original report

Compatibility of Family History Cancer Guidelines With Meaningful Use Standards

abstract

Purpose To evaluate the potential of implementing established family cancer guidelines as clinical decision support within meaningful use (MU)–compliant health information technology systems.

Methods We conducted a systematic analysis of cancer guidelines involving family health history (FHx) published before 2015. By comparing existing cancer guideline statements to current MU FHx standard requirements, we determined whether the cancer guideline statements could be implemented as clinical decision support. For guidelines that could not implemented, we determined the primary reasons for incompatibility.

Results A total of 531 statements from 55 guidelines published by 11 different organizations were reviewed and analyzed. Overall, 18% to 66% of guideline statements could or could not be implemented in MU-compliant health information technology systems, depending on which MU standard was used. Health Level Seven (HL7) models performed better than SNOMED models. Implementability of guideline statements varied by cancer type and guideline organizations. The greatest deficiencies in implementability of statements were largely a result of the fact that MU standards required only first-degree relatives and that FHx terms used in guidelines statements were ambiguous.

Conclusion FHx cancer guidelines and MU-based systems vary widely and are mostly incompatible. We identified sources of incompatibility and made recommendations that could improve the implementability of FHx cancer guidelines. Our findings and recommendations can enhance the use of established FHx cancer risk guidelines in routine clinical workflows.

Clin Cancer Inform.

Ayesha Aziz
Lance Pflieger
Nathaniel O'Connell
Joshua Schiffman
Brandon M. Welch

Author affiliations and support information (if applicable) appear at the end of this article.

A.A. and L.P. contributed equally to this work.

Corresponding author: Brandon M. Welch, PhD, Medical University of South Carolina, 135 Cannon St, 405L, Charleston, SC 29425; e-mail: welchbm@musc.edu.

INTRODUCTION

Family health history (FHx) is one of the strongest known cancer risk factors.[1-4] For example, patients with three or more first-degree relatives with breast or prostate cancers have a four-fold and 11-fold increased risk for those diseases, respectively.[5,6] With a detailed FHx, clinicians can identify these patients early and initiate personalized prevention strategies, such as increased screening, prophylactic surgery, risk-reducing therapeutics, and lifestyle changes during earlier, more treatable stages. Many notable organizations, including the US Surgeon General and the Centers for Disease Control and Prevention, promote the importance of collecting and using FHx.[7-9] Indeed, many clinical guidelines developed by professional organizations, cancer networks, and government entities include FHx.[10]

Several electronic FHx tools, such as the Surgeon General's My Family Health Portrait, are available to help patients collect their FHx as structured data.[9,11-13] Electronic health records (EHRs) now also support FHx as structured data as a result of Meaningful Use (MU) Stage 2 requirements.[14,15] As structured FHx data become more widely available within EHRs, the ability to leverage FHx data to provide computerized clinical decision support (CDS) becomes more feasible.

CDS, which encompasses a variety of tools to enhance decision-making in the clinical workflow, provides clinicians, staff, and patients with knowledge and person-specific information that is intelligently filtered and presented at appropriate times.[16] To run, CDS requires machine-readable clinical knowledge and structured patient data. First, clinical guidelines with clear and explicit language are used to create CDS algorithms. For example, a guideline that states "Patients over 50 years old" can be implemented much more easily than a vague statement such as "Older patients"

because there is less ambiguity. Second, structured patient data are the patient-specific variables used by the CDS algorithm to provide a result. The data can be manually entered into the CDS system or pulled from the patient's EHR. Unstructured data (ie, free text) or nonstandard terms make it more challenging to run CDS effectively because of the need for additional processing. The goal of MU is to encourage standards in EHRs that allow interoperability and reporting, the benefits of which include improved care for patients at lower costs.

