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SYSTEMATIC REVIEW

Informational needs of individuals from families harboring *BRCA* pathogenic variants: A systematic review and content analysis

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ABSTRACT

Purpose: Personalized information is paramount to patient-centered communication and decision-making regarding risk management in hereditary cancer syndromes. This systematic review identified information needs of individuals from families harboring *BRCA* pathogenic variants and compared findings based on gender (women vs men) and clinical characteristics (patients with cancer vs previvors and *BRCA* heterozygotes vs untested relatives).

Methods: We screened 8115 studies identified from databases and citation searching. The quality of selected studies was assessed using the Mixed Methods Appraisal Tool. Narrative synthesis was conducted based on content analysis.

Results: From 18 selected studies including 1063 individuals, we identified 9 categories of information needs. Risk of bias in the selected studies was moderate. Men, untested relatives, and racial and ethnic minorities were underrepresented. Frequently required information was personalized cancer risk and risk-reducing strategies, including decision-making, family implications of hereditary cancers, psychological issues, and cascade testing. Subgroup analyses showed that information needs depended on gender, personal cancer history, and cascade testing in relatives.

Conclusion: We identified comprehensive and detailed informational needs of individuals from families harboring *BRCA* pathogenic variants and gaps in international guidelines. Needs for personalized information varied based on gender, health, and genetic testing status. Findings of this study have implications for genetic counseling, tailoring educational materials, and personalizing interventions.

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Introduction

Identifying a disease-causing germline pathogenic variant can be overwhelming for individuals and families.[1] Women have approximately 70% risk of breast cancer and 12% to 45% risk of ovarian cancer by age 80 years if they carry pathogenic variants in *BRCA1* and *BRCA2* genes (hereafter termed as *BRCA*), respectively.[2] Men with *BRCA* pathogenic variants have a 21% to 27% risk of prostate cancer and 1.2% to 6.8% risk of breast cancer by age 75 years.[3,4] Having a high cancer risk and the possibility of passing on the pathogenic variant to offspring cause significant uncertainty regarding risk management and difficulties in family communication about genetic testing results.[5-9] Women with *BRCA* pathogenic variants face significant challenges in deciding about risk-reducing options, including prophylactic mastectomy and salpingo-oophorectomy,[1,2] because these surgeries have multifarious social and medical effects due to the removal of organs not affected by cancer.[10]

Providing personalized information is an effective strategy for enhancing knowledge about the genetics of cancer risks and managing anxiety and uncertainty in families harboring pathogenic *BRCA* variants.[9,11] Personalized information also enables individuals to make informed decisions and participate in shared medical decision-making.[9,12] Members of these families have unique informational needs based on individual characteristics, eg, gender,[6,13,14] whether they had genetic testing or not,[15] and whether they have a cancer diagnosis associated with Hereditary Breast and Ovarian Cancer Syndrome (HBOC)- or they have a *BRCA* pathogenic variant but they have never been diagnosed with cancer.[6,7,12,13,16] Previous studies highlighted the increased need for personalized information; however, in practice, the most common unmet need of members from these families is the lack of adequate information.[12,14] Although genetic counseling addresses genetic testing, cancer risks, prevention, and risk management, members of families harboring pathogenic *BRCA* variants, including those who had counseling, often require additional information and actively search informational sources, eg, in the internet[5,17-20] and other media.[21]

Because the demand for reliable information addressing all members of families harboring pathogenic *BRCA* variants has been growing,[5,11] a comprehensive synthesis of empirical findings from studies with a broad focus is essential.[16] This systematic review explored the informational needs of individuals from families harboring pathogenic *BRCA* variants and compared the findings based on gender, ie, women vs men; personal cancer history, ie, individuals with an HBOC-associated cancer diagnosis (patients with cancer) vs individuals with a pathogenic variant but without cancer (previvors); and genetic testing status, ie, individuals with a confirmed pathogenic *BRCA* variant vs untested relatives who consider cascade testing. As personalization and tailoring increases the relevance of messages in medical communication,[21] our findings may assist clinicians in various disciplines meet patients' expectations, promote patient-centered communication, and increase the quality of patient care.

Materials and Methods

Design

This systematic review was performed and written following the Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) 2020 guidelines.[22] We also used Sandelowski's mixed-method review methodology to explore the topic across different types of study designs, specifically studies that collected quantitative and/or narrative data.[23] The protocol was prospectively registered in the International Prospective Register of Systematic Reviews (PROSPERO) database under review number CRD42021293285.

Eligibility criteria

Supplemental Table 1 presents detailed eligibility criteria. Studies were included if they examined the information needs of different members of families harboring pathogenic *BRCA*variants, focusing on those with a confirmed pathogenic variant and/or on individuals from families known to harbor an HBOC-associated vaiant who did not have genetic testing. We considered only original studies, including randomized trials, cross-sectional, case-control, retrospective or prospective cohort studies, and case reports. We excluded studies that did not address the outcome of interest, ie, information needs and studies that did not target the populations of interest, ie, solely focused on health care professionals or non−blood relatives, or involved animal or preclinical experiments. To improve the quality of this systematic review, we excluded studies that were not published in peer-reviewed journals, eg, conference abstracts or gray literature, eg, dissertations, white reports, and studies that did not collect primary data, eg, reviews, letters, editorials. Finally, we eliminated studies published in languages other than English because of time and resource limitations.

