Utilization and Outcomes of BRCA Genetic Testing and Counseling in a National Commercially Insured Population

The ABOUT Study

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IMPORTANCE BRCA genetic testing has substantial public health impact, yet little is known of the real-world experiences of more than 100,000 Americans undergoing testing annually.

OBJECTIVE To identify factors associated with use of BRCA testing, assess whether delivery of genetic counseling and testing services adheres to professional guidelines, and measure the impact on patient-reported outcomes.

DESIGN, SETTING, AND PARTICIPANTS The American BRCA Outcomes and Utilization of Testing (ABOUT) Study analyzed data from a consecutive national series of 11,159 women whose clinicians ordered BRCA testing between December 2011 and December 2012. Aetna mailed recruitment information across the United States to commercial health plan members whose clinicians had ordered BRCA testing. A total of 3874 women (34.7%) completed questionnaires. Deidentified clinician-reported data from all respondents and a random sample of 2613 nonrespondents were also analyzed.

MAIN OUTCOMES AND MEASURES The proportion of eligible participants who met testing criteria and respondents’ report of receiving genetic counseling by a genetics clinician and its association with BRCA knowledge, understanding, and satisfaction were assessed.

RESULTS Among 3628 women respondents whose clinicians ordered comprehensive BRCA testing, most were white non-Hispanic (2502 [69.0%]), college educated (2953 [81.4%]), married (2751 [75.8%]), and had higher incomes (2011 [55.4%]). Approximately 16.4% (596) did not meet testing criteria. Mutations were identified in 161 (5.3%) of these women who received comprehensive testing. Only 1334 (36.8%) reported receiving genetic counseling from a genetics clinician prior to testing; the lowest rates (130 [12.3%]) were among patients of obstetrician/gynecologists. The most commonly reported reason for not receiving this clinical service was lack of clinician recommendation. Those who received it demonstrated greater knowledge about BRCA (mean score difference adjusted for demographics and clinician specialty, $\beta = 0.99$ [95% CI, 0.83-1.14]; $P<.001$) and expressed greater understanding ($\beta = 0.47$ [95% CI, 0.41-0.54]; $P<.001$) and satisfaction ($\beta = 2.21$ [95% CI, 1.60-2.81]; $P<.001$).

CONCLUSIONS AND RELEVANCE Despite improved patient knowledge, understanding, and satisfaction among patients who receive genetic counseling provided by a genetics clinician, as well as multiple guidelines emphasizing the importance of genetic counseling, most US women undergoing BRCA genetic testing do not receive this clinical service. Lack of physician recommendation is the most commonly reported reason. These findings demonstrate important gaps in clinical genetics services. Recently mandated coverage of genetic counseling services as a preventive service without patient cost sharing should contribute to improving clinical genetics services and associated outcomes in the future.

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Use and Outcomes of BRCA Testing and Counseling in Insured Women

### At a Glance

- **The goal of this study was to identify factors associated with use of BRCA testing and assess whether delivery of genetic counseling and testing services adheres to professional guidelines.**
- **The majority of commercially insured women undergoing BRCA testing did not have a personal history of breast or ovarian cancer (53.5%) and were white (51.8%) and non-Hispanic (94.5%).**
- **Most respondents (61.8%) did not receive pretest genetic counseling from a genetics clinician.**
- **Lack of physician recommendation was the most commonly reported reason for not receiving counseling from a genetics clinician.**
- **Respondents who received genetic counseling from a genetics clinician demonstrated greater knowledge about BRCA (P < .001) and understanding of the information received (P < .001) and expressed greater satisfaction (P < .001).**

### Methods

**Study Sample and Design**

An overview of the study design is provided in the **Figure**. The study was approved by the University of South Florida (USF) Institutional Review Board (IRB). Collaboration of the multidisciplinary investigative team in study design was described previously. Eligible participants included all 11,426 individuals for whom a BRCA test request was submitted to Aetna between December 2011 and December 2012; however, the following groups were excluded from analyses: men, eligible individuals who could not be contacted, and nonrespondents not selected via randomization.

