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Genomic Landscape of Hereditary Cancer in Cameroon: Comprehensive Analysis of 94 Patients Undergoing Multigene Germline Testing in the GENCAF Program

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Abstract

Background: Hereditary cancer is increasingly recognized as a major contributor to the oncology burden in Africa. Cameroon, like most sub-Saharan countries, had long lacked access to structured genetic counseling and germline testing. Through the Genetic Cancer Families (GENCAF) initiative—whose acceptability was established in a prior feasibility study and whose implementation is described separately—we conducted a systematic multigene panel sequencing of cancer patients in the country. **Methods:** We performed a cross-sectional analysis of 94 consecutive cancer patients enrolled in the GENCAF program between May 2022 and December 2023. All participants received standardized pre-test counseling and saliva-based DNA sampling. Sequencing was performed using a 29-gene hereditary cancer panel. Variants were classified according to American College of Medical Genetics and Genomics (ACMG) criteria. We analyzed the prevalence, spectrum, and clinical correlates of pathogenic/likely pathogenic (P/LP) variants, variants of uncertain significance (VUS), and multi-gene variant profiles across all cancer types. **Results:** Among 94 individuals tested (mean age 43.4 years, 95.7% women), breast cancer was

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the predominant diagnosis (87.2%). Germline P/LP variants were identified in 27.7% (26/94; 25 pathogenic, 1 likely pathogenic). Breast Cancer gene (BRCA) 1 accounted for 69.2% of all P/LP findings. VUS were present in 26.6% of patients across 13 genes. Eight patients (8.5%) carried co-occurring P/LP and VUS in different genes. Pathogenic variants were more common in individuals with a family history (33%) than without (16%; p = 0.12). **Conclusion:** This study provides a detailed genomic mapping of hereditary cancer susceptibility in Cameroon. The high prevalence of BRCA1 P/LP variants, substantial VUS burden across 13 genes, and early age at diagnosis highlight the urgency of integrating genetic counseling and testing into national cancer control strategies.

Keywords

Cameroon, Hereditary Cancer, BRCA1, Genomics, Multigene Panel, Sub-Saharan Africa, VUS, Lynch Syndrome, PALB2

1. Introduction

Cancer epidemiology in sub-Saharan Africa is in rapid transition, with an increasing contribution from non-communicable, non-infectious etiologies and a rise in early-onset disease [1]-[3]. Although environmental and infectious factors remain important, mounting evidence indicates that hereditary predisposition may play a much larger role in African populations than previously recognized [4]-[6]. Studies from Nigeria, South Africa, and Ghana have reported elevated rates of BRCA1/2 and mismatch repair (MMR) gene mutations, as well as unusually young ages at presentation [7]-[10]. Yet genetic counseling and germline testing remain inaccessible to the vast majority of African cancer patients [11]-[13].

Several structural barriers explain this gap: scarcity of trained genetic professionals, absence of dedicated infrastructures, high cost of testing, dependence on foreign laboratories, and underrepresentation of African genomes in reference databases leading to high VUS rates [14]-[17]. In Cameroon like in Africa, reports of strong familial clustering and early-onset breast cancer have raised concerns about a significant but unexplored hereditary cancer burden [18]-[20].

Prior to the GENCAF program, no systematic germline testing was available in the country. The acceptability and feasibility of genetic services among Cameroonian patients were established in a prior cross-sectional study [13]. The present article focuses on the full genomic landscape of hereditary cancer susceptibility in Cameroon, providing a granular characterization of pathogenic germline variants across all 29 tested genes and all cancer types, including the distribution of VUS, multi-variant profiles, and clinico-genomic associations.

2. Methods

2.1. Study Design and Population

We conducted a cross-sectional descriptive study using data from consecutive pa-

tients enrolled in the GENCAF program between May 2022 and December 2023. The program was implemented across three major oncology centers in Yaounde: Yaounde General Hospital, Yaounde Central Hospital, and the Chemotherapy Solidarity Non-Governmental Organization (SOCHIMIO). Together, these institutions account for an estimated 80% of cancer consultations in the capital region.