Providing CDS to a health care provider within the workflow of an MU-compliant EHR is an ideal opportunity to identify patients at increased risk for cancer.[17-19] Patient-specific data for CDS will be available in a standardized format. However, it is not clear how compatible current FHx cancer guidelines will be with MU-compliant EHRs. The goal of this research is to systematically review FHx cancer guidelines to determine their compatibility with FHx standards. This evaluation also identifies gaps in cancer guidelines and/or MU standards that inhibit the use of CDS for FHx.[20] Improving the compatibility of FHx cancer guidelines with EHRs will ultimately improve cancer prevention and personalized care through FHx.

METHODS

To complete our systematic analysis of cancer FHx guidelines and MU standards, we identified eligible FHx cancer guidelines, extracted statements relevant to FHx of cancer, and analyzed each statement to assess whether current data models and standards could support FHx cancer guidelines.

Identifying Cancer Guidelines and Statements

Through expert recommendations, we identified 11 United States–based professional, advocacy, and government organizations that publish cancer guidelines (Appendix Table A1). We limited the scope to eight cancer types with known familial risks: breast, colorectal, stomach (gastric), prostate, skin, uterine, pancreatic, and ovarian (Appendix Table A2). We conducted a keyword text search within available published guidelines from these organizations to identify statements related to FHx. For guidelines with multiple versions, we selected the most recent version (through 2015). Keyword terms used included "family," "familial," "relative," "mother," "father," "sibling," "brother," "sister," "maternal," "paternal," and "degree."

Within each guideline, we extracted one or more logical statements (ie, if-then statements), which are the basis of CDS (Data Supplement). For complex logical statements with inclusive disjunction (eg, X or Y $\Rightarrow$ Z), we separated the statement into multiple simpler statements (eg, X or Y $\Rightarrow$ Z, changing it to two statements: X $\Rightarrow$ Z and Y $\Rightarrow$ Z).

Statement Analysis

We used MU criteria as the benchmark to assess cancer guidelines. To meet MU criteria, providers must record patient FHx as structured data from more than 20% of all unique patients for one or more first-degree relatives (parents, offspring, and siblings).[21] MU also requires EHRs to use either Systematized Nomenclature of Medicine-Clinical Terms (SNOMED CT [International Release, July 2012]) or Health Level Seven (HL7) Clinical Genomics Family Health History (Pedigree) Model for FHx information.[22,23] However, SNOMED and HL7 can support more than first-degree relatives. Because it is important to represent the data models as both the minimum required by MU and the full extent of its potential, we created a minimum class, which involves only first-degree relatives (to meet the MU standard) and a complete class (to meet the data model's full potential).

We used five classification categories to score the statements individually within each of the four MU scenarios (Table 1). Two trained informaticists (A.A. and L.P.) independently reviewed the statements and assigned a classification score for each MU data model scenario. The analysis was completed in multiple rounds, with 75 to 100 statements per round. After each round, the reviewers compared results and discussed discrepancies of their assessments with a third reviewer (B.M.W.) until a consensus on classification assignment was reached.

Summary of Results

Results of the analysis were summarized and compared across MU standards, classification assignments, cancer types, and guideline organizations. For quantitative comparison of classification outcomes, we used a generalized estimating equation to account for the correlation across each of the MU standards for organizations with 20 or more statements. For simplicity of the analysis, the outcome for classification was dichotomized to Y for statements that were compatible or not Y (explicit or derivable; consisting of C [conditional to fuzzy terms], N [not explicit or derivable], and CN [fuzzy and not derivable] grouped together) for statements that were not compatible. Because U (uninformative) classifications were few (five total) and unanimous across models, they were dropped from formal analysis. Results are presented in

Table 1. Assessment Classifications

Classification	Abbreviation	Description
Explicit or derivable	Y	The FHx data required by the statement could be completely implemented, either explicitly or derivatively, within the constraints of the scenario.
Conditional to fuzzy terms	C	The statement contained a vague or fuzzy term that prevented the statement from being implemented. Once the term is better defined, the clearer statement can be implemented.
Not explicit or derivable	N	The data model was not compatible with the statement.
Fuzzy and not derivable	CN	The statement contained a vague term, but even if the term was better defined, the statement would not be compatible with the standard.
Uninformative	U	Not enough information was available to assign an accurate score.

