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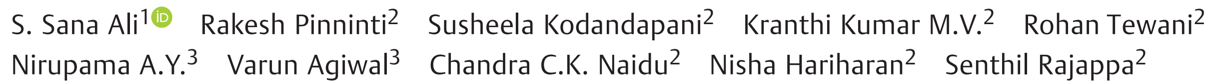

Testing Patterns and Prevalence of gBRCA Mutations among Women with Breast Cancer: A Cross-Sectional Observational Study

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Abstract

Introduction Pathogenic germline mutations in BRCA (*gBRCAm*) genes can heighten the risk of breast cancer (BC) among carriers. Economic constraints and patient testing hesitancy challenge adherence to hereditary germline testing guidelines. As a result, clinicians prioritize hereditary BC screening based on patient willingness, affordability, and therapeutic benefit.

Objectives The objectives of the study were (1) to identify the pattern of hereditary cancer germline testing among women diagnosed with BC and (2) to determine the prevalence of *gBRCAm* among the women with BC who underwent hereditary cancer germline testing.

Materials and Methods A retrospective study was conducted at a cancer hospital between October 2023 and January 2024. We aimed to assess the germline testing patterns of physicians in our hospital by examining the clinical profile of patients with BC who underwent hereditary cancer multigene (30 gene panel) mutation testing using next-generation sequencing between January 2021 and December 2023. A simultaneous analysis was performed with a multiplex ligation-dependent probe amplification to detect deletions and duplications in the *BRCA1* and *BRCA2* genes. The classification of the variants as pathogenic and variants of uncertain significance (VUS) was determined by the American College of Medical Genetics and Genomics guideline.

Results Of the 3,600 patients with BC during this study period, only 325 (9%) underwent germline testing. The testing patterns indicated that the median age of those tested was 48.4 years (standard deviation [SD]: 10.1; range: 20–77), 189 patients (58.2%) were younger than 50 years, and 103 patients (31.7%) had a family history of cancer. Family history of BC was reported in 95 (29.2%) patients. Bilateral BC was noted in 19 patients (5.8%), while ovarian cancer was reported in 9 (2.8%) patients. Triple-negative BC (TNBC), hormone receptor–positive BC, and HER2-positive BC were reported in 52, 42.8, and 17.2% patients, respectively. Pathogenic/likely pathogenic

Keywords
- breast cancer
- germline mutation
- genetic testing
- prevalence
- physician practice patterns
- India

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(P/LP) germline *BRCA* mutations were detected in 48 (14.7%) patients (*BRCA1* in 29/325 [8.9%] patients and *BRCA2* in 19/325 [5.8%] patients). The highest prevalence was seen among TNBC (36/169, 21.3%) patients. P/LP *gBRCAm* prevalence among those with and without notable family history was 27/103 (26.2%) and 21/222 (9.5%), respectively; age less than 50 years and greater than 51 years was noted in 32/189 patients (16.9%) and 16/136 (11.8%) patients, respectively. VUS was noted in 29 patients (*BRCA1* in 4 patients [8.9%] and *BRCA2* in 25 patients).

Conclusions Measures to ensure equitable access to genetic testing can improve testing rates and enhance patient outcomes through personalized care.

Introduction

Breast cancer (BC) is the most common cancer among women in India, with new cases increasing by 39% from 2010 to 2016.[1,2] In 2019, the Global Burden of Disease (GBD) reported a BC prevalence of 163.3 per 100,000 in India.[3] Among the nonmodifiable risk factors, age, ethnicity, high-risk family history, and genetic susceptibility are critical determinants that significantly contribute to overall susceptibility and likelihood of developing BC.

