Development of an Electronic Health Record–Based Clinical Decision Support Tool for Patients With Lynch Syndrome

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ABSTRACT

PURPOSE To develop an electronic health record (EHR)–based clinical decision support (CDS) tool to promote guideline-recommended cancer risk management among patients with Lynch syndrome (LS), an inherited cancer syndrome that confers an increased risk of colorectal and other cancer types.

MATERIALS AND METHODS We conducted a cross-sectional study to determine the baseline prevalence and predictors of guideline-recommended colonic surveillance and annual genetics program visits among patients with LS. Multivariable log-binomial regressions estimated prevalence ratios (PRs) of cancer risk management adherence by baseline sociodemographic and clinical characteristics. These analyses provided rationale for the development of an EHR-based CDS tool to support patients and clinicians with LS-related endoscopic surveillance and annual genetics program visits. The CDS leverages an EHR platform linking discrete genetic data to LS Genomic Indicators, in turn driving downstream clinician- and patient-facing CDS.

RESULTS Among 323 patients with LS, cross-sectional adherence to colonic surveillance and annual genetics program visits was 69.3% and 55.4%, respectively. Patients with recent electronic patient portal use were more likely to be adherent to colonic surveillance (PR, 1.67; 95% CI, 1.11 to 2.52). Patients more recently diagnosed with LS were more likely to be adherent to annual genetics program visits (PR, 0.58; 95% CI, 0.44 to 0.76 for 2-4 years; PR, 0.62; 95% CI, 0.51 to 0.75 for ≥4 compared with <2 years). Our EHR-based CDS tool is now active for 421 patients with LS throughout our health system.

CONCLUSION We have successfully developed an EHR-based CDS tool to promote guideline-recommended cancer risk management among patients with LS.

ACCOMPANYING CONTENT

Appendix

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INTRODUCTION

Lynch syndrome (LS) is an inherited condition that confers an increased risk of colorectal, gastric, endometrial, and multiple other cancers. National guidelines recommend that patients with LS undergo colonic surveillance with lower endoscopies every 1-3 years depending on their age, genotype, previous cancer history, and family history.[1-3] Surveillance guidelines for gastric, endometrial, and other cancers are more heterogeneous and evolve frequently because of less well-established data for these tumor types[4]; as such, it is critical that patients follow regularly with a cancer genetics–trained clinician to ensure tailored and current recommendations on the basis of individual needs, family history, and emerging data.

However, patients and clinicians have reported challenges with coordinating and monitoring LS-related surveillance because of limited familiarity with guideline recommendations, particularly surveillance intervals, and lack of follow-up with cancer genetics–trained clinicians over time.[5] As such, the onus of scheduling follow-up appointments and surveillance activities often falls on individual patients. Integrating LS-specific surveillance recommendations into the electronic health record (EHR) to trigger automated reminders to patients and clinicians may facilitate adherence to guideline-recommended cancer risk management activities.[5,6] Indeed, EHR-based clinical decision support (CDS) has been shown to facilitate management decisions and improve patient receipt of guideline-recommended care in other clinical contexts.[7,8] Furthermore, EHR-based CDS has

CONTEXT

Key Objective

To develop an electronic health record (EHR)–based clinical decision support (CDS) tool to promote guideline-recommended cancer risk management among patients with Lynch syndrome (LS).

Knowledge Generated

In a cross-sectional study of patients with LS, we found that nearly 70% of patients were up to date with colonic surveillance, whereas only 55% were up to date with their annual genetics program visits. These observations provided rationale for the development of an EHR-based CDS tool to support patients and clinicians with LS-related endoscopic surveillance and annual genetics program visit completion.

Relevance

EHR-based CDS may be a promising strategy to help patients with LS and their clinicians coordinate and monitor their cancer risk management care.

been identified as one of the key drivers of implementing and sustaining genomic medicine in routine clinical practice.[9]

In this manuscript, we describe the development of an EHR-based CDS tool to promote guideline-recommended cancer risk management in patients with LS. After determining the baseline prevalence and predictors of guideline-recommended colonic surveillance and annual genetics program visit completion among patients with LS at our institution, we developed a LS-specific, EHR-based CDS tool to support patients and clinicians alike.

MATERIALS AND METHODS

Baseline Adherence to and Predictors of Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome

We conducted a cross-sectional study of patients with LS who received care in the Gastrointestinal Cancer Risk Evaluation Program at Penn Medicine, approved by the University of Pennsylvania Institutional Review Board. Each patient had a confirmed diagnosis of LS on the basis of a pathogenic or likely pathogenic (P/LP) variant in *MLH1*, *MSH2*, *MSH6*, *PMS2*, or *EPCAM*, or was an obligate carrier of a familial P/LP variant in one of these genes. Patients were required to have at least one visit at Penn Medicine within the past 3 years as of the data cutoff date on November 15, 2021, to be included in the study cohort. The electronic medical record of each patient was reviewed for sociodemographic, personal history, family history, and surveillance information. Records for abstraction included medical encounters and endoscopic reports from within and outside our institution.

