Seminars in Oncology Nursing 40 (2024) 151624

Contents lists available at ScienceDirect

Seminars in Oncology Nursing

journal homepage: https://www.journals.elsevier.com/seminars-in-oncology-nursing

Hereditary Cancer Syndrome Carriers: Feeling Left in the Corner

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ARTICLE INFO

Key Words:
Hereditary cancer syndromes
Cancer nurses
Hereditary breast and ovarian cancer
Lynch syndrome
Patient experiences
Hereditary cancer

ABSTRACT

Objectives: There is limited evidence on health promotion interventions in people with hereditary cancer syndromes or on their main sources of support and information. This study aimed to understand these patients' experiences and needs, including their information needs, their views on prevention and mental health, and the support they want from nurses.

Methods: This qualitative study included 22 people (8 previvors and 14 survivors) with hereditary breast and ovarian syndrome or Lynch syndrome from 10 European countries. Participants underwent individual semi-structured interviews, which were recorded and transcribed for reflexive thematic analysis. The patient and public involvement panel provided input on study design and thematic analysis.

Results: Patient experiences were similar regardless of the country and access to testing and screening. Participants reported receiving little information on the importance of health behaviors for cancer risk and expressed their wish to be followed by cancer professionals. They felt compelled to seek support and information from the internet and patient groups. The main themes identified were: (unmet) informational and support needs, seeing life in a different way, and limitations of health care providers.

Conclusions: People with hereditary cancer syndromes need professionals to be involved in their long-term management and to provide reliable information. As genomics are increasingly integrated in oncology, the need for professionals to support these populations will increase.

Implications for Nursing Practice: Nurses are crucial for promoting self-management and advocating for patient decision-making; however, they need skills and knowledge to do so. There is a need for nurses to get more involved in understanding hereditary cancer syndromes and an opportunity to take the lead in the care of these people.

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People with hereditary cancer syndromes (HCSs) have a high risk of developing certain cancers in their lifetime due to a pathogenic gene mutation that confers an increased susceptibility to cancer.[1] The most common HCSs are hereditary nonpolyposis colorectal cancer, also called Lynch syndrome, which entails a higher risk of mainly colon, endometrial, gastric, and small intestine cancer; followed by hereditary breast and ovarian cancer (HBOC), which primarily increases the risk of breast, ovarian and prostate cancer.[2] Even though HCSs are not common, they are linked to around 10% of cancers.[3] Diagnosis is essential in order to plan for long-term follow-up, as HCSs affect entire families, entailing a 50% chance of passing on mutated genes to offspring and a high risk of early onset cancer.[2]

Genetic testing is gradually being implemented in clinical practice, as early diagnosis of these syndromes allows for preventive controls to decrease risk and detect cancer early.[4,5] Interventions include behavioral counselling, surveillance, prophylactic medication, surgeries, and possible modifications in the management and treatment of any cancer that develops.[6] People with HCSs thus require comprehensive, personalized care for them and their families, which poses challenges for the health care system and the person affected alike. HCS carriers have to understand their diagnosis and its implications, get recommendations about preventive controls, be prepared for complex and personal decisions (eg, regarding preventive surgery), and usually take responsibility for disclosing the results to the rest of their family.[7,8] Numerous studies have looked at the psychological impact of HCS diagnosis and related decision-making, which is associated with uncertainty, doubts, and distress.[9-13] These patients have often reported a lack of follow-up and poorly integrated care; they describe being attended by health care professionals with little knowledge of HCSs, leaving them feeling lonely, with nobody to turn to.[10]

The implications of communications with family and the feelings of guilt associated with this process have also been explored.[10,14] People

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https://doi.org/10.1016/j.soncn.2024.151624
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Layperson Summary

What we investigated and why

There are very little studies looking at how actions to improve lifestyle affect people with genetic alterations that increase their risk of having cancer. There is also very little information on where they find information and support. The aim of this study was to understand the needs of people with these genetic alterations and how nurses can help.

How we did our research

We did interviews with 22 people with these genetic alterations (8 never had cancer and 14 had cancer in the past) from 10 different European country. We asked them to have an interview that were recorded and analyzed. Some people with these genetic alterations helped to plan the study, the questions and how we could analyze them.

What we have found

People have similar experiences and problems no matter where they came from. They want more information on lifestyles and they also want to have cancer professionals to follow them up. They also told us they look for information in internet and patients' groups. Our main findings were: they want to be understood, they see life in a different way and they want more from health care professionals.

What it means

After looking at the evidence is important to suggest having professionals that know about genetic alterations to answer their questions and worries. Nurses are probably the best professionals to do it but they need to know more about it as this need is only going to grow.

with an HCS are urged to notify at-risk relatives, so it is left to those with the syndrome to disclose the risks and the implications to family members, often without the guidance of health care professionals.[6]

From the health system perspective, guidelines for the standard of care in HCS carriers, for example, those of the National Comprehensive Cancer Network (NCCN) or the European Society of Medical Oncology, give advice on testing criteria, risk reduction strategies (including screening and surveillance), and reproductive risks.[15-18] However, they rarely address the personal needs or actions of people affected by HCSs, such as psychosocial support or lifestyle behaviors. Guidelines for genetic counselling do acknowledge the need for multidisciplinary team work to cover the personal and social needs and provide resources to HCS carriers.[6,19] The increasing role of genetics in the cancer management poses a challenge to professionals, who may lack knowledge, skills, or confidence when providing care for people with HCSs.[20,21] Interest in educating health care professionals is growing, but this training rarely encompasses comprehensive care.

The delivery of comprehensive care can directly affect self-management and decision-making, including adherence to recommendations, management of appointments, and decision-making about risk reduction actions.[22,23] Self-management is directly affected by the person's perception of their own risk and the extent to which they believe risk reduction actions will work. Adherence to risk management recommendations is therefore not only dependent on the information that health care professionals may provide at HCS diagnosis, but the support and comprehensive care that addresses their risk perception, personal health beliefs, and the psychological effects these have.[24-26] Different studies have explored lifestyle behaviors, behavioral modification interventions,[27,28] risk perception, and psychological impacts in HCS carriers,[8,10-12] but the relationship between mental health and self-management has not been explored and is not part of the recommendations and education currently offered in oncogenetics.

