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Research Article

Dissemination of Genetic Information in Swiss Families with Lynch Syndrome: A Qualitative Exploratory Study

Eléonore Simond[1], Emeline Davoine[2], Maria C. Katapodi[3] and Sheila Unger[2*]

[1]*University of Lausanne, Switzerland*
[2]*Division of Genetic Medicine, Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland*
[3]*Nursing science, Department of Clinical Research, Faculty of Medicine, University of Basel, Switzerland*

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ABSTRACT

In Switzerland, out of respect for privacy and in accordance with federal laws regarding genetic testing, information sharing about hereditary cancer predisposition syndromes is initiated solely by the proband and never from the medical clinic. Thus, an essentially medical task, communication of cancer risk and possible testing, is always delegated, at least initially, to the patient. In order to explore this communication process, its associated difficulties and possibilities for improvements, we have conducted a study with Lynch syndrome families in Western Switzerland. Semi-structured interviews were conducted with 19 participants (12 female, 7 male), either in person or by telephone.

We specifically explored whether participants considered transmission of genetic information a medical or personal responsibility. Other recurrent themes were also identified, including family wisdom and superstitions, emotional responses, and parent-child guilt. The identification of a cancer predisposition and the request to communicate this with family members remained a traumatic experience for many. However, within this group, which may be biased towards better communicators, the information was shared with at risk relatives. Despite inherent difficulties, the majority wish to retain the responsibility for contacting family members. This suggests that in Switzerland, and possibly in other countries with similar rules/attitudes towards privacy, efforts to improve cascade screening should be directed towards facilitating intra-familial communication.

Introduction

Lynch syndrome is the leading cause of hereditary colorectal and endometrial cancers and it accounts for approximately 2% of colorectal cancers [1, 2]. Approximately 1 in 300 people carry a pathogenic mutation in a Lynch syndrome gene [3]. Surveillance guidelines from both American and European organizations recommend colonoscopies every 12 to 24 months and suggest that prophylactic hysterectomy is an option[4, 5]. Lynch syndrome is caused by a heterozygous mutation in one of the mismatch repair genes (*MLH1*, *MSH2*, *PMS2*, *MSH6*), and thus any first degree relative has a 50% probability of having the same mutation. There are three main situations in which an individual might receive genetic counselling pertaining to risk of Lynch syndrome: based on a personal and/or family history of cancer, after detection of microsatellite instability in a tumour, or through cascade genetic testing [6].

In many countries including Switzerland, the latter situation relies on probands as primary communicators with their at-risk family members. This communication strategy has significant limitations in both ensuring contact with the appropriate people and the transmission of accurate information [7, 8]. Moreover, there is limited literature on "sharing

Correspondence to: Sheila Unger, Division of Genetic Medicine, Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland; Tel: +41213143200; Fax: +41213143392; E-mail: sheila.unger@chuv.ch

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results" outside of the research context where dedicated resources and support are included within the framework of the study [8]. This qualitative study aimed to explore intra-familial communication within Lynch syndrome families in a Swiss setting, where federal law clearly states that genetic information belongs to the individual [9]. This means that the responsibility to share genetic test results lies almost exclusively with the proband. Unlike in other countries, such as neighboring France, the proband may not delegate this task to medical professionals. In this context, it was unclear if accurate information was actually transmitted to those at risk, if yes, how effectively it was communicated, and if there were possibilities for improvement.

Methodology

Our research project explored the communication process in general and more specifically to what extent participants considered transmission of genetic information a medical or personal responsibility. We felt the best tool to investigate this was a semi-structured interview format, thus allowing participants to voice experiences and concerns that were not included in our interview guide.

Study Participants and Setting

The study population were known Lynch syndrome carriers identified from the records of the medical genetics department of CHUV. Due to the static nature of the database, no information on the current health status of the contactees was available thus critical illness and/or pregnancy were not de facto exclusion criteria. We identified 86 people who were known mutation carriers and at least 18 years old. They were sent a letter explaining the project and consent forms (available upon request). Those who agreed to participate by return coupon were contacted by telephone or email to set up an interview.

