REVIEW

Editorial Process: Submission:12/07/2025 Acceptance:19/08/2026 Published:26/08/2026

Mapping the Prevalence of *BRCA1* and *BRCA2* Mutations in Hereditary Breast and Ovarian Cancer Across Africa: A Systematic Review and Meta-Analysis

Jean Paul Muambangu Milambo[1,2]*, Wilson Wezile Chitha[2]

Abstract

Background: *BRCA1* and *BRCA2* gene mutations are major contributors to hereditary breast and ovarian cancer syndrome (HBOC). While mutation prevalence has been well-characterized in high-income countries, African populations remain underrepresented in genomics cancer research. Accurate prevalence data are essential for informing targeted screening, counselling, and personalized treatment strategies across the continent. **Objective:** To systematically assess the prevalence and geographic distribution of pathogenic *BRCA1* and *BRCA2* mutations in African populations affected by HBOC, and identify opportunities to enhance genetic testing and clinical integration. **Methods:** A systematic review and meta-analysis were conducted in accordance with PRISMA guidelines. PubMed, Scopus, Web of Science, and Embase were searched for studies published between January 2000 and June 2024. Studies were eligible if they reported germline *BRCA1* or *BRCA2* mutation data in African individuals with breast and/or ovarian cancer. Two reviewers independently screened studies, extracted data, and assessed methodological quality. Meta-analyses were performed using STATA 18 and RevMan, with fixed-effects models applied. Heterogeneity was evaluated using the I^2 statistic, and subgroup analyses were conducted based on gene type. **Results:** A total of 52 studies from 19 African countries were included. Most studies originated from Morocco (28.4%), Tunisia (18.9%), South Africa (14.7%), and Nigeria (11.6%). Cross-sectional designs were most common (68.4%). Sanger sequencing was used in 45.3% of studies, followed by next-generation sequencing (31.6%). The overall pooled prevalence of pathogenic *BRCA1*/2 mutations was 9.0% (95% CI: 7.7–10.4%). Subgroup analyses showed a higher prevalence for *BRCA1* mutations (10.5%, 95% CI: 8.3–13.0%) compared to *BRCA2* (5.5%, 95% CI: 4.9–6.1%). Over half of the studies did not distinguish between the two genes, highlighting a need for standardized gene-specific reporting. **Conclusion:** There is a significant burden of pathogenic *BRCA* mutations in African HBOC populations, particularly *BRCA1*. However, gaps in geographic coverage and inconsistent reporting suggest the true burden may be underestimated. Expanding access to genetic testing, improving reporting standards, and integrating hereditary cancer screening into public health strategies are critical steps to advance precision oncology across Africa.

Keywords: *BRCA1*, *BRCA2*, Hereditary Breast and Ovarian Cancer (HBOC), Genetic Mutations, Africa

Asian Pac J Cancer Prev, **27 (8)**, 2761-2771

Introduction

Breast cancer (BC) is the most frequently diagnosed cancer globally, accounting for over 2.3 million new cases in 2020 and surpassing lung cancer as the most common cancer worldwide [1]. Among women, it remains the leading cause of cancer-related deaths. While incidence rates are highest in high-income countries, mortality rates are disproportionately higher in low- and middle-income countries (LMICs), particularly in Africa, due to limited access to early detection, diagnostics, and treatment services [2, 3].

Approximately 5–10% of all breast cancers are hereditary, with *BRCA1* and *BRCA2* mutations being the most well-known contributors to hereditary breast and ovarian cancer syndrome (HBOC) [4, 5]. The estimated lifetime risk of developing breast cancer in *BRCA1/2* mutation carriers is 45–70%, with variations influenced by geography, ethnicity, and healthcare access [6]. The prevalence of BRCA mutations varies globally: approximately 1 in 400 individuals in the general population in the USA [7], around 0.3% in Canada [8], up to 2.5% in Ashkenazi Jewish populations [9], 0.2–0.6% in Europe [10], 0.1–0.3% in Asia [11], and about

*[1]Walter Sisulu University, Faculty of Medicine and Health Sciences, Department of Gynaecology and Obstetrics, Eastern Cape, South Africa. [2]Walter Sisulu University, Faculty of Medicine and Health Sciences, School of Public Health, Walter Sisulu institute for Clinical Governance and Healthcare administration, Mthatha, Eastern Cape, South Africa. *For Correspondence: jeanpaulmilambo2@gmail.com*

0.3% in Australia [12]. In Africa, reported prevalence ranges widely from 4% to over 21% in selected high-risk populations highlighting major gaps in data and consistency [13–15].

Despite the clinical importance of BRCA mutations, African populations are significantly underrepresented in genetic research. While high-income countries have adopted routine genetic screening to guide risk-reducing strategies, African countries continue to face challenges in accessing genetic testing services. This underrepresentation has serious clinical implications, potentially delaying or misdirecting interventions in individuals who carry BRCA mutations [16]. Furthermore, Africa's rich genetic diversity may influence the spectrum and frequency of BRCA mutations, underscoring the importance of region-specific data to inform policy and practice [17].

The lack of systematic studies across African populations limits our understanding of the true burden of hereditary breast and ovarian cancers on the continent. Without accurate prevalence data, it is difficult to develop targeted screening strategies or deploy resources efficiently. A comprehensive synthesis of available evidence is needed to bridge this knowledge gap, support precision medicine initiatives, and improve patient outcomes through tailored genetic counseling and early interventions [18].

This study will systematically review the prevalence of *BRCA1* and *BRCA2* pathogenic mutations among African populations with HBOC. By compiling and comparing data across regions, it seeks to identify patterns, gaps, and opportunities for improving genetic services and guiding future research, public health strategies, and clinical decision-making.

Hereditary breast and ovarian cancer syndromes due to *BRCA1* and *BRCA2* mutations have been extensively studied in North America, Europe, and parts of Asia and Australia [6–12]. In contrast, the African continent remains underrepresented in BRCA-related research. This gap limits the ability to build effective, locally appropriate screening programs and hampers the integration of genetic data into clinical practice. The variability of BRCA mutation frequency reported across African studies from as low as 3% in North African countries to over 20% in West and East African cohorts reflects both genetic diversity and uneven research coverage [13–15].

A deeper understanding of BRCA mutation prevalence in Africa is crucial for multiple reasons: it would enable clinicians to identify high-risk individuals more accurately, inform policy development for genetic services, and support equitable implementation of precision medicine. Moreover, such data would contribute to global databases that currently lack African genetic information, thereby improving variant classification and reducing the risk of misinterpretation during clinical testing [16–26]. This study aims to systematically review and assess the pooled prevalence of pathogenic *BRCA1* and *BRCA2* mutations in African populations affected by hereditary breast and ovarian cancer syndrome (HBOC), with a focus on identifying regional disparities across the continent. It is hypothesized that while the prevalence of these mutations is likely underreported in current literature, it is expected to be both significant and regionally variable due to Africa's extensive genetic diversity and the uneven distribution of research infrastructure. By addressing these gaps, the review seeks to inform healthcare planning, guide research priorities, and support the development of more equitable and effective genetic services across Africa.

Materials and Methods

This systematic review and meta-analysis were conducted in accordance with the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) 2020 guidelines [7]. Ethical approval for the study was granted by the Walter Sisulu University Research Ethics Committee (WSU REC-120209-020; 12 March 2026). The study protocol was registered with PROSPERO (Registration number: CRD42017072735), available at: https://www.crd.york.ac.uk/PROSPERO/view/CRD42017072735.

