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Characterizing BRCA Mutations in Cameroonian Breast Cancer Patients: Advancing Precision Oncology and Bridging the Genomic Gap in Sub-Saharan Africa

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Abstract

Background: Breast cancer is the most commonly diagnosed malignancy worldwide and represents a growing cause of cancer mortality in sub-Saharan Africa. African populations remain underrepresented in global cancer genomics research, with publications addressing cancer genetics from Africa constituting only 0.016% of the worldwide total. Breast Cancer gene 1 (BRCA1) and BRCA2 mutations, central to hereditary breast cancer, remain understudied in sub-Saharan African populations. Building on prior work demonstrating the willingness of Cameroonian patients to undergo genetic testing and counseling, this study characterizes BRCA1 and BRCA2 mutations among Cameroonian breast cancer patients to address knowledge gaps. **Methods:** This prospective study recruited 82 breast cancer patients from three major oncology referral centers in Yaounde, Cameroon. Following pre-test genetic counseling, saliva samples were collected and analyzed using next-generation sequencing for a panel of 29 cancer-associated genes. Genetic variants were classified according to the American College of Medical Genetics and Genomics (ACMG) 2015 guidelines. Statistical analysis included descriptive statistics, t-tests, chi-

square tests, Fisher's exact test, and linear regression. **Results:** Among 82 breast cancer patients, BRCA1 pathogenic or likely pathogenic (P/LP) variants were identified in 16 patients (19.5%) and BRCA2 in 2 patients (2.4%), giving a combined BRCA1/2 P/LP rate of 22.0%. The most frequent BRCA1 variant was c.4484G>T (p.Arg1495Met), identified in 7 patients (43.8% of BRCA1-positive patients), raising the possibility of a population-specific recurrent variant. Variants of uncertain significance (VUS) were detected in 25.6% of patients. Regression analysis identified younger age at diagnosis ($p = 0.04$) and positive family history ($p = 0.03$) as significant predictors of BRCA1/2 carrier status. **Conclusions:** This study reveals a high prevalence of BRCA1/2 pathogenic variants in Cameroonian breast cancer patients. The recurrence of a specific BRCA1 mutation raises the hypothesis of a population-specific genetic architecture warranting further investigation. These findings reveal the need for inclusive genomic research and equitable implementation of precision oncology in underrepresented communities.

Keywords

BRCA1, BRCA2, Hereditary Breast Cancer, Africa, Cameroon, Precision Oncology, Genomic Inequity, Genetic Counseling, VUS

1. Introduction

Breast cancer has emerged as the most frequently diagnosed malignancy globally and represents a leading cause of cancer-related mortality among women in sub-Saharan Africa [1]. Unlike the relatively stable incidence trends in high-income countries, the burden of breast cancer across Africa is rising, driven by epidemiological transitions, urbanization, and persistently limited access to early detection and treatment [2]. Women in sub-Saharan Africa are diagnosed at younger ages on average and present more frequently with advanced-stage disease [3], reflecting a convergence of biological, socioeconomic, and structural factors.

Among the factors driving this inequity is a profound gap in genomic knowledge. Despite harboring the greatest genetic diversity of any human population [4], African individuals account for a disproportionately small fraction of germline and somatic genomic studies. Publications addressing cancer genetics from African populations constitute a mere 0.016% of the global total [5] [6]. This underrepresentation has cascading consequences: elevated rates of variants of uncertain significance (VUS), inaccurate risk prediction models, population-inappropriate screening guidelines, and constrained access to targeted therapies [5]-[7].

BRCA1 and BRCA2 are the most clinically significant genes in the context of hereditary breast cancer, collectively accounting for the majority of hereditary cases in Western cohorts [8] [9]. Studies from Africa have begun to illuminate the prevalence and spectrum of BRCA mutations in African women, consistently revealing variable yet often substantial pathogenic variant rates alongside elevated VUS rates attributable to underrepresentation in global databases [10] [11].

