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Free testing for BReast CAncer (BRCA1/2) gene mutations in Goiás: Distribution of specialist doctors

Testagem gratuita para mutações dos genes BRCA1/2 em Goiás: Distribuição de médicos especialistas

Prueba gratuita para mutaciones de los genes BRCA1/2 en Goiás: Distribución de médicos especialistas

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Abstract
Objective: This study aimed to assess the number of medical oncologists, mastologists, and geneticists registered with the Regional Medical Council of the State of Goiás (CREMEGO). Methods: This study was conducted in August 2025 as a cross-sectional, quantitative investigation using the public CREMEGO database. Active and inactive registrations of oncologists, mastologists, geneticists, and platform variations were included. Data were tabulated in Excel and analyzed using descriptive statistics. Results: A total of 285 registrations were identified, with 81.4% of them being active. Among active registrations, mastologists accounted for 33.7%, and clinical oncologists accounted for 32.3%. Geneticists were poorly represented (n = 08). A concentration of professionals was observed in Goiânia, with 69.4% of active registrations located in the capital, reaching 100% in some subspecialties. Conclusion: The records indicate that specialists exist in Goiás, but they are centralized in the capital, creating access barriers to genetic testing and counseling for municipalities in the interior and potentially hindering the full implementation of Law Number (No.) 20.707. We propose expanding specialist training in genetic counseling, promoting telemedicine, and decentralizing services to reduce health disparities and inequalities.
Keywords: Breast Neoplasms; Genetic Counseling; Women's Health.

Resumo
Objetivo: o presente estudo verificou o número de médicos oncologistas, mastologistas e geneticistas com registro no Conselho Regional de Medicina do Estado de Goiás (CREMEGO). Metodologia: Estudo observacional, transversal, quantitativo, realizado em agosto de 2025, a partir da base pública do CREMEGO. Foram incluídos registros ativos e

inativos de oncologistas, mastologistas e geneticistas e variações da plataforma. Os dados foram tabulados em Excel e analisados por estatística descritiva. Resultados: Foram identificados 285 registros, 81,4% estão ativos. Entre estes, mastologistas (33,7%) e oncologistas clínicos (32,3%). Houve baixa representatividade de geneticistas (n=08). Observou-se concentração de profissionais em Goiânia, com 69,4% dos ativos, chegando a 100% em algumas subespecialidades. Essa concentração contrasta com a demanda estadual prevista na Lei nº 20.707. Conclusão: Os registros mostram que há especialistas em Goiás, porém centralizados na capital, o que cria barreiras de acesso ao teste genético e ao aconselhamento para municípios do interior e pode dificultar a aplicação plena da Lei nº 20.707. Propõe-se ampliar a capacitação dos especialistas em aconselhamento genético, promover telemedicina e descentralizar serviços para reduzir a desigualdade.
Palavras-chave: Neoplasias da Mama; Aconselhamento Genético; Saúde da Mulher.

Resumen
Objetivo: El presente estudio verificó el número de médicos oncólogos, mastólogos y geneticistas con registro en el Consejo Regional de Medicina del Estado de Goiás (CREMEGO). Metodología: Estudio observacional, transversal y cuantitativo, realizado en agosto de 2025, a partir de la base pública del CREMEGO. Se incluyeron registros activos e inactivos de oncólogos, mastólogos y geneticistas, así como variaciones de la plataforma. Los datos fueron tabulados en Excel y analizados mediante estadística descriptiva. Resultados: Se identificaron 285 registros, de los cuales el 81,4% estaban activos. Entre estos, destacaron los mastólogos (33,7%) y los oncólogos clínicos (32,3%). Se observó una baja representatividad de geneticistas (n = 8). Se constató una concentración de profesionales en Goiânia, con el 69,4% de los registros activos, alcanzando el 100% en algunas subespecialidades. Esta concentración contrasta con la demanda estatal prevista en la Ley n.º 20.707. Conclusión: Los registros muestran que existen especialistas en Goiás, pero centralizados en la capital, lo que genera barreras de acceso a las pruebas genéticas y al asesoramiento para los municipios del interior y puede dificultar la plena aplicación de la Ley n.º 20.707. Se propone ampliar la capacitación de los especialistas en asesoramiento genético, promover la telemedicina y descentralizar los servicios para reducir la desigualdad.
Palabras clave: Neoplasias de la Mama; Asesoramiento Genético; Salud de la Mujer.

