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Exploring the role of genomic literacy on breast cancer prevention

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Dissertação apresentada para cumprimento dos requisitos necessários à obtenção do grau de Mestre em Promoção da Saúde, realizada sob orientação científica da Professora Doutora Ana Rita Pedro

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"Mesmo que a rota da minha vida me conduza a uma estrela, nem por isso fui dispensado de percorrer os caminhos do mundo."
José Saramago

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Abstract

Background

Breast cancer is the most common cancer and one of the leading causes of cancer-related mortality among women in Portugal. Mammography screening has been shown to reduce breast cancer mortality, but participation rates remain low due to barriers such as fear, lack of knowledge, logistical challenges, and health literacy. Recently, the concept of genomic literacy has emerged; however, its specific influence on preventive behaviors, including breast cancer screening adherence, has not been explored. This study aims to examine the association between genomic literacy and breast cancer screening among women in Portugal, while also considering other factors such as health literacy, sociodemographic characteristics, perceived risk, and individual risk level.

Methods

An online self-administered questionnaire collected data on sociodemographic characteristics, breast cancer risk, risk perception, health literacy, and genomic literacy. Breast cancer screening adherence was defined according to national guidelines as having undergone a mammogram in the previous two years (for women aged 45–70). Statistical analyses included T-tests, ANOVA, Chi-square tests, and a logistic regression to identify factors associated with screening adherence.

Results

The final sample included 359 women (mean age=46.2, SD=9.8). Higher education and income levels were significantly associated with higher GL scores ($p<0.001$). However, GL was not a significant predictor of BC screening adherence in bivariate or multivariate analyses. The strongest predictor of non-adherence was the setting where the last mammogram was performed. Women screened in NHS or private facilities were more likely to be non-adherent compared to those screened in mobile units (OR=22.32 and OR=10.65, respectively). Other factors, including HL, income, education, and risk perception, were not significant predictors.

Conclusions

While women with higher education and income showed better genomic literacy, this was not linked to greater screening adherence. Adherence was primarily related to the setting of the last mammogram, with women screened in mobile units showing greater compliance. This highlights the importance of reducing organizational barriers to improve participation. However, given the convenience sampling and overrepresentation of women with higher education in this study,

further research with more representative samples is needed to confirm and expand these findings.

Keywords: genomic literacy, breast cancer, prevention, health literacy, health promotion

Resumo

Contexto

O cancro da mama é o cancro mais comum e uma das principais causas de mortalidade por cancro entre as mulheres em Portugal. Está demonstrado que a mamografia reduz a mortalidade por cancro da mama, mas as taxas de participação continuam baixas devido a barreiras como o medo, falta de conhecimento, desafios logísticos e literacia em saúde. Recentemente, surgiu o conceito de literacia genómica; contudo, a sua influência específica nos comportamentos preventivos, incluindo a adesão ao rastreio do cancro da mama, ainda não foi explorada. Este estudo tem como objetivo analisar a associação entre a literacia genómica e o rastreio do cancro da mama entre mulheres em Portugal, considerando também outros fatores como literacia em saúde, características sociodemográficas, perceção de risco e nível de risco.

Métodos

Foi aplicado um questionário online de autopreenchimento para recolher dados sobre características sociodemográficas, risco de cancro da mama, perceção de risco, literacia em saúde e literacia genómica. A adesão ao rastreio do cancro da mama foi definida de acordo com as orientações nacionais, ou seja, ter realizado uma mamografia nos dois anos anteriores (para mulheres entre os 45 e os 70 anos). As análises estatísticas incluíram testes t, ANOVA, testes do qui-quadrado e uma regressão logística para identificar fatores associados à adesão ao rastreio.

Resultados

A amostra final contou com 359 participantes (idade média=46,2 anos; DP=9,8). Verificou-se que níveis mais elevados de escolaridade e rendimento se associaram significativamente a maiores pontuações de literacia genómica ($p<0,001$). Contudo, a literacia genómica não se revelou um preditor significativo da adesão ao rastreio mamográfico, quer nas análises bivariadas, quer nas multivariadas. O fator mais determinante da não adesão foi o local da última mamografia. As mulheres que realizaram o rastreio no SNS ou em instituições privadas apresentaram maior probabilidade de não adesão, em comparação com as que recorreram a unidades móveis (OR=22,32 e OR=10,65, respetivamente). Outros fatores, como literacia em saúde, rendimento, escolaridade e perceção de risco, não demonstraram impacto significativo na adesão.

