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Investigating the feasibility of cascade genetic testing for *BRCA1/2* in Türkiye: physicians' practices, barriers, and solutions

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Abstract

Cascade genetic testing is a key strategy for identifying individuals at risk of hereditary cancers, yet its implementation remains suboptimal in many healthcare systems. We evaluated physicians' knowledge, attitudes, and clinical practices regarding *BRCA1/2* testing in Türkiye and, from the physicians' perspective, assessed the feasibility of an effective cascade genetic testing model. A nationwide cross-sectional online survey was conducted among physicians involved in the management of *BRCA*-related cancers, covering experience with *BRCA1/2* testing, interpretation of results, and approaches to patient follow-up and family screening. Univariate binary logistic regression examined associations between physicians' characteristics and knowledge, and the likelihood of consistently requesting testing, recommending screening, and referring relatives. Among 206 participants, namely medical oncologists (46.4%), surgeons (35.6%), and medical geneticists (17%), 89.8% frequently or always requested *BRCA1/2* testing when indicated. Higher knowledge was strongly associated with ordering tests (odds ratio: 6.1; 95% confidence interval: 3.2–11.7; $p < 0.001$), recommending screening ($p < 0.001$), and referring first-degree relatives ($p = 0.013$). Despite high physician engagement, the participation of relatives in screening was low (3.4% full; 35.0% partial), as was uptake of prophylactic interventions. Although physicians demonstrated high willingness to use *BRCA1/2* testing, cascade genetic testing remained insufficient, particularly at the level of family engagement and uptake of preventive care. Physicians' knowledge emerged as a key driver of clinical behaviour. Establishing structured, centralised cascade-coordination models integrated into public health systems may improve the identification and management of at-risk individuals.

Keywords: *BRCA1*, *BRCA2*, Cascade genetic testing, Hereditary, Breast cancer, Ovarian cancer, Genetic counseling

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Introduction

Pathogenic germline variants in *BRCA1* and *BRCA2* genes significantly increase the lifetime risk of developing breast, ovarian, prostate, and pancreatic cancers. Identifying individuals with these mutations is critical for guiding treatment and implementing preventive measures, such as regular surveillance and risk-reducing surgeries, which have been shown to reduce cancer risk by over 90%. Despite international guidelines recommending genetic testing for hereditary diseases, its uptake remains limited because of patient misconceptions, limited awareness, and underutilisation by healthcare providers[1–4].

Although hereditary genetic mutations can be inherited from ancestors or occur *de novo*, *de novo* pathogenic variants occur rarely, at a rate of approximately 0.3%[5]. Therefore, the approach of 'cascade genetic testing,' which involves testing the first-degree relatives of *BRCA*-positive cancer patients to identify those at risk and enable early preventive interventions, is essential for detecting individuals who have not yet developed cancer[6–8]. Although public awareness and

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positive perceptions have gradually evolved through increased use of social media, the demand for genetic testing for hereditary diseases is largely dependent on the knowledge, attitudes, and perceptions of physicians. Medical oncologists and surgeons with expertise in cancer care are central to the implementation of cascade genetic testing. A medical oncologist, in collaboration with an expert team, should assess the hereditary risk score of cancer patients and make appropriate referrals for the screening of first-degree relatives. However, several obstacles hinder the optimal application of this system, including the physician's willingness to adopt this approach, the time required to implement it, insurance coverage for the tests, and the lack of a multidisciplinary genetic tumour board. Additionally, effective communication with patients and their first-degree relatives about *BRCA1/2* testing and its implications remains a significant challenge.

This study evaluated physicians' knowledge, attitudes, and clinical practices regarding *BRCA1/2*-associated genetic testing in Türkiye. It also assessed the real-world implementation rate of the cascade genetic testing system, as indirectly gauged via the perspective of physicians. By identifying barriers and proposing solutions, the study seeks to improve the integration of genetic testing into public health systems, enhancing early cancer detection and prevention for high-risk individuals.

