ORIGINAL ARTICLE

Electronic Health Records and the Management of Women at High Risk of Hereditary Breast and Ovarian Cancer

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■ **Abstract:** Currently, management strategies exist that can decrease the morbidity and mortality associated with having a BRCA1 or BRCA2 mutation. Unfortunately, the task of identifying these patients at high risk is a daunting challenge. This problem is intensified because Electronic Health Records (EHRs) today lack the functionality needed to identify these women and to manage those women once they have been identified. Numerous niche software programs have been developed to fill this gap. Unfortunately, these extremely valuable niche programs are prevented from being interoperable with the EHRs, on the premise that each EHR vendor will build their own programs. Effectively, in our efforts to adopt EHRs, we have lost sight of the fact that they can only have a major impact on quality of care if they contain structured data and if they interact with robust Clinical Decision Support (CDS) tools. We are at a cross roads in the development of the health care Information Technology infrastructure. We can choose a path where each EHR vendor develops each CDS module independently. Alternatively, we can choose a path where experts in each field develop external niche software modules that are interoperable with any EHR vendor. We believe that the modular approach to development of niche software programs that are interoperable with current EHRs will markedly increase the speed at which useful and functional EHRs that improve quality of care become a reality. Thus, in order to realize the benefits of CDS, we suggest vendors develop means to become interoperable with external modular niche programs. ■

Key Words: clinical decision support, electronic health records, hereditary breast cancer, hereditary ovarian cancer, risk management

BACKGROUND

The BRCA1 and BRCA2 genes were identified in 1994 and 1995 respectively. Since that time, great strides have been made in understanding the impact that a mutation in one of these genes can have on a person's cancer risk, and many management strategies have been developed to mitigate these risks. However, unless individuals with mutations in these genes are identified and are managed using these strategies, there will be minimal impact on population health. Unfortunately, after 14 years of genetic testing, the vast majority of mutation carriers have yet to be identified. The challenge today is developing an approach that assures that high-risk individuals are identified and managed before they develop cancer, thereby maximizing the chance of preventing cancer, or finding it at an earlier and more treatable stage.

Family history information is likely the simplest and least expensive screening tool for identifying patients who need more intensive counseling and possibly genetic testing. However, the time it takes to collect a family history and the knowledge required to assess risk have been barriers to more effective use. Advancements in computer software have provided a foundation for better data collection and analysis techniques. Ultimately, the widespread adoption of these techniques into the clinical setting and their incorporation into electronic health records (EHRs) are crucial steps toward having a major impact.

It is of concern that currently available EHRs have not been able to incorporate the tools needed to leverage family history information into better patient care. For this reason, the Family Health History Multi-Stakeholder Workgroup, in its presentation to The

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DOI: 10.1111/j.1524-4741.2009.00796.x

Personalized Health Care Workgroup of the American Health Information Community (AHIC) suggested that health care providers involved with EHRs should:

"*…examine the merits of developing a modular family history tool, where collection of family health history is performed within the EHR, followed by messaging of this information to a variety of richer family history tools that perform risk analyses…The enhanced family history and results of these algorithmic calculations could then be returned to the EHR…*" (1).

The practical implication of this statement for risk assessment and population based screening is that rather than having each of the 150 EHR vendors develop a complete family history pedigree drawing and risk assessment tool de novo, we should expect that EHRs could serve as repositories of data capable of using external modules. These modules can use both established techniques as well as new and emerging tools to run analyses and draw pedigrees. Furthermore, as personal health records (PHRs) gain traction as extensions of EHR systems, and also adopt this strategy, patients could be empowered to develop and maintain their family history and to receive basic risk feedback.