Inclusion criteria were: 1) all cancer patients suspected to have a hereditary component based on at least one of the following clinical red flags: breast or ovarian cancer diagnosed at age ≤ 45 years; triple-negative breast cancer at any age; two or more first- or second-degree relatives with breast, ovarian, colorectal, gastric, or prostate cancer; bilateral or multicentric breast cancer; male breast cancer; or personal/family history suggestive of a recognized hereditary syndrome (Lynch syndrome, Li-Fraumeni syndrome, familial adenomatous polyposis); 2) completion of pre-test genetic counseling; and 3) successful saliva-based DNA collection and valid sequencing results. Enrollment was consecutive. Of 123 patients assessed for eligibility, 112 attended pre-test genetic counseling. Of these, 94 (83.9%) proceeded to germline multigene testing. Reasons for non-testing included refusal after counseling (n = 14, primarily citing fear of learning about the existence of mutations), financial barriers (n = 3), and inadequate saliva sample (n = 1). Patients under 18 years of age (n = 5 of those initially approached) were excluded before counseling. All 94 tested patients yielded valid sequencing results.

2.2. Clinical Assessment and Genetic Counseling

Pre-test genetic counseling was performed using a standardized workflow adapted from NCCN and ESMO guidelines to the Cameroonian context, covering hereditary cancer syndromes, possible testing outcomes (P/LP, VUS, negative), treatment and surveillance implications, and psychosocial considerations. Post-test counseling delivered actionable findings and cascade testing advice. Family history of cancer was defined as at least one first- or second-degree relative with a documented cancer diagnosis, regardless of cancer type. Family history was collected through structured interview and three-generation pedigree construction during pre-test counseling.

2.3. Sample Collection and DNA Extraction

DNA was collected from saliva using Oragene OG-600 collection kits, stabilized at room temperature, and shipped to a Clinical Laboratory Improvement Amendments (CLIA)-certified international partner laboratory (Color Genomics, Burlingame, CA, USA). DNA extraction followed manufacturer instructions with fluorometric quantification (Qubit).

2.4. Multigene Panel Sequencing

All samples underwent next-generation sequencing (NGS) using a 29-gene hereditary cancer panel: APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, GREM1, MITF, MLH1, MSH2,

MSH6, MUTYH, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53. Mean coverage was >250×; minimum per-base coverage >20× for >99% of targeted bases. Variant calling followed GATK-based workflows with alignment to GRCh38, annotated via ClinVar, gnomAD (global and African datasets), and LOVD.

2.5. Variant Classification

All variants were classified per American College of Medical Genetics and Genomics and Association for Molecular Pathology (ACMG/AMP) 2015 guidelines [21] into P, LP, VUS, likely benign, or benign. Only P, LP, and VUS variants were reported. Copy-number variant detection was performed using depth-of-coverage algorithms.

2.6. Statistical Analysis

Descriptive statistics were calculated for all variables. Fisher's exact test assessed associations between P/LP status and family history. Age comparisons across gene groups used one-way ANOVA. $p < 0.05$ was considered significant. Analyses were performed using SPSS version 23.

2.7. Ethical Considerations

Ethical approval: N˚2021/12/1424/CE/CNERSH/SP—National Human Health Research Ethics Committee, Cameroon. The study was conducted in accordance with the Declaration of Helsinki. Identifiable data were not used in this analysis.

3. Results

3.1. Cohort Characteristics

A total of 94 patients underwent successful multigene germline testing. The cohort was 95.7% female (n = 90; 4 male). Mean age at diagnosis was 43.4 years (SD ± 11.5; range 19 - 71). Age distribution: <40 years 45.7% (n = 43), 40 - 49 years 22.3% (n = 21), ≥50 years 31.9% (n = 30). Table 1 summarizes cohort characteristics.

Table 1. Cohort characteristics (N = 94).