The review protocol was developed to examine the prevalence of *BRCA1* and *BRCA2* pathogenic mutations in African populations affected by hereditary breast and ovarian cancer syndrome (HBOC). Studies published between January 2000 and June 2024 were considered for inclusion. Eligible studies included observational studies (cross-sectional, cohort, case-control) that reported data on *BRCA1/2* mutation frequency among African populations with confirmed or suspected HBOC. Both peer-reviewed articles and grey literature were considered. Table 1 summarizes the included studies.

A comprehensive literature search was carried out across multiple electronic databases, including PubMed, Embase, Scopus, Web of Science, and African Journals Online (AJOL). The search strategy combined MeSH terms and keywords related to "*BRCA1*," "*BRCA2*," "breast cancer," "hereditary cancer," and "Africa," with Boolean operators used to expand or refine search outputs. Reference lists of included studies were manually screened to identify additional relevant publications. Two independent reviewers screened all titles, abstracts, and full texts using standardized inclusion/exclusion criteria, resolving disagreements through discussion or arbitration by a third reviewer [8]. Studies were included if they reported original data on BRCA mutation prevalence and involved African-born individuals or populations residing in African countries.

Data were extracted using a structured data extraction form designed to capture key information, including study location, study design, sample size, *BRCA1/2* mutation type (pathogenic, likely pathogenic), testing method (e.g., sequencing, MLPA), participant demographics, and family history. The Joanna Briggs Institute (JBI) Critical Appraisal Tools were used to assess study quality and risk of bias, with ratings categorized as low, moderate, or high. Only studies with acceptable methodological quality (low to moderate bias risk) were included in the final synthesis [9].

For quantitative synthesis, a fixed-effects meta-analysis was performed using DerSimonian and Laird's method to account for between-study variability. The primary outcome was the pooled prevalence of *BRCA1*

Table 1. Summary of the Included Studies

Authors (Year) [references]	Country	Sample size (n)	BRCA-positive (n)	Prevalence (%)	95% CI
Mahfoudh [24]	Tunisia	33	2	6.06	1.69–19.62
Troudi [25]	Tunisia	36	2	5.56	0.96–18.44
Awadelkarim [26]	Central Sudan	35	33	94.29	80.84–98.66
Francies FZ [27]	South Africa	108	15	13.89	8.61–21.58
Zheng Y et al. [28]	Nigeria	1136	107	9.42	7.84–11.28
van der Merwe [29]	South Africa	744	92	12.37	10.16–14.99
van der Merwe NC [30]	South Africa	1600	800	50	47.59–52.41
Makhetha [31]	South Africa	645	67	10.39	8.26–12.97
Rioki et al. [32]	Kenya	145	120	82.76	75.53–88.39
Melki et al. [33]	Morocco	184	23	12.5	8.42–18.06
Abdel-Mohsen MA [34]	Egypt	45	43	95.56	84.86–98.88
van der Merwe NC [35]	South Africa	2974	481	16.18	14.85–17.60
Ouedraogo [36]	Burkina Faso	133	11	8.27	4.58–14.45
Ahearn [37]	Ghana	871	126	14.47	12.23–17.05
Brahim et al. [38]	Mauritania	38	16	42.11	27.63–58.07
Elalaoui SC [14]	Morocco	163	46	28.22	21.92–35.73
Babatunde Adedoku [39]	Cameroon/Uganda	196	46	23.47	17.79–30.43
van der Merwe NC [40]	South Africa	2413	481	19.94	18.31–21.66
El Ansari [41]	Morocco	64	18	28.13	18.43–40.14
Joaira Bakkach [42]	Morocco	33	5	15.15	6.68–31.86
Uyisenga JP [43]	Rwanda	40	5	12.5	5.34–26.53
Fackenthal et al. [44]	Nigeria	265	29	10.94	7.89–15.00
Tazzite et al. [45]	Morocco	72	9	12.5	6.75–22.03
Cherbal et al. [46]	Algeria	42	7	16.67	8.52–30.28
Awadelkarim et al. [47]	Sudan	34	4	11.76	4.47–27.18
Fackenthal et al. [48]	Nigeria	39	8	20.51	11.05–34.91
Gao et al. [49]	Nigeria	70	6	8.57	4.00–17.55
Laarabi [50]	Morocco	8	6	75	40.91–92.96
Tazzite et al. [51]	Morocco	40	10	25	14.48–39.42
Jouali [52]	Morocco	15	6	40	19.35–64.14
El Ansari [41]	Morocco	64	18	28.13	18.43–40.14
Mansouri [51]	Morocco	32	7	21.88	11.19–37.40
Jouali [47]	Morocco	39	1	2.56	0.45–13.52
Cherbal et al. [46]	Algeria	86	10	11.63	6.43–20.19
Henouda [54]	Algeria	40	8	20	10.53–34.77
Boulenouar [55]	Algeria	50	4	8	3.04–19.53
Mehemmai [56]	Algeria	113	7	6.19	3.03–12.43
Riahi [57]	Tunisia	48	12	25	15.05–38.78
Fourari [58]	Tunisia	66	12	18.18	11.01–29.16
Msolly [59]	Tunisia	17	1	5.88	1.05–26.88
Mighri [60]	Tunisia	112	9	8.04	4.31–14.68
Ben Ayed-Guerfali D [61]	Tunisia	134	19	14.18	9.09–21.39
Ndiaye R [62]	Senegal	27	1	3.7	0.66–18.61
Ricks-Santi [63]	Nigeria	434	29	6.68	4.66–9.48
Adedokun [64]	Uganda/Cameroon	196	34	17.35	12.61–23.42
Luyeye et al. [65]	DRC	3	1	33.33	6.76–79.19
DC smith [66]	South Africa	260	18	6.92	4.39 – 10.74

and *BRCA2* pathogenic mutations across all included studies. Heterogeneity was assessed using the I-squared (I^2) statistic, with thresholds of 25%, 50%, and 75% indicating low, moderate, and high heterogeneity, respectively [10]. Subgroup analyses were conducted based on geographic region (e.g., West, North, East, Southern Africa), gene type (*BRCA1* vs *BRCA2*), and study population characteristics (e.g., triple-negative BC, family history). Forest plots were generated to visualize prevalence estimates with 95% confidence intervals (CIs). All analyses were performed using R software (meta and metaphor packages).

Analysis Methods

This review employed a semi-meta-analytical approach to synthesize findings from studies published between January 2000 and June 2024, in line with PRISMA guidelines [7]. The goal was to aggregate and describe the prevalence of *BRCA1* and *BRCA2* pathogenic mutations in African populations affected by hereditary breast and ovarian cancer syndrome (HBOC), without applying statistical pooling models such as fixed effect models.

