Original Investigation | Özgün Araştırma

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Investigation of the Level of Awareness and Knowledge of Physicians Regarding the Cancer Susceptibility of Germline *BRCA1* and *BRCA2* Mutations

Germline *BRCA1* ve *BRCA2* Mutasyonlarının Neden Olduğu Kanser Yatkınlığına Dair Hekimlerin Farkındalık ve Bilgi Düzeylerinin İncelenmesi

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ABSTRACT

Objective: The rising incidence and mortality of breast cancer highlight the importance of awareness of *BRCA1/2*-related hereditary breast and ovarian cancer. Appropriate genetic understanding and counseling improve outcomes in disease management.

Methods: This study aimed to evaluate the knowledge and awareness levels of physicians at the Ankara University, Faculty of Medicine using a questionnaire administered to 110 physicians.

Results: Among physicians, 47.7% reported adequately informing patients with *BRCA* mutations, whereas most reported difficulties in accessing genetic testing. Significant associations were observed between specialty knowledge and guideline knowledge, and between self-reported knowledge and actual knowledge. Physicians' ability to identify *BRCA*-associated cancers was correlated with both physicians' specialty and education level.

Conclusion: The results indicate a mismatch between perceived and actual knowledge levels and reveal gaps in both awareness and access to genetic services. The study underlines the need for improved education and integration of *BRCA*-related knowledge into clinical practice, emphasizing that medical education and clinical training in cancer genetics are crucial for early diagnosis and effective patient counseling, benefiting both current and future healthcare professionals.

Keywords: *BRCA1* and *BRCA2*, breast cancer, knowledge and awareness, physician

ÖZ

Amaç: Meme kanserinin artan insidansı ve mortalitesi, *BRCA1/2* ile ilişkili kalıtsal meme ve over kanseri farkındalığının önemini vurgulamaktadır. Uygun genetik yaklaşım ve danışmanlık, hastalık yönetimindeki klinik sonuçları iyileştirmektedir.

Yöntemler: Bu çalışma, 110 hekime uygulanan bir anket aracılığıyla Ankara Üniversitesi, Tıp Fakültesi'nde görev yapan hekimlerin bilgi ve farkındalık düzeylerini değerlendirmeyi amaçlamıştır.

Bulgular: Hekimlerin %47,7'si *BRCA* mutasyonu taşıyan hastaları yeterince bilgilendirdiğini bildirirken, büyük bir çoğunluğu genetik testlere erişimde zorluk yaşadıklarını ifade etmiştir. Uzmanlık alanı bilgisi ile kılavuz bilgisi arasında ve öz bildirime dayalı bilgi düzeyi ile gerçek bilgi düzeyi arasında anlamlı ilişkiler gözlemlenmiştir. Hekimlerin BRCA ile ilişkili kanserleri tanımlama yeteneğinin hem uzmanlık alanları hem de eğitim düzeyleri ile korelasyon olduğu bulunmuştur.

Sonuç: Sonuçlar, algılanan ve gerçek bilgi düzeyleri arasında bir uyumsuzluk olduğuna işaret etmekte, ayrıca hem farkındalık hem de genetik hizmetlere erişim konularındaki eksiklikleri ortaya koymaktadır. Bu çalışma, *BRCA* ile ilişkili bilgilerin klinik uygulamaya entegre edilmesine ve daha iyi bir eğitime duyulan ihtiyacın altını çizmektedir. Kanser genetiği alanındaki tıp eğitiminin ve klinik eğitimin, erken teşhis ve etkili hasta danışmanlığı açısından kritik bir öneme sahip olduğu ve hem mevcut hem de gelecekteki sağlık profesyonellerine fayda sağlayacağı vurgulanmaktadır.

Anahtar Sözcükler: *BRCA1* ve *BRCA2*, meme kanseri, bilgi ve farkındalık, hekim

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INTRODUCTION

Breast carcinoma is the leading cause of cancer mortality in women in Türkiye, mainly due to lack of awareness and delayed diagnosis (1). *BRCA1* and *BRCA2* mutations significantly increase lifetime breast cancer risk (2) and hereditary breast and ovarian cancer (HBOC) syndrome accounts for approximately 10% of breast cancer cases (3). "The *BRCA1* gene, located on chromosome 17, and the *BRCA2* gene, located on chromosome 13, encode proteins that (i) regulate the transcription of other genes, (ii) control the cell cycle and (iii) repair breaks in DNA through homologous recombination (4).

