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Cancer screening behaviors and risk perceptions among family members of colorectal cancer patients with unexplained mismatch repair deficiency

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Abstract

Communication gaps in families with unexplained mismatch repair (MMR) deficiency (UMMRD) could negatively impact the screening behaviors of relatives of individual with UMMRD. We evaluated cancer risk perception, screening behaviors, and family communication among relatives of colorectal cancer (CRC) patients with UMMRD. Fifty-one family members of 17 probands with UMMRD completed a questionnaire about cancer risk perception, adherence to Lynch syndrome (LS) screening recommendations, and communication with relatives. Clinical data about the probands were obtained from medical records. Thirty-eight participants (78%) were worried from having cancer and twenty-one participants (42%) had undergone colonoscopy in the past 2 years, as recommended for LS families. In terms of screening for extracolonic cancers, only two eligible participants (3.9%) were screened for gastric, endometrial (10.0%), and ovarian (9.5%) cancers. Additionally, 5 participants (10%) underwent genetic counseling. Most participants were not told by anyone to be screened for extracolonic cancers (84, 85, and 95% for gastric, ovarian, and endometrial cancers, respectively). A minority of family members of CRC patients with UMMRD follow cancer screening as recommended for LS families. Health care providers should encourage patients with UMMRD to share information on LS-related cancers screening, especially extracolonic cancers, with their relatives.

Keywords

Colorectal cancer; HNPCC; Mismatch repair; Lynch syndrome; Screening

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Conflict of interest The authors declare that they have no conflict of interest.

Introduction

A cancer diagnosis impacts the quality of life for patients as well as their family members. First-degree relatives (FDRs) of colorectal cancer (CRC) patients have an increased risk for developing CRC themselves, and are advised to follow high-risk CRC screening recommendations [1]. Adherence to screening recommendations is particularly important among families with hereditary cancer syndromes, as cancer tends to cluster within several generations of these families.

Lynch syndrome (LS), also called hereditary non polyposis colon cancer (HNPCC), is an autosomal dominant syndrome caused by germline mutations in one of the mismatch repair (MMR) genes, namely *MLH1*, *MSH2*, *MSH6*, *PMS2*, or in the *EPCAM* gene [2, 3]. Persons with LS are predisposed to CRC and extracolonic malignancies, often at a young age [2–4].

Individuals with LS and their affected family members are advised to following screening and preventive measures [5–7], including: (1) colonoscopy every 1–2 years beginning at age 25 or 2–5 years before the youngest age of CRC diagnosis in the family; (2) annual endometrial sampling and transvaginal ultrasound (TVUS) or serum CA-125 [5] *may be considered* for women, and prophylactic hysterectomy and bilateral oophorectomy among women who have completed childbearing *should be considered*; (3) esophagogastroduodenoscopy (EGD) with extended duodenoscopy every 3–5 years [5] or every 2–3 years [7] beginning at age 30–35 years *may be considered* for selected individuals or families, and (4) annual urinalysis beginning at 25–30 *should be considered*.

Up to 70% of patients with CRC or other LS-related tumors that manifest high microsatellite instability (MSI-H) without *MLH1* promoter hypermethylation, and/or show absence of immunohistochemistry staining of MMR proteins, do not have pathogenic germline mutations or deletions in MMR or EPCAM genes. This group of patients has been classified as having as “Lynch-like syndrome (LLS)” or “tumor Lynch” [8, 9]. According to current literature these patients either have an undetected MMR mutation (LS with missed mutation), false positive screening or double somatic mutations (the last two groups are not caused by germline mutation and therefore are not part of the LS spectrum) [9–14]. Since the term LLS is misleading, we will refer to this group of patients with MMR deficiency but without detected germline mutations as “unexplained MMR deficiency (UMMRD)”. Clinically, UMMRD falls in the spectrum midway between LS and sporadic CRC [9]. Patients with LLS/UMMRD have a lower standardized incidence ratio of CRC compared to LS patients. However, the average age of onset of CRC in these patients is similar to that of LS patients (53.7 years for LLS compared with 48.5 years for LS) but is lower than that for sporadic CRC (68.8 years) [8]. Patients with LLS/UMMRD also are less likely to have synchronous or metachronous LS-associated cancer compared with LS patients (7% in LLS vs. 38% in LS) [15].

