# Assessing Risks for Families with Inherited Cancers

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**ABSTRACT**

A family history of disease can be a powerful clinical indicator of risk. The challenges associated with collecting a thorough family history, and maintaining that information over time, prevent the benefits of effective prophylactic treatments from reaching those with the greatest risk. We have introduced patient self-reported data entry using tablet computers, along with clinical decision support algorithms and visualizations for interpretation and analysis, into a clinical workflow capable of large scale screening and management of those at high risk for hereditary breast and ovarian cancer.

**KEYWORDS:** Risk Assessment, Breast Cancer, Clinical Decision Support.

**INDEX TERMS:** J.3 [Life and Medical Sciences]: Health—Medical Information Systems

## 1 INTRODUCTION

One of the great hopes of the genomic age is that by understanding and identifying genetic mutations, we can prevent disease. One of the barriers to genomic technology having a significant impact on public health has been our ability to identify those healthy individuals who are at risk of disease because they have inherited a mutation before they become sick. Until the day when everyone gets full gene sequencing as part of a screening program, we will likely rely on patient self-reported family history as signal for identifying those whom genetic testing would benefit. The difficulty this presents to our current approach is that providers are asked to collect the data in sufficient detail and then identify the wide range of syndromes, often without special training [1]. According to the Online Mendelian Inheritance in Man database of genetic disorders, there are 188 adult hereditary syndromes with at least one adult chronic disease. In this paper we discuss the example of Hereditary Breast and Ovarian Cancer (HBOC), and methods we developed to expand the identification and management of those at risk for its effects.

There are approximately 1,000,000 carriers of mutations in the genes that cause HBOC (BRCA1 and BRCA2) in the US, and of those about 50,000 (about 5%) have been identified to date [2]. We believe this poor performance is likely the highest rate of identification for any adult hereditary cancer syndrome because clinical genetic testing has been available now for thirteen years and we know a great deal about the association between the genes and treating the disease.

Most high risk women are not being identified or referred for counseling, and our risk clinics could not manage the volume if all high risk women were referred [3]. Health Information Technology (HIT) holds the key to increasing the quality of care while decreasing the cost of care [4,5,6,7]. This will be accomplished by increasing efficiency and increasing the use of Clinical Decision Support (CDS) to promulgate evidence based medical care. Thoughtful visualisations will be necessary to synthesize CDS with data from the patient and the clinician to make proper management obvious at the same time as directly supporting the clinical workflow.

We have developed a system that integrates these components into mammographic screening, genetic counseling, and surgical clinic settings.

## 2 LARGE SCALE METHODS

By developing an HIT infrastructure for identifying BRCA carriers, we believe the approach can scale up easily. We also expect the approach can work for many other disease areas. We also believe however it is ultimately necessary for these tools to be interoperable with other clinical systems. Unfortunately, current Electronic Health Record (EHR) systems remain digital copies of paper records, using little of the graphical or organizational power of a computer. As an example, to evaluate the risk of a hereditary condition, one must look in the demographics section, the family history section, the problem list and the lab results section to see all pieces of the puzzle. Pedigree visualizations (Figure 2) can put all this data into a single coherent picture, simplifying the clinician's work [8], but pedigrees and other visualizations remain beyond the capability of EHRs.

The need for CDS is driven by the rate in which providers are being deluged with new information. Knowledge grows exponentially, as seen in part by the dramatic rise in the number of articles in Medline and PubMed [9]. We do not believe it is reasonable to expect that providers will be able to keep up with all the information they need to manage patients. CDS provides the likely solution.

Most importantly, CDS should facilitate the best action as part of normal workflow. Today, CDS is rudimentary at best, both squandering the opportunity to increase quality and producing cynicism among providers as to its utility. EHR vendors uniformly point out alerts for drug-drug interactions and allergies as proof that they know how to accomplish CDS. In reality, these systems have failed as they do not present the information to the provider in a compelling way nor do they help the clinician follow the recommendation within the course of their normal workflow. Isaac et al identified that providers fail to act on 93.4% of drug-drug interaction alerts and 77% of allergy alerts [10].

