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# Clinician-Stakeholders' Perspectives on Using Patient Portals to Return Lynch Syndrome Screening Results

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## Abstract

Test results for genetic conditions, such as Lynch Syndrome (LS), have traditionally been returned by genetic counselors or other providers who can explain results implications and provide psychosocial support. Returning genetic results through an Electronic Health Record's patient portal may increase the efficiency of returning results and could activate patient follow-up; however, stakeholder input is necessary to determine acceptability and appropriate implementation for LS. Twenty interviews were conducted with clinicians from six specialties involved in LS screening that represent a range of settings. Data were analyzed using directed content analysis and thematic analysis across content categories. Participants felt that patient portals could supplement personal calls, but the potential sensitive nature of LS screening results indicated the need for caution. Others felt that LS results could be returned through portals if there were clear explanations of the result, reputable additional information available within the portal, urging follow up confirmatory testing, and a referral to a genetics specialist. Patient portals were seen as helpful for prompting patient follow-up and providing resources to notify at-risk family members. There is potential for patient portals to return LS screening and other genetic results, however we raise several issues to resolve before implementation is warranted.

### Keywords

Lynch Syndrome; Precision Medicine; Patient Portals; Genetic Testing; Return of Results; Hereditary Nonpolyposis Colorectal Cancer; Implementation Science; Ethics

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**CONFLICT OF INTEREST STATEMENT**

Authors Korngiebel, West, and Burke declare that they have no conflicts of interest.

**HUMAN STUDIES AND INFORMED CONSENT STATEMENT**

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all participants for being included in the study.

No animal studies were carried out by the authors for this article.

## INTRODUCTION

Colorectal cancer (CRC) is the second leading cause of cancer deaths in the United States. About 3% of CRCs are attributed to Lynch Syndrome (LS), a heritable condition that confers a high lifetime risk of several types of cancer, particularly CRC (~80%) and endometrial (~60%) (Giardiello et al. 2014). LS is a highly penetrant, autosomal dominant condition, and there are evidence-based interventions for patients and their at-risk family members (Hampel 2014). LS represents a well-evidenced case of genomic medicine, ready for clinical implementation, and universal screening of CRC tumors (UTS) has been recommended (Berg et al. 2009). UTS currently involves immunohistochemistry (IHC), microsatellite instability (MSI) testing, or both. Each screening method has similar sensitivity and specificity: sensitivity for MSI testing is about 89% for *MLH1* and *MSH2* and about 77% for *MSH6* mutations, with specificity of about 90%; sensitivity for IHC testing is about 83% for all three mismatch repair genes, and specificity is about 89% (Palomaki et al. 2009). Positive screens are usually returned to patients in-person, at which time follow-up confirmatory genetic testing is recommended. However, many patients whose tumors screen positive for LS do not follow-up (Beamer et al. 2012; Cragun et al. 2014). While genetic counseling is optimal, the availability of genetic counselors in the U.S. does not meet the service needs in the era of precision medicine, underscoring the importance of additional measures to address patient needs.

Leveraging Information Technology (IT) can support engaging patients in their own healthcare (Maher et al. 2015). One component of health IT with potential implications for patient uptake is returning screening results electronically via an Electronic Health Record (EHR) patient portal. Although there is a robust discussion on the return of genetic results (Knoppers et al. 2015; Wolf et al. 2015), including carrier screening (Korngiebel et al. 2016; Leo et al. 2016), in research (Fullerton et al. 2012; Jarvik et al. 2014), or of incidental findings (Yu et al. 2014), and a growing literature studying patient portal usage (Amante et al. 2014; Clark et al. 2015), there have been few studies concerning the return of genetic results via patient portals (Sweet, Hovick, et al. 2016; Sweet, Sturm, et al. 2016).