The aim of this study was to explore the experience of HCS carriers (both those who have not had cancer, also known as previvors, and those who have had a cancer diagnosis) and their priorities and unmet needs regarding self-management and behavioral counselling during follow-up. A patient and public involvement (PPI) panel collaborated during the project to ensure that the study design was worthwhile for HCS carriers.[29]

Methods

This qualitative study was based on one-on-one virtual interviews with people affected by HCSs in Europe. Semi-structured interviews allow the interviewer to guide the participant to areas of interest but ensure that the flow of the interview is directed by the participant.[30] The consolidated criteria for reporting qualitative research[31] were used to report this study (Appendix 1).

Recruitment

Recruitment was done through posts on social media inviting eligible participants to contact the principal investigator (PI). Posts were promoted in patient support groups and by PPI panel members and were shared by carriers and hereditary cancer professionals via Twitter, Facebook, and LinkedIn. Inclusion criteria were: diagnosis of an HCS (Lynch syndrome or HBOC), regardless of whether any cancer had been diagnosed, residence in a European country, and being able to speak Spanish or English. People who were undergoing cancer treatment at the time of the study, or who were not confirmed carriers of an HCS, were excluded. We used purposive sampling to recruit a demographically and geographically heterogeneous European sample,[32] as data saturation was not considered the appropriate way to determine the sample size.[33] Instead, the final sample was determined by the quality of data.

Interested individuals were sent information about the study, a participant information sheet, and a consent form. Those who replied that they were willing to be part of the study were contacted to set a date for their interviews. The PI conducted the interviews using the university Zoom account. The investigators did not know any of the participants prior to the study. All participants understood the aim of the study, gave written informed consent to take part in the interviews, and agreed to be recorded during the interviews (audio and video). They were informed that only partial quotes from the interviews were going to be used under a pseudonym, and only for study purposes. The university ethics committee approved the study (IRB00003099).

Data Collection

Interviews were conducted from January to June 2022 and lasted 40 to 70 minutes. Audio recordings were transcribed by the PI, using Sonix.ai to facilitate transcription. Field notes, comments, and observations were added to the transcription by the PI.

The interview questions were open and covered questions about different domains: living with high risk of cancer and understanding how participants perceived and managed their risk in terms of lifestyle, mental health, and communication with health care professionals (Table 1). The interview guide was underpinned by a variety of theoretical perspectives informed by nursing and psychology theory. The research team considered the role of mental health in self-care to be important, along with self-care tools in patients at risk, especially in oncology. We also considered that nurses are well positioned to

TABLE 1
Interview Guide with Main Themes

Theme	Topics
Diagnosis: first contact	• When and how were you given information? Who gave it, and what information was given?
Living at risk of cancer	• Needs of yours that haven't been met since your Lynch/BRCA syndrome diagnosis • Issues prioritized in your life
Information	• What information are you missing? • Where do you get and look for the information? • What kind of information you are looking for?
Prevention	• What aspects of your life do you consider important in the risk of cancer? • What things you would like to do to prevent or decrease your risk of cancer?
Mental health	• Has there been any change in your thoughts and mental well-being as a result of the diagnosis? What emotions does it arouse in you? • How important do you think the mental/psychosocial part of the information/diagnosis and follow-up is?
Educational program	• How do you think health professionals could better meet the needs of a person with a hereditary cancer syndrome? • What do you think health professionals should know and prioritize to meet the needs of a person with a hereditary cancer syndrome?

make an impact by promoting mental health, in turn improving self-management. Orem[26] emphasizes the importance of self-care in patients at risk of cancer, Lluch[25] focuses on emotional tools as facilitators of self-care, and the Health Belief Model[24] considers the importance of personal beliefs and knowledge in their decision-making. The interview guide was developed and discussed with expert patients and was piloted by two people from the PPI panel.

Data Analysis

Two researchers independently performed reflexive thematic content analysis on the verbatim transcript, following the six-phase process.[34,35] We chose this method to identify patterns in the data and to involve research participants in the analysis. Firstly, the three authors (a nurse with experience in cancer genetic counselling and working as a research assistant, an experienced researcher working in a cancer hospital, and a professor in nursing and mental health) discussed the patterns found after independent analysis of the interviews, acknowledging their experience and bias when interpreting the transcripts.[34] The team discussed and defined the emerging themes based on the analysis and codes. This method of analysis allows the data to lead the formulation of themes in a collaborative and reflexive way that enriches the interpretation.[34,35] The themes and quotes selected were shared back with some of the participants, refined, and finalized as presented here.

All participants were offered the opportunity to review the themes developed following analysis and three participants agreed. Some expressed concern that they could be recognized based on the ID and the country, so participants' characteristics do not include the participant ID.

Results

The final sample comprised 22 individuals from 10 European countries: Denmark (n = 1, 5%), Germany (n = 3, 14%), Ireland (n = 4, 18%), the Netherlands (n = 1, 5%), Portugal (n = 2, 9%), Poland (n = 1, 5%), Slovenia (n = 2, 9%), Spain (n = 3, 14%), Turkey (n = 1, 5%), and the United Kingdom (n = 4, 18%). There were 14 people (64%) affected by HBOC (10 previously diagnosed with cancer and 4 previvors), and 8 (36%) affected by Lynch syndrome (5 previously diagnosed with cancer and 2 previvors). Seven participants were men (32%), 19 (86%) had a family history of cancer, and 15 (68%) had a personal history of cancer. Among those diagnosed with cancer, four (27%) knew about the HCS prior to developing a tumor. Participants were diagnosed with their HCSs between 2000 and 2021. Their characteristics are displayed in Table 2.