Given the scarcity of hereditary cancer research in Cameroon, Mapoko *et al.* conducted foundational feasibility work demonstrating that Cameroonian patients show a strong willingness to engage with genetic testing and counseling programs, even within a low-resource context [12]—a finding that laid the necessary social and ethical groundwork for the present study. The current investigation provides a genomic characterization of hereditary breast cancer in Cameroon to date, addressing critical gaps in knowledge of BRCA1/2 mutation prevalence, the mutation spectrum, and evidence for potential recurrent variants, the burden of VUS, and the clinico-genomic associations that can guide clinical practice. The full program implementation and comprehensive multi-gene findings across all cancer types are described in companion publications.

2. Methods

2.1. Study Design and Setting

This prospective study was conducted between 2021 and 2023 at three major oncology referral centers in Yaounde, Cameroon: the Yaounde General Hospital, the Yaounde Central Hospital, and the Solidarity Chemotherapy Non-Governmental Organization. Ethical approval was obtained from the Cameroonian National Human Health Research Ethics Committee (N˚2021/12/1424/CE/CNERSH/SP).

2.2. Study Participants

A total of 82 breast cancer patients who had never previously undergone genetic testing were enrolled. Eligibility required a histologically confirmed breast cancer and age of at least 18 years. Demographic, reproductive, and clinical data were collected alongside detailed family histories. Enrollment was consecutive. Of the 111 eligible patients approached, 24 declined participation due to fear of learning about the existence of mutations, and 5 were excluded because they were under 18 years of age. All three centers contributed patients: Yaounde General Hospital, Yaounde Central Hospital, and SOCHIMIO. Family history of cancer was defined as the presence of at least one first- or second-degree relative with a documented cancer diagnosis, including breast, ovarian, colorectal, gastric, uterine, cervical, melanoma, pancreatic and prostate cancer.

2.3. Genetic Counseling

All participants received culturally adapted pre-test genetic counseling delivered by Cameroon's trained cancer genetic counselors. The counseling protocol incorporated both national languages (French and English) and addressed community concerns around hereditary disease, stigma, and implications for family members. Post-test counseling was provided upon return of results.

2.4. Sample Collection and Genetic Sequencing

Five millilitres of saliva were collected from each participant. DNA was extracted and analyzed using next-generation sequencing (NGS) with a panel of 29 cancer

predisposition genes: BAP1, MITF, CDK4, CDKN2A, ATM, CDH1, CHEK2, PTEN, BRCA1, BRCA2, PALB2, BARD1, BRIP1, TP53, STK11, MLH1, MSH2, EPCAM, MSH6, PMS2, RAD51C, RAD51D, APC, BMPR1A, SMAD4, GREM1, MUTYH, POLD1, and POLE. Sequencing was performed at Color Genomics (Burlingame, CA, USA), a College of American Pathologists (CAP)-accredited, Clinical Laboratory Improvement Amendments (CLIA)-certified laboratory. The present article focuses specifically on BRCA1 and BRCA2 findings. Pathogenic or likely pathogenic variants identified in the remaining 27 genes of the panel are reported in full in the companion landscape article. Among the 82 breast cancer patients in this cohort, no participant carried more than one BRCA1/2 P/LP variant; however, 8 patients carried a BRCA1/2 P/LP variant alongside a VUS in a second gene, as detailed in section 3.5.

2.5. Variant Classification

All identified variants were classified as pathogenic (P), likely pathogenic (LP), variant of uncertain significance (VUS), likely benign, or benign in accordance with American College of Medical Genetics and Genomics/Association for Molecular Pathology (ACMG/AMP) 2015 guidelines [13], cross-referenced with ClinVar.