Several ethical questions remain to be resolved regarding the acceptability and appropriateness of using patient portals to return LS results. In addition, the majority of patient portals are add-ons to commercial EHR software packages, often designed without user input (Ratwani et al. 2015). We aim to explore clinician stakeholder perspectives on the use of patient portals tethered to the EHR to return results of Lynch Syndrome screening, as an example of genetic medicine ready for implementation.

## METHODS

### Participants

Twenty key informant interviews were conducted with specialists associated with implementing LS screening in CRC patients: medical geneticists, genetic counselors, pathologists, oncologists, gastroenterologists, and primary care providers (Table I). Participants were identified using purposive sampling to facilitate representation of relevant medical specialties and a range of health care settings, and snowball sampling to include

experts outside of the study team's professional networks. Two to five individuals from each specialty were interviewed.

### Procedures

A semi-structured interview guide was created through expert consensus, including all authors and a project advisory board. The first half of the guide focused on general and technical questions regarding UTS implementation (West et al. 2017), while the second half explored the return of positive LS screen results using a patient portal, including questions on appropriateness, information to be included or excluded with the screening results, and in what ways a patient portal might be leveraged to activate patients to follow up on positive screens (see Table II for sample questions).

The lead investigator (DK) conducted nineteen interviews by phone and one inperson interview between September 2015 and August 2016. Each interview lasted 30–60 minutes. Interviews were audio-recorded, transcribed, and de-identified. The study was approved by the University of Washington Institutional Review Board and all participants provided informed consent.

### Data analysis

All transcripts were coded using Atlas.ti v.7 qualitative analysis software. Two authors (KW and DK) used directed content analysis to deductively code the data based on interview protocol domains, complemented by inductive coding as novel ideas were identified (Hsieh et al. 2005). Textual data were then analyzed across coding categories to identify themes and interpretation verified by consensus (Ryan et al. 2003).

## RESULTS

Participants offered varied insights on the appropriateness of using patient portals to return LS screening results, the content to include with these results, and the potential role of patient portals in facilitating patient and family follow-up for positive screens.

### Appropriateness of returning LS results via patient portals

Participants indicated that returning LS screening results through patient portals should include considerations of whether and in what ways screening results are of a sensitive nature, and how that sensitivity might affect decisions to return results via patient portals. Furthermore, participants discussed how returning results by portal would occur (including consideration of patient preferences).

**Potentially sensitive nature of LS screening results—**Participants felt that sensitive information should be returned by a person, not merely electronically, to ensure psychosocial support if needed. However, some mentioned that it is not clear how sensitive the nature of positive LS screening results are for patients. Participants expressed concern that LS results might be akin to other emotionally fraught test results, or those which would have repercussions beyond the patient—such as other heritable conditions or sexually transmitted diseases. As several participants explained,

> "If they have a cancer…that's a personal call…When I do tell patients about Lynch Syndrome there are a lot of questions that have to be answered before they really fully understand it.…That would be like telling a young woman, 'Gee, your genetics came back. You have Turner's Syndrome. You're infertile. Go forward and have a good life.'…Will the outcome change? No. But they feel supported if they can at least ask questions…Something less sterile than an LED screen." (P20, gastroenterologist)

> "In genetics, our practice has been that we really like to personalize the results for the patients…They [hospital and EMR administrators] want to be able to release any result—as soon as it's available—to the patient. You could have a patient click on an MRI image report that says that they have ten metastatic lesions in their brain. I mean really quite shocking, distressing, life-altering information without the benefit of being able to talk to somebody about it." (P17, clinical geneticist)

> "Email is very, convenient, but it shouldn't be used for everything. The same thing as with the web portal for communication of sensitive results, and HIV results would be an example of that. Just sending email saying, 'Hey, your HIV test was positive. Let me know if you have any questions.' Click…I mention HIV as an extreme example, or same thing as your biopsy's positive for cancer. I don't think all genetic tests have that same sort of implication. I think that would need more review." (P5, primary care provider)

For genetic specialists, the hesitation related to the likelihood of inaccurately communicating results that are both sensitive in nature and complicated, in particular with respect to the need for follow-up after a positive UTS finding.