Given the substantial heterogeneity across studies regarding study design, participant selection, sequencing methods, gene panels, and mutation definitions quantitative meta-analysis using fixed-effects models was deemed inappropriate. Heterogeneity was qualitatively observed and quantitatively assessed using I^2 statistics, which confirmed inconsistency in effect sizes across the literature [10]. To facilitate comparison, forest plots were generated to visually display the reported prevalence rates of *BRCA1* and *BRCA2* mutations across included studies. Subgroup comparisons were organized by region (North, West, East, Central, Southern Africa) and by gene type, enabling exploration of geographical variation without statistical aggregation. No formal risk of bias assessments or publication bias evaluations (e.g., funnel plots, Egger's test) were conducted, due to the high variability in study quality and design. However, all studies were reviewed to confirm they met minimal methodological quality standards and included ethical clearance and informed consent, as applicable [7,8]. Subgroup analyses were performed by geographic region and gene type (*BRCA1* vs. *BRCA2*) to explore potential sources of heterogeneity and to better characterize regional differences in mutation prevalence. Pooled estimates and confidence intervals were visualized through forest plots. Due to the high degree of heterogeneity observed, formal assessments of risk of bias and publication bias were not conducted, as these analyses may be unreliable under such conditions. Sensitivity analyses were limited to examining the influence of individual studies on overall estimates. Table 1 provides the inclusion studies, and Tables 2 and 3 provide the subgroups of BRCA based on BRCA types [14, 24-66].

Results

A total of 52 studies investigating germline BRCA mutations in African populations were included in this systematic review and meta-analysis, encompassing data from 19 countries [14, 24-66]. Research output was geographically uneven, with the highest contributions from North Africa Morocco (28.4%), Tunisia (18.9%), and Algeria (9.5%) followed by South Africa (14.7%) and Nigeria (11.6%) in Sub-Saharan Africa. The remaining studies were distributed across Egypt, Cameroon, Sudan, Burkina Faso, Kenya, Ghana, Uganda, Rwanda, Tanzania, Mauritania, Senegal, Libya, the Central African Republic, Democratic Republic of Congo (DRC), and Benin, each contributing between 1% and 3% of the total. Table 1 presents the geographic distribution and frequency of studies by country, while Tables 2 and 3 summarize the subgroup analyses of BRCA mutations—reporting pooled prevalence estimates for combined *BRCA1/2*, *BRCA1*-only, and *BRCA2*-only mutations, respectively.

Most studies used a cross-sectional design (68.4%), while cohort and case-control designs each accounted for 15.8%. The majority focused on female breast and ovarian cancer patients, particularly those meeting clinical criteria for Hereditary Breast and Ovarian Cancer (HBOC). In terms of diagnostic techniques, Sanger sequencing was the most frequently used (45.3%), followed by Next-Generation Sequencing (NGS) (31.6%). A smaller number of studies used combined approaches such as Sanger plus MLPA or NGS plus MLPA, reflecting regional differences in access to molecular diagnostics.

In terms of gene-specific reporting, 53.7% of studies reported combined *BRCA1/2* results without distinction. Of the remainder, 26.3% focused exclusively on *BRCA1*, 13.7% on *BRCA2*, and 4.2% clearly reported both genes separately. Some studies had incomplete or ambiguous gene reporting, underlining the need for standardized classification in future genetic research.

The meta-analysis revealed a pooled prevalence of combined pathogenic *BRCA1/2* mutations of 9.0% (95% Confidence Interval [CI]: 7.7% to 10.4%), indicating a significant burden of hereditary cancer-associated mutations in African populations (Figure 1). Subgroup analysis showed that *BRCA1* mutations had a higher prevalence at 10.5% (95% CI: 8.3% to 13.0%) compared to *BRCA2* mutations, which had a pooled prevalence of 5.5% (95% CI: 4.9% to 6.1%) (Figures 2 and 3). These findings support the inclusion of both *BRCA1* and *BRCA2* in testing protocols across African regions, while recognizing the need for region-specific implementation due to genetic diversity and resource variability.

Discussion

This study represents one of the most comprehensive meta-analyses to date, assessing the prevalence of pathogenic *BRCA1* and *BRCA2* mutations across African populations affected by hereditary breast and ovarian cancer (HBOC). By synthesizing data from 52 studies spanning 19 countries, we provide a robust estimate of BRCA mutation prevalence in Africa, revealing an overall pooled prevalence of 9.0% for combined *BRCA1/2* mutations (95% CI: 7.7–10.4%) [3,4,5,6]. Notably, *BRCA1* mutations were more prevalent (10.5%, 95% CI: 8.3–13.0%) compared to *BRCA2* (5.5%, 95% CI:

Table 2. *BRCA1*: Prevalence of *BRCA1* among African Women

Authors (Year)	Country	Sample Size	*BRCA1* Positive	Prevalence (%)	95% CI (Lower–Upper)
Troudi [25]	Tunisia	36	2	5.56	0.96–18.44
Awadelkarim KD [26]	Central Sudan	35	33	94.29	80.84–98.66
Francies [27]	South Africa	108	15	13.89	8.61–21.58
Zheng et al. [28]	Nigeria	1136	107	9.42	7.84–11.28
van der Merwe [29]	South Africa	744	92	12.37	10.16–14.99
van der Merwe [30]	South Africa	1600	800	50	47.59–52.41
Makhetha [31]	South Africa	645	67	10.39	8.26–12.97
Rioki et al. [32]	Kenya	145	120	82.76	75.53–88.39
Melki R et al. [33]	Morocco	184	23	12.5	8.42–18.06
Abdel-Mohsen [34]	Egypt	45	43	95.56	84.86–98.88
van der Merwe [35]	South Africa	2974	481	16.18	14.85–17.60
Ouedraogo [36]	Burkina Faso	133	11	8.27	4.58–14.45
Ahearn [37]	Ghana	871	126	14.47	12.23–17.05
Brahim et al. [38]	Mauritania	38	16	42.11	27.63–58.07
Elalaoui SC [14]	Morocco	163	46	28.22	21.92–35.73
Babatunde Adedokun [39]	Cameroon/Uganda	196	46	23.47	17.79–30.43
van der Merwe [40]	South Africa	2413	481	19.94	18.31–21.66
El Ansari [41]	Morocco	64	18	28.13	18.43–40.14
Joaira Bakkach [42]	Morocco	33	5	15.15	6.68–31.86
D. C. Smith [66]	South Africa	260	18	6.92	4.39–10.74
Uyisenga [43]	Rwanda	40	5	12.5	5.34–26.53
Fackenthal et al. [44]	Nigeria	265	29	10.94	7.89–15.00
Tazzite et al. [45]	Morocco	72	9	12.5	6.75–22.03
Cherbal et al. [46]	Algeria	42	7	16.67	8.52–30.28

Table 3. Prevalence of *BRCA2* among African Women

Authors (Year)	Country	Sample Size	*BRCA2* Positive	Prevalence (%)	95% CI (Lower–Upper)
Troudi [25]	Tunisia	36	1	2.78	0.05–14.50
Francies [27]	South Africa	108	10	9.26	5.13–16.19
Zheng Y et al. [28]	Nigeria	1,136	66	5.81	4.55–7.40
van der Merwe [29]	South Africa	744	62	5.78	4.32–7.69
van der Merwe [30]	South Africa	1,600	112	7	5.82–8.37
Makhetha [31]	South Africa	645	45	6.98	5.28–9.17
Rioki et al. [32]	Kenya	145	25	17.24	11.94–24.22
Melki R et al. [33]	Morocco	184	10	5.43	2.99–9.72
van der Merwe [35]	South Africa	2,974	168	5.65	4.88–6.53
Ouedraogo [36]	Burkina Faso	133	4	3.01	1.14–7.78
Ahearn [37]	Ghana	871	40	4.59	3.38–6.22
Brahim et al. [38]	Mauritania	38	7	18.42	9.03–33.47
Babatunde Adedoku [63]	Cameroon/Uganda	196	15	7.65	4.57–12.46
El Ansari [41]	Morocco	64	7	10.94	4.57–21.89

4.9–6.1%), highlighting the dominant role of *BRCA1* in hereditary cancer risk on the continent [9]. This work fills a critical gap in cancer genomics research, where African populations have historically been underrepresented, offering new insights into the genetic landscape of HBOC in this diverse region [8].