1. Introduction

Breast cancer is not only the most common malignancy among women but also one of the leading causes of death among women worldwide (Yan et al., 2020). Early detection approaches for breast cancer aim to diagnose the disease at an initial stage, which leads to a more favorable prognosis and a reduction in treatment-related morbidity; consequently, countries such as Brazil have implemented, since the mid-20th century, control strategies for this highly prevalent cancer (Brasil, 2015; Rodrigues et al., 2021; Almeida et al., 2023).

The genetics of breast cancer has become a fundamental aspect of its management. Genetic information influences screening, follow-up, prophylactic recommendations, and therapeutic decisions for women who carry susceptibility genes for breast cancer. Additionally, it helps identify patient subgroups with different prognoses or treatment responses. Recent studies have shown that germline variants of the *BRCA1/2* genes are clinically relevant for patients already diagnosed with this type of cancer (Criscitiello & Corti, 2022; Dantas Junior et al., 2022; Pantoja et al., 2022).

BRCA stands for “BReast CAncer” gene, indicating its relevance to breast cancer pathogenesis. The gene itself, however, does not induce breast cancer by itself. BRCA1 and BRCA2 are tumor suppressor genes essential for the activation of Deoxyribonucleic acid (DNA) repair in response to cellular stress and for the maintenance of genomic stability. They play important roles in chromatin remodeling, transcriptional control, cell-cycle regulation, and DNA repair processes; their tumor suppressor effects have been primarily attributed to cell-cycle checkpoint control and management of DNA repair. The *BRCA1* gene is located on chromosome 17q21 and comprises twenty-two exons, while *BRCA2* is located on chromosome 13q12-13 and comprises twenty-seven exons (Criscitiello & Corti, 2022; Rivera Rivera et al., 2023).

Pathogenic variants detected in *BRCA1* (MIM no. 113705) and *BRCA2* (MIM no. 600185) are inherited in an autosomal dominant manner and account for 5%–10% of all breast cancer cases. Pathogenic variants in these genes are associated with an increased risk of breast cancer of 55% to 72%, and a 26% to 83% chance of contralateral breast cancer. More than 1,600 variants are known in *BRCA1* and 1,800 in *BRCA2*. Genetic counseling and testing must be individualized

according to each patient's history. Such testing provides greater support and information to patients, assisting decisions regarding lifestyle and/or prophylactic measures (Bokkers et al., 2023; Rivera Rivera et al., 2023).

With growing knowledge about the benefits of genetic testing, the State of Goiás enacted Law No. 20.707 on 14 January 2020, authorizing the implementation—together with the Brazilian Unified Health System (Sistema Único de Saúde, SUS), of *BRCA1* and *BRCA2* mutation testing for women with a personal or family history of breast or ovarian cancer, when requested by a medical geneticist, mastologist or oncologist. Access is conditional on presentation of documentation proving the patient's personal history, or a family history of breast and/or ovarian cancer diagnosed before the age of 50 in two blood relatives in a direct line or collateral up to the third degree, or a personal report of breast and/or ovarian cancer with a primary tumor diagnosed before age 40, or triple-negative tumor diagnosed before age 50 (Goiás, 2020).

This legislation came into effect in October 2023, following a cooperation agreement between the Federal University of Goiás and the State Health Secretariat, establishing Goiás as a pioneer in offering this type of test free of charge in the country. However, at present, the availability of this service is limited to the state capital (Carvalho & Versanna, 2025; Lima et al., 2026).

Given this context, this study aimed to assess the number of medical oncologists, mastologists, and geneticists registered with the Regional Medical Council of the State of Goiás (CREMEGO).

Therefore, this study analyzes the incorporation of genetic testing into the therapeutic strategy for patients with breast cancer in Goiás, since these professionals play a central role in requesting and interpreting the free genetic test provided for by the law. This approach aims to provide evidence to understand how many specialists can operate the new public policy and thus contribute to the effectiveness of early detection of breast cancer in the state.

2. Methodology

This is an observational, cross-sectional, descriptive study with a quantitative approach (Pereira et al., 2018; Risemberg et al., 2026) using descriptive statics with absolute frequency in quantity and relative frequency in percentage (Shitsuka et al., 2014) in data of a population consisting of physicians registered with CREMEGO, regardless of their municipality of practice within the State of Goiás. Data were obtained from the public registry database of CREMEGO, available on the council's official website (https://www.cremego.org.br/).