Literature search strategy

The scientific literature was searched in 3 stages. First, we identified the correct search terms by reviewing 2572 relevant abstracts retrieved from Ovid MEDLINE, Ovid Embase, EBSCO CINAHL, and Cochrane Central Register of Controlled Trials on September 01, 2021. The final search terms were combined using "OR" for similar terms and "AND" for different clusters. The main search terms were "*BRCA*," "hereditary breast-ovarian cancer," "HBOC," "cancer predisposition," "genetic testing," "genetic

counseling," and "informational needs" (Supplemental Table 2). Second, literature search using the predetermined search terms was performed in 5 databases, ie, Ovid MEDLINE, Ovid Embase, EBSCO CINAHL, Cochrane Central Register of Controlled Trials, and Ovid PsycINFO, from database inception to October 06, 2021. The database search was finalized on May 12, 2022. Third, we identified additional articles by manually searching the reference lists of eligible articles using Google Scholar (Figure 1). All publication periods were included in the initial search for the review of titles and abstracts.

After exporting relevant literature from each database to a bibliography management program (EndNote 20; Clarivate Analytics, Inc), we removed ineligible articles and duplicate references using the Bramer method of deduplication.[24] Two team members independently reviewed the titles and abstracts and subsequently selected the eligible studies using Rayyan software.[25] In case of discrepancies, full text of the articles was reviewed. Any disagreements between the 2 reviewers were resolved through discussion with the whole research team. Reasons for the excluding the articles are provided in Supplemental Table 3.

Risk of bias assessment

Two team members independently appraised the quality of selected studies using the Mixed Methods Appraisal Tool (MMAT) version 2018,[26] which is designed to appraise the methodological quality of studies with diverse design, including qualitative, quantitative, and mixed methods.[26] The MMAT uses 2 screening questions "clear research questions" and "collected data allow answering the research questions," and 5 questions related to the study design (Supplemental Table 4).[26] To appraise the quality of selected studies, we chose the appropriate tools based on study design.[26] We rated the 2 screening questions as "yes" or "no" depending on whether or not an appropriate answer was given to the MMAT questions and provided a rating "cannot assess" if the study provided inadequate or inaccurate information. The number of responses ranked as "yes" was summed to calculate the overall score.[26] Details are shown in Supplemental Table 4.

Data extraction

We used Bayesian conversion methods for data extraction and generated summative statements from a meta-aggregation between the collected quantitative and narrative data.[27] For studies with quantitative data, we extracted the questions used in questionnaires and the results pertinent to information needs without mining statistical data, such as *P* values, percentages of correct answers, odds ratios, and hazard ratios.[27] To create compatibility between the quantitative and narrative data, we coded narrative data directly after extraction, whereas we converted quantitative data into a narrative data format before coding.[27]

Two reviewers independently extracted all relevant data from included studies based on the following categories: bibliographic data (eg, first author, publication year, country), study aim, study design, data collection method, sample size, and population characteristics, including gender, race and ethnicity, and personal cancer history, ie, patients with cancer or previvors, genetic testing status, ie, individuals with a *BRCA* pathogenic variant or untested relatives. We also compared the extracted data based on the population characteristics mentioned earlier.

Data analysis

We conducted deductive content analysis to synthesize and compare the extracted quantitative and narrative data to answer the research questions.[28] We created an abstraction tool that randomly used selected studies to group similar variables into themes.[28] After pilot testing and refinement of the initial abstraction tool, 2 reviewers extracted the reported informational needs from eligible studies and coded them independently using a software for narrative data analysis (MAXQDA 2020; VERBI GmbH). All differences in the coding between the 2 reviewers were discussed until an agreement was reached.[28] Codes that did not fit the developed taxonomy were assigned to a new category.[29] Results present the number of studies, the codes of each category and subcategory, their percentage relative to the total number of studies, and the total number of codes.

Results

Characteristics of the selected studies

Figure 1 shows the selection process of studies included in this review. The search strategy including 5 databases (n = 7335) and manual searching (n = 780) resulted in 8115 hits, of which 18 studies were included in this systematic review.[5-8,10,12-20,30-33] Table 1 presents the characteristics of selected studies and Supplemental Table 5 presents the overall summary. The studies included a total of 1063 individuals (range = 12-204, mean = 59.1, SD = 56.4). Although we did not limit the search according to the publication year, most studies (88.9%) were published after 2011 and 27.8% were published in 2021. In terms of data collection methods, 9 studies only collected narrative data and 8 studies solely collected quantitative data. One study used a mixed-methods design to combine quantitative and narrative data. The majority of the studies were conducted in North America (55.6%) followed by Europe (27.8%). Eight studies (44.5%), including either US-based (16.7%)[30-32] or European-based samples (27.8%),[7,10,15,16,19] did not report