Three main themes were developed: (1) (unmet) informational and support needs, (2) seeing life in a different way, and (3)

TABLE 2
Participant Characteristics

Code	Sex	Family history	Personal history	Discovered pre- or postcancer	Cancer	Year DX mutation	Preventive measures	Follow-ups	Behavioral changes
B1	F	Yes	Yes	Post	Breast	2021	Tamoxifen, Mast, BSO	BT, USS	No
L1	M	Yes	Yes	Pre	Bowel	2010	Aspirin	OGD, COL, dermatologist	Yes
B2	M	Yes	Yes	Post	Breast, prostate	2011	Tamoxifen	PSA	No
L2	F	No	Yes	Post	Bowel, breast, pancreas, melanoma, endometrium	2020	Aspirin, tamoxifen, BSO	COL	No
B3	F	Yes	No	N/A	N/A	2019	No	MRI	No
L3	F	Yes	Yes	Post	Endometrial, bowel, breast	1996	No	COL	No
B4	F	Yes	Yes	Post	Ovary	2015	No	MMG, MRI	No
L4	F	Yes	No	N/A	N/A	2017	Partial excision of colon	COL, OGD dermatologist	Yes
B5	F	Yes	No	N/A	N/A	2008	Mast, TAH, BSO	No	Yes
L5	F	Yes	Yes	Post	Bowel	2011	No	COL	Yes
B6	F	Yes	Yes	Post	Breast	2012	Double mast	MRI, USS, BT	Yes
L6	M	Yes	No	N/A	N/A	2019	No	COL	No
B7	F	Yes	Yes	Post	Breast	2013	No	MRI and USS	Yes
L7	M	No	Yes	Post	Bowel	2012	No	COL	No
B8	F	Yes	Yes	Post	Ovary	2012	No	MRI and USS	No
L8	M	Yes	No	N/A	N/A	2018	No	COL	Yes
B9	M	Yes	No	N/A	N/A	2019	Mast, BSO	No	No
B10	F	Yes	No	N/A	N/A	2011	No	Yes	No
B11	M	No	Yes	Pre	Breast	2014	No	PSA	Yes
B12	F	Yes	Yes	Pre	Breast	2019	No	MRI and USS	Yes
B13	F	Yes	Yes	Pre	Breast	2013	Mast	Bex, USS	No
B14	F	Yes	Yes	Post	Breast	2000	Mast, TAH, BSO	No	No

Bex, breast examination; BSO, bilateral salpingo-oophorectomy; BT, blood test; COL, colonoscopy; Mast, mastectomy; MMG, mammography; MRI, magnetic resonance imaging; N/A, not applicable; OGD, gastroscopy; PSA, prostate-specific antigen; TAH, total abdominal hysterectomy; USS, ultrasound.

limitations of health care providers. Each theme is further divided into various subthemes (Fig.), as described below with quotes (L identifies a person with Lynch syndrome and B identifies a person with HBOC).

Theme 1: Unmet Informational and Support Needs

This theme represents the experiences of HCS carriers related to accessing information and support from health care professionals or others. Respondents perceived deficiencies in follow-up care and felt frustration around the need for psychosocial support and information about the implications of the risks and risk reduction techniques. Participants described how they went about looking for information (usually on the internet) and their feeling of relief upon finding others with similar experiences.

Subtheme 1: Finding the "Right" Information

Participants felt that while the information they received during the diagnosis of their HCS was good and generally well supported by a geneticist or genetic counsellor, their real information needs started after they had known/accepted the results. This meant that when they had questions they did not know where to go.

> I've got to say, whilst the counselling was very good, I don't think I totally took it in and realised the implications. Then the questions started but no one was there anymore. **B2**

People living with Lynch syndrome generally felt more distress, as they found a complete lack of information outside the field of genetics.

> [talking about if they got information] No, not a thing. I went to the library. I couldn't find anything.**L3**

Subtheme 2: A Cancer Patient, But Not a Cancer Patient

There was a difference in follow-up between those who had cancer prior to a diagnosis of HCS and those who had never had cancer, as the latter felt like they had the risks, follow-up, and fear but could not access the same services cancer patients could.

> I have a follow-up with my oncologist every six months, but that's not because I've got Lynch, that's almost like a by-product . . . I think I am very lucky to have all of these professionals that follow me up. **L2 (Personal history of cancer)**

> I'm not what's classified as a healthy person. I'm classified as a cancer patient, and I will be for the rest of my life, but I cannot access specific care for cancer because I haven't had an active invasive cancer yet**. L4 (previvor)**

Subtheme 3: Understanding the Dimensions of Risk

Regardless of their syndrome or cancer status, participants all agreed that they did not receive any information on their risks of developing cancers other than the ones primarily affected by their HCSs. Many were not even sure what risks of other cancers they had or if there was any follow-up for these.

> I didn't even realise at the time that I've got an increased chance of getting prostate cancer. **B2**

> The problem I see with Lynch is that it can affect all different parts of your body, so there's no one person to go to. I don't have a Lynch consultant to control all my cancer risks. **L6**

Those who had risk-reducing surgeries perceived a lack of subsequent follow-up and support. A couple of participants explained their fear of not having anyone to go to as they had no further follow-ups, and another participant commented on the risks of other cancers.

> I feel like nobody addresses any of the other cancers. It's like, oh, no, it's not going to happen to you. Don't waste our time with this. And you are kind of left like, well, what do I do? What do I look out for? And they're like, No, you're fine. You've got this surgery done and you have nothing else to worry about. **B9**

They also felt as if health care professionals guided them to the follow-ups and risk-reducing surgeries without fully explaining all the implications.

