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Implementation of Genetic Counseling and Germline Multigene Testing for Hereditary Cancer in Cameroon: Insights from the GENCAF Pilot Program

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Abstract

Introduction: Sub-Saharan Africa faces a rapidly growing cancer burden, yet genetic counseling and hereditary cancer testing remain absent in most national health systems. In Cameroon, no structured oncogenetics services existed prior to 2020, despite the frequent clinical observation of early-onset cancers and strong familial clustering. This project aimed to implement and evaluate the operational outcomes of the first structured genetic counseling and multigene testing program for hereditary cancer in Cameroon. **Methods:** The Genetic Cancer Families (GENCAF) initiative was established as a collaborative North-South (University of Chicago, City of Hope) and South-South (University of Ibadan) program across three oncology centers in Yaounde. Following capacity building and a feasibility study whose results are published separately, the program proceeded to pilot germline testing using next-generation sequencing (NGS) for 29 cancer susceptibility genes in 94 patients. **Results:** The program trained Cameroon's first two certified cancer genetic counselors and 19 supporting health professionals. The feasibility and acceptability out-

comes (including knowledge-score improvement from 41% to 78%, $p < 0.001$) were measured in a prior cross-sectional cohort of 160 patients and are not outcomes of the present testing cohort of 94 patients. Operational sequencing of 94 probands identified pathogenic or likely pathogenic (P/LP) variants in 27.7%, with Breast Cancer gene 1 (BRCA1) being the most prevalent gene (19.1%). Additionally, 26.6% carried variants of uncertain significance (VUS). **Conclusion:** GENCAF demonstrates the feasibility and high acceptability of introducing oncogenetics in Cameroon. The high diagnostic yield (27.7%) confirms a substantial burden of hereditary cancer, necessitating the integration of genetic services into national cancer control plans.

Keywords

Cameroon, Hereditary Cancer, Genetic Counseling, Implementation Science, Africa, BRCA, Multigene Testing, GENCAF

1. Introduction

Cancer incidence is rising across sub-Saharan Africa, driven by demographic transitions, limited screening programs, variable treatment access, and persistent structural health system gaps [1]. While environmental exposures and infection-related cancers remain disproportionately represented, accumulating evidence demonstrates that a substantial subset of cancers in African populations arises from hereditary predispositions, especially in breast, ovarian, colorectal, prostate, and gastric cancers [2]-[4].

Yet genetic counseling and germline testing—now standard of care in high-income settings—are largely unavailable across the region [5]-[7]. Before 2020, Cameroon had no structured service for hereditary cancer counseling or testing, despite the well-recognized presence of early-onset disease, familial clustering, and suspected enrichment of pathogenic variants.

The absence of genomic data across African populations further exacerbates these disparities. Nearly 98% of global genomic datasets originate from non-African ancestry populations, leading to systematic underrepresentation and uncertainty in variant interpretation [8]-[10]. This limits evidence-based management, produces high VUS rates, and restricts the development of population-tailored clinical guidelines.

The GENCAF project was conceived as a pioneering initiative to introduce a structured oncogenetics program in Cameroon, built through North-South collaboration with the University of Chicago and City of Hope National Medical Center, and South-South partnership with the University of Ibadan, Nigeria. The feasibility and acceptability of genetic services were previously established in a dedicated cross-sectional study [11]; the present article reports on the program's capacity-building outcomes and the operational testing phase.

2. Methods

2.1. Study Design and Setting

This study forms part of the GENCAF pilot program conducted between January 2020 and December 2023, using a descriptive pilot implementation design. Primary implementation outcomes assessed included: feasibility (enrollment and counseling completion rates), workforce development (number of professionals trained and certified), fidelity (adherence to standardized counseling protocols), and initial efficacy (P/LP diagnostic yield). Ethical approval was obtained from the National Human Health Research Ethics Committee (N˚2021/12/1424/CE/CNERSH/SP). The program was implemented across three major oncology centers in Yaounde: the Yaounde General Hospital, Yaounde Central Hospital, and the NGO Chemotherapy Solidarity (SOCHIMIO), collectively accounting for an estimated 80% of cancer consultations in the capital region at this period.