According to the National Comprehensive Cancer Network (NCCN) guidelines, no consensus has been reached regarding screening of UMMRD patients, and genetic testing of the corresponding gene could be performed on tumor DNA to assess for somatic mutations, although the efficacy of this method has not yet been proven. Individuals found to have

double somatic mutations/changes in the MMR genes likely do not have LS and management should be based on personal/family history [5]. The United States Multi-Society Task Force on CRC (USMSTF) guidelines recommends that LLS/UMMRD patients *and their relatives* continue LS surveillance [7]. However, no data exist on the screening and surveillance *behaviors* of relatives of individuals with UMMRD.

Since no LS-related mutation can be found in UMMRD patients, relatives of persons with UMMRD are not advised to undergo genetic testing. Therefore, these relatives rely on the proband to communicate cancer risk information about UMMRD and cancer screening recommendations. Prior research in a LS population indicates that there is incomplete communication among patients and their family members in terms of cancer risk and screening recommendations. The lack of communication seen in individuals with LS also may be found in families with UMMRD. Although most LS affected patients share their genetic test results with FDRs, the communication of the results to more distant relatives occur less often [16]. Additionally, the absence of a mutation is negatively associated with communication with distant relatives about genetic counseling and risk for LS [16]. Patients without identified mutations communicated LS genetic counseling and risk information to a smaller number of family members, compared to LS patients with a mutation [17]. It is possible that these communication gaps in families without a known mutation could negatively impact the screening behavior of relatives of individual with UMMRD, however, to our knowledge, this has never been studied. To begin to build the literature in this area, this study examines perceived risk and screening behaviors among family members of CRC survivors with UMMRD.

Materials and methods

Participants and data collection

This study was approved by the Institutional Review Board at The University of Texas MD Anderson Cancer Center (MD Anderson). CRC patients with UMMRD age 18 years or older were recruited between October 2013 and March 2014 from MD Anderson. Patients were mailed a questionnaire and were asked to provide names and contact information for their adult (age 18 and older) first-, second- and third-degree family members so that they also could be invited to participate in the study. All patients underwent genetic counseling. They were recommended to refer their relatives to genetic counseling and to explain them about LS and LLS. Thirty-four patients completed and returned the questionnaire; of those, 17 (50% response rate) provided names and contact information for a total of 104 relatives.

We sent a mailed questionnaire to all 104 relatives. We called non-respondents by phone 3 weeks after the initial mailing; those who were reached were given the option to complete the questionnaire by phone or to have a replacement questionnaire mailed to them. Those who did not respond at 3 and 6 weeks after the initial mailing received another phone call with the option to complete the questionnaire over the phone.

Study measures

Demographic information included age, sex, marital status, number of children, ethnicity, education level, employment, annual income and financial situation. MD Anderson medical records and registry data were used to obtain *medical data* related to the relative with CRC (proband).

Family cancer history was assessed by asking participants to report all known cancers in the family. They also were asked whether they had discussed cancer screening with their family members.

Cancer risk and cancer screening perceptions were assessed using 22 items that we developed for this study which asked participants about the importance of CRC, endometrial cancer (EC), ovarian cancer (OC), and gastric cancer (GC) screening. Responses ranged from not at all to very important. Participants also were asked whether they believed they needed to be screened on the basis of family history or their relative's genetic test results. Participants were asked whether they had been told to be screened and by whom.

Screening adherence was assessed using 18 items that we developed for this study which asked participants about the use of colonoscopy, EGD, endometrial biopsy, and TVUS within the past 6 months, 1, 2 and 3 years or more before the survey (or never). Participants also were asked why they had genetic counseling and testing.

Health insurance coverage was assessed using 7 items that we developed for this study which asked participants about having a health insurance and its coverage of the procedures included in the study (colonoscopy, endometrial biopsy, TVUS, UGI endoscopy, genetic counseling and genetic testing), their preferences about coverage of colonoscopy (billing the insurance or paying out of pocket), and denial by the insurance of screening colonoscopy.

Cancer worry was assessed using three items from the Lerman cancer worry scale [18]. These items measured participants' worries about being diagnosed with cancer. Responses ranged from not at all worried to worried almost all the time. Responses were dichotomized as "worried" or "not worried".

