

Communication of Lynch Syndrome Genetic Test Results:  
Strategies to Facilitate Information Sharing with At-risk Family Members

Master's Thesis

Presented to

The Faculty of the Graduate School of Arts and Sciences  
Brandeis University  
Department of Biological Sciences – Genetic Counseling  
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by  
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## **ABSTRACT**

### **Communication of Lynch Syndrome Genetic Test Results: Strategies to Facilitate Information Sharing with At-risk Family Members**

A thesis presented to the Department of Biological Sciences – Genetic Counseling

Graduate School of Arts and Sciences  
Brandeis University  
Waltham, Massachusetts

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Lynch syndrome is an autosomal dominant cancer disorder responsible for 2-3% of all diagnosed colorectal cancers. Currently, clinical genetic testing can identify a carrier of Lynch syndrome through a mutation in their MMR genes (MLH1, MSH2, MSH6 and PMS2). As Lynch syndrome is a hereditary syndrome, the knowledge that a family member has a gene mutation in one of the MMR genes may impact an individual's health and lifestyle. Previous studies have shown that strategies and interventions to help reduce the barriers to dissemination of genetic test results to at-risk relatives are needed. The aim of this study was to ascertain what strategies and instruments genetic counselors identify as the most useful in helping patients share their positive Lynch syndrome MMR mutation genetic test results with at-risk family members, as well as help inform the family members of their potential risks. We recruited cancer genetic counselors who had counseled patients with Lynch syndrome

within the past five years through the National Society of Genetic Counselors listserv and the NSGC Familial Cancer Risk Special Interest Group listserv to complete an anonymous, online survey. Of the 142 survey respondents, 97.6% reported that their patients expressed interest in sharing information about their diagnosis with their at-risk family members. Among the approaches, strategies and/or materials used, genetic counselors reported that a letter from a genetic counselor to a patient (98.2%) was the most used tool, while an institution's online resource (9.6%) was the least used tool. Overall, most genetic counselors (88%) were satisfied with the tools they currently use. Additionally, genetic counselors reported that the tools they use are both effective and helpful to patients. Future research should look more carefully at patients' perceptions of tools that were successful at helping them share their positive Lynch syndrome genetic test results with at-risk family members.

Keywords: Lynch syndrome, HNPCC, colon cancer, communication, family, genetic testing, hereditary, genetic counseling

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## INTRODUCTION

### *Disease Background*

Lynch syndrome, also known as hereditary nonpolyposis colorectal cancer (HNPCC), is an autosomal dominant cancer disorder responsible for 2-3% of all diagnosed colorectal cancers (CRC) (Stoffel et al., 2008). Individuals diagnosed with Lynch syndrome are predisposed to early onset cancers, including colon, endometrial, ovary, hepatobiliary, ureter, small intestine, kidney and brain cancer (Hadley et al., 2008). In women with Lynch syndrome, up to 60% will present with endometrial cancer (Lynch et al., 2009). Mutations found in four mismatch repair (MMR) genes, most commonly MLH1 and MSH2, are known to cause Lynch syndrome and can be detected through genetic testing (Lynch et al., 2009).

A person who inherits a mutation in one of the MMR genes can have up to a 90% lifetime risk of developing one of the Lynch-associated cancers (Hadley et al., 2008). Due to this cancer predisposition, people with Lynch syndrome are recommended to follow specific cancer screening guidelines, such as more frequent colonoscopies (Mesters et al., 2005). As the average age of onset is 45 years of age, colonoscopies should begin at age 25 and be repeated every 1-2 years until age 40, when they should be repeated annually (Lynch et al., 2009). The high frequency of synchronous and metachronous CRC in an individual with Lynch may necessitate a subtotal colectomy if cancer does occur (Lynch et al., 2009). Currently, clinical genetic testing can identify a carrier of Lynch syndrome through a mutation in their MMR genes (Hadley et al., 2008).

### *Impact of genetic testing on patients*

Knowing that one has Lynch syndrome enables patients and physicians to tailor medical management to the increased cancer risks. The National Comprehensive Cancer Network (NCCN) and other professional societies publish medical management guidelines for people with Lynch syndrome (National Comprehensive Cancer Network, 2011). Following these guidelines has shown to significantly reduce mortality and morbidity through prevention and early detection of cancer (EGAPP, 2009).

Nevertheless, discovering an individual is at an increased risk for developing cancer can have negative psychological outcomes, especially for those who learn they may have passed on their cancer-predisposition to offspring (Gritz et al., 2005). When investigating the psychological impact of a Lynch syndrome test result on both affected and unaffected patients up to one year after test results were disclosed, Gritz et al. determined that, in general, Lynch syndrome genetic testing does not have long-term adverse psychological effects among affected or unaffected patients (2005). However, they did find that unaffected carriers were found to have a more significant increase in anxiety, depression and high-distress immediately following test disclosure (Gritz et al., 2005). In 2007, Bleiker et al. analyzed the long-term psychosocial impact on patients counseled for Lynch syndrome between 1986 and 1998. The authors concluded that there were only a small group of counseled individuals that reported clinically significant levels of stress or social or family problems on the average of four years following counseling. The authors found it encouraging that most people (~95%) viewed genetic counseling for Lynch syndrome as a positive experience.

In 2009, the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group, a project developed by the National Office of Public Health Genomics at the Centers for Disease Control and Prevention, found that there was adequate evidence to recommend offering genetic testing for a Lynch syndrome mutation to all CRC patients because it has been shown to reduce morbidity and mortality. They also found there was no evidence that identifying Lynch syndrome through the use of routine genetic testing would cause adverse psychosocial outcomes.

In a study to examine the impact of family systems on psychological adjustment in patients undergoing BRCA1/2 or Lynch syndrome testing, family functioning, differentiation to parents, hereditary cancer-related family communication and perceived support from relatives were evaluated (Van Oostrom et al. 2007). Through the use of three mailed questionnaires, the authors found that family system characteristics are important in determining psychological distress levels. For example, it was reported that individuals who were hesitant to discuss hereditary cancer with family members were more likely to suffer from psychological distress after test disclosure. These results emphasize the importance for counselors to convey the need for open communication between patients and their families, especially when informing other at-risk family members.

#### *Motivating influences to family communication*

As Lynch syndrome is hereditary, when a patient receives genetic testing results, not only are they uncovering their own risks but the risks of other family members as well. Due to state and federal laws, such as those under the Health Insurance Portability and Accountability Act of 1996 (HIPAA), healthcare providers by and large cannot

notify at-risk relatives; instead they must rely on the patient to inform at-risk relatives of their genetic test results (Offit et al., 2004).

Since major responsibility for disseminating hereditary cancer genetic test results depends on the diagnosed patient, it is critical to identify the ways in which and to whom the proband communicates about testing results. It has been found that when individuals informed family about their hereditary predisposition for Lynch syndrome, most people restricted their disclosure to the nuclear family (Claes et al., 2003; Mesters et al., 2005). Additionally, research has found that two of the main motivations for disclosure to at-risk relatives was because they felt a moral obligation to do so – or that they might feel regret if something happened that could have been prevented (Aktan-Collan et al., 2010; Mesters et al., 2005).

Moreover, it was found that patients were more likely to inform family if health professionals stressed the importance of disclosing test results to family, if the patients were cancer patients themselves or if there was a strong history of cancer in the family, especially if someone had died of colon cancer (Mesters et al., 2005). Not surprisingly, patients were less likely to inform estranged family members about the hereditary predisposition. Gaff (2005) found similar results that support those found by Mesters et al. (2005), and in addition, observed gender differences; the researchers found that men were in more need of guidance when it came to informing their at-risk relatives.

To learn how family cancer experiences and genetic testing information have influenced their hereditary cancer risk perception, Palmquist et al. (2010) studied three families that each have Lynch syndrome documented by a mutation in a mismatch repair gene. Each family was from a different ethnic group (one African-American, one

Mexican-American and one Caucasian), and family communication about cancer experiences and genetic information was evaluated through communication network assessments and semi-structured interviews. Palmquist et al. (2010) concluded that family communication can influence the perceived risk of hereditary disease as well as views of disease prevention by promoting the exchange of health information and encouraging those who are at high risk to seek out screening or preventative measures. This study emphasizes that individuals often absorb and process information in the context of their family relationships.

#### *Barriers to family communication*

In a recent study, Baer et al. (2010), explored the distribution of familial cancers in the general US population and its relation to awareness of familial cancer risk, referral and use of genetic testing. The authors did a cross-sectional analysis of a national survey (2005 National Health Interview Survey) to assess familial cancer risk of BRCA- and Lynch-associated cancers in the US and their association to genetic testing. They found that of the 5.8 million US residents at high risk for familial cancers, only 50% had ever heard of genetic testing and only 15% of these individuals had ever discussed testing with a physician, with 4.5% receiving genetic testing. This study reveals that, based on estimates of the prevalence of familial cancer, many individuals at increased risk for hereditary cancer are not receiving information on genetic testing.

Informing relatives of their chance of a genetic risk, especially that of a cancer predisposition, can be an emotionally challenging process and upsetting to both the patient and the at-risk family member. Due to the sensitivity and significance of the material, there are many factors that can influence the process and impact of relaying

genetic risk information. In a review of family communication about genetic risk within families, it was found that both individual and relationship factors influenced communication patterns (Wiseman et al., 2010). For instance, patients who had little or no contact or a strained relationship with the at-risk relatives were much less likely to disclose genetic testing results (Chivers Seymour et al., 2010; Wiseman, et al., 2010).

Additional inhibiting influences included the perceived “readiness” of the relative for the information, personal feelings of guilt or anxiety of the patient and concerns of genetic discrimination (Chivers Seymour et al., 2010; Wiseman, et al., 2010). A recent Finnish study of parents with Lynch syndrome and communication of genetic risk with their children found that most parents felt it was their responsibility to tell their family, but expressed the desire to have more supportive involvement from a health professional (Aktan-Collan et al., 2010).

It has been found that, most of the time, individuals with an unambiguous Lynch syndrome genetic test result do in fact inform their at-risk first-degree relatives (FDRs) (Stoffel et al., 2008). However, it is much less common for probands to inform more distant relatives, who are also at increased risk (Stoffel et al., 2008). This lack of communication between the proband and distant relatives may be contributing to the number of at-risk individuals who do not receive genetic testing or counseling. Because genetic testing and counseling for Lynch syndrome has been found to reduce morbidity and mortality through screening compliance, it also implies that those who do not receive genetic testing and counseling are less likely to be following high-risk cancer screening guidelines and, in turn, less likely to prevent future cancers (EGAPP, 2009).