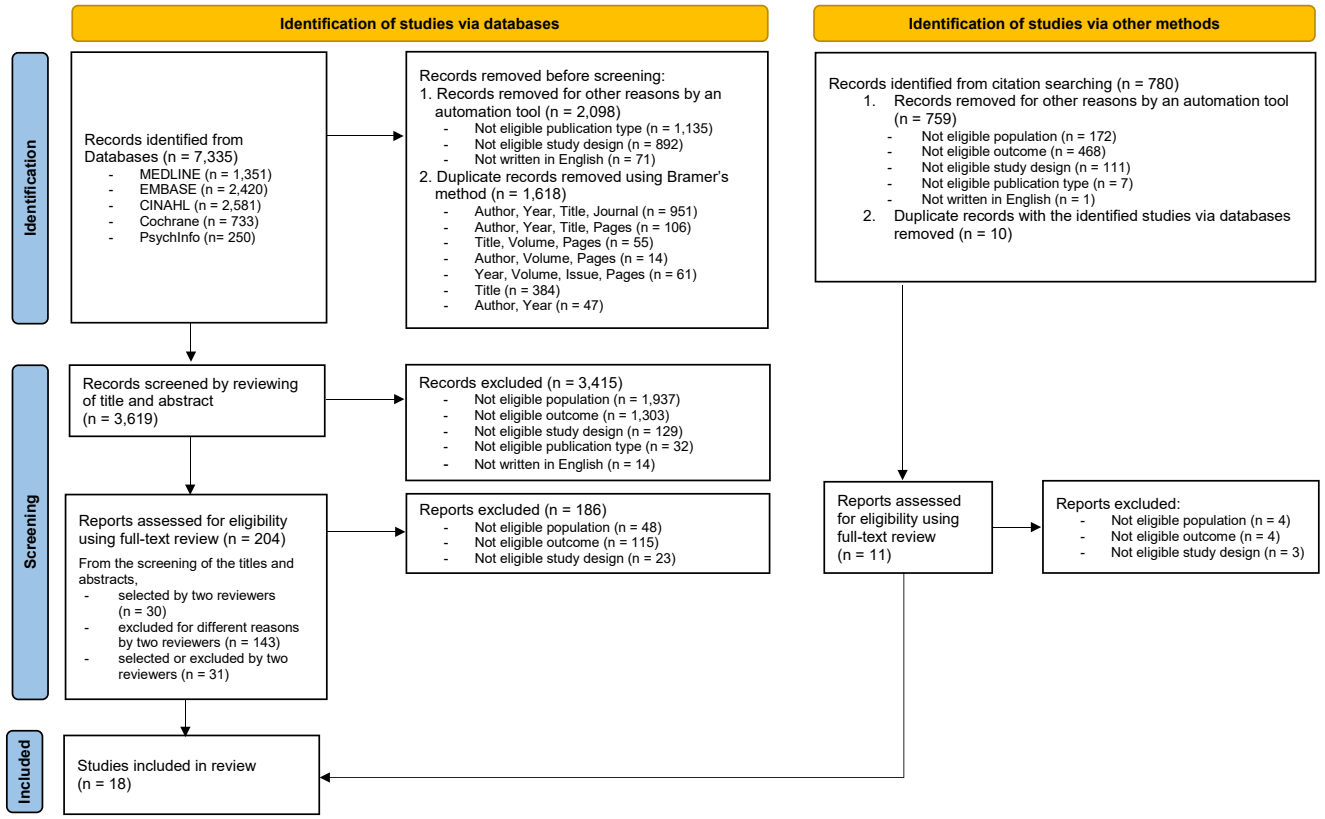

Figure 1 Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) 2020 flow diagram of searching and selection process.

the race or ethnic background of the participants. Among the remaining 10 studies, 6 included only White participants, including participants of Ashkenazi Jewish, European, or French-Canadian backgrounds;[8,12-14,17,33] 2 studies were conducted in Malaysia and Singapore and included participants of Chinese, Malay, or Indian backgrounds,[5,18] whereas the remaining 2 studies included participants of predominantly White race.[6,20] Most studies (61.1%) included exclusively women and individuals with *BRCA* pathogenic variants (66.7%) rather than men (16.7%) and untested relatives (5.6%). More than half included mixed samples of patients with cancer and previvors.

Risk of bias assessment

Supplemental Table 4 presents ratings for each methodological quality criterion. The overall mean quality score for the selected studies was 4.5 (SD = 1.07, range = 0 to 5) on the MMAT.[27] The risk of bias was moderately low, with 72%, 22%, and 6% of the studies meeting 100, ≥ 80, and 40% of the criteria, respectively. In the studies with a quantitative design, the most common risk of bias was a low response rate. No study was excluded based on quality ratings, because this review aimed to explore comprehensive informational from a wide range of studies.

Informational needs among individuals from families harboring pathogenic *BRCA* variants

We extracted 278 codes from the selected studies and grouped them into conceptual categories containing subcategories that described the scope and characteristics of the needed information. This process identified 9 distinct categories and 34 subcategories (Table 2). The most common categories of information needs were about risk-reducing strategies (94.4%), personalized risk assessment (66.7%), family implications of hereditary cancers (55.6%), decision-making for risk-reducing options (44.4%), psychological issues (38.9%), cascade genetic testing (33.3%), the role of *BRCA* genes in hereditary cancers (22.2%), social issues related to genetic testing (16.7%), and cancer treatment/diagnosis (5.6%). Overall, the ordering of information needs was similar to those of the primary studies. However, although the information about cascade testing was the third most frequently mentioned need, it was addressed by few studies.

Cancer risk–reducing strategies

Most individuals from families harboring pathogenic *BRCA* variants needed further information on screening, surveillance, and risk-reducing strategies (n = 17 studies),

Table 1 Main characteristics of selected studies ($N = 18$)