Variable	N (%)
Female gender	90 (95.7%)
Mean age at diagnosis	43.4 years (SD ±11.5)
Age < 40 years	43 (45.7%)
Breast cancer	82 (87.2%)
Colorectal cancer	5 (5.3%)
Gastric cancer	3 (3.2%)
Ovarian cancer	2 (2.1%)
Renal/prostate	2 (2.1%)
Positive family history	57 (60.6%)

According to cancer type distribution, breast cancer was the most represented cancer type at 87.2% (Figure 1). A positive family history of cancer—defined as at least one first- or second-degree relative with a documented cancer diagnosis—was reported by 57 patients (60.6%). Among breast cancer patients, a notably high proportion presented with triple-negative disease—a subtype strongly associated with hereditary BRCA1 mutations. High-risk clinical features were common: 45.7% were diagnosed before age 45, and 60.6% reported a positive family history of cancer.

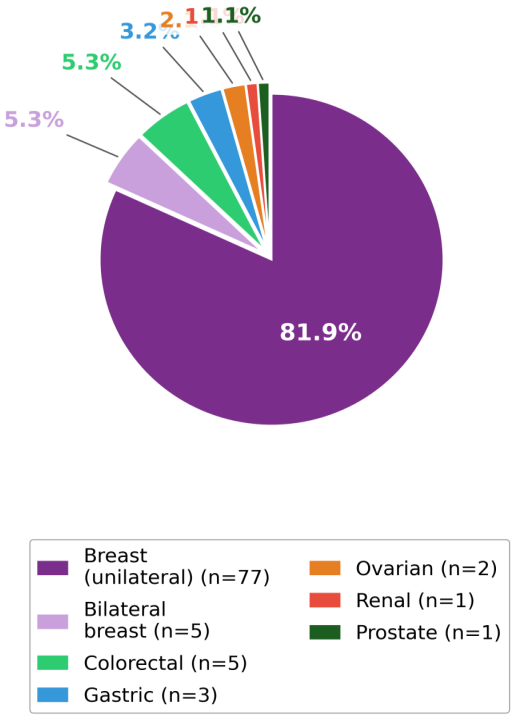


Figure 1. Cancer type distribution among GENCAF patients (N = 94).

3.2. Prevalence and Distribution of P/LP Variants

Among 94 participants, 25 carried at least one pathogenic (P) variant (26.6%) and 1 carried a likely pathogenic (LP) variant (1.1%), for a combined P/LP yield of 26 patients (27.7%). Table 2 presents the distribution of pathogenic/likely pathogenic variants by gene amongst the mutated genes, amongst the population and the syndromes related while Figure 2 presents the distribution of P/LP variants by gene.

Table 2. Distribution of pathogenic/likely pathogenic variants by gene amongst the mutated genes and amongst the population (N = 94).

Gene	n (P/LP)	% of P/LP	% cohort	Hereditary syndrome
BRCA1	18 (17P + 1LP)	69.2%	19.1%	Hereditary breast and ovarian cancer (HBOC)
PALB2	2	7.7%	2.1%	HBOC—elevated breast and pancreatic risk

Continued

BRCA2	2	7.7%	2.1%	HBOC—breast, ovarian, prostate, pancreatic
PMS2	2	7.7%	2.1%	Lynch syndrome—colorectal, endometrial
MSH2	1	3.8%	1.1%	Lynch syndrome—colorectal, endometrial, urinary
ATM	1	3.8%	1.1%	Hereditary breast, pancreatic risk
Total	26	100%	27.7%	