> They [healthcare professionals] need to prepare the patient for the consequences. Not only what this mutation brings. But also, what are the consequences if you remove your breasts? If you remove your ovaries? If you remove your tubes? How hard are the surgeries? I knew nothing about that. **B13**

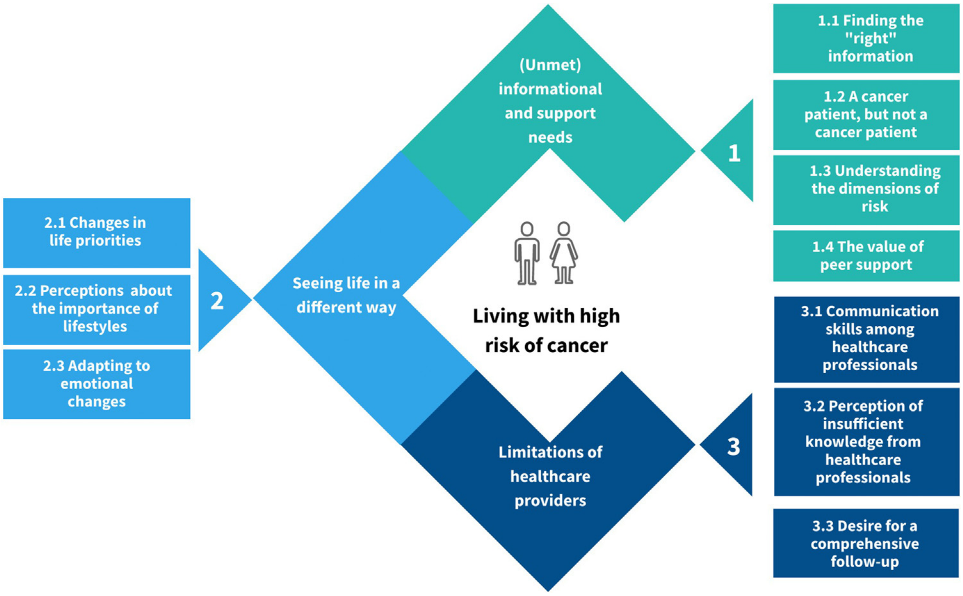

FIG. Themes and subthemes.

> *There is another part they do not inform you, the effects of the surgery on you.* **B1**

Subtheme 4: The Value of Peer Support

Participants found support and understanding in patient support groups. This made them feel more confident about the reliability of the information they were reading, as they felt that knowledgeable people were responsible for curating it. But more than anything, they went to groups to feel understood and to find others with similar worries.

> They have a huge amount of support, so I learn a lot from there. **L6**

> So, I thought, I have to talk to somebody, how they deal with what are they doing, what they are thinking about, how are they dealing with the risk, knowing that they could be sick many times and again and again, that was really hard for me. **B14**

There was a difference between people who took an active role in patient support groups and those who were more passive and felt they were just consumers of information. Those in active roles were more critical of where the information they were seeking came from and felt they could go to other health care professionals (as they built connections via social media) to resolve their doubts. They also felt very helpful when giving others the support that they had trouble finding themselves.

> I found a lot of comfort going into the role of like peer support . . . to give what I did not get. **B9**

At the same time, participants with a more active role in patient supports group felt the responsibility and challenge of supporting others and at times felt they could not share their own burdens and concerns.

> When I go to Facebook groups and in my family, I'm the support, they assume I know what I am talking about. I am not afraid of it (BRCA), but I can't ask them questions, as I am the one that needs to know. **B8**

Theme 2: Seeing Life in a Different Way

The lives of people with HCSs change in a personal and emotional way. Participants had to adjust to the diagnosis, faced decisions on risk reduction, and pondered whether they were healthy and how to be healthier. Throughout the interviews, they also discussed the emotional challenges of living with risk, both for themselves and for their family members.

Subtheme 1: Changes in Life Priorities

Many participants mentioned changes in their perspective about life after being diagnosed. For previvors, this meant a new adjustment to life, with some feeling it did not change their life for the better or give them a more positive perspective and others experiencing it as an opportunity to value life more.

> I do see things differently, probably especially in the last, probably the last year. **L6**

> I try and have more family time, enjoy life a bit more and stress less. I guess it is similar to what would happen with any other major impact on your health. **B12**

For survivors, the diagnosis did not involve the same adjustment, as they felt that their cancer diagnosis was more important than the HCS diagnosis in terms of re-evaluating their life priorities.

> Not really, nothing changed, I guess the shock was bigger when I got the cancer diagnosis (. . .) but still I don't think I made any big changes apart from what I could not do. **L2**

Subtheme 2: Perceptions About the Importance of Lifestyles

When asking about prevention, participants recounted that they had thought about health behaviors, but generally they did not assign much importance to it.

For the most part, participants felt they had received little information about lifestyle and did not see it necessary to make any changes, or they thought that they had such a high risk that behavioral modifications would not impact their cancer risk.

> The doctor didn't tell me anything about lifestyles . . . I didn't change my lifestyle. **B2**

> The doctor said, smoking is not good, you know, but it doesn't cause breast cancer. And, why change? I will probably have cancer at some point. **B14**

In contrast, some participants felt like the HCS status was a motivation to either maintain good habits or improve on them, even if just to face a future cancer from a level of good baseline health.

> I changed my diet, my life, and after the surgery [preventive mastectomy] I started to go to the swimming pool twice a week. **B6**

> I think it's important to be in good health to face cancer. **L1**

Subtheme 3: Adapting to Emotional Changes

Emotions change and evolve over time in those with HCSs.

> I do feel like a different person. From how I was. And, yeah, I think, a change sort of emotionally. On the one hand, I feel quite resilient. And on the other hand, I feel quite vulnerable. **L6**

Participants were very attuned to the changes, but they perceived very little support toward their psychosocial wellbeing. One patient even expressed her gratitude to the interviewer.

> I have never been asked about my feelings, thank you. **B5**

Their coping strategies also varied depending on how long ago they were diagnosed with their HCS, whether they had developed cancer, and the idea they had about HCSs. Most participants moved from the shock of diagnosis:

> It was all a lot to take on board. **B11**

> . . . to a state of uncertainty they described as a *double-edged sword, a loaded die,* or a *bad lottery,* which made them live with fear.

> I cannot have back pain; it has to be cancer. **B4**

Some expressed feelings of defeat after so much constant surveillance.

> Year after year, the same fight to get an appointment, the preparation for a colonoscopy, which is horrible . . . sometimes I just want to stop. **L3**

Some participants felt relief upon diagnosis of HCSs, something that they almost expected. This experience was normally associated with a long family history of cancer.

> In some way, for me, it was it was quite a relief. **L2**

> I was now glad that "I now know my enemy". **L7**

Others felt that HCS was a constant burden, and they needed support.