The first author travelled to participants' homes or to a meeting place of their choice for in person interviews while two were conducted by telephone. Both verbal and written consent was obtained to interview participants, audio record and later transcribe the recordings. Notes were taken during the interviews to aid in asking questions and to clarify participants' responses. Participants were invited to share their experience discovering the existence of Lynch syndrome in the family, discovering their mutation status, and the process of sharing this information within their family. Ethics committee approval for this project was given by the Commission Cantonale d'Ethique de la Recherche sur l'Être Humain (CER-VD).

Results

From the 86 identified potential participants, 20 people were interested in the study. From these 20 potential participants, semi-structured interviews were conducted in May 2017 with 17 individuals from 14 different families. One participant failed to return their signed consent form and thus 16 interviews with 19 participants are included in this analysis. Most participants were female (12 /19); ages ranged from 20 to 74 years. All had a germline pathogenic variant associated with Lynch Syndrome detected between 1 to 10 years prior to the study. Nine participants had had one or more cancer diagnoses related to Lynch syndrome, while ten were asymptomatic. In three instances, a parent and their adult offspring (son or daughter) were interviewed simultaneously. Interviews varied in length, between 20 to 80 minutes. Data saturation was achieved after fourteen interviews, in that no new themes were explored by participants thereafter.

We specifically explored whether transmission of genetic information was considered a medical or personal responsibility. Although not included in our interview guide, many participants also discussed their surveillance practices, hopes for the future and other experiences concerning Lynch syndrome such as problems with insurance. Participants were generally well-informed about Lynch syndrome and almost all had ongoing surveillance by colonoscopy at a frequency of varying between 12 to 24 months. As, the goal of the study was to evaluate communication, we did not make a formal assessment of their surveillance program or of their Lynch syndrome knowledge. Thus, varied topics unrelated to the main study aim could not all be included in this manuscript. The following recurrent themes related to family communication were identified: family wisdom and superstitions, emotional responses, and parent-child guilt.

I A Family Matter?

All participants had informed their first-degree relatives of the risk and/or their test result. Informing family members was seen as a family matter by almost all participants (18 of 19). Most felt equipped to do so and believed they had received sufficient information during genetic counseling to do this. Most participants felt that it was primordial for information to circulate within the family but that it remained an individual choice whether or not to be tested. Many participants were also well informed about the mutation status of their relatives.

"They can do what they want with it, if they don't want to get tested, they don't. But at least they've been told, it's better that than the contrary, to not be told and then "oh but if I had known I would have had the test but now it's too late." (female, 63 years old)

Only two participants had a negative reaction from family members they informed.

"I found myself alone with my news, what I wanted to do was effectively pass on the information, but it was made clear to me that they weren't ready, that I shouldn't annoy them with this." (male, 53 years old)

Possible barriers impeding communication were mentioned, such as the passage of time, physical distance and a strong emotional reaction to one's positive test result.

"It's a branch of the family with which we have lost contact so I couldn't go any further." (male, 74 years old)

"If it was a source of anguish or something that really unsettles me maybe I would have a tendency to hide it." (female, 30 years old)

There was no general consensus regarding the utility of supporting documents:

"I don't think that I would need a brochure or something like that, afterwards well I would explain with my own terms and what I know, what I remembered but, and what I've lived through." (female, 30 years old)

A few would have liked to receive additional written material or diagrams to take home.
"The information that was transmitted to me that day […] I think that I had too much, other preoccupations, my life was too dense at the moment where I received this oral information." (female, 40 years old)

When asked what they thought of medical genetics departments sending information letters to at risk individuals to inform them of their Lynch syndrome risk, answers were quite nuanced. Some felt that communication was vital and thus supported direct contact by the medical professional.
"Yes, why not if it can save lives it's clear" (female, 64 years old)