Materials and methods

Study population

This physician survey forms part of a larger ethical committee-approved research programme (Istinye University Ethics Committee, No. 2024/02, 24-56) that examines hereditary *BRCA1/2* cancer care in Türkiye from complementary perspectives. Two parallel self-administered online questionnaires were developed and distributed, one to physicians ordering *BRCA1/2* tests (analysed here) and one to *BRCA1/2*-positive patients and carriers. To avoid redundant publication, the outputs of this programme report distinct, nonoverlapping analyses: The present article addresses physicians' practices and the feasibility of cascade genetic testing, but preimplantation genetic testing (PGT) awareness across the physician and patient cohorts is reported in a companion article[9]. A separate analysis of patients' and carriers' experiences and barriers to preventive interventions is planned and will be reported separately. Surgeons and oncologists responsible for monitoring and treating cancer patients who worked in private clinics, government institutions, or academic institutions were included. The primary aim of the present survey was to investigate physicians' experiences with individuals carrying likely germline pathogenic *BRCA1/2* variants and their relatives in Türkiye. In Türkiye, patients are typically referred to a clinical geneticist at the treating clinician's discretion; however, cancer patients may also self-refer to clinical genetic services.

The survey questions were designed in alignment with the recommendations of the European Society for Medical Oncology[10] and the National Comprehensive Cancer Network[2] concerning genetic testing and preventive procedures.

Survey questionnaires

The survey items were developed on the basis of existing literature on physicians' knowledge and attitudes toward genetic testing and were prepared by a multidisciplinary expert research team. All items were developed *de novo* by the research team.

Before initiating the survey, physicians were informed about its purpose and significance, and informed consent was obtained (see the Supplementary File 1 and File 2). The survey was designed to gather data on physicians' experiences with *BRCA1/2* testing and their recommendations for additional interventions and follow-up strategies for patients with germline pathogenic *BRCA1/2* variants. Additionally, the feasibility of implementing a cascade genetic testing system was indirectly assessed.

The survey was administered online via Google Forms. The participants' information and informed consent statement appeared on the first screen of the form, and only physicians who actively confirmed their consent could access and proceed to the survey items. Physicians who did not provide consent were unable to view the survey questions or continue; thus, informed consent was a mandatory precondition for participation.

The survey gathered information on each physician's gender, age, geographic location of practice, field of expertise, years of experience, and the type of institution where they worked (state hospital, state university hospital, private university hospital, or private hospital/clinic). The survey also inquired about the availability of *BRCA1/2* testing at their institution or within their city, the facility where *BRCA1/2* testing was performed (private laboratory, private hospital, health ministry institutions, university hospitals, or abroad), the reimbursement status of *BRCA1/2* testing for cancer patients or healthy individuals (covered by the social security institution [sosyal güvenlik kurumu, SGK]), partially covered with additional payment, private insurance, or individual payment), and the time required to obtain *BRCA1/2* test results.

In addition, the following topics were addressed in the survey: the frequency of physicians requesting *BRCA1/2* testing, their knowledge levels of and attitudes toward interpreting *BRCA1/2* test results, the screening modalities they recommend for carriers of a pathogenic *BRCA1/2* variant, the details of their decision-making processes for preventive surgical procedures, and the rates of implementation and acceptance of these procedures by patients or carriers. The survey also explored physicians' knowledge and approaches regarding variants of uncertain significance (VUS) and their understanding and perspectives on preimplantation genetic testing. The specific questions and answer options provided to physicians are detailed in the Supplementary File 2.

High-penetrance genes are defined as those associated with a lifetime risk of developing breast cancer of 30% or greater from the age of 20. In addition to *BRCA1/2*, other high-penetrance genes include *PALB2*, *PTEN*, *TP53*, *CDH1*, and *STK11*, for which prophylactic surgical procedures can also be recommended[2]. Similar to breast cancer, high-penetrance genes are linked to ovarian, prostate, and pancreatic cancers[2]. However, since *BRCA1/2* genes are well-known among physicians, and recommendations for preventive surgical procedures are consistent across guidelines, this study focused exclusively on questions related to *BRCA1/2* genes to avoid confusion.

The participants comprised medical oncologists, surgeons (including gynaecological oncologists, gynaecologists, oncology surgeons, general surgeons, and urologists), medical geneticists, and other physicians involved in oncologic practice (classified under the surgeon category).