This modular approach has tremendous merit, and, if adopted for family history and for other specialty applications, would set the stage for rapid advances in the functionality of EHRs. Today, most EHRs are little more than the sum of the parts that the particular vendor can develop and assemble. Niche software that solves problems specific to small groups of clinicians are not interoperable with EHRs and are generally discouraged (Fig. 1). This deficit in current EHRs is understandable given the lack of a large enough market for niche programs and the typical vendor's lack of expertise in specialty areas. A modular approach, where a niche program would be interoperable with any EHR, would allow the development and updating of these tools outside of the EHR, and make their clinical benefits available to any user of any EHR. Furthermore, as the knowledgebase that drives the development of these tools expands over time, the associated updates and improvements can be centrally maintained by experts in the field and immediately rolled out to clinicians, avoiding the roadblock of waiting for specific vendor acceptance and support. The ultimate goal should be to leverage the value of any niche software application with recognized clinical significance to any clinical situation.

The purpose of this study is to demonstrate the value of this approach as it applies to Hereditary Breast Ovarian Cancer syndrome (HBOC). We will review the current state of the art of family history collection and analysis techniques for this syndrome and examine the elements of Clinical Decision Support (CDS) that have been developed for the identification and management of high-risk patients. Finally, we suggest an approach for the integration of these technologies within the electronic health record to support large scale HBOC risk assessment. While this paper discusses HBOC in detail as an example, the methods discussed are easily extended to other adult hereditary syndromes, both benign and malignant.

CLINICAL DECISION SUPPORT

The inclusion of clinical guidelines, risk algorithms, and other knowledge into tools that help clinicians give better care is known as CDS. These tools are not static; they must evolve over time as new knowledge is gained.

Clinical Decision Support systems have an integral role for both the clinician and the patient in the management of disease risk, and are the key to improving the quality of care that has been promised with EHR and PHR systems. CDS systems can take the information available regarding the patient, and use algorithms, guidelines, and other knowledge to determine the best management strategy. The CDS system should then present that strategy along with supporting data in such a way as to allow the clinician to quickly understand and evaluate the suggested course of action. Furthermore, CDS should help the clinician move to the next step in their workflow by generating orders, prescriptions or managing patient information.

	Patient Data Entry	Risk Algorithms	Pedigree Drawing	Demographic and Clinical Data
Surgeon General's My Family Health Portrait	X		X	
Jameslink	X		X	
My Generations	X		X	
Hughes RiskApps	X	X	X	X
CancerGene		X	X	
Progeny			X	X
Cyrillic			X	

Figure 1. Selected niche applications commonly used by clinicians for the identification and management of patients at high risk for BRCA1/2 mutations.

This presentation and integration into a more efficient workflow is critical to helping the clinician undertake the correct approach. For the patient, CDS should present analysis results and other information in such a way as to enable the patient to understand the benefits of the prescribed care plan and to make an informed decision. Convincing the physician to recommend the correct standard of care will have little value if the patient does not appreciate the risks and benefits sufficiently to make an informed decision (9). Ultimately, such systems that analyze and communicate resulting data empower clinicians and patients to improve the quality of medical care.

Current EHRs and PHRs contain few CDS capabilities. Perhaps, the most advanced CDS involves the rudimentary warnings for drug interactions and allergy notification. While the pharmaceutical knowledge base is fairly complete, the display of information using the standard approach of the "Alert" pop-up has been less than successful. Isaac et al. found that despite the warning, clinicians continued with the same order for 90.8% of potentially harmful drug interactions, and 77% of potential allergic reactions (10). Creative solutions are needed to solve this problem.

Another challenge is that most information in EHRs is unstructured free text, which is undecipherable to most computer software. On one hand, structured data can be labor intensive for a clinician to enter, and often fails to convey the subtleties of prose text that can be critical to the effectiveness of a clinical note. Yet as clinical knowledge becomes formalized into CDS systems or modules, patient data must become structured in order for computers to process it. So, we must find an effective balance in which computer technology adds value to the clinical workflow, and not just in terms of data entry, but in the visualization of the relevant analysis as well. For hereditary risk, this means that the way in which clinical information systems store information about family members, and all of the associated details, must become standardized, and support pushing the work of data entry to the patient. This has been the major motivation behind the development of the currently accepted format for the electronic representation of family history data and pedigree analysis results.

