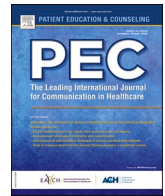


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How hereditary cancer risk disclosure to relatives is handled in practice – Patient perspectives from a Swedish cancer genetics clinic

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ABSTRACT

Objectives: Hereditary cancer risks can be effectively managed if at-risk relatives enroll in surveillance and preventive care. Family-mediated risk disclosure has internationally been shown to be incomplete, selective and leave over a third of eligible at-risk individuals without access to genetic counseling. We explored patients handling of cancer risk information in practice.

Methods: We conducted twelve semi-structured interviews with patients who had completed their genetic counseling and been asked to disclose risk information to relatives. Questions were designed to investigate lived experiences of communicating hereditary risk and focused on disclosure strategies, intrafamilial interactions and emotional responses.

Results: Qualitative content analysis yielded five categories. These span personal fears, shared responsibilities, feeling of empowerment, innovative solutions and unmet needs. Patients put high value on collaboration with their genetic healthcare professionals but also solicited better overview of the counseling process and more personalized, case-tailored information.

Conclusions: Our results add novel insights about the practical strategies employed by genetic counselees and their motivations behind disclosing hereditary risk information to relatives.

Practice implications: A patient-centered cancer genetics care would clarify roles and responsibilities around risk disclosure, inform counselees about the process upfront and tailor information to offer case-specific data with the family's inheritance pattern explained.

1. Introduction

Targeted surveillance of at-risk relatives in families with a hereditary predisposition for cancer is a well-established form of disease prevention [1–3]. This type of preventive approach is dependent on the success of reaching at-risk relatives with relevant risk information and the offer of genetic counselling [4,5].

Traditionally, the responsibility of communicating cancer risk information to at-risk relatives have been placed on the proband; the first family member undergoing evaluation for hereditary cancer. Relying on this mode of risk dissemination, also called family-mediated disclosure has clear limitations. Although most probands convey risk information to some relatives [6], especially to close family, a recent systematic review concluded one third of all at-risk relatives are not reached [7] and only 30–44% of eligible at-risk relatives go through with genetic testing

[3,8,9].

A plausible reason behind the poor uptake of counselling could be personal preferences, i.e. not wanting to know of a hereditary risk. However, this is contradicted by the fact that people in general have a strong desire to receive hereditary cancer risk information [10–13] and both patients and members of the public have similar preferences on this topic [12,14]. Instead, studies report family-mediated risk disclosure as being incomplete, selective, and complex for probands to handle on their own [9,15,16] signifying a lost opportunity for cancer prevention.

Barriers to effective disclosure have been identified at both patient, provider and system levels [2] and factors such as perceived lack of responsibility, emotional burden, degree of relationship with the relative and access to care have been shown to be important [17]. The collected evidence suggests families do require additional forms of support beyond current clinical praxis [3,18,19]. But there is scarce

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evidence on how to facilitate risk disclosure and ensure more equal access to genetic counselling among at-risk relatives [20].

Previous efforts including supportive psychosocial coaching, tailored information aids and augmented counseling [19] have shown moderate effects on counselling uptake. Calls for more proactive approaches with healthcare-assisted disclosure to at-risk relatives have been raised [3, 8, 21–23]. Although studies evaluating direct contact show promising results, the results have not yet been confirmed in a randomized trial and the actual effectiveness in a real-life clinical setting has been a challenge to establish [20,24].

There is thus an evident knowledge gap about how to best tailor counseling to promote effective risk disclosure. Experts in the field have highlighted the need for more research on communication barriers [24], and more focus on qualitative investigation of post-disclosure variables [25]. This study aims to fill that gap by exploring the lived experiences of patients dealing with communication of hereditary cancer risks, with a special focus on handling risk disclosure to at-risk relatives.

