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# Unique perspectives about men's awareness of BRCA1/2 genetic testing in primary care

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**Men with BRCA1/2 genetic mutations have elevated risks for prostate, pancreatic, and breast cancers, yet recommendations for screening and risk assessment are unknown. Referral patterns reveal substantial under-identification and under-testing of at-risk men, with inconsistent documentation of paternal cancer history and low rates of referral to genetic services. Scarce research has examined men's perception of this gene mutation and associated variables. This study aimed to assess men's perception of BRCA1/2 genetic testing status, cancer risks, seriousness and susceptibility, and sources of health information. A cross-sectional survey with a convenience sample of men in the community without a history of cancer or a known BRCA 1/2 genetic mutation status. Susceptibility, seriousness, personal perceived risk, and sources of health information were examined. Among 234 men surveyed, 97% reported healthcare providers as their primary source of health information. Although 69% reported a family history of cancer, over half (58%) were unaware of their own or family's BRCA1/2 status, largely due to not knowing what BRCA1/2 was. While 52% perceived prostate cancer as a high-risk condition for men in general, only 22% viewed their own personal risk for prostate cancer as high. Most participants reported not knowing about BRCA1/2 gene mutations, and that they receive health related information from healthcare providers, therefore, primary care providers play a critical role in identifying those at risk by thorough assessment of patients' family cancer history and providing guidance for screening and surveillance.**

**Keywords** BRCA1/2, Men, Cancer screening, Primary care, Previvors

Globally, every year, over one million men are expected to be diagnosed with cancer[1]. Early cancer detection is strongly associated with increased cancer cure rates and survivorship[2]. Some cancers are caused by gene mutations[3]. These gene mutations are often inherited from either parent, can run in families[4], and are more prevalent in defined populations[3]. Identifying the presence of a cancer susceptibility gene can assist in early diagnosis and treatment strategies[5]. Carriers of cancer-related gene mutations who have not been diagnosed with cancer are now referred to as "previvors"[6]. They should have the opportunity to undergo screening for cancers associated with the mutation, enabling early-stage detection[1,6].

Examples of hereditary cancer-related gene mutations are BRCA1 and BRCA2. These gene mutations are usually associated with breast and ovarian cancers in females[7]. Nonetheless, BRCA1/2 gene mutations are not very prevalent in the general population[8] and therefore are not routinely screened. Moreover, in some populations, such as women and men of Ashkenazi descent, this gene mutation is much more prevalent. Ashkenazi women are screened more often for this mutation[3], yet research shows that Ashkenazi men are not[9]. The detection of a cancer-inducing gene mutation allows for more intensive surveillance and early intervention, which in the case of breast cancer in women with BRCA1/2 gene mutations, has been associated with decreased morbidity and mortality[10].

The BRCA1/2 gene mutation is autosomal dominant, and therefore, both female and male offspring of a carrier have a 50% chance equally of inheriting this gene mutation[11]. However, research has demonstrated that men are screened significantly less for the BRCA1/2 gene mutations than women[9]. BRCA1/2 gene mutations are important to detect because they are associated not only with a higher prevalence of breast cancer in women but also with a higher prevalence of colon cancer in both men and women, and prostate cancer in men. Known reasons for the lower rate of BRCA1/2 gene mutation screening in men include factors related to the men

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themselves and factors related to their healthcare providers[12]. Studies have found that men perceive this gene mutation as specific to females[12–14]. This notion is, at times, shared by some healthcare providers, as studies have reported that a man's family history of cancer, and specifically the detection of a BRCA1/2 gene mutation in family members, is less often assessed[12]. Furthermore, research has found that men tend to believe that women are the "gatekeepers" of genetic testing and information[15].

