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RESEARCH

Exploring family communication preferences in hereditary breast and ovarian cancer and Lynch syndrome: a national Canadian survey

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Abstract

Background Individuals affected with cancer predisposition (CPS) syndromes such as *BRCA1*, *BRCA2* or Lynch syndrome (LS) are at an elevated risk of multiple cancers. Identifying high-risk individuals is important if they are to access risk-reducing strategies. Interventions such as risk-reducing salpingo-oophorectomy in carriers of *BRCA* pathogenic or likely pathogenic (P/LP) variants or regular colonoscopy for carriers of LS P/LP variants are highly effective and reduce mortality. Despite clear evidence that the identification of at-risk relatives has value, the uptake of cascade testing remains at approximately 50%. It is important to understand strategies and barriers to testing to facilitate communication in families identified as haveing a hereditary cancer syndrome, to improve uptake of counselling and testing.

Method A national online survey of both Canadian probands (the first member in a family to have genetic testing and who were variant positive, regardless of a cancer diagnosis) and their at-risk relatives. Respondents were individuals affected with hereditary breast and ovarian cancer (HBOC) and LS. The survey was constructed based on a review of the literature and authors' feedback. Both open and closed-ended questions were used for items on demographic characteristics, risk perception, genetic test results and cancer diagnosis. Items on experiences with hereditary cancer risk communication, communication challenges, preferences and supports required were explored using a 5-point Likert scale.

Results Responses indicated a high level of acceptance for the proband's direct involvement in family communication with the support of a health care provider (67% among the probands given a family letter and 55–57% among those who were not given a family letter). Respondents without a personal history of cancer were more likely to endorse a health care professional's help with family communication compared to those with a personal history of cancer ($p = 0.031$). Preferences for family member outreach also varied by education level, annual income, marital status and geographic location. Similarities were noted between the probands and relatives on communication outreach preferences.

Conclusion While the family-mediated approach to communication remains the standard across many cancer genetics programs, participants note that additional support is necessary for dissemination of result information among relatives. Because family dynamics and communication vary widely, alternative options that retain the probands' involvement in family communication but add support from a health care provider should be explored.

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Background

CPS or hereditary cancer accounts for 5–10% of all cancers (Garber et al. 2005; Wang et al. 2015). There is strong evidence of reduced cancer morbidity and mortality when individuals at high cancer risk due to CPS, such as HBOC and LS, are identified before a cancer is diagnosed due to access to early prevention and screening (Dinh et al. 2010; Guzauskas et al. 2020).

Once a P/LP variant is identified via genetic testing in one family member, their first-degree and second-degree relatives are at 50% and 25% risk, respectively, of carrying the same familial P/LP variants and are eligible for genetic testing (Daly et al. 2021). Cascade screening provides an opportunity to implement cancer risk reduction strategies in healthy people before the onset of cancer, for those who wish them. The impact of preventive care is especially beneficial to women at risk for ovarian cancer, as no screening test is effective for early detection and most cases present at advanced, incurable stages (Gupta et al. 2017; Menon et al. 2011; US Preventive Services Task Force 2018; Thigpen et al. 2011).

In traditional genetics programs (Dheensa et al. 2017; Mendes et al. 2023) the proband is usually given a letter at the time of disclosure of genetic testing results. Such a letter explains the risk of CPS and the benefits of surveillance or preventive management. It is expected that the proband will share these letters with at-risk relatives. However, the literature reveals that only about 50% of first and further-degree relatives seek counselling (Hinchcliff et al. 2019; Loader et al. 2002; Lowery et al. 2010; Menko et al. 2013; Marleen et al. 2019). Some studies report even lower rates, between 20 and 40% (Hodgson et al. 2014; O'Neill et al. 2006; Suthers et al. 2006). Previous research identified that family communication barriers concerning the sharing of cancer risk information may be important to explore to meet a family's communication preferences (Delikurt et al. 2014; Srinivasan et al. 2020a; Vogel et al. 2018).

Although the benefits of genetic testing of relatives in families with a cancer predisposition syndrome are well documented, the question of how best to contact relatives is a complex concern. Issues surrounding legislation, patient and relative privacy and confidentiality, duty to warn and ethics, as well as family relationships and dynamics must be carefully considered in determining optimal relative outreach and communication strategies.

The family-mediated approach (i.e., "the family letter") remains the standard of care in informing individuals of their hereditary cancer risk. Direct contact by a health care provider is typically limited by legal obligations to maintain confidentiality and protect the proband's privacy (Legislative Summary of Bill S-201 2022; CDC 2021; Coleman et al. 1992; Hakonarson et al. 2003; The Government of Iceland 2014a; Henrikson et al. 2021; Lucassen and Hall et al. 2019; Meggiolaro et al. 2020; OECD 2022; Attorney-General's Department 2022; The Government of Iceland 2014b). In such cases, a health care provider may not share genetic test results with the proband's relatives without consent from the relative. The impact of family dynamics and communication patterns also influence how relatives receive information on cancer risk information, and ultimately the decision for counselling and testing (Armstrong et al. 2005; Forrest et al. 2003; Miller et al. 2005; Peters et al. 2005; Silva et al. 2022; Wang et al. 2005). Numerous studies have focused on evaluating the dissemination of the correct risk information to family members—the capacity to share seemingly complex genetic information and how the proband's knowledge and interpretation of risk information affect disclosure (Frey et al. 2019; Young et al. 2020; Baroutsou et al. 2021; Dean et al. 2021). Notwithstanding, the existence of harmonious relationships within the family may even have a more substantial influence on genetic counselling and testing uptake (Di Pietro et al. 2020). Successful communication between probands and their families requires ongoing support (Pollard et al. 2020) and several interventions have been investigated as an alternative to the family-medicated approach. These include video conferencing, group sessions (Hynes et al. 2020; Zilliacus et al. 2010) and mobile health apps (Vogel et al. 2018, 2019; Haas et al. 2021). The importance of a better understanding of attitudes and preferences towards various outreach strategies is underscored by the stated challenges with the acceptable method of practice and the deficiency in information on the success of alternative options that have been researched.

