
Brief Communications

Electronic health records contain dispersed risk factor information that could be used to prevent breast and ovarian cancer

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ABSTRACT

Objective: The genetic testing for hereditary breast cancer that is most helpful in high-risk women is underused. Our objective was to quantify the risk factors for heritable breast and ovarian cancer contained in the electronic health record (EHR), to determine how many women meet national guidelines for referral to a cancer genetics professional but have no record of a referral.

Methods and Materials: We reviewed EHR records of a random sample of women to determine the presence and location of risk-factor information meeting National Comprehensive Cancer Network (NCCN) guidelines for a further genetic risk evaluation for breast and/or ovarian cancer, and determine whether the women were referred for such an evaluation.

Results: A thorough review of the EHR records of 299 women revealed that 24 (8%) met the NCCN criteria for referral for a further genetic risk evaluation; of these, 12 (50%) had no referral to a medical genetics clinic.

Conclusions: Half of the women whose EHR records contain risk-factor information meeting the criteria for further genetic risk evaluation for heritable forms of breast and ovarian cancer were not referred.

Key words: oncology, genetics, electronic health records

INTRODUCTION

Over a quarter-million women are diagnosed with breast and ovarian cancer each year.¹ Of these, 5–10% have cancers linked to inherited pathogenic variants that, if identified before cancer develops, might prompt an intervention to avoid morbidity and fatal disease.² Yet the genetic testing for hereditary breast cancer that is most helpful in women at increased risk for heritable cancer is underused.³ Even among women newly diagnosed with breast cancer, fewer than

half with clinical indications receive a formal genetic risk assessment.⁴ Several reasons that women do not receive formal genetic risk assessments have been identified, including older age at diagnosis, insurance status, distance from genetic services, and patient attitude about the value of genetic services. The single most important factor, however, is the lack of physician referrals to genetics services, even for patients who meet national guidelines for a formal genetic evaluation.⁵ The identification of appropriate candidates for referral at those locations where women receive primary care is essential.

The use of risk assessment tools in the primary care setting has been shown to have moderate to high accuracy in guiding which patients should be referred to a cancer genetics professional, but the use of these tools requires additional time and effort to gather personal and family history information.⁶

Risk factor information is often available in the electronic health record (EHR), because it has been gathered and stored in the course of routine care. The full story of a patient's risk for heritable cancer within their record often does not exist in a single location. It is fragmented across entries created by many authors, over many years, in many locations and formats, and commonly from many different institutions in which women have received care over their lifetimes. As a result, what patients and providers might know from the full content of EHR records differs from what they are acting on today. The focus of our study is on whether or not providers referred patients who met the National Comprehensive Cancer Network (NCCN) criteria.

We define "unrecognized EHR risk-factor information" as information that exists within a patient's EHR record but is not known by current treating providers. If this unrecognized EHR risk factor information could be found and acted on, additional women at high genetic risk could be identified and referred for genetic counseling as a preventative measure, with the goal of improving their health outcomes. The objective of this study is to characterize and quantify the unrecognized EHR risk-factor information related to breast and ovarian cancer, to determine how many women meet national guidelines for referral to a cancer genetics professional based on information in their EHR but have no record of such a referral.

MATERIALS AND METHODS

Population

We identified 9573 women who were ≥ 30 years old and were seen ≥ 5 times or hospitalized ≥ 2 times in the University of Washington (UW) Medicine health system in western Washington state between April 2018 and April 2019, using the University of Washington Enterprise Data Warehouse, and then randomly selected patients for manual review. Given the time available for chart reviews, charts of the first 299 randomly selected patients were reviewed.

Chart review

Six medical students trained in EHR use reviewed UW Medicine inpatient (Cerner) and outpatient (Epic) EHR records of the random subset of this sample to ascertain the presence and location of breast and ovarian cancer risk-factor content within EHR records in notes, reports, orders, outside records, and scanned documents. The review included UW Medicine health system records, documents received from other institutions, handwritten questionnaires completed by patients, and records viewable from other institutions using vendor information exchange tools (CareEverywhere). It included structured data, such as encoded problem lists and the Epic family history tool, and unstructured data, such as the narrative text of progress notes, consultant notes, and imaging requisitions.