2.2. Phase 1: Capacity Building and Training

To address the absence of specialized personnel in cancer genetics, GENCAF utilized a "train-the-trainer" model [12]. Two Cameroonian physicians completed a hybrid four-month intensive oncogenetics immersion program at the City of Hope National Medical Center in California, USA, becoming Cameroon's first formally trained cancer genetic counselors. Following their return in 2021, they conducted onsite training workshops for 19 local health professionals across the three participating institutions, covering pedigree construction, identification of hereditary cancer red flags, and pre/post-test counseling protocols.

A South-South reinforcement mission from the University of Ibadan, Nigeria, provided advanced training in variant classification per American College of Medical Genetics and Genomics and Association for Molecular Pathology (ACMG/AMP) guidelines, management of BRCA-associated cancer syndromes, and cascade testing strategies for at-risk family members [13].

2.3. Phase 2: Feasibility and Acceptability

The feasibility and acceptability of genetic counseling and testing services among Cameroonian cancer patients were evaluated in a prior cross-sectional study among 160 patients, the full methods and results of which have been published separately [11]. That study demonstrated near-universal willingness to undergo counseling and testing, with cost identified as the primary barrier. Its findings provided the social and ethical foundation for the operational phase described here.

2.4. Phase 3: Development of Genetic Counseling Procedures

GENCAF developed Cameroon's first clinical guidelines for hereditary cancer genetic counseling, adapted to the local context. The protocol included standardized procedures for pre-test counseling (covering cancer risk mechanisms, inheritance patterns, penetrance, and psychosocial implications), three-generation pedigree construction, informed consent in both French and English, and saliva sample

collection using Color Genomics collection kits allowing room-temperature storage without cold-chain logistics.

2.5. Phase 4: Operational Implementation of Multigene Panel Testing

Between May 2022 and December 2023, 94 patients were enrolled for germline multigene panel testing. Referral criteria for suspected hereditary cancer included at least one of the following: 1) breast or ovarian cancer at age ≤ 45 years; 2) triple-negative breast cancer at any age; 3) two or more first- or second-degree relatives with breast, ovarian, colorectal, gastric, or prostate cancer; 4) bilateral or multicentric breast cancer; 5) male breast cancer; or 6) personal or family history suggestive of a recognized hereditary syndrome (Lynch syndrome, Li-Fraumeni syndrome, familial adenomatous polyposis). Family history of cancer was defined as at least one first- or second-degree relative with a documented cancer diagnosis. Additional eligibility criteria included: 2) completion of pre-test genetic counseling; and 3) successful saliva-based DNA collection and valid sequencing results.

Saliva samples were shipped to Color Genomics Laboratory (Burlingame, California, USA), a College of American Pathologist-accredited, Clinical Laboratory Improvement Amendments-certified laboratory, for NGS analysis using a 29-gene hereditary cancer panel. Sequencing was performed at minimum coverage >20× for >99% of targeted regions. Variants were classified per ACMG/AMP guidelines into P, LP, VUS, likely benign, or benign. Copy-number variant (CNV) detection was performed using depth-of-coverage algorithms. Pathogenic or likely pathogenic variants in genes other than BRCA1/2 were confirmed by Sanger sequencing. Post-test counseling for P/LP and VUS carriers included result interpretation, cancer risk discussion, enhanced surveillance recommendations based on National Comprehensive Cancer Network (NCCN) guidelines [14], and cascade testing guidance.

2.6. Data Collection and Statistical Analysis

Data were collected on standardized questionnaire forms and entered into a secure REDCap database. Descriptive statistics were used to summarize baseline characteristics and genetic findings. Pre- and post-counseling knowledge scores were compared using paired t-tests. Statistical significance was set at $p < 0.05$. Analyses were performed using SPSS version 23.

3. Results

3.1. Capacity Building Outcomes

The GENCAF program established Cameroon's first multidisciplinary cancer genetics team. Two physicians who completed intensive training in the United States became the country's first formally certified cancer genetic counselors, providing clinical genetic counseling services from late 2020. These counselors subsequently trained 19 additional health professionals (12 oncology residents, 5 nurses, 2 gen-

eral practitioners) across the three participating institutions.

By the end of 2023, the GENCAF team had conducted 96 genetic counseling sessions, constructed 90 detailed three-generation pedigrees, and established standardized counseling protocols. Capacity-building efforts extended to educational presentations at national oncology conferences and training sessions for medical students.

