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ORIGINAL REPORT

Quality of Cancer Family History and Referral for Genetic Counseling and Testing Among Oncology Practices: A Pilot Test of Quality Measures As Part of the American Society of Clinical Oncology Quality Oncology Practice Initiative

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Terms in blue are defined in the glossary, found at the end of this article and online at www.jco.org.

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ABSTRACT

Purpose
Family history of cancer (CFH) is important for identifying individuals to receive genetic counseling/testing (GC/GT). Prior studies have demonstrated low rates of family history documentation and referral for GC/GT.

Methods
CFH quality and GC/GT practices for patients with breast (BC) or colon cancer (CRC) were assessed in 271 practices participating in the American Society of Clinical Oncology Quality Oncology Practice Initiative in fall 2011.

Results
A total of 212 practices completed measures regarding CFH and GC/GT practices for 10,466 patients; 77.4% of all medical records reviewed documented presence or absence of CFH in first-degree relatives, and 61.5% of medical records documented presence or absence of CFH in second-degree relatives, with significantly higher documentation for patients with BC compared with CRC. Age at diagnosis was documented for all relatives with cancer in 30.7% of medical records (BC, 45.2%; CRC, 35.4%; $P \leq .001$). Referall for GC/GT occurred in 22.1% of all patients with BC or CRC. Of patients with increased risk for hereditary cancer, 52.2% of patients with BC and 26.4% of those with CRC were referred for GC/GT. When genetic testing was performed, consent was documented 77.7% of the time, and discussion of results was documented 78.8% of the time.

Conclusion
We identified low rates of complete CFH documentation and low rates of referral for those with BC or CRC meeting guidelines for referral among US oncologists. Documentation and referral were greater for patients with BC compared with CRC. Education and support regarding the importance of accurate CFH and the benefits of proactive high-risk patient management are clearly needed.

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INTRODUCTION

Identification of individuals with a hereditary form of cancer is important for management of that individual and his or her family. Individuals found to have hereditary cancer or cancer predisposition are candidates for increased surveillance (eg, screening breast magnetic resonance imaging, early and more frequent colonoscopy) and prevention options (eg, oophorectomy, bilateral mastectomy, near total colectomy).[1,2] Clues that an individual may have hereditary cancer or cancer predisposition syndrome come from the family history. Those individuals with a significant family history of cancer are candidates for genetic testing.[3,4]

The National Comprehensive Cancer Network (NCCN), US Preventive Services Task Force, and American College of Obstetricians and Gynecologists have specific guidelines regarding referral for genetic counseling based on family history for a number of different settings (eg, hereditary breast and ovarian cancers, Lynch syndrome, Li-Fraumeni syndrome).[2,4,5] The importance of family history in identification of individuals at increased risk for

cancer is also reflected in guidelines for high-risk screening for breast, colon, prostate, and ovarian cancers.[6-9]

Despite the importance of family history in cancer risk assessment, studies have shown that physicians do not always take and/or update a family history[10-12] and that there is a lack of completeness of the documented family history.[13,14] Furthermore, individuals meeting family history–related criteria for referral for genetic counseling and/or testing are often not referred.[14-16]

The genetics community has established a standard that a complete family history should include a three-generation pedigree, obtaining information regarding age or year of birth for each individual, age and cause of death for those deceased, ethnic background of each grandparent, relevant health information, illnesses and age at diagnosis, prior genetic testing, pregnancies, half-siblings, and consanguinity.[17] However, this level of evaluation is labor intensive and thus unlikely to be obtained for every patient in a busy clinical oncology practice. Additionally, there is no evidence that a three-generation pedigree is necessary to obtain the critical information needed to identify candidates for more-intensive screening practices, prevention strategies, and cancer predisposition genetic counseling and testing.[17] Clearly a more concise and accurate family history will likely contain the required information needed to identify those individuals who would benefit from additional screening and/or genetic counseling and testing.

The purpose of the current study was to assess family history taking, genetic counseling, and genetic testing practices among oncology practices participating in the Quality Oncology Practice Initiative (QOPI), with the larger goal of identifying important steps to improve family history taking and referral for cancer genetic counseling and testing.