In this study, we explored probands' and relatives' preferences for the communication of hereditary cancer risk information to help inform risk communication models and policies. Specifically, we aimed to:

1. Explore and describe preferences for various methods of family member outreach among BRCA1/2 and LS P/LP variant carriers and relatives.
2. Explore clinical, social and demographic factors related to outreach preferences.

Methods

Participants

The study targeted participants within Canada above the age of 18 years and included probands with or without a cancer diagnosis who were carriers of *BRCA1* and *2* or LS P/LP variants who had at least one first or second-degree relative at increased risk of CPS. At-risk relatives who were informed of the family's risk by a proband were also included in this study. It was required that participants had the ability to or willingness to provide informed consent and the ability to understand English.

Survey administration and recruitment

Participants were invited to complete an online survey hosted on Qualtrics, the approved secure survey platform of Memorial University. The survey advert and link were shared widely across the Canadian health care system via email and on several social media platforms from October 2021 to January 2022. The ad appealed to individuals who had informed their relatives of the family's risk of HBOC and LS, and to persons who were told about their elevated risk by a member of their family. Interested participants were asked to follow a link to access the survey. Patients attending the Newfoundland (NL) Hereditary Cancer Prevention Clinic and the British Colombia (BC) Gynecologic Cancer Survivorship Clinic were invited to join the study by their physician, a member of the research team. Team members identified other oncology providers, researchers, and patient partners in their networks who were invited to share the study information with eligible participants. Interested patients were encouraged to share study information with their relatives. Investigators are members of the Canadian Cancer Genomics Community of Practice. This group comprises Canadian hereditary cancer providers (geneticists, genetic counsellors, oncologists), patients and researchers. A short presentation about the study was made to group members during a regular meeting in September 2021 and they were asked to share the study information through their networks. An email with the study's advert containing the link to the survey was also shared with the Canadian Association of Genetic Counsellors (CAGC) so that members could share the survey with potential participants.

After approval was obtained from group administrators, the ad and link to the survey were posted by the following Facebook and Instagram groups: the Jacqueline Rush Foundation, lynch syndrome spouses, breastcancersoc, coloncanada, *BRCA* Sisterhood Canada, *BRCA1* And *BRCA2* GENETIC BREAST CANCER AND OVARIAN GENE, *MSH2* Lynch Syndrome Support Group, Lynch syndrome, Lynch Syndrome Support Group/LSI, BCW in action and the SPOR NL Support Unit.

Survey development

Survey development occurred over several months with multiple iterations and team reviews. The survey was constructed from previously developed surveys and studies surrounding family communication and health information (e.g., Cella et al. 2002; DeMarco et al. 2004; Read et al. 2005; McAllister et al. 2011; Nycum et al. 2009) as well as team members' clinical and research design experiences. Items from the Psychological Adaptation to Genetic Information Scale (PAGIS) (Read et al. 2005) influenced survey items on support, certainty, and self-efficacy. Some items that explored the response of relatives to being informed of their risk of hereditary cancer were taken directly from the Multidimensional Impact of Cancer Risk Assessment (MICRA) Questionnaire (Cella et al. 2002). Other items were influenced by the Genetic Counselling Satisfaction Scale (GCSS) (DeMarco et al. 2004), the Perceived Personal Control (PPC) Questionnaire (McAllister et al. 2011) and elements from the ecological model (Nycum et al. 2009). Several content areas were explored, including attitudes and beliefs about risk information sharing, preference for different outreach methods, and sociodemographic characteristics.

Most survey items were measured on a 5-point Likert scale from Strongly Disagree to Strongly Agree, where higher scores indicated greater agreement with attitude and opinion items. Questions directed to the informers (i.e., the probands) ascertained how confident they felt in sharing risk information, how much they felt supported by their family members, their perceived responsibility to share risk information, whether or not they were worried about sharing the information, distress in relatives upon being informed, and how useful they felt the family letter was in facilitating communication. The relatives who were informed, but did not themselves share hereditary cancer risk information, were asked to share their level of satisfaction with being informed using the family-mediated approach. They were also asked about how much they felt in control based on their comprehension of the implications of the information, the management of their elevated risk and their level of distress.

In the survey, the term "informer" was used to refer to the probands and "informed" to refer to the relatives to eliminate any confusion among study participants about which group they belonged to. Before the survey was published, piloting was carried out with three patients known to the clinicians on the research team and in their circle of care, and representatives from Ovarian Cancer Canada known to the team. Minor changes were made to the survey following reviews. For example, one patient suggested an additional option for question 10.

"For Question number 10, could a response of Unsure / Can't recall be added. For example, I was informed regarding LS in 1992/1993. Certainly, some persons may not recall after so many years".