Conclusões

Embora se tenha observado que mulheres com maior escolaridade e rendimento apresentam melhor literacia genómica, tal não se traduziu numa maior probabilidade de adesão ao rastreio do cancro da mama. Os resultados sugerem que a adesão está mais fortemente associada a

fatores organizacionais, nomeadamente ao local onde é realizado o rastreio, com maior cumprimento observado nas unidades móveis. Estes achados destacam a importância de mitigar barreiras de natureza organizacional para promover a participação no rastreio. No entanto, importa sublinhar que a utilização de uma amostra de conveniência, com sobre-representação de mulheres com maior escolaridade, limita a generalização dos resultados, sendo necessária investigação futura com amostras mais representativas.

Palavras-chave: literacia genómica; cancro da mama; rastreio; prevenção; promoção da saúde

Table of Content

Part I – Introduction

1. Breast cancer epidemiology and risk factors

In the latest report from the Global Cancer Observatory, breast cancer (BC) was the second most common cancer diagnosed and the fourth cause of death by cancer, representing 2 296 840 new cases and 666 103 deaths (1). In Portugal, it is the most common type of cancer diagnosed in women and one of the main causes of death by cancer. Additionally, it represents 11% of the treatable causes of mortality, meaning that mortality rates can decrease with adequate screening and treatment (2).

Several factors contribute to the risk of developing BC, both modifiable and non-modifiable. Two very relevant risk factors for BC are non-modifiable – gender and age. Although a man can develop BC, it is a condition a lot more frequent in women due to its etiology (4). Being 50 or older also raises the risk of developing BC (4) because of physiological changes that happen with aging, such as in the mammary gland epithelium and hormone production (5,6). Other non-modifiable factors are the age of menarche and menopause since early menarche and late menopause contribute to greater exposure to estrogen, which promotes tumor growth (7). Having dense breasts and, consequently, more milk ducts also contributes to a higher risk, as BC is usually developed in these structures (8). Lastly, genetic variants in genes such as *BRCA1* and *BRCA2* also contribute to a greater risk. Although only 5-10% of BC cases are hereditary, several studies have shown that variants in these genes contribute to an increased risk of developing BC from 60% to 72% (9–11).

Looking at modifiable factors, some have been deeply explored, such as weight, alcohol consumption, and smoking. A dose-response meta-analysis by Hao *et al*. examined 16 cohort studies, finding that the risk of developing postmenopausal BC increases by 8% per every 5kg of weight gain (12). Concerning alcohol consumption, total alcohol intake, particularly wine, has also been found to increase the risk of postmenopausal BC (13). Finally, smoking, mainly if the woman started smoking more than 10 years before having her first child, increases the risk of developing breast cancer, specifically ER+ (14).

2. Breast cancer prevention

2.1. Traditional Breast Cancer Prevention

As mentioned, several modifiable factors contribute to the risk of developing BC. Consequently, the primary prevention of BC consists of adopting a healthy lifestyle, with an adequate diet such as the Mediterranean diet (15), and practicing physical activity (16).

The secondary prevention of BC is also essential, as it increases the chances of successful treatment and reduces mortality. Secondary prevention consists of an early diagnosis by mammography. In Portugal, the National Program for Oncological Diseases recommends having a mammography every two years in women from 45 to 69 years old. However, according to the National Program for Oncological Diseases Evaluation Report of 2023, only 51% of eligible women attended the screening, and a decrease has been observed in the last years (17). This trend raises concern since this screening strategy has been proven to effectively reduce the mortality rate by BC (18–22). Therefore, it is important to understand the causes underlying the low attendance.

Several studies have explored the barriers to screening, revealing factors such as fear of the test, fear of the results, lack of knowledge, and geographical barriers, for example, being distant from the testing center (23–25). Another factor important to mention is health literacy (HL), which is defined as the ability to "*access, understand, appraise and apply health information in order to make judgments and take decisions in everyday life concerning healthcare, disease prevention and health promotion to maintain or improve quality of life during the life course*" (26). HL has been associated with lower screening awareness (27), lower attendance rates (28), and stronger perceived barriers to screening (29).

Perceived risk is another important factor used in models to explain health behaviors. There is a framework in health behavior change research – The Health Belief Model of Behavior Change – which considers the perceived risk of developing a disease as a precursor to taking preventive measures (30). A study in Ontario has shown that women with higher perceived risks of developing BC were more likely to have taken the screening within the year before, enhancing the contribution of risk perception for screening behaviors (31).