While many others were concerned with the emotional shock of receiving such a letter.
"By letter, terrible. I think that it's violent. […] I'm not convinced." (female, 46 years old)
"It would make me uncomfortable; I find that, I wouldn't like to be told in that way." (male, 74 years old)

A few raised questions about how comprehensible such a letter would be or whether it would violate one's right to privacy. All agreed that a face-to-face discussion with a relative was preferable, for some even if it was a person they disliked.
"Whereas when it's announced by someone you know I think that there, it's, it's, there's no problem, well, very little." (male, 74 years old)

II Family Wisdom and Superstitions

Five different families were aware of their 'bad luck' prior to genetic testing and understood that there was a familial component to the cancer risk.
"And then they had told her no no no no it is never transmissible, my mother would say but really we're an unlucky family" (female, 58 years old)
"We spoke about it sometimes at family reunions because, well the uncles they knew, all those back in the day […] who died pretty young in pain […] but it already existed in the family, simply, they didn't know what it was, there was no name for it" (male, 72 years old)

Even after genetic testing established the diagnosis of Lynch syndrome and following genetic counseling, some families linked Lynch syndrome and/or cancer development to emotions.
"If my brother was the first victim […] it's maybe, he was probably the most vulnerable of us, a musician, an artist, someone pretty sensitive, often anxious" (male, 68 years old)
"…without going into it too much, but I have the feeling that this cancer, carrier, or not a carrier, it begins with emotions." (female, 59 years old)

Four individuals from two families believed cancers grew more slowly with age, and that the risk decreased with age. For others, there seems to be a critical age for cancer development.
"Me it's especially for my daughter that it worries me because well the younger you are the faster it develops, this kind of thing …" (female, 63 years old)
"She died at 46 years old […] so now I have passed this step, then I tell myself that the more time passes the less risk I may have of having it as well" (female, 45 years old)

III Emotional Responses

The identification of a hereditary cancer predisposition and the request to communicate this with family members remains a traumatic experience for many, even many years later.
"I was considerably shocked." (female, 64 years old)
"I can understand one not saying it because it's heavy, because it's not simple, because it's being a carrier of bad news for the whole family and I think that it's a complicated responsibility." (female, 46 years old)

Fear may be a major factor for those who are not getting tested or not following surveillance guidelines.
"We act indifferent, but maybe, behind it all we are maybe a little anxious to know how it might happen." (male, 53 years old)
"I just think he's afraid, but I don't know what of" (female, 30 years old)

Two women had put off genetic testing until they were diagnosed with cancer. One regretted it and one did not.
"For me ([the notion of genetic risk]) was completely abstract" (female, 46 years old)
"My father was positive, the gastroenterologist was encouraging me to get tested I, I put my head in the sand." (female, 61 years old)

In some families, there were taboos that impacted the conversation about Lynch syndrome. Not surprisingly, these concerned death, cancer, and disease.
«There are things unsaid about the dead […] it's incredible, nobody dared to mention him, because mentioning him was mentioning his death, his sickness etc. and it had shocked us so much." (male, 68 years old)
"It's not something we discuss easily actually." (female, 30 years old)

Despite the emotional impact Lynch syndrome had on them, many participants normalized their experience, stating that they were not so different from "normal" people who do not have Lynch syndrome.
"This Lynch syndrome well I live with it it's part of me, like if I don't know I'm tall, I have green eyes, I have Lynch syndrome. It's a genetic characteristic like any other in the end." (male, 53 years old)
"It doesn't keep us from living […] otherwise well, when we don't think about it well, we're really someone like everybody else. Someone normal." (female, 32 years old)

IV Parent-Child Guilt (and Resentment)

Many of the interviewed parents felt guilty about passing on Lynch syndrome to their children.
"Saying well, that I have this crap, alright but that my daughter has it, I find that rather unfair" (male, 53 years old)
"So yes, there is guilt, but it's because you're a parent, before becoming a parent you don't have any." (female, 32 years old)

Several others, however, accepted that genetic transmission was beyond their control.