### *Study aims*

More research is necessary to identify strategies and interventions that can help reduce the barriers to dissemination of genetic test results to at-risk relatives. This study explores what factors or strategies facilitate communication between those diagnosed with Lynch syndrome and their at-risk family members. The aim of this study is to ascertain what strategies and instruments genetic counselors find are the most successful at helping patients share their positive Lynch syndrome mismatch repair (MMR) mutation genetic test results with at-risk family members, as well as help inform the family members of their potential risks. Once tactics that help families communicate about their health risks are identified, healthcare professionals – particularly genetic counselors – can implement these strategies, which will better facilitate communication and help improve cancer surveillance for those at-risk family members.



## METHODS

### *Study design*

The study consisted of a survey of genetic counselors that have experience with counseling in the cancer field using an anonymous, online survey (Appendix A). The survey was administered using [www.qualtrics.com](http://www.qualtrics.com), an online survey tool. The majority of the survey was multiple-choice questions: demographic, Likert scale and forced choice. The survey also included some open-ended questions to allow for qualitative data collection.

The survey collected standard demographic data, such as age, sex and race as well as what year respondents graduated from a genetic counseling program. Participants were asked questions about their experiences counseling patients with a molecular Lynch syndrome diagnosis, including their view of patients' willingness to share their genetic testing results with at-risk family members. Participants were also asked about the effectiveness of specific approaches, strategies and materials that could be used to help patients with a positive Lynch syndrome mutation result inform their at-risk family members of their diagnosis and what the diagnosis means for their at-risk family members.

In order to identify the usefulness of specific approaches, strategies and materials, participants were asked if they use the identified approaches, how often and whether they believe that they increase a patient's willingness to inform their at-risk family members of their positive genetic test results. Additionally, participants were asked about their

satisfaction with the approaches and/or materials they use to help patients disseminate information about their positive mutation results to their at-risk family members.

### *Study sample*

The study included genetic counselors who had some experience with cancer risk assessment for Lynch syndrome cases. For inclusion in the study, the participants must be a genetic counselor who has (a) at least six months of cancer genetic counseling experience, either full or part time, within the last five years and (b) counseled a patient with a molecular Lynch syndrome diagnosis (a mutation in one of the four mismatch repair genes known to cause Lynch syndrome) at least once within the past five years. Participants were not excluded based on age, gender, geographic location or other demographic characteristics.

### *Recruitment*

This project received human subjects approval from the Brandeis University Institutional Review Board. Participants were recruited through the National Society of Genetic Counselors (NSGC) main listserv and the NSGC Familial Cancer Risk special interest group (SIG) listserv.

The recruitment notice (Appendix B) contained information regarding the purpose of the study, criteria for participation and contact information for the student researcher and principal investigator. The recruitment notice also explained that participation was anonymous, confidential and voluntary and that participation could be discontinued at any time. A link provided in the notice directed interested participants to the online survey.

Committee member Judith Tsipis posted the recruitment notice on the NSGC main listserv and committee member Ann Jeffers-Brown posted the recruitment notice on the NSGC Familial Cancer Risk SIG listserv inviting genetic counselors to participate in the study. The recruitment notices were posted in early February. Sixteen days after the initial posting, the recruitment notices were re-posted. The survey was open from February 9, 2011 to March 7, 2011 (~3.5 weeks). After completion of the survey, participants were given the option to be entered into a drawing for a gift card to Amazon.com.

#### *Data collection*

Data was collected through an online, anonymous survey hosted by qualtrics.com. At the completion of the data collection period, the data was downloaded into SPSS (PASW Statistics, v18.0, for Mac).

#### *Data analysis*

Quantitative data was processed and analyzed through the data analysis program SPSS to calculate descriptive statistics, correlations, and single sample t tests. We analyzed open-ended questions by using qualitative methods to identify common themes.

## RESULTS

### *Response rate and demographics*

Overall, 156 individuals responded to the anonymous online survey. Of the 156 respondents, 14 data sets were excluded, as the respondents had not practiced cancer genetic counseling, had practiced less than six months and/or had not counseled a patient with a molecular Lynch syndrome diagnosis within the past five years. After these exclusions, 142 (92%) surveys met all study criteria and were included in the final analysis.

Demographic data is summarized in Table 1. The majority of survey participants were female (97.4%), defined themselves as white or Caucasian (91.2%), were currently full time genetic counselors (84.2%) and nearly half have been cancer genetic counselors for the past four to five years (49.3%). Most participants graduated after 1997 (82.2%) and a plurality worked in a university medical center (40.8%). Cancer genetic counselors that work in a health maintenance organization or a diagnostic laboratory were the least represented group in this sample population as compared to genetic counselors from other work settings. The most common role of participants was overwhelmingly a clinical role (93%). Cancer genetic counselors responded from all geographical regions of the United States.

In a one-year period, these cancer genetic counselors saw an average of 19 patients with a *suspected* clinical or molecular diagnosis of Lynch syndrome, with a maximum of 150 patients. In a one-year period, these same cancer genetic counselors

saw an average of 5 patients with a *confirmed* molecular diagnosis of Lynch syndrome, with a maximum of 50 patients.

**Table 1:** Demographic characteristics of cancer genetic counselor survey participants.

Demographic	Total (N)	Mean	Range	Frequency
Age	84	34	24-62	
Gender	114			Females: 97.4% Males: 2.6%
Ethnicity	114			White or Caucasian: 91.2% Asian: 6.1% Other: 1.8% Black or African American: 0.9%
Year of graduation	107		1979-2010	1979-1984: 7.4% 1985-1990: 3.7% 1991-1996: 6.5% 1997-2002: 20.6% 2003-2010: 61.6%
Full time/Part time work	114			Full time: 84.2% Part time: 14.9% Not currently working: 0.9%
Time within the past 5 years practiced cancer genetic counseling	142			Six months to one year: 7% One to two years: 19% Two to three years: 12% Three to four years: 12.7% Four to five years: 49.3%
Region	134			Region I: 11.9% Region II: 24.6% Region III: 7.5% Region IV: 31.3% Region V: 12.7% Region VI: 11.9%
Work setting	142			University Medical Center: 40.8% Private Hospital/Medical Facility: 21.8% Public Hospital/Medical Facility: 20.4% Cancer Institute: 16.2% Physician's Private Practice: 4.2% Health Maintenance Organization: 2.1% Diagnostic Laboratory: 2.1%
Primary role(s)	142			Clinical: 93% Teaching/Education/Supervising Studies: 20.4% Clinical Coordination: 14.1% Research/Study Coordinator: 13.4% Administrative: 12.7% Management: 4.9% Customer Liaison: 2.1% Lab Support: 2.1% Grant Management: 0%
Number of patients seen with a <i>suspected</i> clinical or molecular diagnosis of Lynch syndrome in the past year	127	19	0-150	
Number of patients seen with a <i>confirmed</i> molecular diagnosis of Lynch syndrome in the past year	127	5	0-50	

*Cancer genetic counselor approach to patient disclosure of testing results to family*

In order to assess a cancer genetic counselor's approach to patient disclosure of positive genetic testing results to at-risk family members, participants were asked how often they discuss certain topics during a results-giving session. Participants were given four topics related to Lynch syndrome results disclosure and were asked to answer how often they discussed the topics when informing a patient that they carry a genetic mutation that predisposes them to cancer. The participants answered how often they discussed the given topics using a 5-point Likert scale ranging from "never" to "always." (Table 2) The average answer for all statements was 4.89 (essentially genetic counselors often or always discuss these topics), which indicates that, overall, genetic counselors are likely to discuss with patients that: (a) their at-risk relatives should follow Lynch syndrome screening guidelines; (b) they should consider sharing their test results with their at-risk relatives; (c) sharing their results with at-risk family members could prevent cancer in their at-risk family members; and (d) their at-risk family members should see a genetic counselor.

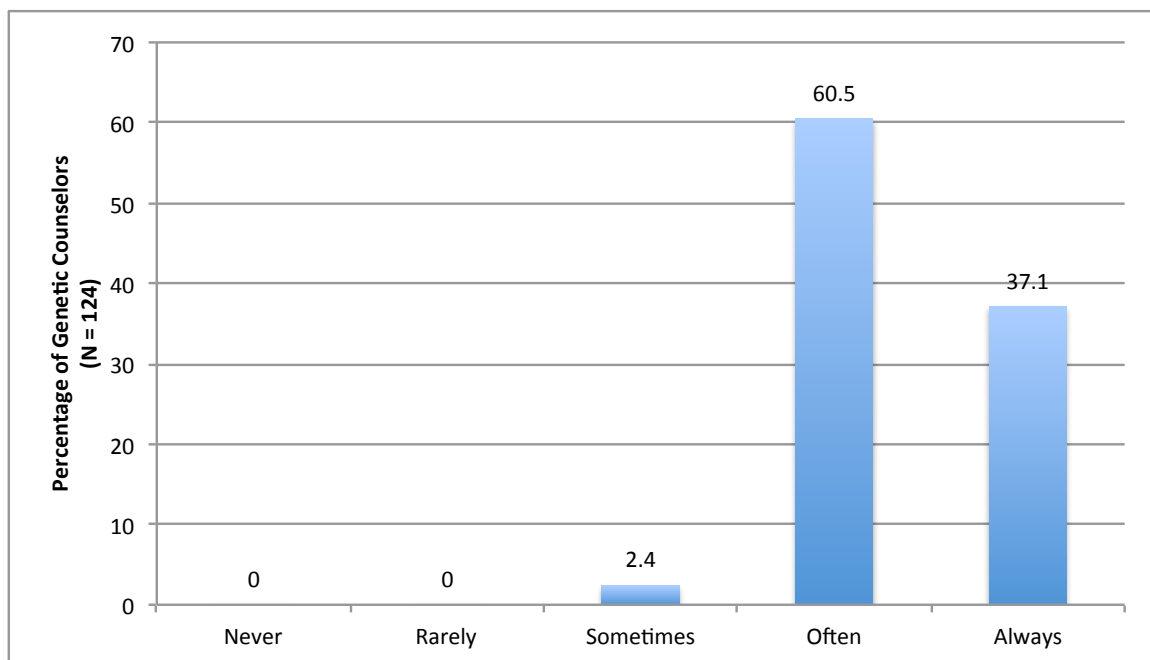
**Table 2:** Discussion topics regarding a cancer genetic counselor's approach to patient disclosure of positive genetic testing results to at-risk family members.