### A prototype solution

The process starts when a patient checks in and is handed a Tablet PC which displays one question per screen in a choice of languages including English, Spanish and Italian (Figure 1). Information from prior sessions pre-fills the answers to most questions, while branching logic moves over questions irrelevant to the patient. She enters risk factors, family history, and an extended review of systems.

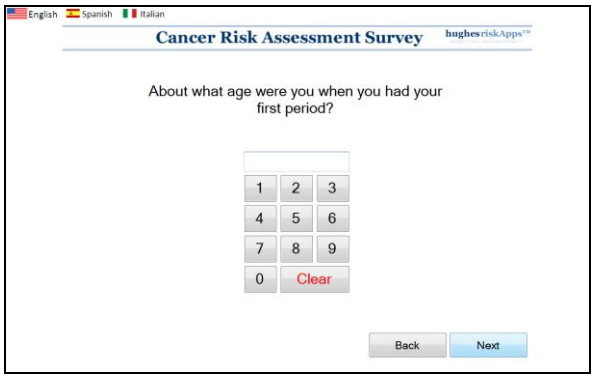


Figure 1. **A sample Hughes RiskApps patient survey question showing a simple interface with easy to read questions and options for multiple languages.**

Upon completion of the survey, risk models for breast cancer are immediately run and a summary printout is generated that displays the patient data in an intuitive form, including a pedigree. Patient information sheets, such as smoking cessation information, are generated for appropriate individuals. The staff reviews the summary printout to confirm accuracy, and makes appropriate corrections.

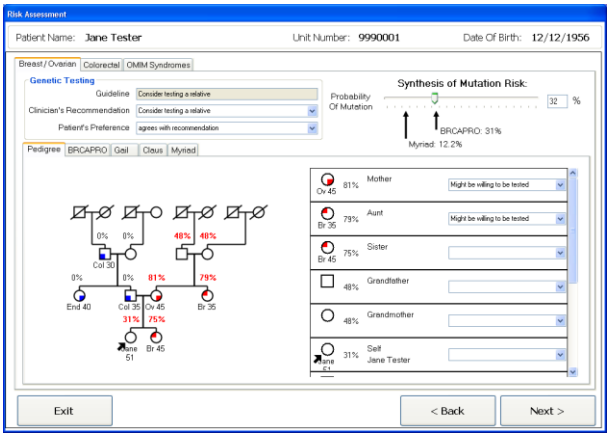


Figure 2. **The pedigree visualization can show risk for a whole family.**

The clinician workflow is based on an intuitive set of tabs that starts with a review of the data entered so far, progresses through the clinical encounter, and ends with all of the necessary documentation and order sheets being generated. In the risk clinic module the genetic counsellor can review the results of the risk models with the patient to help determine what the various options and likely outcomes are, and ultimately if testing is an appropriate course of action (Fig 3).

In addition to viewing the raw data, risk calculation algorithms are run and the results are displayed to the clinician using a visual representation. Graphs show results from BRCAPRO, a standard breast cancer risk assessment algorithm, run multiple times for the same family using different parameters. BRCAPRO is also run for each relevant family member, with the risk of mutation shown for each in a pedigree diagram.

Genetic testing recommendations are made within the same user interface in which the data is shown. In addition, risk of mutation is computed from another risk model, the Myriad model, and results from both BRCAPRO and Myriad are shown on a risk of mutation slider, which the clinician can set manually. Family members are listed in order of likelihood of mutation. The willingness of each to be tested can be recorded.

Lifetime risk of breast or ovarian cancer and several risk management suggestions are shown to the clinician for multiple scenarios: without testing (Current synthesis), as if the patient tested positive, as if the patient tested negative, and the population risk. Gail, Claus, MMPRO and PREMM risk model results are displayed as well. CDS suggests alternative syndromes in order of likelihood, and shows manifestations of the selected syndrome. Double clicking on a syndrome opens its page in the OMIM and Genetests Websites.