Upon reflection and team member feedback, 'prefer not to answer/don't know' was added as a response option to several other survey items as well. Other patients suggested the survey content areas were comprehensive, reflected the issues they faced in communicating with family members about inherited risk and had no additional suggestions.

On completion, the survey comprised a total of 38 questions, of which 35 questions were posed to the probands

(items 1–10, 11–27 and 31–38) and 21 questions were posed to the relatives (items 1–10, 28–30 and 31–38). The survey used skip logic to provide items relevant to probands (the 'informers') and their relatives (the 'informed').

All respondents were asked to share their preference for three different outreach methods:

i. Preference for the family-mediated approach (e.g., the use of the family letter provided by a genetics service),

ii. Preference for active contact taken by a health care provider with or without a follow-up email or phone call, and.

iii. Preference for communicating inherited cancer risk using a website or a mobile health application.

Potential variables related to outreach preferences were also measured and included demographic and clinical items such as self-reported prior experience of cancer, genetic testing result, age, sex, place of residence, etc. The final survey instrument is available in Supplementary file 1.

Data cleaning

Some of the categories for the nominal demographic variables, marital status, level of education and annual income, were aggregated in Table 1. For example, for marital status, married and living common-law were combined. Similarly for annual income, the $ 50, 000 to less than $ 90, 000 income range incorporated an income of $ 50, 000 to less than $ 60, 000, $ 60, 000 to less than $70, 000, $ 70, 000 to less than $ 80, 000 and $ 80, 000 to less than $ 90, 000. Provinces were grouped according to regions in the following manner: Prairie Provinces (Manitoba, Saskatchewan, Alberta), Western Provinces (British Colombia), Atlantic Provinces (Newfoundland, New Brunswick, Prince Edward Island and Nova Scotia) and Central Provinces (Ontario and Quebec).

The Likert scale categories agree strongly and agree somewhat; disagree strongly and disagree somewhat were combined for items on preferences for both probands and relatives. Similarly, items on emotional response and level of satisfaction with the family letter for the relatives were also combined.

Data analysis

SPSS Software 27.0 was used for data analysis. Descriptive statistics were reported for all survey items, including demographic and self-reported clinical, psychosocial and outcome items. The Mann-Whitney U test was used to assess differences between the informers who received a family letter and those who indicated they did not receive a family letter.

Univariate analyses examined the factors that influenced preference for family member outreach using the Mann-Whitney U test and the Kruskal Wallis test for variables with more than two categories. Ordinal variables were further analyzed using the Spearman rank correlation.

Results

A total of 119 survey responses were initiated, and of these, 108 eligible participants indicated the perspective from which they would respond to the survey questions (informer, $N=58$; informed, $N=50$). One hundred and three participants completed the items on cancer risk perception (informer, $N=53$; informed, $N=50$) (Table 2) and 102 participants completed the survey items on genetic test results (Table 3) and cancer diagnosis (informer, $N=52$; informed, $N=50$) (Table 4). Three informers who received a family letter did not complete survey items on experiences with hereditary cancer risk communication, communication challenges and outreach preferences. Two of them had HBOC and the other had LS. Similarly, three informers who did not receive a family letter did not share their experiences with risk communication, its challenges, or their preferences for outreach. Two of them had a LS mutation and were diagnosed with cancer and one had a *BRCA* mutation. Ultimately, 96 respondents (informer, $N=46$; informed, $N=50$) completed the surveys with variable response rates to different survey items. We do not know with how many people the survey link was ultimately shared, nor do we know how many people were aware of the survey but chose not to participate. As a result, the response rate cannot be determined Fig. 1

Demographic and clinical characteristics of study participants

There was a predominance of female participants (informers $N=44/46$, 96%, informed $N=45/48$, 94%). Most study participants were above the age of 45 (mean age of the informers $=56.89\pm16.033$, median $=59.5$; mean age of the informed $=48.21\pm11.362$, median $=45$) and married with children (Table 1).

Individuals were asked about their perceived cancer risk before seeing a genetic specialist (Table 2), their genetic test result (Table 3), and whether they were diagnosed with cancer (Table 4). Most respondents reported perceiving their cancer risk as moderate or high before being seen by genetics specialists, 68% of the informers and over 56% of the informed (Table 2).