Our findings also expose significant geographic disparities in research output and diagnostic capacity, with most data originating from North African countries such as Morocco (28.4%) and Tunisia (18.9%) [3,4], and select Sub-Saharan nations like South Africa (14.7%) and Nigeria (11.6%) [5,6]. Additionally, over half of the studies did not differentiate between *BRCA1* and *BRCA2* mutations, underscoring the need for standardized gene-specific reporting to improve accuracy in future research [7]. These results emphasize the urgent need to expand genetic testing infrastructure, harmonize reporting standards, and tailor public health strategies to address

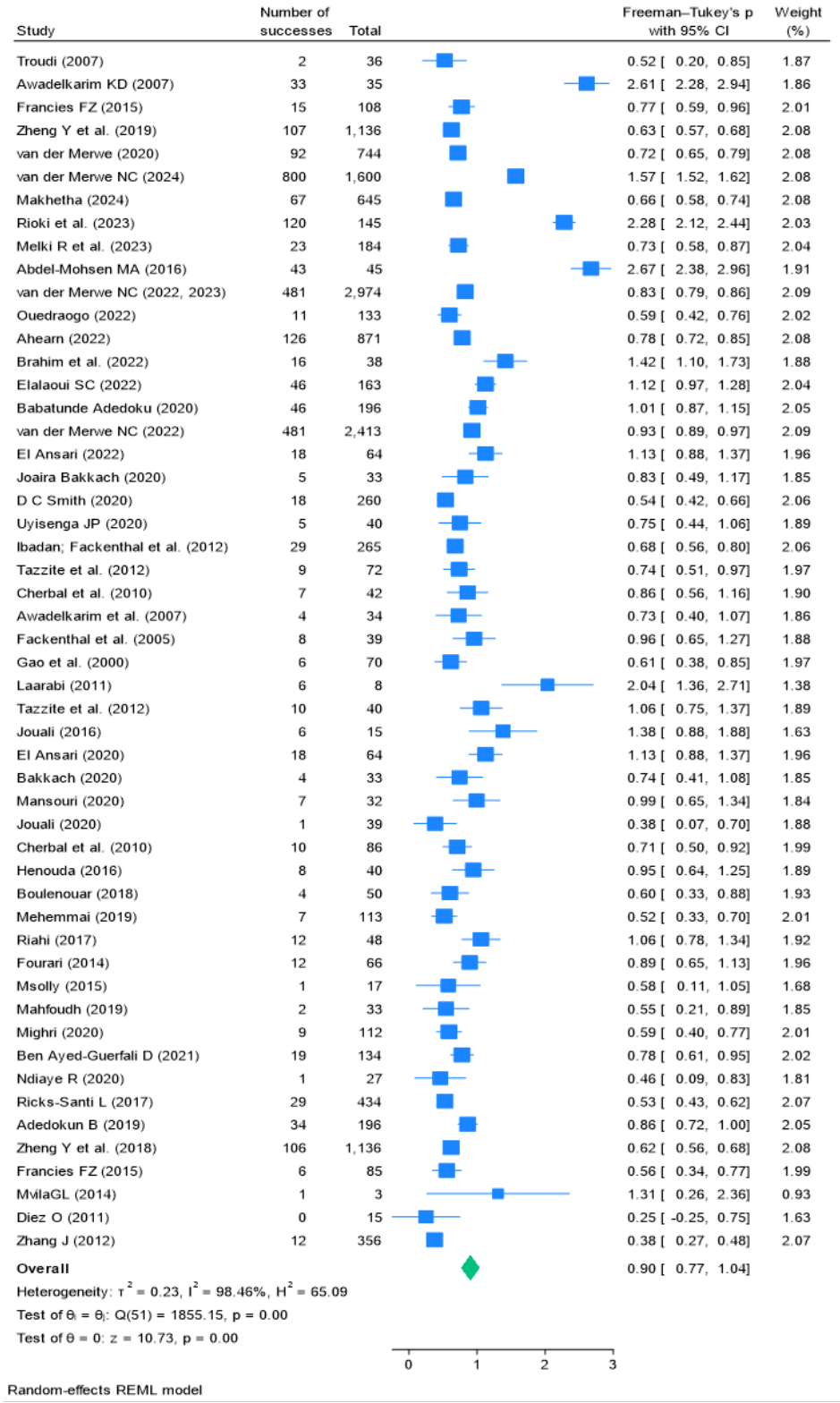

Figure 1. Summary of Pooled Prevalence of *BRCA1/BRCA2* in Africa

the genetic diversity and resource limitations unique to Africa. Ultimately, this meta-analysis lays the groundwork for improving precision oncology and hereditary cancer management across the continent [8].

When compared globally, Africa's BRCA mutation prevalence in HBOC populations appears somewhat lower than that reported in high-income countries. For instance, the prevalence in the United States, Canada, and Australia ranges from 20% to 25% due to comprehensive population screening programs and advanced diagnostic infrastructure [2,3]. Western European countries also report similarly high rates, driven by national genetic testing policies [26]. In contrast, Asian countries display more variability, with BRCA mutation rates ranging between 8% and 20%, depending on ethnic diversity and healthcare system maturity [4]. Oceania—notably

