

## Concerns About Cancer Risk and Experiences With Genetic Testing in a Diverse Population of Patients With Breast Cancer

Reshma Jagsi, Kent A. Griffith, Allison W. Kurian, Monica Morrow, Ann S. Hamilton, John J. Graff, Steven J. Katz, and Sarah T. Hawley

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### ABSTRACT

#### Purpose

To evaluate preferences for and experiences with genetic testing in a diverse cohort of patients with breast cancer identified through population-based registries, with attention to differences by race/ethnicity.

#### Methods

We surveyed women diagnosed with nonmetastatic breast cancer from 2005 to 2007, as reported to the SEER registries of metropolitan Los Angeles and Detroit, about experiences with hereditary risk evaluation. Multivariable models evaluated correlates of a strong desire for genetic testing, unmet need for discussion with a health care professional, and receipt of testing.

#### Results

Among 1,536 patients who completed the survey, 35% expressed strong desire for genetic testing, 28% reported discussing testing with a health care professional, and 19% reported test receipt. Strong desire for testing was more common in younger women, Latinas, and those with family history. Minority patients were significantly more likely to have unmet need for discussion (failure to discuss genetic testing with a health professional when they had a strong desire for testing): odds ratios of 1.68, 2.44, and 7.39 for blacks, English-speaking Latinas, and Spanish-speaking Latinas compared with whites, respectively. Worry in the long-term survivorship period was higher among those with unmet need for discussion (48.7% v 24.9%;  $P < .001$ ). Patients who received genetic testing were younger, less likely to be black, and more likely to have a family cancer history.

#### Conclusion

Many patients, especially minorities, express a strong desire for genetic testing and may benefit from discussion to clarify risks. Clinicians should discuss genetic risk even with patients they perceive to be at low risk, as this may reduce worry.

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### INTRODUCTION

A diagnosis of breast cancer triggers a cascade of increasingly complicated decisions about treatment options. An important consideration for some patients making decisions about the initial course of locoregional treatment is the potential risk of a second primary cancer. Approximately 5% to 10% of patients with breast cancer have **germline mutations** that predispose them to developing additional cancers,<sup>1</sup> and this risk may extend to other relatives who carry the same mutation. *BRCA1/2* mutations have been identified across all racial/ethnic subgroups.<sup>2</sup>

Discovery of a genetic mutation has important implications for a patient's treatment decision making in the context of a new breast cancer diag-

nosis.<sup>3</sup> Patients with high inherited risk of new primary cancers may be more inclined toward mastectomy and contralateral risk-reducing mastectomy; those with *BRCA1/2* mutations may also consider risk-reducing salpingo-oophorectomy; those with Li-Fraumeni syndrome should avoid approaches that incorporate radiotherapy.<sup>4</sup> Implications of genetic mutations also extend to the survivorship period, when high-risk women who did not choose bilateral mastectomy may benefit from additional measures, such as MRI surveillance. Identification of a familial mutation also affects relatives, who may or may not wish to know this information, test for the mutation themselves, and consider risk-reducing interventions.<sup>5</sup>

The rapidly expanding scope and availability of genetic testing for cancer risk<sup>6</sup> motivates research to

Reshma Jagsi, Center for Bioethics and Social Science in Medicine, University of Michigan; Kent A. Griffith, Center for Biostatistics, School of Public Health, University of Michigan; Steven J. Katz, University of Michigan; Sarah T. Hawley, Ann Arbor VA Health Care System, University of Michigan, Ann Arbor, MI; Allison W. Kurian, Stanford University School of Medicine, Stanford; Ann S. Hamilton, Keck School of Medicine, University of Southern California, Los Angeles, CA; Monica Morrow, Memorial Sloan-Kettering Cancer Center, New York, NY; and John J. Graff, Rutgers University, New Brunswick, NJ.

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S.J.K. and S.T.H. contributed equally to this work.

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Corresponding author: Reshma Jagsi, MD, DPhil, Department of Radiation Oncology, University of Michigan, UHB2C490, SPC 5010, 1500 East Medical Center Drive, Ann Arbor, MI 48109-5010; e-mail: [rjagsi@med.umich.edu](mailto:rjagsi@med.umich.edu).