Table 1 Demographic characteristics of survey respondents

Demographic characteristics		
	Proband $N=46$ (%)	Relatives $N=48$ (%)
Age		
34 and younger	3 (6.5)	4 (8.3)
35–44	5 (10.9)	18 (37.5)
45–54	10 (21.7)	14 (29.2)
55 and older	28 (60.9)	12(25)
Mean (SD)	56.89 (16.033)	48.21 (11.362)
Missing	0	0
Marital status		
Single- never married	1 (2.2)	4 (8.3)
Married/Living common-law	39 (84.8)	39 (81.3)
Divorced/Separated/Widowed	6 (13.0)	4 (8.3)
I prefer not to say	0	1 (2.1)
Missing	0	0
Number of children		
0	6 (13.0)	11 (22.9)
1–2	30 (65.2)	29 (60.4)
3 and more	10 (21.7)	8 (16.7)
Mean (SD)	2.09 (0.590)	1.93 (0.633)
Missing	0	0
Gender		
Male	2 (4.3)	2 (4)
Female	44 (95.7)	45 (94)
Non-binary/third gender	0	1 (2)
Missing	0	0
Highest educational level		
High school diploma or less	7 (15.2)	6 (12.8)
Trade or college diploma	15 (32.6)	19 (40.4)
University, undergraduate degree	12 (26.1)	11 (23.4)
University graduate degree	12 (26.1)	11 (23.4)
Missing	0	1
Annual household income		
$ 49, 000 or less	4 (8.9)	3 (6.3)
$ 50, 000 to less than $ 90, 000	9 (20.0)	11 (22.9)
$ 90, 000 to less than $ 150, 000	11 (24.4)	17 (35.4)
$ 150, 000 and over	12 (26.7)	10 (20.8)
I prefer not to say	9 (20.0)	7 (14.6)
Missing	1	0
Province or territory		
Prairie Provinces (MB, SK, AB)	6 (13.0)	8 (16.7)
Western Region (BC)	12 (26.1)	10 (20.8)
Atlantic Provinces (NL, NB, PE, NS)	8 (17.4)	17 (35.4)
Central Provinces (QC, ON)	20 (43.5)	13 (28.1)
Missing	0	0
Urban or rural dwelling		
Small population centre, with a population between 1,000 and 29,999	6 (13.3)	10 (21)
Medium population centre, with a population between 30,000 and 99,999	5 (11.1)	13 (27)
Large urban population centre, with a population of 100,000 or more	27 (60.0)	23 (48)
Rural area	7 (15.6)	2 (4)
I prefer not to say	0	0
Missing	1	0

Table 2 Perception of cancer risk among survey respondents

Perception of cancer risk		
	Informer/ Proband $N=53$ (51.5)	Relatives/ Informed $N=50$ (48.5)
Perceived cancer risk		
High	26 (49.0)	12 (24)
Moderate	10 (18.9)	16 (32)
Low	11 (20.8)	10 (20)
Unknown/insufficient information	6 (11.3)	9 (18)
I was not seen by a genetic specialist	-	3 (6)

Table 3 Genetic test results of the informers and the informed

Genetic test result		
	Informer/ Proband 52 (50.98)	Relatives/ Informed 50 (49.02)
BRCA1/2	29 (55.8)	38 (76)
A mutation that causes Lynch syndrome	19 (36.5)	7 (14)
I did not have genetic testing	-	3 (6)
Other genetic test results	2 (3.9)	2 (4)
A mutation other than *BRCA1* or *BRCA2* that causes hereditary breast and ovarian cancer (HBOC)	2 (3.9)	0

Table 4 The frequency of cancer diagnosis among the informers and the informed

Cancer diagnosis		
	Informer/Proband 52 (50.98)	Informed/ Relatives 50 (49.02)
Yes	31 (59.6)	9 (18)
No	21 (40.4)	41 (82)
Number of years since the first diagnosis	Mean = 2.65; SD = 0.915	Mean = 1.63; SD = 0.518
Cancer that was diagnosed		
	Informer/Proband $N=37$ (80.43)	Informed/ Relative $N=9$ (19.57)
Breast cancer	13 (35.1)	5 (55.6)
Ovarian cancer	5 (13.5)	0
Endometrial cancer	7 (18.9)	2 (22.2)
Colon cancer	9 (24.3)	2 (22.2)
Other	3 (8.1)	0

Ten informers (probands) reported being carriers of the *BRCA1* P/LP variant ($N=10/52$, 19.2%), including one who tested positive for both a *BRCA1* P/LP variant and a single *MUTYH* P/LP variant (Table 3). Nineteen informers carried a *BRCA2* P/LP variant, including one participant with two P/LP variants: *BRCA2* and *ATM* P/LP ($N=19/52$, 36.5%). One informer reported carrying three P/LP variants: *BRCA1*, *BRCA2*, *and RAD51C*. Two other informers reported having a gene variant besides *BRCA1/2* that caused HBOC ($N=2/52$, 3.9%) and 19 informers were carriers of P/LP variants that cause LS ($N=19/52$, 36.5%). 24% of the informed (relatives) reported carrying a *BRCA1* P/LP variant ($N=12/50$), 52% a *BRCA2* P/LP variant ($N=26/50$) and 14% reported carrying a P/LP variant that causes LS ($N=7/50$) (Table 3). Other reported genetic P/LP variants among the informed were the *PMS2* P/LP variant and the *RAD51C* P/LP variant.

Thirty-one informers (probands) reported being diagnosed with cancer ($N=31/52$, 59.6%), while twenty-one informers indicated they were never diagnosed with cancer ($N=21/52$, 40.4%) (Table 4). There were two informers with a diagnosis of both endometrial and colon cancer. One informer was diagnosed with three primary cancers: endometrial, colon and thyroid. Cancers categorized as 'other' were thyroid cancer ($n=1$), adrenal cancer ($n=1$) and gastric cancer ($n=1$) (Table 4). The frequency of cancer diagnosed among the probands was $N=37$ since three informers had more than one cancer diagnosis. Nine of the informed (relatives) reported a previous or current cancer diagnosis ($N=9/50$, 18%) (Table 4). Hereditary breast cancer - core cancer type consistent with cancer predisposition syndrome was most frequently diagnosed in this group ($N=5/9$, 55.6%). Among the informed, other reported cancer diagnoses were colon cancer and endometrial cancer.