When *BRCA1* and *BRCA2* genes are mutated, they can lose their control function and become responsible for the individual's greater hereditary predisposition to neoplastic development. Breast and ovarian cancers are the most frequently occurring neoplasms associated with pathogenic variants of the *BRCA1* and *BRCA2* genes (5). Inherited disease-causing variants in *BRCA1/2* increase risks of breast, ovarian, fallopian tube, primary peritoneal cancers, and in men, breast and prostate cancers; both sexes face elevated pancreatic cancer risk (6).

Genetic testing involves risk assessment through counseling regarding family cancer history, thereby guiding testing decisions. About 13% of women in the general population develop breast cancer, while 55-72% with disease causing *BRCA1* variants and 45-69% with *BRCA2* variants develop breast cancer by age 70-80 (7-9). Risk varies according to individual factors. Women with *BRCA1/2* variants face higher risk of contralateral breast cancer, increasing to 20-30% at 10 years and 40-50% at 20 years post-diagnosis (8-10). The cumulative risk of all types of skin cancer from age 20 to 80 years is shown to be 14.1% for *BRCA1* carriers and 10.7% for *BRCA2* carriers (11).

BRCA1/2 associated cancer susceptibility is relatively high in Türkiye when compared with United States and European countries (3-4% in Europe, up to 15% in United States) (12), according to a study, 15.1% of Turkish patients with a family history of breast and/or ovarian cancer to have *BRCA1* and *BRCA2* mutations (13).

The education of physicians regarding breast cancer and *BRCA*-related cancer can be limited by several factors, including high workload in outpatient clinics, lack of awareness and initiatives focused on breast cancer and patient outcomes, and the absence of comprehensive local guidelines accessible to physicians from different specialties.

Our study revealed a significant lack of knowledge and awareness among general practitioners and specialists regarding genetic counseling for *BRCA*-related cancers, highlighting the need for more education on genetic counseling and for guidelines on referral to genetic testing in Türkiye.

This study is one of the first studies in Türkiye to include an objective assessment of physicians' knowledge and awareness regarding *BRCA1/2* germline mutations. Identifying gaps in understanding of hereditary breast and ovarian cancer would improve medical education and healthcare. Conducted at a leading academic medical center with diverse departments and access to a broad sample of healthcare professionals and trainees, the study is further strengthened by the institution's longstanding commitment to academic excellence, interdisciplinary research, and public health education, enhancing its relevance and potential impact.

MATERIALS AND METHODS

In this study, physician volunteers completed a structured, validated questionnaire to assess their knowledge and awareness of the cancer susceptibility associated with *BRCA1/2* mutations. Surveys were administered online to volunteers, and the data obtained were analyzed using statistical methods. Ethical approval of this study is done by Ankara University, Faculty of Medicine Student Ethics Committee E-72189195-050.03.04-1402268 (date: 13.06.2024, number: 351065). Informed consent were taken from each participant before collecting data.

Methods for Analysis of Questionnaire

For the analysis of "knowledge evaluation questions" in the survey, the possible answers were assigned points prior to analysis according to their relevance to breast cancer guidelines and the literature, thereby making the assessment of knowledge level more objective. Points can be seen in the following table (Table 1). Significant results for the study's outcomes are presented in tables of percentages.

Inclusion and Exclusion Criteria

A cross-sectional study was conducted amongresidents and specialists in oncology, internal medicine, gynecology and obstetrics, intensive care, medical genetics, oncologic surgery, and general surgery at Ankara University, Faculty of Medicine Hospitals between May and June 2025. Physicians from other specialties are excluded from the study.

Statistical Analysis

Statistical analyses to compare different groups and responses were performed using IBM® SPSS (27th version, IBM Corp., 2020, CA, USA). Chi-square tests and Fisher's exact test were used for group and response comparisons, while independent-samples Kruskal-Wallis, One-Way ANOVA, post-hoc Tukey-Duncan, and correlation tests were used to assess question reliability and guide corrections across several questions and variables. When more than 20% of expected cell counts in a data group were less than five, the Fisher-Freeman-Halton exact test with Monte Carlo simulation was used.

RESULTS

The results revealed a statistically significant association between area of specialization and perceived knowledge levels, indicating that these levels differ among specialties.

Higher mean knowledge levels (mean 3.68) were observed in medical genetics, oncologic surgery, and medical oncology specialists, whereas lower levels were observed in gynecologists (Table 2).

61.4% of participants indicated having good knowledge, when measured their knowledge levels, the 42.2% were at intermediate or lower level about mutation type and cancer matching.