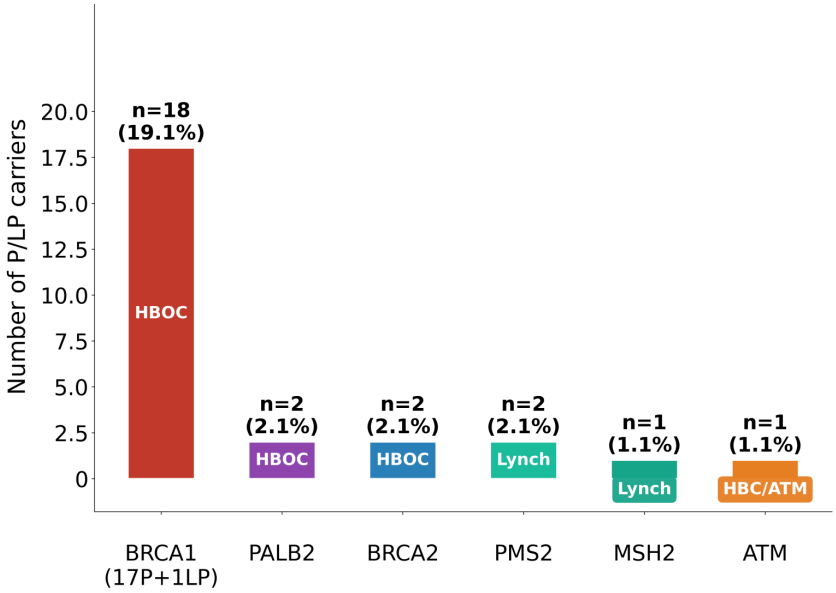


Figure 2. Distribution of P/LP variants by gene and associated syndromes (N = 94; 26 P/LP carriers, 27.7%).

BRCA1 overwhelmingly dominated the mutational landscape, accounting for 69.2% of all P/LP findings.

3.3. Variants of Uncertain Significance—Distribution and Implications

VUS were identified in 25 patients (26.6%), distributed across 13 distinct genes. **Table 3** details VUS distribution by gene, and **Figure 3** details VUS distribution across 13 genes.

Table 3. Distribution of variants of uncertain significance by gene (N = 94).

Gene	n VUS	% cohort	Pathway/reclassification priority
ATM	5	5.3%	DNA damage response—high clinical relevance; reclassification priority
APC	5	5.3%	Familial adenomatous polyposis pathway—colorectal risk

Continued

PALB2	4	4.3%	BRCA2-interacting protein—high priority for breast/pancreatic risk
MSH2	2	2.1%	Lynch syndrome—GI and gynecologic risk
MLH1	2	2.1%	Lynch syndrome—GI and gynecologic risk
PMS2	2	2.1%	Lynch syndrome—GI and gynecologic risk
MUTYH	2	2.1%	Base excision repair; biallelic causes MUTYH-associated polyposis
BARD1	2	2.1%	BRCA1-interacting protein—breast/ovarian risk
BRCA2	2	2.1%	HBOC—high priority for reclassification
TP53	1	1.1%	Li-Fraumeni syndrome—pan-cancer risk
BRIP1	1	1.1%	Fanconi pathway; ovarian cancer risk
CHEK2	1	1.1%	Cell cycle checkpoint—moderate breast cancer risk
RAD51D	1	1.1%	Homologous recombination; ovarian cancer risk