Discussion topic	Mean	SD	Total (N)
Their at-risk relatives should follow Lynch syndrome screening guidelines.	4.82	0.601	125
They should consider sharing their test results with their at-risk relatives.	4.95	0.376	126
Sharing their results with at-risk family members could prevent cancer in their at-risk family members.	4.91	0.284	125
Their at-risk family members should see a genetic counselor.	4.89	0.364	125

Answered on a 5-point Likert scale, with "1" defined as "never" and "5" defined as "always." The average answer (standard deviation and N) to each statement is shown.

*Cancer genetic counselor view of patient interest in sharing information about their diagnosis with at-risk family members*

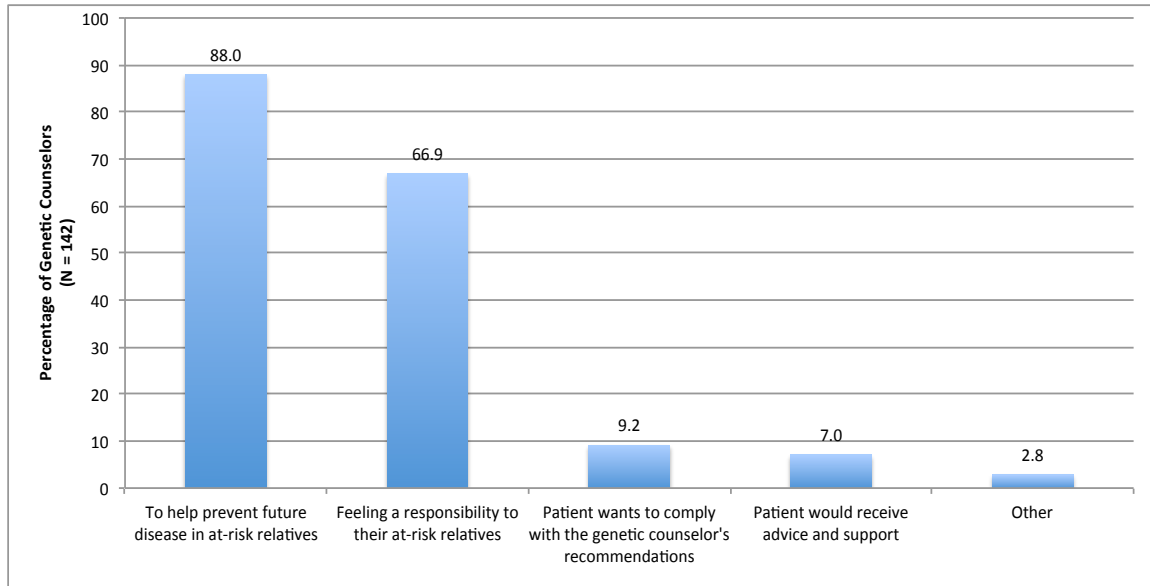
Cancer genetic counselors were asked to report how often patients who have received a molecular diagnosis of Lynch syndrome expressed an interest in sharing information about their diagnosis with their family members. The majority of participants responded that patients often (60.5%) or always (37.1%) express interest in sharing information about their diagnosis to family members (Figure 1).



**Figure 1:** How often patients who have received a molecular diagnosis of Lynch syndrome express interest in sharing information about their diagnosis (and what the diagnosis means for their at-risk family members) with their family members.

Genetic counselors were then asked what were some of the reasons patients gave for wanting to share their positive testing results with an at-risk family member. For those patients who did express interest in sharing information about their diagnosis, genetic counselors noted the most common reasons for sharing were (a) to help prevent future disease in at-risk family members (88%) and (b) that patients felt a responsibility to their at-risk relatives (66.9%) (Figure 2). Some additional reasons genetic counselors

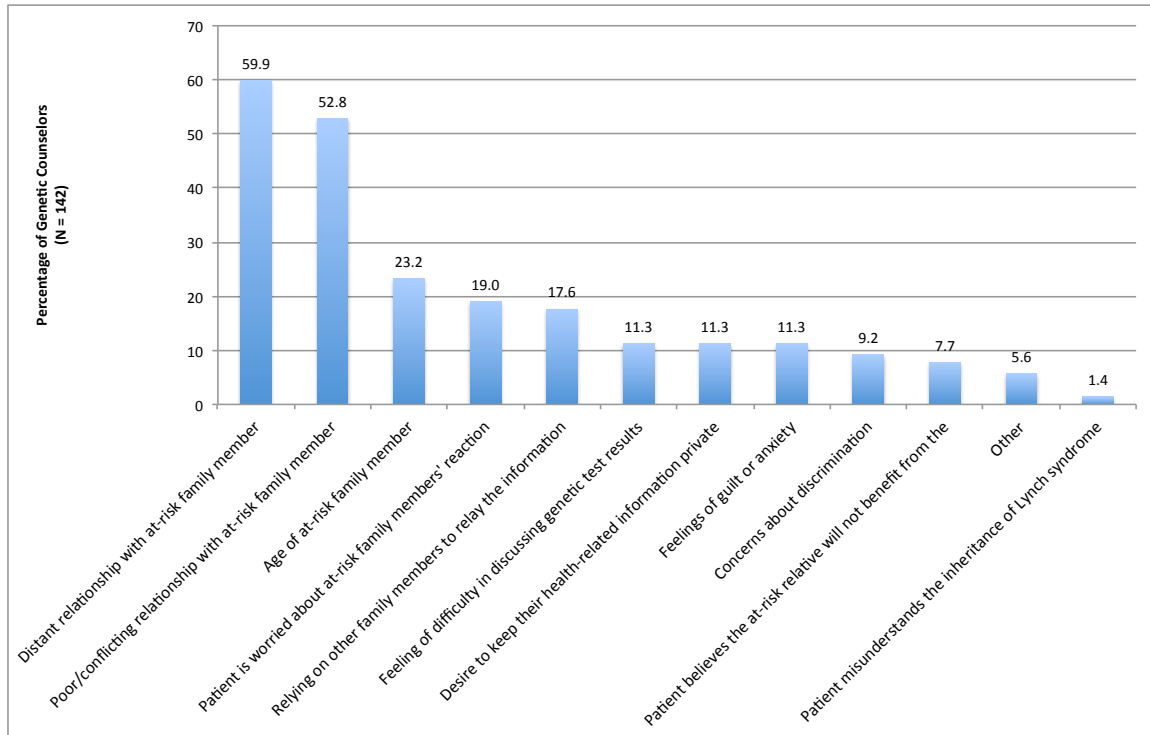
gave that were not listed were: “[Patients] want to see some benefit from their cancer diagnosis, as [a] “silver lining,” and the patients “finally have an explanation for ca[n]cer in [the] family.”



**Figure 2:** Reasons patients give for wanting to share their positive genetic testing results with an at-risk family member. Percents do not add to 100% as respondents could indicate more than one than one reason.

Additionally, participants were asked what were some of the reasons patients gave for *not* wanting to share their positive genetic testing results. Genetic counselors reported that the most common reasons given by patients who did not want to share their diagnosis with an at-risk family member was that they had a distant relationship (59.9%) or a poor/conflicting relationship (52.8%) with the at-risk family member. The least common reason for a patient to be unwilling to share their diagnosis was misunderstanding the inheritance of Lynch syndrome (1.4%). (Figure 3) Other reasons not listed were that the “family member is not insured,” and that the “patient believes relative wouldn't use the information to change their healthcare behaviors.”





**Figure 3:** Reasons patients give for *not* wanting to share their positive genetic testing results with an at-risk family member. Percents do not add to 100% as respondents could indicate more than one than one reason.

### *Tools used to help patients inform their at-risk family members*

Respondents were given eight approaches, strategies and/or materials that could be used to help patients with a positive Lynch syndrome mutation inform their family members of their potential risks. Participants were asked to answer questions about their use of the tools based on their experience (Table 3). Genetic counselors reported that a patient letter (98.2%) was the most used tool while using an online resource designed by the genetic counselor's institution (9.6%) was the least often used tool among respondents.

**Table 3:** Genetic counselors' use of various approaches, strategies and materials to help patients share their positive Lynch syndrome genetic test results with at-risk family members.

Tool	Percent (%)	Frequency (n)	Total (N)
Patient letter	98.2	112	114
Testing lab pamphlet	71.7	81	113
Online educational resource	67.2	80	119
Support group referral	65.8	75	114
Letter addressed to family member	55.3	63	114
Personalized pedigree	54.5	66	121
Role-play	17.8	21	118
Institution's online resource	9.6	11	114

Genetic counselors were then asked, using a 3-point Likert scale ranging from “rarely” to “always,” to rate how often they use specific tools (Table 4). Participants’ indicated that, of the tools used, a patient letter (mean = 2.76, SD = 0.505) was the most often used tool. Role-play (mean = 1.22, SD = 0.475), however, was the tool used the least often, even among those who use role-play from time to time.

**Table 4:** How frequently genetic counselors use the approaches, strategies and materials.

Tool	Mean	SD	Total (N)
Patient letter	2.78	0.479	112
Institution's online resource	2.7	0.483	10
Testing lab pamphlet	2.56	0.548	81
Online educational resource	2.54	0.594	80
Personalized pedigree	2.52	0.588	66
Support group referral	2.27	0.688	74
Letter addressed to family member	2.08	0.685	62
Role-play	1.38	0.59	21

Answered on a 3-point Likert scale, with "1" defined as "rarely" and "3" defined as "always." The average answer (standard deviation and N) to each tool is shown.

A Pearson product-moment correlation coefficient was computed to assess the relationship between the type of tool used and number of patients seen with a suspected clinical or molecular diagnosis of Lynch syndrome, number of patients seen with a confirmed molecular diagnosis of Lynch syndrome and the amount of time within the past five years the counselor has practiced cancer genetic counseling.

There was a negative correlation between use of a pedigree and number of patients seen with a suspected clinical or molecular diagnosis,  $r = -.205$ ,  $n = 120$ ,  $p = 0.025$ . Therefore, as the number of patients seen with a suspected clinical or molecular diagnosis increased, the use of the pedigree tool decreased. There was also a negative correlation between the amount of time within the past five years the counselor has practiced cancer genetic counseling and the use of a Lynch syndrome testing lab pamphlet,  $r = -.200$ ,  $n = 113$ ,  $p = .034$ . So, as the time within the past five years the counselor has practiced cancer genetic counseling increases, the less likely they are to use a testing lab pamphlet.

