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REVIEW

Challenges and opportunities for Lynch syndrome cascade testing in the United States

Lauren E Passero[1] · Megan C Roberts[1]

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Abstract

Lynch syndrome is an underdiagnosed genetic condition that increases lifetime colorectal, endometrial, and other cancer risk. Cascade testing in relatives is recommended to increase diagnoses and enable access to cancer prevention services, yet uptake is limited due to documented multi-level barriers. Individual barriers such as feelings of fear, guilt, and anxiety and limited knowledge about Lynch syndrome as well as interpersonal barriers including complex family dynamics and language barriers limit family communication about Lynch syndrome and prevent uptake of genetic screening for relatives. Organizational and environmental barriers including a shortage of genetics professionals, high costs, and fears of discrimination also reduce cascade testing. These multi-level barriers may disproportionately impact underserved populations in the United States, such as individuals with lower incomes, limited English-speaking proficiency, lower educational attainment, and inadequate access to health systems. Multi-level facilitators of cascade testing include interpersonal support from family members, peers, and healthcare providers, educational resources, and motivation to improve family health. Taken together, these barriers and facilitators demonstrate a need for interventions and strategies that address multi-level factors to increase cascade testing in families with Lynch syndrome and other hereditary cancer conditions. We provide an example of a cascade testing intervention that has been developed for use in individuals diagnosed with Lynch syndrome and discuss the variety of current approaches to addressing these multi-level barriers.

Keywords Lynch syndrome · Cancer · Cascade testing · Barriers · Equity

Introduction

Lynch syndrome is a hereditary cancer condition characterized by up to a 90% lifetime risk of colorectal cancer and a 40% lifetime risk of endometrial cancer due to germline pathogenic variants in DNA mismatch repair genes [1]. Lynch Syndrome-associated cancer risk is best managed through early diagnosis and guideline recommended cancer risk management strategies enabling cost-effective cancer prevention [2]. Unfortunately, most individuals in the US are diagnosed with Lynch syndrome following a significant personal or family history of cancer. Identifying individuals with pathogenic or likely pathogenic variants after a cancer diagnosis signals a missed opportunity in cancer prevention and control. Indeed, up to 98% of individuals with Lynch syndrome remain undiagnosed today [2]. Cascade testing is a guideline-recommended approach for identifying individuals with Lynch syndrome early by systematically testing family members of individuals who were previously diagnosed with Lynch syndrome, starting with first-degree relatives and cascading throughout the family, as needed. In the US, cascade testing typically relies on individuals diagnosed with a pathogenic or likely pathogenic variant to inform at-risk relatives of the need for genetic testing, and then relies on the relatives to seek genetics services individually. Multilevel barriers to facilitating predictive genetic testing among relatives exist such that only 52% of first-degree relatives of patients with Lynch syndrome receive cascade testing, with lower uptake among underserved populations [3]. The presence of barriers to cascade testing at different socio-ecological levels has led to suboptimal use of this strategy for identifying individuals with Lynch syndrome and demonstrates a need for further research and practice

✉ Megan C Roberts
megan.roberts@unc.edu

[1] Division of Pharmaceutical Outcomes and Policy, University of North Carolina at Chapel Hill, Chapel Hill, North Carolina, United States of America

advancement to support probands (i.e., initial family member diagnosed with Lynch syndrome) and relatives in cascade testing [4]. Here, we discuss the multi-level barriers and facilitators to cascade testing present across individual-, interpersonal-, organizational-, and environmental- levels; the need for multi-level interventions to improve the equitable adoption, implementation, and sustainment of cascade testing for Lynch syndrome; and an example of such an intervention developed to address these barriers with insight from invested partners to support cascade testing for Lynch syndrome.

