Published in final edited form as:

Patient Educ Couns. 2018 November ; 101(11): 2011–2017. doi:10.1016/j.pec.2018.07.021.

Patterns of family communication and preferred resources for sharing information among families with a Lynch syndrome diagnosis

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Abstract

Objectives.—To explore patterns of communication among families with a Lynch syndrome diagnosis and understand what resources could facilitate family communication.

Methods.—127 probands (i.e., first person in family with identified mutation) and family members participated in semi-structured interviews about: how they learned about the Lynch syndrome diagnosis, with whom they shared genetic test results, confidence in sharing results with other family members, and helpfulness of educational resources.

Results.—Both probands and family members were most likely to share genetic test results with parents and siblings, and least likely to share results with aunts, uncles, and cousins. Most participants felt very confident sharing their test results with family members, but reported that certain topics such as cancer risk were challenging to convey. Probands reported the most helpful

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Declarations of interest: none

Competing interests statement: the authors declare no competing interests

Contributors (roles):

Study design and conception: JP, CK, YW, KS, MK, PK, AS, WK, KK Data collection: JP, MK

Manuscript drafting and revision: JP, CK, YW, RM, AE, KS, LP, WK, KK

Final manuscript approval: JP, CK, YW, RM, AE, KS, MK, LP, PK, AS, WK, KK

Informed consent statement:

I confirm all patient/personal identifiers have been removed or disguised so the patient/person(s) described are not identifiable and cannot be identified through the details of the story.

resources to be access to a specialty clinic or website, while family members described general printed materials as most helpful.

Conclusions.—Families affected by Lynch syndrome may experience barriers to communication with more distant relatives, and may benefit from receiving specific resources (e.g., websites about Lynch syndrome, print materials) to facilitate family communication.

Practice Implications.—Providers could emphasize the need to share information with more distant family members and provide appropriate supportive resources.

1. Introduction

Lynch syndrome is a hereditary cancer syndrome that substantially increases risk of developing colorectal and endometrial cancer, as well as elevating the risk of developing cancer of the stomach, ovaries, urinary tract, brain, and small bowel [1, 2]. Lynch syndrome is caused by a germline pathogenic variant (i.e., disease-associated mutation) in one of four mismatch repair genes: *MLH1*, *MSH2*, *MSH6*, and *PMS2*. Pathogenic variants in *MSH2* and *MLH1* are associated with up to 74% and 54% lifetime risks for colorectal and endometrial cancer, respectively, while *PMS2* and *MSH6* are associated with up to 22% and 26% lifetime risks for colorectal and endometrial cancers, respectively. Approximately 2–4% of colorectal and endometrial cancer cases are caused by Lynch syndrome [3–5].

A diagnosis of Lynch syndrome is vital since colorectal cancer screening practices can then be better targeted. Individuals with Lynch syndrome are recommended to receive colonoscopies more frequently and beginning at an earlier age as well as other cancer screenings [2, 6, 7]. The importance of diagnosing Lynch syndrome prompted recommendations to begin screening all newly diagnosed colorectal and endometrial cancer patients through tumor testing via immunohistochemistry or microsatellite instability [4, 8]. Universal tumor testing identifies nearly twice as many individuals with Lynch syndrome than would be identified by only screening individuals with an early age of onset of cancer [5, 9]. Moreover, several models have demonstrated the cost-effectiveness of universal tumor screening [10, 11]. However, the effects of such approaches on health and cost outcomes depend on how many additional family members receive cascade testing (i.e., testing biological relatives of individuals with an identified pathogenic mutation) [9, 12–14]. Research has shown that cascade testing remains suboptimal; a review by Sharaf and colleagues (2013) noted that first-degree relatives (children, siblings, and parents) of probands (i.e., first person in family found to have a pathogenic Lynch syndrome variant) may accept genetic testing at a rate of 34–52%, and a majority of the time fewer than three first-degree relatives chose to receive testing [15].