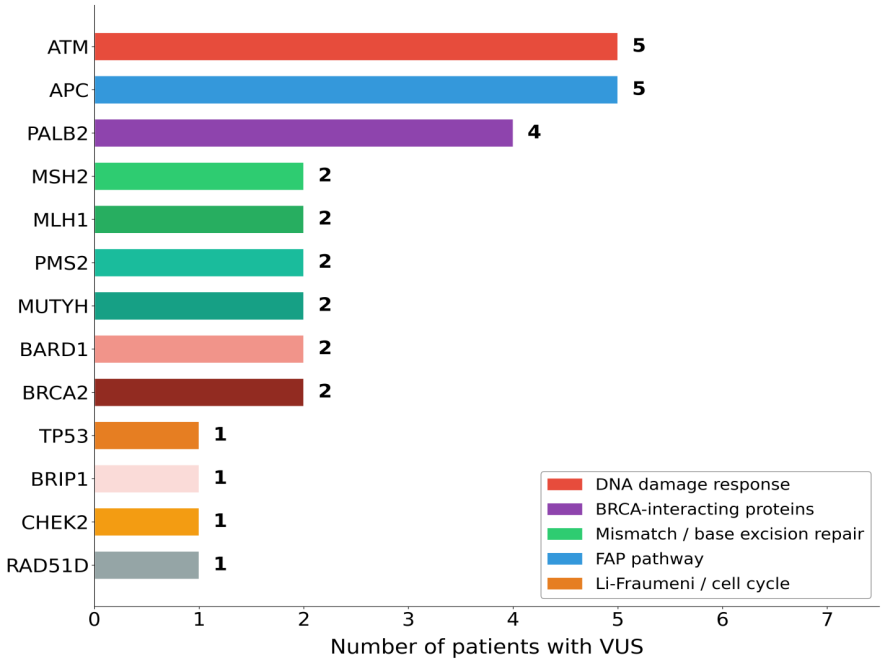


Figure 3. VUS distribution across 13 genes (N = 94; 25 patients with VUS, 26.6%). Colors indicate biological pathway. VUS: variant of uncertain significance; FAP: familial adenomatous polyposis; MMR: mismatch repair.

3.4. Combined Variant Burden

When combining P/LP and VUS findings, 43 patients (45.7%) carried at least one variant of any classification. It is important to note that VUS findings do not constitute evidence of hereditary cancer risk: their clinical significance remains uncertain pending reclassification through functional studies, segregation analyses, or updated population databases. This combined figure is reported to characterize the genomic complexity of the cohort, not to imply pathogenicity of VUS. Fifty-

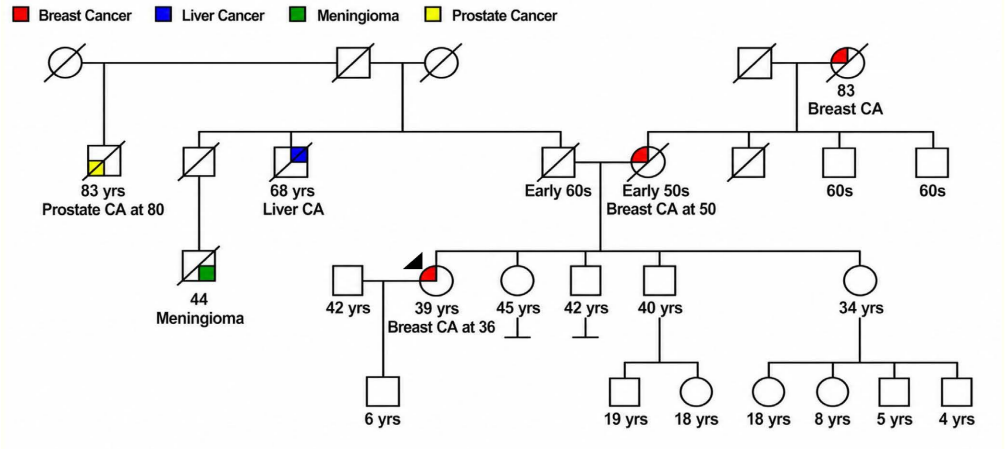
one patients (54.3%) had completely negative results across all 29 genes.

3.5. Multi-Variant Profiles

Eight patients (8.5%) carried a P/LP variant in one gene and a co-occurring VUS in a different gene. Five additional patients carried two VUS in different genes. Representative combinations included BRCA1 P + PALB2 VUS (3 patients), BRCA1 P + ATM VUS (2 patients), BRCA1 P + APC VUS (2 patients), and MSH2 P + PMS2 VUS (1 patient). The recurrence of PALB2 VUS among BRCA1 P/LP carriers warrants longitudinal follow-up and functional characterization.

3.6. Family History and P/LP Status

Among patients with a positive family history of cancer, 33% carried a P/LP variant, compared to 16% among those without family history (Fisher's exact p = 0.12). Although the difference did not reach statistical significance, the trend supports genomic screening even in the absence of a documented family history, given the high rate of early-onset cancers in a context where hereditary cancer has been systematically undiagnosed. Figure 4 presents the pedigree of a 39-year-old female with a positive family history of cancer who presented with right breast cancer and a pathogenic mutation of PMS 2 gene.



CA: *cancer.*

Figure 4. Pedigree of a 39-year-old female with a positive family history of cancer who presented a right breast cancer and a pathogenic mutation of PMS 2 gene.