Reference, Publication Year	Country	Design (Data Collection)	Aim(s)	Sample Characteristics				
				N	Gender	Race and Ethnicity	Result of *BRCA* Testing	Personal Cancer History
Brédart et al,[15] 2021	France	Descriptive cross-sectional (questionnaires)	Explore perceived information received on breast cancer risk factors and related characteristics	161	Women, $n = 161$	NR	Relatives of individuals with *BRCA* and *PALB2*, $n =$ NR	NR
Campfield Bonadies et al,[32] 2011	United States	Descriptive cross-sectional (questionnaires)	Explore perspectives after RRSO	99	Women, $n = 99$	NR	Individuals with *BRCA* pathogenic variants, $n = 99$	NR
Cherry et al,[30] 2013	United States	Qualitative-content analysis (interviews)	Explore needs of individuals with *BRCA* pathogenic variants considering RRSO	12	Women, $n = 12$	NR	Individuals with *BRCA* pathogenic variants, $n = 12$	Patients with cancer, $n = 3$; previvors, $n = 9$
Culver et al,[20] 2011	United States	Qualitative-thematic analysis (focus groups)	Develop a decision aid on risk-reduction options for patients with breast cancer with *BRCA* pathogenic variants	20	Women, $n = 20$	White, $n = 13$; Hispanic, $n = 4$; Asian, $n = 2$	Individuals with *BRCA* pathogenic variants, $n = 20$	Patients with breast cancer, $n = 20$
Dean et al,[12] 2017	United States	Qualitative (interviews)	Explore informational needs for *BRCA* previvors	25	Women, $n = 25$	White, $n = 25$ (Ashkenazi Jewish, $n = 8$)	Individuals with *BRCA* pathogenic variants, $n = 25$	Previvors, $n = 25$
Espenschied et al,[31] 2012	United States	Descriptive cross-sectional (action research activities)	Assess information needs of patients with hereditary cancer	79	Women, $n =$ NR; men, $n =$ NR	NR	Individuals with *BRCA* pathogenic variants, $n =$ NR; high risk for *BRCA*-related cancer, $n =$ NR; family or friends of patients, $n =$ NR	Patients with breast or ovarian cancer, $n =$ NR; previvors, $n =$ NR
Hurley et al,[33] 2012	United States	Qualitative (interviews)	Assess attitudes about preimplantation genetic diagnosis of individuals with *BRCA* pathogenic variants	33	Women, $n = 29$; men, $n = 4$	White, $n = 30$	Individuals with *BRCA* pathogenic variants, $n = 33$	NR

(continued)

Table 1 Continued

Reference, Publication Year	Country	Design (Data Collection)	Aim(s)	Sample Characteristics				
				N	Gender	Race and Ethnicity	Result of *BRCA* Testing	Personal Cancer History
Jacobs et al,[16] 2017	United Kingdom	Descriptive cross-sectional (Delphi consensus, questionnaires)	Identify the key messages required by women with breast/ovarian cancer who undergo genetic testing	16	Women, *n* = 16	NR	Individuals with *BRCA* pathogenic variants, *n* = 16	Patients with breast or ovarian cancer, *n* = 16
Kautz-Freimuth et al,[19] 2021	Germany	Qualitative (focus groups)	Develop a decision aid on risk-reduction options for individuals with *BRCA* pathogenic variants	19	Women, *n* = 19	NR	Individuals with *BRCA* pathogenic variants, *n* = 19	Patients with cancer, *n* = 9; previvors, *n* = 10
Liede et al,[8] 2000	Canada	Descriptive cross-sectional (questionnaires)	Identify unmet needs and describe men's experiences with genetic services	59	Men, *n* = 59	White, *n* = 59 (Ashkenazi Jewish, or European)	Individuals with *BRCA* pathogenic variants, *n* = 59	Patients with cancer, *n* = 12; previvors, *n* = 47
Metcalfe et al,[17] 2000	Canada	Descriptive cross-sectional (questionnaires)	Evaluate needs of individuals with *BRCA* pathogenic variants undergoing genetic counseling	79	Women, *n* = 79	White, *n* = 79 (French-Canadian, Ashkenazi Jewish, European, or Hispanic)	Individuals with *BRCA* pathogenic variants, *n* = 79	Patients with cancer, *n* = 46; previvors, *n* = 33
Modaffari et al,[10] 2019	Italy	Descriptive cross-sectional (questionnaires)	Evaluate expectations and concerns about cancer risk—reducing surgery	204	Women, *n* = 204	NR	Individuals with *BRCA* pathogenic variants, *n* = 192; high risk for *BRCA*-related cancer, *n* = 12	Patients with breast or ovarian cancer, *n* = 100; previvors, *n* = 104
Peshkin et al,[13] 2021	United States	Qualitative (focus groups)	Develop a web-based educational tool for untested men in *BRCA*-positive families	13	Men, *n* = 13	White, *n* = 13	Individuals with *BRCA* pathogenic variants, *n* = 9; untested relatives, *n* = 3; true negative *BRCA*, *n* = 1	Previvors, *n* = 13

(continued)

Table 1 Continued

Reference, Publication Year	Country	Design (Data Collection)	Aim(s)	Sample Characteristics				
				N	Gender	Race and Ethnicity	Result of *BRCA* Testing	Personal Cancer History
Rauscher et al,[14] 2018	United States	Qualitative (interviews)	Examine management of uncertainty and information needs of men about *BRCA*-related cancer risks	25	Men, *n* = 25	White, *n* = 25	Individuals with *BRCA* pathogenic variants, *n* = NR; first degree relatives of individuals with *BRCA* pathogenic variants, *n* = NR	NR
Sa'at et al,[18] 2022	Malaysia	Qualitative-grounded theory with thematic analysis (interviews)	Explore decision-making needs of individuals with *BRCA* pathogenic variants	31	Women, *n* = 31	Asian, *n* = 31 (Chinese, *n* = 17; Malay, *n* = 8; Indian, *n* = 6)	Individuals with *BRCA* pathogenic variants, *n* = 31	Patients with breast cancer, *n* = 28; previvors, *n* = 3
Visser et al,[7] 2016	Netherlands	Descriptive cross-sectional (questionnaires)	Evaluate the efficacy of group medical consultations on yearly breast cancer surveillance of individuals with *BRCA* pathogenic variants	132	Women, *n* = 132	NR	Individuals with *BRCA* pathogenic variants, *n* = 132	Patients with breast cancer, *n* = 10; previvors, *n* = 122
Young et al,[6] 2019	Australia	Mixed methods (interviews, questionnaires)	Clarify and compare information needs of young adults (18-25 vs 26-40 y)	32	Women, *n* = 25; men, *n* = 7	White, *n* = 25; Asian and Arabic, *n* = 7	Individuals with *BRCA* pathogenic variants, *n* = 20; individuals from families with *BRCA* pathogenic variants, *n* = 12	Patients with cancer, *n* = 3; previvors, *n* = 29
Yuen et al,[5] 2020	Singapore	Qualitative (interviews)	Explore informational needs of individuals with *BRCA* pathogenic variants in Asia	24	Women, *n* = 22; men, *n* = 2	Asian, *n* = 24 (Chinese, *n* = 15; Malay, *n* = 3; Indian, *n* = 2; other, *n* = 4)	Individuals with *BRCA* pathogenic variants, *n* = 24	Patients with breast, ovarian, or prostate cancer, *n* = 17; previvors, *n* = 7