Ultimately, it is highly unlikely that over 150 EHR vendors today will each independently develop every possible CDS system and data entry system that our clinicians need and are waiting for. A modular approach that unlocks the creative potential of academics, entrepreneurs and small niche vendors seems much more likely to succeed in the short term future.

CURRENT APPROACHES TO CLINICAL DECISION SUPPORT FOR HEREDITARY BREAST OVARIAN CANCER

Clinical Decision Support for HBOC is an exciting example to consider because much is known about BRCA1/2 mutations in terms of screening, testing, and management. A variety of guidelines, visualizations, and algorithms exist that can aid clinicians in the identification of high-risk individuals and to provide suggestions for genetic testing, MRI screening, chemoprevention, and other management approaches (Fig. 2).

The importance of family history information has been broadly recognized, and there are a variety of clinical tools to support its collection and use. However, almost all of these tools exist outside of EHRs, which lack this functionality. Clinicians are traditionally expected to collect and analyze family history quickly and in real-time during the clinical visit, and then manage high-risk individuals or refer them to appropriate centers. While this is considered good medical care, there are also medicolegal implications. In 1999, Severin noted that as physicians more completely recorded family history, there followed a legal responsibility to act on this information, and pointed to legal contests centered around breast and colon cancers that have had an impact on the identification and treatment for hereditary disease (11). There are

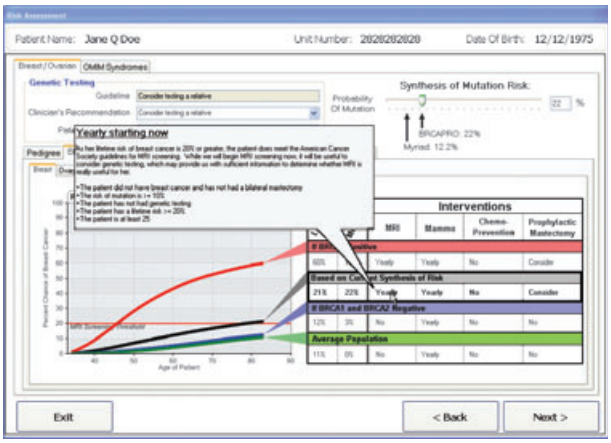


Figure 2. Elements of clinical decision support used by Hughes RiskApps to suggest MRI screening.

several barriers that have prevented this approach from being successful, which include, but are not limited to issues involving data collection, data entry, analysis, and interoperability.

Data Collection

The collection of family history information can be a time consuming process and is often associated with incomplete data. At the primary care level, family histories are only collected in approximately 50% of new patient visits (12). When family history is collected, it is often little more than a list of relatives affected with disease, often lacking the specific data, such as age of diagnosis, essential to risk analysis. The completeness of the family history is determined not just by the amount of information the patient has, but also by the biases of the clinician and the time constraints the clinician is laboring under. In a study by Burke et al., clinicians often missed paternal family history, often missed ovarian and other nonbreast cancers, and obtained age of diagnosis less often in second-degree relatives than in first-degree relatives (13).

In addition, in the ideal world, the family history should be updated at each patient visit to build on its completeness and correctness. This is not common practice today. The notion that old records would be retrieved and reviewed for updating this information is impractical with the current paper based or even the current EHR systems, which often do little more than formalize inefficient paper processes.

Data Entry

Most clinicians record family history as free text within a dictated or typed note, which is only slightly more useful than a paper record. This data is not machine readable for pedigree drawing or analysis, and is not easily updated. To obtain the benefits of CDS in improving quality of care, structured data are necessary.

Structured data are data that have a consistent format and storage location for each constituent element. An example of structured family history would be its collection into a table where specific columns are used for the name of relative, the disease that the relative has had, and the age of diagnosis of that disease in that relative.