Of 112 patients who underwent pre-test genetic counseling between May 2022 and December 2023, 94 proceeded to multigene germline testing (84%). Reasons for non-testing included the fear of the results. Of the 96 counseling sessions reported, 2 involved patients who ultimately declined testing. All 94 tested patients yielded valid sequencing results (**Figure 1**).

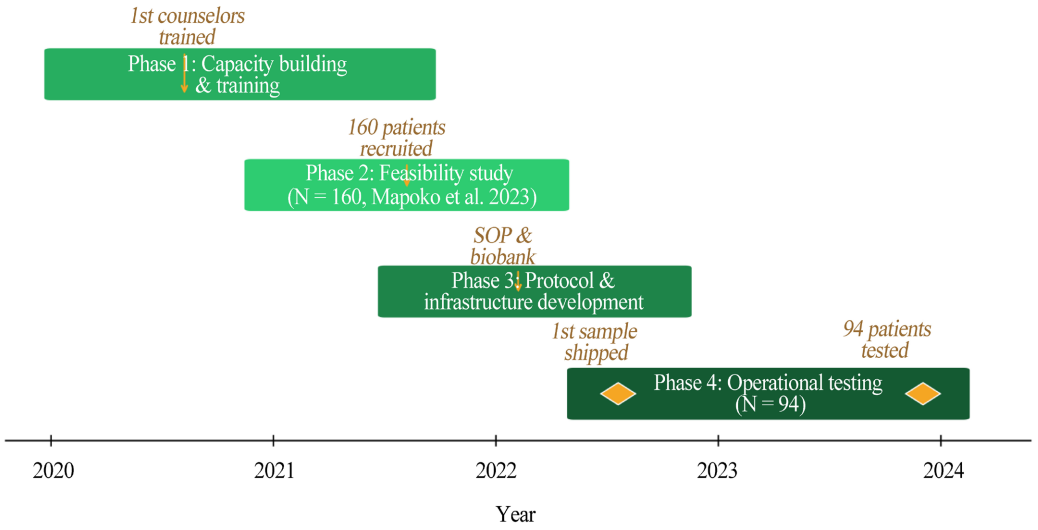


Figure 1. GENCAF program implementation timeline and key milestones (2020-2023). Phases shown as horizontal bars; diamonds indicate key milestones. GENCAF: Genetic Counseling and Cancer Testing in Africa; SOP: standard operating procedure.

3.2. Feasibility and Acceptability

The feasibility and acceptability outcomes (including knowledge-score improvement from 41% to 78%, $p < 0.001$) were measured in a prior cross-sectional cohort of 160 patients and are not outcomes of the present testing cohort of 94 patients [11]. That study documented near-universal willingness to undergo genetic counseling (98%) and testing (96% - 97.5%), high motivation driven by desire to protect family members (81%) and understand cancer origin (84%), and cost as the predominant barrier (91%). These findings confirmed the social readiness of the Cameroonian population for hereditary cancer genetic services and provided the foundational justification for the operational testing phase.

3.3. Genetic Testing Outcomes (N = 94)

Between May 2022 and December 2023, 94 patients underwent germline multigene panel testing. The median age was 45 years (IQR 37 - 53); 95.7% were female.

Cancer types included breast (87.2%, n = 82), colorectal (5.3%, n = 5), gastric (3.2%, n = 3), ovarian (2.1%, n = 2), renal (1.1%, n = 1), and prostate (1.1%, n = 1).

Table 1. Pathogenic and likely pathogenic variants by gene amongst mutation genes and amongst the whole population (N = 94).