Our specific questions (Questions 13, 18, 19, and 20) were selected to assess the physicians' knowledge regarding *BRCA1/2* testing. Each correct answer was awarded 1 point, yielding a total score of 0–4. Scores were then dichotomised into two groups (0–2 and 3–4 points) and compared across physician specialties.

The survey questions were primarily multiple choice, except for those addressing age, gender, geographic location of practice, field of expertise, years of experience, and institution of employment. For certain questions, the participants were allowed to select more than one answer.

Statistical analysis

Descriptive statistics are reported as frequencies (*n*) and percentages (%), whereas continuous variables are presented as means ± standard deviation (SD). In some analyses, participants were categorised as medical geneticists versus other physicians. In other analyses, knowledge levels and clinical approaches were examined by grouping participants into three specialty categories: medical oncologists, surgeons (including other physicians, comprising 1%), and medical geneticists. Potential factors influencing the likelihood of consistently requesting genetic testing were investigated using univariate binary logistic regression analysis. For each comparison, individual variables were entered separately as independent predictors, and the results are reported as odds ratios (OR) with 95% confidence intervals (CI). A *p*-value of ≤ 0.05 was considered to be statistically significant. All analyses were performed using SPSS software, version 22.0 (IBM SPSS Statistics).

The study complied with the principles outlined in the Declaration of Helsinki and received approval from the ethics committees of the participating centres. It was additionally approved as a multicentre study by the Istinye University Ethics Committee (No. 2024/02, 24-56).

Results

General information about physicians, access to, and reimbursement for *BRCA1/2* testing

In total, 206 physicians participated in the survey and completed all the questions. Of the participants, 37.4% (*n* = 77) were women, and the mean age was 42.77 years (SD = 9.6). The duration of medical specialisation was categorised as 0–5 years, 35%; 6–10 years, 19.4%; 10–20 years, 24.7%; and over 20 years, 20.9%. Nearly half of the participants (46.4%) were medical oncologists, and 15% were general surgeons, 15.5% were gynaecologists/obstetricians, 4.1% were urologists, 17% were geneticists, and 1% were from other specialties. Participants were included from various academic and community settings nationwide to ensure social diversity. Of these, 88 participants (42.7%) worked in state university hospitals, 50 (24.3%) in community hospitals, 32 (15.5%) in private or foundation university hospitals, 28 (13.6%) in private facilities, and 8 (3.9%) in private clinics. Access to and reimbursement for *BRCA1/2* testing are detailed in the Supplementary File 3. Most physicians stated that they can access to germline *BRCA1/2* testing either at their workplace or within their city. Additionally, they stated that *BRCA1/2* testing was predominantly reimbursed by the SGK rather than private insurance.

Factors influencing physicians' attitudes toward *BRCA1/2* testing

Among nongenetic physicians, 98.8% reported considering national or international guideline recommendations when ordering *BRCA1/2* testing, whereas all genetic physicians stated that they followed these guidelines. In total, 85.9% of physicians requested the test within the first three months after diagnosis. When asked how frequently they requested *BRCA1/2* testing when indicated, 61.2% of physicians reported 'always,' 28.6% reported 'frequently,' 7.3% reported 'sometimes,' and 2.9% reported 'never'.

As summarised in Table 1, genetic physicians had a significantly higher rate of always requesting *BRCA1/2* testing compared with medical oncologists and surgeons (p = 0.013). Among surgeons, general surgeons had the highest rate of genetic testing. Although not statistically significant, physicians with 0–5 years of experience requested *BRCA1/2* testing less frequently than their more experienced counterparts. Similarly, physicians working in state hospitals exhibited lower rates of always requesting *BRCA1/2* testing compared with those working in universities or private institutions.

Table 1. Parameters influencing the rate of always requesting *BRCA* testing when indicated.