Experience with risk communication

Informers

The informers were asked if they received a family letter to share with their relatives to facilitate risk communication. Twenty-eight informers (53.8%) reported receiving a family letter, eighteen (34.6%) reported that they did not, while six (11.5%) were unable to recall. Some informers who selected "no" or "I do not recall" to this question explained that they were given an ancestry form, informational material, testing protocol information, or their own genetic test result. Of the six informers who were unable to recall being given a family letter, two were later counted among those who received a letter since they went on to complete the survey from that perspective.

Informed

Relatives informed of their risk of hereditary cancer by a proband were asked to give their opinion on their level of satisfaction with the family-mediated approach and share their emotional responses. Most of the informed (relatives) found the family letter helpful ($n=25/32$, 78%) and understood the cause of their own risk ($n=26/33$, 79%) and their eligibility for genetic counselling ($n=33/37$, 89%). They also felt nervous ($n=42/44$, 96%) or concerned ($n=40/44$,

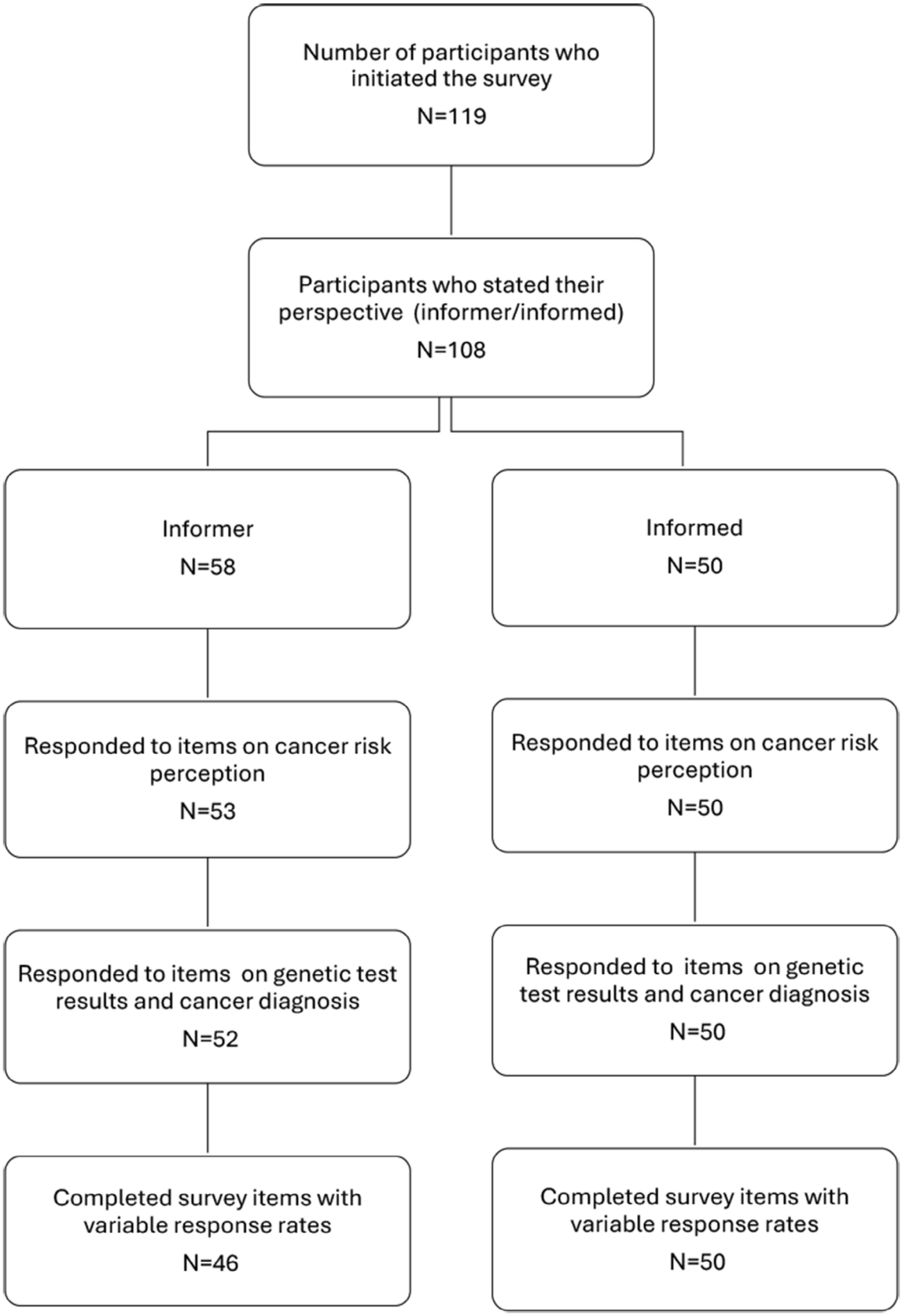

Fig. 1 Flow diagram illustrating the number of study participants

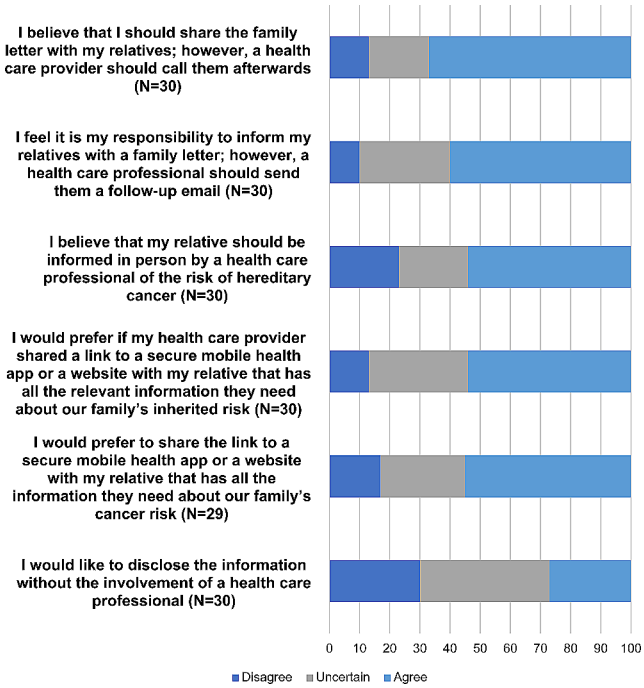

Fig. 2 The preference for sharing hereditary cancer risk information among the informers (probands) who received a family letter

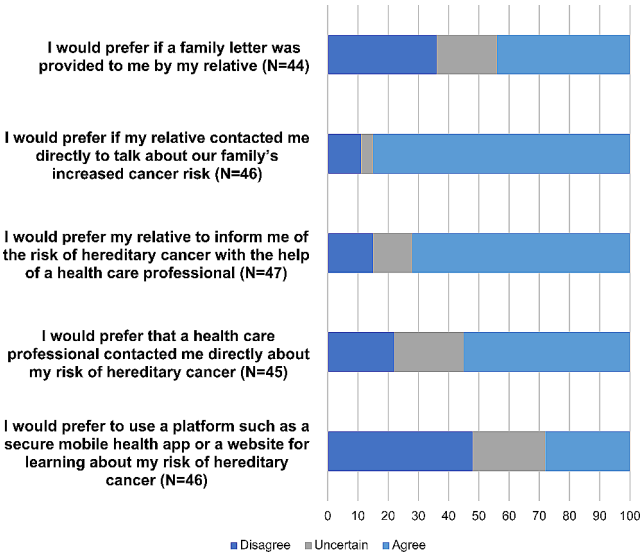

Fig. 3 The preference for receiving hereditary cancer risk information among the informed

91%) upon receiving information about their personal risk. Nevertheless, they were happy that risk information was shared with them ($n=40/45$, 89%).

Preference for hereditary risk communication from the probands' perspective

Informers who were given a family letter agreed they should be involved in communicating hereditary cancer risk information to their relatives, but many would value the support from a health care provider. 67% ($n=20$) of informers agreed that the health care provider should follow up with their relatives via a phone call (Fig. 2). Similarly, 60% ($n=18$) of the informers thought that the health care provider should follow up with their relatives with an email. 53% ($n=16$) of the informers thought that their relatives should be informed in person by a health care provider about their hereditary cancer risk. Even those informers who were not given a family letter similarly agreed to these statements with percentages of 55–57%. Thus, while informers agreed they had a role to play in communicating risk information to relatives, clearly, there were high levels of agreement with a role for health care providers in this process.