#### *Effectiveness of tools used to help patients inform their at-risk family members*

Participants were asked if they found the tools they used were effective at helping patients to understand who in their family was at risk of inheriting Lynch syndrome. All of the respondents ( $N = 11$ ) who used their institution's online resource (100%) reported they found this tool helpful (Table 5). Overall, each tool was found to be helpful by at least 63% of the respondents.

**Table 5:** Percent of genetic counselors who use a particular tool and find it helps a patient understand who in their family is at-risk of inheriting Lynch syndrome.

<b>Tool</b>	<b>Percent (%)</b>	<b>Total (N)</b>
Institution's online resource	100	11
Personalized pedigree	98.5	64
Patient letter	96.4	110
Letter addressed to family member	88.5	61
Online educational resource	77.2	79
Testing lab pamphlet	74.4	78
Role play	66.7	21
Support group	63.2	68

Participants were then asked if they thought the tools they used increased a patient's willingness to disclose their positive test results to their at-risk relatives. Of the respondents who provide their patient with a letter specifically addressed to an at-risk family member (N = 62), 93.7% thought that it helped increase patient motivation to disclose (Table 6). Furthermore, over 50% of all of the respondents for each tool used felt that the tool did increase a patient's willingness to inform their at-risk family member of their positive genetic test result.

**Table 6:** Percent of genetic counselors who use a particular tool and find it increases a patient's willingness to inform their at-risk family members of their positive genetic test results.

<b>Tool</b>	<b>Percent (%)</b>	<b>Total (N)</b>
Letter addressed to family member	93.5	62
Institution's online resource	90.9	11
Role play	90.5	21
Patient letter	88.3	111
Pedigree	77.8	63
Testing lab pamphlet	64.1	78
Online educational resource	53.9	76
Support group	51.5	68

*Tools genetic counselors do not use*

Participants who reported that they did not use any one of the eight approaches, strategies and/or materials were asked what their reasons were for not using the tool. Respondents could choose from a list of five reasons, the final being an “other” category that allowed for an open-ended response. Respondents were permitted to select more than one reason for each response (Table 7).

**Table 7:** Reasons why a genetic counselor does not use a tool.

<b>Tool</b>	<b>Reason not used</b>	<b>Percent (%)</b>	<b>Total (N)</b>
Institution's online resource			103
	Other	47.6	
	Not part of institution's protocol	46.6	
	Too time consuming	4.9	
	Do not find useful to patients	3.9	
Role-play	Not part of role as a genetic counselor	1.0	97
	Other	39.2	
	Do not find useful to patients	36.1	
	Too time consuming	25.8	
	Not part of institution's protocol	5.2	
Personalized Pedigree	Not part of role as a genetic counselor	3.1	55
	Other	38.2	
	Do not find useful to patients	32.7	
	Too time consuming	10.9	
	Not part of institution's protocol	5.0	
Letter addressed to family member	Not part of role as a genetic counselor	1.8	51
	Other	37.3	
	Too time consuming	29.4	
	Not part of institution's protocol	23.5	
	Do not find useful to patients	9.8	
Online educational resource	Not part of role as a genetic counselor	3.9	39
	Other	46.2	
	Do not find useful to patients	25.6	
	Not part of institution's protocol	20.5	
	Too time consuming	2.6	
Support group referral	Not part of role as a genetic counselor	0.0	39
	Other	66.7	
	Do not find useful to patients	17.9	
	Not part of institution's protocol	5.1	
	Too time consuming	0.0	
Testing lab pamphlet	Not part of role as a genetic counselor	0.0	32
	Do not find useful to patients	46.9	
	Other	40.6	
	Not part of institution's protocol	12.5	
	Too time consuming	0.0	
Patient letter	Not part of role as a genetic counselor	0.0	2
	Other	100.0	
	Do not find useful to patients	0.0	
	Too time consuming	0.0	
	Not part of institution's protocol	0.0	

*Institution's online educational resource/website:*

One hundred and three participants reported that they did not provide their patient with an online educational resource/website designed by the institution where they practiced their cancer genetic counseling. Of those respondents who did not use this tool, 46.6% reported this was because it was not part of their institution's genetic counseling protocol. Another common explanation expressed by respondents was that, "Our institution does not have such a website with specific information on Lynch syndrome available."

*Role-playing with the patient:*

The participants who reported that they did not use role-play to help their patient practice sharing their genetic Lynch syndrome diagnosis (N = 97) said that it was not useful to patients (36.1%) and that it was too time-consuming to implement (25.8%). Further reasons not listed (39.2%) that were expressed by respondents included (a) "I've never felt this was necessary," (b) "Many patients would not want to pay for this additional time (that would be billable)," and (c) "Patients usually do not take this kind of approach seriously."

*Personalized pedigree:*

Of the 55 participants who reported that they do not provide the patient with a pedigree that highlights which members of the patient's family are at highest risk for having a Lynch syndrome mutation, nearly one-third (32.7%) chose not to because they do not find it to be useful to patients. Other reasons that were added by participants were, "I think it singles out the specific relatives and places a negative (as in bad) 'red flag' or mark on them." Another counselor stated:

I would have to create an [anonymous] pedigree, and we are often working with very large pedigrees with information from multiple branches, so this would be time-consuming. Also, most people I find don't need a visual aid - they understand who they should share the information with.

*Letter to at-risk family member:*

Fifty-one participants reported that they do not provide their patients with a letter specifically addressed to an at-risk family member that includes a description of Lynch syndrome, which the family member can share with their health care provider. Of them, 29.4% felt that this tool was too time consuming to implement and 37.3% reported other, not listed reasons. Some of the responses included, "Our patient letter is useful for explaining the risk for other family members," and "Would do this in extreme situations if needed, but have not needed to."

*Online educational resource/website:*

Of those participants that do not give patients an online educational resource/website that gives general information about Lynch syndrome (N = 39), 25.6% did so because they thought that the tool was not useful to patients. Approximately 21% percent of respondents also reported that it was not used because it was not part of their institution's genetic counseling protocol. Alternative reasons that were given were: (a) "My patients are usually already aware of these resources," (b) "I just haven't found a website that provides all the information in a patient friendly language/reading level," and (c) "I do not provide these links [because] my patients rarely have Internet access and most are not English speaking."

*Lynch syndrome support group referral:*

Of those respondents who did not provide their patients with information about a support group for individuals with Lynch syndrome (N = 39), the majority (66.7%) had



reasons other than those listed. One respondent stated, “I would if I were asked, but my patients rarely need support groups. Most come from big families with known mutations, and have a lot of support from their relatives already.” Another common response was, “I have not been able to find these resources.”

*Testing lab brochure or pamphlet:*

Thirty-two participants reported that they did not provide their patients with a written brochure or pamphlet from the testing lab where the patients’ sample was tested. Of those participants, 46.9% reported that they did not find this tool useful to patients. Respondents also reported additional reasons that were not listed (40.6%), such as “No such brochure exists from the lab we use,” and “Do not want to give the patient items that are used as marketing tools by the lab.”

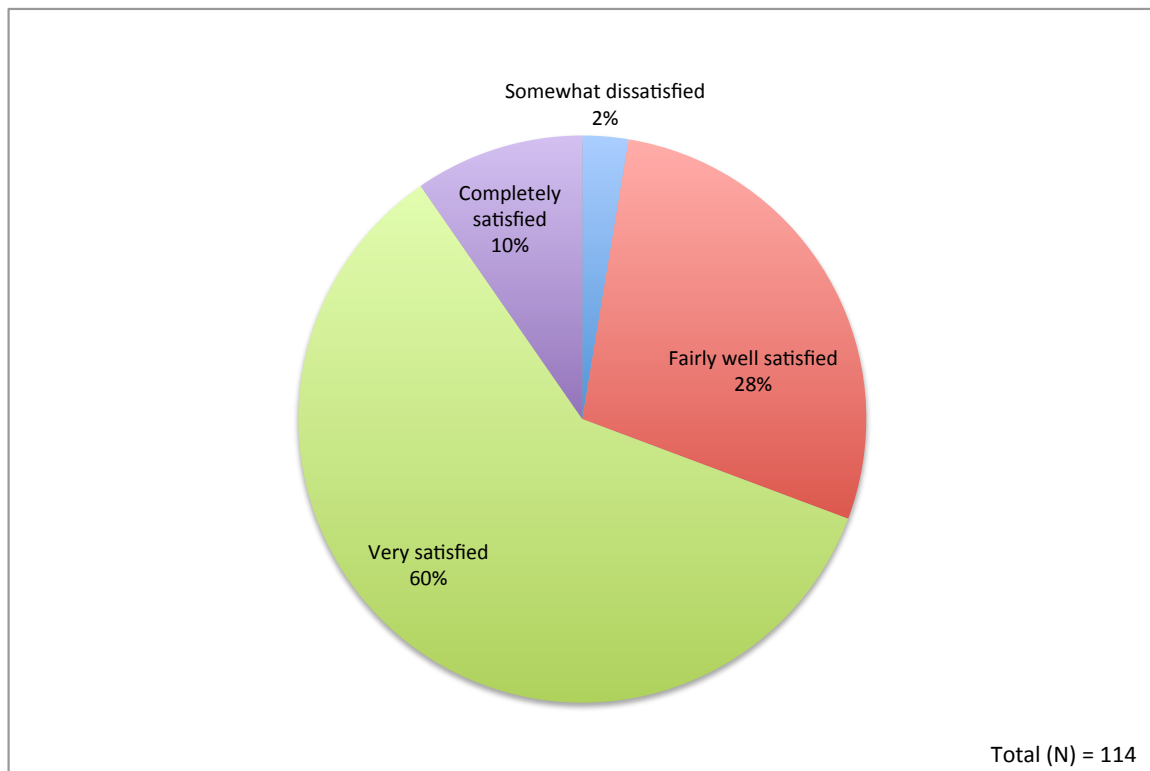
*Patient letter:*

Only two participants reported they did not provide their patient with a letter that included a description of Lynch syndrome, their genetic test results and which of their relatives is at- risk. Both of these respondents reported that there were “other” unlisted reasons that they did not use this tool, however, they did not leave any statements to further explain their answers.

*Genetic counselor satisfaction with tools*

Using a 5-point Likert scale ranging from “very dissatisfied” to “completely satisfied,” participants were asked to answer how satisfied they were with the tools they use to help patients share information about their positive mutation results with their at-risk family members (Figure 4). The majority of respondents reported that they were

either “very satisfied” (60%) or “fairly well satisfied” (28%). No respondents reported that they were “very dissatisfied.”