Multilevel barriers and facilitators

Individual and interpersonal barriers and facilitators

Individual and interpersonal barriers include those at the patient, relative, and provider levels as well as interactions between them during cascade testing. Among family members, low knowledge about Lynch syndrome and the value of genetic testing is a major barrier to cascade testing [4–6]. Qualitative data from individuals with Lynch syndrome have highlighted that low awareness about Lynch syndrome and its elevated lifetime cancer risk may result in low perceived benefit of cascade testing among family members, translating to low uptake of testing [4]. Not only do patients and family members lack this knowledge, but also providers. In particular, data suggest that non-genetics providers may lack knowledge about Lynch syndrome, which in turn hinders the process of cascade testing because providers cannot adequately discuss cascade testing or support patients through the process of testing additional family members [4]. Additionally, confusion among patients about the role of their providers in facilitating cascade testing can lead to delays or disruptions in communication with family members about a Lynch syndrome diagnosis [4].

For patients who are aware of cascade testing and its importance for family health, psychosocial barriers can pose an obstacle for communicating with family members to seek cascade testing. Feelings of guilt and anxiety, privacy concerns, and fears of potential stigmatization due to social norms about illness can cause patients to feel reluctant to disclose their Lynch syndrome diagnosis to family members [5, 6]. Challenging family dynamics such as emotional distance or conflict can further impede communication with family members about Lynch syndrome [5]. Finally, even if patients successfully share their genetic status with family members, feelings of fear and information avoidance among relatives can delay or prevent uptake of genetic testing [5].

Cascade testing for Lynch syndrome is also limited by certain interpersonal communication issues such as language discordance and a lack of open communication channels for family members [5]. A lack of proficient skills in a family member's primary language can prevent effective disclosure of genetic testing results, which is compounded by low genetic literacy broadly [7]. Indeed in the United States, limited proficiency in the English language is also associated with lower probability of awareness of genetic testing [8].

Individual and interpersonal factors can also support the process of cascade testing for relatives. The motivation to inform family members through a sense of duty and desire to support family member health can facilitate communication about a Lynch syndrome diagnosis [4, 6]. Solidarity and psychological support from the diagnosed proband as well as peers in support groups and advocacy organizations can also encourage relatives to overcome concerns about genetic testing [5], suggesting the importance of social networks and social support. Finally, recommendations and educational materials for relatives provided by physicians can facilitate the process of genetic testing for family members [5]. The important role of health professionals in guiding cascade testing emphasizes the need to increase provider education about Lynch syndrome and optimize clinical systems to facilitate patient navigation through testing.

Organizational and environmental barriers and facilitators

Organizational policies and procedures can further reduce accessibility to genetic testing services for family members. Relatives often face logistical challenges in identifying and receiving a referral for a genetic counselor to initiate genetic testing given the workforce shortage in genetics professionals in the United States [5, 6]. Beyond a general workforce shortage, there is also a severe lack of genetics professionals who can provide services in languages other than English, limiting access to the genetics services necessary to receive testing for individuals who speak other primary languages [9]. In cases when a genetic counselor can be identified and a referral obtained, many professionals practice in academic centers or clinics which may require family members living in rural areas to navigate long-distances as well as take time off from work to receive services during typical clinic hours—this may or may not be feasible [5, 6]. Interestingly, the shift to virtual cancer genetic counseling during the COVID-19 pandemic, while removing distance and time costs to accessing care, has been associated with lower willingness to consent to genetic testing among patients compared in-person counseling [10]. Thus, expanding

geographic access remains an important issue in increasing cascade testing among geographically dispersed families.