3.7. Age at Diagnosis by Gene Group

BRCA1 P/LP carriers were diagnosed at a mean age of 37.8 years (n = 17), significantly younger than non-carriers (mean 45.1 years; t-test p = 0.007). The age analysis includes 17 of the 18 BRCA1 P/LP carriers: the one likely pathogenic (LP) carrier (C0066) was excluded from the inferential age comparison due to the lower certainty of her classification, though her age at diagnosis (57 years) is reported descriptively. Age data were available for all 94 participants. Gene groups with n = 1 - 2 (BRCA2, PALB2, ATM, MSH2) are reported descriptively and were not

included in inferential group comparisons. BRCA2 carriers averaged 52.0 years (n = 2), PALB2 carriers 56.0 years (n = 2), ATM carriers 56.0 years (n = 1), and MSH2 carriers 48.0 years (n = 1). These figures are reported descriptively given the small group sizes (Figure 5).

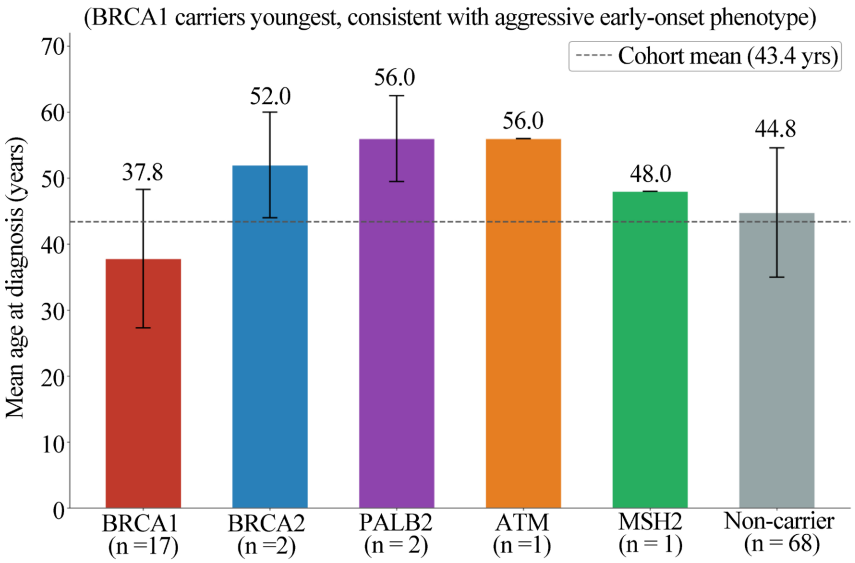


Figure 5. Mean age at diagnosis by gene group.

4. Discussion

This study provides a detailed characterization to date of the hereditary cancer genomic landscape in Cameroon, based on comprehensive germline sequencing of 94 patients enrolled in the GENCAF program. It is important to acknowledge that 87.2% of participants had breast cancer, and the overall P/LP yield is primarily driven by breast cancer cases from tertiary referral centers in Yaounde. Among non-breast cancer patients (n = 12), P/LP variants were identified in the following groups: colorectal (n = 5; 2 P/LP identified: MSH2 in 1, PMS2 in 1), gastric (n = 3; no P/LP), ovarian (n = 2; no P/LP), renal (n=1; no P/LP), prostate (n = 1; no P/LP). Conclusions about hereditary cancer burden across all cancer types, or about Cameroon more broadly, should therefore be drawn with caution from this dataset.

The overall P/LP frequency of 27.7% is considerably higher than the 10% - 15% typically reported in Western populations [22]-[24]. Similar elevated frequencies have been documented in Nigeria, Uganda, and South Africa [4]-[7]. Several factors may contribute: clinic-based selection toward early-onset and family-history-positive patients, possible population-specific variant enrichment, and the high proportion of triple-negative breast cancer in this cohort—a subtype strongly associated with BRCA1 mutations.

The disproportionate prevalence of BRCA1 pathogenic variants (69.2% of all P/LP findings) aligns with studies in West and East Africa [8]-[11].