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### Figure. Overview of American BRCA Outcomes and Utilization of Testing (ABOUT) Study Design in Aetna Member Population

<table>
<thead>
<tr>
<th>BRCA Test Request</th>
<th>11,426 Insured members for whom a BRCA test was requested and Aetna received a TRF completed by the ordering clinician</th>
</tr>
</thead>
<tbody>
<tr>
<td>Members sent institutional review board-approved recruitment materials by Aetna 3 weeks later</td>
<td>11,426 Members were sent institutional review board-approved recruitment materials by Aetna 3 weeks later</td>
</tr>
<tr>
<td>Excluded</td>
<td>267 Excluded</td>
</tr>
<tr>
<td>Returned address undeliverable</td>
<td>37 Returned address undeliverable</td>
</tr>
<tr>
<td>Male</td>
<td>230 Male</td>
</tr>
<tr>
<td>Women (34.7%) completed study questionnaire and returned to USF</td>
<td>3,874 Women (34.7%) completed study questionnaire and returned to USF</td>
</tr>
<tr>
<td>Women (65.3%) did not complete study questionnaire</td>
<td>7,285 Women (65.3%) did not complete study questionnaire</td>
</tr>
<tr>
<td>Aetna provided TRF data</td>
<td>3,874 Aetna provided TRF data</td>
</tr>
<tr>
<td>All female nonrespondents from weeks 1-12</td>
<td>833 All female nonrespondents from weeks 1-12</td>
</tr>
<tr>
<td>Randomly selected female nonrespondents from weeks 13-52</td>
<td>1,780 Randomly selected female nonrespondents from weeks 13-52</td>
</tr>
<tr>
<td>Respondents included in the statistical analysis</td>
<td>3,874 Respondents included in the statistical analysis</td>
</tr>
<tr>
<td>Nonrespondents included in the statistical analysis</td>
<td>2,613 Nonrespondents included in the statistical analysis</td>
</tr>
</tbody>
</table>

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Women with hereditary breast and ovarian cancer susceptibility (HBOC) face high lifetime risks: 24% to 86% for breast cancer and 16% to 67% for ovarian cancer. They often develop cancer at a young age or develop multiple cancers and share elevated risks with family members. Because of these high cancer risks, specific strategies for risk reduction, such as prophylactic mastectomy and oophorectomy, and for early detection through enhanced screening are recommended for women identified as carrying an inherited mutation associated with HBOC. Data show that these strategies can be effective in reducing cancer incidence and stage at diagnosis and, in the case of oophorectomy, can improve cancer-specific and overall survival, especially if offered prior to any cancer diagnosis.

In order to identify and inform women with HBOC, professional guidelines delineate clinical criteria based on personal and family history and recommend, for women who meet the criteria, consultation with a professionally trained, board-certified genetics clinician (GC) for genetic counseling. Genetic testing for HBOC focuses on analysis of 2 genes, BRCA1 and BRCA2. More than 100,000 individuals in the United States undergo BRCA testing annually, and volume is increasing rapidly.

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Questionnaire data were not shared with Aetna. TRF indicates test request form; USF, University of South Florida.

* Under institutional review board–approved waiver of informed consent, deidentified data from the TRF completed by the ordering health care provider, including ordering physician state and specialty, member’s year of birth, ancestry/ethnicity, and state; and test type and ordering criteria, were obtained for all nonrespondents during the first 12 weeks of accrual and from a randomly selected representative sample during weeks 13 through 52 of accrual (described in detail in the Methods section). The TRF data for respondents were not deidentified.

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Little is known of the experiences of Americans undergoing BRCA testing in community settings because previous research has focused almost exclusively on the small subset of individuals counseled and tested at academic medical centers. At such facilities, identification of individuals at risk for HBOC, service delivery, information transfer, decision making, and, quite likely, health and quality-of-life outcomes differ from those in community facilities, where the majority of individuals at high risk receive their health care.

