REMUNOM
ISSN 2178-6925

Received: 13/01/2026 - **Accepted:** 07/05/2026
Vol: 13.08
DOI: 10.66104/qhx4xy11
Pages: 1-18

ADOÇÃO E PERCEPÇÃO DO MAPEAMENTO GENÉTICO NO CÂNCER DE MAMA: UM LEVANTAMENTO PRELIMINAR E EXPLORATÓRIO ENTRE ESPECIALISTAS EM GOIÁS

ADOPTION AND PERCEPTION OF GENETIC MAPPING IN BREAST CANCER: A PRELIMINARY AND EXPLORATORY SURVEY AMONG SPECIALISTS IN GOIÁS

ADOPCIÓN Y PERCEPCIÓN DEL MAPEO GENÉTICO EN EL CÁNCER DE MAMA: UN ESTUDIO PRELIMINAR Y EXPLORATORIO ENTRE ESPECIALISTAS EN GOIÁS

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Resumo

O câncer de mama é uma neoplasia de significativo impacto na saúde pública, com uma parcela

REMUNOM
ISSN 2178-6925

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dos casos associada a fatores hereditários, principalmente mutações nos genes BRCA1 e BRCA2. O estado de Goiás tornou-se pioneiro no Brasil ao instituir, por meio da Lei nº 20.707/2020, o acesso gratuito ao teste genético para essas mutações no SUS. Este estudo investiga a integração desse mapeamento genético na perspectiva dos médicos especialistas locais. Foi conduzido um estudo observacional, transversal, com uma amostra de conveniência de 16 médicos (oncologistas, mastologistas e geneticistas) atuantes em Goiás. Utilizou-se um questionário eletrônico estruturado para avaliar o conhecimento, a frequência de uso e as barreiras percebidas em relação ao mapeamento genético. Os dados foram analisados por estatística descritiva. A maioria dos participantes (93,8\%) relatou conhecer o mapeamento genético, porém apenas 50\% o utilizam frequentemente. Os principais critérios clínicos para solicitação foram diagnósticos antes dos 40 anos, histórico familiar direto e tumor triplo-negativo antes dos 50 anos. As barreiras mais citadas para não utilizar o teste foram a percepção de baixa utilidade (31,3\%) e lacunas no aconselhamento genético. Os resultados apontam para uma discrepância entre o conhecimento teórico sobre a testagem e sua aplicação prática na rotina clínica. Barreiras atitudinais e operacionais, como a falta de familiaridade com o aconselhamento genético, parecem limitar a incorporação efetiva da ferramenta, mesmo com sua disponibilidade legal. Conclui-se que a efetiva implementação do mapeamento genético para câncer de mama em Goiás depende não apenas do acesso ao exame, mas também da superação de barreiras práticas, mediante capacitação profissional e estruturação de fluxos de aconselhamento genético.
Palavras-chave: Câncer de Mama; Genes BRCA1; Genes BRCA2; Testes Genéticos.

Abstract

Breast cancer is a neoplasm with a significant public health impact, with a proportion of cases associated with hereditary factors, mainly mutations in the BRCA1 and BRCA2 genes. The state of Goiás became a pioneer in Brazil by establishing, through Law No. 20,707/2020, free access to genetic testing for these mutations within the Brazilian Unified Health System (SUS). This study investigates the integration of genetic mapping from the perspective of local specialist physicians. An observational, cross-sectional study was conducted with a convenience sample of 16 physicians (oncologists, breast specialists, and geneticists) practicing in Goiás. A structured electronic questionnaire was used to assess knowledge, frequency of use, and perceived barriers related to genetic mapping. Data were analyzed using descriptive statistics. Most participants (93.8%) reported being aware of genetic mapping, but only 50% used it frequently. The main clinical criteria for ordering the test were diagnosis before the age of 40, direct family history, and triple-negative tumor before the age of 50. The most frequently cited barriers to not using the test were the perception of low usefulness (31.3%) and gaps in genetic counseling. The results point to a discrepancy between theoretical knowledge about testing and its practical application in clinical routine. Attitudinal and operational barriers, such as unfamiliarity with genetic counseling, appear to limit the effective incorporation of this tool, even when it is legally available. It is concluded that the effective implementation of genetic mapping for breast cancer in Goiás depends not only on access to the test, but also on overcoming practical barriers through professional training and the structuring of genetic counseling workflows.
Keywords: Breast Cancer; BRCA1 Genes; BRCA2 Genes; Genetic Testing.