> "I don't think people understand necessarily the difference between a screen and a positive test." (P6, clinical geneticist)

**Positive Lynch screens could be returned via a patient portal, with other support—**A few participants offered that electronic portals could provide results, as long as there were additional methods for patients to receive personal support, either before or after results were available electronically.

> "I think it certainly could be the first line. Then…they either go and get in touch with genetics or, vice versa, they talk to genetics first and [then] they have access… through the patient portal." (P11, genetic counselor)

> "I usually err on the side of calling my patients when it's a result that I want to make sure I explain to them fully…I will call the patient first, and then send them the results via their patient portal so that they have it in their records officially, which I think is where that is really useful for them. I think yes, you want to make it available to them on their patient portal, but I don't want that to be the only contact they get with it." (P9, primary care provider)

**Accommodation of patient preferences for mode and timing of communication—**Several participants felt that patients' varying circumstances and communication preferences should inform their return of results experience and offered

differing thoughts on how patients may prefer to receive screening results. For example, for patients undergoing intensive treatment, it is likely that sending results through a portal would not be effective and, even if read, could layer on additional worries:

> "That would be a bad time to point that out…'You just had surgery. Now, we're worried about all these other cancers.' " (P12, genetic counselor)

By contrast, if the patient were not undergoing extensive treatment, then return of results via portal might be appropriate:

> "The person, on the other hand, with a polyp that's removed during colonoscopy,… I suppose you could [return results] after that person has been in, had that done, gone back home. They'll maybe be looking at lots of results on the patient portal for many other things. Maybe for them, it would be okay." (P4, primary care provider)

One geneticist explained that, in their practice, patients want breast cancer-related genetic results electronically:

> "Now people just want to know their results of breast cancer testing by their secured email…which I find astounding…Breast cancer patients…we see them once, we sent them information before they get to our clinic… We will let them know by secure email and then we will follow-up as necessary." (P6, clinical geneticist)

A primary care clinician preferred to return concerning results personally but also accommodated patient communication preferences.

> "Sometimes when that happens, if I know a patient well and I can't reach them for some reason, [and] they want to communicate through the portal, I will." (P10, primary care provider)

One concern regarding using the portal as the sole means of results conveyance included generational differences; younger adults might prefer to use portals whereas older adults might not:

> "My mom would never in a million years think about going to a patient portal to get any results, and yet, I'm sure that's what my kids would do without a second thought." (P16, genetic counselor)

## Content to Include with the Electronic Return of Results

Participants were asked what content they would recommend accompany LS screening results returned via a patient portal. Their responses focused on ensuring there was a qualified person identified would could provide follow-up and recommendations for the type and amount of content to include.

**Providing an appropriate communication point person—**Participants indicated that determining the appropriate person to provide follow-up personal communication could be a significant challenge when the patient portal is the primary means for results conveyance. While some felt genetic counseling should be the next referral, it was acknowledged that

upon electronic receipt of results, many patients are likely to contact the provider they have already seen for treatment, even if that provider is not the recommended contact to discuss results. One pathologist posited,

> "If you're a cancer patient, you have a special relationship with whoever's treating your cancer…You're going to talk to your cancer provider first…Even if it says call medical genetics, that's what going to actually happen." (P7, pathologist)

**Amount and type of information to return—**Participants commented on both the quantity of information returned with LS results as well as key content to include. Some felt that positive LS screens could be returned by patient portal if certain informational conditions were met, such as clearly explaining the difference between a screen and a diagnostic test, with links to reputable information sources, and next steps. Still, balancing accuracy with accessible information for patients was seen as a challenge.