In collaboration with the nation’s third-largest commercial health insurer (Aetna), we used a novel study design to investigate the characteristics and experiences of individuals undergoing BRCA genetic testing in the community setting. We also examined disparities in the use of genetic counseling and testing services based on key sociodemographic and ordering physician characteristics. To our knowledge, the American BRCA Outcomes and Utilization of Testing (ABOUT) study is the first research effort based on a national sample drawn from the community setting. We hypothesized that genetics services in the community setting are not being delivered according to published guidelines. Specifically, we predicted that most individuals do not receive genetic counseling from a GC, tests are often not ordered according to established criteria, and patient knowledge, understanding, and satisfaction are suboptimal.

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### At a Glance

- **The goal of this study was to identify factors associated with use of BRCA testing and assess whether delivery of genetic counseling and testing services adheres to professional guidelines.**
- **The majority of commercially insured women undergoing BRCA testing did not have a personal history of breast or ovarian cancer (53.5%) and were white (51.8%) and non-Hispanic (94.5%).**
- **Most respondents (61.8%) did not receive pretest genetic counseling from a genetics clinician.**
- **Lack of physician recommendation was the most commonly reported reason for not receiving counseling from a genetics clinician.**
- **Respondents who received genetic counseling from a genetics clinician demonstrated greater knowledge about BRCA (P < .001) and understanding of the information received (P < .001) and expressed greater satisfaction (P < .001).**

### Methods

**Study Sample and Design**

An overview of the study design is provided in the **Figure**. The study was approved by the University of South Florida (USF) Institutional Review Board (IRB). Collaboration of the multidisciplinary investigative team in study design was described previously. Eligible participants included all 11,426 individuals for whom a BRCA test request was submitted to Aetna between December 2011 and December 2012; however, the following groups were excluded from analyses: men, eligible individuals who could not be contacted, and nonrespondents not selected via randomization.
Recruitment
Aetna’s commercial health plans cover genetic counseling services by a GC, whether in person or by telephone, as well as genetic testing for members who meet Aetna’s evidence-based medical policy criteria. Aetna is notified of a member’s request for BRCA testing when the blood sample arrives at the laboratory along with a standard test request form (TRF) completed by the ordering clinician. The TRF specifies the test being ordered and includes the patient’s contact information, age, ancestry/ethnicity, and personal and family cancer history. This information is reviewed by Aetna to determine approval/authorization for coverage of testing. Some participants for whom BRCA testing was ordered by their clinician and their sample submitted to the laboratory did not meet testing criteria and, thus, did not receive results; however, they were still eligible for the study.

When Aetna received notification of a member’s BRCA test request, they assigned a unique eligible participant code. Identifying information about eligible participants was not shared with study investigators outside Aetna unless the individual provided informed consent. Aetna mailed IRB-approved study materials to each eligible participant, along with a prepaid reply envelope addressed to the USF researchers. In addition to permission to use data from the participant’s completed questionnaire, TRF, and related documents from Aetna, informed consent also included permission to recontact the participant and optional permissions to (1) obtain BRCA test results from the testing laboratory, (2) obtain relevant medical records, and claims data from Aetna, and (3) use data in future IRB-approved research.

Materials were mailed 3 weeks after the sample arrived at the laboratory to ensure receipt by each eligible participant within 3 weeks after receiving test results for those who did receive results. A reminder postcard was mailed 3 weeks later. A toll-free telephone number was provided to permit contacting study staff for assistance. The packet included a $5 bill as a prepaid token of appreciation. In addition to permission to use data from the participant’s completed questionnaire, TRF, and related documents from Aetna, informed consent also included permission to recontact the participant and optional permissions to (1) obtain BRCA test results from the testing laboratory, (2) obtain relevant medical records and claims data from Aetna, and (3) use data in future IRB-approved research.