Abbreviation: FHx, family health history.

terms of their significance, odds ratios (ORs), and 95% CIs.

RESULTS

We analyzed a total of 531 statements from 55 guidelines published by 11 different organizations. In total, 209 statements from 14 guidelines published by eight organizations were specific to breast cancer, 151 statements from 12 guidelines published by eight organizations were specific to colorectal cancer, 67 statements from 11 guidelines published by five organizations were specific to ovarian cancer, and 103 statements from 28 guidelines published by eight organizations targeted other cancers. The most prolific guideline developing organizations (those with $>$ 20 statements) include the National Comprehensive Cancer Network (NCCN) with 199 statements, American College of Medical Genetics (ACMG) with 72, Society of Gynecologic Oncology (SGO) with 52, American Cancer Society (ACS) with 49, National Cancer Institute (NCI) with 43, American Congress of Obstetricians and Gynecologists (ACOG) with 42, and US Preventive Services Task Force (USPSTF) with 26 (Data Supplement).

Comparison of FHx MU Data Model Scenarios

We found a significant difference in the ability of MU standards to support cancer guidelines. Overall, the complete standards were better than using the minimum standards for both HL7 and SNOMED ($P < .01$). SNOMED minimum and HL7 minimum were compatible (Y) with 18% and 24% of cancer guidelines statements, respectively. SNOMED full and HL7 Pedigree full were compatible with 50% and 66% of statements, respectively. An inverse relationship exists between statements compatible with MU standards compared with statements that were not compatible (N) for each scenario, ranging from 4% in HL7 Pedigree full to 51% in SNOMED minimum. HL7 models were better than SNOMED models in both complete and minimum scenarios ($P < .01$; Fig 1).

One quarter of the statements were C in both SNOMED scenarios (24% SNOMED minimum, 25% SNOMED full), whereas 26% and 28% of statements were conditional to HL7 Pedigree minimum and complete, respectively. Five percent of statements were CN for SNOMED minimum and SNOMED full, 3% for HL7 Pedigree minimum and 1% for HL7 Pedigree full. Finally, only 1% of statements across all scenarios were found to be U.

For the analysis of scenarios aggregated over organization and cancer type, we assumed an unstructured covariance matrix to model correlation between the scenarios. We found that odds were greater for full versus minimum (OR, 4.66 to 7.34) and HL7 versus SNOMED (OR, 1.31 to 1.72), with the greatest odds for HL7 Pedigree full versus SNOMED minimum (OR, 10.46; Table 2).

Comparison of Guideline-Developing Organizations

We found significant differences in the compatibility of guidelines with MU standards across organizations. Guidelines published by ACMG provided the highest percentage statement coverage (Y) across all four data model scenarios, ranging from 31% to 94%, and least amount of fuzzy terms (C), between 3% and 4%. Conversely, ACS had the lowest percentage of statement compatibility (Y), ranging from 24% to 42%, and the highest rate of uncertain statements (C), 47% to 53%. The trend of HL7 and complete being more compatible with guidelines than SNOMED and minimum tended to continue across organizations (Fig 2).

Comparison of Statements by Cancer Type

We found that the cancer focus of the guidelines also affected the compatibility with MU standards. Guidelines for breast cancer with SNOMED minimum had the least with only 11% compatibility compared with 23% to 24% for colorectal and other cancers. HL7 Pedigree full was compatible with 67% of breast cancer statements, 63% of colorectal cancer statements, 87% of ovarian