There is a significant difference in the distribution of physicians' experience levels and their stated levels of knowledge regarding *BRCA*-related carcinogenesis (Table 3).

Participants' stated knowledge of breast cancer risk factors differed significantly across responses related to *BRCA1/2* mutation-associated cancers ($p < 0.05$) (Figure 1).

Table 1. The scoring criteria were primarily derived from the NCCN Guidelines, while the Manchester scoring system was used as supplementary background information.

Questions	Answers	Points (question number. points)
Indicate the key diagnostic and prognostic criteria in breast cancer (3,14)	1. Triple breast cancer diagnosis before age of 60	1. +3
	2. Lobular breast cancer seen with diffuse gastric cancer	2. +2
	3. Ancestry of Ashkenazi Jew	3. +2
	4. Two or more than two primary breast cancer in the same person	4. +2
	5. Breast, ovarian, pancreas or/and agressive prostate cancer in 3 or more than 3 relatives including patient	5. +3
Indicate the *BRCA1* and *BRCA2* related cancers (15,16)	1. Breast cancer	1. +1
	2. Ovarian cancer	2. +1
	3. Prostate cancer	3. +1
	4. Pancreatic cancer	4. +1
	5. Melanoma	5. +1
	6. Colorectal cancers	6. 0
	7. Lung cancer	7. 0
	8. Leukemia	8. 0
In which pathological types of breast cancer would you think about hereditary breast-ovarian cancer syndrome?	1. Triple negative	1. +3
	2. HR+	2. +2
	3. Ductal	3.+2
	4. Luminal A	4. +1
	5. Luminal B	5. +1

NCCN: National Comprehensive Cancer Network, HR: Hormone receptor.

Table 2A. Evaluate your level of knowledge regarding the evidence and recommendation levels in *BRCA1/2* genetic testing guidelines.

Specialty/evaluation	1	2	3	4	5	Total
Oncologic surgery	0	0	1	4	4	9
General surgery	2	4	10	10	7	33
General internal medicine	3	3	7	8	0	21
Gynecology and obstetrics	1	3	6	7	7	24
Medical genetics	0	0	0	3	7	10
Medical oncology	0	1	2	4	6	13
Total	**6**	**11**	**26**	**36**	**31**	**110**

Table 2B. Chi-square tests.

	Value	df	Asymptotic significance (2-sided)	Monte Carlo sig. (2-sided)		
				Significance	99% confidence interval	
					Lower bound	Upper bound
Pearson chi-square	78,863[a]	40	0.000	0.047[b]	0.030	0.064
Likelihood ratio	78,952	40	0.000	0.000[b]	0.000	0.005
Fisher-Freeman-Halton exact test	77,486			0.000[b]	0.000	0.005
N of valid cases	110					

[a]: Forty-seven cells (87.0%) have expected count less than 5. The minimum expected count is 0.02, [b]: Based on 1000 sampled tables with starting seed 1314643744.

Table 3. Relations of objectively evaluated and stated knowledge levels.

Objective (vertical, 0-5)/stated (transverse, 1-5)	1	2	3	4	5	Total
0	0	0	0	0	1	1
1	1	0	0	1	2	4
2	0	4	8	6	2	20
3	1	7	7	4	2	21
4	1	2	8	10	7	28
5	0	0	3	14	18	35
Total	**3**	**13**	**26**	**35**	**32**	**109**

Chi-Square Tests

			Asymptotic Significance (2-sided)	Monte Carlo Sig. (2-sided)			Monte Carlo Sig. (1-sided)		
					99% Confidence Interval			99% Confidence Interval	
	Value	df		Significance	Lower Bound	Upper Bound	Significance	Lower Bound	Upper Bound
Pearson Chi-Square	34,807[a]	20	,021	,046[b]	,029	,063			
Likelihood Ratio	30,181	20	,067	,042[b]	,026	,058			
Fisher-Freeman-Halton Exact Test	30,191			,032[b]	,018	,046			
Linear-by-Linear Association	7,993[c]	1	,005	,004[b]	,000	,009	,003[b]	,000	,007
N of Valid Cases	110								

a. 20 cells (66,7%) have expected count less than 5. The minimum expected count is ,02.

b. Based on 1000 sampled tables with starting seed 1502173562.

c. The standardized statistic is 2,827.