Resumen

El cáncer de mama es una neoplasia de impacto significativo en la salud pública, con una parte de los casos asociada a factores hereditarios, principalmente mutaciones en los genes BRCA1 y BRCA2. El estado de Goiás se convirtió en pionero en Brasil al instituir, por medio de la Ley n.º 20.707/2020, el acceso gratuito a la prueba genética para estas mutaciones en el Sistema Único de Salud (SUS). Este estudio investiga la integración de este mapeo genético desde la perspectiva de los médicos especialistas locales. Se realizó un estudio observacional, transversal, con una muestra por conveniencia de 16 médicos (oncólogos, mastólogos y genetistas) que ejercen en Goiás. Se

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utilizó un cuestionario electrónico estructurado para evaluar el conocimiento, la frecuencia de uso y las barreras percibidas en relación con el mapeo genético. Los datos se analizaron mediante estadística descriptiva. La mayoría de los participantes (93,8%) informó conocer el mapeo genético, pero solo el 50% lo utiliza con frecuencia. Los principales criterios clínicos para solicitar la prueba fueron diagnóstico antes de los 40 años, antecedente familiar directo y tumor triple negativo antes de los 50 años. Las barreras más citadas para no utilizar la prueba fueron la percepción de baja utilidad (31,3%) y las lagunas en el asesoramiento genético. Los resultados señalan una discrepancia entre el conocimiento teórico sobre la prueba y su aplicación práctica en la rutina clínica. Barreras actitudinales y operativas, como la falta de familiaridad con el asesoramiento genético, parecen limitar la incorporación efectiva de esta herramienta, incluso con su disponibilidad legal. Se concluye que la implementación efectiva del mapeo genético para cáncer de mama en Goiás depende no solo del acceso a la prueba, sino también de la superación de barreras prácticas mediante capacitación profesional y la estructuración de flujos de asesoramiento genético.
Palabrasclave: Cáncer de Mama; Genes BRCA1; Genes BRCA2; Pruebas Genéticas.

1. Introduction

Although breast cancer is currently the second most common cancer and the fourth leading cause of cancer-related death, in 2020 it surpassed lung cancer in the number of diagnoses. Although this pattern has reversed in subsequent years, the World Health Organization (WHO) and the oncology community were compelled to act urgently to address this neoplasm and confront the growing global burden of the disease, which is placing increasing strain on individuals, communities, and health systems worldwide (INTERNATIONAL AGENCY FOR RESEARCH ON CANCER, 2022.) (ORGANIZAÇÃO PAN-AMERICANA DA SAÚDE, 2020.).

Breast cancer is one of the leading causes of cancer-related death worldwide. According to the WHO, an estimated 2.3 million cases were diagnosed globally in 2022, representing approximately 11.5% of all new cancer cases. In addition, breast cancer was responsible for approximately 666,103 deaths in the same year (ORGANIZAÇÃO PAN-AMERICANA DA SAÚDE, 2020.) (WORLD HEALTH ORGANIZATION, 2021).

In Brazil, breast cancer is the leading cause of mortality among women, with a higher incidence in the South and Southeast regions. Excluding non-melanoma skin tumors, it is the most frequently diagnosed cancer in all regions of the country, accounting for about 28% of new cancer cases in women (BRASIL, 2022.).

Breast cancer is a heterogeneous disease, with several distinct subtypes.

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Some progress rapidly, while others follow a slower course. However, most cases have a favorable prognosis when diagnosed early and treated appropriately. Although it is relatively rare before the age of 35, its incidence increases progressively, especially after the age of 50. For the 2023–2025 triennium, 73,610 new cases are projected in Brazil, resulting in an adjusted incidence rate of 41.89 cases per 100,000 women (BRASIL, 2022.) (WORLD HEALTH ORGANIZATION, 2021).