The Middle Eastern and North African (MENA) group is a designated minority subgroup with evidence indicating specific BRCA1/2 gene mutations that occur more often in this group than in the general population at large[16]. Yet, this group is underrepresented in general cancer screening research[17]. An additional population at risk are Ashkenazi Jews having up to ten times higher incidence than the general population[18,19]. According to the Central Bureau of Statistics (CBS), at the end of 2024, the population in Israel comprised approximately 9.195 million people, 7.632 million (77%) of whom are Jewish, and 2.067 million are Arab. The majority of the Arab population are Muslim (1.809 million [18%][20,21]. Due to the increased prevalence of the BRCA 1/2 genetic mutation in these population, Israel's Ministry of Health offers the test for BRCA1/2 gene mutation free of charge. The test is recommended for *all* women, and for men *at risk*.

In general, there is an underrepresentation in cancer screening studies among Middle Eastern and North African (MENA) individuals, a group that includes both Jewish and Arab populations[22]. Specifically, little is known regarding the testing practices of men. Additionally, men's knowledge and beliefs about cancer screening, including their understanding of susceptibility and seriousness, are important factors motivating cancer screening and early detection[5,13]. However, a lack of information interferes with their ability to make informed decisions. Understanding the link between risk perception and cancer screening is a long-standing challenge, as is understanding men's knowledge about their own and family BRCA1/2 gene mutation status and their sources of health information.

The rationale for the present study is to provide evidence pertaining to the gap about men's perception of BRCA 1/2 gene mutations and their needs. Therefore, the current study aims to assess Jewish and Muslim men's perception of BRCA1/2 gene mutation status, perceived risk of cancer seriousness and susceptibility, sources of health information, and associations with selective background variables.

## Methods

### Study design and participants

The study employed a cross-sectional design examining a convenience sample of men recruited online and in person (on ground). Inclusion criteria included men ages 18 years or older who were proficient in English, Hebrew, or Arabic. Exclusion criteria were men who had been diagnosed with cancer. The sample size was determined both to reflect the percentage of the populations in the country and through a power analysis, where power was set at 80%, assuming a medium effect and $p \leq 0.05$.

### Procedure

Data were collected between 2017 and 2022 via electronic and printed questionnaires. Electronic questionnaires were distributed through social media sites such as Facebook and printed versions were distributed in community centers, since some of the study population refrains from using the internet (due to cultural, religious, and generational reasons).

### Instrumentation

The Andrews & Noble Cancer-Cultural Assessment Tool (C-CAT) included selected questions from Cohen & Azaiza's Arab Cultural-Specific Barriers (ACSB) tool for breast cancer screening and were adapted for men and BRCA 1/2 gene mutation screening[23]. The questions included: BRCA knowledge of status, and personal and general perceived risk of BRCA genetic testing, cancer susceptibility, cancer seriousness, general and personal perceived risk of cancer, and sources of health information using 5-point Likert scale. The demographic questions regarding age, family status, religion, religiosity and education were adapted from Spector & Noble's Israel Heritage Assessment Tool (IHAT)[24]. The C-CAT was validated by expert Oncology and Family clinicians and the questionnaire was available in English, Arabic and Hebrew.

### Ethical considerations

The study was conducted in accordance with the principles of the Helsinki Declaration, and tacit informed consent was obtained from all participants. Participation was voluntary, no incentives were provided, and no identifying information was collected. Ethical approval was obtained from Hadassah Medical Center (#0622-12-HMO) and the Hebrew University Ethics Committee (#25112022).