Gene	n (P/LP)	% gene	% 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, pancreatic risk
BRCA2	2	7.7%	2.1%	HBOC—breast, ovarian, 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 P/LP	26	100%	27.7%	

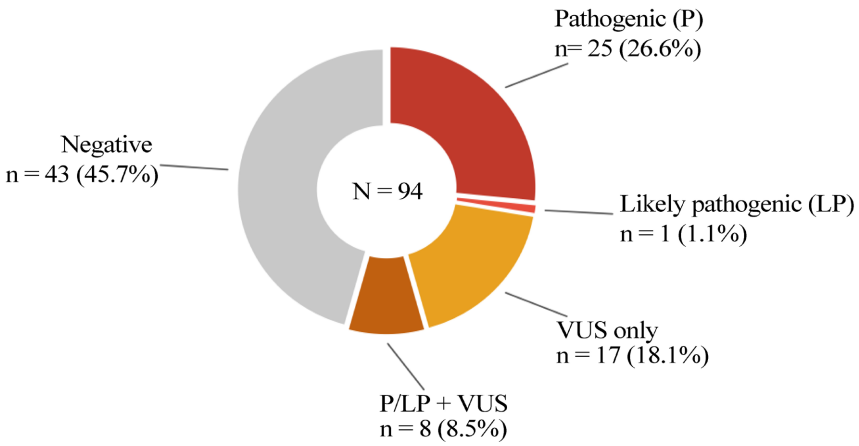


Figure 2. Variant classification distribution (n = 94).

Table 1 shows the pathogenic and likely pathogenic variants by gene amongst mutation genes and amongst the whole population, and the hereditary syndromes. Among the 94 tested patients, 57 (60.6%) reported a positive family history of cancer (at least one first- or second-degree relative with a documented malignancy). Early-onset disease was common, with 43 patients (45.7%) diagnosed before age 45. The primary indication for testing was breast cancer (n = 82, 87.2%), followed by colorectal (n = 5, 5.3%), gastric (n = 3, 3.2%), ovarian (n = 2, 2.1%), renal (n = 1, 1.1%), and prostate (n = 1, 1.1%) cancer. The sequencing success rate was 100%. P/LP variants were identified in 26 patients (27.7%; 25 pathogenic, 1 likely pathogenic). VUS were detected in 25 patients (26.6%). Overall, 43 patients (45.7%) carried at least one variant of any classification. Eight patients carried both a P/LP variant and a VUS in different genes. Figure 2 and Figure 3 summa-

rize overall the variants distribution and pathogenic/likely pathogenic variants by gene.

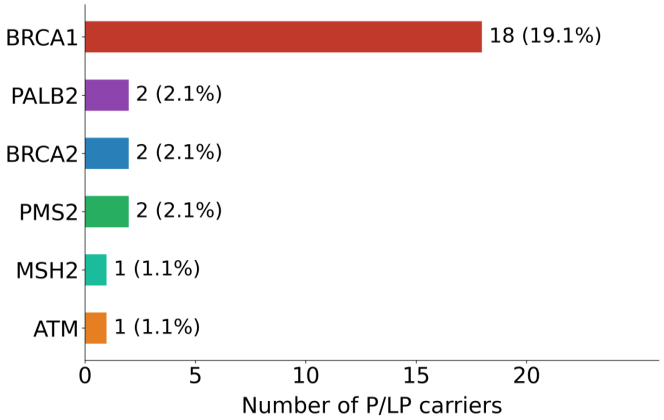


Figure 3. Distribution of Pathogenic/Likely Pathogenic carriers per gene (26 carriers).

BRCA1 was the most frequently mutated gene (19.1% of the cohort; 17 pathogenic + 1 likely pathogenic), followed by PALB2 (2.1%), BRCA2 (2.1%), PMS2 (2.1%), MSH2 (1.1%), and ATM (1.1%). **Figure 4** and **Figure 5** illustrate the pedigree and the results of a 57-year-old female with risk factors of breast cancer and a positive family history who presented an ATM mutation.