Regarding mortality, in 2021, the death rate from breast cancer in the global population was 11.71 deaths per 100,000 women. The Southeast and South regions showed the highest mortality rates, with 12.43 and 12.69 deaths per 100,000, respectively, followed by the Northeast (10.75 deaths per 100,000), Central-West (10.90 deaths per 100,000), and North (8.59 deaths per 100,000) (INSTITUTO NACIONAL DE CÂNCER, 2022) (WORLD HEALTH ORGANIZATION, 2021).

The disease is multifactorial, and its risk factors can be categorized into three main groups. Behavioral and environmental factors include postmenopausal obesity, sedentary lifestyle, alcohol consumption, and exposure to ionizing radiation. Factors related to reproductive and hormonal history include early menarche, nulliparity, first pregnancy after the age of 30, late menopause, prolonged use of oral contraceptives, and postmenopausal hormone replacement therapy. In addition, hereditary/genetic factors, such as a family history of ovarian or breast cancer, especially before the age of 50, and alterations in the BRCA1 and BRCA2 genes, also significantly increase the risk of developing the disease (MOMOZAWA *et al*., 2022) (INSTITUTO NACIONAL DE CÂNCER, 2018).

This hereditary predisposition is an important epidemiological factor in the development of breast cancer. Approximately 5% to 10% of all cases are related to inherited genetic mutations. Among the most well-known and extensively studied genetic alterations are those involving the BRCA gene family, which are associated with a 60% to 80% increased risk of developing breast neoplasms and a 60% risk of ovarian neoplasms (LI et al., 2022; BRASIL, 2015).

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Breast cancer is characterized by broad genetic heterogeneity, resulting in differences in treatment response and prognosis. Precision medicine, based on genomic sequencing, offers personalized approaches for the prediction, diagnosis, and treatment of tumors. In addition, preventive strategies such as risk-reducing surgery, including bilateral prophylactic mastectomy, may be considered for women with predisposing genetic variants. In this regard, it is important that health professionals are prepared to make decisions based on genetic mapping, especially of the BRCA family genes, which are among the best known and will be addressed in this study, while considering the complexity of decision-making in the management of hereditary breast cancer (BRASIL, 2015; CRISCITIELLO; CORTI, 2022).

The state of Goiás was a pioneer in integrating free genetic testing for BRCA1 and BRCA2 genes for patients with risk factors for these mutations, even though this approach has not yet been incorporated into national guidelines. State Law No. 20,707, of January 14, 2020, guarantees free testing for genetic mutations in these genes for women with a family history of breast or ovarian cancer. This measure aims to facilitate access to early diagnosis, enabling more effective follow-up and the adoption of appropriate preventive measures. Through this law, the Executive Branch, in cooperation with the Unified Health System (Sistema Único de Saúde — SUS), ensures that women with a personal history of breast and/or ovarian cancer diagnosed before the age of 40 or 50, as well as those with a family history of these diseases, have access to testing upon referral by a geneticist, breast specialist, or oncologist. This initiative represents a significant advance in the fight against breast and ovarian cancer, enabling a preventive and personalized approach for women at higher genetic risk for these diseases. Currently, Brazilian guidelines for early detection of breast cancer prioritize imaging tests and do not include genetic screening (BRASIL, 2015; GOIÁS, 2020).

This legislation came into effect in October 2023, following a cooperation agreement between the Federal University of Goiás (UFG) and the State Health Secretariat (SES), establishing Goiás as a leading state in the provision of this type

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of free testing in the country, serving as an example of prevention and control of such a prevalent burden, even though it is not the region with the highest incidence. However, the availability of this service is currently limited to the state capital (GOIÁS, 2020; CARVALHO, 2023).

Because this predisposition is inherited, early diagnosis is essential for prevention, family planning, and treatment selection, and may influence a patient's prognosis and quality of life (NEVES et al., 2022).

With this in mind, this study aims to analyze the perception and patterns of use of genetic mapping, specifically of the BRCA1 and BRCA2 genes, among oncologists, breast specialists, and geneticists in Goiás, identifying professional perspectives while considering the recent availability of this test through the SUS in the state. In doing so, it seeks to contribute to a deeper understanding that may enhance the prevention and treatment of this significant condition in the region.