Parameters	The rate of always requesting the test	OR	95% CI	*p*-Value
Medical branch				
Medical oncologist	53.3%	1 (reference)		
Surgeon (general surgeon; gynaecologist; urologist)	56.3% (69.7; 56.7; 0)	1.12	0.6–1.66	0.707
Medical geneticist	78.8%	3.22	1.3–8.3	0.013
Years of experience				
0–5 years	51.4%			0.132
6–10 years	70.0%			
11–20 years	69.7%			
Over 20 years	64.5%			
Institution				
State hospital	48.8%			0.777
State university	63.6%			
Private hospital/clinic	61.1%			
Private university/foundation	68.8%			
Availability of germline *BRCA* testing				
Available	62.6%			0.17
Not available	51.2%			
Test reimbursement				
Covered by SGK	61.9%			0.36
Private/additional payment	54.3%			
Knowledge level				
Very sufficient/sufficient	72.9%	6.1	3.2–11.7	< 0.001
Not sufficient	30.6%			

OR, odds ratio; CI, confidence interval; knowledge level (interpreting *BRCA* test results).

Physicians who had access to *BRCA1/2* testing at their institutions showed a higher (though not statistically significant) rate of always requesting *BRCA1/2* testing compared with those without direct access. Additionally, full reimbursement for *BRCA1/2* testing by the SGK was associated with higher rates of test orders compared with cases requiring additional patient payment, though this difference was not statistically significant.

The most influential parameter was physicians' knowledge level in interpreting genetic test results, which was strongly associated with always requesting *BRCA1/2* testing (odds ratio [OR]: 6.1; 95% confidence interval [CI]: 3.2–11.7, $p < 0.001$) (Table 1).

Knowledge levels and approaches toward *BRCA1/2* testing and results by physician specialty

Physicians were asked to assess their knowledge level in interpreting *BRCA1/2* test results. Among oncologists, 58.9% rated their knowledge as sufficient or very sufficient, whereas 41.1% considered it insufficient or somewhat sufficient. Similarly, 63.4% of surgeons reported their knowledge as sufficient or very sufficient, with the remainder rating it as insufficient or somewhat sufficient. In contrast, 92% of medical geneticists rated their knowledge as sufficient or very sufficient.

Physicians also evaluated their knowledge regarding the diagnosis, treatment, and follow-up of *BRCA*-associated cancers. Among medical oncologists, 64.5% rated their knowledge as sufficient or very

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sufficient, but the remainder considered it insufficient or somewhat sufficient. For surgeons, 69.0% reported sufficient or very sufficient knowledge, whereas the rest rated it as insufficient or somewhat sufficient. Notably, 81.3% of geneticists reported sufficient or very sufficient knowledge, but the remainder rated it as somewhat sufficient. The physicians' knowledge about VUSs and PGT are detailed in the Supplementary File 3.

When asked 'Do you personally provide information to your *BRCA*-positive patients regarding their follow-up and treatment?', 83.3% of nongenetic physicians and 90.6% of geneticists answered affirmatively. Regarding the question 'Do you ensure that your *BRCA*-positive patients receive genetic counseling regarding their follow-up and treatment?', 96.9% of nongenetic physicians responded 'yes'.

Physicians were also asked about decision-making processes for prophylactic procedures. Table 2 shows a summary of prophylactic surgery decisions.

When asked 'Do you recommend screening programs for *BRCA*-positive patients?', 66% of participants reported always recommending them, 27% frequently recommended them, and the remainder recommended them occasionally.

When asked 'Do you request *BRCA1/2* testing for the relatives of your *BRCA*-positive patients?', 9.9% of nongenetic physicians reported always requesting it, 19.1% frequently requested it, and 59.3% referred patients for genetic counseling. Among geneticists, 75% always requested *BRCA1/2* testing; the remainder frequently requested it.

As summarised in Table 3, physicians with sufficient or very sufficient self-reported knowledge (corresponding to higher scores on the four-item knowledge assessment described in the Methods) were significantly more likely to recommend screening for *BRCA*-positive patients (univariate analysis: $p < 0.001$) and to refer first-degree relatives for *BRCA* testing ($p = 0.013$).

Table 2. Prophylactic decision-making.