2. Methods

Eligible participants were recruited in collaboration with a cancer genetics clinic in Northern Sweden. The cancer genetics clinic is a specialized care unit at a regional university hospital where individuals are offered genetic counseling for different hereditary cancer risk syndromes. Counselees consist of either referred cancer patients, healthy individuals who want to know if they have an increased hereditary risk of cancer or healthy at-risk relatives who already know of a pathogenic variant in the family and want to undergo pre-symptomatic testing. Hence, in this study, the concept of *family-mediated risk disclosure* includes all topics of intrafamilial communication about results from genetic counseling. Both information about a known pathogenic variant and about genetic screening being negative but the family has a “familial cancer syndrome” where surveillance is offered based on a cancer risk (estimated from the pedigree as well as number and characteristics of cancer cases).

We employed consecutive enrolment and used convenience sampling. The staff were instructed to invite individuals with at least one at-risk-relative fulfilling three inclusion criteria; they had completed their last genetic counseling appointment, they had been investigated for a hereditary risk of breast, ovarian or colorectal cancer and they were over 18 years of age. If the patient accepted the oral invitation, the patient information leaflet and a consent form were mailed to the individual by post. Individuals who consented to participate in writing were contacted by phone by the first author and scheduled for a telephone interview.

Of the 14 returned consent forms, two persons could not be reached through the contact details provided. The final sample included twelve individual interviews with patients who had personal experiences of undergoing genetic counseling and being faced with the task of informing at-risk relatives (Table 1). The sample included eight women and four men between the ages of 22 and 73 years (median 57 years). Six of the twelve respondents had at some time in their life been diagnosed with cancer. All participants had some experiences relating to cancer disease in the family and several had experienced loss of a loved one due to cancer. The time since their last counseling session varied between a few months to almost a year, at the time of data collection which took place between 22 of May to 18 of December 2018.

A semi-structured interview protocol was used (Appendix 1). Sound was captured with a voice recorder and microphone especially adapted for telephone interviews. The interviews lasted between 36 and 60 min, with a median length of 47 min. Voice recordings were transcribed verbatim by an experienced medical secretary and then checked by the first author against voice recordings, anonymized, and exported as text files to the software Open Code, version 4.03, copyright of ITS and the Division of Epidemiology and Global Health, Department of Public Health and Clinical Medicine (2022) at Umeå University, Sweden [26].

All interviews started with a broad question asking the respondent to

Table 1
Characteristics of patients included.

Characteristics of interviewees (n = 12)	
<i>Sex</i>	
Female	8
Male	4
<i>Role (main perspective)</i>	
Proband	7
At-risk relative	5
<i>Diagnose</i>	
Pathogenic variant in BRCA1 (n = 1), BRCA2 (n = 3), MSH2 (n = 1)	5
No pathogenic variant, but surveillance recommended	7
<i>Personal cancer history</i>	
Yes	6
No	6
<i>Family situation</i>	
Married/cohabitating with children	9
Single without children	3
<i>Living area</i>	
City	6
Small town	4
Rural	2
<i>Housing situation</i>	
Own house	7
Condominium flat	1
Rental apartment	4

recount their personal story of encountering the cancer genetics clinic and learning about a possible hereditary risk in their family. This allowed the research team to get a sense of each individual’s context and background, as well as letting the respondent frame the chain of events in their own way and using their own narrative. The first set of targeted questions concentrated on what risk information they had been given and how they understood the information.

The next section inquired about how they experienced the process of genetic counseling, and how they managed the knowledge in relation to other family members. Details on when, why, and what they decided to share with family and relatives were discussed in detail. The order of questions discussed were at times adapted to fit the participant’s choice of topic and content area. If a participant paused, or seemed to have more to say, the interviewer used follow-up questions prompting the respondent to elaborate on experiences relevant for the aim of the study. All interviews were held in Swedish.

Qualitative content analysis according to Graneheim and Lundman [27] was used to analyze interviews. Each interview was listened to, and full transcripts were read to establish a sense of the whole. The analysis procedure first decontextualized interview data and later recontextualized findings relevant for the study aim. The full interview transcripts were used as unit of analysis. Text fragments with a bearing on the study aim were identified as meaning units, and subsequently condensed and coded (examples in Table 2). Recurrent topics were noted continuously as memos, in part to assess data saturation [28]. Tentative sub-categories were drafted based on the codes to assemble pieces of data with similar meaning further distinguishing nuances and variations. We analyzed both manifest and latent content in our data.