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examine patients' perspectives and experiences with testing. Most studies to date have focused on general or high-risk populations and have raised concerns about racial/ethnic disparities in knowledge and access.<sup>7</sup> The few studies that included cancer patients have been limited by smaller convenience samples, low participation rates, and few racial/ethnic minority patients. These studies have yielded conflicting results regarding whether patients with a personal history of cancer are more likely than others to desire or receive testing. In a 2002 survey of women with early-onset breast cancer who were identified through two breast cancer support groups, among the 21% completing questionnaires, 83% were aware of *BRCA* testing, and 12.5% had received testing.<sup>8</sup> More generalizable research is needed to understand which women diagnosed with breast cancer in the community desire and receive genetic testing.

Given gaps in knowledge about desire for and receipt of testing in patients with breast cancer, particularly racial/ethnic minorities and those not at high risk, we considered a diverse cohort of breast cancer survivors identified through population-based registries to examine patients' self-reported desire to receive genetic testing, whether they had unmet need for discussion (failed to discuss genetic testing with a health professional when they had a strong desire for testing), and whether they had received testing. We described reasons for test receipt and nonreceipt and explored associations between unmet need for discussion and worry about breast cancer during survivorship. Finally, we evaluated the correlates of desire for genetic testing, unmet need for discussion, and test receipt.

## METHODS

### Study Sample

Our study sample originated from a longitudinal cohort study of women diagnosed with breast cancer in metropolitan Los Angeles and Detroit. We included patients age 20 to 79 years who were diagnosed with stage 0-III breast cancer between June 2005 and February 2007, as reported to the National Cancer Institute's *Surveillance, Epidemiology, and End Results (SEER)* population-based program registries in those regions.

We excluded patients with stage IV breast cancer and those who could not complete a questionnaire in English or Spanish. Asian women in Los Angeles were excluded because of enrollment in other studies. Latina (in Los Angeles) and black (in both Los Angeles and Detroit) patients were oversampled to ensure sufficient minority representation.

### Questionnaire Design and Content

Questionnaires (Data Supplement) were based on existing literature, measures previously developed to assess relevant constructs, and theoretical models. We utilized standard techniques of content validation,<sup>9</sup> including systematic review by design experts<sup>10-12</sup> and pretesting with 40 patients in three waves, including 12 detailed cognitive interviews.<sup>13,14</sup>

### Data Collection

After IRB approval, patients were identified via rapid case ascertainment and surveyed a mean of 9 months after diagnosis (mean, 288 days; SD, 100), and again approximately four years later (mean, 1,524 days; SD, 143). To encourage response, we provided a \$10 cash incentive and used a modified Dillman method.<sup>15</sup> All materials were sent in English and Spanish to those with Spanish surnames.<sup>16</sup> The response rate to the baseline survey was 73%, and the response rate to the follow-up survey was 68%. Survey responses were combined into a single data set, into which clinical data from SEER were merged. More details regarding the flow of patients into the sample are provided in Appendix Figure A1 (online only).

### Measures

SEER records provided clinical stage. In the baseline survey, we measured age, race/ethnicity (white, black, English-speaking Latina, and Spanish-speaking Latina), education ( $\leq$  high school *v* at least some college), insurance status (none, private, Medicare, or Medicaid), and family history of breast and/or ovarian cancer (in none *v*  $\geq$  1 first-degree relatives). We measured worry about implications for family in the baseline survey, by asking how true it was that the respondent had "worry that other members of my family might someday get the same illness I have" in the past 7 days.

We introduced the concept of genetic testing for cancer risk in the follow-up survey by describing tests that "look for gene mutations or changes, to see if women and their families have a greater risk of developing breast cancer in the future." We evaluated desire for testing by asking "How much did you want to have a genetic test for breast cancer risk?" We dichotomized responses for analysis (defining a "strong desire" as responses of "quite a bit" or "very much" rather than "somewhat," "a little bit," or "not at all"). We evaluated whether the patient had discussed testing with a health care professional through an item that inquired: "Did a genetic counselor, doctor, or other health professional talk with you about having a genetic test for breast cancer risk?" We further defined "unmet need for discussion" by using responses from these two items to define a subset of patients who expressed strong desire for testing but denied discussion with a health care professional. Finally, we evaluated test receipt with an item that inquired: "Have you ever had a genetic test for breast cancer risk?"

