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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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%; \( P < .001 \)). 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.

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 history10–12 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.

### 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 (cancer 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 family history–related criteria for referral for genetic counseling and testing.18

Table 1. Criteria for Increased Hereditary Risk

<table>
<thead>
<tr>
<th>Criteria for Increased Hereditary Risk</th>
<th>Breast Cancer</th>
<th>Colorectal Cancer</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient history</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Diagnosed at age ≤ 45 years</td>
<td>Diagnosed at age &lt; 50 years</td>
<td></td>
</tr>
<tr>
<td>History of epithelial ovarian, fallopian tube, or primary peritoneal cancer</td>
<td>History of another cancer consistent with HNPCC syndrome</td>
<td></td>
</tr>
<tr>
<td>Two breast primaries with initial diagnosis at age &lt; 50 years</td>
<td>High results on MSI testing (MSI-H) and diagnosed at age &lt; 60 years</td>
<td></td>
</tr>
<tr>
<td>Family history</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male blood relative with breast cancer</td>
<td>First-degree blood relative with colorectal, ovarian, or endometrial cancer diagnosed at age &lt; 50 years</td>
<td></td>
</tr>
<tr>
<td>Blood relative diagnosed with cancer (when patient diagnosed at age ≤ 50 years)</td>
<td>Two related first- or second-degree blood relatives with colorectal, ovarian, or endometrial cancer</td>
<td></td>
</tr>
<tr>
<td>Blood relative with known mutation in breast cancer susceptibility gene (BRCA1, BRCA2, PTEN, TP53, CDH1)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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

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 χ² 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
family histories and documented age at cancer diagnosis.

**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 vs 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 2. Characteristics of Participating Practices

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Breast Cancer Pilot Questions (n = 199)</th>
<th>Colorectal Cancer Pilot Questions (n = 147)</th>
<th>$P^*$</th>
</tr>
</thead>
<tbody>
<tr>
<td>No. of medical oncologists</td>
<td>Mean 8.2, SD 7.7</td>
<td>Mean 9.7, SD 9.3</td>
<td>.51</td>
</tr>
<tr>
<td></td>
<td>Mean 9.7, SD 9.3</td>
<td>Mean 10.2, SD 10.2</td>
<td></td>
</tr>
<tr>
<td>No. of office locations</td>
<td>Mean 2.6, SD 2.6</td>
<td>Mean 2.6, SD 2.6</td>
<td>.76</td>
</tr>
<tr>
<td></td>
<td>Mean 2.6, SD 2.6</td>
<td>Mean 2.5, SD 2.4</td>
<td></td>
</tr>
<tr>
<td>Practices with genetic counselor, %</td>
<td>Mean 36.5, SD 33.9</td>
<td>Mean 38.5, SD 33.9</td>
<td>.68</td>
</tr>
<tr>
<td></td>
<td>Mean 33.9, SD 38.5</td>
<td>Mean 38.6, SD 38.5</td>
<td></td>
</tr>
<tr>
<td>Census region, %</td>
<td>Mean 40.1, SD 42.1</td>
<td>Mean 40.8, SD 40.8</td>
<td>.94</td>
</tr>
<tr>
<td></td>
<td>Mean 42.1, SD 40.8</td>
<td>Mean 40.8, SD 40.8</td>
<td></td>
</tr>
<tr>
<td>Practice type, %</td>
<td>Mean 10.1, SD 9.4</td>
<td>Mean 13.1, SD 13.6</td>
<td>.67</td>
</tr>
<tr>
<td>Private with academic affiliation</td>
<td>Mean 55.0, SD 57.3</td>
<td>Mean 51.7, SD 51.7</td>
<td></td>
</tr>
<tr>
<td>Private independent</td>
<td>Mean 18.2, SD 19.3</td>
<td>Mean 17.9, SD 17.9</td>
<td></td>
</tr>
<tr>
<td>Employee</td>
<td>Mean 12.9, SD 11.5</td>
<td>Mean 13.1, SD 13.6</td>
<td></td>
</tr>
<tr>
<td>Academic full time</td>
<td>Mean 3.8, SD 2.6</td>
<td>Mean 4.1, SD 4.1</td>
<td></td>
</tr>
</tbody>
</table>