Sub-categories were scrutinized in detail for a coherent level of abstraction. The wording of the categories was iterated several times in joint discussion between the authors and adjusted to represent the essence of the accumulated codes. Sub-categories were also compared to similar ones, analyzed regarding differences in meaning, semantics and scope and finally interpreted to form categories. Recurrent consensus discussions throughout the coding process resulted in the final table of categories.

3. Results

The analysis of patient interviews resulted in five main categories (Table 3). Of note is that a large part of the interview data concerned

Table 2

Examples from the decontextualization phase of content analysis. From left, interview number, identified meaning unit corresponding with the study aim in the transcript, the condensation of meaning unit and the applied code. Right-most column shows the sub-category each code was grouped into at this stage of the analysis process.

Interview	Meaning unit	Condensation	Code	Subcategory
Interv. 2	...it's filled with anxiety but after discussing with both the physician and the nurse I thought it's only reasonable (they were informed) ...	Feel anxiety at first but reconsider	Struggle with difficult feelings	<i>Struggling to bring it up</i>
Interv. 7	...read this information folder so I won't have to explain 'cause I don't possess that kind of knowledge... I'm just passing on what I was told...	Would like an information folder as support to explain Not possess the knowledge – just pass on risk information	Want more written information Feeling inadequate to inform	<i>Envisioned support and services</i> <i>Limited capacity to inform</i>
Interv. 3	...I found out that it would be a lot up to me to relay this information to relatives. I became a sort of informant, standing between the facts and my family...	Becoming the informant between healthcare and family	Unexpectedly becoming the informant	<i>Lack of information about the process</i>

Table 3

Table of categories. Results of the qualitative content analysis as five categories shown with respective sub-categories.

Sub-categories	Main categories
Struggling to bring it up Being put in unexpected role Hearing criticism via others Respecting autonomy in myself and others Understand risk and concern for others Sharing a norm of the right thing to do Handing over to the next in line No big deal to communicate risk Gratitude for access to genetic counseling Encouragement from healthcare meaningful Preferred modes of contact Relief from finding out result Cooperate with relatives	Confront own fear and family resistance
Forwarding info from the cancer genetics unit Designing homemade risk info Lack of information about the process Uncertainty about healthcare's mandate Limited capacity to inform Envisioned support and services	Recognize a shared responsibility
	Feel empowered by healthcare contact
	Finding creative pathways to inform
	Recognize need for improved support

how our respondents chose to act and manage risk information in relation to the support they were given, the things they were told by the healthcare staff and in relation to what and how they themselves had interpreted the results of genetic counseling.

We also identified a persistent dual perspective in the form of two counteracting forces in the dataset; the motivation to share risk and the reluctance to do so in order to protect others. This emotional struggle evidently causes discomfort and hesitation and highlights the challenging situation of knowing one may induce harm and worry in loved ones, but in parallel acknowledge that risk information may be both appreciated and beneficial.

The findings range across topics from the individuals' feelings of inadequacy to empathetic, altruistic considerations of relatives' reactions and finally to the experiences and limitations in current risk information support.

3.1. Confront own fear and family resistance

The first category echoes personal difficulties experienced when encountering affective and cognitive hurdles in the process of sharing hereditary risk with family. The first sub-category *Struggling to bring it up* describes how some respondents or their relatives put off or delay risk disclosure. This topic also includes codes on lingering cancer worries, anxiety due to uncertainty, and fear of making loved ones upset by sharing risk information. Mentions revealed an awareness of complex emotions like guilt and being the messenger of bad news.

However, there were also expressions of resilience and overcoming challenging emotions, with tendencies to bring up the rationale and importance of going through the fear for a greater good (see example quotes in Table 4).