NR, not reported; *RRSO*, risk-reducing salpingo-oophorectomy.

Table 2 Informational needs of individuals from families harboring pathogenic *BRCA* variants (N = 18 studies, 278 codes)

Informational Needs	Codes of Needs, N (%)[a]	Studies, N (%)[b]	References
Cancer risk—reducing strategies	132 (47.5)	17 (94.4)	5-8,10,12-20,30-32
Types of cancer risk—reducing options	9 (3.2)	6 (33.3)	6,7,12,16,17,30
Risk-reducing bilateral salpingo-oophorectomy	40 (14.4)	11 (61.1)	6,7,10,12,15,16,18-20,30,32
Additional information	5 (1.8)	4 (22.2)	17-20
Timing	3 (1.1)	3 (16.7)	6,12,30
Benefit	5 (1.8)	4 (22.2)	6,16,20,32
Limitation	21 (7.6)	10 (55.6)	6,7,10,15,16,18-20,30,32
Cost and insurance coverage	4 (1.4)	2 (11.1)	6,12
Operation procedures	2 (0.7)	2 (11.1)	18,32
Risk-reducing bilateral mastectomy	25 (9.0)	8 (44.4)	6,7,10,12,16,17,19,20
Additional information	3 (1.1)	3 (16.7)	17,19,20
Timing	2 (0.7)	2 (11.1)	6,12
Benefit	6 (2.2)	3 (16.7)	6,16,20
Limitation	4 (1.4)	3 (16.7)	6,10,20
Cost and insurance coverage	4 (1.4)	2 (11.1)	6,12
Operation procedures and breast reconstructions	6 (2.2)	4 (22.2)	6,7,16,19
Screening and surveillance	36 (12.9)	11 (61.1)	5-7,10,12,13,16,17,19,20,31
Additional information	2 (0.7)	2 (11.1)	17,20
Timing	5 (1.8)	4 (22.2)	7,12,16,19
Benefit	5 (1.8)	5 (27.8)	6,12,19,20,31
Limitation	8 (2.9)	7 (38.9)	6,10,12,16,19,20,31
Methods	13 (4.7)	8 (44.4)	5-7,10,12,13,16,19
Cost and insurance coverage	3 (1.1)	3 (16.7)	6,12,19
Other risk-reducing options: chemoprevention, contraceptives	5 (1.8)	4 (22.2)	6,7,12,20
Cancer risk for men, risk-reducing options	10 (3.6)	3 (16.7)	8,13,14
Lifestyle behaviors	10 (3.6)	4 (22.2)	8,10,15,30
Personalized cancer risk	37 (13.3)	12 (66.7)	5-7,10,12-14,16,18,19,30,31
Personalized cancer risk: timing of cancer development and types of cancer based on age and family history	13 (4.7)	8 (44.4)	5-7,10,16,18,30,31
Previvors' cancer risk	10 (3.6)	5 (27.8)	6,7,12,13,16
Survivors' recurrence, future cancer risk	6 (2.2)	2 (11.1)	16,19
Men's cancer risk: prostate, male breast cancer, etc	8 (2.9)	3 (16.6)	6,13,14
Family implications of hereditary cancer	25 (9.0)	10 (55.6)	5-8,12,13,15,16,31,33
Disclosing genetic test results and communication with family/relatives	8 (2.9)	6 (33.3)	5-8,12,13
Inheriting cancer risk for family/relatives	5 (1.8)	5 (27.8)	6,7,13,16,31
Inheriting cancer risk for current/future children	5 (1.8)	3 (16.7)	6,13,16
Reproductive issue	7 (2.5)	4 (22.2)	6,7,15,33
Decision-making for risk-reducing options	14 (5.0)	8 (44.4)	5-7,12,17,18,20,30
Need support	6 (2.2)	5 (27.8)	5-7,12,20
Need peer's experience	7 (2.5)	5 (27.8)	5,12,17,18,30
Need recommendations from health care providers based on guidelines	1 (0.4)	1 (5.6)	12
Psychological issues	22 (7.9)	7 (38.9)	6,7,12,15,17-19
Emotional management and coping	16 (5.8)	7 (38.9)	6,7,12,15,17-19
Additional emotional support	2 (0.7)	2 (11.1)	17,18
Peer support	2 (0.7)	2 (11.1)	12,18
Referral to psychologist	2 (0.7)	1 (5.6)	6
Cascade genetic testing	27 (9.7)	6 (33.3)	5,6,12-14,16
Implication	9 (3.2)	4 (22.2)	6,13,14,16
Benefit	3 (1.1)	2 (11.1)	6,16
Cost and insurance coverage	6 (2.2)	3 (16.7)	6,12,13
Process	3 (1.1)	2 (11.1)	6,12
Understanding test results	5 (1.8)	2 (11.1)	5,6
Terminology	1 (0.4)	1 (5.6)	6