The idea of the health care provider directly sharing the link to a website or a mobile health app with relatives that had additional information was somewhat supported by the informers who received a family letter (53%, $n=16$) (Fig. 2) while less than half of the informers who were not given a family letter agreed to this statement. 55% ($n=16$) of the informers who were given a family letter thought they should share the link themselves and only 27% ($n=8$) of them thought their relatives should be informed without the assistance of a health care professional. The informers who were not given a family letter shared similar views on these two items.

The informers were asked to share any other suggestions for informing their relatives. Very few suggestions were left in open comments. One informer shared that they informed family members via text messaging and asked another family member to spread the word to distant relatives via Facebook.

Preference for hereditary risk communication from the perspective of the informed (relative)

The informed (relatives) were also asked to share their opinions on how they would prefer to be informed of their hereditary cancer risk. Again, a high percentage of relatives preferred to be informed with the help of a health care professional (72%, $n=34$), even though the majority, 85% ($n=39$) thought that the informer (proband) should contact them directly (Fig. 3). 56% ($n=25$) of them thought that a health care professional should contact them directly. On the other hand, only 28% ($n=13$) agreed with using a mobile health app or website (Fig. 3), which is less than what was observed for the informers (55% and 38% for informers who received a family letter and those who did not, respectively).

Factors that potentially influence preference for risk communication

Informers (probands)

Informers with no cancer history ($n=19$) were significantly more likely to agree that they should share the family letter with their relatives with a follow-up phone call from the health care professional ($p=0.031$) compared to those with a personal history of cancer ($n=28$). Informers who were never diagnosed with cancer ($n=19$) also thought it would be appropriate for the health care professional to send their relatives a follow-up email after the family letter was shared, $p=0.030$, compared to those with a cancer diagnosis ($n=28$).

The demographic variables, income, marital status, and age were associated with the endorsement of the health care provider in sharing hereditary risk communication with relatives. Informers who earned $49,000 or less were significantly more likely to agree that they should share hereditary cancer risk information with their relatives and that a health care professional should follow-up with a phone call ($p=0.025$), compared to those with an annual income of $50,000 to less than $90,000.

Similarly, informers with an annual income of $49,000 or less ($n=4$) were more likely to agree that the health care professional should send their relatives a follow-up email ($p=0.044$) compared to those whose annual income was $50,000 to less than $90, 000 ($n=9$). Marital status was also related to email outreach by a health care provider. Informers who were divorced/separated/widowed ($n=6$) were more likely to agree with this outreach method ($p=0.032$) than respondents who were married/living common-law ($n=39$).