Entering structured family history into a computer is time consuming. Structured data entry into niche programs by genetic specialists is common, as these programs help curate data, draw pedigrees, and run analyses. On the other hand, the entry of structured data into an EHR is almost nonexistent for two reasons: (a) few EHR systems allow structured family history collection and storage, and (b) few clinicians find it either practical or worthwhile to enter structured data into an EHR when the capability to do so exists. The additional work needed to enter structured family history data into an EHR is not considered a priority by most clinicians as there is little or no return on that investment (e.g., pedigree drawing or CDS). Thus, structured family history data tables are usually left empty and the family history appears as free text within clinic notes.

Allowing patient data entry for family history can help alleviate some of the time and cost of data collection and entry, thus freeing the practitioner to review and analyze the information rather than passively transcribe it. The literature on patient entered data would suggest that it is at least as accurate as that collected by interview for many of the more common diseases (14). An approach would be for patient entered data to be displayed to the clinician for editing and approval. Once it is accepted by the clinician, it would then be uploaded into the EHR. While Patient Portals to EHRs have been described as an option for patient self entry of family history, there are no examples in use today that we are aware of.

This problem has been identified and addressed by the development of several stand alone software applications that could easily become modules for EHRs. Some examples include systems that allow patient data entry via Tablet PC, such as HughesRiskApps, a program developed at the Massachusetts General and Newton-Wellesley Hospitals, systems that allow patient data entry via Kiosk, such as Jameslink from Ohio State University, and systems that allow data entry via website, such as MyGenerations from the North Shore University Health System, and ''My Family Health Portrait'' from the office of the Surgeon General. While several of these systems have adopted the HL7 standards (Described after), none have been adopted by existing EHRs for patient data entry. Early steps toward this goal are being taken by the Indian Health Service's EHR in their work with the Surgeon General's tool and Microsoft's HealthVault Project.

Data Analysis

Analyzing family history information can be a daunting task, and many clinicians lack the skills needed to do this well. This is not surprising when con-

sidering the broad array of syndromes that a clinician must know. Schuener et al., in reviewing syndromes with common adult chronic diseases, found 188 hereditary syndromes listed in the Online Mendelian Inheritance in Man (OMIM) database. There are 153 of these with various benign clinical manifestations, 32 involving cancer, and three showing both. It is unreasonable to think that a clinician will remember the details of 188 possible syndromes and be able to identify possible candidate patients in the midst of a busy clinic.

Traditionally, clinician education has been stressed as the way to overcome these barriers. For instance, in 1996, the American Society of Clinical Oncology adopted a policy statement in which they express their commitment to "…providing educational opportunities for physicians concerning methods of quantitative cancer risk assessment, genetic testing, and pre- and post-test genetic counseling…" (15). This approach has not been as successful as we hoped.

In 1999, Fry et al. published a survey of general practitioners in which over 60% of responding doctors agreed that taking a detailed family history from the patient was part of their role, yet 84% disagreed that this should include calculating the risk associated with a family history of cancer (16). In that same study they found 38% of doctors felt comfortable taking a detailed family history, and yet only 0.3% felt comfortable calculating the associated risks. By 2002, computers had started to take a more progressive role in some risk assessment programs, with Sweet et al. having used the Jameslink program (mentioned above) in a comprehensive cancer center to collect patient entered family history. They categorized 101 out 362 selected patients as high risk, but found that only 14 of these had documentation of physician risk assessment in the medical record, with only seven patients having been referred for genetic counseling (17). Burke et al. found that even when sufficient family history is collected, many primary care providers often miss the essential elements of HBOC as presented by a set of standardized patient cases. They suggest "… educational efforts will be most successful when they link collection of family history and referral to genetic counseling for consideration of BRCA testing to specific risk interventions." While this may be true, it adds an understanding of all of the associated risk mitigating interventions for HBOC onto the already overburdened educational goals.