**Figure 4:** Genetic counselor satisfaction with the tools used to help patients disseminate information about their positive mutation results to their at-risk family members.

In the survey, counselors had an opportunity to explain and discuss their level of satisfaction with the approaches, strategies and materials they use to help patients disseminate information about their positive mutation results to their at-risk family members. Some respondents commented that they do feel satisfied with current tools they use now. One counselor stated:

Overall I am very satisfied with the materials I use for the men/women I have counseled with suspected or confirmed Lynch syndrome. I feel the tools/materials that I use are very effective with most patients. However, I also find that most patients are already motivated to share their results with their family members.

A second counselor also commented:

I think my methods have been more than sufficient for facilitating communication with family members for the few clients I have had. For the most part, my

patients with a molecular diagnosis have been close with their family members and willing to talk to them about the potential cancer risk. I am not sure the methods I use would be as effective with a less willing patient, which is why I consider myself "very" rather than "completely" satisfied.

Other counselors commented on not being satisfied with the tools available. One counselor commented, "I think that in general the resources are poor for aiding patients with the process of communicating genetic information to family members." This opinion was further supported by a second counselor who stated, "I think there is a lack of good patient-oriented (non-lab) materials on Lynch syndrome - both print and on-line." A third counselor stated:

"I am uncertain if it is our way of disseminating the info, or a resistance on the part of the recipients. Why do so few family members seek genetic services[?] I think it may be due to a cultural bias about "colon" issues. The colon is not a socially important organ! We don't see the resistance in the HBOC families, at least not to the same extent. Different organ, different levels of concern about risk."

#### *Importance of tools for patients*

Counselors were asked, based on their experience, what percent of patients need one or more of the tools to help them share their molecular diagnosis of Lynch syndrome with at-risk family members. The majority of respondents (n = 67, 60.3%) reported that they think 81-100% of patients need at least one of these tools. (Table 8)

**Table 8:** Percent of all patients that genetic counselors think need one or more of the tools to help them share their molecular diagnosis of Lynch syndrome with at-risk family members.

Percent of patients that need tool	Percent (%)	Frequency (n)	Total (N) = 111
0-20%	8.1	9	
21-40%	0.9	1	
41-60%	13.5	15	
61-80%	17.1	19	
81-100%	60.3	67	

### *Additional resources for patients*

Participants were asked, if time and cost were not a factor, what additional resources they would like for their patients to help them share their molecular diagnosis of Lynch syndrome with at-risk family members. One genetic counselor explained that they did not need any additional resources, stating, “Nothing [is needed], a good results appointment should provide a patient with the necessary resources and information to talk to their family.” Other counselors, however, felt that additional resources would be helpful, such as an “organization like FORCE, but dedicated to those with Lynch.” Some of the counselors described the resources they would like to have:

Would like to be able to offer a local support group for individuals with Lynch syndrome to allow patients to talk to each other and share information with each other.

A family tree that has the person's risk on it, with some kind of check box for each relative to mark off when the information has been shared with them. Also, giving the patients many copies of the family letter and results would be nice so that the burden of copying does not fall on them.

A CD they could take home or website that would describe Lynch syndrome using patient driven and patient appropriate materials, education, so that they can go over it and share with other family members.

Perhaps have our institution create a packet specific for patients with positive Lynch syndrome results that describes the condition, lays out who in the family is at-risk, and provides contact information for a local GC or GC service for each of their family members, specific to where they live.

## DISCUSSION

### *Sharing Lynch syndrome genetic diagnosis with at-risk family members*

In this study, we found that cancer genetic counselors do discuss the implications that their patient's positive testing results can have for their at-risk family members. Specifically, most genetic counselors often, or always, discuss with their patients that (a) their at-risk relatives should follow Lynch syndrome screening guidelines; (b) they should consider sharing their test results with their at-risk relatives; (c) sharing their results with at-risk family members could prevent cancer in their at-risk family members; and (d) their at-risk family members should see a genetic counselor. This shows that genetic counselors are fulfilling an essential role in helping patients understand the value of informing their at-risk relatives. This is particularly significant in light of the research by Mesters et al., (2005) that revealed patients were more likely to inform family if health professionals stressed the importance of disclosing test results to family.

Not only were patients typically made aware of the importance of telling at-risk relatives their results, but they also showed interest in doing so. Nearly all of the genetic counselors (97.6%) indicated that their patients expressed interest in sharing information about their diagnosis with their at-risk family members. Also notable is the fact that no genetic counselors reported their patients "never" or "rarely" showed interest in sharing their diagnosis. These results correlate with those of Stoffel et al. (2008) and demonstrate that individuals with an unambiguous Lynch syndrome genetic test result do in fact have an interest in informing their relatives.

When asked what some of the reasons were behind a patient's interest in sharing their positive testing results with an at-risk family member, 88% of genetic counselors responded that it was in order to help prevent future disease in at-risk family members. Additionally, 66.9% noted that patients felt a responsibility to their at-risk relatives. These results imply that patients do understand the risks associated with their diagnosis as well as what their diagnosis means to their relatives and their relatives' health. One counselor explained that patients want to see a "silver lining" to their diagnosis by being able to warn their relatives and to help prevent the occurrence of Lynch syndrome related cancers in their family members. Another counselor pointed out that patients tell their family because they now have an explanation for the cancer running in the family.

For the patients who expressed some level of resistance to sharing their positive genetic testing results with at-risk family members, more than half of genetic counselors reported that this was due to a distant (59.9%) or poor/conflicting (52.8%) relationship between the patients and the family member. These results support the literature regarding communication about genetic risk within families, specifically that relationship factors strongly influence communication patterns and have been found to significantly influence whether or not an at-risk family member is informed of an individual's positive test result (Wiseman et al., 2010). Misunderstanding the inheritance of Lynch syndrome (1.4%) was the least common reason given by genetic counselors for patient resistance. This is reassuring as it implies that it is not that patients are misunderstanding the material, but that other external elements are influencing their decision to not inform their at-risk family members.

Other factors that were noted by counselors were related to the patients' perception of how the at-risk family member would use the information given to them. If a patient felt their family member was not going to use the information to guide their health care behaviors then the patient was less inclined to want to discuss their test results. One counselor noted that an at-risk family member was not insured so the patient was feeling less motivated to tell them, as the family member would not have the ability to get genetic testing or access to the recommended screening procedures.

*Approaches, strategies and materials used by genetic counselors*

Of the eight approaches, strategies and/or materials listed that could be used to help patients with a positive Lynch syndrome mutation inform their family members of their potential risks, genetic counselors reported that a patient letter (98.2%) was the tool used by the most genetic counselors. Moreover, when asked how frequently the tools were used, genetic counselors reported that a patient letter was used the most often. These results probably reflect the fact that a patient letter is a part of a typical cancer genetic counseling protocol at most institutions. Only 9.6% of genetic counselors, however, reported ever having used an online educational resource from the genetic counselors' own institution. Role-play, used by 17.8% of genetic counselors, was the tool reportedly used the least frequently among those genetic counselors that reported ever having used it.

Personalized pedigrees were used by 54.5% of genetic counselors and we found a negative correlation between the use of a personalized pedigree and number of patients seen with a suspected clinical or molecular diagnosis: as the number of patients seen with a suspected clinical or molecular diagnosis increased, the use of the personalized

pedigree tool decreased. We speculate that this correlation is due to a personalized pedigree being a time-consuming tool to create. As the number of patients being seen increases, the practice itself may be getting busier and the time available to spend with a patient during a genetic counseling session may be getting shorter as well, making it more difficult to use such a tool.

We also found a negative correlation between the amount of time within the past five years the counselor has practiced cancer genetic counseling and the use of a Lynch syndrome testing lab pamphlet. Therefore, as the time within the past five years the counselor has practiced cancer genetic counseling increased, the less likely they were to use a pamphlet from the testing lab where the patient's sample was tested. This correlation may be attributed to counselors who have practiced more within the past five years being more likely to have their own, possibly more personalized, resources that they prefer to use.

In this study, genetic counselors indicated that, overall, the tools they use are both effective and helpful to patients. Of the tools discussed in this research, every tool was reported to be effective at helping patients to understand who in their family was at risk of inheriting Lynch syndrome by at least 63% of genetic counselors. Furthermore, over 50% of all of the respondents who used a given tool felt that the tool did increase a patient's willingness to inform their at-risk family member of their positive genetic test result.

#### *Reasons why genetic counselors do not use certain approaches, strategies and materials*

Genetic counselors reported various reasons for not using certain tools. For instance, some genetic counselors felt that a personalized pedigree was not helpful for



patients because it could single out family members in a negative manner. Additionally, counselors felt that it was an inefficient use of their time, as patients already understood which family members were at-risk. Some genetic counselors expressed that an online educational resource/website was not useful because they have yet to find a suitable, patient-friendly website to give out to patients. Additionally, genetic counselors expressed that electronic resources overall were not helpful, as many of their patients do not have access to the Internet.

Only two genetic counselors reported not using a patient letter, though they did not explain why. Those genetic counselors who reported not using a letter to an at-risk family member responded that it was too time-consuming to implement and that the actual patient letter was just as useful to explain the risk for other family members. One genetic counselor wrote that they would provide such a letter, but only if they were asked by a patient to do so.

With role-play, counselors explained that it was too time-consuming and that the patients did not take this approach seriously. A testing lab brochure or pamphlet was noted not to be useful, as some of the genetic counselors' testing labs did not have a brochure or pamphlet to give out to patients. Also, some genetic counselors expressed that they did not want to give a patient materials that were also used as lab marketing tools.

Of those respondents who did not provide their patients with information about a Lynch syndrome support group for individuals with Lynch syndrome, genetic counselors stated that they would like to use this tool for their patients, but that they have not been able to find a support group in their geographical location. Another genetic counselor

noted that they did not feel this resource was necessary as their patients already had family support.

#### *Genetic counselor satisfaction with approaches, strategies and materials*

According to this study, genetic counselors (88%) are generally “very satisfied” or “fairly well satisfied” with the tools they use to help patients share information about their positive mutation results with their at-risk family members. A common theme expressed by genetic counselors was that they were very satisfied, but that their patients typically are already motivated to discuss their results with their at-risk family members.

No genetic counselors reported that they were “very dissatisfied” with the tools that they use, though 2% did respond that they were “somewhat dissatisfied.” In their responses, though, genetic counselors expressed frustration with the lack of patient-friendly tools, such as websites and non-laboratory related brochures or pamphlets. Counselors also noted that access to local support groups was significantly lacking.