Family members also demonstrate reluctance to complete cascade testing due to concerns about affordability and fears of discrimination in employment, health or life insurance [4–6]. The high cost of genetic testing and a lack of insurance coverage have been reported by patients as reasons for declining genetic testing [4]. Free testing programs offered by laboratories for relatives of newly diagnosed Lynch syndrome patients have been found to address financial concerns and increase uptake of cascade testing by family members [4]. Yet qualitative data has indicated that this alone does not fully relieve concerns about the perceived costs related to cascade testing and potential costs related to management of Lynch syndrome [4]. Along these lines, family members have also expressed concerns about the indirect costs of a Lynch syndrome diagnosis. Concerns that career advancement or employment in jobs with physical or medical requirements could be compromised by a positive test result preclude some family members from pursuing genetic testing after being informed of their need for cascade testing. Additionally, patients also report barriers to receiving testing due to uncertainty about eligibility for medical or life insurance after having Lynch syndrome listed as a pre-existing condition on medical records [4]. While the Affordable Care Act provides protections for medical insurance in the United States, protections from life insurance discrimination remain a concern.

Finally, while evidence suggests that centralized cascade testing and provider-mediated cascade testing may be effective means for implementing cascade testing, there is not yet a centralized cascade testing program for Lynch syndrome in the US. Such a program could be especially challenging in a fragmented healthcare system. Further, Health Insurance Portability and Accountability Act and variation in state genetic privacy laws to protect patient privacy may limit direct disclosure of Lynch syndrome risk to relatives by healthcare providers [3]. However, despite these implementation barriers, there is increasing interest in establishing the feasibility of such an approach as is being done for other Tier 1 genetic conditions [11].

A cross cutting barrier: health inequities

Barriers to cascade testing may be heightened among underserved populations who experience additional barriers to the health care system due to historical injustices imposed by the healthcare systems [12]. Recent research has identified unique challenges faced by non-English-speaking patients in the use of genetics services. Language concordance is known to support patient-provider communication and improve understanding and decision-making in genetic counseling, yet patients have reported limited access to genetics professionals practicing in non-English languages [4, 13]. As noted previously, limited English proficiency is also correlated with low awareness of genetic testing and a lack of informational resources available in non-English languages is a known barrier to the uptake of genetic cancer risk assessments [8, 14].

Strategies to overcome multilevel barriers

Given the multilevel barriers associated with cascade testing, there is a need for effective interventions that can operate at multiple socio-ecological levels to eliminate barriers and improve the adoption of cascade testing. However, most current interventions to support Lynch syndrome cascade testing target only barriers at a one or two socio-ecological levels. For example, educational materials for patients can address barriers such as a lack of knowledge and communication skills [15]; However, patient education does not support relatives in locating a genetics provider or affording testing costs. Interventions such as physician referral letters recommending genetic testing for at-risk relatives can provide critical information about Lynch syndrome and simplify health system navigation for relatives, but do not offer psycho-social support to patients and relatives who feel fear or concerns about their current or potential diagnosis [16]. Additionally, many current interventions do not specifically account for unique challenges faced by patients across underserved communities who have lower access to healthcare systems and resources to engage in cascade testing and benefit from cancer prevention. Finally, a prior scoping review of cascade testing interventions more broadly found that few rigorous evaluations of cascade testing models and interventions have been conducted [6].

An example strategy

In response to multilevel barriers to cascade testing, our research team developed a theory-based, intervention informed by key parties in cascade testing that considers patient, family, provider, and system barriers for cascade testing for LS through an intervention called “Let’s Talk about Lynch syndrome and your family” (Let’s Talk) [17]. Let’s Talk is a theory- and evidence-based cascade testing intervention developed to systematically address multilevel needs of this population. It was designed to be delivered to the proband in the format of an interactive workbook. We used intervention mapping – a 6 step process for developing an intervention that integrates the perspectives of key leaders and implementing partners in intervention design. Let’s Talk was informed by qualitative interviews (among patients, providers, administrators, policy-makers, and

patient advocacy organizations, $n = 60$) [4], a systematic literature review [5], and an Advisory Panel. The intervention includes four core components: information chunking, guided practice for family communication, planning coping responses, and gain-framing messaging for patients, their provider and family members. Let's Talk content also addresses obstacles to cascade testing including interpersonal dynamics (e.g., identifying relatives that the proband needs help contacting either by a friend, family member or provider), limited knowledge about Lynch syndrome (e.g., educational materials, glossary), fear, guilt or stigma about Lynch syndrome (e.g., planning coping responses, gain frame messaging), low social support (e.g., linked resources for social support groups), and low provider access (e.g., linked resources for identifying genetics providers across geographic areas). Initial paper-based content of Let's Talk has been tested among patients for its usability ($n = 10$) and was found to be highly acceptable, easy to use, and superior to existing resources [17]. Ongoing work seeks to make this intervention more accessible and interactive in an online version.