"In any case, no one can blame anyone, it's random." (female, 63 years old)

All participants agree that while Lynch syndrome is part of decisions regarding childbearing, it did not definitively influence whether or not they chose (or will choose) to have children.
*"But I understand that a young couple is still going to ask themselves the question, but after you'll tell me that there are families with depression, families with, I think that if all these families, then, stop having children there won't be any left. There is a weak spot everywhere." (*female, 58 years old)

Adult offspring rarely expressed resentment towards their Lynch syndrome parent unless their relationship was already strained.
"I phoned my father I told him at any rate, if this is the only thing I've inherited from you, well I was very angry" (female, 64 years old)

For some participants, getting a positive result strengthened their relationship with their affected parent.
"It really made me feel closer to my father actually […] to tell myself well decidedly I have a lot of him in me." (female, 40 years old)
"Now when I tell them ([my children]), you're getting on my nerves, they tell me genetically we belong to you." (female, 58 years old)

Survivor's guilt was common amongst those who were not mutation carriers.
«My brother I think was quite touched because finally it brought him back to his negative test result and, and, and probably a little to this notion of a lottery" (female, 46 years old)

Discussion

Participants in our study were reserved about direct contact between medical professionals and their family members to disseminate awareness of genetic risk. However, even though face-to-face communication by family members was preferred, many participants readily admitted that this was likely not possible for all concerned persons. Participants were globally supportive of communication within the family, as documented in the available literature [10-14]. This is congruent with the joint account model of confidentiality, where genetic information is defined as familial rather than individual, and therefore belonging to all possibly affected relatives [12]. However, outside the nuclear family of first-degree relatives, communication sometimes proved difficult. Our results were aligned with previous quantitative studies; more distant relatives are not always contacted [15].

Despite the small study size, several common barriers to communication were mentioned by various individuals. Given the existence of these barriers, much of the literature concurs that interventions are necessary in order to improve intra-familial communication [6, 15, 16]. Some barriers such as misinformation and poor recall have more obvious and easily implementable solutions, such as web-based information, letters to family, or a high-risk clinic for carriers [7]. Other problems more intrinsic to family life are more difficult to resolve, but simply addressing them during genetic counseling could diminish their impact. Since normalization is a recurrent coping mechanism, this could be promoted during genetic counseling in order to facilitate intra-familial communication, while keeping in mind that the different responses found in our study highlight the individual nature of genetic counselling, encouraging us to tailor the consultation to individual needs.

Several studies found gender differences in communication patterns, but our study did not have the scope to confirm this [7, 8, 13, 14]. These studies suggest that women have traditionally had a responsibility regarding familial health and are therefore more likely to communicate their results. Directly enlisting a female family member when Lynch syndrome is identified in a male proband might serve to improve communication. While this was not the aim of the study, participants spontaneously shared their surveillance experiences, and some of these suggested that, as for hereditary breast and ovarian cancer carriers in Switzerland, Lynch syndrome carriers' surveillance was not always congruent with international guidelines and was not even homogenous within the group [17]. Although no participants expressed a desire to meet Lynch syndrome carriers outside their family, many were interested to hear about how many people we had contacted or were meeting. Additionally, all participants expressed a desire to receive a copy of the study results. However, our group of participants is likely different from those who did not respond to our invitation letter; non-communicators were, by their nature, absent.

Conclusion

In these days of multimedia, people have access to apps, internet videos and more, but in our group face to face communication by family members remained important, maintaining privacy and a personal vibe, when discussing Lynch syndrome results. This implies that strategies designed to improve the efficacy of cascade screening should address good preparation of the proband for their role as a communicator and this may deserve a special training/counselling session distinct from the test result announcement. The study also highlighted the need to communicate accurately with the person at risk as they are the principal source of initial information or misinformation for the extended family. It is clear that many emotions were stirred by this process and remain so for many years regardless of when genetic testing has occurred. This aspect should not be neglected in the genetic counselling session and general practitioners should also be made aware of these long-term implications and lingering issues.

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Conflicts of Interest

The authors declare no conflicts of interest regarding this study.

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