Statistical analysis

Primary outcomes were cancer screening behaviors for CRC, EC, OC, and GC as well as having genetic counseling. Secondary outcomes were need and importance of being screened for the above cancers, and perceived risk of and worry about being diagnosed with cancer. Summary statistics were calculated for continuous variables; frequencies were calculated for categorical variables. Exploratory association tests were conducted using the Fisher exact test to compare 2 categorical variables, the Kruskal–Wallis test to compare categorical and continuous variables, and Spearman correlation to compare 2 continuous variables. $p < .05$ was considered statistically significant. Stata v13.1 software (StataCorp LP, College Station, TX) was used for all analyses. We considered both random-effects model and generalized estimating equation (GEE) to account for possible correlations among participants from the same family. However, due to small sample size, these approaches failed to converge for the majority of outcome variables. For a few variables that

the model converged, the estimate of correlation was low and the results were very similar to those assuming independence, suggesting that the within-family correlation may be negligible. Based on these considerations, our analysis assumed independence to obtain more reliable estimates.

Results

A total of 51 family members (49% response rate) from 17 probands responded to our survey. Thirty-seven were FDRs, 7 were second-degree relatives (SDRs), and 7 were third-degree relatives (TDRs). Participants' demographic characteristics are summarized in Table 1.

Thirty-two participants out of 49 who reported having a family member with CRC (65%) sought information for cancer screening after hearing about the CRC diagnosis in their family. FDRs and SDRs asked for screening information more than TDRs ($p = .003$). The same number of participants (32) reported having ever heard about LS, most of them FDRs of CRC patients with UMMRD ($p = .038$). Only 9 participants (18%) were referred for genetic counseling and the same number was referred for genetic testing. Only 5 of the 51 (10%) actually underwent genetic counseling.

Cancer risk perceptions and worry

Most participants estimated their risk of developing CRC to be higher than people their age, based on family history (64%) and their relative's genetic test results (58%). Females ($p = .025$), participants with a personal history of any cancer ($p = .026$), and those who were familiar with the term LS ($p = .04$) estimated their CRC risk to be higher based on their family history. Being a relative of a proband with a synchronous tumor also was associated with higher estimated CRC risk ($p = .008$). Thirty-eight (78%) participants were worried about being diagnosed with cancer. This affected the mood of only 6 (12%) participants and only 2 (4%) participants reported that this impacted their ability to perform daily activities.

Screening behavior

Colorectal cancer screening—Forty-five participants (92%) indicated that screening for CRC is somewhat or very important for them, however only 21 (42%) had undergone colonoscopy in the past 2 years (Table 2). Eighteen participants (35%) believed they should have a colonoscopy every 1–2 years, as recommended for LS families. Being female ($p = .011$), having a higher education ($p = .002$), having heard about LS ($p = .009$), and having a relative with a synchronous tumor were associated with believing in the importance of CRC screening ($p = .022$); however, no association was found between these parameters and adherence to CRC screening recommendations. Not having children ($p = .017$), having a job ($p = .012$), and left-sided cancer and rectal cancer in the proband ($p = .015$) were negatively associated with colonoscopy completion in the last 2 years. The most common reasons that participants cited for undergoing colonoscopy were increased risk in the family (35%) or physician recommendation (25%). Of note, 14 participants out of 36 who had had a colonoscopy (39%) were found to have polyps.

Gastric cancer screening—Thirty-six (71%) of our participants have never had an EGD. Among 15 participants who underwent the procedure, 11 (73%) stated that the reason was problems or symptoms and 2 underwent EGD as part of routine cancer screening or because of increased risk of cancer in the family.

Gynecological cancer screening—Seven female participants underwent hysterectomy, oophorectomy, or both. One participant was diagnosed with cancer and one more had it prophylactically because of LS in her family. Only 1 female participant (5%) had been recommended for EC screening, and 3 (14%) for OC screening. Four participants ever underwent endometrial biopsy. Two of them (10%) had undergone this screening procedure in the last year—both were FDRs of CRC patients with UMMRD ($p = .03$). No other factors were associated with having an endometrial biopsy. The most common reason for not undergoing endometrial biopsy during the past 2 years was a lack of information about screening (84.2%).

Ten women (48%) underwent TVUS, 2 of them (10%) during the last year, and only one as part of routine cancer screening.

Table 2 summarizes participants' beliefs about cancer screening and their screening behavior. We found statistically significant differences in beliefs about the importance of screening by cancer type ($p = .001$). Recommendation to be screened also differed by cancer type ($p < .001$), as well as screening behaviors ($p = .001$).

Health insurance

Forty-eight participants (94%) had health insurance. Including what is paid by the insurance 78% of our participants could afford colonoscopy every 1–2 years and 57–59% could afford endometrial biopsy, TVUS and UGI endoscopy. Forty-two participants (84%) would choose to bill their insurance for colonoscopy and only 2 patients have been denied coverage for screening colonoscopy by their insurance. Most participants (82, 84%) did not know if genetic counseling and genetic testing, respectively, are covered benefits by their health insurance. Screening beliefs and behavior were not associated with any health insurance related statement.