Germline mutations in the BRCA (*gBRCAm*) gene give rise to *BRCA1* and *BRCA2* variants, heightening the risk of developing BC among their carriers.[4] It has been estimated that approximately 5 to 10% of all BC cases are familial, with germline mutations in the BRCA1 or BRCA2 genes accounting for 15 to 20% of the observed risk.[5] These mutations are more sensitive to certain therapies, necessitating tailored interventions with poly-ADP ribose polymerase (PARP) inhibitors.[6]

The prevalence of *gBRCAm* varies by age, family history of BC or ovarian cancer, and BC types: hormone receptor–positive BC (HR+), triple-negative BC (TNBC), and human epidermal growth factor receptor 2 positive (HER2+). The National Comprehensive Cancer Network (NCCN) broadly endorses testing for women diagnosed with BC based on epidemiological, clinical, and biological factors as well as indications for the use of PARP inhibitors.[7] The prevalence of pathogenic *BRCA1/2* and TNBC is higher in India, with the *BRCA1/2* mutation frequencies ranging from 2.9 to 24% among Indian familial patients with BC.[1,8] Additionally, genetic, ethnic, and cultural diversity within India complicates accurate representation of the *gBRCAm* burden in BC.[9,10] Economic constraints in testing, stigma, and the psychological burden experienced by the patients further challenge adherence to testing criteria such as the NCCN guidelines.[11,12] As a result, clinicians prioritize hereditary BC screening based on patient willingness, affordability, and therapeutic benefit.

The objectives of this study are the following:

- To identify the pattern of hereditary cancer germline testing among women diagnosed with BC.
- To determine the prevalence of gBRCA mutation among women with BC who underwent hereditary cancer germline testing.

Materials and Methods

A retrospective study was conducted at the Basavatarakam Indo-American Cancer Hospital & Research Institute (BIACHRI) between October 2023 and January 2024.

Inclusion Criteria

The study population included all women who had been diagnosed with BC and had undergone testing for germline *BRCA1/BRCA2* mutation between January 2021 and December 2023.

Exclusion Criteria

Male patients with BC were excluded.

The data were obtained from the electronic medical records and laboratory records. The collected data included the following: age at the time of diagnosis, family history of any type of cancer and clinical characteristics including type of *BRCA* mutation (*BRCA1* or *BRCA2*); variant category (pathogenic, likely pathogenic, variant of uncertain significance [VUS]); metastatic status; status of estrogen receptor (ER); progesterone receptor (PR); and HER2 status.

Primary and Secondary Outcomes

The primary outcome of the study was the prevalence of *gBRCAm* among women with BC between October 2023 and January 2024. The secondary outcome was to describe the patterns of germline hereditary breast and ovarian cancer (HBOC) testing by analyzing the descriptive characteristics of the patients who were tested. A notable family history, as per the NCCN criteria, was defined as BC at any age and $\geq$1 close blood relative (first-, second-, or third-degree relative) with BC, male BC, ovarian cancer, pancreatic cancer, and prostate cancer.

Statistical Analysis

Data were entered in Microsoft Excel and analyzed using Stata Statistical Software (Release 14, StataCorp LP, College Station, TX, United States). The prevalence of *gBRCAm* was expressed as a percentage with a 95% confidence interval (CI), calculated using the binomial exact test. The analysis was shown for age in mean with standard deviation (SD) and family history of the study population, and the description of *gBRCAm* according to the clinical parameters was expressed in frequency and percentage. Cases with *gBRCAm* were

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analyzed using the chi-squared test. A *p*-value of less than 0.05 was considered statistically significant.

Ethical Approval

Ethical approval was obtained from the institutional ethics committee at Basavatarakam Indo-American Cancer Hospital & Research Institute (BIACHRI; IEC/2023/281) on October 30, 2023. All data were anonymized and the study was conducted in accordance with the ethical standards of the institute and the Helsinki Declaration of 1964 and its later amendments.