Data Collection
Deidentified Existing Data on Eligible Participants
To characterize the national population of individuals whose clinicians are requesting BRCA testing through commercial health insurance, Aetna provided deidentified data from TRFs received for test preauthorization from December 2011 to December 2012. The deidentified data included ordering physician state and specialty, as well as member information that included year of birth, state, ancestry/ethnicity, ordering criteria (ie, relevant personal and/or family cancer history), and test type. Test type is defined as comprehensive if the participant is tested by full sequencing and analysis for common re-arrangements in BRCA1 and BRCA2 or by testing for 3 common mutations found in the Ashkenazi Jewish population (appropriate for individuals of Ashkenazi Jewish ancestry)18-21; test type is defined as family mutation if the participant is tested for a specific mutation previously identified in the family. Additional deidentified data included whether the eligible participant met testing criteria. The TRF collects ancestry/ethnicity data using the following categories: Western/Northern Europe, Central/Eastern Europe, Africa, Near East/Middle East, Ashkenazi, Latin American/Caribbean, Asia, Native American, and other. The questionnaire used the Office of Management and Budget Standards for Federal Statistics and Administrative Reporting race categories: American Indian or Alaska Native, Asian, black, Native Hawaiian or other Pacific Islander, and white, as well as Hispanic and Ashkenazi Jewish ethnicities. In the analyses, these same race and ethnicity categories were used for nonrespondents based on the clinician-reported TRF ancestry/ethnicity as follows: Western/Northern Europe, Central/Eastern Europe, Near East/Middle East: white; Africa: black; Asia: Asian; Native American: American Indian or Alaska Native; Ashkenazi: Ashkenazi Jewish; and Latin American/Caribbean: Hispanic.

Manual redaction of identifiable data from the handwritten TRFs completed by the ordering clinicians was labor intensive, and it was not feasible for Aetna to perform this for all 7285 nonrespondents. In order to develop an appropriate random sampling schema for analysis of nonrespondent data, Aetna first redacted all nonresponder TRF data for recruitment weeks 1 through 12 (response rate, 961 of 2622 [36.7%]), and we used the data to inform our sampling strategy. An additional 1780 nonrespondents were randomly selected during the remaining 40 weeks of accrual using SPSS and a randomization schema to ensure representation across age, race/ethnicity, and week of recruitment, for a total of 2613 nonrespondents in the analyses. A separate analysis showed no characteristic differences between nonrespondents from weeks 1 through 12 and those randomly selected from weeks 13 through 52.

Data From Respondents
The study questionnaire assessed sociodemographic factors, personal and family cancer history, and whether the individual received genetic counseling by a GC, as well as genetic testing results. Participants were asked what types of information they received prior to testing and the reason(s) if they did not receive genetic counseling by a GC.

We measured BRCA1 and BRCA2 knowledge with the commonly used 11-item knowledge scale developed by the National Center for Human Genome Research Cancer Genetics Studies Consortium. Total score was the number of correct responses.

Understanding of the information received prior to testing was measured using a 5-point Likert scale and summed. Higher scores indicate greater patient-reported understanding.

Satisfaction with information received prior to testing and the encounter was measured across 8 domains using a 4-point Likert scale and summed. Higher scores indicate greater satisfaction.
For eligible participants who completed the study questionnaire and provided informed consent for release of identifiable data (ie, respondents), Aetna provided TRFs without redaction. The testing laboratory provided test results for participants who provided informed consent. No identifiable questionnaire data or genetic test results were shared with Aetna.

Statistical Analyses
We used TRF data to characterize the eligible participant population and compare demographic characteristics and personal and family cancer history between participants who completed the questionnaire (respondents) and those who did not complete the questionnaire (nonrespondents). Prior to conducting these analyses, we compared the demographic data that respondents provided to us in their questionnaire with the demographic data that their clinician had provided on their TRF; our results indicated that the TRF data were 97% correlated with patient-reported data; thus, TRF data were used to compare characteristics of respondents to nonrespondents. We used χ² tests to assess whether the distribution of each characteristic differed between respondents and nonrespondents. Multiple logistic regression analysis identified predictors of response and questionnaire completion after adjusting for study week.