Table 1. Criteria for Increased Hereditary Risk

Criteria	Breast Cancer	Colorectal Cancer
Patient history	Diagnosed at age ≤ 45 years	Diagnosed at age < 50 years
	History of epithelial ovarian, fallopian tube, or primary peritoneal cancer	History of another cancer consistent with HNPCC syndrome
	Two breast primaries with initial diagnosis at age < 50 years	High results on MSI testing (MSI-H) and diagnosed at age < 60 years
Family history	Male blood relative with breast cancer	First-degree blood relative with colorectal, ovarian, or endometrial cancer diagnosed at age < 50 years
	Blood relative diagnosed with cancer (when patient diagnosed at age ≤ 50 years)	≥ Two related first- or second-degree blood relatives with colorectal, ovarian, or endometrial cancer
	Blood relative with known mutation in breast cancer susceptibility gene (*BRCA1*, *BRCA2*, *PTEN*, *TP53*, *CDH1*)	
	≥ Two blood relatives from same side of family (maternal or paternal) with breast, epithelial ovarian, fallopian tube, or primary peritoneal cancer (when patient diagnosed at age ≤ 50 years)	

Abbreviations: HNPCC, hereditary nonpolyposis colorectal cancer; MSI, microsatellite instability.

METHODS

The American Society of Clinical Oncology (ASCO) established QOPI in 2002.[18] QOPI is an oncologist-led, practice-based quality assessment and improvement program with the goal of promoting excellence in cancer care by helping practices create a culture of self-examination and improvement. Participating practices abstract information from patient medical records to determine compliance with measures covering core areas (care documentation, chemotherapy administration, pain management, smoking cessation, and psychological support) in disease-specific domains (breast cancer, colorectal cancer, non-Hodgkin lymphoma, and non–small-cell lung cancer) and domain-specific areas (end-of-life care, symptom and toxicity management). Practices are required to collect data in at least two modules, along with all of the core measures. Medical records included in the QOPI sample must be for patients diagnosed within the 2 preceding years who have been seen at least twice in the recent 6-month period.

To develop questions regarding family history taking and genetic counseling and testing practices, an expert team composed of individuals from the ASCO Cancer Prevention (M.E.W., K.H.L., J.N.W., K.S.H.) and Quality of Care (M.N.N.) Committees was assembled. The team focused on developing measures for breast or colon cancer, where the role of genetics is the most clinically advanced, with available and well-publicized practice guidelines.[19,20] Measures were developed around two domains: family history taking and genetic counseling and testing. Questions were designed and modified by consensus using an iterative process of meetings, conference calls, and document reviews. Measures of family history taking included: documentation of first- and second-degree cancer family histories, and age at cancer diagnosis for family members with cancer. Measures of genetic counseling and testing practice included: referral for genetic counseling and/or genetic testing for any patient with breast or colorectal cancer and for those most likely to have a hereditary form (criteria listed in Table 1) of breast or colorectal cancer, and referral for individuals who underwent genetic testing within the practice, documentation of counseling, informed consent, and result disclosure. Consensus on criteria for hereditary breast or colorectal cancer was achieved through examination and evaluation of available guidelines to generate a simple list for data extraction (Table 1). For hereditary breast cancer, criteria from ASCO,[4,21] NCCN,[20] and the US Preventative Services Task Force[22] were used, and for hereditary colon cancer, criteria from ASCO[4] and NCCN[19,23] were used.

Once completed, the consensus measures were incorporated as pilot measures into QOPI during the fall 2011 collection round. Practices were provided notes and instructions on what information to abstract for all measures. As pilot measures, participation was optional; practices could choose to answer or skip these questions for each medical record abstracted.

Characteristics of practices and patients were compared using descriptive measures. Differences between patients with breast and colorectal cancers and between patients referred and not referred for genetic counseling and testing were assessed by t tests for continuous variables and χ^2 tests for categorical variables. Multivariate logistic regression was used to assess referral rate differences between patients with breast and colorectal cancers, adjusting for age at diagnosis, stage at diagnosis, and chemotherapy status. Analyses were conducted using SAS software (version 9.3; SAS Institute, Cary, NC).