### Data analysis

Data were entered into a shared Excel file, cleaned and coded (missing data were not treated or imputed, such as by mean substitution), prior to analysis using SPSS version 29. All variables were examined using univariate descriptive statistics. Parametric and non-parametric tests were employed based on data distribution and skewness. To compare "knowing what BRCA1/2 is" with "not knowing" the two items "yes/no" have a family history of BRCA1/2 were combined into one variable that represented "knowing". Subsequently, a logistic regression was used to examine variables associated with "knowing" what BRCA1/2 was. In analyses, statistical significance was set at $p < 0.05$.

| | *n* (%) |
|---|---|
| Age (*n* = 234) | |
| ≤ 40 | 76 (33%) |
| 41–59 | 94 (40%) |
| 60 and above | 64 (27%) |
| Family status (*n* = 224) | |
| Married / Live with partner | 167 (74%) |
| Single | 38 (17%) |
| Divorced | 13 (6%) |
| Widowed | 6 (3%) |
| Religion (*n* = 210) | |
| Jewish | 129 (61%) |
| Muslim | 75 (36%) |
| Christian | 5 (2.5%) |
| Other | 1 (0.5%) |
| Religiosity (*n* = 215) | |
| Not religious | 115 (53%) |
| Religious | 100 (47%) |
| Education (*n* = 201) | |
| Elementary | 6 (3%) |
| High school | 47 (23%) |
| Vocational | 14 (7%) |
| BA | 71 (35%) |
| MA | 46 (23%) |
| PhD | 17 (9%) |

**Table 1**. Background Information.

| | Very high | High | Medium | Low | No risk | Chi2 |
|---|---|---|---|---|---|---|
| Prostate cancer (*n* = 218) | | | | | | |
| Personal risk | 4 (2%) | 43 (20%) | 73 (33%) | **71 (33%)** | **27 (12%)** | 60.54 (df = 16) $p < 0.001$ |
| General risk | **38 (17%)** | **79 (35%)** | 75 (33%) | 33 (14%) | 2 (1%) | |
| Colon cancer (*n* = 211) | | | | | | |
| Personal risk | 5 (2%) | 23 (11%) | 72 (34%) | **81 (39%)** | **30 (14%)** | 74.92 (df = 16) $p < 0.001$ |
| General risk | **25 (11%)** | **62 (28%)** | 92 (41%) | 42 (19%) | 2 (1%) | |

**Table 2**. General versus personal perceived risk of prostate and colon cancer.

## Results

A total of 234 men participated in the study. As depicted in Table 1, most men were married or lived with a partner and two thirds had an academic education.

### Perceived cancer susceptibility (risk and history)

*Perceived risk*

Given the established increased risk of prostate and colorectal cancers among male BRCA1/2 mutation carriers, participants (not identified as carriers) were asked to report their perceptions of both the general risk for men and their own personal risk of developing these cancers. As presented in Table 2, a statistically significant discrepancy was observed between the perceived general and personal cancer risk for both prostate and colorectal cancers. For instance, while 52% of respondents rated the general male risk for prostate cancer as "high" or "very high," only 22% perceived their own personal risk to fall within these categories. This pattern was evident despite that 69% of participants reported a family history of cancer, and a substantial majority (*n* = 192; 92%) characterized cancer as a "very serious" or "serious" health condition.

*Awareness of family BRCA1/2 history*

When asked about being aware (knowing) of a family history of BRCA1/2 gene mutation, of the 234 participants, 10 responded "yes," 86 responded "no," and 130 (58%) reported that they "did not know what BRCA1/2 gene mutation was". To examine associations between knowing if there was a family history of BRCA1/2 and background variables, responses of "yes" and "no" to family history were grouped together as "knowing," and compared with those who indicated they "did not know what BRCA1/2 was". A logistic regression analysis

| | Very unlikely *n* (%) | Unlikely *n* (%) | Likely *n* (%) | Very likely *n* (%) |
|---|---|---|---|---|
| Doctors, Nurses etc. ($n = 227$) | 5 (2%) | 3 (1%) | **50 (22%)** | **169 (75%)** |
| Lectures, Workshops etc.($n = 215$) | 26 (12%) | 58 (27%) | **74 (34%)** | **57 (27%)** |
| People around you ($n = 218$) | 21 (10%) | 32 (15%) | **102 (47%)** | **63 (28%)** |
| Radio, TV, Newspapers etc. ($n = 216$) | 32 (15%) | 48 (22%) | **85 (39%)** | **51 (24%)** |
| Internet ($n = 219$) | 27 (12%) | 30 (14%) | **73 (33%)** | **89 (41%)** |
| Spiritual leader ($n = 214$) | **102 (48%)** | **65 (30%)** | 36 (17%) | 11 (5%) |