We performed multiple logistic regression analyses to examine associations between participants who met criteria for comprehensive testing and those who did not (ie, were not tested) with regard to (1) demographic factors, personal history of breast or ovarian cancer, and pretest genetic counseling by a GC, and (2) the association between ordering clinician specialty and receiving pretest genetic counseling by a GC.

In separate multiple linear regression models, we examined the association of pretest genetic counseling by a GC with each study outcome after adjusting for demographic factors, personal breast and ovarian cancer history, and ordering clinician specialty. We excluded income from final multivariate models because most of the effect was explained by education. P < .05 was considered statistically significant. Statistical analyses were performed using SPSS, version 222013, and SAS, version 9.32011.

As shown in the Figure, IRB-approved recruitment materials were delivered to 11 159 women. Demographic information was available for all respondents (3874 of 11 159 [34.7%]) and a sample of 2613 of the 7285 nonrespondents. To summarize demographic information for the entire 11 159 population, we weighted statistics (medians, interquartile ranges, and percentages) based on the total percentage of respondents (weight = 0.35) and nonrespondents (weight = 0.65).

Results
Over the 1-year accrual period, BRCA testing was requested for 11 426 individuals through Aetna commercial health plans, including 11 159 women to whom study packets could be delivered. The median (interquartile range) age among these 11 159 women was 48.4 (42.4-57.1) years. The majority (estimated 53.3%) had no personal history of breast or ovarian cancer; an estimated 43.3% had a personal history of breast cancer; 2.9% had a personal history of ovarian cancer, and 0.5% had a personal history of breast and ovarian cancer.

In a multivariate logistic regression analysis that included week of accrual and all variables in Table 1, eligible women were more likely to complete the questionnaire if they were older than 45 years (odds ratio [OR], 1.17 [95% CI, 1.05-1.30]; P = .005); had a personal history of breast or ovarian cancer (OR, 1.36 [95% CI, 1.22-1.51]; P < .001), or were of Ashkenazi ethnicity (OR, 1.22 [95% CI, 1.01-1.47]; P = .04) or white race (OR, 1.26 [95% CI, 1.12-1.41]; P < .001). Individuals of Asian (OR, 0.53 [95% CI, 0.41-0.68]; P < .001), black (OR, 0.66 [95% CI, 0.53-0.83]; P < .001), and Hispanic (OR, 0.80 [95% CI, 0.64-1.00]; P = .049) race/ethnicity were less likely to respond.

Table 2 shows test results and characteristics of respondents who underwent testing and those who did not meet criteria for comprehensive testing (ie, did not receive results). A total of 596 (16.4%) of respondents did not meet evidence-based criteria for comprehensive testing and were not tested. Among those individuals who did not meet criteria, 483 (81.0%) had not met with a GC prior to the test being ordered; overall, only 1481 (38.2%) of all female respondents reported receiving genetic counseling from a GC prior to testing. The majority of individuals who met criteria for comprehensive testing had a personal history of breast or ovarian cancer, in contrast to a majority of unaffected individuals among those who did not meet testing criteria. There were no differences in meeting testing criteria based on education, income, race, or ethnicity. Rates of ineligibility were higher among individuals younger than 40 years (mostly unaffected) compared with older age groups in which the proportion of individuals with cancer was higher.

As expected, most of the relatively small group of individuals who underwent testing for a family mutation (n = 246) did not have a personal history of cancer and saw a GC prior to testing. Because a mutation had been previously identified in the family, this group had an expected high rate of positive results (41.8%).