(continued)

Table 2 Continued

Informational Needs	Codes of Needs, *N* (%)[a]	Studies, *N* (%)[b]	References
The role of *BRCA* genes in hereditary cancers	11 (4.0)	4 (22.2)	6,13,16,31
General information of *BRCA*: mechanisms, prevalence, and penetrance	8 (2.9)	4 (22.2)	6,13,16,31
Inheritance likelihood of *BRCA*	3 (1.1)	3 (16.7)	13,16,31
Social issues related to genetic testing	8 (2.9)	3 (16.7)	6,12,13
Disclosing test results to friends	4 (1.4)	2 (11.1)	6,12
Future health insurance and employment	2 (0.7)	2 (11.1)	12,13
Laws for genetic discrimination	1 (0.4)	1 (5.6)	13
Social resource	1 (0.4)	1 (5.6)	12
Cancer treatment and prognosis	3 (1.1)	2 (11.1)	17,19

[a]The percentage of codes in informational needs was determined by dividing the number of codes from each unique subcategory by the sum of all codes ($N = 278$).

[b]The percentage of reported studies was determined by dividing the number of total studies by the number of selected articles in this review ($N = 18$).

including salpingo-oophorectomy and bilateral mastectomy and nonsurgical methods, ie, chemoprevention and contraceptives and lifestyle.[5-7,12,16,17,20,30] They requested detailed information about timing and benefits, effect on cancer risk, limitations, costs and insurance coverage, and specific procedures. Among risk-reducing strategies, surgical methods ($n = 12$ studies) were discussed more than chemoprevention and contraceptives ($n = 4$ studies) and lifestyle ($n = 4$ studies).

Personalized cancer risk

A total of 12 studies reported that individuals from families harboring pathogenic *BRCA* variants required information on the risk of developing cancer, the timing of cancer onset, and the types of cancers associated with the specific variants.[5-8,10,12-14,16,18,19,30,31] The needs for personalized information depended on age, gender, and personal and family cancer history.[5-8,10,12-14,16,18,19,30,31]

Family implications of hereditary cancers

Individuals from families harboring pathogenic *BRCA* variants required extensive information regarding family implications of hereditary cancers, such as how to disclose test results and cancer risk to children, partners, and relatives ($n = 6$ studies).[5-8,12,13] They also requested information about the risk of child(ren) and relatives inheriting the pathogenic variant and developing cancer ($n = 5$ studies).[6,7,13,16,31] Four studies reported information needs regarding *BRCA*-related fertility preservation and contraception, preimplantation genetic diagnosis, timing to start a family, and breastfeeding.[6,7,15,33]

Decision-making for cancer risk–reducing strategies

Eight studies reported that individuals from families harboring pathogenic *BRCA* variants needed decisional support regarding risk-reduction options,[5-7,12,20] primarily from peers[5,12,17,18,30] but also from health care providers and medical guidelines.[12]

Psychological issues

There was significant uncertainty and fear caused by the pressure to find a partner and start a family, feelings of parental or familial guilt, and diagnosis of cancer in relatives.[6,7,12,15,17-19] Feelings of vulnerability created the need to manage negative emotions by seeking support from peer or patient groups[12,18] and gathering information about referrals for psychological support.[6]

Cascade genetic testing

Individuals with pathogenic *BRCA* required detailed information on cascade testing of relatives,[5,6,12-14,16] including the appropriate age and time for testing,[6,13,14,16] benefits,[6,16] scheduling,[6,12] costs,[6,12,13] and insurance coverage.[6,12,13] They requested information on the meaning of test results such as positive or negative, identifying a variant of uncertain significance,[5,6] and additional terminology related to genetic testing.[6]

The role of *BRCA* genes in hereditary cancer

Four studies reported that individuals from families harboring pathogenic *BRCA* variants needed general information about HBOC, which included basic genetic information, the frequency of pathogenic variants in *BRCA* genes, their association to hereditary cancer, and the patterns of inheritance.[6,13,16,31]

Social issues related to genetic testing

Social issues included ways to disclose testing results to friends,[6,12] issues of genetic discrimination, future health insurance and employment,[12,13] related legislation for genetic discrimination,[13] and how to find social support groups.[12]

Cancer treatment and prognosis

In 2 studies, patients with cancer requested additional information about various treatment modalities and prognosis,[17,19] specifically treatment information according to immunohistochemical subtype.[19]