Given concerns around the availability of non-English language resources, we recently adapted Let's Talk into Spanish (i.e., Hablemos) to assist Spanish-speaking patients in overcoming challenges to cascade testing with family members given the limited availability of language-concordant care, and genetic resources. Hablemos content was professionally translated into Spanish as a direct translation of the English content and reorganized into a paper- and pdf-based workbook format. We conducted a qualitative usability study with five bilingual adults diagnosed with Lynch Syndrome to examine the preliminary usability of Hablemos for Spanish-speaking patients and to gather data to enable future areas for cultural tailoring (Supplemental File 1).

Similar to findings in English-speaking patients [16], we found that all participants liked the educational messaging in the workbook explaining the definition of Lynch syndrome, the cancer risks associated with the condition, and strategies for cancer prevention. Most participants further commented positively on the motivational content in the workbook describing the benefits of Lynch syndrome genetic testing and encouraging the uptake of cascade testing. All participants emphasized the value of the interactive workbook activities in directing communication about Lynch syndrome and genetic testing with family members. Consistent with other qualitative work highlighting limited knowledge of Lynch syndrome cascade testing and appropriate educational resources, most participants had not previously seen a similar resource for Lynch syndrome family cascade testing delivered in Spanish [5, 17] and expressed that they would recommend the workbook for use with patients in clinical settings. The participants also had several suggestions for improving the workbook including increasing the font size, reducing the number of pages, simplifying the language, and adding more graphics to convey the meaning of difficult concepts. This feedback around the visual formatting, length, and language complexity of Let's Talk/Hablemos was unique among the bilingual participants compared to prior usability testing among those for whom English was their primary language, suggesting potential areas for further co-design and tailoring. Similarly, work is underway to adapt and tailor Let's Talk for other hereditary conditions, including hereditary breast and ovarian cancer syndrome and familial hypercholesterolemia. Thinking forward, most participants expressed interest in an electronic delivery format, primarily a web-based version, and emphasized the value of providing the workbook in multiple formats to meet the needs of different users. These findings aligned with usability testing for the English-language version as well.

While Let's Talk's design takes into consideration multiple participants and multilevel barriers related to cascade testing, it is primarily a patient-level intervention with the option of provider-led support in (1) identifying which relatives to contact and (2) contacting those relatives who the proband does not feel comfortable reaching out to or having a family member or friend reach out to (activity 1 in the workbook). This flexibility offers the opportunity for this intervention to target multiple partners in cascade testing, including family, friends, and providers but still heavily relies on patient engagement.

A path forward

Let's Talk/Hablemos represents a theory and evidence-based cascade testing intervention aimed at addressing multiple barriers for cascade testing in English and Spanish. In our initial work, our advisory panel prioritized barriers and facilitators based on their importance and changeability. As a result, not all barriers were addressed, and thus many opportunities remain to improve the adoption and implementation of cascade testing in the US beyond the development of supportive interventions delivered directly to patients and providers.

Other approaches to cascade testing in the US have expanded beyond proband initiated approaches to hybrid approaches engaging relatives as well. For example, Caswell-Jin and colleagues developed an online program in which both known carriers and first-degree relatives of carriers can enroll in an online family testing program including a pre-test video and online consent process [18]. Offered by a genetic testing laboratory, individuals who have a pathogenic variant can provide email addresses for their

relatives who then receive an email with the pre-test video and consent information directly from the program. Those first-degree relatives who are enrolled then receive results and these results are provided by a genetic counselor if positive. In parallel, first-degree relatives who learn about the program can directly enroll into the program.