> "Explaining as succinctly as possible but as accurately as possible what this means. If the main salient points are… it's screening. It's not diagnostic; it's only a risk prediction tool. Even then I think some people will have a hard time understanding. 'What does that mean, I'm at high risk?'" (P1, gastroenterologist) "It would have to be really, really clear information, like 'this does not mean you have anything'… You'd probably want to have numbers associated with it…some ranges of this result means it's about an X percent chance that you might have Lynch Syndrome. Something really concrete…Then really clear instructions for…follow-up." (P7, pathologist)

However, other participants warned that less, but still accurate, information would provide meaningful context without overwhelming the patient:

> "A little bit about what Lynch Syndrome is, just the bullet points. Emphasize that this is a screening test that they had. It is not diagnostic…If you use a patient portal, then you definitely have to put in a one-page thing… Otherwise, patients are going to get overwhelmed and freaked out." (P12, genetic counselor)

> "We tend to do a poor job of really explaining things thoroughly, especially when rushed for time or space. That's why I think leaving it simple to say, 'We're looking for hereditary causes for colorectal cancer. This is rare, but as part of our comprehensive care, we're screening you as well. We would recommend a genetic counselor to help explain this more thoroughly.' " (P19, medical oncologist)

For many, keeping information basic but offering links to additional, reputable resources for patients to reference was a reasonable option to satisfy a range of patient information needs.

> "I think it is good to, as much as you can, explain what those results mean as part of that result popup, and then a link to reputable information. I know, like with a cholesterol screening,…it has a link to the National Institutes of Health if people are looking for more information on what that means. That comes with that result." (P9, primary care provider)

Regardless of the amount of information conveyed, the information must be meaningful to the patient, so some participants recommended having multiple specialists co-create and

standardize the language, relieving individual providers (especially non-genetic specialists) from having to explain positive screen results.

> "The language for that result…should be developed by a genetic counselor, and perhaps a pathologist together, so that the person doesn't see that and assume that their whole family's at risk…I think you have to be able to provide that result, with the appropriate language to make it meaningful." (P8, pathologist)

> "In a patient portal you can imagine…not having people rewrite it every time… so some sort of set language. Again, having that standardized so it's not dumbed down, or editorialized by the gastroenterologist, or the surgeon, or the family physician who doesn't quite understand the significance of it." (P4, primary care provider)

### Using patient portals to leverage the patient as the key actor

Given the complexity of the healthcare delivery system, the patient may be the best individual for ensuring that follow-up testing remains on the radar. Participants agreed that the portal could be leveraged to support patient follow-up and assist in family outreach. The portal can send reminders and offer easy links to scheduling.

> "I think that ultimately the patient is the only consistent actor in the healthcare delivery system. Communication between providers and healthcare systems is always prone to failure of communication…That has led us to think much more about how we engage the patients to have a successful role as the common actor in the healthcare system, as being the prompt and the recipient so this information… wouldn't completely fall through the cracks." (P14, medical oncologist)

> "If they get it it's like, 'Click here to schedule your appointment with a genetic counselor.' That would be fantastic." (P13, gastrointestinal oncologist)

Likewise, the patient may also be the best actor to reach at-risk relatives, and the portal can provide resources to prompt and encourage family outreach. One genetic counselor saw an opportunity for patient portals to ask patients to,

> "'Fill in the cancers in your family.' That might get them to thinking, 'Gee, maybe this really is an important issue because I did have those two uncles who died of stomach cancer when they were 50.'…That might encourage them to follow-up. I would think you would want to also—if you don't hear back from the patient—have the system automated to re-contact them." (P2, genetic counselor)

A pathologist saw the portal as filling an important care need, which providers may not have time to address:

> "One of the challenging things is that medical geneticists and genetic counselors and oncologists spend a lot of time on… and they might not spend as much time as they could on, just good counseling on how to contact the family members…I don't know how to put it in the patient portal but that would be really cool." (P18, pathologist)

## DISCUSSION

Providers involved in LS screening were asked to discuss using patient portals to return LS screening results. Generally, the use of patient portals was considered inevitable, particularly given that patients have a legal right to their medical records, and will have the ability to see their test results, regardless of their providers' views on how those results are returned. Our study raised several unresolved issues that must be addressed for appropriate use of patient portals for returning LS results, with implications for other genetic results. Broadly, these areas include sensitive nature of the results, what constitutes appropriate information to contextualize results, accommodating patients' varying usage of portals, and leveraging portals for patient activation.