Regardless of how the respondents had come in contact with the cancer genetics unit, several describe the unexpected role of suddenly being an intermediate actor in the information process. *Being put in unexpected role* reflects the psychologically demanding situation of being appointed with a task to inform others and be placed in between the family and healthcare services. Respondents described the awkward situation of overseeing important but highly complex health information. For some respondents, coping with this situation involved prompt action – a task to swiftly complete and “get it over with”. Others treated the task to inform as a balancing act weighing the different aspects like benefits of disclosure against the risk of relatives reacting negatively during the process.

Additional expressions related to the influence of family dynamics on risk disclosure. *Hearing criticism via others* is a sub-category which stems from interviewees hearing about relatives' opinions by word-of-mouth. They might not have been told directly by a certain relative when they disapproved of their actions relating to risk disclosure, but instead they were later told through someone else.

The last sub-category named *Respecting autonomy in myself and others* contains statements on how the interviewees delineated their responsibilities from others, how they considered the wellbeing and preferences of relatives for the timing of risk disclosure and how the fundamental task was to offer relatives the knowledge necessary to make their own choice, no matter which way they decided to act.

3.2. Recognize a shared responsibility

The second category revolves around understanding the abstract concept of risk. The sub-category *Understand risk and concern for others* summarizes expressions about if and how relatives and family beside oneself may be affected and how inheritance patterns work. A prominent code in this category underscores the fact that risk is not equivalent to disease. Respondents explicitly soften and tone-down the seriousness of receiving risk information.

This category also includes acknowledgements of gender differences regarding cancer risks and considerations of how important it is to reach

Table 4
Example quotes from each category, with meaning units from selected sub-categories.

Cat.	Sub-category	Example quotes
Confront own fear and family resistance	Struggling to bring it up	...I'm not saying it is like that, but I think many people may feel guilt of having a cancer that is genetic and being the messenger of this disease /.../ it's quite irrational really, but I believe it's rather common, well that there is a sense of guilt in it... (Interview 8, female, 52 years, breast cancer, family history of breast cancer)
	Hearing criticism via others	...it was like she didn't want me to do it because...we will know we have it, I would kind of embarrass the whole family by revealing we got this...there was a resistance towards me going forward with it - but that was the only resistance I encountered... (Interview 7, female, 22 years, no cancer, family history of breast cancer)
	Respecting autonomy in me and others	...it's good that we get to do it if we want to, but she also said...it was a calm and pleasant talk, she was not upset or anything...we talked calmly, but she made clear she does not want to know and instead she will choose to close her eyes to what's ahead... (Interview 12, female, 35 years, no cancer, family history of breast cancer)
Recognize a shared responsibility	Handing over to the next in line	...well, if it runs in one generation who receives this information, then it is their duty to pass it on and continue this... (Interview 3, male, 70 years, no cancer, BRCA2-carrier)
	No big deal to communicate risk	I never hesitated. Since I had this knowledge, it wasn't a thing I could harbor in myself. What would happen if she would be affected (by cancer) and said: Why did you not tell me when you knew?! (Interview 2, male, 73 years, prostate cancer, BRCA2-carrier)
Feel empowered by healthcare contact	Gratitude for access to genetic counseling	...the available exams, I don't think I'm afraid of that, I think it is good I get to do them, it is very valuable. Whatever it shows I prefer to have it done so to say... (Interview 6, female, 69 years, colorectal cancer, family history of colorectal cancer)
	Encouragement from healthcare meaningful	I was probably affected by the physician, because implicitly there is this matter of sharing this knowledge...so naturally I was affected by it, and I think it's correct, I think knowledge is important. (Interview 2, male, 73 years, prostate cancer, BRCA2-carrier)
	Preferred modes of contact	...would after all want to know /.../ I know healthcare services are not allowed to make contact but for those who do carry a cancer gene, well I don't know...I think I would have wanted to know no matter if I would find out from healthcare or from a relative.