Because U.S. health care providers cannot directly contact biological family members, cascade testing is reliant on effective communication about Lynch syndrome between family members to promote genetic testing within families [16]. Previous studies have demonstrated substantial gaps in communication about genetic risk within families. While individuals who have received genetic testing for Lynch syndrome are likely to share their test results with first-degree relatives, they are less likely to share this information with extended family such as second- or third-degree relatives (e.g., aunts, uncles, nieces,

nephews, cousins) [17–20]. Other factors that may influence whether or not genetic information is shared include age of family member and extent of experience with familial cancer [21]. Barriers to sharing genetic information may include a lack of close relationships with more distant relatives, or not wanting to cause worry in family members who may be at risk [17, 18, 21–23]. Individuals may also find sharing information about a Lynch syndrome diagnosis with family members to be burdensome [24]. However, few studies have investigated what resources can support family communication [18, 25]. Additional research is therefore needed to understand family communication patterns and the resources that may best support family communication about Lynch syndrome.

To address these needs, this study therefore aimed to understand communication patterns within families with a diagnosis of Lynch syndrome and determine their preferences for receiving information at the time of diagnosis. We approached this question by investigating how family members first heard of a diagnosis of Lynch syndrome in their family, with which family members individuals shared their genetic test results, and what resources they received and would have preferred to receive at the time of diagnosis. This information is critical to guide approaches to facilitate family communication about Lynch syndrome, and enable healthcare providers to deliver appropriate resources to support family communication and subsequent cascade testing.

2. Methods

2.1. Participants and Procedure

We conducted semi-structured interviews with 127 participants recruited from the Huntsman Cancer Institute (Salt Lake City, Utah) clinical genetics service and registries for patients with confirmed or suspected hereditary cancer, or referred to the study by family members. Individuals were eligible to participate if they were a member of a family with a confirmed pathogenic Lynch syndrome variant. Because of the importance of communicating both positive and negative genetic test results within the family to inform clinical care, individuals could have tested positive (i.e., carried variant) or negative for the familial pathogenic variant, or not had genetic testing. Participants were 18 years or older, and were able to speak and understand English.

Individuals were initially notified of the study by letter. Individuals who did not respond received two follow-up phone calls. Invitation letters were mailed to 298 individuals, and 127 enrolled (43%). Reasons for not participating were: unable to contact (n=143, 84%), declined (n=18, 11%), deceased (n=6, 4%), and ineligible (n=4, 2%). Of those contacted and eligible, 88% agreed to participate and were scheduled for an interview. Participants were classified as:

- Probands (n=32): first person in the family found to have a pathogenic Lynch syndrome variant or
- Family members (n=95): Members of families with Lynch syndrome other than probands, regardless of positive, negative, or untested status

The semi-structured interviews were conducted by telephone and digitally recorded. All participants completed the informed consent process by telephone. Each interview lasted approximately 30–45 minutes. Separate interview protocols were used for probands and family members (see Supplementary materials for interview questions); most questions were similar and differences are noted below. Participant answers were entered into an online survey tool by the interviewer during the interview. In this analysis, we examined responses to open-ended questions designed to generate categorical data (e.g., “What resources were you given?”). For these questions, the study team developed a list of possible answers, and these were built into the survey tool and could be selected if the participant gave one of these answers. An “other” option was available for participant answers that did not match an existing answer; additional categories were added to the a priori categories as appropriate based on the data. A second coder reviewed 20% of the interviews and independently coded responses. Participants who completed an interview received a \$20 gift card. This study was approved by the Institutional Review Board at the University of Utah.

2.2. Measures

Family communication of genetic test results.—To examine family communication about genetic test results related to Lynch syndrome, participants were asked, “Have you told any of the following relatives about your test results?” for the following types of relatives: parents, siblings, children, aunts/uncles, and cousins. They were then asked with open-ended questions why they had or had not shared that information with particular relatives. They were also asked whether they had shared their genetic test results with any other relatives and whether there were any other relatives with whom they had decided not to share results.

Confidence with family communication.—To assess their confidence in communication, participants were asked “How confident are you that you could explain your Lynch syndrome test results to other family members?” and answered on a three-point scale: “Not Confident”, “Somewhat Confident”, or “Very Confident.” They then were asked an open-ended question: “What information regarding Lynch syndrome did you find most challenging to describe and share with your family members?”

Resources received.—To examine resources received at the time participants first learned about a Lynch syndrome diagnosis, we asked open-ended questions: “Were you given any resources such as brochures, letters, or a website to go to for more information?” If yes, “What resources were you given?” We also asked, “Did you search for more information on your own?” If yes, “Where did you search for that information?”