2.6. Statistical Analysis

Descriptive statistics were calculated as mean ± SD for continuous variables and frequencies with percentages for categorical variables. Independent samples t-tests compared the mean age at diagnosis between BRCA1/2 carriers and non-carriers. Chi-square tests and Fisher's exact test assessed associations between mutation status and categorical variables. Linear regression identified predictors of BRCA1/2 carrier status. All tests were two-sided; $p < 0.05$ was considered statistically significant. The multivariable regression model included the following covariates: age at diagnosis (continuous), family history of cancer (binary: yes/no), menopausal status (binary), and tumor stage at diagnosis (ordinal: I - IV). Missing data were handled by complete-case analysis (listwise deletion). This analysis used a 29-gene cancer predisposition panel (see section 2.4); pathogenic or likely pathogenic variants identified in genes other than BRCA1/2 are reported in the companion landscape article.

3. Results

3.1. Patient Characteristics

The cohort comprised 82 breast cancer patients. The mean age at diagnosis was 43.4 ± 11.5 years (range 19 - 71). Forty-three patients (52.4%) were diagnosed before age 45. **Figure 1** summarizes the age distribution at diagnosis in breast cancer patients. A family history of cancer was reported by 57 patients (69.5%). Among patients reporting family history, the most commonly reported malignancy in relatives was breast cancer (n = 44), followed by cervical (n = 11), pros-

tate (n = 10), uterine (n = 5), ovarian (n = 4), gastric (n = 4), and colorectal (n = 3) cancers.

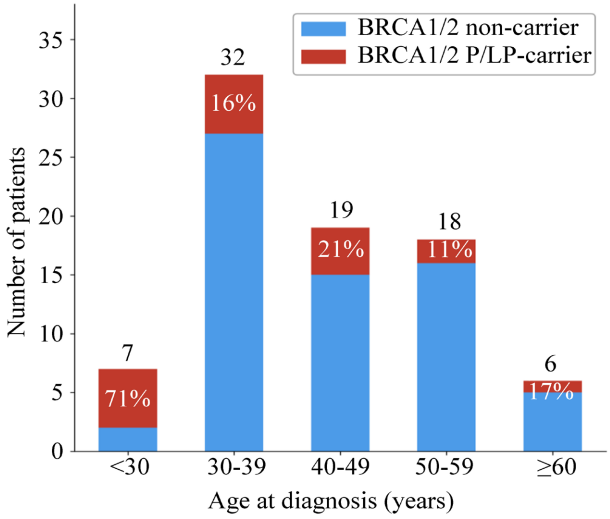

Figure 1. Age distribution at diagnosis in breast cancer patients (N = 82) by BRCA1/2 P/LP carrier status.

3.2. Prevalence and Distribution of Pathogenic BRCA1/2 Variants

Pathogenic (P) BRCA1 variants were identified in 15 patients (18.3% of the breast cancer cohort) and one likely pathogenic (LP) BRCA1 variant in an additional patient, giving a total of 16 BRCA1 P/LP carriers (19.5%). Two patients carried pathogenic BRCA2 variants (2.4%). Combined, BRCA1/2 P/LP variants were present in 18 breast cancer patients (22.0%). Table 1 presents the complete BRCA1/2 variant spectrum.

Table 1. Frequency and distribution of pathogenic BRCA1 and BRCA2 mutations (N = 82).

Gene	Nucleotide change	Protein change	Class.	n	% gene	% cohort
BRCA1	c.4484G>T	p.Arg1495Met	P	7	43.8%	8.5%
BRCA1	c.5155dup	p.Val1719Glyfs*6	P	3	18.8%	3.7%
BRCA1	c.38del	p.Asn13Metfs*10	P	1	6.3%	1.2%
BRCA1	c.66dup	p.Glu23Argfs*18	P	1	6.3%	1.2%
BRCA1	c.2157dup	p.Glu720Argfs*6	P	1	6.3%	1.2%
BRCA1	c.2517_2518del	p.His839Glnfs*12	P	1	6.3%	1.2%
BRCA1	c.3607C>T	p.Arg1203*	P	1	6.3%	1.2%
BRCA1	c.4676-1G>C	Splice acceptor	P	1	6.3%	1.2%
BRCA1	c.5155dup	p.Val1719Glyfs*6	LP	1	—	1.2%
BRCA2	c.1813dup	p.Ile605Asnfs*11	P	1	50.0%	1.2%
BRCA2	c.5572del	p.Thr1858Glnfs*5	P	1	50.0%	1.2%