Table 4 (continued)

Cat.	Sub-category	Example quotes
Find creative pathways to inform	Relief from finding out result	(Interview 13, female, 32 years, no cancer, MSH2-carrier) ...there is a somewhat increased risk but there is nothing saying you'll get cancer and that's why I think it had a soothing effect when they informed us.... (Interview 11, male, 39 years, no cancer, BRCA2-carrier)
	Cooperate with relatives	...but then if the two youngest brothers go and take a test and they would be carriers then one may end up in a new situation... but where we are today, I don't feel it is anything I need to keep talking about or informing... (Interview 11, male, 39 years, no cancer, BRCA2-carrier)
Recognize need for improved support	Designing homemade risk info	...well, I wrote this information and then I multicopied it and cut over a hundred of those leaflets which I distribute and still have, I haven't reached everyone... (Interview 3, male, 70 years, no cancer, BRCA2-carrier)
	Lack of information about the process	I really wondered what was going to happen...if we would get a result now, or it would take a long time, or how fast it would go, I had no clue about that either... (Interview 7, female, 22 years, no cancer, family history of breast cancer)
	Uncertainty about healthcare's mandate	...the physician, if I remember correctly, talked about healthcare services not having the possibility or the mandate to inform, if I understood it correctly (Interview 2, male, 73 years, prostate cancer, BRCA2-carrier)
Feel empowered by healthcare contact	Limited capacity to inform	I couldn't describe what was going to happen, so I just told them you have to do this...it is difficult to explain why one should do something if one does not know the...outcome...what the solution will be... (Interview 8, female, 68 years, breast cancer, family history of breast cancer)
	Envisioned support and services	It feels really, really important... and a little, well it would feel pretty good if healthcare services made contact and not like it is now - when someone [genetic staff] says 'good that you called because we're not allowed to... (Interview 5, female, 68 years, breast cancer, family history of breast cancer)

out, motivating action to care for others first-hand and acting altruistic despite one's own temporary discomfort. Patients also express an awareness of how their own decisions will have repercussions on future events and the choices available for people around them.

Sharing the norm of the right thing to do is a rich sub-category with expressions related to the accepted norms about what is "right to do". Often respondents connected the possibility of accessing preventive measures first after being made aware of risk information. One patient referred to the family culture of openness as "we talk about everything between heaven and earth" as a way to express why they consider risk disclosure to be something of paramount importance over other factors, such as the right not to know.

Handing over to next in line refers to the feelings of a stepwise task performed by one generation, or in one branch of the family at a time. It could describe a person telling a cousin, and then letting him or her take the information further or sending a written notice to a distant relative and consider one's own part of disclosure as completed. Finally, the sub-category *No big deal to communicate risk* consists of a variety of straightforward statements on how obvious it is to go through with disclosure. Some were highly motivated by the urgency of the task and potential consequences, while others had experienced relief about how it had been "surprisingly easy to talk to" a sibling or a grown-up daughter.

3.3. Feel empowered by healthcare contact

The third category highlights the experiences of undergoing genetic counseling. The sub-category *Gratitude for access to genetic counseling* reflects an appreciation for the service provided, describing the availability of staff, the kind and empathetic appointments and the unexpected contacts and highly valued follow-up calls received months later. A gratitude for gaining access to and receive genetic counseling was recurrently voiced.

Expressions also revealed opinions that people "like themselves" should be given the opportunity to clarify their genetic status. Spontaneous suggestions included public information campaigns to raise public awareness of the services available. Patients also described in many ways how they found the *Encouragement from healthcare meaningful*. The feedback motivated them and affirmed their planned actions, thus prompting them to continue their mission to reach the relatives they had identified at risk.

Preferred modes of contact is a category which includes expressions about both personal wishes, and those of how respondents imagined relatives would want to be approached about a hereditary risk. Scenarios discussed ranged from preferences to sit down with someone face-to-face, forwarding a personalized family letter from healthcare, or picking up the phone to call a relative right away. Statements included both references to past experiences of counseling, but also envisioned future alternatives to current practice for making contact. In some cases, patients intuitively considered how they would do themselves if a direct contact alternative would have been available.