Baseline adherence to guideline-recommended cancer risk management was evaluated for colonic surveillance and annual genetics program visits and defined using a previously described approach to evaluating the currency of cancer screening.[10] Patients were considered up to date with their colonic surveillance on the basis of local institutional practice for managing patients with LS, which takes into account National Comprehensive Cancer Network guideline recommendations incorporating patients' age and personal history of colorectal cancer (CRC; Table 1); patients were considered up to date with their annual genetics program visits if they had a documented encounter with a cancer genetics specialist within the past year. Sociodemographic and clinical characteristics were evaluated as potential predictors of adherence to guideline-recommended cancer risk management; these variables included age, sex, race, ethnicity, insurance status, residence in a medically underserved area (MUA),[11] electronic patient portal use, time since LS diagnosis, LS gene, personal history of CRC, family history of CRC, and personal history of colonic resection.

We used standard descriptive statistics to evaluate baseline patient characteristics. Spline regressions assessed for nonlinearity in the association between cancer risk management adherence and continuous variables such as patient age and time since LS diagnosis. Univariable log-binomial regressions estimated the prevalence ratio (PR) of cancer risk management adherence for the predictors described above. Variables with $P < .1$ on univariable models were retained in final multivariable models to estimate the adjusted PR of cancer risk management adherence by patient and tumor characteristics. Tests of statistical significance were two-sided, and significance was defined as $P < .05$. All analyses were performed using STATA version 17 (StataCorp LLC, College Station, TX).

Multiple post hoc sensitivity analyses were conducted to assess the robustness of our findings. First, we lengthened the recommended surveillance intervals for colonic

TABLE 1. Adherence Definitions for Cancer Risk Management Activities Among Patients With Lynch Syndrome

Cancer Risk Management Activity	Patient Characteristics	Up to Date Definition
Colonic surveillance	Previous history of colorectal cancer	Within the past 1 year
	25-40 years	Within the past 2 years
	>40 years	Within the past 1 year
Annual genetics program visits	All patients	Within the past 1 year
Upper endoscopy surveillance[a]	≥30 years	Within the past 2 years

NOTE. Definitions for colonic surveillance were based on both local institutional practice and National Comprehensive Cancer Network guideline recommendations for managing patients with Lynch syndrome.
[a]Not included in cross-sectional analysis of baseline adherence to guideline-recommended cancer risk management.

surveillance and genetics program visits by 2 months to evaluate the impact of calendar year–based insurance stipulations for procedures and specialist visits. Second, we evaluated a 24-month interval for colonic surveillance and genetics program visits to account for delays in care due to the COVID-19 pandemic. Finally, we conducted an analysis of colonic endoscopic surveillance adherence stratified by genetics adherence status to determine whether the groups were intrinsically different with respect to their surveillance behaviors.

The results of this cross-sectional study provided rationale for the development of an EHR-based CDS tool for patients with LS and will serve as a baseline to which future evaluations of the CDS tool will be compared.

Development of an EHR-Based CDS Tool to Promote Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome

We developed an EHR-based CDS tool for patients with LS according to established technical desiderata for CDS,[12-15] with a particular emphasis on the five rights of CDS previously described by Osheroff et al (Table 2). We leveraged existing infrastructure established by the PennChart Genomics Initiative at the University of Pennsylvania, a multidisciplinary collaborative that has successfully linked orders and results from genetic testing laboratories with discrete genetic data in our EHR (PennChart; Epic Systems Corporation, Verona, WI).[16,17] All discrete variants imported into PennChart are processed by Epic's Genomic Translational Engine to link P/LP variants to genomic indicators, which are then leveraged to facilitate downstream CDS. Variant data, genomic indicators, and CDS assignments can also be manually entered into discrete fields for patients with genetic data that were previously entered into the EHR in unstructured format.

A LS genomic indicator was automatically applied to any patient in Penn Medicine who had discrete genetic variant results corresponding to a P/LP variant in *MLH1*, *MSH2*, *MSH6*, *PMS2*, or *EPCAM* and for whom mosaicism was not detected. LS-specific CDS was built for colonic surveillance using the intervals described in Table 1 and annual genetics program visits. In light of evolving data supporting upper endoscopic surveillance for patients with LS,[18-20] we also elected to build CDS for upper endoscopic surveillance using institution-specific recommendations for upper endoscopies every 2 years for patients age 30 years and older. We did not, however, develop CDS for endometrial

TABLE 2. Application of the Five Rights of CDS to Lynch Syndrome

Five Rights of CDS	Applications to Lynch Syndrome
Right information	Provision of cancer risk management recommendations on the basis of local institutional practice, which accounts for National Comprehensive Cancer Network guideline recommendations for managing patients with Lynch syndrome
Right person	Provision of information to clinicians who can advise their patients to complete their cancer risk management activities Provision of information to patients with Lynch syndrome who can then schedule their cancer risk management activities at their convenience
Right intervention format	Alerts that unobtrusively notify clinicians when their patients are due for cancer risk management activities Patient-monitoring system that allows clinicians to aggregate all the patients in their panel who are due for cancer risk management activities Alerts that remind patients to schedule their cancer risk management activities on the basis of their information preferences (eg, whether they have opted in to email communications)
Right channel	Delivery of clinician-facing CDS through the EHR, which already contains relevant patient information and includes order entry, messaging, and scheduling functionalities to facilitate the completion of cancer risk management activities Delivery of patient-facing CDS through the electronic patient portal, which is HIPAA-compliant and includes messaging and scheduling functionalities to facilitate the completion of cancer risk management activities
Right time in workflow	Delivery of clinician-facing CDS at the point of care when a clinician is already in an individual patient's chart Delivery of patient-facing CDS when patients become due for a given cancer risk management activity

Abbreviations: CDS, clinical decision support; EHR, electronic health record; HIPAA, Health Insurance Portability and Accountability Act.

and other cancers since surveillance guidelines are less well established for these tumor types. Encounter and procedure details documented within PennChart were used to update CDS topics in real time. Clinician-facing CDS was composed of both patient- and population-level displays using Epic's Health Maintenance and SlicerDicer functionalities, respectively, whereas patient-facing CDS was accessible in the Preventive Care section of the electronic patient portal.