Cancer screening discussion in the family

Table 3 summarizes cancer screening discussion in the families. Almost all participants discussed cancer screening with their siblings (97%, $N = 36$). More than half discussed it with their parents or children, and less than half discussed it with their SDRs and TDRs.

Discussion

UMMRD (previously called LLS) is a recently-used definition for patients who present with MMR-deficient tumors, but do not have identifiable mutations upon germline sequencing. UMMRD comprises patients with both hereditary and sporadic cancers [9–14]. The only way to distinguish between these two groups is by sequencing the tumor's DNA MMR genes, which is still not routinely done. As a result, providing screening recommendations to cancer patients with UMMRD and their family members is often challenging both for the

health care providers and for the patients and families. Some guidelines still recommend that UMMRD patients and their relatives be screened as if they have LS [7], while the latest guidelines emphasize the importance of somatic DNA testing (although its efficacy has not yet been proven) since growing evidence suggests that the majority of UMMRD patients have double somatic mutations/changes in the MMR genes [5]. Previous studies have demonstrated that many physicians do not always make the correct recommendations even for patients with a clear LS diagnosis [19–22]; therefore it is not surprising that our study shows that most relatives of UMMRD patients were not told by anyone to be screened for cancers other than CRC (84, 85 and 95% for GC, OC and EC, respectively). Additionally, our data showed that although family members of patients with UMMRD are worried about developing cancer, perceive LS-related cancer screening to be important, and discuss cancer screening with their FDRs, only 40% of them follow LS screening recommendation for CRC, and 9–12% for extracolonic cancers. This is particularly surprising given that almost 70% of our participants perceived their risk of developing CRC to be higher or much higher than other people their age based on their family history. This rate is concordant with the 62% previously described by Grover et al. [23] among individuals who met the Revised Bethesda Guidelines and had an indeterminate genetic test result; however, it is lower than the 90% who perceived high or very high risk for having CRC among patients with pathogenic mutation in the MMR genes.

Adherence to screening guidelines is crucial for both the probands and their family members. Hence it becomes imperative for physicians and genetic counselors to discuss screening recommendations with probands and their family members [20]. It is equally important for index patients to share information about their cancer diagnosis and genetic test results with their family members and encourage them to get screened. According to the literature, family members believe that genetic information should be shared among families. In a study that described LS family members' perceptions about sharing genetic information, the majority of family members indicated that everyone in the family has the right to learn about a cancer diagnosis and the presence of a mutation in the family [24].

In most cases, it is the responsibility of the probands to share information about their cancer diagnosis and advise family members about screening recommendations. We have previously shown that most CRC patients with UMMRD adhere to LS screening recommendations (94, 86, 71 and 56% for CRC, EC, OC and GC, respectively) and inform their relatives about the CRC diagnosis as well as advise them to undergo CRC screening; however, only 50% advise female relatives to be screened for endometrial cancer and only one-third advise relatives to have genetic counseling [19]. Therefore, it is expected that most of our participants did report a discussion about cancer screening within their families; however, the low adherence rate to screening recommendations among our participants is surprising (42% for CRC and 10, 9.5 and 11.7% for EC, OC and GC, respectively).

One explanation for these low screening rates is a lack of understanding regarding the meaning of the genetic test results. In our prior study, only 41% of patients correctly interpreted their genetic tests [19]. In the current study only nine participants (18%) were referred for genetic counseling and only 5 (10%) actually had it. It is possible that the lack of knowledge by the proband about whether his/her UMMRD diagnosis has a genetic basis

can lead to underestimation of the need for routine enhanced screening among relatives. Therefore, it is important for the probands with UMMRD to have appropriate genetic counseling.

Appropriate screening of UMMRD families is still an unresolved issue, therefore it is understandable that, despite the existence of formal screening recommendations, not all physicians emphasize the importance of family member screening to their patients. As a result, this information may not be accurately shared among family members. The relatives, who are not familiar with UMMRD and with the higher risk of cancer, may contribute to the low rate of screening recommendation adherence as well. Hence, it is important that health care providers educate and disseminate accurate information of genetic test results, cancer risk, and appropriate screenings to both probands and their relatives and encourage their patients to share this information with their family members.