Patients who indicated they did receive the test were asked to check all that applied from among a list of reasons for desiring testing ("my doctor thought I should get tested," "I wanted more information about my own health," "I wanted more information for my family members," "my family wanted me to be tested," and "other"). Those who indicated they did not receive testing (from among "my doctor didn't recommend it," "I didn't want it," "my family didn't want me to get it," "it was too expensive," and "other").

We measured worry about breast cancer in the follow-up survey by asking how much the respondent worried about breast cancer coming back in the same breast, occurring in the other breast, or spreading to other parts of her body. Responses to the worry items were dichotomized for analysis as "very much" or "quite a bit" versus "somewhat," "a little bit," or "not at all."

### Analytic Approach

To allow statistical inferences to represent the original targeted population, we applied complex survey weights to the calculation of percentages and regression analyses. Design weights compensated for the oversampling of minorities and disproportionate selection across SEER sites; nonresponse weights compensated for the fact that women with certain characteristics were not as likely to respond to the surveys at each time point (Data Supplement). Analyses were conducted using SAS 9-2 (Cary, NC).

We first generated descriptive statistics for the sample, including rates of strong desire for testing, discussion of testing with a health care professional, and receipt of testing. We then evaluated correlates of expressing a strong desire for genetic testing, unmet need for discussion (a strong desire for testing but without a discussion with a health care professional), and genetic testing receipt. In each set of analyses, we first considered the following independent variables on bivariable analyses: age, race/ethnicity (white, black, English-speaking Latina, and Spanish-speaking Latina), disease type (in situ *v* invasive), education (high school or less *v* at least some college), insurance status (none, private, or governmental—Medicaid/Medicare), and family history of breast and/or ovarian cancer (in none *v* one or more first-degree relatives). Best multiple variable models were constructed using a backward elimination strategy, with all covariates first offered to the model, and covariates iteratively removed and the model recalculated, after consideration of the covariate's significance, its possible effect modification on the remaining covariates, and its impact on the overall model fit as computed by Akaike's information criterion for nested models. We also described concerns about impact on family, reasons for receipt and nonreceipt of testing, and evaluated associations between unmet need for discussion and worry about breast cancer during survivorship. Frequencies

between groups were compared statistically using the Rao-Scott  $\chi^2$  test. For all statistic tests,  $P$  values  $\leq .05$  were considered significant.

## RESULTS

Of 3,133 women surveyed, 2,290 (73%) completed the baseline survey. Of these, 1,536 (68%) completed the follow-up survey and constituted the analytic sample. Table 1 reports the characteristics of these 1,536 patients.

**Table 1.** Patient Demographic and Clinical Characteristics

| Characteristic  | No.  | %    | Weighted Mean or Weighted % |
|---|------|------|-----------------------------|
| <b>SEER site</b>  |      |      |                             |
| Detroit   | 694  | 45.2 |                             |
| Los Angeles   | 842  | 54.8 |                             |
| <b>Age at time of baseline survey, years</b>  |      |      |                             |
| Mean  | 57.5 |      | 57.2                        |
| SD  | 11.2 |      |                             |
| <b>Race/ethnicity</b>   |      |      |                             |
| White   | 728  | 47.4 | 41.8                        |
| Black   | 380  | 24.7 | 17.1                        |
| Latina, English speaking  | 191  | 12.4 | 19.2                        |
| Latina, Spanish speaking  | 203  | 13.2 | 20.0                        |
| Other   | 34   | 2.2  | 1.8                         |
| <b>Education</b>  |      |      |                             |
| High school or less   | 564  | 36.7 | 41.7                        |
| At least some college   | 945  | 61.5 | 56.1                        |
| Missing/unknown   | 27   | 1.8  | 2.2                         |
| <b>Insurance status</b>   |      |      |                             |
| None  | 104  | 6.8  | 9.2                         |
| Private   | 1054 | 68.6 | 63.6                        |
| Medicaid  | 120  | 7.8  | 9.6                         |
| Medicare  | 202  | 13.2 | 13.5                        |
| Missing/unknown   | 56   | 3.6  | 4.1                         |
| <b>Disease stage</b>  |      |      |                             |
| DCIS  | 380  | 24.7 | 18.1                        |
| 1   | 553  | 36.0 | 34.3                        |
| 2   | 425  | 27.7 | 32.7                        |
| 3   | 141  | 9.2  | 11.7                        |
| Missing/unknown   | 37   | 2.4  | 3.2                         |
| <b>Family history of breast and/or ovarian cancer</b>   |      |      |                             |
| None  | 964  | 62.8 | 62.6                        |
| $\geq$ One first-degree relative  | 504  | 32.8 | 32.4                        |
| Missing/unknown   | 68   | 4.4  | 5.0                         |
| <b>Strongly wanted testing</b>  |      |      |                             |
| Yes   | 493  | 32.1 | 35.3                        |
| No  | 966  | 62.9 | 60.1                        |
| Missing/unknown   | 77   | 5.0  | 4.6                         |
| <b>Discussed genetic testing with a genetic counselor, physician, or other health professional?</b> |      |      |                             |
| Yes   | 432  | 28.1 | 27.9                        |
| No  | 957  | 62.3 | 62.5                        |
| I don't know  | 136  | 8.9  | 8.8                         |
| Missing/unknown   | 11   | 0.7  | 0.9                         |
| <b>Received genetic testing</b>   |      |      |                             |
| Yes   | 269  | 17.5 | 18.6                        |
| No  | 1118 | 72.8 | 71.9                        |
| I don't know  | 105  | 6.8  | 7.6                         |
| Missing/unknown   | 44   | 2.9  | 2.0                         |