2.2. Personalized Prevention of Breast Cancer

Mammography has all the characteristics needed for a screening test, such as being widely available and being able to detect BC at an early stage (32). Therefore, it is a suitable test for organized screening programs. However, these organized programs are insufficient, as age is the only inclusion factor. For women at high risk, particularly those with *BRCA1/2* variants, starting the screening at age 45 through organized programs is irrelevant, as most get diagnosed before that age (33).

Is it recommended that women are submitted to a risk assessment by the age of 25-30, to begin screening if necessary (34,35). This can be done in a Primary Care setting, or at a Gynecology consult, by gathering all the family history and other risk factors (36). With this risk assessment, the physician can determine if a woman has an average, moderate, or high risk for developing BC.

For those at high risk, genetic counseling can be suggested (36). Genetic testing is a key tool in addressing hereditary cancer syndromes. Detecting pathogenic variants, such as *BRCA1* or *BRCA2*, can significantly impact prevention and treatment decisions. For example, *BRCA1* variants are strongly associated with aggressive phenotypes like triple-negative breast cancer (37), highlighting the importance of personalized prevention approaches based on genetic information. However, genetic testing is still not a current practice for prevention. It is performed mainly when a woman is diagnosed with specific BC types, such as bilateral or triple-negative, or when the woman is diagnosed at an early age and has a family history (38).

In 2019, the US recommended expanding preventive measures for women at high risk of hereditary cancer, even without a formal diagnosis (39). Similarly, in Portugal, interest in genetic testing for high-risk women has been increasing. As part of the National Strategy Against Cancer 2021-2030, several national objectives and strategies address hereditary cancer syndromes. Notably, the strategy includes a goal to publish guidelines for the early detection of Hereditary Breast Cancer Syndrome linked to *BRCA1* or *BRCA2* mutations.

3. Genomic literacy

Genetic testing provides an opportunity to tailor prevention and treatment strategies, particularly for individuals at high risk of BC. However, the effective implementation of these personalized approaches relies not only on the availability of genetic tools but also on the ability of individuals and healthcare professionals to understand and apply genetic information. This brings focus to the concept of genomic literacy (GL). GL is defined as "*the knowledge needed to:*

1) make an informed decision for genomic testing, 2) appropriately apply genomic technologies and accurately interpret genomic data, and 3) participate in decisions about genetics and genomics policy questions as a member of society" (40).

Since lower HL has been associated with poor health status, Nakamura *et al*. hypothesized that GL might also correlate with health status. These authors were the first ones to explore such a relationship. In their study, individuals with lower GL presented a higher risk of hypertension and obesity, suggesting that GL might correlate with the risk for non-communicable diseases associated with a genetic risk (41).

Given the significant genetic predisposition associated with BC, GL may have a role in its prevention. Women with a family history of BC will significantly benefit from more effective preventive measures if they have an adequate level of GL.

Since no studies have explored the correlation between GL and BC prevention, this study aims to bridge this gap and evaluate this association in the context of the Portuguese population. To achieve this general goal, the study also seeks to address the following specific objectives:

1. Assess the level of genomic literacy among women living in Portugal aged 25 to 70 years;
2. Identify factors that contribute to genomic literacy in women living in Portugal aged 25 to 70 years;
3. Investigate determinants that influence the adoption of BC prevention practices among women living in Portugal aged 25 to 70 years;
4. Characterize genomic literacy levels across different BC risk categories within the study population.

In an era where the potential of personalized prevention has started to gain focus, this study ultimately aims to contribute to the understanding of GL within the Portuguese population. By assessing GL levels and exploring their relationship with BC prevention, the findings of this study may serve as a foundation for future studies, potentially guiding initiatives focused on education and awareness in this area. This research aligns with the United Nations' Sustainable Development Goal 3 (SDG 3), which seeks to ensure healthy lives and promote well-being for all. By generating knowledge about population-level GL and its determinants, this study contributes to identifying needs and inequalities that may influence access to information and participation in preventive health measures. This kind of evidence is essential to support the design of inclusive, equitable, and literacy-sensitive health promotion strategies, particularly in a context where genomic information is expected to play an increasingly important role in public health and cancer prevention.