RESULTS

Participants

Between September and October 2011, 271 practices participated in QOPI, and 212 practices (78%) chose to participate in the family

Table 2. Characteristics of Participating Practices

Characteristic	Practices (n = 212)	Breast Cancer Pilot Questions (n = 195)	Colorectal Cancer Pilot Questions (n = 147)	P*
No. of medical oncologists				.51
Mean	8.2	7.7	8.5	
SD	9.7	9.3	10.2	
No. of office locations				.76
Mean	2.6	2.6	2.5	
SD	2.9	2.9	2.4	
Practices with genetic counselor, %	36.5	33.9	38.5	.68
Census region, %				.94
Midwest	40.1	42.1	40.8	
Northeast	18.4	16.4	19.1	
South	27.8	27.7	26.5	
West	13.7	13.8	13.6	
Practice type, %				.67
Private with academic affiliation	10.1	9.4	13.1	
Private independent	55.0	57.3	51.7	
Employee	18.2	19.3	17.9	
Academic full time	12.9	11.5	13.1	
Fellowship program	3.8	2.6	4.1	

Abbreviation: SD, standard deviation.
*P value corresponds to differences between breast and colorectal cancers.

Table 3. Patient Demographic and Clinical Characteristics by Cancer Type

Characteristic	Total		Breast Cancer		Colorectal Cancer		P*
	No.	%	No.	%	No.	%	
Age at diagnosis, years							< .001
Mean	59.5		58.0		61.9		
SD	13.2		13.2		13.2		
< 40	659	6.3	471	7.2	188	4.8	
40-49	1,838	17.6	1,341	20.4	497	12.8	
50-59	2,760	26.3	1,796	27.3	964	24.7	
60-69	2,732	26.1	1,681	25.6	1,051	27.0	
≥ 70	2,477	23.7	1,280	19.5	1,197	30.7	
Female sex	8,439	80.6	6,569	100.0	1,870	48.0	
Disease stage							< .001
I	2,186	24.8	2,037	35.8	149	4.8	
II	3,031	34.5	2,323	40.8	708	22.8	
III	2,428	27.6	972	17.1	1,456	46.8	
IV	1,151	13.1	356	6.3	795	25.6	
Received chemotherapy	8,079	77.2	4,706	71.6	3,373	86.6	< .001

Abbreviation: SD, standard deviation.
*P value corresponds to differences between breast and colorectal cancers.

history pilot. For pilot testing, 10,466 medical records of patients with breast or colorectal cancer were reviewed. Practices that chose to participate in the pilot test measures were not different from those that did not with respect to number of medical oncologists in the practice, number of practice sites, affiliation, and geographic location (data not shown). Similarly, practices participating in the breast cancer pilot questions were not different from practices participating in the colorectal cancer pilot questions (Table 2). Medical records of patients diagnosed between August 2009 and August 2011 and seen twice within a 6-month period were reviewed. Table 3 lists characteristics of those patients. Family history and genetic counseling information noted in the medical record at any time from a patient's initial diagnosis could be used for abstraction.

Family History Taking

In terms of family structure, 79.8% of patients with breast or colorectal cancer had a first-degree family history (with or without cancer) documented in their medical record, and 64.6% of patients with breast or colorectal cancer had a second-degree family history (with or without cancer) documented. Both first- and second-degree family histories were significantly more commonly documented for those with breast cancer compared with colon cancer ($P < .001$; Table 4). After accounting for identified differences between patients with breast and colorectal cancers (such as age, stage, and receipt of chemotherapy), there remained a significant difference in documentation of first- and second-degree histories between the two groups of patients.