Data collected

Chart reviewers recorded all risk-factor information, as defined in the NCCN Guidelines version 3.2019 criteria,⁷ using the REDCap⁸ electronic data capture tool hosted at the University of Washington. NCCN Guidelines include personal and family histories of many types of cancer, along with age of onset and degree of relatedness; Ashkenazi Jewish ancestry; known pathogenic/likely pathogenic var-

Table 1. Data collected during manual chart review

<i>Patient ID</i>
<i>Zip code</i>
<i>Race</i>
<i>Ethnicity</i>
<i>Primary care provider</i>
<i>Clinic visits last year</i>
<i>Hospitalizations last year</i>
Referred to genetics clinic (Y/N)
Date seen in genetics clinic
BMI
Age of menarche
Menopause reached (Y/N)
Menopause age
Gravida
Parity
Age at first childbirth
Hormone replacement therapy (Y/N)
Prior breast biopsy (Y/N)
Findings of breast biopsy
Breast density
BiRad
Breast cancer diagnosis (Y/N)
–For each: age at diagnosis, source of breast cancer diagnosis, triple negative (Y/N), lobular (Y/N)
Cowden Syndrome criteria (Y/N)
Personal history of pancreatic cancer (Y/N)
Personal history of ovarian, fallopian or primary peritoneal cancer (Y/N)
Ashkenazi Jewish ancestry (Y/N)
Founder mutation in relative (Y/N)
Known pathogenic/likely pathogenic variant in a cancer susceptibility gene found on tumor testing in the family
Family history of cancer (list)
–For each: information source, age of onset, relatedness, type (17 listed types + other)

Note: The list consists of data used in criteria for further genetic risk evaluations in the National Comprehensive Cancer Network Version 3.2019, Breast and/or Ovarian Cancer Genetic Assessment.⁶ The italicized text indicates data that were extracted from the Enterprise Data Warehouse rather than from a manual chart review but that were included to confirm the patient's identity during the chart review.

BiRad: Breast Imaging Reporting and Data System; BMI, body mass index; N: no; Y: yes.

iants in a cancer susceptibility gene; and other factors. A complete list of the risk-factor information recorded is in the [Table 1](#).

Determination of criteria for referral

The NCCN criteria for further genetic risk evaluation for breast and/or ovarian cancer were applied to determine which women met the criteria for referral for a genetic consultation. Of these, we noted what percentage had a record of a referral to a medical geneticist or genetic counselor. We randomly selected 10% of these charts to be reviewed a second time by another reviewer, to measure interviewer agreement. The number of charts reviewed was constrained by the time required for a manual EHR review and the project timeline. The project was approved by the University of Washington Institutional Review Board.

RESULTS

We reviewed the complete EHR records of 299 women. Each review required up to 1 hour and included a detailed review of UW Medi-

A

Hyperspace - 13 : Canceled Ord

Legal Name: [Redacted] MRN: U [Redacted] Language: English eCare: Active T/P/R: 9
 Preferred Pronoun: she/her/hers Primary Loc: Uwnc Northgate Need Intep: No ACN BP: 124
 Sex: Female, 60 year old, Adv Dir: None ACN Registry, Behavioral... Weight: [Redacted]
 PCP: [Redacted], MD, Infectio: None Care Pathways: None BMI: 23.1
 PCP InBasket Access: Yes Allergies: Penicillins, P... HM Due?: Due

Report

Physician [Redacted] MD
 Family Practice
 Progress Notes Encounter Date: 12/23/
 Signed

[Redacted] is a [Redacted] year old female here to discuss the following:

1. Breast lump - L breast, partner felt a lump above the nipple. First noticed a couple of weeks ago, not sure if it's changed. No nipple discharge. Mom had a breast bx and getting a lumpectomy. Unclear exactly the diagnosis, this is happening right now. Mom's family is ashkenazi. Other fhx cancer.
2. PT referral - R ulnar neuropathy. Did PT for this, completed 2-3 months ago, but since ending, sx have returned. Also has bumps on the arm near where the nerve passes, had considered doing u/s to further evaluate. 4th and 5th fingers get numb. Works as a barber. Has been hurting since getting in a car accident in 9/2014.
3. Finger lac - cut finger with scissors at work 1.5 weeks ago. Washed it out and glued it, and has been washing it regularly. No fevers or drainage. Just wants it checked.