We evaluated the sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV) of our CDS logic for colonic surveillance using procedure results and clinician-recommended surveillance intervals reported in the EHR. Given that we were unable to incorporate unstructured procedure results into our CDS logic, this analysis enabled us to evaluate the robustness of our predefined logic in ascertaining which patients were up to date versus due for their colonic surveillance. Finally, we collected qualitative feedback from clinician end users regarding their overall impressions of the CDS tool, its usability, suggestions for improvement, and lessons that may inform similar efforts at other institutions.

RESULTS

Patient Population

Our cross-sectional study included 323 patients with LS with at least one visit at Penn Medicine within the past 3 years as of November 15, 2021. The median age was 50.4 years, and most patients were female (63.2%), White (87.3%), not a resident in an MUA (85.8%), and commercially insured (78.3%; Table 3, Appendix Table A1). Most patients were active on the electronic patient portal (89.8%) and had logged in at least once in the preceding 3 months (82.0%). The median number of years since a LS diagnosis was 4.2. Personal history was notable for previous CRC in 24.1% of patients, previous colonic resection in 24.5% of patients, and documentation of a first-degree relative with CRC in 47.1% of patients.

Baseline Adherence to and Predictors of Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome

As of November 15, 2021, 224 (69.3%) of patients were up to date with their colonic surveillance, 179 (55.4%) with their annual genetics program visits, and 136 (42.1%) with both cancer risk management activities. Rates of adherence to colonic surveillance increased to 79.6% and genetics program visits to 74.9% on post hoc analyses in which the surveillance interval was increased to 24 months.

On multivariable analyses, there were no appreciable differences in adherence to colonic surveillance or annual

TABLE 3. Baseline Characteristics of Patients in the Cross-Sectional Study of Adherence to and Predictors of Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome

Baseline Characteristic	All Patients (n = 323)
Age, years, median (IQR)	50.4 (39.1-61.6)
Sex, No. (%)	
Male	119 (36.8)
Female	204 (63.2)
Race, No. (%)	
White	282 (87.3)
Black	13 (4.0)
Asian	12 (3.7)
Other	7 (2.2)
Unknown	9 (2.8)
Ethnicity, No. (%)	
Not Hispanic/Latino	221 (68.4)
Hispanic/Latino	5 (1.5)
Unknown	97 (30.0)
Residence in a MUA, No. (%)	
No	277 (85.8)
Yes	46 (14.2)
Insurance, No. (%)	
Commercial	253 (78.3)
Medicare	56 (17.3)
Medicaid	8 (2.5)
Self-pay or unknown	6 (1.9)
Electronic patient portal enrollment, No. (%)	
No	33 (10.2)
Yes	290 (89.8)
Electronic patient portal login within the past 3 months, No. (%)	
No	58 (18.0)
Yes	265 (82.0)
Years since Lynch syndrome diagnosis, median (IQR)	4.2 (2.1-6.9)
Lynch syndrome mutation, No. (%)	
MLH1	72 (22.3)
MSH2	92 (28.5)
MSH6	82 (25.4)
PMS2	71 (22.0)
EPCAM	6 (1.9)
History of colorectal cancer, No. (%)	
No	245 (75.9)
Yes	78 (24.1)
History of metastatic colorectal cancer, No. (%)	
No	309 (95.7)
Yes	14 (4.3)
First-degree relative with colorectal cancer, No. (%)	
No	163 (50.5)
Yes	152 (47.1)
Unknown	8 (2.5)

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TABLE 3. Baseline Characteristics of Patients in the Cross-Sectional Study of Adherence to and Predictors of Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome (continued)

Baseline Characteristic	All Patients (n = 323)
History of colon resection, No. (%)	
None	244 (75.5)
Partial or hemicolectomy	55 (17.0)
Subtotal colectomy	23 (7.1)
Total proctocolectomy	1 (0.3)

Abbreviation: MUA, medically underserved area.

genetics program visits by patient age, sex, race, ethnicity, residence in an MUA, insurance status, personal or family history of CRC, or history of colonic resection. The sole independent predictor of adherence to colonic surveillance was electronic patient portal use within the preceding 3 months (PR, 1.67; 95% CI, 1.11 to 2.52). The relationship between annual genetics program visit adherence and time since LS diagnosis was nonlinear (Appendix Fig A1). After multivariable adjustment, time since LS diagnosis was the sole negative predictor of adherence to annual genetics program visits (PR, 0.58; 95% CI, 0.44 to 0.76 for 2-4 years; PR, 0.62; 95% CI, 0.51 to 0.75 for ≥4 compared with <2 years). These associations remained similar in direction and magnitude on post hoc sensitivity analyses in which the recommended surveillance interval was extended to 24 months (Appendix Tables A2 and A3).