> I think they're both equally hard, really living, getting the diagnosis wasn't nice, and then living with it. **B10**

> I basically I feel like no one was there to guide me and to hold my hand. **L1**

Theme 3: Limitations of Health Care Providers

This theme encompasses perceived shortcomings in health care professionals' knowledge and communication skills around HCSs (as opposed to unmet informational support needs reported in theme 1). They described difficulties when facing different professionals and a desire for a less disjointed health care experience.

Subtheme 1: Communication Skills Among Health Care Professionals

Most participants described communication with health care professionals about HCSs as a real, unmet need, with many wishing for more active listening and an empathetic attitude from their health care providers.

> You have to listen, what is this person thinking about now? What are they understanding now? What is the knowledge now? What do they know? What do they not know? And then listen. **B14**

> They're great at telling us how often we need a colonoscopy and how often we need to have guidance screening. But they're not really good at sitting and listening and explaining what you want to know. **L4**

Subtheme 2: Perception of Insufficient Knowledge from Health Care Professionals

Participants felt that health professionals lacked an understanding of HCSs. There was a common feeling of deficits in communication in the health care system, which created a burden for patients, as they had to repeatedly share information about their condition to different professionals.

> Any time I spoke to somebody medically and said I had Lynch syndrome, they looked so lost themselves. I think they thought I was making it up. So, I stopped telling anybody. **L4**

> I think it's terrible, though, that it comes down to the luck of the draw. Whether you are lucky enough to meet a medical professional that knows enough about it. **L8**

While they understood that health care professionals cannot know everything, they wished their providers knew more about HCSs. This need was more relevant for people with Lynch syndrome, who found that many health care professionals had not heard about this syndrome, while HBOC carriers felt that the BRCA mutation was understood, but only in terms of breast and ovarian risks, not other cancer risks. They found it difficult to secure the follow-up appointments recommended for their syndrome, meaning that the patients themselves had to be on top of their own surveillance, as there is no coordination within the health care system.

> You know, you have to fight for some things. I mean, fight to get a colonoscopy every two years, even though that is standard. Because some doctors will be like,… Oh, well, you know, you don't need it now. **L5**

> *I have my gynaecologist. Yeah. So, uh, he's not, he's not interested either. So it's totally up to me to make sure that I'm tested regularly.* ***B3***

> *I also was not given any sort of plan, monitoring plan or whatever it was.* **L1**

Subtheme 3: Desire for a Comprehensive Follow-Up

Participants—especially HBOC previvors and those with Lynch syndrome—commonly wished they had a health professional who would be available at their follow-ups. On many occasions, they identified nurses and specifically nurses working in oncology as their ideal person to talk to on a regular basis and use as a focal point with the health care system in case any doubts arose.

> People feel lost. We feel that there is a need to have an annual check-up with a consultant or nurse. Some of us are scared, even though we know there's a very low chance of finding anything after mastectomy. **B5**

Interviewees wished that health providers would give them a list of services, support groups, and written information to help them navigate their needs and queries. Another participant recommended health professionals use social media to reach young people.

> For example, a health professional could tell you the news and say, we've got this website with this information, and this is what you can do to reduce your risks, and be very clear and supportive. **L6**

> You can and give guidance and counselling and education, getting involved in social media like an internal app. **B7**

Discussion

Our results indicate that people with HCSs from around Europe perceive that health professionals do not have complete and exhaustive knowledge about HCSs and, consequently are not providing the support they need. While the information needs of previvors and those that have had cancer are different,[9] they all reported unmet needs. They received some information pre- and post-test but little to no information thereafter, regardless of the country where they were receiving care. Other studies, both in and outside of Europe, have drawn similar conclusions about the need for information.[10,36]

On top of their unmet needs in the information domain, they also described lack of coordination in their follow-up, difficulties that have also been shared in other studies.[11,37] HCS carriers feel unsupported, and this affects their ability to cope with the diagnosis and their decision-making. During the interviews, they sometimes wondered if their decisions would have been different had they received more information.

Participants also expressed an interest in having cancer nurses involved in their care. These nurses generally perform a lot of educational interventions, but the programs are normally targeted to people undergoing cancer treatment, not to previvors, and the education mainly focuses on the side effects of treatment in survivors.[38] HCS carriers differ from the general patient population, who generally demonstrate great trust in their health care professionals, especially nurses,[39] in that their trust in health care professionals is undermined by their perceived lack of knowledge about HCSs.[9]

In line with these perceived shortfalls, HCS carriers seek information and peer support elsewhere, especially the internet, where they often find solace from peers affected by the same syndrome. Our study also suggests that knowledge shared by peers with similar experiences is greatly valued. Health professionals should recognize that value providing information that people need and facilitating access to support groups. Social media platforms have become popular avenues to seek health care information and support among cancer patients.[40] There is a lack of research on how HCS support groups and social media are used by this population and the impact it has on them. In 2016, a couple of genetic counsellors commented on the benefits of social media and support groups[41] and the need for health care professionals to get involved in social media,[42] but there has been no subsequent research on interventions or impact. A Cochrane Review in breast cancer patients and support group interactions[43] concluded that being part of a support group can relieve anxiety and even improve quality of life, and in interviews with HBOC previvors,[44] participants shared that writing down their experience was helpful to process their own feelings. In this study, we found that HCS carriers felt a sense of belonging when participating in these groups, but there was also some sense of burnout in those providing the most support. While many studies have explored the benefits, few have investigated the negative impact of social media; one qualitative study in young cancer patients[45] revealed that they felt some level of burden and negative impact from reading the experience of others. Future research should explore the role of social media for HCS carriers as well as the emotional burden of supporting peers.