REVIEW OF SYSTEMS:
Per HPI

OBJECTIVE:
BP 107/70 mmHg | Pulse 80 | Temp(Src) 98.1 °F (36.7 °C) (Temporal) | Resp 16 | Wt 139 lb (63.05 kg) | SpO2 100%
Last blood pressure recorded in this encounter: 107/70

PHYSICAL EXAM:
General: healthy, alert, no distress
Skin: has some soft, mobile nodules on the radial side of the R arm just distal to the elbow.
Ext: very tender to palpation over R ulnar groove. L 3rd finger has a 1cm healing lac over DIP. No surrounding erythema, edema, drainage
Breast: symmetrical. No drainage. Breasts are dense and fibrous. No suspicious lesions. No axillary LAD

ASSESSMENT AND PLAN:
(N63) Lump in female breast (primary encounter diagnosis)
Plan: MAMMOGRAM, SCREENING
Breast exam is reassuring, but given age, mom with breast cancer, and Ashkenazi background, recommend mammogram screening
(G56.21) Ulnar neuropathy of right upper extremity
Plan: UWNC NORTHGATE CLINIC INTERNAL REFERRAL
Refer to sports med for discussion of next steps
(S61.219S) Finger laceration, sequela
Plan: healing well. Recommend continuing supportive care.

[Redacted] MD

Office Visit on 12/23/2015 Detailed Report Note shared with patient

Figure 1. Examples of EHR documents (deidentified) containing risk-factor information listed in NCCN Guidelines Version 3.2019. (A) EHR note that includes both family history of breast cancer and Ashkenazi Jewish heritage in narrative text. (B) Outside EHR dermatology note that includes information on family history of breast and uterine cancer (arrow). (C) Scanned outside imaging requisition that includes family history of cancer (arrow). EHR: electronic health record.

cine EHRs and outside records. Risk-factor information was found in many locations within the EHRs, including scanned notes and imaging requisitions, outside notes, the family history module, and in note narrative text (Figure 1).

Using the risk-factor information in the EHR, 24 women (8%) met the NCCN criteria for referral for a further genetic risk evaluation; of these, 12 (50%) had no record of a referral to a medical genetics clinic. The location of risk-factor information for these 12 patients is shown in Figure 2. The most common risk factors

were a family history or personal history of ovarian cancer or breast cancer. Family history information was frequently found in note narrative text rather than in the EHR family history tool. Agreement between 2 independent chart reviewers was high for quantitative risk factors, including body mass index, age at the first birth, and age at menarche (kappa values = 0.93, 0.99, and 0.98, respectively; P -values much smaller than .001), but was moderate for family history of cancer (kappa = 0.55; P -value = .0038).