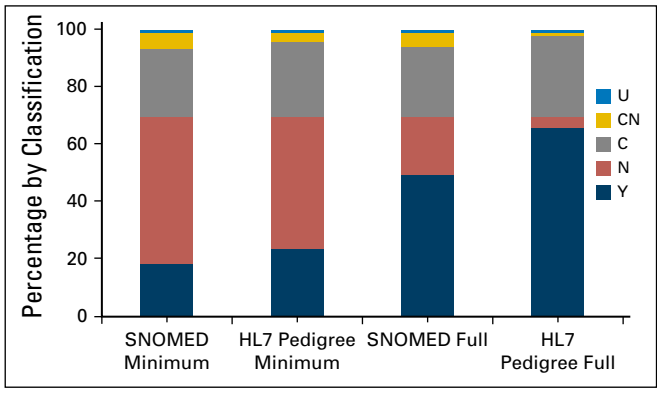

Fig 1. Comparison of compatibility by meaningful use standard. HL7, Health Level Seven; SNOMED, Systematized Nomenclature of Medicine. SNOMED Minimum, the minimum data required by meaningful use to be collected with SNOMED; HL7 Pedigree Minimum, the minimum data required by meaningful use to be collected with HL7; SNOMED Full, the full data set capable of being collected by SNOMED; HL7 Pedigree Full, the full data set capable of being collected by HL7. C, conditional to fuzzy terms; CN, fuzzy and not derivable; N, not explicit or derivable; U, uninformative; Y, explicit or derivable.

cancer statements, and 57% of other cancer statements (Fig 3).

Sources of Inadequacy

We identified several common factors that contributed to inadequacy in guidelines (C and CN) or inadequacy in MU standards used (N or CN). Inadequately defined or fuzzy terms (C and CN) used in guidelines accounted for 24% to 28% of statement incompatibility. The most common cause of guideline inadequacy was related to the vague definition of FHx used in many guidelines, accounting for three (74%) of four guideline inadequacies. For instance, the common phrase "strong family history" could not be implemented computationally unless a clear definition of the term was available somewhere in the guideline.[24] Disease risk, such as "increased lifetime risk" or "high suspicion of hereditary cancer," with no value or clear definition contributed to 13% of statement incompatibility. Genetic terms like "known genetic mutation" without any clear definition or criteria for interpretation (ie, pathogenic variant *v* variant of uncertain significance) contributed to 9% of statement incompatibility. Other examples include cancer-specific terms or modifiers such as "other cancers" or "sporadic cancer" as well as phrases involving age such as "younger age." Approximately 6% of the inadequate statements contained multiple fuzzy terms.

We also identified causes of incompatibility within the MU standards (Table 3). The most common cause among SNOMED minimum and HL7 minimum, affecting 246 statements, was that they failed to represent relations beyond first-degree relatives. No inadequacies related to degree of relation were found for HL7 Pedigree full. However, SNOMED full had 16 incompatible statements because the standards were unable to represent extended and complex relationships. The next most common problem was related to negation, such as statements like "without known mutation" and "genetic testing has not been performed," affecting 22 statements. Other data model inadequacies include the use of genetic terms, disease risk, laboratory results, and terms with missing SNOMED codes.

Table 2. ORs for Guideline Statement Compatibility by Data Model

Comparison Groups	OR for Compatibility	95% CI
SNOMED full *v* minimum	4.73	3.79 to 5.92
HL7 Pedigree full *v* minimum	7.39	5.87 to 9.29
HL7 minimum *v* SNOMED minimum	1.42	1.24 to 1.61
HL7 Pedigree full *v* SNOMED full	2.21	1.90 to 2.57
HL7 Pedigree full *v* SNOMED minimum	10.46	8.10 to 13.51

NOTE. Because of sample size constraints, only organizations with more than 20 statements were included in statistical analysis. (American College of Physicians, American Society of Clinical Oncology, American Society of Colon and Rectal Surgeons, and American Urological Association were not included.)

Abbreviations: HL7, Health Level Seven; OR, odds ratio; SNOMED, Systematized Nomenclature of Medicine.