2. Literature Review

2.1 Clinical criteria for indicating genetic mapping in breast cancer

Genetic testing for hereditary breast cancer plays a central role in identifying patients and family members at higher risk of pathogenic variants in genes such as BRCA1 and BRCA2. Current clinical guidelines indicate that test ordering should be guided by specific criteria such as early-age diagnosis, relevant family history, including multiple cancer cases in the same family, and triple-negative tumors. Thus, BRCA family mapping should not be understood as a routine screening test, but rather as a tool for risk stratification and support in clinical follow-up decisions, especially in patients suspected of having a hereditary syndrome. In this context, the literature reinforces that appropriate indication depends on specialists recognizing these criteria and being able to integrate them into the individual patient assessment. In systems where testing is available, as in Goiás, but is still only minimally incorporated into routine care, adherence tends to increase when professionals perceive the practical utility of the test and associate its results with concrete changes in clinical management, such as more intensive and specific

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surveillance, planned family counseling, and risk-reducing measures (AHMED, 2022; WANG; WANG, 2023; NATIONAL COMPREHENSIVE CANCER NETWORK, 2025).

2.2 Genetic counseling and professional training

Despite the expansion of genomic medicine and its incorporation into BRCA mutation mapping, the literature indicates that the integration of genetic testing into clinical practice still faces barriers related to professionals' genetic knowledge. Many non-geneticist physicians report low literacy on the subject, leading to uncertainty in ordering, interpreting, explaining results, and planning follow-up, which limits the effective use of the test. This gap is highly relevant, since genetic counseling is an essential step in the process, from indication to result interpretation and the follow-up of the patient and their family members (OWENS et al., 2019; PILARSKI, 2021).

Studies show that limited familiarity with genetic counseling, combined with the absence of specialized training and well-defined referral pathways, contributes to low testing uptake in oncology. Therefore, the literature supports that physician training should not be limited to theoretical knowledge about BRCA1 and BRCA2, but should also include practical skills such as ordering the test, explaining its clinical significance, and guiding the patient within an integrated care network (GAMBOA et al., 2026; DESAI et al., 2020).

2.3 Implementation in public health systems and access barriers

In public health systems, the availability of genetic testing depends not only on a legal framework, but also on investment in infrastructure, integrated organization, and equitable territorial distribution of services. Even when the test is offered free of charge, its effectiveness may be limited by centralization in capital cities, referral difficulties, delays in access, and the absence of integrated pathways between primary care, specialists, and laboratories. These limitations strongly affect actual access to the test, unlike the formal access guaranteed on paper (KHOURY et al., 2022).

The literature on public health implementation shows that institutional and

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logistical barriers have a major impact on the incorporation of genomic technologies that analyze BRCA1 and BRCA2. In regions where services are centralized in capital cities, geographic distance, fragmented care networks, and poor communication between levels of care may reduce test ordering even among professionals who recognize its relevance. Thus, implementing BRCA family genetic mapping requires not only test availability, but also referral structure, result reporting, and longitudinal support (LIMA et al., 2026; SCHAIBLEY et al., 2022).

3. Methodology

Study design and setting

his was an observational, cross-sectional, exploratory study conducted with oncologists, breast specialists, and geneticists working in the state of Goiás who care for patients with breast cancer.

Population and sample

The target population included professionals registered with the CRM-GO holding a specialty qualification (RQE) in oncology, mastology, and/or medical genetics. Considering the local reference institutions — Unidade de Combate ao Câncer Única de Anápolis, Associação de Combate ao Câncer em Goiás (ACCG), and Hospital de Câncer Araújo Jorge — an initial base of approximately 24 specialists was estimated; however, the sample was expanded through invitations to all eligible physicians in the state, using a non-probabilistic convenience sampling approach.