Of those with a positive family history, 41.7% had age at cancer diagnosis of the relative with cancer documented in their medical record. Age at diagnosis was significantly more commonly recorded for patients with breast cancer than for those with colorectal cancer (Table 4); again, this significant difference persisted even after accounting for known differences between the groups. A documented complete cancer family history existed for 32.9% of patients with breast cancer and 22.0% of those with colorectal cancer (defined as containing documentation of first- and second-degree family histories and age at cancer diagnosis for family members with cancer).

Referral for Genetic Counseling and/or Testing

Referral for genetic counseling and/or testing occurred for 25.6% of all patients with breast or colorectal cancer. Examination of patients referred for genetic counseling/testing compared with those who were not referred revealed that those referred were significantly younger, more likely to be women, and more likely to have earlier-stage cancer (stage I or II *v* III or IV; data not shown). Of the 2,457 patients (23.5% of all patients) at risk for hereditary cancer (using criteria summarized in Table 1), only 43% were referred for genetic counseling and/or

Table 4. Extent of Family History Documentation in Medical Records of Patients With Breast or Colorectal Cancer

Family History Documented	Total (N = 10,466)	Breast Cancer (n = 6,569)	Colorectal Cancer (n = 3,897)	P*
First degree, %	79.8	81.2	77.4	< .001
Second degree, %	64.6	68.9	57.3	< .001
	n = 7,714	n = 4,984	n = 2,730	
Documented age at diagnosis, %	41.7	45.1	35.4	< .001
Complete family history, %†	29.1	32.9	22.0	< .001

*P value corresponds to differences between breast and colorectal cancers.
†Complete family history is defined as presence of first- and second-degree family histories and documented age at cancer diagnosis.

Table 5. Referral of Patients With Breast or Colorectal Cancer for Genetic Counseling and/or Testing

Referral	Total (N = 10,466)	Breast Cancer (n = 6,569)	Colorectal Cancer (n = 3,897)	P*
Referred for genetic counseling and/or testing, %	25.6	29.1	19.6	< .001
	n = 2,457	n = 1,556	n = 901	
Positive family history and referred, %	42.7	52.2	26.4	< .001

*P value corresponds to differences between breast and colorectal cancers.

testing. Referral and testing rates were higher for those with breast compared with colorectal cancer ($P < .001$; Table 5). These differences remained significant even after accounting for factors known to be different between patients with breast and colorectal cancers in this group. For patients with breast or colorectal cancer who were tested within the practice, 77.7% had consent documented, and 78.8% had result disclosure discussion documented.

DISCUSSION

We have shown that oncologists participating in the ASCO QOPI document first- and second-degree family histories fairly consistently in their patients with breast or colorectal cancer. However, they do significantly less well identifying age at cancer diagnosis of individuals in families and collect a complete cancer family history in < 40% of patients with breast or colorectal cancer. Low rate of documentation of the complete cancer family history is not a new finding and has been identified by several other groups[13,24-26]; however, this sample is by far the largest and most geographically diverse group of providers sampled, to our knowledge. Our study highlights the continued need for education and support for oncology providers on gathering and documenting family history. Getting an adequate family history into the medical records is the first step in ensuring that the proper patients are referred for genetic counseling/testing.

The significant difference in documentation of family history between patients with breast and colon cancers is striking and persisted even after accounting for the fact that those with colorectal cancer in this data set were older, more likely to have advanced-stage disease (stage III and IV), and more likely to have received chemotherapy. Provider-related factors (eg, presence of genetic counselor, academic affiliation, or geographic location) between the two groups of patients were not different. It is not clear why there would be differences in family history documentation between patients with breast and colorectal cancers other than maybe awareness of hereditary risks related to either type of cancer. Carroll et al[27] found that 91% of primary and specialty physicians were aware of genetic testing for breast and ovarian cancers, but only 60% of this group was aware of testing for colorectal cancer. Provider- and practice-related characteristics should be explored further to see if there are identifiable practice barriers that if overcome could result in better family histories being documented.