DISCUSSION

To understand the level of compatibility between FHx cancer guidelines and MU standards, we extracted and analyzed 531 FHx cancer guideline statements and found that when guidelines are well defined and standards are complete (as in the case of ACMG guidelines using HL7 Pedigree), compatibility can reach 94%. However, limitations were found in currently available MU standards as well as the FHx cancer guidelines, accounting for 5% to 56% and 29% to 30% incompatibility, respectively. Unfortunately, as it is currently written, MU requires a minimum of first-degree FHx using SNOMED or HL7. If EHRs adhered to this minimum MU standard implementation of first-degree relatives only, they would be compatible with only 18% (for SNOMED) to 24% (for HL7) FHx cancer guidelines. Furthermore, only one in 10 breast cancer FHx statements we reviewed were compatible with the SNOMED minimum MU standard: that 20% of the patient population have their FHx recorded. Given this, in combination with the minimum first-degree relatives requirement, MU-compliant EHRs will miss the vast majority of patients who could be helped by cancer guidelines and genetic testing. So, although the HL7 Pedigree full is the most complete data model scenario, only two thirds of statements were compatible because of insufficiencies in the FHx cancer guidelines themselves. Through our analysis, we identified several areas of improvement and have created several recommendations that could

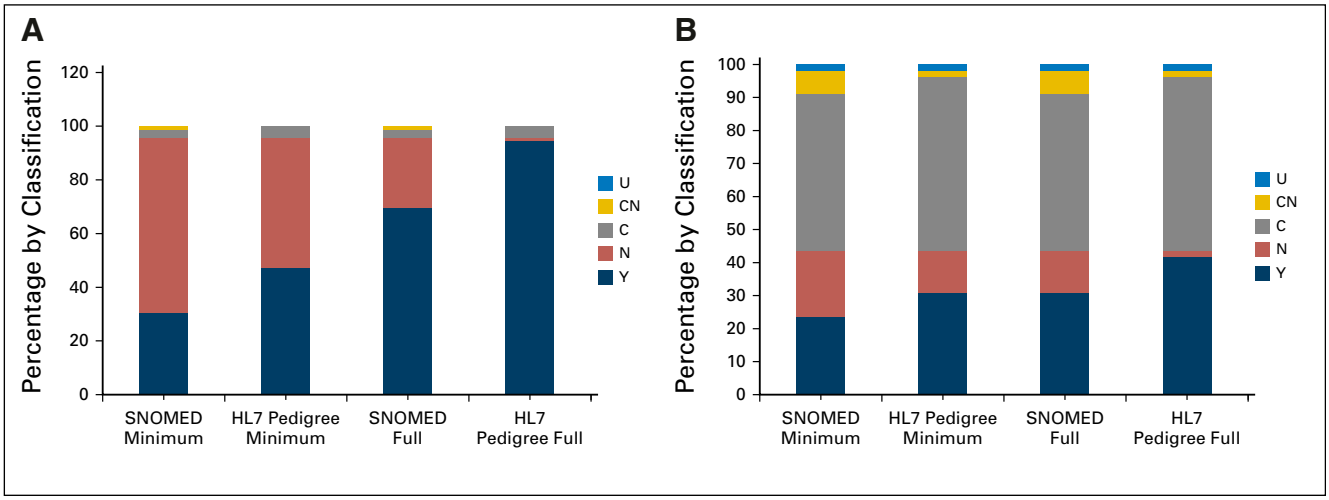

Fig 2. Comparison by guideline compatibility between American College of Medical Genetics (n = 72) and the American Cancer Society (n = 49). SNOMED Minimum, the minimum data required by meaningful use to be collected with SNOMED; HL7 Pedigree Minimum, the minimum data required by meaningful use to be collected with HL7; SNOMED Full, the full data set capable of being collected by SNOMED; HL7 Pedigree Full, the full data set capable of being collected by HL7. HL7, Health Level Seven; SNOMED, Systematized Nomenclature of Medicine. C, conditional to fuzzy terms; CN, fuzzy and not derivable; N, not explicit or derivable; U, uninformative; Y, explicit or derivable.

improve the compatibility of cancer guidelines with MU-compliant systems.