Lastly, the sub-category *Relief from finding out result* describe the feeling of being unburdened as the result was finally returned. This included both positive and negative results, where carriers described feelings of "always suspecting I also had it" but then experiencing relief to have a definitive answer and get the result confirmed in order to move on and handle the informative knowledge.

3.4. Finding creative pathways to inform

In this category the dedication and creativity of the patients is highlighted. *Cooperate with relatives* describes how unexpected alliances within families were formed to reach certain relatives, and how some go through the entire process of counseling together with a family member. Mainly to secure emotional support but also to be able to recall the correct information if the respondents themselves would not be able to. For some, the disclosure process was a task that could actually be completed, while others considered their own acts as dependent on future events which may alter the situation and their approach to finding out their own risk or sharing risk with relatives at some point in the future.

The sub-category *Forwarding info from the cancer genetics unit* give insights on how the offered family letters or other written information is used in practice, either copied and distributed or read directly to relatives. *Designing homemade risk info* highlights how some patients felt a need to take tailored information materials into their own hands by compiling the information they saw suitable and sharing it with at-risk relatives.

3.5. Recognize need for improved support

The fifth and final category describes identified gaps in the current practice from a patient perspective. The sub-category *Lack of information about the process* reflects knowledge gaps related to both personal and family risks, sometimes regarding which relatives are eligible for testing, and confusion about what type of risk the family had. Familial cancer syndromes, where no pathogenic variant has been identified, but there is still a suspected increase in hereditary risk were described in especially contradictory terms.

Some interviewees stressed the fact that better information is not equal to more information. They would rather get the right information adapted to the recipient's understanding and circumstances. Information counselees missed also included insights into the counseling process itself with several patients voicing that it would have been much easier if they had known from the beginning how long the investigation will take, how many relatives would be involved and what would be required of them in terms of informing others.

Uncertainty about healthcare's mandate include statements of insecurity about clinical praxis, legal context, and what is possible for patients to ask their assigned genetic healthcare professional for when they feel unable, or struggle with disclosing risk. Often phrased as a type of question and statement simultaneously, these meaning units seem to include an ambition to confirm one's own understanding of current practice but at the same time being a little perplexed if this is indeed the way "things are done". Other statements display disappointment over the perceived barriers imposed by current legislation. A respondent describes it being a pity we cannot use what "would be the easiest way" when referring to direct disclosure from healthcare professionals by email to at-risk relatives.

The next sub-category is more descriptive of the feelings of perceived personal inadequacy when respondents discussed the role of the informant. In *Limited capacity to inform* respondents pointed out the skill and professional guidance of their genetic counsellor in relation to their own lack of skills. Their concern revolved mainly around not being able to answer relatives' questions and not remembering all the facts they were meant to convey.

Finally, the sub-category *Envisioned support and services* pinpoint several aspects of counseling which respondents felt could be clarified, improved, or developed – mainly concerned what type of support healthcare services should offer. Besides adding the possibility for probands to hand over disclosure to a healthcare professional, some interviewees envisioned signing a contract prior to testing regarding who should inform which relatives given that the test comes back positive. This was seen as a way to share the task of disclosure between the proband and the genetic healthcare professional.

Other respondents elaborated on different types of graphic information they saw would be useful, with "family trees with red lines" or colorful pie-chart diagrams. A few still thought healthcare could simply send brief information with an offer of an appointment to eligible relatives and commented that those interested could simply attend the appointment to find out more.

4. Discussion and conclusion

This qualitative study explores patient experiences of undergoing genetic counseling and handling information about hereditary cancer risk in northern Sweden. Our findings describe a duality in how patients experience the task of communicating risk with their relatives. Respondents place a high value on access to genetic counseling and feel empowered by contact with their genetic healthcare professional. Hereditary risk information was described as both useful and unsettling as their experiences elicited conflicting emotions. Despite this, and the sometimes-challenging family dynamics respondents felt a sense of individual responsibility to reach those at risk, but solicited better guidance and collaborative support by healthcare professionals in that

process.