The CDS system helps the clinician find all mutation carriers by enabling the clinician to visually document the testing of family members. The tool then shows the number tested versus number of living relatives age 18 or older with a mutation risk of 10% or greater.

In the surgical module, the clinician adds details about the exam and completes collection of information using an interface tailored for this encounter (Fig 4). The immediate payback for this work is how the CDS helps the clinician develop an impression and plan. After discussion with the patient, the record is finalized and the software generates a history and physical, a letter to the referring provider, patient information sheets appropriate to the diagnosis, and a consent form if surgery is planned.

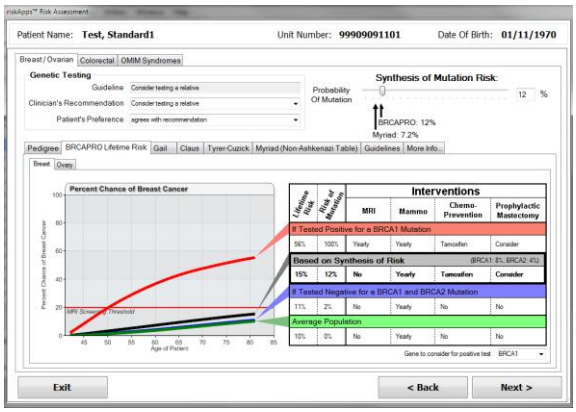


Figure 3. **Clinical Decision Support guides the workflow through making a decision about genetic testing.**

On subsequent visits, pathologic results are entered which help generate additional summary documents, and generate further suggested orders (e.g., if Estrogen receptor negative, obtain Medical Oncology consultation). The approach is to keep the surgeon in line with the appropriate quality measures in real-time, rather than tell them at the end of the year how often they were not compliant.

Street Address and Electronic Mail Address

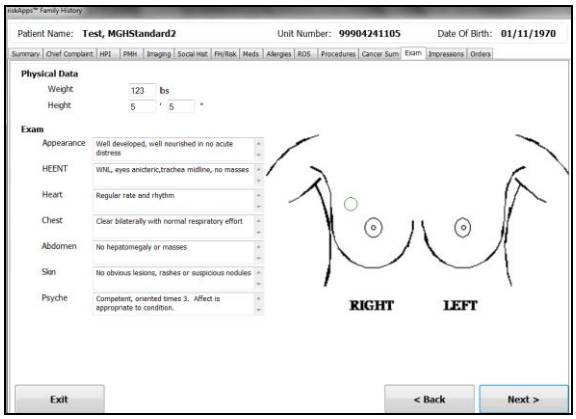


Figure 4. **The data entry screen for a breast exam in the RiskApps surgical clinic module helps entering encounter-specific information.**

## 2.1 More Women Identified

Once high risk patients are identified, the next challenge is to improve the efficiency of the risk clinic to manage the influx of patients. Our challenges are to minimize clinician work, minimize redundant data entry, and minimize dictation and editing tasks.

| Task | Traditional (minutes) | Our Approach (minutes) |
|---|---|---|
| Clinician collects family history | 0 to 10 | 0 |
| Data entered in risk calculator app | 5 to 10 | 0 |
| Data entered into pedigree drawing app | 10 to 20 | 0 |
| Risk level assessed | 5 to 10 | 5 |
| Fae to face counseling | 30 to 60 | 30 to 60 |
| Letters/notes generated | 20 to 40 | 10 |
| **Total** | **70 to 150** | **45 to 75** |

Table 1. **Comparative time costs using the traditional approach versus the Hughes RiskApps approach.**

At the Newton Wellesley Hospital Breast Center between April of 2007 and December of 2010, 49,758 unique family histories were collected and analyzed. Of those, there were 2,255 patients whose risk of mutation were greater than ten percent and were referred for counseling. The system maintains several mechanisms for tracking those identified, including a specialized queue interface listing all at risk individuals with quick access to their family history, contact information and pending appointments at the screening center. Each identified woman is also mailed a letter that explains the risks of cancer and the testing process. This letter is copied to her primary care physician as well.