Resources preferred.—To investigate what resources participants would have preferred when learning about Lynch syndrome they were asked: “Tell me whether any of these things may have been helpful when your family was learning about Lynch syndrome” and rated a series of resources on a 5-point Likert-type scale from “Not Helpful” to “Extremely Helpful.” Resources rated were: general printed materials about Lynch syndrome, printed materials specific to the familial mutation, contact information for a genetics specialty clinic, organizing a family meeting, website with general information about Lynch syndrome,

general social media resources such as Facebook or twitter, social media resources specifically for people with Lynch syndrome, support group for people with Lynch syndrome, and access to a specialty Lynch syndrome clinic or specialists knowledgeable about Lynch syndrome (this item only asked of probands). After rating each item, participants were asked, “Of all the resources we just discussed, which do you think would have been the most helpful?”

Demographic and clinical characteristics.—Participants were asked demographic information including age, gender, race/ethnicity, cancer status, marital status, and insurance status. Familial pathogenic variant was determined via medical record.

2.3. Analysis

We examined descriptive statistics for all variables. For variables assessed using closed-ended questions (i.e., family communication of genetic test results, helpfulness, confidence), we compared responses between probands and family members using z tests to compare proportions. For the helpfulness items, the “Very Helpful” and “Extremely Helpful” responses were combined. We examined bivariate associations with mutation status and cancer status; significant associations are presented below. Relatives (e.g., parents, siblings) were excluded from analysis if they were reported to be deceased. Participants without a particular relationship type (e.g., siblings) were excluded from the calculation of family communication for that type (e.g., proportion of siblings told). Analyses were conducted using SAS/STAT 9.4 (Cary, NC). Statistical significance was assessed as $p < 0.05$.

3. Results

3.1 Participant characteristics

As shown in Table 1, the majority of participants in the proband and family member groups were female (78.1% and 67.4%, respectively), Caucasian (93.8% and 94.7%), and married (78.1% and 73.7%). Almost all (90.6%) probands had been diagnosed with cancer, while 23.2% of family members had been. Most probands were over the age of 40 (87.5%), while about half (53.7%) of family members were over age 40.

Of the family members interviewed, 86 (90.5%) had pursued genetic testing for Lynch syndrome themselves; 59.0% received a positive test result and 31.6% received a negative test result. Approximately 9.5% ($n=9$) had not pursued genetic testing. Of all participants, the majority (63.8%) came from a family with a pathogenic mutation in the genes *MLH1* or *MSH2*.

3.2. Family communication patterns

Both probands (96.7%) and family members (87.1%) most often reported sharing their Lynch syndrome genetic test results with a sibling (Figure 1). Test results were often shared with at least one parent (90.0% of probands and 85.9% of family members). While almost all probands (96.2%) reported sharing their test results with at least one child, a significantly smaller proportion of family members (62.0%) had done so ($p=0.003$). The majority of probands reported sharing their test results with more distant relatives; 70% reported

informing at least one aunt or uncle, and at least one cousin of their test results. In contrast, significantly fewer family members reported sharing test results with second- and third-degree relatives, with 34.2% informing at least one aunt or uncle ($p=0.004$) and 35.6% informing at least one cousin ($p=0.001$).

The majority of first-degree relatives to the proband (80.3%) reported being informed about the diagnosis of Lynch syndrome in the family by the proband (Table 2). In contrast, only 28.6% of second-degree relatives to the proband were informed of the Lynch syndrome diagnosis by the proband; the majority (61.9%) were informed by one of their own first-degree relatives instead. Although the number of third-degree relatives (e.g., cousins) interviewed was small ($n=5$), these participants reported being told by various family members included the proband ($n=1$), a different first-degree relative ($n=2$), or a different second- or third-degree relative ($n=2$).

We also examined participants' reasons for sharing or not sharing their genetic test results with family members. The most common reasons why participants chose to share information about their genetic test results were to receive support, knowing that their relatives may be at risk, and knowing that their relatives may want testing. The most commonly reported reasons for not sharing genetic test results included not having a close relationship or being estranged, believing that the relative had already heard the information from another relative, and, in the case of children, believing that they are too young to hear the information.