Despite over a decade of clinician education, there has been only a modest impact in the identification of patients with HBOC, and their referral for genetic testing and risk mitigating strategies. Similarly, when considering the Hereditary NonPolyposis Colorectal Cancer, Domanska et al. found that at risk individuals had similar levels of knowledge concerning key aspects of the syndrome (18).

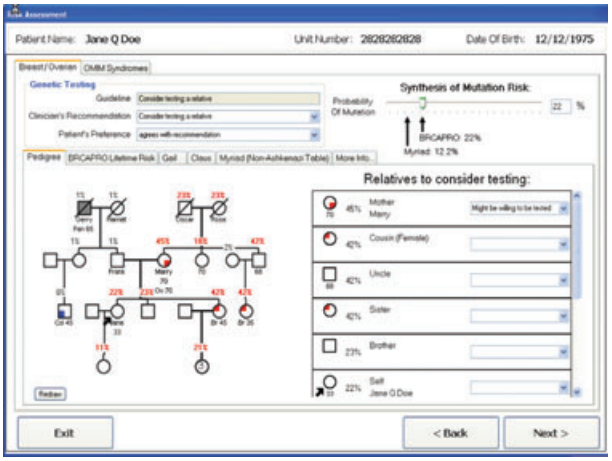


Figure 3. A pedigree used to show the pattern of disease in the family and the mutation probabilities for every family member in the HughesRiskApps program.

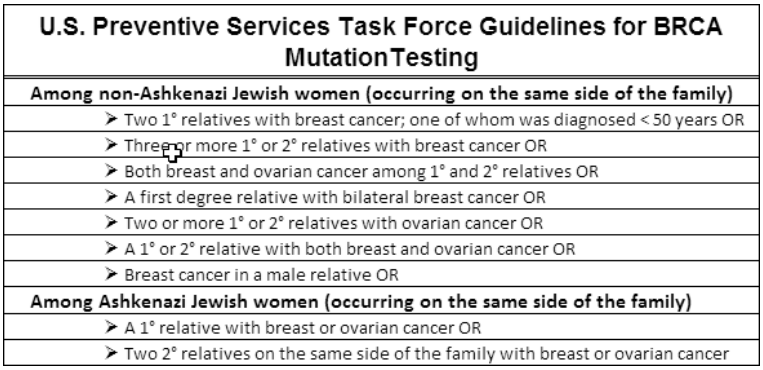
In addition to education, numerous tools are available to the clinician to simplify family history interpretation, including pedigrees, guidelines, and risk algorithms. However, these are still not easy to use in a typical office setting.

Pedigrees

Pedigrees are visual representations of the family structure that help clinicians identify patterns that may indicate hereditary disease. These visualizations can help to make obvious the pattern of Mendelian inheritance. The pedigree can also be a touchstone for the clinician or patient to support the building and curating of a family history. Unfortunately, Acheson found that only approximately 29% of primary care physicians feel prepared to collect family history and draw pedigrees.

Pedigrees can be hand drawn, or generated by computer software, such as CancerGene, Progeny, HughesRiskApps, My Family Health Portrait or others (Fig. 3). A surprising gap in computer science research is the lack of an open source pedigree drawing software package.

Despite the obvious value of being able to view family history as a pedigree, no current EHR can



U.S. Preventive Services Task Force Guidelines for BRCA Mutation Testing
Among non-Ashkenazi Jewish women (occurring on the same side of the family)
➢ Two 1° relatives with breast cancer; one of whom was diagnosed < 50 years OR
➢ Three or more 1° or 2° relatives with breast cancer OR
➢ Both breast and ovarian cancer among 1° and 2° relatives OR
➢ A first degree relative with bilateral breast cancer OR
➢ Two or more 1° or 2° relatives with ovarian cancer OR
➢ A 1° or 2° relative with both breast and ovarian cancer OR
➢ Breast cancer in a male relative OR
Among Ashkenazi Jewish women (occurring on the same side of the family)
➢ A 1° relative with breast or ovarian cancer OR
➢ Two 2° relatives on the same side of the family with breast or ovarian cancer

Figure 4. Example of a guideline for the identification of a patient who might benefit from genetic testing (19).

display family history in this manner, and no EHR is interoperable with the pedigree drawing software currently available. Often the only way to make a computer drawn pedigree available in an EHR is to undertake the data entry and drawing in a standalone program, and then to use a cumbersome cut and paste process to make it part of the note.