Decision-making context	Prophylactic mastectomy (*n*)	Prophylactic oophorectomy (*n*)
Tumour boards	63	60
Joint decision with geneticists	37	41
Multidisciplinary consultations	30	30
Geneticist-only consultation	10	8
Individual decisions	66	67

Assessing physicians' knowledge levels

Questions 13, 18, 19, and 20 were selected to assess physicians' knowledge regarding *BRCA1/2* testing. It was observed that 68% of physicians selected the correct answer per question, on average, according to international guidelines (i.e., 68% accuracy rate per individual question across all respondents). For these four questions, each correct approach was awarded 1 point, resulting in an average knowledge score of 2.66 (±1.06). Physicians scoring 3–4 points were compared with those scoring 0–2 points by specialty. Among oncologists, 68.9% scored 3–4 points, compared with 47.9% of surgeons (vs oncology, $p = 0.08$), and 51.5% of medical geneticists (vs oncology, $p = 0.078$). The answers of the physicians are shown in detail in the Supplementary File 3.

Patients' and relatives' approach

From the physicians' perspective, the rates at which patients and their relatives undergo screening and surgical procedures are summarised in Table 4.

Discussion

We surveyed physicians ordering *BRCA1/2* testing for cancer patients and their relatives to examine their counseling, testing, and recommendation practices for women with the pathogenic *BRCA1/2* variant. Participants represented all geographic regions of Türkiye and included medical oncologists, surgeons, and medical geneticists from both academic and nonacademic institutions. Our study provides valuable insights into the current attitudes, knowledge levels, and practices of physicians regarding *BRCA1/2* genetic testing in Türkiye. In addition, the extent to which physicians adopted the cascade genetic system was indirectly analysed by asking them about their attitudes towards referring relatives of individuals with pathogenic *BRCA1/2*

Table 3. Relationship between level of knowledge on *BRCA* testing and screening recommendations.

Parameters	Knowledge level		*p*-Value
	Very sufficient/sufficient ($n = 144$)	Insufficient ($n = 62$)	
The rate of always recommending screening for *BRCA*-positive cancer patients	77.1% (111)	40.3% (25)	< 0.001
The rate of always referring relatives of *BRCA*-positive patients for *BRCA* testing	19.4% (28)	11.3% (7)	0.013
The rate of referring relatives of *BRCA*-positive patients to genetic counseling for *BRCA* testing	56.9% (82)	45.2% (28)	
The rate of rarely or never referring relatives of *BRCA*-positive patients for *BRCA* testing.	23.6% (34)	43.5% (27)	

Table 4. Rates of patients and relatives undergoing screening and surgical procedures.

Questions	All participate	Most participate	Some participate/ do not participate
BRCA-positive patients' participation in screening:	4.9%	51.0%	44.2%
Is participation regular?	4.9%	46.1%	49%
Participation of *BRCA*-positive carriers in screening:	3.4%	35.0%	61.7%
Is participation regular?	4.4%	35.0%	62.1%
	Always/very frequently	Very frequently/frequently	Rarely/never
Rates of prophylactic surgery in *BRCA*-positive breast cancer patients for ovarian cancer risk	21.4%	40.3%	38.4%
Rates of prophylactic surgery in *BRCA*-positive ovarian cancer patients for breast cancer risk	12.7%	39.3%	48.0%
Rates of prophylactic surgery in *BRCA*-positive carriers for breast cancer risk	13.6%	28.6%	55.3%
Rates of prophylactic surgery in *BRCA*-positive carriers for ovarian cancer risk	16.9%	28.2%	51.9%

mutations for genetic screening. The findings reveal a high level of willingness among physicians (89.8%) to request *BRCA1/2* testing when indicated, particularly among oncologists and geneticists. Despite this promising level of engagement, our findings highlight notable gaps in practice. Participation rates in screening programs among first-degree relatives of cancer patients with a pathogenic *BRCA* variant were alarmingly low. These findings reflect a fundamental inadequacy in the implementation of the cascade genetic testing system. Similarly, the uptake of prophylactic surgical interventions, such as mastectomy and oophorectomy, remained suboptimal.