Participants had very different views on the perceived benefits of their own actions such as lifestyle behaviors. Health care professionals have the ability to influence those beliefs,[24] but while health professionals are knowledgeable about cancer prevention, they do not promote literacy on cancer prevention and lifestyle behaviors.[46,47] The current lack of engagement from health care professionals in follow-up and health behaviors, together with the dearth of behavioral research and interventions to address lifestyle behaviors,[27,28] is affecting the self-management and actions of HCS carriers. There is also a lost opportunity regarding the potential to use social media and patient support groups to promote cancer prevention and healthy behaviors.[48,49]

In order to feel engaged in their self-management and self-care, HCS carriers need to have their psychosocial needs met and be able to accept and process the storm of feelings brought on by an HCS diagnosis. Participants in the interviews, and the existing evidence, reinforce the need to improve patients' experience.[9-12,36,50] Regardless of whether they have been diagnosed with cancer, finding out about a genetic alteration of this kind takes some getting used to.[37,51] From assimilating the concerns from and about their family to understanding the myriad impacts of the different management strategies they are offered, HCSs carriers have a real need for psychosocial support,[51] a trusting relationship with the health care system, and health services that promote healthy behaviors. In this line, the six factors formulated in the Multifactorial Positive Mental Health Model[25]: personal satisfaction, prosocial attitude, self-control, autonomy, problem-solving and self-actualizations, and interpersonal relationships; have been proven effective in different intervention programs,[52,53] with a positive impact on self-care.[26]

Our findings are suggestive of a generalized need for more nurse education on HCSs in Europe. Health care professionals should be more involved in the follow-up of HCS carriers, who in turn need to be empowered to take a lead role in their own care. Closer involvement of health systems in satisfying these needs would allow patients to feel more supported and empowered.

Study Strengths and Limitations

A key strength of this study was the involvement of a PPI panel during the planning and design stage. PPI is very important in cancer research and more so in PhD projects to ensure that the studies and research questions are pertinent for them.[28,54,55] Our inclusion of both previvors and survivors also means our results are generalizable to all HCS carriers, without neglecting the differences that may exist according to their cancer status or country. While systems, opportunities, and access to genetic counselling vary in these countries,[56] HCS carriers have common needs and experiences.

This study also has some limitations. Participants were recruited via social media and patient support groups, so the views and needs of HCS carriers that do not even have the information and support from these groups are not included; therefore, we may be leaving out an important group to explore. Also, while we had the views of both men and women with BRCA and Lynch syndrome, far fewer men were in our sample, meaning we may have overlooked some of their needs by not including a large enough sample.

Conclusions and Clinical Implications for Nurses

This qualitative study provides insight into the perspectives and needs of HCS carriers on their long-term management. People with HCSs need a health professional they can go to in order to ask questions and who can help them navigate the system and meet their needs. Moreover, health care professionals should have a role in the follow-up and long-term management of these patients. Nurses are well placed to promote self-management and advocate for patient decision-making; however, they need to have adequate skills and knowledge to effectively perform this role.

In light of how many of our participants were actively supporting others on social media, future studies should look further into the involvement of HCS carriers in social media and the emotional burden that they feel.

People with HCSs are asked to make very difficult decisions on surveillance and management. Building professional capacity and conducting more research on lifestyle behaviors and behavioral theories would help enable these patients to make informed choices. With the rapid adoption of genomics in cancer care, there is and will be more demand for genetic testing, which will increase the need for professionals to guide and support this population.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

CRediT authorship contribution statement

Celia Diez de los Rios de la Serna: Conceptualization, Methodology, Validation, Formal analysis, Investigation, Resources, Data curation, Writing – original draft, Writing – review & editing, Visualization, Project administration, Funding acquisition. **Maria Teresa Lluch-Canut:** Conceptualization, Methodology, Validation, Resources, Writing – original draft, Writing – review & editing, Supervision. **Maria Paz Fernández-Ortega:** Conceptualization, Methodology, Validation, Formal analysis, Resources, Data curation, Writing – original draft, Writing – review & editing, Supervision.

Funding

This work was supported by an education grant award from the International Society of Nurses in Genetics (ISONG).

Acknowledgments

The authors would like to sincerely thank the PPI panel for their support, ideas and encouragement. The authors are also immensely grateful to the participants who were so open and shared their experiences and struggles. The authors also want to thank all the professionals, patients and patient organizations that supported and shared this study.

Supplementary materials

Supplementary material associated with this article can be found in the online version at doi:10.1016/j.soncn.2024.151624.