Inclusion and exclusion criteria and recruitment process

Professionals older than 18 years, with an active CRM-GO registration and a compatible RQE, who agreed to participate and signed the Informed Consent Form (ICF), were included. Incomplete questionnaires or those containing identifying information were excluded. Recruitment was carried out through electronic invitations (e-mail, WhatsApp) and via a link shared on the project's Instagram page (@projetoBRCA), containing information about the study objective and access to the ICF and the electronic questionnaire (Google Forms). After virtual acceptance of the ICF, participants were redirected to the standardized

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form.

Data collection instrument

The standardized questionnaire developed for the study population was constructed based on data obtained from a literature review on the topic conducted by the authors of this article. However, the questionnaire had no formal validation by specialists, which constitutes a methodological limitation of this study.

The questionnaire was structured in sections, beginning with the Informed Consent Form (ICF), in which the participant is informed about the research and decides whether to participate. If the participant declines, the questionnaire is terminated. If they agree, the form proceeds to the sociodemographic and professional characterization section, where information is collected on medical specialty, sex, age group, and place of practice, including the possibility of working in more than one city in the state of Goiás. The instrument then investigates general knowledge about genetic mapping applied to breast cancer, with questions about the participant's familiarity with the topic and awareness of the free testing of BRCA1 and BRCA2 genes in the state. Based on these responses, the questionnaire directs participants to different flows: those who report frequent use of genetic mapping indicate the reasons that justify this practice in clinical routine, whereas those who do not use it or use it infrequently are asked to identify the reasons for not incorporating it, such as cost-effectiveness, accessibility, or knowledge about genetic counseling. In addition, participants unfamiliar with the topic receive a brief informational material before answering whether they would incorporate this tool into clinical practice after reading it. At the end, the questionnaire concludes with a thank-you message and an invitation to share the study with other professionals in the field.

Data analysis

Data were tabulated in electronic spreadsheets (Microsoft Excel) and analyzed using descriptive statistics, with frequency and percentage calculations.

Ethical aspects

This study complied with the guidelines of Resolution CNS 466/2012 on

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research involving human subjects and was approved by the Research Ethics Committee of the Evangelical University of Goiás (CAAE 84082424.9.0000.5076, opinion 7,413,928).

4. Results

It should be emphasized that the results presented here are exploratory in nature. The sample is small (n = 16) and does not constitute a representative basis, particularly for categories with minimal counts, such as geneticists, for whom only one participant from that specialty was included. These characteristics prevent comparative analysis across specialties and preclude the reliable use of Pearson's chi-square test, as well as compromising the stability of inferential estimates (ORs with wide CIs). In addition, some variables show little explanatory variability, such as knowledge about genetic mapping, which was nearly unanimous (15/16), making them of limited value for explaining differences in practice. The responses regarding reasons allowed multiple mentions per participant, generating non-independence between categories and making traditional multivariable models difficult to apply. For these reasons, it is more appropriate to treat the findings as descriptions of trends and hypothesis generation, useful for guiding future research with larger samples, qualitative studies, or multilevel analyses, rather than as statistical proof of association.

Among the 16 physicians who responded to the questionnaire, the largest number of participants were oncologists, followed by breast specialists and geneticists. The sample was predominantly male, and the most frequent age group was 31–40 years (Table 1).

Table 1. Sociodemographic profile of the study participants.

Variables	**Total (n=16)**		**Female (n=06)**		**Male (n=10)**	
	n	**%**	**n**	**%**	**n**	**%**
Age group (years)						
21 - 30	01	6,25	0	-	01	6,25
31 - 40	08	50,0	06	37,5	02	12,5
41 - 50	03	18,75	0	-	03	18,75
51 - 60	04	25,0	0	-	04	25,0
Specialty						
Clinical oncology	08	50,0	04	25	04	25,0
Breast surgery	07	43,75	02	12,5	05	31,25

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Medical genetics	01	6,25	0	-	01	6,25

Source: research data.

Regarding knowledge of BRCA genetic mapping applied to breast cancer, 15 participants (93.8%) stated that they knew what it is. Concerning practical use of this mapping, 08 (50%) reported frequent use, 03 (18.8%) occasional use, 03 (18.8%) rare use, and 01 (6.2%) had never used the test.