In a post hoc sensitivity analysis of adherence to colonic surveillance stratified by genetics adherence status, 136 (76.0%) patients who were up to date with their annual genetics program visits were also up to date with their colonic surveillance, whereas 88 (61.1%) patients who were not up to date with their annual genetics program visits were up to date with their colonic surveillance. On multivariable analyses, there were no significant predictors of adherence to colonic surveillance among patients who were also up to date with their annual genetics program visits, but recent electronic patient portal use within the preceding 3 months was identified as the sole significant predictor of colonic surveillance adherence among patients who were not up to date with their annual genetics program visits (PR, 2.15; 95% CI, 1.11 to 4.17; Appendix Table A4).

Development of an EHR-Based CDS Tool to Promote Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome

To address the gaps that we identified in LS-related surveillance, we built an EHR-based CDS tool to promote guideline-recommended cancer risk management in patients with LS. Before implementation, we disseminated educational tip sheets and provided live demonstrations among clinician end users. Our CDS tool went live for patients with LS on November 15, 2021, and is active for 421 patients as of December 2022. Clinician-facing CDS is available at the point of care in each patient's chart and includes a listing of cancer risk management activities on the basis of the logic previously described (Fig 1). These entries are automatically updated whenever a procedure or encounter takes place within the Penn Medicine system. Clinicians also have the option to manually edit these entries as they see fit, as may be appropriate if a patient completes a procedure at an outside institution or requires closer follow-up than dictated by the CDS logic. Clinicians can also access a population-level display to identify all the patients in their panel who are due for a given cancer risk management activity at any point in time (Fig 2).

Patient-facing CDS consists of reminders for cancer risk management activities in the Preventive Care section of the electronic patient portal (Fig 3). Patients who have opted in to email communications also receive automated emails

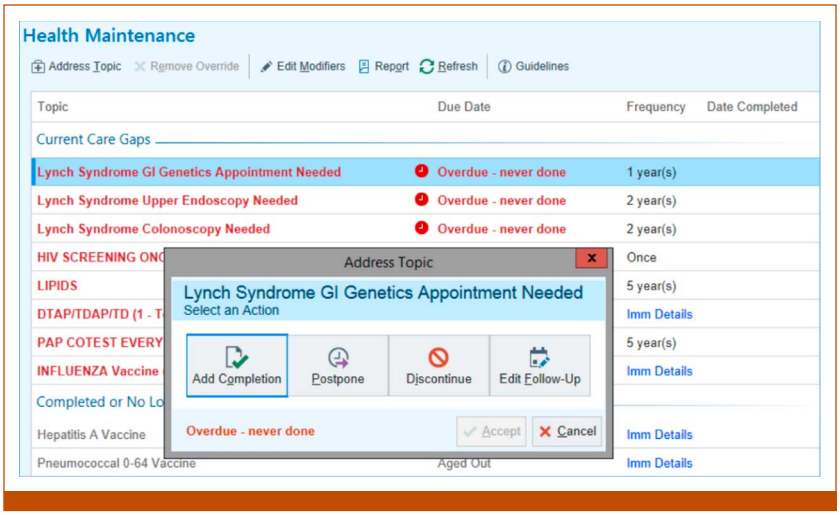

FIG 1. Clinician-facing, patient-level clinical decision support for Lynch syndrome. Printed with permission from Epic Systems Corporation, Verona, WI.

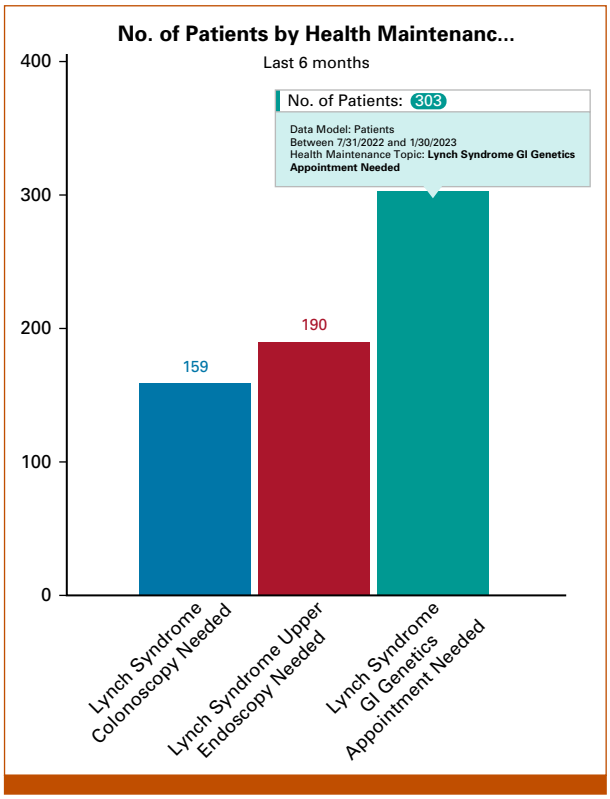

FIG 2. Clinician-facing, population-level clinical decision support for Lynch syndrome. Printed with permission from Epic Systems Corporation, Verona, WI.

anytime they are due for a given cancer risk management activity.