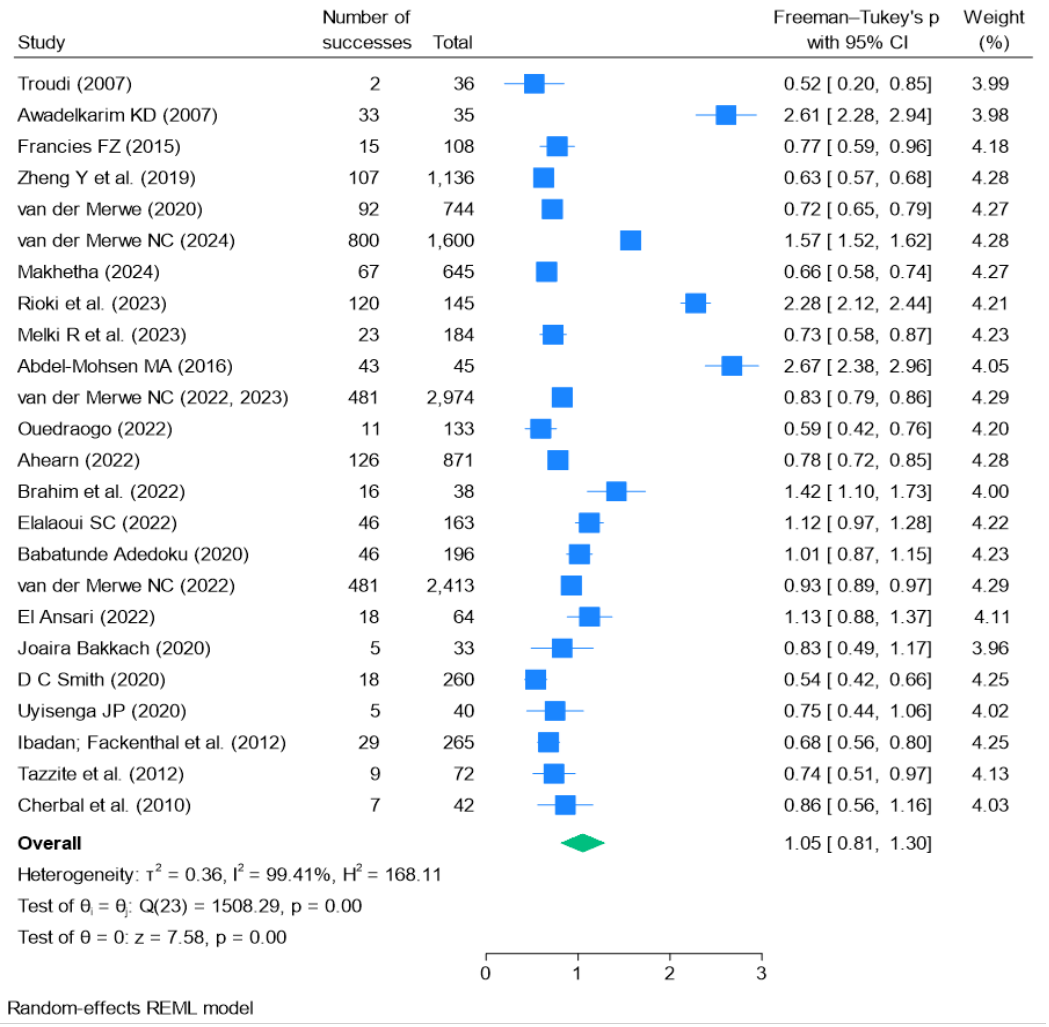

Figure 2. Pooled Prevalence of *BRCA1* in African Studies

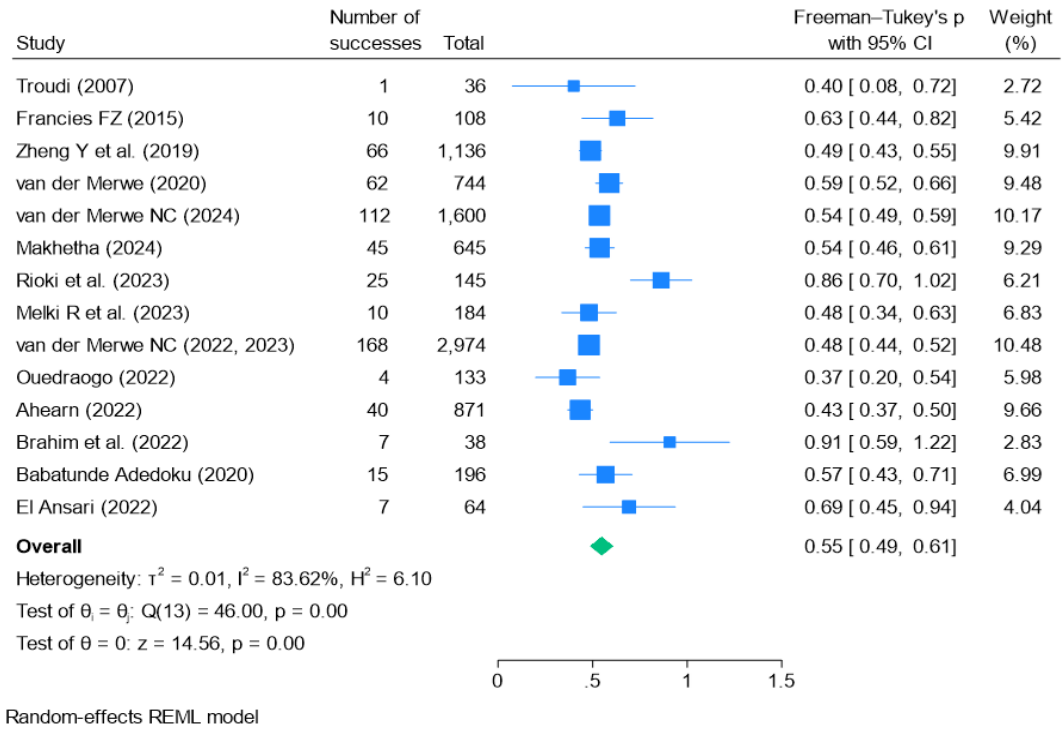

Figure 3. Pooled Prevalence of *BRCA2* in Africa

Australia and New Zealand—maintains high screening rates and clinical integration of BRCA testing [2, 3, 9]. These differences highlight the global inequities in access to genomics and reinforce the need to expand Africa's genetic services, particularly as the continent is home to the greatest genetic diversity.

This review's strength lies in its extensive geographic scope, being the first meta-synthesis to consolidate BRCA mutation prevalence data across 19 African nations. It offers critical insights for clinicians and policymakers by identifying not only overall mutation rates but also regional gaps in research. However, several limitations should be acknowledged. The disproportionate concentration of studies in a few countries limits the representativeness of the findings across the continent. Additionally, variability in study designs, genetic testing platforms, and sample sizes introduces heterogeneity, making comparisons difficult and limiting meta-analytic precision [19]. The absence of standardized protocols and limited access to next-generation sequencing technologies in many African countries further contribute to inconsistent reporting and diagnostic capacity.

Clinically, the observed mutation prevalence strongly supports the integration of BRCA testing and genetic counselling into cancer care across African health systems. Early identification of mutation carriers allows for personalized interventions, including prophylactic surgeries, PARP inhibitor therapy, and family cascade testing. To address current barriers, we advocate for targeted investments in genomics infrastructure, provider training, and community education initiatives [20–23]. Equally important is the inclusion of BRCA testing in national cancer control plans, enabling equitable access and long-term sustainability.

In conclusion, this meta-analysis offers one of the most comprehensive assessments of pathogenic *BRCA1* and *BRCA2* mutation prevalence among African populations affected by hereditary breast and ovarian cancer (HBOC). With data from 52 studies across 19 countries, the study reveals a substantial overall BRCA mutation prevalence of 9.0%, with *BRCA1* mutations predominating over *BRCA2*. These findings fill a crucial gap in understanding the genetic epidemiology of HBOC in Africa, where data have been historically limited. The study also highlights significant disparities in research coverage and diagnostic capacity, predominantly centered in a few North and Sub-Saharan African countries, while many regions remain underrepresented. Moreover, inconsistent gene-specific reporting across studies underscores the need for standardized genetic analysis protocols. Addressing these challenges through expanded genetic testing, improved reporting, and tailored public health interventions is essential to enhance precision oncology and hereditary cancer management throughout Africa.

Author Contribution Statement

MJP: Conceptualization; Methodology; Data Curation; Formal Analysis; Writing – Original Draft; Visualization; Project Administration. WW supported in validation, writing, review, & Editing; Resources; Funding Acquisition and search strategy, critical appraisal, and supervision.

