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SHORT COMMUNICATION

Patient-reported hereditary breast and ovarian cancer in a primary care practice

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Abstract Identifying women appropriate for cancer genetic counseling referral depends on patient-reported family history. Understanding predictors of reporting a high-risk family is critical in ensuring compliance with current referral guidelines. Our objectives were to (1) assess prevalence of candidates for *BRCA1* and *BRCA2* counseling referral in a primary care setting, (2) explore associations with high-risk status and various patient (e.g., race) and family structure (e.g., number of relatives) characteristics, and (3) determine whether high-risk patients had genetic counseling and/or testing. Survey and pedigree data were collected between 2010 and 2012 for 486 Women's Health Clinic patients. Analyses in 2013 investigated perceived cancer risk and worry, family structure, and receipt of genetic counseling. We explored whether these were associated with meeting USPSTF guidelines for genetic counseling referral. Twenty-two (4.5 %) women met the criteria for *BRCA* referral. Only one of these women had previous genetic counseling, and one reported prior genetic testing. Older women were more likely to meet *BRCA* referral criteria ($P<0.001$). Although perceived risk was higher among high-risk women, 27 % of high-risk women felt their breast cancer risk was "low", and 32 % felt their risk was lower than average. About one in 22 women in primary care may require genetics services for hereditary breast and ovarian cancer, but alarmingly, few actually receive these services. Also, a significant proportion do not perceive that they are at increased risk. Educational interventions may be needed for both providers and patients to increase awareness of familial risk and appropriate genetic counseling services.

Keywords Hereditary breast and ovarian cancer syndrome · Primary health care · Women's health · Genetic counseling

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Introduction

Family history is a major risk factor for breast and ovarian cancers. Hereditary risk is often linked to mutations in the *BRCA1* and *BRCA2* genes. Identifying and referring genetic testing candidates in primary care is recommended by the United States Preventive Services Task Force (USPSTF) and an associated Healthy People 2020 objective (US Department of Health and Human Services 2013; US Preventive Services Task Force 2005).

Unfortunately, most primary care practices do not collect adequate family histories for risk triage (Flynn et al. 2010; Murff et al. 2004, 2007; Sifri et al. 2002). Lack of systems for family history collection, patients' lack of knowledge and/or interest, limited time for collection and interpretation, lack of guideline knowledge, and insurance concerns contribute to this deficit (Brandt et al. 2008; Qureshi et al. 2007; Wood et al. 2008). Family history collection tools continue to be developed, but no clear winner has emerged (Qureshi et al. 2009). Regardless, any family history collection tool will likely depend on information provided by the patient.

The validity of these tools can only be as good as the family histories patients report.

Understanding the prevalence and predictors of patient-reported familial breast and ovarian cancers can help in deciding how best to collect and interpret this. For example, lower-than-expected reporting of familial breast cancer in clinic could mean that patients are not fully aware of their family history. These patients could be the focus for educational interventions.

We present patient-reported family history information in a primary care Women's Health Clinic, relevant for referral for *BRCA1* and *BRCA2* genetic counseling. Our analyses benefit from a study sample that is diverse and not preselected for high-risk status. Our objectives were to (1) assess the prevalence of candidates for *BRCA1* and *BRCA2* counseling referral in a primary care setting, (2) explore the associations with high-risk status and various patient (e.g., race) and family structure (e.g., number of relatives) characteristics, and (3) determine whether high-risk patients had genetic counseling and/or testing.

Research methods

We analyzed the baseline data collected from the Kin Fact Study, a longitudinal randomized controlled trial that will test the effects of a brief intervention on family communication about hereditary cancer risk. Participants (N=490) were adult Women's Health Clinic patients at Virginia Commonwealth University Health System (VCUHS) who were enrolled in the Kin Fact Study (R01 CA140959).

Recruitment occurred 4 days per week for 18 months in the waiting areas of the two academic Women's Health Clinics, a faculty practice and a resident practice, in downtown Richmond, Virginia, at VCUHS. Women presenting for care who did not appear to be acutely ill were consecutively approached by the study coordinators in the waiting room about study participation. In 2011, nearly 30,000 patient visits occurred at these clinics, and 63 % of these visits were with African–American women. Typically, only one recruiter was present in the clinic at a time, so not all patients could feasibly be invited. Of those approached within the residents' clinic, 69 % were African–American, 24 % were Caucasian, and 7 % included all other races. Of those approached within the faculty practice, 49 % were African–American, 42 % were Caucasian, and 9 % included all others. These differences are consistent with the patient demographics of both clinics.

Between July 2010 and January 2012, 1,046 women were approached for study participation. Of 874 eligible women, 518 enrolled in the study. Twenty-eight of these women (5 %) did not complete baseline measures, resulting in a final sample of 490 women.