The study also aligns with the frame of the Master's in Health Promotion, which is grounded in multidisciplinary approaches, evidence-based practice, and ethical values. By addressing emerging challenges in health literacy and cancer prevention, this research reflects the program's commitment to generating knowledge that supports more equitable, informed, and sustainable health promotion strategies at both individual and community levels.

Part II – Article

Title: Adherence to Breast Cancer Screening: Exploring the Impact of Genomic Literacy in the Portuguese population

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Abstract

Background

Breast cancer is the most common cancer and one of the leading causes of cancer-related mortality among women in Portugal. Mammography screening has been shown to reduce breast cancer mortality, but participation rates remain low due to barriers such as fear, lack of knowledge, logistical challenges, and health literacy. Recently, the concept of genomic literacy has emerged; however, its specific influence on preventive behaviors, including breast cancer screening adherence, has not been explored. This study aims to examine the association between genomic literacy and breast cancer screening among women in Portugal, while also considering other factors such as health literacy, sociodemographic characteristics, perceived risk, and individual risk level.

Methods

An online self-administered questionnaire collected data on sociodemographic characteristics, breast cancer risk, risk perception, health literacy, and genomic literacy. Breast cancer screening adherence was defined according to national guidelines as having undergone a mammogram in the previous two years (for women aged 45–70). Statistical analyses included t-tests, ANOVA, chi-square tests, and a logistic regression to identify factors associated with screening adherence.

Results

The final sample included 359 women (mean age=46.2, SD=9.8). Higher education and income levels were significantly associated with higher GL scores ($p<0.001$). However, GL was not a significant predictor of BC screening adherence in bivariate or multivariate analyses. The

strongest predictor of non-adherence was the setting where the last mammogram was performed. Women screened in NHS or private facilities were more likely to be non-adherent compared to those screened in mobile units (OR=22.32 and OR=10.65, respectively). Other factors, including HL, income, education, and risk perception, were not significant predictors.

Conclusions

While women with higher education and income showed better genomic literacy, this was not linked to greater screening adherence. Adherence was primarily related to the setting of the last mammogram, with women screened in mobile units showing greater compliance. This highlights the importance of reducing organizational barriers to improve participation. However, given the convenience sampling and overrepresentation of women with higher education in this study, further research with more representative samples is needed to confirm and expand these findings.

Keywords: genomic literacy, breast cancer, prevention, health promotion, health literacy

Background

Breast cancer (BC) is the leading cause of cancer among women in Portugal and one of the main causes of death by cancer, representing 11% of the treatable causes of death in the country (1). Several factors contribute to the risk of developing BC. Some, like age (2), sex (2), age of menarche and menopause (3), and the presence of genetic variants in *BRCA1* and *BCRA2* (4-6) are not modifiable. While others, such as weight (7), alcohol consumption (8), and smoking (9), are modifiable.

The primary prevention of BC consists of controlling these modifiable factors by embracing a healthy lifestyle, such as adopting a Mediterranean diet (10) and practicing physical activity (11). Secondary prevention lies in the early diagnosis by mammography, which has been proven to reduce the mortality rate by BC (12 - 17). However, conventional screening programs may be inadequate for high-risk women, who are often diagnosed earlier (18). Current organized programs in Portugal are age-based, potentially overlooking individual risk profiles. Therefore, assessing individual BC risk, through personal and family history, is essential to tailor prevention and screening recommendations more effectively.

Several studies have explored the barriers to screening, identifying factors such as fear of the test or its results, lack of knowledge, and logistical challenges, including geographical distance

from screening centers (19-21). Health literacy (HL), defined as the ability to "*access, understand, appraise and apply health information in order to make judgments and take decisions in everyday life concerning healthcare, disease prevention and health promotion to maintain or improve quality of life during the life course*" (22), has been associated with lower screening awareness (23), lower attendance rates (24), and stronger perceived barriers to screening (25).

With advancements in science, personalized prevention has gained relevance. Genetic testing provides an opportunity to tailor prevention and treatment strategies, particularly for individuals at high risk of BC. However, the effective implementation of these personalized approaches relies not only on the availability of genetic tools but also on the ability of individuals to understand and apply genetic information. This brings focus to the concept of genomic literacy (GL). GL is defined as "*the knowledge needed to: 1) make an informed decision for genomic testing, 2) appropriately apply genomic technologies and accurately interpret genomic data, and 3) participate in decisions about genetics and genomics policy questions as a member of society*" (26).