Table 3 presents characteristics of individuals who reported receiving genetic counseling by a GC prior to comprehensive or Ashkenazi Jewish panel testing being requested, compared with those who did not receive genetic counseling prior to testing. Individuals undergoing testing for a family mutation were excluded.

Individuals who reported receiving pretest genetic counseling by a GC were more than twice as likely to meet testing criteria. The likelihood of receiving genetic counseling by a GC prior to testing was higher among individuals with some college education but did not vary by race or ethnicity, with the exception that Jewish individuals were more likely to receive it. Individuals with a personal history of breast or ovarian cancer were nearly twice as likely to have received genetic counseling by a GC compared with unaffected individuals. This is consistent with the finding that receiving genetic counseling varied substantially by the specialty of the clinician ordering the test, with low rates among obstetrician/gynecologists compared with all other clinicians and relatively higher rates among oncologists and internal medicine specialists. Among indi-
Individuals who reported not receiving genetic counseling by a GC, the most common reason was “doctor or health professional did not recommend it.”

Table 4 displays outcomes among individuals who reported receiving pretest genetic counseling from a GC vs those who did not. Only individuals whose clinician had requested comprehensive or Ashkenazi Jewish panel testing were included (individuals tested for a family mutation were excluded).

Individuals who received genetic counseling from a GC demonstrated higher BRCA testing knowledge scores and expressed greater self-reported understanding of the information they received prior to testing. Those who reported receiving genetic counseling from a GC were also more satisfied than those who did not across all measured parameters, including whether their GC or other clinician “explained things clearly,” “listened to what I had to say,” “used language I understood,” “provided new information,” “really understood my concerns,” “cares for me,” “lessened my worries,” and “helped me cope better.”

Discussion
To our knowledge, the present study provides the first characterization of a national sample of commercially insured individuals whose clinician has ordered genetic testing for HBOC, as well as their pretest and posttest experiences. Female Aetna members whose clinicians requested BRCA testing were mainly white non-Hispanic, college-educated, married or partnered, and had higher household incomes. Compared with black and Hispanic women, a greater proportion of white non-Hispanic women also received genetic counseling from a GC prior to genetic testing, which suggests that there are racial and ethnic differences in genetic testing and counseling experiences.
Table 2. Characteristics of Respondents Who Met Aetna’s Criteria for Testing (Tested) and Those Who Did Not (Not Tested)