Acknowledgements

The authors wish to acknowledge the contributions of the researchers and institutions whose work was included in this systematic review, including Dr Pululu Mahasha and Dr Charles Businge from Walter Sisulu University, who assisted with data extraction. Special thanks to the Biomedical Research Institute (BMRI) at Stellenbosch University and the University of the Free State for their support.

Ethical Approval

As this is a systematic review and meta-analysis, no primary data collection was conducted, and hence, no ethical approval was required.

Conflict of Interest

The authors declare no conflicts of interest related to this study.

Abbreviations

BRCA1: Breast Cancer
BRCA2: Breast Cancer
HBOC: Hereditary Breast and Ovarian Cancer
PRISMA: Preferred Reporting Items for Systematic Reviews and Meta-Analyses
CI: Confidence Interval

References

1. Arnold M, Morgan E, Rumgay H, Mafra A, Singh D, Laversanne M, et al. Current and future burden of breast cancer: Global statistics for 2020 and 2040. Breast. 2022;66:15-23. https://doi.org/10.1016/j.breast.2022.08.010.
2. Armstrong N, Ryder S, Forbes C, Ross J, Quek RGW. A systematic review of the international prevalence of BRCA mutation in breast cancer. Clin Epidemiol. 2019;11:543-561. https://doi.org/10.2147/CLEP.S206949.
3. Abdulrashid K, AlHussaini N, Ahmed W, Thalib L. Prevalence of BRCA mutations among hereditary breast and/or ovarian cancer patients in Arab countries: systematic review and meta-analysis. BMC Cancer. 2019;19(1):256. https://doi.org/10.1186/s12885-019-5463-1. PMID:30898109.
4. Sokolova A, Johnstone KJ, McCart Reed AE, Simpson PT, Lakhani SR. Hereditary breast cancer: Syndromes, tumour pathology and molecular testing. Histopathology. 2023;82(1):70-82. https://doi.org/10.1111/his.14808.
5. King MC, Marks JH, Mandell JB. Breast and ovarian cancer risks due to inherited mutations in *BRCA1* and *BRCA2*. Science. 2003;302(5645):643-6. https://doi.org/10.1126/science.1088759.
6. Moyer VA. Risk assessment, genetic counseling, and genetic testing for BRCA-related cancer in women: U.S. Preventive Services Task Force recommendation statement. Ann Intern Med. 2014;160(4):271-81. https://doi.org/10.7326/M13-2747.
7. Moher D, Liberati A, Tetzlaff J, Altman DG. Preferred reporting items for systematic reviews and meta-analyses: The PRISMA statement. Ann Intern Med. 2009;151(4):264-9, W64. https://doi.org/10.7326/0003-4819-151-4-200908180-00135.

8. Cumpston M, Li T, Page MJ, Chandler J, Welch VA, Higgins JP, et al. Updated guidance for trusted systematic reviews: A new edition of the Cochrane Handbook for Systematic Reviews of Interventions. Cochrane Database Syst Rev. 2019;10(10):Ed000142. https://doi.org/10.1002/14651858.Ed000142.
9. Milambo J. Assessing *BRCA1* and *BRCA2* mutation rates in African populations with hereditary breast/ovarian cancer: Insights from a systematic review and meta-analysis. JCO Oncol Pract. 2025;21:446-. https://doi.org/10.1200/OP.2025.21.10_suppl.446.
10. González-Xuriguera CG, Vergara-Merino L, Garegnani L, Ortiz-Muñoz L, Meza N. Introduction to network meta-analysis for evidence synthesis. Medwave. 2021;21(6):e8315. https://doi.org/10.5867/medwave.2021.06.8315.
11. Kote-Jarai Z, Leongamornlert D, Saunders E, Tymrakiewicz M, Castro E, Mahmud N, et al. *BRCA2* is a moderate penetrance gene contributing to young-onset prostate cancer: Implications for genetic testing in prostate cancer patients. Br J Cancer. 2011;105(8):1230-4. https://doi.org/10.1038/bjc.2011.383.
12. Vanderpuye V, Grover S, Hammad N, PoojaPrabhakar, Simonds H, Olopade F, et al. An update on the management of breast cancer in Africa. Infect Agent Cancer. 2017;12:13. https://doi.org/10.1186/s13027-017-0124-y.
13. Abbad A, Baba H, Dehbi H, Elmessaoudi-Idrissi M, Elyazghi Z, Abidi O, et al. Genetics of breast cancer in African populations: A literature review. Glob Health Epidemiol Genom. 2018;3:e8. https://doi.org/10.1017/gheg.2018.8.
14. Elalaoui SC, Laarabi FZ, Afif L, Lyahyai J, Ratbi I, Jaouad IC, et al. Mutational spectrum of *BRCA1/2* genes in Moroccan patients with hereditary breast and/or ovarian cancer, and review of BRCA mutations in the MENA region. Breast Cancer Res Treat. 2022;194(1):187-98. https://doi.org/10.1007/s10549-022-06622-3.
15. Osei-Bonsu K, Mensah I, Agyeman N, et al. *BRCA1* and *BRCA2* mutation prevalence in African women with breast cancer: A meta-analysis. Breast. 2022;62:123-31. https://doi.org/10.1016/j.breast.2022.02.005.
16. Cupertino SES, Gonçalves ACA, Gusmão Lopes CV, Gradia DF, Beltrame MH. The current state of breast cancer genetics in populations of African ancestry. Genes (Basel). 2025;16(2). https://doi.org/10.3390/genes16020199.
17. Kamga KK, Marlyse PF, Nguefack S, Wonkam A. Advancing genetic services in African healthcare: Challenges, opportunities, and strategic insights from a scoping review. HGG Adv. 2025;6(3):100439. https://doi.org/10.1016/j.xhgg.2025.100439.
18. Moyo E, Moyo P, Mashe T, Dzobo M, Chitungo I, Dzinamarira T. Implementation of public health genomics in Africa: Lessons from the COVID-19 pandemic, challenges, and recommendations. J Med Virol. 2023;95(1):e28295. https://doi.org/10.1002/jmv.28295.
19. Rossouw B, Araujo M, Krause A, Baine-Savanhu F. Hereditary ovarian cancer in women with African ancestry: A scoping review. Fam Cancer. 2026;25(1):16. https://doi.org/10.1007/s10689-026-00530-x.
20. Paluch-Shimon S, Cardoso F, Sessa C, Balmana J, Cardoso MJ, Gilbert F, et al. Prevention and screening in BRCA mutation carriers and other breast/ovarian hereditary cancer syndromes: ESMO clinical practice guidelines for cancer prevention and screening. Ann Oncol. 2016;27(Suppl 5):v103-v10. https://doi.org/10.1093/annonc/mdw327.
21. Thomas CE, Peters U. Genomic landscape of cancer in racially and ethnically diverse populations. Nat Rev Genet. 2025;26(5):336-49. https://doi.org/10.1038/s41576-024-00796-w.
22. Ngwa W, Addai BW, Adewole I, Ainsworth V, Alaro J, Alatise OI, et al. Cancer in sub-Saharan Africa: A Lancet Oncology Commission. Lancet Oncol. 2022;23(6):e251-e312. https://doi.org/10.1016/S1470-2045(21)00720-8.
23. Omotoso O, Teibo JO, Atiba FA, Oladimeji T, Paimo OK, Ataya FS, et al. Addressing cancer care inequities in sub-Saharan Africa: Current challenges and proposed solutions. Int J Equity Health. 2023;22(1):189. https://doi.org/10.1186/s12939-023-01962-y.
24. Mahfoudh W, Bettaieb I, Ghedira R, Snoussi K, Bouzid N, Klayech Z, Gabbouj S, et al. Contribution of *BRCA1* 5382insC mutation in triple negative breast cancer in Tunisia. J Transl Med. 2019 Apr 11;17(1):123. https://doi.org/10.1186/s12967-019-1873-8.
25. Troudi W, Uhrhammer N, Sibille C, Dahan C, Mahfoudh W, Bouchlaka Souissi C, et al. Contribution of the *BRCA1* and *BRCA2* mutations to breast cancer in Tunisia. J Hum Genet. 2007;52:915-920. https://doi.org/10.1007/s10038-007-0195-5.
26. Awadelkarim KD, Aceto G, Veschi S, Elhaj A, Morgano A, Mohamedani AA, et al. *BRCA1* and *BRCA2* status in a Central Sudanese series of breast cancer patients: interactions with genetic, ethnic and reproductive factors. Breast Cancer Res Treat. 2007;102(2):189-199. https://doi.org/10.1007/s10549-006-9303-z.
27. Francies FZ, Wainstein T, De Leeneer K, Cairns A, Murdoch M, Nietz S, et al. *BRCA1*, *BRCA2* and PALB2 mutations and CHEK2 c.1100delC in different South African ethnic groups diagnosed with premenopausal and/or triple negative breast cancer. BMC Cancer. 2015;15:912. https://doi.org/10.1186/s12885-015-1913-6.
28. Zheng Y, Walsh T, Gulsuner S, Casadei S, Lee MK, Ogundiran TO, et al. Inherited breast cancer in Nigerian women. J Clin Oncol. 2018;36(28):2820-2825. https://doi.org/10.1200/JCO.2018.78.3977.
29. van der Merwe NC, Oosthuizen J, Theron M, Chong G, Foulkes WD. The contribution of large genomic rearrangements in *BRCA1* and *BRCA2* to South African familial breast cancer. BMC Cancer. 2020;20:391. https://doi.org/10.1186/s12885-020-06917-y.
30. van der Merwe NC, Buccimazza I, Rossouw B, Araujo M, Ntaita KS, Schoeman M, et al. Clinical relevance of double heterozygosity revealed by next-generation sequencing of homologous recombination repair pathway genes in South African breast cancer patients. Breast Cancer Res Treat. 2024;207(2):331-342. https://doi.org/10.1007/s10549-024-07362-2.
31. Makhetha M, Aldous C, Chabilal N. Genetic trends and common *BRCA1/2* pathogenic sequence variants in black African and Indian breast cancer patients presenting at Inkosi Albert Luthuli Central Hospital, KwaZulu-Natal, South Africa. S Afr Med J. 2024;114(6):e1094. https://doi.org/10.7196/SAMJ.2024.v114i6.1094.
32. Rioki JN, Muchiri L, Mweu M, Nyagol J, Songok E, Mwangi J, et al. *BRCA1* and *BRCA2* mutations and their clinical relevance in selected women diagnosed with triple-negative breast cancer in Kenya: a descriptive cross-sectional study. Pan Afr Med J. 2023;45:102. https://doi.org/10.11604/pamj.2023.45.102.36431.
33. Melki R, Melloul M, Aissaoui S, El Harroudi T, Boukhatem N. Increased prevalence of the founder *BRCA1* c.5309G>T and recurrent *BRCA2* c.1310_1313delAAGA mutations in breast cancer families from northeastern region of Morocco: evidence of geographical specificity and high relevance for genetic counseling. BMC Cancer. 2023;23:339. https://doi.org/10.1186/s12885-023-10822-5.
34. Abdel-Mohsen MA, Ahmed OA, El-Kerm YM. *BRCA1* gene