3.3. BRCA1 Mutation Spectrum and Evidence of a Possible Recurrent Variant

Eight distinct P/LP BRCA1 variants were identified in 16 BRCA1-positive patients (Figure 2). The most frequent was c.4484G>T (p.Arg1495Met), detected in 7 patients, representing 43.8% of all BRCA1 P/LP carriers and 8.5% of the total breast cancer cohort. The second most frequent BRCA1 variant was c.5155dup (p.Val1719Glyfs*6), found in 4 patients (including 1 LP). Additional BRCA1 variants—c.2157dup, c.2517_2518del, c.3607C>T, c.38del, c.4676-1G>C, and c.66dup—were each observed in a single patient. The variant c.4484G>T (p.Arg1495Met) is classified as pathogenic (Class 5) in ClinVar based on ACMG/AMP criteria: PS1 (same amino acid change as established pathogenic variant), PS3 (functional studies demonstrating loss of BRCA1 function), PM2 (absent from gnomAD), and PP3 (multiple computational evidence of deleteriousness). The variant c.5155dup (p.Val1719Glyfs*6) was classified as pathogenic in 3 patients and as likely pathogenic in 1 patient. This discrepancy reflects [case-level reinterpretation based on individual clinical data/laboratory re-evaluation between testing dates/slight differences in allele-specific evidence]. The 4 patients are counted separately in the variant frequency table.

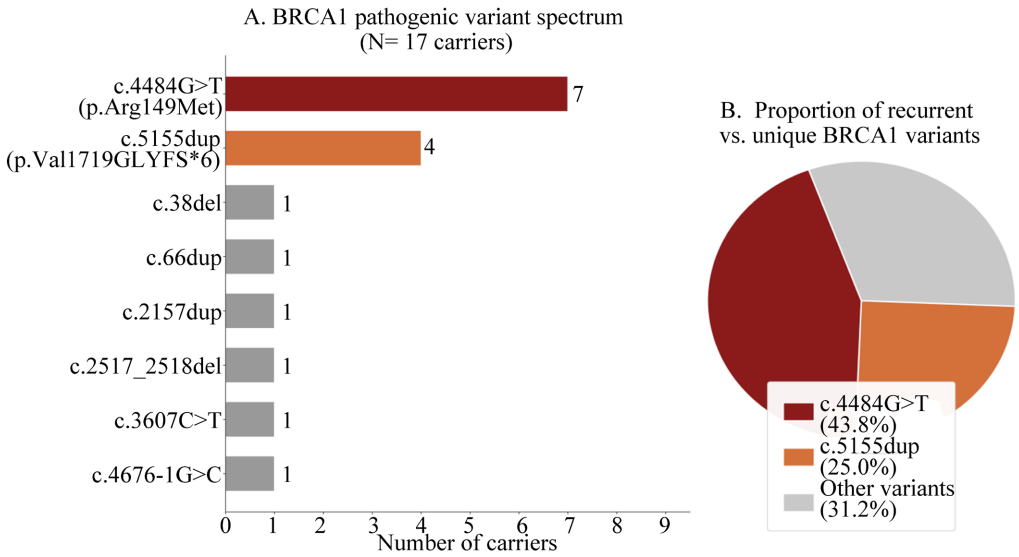

Figure 2. BRCA1 pathogenic variant spectrum (N = 16 BRCA1 P/LP carriers).

3.4. BRCA2 Mutation Spectrum

Two pathogenic BRCA2 variants were identified: c.1813dup (p.Ile605Asnfs*11) and c.5572del (p.Thr1858Glnfs*5), each found in one patient, each comprising 50% of all BRCA2 pathogenic findings.