References

1. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med*. 2015;17(5):405–424. https://doi.org/10.1038/GIM.2015.30.
2. Imyanitov EN, Kuligina ES, Sokolenko AP, et al. Hereditary cancer syndromes. *World J Clin Oncol*. 2023;14(2):40. https://doi.org/10.5306/WJCO.V14.I2.40.
3. Garutti M, Foffano L, Mazzeo R, et al. Hereditary cancer syndromes: a comprehensive review with a visual tool. *Genes (Basel)*. 2023;14(5):1025. https://doi.org/10.3390/GENES14051025/S1.
4. De Keersmacker S, Meder S, Cassidy D. Europe's beating cancer plan: a new EU approach to prevention, treatment and care. *Press Release Eur Comm*. 2021:(February):1–3.
5. Berger MF, Mardis ER. The emerging clinical relevance of genomics in cancer medicine. *Nat Rev Clin Oncol*. 2018;15(6):353–365. https://doi.org/10.1038/s41571-018-0002-6.
6. Riley BD, Culver JO, Skrzynia C, et al. Essential elements of genetic cancer risk assessment, counseling, and testing: updated recommendations of the National Society of Genetic Counselors. *J Genet Couns*. 2012;21(2):151–161. https://doi.org/10.1007/s10897-011-9462-x.
7. Knapp ST, Revette A, Underhill-Blazey M, et al. MyLynch: a patient-facing clinical decision support tool for genetically-guided personalized medicine in Lynch syndrome. *Cancers (Basel)*. 2023;15(2):391. https://doi.org/10.3390/CANCERS15020391/S1.
8. Dean M, Fisher CL. Uncertainty and previvors' cancer risk management: understanding the decision-making process. *J Appl Commun Res*. 2019;47(4):460–483. https://doi.org/10.1080/00909882.2019.1657236.
9. Dean M, Scherr CL, Clements M, Koruo R, Martinez J, Ross A. "When information is not enough": a model for understanding BRCA-positive previvors' information needs regarding hereditary breast and ovarian cancer risk. *Patient Educ Couns*. 2017;100(9):1738–1743. https://doi.org/10.1016/J.PEC.2017.03.013.
10. Warner NZ, Gleeson C, Fahey P, Horgan R, Groarke AM. Experiences of living with Lynch syndrome: a reflexive thematic analysis. *Eur J Oncol Nurs*. 2022;58: 102117. https://doi.org/10.1016/J.EJON.2022.102117.
11. Warner NZ, Groarke AM. A qualitative reflexive thematic analysis into the experiences of being identified with a BRCA1/2 gene alteration: "So many little, little traumas could have been avoided.". *BMC Health Serv Res*. 2022;22(1):1–12. https://doi.org/10.1186/S12913-022-08372-W/TABLES/2.
12. Bartley N, Napier C, Best M, Butow P. Patient experience of uncertainty in cancer genomics: a systematic review. *Genet Med*. 2020;22(9):1450–1460. https://doi.org/10.1038/S41436-020-0829-Y.
13. Oliveira FFB, de Barros Silva PG, de Sant'Ana RO, et al. How does genetic testing influence anxiety, depression, and quality of life? A hereditary breast and ovarian cancer syndrome suspects trial. *Support Care Cancer*. 2020;29(7):3521–3530. https://doi.org/10.1007/S00520-020-05867-2.
14. Pollard S, Kalloger S, Weymann D, et al. Genetic testing for hereditary cancer syndromes: patient recommendations for improved risk communication. *Health Expect*. 2020;23(4):884–892. https://doi.org/10.1111/hex.13062.
15. Stjepanovic N, Moreira L, Carneiro F, et al. Hereditary gastrointestinal cancers: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up. *Ann Oncol*. 2019;30(10):1558–1571. https://doi.org/10.1093/annonc/mdz233.
16. National Comprehensive Cancer Network. NCCN genetic/familial high-risk assessment: breast, ovarian, and pancreatic. Available at: https://www.nccn.org/professionals/physician_gls/pdf/genetics_bop.pdf Accessed February 23, 2024.
17. National Comprehensive Cancer Network. NCCN genetic/familial high-risk assessment: colorectal. Available at: https://www.nccn.org/professionals/physician_gls/pdf/genetics_colon.pdf Accessed February 23, 2024.
18. Balmaña J, Balaguer F, Cervantes A, Arnold D. Familial risk-colorectal cancer: ESMO clinical practice guidelines. *Ann Oncol*. 2013;24(Suppl.6):vi73–vi80. https://doi.org/10.1093/annonc/mdt209.
19. Berliner JL, Cummings SA, Boldt Burnett B, Ricker CN. Risk assessment and genetic counseling for hereditary breast and ovarian cancer syndromes—practice resource of the National Society of Genetic Counselors. *J Genet Couns*. 2021;30(2):342–360. https://doi.org/10.1002/JGC4.1374.
20. Calzone KA, Kirk M, Tonkin E, Badzek L, Benjamin C, Middleton A. Increasing nursing capacity in genomics: overview of existing global genomics resources. *Nurse Educ Today*. 2018;69:53. https://doi.org/10.1016/J.NEDT.2018.06.032.
21. Papaioannou K, Kampourakis K. Health-care professionals' awareness and understanding of genomics. *Appl Genomics Public Heal*. 2020:225–242. https://doi.org/10.1016/B978-0-12-813695-9.00012-1.
22. Connors LM, Voian N, Shi Y, Lally RM, Edge S. Decision making after BRCA genetic testing. *Clin J Oncol Nurs*. 2014;18(3):E58–E63. https://doi.org/10.1188/14.CJON.E58-E63.
23. Moreno L, Linossi C, Esteban I, et al. Germline BRCA testing is moving from cancer risk assessment to a predictive biomarker for targeting cancer therapeutics. *Clin Transl Oncol*. 2016;18(10):981–987. https://doi.org/10.1007/s12094-015-1470-0.
24. Glanz K, Rimer BK, Viswanath K, eds. *Health Behavior and Health Education: Theory, Research, and Practice*. 4th ed. San Francisco: Jossey-Bass; 2008.
25. Lluch Canut MT. *Construcción de una escala para evaluar la salud mental positiva. Doctoral thesis*. Barcelona, Spain: Faculty of Psychology, University of Barcelona; 1999.
26. Naranjo Hernández Y, Concepción Pacheco JA, Rodríguez Larreynaga M. The Self-Care Deficit Theory: Dorothea Elizabeth Orem. *Gac Méd Espirit*. 2017;19(3):89–100.
27. Donato KM, Moore K, Parker WM, et al. Health and lifestyle behaviors in colorectal cancer survivors with and without Lynch syndrome. *J Community Genet*. 2019;11(1):59–63. https://doi.org/10.1007/s12687-019-00421-y.
28. Diez de los Rios de la Serna C, Fernández-Ortega P, Lluch-Canut T. Lifestyle behavior interventions for preventing cancer in adults with inherited cancer syndromes: systematic review. *Int J Environ Res Public Health*. 2022;19(21). https://doi.org/10.3390/ijerph192114098.