Abbreviations: DCIS, ductal carcinoma in situ; SD, standard deviation.

The sample was diverse, with 17% black and 39% Latina. A total of 42% had a high school education or less, 52% had stage 0-I disease, and 32% reported a family history of breast or ovarian cancer in a first degree relative. About a third of patients (35%) expressed a strong desire for genetic testing; 28% reported discussing testing with a health care professional; and 19% reported receipt of genetic testing. Figure 1 depicts variations by race/ethnicity in these responses.

Table 2 presents a multivariable model of the correlates of a strong desire for genetic testing. Strong desire for genetic testing was more common in younger women, Spanish-speaking Latinas, and those with a family cancer history. Of note, the strong desire for testing among Latinas was highly consistent with racial/ethnic differences in the expression of worry about implications for family members: 31.1% of blacks, 38.4% of whites, 56.7% of English-speaking Latinas, and 83.1% of Spanish-speaking Latinas ( $P < .001$ ) reported that they were quite a bit or very worried that other members of the family might get breast cancer in the future.

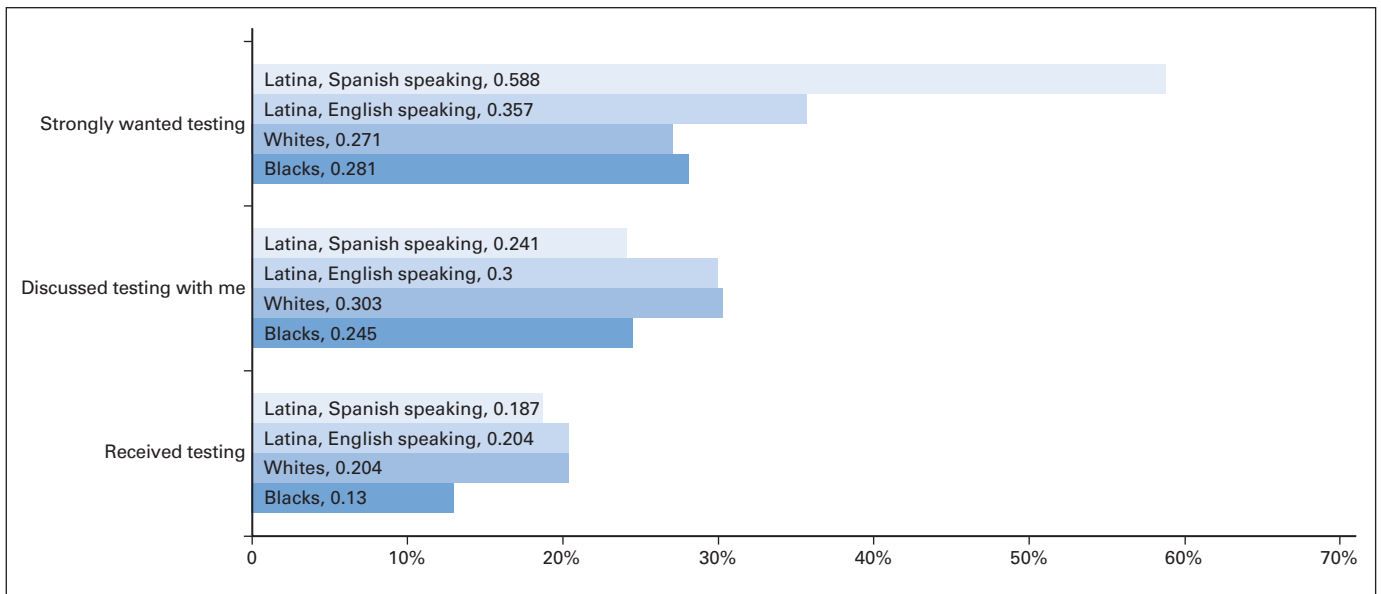
Of the 493 patients who expressed a strong desire for testing, 196 (43.4%) did not have a relevant discussion with a health care professional ("unmet need for discussion"). Table 3 shows that minority patients were more likely to express unmet need for discussion after controlling for other factors (see also Fig 2). Spanish-speaking Latinas were nearly five times more likely to have unmet need for discussion about testing than white non-Latina patients. Worry about local recurrence, contralateral new primary, and/or distant metastases during survivorship was considerably higher among those who had unmet need for discussion: 48.7% of those with unmet need for discussion worried about breast cancer compared with 24.9% of those without unmet need ( $P < .001$ ).