We identified perceived barriers on several levels as seen in previous research; on individual/personal, institutional/community, and systematic/societal level [2,29]. A novel barrier in our data is the sub-category *Lack of information about the process*. Respondents described the lack of overview of events during counselling and testing as something creating uncertainty. The long timeframe created additional insecurities and undermined their perceived ability to disclose risk information. A related result is the *Uncertainty about healthcare's mandate*, which included expressions of frustration of not knowing what support one can ask for. Patients may find it hard to verbalize this need as it requires display of vulnerability, and the desired support may not even be offered within the current praxis.

Individual barriers to disclosing risk information are well described [17] and in line with our finding *Limited capacity to inform*. This category is multi-faceted encompassing everything from perceived lack of expertise and knowledge to awareness of not having the right tools to deal with strong psychological reactions.

Other challenges when communicating risk information with family relates to negative expectations about familial reactions, assumptions about relatives already being informed, low health-literacy or dysfunctional family dynamics [30,31]. In our data, the sub-category *Respecting autonomy in me and others* underlined the importance of knowing the preferences among relatives. This factor, elsewhere described as "Perceived readiness of relatives" is known to affect disclosure [17], and knowing a relative's preferences can increase the likelihood of risk disclosure [32]. A development in genetic counseling could therefore be to routinely explore patients' family communication culture and discuss how prone family members are to share health-related information. Clarifying the communicative context surrounding the counselee could increase understanding of the proband's capacity to disclose risk effectively and identify necessary support.

The category *Recognize need for improved support* also correlate well with existing research. A recent qualitative study with members from ten European countries identified "Limitations of health care providers" as one of three main themes [19]. Our group also identified the category "Depend on healthcare to take main responsibility" in a focus group study with members of the Swedish public [33]. Those informants conceptualized a "missing link" among the three parties involved in risk disclosure: the healthcare professional, the patient, and the at-risk relative. Studies internationally have also reported a desire for more proactive healthcare-assistance in risk disclosure management [21,34,35] in both patients [36,37], citizens [12] and at-risk relatives [22]

Overall, there is mounting evidence that a more proactive genetic counseling approach may be warranted when discussing disclosure of hereditary cancer risk in families [38]. Previous research on cascade testing have reported high expectations on the support and follow-up of hereditary risk disclosure [33,37], and a recent mixed-methods study concluded that genetic counselors should consider encouraging disclosure besides exploring existing family dynamics in order to offer the right strategies for each family [31]. Our study adds to these conclusions and present some concrete improvements suggested by patients; better tailored information not only about the risk, but also about the process of genetic counseling and testing, options and agreement on who is to approach which relatives as well as offering options on the mode of contact, i.e. by phone, letter or digital communication.

4.1. Theory-related constructs

In a post-analysis mapping against the theory of planned behavior we noted a significant degree of correlation between theory-derived constructs and our sub-categories and categories [39]. "Self-efficacy" plays a central role in several motivational theories and has been shown to be a significant predictor of intentions and motivation to act [39]. Our sub-category of *Limited capacity to inform* reveals an awareness and fear of not having the right skills to perform risk disclosure in a manner that

will be optimal for the relative. This concept is likely very close to that of "self-efficacy" and adds further support of such a factor being instrumental in the decision making involved in risk disclosure.

Two constructs added in an expanded version of the theory, are "self-identity" and "moral norms". The construct "self-identity" can have a significant influence on individuals who identify strongly with a social group [40]. Something we may assume would be the case in the situation of belonging to a family. Our sub-categories *Sharing the norm of the right thing to do* and *No big deal to communicate risk* are probably related to these constructs, as they refer to a common belief system or collectively held convictions in a certain family. These concepts were in our study described by respondents as "we are an outspoken family" or "no, in our family we don't discuss personal health issues".