B

The screenshot displays the Epic EHR interface for a patient's medical record. The top navigation bar includes 'Hyperspace - UWMC GENERAL IM - Production' and various utility icons. The patient's demographic and clinical information is shown at the top, including MRN, Language (English), eCare status (Active), T/P/R (98.4 °F, 75, 16), BP (108/73), Weight (1256 lb, 14.4 oz), and BMI (40.24 kg/m²). The patient's sex is listed as Female, and allergies include Bee Venom. The main content area is titled 'Office Visit' and shows a 'Request Document Update' button. The 'Location' field is blank. The 'A/P:' field contains '(L71.9) Rosacea (primary encounter diagnosis)'. The 'Comment' field contains the following text: 'severe symptoms with cysts and excoriations that are painful'. The 'Discussed diagnosis and treatment options, side effects or risks of treatment options, written handout/pamphlet given' field contains: 'Discussed diagnosis and treatment options, side effects or risks of treatment options, written handout/pamphlet given'. The 'Plan' field contains: 'pt is also with abnormal periods (perimenopausal) -pt has apt to discuss with women's health Given ftx of breast cancer and uterine cancer may not be best candidate for hormones'. The 'Pt is no longer tolerating oral antibiotics has been on many types for months with incomplete control Has also been tried on numerous topicals (microcream etc)' field contains: 'Pt is no longer tolerating oral antibiotics has been on many types for months with incomplete control Has also been tried on numerous topicals (microcream etc)'. The 'Recommend trial of isotretinoin-full packet given' field contains: 'Recommend trial of isotretinoin-full packet given'. The 'Baseline labs' field contains: 'Baseline labs HEPATITIS B CORE AB TOTAL, HEPATITIS B SURFACE ANTIGEN, HEPATITIS B CORE-IGM, HEPATITIS B SURFACE ANTIBODY, HEPATITIS C SCREENING (REFLEX) (GHC), LIPOPROTEIN PANEL (GHC), CBC/PLT/DIFF (GHC), FASTING GLUCOSE (GHC), HEMOGLOBIN A1C, HCG, QUANTITATIVE FOR PREGNANCY, COMP, METABOLIC PANEL, HERPES 1 & 2 BY EIA, ANA SCREEN (WITH TITER IF INDICATED) (GHC)'. The 'F/u' field contains: 'F/u 1-2 months to start isotretinoin'. The 'Pt to send me secure message when fasting labs drawn to get registered in i-pledge-pt is very symptomatic so wants to get started on therapy and has few other options' field contains: 'Pt to send me secure message when fasting labs drawn to get registered in i-pledge-pt is very symptomatic so wants to get started on therapy and has few other options'. The 'Pt should also get clearance from behavior health' field contains: 'Pt should also get clearance from behavior health'. The '(L73.9) Folliculitis' field contains: '(L73.9) Folliculitis Comment: scalp severe, bx proven with necrotizing change that can also be seen as viral effect Discussed diagnosis and treatment options. side effects or risks of treatment options. written handout/pamphlet given'. A black arrow points to the 'Pt is no longer tolerating oral antibiotics...' text.

Figure 1. Continued

DISCUSSION

People at increased risk for heritable forms of breast and ovarian cancer benefit from referral to a cancer genetics professional. Comprehensive genetic counseling has been shown to reduce breast cancer–related worry and depression, increase patient understanding of risks, and reduce the intention for inappropriate genetic testing.⁴ In this random sample of women selected from among those cared for in our health-care system, we were able to identify many women whose EHRs contain risk-factor information meeting national guidelines for further genetic risk evaluation, yet half of these women had no record of a referral in their EHR. Identifying these women did not require additional surveys, visits, or outreach efforts: the risk-factor information was already in their records, but it was dispersed in notes and other locations within the EHR, so current treating physicians may not have been aware of it. Finding this information took trained reviewers far more time than most busy clinicians can reasonably devote to a chart review. However, had

the scattered risk factors for each patient been presented together to a treating provider with knowledge of NCCN guidelines, more women might have been referred to a medical geneticist or genetic counselor, and might have engaged appropriately in a discussion of the risks and benefits of genetic testing. For this reason, the dispersion of risk-factor information in the record may pose a barrier to recognizing an enhanced risk for cancer. It is a missed opportunity amenable to improvement by better methods to search and summarize EHR risk-factor information and prompt patients and physicians for additional information that could result in the identification of women who would benefit from an appropriate referral to a cancer genetics clinic for counseling and testing.

We chose to focus on hereditary breast ovarian cancer risk for 3 reasons: it is common,⁹ there are widely accepted guidelines supporting genetic evaluation,⁷ and diagnosis is linked to recommendations for treatment, such as the availability of poly-ADP ribose [PARP] inhibitors for the treatment of ovarian cancer with a BRCA1 or 2 pathogenic variant.¹⁰ The same case could be made for

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1041 190th Ave, N.E. - Screening
1135 116th Ave, N.E. Suite 2E0 - Diagnostic
Bellevue, WA 98004
6620 226th Place S.E. - Screening
Issaquah, WA 98027
P (425) 888-5985 F (425) 467-3885

PATIENT:

REFERRED BY:

Phone:

BILATERAL DIGITAL SCREENING MAMMOGRAM WITH CAD: 3/14/2009

CLINICAL: Asymptomatic. Very strong family history of breast cancer (mother, diagnosed age 42; sister, diagnosed age 53).

COMPARISON: 11/3/2007 mammogram - Overlake Breast Screening Center and 2/25/2006 mammogram - Overlake Breast Center. Views obtained Bilateral CC and MLO.

FINDINGS: There are scattered fibroglandular elements in both breasts that could obscure a lesion on mammography. No significant masses, calcifications or other findings are seen in either breast. There has been no significant interval change.