3.3. Confidence and challenges in sharing Lynch syndrome test results within families

When asked how confident they felt about sharing their Lynch syndrome test results with other family members, 75% of probands and 59% of family members reported feeling very confident; this difference was not significant. We found that participants with a positive genetic test result were significantly more confident in sharing their test results compared to those with a negative result ($p<0.001$). Two topics were identified by both probands and family members as being most challenging in family communication about Lynch syndrome: details of genetic results and associated cancer risks.

3.4. Resources received and preferred during disclosure of a Lynch syndrome diagnosis.

75.0% of probands and 50.5% of family members reported receiving an informational resource from any source when first learning about Lynch syndrome. A brochure/pamphlet (50.0% probands, 26.3% family members) or website (28.1% probands, 15.8% family members) were most commonly received. 18.8% of probands received a letter while 11.6% of family members did. Some probands (9.4%) and family members (2.1%) also received a referral to a specialist. In addition, 71.9% of probands and 50.5% of family members reported searching for more information themselves after first hearing about their own or a family member's diagnosis of Lynch syndrome. The internet was the resource most commonly utilized by people who looked for more information themselves, with 98.6% of these participants choosing to research Lynch syndrome online. Other sources utilized by family members during information seeking included a Lynch syndrome specialist (2.1%), primary care provider (2.1%), and magazine (1%).

When asked to rate the helpfulness of various resources when first learning about Lynch syndrome, 87.5% of probands and 72.6% of family members said that a website with general information would be very or extremely helpful (Figure 2). Most probands and family members also reported that general printed materials about Lynch syndrome (81.2% and 65.3%, respectively), specific printed materials about the mutation found in the family (78.1% and 71.6%), and contact information for a genetic specialty clinic (81.2% and 70.5%) would be very or extremely helpful. In contrast, fewer probands and family members thought that general social media resources (34.4% and 26.3%, respectively) or Lynch syndrome specific social media resources (46.9% and 41.1%) would be very or extremely helpful. We found no significant differences between probands and family members on ratings of helpfulness of resources.

When asked to identify which resource they thought would have been the most helpful when first learning about Lynch syndrome, probands most commonly reported that a website with general information about Lynch syndrome (28.1%) and access to a specialty Lynch syndrome clinic or specialists knowledgeable about Lynch syndrome (31.3%) would be most helpful (Table 3). In contrast, family members reported that general print materials with information about Lynch syndrome (30.2%) and organizing a family meeting (18.6%) would be most helpful. Few probands or family members reported that general social media resources (3.1% and 2.3%, respectively) or a support group for people with Lynch syndrome (3.1% and 2.3%) would have been the most helpful resource when first learning about Lynch syndrome.

4. Discussion and Conclusion

4.1 Discussion

In this study of 127 members of Lynch syndrome families, we found that participants most often shared their test results with first-degree family members such as siblings, children, and parents, and less often reported sharing their results with more distant family members such as aunts, uncles, and cousins. This is consistent with previous studies on family communication about Lynch syndrome genetic test results, which showed that individuals share more information with first-degree relatives [18–20]. Our findings also extend the existing literature in several ways. Because we were able to enroll both probands and family members, we were able to compare their patterns of family communication. We found greater sharing of test results with second- and third-degree family members by probands compared with family members. In addition, our results showed that second-degree relatives of a proband were more likely to hear about Lynch syndrome from one of their own first-degree relatives rather than from the proband themselves. These findings therefore indicate that genetic information most often travels from first-degree relative to first-degree relative in moving from a proband to more distant relatives.

We found several barriers that may prevent individuals from sharing information with more distant relatives. Participants reported that lack of a close relationship was a factor, which is consistent with prior studies [17, 18, 21]. In our study, participants also reported not sharing information about their genetic test results if they believed that a relative had already heard about Lynch syndrome from other family members. However, test results for each family

member are important to share since the pattern of results may direct cascade testing and determine the appropriate individuals to test [14]. For example, testing would only be indicated for a proband's cousin if the second-degree relative connecting them to the proband had tested positive for the family variant, was deceased prior to testing, or refused testing.