Table 4 shows the correlates of receipt of genetic testing in this sample. Patients who ultimately received genetic testing were younger, more likely to be white than black, and more likely to have a family history of breast and/or ovarian cancer. Among patients who expressed strong desire for testing, 41.3% had testing. Of the 269 patients who had testing, 20.5% did not express a strong desire for testing. Those who received testing endorsed various reasons (Appendix Fig A2A, online only). These included perceived physician recommendation (65.2%), patients' desire for information relevant to family members (53.6%). Those who did not receiving testing also indicated a variety of reasons (Appendix Fig A2B, online only). These included physician recommendation (64.9%) and personal choice (8.9%). Financial expense was cited as a reason for nonreceipt by 7.0%.

## DISCUSSION

In this study of diverse patients with breast cancer identified through population-based registries, about one-third strongly desired genetic testing. One in five reported test receipt, which was more common in whites than blacks, as well as those who were younger and had a family cancer history. Minority patients were significantly more likely to have unmet need for discussion in this context, and those with this unmet need were much more likely to express worry about breast cancer as long-term survivors.

Previous studies of desire for and uptake of genetic counseling in cancer-free patients have largely focused on populations of affluent white women, but noteworthy exceptions have identified racial/ethnic differences in attitudes, preferences, and decisions regarding genetic



**Fig 1.** Weighted percentages for wanting, discussing, and receiving genetic testing for breast cancer risk by race/ethnicity in a survey sample of 1,536 women diagnosed with early-stage breast cancer between 2005 and 2007 and reported to the population-based SEER registries of metropolitan Los Angeles and Detroit. Weighting includes both design weights compensating for oversampling of minorities and disproportionate selection across SEER sites, as well as nonresponse weights, as detailed in Methods and the Data Supplement.

testing.<sup>17-24</sup> One study found that African American women with a family history of breast or ovarian cancer were less likely to receive genetic counseling than white women with a similar family history, even after adjustment for socioeconomic status, estimated probability of *BRCA1/2* mutation carriage, risk perception and worry, attitudes

about testing, and primary care physician discussion.<sup>7</sup> Studies of ovarian cancer patients have documented lack of awareness about *BRCA1/2* mutation testing, particularly in minorities.<sup>25</sup> Our findings suggest marked unmet need for discussion, particularly among Latinas with breast cancer, a group who may have elevated risk of *BRCA1*