**Table 3**. Cancer information Sources.

yielded a statistically significant model, $\chi^2(5) = 19.84$, $p < 0.01$. Among the predictors, only education level was statistically significant ($p < 0.001$), with an odds ratio of 1.60 (95% CI [1.23, 2.08]), indicating that individuals with higher education levels had significantly greater odds of reporting "knowing" their status. Other variables, including family status, religion, religiosity, and age group were not significantly associated with "knowing".

*Cancer information sources*

As shown in Table 3, doctors and nurses were the most trusted sources of cancer information, with 97% of participants reporting they were *very likely* or *likely* to rely on them. The Internet (74%) and people around them (75%) were also commonly relied upon. Traditional media (radio, TV, newspapers) were moderately used. Lectures and workshops showed mixed responses. Although 47% of the participants were religious, spiritual leaders were the least used source, with 78% reporting they were *unlikely* or *very unlikely* to rely on them.

## Discussion

In this sample of 234 men *without* cancer or a history of the BRCA 1/2 gene mutation, over half demonstrated limited knowledge of BRCA1/2 gene mutation, thus many were unfamiliar with the term entirely and unable to assess their own cancer risk. Despite 69% reporting a family history of cancer, 58% did not know what BRCA1/2 gene mutation was, indicating a critical gap in personal awareness. While 52% rated prostate cancer risk as high for men in general, only 22% considered themselves at high risk, suggesting a disconnect between general cancer knowledge and personal risk perception.

Healthcare providers were cited as the primary source of health information by 97% of respondents, far outweighing reliance on books, media, the internet, or spiritual leaders. These findings underscore the essential role of primary care providers in educating patients, identifying hereditary risk through family history, and recommending appropriate genetic counseling and testing. This study, however, did not address primary care practices, rather the beliefs among the men in the sample that their health information is expected to come to them from primary care.

That said, current evidence suggests that even though primary care practitioners act as key gatekeepers for male genetic risk, available guidelines and practice patterns are heavily shaped by a historical focus on women[25] leaving primary care providers to extrapolate from women's guidelines about how to manage men. Furthermore, BRCA 1/2 gene mutation-related recommendations for men, are largely derived from oncology and genetics guidelines (e.g., earlier prostate cancer screening and breast awareness in male BRCA carriers) found in the National Cancer Comprehensive Network (NCCN) guidelines[26]. These cancer prevention guidelines now acknowledge male BRCA 1/2 gene mutation carriers (previvors), yet primary care guidance remains centered on women, as there is a dearth of studies reflecting general assessment aimed to men who may also be carriers. Moreover, despite these recommendations, referral to genetic services is inconsistent, and men are tested for BRCA 1/2 gene mutations at far lower rates than women[27], even when they have comparable family histories or personal cancer diagnoses.

Studies of primary care consistently identify inadequate genetics knowledge, lack of confidence, time pressure, and unclear referral pathways as barriers to incorporating genetic risk assessment into routine care[28], with additional male-specific challenges such as low awareness of BRCA-related risk among men, limited attention to paternal family history, and assumptions that BRCA 1/2 gene mutations are "a women's issue"[4]. Dekanek et al., (2020)[29], found that primary care physicians in the United States report that they lack formal cancer genetics training, and few reported ever sending patients for genetic testing. These studies allude to the lack of medical education regarding both men and women and BRCA 1/2 genetic mutation testing.