Since lower HL has been associated with poor health status, Nakamura *et al*. hypothesized that GL might also correlate with health status. These authors were the first ones to explore such a relationship. In their study, individuals with lower GL presented a higher risk of hypertension and obesity, suggesting that GL might correlate with the risk for non-communicable diseases associated with a genetic risk (27).

Additionally, perceived risk has been shown to influence health behaviors. According to the Health Belief Model, individuals who perceive themselves as being at high risk are more likely to engage in preventive actions (28). Evidence suggests that a higher perceived risk of BC is associated with greater likelihood of undergoing screening (29). Despite this, the combined influence of perceived risk, risk level, and GL on BC prevention has not yet been explored.

This study aims to bridge this gap by examining the association between GL and BC prevention behaviors among women living in Portugal, while considering how other factors, such as demographic characteristics, HL, perceived risk, and actual risk levels, influence these behaviors.

Methods

Aim, design and setting of the study

This study aims to explore the relationship between GL and BC prevention behaviors among women living in Portugal. A self-administered questionnaire was developed and distributed online between February 17th and March 31st, 2025. The research was conducted as an observational and descriptive design, adopting a cross-sectional perspective with a quantitative approach.

Participants

The target population consisted of women residing in Portugal, aged between 25 and 70 years, consisting of 3 261 400 women (30). Using a 90% confidence level and a margin of error of 5%, a minimum sample size of 273 was calculated based on sample size calculation.

Due to time and resource constraints, a convenience sampling method was adopted, and participants were recruited through social media. The inclusion criteria were:

1. Female sex;
2. Age between 25 and 70 years;
3. Residency in Portugal.

All women with a previous BC diagnosis were excluded.

Data collection and measurements

The survey contained three sections. In the first one, risk stratification was made according to BC family history, and BC screening behaviors were assessed, based on national recommendations. The risk assessment was grounded in the recommendations of the National Association of General Practitioners (31) and was validated through a discussion with a radiology doctor specialist in BC. This section also explored BC risk perception and reasons for lower mammography attendance. The second section contained the GKnowM Genomics Knowledge Scale (26), and the third included all characterization questions and the HLS-EU-Q6 instrument for HL (32).

Genomic literacy scale

GKnowM Genomics Knowledge Scale was translated to Portuguese using the translation-retroversion method (33). First, two independent bilinguals translated the scale into Portuguese,

followed by a comparison of the two versions by the investigator. Lastly, a retroversion was made, the differences were analyzed, and any necessary adjustments were made.

Data analysis

The statistical analysis was performed using Jamovi, an open statistical software. For descriptive statistics, mean, median, standard deviation (SD), and frequencies were calculated. To identify which factors influenced GL, t-tests were made between the GL level and HL level, and age. ANOVAs were done with the level of education, income, risk perception, and risk level.

Regarding BC screening behavior, a binary variable was created to indicate whether participants followed the screening guidelines, according to the national recommendations of the National Breast Cancer Screening Program, which advises biennial screening for women aged 45–74 (34). For HL, the HLS-EU-Q6 scores were dichotomized into inadequate (0–25) and adequate (26–50) levels, generating a binary variable for analysis.

To examine factors associated with screening adherence, independent samples t-tests were used to compare GL, age, and HL between those who complied with recommendations and those who did not. Chi-square tests were performed to assess associations between screening adherence and level of education, income, risk perception, risk level, and the setting in which the mammogram was conducted (NHS, private, or in a mobile screening unit operated by the Portuguese League Against Cancer). Lastly, a logistic regression model was performed to further examine predictors of screening attendance, including GL score, HL index, education level, income, risk level, and screening setting as independent variables.

Results

Descriptive Statistics

A total of 359 women responded to the survey, with a mean age of 46.2 years (SD=9.8). Most participants were from the districts of Setúbal (59.2%) and Lisbon (25.7%). Regarding education, 61.8% had higher education, 32.3% had completed secondary education, and 5.8% had only basic education. Table 1 summarizes the sociodemographic characteristics of the study population.

Although 359 participants were included in the study, as part of the survey, participants were asked whether they had ever undergone a mammogram following a doctor's recommendation. Younger women, who had not yet had a mammogram, were excluded from all the analyses of compliance with screening recommendations. Therefore, the total number included in these analyses is lower than the initial sample.

