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Gynecologic cancer screening and communication with health care providers in women with Lynch syndrome

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Abstract

We evaluated knowledge of gynecologic cancer screening recommendations, screening behaviors, and communication with providers among women with Lynch syndrome (LS). Women aged ≥25 years who were at risk for LS-associated cancers completed a semi-structured interview and a questionnaire. Of 74 participants (mean age 40 years), 61% knew the appropriate age to begin screening, 75–80% correctly identified the recommended screening frequency, and 84% reported no previous screening endometrial biopsy. Women initiated discussions with their providers about their LS cancer risks, but many used nonspecific terms or relied on family history. Most were not offered high-risk screening options. While many women were aware of risk-appropriate LS screening guidelines, adherence was suboptimal. Improving communication between women and their providers regarding LS-related gynecologic cancer risk and screening options may help improve adherence.

Keywords

endometrial cancer; Lynch syndrome; mixed methods; ovarian cancer; patient; physician communication

Few studies have examined adherence to gynecologic screening, knowledge of screening recommendations, and understanding of gynecologic cancer risks among women with Lynch syndrome (LS). LS mutation carriers may not correctly estimate their personal cancer risks and may not universally adopt gynecologic cancer screening recommendations, which are

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Conflict of interest
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based on expert consensus and not empirical data (1-3). Reductions in cancer incidence and improvements in survivorship could be achieved if health care providers implemented the current and best practices for LS medical management (Table 1) (3-6).

To identify and manage individuals with LS, providers should be knowledgeable about the characteristics of LS, obtain updated medical and family histories, initiate referrals to genetic counseling and testing services, and recommend appropriate medical management (3, 7-9). However, 65% of obstetricians/gynecologists have never received formal training in DNA-based genetic testing (10) and do not feel confident in their knowledge of genetics (11). LS-affected individuals have identified the perceived knowledge of providers as a key factor in determining appropriate medical management, and those who did not perceive their providers as knowledgeable about LS were less likely to engage in appropriate screening (12). LS mutation carriers placed more *trust* in providers familiar with their family history and more *distrust* in those not aware that cancer age of onset is a determinant of screening recommendations (12). When LS-affected individuals believe their providers are uninformed about LS, they may choose to seek health information on their own and share this knowledge with the providers (12).

Using a mixed-methods approach, we evaluated the prevalence of gynecologic cancer screening among women with LS, their knowledge of LS risk and screening recommendations, and their perceptions regarding communication about LS with providers. As a reference point, colorectal cancer (CRC) knowledge and screening also are evaluated.

Materials and methods

Participants

This study was approved by The University of Texas MD Anderson Cancer Center (MDACC) Institutional Review Board. Participants (n =74) were recruited through the MDACC LS registry, MDACC physician referral, or self-referral through family members. Women aged ≥25 years who had not previously had a gynecologic cancer diagnosis were eligible if they were a LS mutation carrier (n =59) or met the revised Amsterdam II criteria (n =15). All participants had undergone genetic counseling for LS risk with a licensed genetic counselor (n =63), which included information on gynecologic screening recommendations, and/or had learned about their LS risk from a family member (n =9) prior to participating in this study. Screening tests were not provided as part of this study.

Data collection

Women completed an audio-taped semi-structured interview. Participants were asked how they had discussed their family's cancer history and/or LS with providers, and to identify who initiated these discussions, the reasons for initiation, and the gynecologic cancer screening recommendations made by the providers. Study participants also completed a questionnaire that assessed personal and medical characteristics, knowledge of LS screening recommendations, and perceived cancer risk. Participants were asked if they had ever had an endometrial biopsy, transvaginal ultrasound (TVU) or colonoscopy and, if so, when it was

performed and whether it had been through routine care or participation in a research protocol.

Data analysis

To evaluate participants' knowledge, risk perception, and screening behaviors, we computed descriptive statistics using SPSS. Participants were classified as adherent or non-adherent to endometrial, ovarian, and CRC screening based on whether they had received their most current test within a time interval consistent with LS recommendations (3). Participants were further stratified based on whether they had received these screenings through routine care or research participation. Chi-squares were computed to examine whether women were more likely to be adherent to screening through routine care or research. For each cancer risk, women were classified as having accurate, over-, or under-estimations of risk (3).

To evaluate patient-provider communication responses, we qualitatively analyzed transcribed interviews using a grounded theory approach (13). Using an interactive coding process, two coders (AB-C and SRH) independently developed and refined a list of major themes that emerged from the data on a subset of interview transcripts. This coding scheme was then applied to all transcripts by the coders, allowing for expansion and further refinement of the coding structure. The coders had ≥80% agreement on all data and met to resolve differences and to identify major themes.