3.5. Variants of Uncertain Significance

VUS were detected in 21 breast cancer patients (25.6%). Among BRCA1/2 P/LP carriers, VUS co-occurrence was notable: 8 of 18 BRCA1/2 carriers (44.4%) had an associated VUS in a second gene. Two patients with the recurrent c.4484G>T

BRCA1 variant had a co-occurring VUS in PALB2 (c.365A>G, p.Asp122Gly); both were under 30 years of age and reported multiple first- and second-degree relatives with breast or ovarian cancer. Two other patients carried a VUS in APC (c.3760A>G, p.Ile1254Val). No VUS was identified within BRCA1 itself; two VUS were detected in BRCA2.

3.6. Clinico-Genomic Associations

The mean age at diagnosis was 35.84 ± 9.92 years among BRCA1/2 P/LP carriers, compared to 40.54 ± 10.65 years in non-carriers. Family history of cancer was reported in 76.5% of BRCA1/2 carriers versus 57.6% of non-carriers. Regression analysis identified younger age at diagnosis (p = 0.04) and positive family history (p = 0.03) as significant independent predictors of BRCA1/2 carrier status (**Figure 3**).

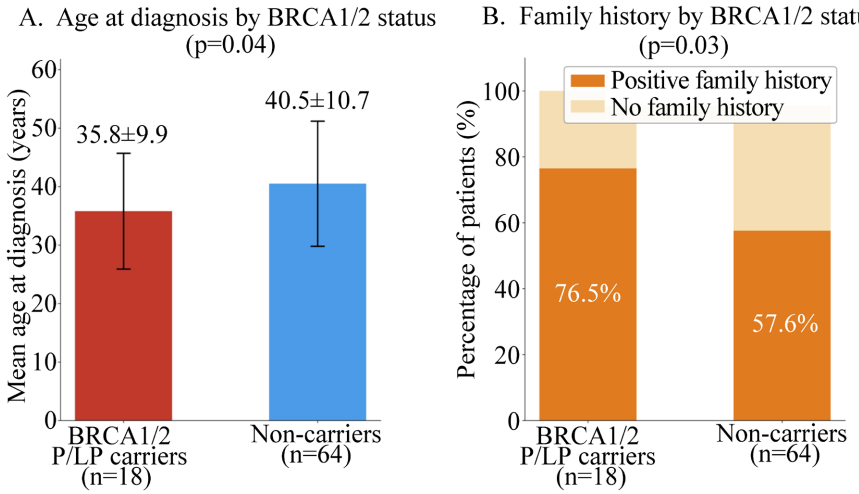

Figure 3. Clinical predictors of BRCA1/2 P/LP carrier status. (A) Mean age at diagnosis ± SD by carrier status (p = 0.04, t-test); (B) Proportion with positive family history by carrier status (p = 0.03, chi-square).

4. Discussion

The high prevalence of pathogenic BRCA1/2 variants in this Cameroonian breast cancer cohort is among the highest reported in any African population study and rivals figures typically found in high-risk hereditary cancer clinics in Western countries [8] [9]. Although preliminary, this may suggest that hereditary breast cancer is underdiagnosed in Cameroon. The magnitude of this prevalence challenges prevailing assumptions that attribute the African breast cancer burden predominantly to environmental and reproductive factors [2], and reveals the need to integrate germline genetic evaluation into standard oncological workup.

This finding must be interpreted with caution and confirmed in larger cohorts including controls. Authors from West Africa with larger cohorts report rates ranging from 6 to 7% [14] [15]. However, Manirakiza *et al.* in Rwanda demonstrated BRCA mutation rates as high as 18.3% in female breast cancer [16], which may suggest regional genetic variation across Africa [4], warranting larger studies

across more representative African regions. Selection bias also warrants consideration: cohorts recruited from tertiary oncology centers may disproportionately represent younger patients and those with more aggressive phenotypes—precisely the profile associated with hereditary breast cancer.

One of the most significant findings of this study is the recurrence of BRCA1 c.4484G>T (p.Arg1495Met) in 7 patients, comprising 43.8% of all BRCA1 pathogenic variants detected. This pattern raises the possibility of a founder or otherwise recurrent variant specific to the Cameroonian population. If confirmed, the identification of such a variant could enable the development of targeted, low-cost, population-specific genetic screening tests that do not require comprehensive panel sequencing, dramatically reducing costs in resource-constrained settings [17].