Despite these barriers, a majority of both probands and family members reported feeling very confident that they could communicate their Lynch syndrome test results to other family members. The greater proportion who reported being very confident among probands may be because disclosure of a diagnosis of Lynch syndrome to a proband by a genetic counselor includes discussion of the importance of notifying at-risk family members of the diagnosis and strategies for passing on the information. In addition, the probands were all positive for a pathogenic mutation while family members were not. Our data also suggests that individuals with negative results are less confident in sharing test results about Lynch syndrome. We found that both probands and family members identified details of genetic test results and associated cancer risks as the most challenging topics to communicate about with family members. Educational and communication interventions may be needed to address how to share this information, as both of these topics are critical in order for family members to make informed decisions about genetic testing as well as cancer screenings for themselves.

This study also aimed to understand the resources that are most helpful to individuals when first learning about Lynch syndrome in order to support family communication and cascade testing, an area in which little prior research has been conducted [18, 25, 26]. The resources that probands found to be the most helpful were access to a specialty Lynch syndrome clinic and a website with general information about Lynch syndrome. In contrast, family members identified the most helpful resources as general print materials about Lynch syndrome and organizing a family meeting. These differences may be due to the different experiences of probands and family members. While almost all of the probands had a personal history of cancer, only about one quarter of the family members did. A personal history of cancer may have led probands to prefer access to clinicians who are experienced in treating cancer, while family members instead may feel a stronger need for a family meeting to discuss implications for the family and their risk of having Lynch syndrome or developing cancer. Family members may also not know where to seek additional information if they have not been connected with a specialty healthcare professional previously, so a family meeting may seem like an accessible means to gain additional information. In addition, probands by definition had received a positive genetic test result, while only about half of family members had.

When probands and family members searched for more information about Lynch syndrome on their own, almost all chose to use the internet to learn more. While the internet was used to seek information, however, neither general social media nor Lynch syndrome specific social media were considered to be helpful by many probands or family members. This suggests that while technology is considered to be a useful source of information about Lynch syndrome, individuals consider some sources to be more helpful than others and distinguish between websites and social media. These findings may have been impacted by

age, as the majority of participants in this study were 40 years of age or older. Social media may become a more popular resource in the future, as younger age cohorts are greater users of social media [27].

The limitations of this study should be considered in interpreting the results. Although we were able to recruit a large sample of probands diagnosed with Lynch syndrome and family members, the majority were Caucasian and associated with an academic cancer center. Responses may differ in other populations. In addition, family members who were not reached, as well as those individuals who were not willing to participate in a telephone interview, may have different family communication patterns or informational needs. Because we did not have data on the total size of the family network, we were not able to examine the proportion of family members who learned about the Lynch syndrome diagnosis. We also were not able to capture the content of family communication about Lynch syndrome, which is important in future research to develop interventions to support such communication. To reduce participant burden, we examined communication with different types of family members but did not collect data on with how many relatives of each type the results were communicated or barriers to communication for each individual relative (e.g., age, estranged relationship). Social network analysis may be useful in addressing these issues. Finally, perceptions of the helpfulness of different resources may be different than cognitive and behavioral responses to resources.

4.2. Conclusion

Though individuals frequently communicate Lynch syndrome genetic test results with their first-degree family members, there may be barriers to sharing this information with second and third-degree relatives. Probands described the most helpful resources when first learning about Lynch syndrome as websites with general information about Lynch syndrome and access to a specialty clinic or specialists knowledgeable about the condition. Family members reported general print materials and organizing family meetings as the most helpful resources.

4.3. Practice Implications

While family communication between first-degree relatives happens frequently, communication with second and third-degree relatives remains sub-optimal. Healthcare providers involved with genetic testing could stress the importance of communicating test results to more distant relatives, even if they do not have a close relationship. In addition, educational resources [28, 29] and support from multidisciplinary teams could be provided to facilitate the flow of information between different relatives.

The majority of probands, although only about half of family members, reported receiving a resource when they were first learning about Lynch syndrome. The majority of participants in both groups reported that resources such as general websites, general print materials about Lynch syndrome, materials about a specific mutation, and contact for a specialty clinic would be helpful to receive when first learning about Lynch syndrome. Practitioners may consider offering at least one of these resources to probands in addition to standard of care, which is often a family letter. They may also consider giving resources to probands to pass

on to family members, such as print materials that can be easily shared. Materials that emphasize details about the genetic mutation and associated cancer risks may be particularly important to facilitate family communication. System-level approaches to enhance family communication (e.g., specialty clinics, support organizations) that are financially sustainable are also needed. Cost-effectiveness data that examine the impact of such approaches on family communication and cascade testing may provide support. In addition, with the accessibility and popularity of Lynch syndrome information on the internet, providers may direct patients to informative websites. As not all online resources provide reliable information, providers may play a role in helping patients identify how and where to find reputable sources. By providing supportive resources, providers may therefore facilitate family communication about Lynch syndrome.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgements

The study team would like to thank the individuals who participated in this study for their time and their stories.