Figure 1. A positive association between perceived knowledge and accurate identification of BRCA-related cancers*

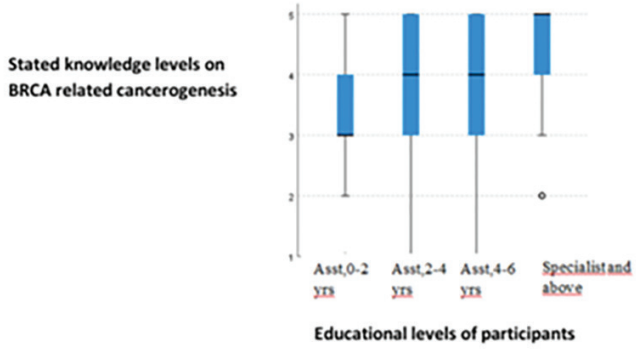

Figure 2. Figure 1 shows the Kruskal-Wallis distribution of knowledge on breast cancer risk factors and evaluated knowledge levels on *BRCA* mutations. Points are given according to Table 1 (0 to 5 points), while self-assessment is done from 1 (very low) to 5 (very high).

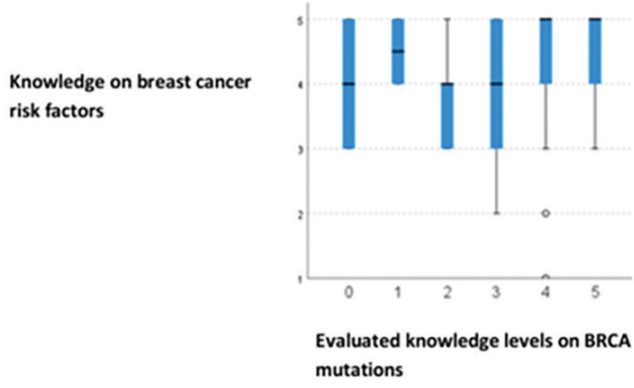

Figure 3. Shows the Kruskal-Wallis distribution of stated knowledge levels on *BRCA*-related carcinogenesis and educational levels of participants. Self-assessment is done from 1 (very low) to 5 (very high) and educational level is from demographic data.

Figure 1 shows the Kruskal-Wallis distribution of knowledge about breast cancer risk factors and evaluated knowledge levels regarding *BRCA* mutations. Points are assigned according to Table 1 (0 to 5 points), and self-assessment is rated on a scale from 1 (very low) to 5 (very high).

There is a significant difference in the distribution of physicians' experience levels and stated levels of knowledge regarding *BRCA*-related carcinogenesis (asymptotic significance = 0.024) (Figure 2).

There is a statistically significant difference in participants' stated knowledge of breast cancer risk factors across their responses related to *BRCA1/2* mutation-associated cancers ($p < 0.05$)

There was no significant relationship between physicians' information-giving attitudes and their knowledge levels regarding breast cancer (Table 4).

Five of six physicians report difficulty accessing genetic testing.

47.7% of physicians stated that they appropriately inform their patients with *BRCA1/2* mutations. 7.2% of physicians stated that they almost never inform their patients about BRCA mutations.

Only 17.3% of respondents believe that breast cancer awareness initiatives are fully sufficient, while 20% of respondents find them clearly insufficient.

DISCUSSION

Our findings demonstrate that, while there is a general understanding of hereditary cancer concepts among participants, significant gaps remain in practical knowledge, application, and awareness, particularly concerning genetic testing guidelines, cancer association, and risk communication.

It was seen that large proportions of the participants reported little or no awareness of genetic testing and services, demonstrating a need to educate physicians about awareness on genetic conditions of their patients (17), but in contrast, in our study, the stated knowledge levels are relatively high when compared with evaluated knowledge levels. These findings indicate that the physicians in our cohort perceive their knowledge to be higher than their actual knowledge.

While most participants rated themselves as having good knowledge, objective assessments showed that many participants lacked adequate understanding, especially regarding mutation-specific cancer associations. Although 32.4% of participants achieved full scores on *BRCA*-related cancer knowledge, a larger proportion of participants remained at intermediate or lower levels. This discordance between perceived and actual knowledge is significant, as it may lead to overconfidence and misinformed clinical decisions**.** Other studies revealed that there is a low level of perceived knowledge indicating a need for a further education for physicians (18) in context of increasing importance of up to date knowledge (19).

Appropriate risk evaluation in genetic counseling is important both to prevent unnecessary genetic testing and to contribute to positive prognostic outcomes (20). In our study, the majority of respondents strongly agreed with the need to inform at-risk relatives of *BRCA1/2* mutation carriers. In this setting, approximately half of physicians stated that they inform their patients with *BRCA1/2* mutations appropriately, and 7.2% of physicians stated that they almost never inform their patients about *BRCA* mutations.