Although use of the eight approaches, strategies and/or materials varies, overall, the majority of genetic counselors think that patients need at least one or more of these tools to help them share their molecular diagnosis of Lynch syndrome with at-risk family members. Genetic counselors also expressed that the addition of patient-friendly multimedia resources (such as a Lynch syndrome DVD or a website that could be shared with family members) and local support groups would help their patients share their molecular diagnosis of Lynch syndrome with at-risk family members.

#### *Study limitations*

Since survey participants were required to have seen patients for Lynch syndrome diagnosis, demographics could not be compared to the 2010 NSGC Professional Status

Survey (PSS). The PSS includes responses from all genetic counselors, and is not divided by the type of cases they see, so we are unable to determine if this study sample is representative of cancer genetic counselors who see Lynch syndrome patients. Additional sample limitations include the fact that only genetic counselors who are members of NSGC and elected to receive the listservs during the recruitment period received information about the study. This may have limited the sample size.

Online survey design, by nature, has specific flaws. The survey required participants to determine how helpful the approaches, strategies and/or materials were for patients and whether they increase a patient's willingness to disclose their test results. This may have been difficult for participants to ascertain. Some participants felt that they did not have a good way to measure these values, and therefore may have skipped answering these questions. Additionally, genetic counselors were asked how helpful and effective they thought the tools were for their patients. In doing so, we measured genetic counselors' *perception* of the effectiveness of the tools but did not assess the *actual* effectiveness of the tools themselves. Finally, participants were not required to answer all questions, which may also have factored in to skewed data.

## **CONCLUSION**

The aim of this study was to identify what strategies and instruments genetic counselors find to be the most successful in helping patients share their positive Lynch syndrome mismatch repair (MMR) mutation genetic test results with at-risk family members. This is particularly important as improved communication of positive test results will inform the family members of their potential risks as well as help improve cancer surveillance for those at-risk family members. This study shows that cancer genetic counselors use a variety of tools to help Lynch syndrome patients share the positive genetic test results with their family members and that, overall, most genetic counselors (88%) are satisfied with the tools they currently use. Additionally, genetic counselors reported that, by and large, they feel the tools they use are both effective and helpful to patients.

Despite satisfaction with current tools, genetic counselors do think additional tools would benefit Lynch syndrome patients during their genetic counseling sessions. Future research should focus on carefully assessing patients' perceptions of the tools that successfully helped them share their positive Lynch syndrome genetic test results with at-risk family members. Results can then be used to develop new tools such as more patient-friendly online resources similar to those offered by the hereditary breast and ovarian cancer organization, FORCE.

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## APPENDIX A: Survey Questions

### *Introduction:*

Thank you for accepting the invitation to participate in my Master's thesis research project.

The survey should take approximately 20 minutes to complete. Please note that participation in this study is completely voluntary and anonymous. You may stop at any time. All information collected will remain anonymous and no identifying information will be linked to your answers. This research has been approved by the Brandeis University IRB (irb@brandeis.edu).

By clicking "*Next Page*," you acknowledge that you have read the above information and wish to participate in this survey.

If you would like to enter the drawing for one of two \$50 gift cards to Amazon.com, please complete the survey and follow the directions at the end.

If you have any questions or comments please do not hesitate to contact me or the Principal Investigator of this study, Judith Tsipis, PhD at: [tsipis@brandeis.edu](mailto:tsipis@brandeis.edu).

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### *Survey Questions:*

1. Have you practiced cancer genetic counseling, either full or part time, within the last five years (i.e. since 2006)?
  - a. Yes
  - b. No (*If answer 'No,' will be excluded from survey*)

What a participant will see if they have been excluded: "*Thank you for your participation, but you do not meet the enrollment criteria for this study.*"

2. How much time within the past 5 years have you practiced cancer genetic counseling, either full time or part time?
  - a. Less than six months (*If answer 'a,' will be excluded from survey*)
  - b. Six months to one year
  - c. One to two years
  - d. Two to three years

- e. Three to four years
  - f. Four to Five years
3. Have you counseled a patient with a molecular Lynch syndrome diagnosis (a mutation in one of the four mismatch repair (MMR) genes known to cause Lynch syndrome) within the past five years?
    - a. Yes
    - b. No (*If answer 'No,' will be excluded from survey*)
  4. Do you currently see patients for cancer risk assessment or counsel patients for hereditary cancer as part of your job?
    - a. Yes
    - b. No  
(*If answer "No," then will be directed to a set of questions about their "last" or "most recent" cancer counseling experience*)
  5. What is your current cancer genetic counseling work setting? (Check all that apply)
    - a. Cancer Institute
    - b. University Medical Center
    - c. Private Hospital/Medical Facility
    - d. Public Hospital/Medical Facility
    - e. Physician's Private Practice
    - f. Health Maintenance Organization
    - g. Diagnostic Laboratory - Commercial
    - h. Other (please specify)
  6. Please describe your primary role(s) in your current position as a cancer genetic counselor (you may indicate more than one):
    - a. Clinical
    - b. Research/Study Coordinator
    - c. Administrative
    - d. Clinical Coordination
    - e. Management
    - f. Teaching/Education/Supervising Students
    - g. Customer Liaison
    - h. Lab Support
    - i. Grant management
    - j. Other
  7. In what U.S. region do you practice cancer genetic counseling?
    - a. Region I: CT, MA, ME, NH, RI, VT
    - b. Region II: DC, DE, MD, NJ, NY, PA, VA, WV, Puerto Rico, Virgin Islands
    - c. Region III: AL, FL, GA, KY, LA, MS, NC, SC, TN
    - d. Region IV: AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI



- e. Region V: AZ, CO, MT, NM, TX, UT, WY
- f. Region VI: AK, CA, HI, ID, NV, OR, WA

8. In the past year, approximately how many patients with a *suspected* clinical or molecular diagnosis of Lynch syndrome have you seen? Please provide a specific number.
9. In the past year, approximately how many patients with a *confirmed* molecular diagnosis of Lynch syndrome (a mutation in one of the four mismatch repair (MMR) genes known to cause Lynch syndrome) have you seen? Please provide a specific number.
10. In a typical Lynch syndrome result-giving session where you inform a patient that they carry a genetic mutation that predisposes them to cancer, how often do you discuss that:

	Always	Often	Sometimes	Seldom	Never
Their at-risk relatives should follow Lynch syndrome screening guidelines					
They should consider sharing their test results with their at-risk relatives					
Sharing their results with at-risk family members could prevent cancer in their at-risk family members					
Their at-risk family members should see a genetic counselor					

11. In the last five years, how often do patients who have received a molecular diagnosis of Lynch syndrome express an interest in sharing information about their diagnosis (and what the diagnosis means for their at-risk family members) with their family members?
  - a. Always
  - b. Often
  - c. Sometimes
  - d. Seldom
  - e. Never
12. For patients who express an interest in sharing information about their diagnosis, what are some of the reasons they give for wanting to share their positive genetic testing results with an at-risk family member? (Please mark all that apply)
  - a. Feeling a responsibility to their at-risk relatives
  - b. To help prevent future disease in at-risk relatives
  - c. Patient wants to comply with the genetic counselor's recommendations
  - d. Patient would receive advice and support

- e. Other: Please list
13. In the last five years, how often do patients who have received a molecular diagnosis of Lynch syndrome express *resistance* to sharing information about their diagnosis (and what the diagnosis means for their at-risk family members)?
- Always
  - Often
  - Sometimes
  - Seldom
  - Never
14. For patients who express *resistance* to sharing information about their diagnosis, what are some of the reasons they give for *not* wanting to share their positive genetic testing results with an at-risk family member? (Please mark all that apply)
- Age of at-risk family member
  - Distant relationship with at-risk family member
  - Poor/conflicting relationship with at-risk family member
  - Feeling of difficulty in discussing genetic test results
  - Desire to keep their health-related information private
  - Relying on other family members to relay the information
  - Patient is worried about at-risk family members' reaction
  - Patient misunderstands the inheritance of Lynch syndrome
  - Patient believes the at-risk relative will not benefit from the information
  - Feelings of guilt or anxiety
  - Concerns about discrimination
  - Other: Please list
15. Approximately how often do your patients who have a molecular diagnosis of Lynch syndrome *ask for help* relaying information about their diagnosis (and what the diagnosis means for their at-risk family members) to their family members?
- Always
  - Often
  - Sometimes
  - Seldom
  - Never

The following are some specific approaches, strategies and materials that could be used to help patients with a positive Lynch syndrome mutation result inform their at-risk family members of their diagnosis and what the diagnosis means for their at-risk family members. Please answer the questions following each approach, strategy or material based on your experience.

16. Providing the patient with a three-generation pedigree that highlights which members of the family are at highest risk for having inherited the Lynch syndrome mutation.

- a. Do you ever use this approach?
  - i. Yes
  - ii. No

**If answer yes to ‘a’ then will be asked question b-f. If answer no, then will be asked g.**

- b. How often do you use this approach?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. When using this approach, do you include the numerical risks for each at-risk family member of inheriting the Lynch syndrome mutation?
  - i. Yes
  - ii. No
- d. Overall, do patients find this approach helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- e. Do you think this approach increases a patient’s willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- f. Optional: Please use this space to further discuss/explain your previous answers to a-e
- g. What are some of the reasons why you do not use this approach?
  - i. Do not find it to be useful to patients
  - ii. Is not part of my institution’s genetic counseling protocol
  - iii. Too time consuming for me to implement
  - iv. Using this approach is not part of my role as a genetic counselor
  - v. Other: Please list
- h. Optional: Please use this space to further discuss/explain your previous answer to g

17. Providing the patient with an online educational resource/website that gives general information about Lynch syndrome (description of the disease, genetics and inheritance) such as the Genetics Home Reference (<http://ghr.nlm.nih.gov/condition/lynch-syndrome>) or the American Cancer Society (<http://www.cancer.org/Cancer/ColonandRectumCancer/DetailedGuide/index>).
  - a. Do you provide patients with this information?

- i. Yes
- ii. No

**If answer *yes* to ‘a’ then will be asked question b-e. If answer *no*, then will be asked f.**

- b. How often do you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, do patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this information increases a patient’s willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What are some of the reasons why you do not give patients this information?
  - i. Do not find it to be useful to patients
  - ii. Is not part of my institution’s genetic counseling protocol
  - iii. Too time consuming for me to implement
  - iv. Providing this information is not part of my role as a genetic counselor
  - v. Other: Please list
- g. Optional: Please use this space to further discuss/explain your previous answers to f

18. Role-playing with the patient. You act as the patient’s at-risk family member and the patient practices sharing their genetic Lynch syndrome diagnosis with you.