Guidelines

Guidelines are useful in determining the significance of cancers in the family. McClain et al. identified six different sets of guidelines published by a Society or Government to identify women who might benefit from counseling and possible genetic testing for HBOC. At least seven other guidelines also exist that did not meet their criteria (20).

The clinician must not only decide which set of guidelines to follow, but must also either commit those guidelines to memory or refer to a written document in the midst of a busy clinic. A quick look at a representative guideline (Fig. 4) should explain why this is not feasible, and thus why most patients who meet these criteria are not referred for counseling. The use of CDS software to flag those patients who meet the guidelines for referral and possible testing would facilitate increased identification. No current EHR can use any of the established HBOC guidelines to identify high-risk individuals.

Algorithms and Tables

Algorithms (Mathematical formulas such as BRCAPRO (21), Tyrer-Cuzick (22), or Boadicea (23)) or tables (such as Claus (24), Myriad (25), or FHAT (26)) can be used to determine the risk of having a BRCA1 or BRCA2 mutation and/or the future risk of developing breast or ovarian cancer. These results must then be translated into an action. For genetic testing, a reasonable, though arbitrary, threshold is that the risk of a BRCA 1 or BRCA2 mutation should be 10% or greater. Another example is the American Cancer Society suggestion that MRI breast screening is appropriate for a woman with a lifetime risk of breast cancer of 20% or greater by Claus, BRCAPRO, Tyrer-Cuzick or any model that predominantly uses hereditary factors in its calculations (27).

Although these algorithms might appear simple on the surface, the required computations, and interpretation of the results, necessitates using a computer with specialty software. The table approach is interpretable without a computer, but can be time consuming and often is not feasible in a busy clinic. Thus, software (Such as CancerGene, BRCAPRO, and HughesRiskApps) has been developed to run the algorithms and/or access the tables to help the clinician make better decisions. No EHR today can run accepted risk algorithms, or interpret accepted tables to help identify high-risk individuals.

Interoperability

The advantage of having a variety of niche tools for the various components of risk assessment and management is that it facilitates individual research institutions to develop and maintain work that represents the current state of the art in clinical knowledge. Correspondingly, the disadvantages include the requirement that clinicians be familiar with a disparate set of software applications, and must enter the same data over and over again. Redundent data entry can be avoided by mechanisms for information flowing freely between the producers and consumers of family history data and analysis results. This is the concept of interoperability.

Software that Combines Multiple Functions

Software for computing mathematical algorithms typically comes first from the research organizations

that developed the model. Examples can be seen in the cases of the BayesMendel software built for running BRCAPRO, and the IBIS Risk Evaluator built to run the Tyrer-Cuzick model. The interfaces for actually computing results are typically inappropriate for clinical workflows, therefore their use is limited.

Applications that bundle risk algorithms and tables, that provide interfaces for simplified data entry, and that display results in an easy to understand visualization are the beginnings of CDS in this area. One example is CancerGene, likely the best known method for calculating breast cancer risk probabilities. A single interface allows entry of data needed for multiple models and tables. The data is then used and reused to run multiple algorithms, with the result displayed on tabbed pages, along with a pedigree. A monumental achievement in its own right, CancerGene also implements CDS in the form of a syndrome suggestion, and a yes/no recommendation for considering chemoprevention based on the Gail model.