For first-degree relatives to participate directly with the program, the proband must have disclosed receiving a pathogenic variant to the relative from the outset. Thus, in a sense, this pathway, while directly targeting first degree relatives, does still rely on information flow from the proband. This gap is addressed by also intervening with probands and providing direct-contact to first degree relatives by the program. This approach circumvents provider and health system barriers related to competing demands and relative priority of cascade testing by delivering it through a genetic testing company and connecting individuals with a positive result directly to a genetic counselor. Understanding the extent to which relatives are connected back to their health care teams (or new care teams) would be an interesting future direction.

A direct-contact strategy for cascade testing removes the burden of cascade testing from the patient and utilizes an outside party (often a provider service) to contact relatives of individuals recently diagnosed with a hereditary condition. Primarily based on non-US studies, a recent meta-analysis suggests that compared with patient-mediated contact, direct-contact strategies for cascade testing are associated with increased rates of genetic counseling and testing among relatives [19]. Recent work by Henrikson and colleagues has looked at patient and family preferences for a health system-led direct contact approach for cascade testing for hereditary cancer syndromes (hereditary breast and ovarian cancer and Lynch syndromes) [20]. Using a qualitative human-centered design approach, the team identified a draft set of requirements for a direct-contact cascade testing strategy aligned with patient and family preferences. These requirements included: patient consent prior to a provider contacting their relatives, autonomy for relatives to decide what information is received and how to use it, multiple lines of communication with relatives, specific communication points for contact with relatives, a clear recommendation for testing and follow-up, and resources for the proband to share with their relatives [20]. Patients and families felt that communication to the relative should include notification that their relative gave permission for the provider to call, the reason for contact, and information about inherited cancer risk, cost, coverage, privacy, and non-discrimination laws. The strength of this approach is that it helps to address proband- and interpersonal-level barriers to cascade screening and directly provides the necessary information to relatives. Health care system approaches do not, however, address health system level barriers such as competing demands and low relative priority related to cascade testing. Thus, the feasibility and sustainability of such programs must be considered and further studied.

Other approaches to direct-contact include centralized cascade testing models, which include a coordinating center that directs cascade testing. Qualitative data from providers, administrators and patient advocates have pointed to centralized cascade testing as a gold standard for increasing cascade testing rates [4]. Deemed one of the most successful strategies for cascade testing, the Netherlands implemented a national cascade testing program for familial hypercholesterolemia with over 80% of eligible relatives receiving testing [21]. Despite the success of centralized cascade screening for FH in the Netherlands, after twenty years, government funding of the program was discontinued. However, with buy-in for centralized cascade screening across key partners, a non-profit organization was developed and took over national coordination of cascade testing, demonstrating yet another model for funding such a national, centralized program [21].

Increasingly consideration of how to adapt such a model to the US context has been considered. A recent proof of concept study for a centralized cascade testing model for familial hypercholesterolemia showed the potential feasibility of this strategy in the US. McGowan and colleagues adapted the Dutch model for FH cascade testing to the US context in partnership with the Family Heart Foundation in which individuals diagnosed with FH are referred to the Family Heart Foundation [11]. In turn the Family Heart Foundation educated the proband about the need for cascade testing, and upon consent, supported conversations between probands and their relatives as well as directly contacted relatives for cascade testing, provided education to relatives, and coordinated cascade testing and lipid testing for consenting relatives [11]. As a final step in this approach, the Family Heart Foundation then supported relatives with a positive result to identify specialty care and engage in additional cascade testing in their family. Ongong work is examining the feasibility of this strategy further (Department of Defense Award Number: W81XWH-21-1-0645, PI Ahmad), and if successful this approach could be adapted for Lynch syndrome in partnership with other advocacy organizations.