Age was significantly related to the preference for relatives to receive a phone call from a health care professional informing them of the risk of hereditary cancer ($p=0.040$). There was a trend for informers 34 and younger ($n=3$) to be more likely to prefer this method when compared to those who were 55 and older ($n=28$), though the pairwise comparison between the two groups was not significant.

Informers living in a small population centre ($n=6$) were more likely to agree with in-person communication from a health care provider ($p=0.032$) than those from a larger population centre ($n=5$).

Informed (relatives)

Agreement with the item suggesting the proband share hereditary cancer risk information with the help of the health care professional was supported by relatives with a LS P/LP variant ($n=7$) significantly more ($p=0.035$) than relatives with a *BRCA1* P/LP variant ($n=12$). There was no association between this item and carriers of the *BRCA2* P/LP variant.

Relatives with a university graduate degree ($n=10$) and those with a trade or college diploma ($n=19$) significantly preferred that their relatives contact them directly about the family's increased cancer risk ($p=0.005$) when compared to respondents with a high school diploma or less ($n=5$). Annual income was also significantly associated with direct contact from probands without a health care provider's involvement ($p=0.025$). Relatives with an income of $50,000 to less than $90,000 were more likely to endorse this statement ($n=11$) than those earning $49,000 or less ($n=3$).

Relatives residing in an Atlantic province (NL, NB, PE, NS) ($n=17$) preferred the health care professional's involvement when being informed of their hereditary cancer risk. They were significantly more likely to prefer being informed of the risk of hereditary cancer with the help of the health care professional ($p=0.035$) than those living in Prairie provinces (AB, SK, MB) ($n=8$). Similarly, they preferred that the health care professional contact them directly ($p=0.038$) ($n=16$) compared to those from the Prairie provinces ($n=8$).

Relatives living in a rural area ($n=2$) were also more likely to support being informed of their risk of hereditary cancer with the help of a health care professional ($p=0.024$) and this method of outreach was least supported by relatives who lived in a large urban population centre ($n=23$).

Among the informed/relatives, there were no significant associations among clinical and demographic characteristics for receiving the family letter from the proband (the informer) or the preference for the use of a platform such as a secure mobile health app or a website for learning about the risk of hereditary cancer.

Discussion

This national cross-sectional study explored the preferences for CPS risk communication within families affected by HBOC and LS. This study demonstrates that patients and their families report variability in the preferred approach for contacting relatives about genetic results.

Study results could be useful to health care providers in supporting probands to effectively communicate risk information within their families. Having a better understanding of how affected families would like to be advised of their cancer risks could potentially lead to an increase in the percentage of relatives who attend genetic counselling from the 50% or less reported in the literature (Loader et al. 2002; Lowery et al. 2010; Hinchcliff et al. 2019; Marleen et

al. 2019; Menko et al. 2013). By increasing the completion of genetic counselling visits, a greater proportion of at-risk individuals may benefit from preventative risk management.

First and notably, one-third of the probands reported not receiving a family letter despite this being the standard of care in Canada. It is unknown whether no letter was actually provided or if this finding is the result of a simple recall issue. It is noteworthy - if probands did not receive a letter or forgot they were given one, this has implications for whether it can be shared with relatives and supports the exploration of other methods of family member outreach.

The results showed that there was generally a high acceptance among the probands for participating in risk communication. Nonetheless, there remained high agreement with items measuring opinions on assistance from a health care professional, similar to other studies (Andersson et al. 2020; Henrikson et al. 2021). There was little acceptance for using web-based approaches such as mobile health apps and websites for sharing hereditary cancer risk information. These findings suggest probands require assistance with family communication outside the scope of telegenetics and devoted discussion of this issue during counseling sessions would likely be appreciated by patients.

Despite few studies exploring the effects of a cancer diagnosis on risk communication within affected families, this factor appears to be a facilitator rather than a barrier (Taber et al. 2014). The results suggest that individuals without a cancer diagnosis desire the health care professional's involvement after first contacting their relatives about their inherited risk. Probands unaffected by cancer are more likely to want the health care professional to give their relative a follow-up phone call or send them a follow-up email. The findings imply that the family-mediated approach limits risk communication among probands with no cancer diagnosis. This finding highlights a potential subgroup of patients who prefer additional support with communication from health care professionals.

Probands affected by LS were more likely to indicate they wished for the assistance of a health care professional than probands affected by the *BRCA1* P/LP variant. The management of LS is more complex since several pathogenic variants are implicated, the list of malignancy-related risks is more extensive, and specific surveillance recommendations are nuanced per gene (Cohen et al. 2014; Rosenblum et al. 2020). Therefore, probands may be more likely to need a health care professional's assistance in communicating risk information on LS, given its complexity and the variable penetrance and expressivity. This finding could have direct implications for practice as it highlights a subset of patients with LS who may benefit from additional communication support tools.