29. Briefing notes for researchers - public involvement in NHS, health and social care research | NIHR. Available at: https://www.nihr.ac.uk/documents/briefing-notes-for-researchers-public-involvement-in-nhs-health-and-social-care-research/27371 Accessed July 27, 2023.
30. Kallio H, Pietilä AM, Johnson M, Kangasniemi M. Systematic methodological review: developing a framework for a qualitative semi-structured interview guide. *J Adv Nurs*. 2016;72(12):2954–2965. https://doi.org/10.1111/JAN.13031.
31. Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. *Int J Qual Health Care*. 2007;19(6):349–357. https://doi.org/10.1093/INTQHC/MZM042.
32. Robinson OC. Sampling in interview-based qualitative research: a theoretical and practical guide. *Qual Res Psychol*. 2014;11(1):25–41. https://doi.org/10.1080/14780887.2013.801543.
33. Braun V, Clarke V. To saturate or not to saturate? Questioning data saturation as a useful concept for thematic analysis and sample-size rationales. *Qual Res Sport Exerc Heal*. 2021;13(2):201–216. https://doi.org/10.1080/2159676X.2019.1704846.
34. Braun V, Clarke V. One size fits all? What counts as quality practice in (reflexive) thematic analysis? *Qual Res Psychol*. 2020;18(3):328–352. https://doi.org/10.1080/14780887.2020.1769238.
35. Braun V, Clarke V, Hayfield N, Terry G. Thematic analysis. *In: Handbook of Research Methods in Health Social Sciences*. Singapore: Springer; 2019:843–860. https://doi.org/10.1007/978-981-10-5251-4_103.
36. Wellman ML, Holton AE, Kaphingst KA. "Where do I go? Who do I go to?": BRCA previvors, genetic counselors and family planning. *PEC Innov*. 2023;2:100157. https://doi.org/10.1016/J.PECINN.2023.100157.
37. Campbell-Salome G, Buchanan AH, Hallquist MLG, Rahm AK, Rocha H, Sturm AC. Uncertainty management for individuals with Lynch syndrome: identifying and responding to healthcare barriers. *Patient Educ Couns*. 2021;104(2):403–412. https://doi.org/10.1016/J.PEC.2020.07.017.
38. Charalambous A, Wells M, Campbell P, et al. A scoping review of trials of interventions led or delivered by cancer nurses. *Int J Nurs Stud*. 2018;86:36–43. https://doi.org/10.1016/J.IJNURSTU.2018.05.014.
39. Dinç L, Gastmans C. Trust in nurse–patient relationships. *Nurs Ethics*. 2013;20(5):501–516. https://doi.org/10.1177/0969733012468463.
40. Gandamihardja TAK, Liyanage S, Coutee T, Peled AW, Masannat YA. The role of social media and breast cancer: how does it impact patients? *Breast Care*. 2023;18(3):203–208. https://doi.org/10.1159/000530433.
41. Stefansdottir V. Experience of social media support group for BRCA carriers. *J Genet Couns*. 2016;25(6):1342–1344. https://doi.org/10.1007/S10897-016-0009-Z.
42. Gallagher L, McCuaig J, Benoit L, Davies C. It's time for the genetic counseling profession to embrace social media. *J Genet Couns*. 2016;25(6):1338–1341. https://doi.org/10.1007/S10897-016-9950-0/METRICS.
43. Mccaughan E, Parahoo K, Hueter I, Northouse L, Bradbury I. Online support groups for women with breast cancer. *Cochrane Database Syst Rev*. 2017;2017(3): CD011652. https://doi.org/10.1002/14651858.CD011652.pub2.
44. Wellman ML, Holton AE, Kaphingst KA. Previvorship posting: why breast cancer previvors share their stories on social media. *Health Commun*. 2023;38(11):2441–2449. https://doi.org/10.1080/10410236.2022.2074780.
45. Stage C, Klastrup L, Hvidtfeldt K. Ugly media feelings: Negative affect in young cancer patients' experiences of social media. *First Monday*. 2021;26(7). https://doi.org/10.5210/fm.v26I7.11093.
46. Fleary SA, Paasche-Orlow MK, Joseph P, Freund KM. The relationship between health literacy, cancer prevention beliefs, and cancer prevention behaviors. *J Cancer Educ*. 2019;34(5):958–965. https://doi.org/10.1007/S13187-018-1400-2.
47. Albada A, Vernooij M, van Osch L, Pijpe A, van Dulmen S, Ausems MGEM. Does and should breast cancer genetic counselling include lifestyle advice? *Fam Cancer*. 2014;13(1):35–44. https://doi.org/10.1007/S10689-013-9672-5.
48. Lauckner C, Whitten P. The differential effects of social media sites for promoting cancer risk reduction. *J Cancer Educ*. 2016;31(3):449–452. https://doi.org/10.1007/s13187-015-0881-5.
49. Han CJ, Lee YJ, Demiris G. Interventions using social media for cancer prevention and management: a systematic review. *Cancer Nurs*. 2018;41(6):E19–E31. https://doi.org/10.1097/NCC.0000000000000534.
50. Dean M, Davidson LG. Previvors' uncertainty management strategies for hereditary breast and ovarian cancer. *Health Commun*. 2018;33(2):122–130. https://doi.org/10.1080/10410236.2016.1250187.
51. Yanes T, Willis AM, Meiser B, Tucker KM, Best M. Psychosocial and behavioral outcomes of genomic testing in cancer: a systematic review. *Eur J Hum Genet*. 2019;27(1):28–35. https://doi.org/10.1038/s41431-018-0257-5.
52. Puig Llobet M, Sánchez Ortega M, Lluch-Canut M, Moreno-Arroyo M, Hidalgo Blanco MÀ, Roldán-Merino J. Positive mental health and self-care in patients with chronic physical health problems: implications for evidence-based practice. *Worldviews Evidence-Based Nurs*. 2020;17(4):293–300. https://doi.org/10.1111/WVN.12453.

53. Sánchez-Ortega MA, Lluch-Canut MT, Roldán-Merino J, et al. Nursing intervention to improve positive mental health and self-care skills in people with chronic physical health conditions. *Int J Environ Res Public Health*. 2022;20(1):528. https://doi.org/10.3390/IJERPH20010528.
54. Colomer-Lahiguera S, Steimer M, Ellis U, et al. Patient and public involvement in cancer research: a scoping review. *Cancer Med*. 2023;12(14):15530–15543. https://doi.org/10.1002/cam4.6200.
55. Tanay MA, Diez de los Rios de la Serna C, Boland V, et al. Patient and public involvement in research: reflections and experiences of doctoral cancer nurse researchers in Europe. *Eur J Oncol Nurs*. 2023;64:102351. https://doi.org/10.1016/J.EJON.2023.102351.
56. Abacan MA, Alsubaie L, Barlow-Stewart K, et al. The global state of the genetic counseling profession. *Eur J Hum Genet*. 2019;27(2):183–197. https://doi.org/10.1038/s41431-018-0252-x.