In our previous study we have shown that all UMMRD participants with CRC ($n = 34$) reported their cancer diagnosis to at least 1 relative, and only one patient did not inform his relative about genetic test results [19]. According to the probands' responses, only 2% of participants' FDRs were not informed of the CRC diagnosis; in contrast, about 30% of SDRs and 40% of TDRs were not informed. Regarding screening recommendations, according to the probands' reports, 74% of their living relatives were recommended to undergo screening for CRC. Our results, taken from a selected group of these relatives, indicate that indeed 96% of them knew they had a relative with CRC and 75% were recommended to undergo CRC screening; however, while 50% of the probands reported that they recommended EC screening to their female family members and 33% advised all of their relatives to receive genetic counseling, only one of our female participants reported that someone had told her to be screened and <20% of all relatives were referred for genetic counseling. Therefore, while disclosing a diagnosis of UMMRD to family members, index patients should emphasize not only CRC, but also extracolonic cancers and genetic counseling [25]. Additionally, involving a health care professional in this process may be beneficial to everyone.

The major strength of our study is its innovation. It is the first study reporting screening behavior of family members of UMMRD patients. Our response rate of 49% among the family members is considerably high, reflecting the motivation of the relatives to participate in the study. We were able to assess risk perception and cancer screening behaviors in this population of relatives of cancer patients with UMMRD; we also were able to assess communication barriers within these families. Many studies have looked at these parameters in sporadic CRC population and in LS cohorts, but none in UMMRD families.

Our study also has some limitations. It is based on a limited number of participants and does not directly compare screening behavior between UMMRD and LS families, or between cancer patients with UMMRD and their relatives. As with any survey-based study, information bias and self-reported data are common limitations. Since the included participants were recruited through their relatives with CRC, our results may overemphasize the rate of screening adherence in the "real world."

Taken together, our results indicate that family members of CRC patients with UMMRD are worried about their familial cancer risk; however, less than half underwent CRC screening as recommended for LS families, and only a minority were screened for other LS-associated cancers or participated in genetic counseling. A better understanding of the genetic basis and natural history of UMMRD is needed to modify screening recommendations for families with UMMRD. However, until the genetic basis of UMMRD is established, physicians and genetic counselors should help patients to better understand the meaning of this diagnosis and its implication on their families, while also encouraging patients with UMMRD to share information on LS-related cancer screening, especially extracolonic cancers, with their relatives.

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Table 1

Participant demographic characteristics (n = 51)

Characteristic	N (%)
Mean age (SD, range)	50 years (15.9, 18–82)
Female	28 (55)
White	50 (98)
Married	40 (78)
Education > High School	40 (77)
Income >\$50 K	36 (75)
Currently Employed	33 (65)
Diagnosed with any cancer	11 (22)

Table 2

Screening behavior of relatives of UMMRD patients

Item	Response	% (n)				<i>p</i> value
		OC ^b	EC ^b	UGI ^a	CRC ^a	
Importance of screening	Somewhat + very important	71.4 (15)	75.0 (15) ^d	57.1 (28)	91.8 (45)	0.001
Believing that should be screened based on family history	Yes	28.6 (6)	40 (8) ^d	25.5 (13)	NA	0.457
Told by someone to be screened	Yes	14.3 (3)	4.8 (1)	15.7 (8)	75.5 (37) ^c	<0.001
Time of last screening test ^e	<i>e</i>	9.5 (2)	10.0 (2) ^d	11.8 (6)	42.0 (21)	0.001

^a n = 51 (eligible patients)^b n = 21 (eligible patients)^c For any type of cancer (after realizing that relative was diagnosed with CRC)^d n = 20 (answered these questions)^e For colonoscopy—within the last 2 years; for EGD—within the last 3 years; for endometrial biopsy—within the last year; for TVUS—within the last year

Table 3

Cancer information sharing with family members (FDR/SDR/TDR)

Characteristics	N (%)
<i>Have you discussed cancer screening with your FDRs?</i>	
Parents	19 (79)
Siblings	36 (97)
Children 18+	18 (86)
Children <18 years	10 (56)
<i>Have you discussed cancer screening with your SDRs and TDRs?</i>	
Grandparents	4 (26)
Grandchildren >18 years	2 (40)
Uncles/Aunts	13 (39)
Niece/Nephews	15 (48)
Cousins	16 (44)
Heard about Lynch syndrome	32 (63)
Referred for genetic counseling	9 (18)
Had genetic counseling	5 (56)

FDR First degree relative, *SDR* Second degree relative, *TDR* Third degree relative