**Table 3. Patient Demographic and Clinical Characteristics by Cancer Type**

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Total</th>
<th>Breast Cancer</th>
<th>Colorectal Cancer</th>
<th>$P^*$</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at diagnosis, years</td>
<td>Mean 59.5, SD 61.9</td>
<td>Mean 61.9, SD 61.9</td>
<td>Mean 61.9, SD 61.9</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Disease stage</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Stage IV</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Stage III</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Stage II</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Stage I</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>Mean 49.7, SD 49.7</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Female sex</td>
<td>Mean 84.9, SD 84.9</td>
<td>Mean 84.9, SD 84.9</td>
<td>Mean 84.9, SD 84.9</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Contingency table</td>
<td>Mean 84.9, SD 84.9</td>
<td>Mean 84.9, SD 84.9</td>
<td>Mean 84.9, SD 84.9</td>
<td>&lt; .001</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>Family History Documented</th>
<th>Total (N = 10,466)</th>
<th>Breast Cancer (n = 6,569)</th>
<th>Colorectal Cancer (n = 3,897)</th>
<th>$P^*$</th>
</tr>
</thead>
<tbody>
<tr>
<td>First degree, %</td>
<td>Mean 79.8, SD 81.2</td>
<td>Mean 79.8, SD 81.2</td>
<td>Mean 79.8, SD 81.2</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Second degree, %</td>
<td>Mean 64.6, SD 68.9</td>
<td>Mean 64.6, SD 68.9</td>
<td>Mean 64.6, SD 68.9</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>Documented age at diagnosis, %</td>
<td>Mean 41.7, SD 45.1</td>
<td>Mean 41.7, SD 45.1</td>
<td>Mean 41.7, SD 45.1</td>
<td>&lt; .001</td>
</tr>
</tbody>
</table>

*Abbreviation: SD, standard deviation.

$^*$ $P$ 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\(^3\),\(^24\)\-\(^26\); 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/test.

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.\(^24\) 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.

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

<table>
<thead>
<tr>
<th>Referral</th>
<th>Total (N = 10,466)</th>
<th>Breast Cancer (n = 6,889)</th>
<th>Colorectal Cancer (n = 3,577)</th>
<th>P (^{*})</th>
</tr>
</thead>
<tbody>
<tr>
<td>Referral for genetic counseling and/or testing, %</td>
<td>25.6</td>
<td>29.1</td>
<td>19.6</td>
<td>&lt; .001</td>
</tr>
<tr>
<td>n = 2,457</td>
<td>n = 1,566</td>
<td>n = 901</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Positive family history and referred, %</td>
<td>42.7</td>
<td>52.2</td>
<td>26.4</td>
<td>&lt; .001</td>
</tr>
</tbody>
</table>

\(^*P\text{ value corresponds to differences between breast and colorectal cancers.}\)
AUTHORS’ DISCLOSURES OF POTENTIAL CONFLICTS OF INTEREST

Although all authors completed the disclosure declaration, the following author(s) and/or an author’s immediate family member(s) indicated a financial or other interest that is relevant to the subject matter under consideration in this article. Certain relationships marked with a “U” are those for which no compensation was received; those relationships marked with a “C” were compensated. For a detailed description of the disclosure categories, or for more information about ASCO’s conflict of interest policy, please refer to the Author Disclosure Declaration and the Disclosures of Potential Conflicts of Interest section in Information for Contributors.

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Data analysis and interpretation: Marie E. Wood, Trang H. Pham, Karen H. Lu, Michael N. Neuss, Kevin S. Hughes

Manuscript writing: All authors

Final approval of manuscript: All authors

REFERENCES


**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.