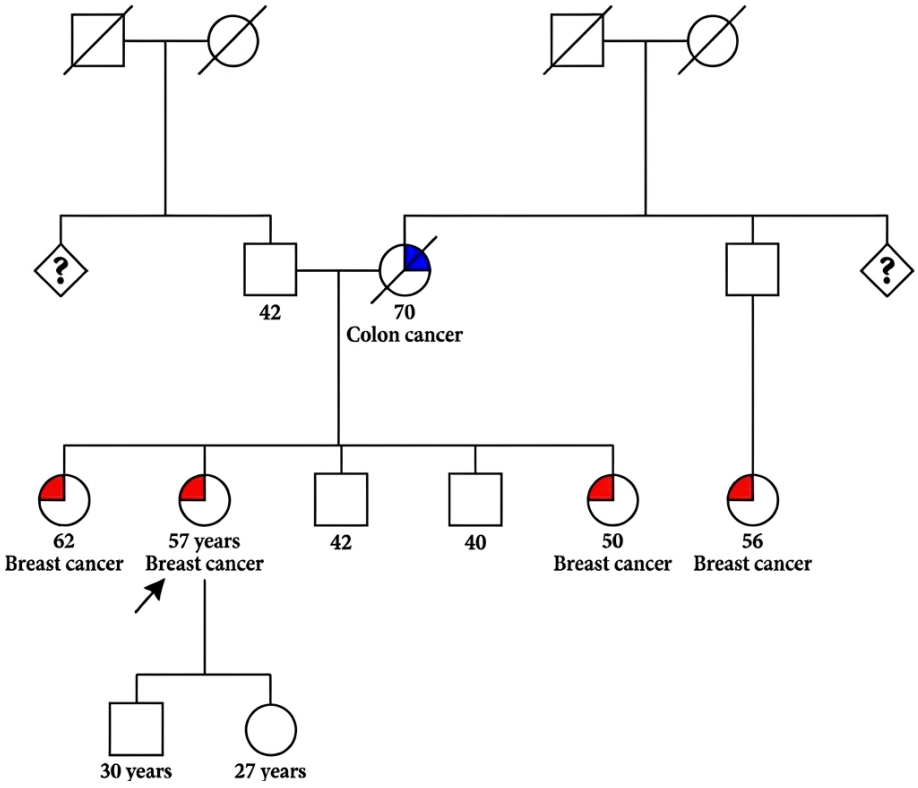
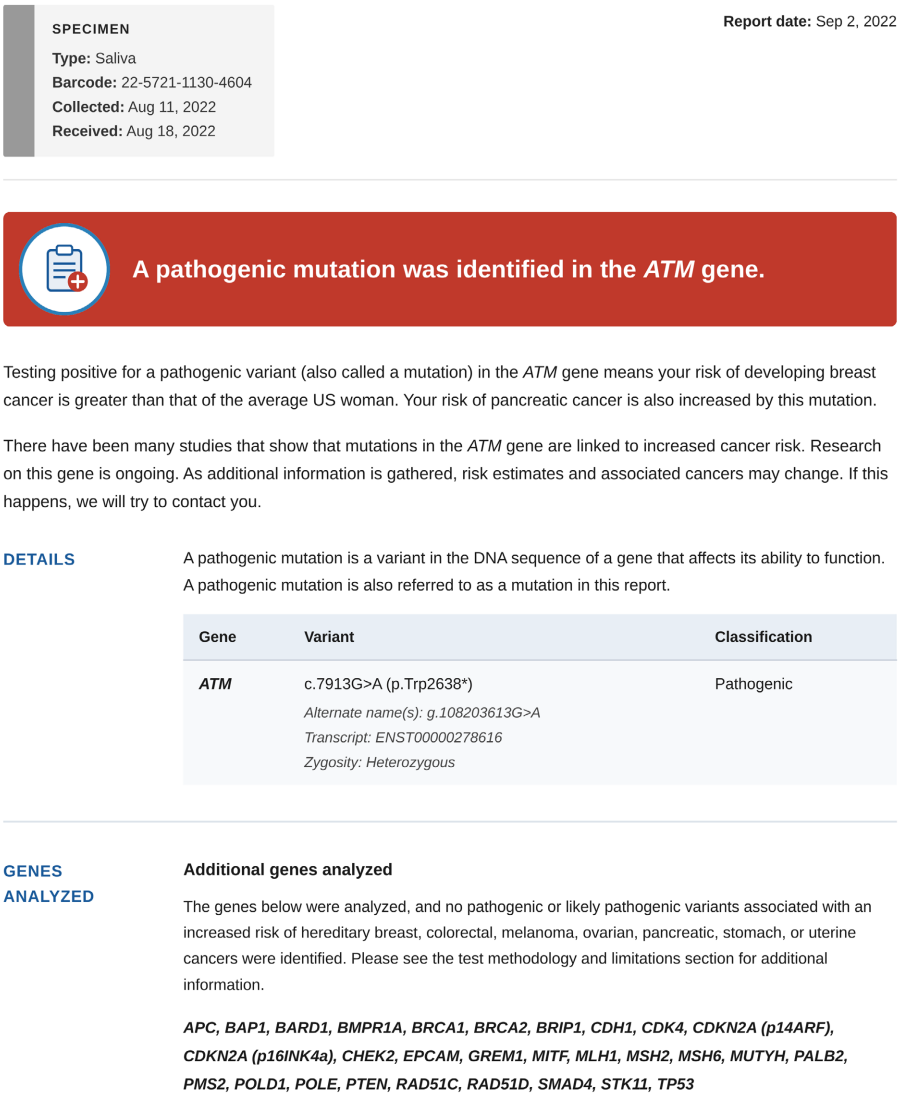