The main reasons for requesting research into BRCA gene mutations through genetic mapping were breast or ovarian cancer diagnosed before the age of 40, direct family history of breast/ovarian cancer, and triple-negative tumors diagnosed before the age of 50. Among the reasons for not incorporating the test into clinical practice, the following stood out: “I do not think it is worth ordering this type of test,” “low cost-effectiveness,” and “limited understanding of genetic counseling.”

Table 2. Main reasons for requesting or not requesting genetic mapping.

Motivation	**Total**		**Male**		**Female**	
Reason for use	**n=16**	**%**	**n=10**	**%**	**n=6**	**%**
Breast or ovarian cancer diagnosed before age 40	08	50,0	05	50,0	03	50,0
Direct family history of breast or ovarian cancer	07	43,8	04	40,0	03	50,0
Triple-negative tumor diagnosed before age 50	07	43,8	04	40,0	03	50,0
Patient concern and planning	01	6,3	01	10,0	0	-
Reasons for non-use						
I do not think it is worth ordering this type of test	05	31,3	03	30,0	02	33,3
Low cost-effectiveness	02	12,5	02	20,0	0	-

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Limited understanding of genetic counseling	02	12,5	01	10,0	01	16,7
I have few patients with breast cancer	02	12,5	01	10,0	0	-

Source: research data. *Because the questions allowed multiple responses, percentage totals by sex exceed 100%.

5. Discussion

The main finding of our study was the clear discrepancy between knowledge and practice, since although 93.8% of physicians reported being familiar with BRCA genetic mapping, only half use it routinely. This difference reflects multifactorial barriers already described in the international literature. For instance, studies have shown that most physicians were aware of the availability of BRCA testing, but only a small proportion would actually request it. Other studies reinforce that the exponential demand for genetic testing faces obstacles that go beyond theoretical knowledge, including infrastructure limitations, institutional support, various levels of public administration, and perceptions of clinical utility (Dubsky et al., 2024; Armstrong, 2019).

Despite the high rate of self-reported knowledge in our study, these preliminary and limited data may suggest gaps in the depth of this knowledge and in the ability to apply it in practice. Many physicians are familiar with the test in theory but report uncertainty in interpreting results and choosing individualized management. Studies emphasize the need for continuous education and greater support resources for appropriate genetic counseling, since deficiencies in professional training may result in patients with relevant genetic mutations, such as BRCA, not receiving the appropriate referral in clinical practice. In other words, superficial knowledge does not automatically translate into practical action (Dubsky et al., 2024; Arun et al., 2024).

A second set of barriers involves the structure of the health system and patient flow. The physical availability of genetic tests is quite limited, concentrated in a few laboratories or specialized centers, mainly in capital cities. Publications

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highlight limited access to testing facilities and genetic counselors as key barriers, along with the possible financial burden. In other countries, poorly completed medical records and the need for bureaucratic authorization by health systems have been shown to delay the process (Dekanek et al.).

In addition, the absence of defined referral pathways hinders test implementation. Without clear protocols, the test remains restricted to isolated consultations, and most patients seen in primary care end up not using it because they do not know how to access it. Although our study did not address these protocols, the literature suggests that integrated care structures significantly increase the use of BRCA gene mapping. In short, the availability of the test in the SUS is only the first step; integrated, rapid, and well-coordinated care pathways are essential to ensure that the test is effectively used by the largest possible proportion of the target population (Matalon et al., 2023; Lasta; Groto; Paula, 2023).

Another important axis is the individual perception and attitude of each professional within clinical practice. Our study preliminarily suggests that some physicians view BRCA genetic mapping as of little use if the test results do not affect clinical management. One study reported an initial degree of skepticism among physicians; this tendency to wait for evidence-based studies before adopting the new technology led to a healthy skepticism regarding the benefit of BRCA1 and BRCA2 testing (Manolio et al., 2019; Zawatsky et al., 2023).

Finally, the fact that the testing service is primarily located in the capital adds logistical barriers to the previous ones. In Goiás and other states, physicians in inland areas must refer cases to the capital, which causes delays and discouragement. The literature suggests that geographic inequalities intensify existing barriers; remote areas tend to have fewer trained professionals and greater difficulty maintaining longitudinal follow-up of results. Although our study did not directly address this disparity, literature analysis shows that many regions far from capital cities have few or no genetics services, preventing many patients from being adequately assessed. In addition, the lack of teleconsultation and

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transportation protocols makes it difficult for physicians from inland areas to refer patients efficiently (Matalon et al., 2023; Best et al., 2022).