Testing Methods

Hereditary cancer multigene mutation testing was done using next-generation sequencing (NGS) at BIACHRI. A simultaneous analysis was performed with multiplex ligation-dependent probe amplification for *BRCA1* and *BRCA2*, deletions, and duplications. The genomic DNA was extracted from the peripheral blood samples using the QIAamp DNA Mini Kit (QIAGEN, Victoria, Australia) according to the manufacturer's instructions.[13] The DNA quality was confirmed using a Qubit dsDNA HS Assay kit (Life Technologies) on a Qubit4.0 Fluorometer (Life Technologies).[14]

Next-Generation Sequencing

NGS was performed in the following steps. The library preparation was performed using the Oncomine *BRCA* Research Assay and was sequenced using an Ion GeneStudio S5 Plus Platform (Life Technologies).[15] The resulting sequence reads were initially analyzed for variant detection using the Ion Torrent Variant Caller (available at the Life Technology Torrent Browser Plugin store by aligning to the human genome reference [hg19]).[16] Visual confirmation of the identified variants was accomplished with Integrative Genomics Viewer software from the Broad Institute (Cambridge, MA, United States).[17] Finally, the variants (related to the phenotype) were scored and reported according to the American College of Medical Genetics and Genomics (ACMG) standards and guidelines for sequence variant interpretation.[18]

Multiplex Ligation-Dependent PCR Amplification

Multiplex ligation-dependent PCR amplification (MLPA) was performed using the following kits: P002_BRCA1 for the BRCA1 gene and P045_BRCA2 for the BRCA2 gene (MRC Holland, the Netherlands).[19] An optimized 100 ng of input DNA was utilized for the MLPA reaction and the process was performed at the applied condition for initial denaturation of the sample DNA; a mixture of MLPA probes (BRCA1 and BRCA2 probes) was added separately to the sample. In general, each MLPA probe consists of two oligonucleotides as instructed and designed by the manufacturer. The fragment analysis was performed on the ABI PRISM 3730XL Genetic Analyzer (Applied Biosystems, United States) using LIZ 500 (Applied Biosystems) as a standard size.[20] For statistical analysis, the MLPA ratios (dosage quotient) of below 0.7 or above 1.3 are indicative of a deletion (copy number change from 2 to 1) or duplication (copy number change from 2 to 3), respectively. A dosage quotient of 0.0 indicates a homozygous deletion; a dosage quotient of 0.35 to 0.65 indicates heterozygous deletion, a dosage quotient of 1.35 to 1.55 indicates heterozygous duplication, and a dosage quotient of 1.7 to 2.2 indicates homozygous duplication.

Results

Of the cohort of 3,600 patients with BC that presented to our hospital between January 2021 and December 2023, 325 patients (9%) underwent germline multigene (30 gene panel) mutation testing using NGS for germline BC. Pre- and post-test genetic counseling was provided to all the patients by the consulting oncologist.

Characteristics of Patients Tested

The mean age of the cohort was 48.4 years (SD: 10.1; range: 20–77; ►**Table 1**). A notable family history was present in 103 (31.7%) of the patients tested, including 95 (29.2%) with a family history of BC. Other cancers were reported in 21 patients (6.4%). With regard to molecular subtypes of BC, TNBC, HR +, and HER2+ were reported in 169 (52%), 139 (42.8%), and 56 (17.2%) patients, respectively.

Prevalence of P/LP gBRCA1/2m

Of the patients tested, pathogenic/likely pathogenic (P/LP) gBRCA1/2m were detected in 48/325 (14.7%) patients (►**Table 2**). The afflicted genes were discovered as *BRCA1* in 29/325 patients (8.9%; $p = 0.001$), while 19 patients were found to have BC of *BRCA2* origin (19/325; 5.8%; $p = 0.001$). The majority of P/LP gBRCA1/2m were in patients with TNBC (26/29; 89.7%; $p < 0.001$). The variants of VUS were detected in 29 patients (8.9%); 4 (1.2%) in *BRCA*1 and 25 (7.7%) in *BRCA*2. The P/LP non-BRCA germline mutations detected were two each in RAD50 and MUTYH, and one in MRE1.