Results

Seventy-eight women met eligibility criteria and were invited to participate; four did not respond or declined, resulting in a 95% response rate. Participant characteristics, LS knowledge, and adherence are reported in Tables 2 and 3. There were no differences in these variables between individuals with a confirmed LS gene mutation and those who met the Amsterdam II criteria. The majority of participants knew the recommended age to begin gynecologic screening and screening frequency and had accurate perceptions of their colorectal and endometrial cancer risks. Those adherent to endometrial biopsy and TVU recommendations were more likely to have received screening in a research protocol offering no-cost screening versus routine care ($p<0.05$). Those adherent to colonoscopy were more likely to have undergone screening through routine clinical care ($p<0.05$).

Qualitative data showed that participants discussed LS risk and screening with up to 1–5 providers, representing 12 specialties. Over half discussed LS or their family cancer history with a gynecologist/gynecologic oncologist, most discussed this information with a primary care provider, and almost half discussed this with a gastroenterologist. A minority of women reported they either had not discussed LS or family history with a provider or had minimal discussion and did not discuss gynecologic cancer risk. One participant said, '[My doctor] recommended that I have a colonoscopy, but we've never actually had any chance to, like, sit down and figure out what to do next'.

For most participants, the primary reason for initiating discussions with providers about LS or family history was to increase provider awareness of their cancer risk. Patients often viewed these discussions as their responsibility. While women reported various reasons for

introducing the topic, about half did so to make providers more aware of their family history or genetic testing results, or to seek information and advice regarding risk and appropriate screening. One participant initiated the discussion ‘ … so [my doctor would] be sure that I got a good check-up for cancer’ while another did so to ensure the provider did not ‘let me slack off’ on screening. Some women initiated discussions of LS risk after a provider’s request for family health information, while others brought it up to obtain referrals, participate in research, report symptoms, or share updated family history information.

For some women, decisions to discuss LS or family cancer history depended on the type of provider or the circumstances regarding their visit. One woman said: ‘My decision to bring it up would be based on how important I feel it is based on the condition I am being treated for’. Another said: ‘I would bring it up to the extent that I thought that it was necessary to the scope of appointment’.

Among women who had discussed LS with providers, most used nonspecific terms or reported only their family cancer history, using phrases such as ‘I have a cancer gene’ or ‘I have a family history of colon cancer’. Only about one fourth directly told providers of their LS mutation status, and about half relied on their providers to recommend screening based on personal or family cancer history (primarily CRC). One participant stated: ‘I don’t tell [my doctors] it’s [LS], I tell them about my family history for CRC’. For the minority who directly communicated their LS status, many brought copies of their genetic test results or other paperwork indicating the high-risk screening guidelines.

About half of participants reported that their providers did not inform them about high-risk gynecologic cancer screening recommendations. One woman noted: ‘They don’t know a lot about [LS]. None of the doctors I spoke to knew more than I did’. When some providers were aware of the family history, women reported that many did not consistently acknowledge or recognize an increased cancer risk. One woman noted: ‘My [gynecologist] noticed my family history … but did not register that it implied risk for me’. About one tenth of women reported that their providers did not know about LS and the appropriate screening guidelines or did not indicate concern when the topic was introduced. One woman said: ‘The last time I got a colonoscopy, the [doctor] told me that he never heard of such a thing about getting [endometrial or ovarian cancer] screenings on an annual basis’.

More than half who discussed LS or cancer family history with internists or family practitioners did not recall receiving LS-specific gynecologic screening recommendations, although some received referrals to gynecologists or other specialists to discuss gynecologic screening. About one third of women recalled their providers recommending the need for high-risk screening; however, these recommendations were not uniformly consistent with LS screening guidelines. One woman stated: ‘They [her doctors] would start watching for it [gynecologic cancer] as I turn to 40 or so. I am 37’. In terms of surgical risk-reduction options, only a few women reported having discussed total abdominal hysterectomy with bilateral saphingo-oophorectomy (TAHBSO) with their gynecologists, and it was presented as an option after menopause or childbearing. As one woman said: ‘My doctors are encouraging me to have a [TAHBSO] when I’m done having kids’.

Discussion

Women in our study were knowledgeable about their risks for LS-associated cancers and the recommended frequency of cancer screening, but this knowledge was not entirely reflected in their self-reported screening behaviors. Almost half of participants were non-adherent to LS screening recommendations for gynecologic cancers, and nearly one fourth were non-adherent to CRC screening recommendations. In this study, women may not have sought or been offered gynecologic cancer screening for LS during routine care. Among those adherent to screening recommendations, many underwent screening as part of research protocols, suggesting that research participation presents an opportunity to reinforce the importance of continued cancer surveillance.

Although participants did not express concerns about the overall care they received from providers, a minority had received risk-appropriate recommendations about gynecologic cancer screening. This finding suggests that awareness of LS-associated cancer risks and management guidelines can be improved among providers, which is consistent with other studies that have shown limitations in providers' knowledge of the management of hereditary cancer syndromes (10, 11, 14). Given that physician recommendation is a consistent predictor of adherence to cancer screening behaviors (15, 16), continued efforts to educate providers about cancer screening recommendations for high-risk patients are very important.