Table 1: Sociodemographic characterization of the study population (n=359)

	N	%
District		
Lisboa	92	25.7
Setúbal	212	59.2
Porto	13	3.6
Other	41	11.5
Level of Education		
Basic Education	21	5.8
Secondary Education	116	32.3
Higher Education	222	61.9
Income*		
Up to 870€	55	15.6
871€ to 1740€	202	57.2
1741€ to 2610€	70	19.8
More than 2610€	26	7.4

*Different n due to missing data

Associations with Genomic Literacy

Table 2 presents the associations performed with GL. The analysis showed statistically significant differences according to educational level, income, and HL. Participants with higher education had a higher mean GL score (M=43.5, SD=7.8) compared to those with secondary education (M=39.1, SD=6.0) and basic education (M=36.7, SD=5.0) ($p<0.001$). Similarly, participants with higher income presented higher GL. Those earning more than 2610€ had the highest mean score (M=45.9, SD=7.8), followed by those earning between 1741€ and 2610€ (M=43.5, SD=7.4), 871€ to 1740€ (M=41.3, SD=7.2), and up to 870€ (M=38.7, SD=5.6) ($p<0.001$). No statistically significant differences were observed regarding GL according to individual risk level (average, slightly increased, or high risk) ($p=0.839$).

Significant differences were also found between GL and HL ($p = 0.042$), with participants classified as having adequate HL showing slightly higher GL scores than those with inadequate HL (M=42.6 VS M=41.0).

Compliance with Breast Cancer Screening Recommendations

Table 3 presents the bivariate analysis of compliance with national screening recommendations. Regarding age, women who complied with the National Program recommendations had a higher mean age (M=53.7, SD=5.8) than those who did not comply (M=51.7, SD=6.9); however, this difference was not significant (p=0.108).

No significant differences were found in educational level between compliant and non-compliant women (p=0.918). Similarly, income did not show significant associations with compliance (p=0.436). Mean HL scores were slightly higher among compliant women compared to non-compliant women; however, these differences were not statistically significant (p=0.123). As for risk perception, no statistically significant differences were observed between compliant and non-compliant women (p=0.335). The setting where the last mammogram was done was the only factor associated with compliance. Women who did not comply were more frequently screened in the NHS (58.3% vs 25.6%; p=0.001).

Predictors of Screening Adherence: Logistic Regression Analysis

Lastly, a logistic regression analysis was performed to explore predictors of screening adherence (Table 4). The place where the mammogram was performed was a significant predictor: women who had their last mammogram in the NHS (OR=22.32, p=0.004) and in the private sector (OR=10.65, p=0.032) had higher odds of being non-adherent compared to those screened in the mobile units of the Portuguese League Against Cancer. Other predictors, including HL index, GL score, education level, income, and risk level, were not statistically significant.

Table 2: Bivariate analysis between genomic literacy and educational level, income, risk level, and health literacy (n=359)

	N (%)	Genomic Literacy Score Mean (SD)	p-value
Level of education			
Basic Education	21 (5.8)	36,7 (5.0)	**<0,001**
Secondary Education	116 (32.3)	39,1 (6.0)	
Higher Education	222 (61.9)	43,5 (7,8)	
Income*			
Up to 870€	55 (15.6)	38,7 (5,6)	
871€ to 1740€	202 (57.2)	41,3 (7,2)	**<0,001**
1741€ to 2610€	70 (19.8)	43,5 (7,4)	
More than 2610€	26 (7,4)	45,9 (7,8)	
Risk level*			
Average risk	259 (74)	41,9 (7,7)	0,839
Slightly increased risk	85 (24.3)	41,4 (6,8)	
High risk	6 (1.7)	41,1 (6,3)	
Health literacy*			
Adequate	186 (53.1)	42.6 (7.39)	**0.042**
Inadequate	164 (46.9)	41.0 (7.40)	

*Different n due to missing data

Table 3: Bivariate analysis between breast cancer screening compliance and age, education level, income, health literacy, genomic literacy, risk perception, and risk level (n=185)