Current study was also evaluated with a Computer Aided Detection (CAD) system.

Figure 1. Continued

hereditary colorectal cancer. For other types of cancer, the evidence for a germline genetic cause is low (lung cancer) or the hereditary forms are rare and do not commonly affect treatment (sarcoma, clear cell renal carcinoma), so the benefits of this approach on health outcomes for these cancers are not clear, but may change either because of new genetic discoveries or because of changes in treatment recommendations.

Over the last decade, most US medical records have switched from paper to electronic form,¹¹ resulting in an enormous corpus of medical information in machine-readable form that did not exist when clinical cancer genetic testing was first available.

To our knowledge, this is the first report showing the amount of cancer risk-factor information in the EHR and seeking to determine its potential impact on patient referrals. Other work has focused on information in family history tools, including the use of natural language processing to find information in comments,¹² but this did not consider information in outside records, scanned documents, or in other parts of the record, nor other risks, such as Ashkenazi Jewish ancestry. As others have noted, primary care providers are overwhelmed with other clinical priorities that prevent the systematic documentation and use of family health history tools.^{13,14} If providers gather risk-factor information in the course of care—and often they do not¹⁵—it may be entered in a fashion most expeditious for the pace of clinical practice; specialized structured data capture tools may not be used because of the time required to use them.¹⁶

There have been numerous efforts to improve and centralize the capture of family history information in the EHR, using both structured (checkbox, grid and other) and unstructured methods.^{13,17,18} Though structured methods are often available to enter family history data, many providers use unstructured methods, such as narrative text, and the data entry task usually falls to the physician.¹⁹ The focus of most of these methods is family history, which is an important risk factor, but other risk factors, such as personal cancer history and ancestry, may be recorded elsewhere in the EHR outside of a dedicated family history tool.

A limitation of our study is that we focused on referrals, and not completed counseling. Though referrals are the single most important factor,⁵ there are other reasons women do not receive counseling and appropriate testing for pathogenic variants that increase the risk for cancer.

Methods exist to find this scattered risk-factor information automatically, without requiring the manual review of hundreds of pages, but are not yet a feature of most commercial EHRs. Our findings suggest that a different approach may be helpful: gathering risk-factor information from the wide range of locations and formats in which it is recorded may increase the number of women at risk who will be identified. Our results should lend impetus to apply these methods, which include image²⁰ and natural language processing,²¹ to finding important, actionable information dispersed within the records.

	UW MEDICINE EHR			OUTSIDE EHR
	Outpatient	Inpatient	FH tool	
1	● Ashkenazi in note text only			
2		● Consultant note (PDF)		● Consultant note (PDF)
3	○		●	
4		● Narrative text inpatient note	○	●
5		● Narrative text inpatient note	○	
6		● Narrative text inpatient note		
7			○	● Imaging requisition (PDF)
8	●			
9				●
10			●	● Imaging requisition (PDF)
11				●
12	●		●	●

Figure 2. Location of risk-factor information within UW Medicine and outside EHR. Each row represents a patient whose record contained information meeting the NCCN 3.2019 criteria for referral to a cancer genetics professional but who had no record of a referral. The black circles indicate the source contained information meeting the NCCN 3.2019 criteria for referral. The gray circles indicate risk-factor information not meeting the NCCN criteria, such as a family history of cancer without a mention of age. EHR: electronic health record; FH: Family history; NCCN: National Comprehensive Cancer Network; UW: University of Washington.

CONCLUSIONS

We found that half of women whose EHR record contained risk-factor information meeting criteria for further genetic risk evaluation for heritable forms of breast and ovarian cancer were not re-

ferred, and that this risk-factor information was often dispersed in the EHR in locations other than the family history tools designed to collect it. If this were gathered and presented, it could lead to discussions between women and their providers that could lead to testing that might avert new incidence, morbidity, and mortality from heritable breast and ovarian cancer.

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AUTHOR CONTRIBUTORS

All authors made substantial contributions to the conception or design of the work; contributed to the acquisition, analysis, or interpretation of data for the work; participated in drafting the work and revising it critically for important intellectual content; have finally approved the version to be published; and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

CONFLICT OF INTEREST

None declared.

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