Another package is HughesRiskApps, developed at Massachusetts General Hospital and the Newton Wellesley Hospital. Similar to CancerGene, HughesRiskApps provides an interface for data entry by the clinician, but also allows data entry by the patient using a Tablet PC. In addition, HughesRiskApps can import data from other packages, such as My Family Health Portrait via an HL7 interface. Data is then analyzed by a suite of algorithms and tables, including BRCAPRO, Myriad, Claus, and Gail models, as well as a rudimentary algorithm (Under development) to identify any of the cancer syndromes identified by Scheuner et al. Furthermore, HughesRiskApps uses CDS to help identify the need for genetic testing, intensive screening, chemoprevention, and prophylactic surgery for both the breast and ovary. Its presentation strategy is meant to be a framework for synthesizing the relevant data and presenting the results in direct support of choosing management options.

Attempts have been made to integrate Progeny with risk assessment algorithms to perform quantitative risk assessment (28). The downside of this approach is that each Progeny user is left to develop their own set of data to collect, and therefore each attempt to integrate with other data stores or risk calculators must be developed anew, and will only work for that single site.

The largest challenge to the establishment of interoperability between tools used for risk assessment is the heterogeneity of the data. While most programs collect the same set of data points (relationship type, disease, age of onset, etc.), each uses a different name and data structure for each element, and uses different codes or names for the data entries. For example, one program might use relationship as the field and GM as the code, but another will use relative as the field name and Grandmother as the code. Some programs capture actual age and other capture age ranges.

These challenges are currently being met by developing a standard intermediary defined by HL7 (ref), an international organization dedicated to standardization of health related data. Within HL7, the Clinical Genomics Special Interest Group (CG SIG) has designed and developed a message for transmitting family history information, known as the CG SIG Pedigree Model. This has been approved by the American National Standards Institute (ANSI) and accepted by the Healthcare Information Technology Standards Panel (HITSP). This model has been implemented by early adopters (HughesRiskApps, My Family Health Portrait, CancerGene) and used to demonstrate the power of interoperability between family history data collection and analysis applications. Unfortunately, no EHR vendor is currently able to send or receive this HL7 message.

American Health Information Community has developed a core data set (29) that defines what data elements (Table) are needed in an EHR or PHR) to store family history data in a manner that will allow CDS and pedigree drawing. However, while this core data set is HITSP approved, no EHR vendor to date has adopted these standards as of yet. Most EHRs today either collect family history as free text, or have a rudimentary data structure that is inadequate for CDS or pedigree drawing. Adoption of the AHIC core data set and compatibility with the HL7 standard for family history transmission by every vendor is critical.

SUMMARY OF THE CURRENT SITUATION

Electronic Health Records today lack the functionality needed to identify women at high risk for HBOC or to manage those women once they have been identified. Numerous niche programs have been developed to fill this gap. Clinicians interested in these areas use this niche software to identify patients, refer them for consultation, run risk analyses and collect data to

continue assessing and improving results. Unfortunately, these extremely valuable niche programs are prevented from being interoperable with the EHRs, on the premise that each vendor will build their own programs. This effort would attempt to reinvent the wheel, stifling an area of innovation in quality medical care. Essentially, if we depend on the vendors, it will take years to get to functional and useful solutions. As no EHR can currently reproduce this functionality and as no EHR can accept the structured data and analyses these programs produce, clinicians are forced to dictate or type family history and analyses into free text notes. In this form, the data cannot be used for improving the quality of medical care for the individual, it cannot be searched to identify patients who may benefit from new tests or treatments, and it cannot be used for medical research, except by expensive and time consuming chart reviews. This is not the electronic age we were promised.

Unfortunately, in our zeal to adopt EHRs, we have lost sight of the fact that they can only have a major impact on quality of care if they contain structured data and if they interact with robust CDS tools. This lesson of usability applies not just too family history, but too many other aspects of patient care as well.