MU Should Require EHRs to Collect a Complete FHx Pedigree

To be compliant with MU, EHRs need to collect only a minimum of first-degree relatives. However, nearly half (48%) of the cancer guidelines analyzed required relations beyond first-degree relatives (ie, only parents, siblings, and children). This limitation resulted in the largest source of incompatibility caused by the MU standard. Furthermore, the most clinically valuable familial information often resides in second-degree relatives (eg, aunts, uncles, and grandparents), suggesting that MU should require that EHRs support at least second-degree relatives. Doing so would increase compatibility with guidelines, particularly with regard to breast cancer. Addressing this would have the greatest impact on improving the compatibility of FHx cancer guidelines with MU standards. In fact, if FHx data were available in the EHR, several EHRs could support second-degree relatives, and thus could run many of the cancer guidelines at a rate higher than the minimum.

Guideline-Developing Organizations Need to Reduce Ambiguous Terms in Guidelines

Organizations that develop guidelines need to clearly define terminology. In particular, 74% of ambiguous terms were the result of an inadequate definition of family history. Without a clear definition of who and how many relatives constitute a family history, family history is challenging to properly implement computationally. Other vague terms used in guidelines were related to imprecise definition of age (eg, younger) and representation of risk (eg, increased risk of), hindering the compatibility with MU standards. Given the complexity of cancer risk, it is understandable if guideline-developing organizations keep recommendations vague to provide flexibility. However, doing so hinders the implementation of the guidelines within computerized CDS. Therefore, we recommend that guidelines provide a clear definition of family history and other vague terms to improve compatibility with MU standards.

EHRs Should Collect FHx According to the HL7 Pedigree Model

The HL7 Pedigree model was created by genetics and informatics experts with the intention of its being used for decision support. As a result, the HL7 Pedigree model performed better than SNOMED in all cases, largely because of its ability to represent family members discretely. MU allows the use of SNOMED alone; however, our analysis shows that using SNOMED is not ideal for compatibility with FHx cancer guidelines. It is worth noting that family history can be represented through terminology (eg, SNOMED) or through the information model (eg, HL7), although the information model requires terminology to specify the terms used within it. Because SNOMED can provide the terminology used within HL7, we suggest that EHRs use the HL7 Pedigree model to represent FHx (with or without SNOMED), as opposed to using SNOMED alone.

Our analysis reveals that there can be further recommendations to improve compatibility of cancer guidelines with MU standards. For example, organizations in charge of developing guidelines should ensure that specific clinical terms (eg, lobular breast cancer), negation, risk, and genetic terms are used by SNOMED or are compatible