Table 1 Patient characteristics

Parameters	Estimate
Age (y)[a]	48.4 (10.1)
Comorbidities	123 (37.8)
Stage 4	64 (19.7)
Bilateral breast cancer	19 (5.8)
Significant family history[b]	103 (31.7)
Other cancers (excluding bilateral breast cancer)	
Contralateral breast	6 (1.8)
Carcinoma ovary	9 (2.8)
Thyroid cancer	3 (0.9)
AML	1 (0.3)
CML	1 (0.3)
Lung cancer	1 (0.3)

Abbreviations: AML, acute myeloid leukemia; CML, chronic myeloid leukemia.
[a]Mean (standard deviation).
[b]Following NCCN criteria: Breast cancer (BC) at any age and ≥1 close blood relative (first-, second-, or third-degree relative) with BC, male BC, ovarian cancer, pancreatic cancer, and prostate cancer.

Table 2 BRCA1 and BRCA2

Variants	*n* (%)	BRCA mutation	
		BRCA1	BRCA2
Pathogenic/likely pathogenic	48 (14.7)	29 (8.9)	19 (5.8)
Variants of uncertain significance	29 (8.9)	4 (13.7)	25 (86.2)

Association of P/LP *gBRCAm* with Patient Characteristics

The P/LP gBRCA1/2m prevalence in patients with ER/PR positive, HER2 positive, and TNBC were 11/139 (7.9; 95% CI 4–13), 4/56 (7.1%; 95% CI 2.6–17) and 36/169 (21.3%; 95% CI 15.7–28), respectively (►**Table 3**). The *gBRCAm* prevalence in patients with a notable family history was 27/103 (26.2%; 95% CI 18.5–35.6). There was no significant association between age of onset and *gBRCAm*.

Discussion

The cross-sectional study aimed to determine the prevalence of *gBRCAm* among BC patients and identify the testing patterns for HBOC. We found a 14.7% prevalence of *gBRCAm* after testing less than 10% of the population over the 3-year study period. The testing patterns showed that patients who underwent HBOC testing typically had a notable family history of breast or ovarian cancers, were diagnosed at a young age, or had TNBC. Upon comparing our findings with other similar studies, 32 (16.9%) young women (<50 years) in our study had a *gBRCAm* compared with 10.3% of the patients with BC in a multicenter study.[21] Germline BRCAm was reported in 27 (26.2%) patients with notable family history, closely matching the 28% reported in the same study.[21] Additionally, *gBRCAm* was detected in 36 (21.3%) patients with TNBC, compared with a range of 14.8 to 62.5% patients with TNBC reported in other studies.[21,22] Applying stringent criteria for HBOC testing was expected to select a BRCA-enriched population and thus reveal a higher positivity rate. Indian studies report a *gBRCAm* prevalence of 18 to 23% in patients with BC meeting the NCCN criteria[22–24] and 8 to 21% in unselected cohorts.[21,23,25,26] In our study, selecting patients with a high likelihood of a positive result yielded a detection rate comparable to that achieved by following the NCCN criteria or testing all consecutive patients with BC. Our perspective is that given the high pretest probability, challenges in applying testing criteria and low uptake of testing services have impacted detection rates. Furthermore, testing only 9% of patients with BC indicated a low overall testing rate. A significant barrier to adequate referral and

Table 3 Association of P/LP *gBRCAm* with patient characteristics

Parameters	*n* (%)	BRCA1 positive ($n = 29$)	BRCA2 positive ($n = 19$)	BRCA1/2 positive ($n = 48$)	*p*-value
Receptor subtype					
HR (ER/PR) positive	139 (42.8)	3 (2.2)	8 (5.8)	11 (7.9)	0.003
HER2 positive	56 (17.2)	1 (1.8)	3 (5.4)	4 (7.1)	0.07
TNBC	169 (52)	26 (15.4)	10 (5.9)	36 (21.3)	< 0.001
Bilateral breast cancer					
Present	13 (4)	1 (7.7)	2 (15.4)	3 (23.1)	0.4
Absent	312 (96)	28 (9)	17 (5.4)	45 (14.4)	
Significant family history[a]					
Present	103 (31.7)	14 (13.6)	13 (12.6)	27 (26.2)	< 0.001
Absent	222 (68.3)	15 (6.7)	6 (2.7)	21 (9.5)	
Age of onset (y)					
≤50	189 (58.2)	22 (11.6)	10 (5.3)	32 (16.9)	0.2
≥51	136 (41.8)	7 (5.1)	9 (6.6)	16 (11.8)	
Disease characteristic					
Metastatic	64 (20.6)	6 (9.4)	6 (9.4)	12 (18.8)	0.3
Nonmetastatic	261 (80.3)	23 (8.8)	13 (5)	36 (13.8)	