According to the present survey, most physicians stated that they can access to germline *BRCA1/2* testing either at their workplace or within their city. Unlike many European countries and the United States, *BRCA1/2* testing in Türkiye is predominantly reimbursed by the SGK rather than by private insurance[11,12]. Physicians with less than five years of experience and those in state hospitals were less likely to consistently request testing than their more experienced counterparts and those in academic or private institutions. These differences did not reach statistical significance, potentially because of the unequal distribution of physicians across groups and the relatively modest overall sample size (Table 1). Though access to testing and SGK reimbursement had a minimal impact on ordering testing, the most significant factor was sufficient knowledge of *BRCA*-related diagnosis, treatment, and follow-up. A previous study reported that only 15.1% of ovarian cancer patients discussed genetic testing with their physician, and only 10.5% underwent hereditary testing. Similarly, only 29% of breast cancer patients discussed testing with their physician, and 15.3% underwent genetic testing[4]. The rates observed in our study do not directly reflect the real genetic testing rates in Türkiye but rather highlight physicians' attitudes, which may result in higher testing tendencies. Additionally, over the years, *BRCA1/2* testing has evolved beyond its hereditary implications to serve as a treatment-guiding tool, potentially increasing physicians' tendency to request the test[13–15].

In our study, the knowledge levels of physicians were assessed and scored on the basis of four questions related to the cancers for which screening is recommended in cases of a pathogenic *BRCA1/2* variant and the screening methods used for the follow-up of breast and ovarian cancers. Between 55% and 72% of physicians answered the individual knowledge questions correctly. Similar to findings in the literature, it was observed that oncologists and medical geneticists had a more guideline-compliant and accurate approach to cancer screening compared with surgeons when pathogenic *BRCA1/2* variants were detected[16,17]. However, our study also showed that over time, both oncologists and surgeons demonstrated improved knowledge levels compared with previous years[18]. Among the questions assessing knowledge levels, breast and ovarian cancer screening were more widely known, although screening for prostate and pancreatic cancers was less recognised, consistent with the literature[19,20].

Physicians with sufficient knowledge were significantly more likely to recommend screening for patients with a pathogenic *BRCA* variant and refer their first-degree relatives for genetic counseling (Table 3). Evidence-based guidelines emphasise cascade genetic testing for hereditary cancer syndromes, enabling early detection and preventive interventions for relatives. Effective management of hereditary cancer syndromes extends beyond the patient to their family members, as genetic test results have implications for the entire family. Cascade genetic testing remains the most effective approach for identifying individuals with hereditary disorders in a population. According to our survey, 66% of physicians always recommended screening programs to cancer patients with a pathogenic *BRCA1/2* variant, whereas only 9.9% reported always recommending screening programs to the first-degree relatives of cancer patients. It was observed that a significant majority of physicians (96.9%) referred individuals with a pathogenic *BRCA* variant for genetic counseling. Despite the high rate of referrals to genetic counseling, according to physicians' experiences, the participation rate of carrier individuals without cancer in screening programs was low (3.4% reported that all participate; 35.0% reported that most participate), as was the rate of regular participation in screenings (4.4% always/most of the time). Additionally, the rates of undergoing prophylactic mastectomy (13.6% all; 28.6% most) and prophylactic oophorectomy (16.9% all; 28.2% most) were similarly low, consistent with the published literature[21,22]. These findings underscore that referral of all *BRCA1/2*-positive patients to a medical geneticist should be considered an essential and mandatory step in clinical practice. Medical geneticists, by virtue of their specialised training, are uniquely positioned to interpret genetic test results comprehensively, provide patients with clear and accurate information about their findings, and counsel them on the critical importance of informing their first-degree relatives. Ensuring that this referral pathway is consistently followed—regardless of the treating physician's specialty—would create a standardised foundation for cascade genetic testing and help bridge the gap between genetic diagnosis and family-level prevention.

In the USA, the Office of Public Health Genomics (OPHG) at the Centers for Disease Control and Prevention (CDC) partners with state public health departments to integrate evidence-based genomic applications and discoveries into disease prevention and population health[23]. To achieve this, the OPHG has suggested implementing the 'identify, inform, and integrate' model. First, establishments should identify evidence-based genomic applications; second, they should inform the related institutions and units about these applications; and finally, they should integrate these applications into public health at the local and national levels. Cascade genetic testing for hereditary breast and ovarian cancer and Lynch syndrome is categorised as a Tier 1 genomic application by the CDC because of its significant positive impact on public health. Cascade testing has also been found to be cost-effective in addition to enabling early protective risk management for healthy individuals in hereditary cancer settings.