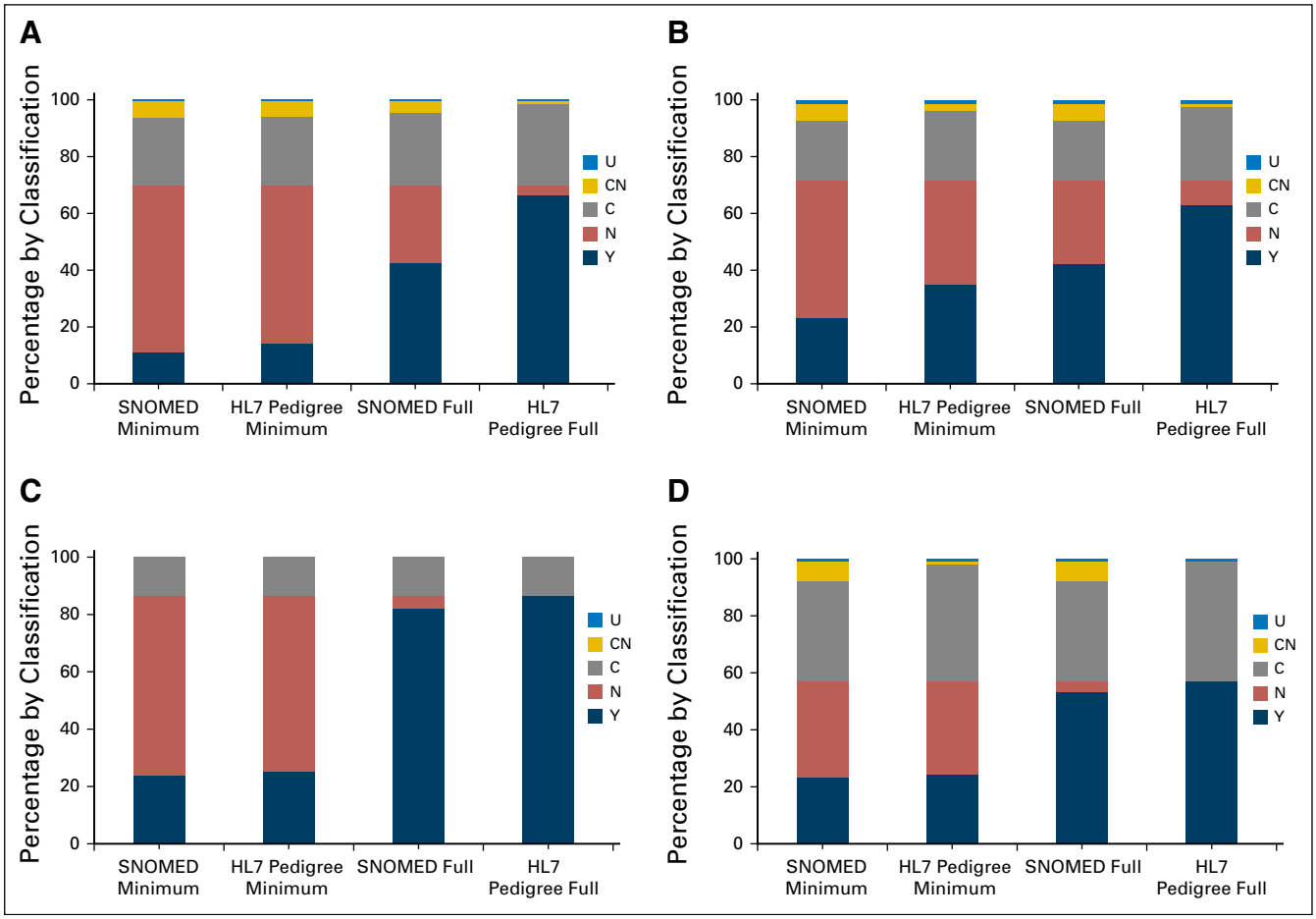

Fig 3. Comparison by cancer type for (A) breast cancer (n = 227), (B) colorectal cancer (n = 162), (C) ovarian cancer (n = 67), and other cancer (n = 186). SNOMED Minimum, the minimum data required by meaningful use to be collected with SNOMED; HL7 Pedigree Minimum, the minimum data required by meaningful use to be collected with HL7; SNOMED Full, the full data set capable of being collected by SNOMED; HL7 Pedigree Full, the full data set capable of being collected by HL7. HL7, Health Level Seven; SNOMED, Systematized Nomenclature of Medicine. C, conditional to fuzzy terms; CN, fuzzy and not derivable; N, not explicit or derivable; U, uninformative; Y, explicit or derivable.

with HL7. To do this, guideline-developing organizations and standards-development organizations should work together to ensure that guidelines and standards are compatible.

Limitations

We limited the search to eight of the most common hereditary and familial cancers. Although this is not a comprehensive list of all cancers, it represents the common cancers for which familial guidelines are most likely to exist. Second, clinical guidelines are updated periodically, and our analysis was based on versions of guidelines before June 2015. There have been updates in guidelines since our analysis, but even with these updates, we do not anticipate significant changes to our conclusions and recommendations. Third, the reviewers for this analysis are informatics experts with experience in CDS and data models, not trained clinicians. However, we felt it was necessary to use informaticists to evaluate computational implementability of these guidelines against data models. For any clinically related questions, we consulted a practicing clinical oncologist (J.S.) as needed.