	Complies		Does not comply		p-value
	N (%)	Mean (SD)	N (%)	Mean (SD)	
Age	160 (86,5)	53,7 (5,8)	25 (13,5)	51,7 (6,9)	0,108
Level of education					
Basic Education	12 (7,5)	N/A	2 (8,0)	N/A	0,918
Secondary Education	58 (36,3)		8 (32,0)		
Higher Education	90 (56,3)		15 (60,0)		
Income					
Up to 870€	25 (15,9)		1 (4,2)		
871€ to 1740€	79 (50,3)	N/A	14 (58,3)	N/A	0,436
1741€ to 2610€	36 (22,9)		7 (29,2)		
More than 2610€	17 (10,8)		2 (8,3)		
Health literacy index					
Adequate	91 (59.1)	N/A	11 (44)	N/A	0,157
Inadequate	63 (40.9)		14 (56)		
Genomic literacy score	160 (86,5)	40.8 (7.3)	25 (13,5)	43,3 (7.1)	0,122
Risk perception					
No risk	9 (5,9)	N/A	2 (8,3)	N/A	0,335
Average risk	121 (79,1)		21 (87,5)		
High risk	23 (15)		1 (4,2)		
Risk level					
Average risk	112 (70,4)	N/A	19 (79,2)	N/A	0,595
Slightly increased risk	44 (27,7)		5 (20,8)		
High risk	3 (1,9)		0 (0,0)		
Setting					
NHS	41 (25,6)	N/A	14 (58,3)	N/A	**0,001**
Private	54 (33,8)		8 (33,3)		
Mobile Units	65 (40,6)		2 (8,3)		

N/A – Not applicable

Table 4. Logistic regression for the predictors of not adhering to breast cancer screening (n=172)

Predictor	OR (CI 95%)	p-value
Genomic literacy score	1.022 (0.949 – 1.100)	0.572
Health literacy		
Inadequate - Adequate	1.493 (0.556 – 4.007)	0.426
Education level		
Higher education – Basic education	0.476 (0.066 – 3.443)	0.462
Secondary education – Basic education	0.606 (0.085 – 4.338)	0.627
Income		
Up to 870€ - 871€ to 1740€	0.183 (0.019 – 1.739)	0.139
1741€ to 2610€ - 871€ to 1740€	0.912 (0.261 – 3.193)	0.885
More than 2610€ - 871€ to 1740€	0.285 (0.030 – 2.693)	0.273
Risk level		
High risk – Average risk	6.22e-7 (0.000 – ∞)	0.991
Lightly increased risk – Average risk	0.447 (0.132 – 1.520)	0.197
Setting		
Private – Mobile Units	10.472 (1.207 – 90.870)	**0.033**
NHS – Mobile Units	23.349 (2.844 – 191.726)	**0.003**

Discussion

This study aimed to explore the relationship between GL and BC screening behaviors among women living in Portugal, based on the National Breast Cancer Screening Program recommendation. It also aimed to investigate a range of factors that may contribute to GL as well as determinants that can contribute to BC screening adherence.

Regarding the factors that contribute to GL, the results revealed that women with higher education and income levels present higher GL scores. While research exploring the drivers of GL is lacking, several studies on HL have identified comparable socio-demographic influences. For example, Svendsen *et al.* conducted a national cross-sectional survey for people aged ≥25 years, and identified that individuals with basic education or an income below the national average presented higher odds of having inadequate HL (35). Berete *et al.* found similar results, observing that lower educational attainment and lower income were associated with having an

insufficient level of HL (36). Other international studies reinforce this evidence. A large-scale survey in China found that adults with at least 13 years of education were more than twice as likely to have adequate HL compared to those with less than nine years, and those with higher family average income also reported significantly higher HL levels (37). These findings suggest that higher socioeconomic status, especially higher education and income, consistently supports greater HL, and by extension, may also contribute to higher levels of GL.

Concerning BC screening adherence, GL was not identified as a significant predictor in our analysis. To our knowledge, there are currently no published studies that explore the relationship between GL and adherence to BC screening, which limits the possibility of comparing our results.

One possible explanation for this lack of association may lie in the overall low levels of GL observed in our sample. For instance, Linderman *et al.*, who developed and validated the GL scale, reported average scores of 49.8 and 47.1 among participants with adequate and inadequate HL, respectively (26). In contrast, the mean scores observed in our study were lower (42.6 and 41.0) and closer to each other, which may have reduced the ability to detect meaningful differences in screening behaviors.

The most notable result was the strong association between the setting where the last mammogram was performed and screening adherence. Women who had mammograms in NHS or private facilities were significantly more likely to be non-adherent compared to those screened in the mobile units of the Portuguese League Against Cancer.

This finding suggests that accessibility and convenience may play a more decisive role in promoting adherence than literacy alone. Mobile units are typically located closer to the population, and by receiving an invitation directly from the Portuguese League Against Cancer, without the need to schedule an appointment, potential barriers such as distance, time constraints, and organizational effort are effectively reduced.