Our study found that specialization significantly influenced knowledge levels. Participants in medical genetics, medical oncology, and oncologic surgery have greater awareness and understanding of *BRCA* testing guidelines and related cancers. This suggests that specialty-specific exposure and training shape familiarity with hereditary cancer concepts. Similarly, knowledge improved with clinical experience but not necessarily with educational attainment, underscoring the value of practical, case-based learning in genetics education. As it becomes more important to support the collaboration of genetic services in the clinic (21). However, in the evaluation of *BRCA1/2*-related cancers, gynecologist participants scored lower than geneticists and other specialists. It can be an indicator for these groups to increase their knowledge on *BRCA* related cancers other than ovarian cancer, which is an important eligibility criterion for hereditary cancer syndromes related with BRCA mutations (22).

Referring patients to medical genetics departments because collaboration between medical genetics and other specialties is an important aspect of clinical genetics practice.

Table 4A. Do you contact with your patients about their genetic conditions? (yes/no).

Objective evaluation points/answers	Yes	No	Total
0	1	1	2
1	4	0	4
2	18	2	20
3	19	1	20
4	27	0	27
5	29	6	35
Total	**98**	**10**	**108**

Table 4B. Do you contact with your patients about their genetic conditions? (yes/no).

	Value	df	Asymptotic significance (2-sided)	Exact sig. (2-sided)
Pearson chi-square	10,148[a]	5	0.071	0.071
Likelihood Ratio	10,848	5	0.054	0.057
Fisher-Freeman-Halton exact test	9,058			0.066
N of valid cases	108			

[a]: Eight cells (66.7%) have expected count less than 5. The minimum expected count is 0.19.

No relationship was observed between physicians' information-giving attitudes and knowledge levels. Even though in the literature, a correlation was shown (23), our study does not indicate for such correlation and our findings show that it become rather a personal choice of physician during practice, who are from different specialties, experience and knowledge levels.

The majority of physicians stated that there should be a referral guideline for medical genetics, indicating a need for such a guideline in Türkiye. Guidelines are important for physicians to guide patient better and make more comprehensive evaluations (20).

Study Limitations

Despite these findings, the study has limitations. Because it is a cross-sectional survey based on self-reported data, it is subject to recall and social desirability bias. Actual clinical behaviors were not directly observed. While we assessed general awareness and perceived competence, more in-depth evaluations of clinical application and guideline adherence would be beneficial for future research. Even points that are given to answers are used to evaluate and compare the knowledge levels appropriately, only literature and evidence-based data can be used to prepare a evaluation scale due to lack of pre-established system

CONCLUSION

This study highlights the need for more standardized, interdisciplinary education for physicians on hereditary cancers, especially those related to *BRCA* mutations.

Targeted educational programs, collaborations between clinicians and greater emphasis on awareness of physicians on genetic topics are critical steps to close the gap between awareness and action in hereditary cancer prevention and management in future. More breast cancer awareness studies and practice-based studies will underscore the implications of knowledge about *BRCA* mutations and breast cancer, leading to applications that yield more positive outcomes for patients. Lifelong learning initiatives regarding the practice of genetics in different fields of clinical medicine, along with increased awareness and knowledge of breast-ovarian cancer syndromes, can enable better management of these syndromes through collaboration between the healthcare service team, the patient, and their relatives.

Guidelines on breast cancer and other cancers with related mutations in Türkiye would help physicians make comprehensive evaluations of patients and improve recognition of breast cancer in a more scientific manner.

Ethics

Ethics Committee Approval: Ethical approval of this study is done by Ankara University, Faculty of Medicine Student Ethics Committee E-72189195-050.03.04-1402268 (date: 13.06.2024, number: 351065).

Informed Consent: Informed consent were taken from each participant before collecting data.

Footnotes

Authorship Contributions

Surgical and Medical Practices: Ş.A., Concept: S.K., B.A., M.S.Y., Ş.A., Design: S.K., B.A., M.S.Y., E.K.D., Ş.A., Data Collection or Processing: S.K., M.S.Y., B.D., Ş.A., Analysis or Interpretation: S.K., M.S.Y., B.D., Ş.A., Literature Search: S.K., M.S.Y., E.K.D., Ş.A., Writing: S.K., M.S.Y., Ş.A.

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