These strategies are but a few of many cascade testing approaches being tested and used in practice. For example, other innovative approaches focus on (1) supporting the connections between probands and close relatives as well as distant relatives through the use of social networks [22], (2) using interactive videos to model family discussions [23], and (3) chatbots to support education about genetic testing [24]. While each of these strategies addresses subsets of implementation barriers, the comparative and cost

effectiveness of these approaches remains unknown. Future work to understand which strategies and components of strategies are optimal (e.g., educational v direct contact) to meet the needs of different patient groups is needed to advance cascade testing.

Finally, because cascade testing relies on the initial diagnosis of a proband in a family, improving initial genetic testing is another opportunity to advance cascade testing uptake. Genetic testing in probands remains suboptimal limiting the potential impact of cascade testing in family members. Efforts to improve identification of individuals through universal tumor screening for Lynch syndrome among patients with colorectal and endometrial cancers as well as emerging calls for population genetic screening for LS and other conditions may enhance these identification efforts. These efforts should be paired with cascade testing strategies as well as interventions focused on ensuring appropriate follow up for family members who test positive.

Though little has been done in this area, ideal implementation of cascade testing will support all aspects of care from proband diagnosis, to communication of risk status to family members, to genetic testing in relatives, and to follow-up programs to ensure appropriate risk management for cancer. Thus the development of interventions or linkages between interventions across this continuum will be crucial for ensuring improved outcomes for these high risk families. Finally, as more tools are developed in cascade testing, and other aspects of the care continuum (proband diagnosis and long-term risk management), the call must shift from developing *more* tools to *implementing* the tools that we know work. The field of implementation science can support the integration of cascade testing best practices and evidence-based interventions into clinical and public health settings. Through comparative effectiveness studies, implementation scientists can identify which strategies are efficient and cost-effective for delivering and sustaining cascade testing in practical settings. Yet, the field of implementation science in cascade testing, while growing, is still underdeveloped and in need of dedicated research to advance the field [25]. Numerous calls for implementation science in precision public health propose that the field can contribute towards realizing the public health impact of interventions like cascade testing [26, 27].

Finally, current research has also emphasized the need for ongoing improvement as it relates to the development of tailored, culturally- and linguistically-appropriate interventions for cascade testing for Lynch syndrome. Public health leaders recently called for increased action to address disparities in access and utilization of genomics-based healthcare including through the development of culturally and community-tailored genomic educational materials for patients and families [12, 28]. Answering this call is critical for ensuring precision public health.

Conclusion

The numerous barriers to cascade testing for Lynch syndrome at individual, interpersonal, community, and policy levels emphasize the need for multi-level interventions to facilitate the equitable implementation of cascade testing for Lynch Syndrome across the care continuum. There is a need for the development of multi-level solutions, both at an individual and interpersonal levels, as well as at the organizational and policy levels to overcome the multi-level barriers to cascade testing faced by patients, particularly in underserved communities. Research to develop innovative and effective interventions that support the cascade testing process at all socio-ecological levels is an important step toward addressing the critical and timely need to improve health equity in the implementation of Lynch syndrome cascade testing and other applications of genomic medicine.

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Data availability The data that support the findings of this study are available from the corresponding author upon reasonable request.

Declarations

Ethical approval The qualitative study reported in this paper was granted an exemption from human subjects review by the Office of Human Research Ethics at the University of North Carolina at Chapel Hill (Study Number 21–0643).

Informed consent All participants provided verbal informed consent prior to study enrollment. All procedures followed were in accordance with US Federal Policy for the Protection of Human Subjects.

Conflicts of interest Lauren Passero is a predoctoral fellow funded by Bristol Myers Squibb. Dr. Megan Roberts' spouse hold stock in Merck and Thermo Fisher Scientific. The content is solely the responsibility of the authors and does not necessarily represent the official view of

the NIH.

Animal studies No non-human animal studies were carried out by the authors for this article.

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