The query was performed directly using the public consultation/physician portal of CREMEGO. Using the "Search for physicians" tool under "Citizen services" on the site, we applied the following filter combination: State: GO (Goiás); Municipality: All; Registration type: All; Status: Active; Specialties: Oncology, Clinical Oncology, Cancerology, Surgical Oncology; Mastology; Medical Genetics, Laboratory Genetics, and Clinical Genetics. The process was repeated to search inactive registrations and then active registrations restricted to the municipality of Goiânia. The extracted data corresponds to the official records maintained by the institution up to August 2025 and were tabulated in spreadsheets.

Included were records of physicians with either active or inactive registration in CREMEGO whose primary specialty, at the date of extraction, was registered as Oncology, Mastology, or Medical Genetics, including the specialty variations available in the platform filters (Clinical Oncology, Cancerology, Surgical Oncology; Laboratory Genetics and Clinical Genetics). Registrations for physicians practicing in states other than Goiás were excluded.

Data processing began with characterization of the sample, grouping, data slicing, and compilation into thematic units for interpretation and discussion of the findings. Analyses were performed using descriptive statistics expressed as percentages. This study used publicly available data posted on the council's official website. No data identifying professionals or additional contact information was collected; therefore, all confidentiality norms were respected. This study is part of a larger project and

was approved by the Research Ethics Committee of the Evangelical University of Goiás (CAAE 84082424.9.0000.5076, process 7.413.928).

3. Results

This study examined the registry categories or specialties that CREMEGO groups as oncologists, mastologists, and geneticists. Table 1 presents the observed values in the analysis, which focused on quantifying active and inactive registrations by category/specialty based on the total number of records, which was 285. Of this total, 81.4% were active and 18.6% inactive.

Mastology occupied first place in the number of active registrations, with 33.68%, followed by clinical oncology with 32.28%; however, this ranking is reversed among inactive registrations, where clinical oncology had the highest number (8.07%) and mastology the second highest (5.61%). It is also notable that the variety of categories established by CREMEGO results in some specialties with no registrations (such as Oncology and Laboratory Genetics), or very low numbers compared with the total, Clinical Genetics had only one physician with an active registration, and Cancerology had two. Considering only the broad categories of oncologists, mastologists, and geneticists, oncologists account for the largest number of registrations, with a substantial difference compared with geneticists, who present the smallest number, highlighting the numerical disparity among the different specialties addressed in Law No. 20.707.

Table 1 - Distribution of active and inactive physicians registered with CREMEGO by category/specialty.

CATEGORY / SPECIALTY	ACTIVE		INACTIVE	
	n	%*	n	%*
Oncologists				
Oncology	0	0.00	0	0.00
Clinical Oncology	92	32.28	23	8.07
Cancerology	02	0.70	01	0.35
Cancerology / Surgical Cancerology	34	11.93	10	3.51
Mastologists				
Mastology	96	33.68	16	5.61
Geneticists				
Medical Genetics	07	2.46	03	1.05
Laboratory Genetics	0	0.00	0	0.00
Clinical Genetics	01	0.35	0	0.00
TOTAL	**232**	**81.40**	**53**	**18.60**

* % of the overall total (n = 285)
Source: CREMEGO website (August 2025).

Considering only active physicians, a strong concentration was observed in the state capital. Table 2 shows that, of the 232 active professionals, 161 were registered in Goiânia, while 71 were distributed across other municipalities in Goiás. This concentration pattern was particularly evident in certain specialties, such as Cancerology, in which all active practitioners worked exclusively in the capital, and Cancerology / Surgical Cancerology, in which more than 90% of registrations corresponded to Goiânia. In the broader breakdown, 97 of the 128 active oncologists are based in the capital, as are 60 of the 96 mastologists and 4 of the 8 geneticists. Additionally, 57% of active specialists in Medical Genetics are concentrated in the state capital, Goiânia.

Table 2 - Distribution of active physicians in the State of Goiás and in the capital Goiânia registered with CREMEGO by category/specialty.

CATEGORY / SPECIALTY	ASSETS IN THE STATE	ASSETS IN THE CAPITAL	
	Goiás (n=232)	Goiânia (n=161)	% Active
Oncologists			
Oncology	0	0	0.00
Clinical Oncology	92	64	69.57
Cancerology	02	02	100.00
Cancerology / Surgical Cancerology	34	31	91.18
Mastologists			
Mastology	96	60	62.50
Geneticists			
Medical Genetics	07	04	57.14
Laboratory Genetics	0	0	0.00
Clinical Genetics	01	0	0.00

Source: CREMEGO website (August 2025).

