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

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ABSTRACT

Objectives: To explore patterns of communication among families with a Lynch syndrome diagnosis and understand what resources could facilitate family communication.
Methods: 127 probands (i.e., first person in family with identified mutation) and family members participated in semi-structured interviews about: how they learned about the Lynch syndrome diagnosis, with whom they shared genetic test results, confidence in sharing results with other family members, and helpfulness of educational resources.
Results: Both probands and family members were most likely to share genetic test results with parents and siblings, and least likely to share results with aunts, uncles, and cousins. Most participants felt very confident sharing their test results with family members, but reported that certain topics such as cancer risk were challenging to convey. Probands reported the most helpful resources to be access to a specialty clinic or website, while family members described general printed materials as most helpful.
Conclusions: Families affected by Lynch syndrome may experience barriers to communication with more distant relatives, and may benefit from receiving specific resources (e.g., websites about Lynch syndrome, print materials) to facilitate family communication.
Practice implications: Providers could emphasize the need to share information with more distant family members and provide appropriate supportive resources.

1. Introduction

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

A diagnosis of Lynch syndrome is vital since colorectal cancer screening practices can then be better targeted. Individuals with Lynch syndrome are recommended to receive colonoscopies more frequently and beginning at an earlier age as well as other cancer screenings [2,6,7]. The importance of diagnosing Lynch syndrome prompted recommendations to begin screening all newly diagnosed colorectal and endometrial cancer patients through tumor testing via immunohistochemistry or microsatellite instability

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[4,8]. Universal tumor testing identifies nearly twice as many individuals with Lynch syndrome than would be identified by only screening individuals with an early age of onset of cancer [5,9]. Moreover, several models have demonstrated the cost-effectiveness of universal tumor screening [10,11]. However, the effects of such approaches on health and cost outcomes depend on how many additional family members receive cascade testing (i.e., testing biological relatives of individuals with an identified pathogenic mutation) [9,12–14]. Research has shown that cascade testing remains suboptimal; a review by Sharaf et al. noted that first-degree relatives (children, siblings, and parents) of probands (i.e., first person in family found to have a pathogenic Lynch syndrome variant) may accept genetic testing at a rate of 34–52%, and a majority of the time fewer than three first-degree relatives chose to receive testing [15].

Because U.S. health care providers cannot directly contact biological family members, cascade testing is reliant on effective communication about Lynch syndrome between family members to promote genetic testing within families [16]. Previous studies have demonstrated substantial gaps in communication about genetic risk within families. While individuals who have received genetic testing for Lynch syndrome are likely to share their test results with first-degree relatives, they are less likely to share this information with extended family such as second- or third-degree relatives (e.g., aunts, uncles, nieces, nephews, cousins) [17–20]. Other factors that may influence whether or not genetic information is shared include age of family member and extent of experience with familial cancer [21]. Barriers to sharing genetic information may include a lack of close relationships with more distant relatives, or not wanting to cause worry in family members who may be at risk [17,18,21–23]. Individuals may also find sharing information about a Lynch syndrome diagnosis with family members to be burdensome [24]. However, few studies have investigated what resources can support family communication [18,25]. Additional research is therefore needed to understand family communication patterns and the resources that may best support family communication about Lynch syndrome.

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

2. Methods

2.1. Participants and procedure

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

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

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

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

2.2. Measures

2.2.1. Family communication of genetic test results

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

2.2.2. Confidence with family communication

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

2.2.3. Resources received

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

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