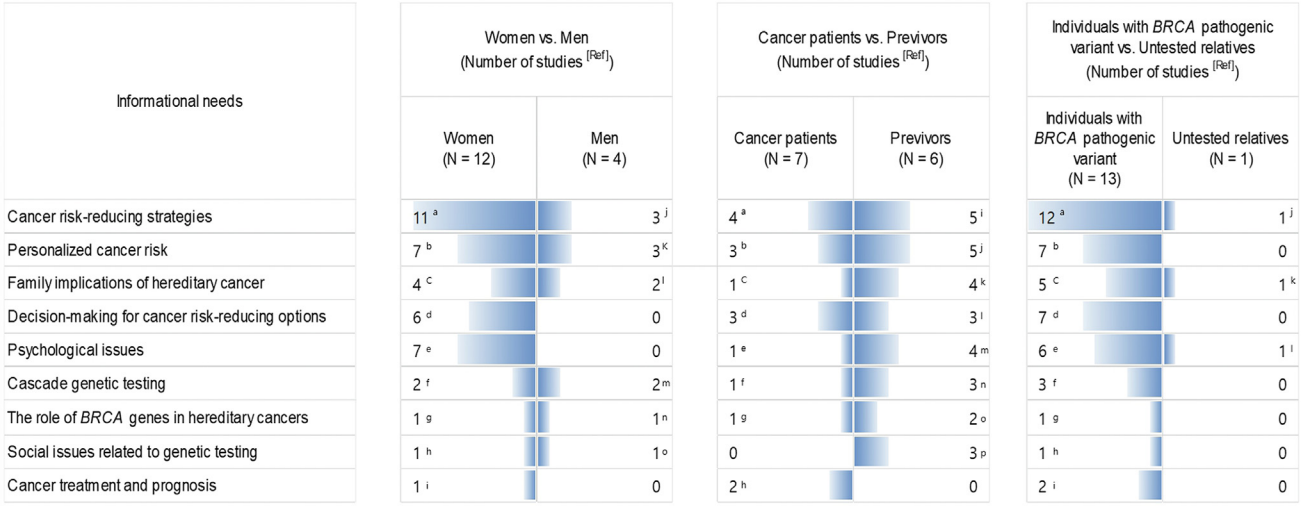

Figure 2 Subgroup analyses: Differences in informational needs according to gender and clinical characteristics of individuals from families harboring pathogenic *BRCA* variants. Women vs men:[a] 7,10,12,15-20,30,32; [b] 5,7,10,12,16,18,30; [c] 7,12,15,16; [d] 7,12,17,18,20,30; [e] 7,12,15,17-19,30; [f] 12,16; [g] 16; [h] 12; [i] 19; [j] 8,13,14; [k] 6,13,14; [l] 8,13; [m] 13,14; [n] 13; [o] 13. Patients with cancer vs previvors: [a] 16,18-20; [b] 10,16,18; [c] 16; [d] 6,18,20; [e] 18; [f] 16; [g] 16; [h] 17,19; [i] 6,7,12,13,19; [j] 6,7,12,13,16; [k] 6,7,12,13; [l] 6,7,12; [m] 6,7,12,19; [n] 6,12,13; [o] 6,13; [p] 6,12,13. Individuals with pathogenic *BRCA* variants vs untested relatives: [a] 5,7,8,10,12,16-20,30,32; [b] 5,7,10,12,16,18,30; [c] 5,7,12,16,33; [d] 5,7,12,17,18,20,30; [e] 7,12,17-19,30; [f] 5,12,16; [g] 16; [h] 12; [i] 17,19; [j] 15; [k] 15; [l] 15.

Comparison of informational needs according to gender and clinical characteristics

Figure 2 presents specific information needs based on gender (women vs men) and clinical characteristics (patients with cancer due to a *BRCA* pathogenic variant vs previvors and individuals with *BRCA* pathogenic variants vs untested relatives). We included studies with homogenous samples, eg, only individuals with pathogenic *BRCA* variants or only untested relatives, and we excluded studies with mixed samples because we could not differentiate the findings for each of the subgroups. We did not examine information needs based on race or ethnic background because most studies either did not provide this information or they did not provide differential findings based on race and ethnicity because of small sample sizes.

We identified 12 studies including only women[5,7,10,12,15-20,30,32] and 4 studies including only men.[6,8,13,14] Women needed a wide range of information in all categories,[5,7,10,12,15-20,30,32] particularly decision-making for risk-reducing options[7,12,17,18,20,30] and emotional management and coping strategies,[7,12,15,17-19,30] which were not reported for men. Men required gender-specific information about prostate and male breast cancer risks[6,13,14] and risk management strategies.[8,14]

Patients with cancer (n = 7 studies)[6,10,16-20] and previvors (n = 6 studies)[6,7,12,13,16,19] required similar information regarding the role of pathogenic *BRCA* genes, personalized cancer risk, psychological issues, familial issues, and cascade genetic testing of relatives. However, previvors required further information about the risk of developing cancer in the future[6,7,12,13,16] and risk-reducing strategies.[6,7,12,13,19] Patients with cancer required more information about the risk of recurrence on the unaffected side and risk for cancer in other organs,[16,19] whereas this information was not relevant for previvors.

Most studies (n = 13 studies) focused on individuals identified with a pathogenic variant[5,7,8,10,12,16-20,30,32,33] rather than untested relatives from families harboring pathogenic *BRCA* variants (n = 1 study).[15] The most frequently reported informational needs of individuals with *BRCA* pathogenic variants were personalized cancer-risk[5,7,10,12,16,18,30] and risk-reducing options,[5,7,8,10,12,16-20,30,32] whereas the least frequently reported ones were social issues related to genetic testing[12] and the role of pathogenic *BRCA* genes in developing hereditary cancer.[16] Untested relatives focused on risk-reducing salpingo-oophorectomy and lifestyle, information on reproduction, and psychological issues, eg, emotional management and coping.[15]

Discussion

This comprehensive review examined the information needs of diverse individuals from families harboring pathogenic *BRCA* variants. We identified 9 categories of informational needs from 18 studies based on gender, personal cancer history, and genetic testing status. Although the selected studies included an overall large sample, men, untested relatives, and racial or ethnic minorities were underrepresented. In the following sections, we compare our findings with prior literature and with international guidelines to shed light on implications for research and practice.