Future Direction

Although this study was a theoretical exercise designed to demonstrate the impact of different MU data model scenarios on CDS implementations, the results assume complete and accurate documentation of FHx and do not reflect the reality that FHx information is often incomplete or wrong.[25,26] Therefore, we plan to simulate missing or incorrect FHx information to understand the impact on running FHx cancer guidelines. We also intend to implement many of these cancer guidelines as CDS for use within MU-compliant health information technology (IT) tools to promote cancer prevention. In the future, we intend to expand this evaluation beyond cancer to other illnesses. It is also worth noting that an emerging standard, HL7's Fast Healthcare Interoperability Resources (FHIR), is quickly gaining traction in the health IT community. Although it is not currently an MU standard for FHx, it could become one in the future. Because of this, future work will explore the compatibility of cancer guidelines with HL7's FHIR Family Member History specification.[27]

Table 3. Categories of Inadequacies for Each MU Standard

Category	SNOMED Minimum (n = 300)	HL7 Minimum (n = 261)	SNOMED Full (n = 132)	HL7 Pedigree Full (n = 24)
Degree relation	246	246	16	0
Negation	22	22	22	22
Genetic	30	0	30	0
Risk	13	0	13	1
Missing SNOMED terms	28	0	24	0
Laboratory result	19	2	19	2

NOTE. Some statements contain multiple inadequacies. SNOMED Minimum, the minimum data required by meaningful use to be collected with SNOMED; HL7 Pedigree Minimum, the minimum data required by meaningful use to be collected with HL7; SNOMED Full, the full data set capable of being collected by SNOMED; HL7 Pedigree Full, the full data set capable of being collected by HL7.
Abbreviations: HL7, Health Level Seven; MU, meaningful use; SNOMED, Systematized Nomenclature of Medicine.

In conclusion, our analysis, which highlights opportunities and challenges in implementing cancer FHx guidelines in the context of MU-compliant CDS, provides several recommendations for guidelines and MU standards to improve compatibility. By improving the ability of MU-compliant health IT systems to leverage established FHx cancer guidelines through CDS, a larger impact on cancer control and prevention is possible.

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AUTHOR CONTRIBUTIONS

Conception and design: Ayesha Aziz, Lance Pflieger, Joshua Schiffman, Brandon M. Welch
Collection and assembly of data: Ayesha Aziz, Lance Pflieger, Brandon M. Welch
Data analysis and interpretation: All authors
Manuscript writing: All authors
Final approval of manuscript: All authors
Accountable for all aspects of the work: All authors

AUTHORS' DISCLOSURES OF POTENTIAL CONFLICTS OF INTEREST

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Ayesha Aziz
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Lance Pflieger
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Nathaniel O'Connell
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Joshua Schiffman
Stock and Other Ownership Interests: ItRunsInMyFamily.com, PEEL Therapeutics
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Consulting or Advisory Role: Omicia, N-of-One

Brandon M. Welch
Leadership: Doxy.me, ItRunsInMyFamily.com
Stock and Other Ownership Interests: Doxy.me, ItRunsInMyFamily.com
Travel, Accommodations, Expenses: Doxy.me, ItRunsInMyFamily.com

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Affiliations

Ayesha Aziz, **Nathaniel O'Connell**, and **Brandon M. Welch**, Medical University of South Carolina, Charleston, SC; and **Lance Pflieger** and **Joshua Schiffman**, University of Utah, Salt Lake City, UT.

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APPENDIX

Table A1. Organizations Whose Guidelines Were Reviewed

Organization	Acronym	Guidelines	Statements
American College of Medical Genetics and Genomics	ACMG	1	72
American Congress of Obstetricians and Gynecologists	ACOG	6	42
American College of Physicians	ACP	3	16
American Cancer Society	ACS	15	55
American Society of Clinical Oncology	ASCO	2	11
American Society of Colon and Rectal Surgeons	ASCRS	1	12
American Urological Association	AUA	1	1
National Comprehensive Cancer Network	NCCN	10	199
National Cancer Institute	NCI	8	43
Society of Gynecologic Oncology	SGO	3	52
US Preventive Services Task Force	USPSTF	5	26

Table A2. Cancer Types With Number of Guidelines and Statements Reviewed

Cancer Type	Guidelines	Statements
Breast	14	234
Colorectal	12	164
Gastric	4	36
Prostate	8	24
Skin	8	27
Uterine	3	7
Pancreatic	4	24
Ovarian	10	72