Participants were first asked to complete a self-administered pen-and-paper survey. We excluded from the analyses women (N=4) with personal histories of breast or ovarian cancer. Participants were asked, "A genetic counselor is a health care professional who works with people to identify risks for various genetic conditions including cancer. Have you ever seen a genetic counselor to discuss cancer risks?" and "Have you ever had genetic testing for cancer risk? ('Genetic testing' refers to a blood test that looks for a marker of cancer risk that can be passed on in families like other traits.)" Other survey items preselected for analyses were three measures of perceived risk for breast cancer; worry about breast cancer as assessed by the Cancer Worry Scale (Lerman et al. 1991), and the Cancer Worry Chart (Gramling et al. 2007), race, and age. For perceived risk, participants were asked three questions assessing numeric, verbal, and comparative risk, respectively, as follows: (1) "Of 100 women with your same breast cancer risk, how many will get breast cancer by the time they are 90 years old?", (2) "How would you rate your risk of developing breast cancer" (low, moderate, high), and (3) "Compared to most women your age and race/ethnicity, what do you think your chances are that you will get breast cancer? (much lower than average, a little lower than average, average, a little higher than average, much higher than average—which were analytically collapsed to three responses: lower, average, higher). The Breast Cancer Worry Scale consists of four items. Frequency of worry (rarely or never, sometimes, often, all the time) was assessed by three questions as follows: (1) "How frequently do you worry about getting breast cancer?", "How frequently does worry about breast cancer affect your mood?", "How frequently does worry about breast cancer affect your daily functioning?" One additional item in the scale asked, "How concerned are you about breast cancer?" (not at all, somewhat, moderately concerned, or very concerned). The Cancer Worry Chart rates worry on a scale from 1 (low) to 5 (high) with corresponding pictures of faces, similar to a pain scale.

After completing the survey, two recruiters (one a certified genetic counselor and the other a doctoral student trained in family history taking) obtained a family pedigree noting all breast and ovarian cancers among first- and second-degree relatives. They also administered the rapid estimate of adult literacy in genetics, which correlates highly with other measures of health literacy and general educational background and may be related to awareness of family health history (Erby et al. 2008; Yoon et al. 2004).

Statistical analyses

Data were analyzed in 2013 using the R statistical package (v2.15.1) and SAS (v9.3). Family histories were categorized as meeting (or not) USPSTF criteria for *BRCA* genetic counseling referral (US Preventive Services Task Force 2005). Bivariate associations were investigated for high-

risk status with each dependent variable (having had genetic counseling and/or testing for cancer risk, perceived cancer risk, and cancer worry) and potential confounders (age, race, genetic literacy, family size, and difference between number of maternal and paternal relatives). Chi-square testing was used for categorical variables, and *t* tests were used for continuous variables. Regression models (linear or ordinal logistic, as appropriate) were fitted controlling for confounders that had at least nominally significant associations ($P<0.10$) with high-risk status in bivariate tests. In regression models, *P* values less than 0.05 were considered statistically significant. We did not correct alpha for multiple statistical testing.

Results

Table 1 gives participant characteristics. Average age was 33 years. Most (59 %) participants were African–American. Twenty-two (4.5 %) met the criteria for *BRCA* genetic counseling referral (Table 2). The mean age for women with genetic cancer risk was 40 years compared with 33 years for average-risk women ($P=0.007$) There was no significant difference in the number of maternal and paternal relatives reported in high-risk women versus those not meeting USPSTF criteria. Race was nominally significant ($P=0.054$), with 3 % of African–American participants being high risk, 8 % of Caucasians, and 0 % of women reporting other race.

Among the 22 participants meeting the criteria for *BRCA* counseling and testing, only one self-reported meeting with a genetic counselor to discuss cancer risks occurred, and this person did not have genetic testing. Another person meeting high-risk criteria reported having genetic testing for cancer risk, but did not meet with a genetic counselor.

Table 1 Participant characteristics ($N=486$)

Variable	No. (%)
Age (years) (M, SD)	33.2, 11.8
Race	
Black or African–American	287 (59.0)
White or Caucasian	160 (32.9)
Other race	39 (8.0)
REAL-G score (M, SD), range 0 to 8[a]	5.8, 2.3
Family size (First- and second-degree relatives)	
Total (M, SD)	17.5, 7.0
Paternal relatives[b]	5.5, 3.7
Maternal relatives[c]	7.0, 3.3

[a] REAL-G score was not obtained for three participants

[b] Thirty-two women reported no paternal family members

[c] Three women reported no maternal family members

Table 2 Women's Health Clinic patients ($N=486$) recommended for *BRCA* genetic counseling referral