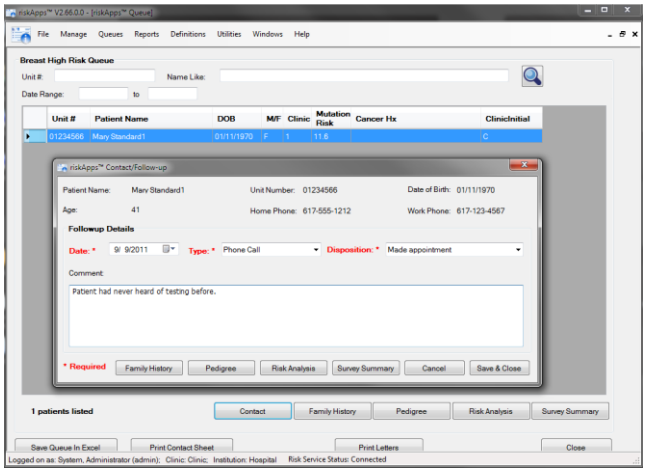


Figure 5. **The high-risk patient queue gives a clinic-wide view of which patients are at the highest risk and would benefit the most from testing.**

## 3 Document Generation

RiskApps currently generates over 80 different clinical documents specialized for a variety of workflows, saving time on dictation and cost of transcription. In the risk clinic setting, these include: a letter to the referring doctor, a letter to the patient, a progress note for the patient's chart, a letter to relatives who need testing, a letter of Medical Necessity for the patient's insurance company justifying genetic testing, and a document justifying an MRI.

The data can be easily summarized into reports required for quality measurement. The system can be set up to automatically produce performance based measures used by quality programs such as the NAPBC, NCBC, and QOPI. The system can help improve quality in 3 ways: 1.) help the clinician follow quality standards in real-time, 2.) run quality reports daily or weekly, identifying activities that do not meet the standards, or 3.) run the report yearly or when the recertification application is due.

## 3.1 Structured data and Standards

While EHRs do allow multiple clinicians to share data, the majority of meaningful information in the EHR is free text in encounter-based unstructured notes. As such, it cannot easily be organized, it typically cannot be used by CDS, and it is difficult to extract for research or quality initiative reporting. This emphasizes the need for structured data.

Structured data is data recorded in predefined fields (placeholders) using coding systems (ICD-9, ICD-10, CPT, SNOMED, etc.). As such, it is made machine readable. The beauty of structured data is that it allows the development of unified methods to view and interpret data. In today's EHRs, few structured data elements exist, and those that exist are mostly unpopulated. Most Clinicians will not take the time to enter structured data into an EHR because there is little return on investment.

As an example, the family history data elements needed by EHRs were published by AHIC in 2008 [11], and were these elements adopted and implemented there would be tremendous opportunity for visualizations (e.g., pedigrees) and risk algorithms. Instead, in practice the vast majority of recorded family history information is found as multiple dictated notes

made by multiple clinicians, while the family history section of the EHR remains mostly ignored.

Hughes RiskApps complies with the HL7 standard for representing health records. Data from our software can be shared with any HL7 compliant software. Data can be uploaded or downloaded to any EHR that has a complete family history section and that is HL7 compliant.

## 4 Conclusion and Future Work

HughesRiskApps can help us realize the promise of the genomic age on a population level. As this tool is becoming more widely used, more high risk women are being identified, family history is being integrated into normal clinic workflow, more women are being cared for by risk clinics, and risk counselors are able to act with much more efficiency.

We believe the future of RiskApps, and that of all successful EHRs, will be a modular approach. Niche vendors will be able to develop approaches specific to the needs of each specialty, and then use these as frontends to any EHR [12]. In this approach, the EHR would increase its database to house common data elements, and provide the more ubiquitous functions of allergies, ePrescribing, etc. Domain specific user interfaces provide the presentation and the organization of information specific to that specialty.

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