Previous research consistently shows that mobile mammography helps reduce inequalities in screening participation. For instance, Guillaume *et al.* (2017) demonstrated that mobile mammography services can help overcome social and geographic disparities in access to BC screening (36). Similarly, a systematic review by Vang *et al.* (2018) found that mobile units significantly improve participation rates among medically underserved women, further highlighting the importance of addressing structural barriers to promote equitable access (37).

Taken together, these findings suggest that structural and organizational factors, such as service location and reduced need for individual initiative, may have a stronger influence on screening adherence than individual-level factors such as literacy. This highlights the importance of

designing screening programs that actively remove practical barriers and bring services closer to communities, thereby promoting more equitable participation.

An additional finding is that, despite the sample being highly educated and more than half of the participants presenting adequate HL, the overall GL levels remained low. This apparent discrepancy reinforces that while HL is influenced by general education and reflects broader competencies in accessing, understanding, and using health information, GL requires more specific knowledge and skills related to genetics and genomics, which may not be typically covered in higher education unless directly related to health or biological sciences. Therefore, these results highlight a potential gap between general and domain-specific literacy, suggesting that even populations with high education levels and adequate HL may still lack the competencies needed to make informed decisions in contexts where genetic information plays a role.

This study had several limitations that should be acknowledged. Firstly, the recruitment strategy relied on convenience sampling through social media platforms, due to time constraints. This has introduced selection bias, as the sample consisted predominantly of women with higher education levels. As a result, the proportion of participants who adhered to screening recommendations was relatively high, limiting the representation of non-adherent women and affecting the validity of the findings and the statistical power of certain subgroup analyses. For example, only 8 and 2 non-adherent women reported attending private services or mobile units, respectively, potentially affecting the validity of these findings.

Another limitation of the present study is the fact that some individuals are not included in the study but are within the recommended age range, since we included only women up to 70 years of age, whereas the national recommendation includes women up to 74. Additionally, there is currently limited evidence regarding GL both in general and in the context of BC screening, which makes it challenging to interpret and contextualize the GL scores obtained in this study.

Despite these limitations, the study has notable strengths. It is, to our knowledge, the first to explore the relationship between GL and BC screening behaviors, providing novel insights into a largely understudied area. Future research is needed to include more diverse and representative samples, particularly women who do not adhere to screening recommendations, to better understand the influence of GL in adherence.

Conclusions

This study found that although more than half of the participants had adequate levels of health literacy, overall genomic literacy scores were low, particularly among individuals with lower income and education levels, highlighting important inequalities and the need to strengthen GL in the general population.

We identified that the strongest predictor of non-adherence to breast cancer screening in Portugal was the setting where the last mammogram was performed, with community-based units being the setting associated with higher adherence. This finding suggests that structural and organizational factors, such as accessibility and proactive invitation, may have a more decisive influence on screening behaviors than individual literacy alone.

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Part III – Future Studies

The present study relied on convenience sampling through social media, which resulted in a highly educated sample and underrepresentation of non-adherent women. Future research should use mixed recruitment strategies to reach women across different socioeconomic groups, geographical areas (urban vs. rural), and education levels. This would help capture the perspectives of harder-to-reach populations who may face greater barriers to screening. Also regarding sampling, since this study only included women up to 70 years old, future work should encompass the entire target population (45–74 years old) to improve comparability with the National Breast Cancer Screening Program.

This study focused on women eligible for population-based BC screening under the national screening program. However, women at increased genetic risk (e.g., carriers of *BRCA1/2* mutations or with strong family history) are recommended to begin screening at younger ages. Including this group in future studies would provide valuable insights into the role of GL in contexts where decisions about genetic testing and earlier screening are required. Nevertheless, the current study design and recruitment strategy made it difficult to reach such women, since there is no national policy to identify individuals who should begin screening earlier. Future research should therefore consider alternative recruitment methods, such as collaboration with genetic counselling services, oncology clinics, or patient associations, to ensure the inclusion of high-risk women.

Lastly, given the cross-sectional nature of this study, causal inferences are limited. Longitudinal studies could clarify whether improving GL over time influences screening behaviors. Additionally, intervention studies, such as educational programs targeting GL or structural changes in screening delivery, could provide evidence on effective strategies to promote informed decision-making and screening adherence.

Although the study's recruitment method and sample composition limit the generalizability of these results, it provides valuable insight into an understudied area in the Portuguese context.

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