We used our cross-sectional cohort of 323 patients with LS to evaluate our colonic surveillance CDS logic against a gold standard of procedure results and clinician-recommended surveillance intervals reported in the EHR. In this analysis, our CDS tool had a sensitivity of 96.4%, specificity of 91.0%, PPV of 96.0%, and NPV of 91.9%. There were 17 (5.3%) discrepancies in up-to-date designations, 15 (88.2%) of which were due to clinician-recommended follow-up intervals that differed from the designations made in our CDS logic.

We received feedback on the CDS tool from two genetic counselors, one gastroenterologist, and one medical oncologist (Appendix Table A5). The CDS tool was described as easy to use and appeared to have a positive impact on both clinicians and patients. The primary suggestion for improvement was to incorporate active CDS reminders outside of the Health Maintenance tab. Lessons learned included building CDS logic that incorporated guideline recommendations that could be universally applied to the population of interest and that could be easily updated over time.

DISCUSSION

We found that nearly 70% of patients with LS were up to date with colonic surveillance but that only 55% were up to date with their annual genetics program visits, highlighting the need for tools to improve patient participation in this critical aspect of their cancer risk management care. Adherence to colonic surveillance was positively associated with recent electronic patient portal use, whereas adherence to annual genetics program visits was negatively associated with time since initial LS diagnosis. We have since developed what is, to our knowledge, the first EHR-based CDS tool to support patients and clinicians with LS-related endoscopic surveillance and annual genetics program visit completion.

Previous studies have demonstrated adherence with colonic surveillance ranging from 73% to 82% in LS, with greater adherence observed among younger patients, females, individuals with a personal or family history of CRC, and those who have undergone a genetic evaluation.[21-23] In this study limited to patients who receive care in the Gastrointestinal Cancer Risk Evaluation Program at Penn Medicine, we observed a similarly high rate of adherence with colonic surveillance and did not observe any differences by sociodemographic or clinical characteristics. However, cross-sectional adherence to annual genetics program visits was suboptimal and negatively associated with time since initial LS diagnosis, likely due to competing priorities as patients contend with their increased cancer risk. A recent study by Baert et al[24] also demonstrated that only 27% of patients with inherited CRC syndromes had systematic follow-up for surveillance coordination and monitoring. Regular genetics program visits play a critical role in tailoring nuanced and evolving surveillance recommendations for patients with LS, particularly for extracolonic tumors; as such, innovative strategies such as EHR-based CDS are needed to promote regular follow-up with cancer genetics-trained clinicians.

We also found electronic patient portal use to be significantly associated with adherence to colonic surveillance, particularly among those who were not concomitantly up to date with their annual genetics program visits. This observation likely reflects overall engagement with the health system rather than a causal relationship between electronic patient portal use and adherence. Indeed, studies have shown conflicting data regarding the impact of electronic patient portal use on patient activation and health care utilization, although patients have reported finding the portal helpful with improving access to their own health information and enhancing communication with their providers.[25-27] Additional research is needed to determine the impact of electronic patient portals, automated reminder messages, and other aspects of patient-facing CDS on patient engagement, care coordination, and downstream clinical outcomes.

Our development of an EHR-based CDS tool for patients with LS adds to a growing body of work aiming to leverage health information technology to implement genomic medicine in routine clinical practice.[28] CDS tools have been developed to identify patients in need of germline genetic risk assessments on the basis of their family history,[29] facilitate pharmacogenetic-guided medication prescribing,[30] and match patients to targeted therapies on the basis of their

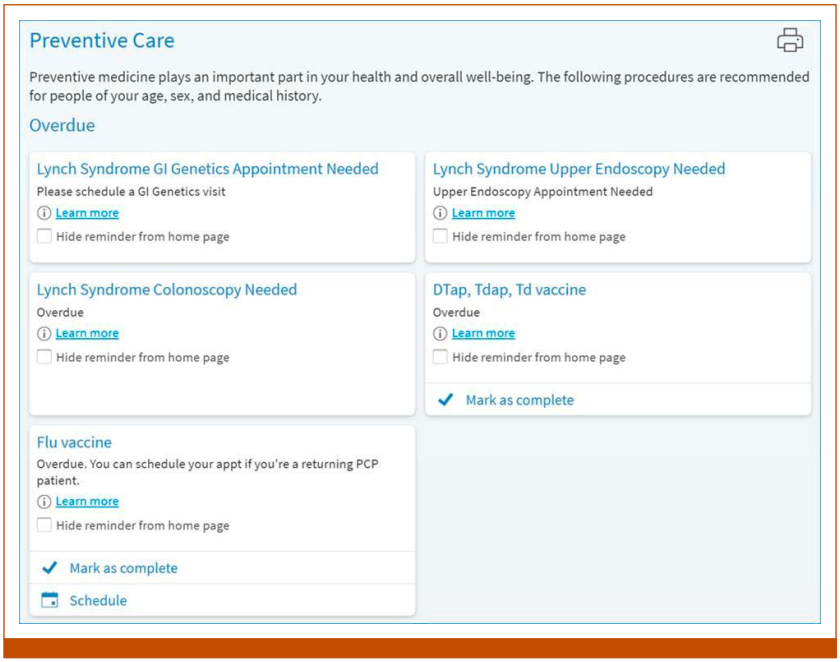

FIG 3. Patient-facing clinical decision support for Lynch syndrome. Printed with permission from Epic Systems Corporation, Verona, WI.

genomic testing results.[31] We applied many of the lessons learned from these previous efforts to our own CDS build, particularly the use of effective implementation strategies revolving around stakeholder engagement and education.[32] One additional goal of our work was to keep the CDS logic as simple as possible by integrating only discrete data elements that were already readily available in the EHR. Ultimately, our colonic surveillance CDS logic performed with >90% sensitivity and specificity when compared with a more complex approach incorporating unstructured procedure results and clinician-recommended surveillance intervals. This encouraging observation will be especially important as we move toward broader dissemination and scaling of this tool to other institutions and clinical settings.