- a. Do you use this method?
  - i. Yes
  - ii. No

**If answer *yes* to ‘a’ then will be asked question b-e. If answer *no*, then will be asked f.**

- b. How often do you use this method?
  - i. Always
  - ii. Sometimes

- iii. Rarely
  - c. Overall, do patients find this method helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
    - i. Yes
    - ii. No
  - d. Do you think this method increases a patient's willingness to inform their at-risk family members of their positive genetic test results?
    - i. Yes
    - ii. No
  - e. Optional: Please use this space to further discuss/explain your previous answers to a-d
  - f. What are some of the reasons why you do not use this method?
    - i. Do not find it to be useful to patients
    - ii. Is not part of my institution's genetic counseling protocol
    - iii. Too time consuming for me to implement
    - iv. Using this method is not part of my role as a genetic counselor
    - v. Other: Please list
  - g. Optional: Please use this space to further discuss/explain your previous answers to f
19. Providing the patient with a letter written to the patient that includes a description of Lynch syndrome, the patient's genetic test results and which of their relatives are at-risk of having inherited the Lynch syndrome mutation.
- a. Do you use this approach?
    - i. Yes
    - ii. No

**If answer yes to 'a' then will be asked question b-e. If answer no, then will be asked f.**

- b. How often do you use this approach?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, do patients find this approach helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this approach increases a patient's willingness to inform their at-risk family members of their positive genetic test results?

- i. Yes
- ii. No

- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What are some of the reasons why you do not use this approach?
  - i. Do not find it to be useful to patients
  - ii. Is not part of my institution's genetic counseling protocol
  - iii. Too time consuming for me to implement
  - iv. Using this approach is not part of my role as a genetic counselor
  - v. Other: Please list
- g. Optional: Please use this space to further discuss/explain your previous answers to f

20. Providing the patient with an online educational resource/website designed by the institution where you practice your cancer genetic counseling.

- a. Do you provide patients with this information?
  - i. Yes
  - ii. No

**If answer *yes* to 'a' then will be asked question b-e. If answer *no*, then will be asked f.**

- b. How often do you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, do patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this information increases a patient's willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What are some of the reasons why you do not give patients this information?
  - i. Do not find it to be useful to patients

- ii. Is not part of my institution's genetic counseling protocol
- iii. Too time consuming for me to implement
- iv. Providing this information is not part of my role as a genetic counselor
- v. Other: Please list

g. Optional: Please use this space to further discuss/explain your previous answers to f

21. Providing the patient with information about a Lynch syndrome support group for individuals with Lynch syndrome such as Lynch Syndrome International (<http://www.lynchcancers.org/>)

- a. Do you give patients this information?
  - i. Yes
  - ii. No

**If answer yes to 'a' then will be asked question b-e. If answer no, then will be asked f.**

- b. How often do you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, do patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this information increases a patient's willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What are some of the reasons why you do not give patients this information?
  - i. Do not find it to be useful to the patient
  - ii. Is not part of my institution's genetic counseling protocol
  - iii. Too time consuming to implement
  - iv. Providing this information is not part of my role as a genetic counselor
  - v. Other: Please list

- g. Optional: Please use this space to further discuss/explain your previous answers to f

22. Providing the patient with a written brochure or pamphlet from the testing lab where their sample was tested or from your institution, that can be used to educate the patient and that can be shared with at-risk relatives.

- a. Do you give patients this information?
  - i. Yes
  - ii. No

**If answer yes to ‘a’ then will be asked question b-e. If answer no, then will be asked f.**

- b. How often do you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, do patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this information increases a patient’s willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What are some of the reasons why you do not give patients this information?
  - i. Do not find it to be useful to patients
  - ii. Is not part of my institution’s genetic counseling protocol
  - iii. Too time consuming for me to implement
  - iv. Providing this information is not part of my role as a genetic counselor
  - v. Other: Please list

- g. Optional: Please use this space to further discuss/explain your previous answers to f

23. Providing the patient with a letter specifically addressed to an at-risk family member that includes a description of Lynch syndrome which the family member can share with their health care provider.

- a. Do you use this approach?



- i. Yes
- ii. No

**If answer *yes* to ‘a’ then will be asked question b-e. If answer *no*, then will be asked f.**

- b. How often do you use this approach?
    - i. Always
    - ii. Sometimes
    - iii. Rarely
  - c. Overall, do patients find this approach helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
    - i. Yes
    - ii. No
  - d. Do you think this approach increases a patient’s willingness to inform their at-risk family members of their positive genetic test results?
    - i. Yes
    - ii. No
  - e. Optional: Please use this space to further discuss/explain your previous answers to a-d
  - f. What are some of the reasons why you do not use this approach?
    - i. Do not find it to be useful to the patient
    - ii. Is not part of my institution’s genetic counseling protocol
    - iii. Too time consuming to implement
    - iv. Using this approach is not part of my role as a genetic counselor
    - v. Other: Please list
  - g. Optional: Please use this space to further discuss/explain your previous answers to f
24. How satisfied are you with the approaches, and/or materials you use to help patients disseminate information about their positive mutation results to their at-risk family members?
- a. Completely satisfied
  - b. Very satisfied
  - c. Fairly well satisfied
  - d. Somewhat dissatisfied
  - e. Very dissatisfied
  - f. Optional: Please use this space to further discuss/explain your previous answer

25. In your experience, approximately what percent of all patients need one or more of these tools to help them share their molecular diagnosis of Lynch syndrome with at-risk family members?
- There will be a scale of percentages from 0-100% that the participant can choose from
26. If time and cost were not a factor, what additional resources would you like to have for your patients to help them share their molecular diagnosis of Lynch syndrome with at-risk family members?
- Open-ended question

**Demographic questions:**

D1. Please indicate your age:

D2. Please indicate which gender you most identify with:

- Male
- Female

D3. What best describes your ethnic background?

- Black or African American
- Asian
- White or Caucasian
- Native Hawaiian or Other Pacific Islander
- American Indian or Alaskan Native
- Other (please specify)

D4. Do you currently work full time or part time?

- Full time
- Part time
- Not currently working

D5. In what year did you graduate from a genetic counseling masters program?

END OF SURVEY:

Thank you for participating in this research project. The answers and insight you shared will be very helpful. Please feel free to contact me with any questions.

Tara Biagi

Brandeis University Genetic Counseling, Class of 2011

[tbiagi@brandeis.edu](mailto:tbiagi@brandeis.edu)

If you would like to enter the drawing for one of two \$50 gift cards to Amazon.com please click on the link below. Your entry will not be connected with your responses to the survey.

LINK

*The link will take participants to a new page where they will enter their email address.*

*This information will not be linked with participants' responses to the survey.*

END

Survey questions #5-26 formatted for GCs who are not currently cancer counselors:  
Questions:

5. What was your most recent cancer genetic counseling work setting? (Check all that apply)
  - a. Cancer Institute
  - b. University Medical Center
  - c. Private Hospital/Medical Facility
  - d. Public Hospital/Medical Facility
  - e. Physician's Private Practice
  - f. Health Maintenance Organization
  - g. Diagnostic Laboratory - Commercial
  - h. Other (please specify)
6. Please describe your primary role(s) in your most recent position as a cancer genetic counselor (you may indicate more than one):
  - a. Clinical
  - b. Research/Study Coordinator
  - c. Administrative
  - d. Clinical Coordination
  - e. Management
  - f. Teaching/Education/Supervising Students
  - g. Customer Liaison
  - h. Lab Support
  - i. Grant management
  - j. Other
7. In what U.S. region did you practice cancer genetic counseling?
  - a. Region I: CT, MA, ME, NH, RI, VT
  - b. Region II: DC, DE, MD, NJ, NY, PA, VA, WV, Puerto Rico, Virgin Islands
  - c. Region III: AL, FL, GA, KY, LA, MS, NC, SC, TN
  - d. Region IV: AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI
  - e. Region V: AZ, CO, MT, NM, TX, UT, WY
  - f. Region VI: AK, CA, HI, ID, NV, OR, WA
8. In a one-year period, approximately how many patients with a *suspected* clinical or molecular diagnosis of Lynch syndrome did you see? Please provide a specific number.
9. In a one-year period, approximately how many patients with a *confirmed* molecular diagnosis of Lynch syndrome (a mutation in one of the four mismatch repair (MMR) genes known to cause Lynch syndrome) did you see? Please provide a specific number.

10. In a typical Lynch syndrome result-giving session where you informed a patient that they carry a genetic mutation that predisposes them to cancer, how often did you discuss that:

	Always	Often	Sometimes	Seldom	Never
Their at-risk relatives should follow Lynch syndrome screening guidelines					
They should consider sharing their test results with their at-risk relatives					
Sharing their results with at-risk family members could prevent cancer in their at-risk family members					
Their at-risk family members should see a genetic counselor					

11. In the last five years, how often did patients who received a molecular diagnosis of Lynch syndrome express an interest in sharing information about their diagnosis (and what the diagnosis means for their at-risk family members) with their family members?
- Always
  - Often
  - Sometimes
  - Seldom
  - Never
12. For patients who expressed an interest in sharing information about their diagnosis, what were some of the reasons they gave for wanting to share their positive genetic testing results with an at-risk family member? (Please mark all that apply)
- Feeling a responsibility to their at-risk relatives
  - To help prevent future disease in at-risk relatives
  - Patient wants to comply with the genetic counselor's recommendations
  - Patient would receive advice and support
  - Other: Please list
13. In the last five years, how often did patients who had received a molecular diagnosis of Lynch syndrome express *resistance* to sharing information about their diagnosis (and what the diagnosis means for their at-risk family members)?
- Always
  - Often
  - Sometimes
  - Seldom
  - Never

14. For patients who expressed *resistance* to sharing information about their diagnosis, what were some of the reasons they gave for *not* wanting to share their positive genetic testing results with an at-risk family member? (Please mark all that apply)
- a. Age of at-risk family member
  - b. Distant relationship with at-risk family member
  - c. Poor/conflicting relationship with at-risk family member
  - d. Feeling of difficulty in discussing genetic test results
  - e. Desire to keep their health-related information private
  - f. Relying on other family members to relay the information
  - g. Patient is worried about at-risk family members' reaction
  - h. Patient misunderstands the inheritance of Lynch syndrome
  - i. Patient believes the at-risk relative will not benefit from the information
  - j. Feelings of guilt or anxiety
  - k. Concerns about discrimination
  - l. Other: Please list
15. Approximately how often did your patients who had a molecular diagnosis of Lynch syndrome *ask for help* relaying information about their diagnosis (and what the diagnosis means for their at-risk family members) to their family members?
- a. Always
  - b. Often
  - c. Sometimes
  - d. Seldom
  - e. Never

The following are some specific approaches, strategies and materials that could be used to help patients with a positive Lynch syndrome mutation result inform their at-risk family members of their diagnosis and what the diagnosis means for their at-risk family members. Please answer the questions following each approach, strategy or material based on your experience.