4. Discussion

This study sought to evaluate the incorporation of genetic testing into the therapeutic strategy for patients with breast cancer in Goiás by verifying the number of oncologists, mastologists, and geneticists registered with CREMEGO, since these professionals may request genetic testing and provide the required report under Law No. 20.707. The results of this study are consistent with international literature highlighting an insufficient number of specialists in the addressed areas and their concentration in state capitals (Goiás, 2020; Carvalho & Versanna, 2025; CREMEGO, 2025; Silva et al., 2025; Zegarra-López et al., 2025; Lima et al., 2026).

The sample comprised 285 specialists listed on CREMEGO's official website. The majority were active physicians, and the largest single specialty was mastology. Regarding geographic distribution within the state, a greater number of professionals practice in Goiânia, which can be understood because of the distribution of residency programs in Latin America, most of which are in state capitals (Zegarra-López et al., 2025).

The findings of this work contribute to research showing the unequal distribution of physicians and specialists between urban and rural areas, particularly concentrated in capitals, even when compared with other Organization for Economic Co-operation and Development (OECD) countries. Some studies go further, describing how oncology fields suffer from poor distribution of medical residency positions, which hinders access to specialized services for populations in rural regions (OECD, 2021; Horovitz et al., 2024; Zegarra-López et al., 2025).

This predominance of active specialists concentrated in the capital, Goiânia, may hinder full implementation of Law No. 20.707. Although the law allows the State to propose technical cooperation with municipalities for performing genetic testing, initiation of the process requires a report from an oncologist, mastologist, or geneticist. Because these professionals are largely concentrated in the capital, obtaining such reports may be difficult for people living in other cities (Goiás, 2020, Lima et al., 2026).

One avenue to overcome geographic barriers in oncologic follow-up is telemedicine. A study that analyzed data

collected during the COVID-19 pandemic, when geographic issues had to be rapidly addressed, concluded that telemedicine is indeed a valuable alternative to improve the distribution of specialized care across countries; however, this requires regulations and supporting public policies (Garavand et al., 2023).

Another concern is the small number of specialists qualified to publish the reports recommended by the law. Many may be unaware of the new legislation or not trained in genetic counseling, which is expected to occur in conjunction with testing; this underscores the need to train physicians so that the law can be used optimally to individualize treatments (Goiás, 2020; Silva Filho et al., 2020; Ashton-Prolla et al., 2024; Silva et al., 2025).

Moreover, the proportion of inactive registrations (18.6%) may be high: studies indicate that in states with few specialists, a rising number of inactive physicians could worsen workforce shortages over time, since working conditions and mental health burdens among oncology professionals can contribute to reduced active workforce through reduced hours or withdrawal from practice (Mendes et al., 2024).

This is the first study to analyze the number of physicians eligible to operate under Law No. 20.707 for the incorporation of genetic testing in Goiás. No publications were identified that reference this law and directly describe the distribution of specialists able to contribute. Thus, the results are clearly relevant, highlighting the importance of a more detailed analysis of geographic barriers between professionals and Law No. 20.707, and emphasizing the need for physicians to be aware of the availability of free genetic testing to reduce barriers to testing (Goiás, 2020; Lima et al., 2026).

The way CREMEGO's website categorizes and displays medical specialties was considered a limitation, as it encompasses multiple nomenclatural variations that may be misinterpreted as specialties or subspecialties. The lack of filters such as "sex" and "age" also hinders more in-depth analyses of the sociodemographic profiles of registered physicians. Future studies should be conducted with scientific rigor and should seek solutions for the geographic and informational distribution of the medical workforce in Goiás, Brazil, and internationally.

5. Conclusion

This study showed that most recordings of oncologists, mastologists, and geneticists in Goiás are active but concentrated in the state capital. This indicates an unequal distribution of specialists available to provide genetic testing in the state. Such disparity may impede access to genetic testing and genetic counseling in inland municipalities, complicating the implementation of Law No. 20.707. We recommend broad training in genetic counseling for specialist physicians, adoption of telemedicine, and decentralization of specific services. As a limitation, this study relied on the nomenclature used by CREMEGO on its website, and it was not possible to assess actual competency for counseling or patients' effective access to specialist physicians.

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