Figure 4. Pedigree of a 57-year-old female with risk factors of breast cancer and a positive family history who presented an ATM mutation.



SPECIMEN
Type: Saliva
Barcode: 22-5721-1130-4604
Collected: Aug 11, 2022
Received: Aug 18, 2022

Report date: Sep 2, 2022

A pathogenic mutation was identified in the *ATM* gene.

Testing positive for a pathogenic variant (also called a mutation) in the *ATM* gene means your risk of developing breast cancer is greater than that of the average US woman. Your risk of pancreatic cancer is also increased by this mutation.

There have been many studies that show that mutations in the *ATM* gene are linked to increased cancer risk. Research on this gene is ongoing. As additional information is gathered, risk estimates and associated cancers may change. If this happens, we will try to contact you.

DETAILS

A pathogenic mutation is a variant in the DNA sequence of a gene that affects its ability to function. A pathogenic mutation is also referred to as a mutation in this report.

Gene	Variant	Classification
ATM	c.7913G>A (p.Trp2638*) *Alternate name(s): g.108203613G>A* *Transcript: ENST00000278616* *Zygosity: Heterozygous*	Pathogenic

GENES ANALYZED

Additional genes analyzed

The genes below were analyzed, and no pathogenic or likely pathogenic variants associated with an increased risk of hereditary breast, colorectal, melanoma, ovarian, pancreatic, stomach, or uterine cancers were identified. Please see the test methodology and limitations section for additional information.

APC, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A (p14ARF), CDKN2A (p16INK4a), CHEK2, EPCAM, GREM1, MITF, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53

Figure 5. Genetic testing result of a 57-year-old female with risk factors of breast cancer and a positive family history who presented an ATM mutation.

4. Discussion

The GENCAF pilot program represents an operational effort to integrate hereditary cancer genetic counseling and multigene germline testing into clinical oncology services in Cameroon. The present findings confirm that such an initiative is both feasible and highly accepted in a resource-limited health system, while revealing a substantial burden of hereditary cancer susceptibility variants.

A key strength of GENCAF was its sequential, capacity-building approach. Rather than relying exclusively on external expertise, the program invested in creating lasting local infrastructure through the "train-the-trainer" model, aligned with implementation science frameworks emphasizing local ownership [12]. The South-South collaboration with Nigeria further reinforced this model, demonstrating the potential for regional knowledge exchange across African countries facing similar challenges [13]. The successful training of 19 additional health pro-

fessionals extended the program's reach, creating a broader network of healthcare providers with basic genetic literacy.

The high acceptability of genetic services documented in the prior feasibility study [11] substantially exceeds early acceptability levels reported in several high-income countries during initial implementation phases [15]. This challenges assumptions that genetic testing may face cultural resistance in African settings. The dominant family-oriented motivation—to protect children from cancer—presents a genuine opportunity for cascade testing programs, though the gap between expressed interest and actual cascade testing completion highlights implementation challenges that must be addressed.

The P/LP yield of 27.7% substantially exceeds typical rates of 10% - 15% in unselected cancer populations in high-income countries [2]-[4] [16], reflecting clinical enrichment toward early-onset disease and positive family history. BRCA1 dominated the mutational landscape (19.1%), consistent with global data showing BRCA1 predominance among women of African ancestry [2]-[4]. The identification of non-BRCA genes—PALB2, PMS2, MSH2, and ATM—validates the use of multigene panel testing rather than single-gene approaches. Lynch syndrome genes (PMS2, MSH2) carry implications beyond breast cancer, requiring colorectal and endometrial cancer surveillance [17].