**Table 2.** Characteristics Associated With a Strong Desire for Genetic Testing

| Characteristic                             | Bivariate Associations |              |        | Best Multivariable Model |              |        |
|--|------------------------|--------------|--------|--------------------------|--------------|--------|
|  | OR                     | 95% CI       | P      | OR                       | 95% CI       | P      |
| Age at diagnosis (+ 1 year)*               | 0.95                   | 0.93 to 0.96 | < .001 | 0.95                     | 0.93 to 0.96 | < .001 |
| Race/ethnicity                             |                        |              | < .001 |                          |              | < .001 |
| White                                      | 1.00                   |              |        | 1.00                     |              |        |
| Black                                      | 1.14                   | 0.82 to 1.58 |        | 0.96                     | 0.67 to 1.67 |        |
| Latina, English speaking                   | 1.57                   | 1.07 to 2.30 |        | 1.32                     | 0.86 to 2.03 |        |
| Latina, Spanish speaking                   | 4.32                   | 2.95 to 6.35 |        | 3.64                     | 2.34 to 5.66 |        |
| Other                                      | 1.00                   | 0.38 to 2.64 |        | 1.09                     | 0.30 to 3.95 |        |
| Disease type                               |                        |              | .040   |                          |              |        |
| In situ                                    | 1.00                   |              |        |                          |              |        |
| Invasive                                   | 1.38                   | 1.02 to 1.87 |        |                          |              |        |
| Family history of breast or ovarian cancer |                        |              | .003   |                          |              | < .001 |
| No   | 1.00                   |              |        | 1.00                     |              |        |
| Yes  | 1.57                   | 1.17 to 2.11 |        | 1.73                     | 1.26 to 2.39 |        |
| Education                                  |                        |              | .011   |                          |              |        |
| High school or less                        | 1.00                   |              |        |                          |              |        |
| At least some college                      | 0.71                   | 0.54 to 0.93 |        |                          |              |        |
| Insurance                                  |                        |              | < .001 |                          |              |        |
| None                                       | 2.43                   | 1.52 to 3.89 |        |                          |              |        |
| Private                                    | 1.00                   |              |        |                          |              |        |
| Medicaid                                   | 1.40                   | 0.88 to 2.23 |        |                          |              |        |
| Medicare                                   | 0.66                   | 0.43 to 1.01 |        |                          |              |        |

Abbreviation: OR, odds ratio.

\*Another way of summarizing the information regarding the association between age and strong desire would be that for a woman 20 years older than another—for example, a 65-year-old versus a 45-year-old woman—the OR for having strong desire would be 0.33 (95% CI, 0.26 to 0.43) in the bivariable analysis and 0.34 (95% CI, 0.25 to 0.46) in the multivariable analysis.



**Table 3.** Characteristics Associated With Unmet Need for Discussion (a strong desire for testing but without a discussion of genetic testing with a medical professional)

| Characteristic                             | Bivariate Associations |               |        | Best Multivariable Model |               |        |
|--|------------------------|---------------|--------|--------------------------|---------------|--------|
|  | OR                     | 95% CI        | P      | OR                       | 95% CI        | P      |
| Age at diagnosis (+ 1 year)                | 0.99                   | 0.97 to 1.00  | .075   |                          |               |        |
| Race/ethnicity                             |                        |               | < .001 |                          |               | < .001 |
| White                                      | 1.00                   |               |        | 1.00                     |               |        |
| Black                                      | 1.68                   | 1.00 to 2.80  |        | 1.64                     | 0.94 to 2.88  |        |
| Latina, English speaking                   | 2.44                   | 1.41 to 4.23  |        | 2.59                     | 1.44 to 4.63  |        |
| Latina, Spanish speaking                   | 7.39                   | 4.58 to 11.93 |        | 7.08                     | 4.22 to 11.89 |        |
| Other                                      | 3.36                   | 1.00 to 11.29 |        | 4.41                     | 1.10 to 17.62 |        |
| Disease type                               |                        |               | .686   |                          |               |        |
| In situ                                    | 1.08                   | 0.73 to 1.61  |        |                          |               |        |
| Invasive                                   | 1.00                   |               |        |                          |               |        |
| Family history of breast or ovarian cancer |                        |               | .028   |                          |               | .060   |
| No   | 1.60                   | 1.05 to 2.42  |        | 1.57                     | 0.98 to 2.50  |        |
| Yes  | 1.00                   |               |        | 1.00                     |               |        |
| Education                                  |                        |               | < .001 |                          |               |        |
| High school or less                        | 2.46                   | 1.73 to 3.51  |        |                          |               |        |
| At least some college                      | 1.00                   |               |        |                          |               |        |
| Insurance                                  |                        |               | < .001 |                          |               |        |
| None                                       | 3.76                   | 2.25 to 6.29  |        |                          |               |        |
| Private                                    | 1.00                   |               |        |                          |               |        |
| Medicaid                                   | 1.79                   | 1.01 to 3.15  |        |                          |               |        |
| Medicare                                   | 1.06                   | 0.59 to 1.90  |        |                          |               |        |

Abbreviation: OR, odds ratio.