4.2. Study limitations

This study has a number of limitations. We acknowledge the risk of selection bias as we can presume individuals with stronger opinions or convictions about the topic of risk disclosure may be more prone to participate in the study. Efforts to reduce this risk included a neutrally phrased patient information leaflet with a generally described research aim outlining only that participation would benefit future work to improve cancer genetic risk assessment and care. Respondents were also interviewed at varying times after their genetic testing and genetic counseling. This may be seen as a limitation, possibly introducing recall bias for some, but may also have contributed to diversify the data as respondents had different amounts of time to process and reflect on their experiences.

As participants were recruited at one university hospital our results have a limited transferability to other contexts. However, our group of participants was sampled from the cancer genetic clinic serving almost half of Sweden's geographical area. Adding participants via one of the other specialized clinics may have added more diversity in terms of ethnicity, socioeconomic class and cultural background to our sample. Finally, the cultural context in which this study is conducted may play a role in how communications are viewed, and which moral and societal norms are referred to. Our findings may therefore be valid in a Scandinavian context but may need to be interpreted with caution in countries and societies with significantly different governance, healthcare systems and social welfare structure.

4.3. Conclusions

Our results add new insights into patients' own capacity of handling hereditary risk disclosure as well as unmet needs in this process. Former genetic counselees express a strong respect for their relatives' personal preferences and autonomy in this study. They handled risk disclosure by employing creative strategies to fulfil their part of the information chain. Difficult emotions are a struggle, but not necessarily a barrier to risk disclosure in our data. A close collaboration with and encouragement from healthcare staff was viewed as a significant facilitator for family-mediated risk disclosure.

Our findings also describe how patients co-create solutions to handle risk disclosure in a manner agreeable to themselves and their relatives. Unmet needs concerned better guidance in the genetic counseling process and a more transparent division of responsibilities around sharing risk information with relatives. Finally, more case-tailored, visual and personalized information was desired to allow counselees to easier navigate the complexities around hereditary cancer risk disclosure to at-risk relatives.

4.4. Practice Implications

This study presents a number of opportunities for improving risk disclosure routines more in line with a patient-centered practice. Here we present a number of suggestions extrapolated from our data, which

can be viewed as promising hypothesis in need of future verification.

Promotion of effective risk disclosure should focus on providing accessible hands-on support from genetic health care professionals coupled with understandable and tailored written information. This should include graphics as communication aids when suitable and focus less on plain risk numbers, and more on the options, actions or decisions the counselee may be faced with in the future as well as practical disclosure recommendations. Written information templates for each patient group is a good starting point allowing for additional tailoring by adding family- or individual-specific messages on which relatives may be at risk and what part of the family is not concerned with the reported genetic results.

A clear improvement aiming at reducing uncertainty and ambiguity in risk information would be achieved if healthcare institutions more clearly defined the responsibilities between patients and healthcare regarding the task of informing at-risk relatives beforehand. Another improvement would be to early on outline the counselees expected counselling process. A transparent process enables patients to mentally prepare for each stage of counseling, such as pre-testing information, result disclosure, and future family communication. Patients would more likely feel engaged and have a sense of control amid the often-unexpected situation.

Author contributions

AR and SH conceptualized the study. AR applied for ethical approval, led the recruitment of patients, and funded the data-collection and analysis. CH interviewed patients with assistance of SH. CH coded all transcripts in close collaboration with SH and input from AR. CH headed the data analysis in collaboration with SH. CH drafted the manuscript and was in charge of edits and revisions, SH and AR provided feedback and helped edit the manuscript. All authors read and approved the final draft.

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.

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Human studies and informed consent

The authors confirm all patients' personal identifiers have been removed or disguised so the persons described are not identifiable and cannot be identified through the details of the story. This study was approved by the Regional Ethical Review Board in Umeå, ref no: 2016/345–31 M and 2017–472–32 M. Eligible patients were invited by health care professionals after their last appointment with the cancer genetics clinic. Those with interest of participating were sent a patient information leaflet. Written informed consent was collected for each participant prior to the commencement of the interviews.

Animal studies

Not applicable in this research.

Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at doi:10.1016/j.pec.2024.108319.

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