This study has several limitations. First, we describe our experience with a predominantly non-Hispanic White population at a single academic institution. This lack of racial and ethnic diversity likely reflects the lower germline genetic testing and LS detection rates that have been observed among non-White patients worldwide.[33-35] Additional work is needed to improve the uptake of germline genetic testing among all eligible patients and determine whether our findings are generalizable to other populations and clinical settings. Second, we recognize the potential for misclassification of our study outcomes due to endoscopic procedures or annual genetics program visits that may have been conducted outside of the Penn Medicine system. To address this potential source of bias, we restricted our analyses to patients who had at least one visit in the Penn Medicine system within the past 3 years and manually reviewed all scanned and linked records from outside institutions that were available in our EHR. Third, the cross-sectional nature of this study limited our ability to evaluate longitudinal adherence to guideline-recommended cancer risk management activities, which is arguably more challenging for patients and clinicians to achieve over time. Finally, data are still lacking with respect to the impact of our EHR-based CDS tool on long-term clinical outcomes relative to the baseline adherence rates that we observed in our cross-sectional study.

In conclusion, we have successfully developed an EHR-based CDS tool to promote guideline-recommended cancer risk management among patients with LS. Ongoing efforts aim to refine this tool on the basis of stakeholder feedback, ensure that it remains up to date as clinical guidelines evolve, and adapt it for patients with other inherited cancer syndromes.

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DATA SHARING STATEMENT

The data that support the findings in this manuscript are available on request from the corresponding author, B.W.K. Additional information about our clinical decision support build is available on the PennChart Genomics Initiative website (https://www.med.upenn.edu/pgi/) and in the Epic Community Library (https://comlib.epic.com/).

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AUTHORS' DISCLOSURES OF POTENTIAL CONFLICTS OF INTEREST

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No other potential conflicts of interest were reported.

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APPENDIX

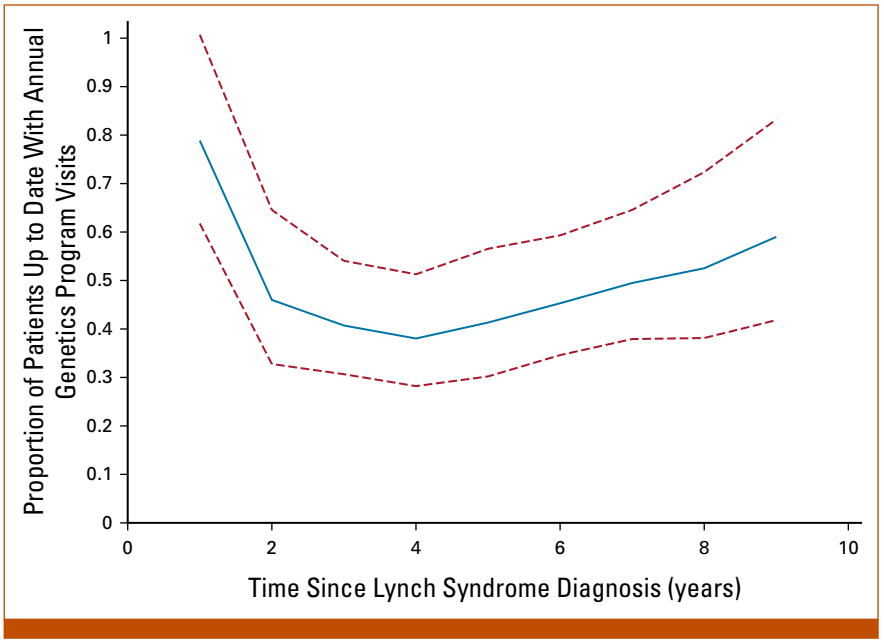

FIG A1. Spline regression of the prevalence of annual genetics program visits by time since initial Lynch syndrome diagnosis in years. Dashed lines = 95% CI.

TABLE A1. Baseline Characteristics of Patients in the Cross-Sectional Study of Adherence to and Predictors of Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome

Baseline Characteristic	All Patients (n = 323)	Colonic Surveillance		Annual Genetics Program Visits	
		Up to Date (n = 224)	Not Up to Date (n = 99)	Up to Date (n = 179)	Not Up to Date (n = 144)
Age, years, median (IQR)	50.4 (39.1-61.6)	47.8 (35.7-59.9)	56.0 (44.4-63.7)	51.6 (39.3-62.2)	48.4 (38.9-59.8)
Sex, No. (%)					
Male	119 (36.8)	76 (33.9)	43 (43.4)	58 (32.4)	61 (42.4)
Female	204 (63.2)	148 (66.1)	56 (56.6)	121 (67.6)	83 (57.6)
Race, No. (%)					
White	282 (87.3)	197 (87.9)	85 (85.9)	151 (84.4)	131 (91.0)
Black	13 (4.0)	8 (3.6)	5 (5.1)	10 (5.6)	3 (2.1)
Asian	12 (3.7)	8 (3.6)	4 (4.0)	7 (3.9)	5 (3.5)
Other	7 (2.2)	5 (2.2)	2 (2.0)	5 (2.8)	2 (1.4)
Unknown	9 (2.8)	6 (2.7)	3 (3.0)	6 (3.4)	3 (2.1)
Ethnicity, No. (%)					
Not Hispanic/Latino	221 (68.4)	153 (68.3)	68 (68.7)	140 (78.2)	81 (56.3)
Hispanic/Latino	5 (1.5)	2 (0.9)	3 (3.0)	2 (1.1)	3 (2.1)
Unknown	97 (30.0)	69 (30.8)	28 (28.3)	37 (20.7)	60 (41.7)
Residence in a MUA, No. (%)					
No	277 (85.8)	190 (84.8)	87 (87.9)	154 (86.0)	123 (85.4)
Yes	46 (14.2)	34 (15.2)	12 (12.1)	25 (14.0)	21 (14.6)
Insurance, No. (%)					
Commercial	253 (78.3)	175 (78.1)	78 (78.8)	136 (76.0)	117 (81.3)
Medicare	56 (17.3)	40 (17.9)	16 (16.2)	36 (20.1)	20 (13.9)
Medicaid	8 (2.5)	4 (1.8)	4 (4.0)	3 (1.7)	5 (3.5)
Self-pay or unknown	6 (1.9)	5 (2.2)	1 (1.0)	4 (2.2)	2 (1.4)
Electronic patient portal enrollment, No. (%)					
No	33 (10.2)	20 (8.9)	13 (13.1)	14 (7.8)	19 (13.2)
Yes	290 (89.8)	204 (91.1)	86 (86.9)	165 (92.2)	125 (86.8)
Electronic patient portal login within the past 3 months, No. (%)					
No	58 (18.0)	26 (11.6)	32 (32.3)	28 (15.6)	30 (20.8)
Yes	265 (82.0)	198 (88.4)	67 (67.7)	151 (84.4)	114 (79.2)
Years since Lynch syndrome diagnosis, median (IQR)	4.2 (2.1-6.9)	4.1 (2.1-6.7)	4.7 (2.1-7.3)	3.4 (0.9-7.1)	4.5 (2.7-6.4)
Lynch syndrome mutation, No. (%)					
MLH1	72 (22.3)	54 (24.1)	18 (18.2)	39 (21.8)	33 (22.9)
MSH2	92 (28.5)	69 (30.8)	23 (23.2)	55 (30.7)	37 (25.7)
MSH6	82 (25.4)	51 (22.8)	31 (31.3)	44 (24.6)	38 (26.4)
PMS2	71 (22.0)	45 (20.1)	26 (26.3)	37 (20.7)	34 (23.6)
EPCAM	6 (1.9)	5 (2.2)	1 (1.0)	4 (2.2)	2 (1.4)
History of colorectal cancer, No. (%)					
No	245 (75.9)	167 (74.6)	78 (78.8)	136 (76.0)	109 (75.7)
Yes	78 (24.1)	57 (25.4)	21 (21.2)	43 (24.0)	35 (24.3)
History of metastatic colorectal cancer, No. (%)					
No	309 (95.7)	216 (96.4)	93 (93.9)	176 (98.3)	133 (92.4)
Yes	14 (4.3)	8 (3.6)	6 (6.1)	3 (1.7)	11 (7.6)

(continued on following page)

TABLE A1. Baseline Characteristics of Patients in the Cross-Sectional Study of Adherence to and Predictors of Guideline-Recommended Cancer Risk Management Among Patients With Lynch Syndrome (continued)

Baseline Characteristic	All Patients (n = 323)	Colonic Surveillance		Annual Genetics Program Visits	
		Up to Date (n = 224)	Not Up to Date (n = 99)	Up to Date (n = 179)	Not Up to Date (n = 144)
First-degree relative with colorectal cancer, No. (%)					
No	163 (50.5)	117 (52.2)	46 (46.5)	93 (52.0)	70 (48.6)
Yes	152 (47.1)	101 (45.1)	51 (51.5)	82 (45.8)	70 (48.6)
Unknown	8 (2.5)	6 (2.7)	2 (2.0)	4 (2.2)	4 (2.8)
History of colon resection, No. (%)					
None	244 (75.5)	168 (75.0)	76 (77.6)	134 (74.9)	110 (76.4)
Partial or hemicolectomy	55 (17.0)	39 (17.4)	16 (16.3)	32 (17.9)	23 (16.0)
Subtotal colectomy	23 (7.1)	17 (7.6)	6 (6.1)	13 (7.3)	10 (6.9)
Total proctocolectomy	1 (0.3)	—[a]	—[a]	0 (0.0)	1 (0.7)

Abbreviation: MUA, medically underserved area.
[a]Colonic surveillance was not applicable for the one patient who had undergone total proctocolectomy. This patient was categorized as "Not Up to Date" for the remaining baseline characteristics.