In this study, 25.6% of patients with breast or colon cancer were referred for genetic counseling and/or testing, and more than three quarters had consent and disclosure discussions documented, which is encouraging. However, we also found that less than half (42.7%) of those who were likely to have a hereditary form of breast or colorectal cancer were referred, again with higher rates of referral for those with breast (52%) compared with colorectal cancer (26%). Ours is not the first study to document low rates of referral of eligible patients for consideration of genetic testing.[16,28-30] Reasons for low referral are varied and range from provider knowledge, provider time, and poor patient knowledge of family history.[31] Drohan et al[32] estimated that ≤ 5% of unaffected *BRCA1* and *BRCA2* mutation carriers are being identified, whereas only 1% of Lynch syndrome carriers are being identified. Improvements in documentation and interpretation of family history will be required to assure that greater numbers of at-risk individuals are being identified.

This study would also suggest that approximately 60% of patients referred for counseling and testing did not meet the minimal guidelines for referral (Table 1) used in this measure. Although many of these patients may have met more-thorough guidelines, it is also possible that many of these were unnecessary referrals that may have led to unnecessary testing and health care costs. The risks of both over- and undertesting should be addressed.

Deeker et al[33] found that 23% of patients with colorectal cancer at low familial risk were referred for preventative measures. Trivers et al[28] found that only 71% of physicians would adhere to recommendations against genetic counseling or testing for average-risk women in a vignette-based survey of 3,000 US primary care providers. Unfortunately, this suggests that 29% of average-risk women would be referred. Although these referred individuals may gain reassurance from counseling and/or testing, it is important to consider the resources necessary to provide this level of reassurance.

Strengths of this study include its large sample size of oncologists across the United States and the unbiased use of assessment through ASCO QOPI. One bias in this study is that quality-focused practices are more likely to participate in QOPI. However, the identified gaps in care might be expected to be greater among nonparticipating community practices.

In conclusion, family history is key to identifying individuals at risk for both primary and secondary cancers and to identifying those individuals most likely to benefit from genetic counseling and/or testing. In this pilot test of QOPI measures, we identified a higher rate of cancer family history documentation than expected, and we identified significant genetic counseling and testing activity. There is significant room for improvement with respect to both documentation of family history and appropriate referral for genetic counseling and testing. Professional education about the important elements of an accurate cancer family history (first- and second-degree histories, including age of diagnosis for those relatives with cancer) and the benefits of proactive high-risk patient management may improve the appropriate referral rate and improve the identification and management of patients at high risk. Additionally, systems improvements such as improved health information technology are needed to help providers achieve better family history taking and genetic counseling and testing practices in the future.

AUTHORS' DISCLOSURES OF POTENTIAL CONFLICTS OF INTEREST

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GLOSSARY TERMS

***BRCA1*:** A tumor suppressor gene, the breast cancer 1 susceptibility gene is known to play a role in repairing DNA breaks. Mutations in this gene are associated with increased risks of developing breast or ovarian cancer.

***BRCA2*:** Known as breast cancer 2 early onset gene, *BRCA2* is a tumor suppressor gene whose protein product is involved in repairing chromosomal damage. Although structurally different from *BRCA1*, *BRCA2* has cellular functions similar to *BRCA1*. *BRCA2* binds to RAD51 to fix DNA breaks caused by irradiation and other environmental agents.

***PTEN* (phosphatase and tensin homolog):** *PTEN* is a tumor suppressor gene with a gamut of regulatory activities. The gene product is a multifunctional molecule. The predominant activity identified for *PTEN* is its lipid phosphatase activity that converts inositol trisphosphates into inositol bisphosphates, thus inhibiting survival and proliferative pathways that are activated by inositol trisphosphates. *PTEN* acts to maintain arrest in the G1 phase of the cell cycle and enable apoptosis through an AKT-dependent mechanism.

***TP53*:** Gene encoding p53, a nuclear protein, which plays an essential role in the regulation of cell cycle. Mutations in *p53*, resulting in proteins that fail to bind DNA, frequently occur in a number of different human cancers, resulting in a loss of tumor-suppressor activity. Alterations of the *TP53* gene occur as somatic mutations in human malignancies and as germline mutations in some cancer-prone families with Li-Fraumeni syndrome.