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Family Mutation(^a)</th>
<th>Requested Comprehensive Testing(^b)</th>
<th>% Tested (n = 2032)</th>
<th>Not Tested (n = 596)</th>
<th>Odds Ratio (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Total Tested, No. (n = 246)</td>
<td>Total Requesting, No. (n = 3628)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Personal history of breast or ovarian cancer</td>
<td>No</td>
<td>221</td>
<td>1601</td>
<td>73.0</td>
<td>27.0</td>
</tr>
<tr>
<td></td>
<td>Yes</td>
<td>25</td>
<td>2015</td>
<td>92.1</td>
<td>7.9</td>
</tr>
<tr>
<td>Saw genetics clinician prior to testing</td>
<td>No</td>
<td>95</td>
<td>2247</td>
<td>78.5</td>
<td>21.5</td>
</tr>
<tr>
<td></td>
<td>Yes</td>
<td>147</td>
<td>1334</td>
<td>92.4</td>
<td>7.6</td>
</tr>
<tr>
<td>Education</td>
<td>No</td>
<td>33</td>
<td>583</td>
<td>84.0</td>
<td>16.0</td>
</tr>
<tr>
<td></td>
<td>Some college</td>
<td>208</td>
<td>2953</td>
<td>83.6</td>
<td>16.4</td>
</tr>
<tr>
<td>Yearly income, $</td>
<td>≤35 000</td>
<td>28</td>
<td>325</td>
<td>83.4</td>
<td>16.6</td>
</tr>
<tr>
<td></td>
<td>35 001-50 000</td>
<td>21</td>
<td>389</td>
<td>83.0</td>
<td>17.0</td>
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<tr>
<td></td>
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<td>42</td>
<td>569</td>
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<td></td>
<td>&gt;75 000</td>
<td>127</td>
<td>2011</td>
<td>84.0</td>
<td>16.0</td>
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<tr>
<td>Race/ethnicity(^e)</td>
<td>Ashkenazi Jewish</td>
<td>21</td>
<td>355</td>
<td>95.2</td>
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<tr>
<td></td>
<td>Asian</td>
<td>6</td>
<td>105</td>
<td>91.4</td>
<td>8.6</td>
</tr>
<tr>
<td></td>
<td>Black</td>
<td>3</td>
<td>255</td>
<td>81.2</td>
<td>18.8</td>
</tr>
<tr>
<td></td>
<td>Native American(^f)</td>
<td>2</td>
<td>56</td>
<td>87.5</td>
<td>12.5</td>
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<tr>
<td></td>
<td>White Hispanic</td>
<td>7</td>
<td>146</td>
<td>80.0</td>
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<td>White non-Hispanic</td>
<td>196</td>
<td>2502</td>
<td>82.1</td>
<td>17.9</td>
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<tr>
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<td>Other/not sure</td>
<td>3</td>
<td>101</td>
<td>80.2</td>
<td>19.8</td>
</tr>
<tr>
<td>Ordering clinician specialty</td>
<td>Family practice</td>
<td>8</td>
<td>66</td>
<td>83.8</td>
<td>16.7</td>
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<td>7</td>
<td>152</td>
<td>88.8</td>
<td>11.2</td>
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<td>1075</td>
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<td>62</td>
<td>868</td>
<td>93.0</td>
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<td>87</td>
<td>77.0</td>
<td>23.0</td>
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<td>26</td>
<td>582</td>
<td>91.9</td>
<td>8.1</td>
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<td>197</td>
<td>96.4</td>
<td>3.6</td>
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<tr>
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<td>Unknown</td>
<td>24</td>
<td>119</td>
<td>83.2</td>
<td>16.8</td>
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<tr>
<td></td>
<td>Missing</td>
<td>10</td>
<td>482</td>
<td>75.9</td>
<td>24.1</td>
</tr>
<tr>
<td>Marital status</td>
<td>Married or living together</td>
<td>177</td>
<td>2751</td>
<td>83.9</td>
<td>16.1</td>
</tr>
<tr>
<td></td>
<td>Separated/divorced/widowed</td>
<td>25</td>
<td>483</td>
<td>82.0</td>
<td>18.0</td>
</tr>
<tr>
<td></td>
<td>Single, never married</td>
<td>37</td>
<td>299</td>
<td>83.6</td>
<td>16.4</td>
</tr>
<tr>
<td>Age of participant, y</td>
<td>&lt;45</td>
<td>141</td>
<td>1205</td>
<td>82.7</td>
<td>17.3</td>
</tr>
<tr>
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<td>45-59</td>
<td>72</td>
<td>1736</td>
<td>85.1</td>
<td>14.9</td>
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<tr>
<td></td>
<td>≥60</td>
<td>23</td>
<td>567</td>
<td>81.3</td>
<td>18.7</td>
</tr>
<tr>
<td>Results</td>
<td>Positive</td>
<td>100</td>
<td>161</td>
<td>100</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Negative</td>
<td>137</td>
<td>2600</td>
<td>100</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Variant of uncertain significance</td>
<td>2</td>
<td>65</td>
<td>100</td>
<td>N/A</td>
</tr>
<tr>
<td></td>
<td>Not sure</td>
<td>0</td>
<td>33</td>
<td>100</td>
<td>N/A</td>
</tr>
</tbody>
</table>

Abbreviation: N/A, not applicable.

\(^a\) All individuals with a family mutation meet criteria for testing.