Two other key areas were explored in this study regarding the lack of knowledge about BRCA1/2 gene mutation status: (1) perceived cancer susceptibility—defined as personal and general perceived risk of cancer, family history of cancer, or family history of BRCA 1/2 gene mutation—and (2) demographic variables, including education level, religion, religiosity, and family status. Among these, education level was the only demographic variable significantly associated with knowledge. Although lower education has been linked to lower awareness in previous studies[30], most participants in the present study had post–high school or academic degrees. This finding suggests that higher education alone does not ensure understanding of BRCA1/2 gene mutations as differs from previous research[31–33]. Despite expectations that greater educational attainment would correspond with higher knowledge, the results indicate that even highly educated individuals may lack sufficient awareness of BRCA1/2 gene mutation risks.

The findings of the current study highlight a notable gap between general awareness of cancer as a major health concern and low personal perceived risk. This divergence is consistent with previous literature[14] and

can be understood through several psychological and cognitive mechanisms. One explanation is the well-documented phenomenon of optimism bias[34], wherein individuals tend to believe they are less likely than others to experience negative events, such as a cancer diagnosis. Relatedly, many participants may hold an illusion of control[34], attributing their perceived lower risk to personal behaviors such as healthy eating, regular exercise, or not smoking—despite evidence that lifestyle alone does not eliminate cancer risk. Furthermore, denial or avoidance coping[35] strategies may contribute to the minimization of personal risk, as acknowledging vulnerability can provoke anxiety[35]. The lack of direct personal experience with cancer, such as having a close family member or friend affected, may also reduce an individual's sense of susceptibility. Additionally, misinterpretation or lack of awareness regarding epidemiological statistics may lead people to underestimate their actual risk. For example, a lifetime risk statistic may appear abstract or non-immediate, especially when cancer is viewed as a disease primarily affecting older individuals.

Cultural beliefs influence genetic and cancer screening and the extent to which family members discuss personal risk to developing cancer[36–38]. This may further decrease acknowledgment of personal risk in certain populations. Collectively, these factors suggest that while general awareness campaigns have effectively communicated the societal burden of cancer, personalized risk communication strategies may be needed to enhance accurate risk perception and encourage preventive behaviors such as screening and lifestyle modification. These findings suggest that while public messaging has increased general awareness, targeted communication strategies may be needed to improve personal risk perception and encourage proactive health behaviors[39].

## Clinical implications

Numerous studies over a long expanse of time report an association between carrying the BRCA1/2 gene mutation and a higher risk of developing prostate cancer[40–42]. Previvors, carriers of a cancer-related gene, once aware of their carrier status, have a unique opportunity to increase the chances of their own survival as well as the survival of other family members, especially their offspring. Carriers of cancer-related gene mutations can assist in promoting lifestyle modifications, alongside screening for associated cancers, diagnosing, and initiating treatment at an early stage[1,6].

Nonetheless, studies have found that men are often not informed of their potential hereditary risk through female relatives[40,43,44]. Primary care providers are the gatekeepers for identifying and referring patients at risk[4]. Therefore, primary care providers are encouraged to consider obtaining a comprehensive personal and family health history, including cases of cancer, to ensure that they detect cases that would otherwise be missed or unintentionally dismissed. Genetic testing that includes BRCA1/2 gene mutation should be offered to males identified having a personal or family history or are at high risk for genetic counseling and testing that would allow earlier identification of male carriers, appropriate risk management and appropriate preventative care to the male members and their families[4].

## Case examples

As mentioned, BRCA1/2 gene mutations are commonly associated with breast cancer in women, and therefore screening men for BRCA1/2 mutations may be less intuitive. We present two case examples that underscore the critical importance of obtaining a comprehensive personal and family health history, particularly regarding cancers less commonly associated with the patient being screened. In the first example, the focus is on the man's awareness of his BRCA1/2 status, with significant implications not only for his own health but also for future generations:

A 77-year-old man was diagnosed with metastatic prostate cancer. He had participated in a random genetic screening study a decade earlier, which revealed that he carried the BRCA2 genetic mutation. Following his diagnosis of prostate cancer, he and his wife consulted their primary care physician (PCP) to discuss the potential need for genetic testing for their two daughters, aged 19 and 26. The PCP dismissed the concern, stating, "What good would this knowledge provide?" Nevertheless, the couple insisted on testing their daughters. Both were found to be BRCA2 positive and subsequently initiated early surveillance for breast and ovarian cancer, demonstrating the critical importance of hereditary cancer risk awareness not only for the patient himself but for future generations.