High-risk criterion[a]	No. (%)
Two first-degree relatives with breast cancer, at least one diagnosed at age 50 or younger	1 (0.2)
Three or more first- or second-degree relatives with breast cancer	11 (2.3)
Breast and ovarian cancer among first- and second-degree relatives	5 (1.0)
Bilateral breast cancer in a first-degree relative	3 (0.6)
Two or more first- or second-degree relatives with ovarian cancer	1 (0.2)
Single relative with breast and ovarian cancer at any age	3 (0.6)
Male breast cancer in a relative	1 (0.2)
Jewish ancestry and a first-degree relative with breast or ovarian cancer	4 (0.8)
Jewish ancestry and at least two second-degree relatives on the same side of the family with breast or ovarian cancer	1 (0.0)
Meets any criterion for *BRCA* genetic counseling referral	22 (4.5)

[a] Based on the United States Preventive Services Task Force recommendation, 2005

Overall, perceived cancer risk was higher among women with familial cancer risk (Table 3). Still, 27 % of women with familial breast/ovarian cancer felt their risk was "low", and 32 % felt their risk was lower than average. Cancer worry was similar between groups.

Discussion

The findings from this study suggest that 1 in 22 primary care patients may meet referral guidelines for *BRCA* genetic counseling. Two recent reports suggested a similar prevalence—one in general population (Office of Disease Prevention and Health Promotion, Department of Health and Human Services) and another in a large health system (Bellcross et al. 2013). A progress review of the Healthy People 2020 genomics objectives suggested that the *BRCA* referral target rate was met, with more than half fulfilling this objective. However, this was based on eligible individuals confirming that they had "ever discussed the possibility of getting a genetic test for cancer risk with a health care provider." Another study suggested that 90 % of eligible women reported having talked with their provider about family history, but fewer than 20 % were actually referred for genetic counseling. Our data suggest that an even smaller proportion (2 out of 22) actually received genetic counseling or genetic testing. In sum, there appears to be a critical gap between women eligible for *BRCA* counseling and those receiving the services. Our study did not assess why these high-risk women had not received genetic counseling. We do not know,

Table 3 Association of familial breast/ovarian cancer with perceived breast cancer risk and worry[a]

Variable	Familial breast/ovarian cancer	No familial risk	*P* value
Numeric lifetime risk (0 to 100)[b]	43.0 (32.7, 53.2)	29.5 (26.5, 32.5)	0.010
Verbal risk description[c]	OR=3.8 (1.6, 8.7)	Ref	0.002
Low[d]	6 (27.3)	280 (60.5)	
Moderate[d]	12 (54.5)	159 (34.3)	
High[d]	4 (18.2)	24 (5.2)	
Risk compared to average[c]	OR=3.5 (1.5, 7.8)	Ref	0.003
Lower[d]	7 (31.8)	264 (57.0)	
Average[d]	5 (22.7)	137 (29.7)	
Higher[d]	10 (45.4)	62 (13.2)	
Breast cancer worry scale (4 to 16)[b]	6.7 (5.8, 7.6)	6.0 (5.8, 6.3)	0.133
Breast cancer worry chart (1 to 5)[b]	1.6 (1.3, 1.9)	1.4 (1.3, 1.5)	0.313

[a] Adjusted for age and race
[b] Least squares mean (95 % CI)
[c] Odds ratio (95 % CI)
[d] Frequency (%)

for example, if these services were in fact recommended by their doctors and the women elected not to receive genetic counseling. Furthermore, receipt of genetic services was only measured by self report and not confirmed by medical records review.

We found age and race to be potentially associated with patient-reported familial cancer. Association with age is consistent with at least one earlier large study, and this makes sense, considering the chance for breast cancer increases with age (Ziogas et al. 2011). Primary care providers often see patients repeatedly over many years. While cancer screening is recommended with some periodicity (e.g., yearly mammograms), family history taking might not be updated at regular intervals (ASHG Statement 1998; Acheson et al. 2000). This omission could miss hereditary risk in women as they get older. Association between race and high-risk status was nominally significant. If our finding is not due to random sampling variation, this difference could reflect a lower incidence of breast and ovarian cancers among African–American women in the general population, or perhaps under/overreporting of family history by race group. Given the known race disparities in genetic counseling (e.g., Thompson et al. 2012), this finding should be followed up in future studies.

Importantly, our study involved a sociodemographically diverse population, and we found no significant differences by genetic literacy or family structure, suggesting that these should not be barriers to equitable family history collection, assessment, and referral. On the other hand, our study involved a relatively small number of high-risk women (N=22), limiting statistical power to detect associations. While our study demonstrated that high-risk women based on family history were more likely to perceive themselves at elevated risk, a sizeable proportion still considered themselves to be at low risk—an opportunity for patient education.

Our study has limitations. Our sample was derived from a single institution. The accuracy of reported family histories was not verified, and thus the classification of high-risk women may not be valid (Mai et al. 2011; Ziogas and Anton-Culver 2003). Our sample included volunteers recruited from a Women's Health clinic. Although our sample characteristics closely reflect those of typical patients at this clinic, there is the possibility of respondent bias.

These limitations notwithstanding, this study provides compelling evidence that a significant proportion of primary care patients require genetic counseling referral, yet few are receiving these services.

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