The VUS rate of 26.6% is consistent with reports from other African populations [8] [10] [18]-[20], reflecting the fundamental genomic divide in global databases. Resolving these VUS requires substantial investments in African genomic research and population-specific variant databases.

Limitations

Several limitations warrant consideration. First, the study was conducted at urban tertiary centers in Yaounde, potentially limiting generalizability to rural populations. Second, the sample size (N = 94) limits statistical power for comparing variant rates across cancer types. Third, the use of an international laboratory introduces delays and costs not representative of a mature national program. Fourth, the absence of local sequencing capacity limits the long-term sustainability of the program without external funding.

5. Conclusion

The GENCAF pilot program demonstrates that cancer genetic counseling and multigene panel testing can be successfully implemented in Cameroon through strategic capacity building, international partnerships, and culturally adapted service delivery. A diagnostic yield of 27.7% in this clinically selected, predominantly breast-cancer cohort confirms a substantial hereditary cancer burden in the patients most likely to benefit from genetic services. These figures should not be extrapolated to the general Cameroonian cancer population without population-based data. The establishment of a trained national workforce makes this a pioneering initiative for Central Africa. Integration of genetic services into Cameroon's national cancer control plan, supported by sustainable financing and ex-

panded counseling capacity, represents an essential next step toward equitable precision oncology in the region.

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Conflicts of Interest

The authors declare no conflicts of interest.

References

[1] Morhason-Bello, I.O., Odedina, F., Rebbeck, T.R., Harford, J., Dangou, J., Denny, L., *et al.* (2013) Challenges and Opportunities in Cancer Control in Africa: A Perspective from the African Organisation for Research and Training in Cancer. *The Lancet Oncology*, **14**, e142-e151. https://doi.org/10.1016/s1470-2045(12)70482-5

[2] Fackenthal, J.D. and Olopade, O.I. (2007) Breast Cancer Risk Associated with BRCA1 and BRCA2 in Diverse Populations. *Nature Reviews Cancer*, **7**, 937-948. https://doi.org/10.1038/nrc2054

[3] Adedokun, B., Zheng, Y., Ndom, P., Gakwaya, A., Makumbi, T., Zhou, A.Y., *et al.* (2020) Prevalence of Inherited Mutations in Breast Cancer Predisposition Genes among Women in Uganda and Cameroon. *Cancer Epidemiology, Biomarkers & Prevention*, **29**, 359-367. https://doi.org/10.1158/1055-9965.epi-19-0506

[4] Hayat, M., Chen, W.C., Brandenburg, J., Babb de Villiers, C., Ramsay, M. and Mathew, C.G. (2021) Genetic Susceptibility to Breast Cancer in Sub-Saharan African Populations. *JCO Global Oncology*, **7**, 1462-1471. https://doi.org/10.1200/go.21.00089

[5] Rotimi, S.O., Rotimi, O.A. and Salhia, B. (2021) A Review of Cancer Genetics and Genomics Studies in Africa. *Frontiers in Oncology*, **10**, Article 606400. https://doi.org/10.3389/fonc.2020.606400

[6] Bentley, A.R., Callier, S.L. and Rotimi, C.N. (2020) Evaluating the Promise of Inclusion of African Ancestry Populations in Genomics. *npj Genomic Medicine*, **5**, Article No. 5. https://doi.org/10.1038/s41525-019-0111-x

[7] Ginsburg, O. and Narod, S.A. (2018) Clinical Cancer Genetics in a Lower-Middle Income Country: Considerations for Policymaking. *JCO Global Oncology*, **4**, JGO.18.00081.