In the second example, involving a young woman and her own personal saga regarding BRCA testing, she also had random testing and discovered her BRCA status.

> A 40-year-old woman with two children underwent random genetic screening and was found to carry a BRCA 2 mutation. Her mother tested negative. When she approached her father, she learned that his mother had died at age 36 from breast and ovarian cancer—vital family history that had not previously been connected to hereditary cancer risk. Her sister was also found to carry the mutation. Following her diagnosis, the woman chose to undergo bilateral mastectomy and bilateral oophorectomy, while her sister opted for close surveillance.

These illustrative cases underscore the findings of our study. These cases emphasize that men may be silent carriers of critical genetic information, and yet they often remain uninformed—potentially limiting opportunities for preventive care across generations. Ultimately, the true beneficiaries of BRCA awareness and testing may be the offspring of these men, whose early detection and surveillance strategies can significantly impact cancer outcomes. Ensuring that men are educated about their potential genetic risk is therefore not only a matter of personal health, but a public health imperative.

## Study limitations

The limitations of the present study are that it included a convenience sample of men, their actual family history of BRCA1/2 carrier status was unknown and we relied on self-report. In addition, this was not a culturally

diverse population, as noted globally, but represents only the culture of a small country. In addition, the sample was highly educated, further reducing the generalizability of the results.

Furthermore, the sample did not include a confirmed group of BRCA 1/2 gene mutation carriers. Additionally, the sample size, which was more than adequate for the analysis, may not reflect a broader perspective of men which limit the external validity of the findings. Future studies are encouraged to examine these and similar aspects in relation to BRCA1/2 and other cancer-related gene mutations.

## Conclusions

Although our sample did not include a confirmed group of BRCA 1/2 gene mutation carriers, the widespread lack of awareness is striking and concerning. The current study highlights the necessity for heightened clinical vigilance and the proactive collection of comprehensive familial health histories, including both paternal and maternal histories, by healthcare professionals, especially primary care providers, who are the drivers of health information for many men. Furthermore, the findings underscore the urgent need to address existing knowledge gaps among men regarding their own hereditary cancer risks, as well as the potential implications for their descendants. Timely identification and communication of BRCA1/2 gene mutation carrier status can facilitate early intervention strategies, ultimately improving long-term health outcomes for at-risk individuals and their families. For primary care providers, each life saved through such vigilance represents the preservation of an entire world.

## Data availability

Research data supporting the findings from this study will be available by reasonable request.

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## Author contributions

Author ContributionsCSA, AN, RYZR: Conceptualization, Methodology. Writing- Reviewing and Editing.CSA, AN, RYZR, AI: Data curation, Writing- Original draft preparation. II, YBB: Data CollectionCSA, AN, RYZR, II, YBB: Data AnalysisAN, RYZR-Rony: VisualizationCSA, AN, RYZR: Project Administration CSA, AN, RYZR: Funding Acquisition All authors reviewed the manuscript.

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## Declarations

## Competing interests

The authors declare no competing interests.

## Ethical approval

Ethical Approval was obtained from Hadassah Medical Center (#0622-12-HMO) and Hebrew University ethics committee (#25112022). All participants were anonymous. Consent was implied with the participant completing the survey. Information on participant's rights were included in the email cover sheet. The researchers had no access to identifying information on the pen & paper or computer format nor the ability to track computer information.

## Additional information

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