16. Providing the patient with a three-generation pedigree that highlights which members of the family are at highest risk for having inherited the Lynch syndrome mutation.
- a. Did you ever use this approach?
    - i. Yes
    - ii. No

**If answer yes to 'a' then will be asked question b-f. If answer no, then will be asked g.**

- b. How often did you use this approach?
  - i. Always
  - ii. Sometimes
  - iii. Rarely

- c. When using this approach, did you include the numerical risks for each at-risk family member of inheriting the Lynch syndrome mutation?
    - i. Yes
    - ii. No
  - d. Overall, did patients find this approach helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
    - i. Yes
    - ii. No
  - e. Do you think this approach increased a patient's willingness to inform their at-risk family members of their positive genetic test results?
    - i. Yes
    - ii. No
  - f. Optional: Please use this space to further discuss/explain your previous answers to a-e
  - g. What were some of the reasons why you did not use this approach?
    - i. Did not find it to be useful to patients
    - iii. Was not part of my institution's genetic counseling protocol
    - iv. Too time consuming for me to implement
    - v. Using this approach was not part of my role as a genetic counselor
    - vi. Other: Please list
  - h. Optional: Please use this space to further discuss/explain your previous answer to g
17. Providing the patient with an online educational resource/website that gives general information about Lynch syndrome (description of the disease, genetics and inheritance) such as the Genetics Home Reference (<http://ghr.nlm.nih.gov/condition/lynch-syndrome>) or the American Cancer Society (<http://www.cancer.org/Cancer/ColonandRectumCancer/DetailedGuide/index>).
- a. Did you provide patients with this information?
    - i. Yes
    - ii. No

**If answer yes to 'a' then will be asked question b-e. If answer no, then will be asked f.**

- b. How often did you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely

- c. Overall, did patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
    - i. Yes
    - ii. No
  - d. Do you think this information increased a patient's willingness to inform their at-risk family members of their positive genetic test results?
    - i. Yes
    - ii. No
  - e. Optional: Please use this space to further discuss/explain your previous answers to a-d
  - f. What were some of the reasons why you did not give patients this information?
    - i. Did not find it to be useful to patients
    - ii. Was not part of my institution's genetic counseling protocol
    - iii. Too time consuming for me to implement
    - iv. Providing this information was not part of my role as a genetic counselor
    - v. Other: Please list
  - g. Optional: Please use this space to further discuss/explain your previous answers to f
18. Role-playing with the patient. You act as the patient's at-risk family member and the patient practices sharing their genetic Lynch syndrome diagnosis with you.
- a. Did you use this method?
    - i. Yes
    - ii. No

**If answer yes to 'a' then will be asked question b-e. If answer no, then will be asked f.**

- b. How often did you use this method?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, did patients find this method helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - iii. No
- d. Do you think this method increased a patient's willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes

- ii. No
  - e. Optional: Please use this space to further discuss/explain your previous answers to a-d
  - f. What were some of the reasons why you did not give patients this information?
    - i. Did not find it to be useful to patients
    - ii. Was not part of my institution's genetic counseling protocol
    - iii. Too time consuming for me to implement
    - iv. Providing this information was not part of my role as a genetic counselor
    - v. Other: Please list
  - g. Optional: Please use this space to further discuss/explain your previous answers to f
19. Providing the patient with a letter written to the patient that includes a description of Lynch syndrome, the patient's genetic test results and which of their relatives are at-risk of having inherited the Lynch syndrome mutation.
- a. Did you use this approach?
    - i. Yes
    - ii. No

**If answer yes to 'a' then will be asked question b-e. If answer no, then will be asked f.**

- b. How often did you use this approach?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, did patients find this approach helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this approach increased a patient's willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What were some of the reasons why you did not use this approach?
  - i. Did not find it to be useful to patients



- ii. Was not part of my institution's genetic counseling protocol
- iii. Too time consuming for me to implement
- iv. Using this approach was not part of my role as a genetic counselor
- v. Other: Please list

- g. Optional: Please use this space to further discuss/explain your previous answers to f

20. Providing the patient with an online educational resource/website designed by the institution where you practice your cancer genetic counseling.

- a. Did you provide patients with this information?
  - i. Yes
  - ii. No

**If answer yes to 'a' then will be asked question b-e. If answer no, then will be asked f.**

- b. How often did you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, did patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this information increased a patient's willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What were some of the reasons why you did not give patients this information?
  - i. Did not find it to be useful to patients
  - ii. Was not part of my institution's genetic counseling protocol
  - iii. Too time consuming for me to implement
  - iv. Providing this information was not part of my role as a genetic counselor
  - v. Other: Please list
- g. Optional: Please use this space to further discuss/explain your previous answers to f

21. Providing the patient with information about a Lynch syndrome support group for individuals with Lynch syndrome such as Lynch Syndrome International (<http://www.lynchcancers.org/>)

- a. Did you give patients this information?
  - i. Yes
  - ii. No

**If answer yes to ‘a’ then will be asked question b-e. If answer no, then will be asked f.**

- b. How often did you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, did patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this information increased a patient’s willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What were some of the reasons why you did not give patients this information?
  - i. Did not find it to be useful to the patient
  - ii. Was not part of my institution’s genetic counseling protocol
  - iii. Too time consuming to implement
  - iv. Providing this information was not part of my role as a genetic counselor
  - v. Other: Please list
- g. Optional: Please use this space to further discuss/explain your previous answers to f

22. Providing the patient with a written brochure or pamphlet from the testing lab where their sample was tested or from your institution, that can be used to educate the patient and that can be shared with at-risk relatives.

- a. Did you give patients this information?
  - i. Yes
  - ii. No

**If answer *yes* to ‘a’ then will be asked question b-e. If answer *no*, then will be asked f.**

- b. How often did you give patients this information?
  - i. Always
  - ii. Sometimes
  - iii. Rarely
- c. Overall, did patients find this information helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
  - i. Yes
  - ii. No
- d. Do you think this information increased a patient’s willingness to inform their at-risk family members of their positive genetic test results?
  - i. Yes
  - ii. No
- e. Optional: Please use this space to further discuss/explain your previous answers to a-d
- f. What were some of the reasons why you do not give patients this information?
  - i. Did not find it to be useful to patients
  - ii. Was not part of my institution’s genetic counseling protocol
  - iii. Too time consuming for me to implement
  - iv. Providing this information was not part of my role as a genetic counselor
  - v. Other: Please list
- g. Optional: Please use this space to further discuss/explain your previous answers to f

23. Providing the patient with a letter specifically addressed to an at-risk family member that includes a description of Lynch syndrome which the family member can share with their health care provider.

- a. Did you use this approach?
  - i. Yes
  - ii. No

**If answer *yes* to ‘a’ then will be asked question b-e. If answer *no*, then will be asked f.**

- b. How often did you use this approach?
  - i. Always
  - ii. Sometimes
  - iii. Rarely

- c. Overall, did patients find this approach helpful to their understanding of whom in their family is at-risk of inheriting Lynch syndrome?
    - i. Yes
    - ii. No
  - d. Do you think this approach increased a patient's willingness to inform their at-risk family members of their positive genetic test results?
    - i. Yes
    - ii. No
  - e. Optional: Please use this space to further discuss/explain your previous answers to a-d
  - f. What were some of the reasons why you did not use this approach?
    - i. Did not find it to be useful to the patient
    - ii. Was not part of my institution's genetic counseling protocol
    - iii. Too time consuming to implement
    - iv. Using this approach was not part of my role as a genetic counselor
    - v. Other: Please list
  - g. Optional: Please use this space to further discuss/explain your previous answers to f
24. How satisfied were you with the approaches, and/or materials you used to help patients disseminate information about their positive mutation results to their at-risk family members?
- a. Completely satisfied
  - b. Very satisfied
  - c. Fairly well satisfied
  - d. Somewhat dissatisfied
  - e. Very dissatisfied
  - f. Optional: Please use this space to further discuss/explain your previous answer
25. In your experience, approximately what percent of all patients needed one or more of these tools to help them share their molecular diagnosis of Lynch syndrome with at-risk family members?
- a. There will be a scale of percentages from 0-100% that the participant can choose from
26. If time and cost were not a factor, what additional resources would you have liked to have for your patients to help them share their molecular diagnosis of Lynch syndrome with at-risk family members?
- a. Open-ended question

## APPENDIX B: Recruitment Notice

Do you have experience as a cancer genetic counselor? If so, I invite you to participate in a research study regarding family communication between Lynch syndrome patients and their at-risk relatives.

Dear genetic counselor,

My name is Tara Biagi and I am a second year graduate student in the Genetic Counseling Program at Brandeis University. I am seeking volunteers to participate in a research project that will be the basis of my Master's Thesis. The goal of the project is to assess genetic counselors' experiences with family communication between Lynch syndrome mutation carriers and their family members and to learn what strategies they have found most useful in accomplishing this goal.

This study is open to all genetic counselors who:

- Have at least six months of cancer genetic counseling experience, either full or part time, within the last five years.
- Have counseled a patient with a molecular Lynch syndrome diagnosis at least once within the past five years.

Study participation involves an online, anonymous survey that will take approximately 20 minutes to complete. Participation in the study is completely confidential and voluntary.

As a thank you for participating in the survey, you will be offered entry into a drawing for one of two \$50 gift cards to Amazon.com

Please follow the link below to access the online survey:

LINK

Please note:

- Participation in this study is **voluntary** and **anonymous**
- The Brandeis University IRB (Institutional Review Board), Waltham, MA, has approved this research study.

If you have any questions or comments regarding this study, please feel free to contact me by email at: [tbiagi@brandeis.edu](mailto:tbiagi@brandeis.edu) or the Principle Investigator, Judith Tsipis, at [tsipis@brandeis.edu](mailto:tsipis@brandeis.edu). Thank you in advance for your participation!

Sincerely,

Tara Biagi, BS

Brandeis University Genetic Counseling Program, Class of 2011