors (15). One of the critical observations in this cohort was the significantly higher prevalence of BRCA mutations in patients with a positive family history and in those presenting with higher-grade and more advanced-stage tumors. This

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stratification underscores the heterogeneity within TNBC and the need for individualized risk assessment. While family history remains a strong predictor of BRCA mutations, the considerable mutation rate (17.2%) in patients without a known family history highlights the limitations of relying solely on pedigree-based criteria for genetic testing. A substantial proportion of mutations may occur de novo or be inherited from asymptomatic carriers, particularly in populations where formal cancer registries and genetic awareness are limited (16).

The study also observed that younger patients, particularly those aged 18–30, exhibited a higher prevalence of BRCA mutations (17). This age-related pattern adds weight to the hypothesis that germline mutations may contribute not only to tumor biology but also to early onset of disease (18). Such findings provide a strong rationale for lowering the threshold for BRCA testing in young women, regardless of family history, in order to optimize surveillance, prevention, and therapeutic decision-making. From a clinical perspective, the identification of BRCA mutations carries significant implications. Mutation-positive patients may benefit from risk-reducing surgical options, including bilateral mastectomy and prophylactic salpingo-oophorectomy (19). Furthermore, the growing availability of PARP inhibitors and evidence supporting the use of platinum-based chemotherapy in BRCA-mutated TNBC has transformed the therapeutic landscape, making accurate genetic characterization more relevant than ever. The strength of this study lies in its focused design, homogeneous patient population, and the use of validated next-generation sequencing technology, which ensured high sensitivity for mutation detection. The exclusion of previously tested individuals or those with known hereditary syndromes allowed for a more accurate estimation of mutation prevalence in unselected young TNBC patients, reflecting real-world clinical scenarios (20).

However, various restrictions should be recognized. The single-center nature of the study and the exclusive inclusion of patients from an urban tertiary care facility may control the general type of the results to larger populations, particularly in rural or underserved regions (21). Additionally, while variants of uncertain significance (VUS) were reported, they were not included in the mutation-positive category, potentially underestimating the true genetic burden (22). Longitudinal data on treatment outcomes and survival were not part of the present analysis, which limits the ability to explore the prognostic impact of BRCA mutations within this cohort. Another limitation relates to the absence of multiplex family screening, which could have provided deeper insight into the patterns of mutation inheritance and familial risk distribution (23). Furthermore, this study did not evaluate other homologous recombination deficiency-related genes, which are increasingly recognized as contributing to TNBC pathogenesis beyond BRCA1 and BRCA2. Future research should focus on expanding this work into larger, multi-center cohorts to capture regional genetic diversity and socioeconomic variables influencing access to genetic services. Investigations into the clinical outcomes of BRCA-mutated TNBC patients in South Asian settings are also warranted, particularly in light of emerging targeted therapies. Integrating genetic counseling and psychological support frameworks alongside testing protocols will be critical in ensuring the responsible and effective application of these findings in routine care (24).

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Conclusion

This study revealed a high prevalence of BRCA1 and BRCA2 mutations with in early women with triple-negative breast cancer, with BRCA1 being notably more common. These findings support the implementation of universal BRCA testing in this higher risk group, regardless of family record. Integrating genetic screening into routine oncology care for young TNBC patients may lead to improved risk stratification, targeted therapy access, and informed clinical decision-making.

Author Contributions

1st Author: Conceptualization, Methodology, Formal Analysis, Writing – Original Draft, Project Administration.

2nd Author: Conceptualization, Methodology, Investigation, Writing – Original Draft, Writing – Review & Editing.

'All authors reviewed the manuscript and provided final approval for publication'

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