Most felt that LS screening results should be considered results sensitive in nature that warrant person-to-person return. Most of the comparisons participants provided to demonstrate similarly sensitive results involved sexually transmitted diseases (e.g., HIV, Chlamydia), diseases that physicians are required to report (Chorba et al. 1989) and for which there are public health measures to notify contacts. Both genetic results and infectious disease results could have implications beyond the patient. Australia's recent guidelines for returning heritable condition results to family members are rooted in a concern for this reality (Otlowski 2015). However, in the United States, unresolved debates about a "duty to warn" have generated uncertainty about returning genetic screening and test results to others potentially affected (Weaver 2016; Wouters et al. 2016).

Non-genetic screening tests are routinely delivered by patient portals, despite many of them testing for conditions with recognized heritability (e.g., hypertension). Further, in an assessment of using portals to return abnormal (and potentially concerning) radiology results, patients did not express anxiety about this method of conveyance (Reicher et al. 2016). Interestingly, according to some of our participants, returning breast cancer genetic results electronically is considered acceptable to some patients, pointing to a potential shift toward the normalization of electronic return (although it is possible that breast cancer is a special case due to public awareness campaigns). Questions remain about whether patients consider LS results to be sensitive in nature or whether this shift among patients toward acceptance of receiving genetic results electronically is becoming widespread, and which factors support patient acceptance. If this becomes normalized, returning results via patient portals under certain circumstances, and in keeping with patient preferences, may also become more acceptable.

We found limited consensus on the appropriate amount and content of information to accompany LS results. Most agreed that this should include the meaning of a screening result, implications for relatives, links to reputable, patient-friendly information, and explicit next steps. Several participants acknowledged the significant challenge of effectively conveying the difference between a screening and a diagnostic test, a concern that has been described in the prenatal screening literature (Williams et al. 2015). However, there were differing views on the level of detail to provide. The literature concerning returning results bears out this tension between protecting the patient from being overwhelmed with or confused/worried by data, to crossing the line into paternalism by withholding information

(Fernandez 2008). Finding the balance for the return of electronic genetic results will be challenging and cannot be determined without diverse patient input.

In addition, provider communication styles on portals have been shown to vary widely, from too much medical language to overly brief messages (North et al. 2014), and it has been suggested that as IT becomes a larger part of clinical care and patient-provider interactions, that how to effectively communicate with patients through portals should be incorporated into medical education (Keplinger et al. 2013). To address these types of issues, the potential for a portal to provide vetted information sources is being explored (Borbolla et al. 2014) and work on best practices is underway (McDougald Scott et al. 2013).

Some participants expressed concerns that using the portal as the primary means of results conveyance could exclude certain patient populations (e.g., older patients, patients located in rural or under-resourced areas). This concern is supported by a study conducted across four nephrology clinics, which reported that predictors of nonportal use were older age, African-American race, Medicaid enrollment, and lower neighborhood median household income (Jhamb et al. 2015). This study showed a modest correlation between portal use and blood pressure control, which could be concerning, as low use of portals among these populations may exacerbate health disparities. Another study of portals deployed in primary care showed that African-American and Hispanic patients were less likely to use portals but in contrast to the previous study, increased age was associated with increased portal use to manage health conditions (Krist et al. 2014). A trend among older adults to access and use patient portals is borne out by other small studies; however older users are still not the largest groups of patient portal users (Crotty et al. 2015; Turner et al. 2015). To be mindful of the disparate use of patient portals across population groups, results return should incorporate the needs of many potential populations in the content and information delivery modes and use patient friendly language that considers varying health literacy and numeracy levels (Apter 2014; Gu et al. 2015).