Persons with LS may be willing to take an active role in communicating personal and family risk to their providers (12), but our results showed that women often did not effectively convey essential details about the condition. This may limit providers' ability to comprehensively assess cancer risks and provide management recommendations. Our findings suggest that women may perceive that providers have a greater understanding of LS than they actually do or assume that providing information about their family cancer history is adequate to convey LS risk status. In the absence of an integrated health care system with universal access to medical records, it is mainly the responsibility of patients to effectively communicate or inform providers regarding their personal and familial health risks and conditions. Thus, patients may benefit from interventions aimed at improving information provision so that they are able to effectively communicate hereditary cancer risk information to their providers (17).

This study provided important insights regarding awareness of gynecologic cancer risks and adherence to screening among women with LS, and identified challenges posed in communicating risk information to providers. However, some study limitations should be noted. This study was conducted with a small sample in a large comprehensive cancer center setting, which may limit generalizability and may not reflect the experience of patients seen in settings outside of such centers. The date of the genetic counseling session and socioeconomic variables other than education were not collected, therefore we were unable to examine the relationship between these variables and our study measures. Screening behaviors were self-reported, although studies have supported the validity of self-reported screening information (18). Data on the efficacy of gynecologic cancer screening tests for

women with LS also are limited, which may affect providers' willingness to make screening recommendations.

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Table 1

Colorectal and gynecologic medical management recommendations for at-risk members of families with Lynch syndrome[a]

Type of intervention	Recommendation	Quality of evidence
Colorectal cancer Screening		
Colonoscopy	Annual or biennial beginning at 20–25 years old or 10 years younger than the youngest age at diagnosis in the family, whichever comes first	Evidence includes consistent results from well-designed, well-conducted studies in representative populations that directly assess effects on health outcomes
History and examination with detailed review of systems, education, and counseling regarding LS	Annual beginning at 21 years old	Evidence is insufficient to assess the effects on health outcomes
Prophylactic surgery		
Colorectal resection	For at-risk persons without a previous diagnosis of CRC: generally not recommended, discuss as alternative to regular colonoscopy, with preferences for well-informed patient actively elicited	Evidence is insufficient to assess the effects on health outcomes
	For persons with a diagnosed CRC or polyp not resectable by colonoscopy, subtotal colectomy favored with preferences of well-informed patient actively elicited	
Gynecologic cancer *Screening*		
Endometrial biopsy	Annual beginning at 30–35 years old	Evidence is insufficient to assess the effects on health outcomes
Transvaginal ultrasound	Annual beginning at 30–35 years old	Evidence is insufficient to assess the effects on health outcomes
History and examination with detailed review of systems, education, and counseling regarding LS	Annual beginning at 21 years old	Evidence is insufficient to assess the effects on health outcomes
Prophylactic surgery		
Hysterectomy or oophorectomy	Discuss as option after childbearing is complete	Good–fair

[a] Adapted from Lindor et al. (3).

Table 2

Participants' demographic and medical history characteristics (*n*=74)

Characteristic	% (*n*)
Mean age (SD, range), years	40 (8.7, 25–64)
Race, White	92% (68)
Education, college graduate or higher	62% (46)
Married	72% (53)
≥1 child	77% (57)
Prior colon cancer diagnosis or polyps	41% (30)
Family history of any gynecologic cancer	64% (47)
LS mutation carrier	78% (59)
Prior LS genetic counseling	85% (63)

Table 3

Participants' adherence to and knowledge of LS screening recommendations and perceived lifetime cancer risks

	Non-adherent % (*n*)	Adherent % (*n*)	
Screening test		**Through research**	**Through routine care**
Endometrial biopsy[a]	48 (32)	42 (28)**	10 (7)**
Transvaginal ultrasound[b]	49 (33)	31 (21)*	21 (14)*
Colonoscopy[c]	24 (13)	26 (14)*	51 (28)*
Screening recommendations (*n*=74)			Correct % (*n*)
Age range to begin gynecologic screening			61 (45)
Recommended screening frequency for:			81 (60)
Endometrial cancer			
Ovarian cancer			74 (55)
Colorectal cancer			82 (61)
Perceived lifetime cancer risks (*n*=74)			
	Under-estimation % (*n*)	Over-estimation % (*n*)	Accurate estimation % (*n*)
Endometrial cancer[d]	8 (6)	30 (22)	53 (39)
Ovarian cancer[d]	3 (2)	89 (66)	7 (5)
Colorectal cancer[d]	19 (14)	20 (15)	61 (45)

[a] *n*=67; excludes women who had a hysterectomy (with or without oophorectomy).

[b] *n*=68; excludes women who had an oophorectomy (with or without hysterectomy).

[c] *n*=55; excludes women with a prior CRC diagnosis.

[d] Assessment of correct lifetime cancer risk estimates were based on the data provided during the genetic counseling session (3).

* $p>0.05$ ($\chi^2 = 16.15$, $p<0.0001$).

** $p>0.0001$.