[8] Rotimi, C.N., Bentley, A.R., Doumatey, A.P., Chen, G., Shriner, D. and Adeyemo, A. (2017) The Genomic Landscape of African Populations in Health and Disease. *Human Molecular Genetics*, **26**, R225-R236. https://doi.org/10.1093/hmg/ddx253

[9] Fatumo, S., Chikowore, T., Choudhury, A., Ayub, M., Martin, A.R. and Kuchenbaecker, K. (2022) A Roadmap to Increase Diversity in Genomic Studies. *Nature Medicine*, **28**, 243-250. https://doi.org/10.1038/s41591-021-01672-4

[10] Popejoy, A.B. and Fullerton, S.M. (2016) Genomics Is Failing on Diversity. *Nature*,

538, 161-164. https://doi.org/10.1038/538161a

[11] Esson Mapoko, B.S., Chi Ndi, K., Tabola, L., Mouaye, V., Douanla, P., Nsangou, N., *et al.* (2023) Feasibility of Cancer Genetic Counselling and Screening in Cameroon: Perceived Benefits and Barriers. *eCancerMedicalScience*, **17**, Article 1588. https://doi.org/10.3332/ecancer.2023.1588

[12] Proctor, E., Silmere, H., Raghavan, R., Hovmand, P., Aarons, G., Bunger, A., *et al.* (2011) Outcomes for Implementation Research: Conceptual Distinctions, Measurement Challenges, and Research Agenda. *Administration and Policy in Mental Health and Mental Health Services Research*, **38**, 65-76. https://doi.org/10.1007/s10488-010-0319-7

[13] Binagwaho, A., Kyamanywa, P., Farmer, P.E., Nuthulaganti, T., Umubyeyi, B., Nyemazi, J.P., *et al.* (2013) The Human Resources for Health Program in Rwanda—A New Partnership. *New England Journal of Medicine*, **369**, 2054-2059. https://doi.org/10.1056/nejmsr1302176

[14] Daly, M.B., Pal, T., Maxwell, K.N., Churpek, J., Kohlmann, W., AlHilli, Z., *et al.* (2023) NCCN Guidelines® Insights: Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic, Version 2.2024. *Journal of the National Comprehensive Cancer Network*, **21**, 1000-1010. https://doi.org/10.6004/jnccn.2023.0051

[15] Braithwaite, D., Emery, J., Walter, F., Prevost, A.T. and Sutton, S. (2004) Psychological Impact of Genetic Counseling for Familial Cancer: A Systematic Review and Meta-Analysis. *JNCI Journal of the National Cancer Institute*, **96**, 122-133. https://doi.org/10.1093/jnci/djh017

[16] Couch, F.J., Shimelis, H., Hu, C., Hart, S.N., Polley, E.C., Na, J., *et al.* (2017) Associations between Cancer Predisposition Testing Panel Genes and Breast Cancer. *JAMA Oncology*, **3**, 1190-1196. https://doi.org/10.1001/jamaoncol.2017.0424

[17] Hampel, H., Pearlman, R., Beightol, M., Zhao, W., Jones, D., Frankel, W.L., *et al.* (2018) Assessment of Tumor Sequencing as a Replacement for Lynch Syndrome Screening and Current Molecular Tests for Patients with Colorectal Cancer. *JAMA Oncology*, **4**, 806-813. https://doi.org/10.1001/jamaoncol.2018.0104

[18] Ndugga-Kabuye, M.K. and Issaka, R.B. (2019) Inequities in Multi-Gene Hereditary Cancer Testing: Lower Diagnostic Yield and Higher VUS Rate in Individuals Who Identify as Hispanic, African or Asian and Pacific Islander as Compared to European. *Familial Cancer*, **18**, 465-469. https://doi.org/10.1007/s10689-019-00144-6

[19] Ricks-Santi, L., McDonald, J.T., Gold, B., Dean, M., Thompson, N., Abbas, M., *et al.* (2017) Next Generation Sequencing Reveals High Prevalence of BRCA1 and BRCA2 Variants of Unknown Significance in Early-Onset Breast Cancer in African American Women. *Ethnicity & Disease*, **27**, 169-178. https://doi.org/10.18865/ed.27.2.169

[20] Manrai, A.K., Funke, B.H., Rehm, H.L., Olesen, M.S., Maron, B.A., Szolovits, P., *et al.* (2016) Genetic Misdiagnoses and the Potential for Health Disparities. *New England Journal of Medicine*, **375**, 655-665. https://doi.org/10.1056/nejmsa1507092

List of Abbreviations

Abbreviation	Definition
ACMG/AMP	American College of Medical Genetics and Genomics/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 Cancer Families
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
REDCap	Research Electronic Data Capture
SSA	Sub-Saharan Africa
VUS	Variant of uncertain significance