This survey revealed that in Türkiye, physicians frequently make independent decisions regarding the indications for hereditary genetic testing in cancer patients. Although preventive surgical modalities are highly recommended for cases with a pathogenic *BRCA1/2* variant, decisions are often made individually or through consultations between departments, mainly because of the lack of a multidisciplinary tumour board including geneticists, which may disrupt patient–physician communication. Moreover, the rate of always requesting genetic testing for first-degree relatives of cancer patients with hereditary genetic defects was low. This gap can be attributed, in part, to a structural absence: in Türkiye. Although cancer screening and surgical recommendations for patients are primarily the responsibility of the medical oncologist, no dedicated clinical unit exists for the follow-up of cancer-free carriers of germline pathogenic variants. Individuals identified with a pathogenic *BRCA1/2* variant are informed about their inherent cancer risks by geneticists but are not followed up by them. Instead, they are referred to other internal or surgical specialties for monitoring their cancer risk. Centralising the diagnosis, screening, and follow-up of both cancer patients and carriers under a single unit would be more effective. The integration of genetic risk assessment into public health remains insufficient in our country. A system in which a public health unit takes responsibility, oversees follow-ups, and conveys decisions made by a multidisciplinary council to the patient would ensure shared responsibility among physicians and provide a more satisfactory healthcare service to individuals. This

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approach would also enable direct management of cascade genetic testing by the public health unit. We propose that such a centralised cascade coordination unit could operate through the following stepwise pathway: (1) the treating oncologist or surgeon identifies a patient with a confirmed germline pathogenic *BRCA1/2* variant and refers the patient to the medical geneticist; (2) the medical geneticist counsels the patient, obtains written consent for contact with first-degree relatives, and notifies the cascade coordination unit; (3) the unit proactively contacts consenting relatives, offers genetic counseling appointments, and tracks uptake; (4) relatives who test positive are enrolled in a structured surveillance and prevention programme managed jointly by the unit and relevant clinical departments; and (5) aggregated data on referral rates, uptake, and outcomes are reported to the public health authority to guide policy. Reimbursement for cascade counseling and testing could be integrated within the existing SGK framework by classifying cascade genetic counseling as a preventive health service, an approach already adopted in several European health systems.

In fact, in Türkiye, the Cancer Early Diagnosis Screening and Education Center (KETEM), managed by the public health unit, conducts cancer screening for healthy individuals following international guidelines and the frequency of cancer types in our country. However, this unit's services do not include a dedicated programme for screening and following up healthy individuals with hereditary pathogenic genetic disorders. The recommended starting age, screening frequency, imaging modalities, and range of cancers monitored differ substantially between individuals with and without hereditary pathogenic variants. By integrating the public health unit into the cascade genetic testing system, this gap can be closed, and communication issues among departments can be resolved.

In cascade genetic testing, the common implementation method is to contact cancer patients and encourage their relatives (patient-mediated contact) to undergo hereditary cancer testing. However, this approach is often complicated by the challenge of communicating complex cancer-related information. Cancer patients may also be coping with a recent cancer diagnosis that prompted their genetic testing. Additionally, strained family relationships may further limit the ability to convey genetic testing recommendations to other family members. According to an analysis, only half of cancer patients have informed their relatives about genetic testing[24]. Although direct contact with relatives by physicians or testing laboratories yields better results in most studies, there are also conflicting results; in addition, applicability is limited by privacy laws and a lack of infrastructure[25–30]. A meta-analysis of 87 studies revealed that 48% of cancer patients' relatives underwent cascade genetic counseling and 41% underwent cascade genetic testing. These rates were higher with direct relative contact compared with the patient-mediated approach: genetic counseling for first-degree relatives (63%, 95% CI: 49–75 vs 35%, 95% CI: 24–48) and genetic testing for first-degree relatives (62%, 95% CI: 49–73 vs 40%, 95% CI: 32–48)[25]. Further complicating matters, healthcare systems and policies have primarily focused on disease treatment rather than prevention. In this context, integrating the public health unit into the screening system could allow direct communication with cancer patients' relatives, facilitated by obtaining consent from patients during one-on-one interactions. This approach could enhance outreach and increase the participation of relatives in genetic testing and counseling programs.