## Practice Implications

Given the increase in the use of genetic testing in clinical practice in the U.S., the potential for using electronic methods to facilitate return of genetic results is worth considering. Specialty ownership for implementing universal tumor screening for Lynch Syndrome is highly complex (West et al. 2017), and patient activation may be important for ensuring follow-up care. Our data show that although interacting with genetic specialists is ideal, providers anticipate that patient portals could be used to encourage follow-up of positive screen results. However, clinicians play a crucial role in encouraging patients to enroll in and use portals (Amante et al. 2014; Black et al. 2015). In addition, some studies demonstrate that patients respond to electronic reminders sent through the portal, to schedule screenings and other preventive services (Irizarry et al. 2015). Yet vulnerable patients and those with limited health literacy need engagement methods that assist them in effective portal and information use (Tieu et al. 2015; Tieu et al. 2016). Research findings in these areas, combined with the Precision Medicine Initiative and various inducements to support meaningful use of EHRs (Ahern et al. 2011), may foster a much-needed focus on the most appropriate ways to return genetic-related results electronically.

### Study Limitations

This study is exploratory and is limited to providing empirical data to inform implementing return of one type of genetic screening results to patients using an EHR portal. Further research is needed to determine the applicability of the themes we report here to other cases of genetic medicine. The study sample was limited to provider perspectives, but any decisions about electronic return of genetic results must include diverse patient input concerning thresholds for the sensitive nature, content, and delivery mode and timing.

### Research Recommendations

If the portal does not meet broad patient information and activity needs, then adoption will be hindered (Otte-Trojel et al. 2015). Therefore, we recommend research to explore: diverse patient perspectives on LS genetic result's sensitive nature, patient information needs, and what portal content would support patient activation. Further investigation should include whether genetic tests, in general, differ from other potentially upsetting non-genetic test results returned electronically (e.g., radiology reports), whether electronic return of results can adequately address the nuances of genetic results without an in-person consult, which formats are accessible to patients (e.g., textual, visual, or audiovisual), and how genetic information might be provided through patient portals, such as via external hyperlinks to an outside resource like MedlinePlus (Borbolla et al. 2014)—thus enhancing generalizability beyond the specific software used. The above topics could be explored through a user-centered design approach, identifying patient information needs to guide creation of genetic report templates (and/or externally linked material).

## CONCLUSION

With precision medicine, opportunities increase for patients to manage their health information electronically and for clinicians and healthcare organizations to leverage patient-centered IT. Our data show that clinicians are cautiously open to leveraging EHR-based patient portals to return LS genetic screening results, provided several key issues are resolved.

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**TABLE I**

Participant Demographics (n=20)

| | |
|---|---|
| **Gender** | |
| Male | 11 |
| Female | 9 |
| **Medical specialty** | |
| Medical geneticist | 3 |
| Genetic counselor | 4 |
| Pathologist | 3 |
| Oncologist | 3 |
| Gastroenterologist | 2 |
| Primary care provider | 5 |
| **Setting** | |
| *Academic medical center* | 13 |
| Past or present UTS implementation | 5 |
| No UTS Implementation | 8 |
| *Community-based clinic* | 7 |
| Past or present UTS implementation | 2 |
| No UTS Implementation | 5 |

## TABLE II

Sample Interview Questions

| **Sample leading questions** |
| --- |
| Could, or should, a patient portal be used to electronically return positive screen results to patients? |
| IF YES or MAYBE, could you explain your position? |
| If NO, could you say more about that? |
| If screening results are returned using a patient portal, then what other information do you think should also be provided along with the results? |
| **Follow up prompts** |
| Information about Lynch Syndrome? |
| Information about local counseling resources? |
| Information about local genetic testing resources? |
| Information about implications for family members? |
| A brief statement that genetic counseling is recommended (or not recommended) and why (i.e., what a positive screen means)? |
| The contact details for scheduling genetic counseling? |
| The contact details for a specific genetic counselor (e.g., a more personal approach than the previous choice in this case, the counselor would be assigned beforehand to this particular patient)? |