The recurrence of this variant provides a compelling rationale for expanded population-based genotyping studies in Cameroon and neighboring Central African countries, which share population genetic histories and may harbor the same or related variants [4]. Future studies should integrate genome-wide single nucleotide polymorphism (SNP) data and haplotype analysis to assess whether this recurrence reflects a common ancestral haplotype. Until such analyses are performed, this observation should be considered a preliminary signal requiring replication.

The detection of VUS in 25.6% of breast cancer patients—and in 44.4% of confirmed BRCA1/2 carriers—reflects a well-recognized consequence of the chronic underrepresentation of African populations in global genomic reference databases [4]-[6]. African populations carry a disproportionate number of rare variants absent or underrepresented in databases built predominantly on European and East Asian data [18], leading to diagnostic uncertainty, potential misclassification, and reduced access to targeted therapies [7] [19] [20].

The co-occurrence of VUS with confirmed pathogenic mutations adds further complexity to patient management, as illustrated by the two patients carrying both the BRCA1 c.4484G>T variant and a PALB2 VUS. Addressing the VUS burden in African populations is not merely a technical challenge; it is a matter of health equity [7] [19]. Resolving these variants requires systematic inclusion of African genomes in global reference consortia, creation of African-specific variant databases, and sustained investment in local genomic infrastructure.

The high mutation yield observed strongly supports extending genetic testing to breast cancer patients in Cameroon, particularly those presenting at a young age or with significant family history. Realizing the full potential of these findings requires concerted investment: genetic counseling certification programs, local laboratory sequencing capacity, collaboration with African and global genomic reference centers, and integration of PARP inhibitors into national treatment formularies. The hereditary cancer DNA biobank established through this study represents a foundational infrastructure investment for future mutational studies and familial segregation analyses across Central Africa.

Limitations

Several limitations warrant consideration. First, the study was conducted at urban

tertiary centers in Yaounde, potentially limiting generalizability to rural populations and other regions. Second, the sample size (N = 82) limits statistical power for subgroup analyses; larger, population-representative studies are needed. Third, haplotype analysis was not performed to confirm the putative recurrent status of c.4484G>T; this remains a working hypothesis. Fourth, clinical outcome data (receptor status, survival) were unavailable for most patients, precluding genotype-phenotype analyses. Finally, the use of an international laboratory introduces logistical delays and costs not representative of a mature national program.

5. Conclusion

This study provides a characterization of BRCA1 and BRCA2 mutations in Cameroonian breast cancer patients, filling a critical gap in the African genomic landscape. The high prevalence of pathogenic BRCA variants reveals a substantial hereditary cancer burden that has remained largely invisible to clinical practice. The recurrence of a specific BRCA1 mutation in a significant proportion of carriers raises a compelling hypothesis of a population-specific variant with implications for targeted screening programs—a hypothesis that must be confirmed through haplotype studies and replication in larger cohorts. The high rate of co-occurring VUS highlights the urgent need to increase African representation in global genomic reference databases. Taken together, these findings make a compelling case for the integration of hereditary breast cancer genetic testing into oncological care in Cameroon.

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Conflicts of Interest

The authors declare no conflicts of interest.

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List of Abbreviations

Abbreviation	Definition
ACMG	American College of Medical Genetics and Genomics
AMP	Association for Molecular Pathology
BRCA1/2	Breast Cancer gene 1/2
CAP	College of American Pathologists
CLIA	Clinical Laboratory Improvement Amendments
DNA	Deoxyribonucleic acid
GENCAF	Genetic Counseling and Cancer Testing in Africa
LP	Likely pathogenic
NGS	Next-generation sequencing
P	Pathogenic
P/LP	Pathogenic or likely pathogenic
PARP	Poly(ADP-ribose) polymerase
SNP	Single nucleotide polymorphism
SSA	Sub-Saharan Africa
VUS	Variant of uncertain significance