Reproductive concerns are also relevant to the uptake of cascade testing: informing carriers about PGT as a means of preventing transmission to their offspring may increase engagement with hereditary cancer screening, and carriers themselves have been reported to regard PGT as a justified option[31]. In the present survey, physicians' awareness of PGT was low (approximately 30%), with some physicians raising social and ethical concerns about the procedure. Because PGT awareness was examined in depth across both physicians and patients in a companion article arising from this research programme[9], it is only briefly noted here; nonetheless, improving PGT knowledge among clinicians and affected individuals warrants incorporation into multidisciplinary genetic counseling.

This study has several limitations that should be considered when interpreting the findings. First, the surveyed physicians were predominantly from oncology-related specialties and academic or private institutions, which may not represent the broader physician population across Türkiye, particularly those in primary care or rural settings. The higher than expected knowledge levels observed are likely to reflect this selection bias, and the true knowledge gap in the general physician population may be considerably wider. Second, the study relied entirely on self-reported data. Physicians may overestimate their own knowledge and referral practices as a result of social desirability bias, and the absence of corroborating hospital registry data means that reported behaviours cannot be verified. Third, the feasibility of cascade genetic testing was assessed only indirectly through physicians' perceptions rather than actual data tracking patients and their families; this approach probably underestimates real-world barriers such as patients' reluctance, family communication difficulties, and structural gaps in follow-up. Fourth, the assessment of physicians' knowledge was based on only four survey questions, which may not fully capture the breadth of clinical knowledge relevant to *BRCA1/2* testing and managing hereditary cancer; a more comprehensive, validated instrument would strengthen these conclusions in future work. Despite these limitations, the study provides a nationwide perspective by including physicians from diverse geographic regions and practice settings, and identifies actionable gaps in knowledge, practice, and infrastructure to guide improvements in cascade genetic testing across Türkiye.

Conclusions

Although physicians in Türkiye demonstrate high willingness to utilise *BRCA1/2* testing, the implementation of cascade genetic testing remains insufficient, particularly at the level of family engagement and the uptake of preventive care, and physicians' knowledge emerged as a key driver of clinical behaviour.

Future efforts should prioritise the integration of genetic testing and counselling into public health infrastructure through dedicated centralised coordination units that streamline surveillance and follow-up for patients and their families. Establishing multidisciplinary tumour boards that include medical geneticists, and expanding physicians' education on advanced genetic tools such as PGT would further improve clinical outcomes. Implementing consent-based direct outreach to relatives of *BRCA*-positive patients could substantially increase the uptake of cascade testing. Addressing these systemic gaps through targeted policy and resource allocation is essential to optimizing the prevention and early detection of hereditary cancers in Türkiye.

Ethical statements

The study was conducted in accordance with the principles of the Declaration of Helsinki. The study protocol was approved by the Istinye University Ethics Committee (Approval No. 2024/02, Decision No. 24-56). Electronic informed consent was obtained from all participants before completion of the survey. During the preparation of this manuscript, the authors used ChatGPT (OpenAI, GPT-5 series) exclusively for English language refinement and editorial assistance.

All AI-assisted suggestions were carefully reviewed, critically evaluated, and edited by the authors. The authors verified the accuracy of the final manuscript, take full responsibility for its content, and confirm that no artificial intelligence tool was used for data collection, data analysis, interpretation of the results, or formulation of the scientific conclusions.

Author contributions

The authors confirm their contribution to the paper as follows: study conception and design: Cicin I; data acquisition: Cicin I, Kus T, Eralp Y, Urun Y, Demirci U, Bahsi T, Bozkurt M, Sendur MAN, Kaplan MA, Bilici A, Goksu SS, Karanlık H, Olmez OF, Arslan C, Kose F, Karadurmus N, Comba C, Suer E, Arık Z, Kılıçkap S; analysis and interpretation of results: Cicin I, Kus T, Eralp Y, Urun Y, Demirci U, Bahsi T; draft manuscript preparation: Cicin I. All authors reviewed the results and approved the final version of the manuscript.

Data availability

The datasets generated and/or analysed during the current study are not publicly available because they contain information that could compromise participants' confidentiality but are available from the corresponding author on reasonable request.

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Conflict of interest

The authors declare that they have no conflict of interest.

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