Demographic variables such as age and marital status influenced the preference for hereditary cancer risk communication among probands. Other demographic variables such as annual income, educational level, population settlement and regional location within Canada significantly influenced communication preferences among both the proband and relatives. Younger informers were more likely to indicate wanting a health care provider to contact their relatives directly than older informers. This could be because older individuals have more experience with communication and a better understanding of the disease by virtue of having lived with it longer. Interestingly, there was no association between informers of a younger age group and the preference for using an electronic platform for risk communication. Individuals who are married/living common law may not prefer an approach that does not allow for face-to-face contact, such as having the health care professional send their relatives an email. On the other hand, probands who are divorced/separated/widowed may be in agreement with an indirect form of risk communication. Coupling or marriage provides for more encouragement and social support. These associations are in keeping with findings from studies which outlined the importance of family dynamics in risk communication (Armstrong et al. 2005; Miller et al. 2005; Peters et al. 2005; Wang et al. 2005).

There seems to be a higher acceptance of a direct contact approach for genetic counselling among low-income relatives and probands, while those with a higher annual income were less accepting of the involvement of the health care professional. Similarly, relatives with an advanced education also preferred that the proband contact them directly without the health care professional's involvement, perhaps indicating they may be better able to understand risk information and perhaps perceive they do not need additional support. In general, individuals of a lower socioeconomic status are at greater risk of suffering poorer health outcomes and have fewer resources (Leonard et al. 2016) and might benefit greatly from the involvement of the health care provider.

Both probands and relatives from rural and small populations settings preferred the involvement of the health care professional. Little information was found in the literature about how implicated individuals from a rural or urban setting would like to be informed about the risk of hereditary cancer. It has been shown, however, that decreased rates of genetic testing in rural areas result from not having a nearby clinic and decreased access to genetic counsellors (Fogleman et al. 2019). Furthermore, within different jurisdictions, there are variations in the family-mediated approach. Relatives from the Atlantic provinces preferred that the health care professional contact them directly or that the proband inform them with the help of the health care professional. On the other hand, relatives from the Prairie provinces

disagreed. The literature shows that there are approximately nine genetic counsellors in Newfoundland (NL), 21 in Nova Scotia (NS), 1 in Prince Edward Island (PEI) and 0 in New Brunswick (NB), compared to 45 in Alberta (AB), 8 in Saskatchewan (SK) and 14 in Manitoba (MB) (Lambert et al. 2021). The desire for the health care professional's involvement in hereditary risk communication by relatives from the Atlantic regions could be due to fewer regulated specialists in these provinces and the need for an improvement in this area. In addition, there is a shortage of genetic specialists with an increasing demand for their services in North America and generally worldwide (Etchegary et al. 2021; Haga et al. 2013). Again, this highlights the shortage of genetic counsellors more broadly and the benefit of exploring alternative models for facilitating risk communication in families affected by cancer predisposition syndromes so that an increased number of at-risk relatives may avail themselves of life-saving treatment modalities if they so choose.

In summary, the observed trends in family communications preferences showed that there were varying degrees of acceptance for the involvement of the health care professionals through follow-up emails and phone calls once the proband made initial contact with their relatives. The study showed that participants had little threshold for using web-based platforms such as a website or mobile health apps during risk communication, indicating these alternatives to in-person counselling will not be acceptable to all. The findings highlight the need to explore alternative models for facilitating risk communication in implicated families. In an era of a "menu of options," a more practical approach might be to allow all options to be available to individuals so they may tailor their communication preferences.

Findings are limited by the relatively small sample size and the predominance of female participants. We suspect the COVID-19 burden contributed to the low response rate, both in terms of provider ability and time to help recruit eligible patients and respondent interest in research during pandemic concerns and lockdowns. This study used the Likert scale for responses on preferences without allowing for open-ended responses which might have given a better understanding of how affected individuals felt about the topic. Based on these limitations, the results might not be generalizable to all patients affected by HBOC and LS. Another limitation was that twelve probands who stated the perspective from which they would respond to the survey items did not share their input on outreach preferences. Six of them however completed different survey items of background information such as cancer risk perception, genetic test results and cancer diagnosis. The exact reason for this is unknown, however, we were able to capture information on communication preferences from an almost equal number of probands and relatives.

In addition, there was the potential for familial clustering in survey responses; however, whether the participants were formally related could not be measured. Individuals who participated in the survey are those most likely to seek health information or be most interested in discussing hereditary risk. This, therefore, means that little feedback would have been obtained from individuals who are among the 50% of relatives who fail to attend genetic counselling. Such an assumption could also explain why only two male relatives participated in the survey since studies have shown that men may be less aware of their inherited cancer risks or have less interest in risk information than women (Rauscher et al. 2018; Daly et al. 2016; Suttman et al. 2018).

Despite these limitations, this study contributes to the body of literature that focuses on how individuals affected by cancer predisposition syndrome would like to inform their relatives of the risk of hereditary cancer and how relatives would like to be informed. Few investigations have explored specific communication preferences of probands, and even fewer still explored how relatives prefer to receive risk information. New knowledge about specific outreach preferences of both probands and relatives has been generated by this work and should assist providers in their efforts to support families communicate about inherited cancer risk. Ultimately, it is hoped that effective risk communication can help increase the number of at-risk relatives who undergo genetic testing and subsequent life-saving risk management.

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Data availability No datasets were generated or analysed during the current study.

Declarations

Ethical approval Ethics approval was granted from the provincial Health Research Ethics Board in St. John's, NL, Canada. (Ref # 2021.154). All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all participants included in the study.

Consent to participate Survey consent was implied if respondents submitted a survey.

Consent for publication Not applicable.

Competing interests The authors declare no competing interests.

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