An accelerating number of studies focused on individual members of families harboring pathogenic *BRCA* variants.[12,34] Our findings showed that in recent years, research has focused on patient needs and patient-centered care,[34,35]

following an increasing demand for genetic counseling and testing.[36] Our findings offer salient directions for future research, focusing on the information needs of men and untested relatives and addressing limitations of prior studies, especially the limited representation of individuals and families from minority racial and ethnic backgrounds.

Families harboring pathogenic *BRCA* variants required a broad scope of information, primarily related to living with cancer risk and the risk of child(ren) and relatives inheriting the familial pathogenic variant. This is different from the information needs of patients without hereditary cancer,[21,34] who focus primarily on prognosis, treatment and side effects, and rehabilitation.[11,12] Families harboring pathogenic *BRCA* variants require unique information to cope with the challenges originating from "living with *BRCA*." Their informational needs are dynamic, changing according to the individual life trajectories, cancer status, and genetic testing status. These findings are consistent with international guidelines regarding the relevance of genetic counseling before a cancer diagnosis, during cancer care, and during rehabilitation.[37] Thus, it is crucial to evaluate the information needs of individuals with genetic risks over extended periods of time.[35] Finally, our findings reveal important knowledge gaps regarding the information needs of at-risk relatives, who may consider cascade genetic testing, and make a significant contribution to promoting equity in genomic health care.[38]

The greatest concern of families harboring pathogenic *BRCA* variants included cancer risks and risk management, which have been emphasized as major topics of discussion in genetic counseling following international guidelines by the National Comprehensive Cancer Network (NCCN), American Society of Clinical Oncology (ASCO), European Society for Medical Oncology (ESMO), and US Preventive Services Task Force (USPSTF).[37,39] This is not surprising considering that pathogenic *BRCA* variants have high penetrance and are associated with the development of multiple types of cancer.[2] However, our findings revealed gaps in the treatment and risk management guidelines when comparing the actual information needs of individuals from these families who require a broad spectrum of detailed information, including timing, benefits, limitations, costs and insurance coverage of surgical procedures, details about nonsurgical options that could replace or delay risk-reducing surgeries, and decisional support based on peer experiences. With the exception of NCCN guidelines, which emphasize the importance of covering cancer risk−reduction options and family implications of hereditary cancer, most guidelines do not provide guidance regarding the specific content or the format of genetic counseling sessions.[37,39] Our findings can be used to enrich current guidelines and improve patient satisfaction with genetic care, because they have implications for personalized counseling of different members from families harboring pathogenic *BRCA* variants.[40]

Our findings can also be used to enhance the content and structure of tailored educational materials and technology-based informational sources, eg, chatbots and patient portals for these families.[41] They can also inform assessment tools that evaluate information needs before counseling, not only for families harboring HBOC-associated variants but also for possibly other hereditary syndromes. Communication during genetic counseling tends to be primarily unidirectional and tends to provide large amounts of information during mostly 1 or 2 sessions.[42] Our findings can facilitate tailoring and effective communication during genetic counseling, eg, the consultation could focus on patients' main concerns and cover unaddressed information with educational materials.

Finally, individuals from families harboring pathogenic *BRCA* variants need additional information and support regarding the management of psychosocial issues. Primary studies reported concerns about family implications of hereditary cancer and the psychological and social issues related to the testing, indicating the significant interpersonal aspects of genetic risk.[35] Determining the psychosocial needs during the care trajectory can further clarify the type of intervention and resources that will be most effective and valuable for a particular population.[35]

One limitation of this study was that it may not include meaningful content published in languages other than English. Most studies were conducted in North America and Europe, and the results may not be generalizable to families harboring pathogenic *BRCA* variants in other countries. Although the overall sample size was substantial, results from subgroup analyses in certain domains with small sample sizes, ie, cascade genetic testing of relatives, the role of *BRCA* genes in hereditary cancers, and social issues related to genetic testing, should be interpreted with caution. We could not identify information needs based on race and ethnic background because of the large amount of missing or mixed data regarding race and ethnicity in primary studies. We did not include primary studies of information needs of individuals with pathogenic variants in other high-penetrance genes, eg, *PALB2*, because the scientific literature on this topic is extremely scarce. Finally, only 1 study moved beyond individuals identified with a pathogenic *BRCA* variant, and our findings may not reflect informational needs among untested relatives from these families.

Despite the aforementioned limitations, the strength of this systematic review is that we investigated the information needs directly reported by individuals from families harboring pathogenic *BRCA* variants by including a large number of studies that collected narrative data and by converting the survey data into compatible narrative codes. Informational needs of individuals from these families are unique and dynamic, depending on life trajectories and cancer status. Our findings indicate the need for multiple genetic counseling sessions over a prolonged period of time along the care continuum, because members of these families live with cancer risk their entire lives. In light of the costs and feasibility of providing genetic counseling in the long term, efforts should focus on developing and using alternative counseling methods, eg, telephone and technology-based methods that are equally

acceptable by patients and can supplement in-person counseling.[43,44] Notably, international guidelines do not address the content and format of genetic counseling, and our findings can be used to address this gap in clinical practice. Healthcare professionals and specialists should provide tailored and detailed genetic information based on gender, cancer status, and genetic testing status; address psychosocial concerns; and provide support according to individual life stages. The identified typology, including the content and structure of informational needs, has implications for guiding genetic counseling sessions and for developing tailored educational materials, personalized interventions, and assessment tools to gauge information needs before genetic counseling sessions.

Data Availability

The data sets generated and/or analyzed during this study are available from the corresponding author on request.

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Ethics Declaration

This study did not include human subjects or animal research.

Conflict of Interest

The authors declare no conflicts of interest.

Additional Information

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