Funding:

This work was supported by the Enhancing Best Practices through Education, Surveillance, and Policy grant awarded to the Utah Department of Health by the Centers for Disease Control, the Huntsman Cancer Foundation, the Colon Cancer Disease-Oriented Research Team at Huntsman Cancer Institute, and the National Cancer Institute of the National Institutes of Health under Award Number P30CA042014, which supports the Genetic Counseling, Biostatistics, and Research Informatics Shared Resources which were used for this project, and Award Number K07CA196985 (YW). This study was also supported by grant 8UL1TR000105 (formerly UL1RR025764) from NCATS/NIH. The content is solely the responsibility of the authors and does not necessarily represent the official views of the NIH.

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Highlights

- Individuals most often share Lynch syndrome test results with first-degree relatives
- Details of genetic results and associated cancer risks most challenging topics
- Probands and family members find different resources most helpful for learning
- Providers may distribute resources to encourage family communication

Figure 1.
Disclosure of Lynch syndrome genetic test results to relatives of probands and family members. *** $p < .001$

Figure 2.

Resources rated as very or extremely helpful when first learning about Lynch syndrome. a.

Access to specialty clinic option was only asked of probands.

Table 1.**Participant characteristics.**

Characteristic	Probands		Family Members	
	n	%	n	%
Gender				
Male	7	21.9	31	32.6
Female	25	78.1	64	67.4
Race/Ethnicity				
Non-Hispanic, Caucasian	30	93.8	90	94.7
Other	2	6.3	5	5.3
Age				
<40 years of age	4	12.5	43	45.3
40 years of age	28	87.5	51	53.7
missing	0	0.0	1	1.1
Cancer status				
Prior cancer	29	90.6	22	23.2
No prior cancer	3	9.4	73	76.8
Gene with pathogenic variant in family				
<i>MLH1</i>	6	18.8	24	25.3
<i>MSH2</i>	10	31.3	41	43.2
<i>MSH6</i>	10	31.3	9	9.5
<i>PMS2</i>	6	18.8	21	22.1
Personal pathogenic variant status				
Positive	32	100.0	56	59.0
Negative	0	0.0	30	31.6
Not tested	0	0.0	9	9.5
Marital status				
Married/living as married	25	78.1	70	73.7
Widowed/divorced/other	6	18.8	12	12.6
Single	1	3.1	13	13.7
Insurance status				
Private	17	53.1	63	66.3
Public	15	46.9	29	30.5
No insurance	0	0.0	3	3.2

Table 2.

Disclosure of Lynch syndrome test results to family members.

Relationship to the Proband	Informed by proband	Informed by a different FDR	Informed by other SDR/TDR	Informed by healthcare provider
FDR (n=61)	49 (80.3%)	9 (14.8%)	0 (0.0%)	3 (4.9%)
SDR (n=21)	6 (28.6%)	13 (61.9%)	1 (4.8%)	1 (4.8%)
TDR (n=5)	1 (20%)	2 (40%)	2 (40%)	0 (0.0%)

Table 3.

Resources identified as most helpful when first learning about Lynch syndrome.

Resource	Proband (n=32) (%)	Family Member (n=95) (%)
General printed materials about Lynch syndrome	9.4	30.2
Organizing a family meeting	0	18.6
Printed materials specifically about the genetic mutation found in your family	0	15.1
A website with general information about Lynch syndrome	28.1	10.5
Social media resources specifically for people with Lynch syndrome, i.e a Facebook page for people with Lynch syndrome	3.1	10.5
Contact information for a genetics specialty clinic	6.3	8.1
General social media resources such as Facebook or Twitter	3.1	2.3
A support group for people with Lynch syndrome	3.1	2.3
Access to a specialty Lynch syndrome clinic or specialists knowledgeable about Lynch syndrome ^a	31.3	0

^a.This option was only asked of probands.