This exploratory study identified that, among oncologists, breast specialists, and geneticists working in Goiás, clinical criteria such as diagnosis of neoplasia at an early age, direct family history of breast/ovarian cancer, and the presence of tumors with aggressive characteristics were the main factors leading to the request for BRCA gene mapping. This fully corroborates national and international guidelines and other studies that have shown these criteria to be strong motivators for requesting this genetic test in breast cancer, reinforcing that such factors can be maintained in guidelines as indicators of high risk (Herzog et al., 2021; National Comprehensive Cancer Network, 2025).

The findings precisely align with the clinical logic adopted by national and international guidelines, which recommend prioritizing genetic testing of these BRCA family genes in patients with clear risk criteria, such as those diagnosed before age 40, those with significant family history, or those with aggressive tumor phenotypes, indicating that professionals recognize the value of mapping for individualized management. However, Brazilian studies show that, despite relatively high knowledge of hereditary breast and ovarian cancer syndrome among specialists, this theoretical preparation does not always translate into established practice of test ordering or follow-up. This study describes how such discrepancy suggests that test availability alone does not guarantee clinical use, since structural and attitudinal barriers, such as lack of counseling infrastructure, defined pathways, and training in result interpretation, play a central role in low practical adoption, thus corroborating the previous discussions (Lasta; Groto; Paula, 2023; Bilenduke et al., 2023; National Comprehensive Cancer Network, 2025).

In our sample, it was preliminarily identified that, despite recognition of the clinical criteria for test indication, barriers remain in routine incorporation, especially those related to understanding the risks and proper clinical management. In this context, strengthening pre- and post-test genetic counseling

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emerges as a need highlighted by the professionals themselves. International literature agrees that genetic counseling is a fundamental component of the responsible and effective use of BRCA testing, as it improves risk understanding, supports decision-making, and guides appropriate clinical management (Van et al., 2024).

Moreover, we observed that, although there is local regulatory movement, such as Law No. 20,707, to expand access to the test, its practical implementation remains incomplete, and this issue is present in current discussions about the ideal scope of testing. Recent guidelines from scientific societies have proposed offering BRCA testing to broad subgroups of breast cancer patients, even in the absence of a classic family history, with the aim of increasing the therapeutic and preventive impact of this strategy (Assad et al., 2024).

The cross-sectional design, convenience sample, and small number of respondents limit generalizability and the inferential power of the study. The presence of categories with very small counts compromises the stability of statistical estimates and prevents causal conclusions. In addition, the responses were self-reported, which may introduce recall or interest bias. Some key variables showed little variability, such as knowledge about mapping, reducing their usefulness for explaining differences in clinical practice.

For these reasons, the results should be interpreted as descriptions of trends and as a source of hypotheses to guide further investigations and primary interventions, rather than as definitive evidence of associations or effects.

5. Conclusion

This exploratory study preliminarily showed that, among oncologists, breast specialists, and geneticists in Goiás, genetic mapping (BRCA1/BRCA2) is widely recognized and is mainly requested in the presence of classic clinical criteria such as early diagnosis, family history, and aggressive tumor phenotypes. However, routine incorporation of the test into clinical practice remains partial, being limited by attitudinal and operational barriers, such as perceived low benefit, uncertainty about cost-effectiveness, and gaps in genetic counseling.

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ISSN 2178-6925

Received: 13/01/2026 - **Accepted:** 07/05/2026
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Given the descriptive nature and small sample size, the findings should be interpreted as preliminary indications of trends and as a basis for intervention hypotheses, not as conclusive evidence. For the normative availability of the test to translate into effective benefit for patients, it is recommended that multicenter studies with larger samples and a qualitative approach be prioritized. This would allow evaluation of whether professional training initiatives, strengthening of genetic counseling, standardization of care pathways, and implementation assessments that monitor service coverage and quality are feasible proposals.

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