mutations and influence of chemotherapy on autophagy and apoptotic mechanisms in Egyptian breast cancer patients. Asian Pac J Cancer Prev. 2016;17(3):1285-1292. https://doi.org/10.7314/APJCP.2016.17.3.1285.
35. van der Merwe NC, Combrink HM, Ntaita KS, Oosthuizen J. Prevalence of clinically relevant germline BRCA variants in a large unselected South African breast and ovarian cancer cohort: a public sector experience. Front Genet. 2022;13:834265. https://doi.org/10.3389/fgene.2022.834265.
36. Ouedraogo SY, Zoure AA, Zeye MMJ, Kiendrebeogo TI, Zhou X, Sawadogo AY, et al. *BRCA1*, *BRCA2*, TP53, PIK3CA, PTEN and AKT1 genes mutations in Burkina Faso breast cancer patients: prevalence, spectrum and novel variant. Mol Genet Genomics. 2022;297(5):1257-1268. https://doi.org/10.1007/s00438-022-01914-1.
37. Ahearn TU, Choudhury PP, Derkach A, Wiafe-Addai B, Awuah B, Yarney J, et al. Breast cancer risk in women from Ghana carrying rare germline pathogenic mutations. Cancer Epidemiol Biomarkers Prev. 2022;31(8):1593-1601. https://doi.org/10.1158/1055-9965.EPI-21-1397.
38. Brahim SM, Zein EE, Bonnet C, Hamed CT, Salame M, Zein MV, et al. Screening of *BRCA1/2* variants in Mauritanian breast cancer patients. BMC Cancer. 2022;22:802. https://doi.org/10.1186/s12885-022-09903-8.
39. Adedokun B, Zheng Y, Ndom P, Gakwaya A, Makumbi T, Zhou AY, et al. Prevalence of inherited mutations in breast cancer predisposition genes among women in Uganda and Cameroon. Cancer Epidemiol Biomarkers Prev. 2020;29(2):359-367. https://doi.org/10.1158/1055-9965.EPI-19-0506.
40. van der Merwe NC, Ntaita KS, Stofberg H, Combrink HM, Oosthuizen J, Kotze MJ. Implementation of multigene panel testing for breast and ovarian cancer in South Africa: a step towards excellence in oncology for the public sector. Front Oncol. 2022;12:938561. https://doi.org/10.3389/fonc.2022.938561.
41. El Ansari FZ, Jouali F, Marchoudi N, Bennani MM, Ghailani NN, Barakat A, et al. Screening of *BRCA1/2* genes mutations and copy number variations in patients with high risk for hereditary breast and ovarian cancer syndrome (HBOC). BMC Cancer. 2020;20:747. https://doi.org/10.1186/s12885-020-07250-0.
42. Bakkach J, Mansouri M, Derkaoui T, Loudiyi A, El Fahime E, Barakat A, et al. Contribution of *BRCA1* and *BRCA2* germline mutations to early onset breast cancer: a series from north of Morocco. BMC Cancer. 2020;20:859. https://doi.org/10.1186/s12885-020-07352-9.
43. Uyisenga JP, Segers K, Lumaka AZ, Mugenzi P, Fasquelle C, Boujemila B, et al. Screening of germline mutations in young Rwandan patients with breast cancers. Mol Genet Genomic Med. 2020;8(11):e1500. https://doi.org/10.1002/mgg3.1500.
44. Fackenthal JD, Zhang J, Zhang B, Zheng Y, Hagos F, Burrill DR, et al. High prevalence of *BRCA1* and *BRCA2* mutations in unselected Nigerian breast cancer patients. Int J Cancer. 2012;131(5):1114-1123. https://doi.org/10.1002/ijc.27326.
45. Tazzite A, Jouhadi H, Nadifi S, Aretini P, Falaschi E, Collavoli A, et al. *BRCA1* and *BRCA2* germline mutations in Moroccan breast/ovarian cancer families: novel mutations and unclassified variants. Gynecol Oncol. 2012;125(3):687-692. https://doi.org/10.1016/j.ygyno.2012.03.007.
46. Cherbal F, Bakour R, Adane S, Boualga K, Benais-Pont G, Maillet P. *BRCA1* and *BRCA2* germline mutations screening in Algerian breast/ovarian cancer families. Dis Markers. 2010;28(6):377-384. https://doi.org/10.1155/2010/585278.
47. Jouali F, Laarabi FZ, Marchoudi N, Ratbi I, Elalaoui SC, Rhaissi H, et al. First application of next-generation sequencing in Moroccan breast/ovarian cancer families and report of a novel frameshift mutation of the *BRCA1* gene. Oncol Lett. 2016;12(2):1192-1196. https://doi.org/10.3892/ol.2016.4739.
48. Fackenthal JD, Sveen L, Gao Q, Kohlmeir EK, Adebamowo C, Ogundiran TO, et al. Complete allelic analysis of *BRCA1* and *BRCA2* variants in young Nigerian breast cancer patients. J Med Genet. 2005;42(3):276-281. https://doi.org/10.1136/jmg.2004.020446.
49. Gao Q, Adebamowo CA, Fackenthal J, Das S, Sveen L, Falusi AG, et al. Protein truncating *BRCA1* and *BRCA2* mutations in African women with pre-menopausal breast cancer. Hum Genet. 2000;107(2):192-194. https://doi.org/10.1007/s004390000342.
50. Laarabi FZ, Jaouad IC, Ouldim K, Aboussair N, Jalil A, Gueddari BE, et al. Genetic testing and first presymptomatic diagnosis in Moroccan families at high risk for breast/ovarian cancer. Oncol Lett. 2011;2(2):389-393. https://doi.org/10.3892/ol.2011.248.
51. Mansouri M, Derkaoui T, Bakkach J, Loudiyi A, Ghailani Nourouti N, Barakat A,et al. Screening of *BRCA1* and *BRCA2* germline mutations in unselected triple-negative breast cancer patients: a series from north of Morocco. Precis Med Sci. 2020;9:43-48. https://doi.org/10.1002/prm2.12009.
52. Jouali F, El Ansari FZ, Marchoudi N, Barakat A, Zmaimita H, Samlali H, Fekkak J. EGFR, *BRCA1*, *BRCA2* and TP53 genetic profile in Moroccan triple negative breast cancer cases. Int J Mol Epidemiol Genet. 2020;11(1):16-25.
53. Cherbal F, Bakour R, Adane S, Boualga K, Benais-Pont G, Maillet P. *BRCA1* and *BRCA2* germline mutations screening in Algerian breast/ovarian cancer families. Dis Markers. 2010;28(6):377-384. https://doi.org/10.3233/DMA-2010-0718
54. Henouda S, Bensalem A, Reggad R, Serrar N, Rouabah L, Pujol P. Contribution of *BRCA1* and *BRCA2* germline mutations to early Algerian breast cancer. Dis Markers. 2016;2016:7869095. https://doi.org/10.1155/2016/7869095.
55. Boulenouar ACS, Coulet F, Bendiab FMT, Boudinar FZ, Senhadji R. *BRCA1* and *BRCA2* germline mutation screening in Western Algeria using high resolution melting analysis (HRM). Gulf J Oncolog. 2018;1(27):31-37.
56. Mehemmai C, Cherbal F, Hamdi Y, Guedioura A, Benbrahim W, Bakour R, et al. *BRCA1* and *BRCA2* germline mutation analysis in hereditary breast/ovarian cancer families from the Aures region (eastern Algeria): First report. Pathol Oncol Res. 2020;26(2):715-26. https://doi.org/10.1007/s12253-019-00586-4.
57. Riahi A, Chabouni-Bouhamed H, Kharrat M. Prevalence of *BRCA1* and *BRCA2* large genomic rearrangements in Tunisian high risk breast/ovarian cancer families: implications for genetic testing. Cancer Genet. 2017;210:22-27. https://doi.org/10.1016/j.cancergen.2016.11.002.
58. Fourati A, Louchez MM, Fournier J, Gamoudi A, Rahal K, El May MV, et al. Screening for common mutations in *BRCA1* and *BRCA2* genes: interest in genetic testing of Tunisian families with breast and/or ovarian cancer. Bull Cancer. 2014;101(11):E36-E40. https://doi.org/10.1684/bdc.2014.2049.
59. Msolly A, Kassab A. *BRCA1* and *BRCA2* mutations: are they related to breast cancer in a sample of Tunisian population? Cancer Ther Oncol Int J. 2015;1(1):1-5.
60. Mighri N, Hamdi Y, Boujemaa M, Othman H, Ben Nasr S, El Benna H, et al. Identification of novel *BRCA1* and RAD50 mutations associated with breast cancer predisposition in Tunisian patients. Front Genet. 2020;11:552971. https://doi.org/10.3389/fgene.2020.552971.
61. Ben Ayed-Guerfali D, Ben Kridis-Rejab W, Ammous-