THE FUTURE

We are at a crossroads. As we push adoption of the current generation of EHRs, we stand to lose much of the progress made by individuals, academic centers, and companies that have developed functional niche software that fits into workflow and uses CDS to improve patient outcomes.

While software vendors have been unable or unwilling to be interoperable with external niche programs, they have also been unable to replace these existing niche programs within their own products. Unfortunately, after spending millions of dollars implementing EHRs, many hospitals feel compelled to only use these systems for clinical care, and to exclude niche programs from their plans. As such, clinicians who use patient data entry, pedigree drawings, risk algorithms, and other aids are being asked to give them up in favor of an EHR that can do none of these.

The more effective approach is to follow the AHIC recommendations, which positions EHRs as data repositories that exchange data with robust external modules. Under this paradigm, it would be these external modules that draw pedigrees, run algorithms, decipher guidelines, and undertake other functions that improve medical care. This design would also draw on the strength of each entity; EHR vendors are not experts in the myriad of clinical decision support systems that are required for quality medical care and small niche software cannot replace the massive data repository that is the current EHR. Here is room for tremendous synergy.

Such a design can be accomplished if every EHR vendor (a) adopts the AHIC core data set for family history, and (b) is able to send and receive messages using the HL7, ANSI approved message. EHR vendors should make their data (which is really patient data or institutional data) accessible for manipulation by external modular programs and allow the completed product of those external programs to be reincorporated into the EHR. An HL7 message is the ideal format for pushing data from an EHR into an external modular system and then taking the results in a second message back into the EHR.

This will jump start EHR functionality in HBOC, but also open the floodgates of innovation by the vast number of clinicians with expertise in highly specialized areas throughout medicine, not just in the hereditary arena. This approach will increase the likelihood of functional, efficient EHRs that improve quality of care within our lifetime.

CONCLUSION

The evolution of CDS mechanisms for the identification and management of patients at high risk for HBOC has mirrored the growth in our understanding of this syndrome. As our mission in caring for these patients expands from the high risk center into the broader primary care setting, this growth must continue to support the expanded demand placed on clinicians. Fifteen years after the identification of the BRCA1 gene we have come far, but with many clinicians still struggling to identify the essential elements of HBOC, we have a long way to go.

Electronic Health Records currently can be seen as collections of free text documents that describe independent events in the care of the patient. Synthesizing and organizing these fragments into a coherent whole takes significant cognitive work by the clinician. The future of the EHR is to contain structured data that can be synthesized and evaluated using Clinical Decision Support, and presented to the clinician as a

coherent whole that is easily and intuitively understood and acted upon. How we move from where we are to this higher plain is a matter of debate.

Using family history as an example, the EHR today contains multiple records of family history recorded by multiple clinicians in multiple notes. The data is unstructured, no visualizations are present, and, for all practical purposes, no risk calculations or guidelines are available. Several niche programs have been devised to fill the EHR deficits by drawing pedigree visualizations, recording structured data (entered by the patient or the clinician), running risk algorithms, and accessing guidelines. The success of these systems has demonstrated the benefits of CDS. We now stand at a crossroads, where we must decide to abandon all the work done to date in niche software, putting all our eggs in the EHR basket, or whether we leverage external niche modules by making them interoperable with current EHRs.

It seems highly unlikely that 150 EHR vendors will develop independently all the functionality of these various niche programs within the foreseeable future. A modular approach to development of niche software will speed this process enormously. Thus, to see the benefits of CDS now, we strongly suggest vendors develop means to become interoperable with external modular niche programs. We have presented here the rationale as it relates to hereditary syndromes. It takes little imagination to see how this applies to almost all aspects of clinical care.

Acknowledgments

This work was supported by Philanthropic Funds from the MGH and the NWH.

Disclosure

KSH is on the Speaker's Bureau for Myriad Genetics. He is the developer of HughesRiskApps, an open source software package for managing hereditary risk. BD and EMO declare no conflicts of interest.

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