The VUS rate of 26.6% across 13 genes is consistent with reports from African

cohorts [14]-[16] [12], reflecting the severe underrepresentation of African genomes in global databases [17]. This breadth—spanning DNA damage response, mismatch repair, base excision repair, and BRCA-interacting pathways—is itself a key finding. The development of African-specific variant registries, participation in global reclassification consortia, and systematic sharing of data with ClinVar and LOVD represent strategic priorities. The 5 APC VUS are particularly noteworthy: reclassification as pathogenic would indicate familial adenomatous polyposis risk with major implications for affected families.

Approximately 8.5% of patients carried co-occurring P/LP and VUS variants in different genes—multi-nearly configurations warrant longitudinal follow-up but their combined clinical significance has not been established in this cohort. The recurrence of PALB2 VUS among BRCA1 P/LP carriers merits further functional characterization given the established BRCA1-PALB2 interaction in homologous recombination.

The identification of Lynch syndrome mutations (PMS2, MSH2) in patients presenting with breast cancer—rather than the classically expected colorectal or endometrial cancer—illustrates the phenotypic complexity that multigene panel testing reveals. These patients require expanded surveillance protocols that their initial breast cancer diagnosis alone would not have prompted.

Given the young age at diagnosis (45.7% below age 40), screening guidelines should be adapted: women with BRCA1 variants or strong family histories may require enhanced surveillance from the late twenties or early thirties. Knowledge of BRCA1/2 status enables utilization of PARP inhibitors, platinum chemotherapy optimization, and tailored surveillance. Cascade testing of first-degree relatives—currently initiated in only a small minority of families due to cost and logistical barriers—is essential to amplify the public health benefits of each genetic diagnosis.

African genomes remain massively underrepresented (~2%) in global sequencing datasets [17]-[19]. Studies like GENCAF provide critical data that enrich variant interpretation globally, reduce misclassification, and contribute to equity in precision medicine [20]. The hereditary cancer biobank established by GENCAF at the University of Yaounde I—the first such repository in Cameroon—is positioned to generate longitudinal clinical and functional data for VUS reclassification.

Limitations

Several limitations warrant consideration. First, single-city recruitment at tertiary centers in Yaounde limits generalizability to rural and regional populations. Second, breast cancer is overrepresented (87.2%), limiting power for inferential analyses across other cancer types. Third, the absence of matched tumor sequencing prevents direct comparison of somatic and germline findings. Fourth, statistical power for subgroup analyses is limited by the sample size (N = 94). Fifth, functional validation of novel VUS was not performed. Despite these limitations, this dataset represents a foundational reference for hereditary cancer genomics in Central Africa.

5. Conclusion

This study presents a comprehensive map of hereditary cancer susceptibility in Cameroon using multigene germline sequencing. The high prevalence of BRCA1 pathogenic variants (27.7% P/LP combined yield), the substantial VUS burden across 13 genes, and the early age at diagnosis highlight the urgency of integrating genetic counseling and testing into national cancer control strategies. Expanding access, improving variant interpretation through African-specific databases, and developing local genomic capacity will be essential to achieving equitable precision oncology in the region. The GENCAF program provides a scalable model for hereditary cancer risk assessment in sub-Saharan Africa, and these findings lay the groundwork for future regional genomic epidemiology initiatives and reclassification efforts.

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Conflicts of Interest

The authors declare no conflicts of interest.

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List of Abbreviations

Abbreviation	Definition
ACMG/AMP	American College of Medical Genetics and Genomics/Association for Molecular Pathology
ATM	Ataxia-telangiectasia mutated
BRCA1/2	Breast Cancer gene 1/2
CAP	College of American Pathologists
CLIA	Clinical Laboratory Improvement Amendments
DNA	Deoxyribonucleic acid
GENCAF	Genetic Cancer Families
GATK	Genome Analysis Toolkit
LP	Likely pathogenic
MMR	Mismatch repair
NCCN	National Comprehensive Cancer Network
NGS	Next-generation sequencing
P	Pathogenic
P/LP	Pathogenic or likely pathogenic
PALB2	Partner and localizer of BRCA2
PARP	Poly(ADP-ribose) polymerase
SSA	Sub-Saharan Africa
VUS	Variant of uncertain significance