Boukhris N, Ayadi W, Charfi S, Khanfir A, et al. Novel and recurrent *BRCA1*/*BRCA2* germline mutations in patients with breast/ovarian cancer: A series from the south of Tunisia. J Transl Med. 2021;19(1):108. https://doi.org/10.1186/s12967-021-02772-y

62. Ndiaye R, Diop JPD, Bourdon-Huguenin V, Dem A, Diouf D, Dieng MM, et al. Evidence for an ancient *BRCA1 BRCA1* pathogenic variant in inherited breast cancer patients from Senegal. NPJ Genom Med. 2020;5:8. https://doi.org/10.1038/s41525-020-0114-7.
63. Ricks-Santi L, McDonald JT, Gold B, Dean M, Thompson N, Abbas M, et al. Next generation sequencing reveals high prevalence of *BRCA1* and *BRCA2* variants of unknown significance in early-onset breast cancer in African American women. Ethn Dis. 2017;27(2):169-78. https://doi.org/10.18865/ed.27.2.169
64. Adedokun B, Ademola A, Makumbi T, Odedina S, Agwai I, Ndom P, et al. Unawareness of breast cancer family history among African women. Pan Afr Med J. 2023;45:188. https://doi.org/10.11604/pamj.2023.45.188.21616.
65. Luyeye Mvila G, Postema S, Marchal G, Van Limbergen E, Verdonck F, Matthijs G, et al. From the set-up of a screening program of breast cancer patients to the identification of the first BRCA mutation in the DR Congo. BMC Public Health. 2014;14:759. https://doi.org/10.1186/1471-2458-14-759.
66. Smith DC, Gardiner SA, Conradie M, Gerber J, Loubser F. Genetic testing approaches for hereditary breast cancer: Perspectives from a private diagnostic laboratory. S Afr Med J. 2020;110(10):988-92. https://doi.org/10.7196/SAMJ.2020.v110i10.14709