Abbreviations: ER, estrogen receptor; HER, human epidermal growth factor receptor 2; HR, hormone receptor; PR, progesterone receptor; TNBC, triple negative breast cancer.

[a]Following NCCN criteria: Breast cancer (BC) at any age and ≥1 close blood relative (first-, second-, or third-degree relative) with BC, male BC, ovarian cancer, pancreatic cancer, and prostate cancer.

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underutilization of HBOC testing services is the ethical challenge faced by physicians in recommending tests that many patients cannot afford.[11,27] The cost of genetic testing and subsequent treatments, such as PARP inhibitors and risk reduction surgeries are often prohibitive for most patients. This financial burden creates an ethical dilemma for health care providers who must balance the necessity of these tests with the potential financial strain on their patients. Additionally, informing family members of potentially distressing test results, which are crucial for their screening, presents another ethical challenge.[28] The lack of dedicated genetic counsellors in a culturally diverse country like India complicates the communication of genetic testing benefits in a manner that respects the sociocultural norms and values.[29] High financial costs remain the main barrier to the uptake of genetic testing services, with genetic testing affordable for only 15% of newly diagnosed patients with BC.[30–32] Awareness and interest in genetic counseling and testing are low, with many patients disinclined to screen themselves or their families for hereditary BC.[29,30] Factors such as paternalistic attitudes, sociocultural influences, familial tensions, and a low perceived benefit of testing hinder uptake of genetic testing.[11,12,33,34] Social stigma affecting both patients and future generations, coupled with the absence of laws against genetic discrimination in the workplace or society at large, contributes to the reluctance to pursue genetic testing. To improve testing patterns, enacting laws against genetic discrimination could address policy-level issues and alleviate bias concerns. Insurer support is essential to mitigate the costs of genetic testing and treatment, making these services more accessible. BC awareness campaigns should highlight the unique role of genetic testing in precision treatment, distinguishing it from other diagnostic tests. Establishing training programs in genetic counseling and integrating these services into mainstream health care are vital steps forward in bridging the health care gap and ensuring that patients receive comprehensive care that includes genetic testing.

Limitations

The data for this study were acquired from the hospital records. It reflects the patient population that seeks care at this hospital and may not be representative of the general population. Despite this, the study provides a valuable assessment of testing pattern in our institute and the burden of *gBRCAm* among patients with BC and can be used as a reference for future research.

Conclusion

In our cohort of BC patients, germline mutation testing identified P/LP *BRCA1/2* mutations in 14.7% of cases, with a notable association observed in patients with TNBC and those with a notable family history. Available guidelines and sociocultural and economic constructs are key determinants for the referral and uptake of hereditary germline testing for BC. Measures to ensure equitable access to genetic testing can improve testing rates and enhance patient outcomes through personalized care.

Authors' Contributions

Study concept and design were developed by S.S.A., R.P., N.H., S.R. Literature search was performed by S.S.A. Data acquisition and data analysis were done by S.S.A. and V.A. Manuscript preparation was done by S.S.A., K.K.M.V., N.A.Y., and V.A. Manuscript editing and manuscript review were done by S.S.A., R.P., N.H., S.R., S.K., R.T., N.A.Y., and C.C.K.N. The manuscript has been read and approved by all the authors. The requirements for authorship have been met. Each author believes that the manuscript represents honest work.

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Conflict of Interest

None declared.

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