\(^b\) Comparison of individuals who did not meet criteria for comprehensive testing and were not tested vs individuals who met criteria for comprehensive testing and were tested. Includes Ashkenazi Jewish individuals undergoing Jewish panel testing.

\(^c\) Number of respondents providing this answer to the item; does not include respondents who did not answer the item.

\(^d\) Multivariate model includes all applicable covariates in the table: personal history of breast or ovarian cancer, saw genetics clinician prior to testing, education, yearly income, race/ethnicity, marital status, age, results.

\(^e\) Four participants whose race was Native Hawaiian or other Pacific Islander were not included. All 4 had comprehensive testing.

\(^f\) Includes Native American or Alaska Native.
disparities in access to genetic counseling, which is consistent with previous research.24

In contrast to previous reports from academic medical center settings, most individuals did not receive pretest genetic counseling from a GC. This low rate of pretest genetic counseling is particularly surprising given clear professional society guidelines and published research that consistently document its importance in informed decision making and facilitating better patient outcomes. The specialty of the ordering clinician was the primary factor associated with whether an individual received genetic counseling services from a GC.

As in previous studies, lack of physician recommendation was the most common reason reported for low rates of genetic counseling by a GC. Our findings that GC referrals are low among obstetrician/gynecologists is particularly concerning, especially given the high rate of tests being ordered by this clinician group for individuals with no personal cancer history and who do not meet criteria for testing. The most appropriate individual to test first in any family is an individual who has had breast, ovarian, or another related cancer because the results are more likely to be informative. Following recent aggressive marketing by testing laboratories,25 there has been rapid growth in tests ordered for unaffected individuals. In response, national guidelines panels have added specific guidance that testing unaffected individuals should be performed only when there is no living affected family member available for testing.14

Previous studies have consistently demonstrated that genetic counseling by a GC is associated with improved patient knowledge, understanding, and satisfaction.13,26 Better outcomes when genetic counseling is performed by a GC compared with nongenetics physicians are not unexpected, given that nongenetics physicians often self-report little or no genetics training and low competence in genetics.27-29 Compounding factors include increasing demands on physician time, shorter encounters with patients, and limited mechanisms for clinician training in genetics.30,31 These trends are concerning, especially given the rapidly increasing complexity of genetic science and tests.32

Recent trends in health care policy may begin to increase the number of individuals who are able to access and receive pretest genetic counseling services by a GC. Under the Pa-
Despite the study’s unique national and community-based scope, there were some limitations. We were unable to survey clinicians directly to obtain additional information about the potential underlying causes for the suboptimal genetic counseling referral patterns and relatively high rate of inappropriate test requests. The response rate was less than ideal but was tempered by our ability to address potential participation bias by analyzing critical data on the national ascertainment pool of individuals whose clinician had ordered a BRCA test. Our findings showed statistically significant differences in demographic characteristics between respondents and nonrespondents. Thus, our findings that are based on respondents should be interpreted with caution because they may be less representative of nonrespondents. However, because the findings also indicate that nonrespondents were less likely to meet testing criteria, the differences in outcomes that we report between individuals who were or were not tested on the basis of whether they met testing criteria are likely to be conservative. Lower response rates among certain racial and ethnic groups, including blacks and Hispanics, were also a limitation, a challenge that has been noted in research overall.

### Conclusions

Evaluation of a national consecutive series of commercially insured individuals whose clinicians recently ordered BRCA testing provides a unique snapshot of genetic counseling and testing services in communities across the United States. It reveals overall low rates of referral to GCs for genetic counseling as a critical issue affecting outcomes. Referral patterns...
also vary widely across physician specialties. The lowest referral rates to GCs were noted among patients of obstetrician/gynecologists, with high rates of tests ordered for unaffected individuals who do not meet criteria for testing based on national guidelines. Similar to previous studies, patients who reported receiving genetic counseling by a GC were more likely to meet criteria for testing and to demonstrate greater knowledge, understanding, and satisfaction.

REFERENCES