TABLE A2. Multivariable Analyses of Determinants of Guideline-Recommended Colonic Surveillance (1) in the Primary Analysis, (2) Lengthening the Recommended Surveillance Interval by 2 Months, and (3) Using a 24-Month Surveillance Interval for All Patients

Determinant	Primary Analysis			Additional 2 Months			24-Month Interval		
	PR	95% CI	*P*	PR	95% CI	*P*	PR	95% CI	*P*
Age (ref: <50 years)									
≥50 years	0.78	0.60 to 1.02	.069	0.80	0.62 to 1.03	.086			
Electronic patient portal login within the past 3 months (ref: No)									
Yes	1.67	1.11 to 2.52	**.014**	1.59	1.08 to 2.34	**.019**	1.49	1.18 to 1.87	**.001**

NOTE. Adjusted prevalence ratios were estimated using multivariable log-binomial regression models. Bold text indicates $P < .05$.
Abbreviation: PR, prevalence ratio.

TABLE A3. Multivariable Analyses of Determinants of Guideline-Recommended Genetics Program Visits Using (1) the Recommended 12-Month Visit Interval (primary analysis), (2) Lengthening the Recommended Surveillance Interval by 2 Months, and (3) a 24-Month Interval

Determinant	Primary Analysis			Additional 2 Months			24-Month Interval		
	PR	95% CI	*P*	PR	95% CI	*P*	PR	95% CI	*P*
Time since Lynch syndrome diagnosis (ref: <2 years)									
2-4 years	0.58	0.44 to 0.76	**<.001**	0.57	0.44 to 0.74	**<.001**	0.69	0.49 to 0.99	**.042**
≥4 years	0.62	0.51 to 0.75	**<.001**	0.65	0.55 to 0.77	**<.001**	0.70	0.52 to 0.93	**.014**
Electronic patient portal login within the past 3 months (ref: No)									
Yes							1.45	1.00 to 2.12	.052

NOTE. Adjusted prevalence ratios were estimated using multivariable log-binomial regression models. Bold text indicates $P < .05$.
Abbreviation: PR, prevalence ratio.

TABLE A4. Multivariable Analyses of Determinants of Guideline-Recommended Colonic Surveillance (1) in the Primary Analysis and (2) Stratified by Annual Genetics Program Visit Adherence Status

	Primary Analysis (n = 323)			Up to Date With Annual Genetics Program Visit (n = 179)			Not Up to Date With Annual Genetics Program Visit (n = 144)		
Determinant	PR	95% CI	*P*	PR	95% CI	*P*	PR	95% CI	*P*
Age (ref: <50 years)									
≥50 years	0.78	0.60 to 1.02	.069	0.76	0.54 to 1.07	.111			
Electronic patient portal login within the past 3 months (ref: No)									
Yes	1.67	1.11 to 2.52	**.014**	1.35	0.79 to 2.28	.270	2.15	1.11 to 4.17	**.023**
First-degree relative with colorectal cancer (ref: No)									
Yes				0.86	0.60 to 1.22	.386			
Unknown				1.07	0.33 to 3.44	.907			
Lynch syndrome mutation (ref: *MLH1*)									
MSH2							1.23	0.71 to 2.12	.460
MSH6							0.67	0.36 to 1.24	.203
PMS2							0.70	0.37 to 1.32	.268
EPCAM							0.65	0.09 to 4.80	.671

NOTE. Adjusted prevalence ratios were estimated using multivariable log-binomial regression models. Bold text indicates $P < .05$.
Abbreviation: PR, prevalence ratio.

TABLE A5. Clinician End-User Feedback on CDS Tool for Patients With Lynch Syndrome

Topic	Representative Quotations
Overall impressions	"Prior to the CDS tool, patients would inquire whether our program had the ability to send reminder messages for annual follow-up visits and/or cancer screenings, but this wasn't feasible due to lack of patient-specific tracking & communication tools. CDS enables patients to be informed and empowers them to engage in their health maintenance." (genetic counselor) "The CDS tools in the EHR allow for easy data collection regarding patients with Lynch syndrome and whether or not they are overdue for recommended screenings and follow up visits." (genetic counselor)
CDS usability	"The CDS is very easy to use and update, and given that it appears in the Health Maintenance tab, it is clearly visible to all health care providers who interact with that tab." (gastroenterologist) "Overall, the CDS is easy to create, update and track. However, there is still some manual effort required on an individual patient level when further personalization is needed." (genetic counselor)
Suggestions for improvement	"It would be helpful if the CDS could directly flag providers about overdue Lynch syndrome screening without the provider needing to access the Health Maintenance tab, as some providers, such as oncologists, surgeons, or gastroenterologists, may not utilize the Health Maintenance tab as much as other providers such as primary care physicians." (gastroenterologist) "Automated reminders with more detailed scheduling instructions may prove to be more effective than passive reminders to patients with overdue screenings." (genetic counselor)
Lessons learned	"When developing CDS for all individuals with a hereditary cancer risk syndrome, it is only feasible to include types of screening or other cancer risk reducing initiatives that are universally recommended to all of these patients. Screening tests that are only recommended on an individualized basis without consistent criteria are difficult to incorporate into a universal CDS." (gastroenterologist) "When building a CDS tool, it is important to think about how the tool will be maintained and sustained in the long run. Building logic that is easy to update as errors are identified or guidelines are updated is ideal." (medical oncologist)

Abbreviations: CDS, clinical decision support; EHR, electronic health record.