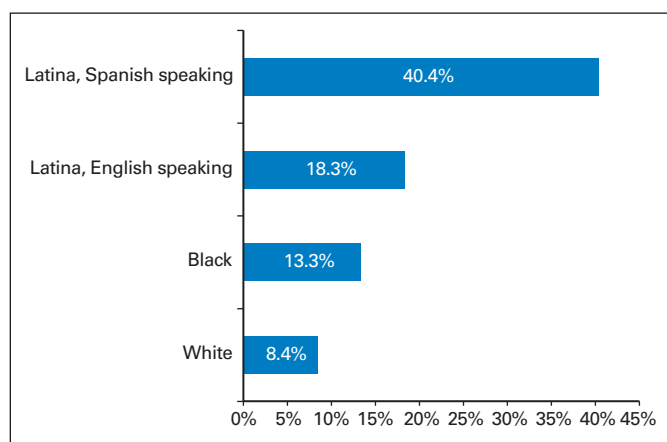
mutation carriage.<sup>26</sup> Attention to this disparity is necessary to ensure that all women diagnosed with breast cancer can make informed and preference-concordant decisions.

Previous studies have explored barriers to genetic testing, including affordability and insurance concerns,<sup>27</sup> including coverage. In one study, cost was described as very important by 23% of patients in a cancer risk assessment clinic<sup>20</sup>; numerous others reported that cost appears to influence testing uptake.<sup>25,28,29</sup> A survey of patients who

experienced cancer genetic counseling identified misperceptions about insurance coverage.<sup>30</sup> A relatively small proportion of our respondents endorsed financial concerns as a reason for nontesting, although it is possible that some women who had not discussed testing were unaware of the potential costs.

Of note, not all patients who express a strong desire for testing have significant probability of carrying a genetic mutation,<sup>5</sup> and some may misunderstand the distinction between a second primary cancer and a recurrence of the first cancer; simply discussing these issues might alleviate anxiety and not necessarily require testing to follow. Furthermore, studies have documented that patients tend to overestimate risk before genetic counseling.<sup>31</sup> Therefore, some patients with unmet need for discussion may harbor anxiety resulting from inaccurate risk perception, which might be resolved by provider discussion when testing is not clinically indicated. Our observation that patients with unmet need for discussion are more likely to express elevated levels of worry during survivorship supports this idea. Given the prevalence of misconceptions about genetic testing that may distort its use,<sup>32</sup> the infrequency of relevant discussion in breast cancer decision making that we observed in our 2006 cohort is concerning. These findings are even more relevant today given the exponential growth in news about genetic risk and rapidly increasing access to an expanded array of available genetic tests. Especially in today's climate, our results suggest that clinicians should proactively discuss genetic risk even with patients whom they perceive to be at low risk. Addressing this potentially missed clinical opportunity may alleviate worry and reduce confusion about the risks of subsequent primary cancers versus recurrence of the incident cancer.

Our finding that genetic testing receipt correlated with younger age and family cancer history is reassuring, since these are among the



**Fig 2.** Weighted percentages, by race/ethnicity, of unmet need for discussion (reporting a strong desire for genetic testing but failure to discuss this with a genetics counselor, physician, or other health professional) from a survey sample of 1,536 women diagnosed with early-stage breast cancer between 2005 and 2007 and reported to the population-based SEER registries of metropolitan Los Angeles and Detroit. Weighting includes both design weights compensating for oversampling of minorities and disproportionate selection across SEER sites, as well as nonresponse weights, as detailed in Methods and the Data Supplement.

**Table 4.** Characteristics Associated With Receipt of Genetic Testing

| Characteristic                             | Bivariate Associations |              |        | Best Multivariable Model |              |        |
|--|------------------------|--------------|--------|--------------------------|--------------|--------|
|  | OR                     | 95% CI       | P      | OR                       | 95% CI       | P      |
| Age at diagnosis (+ 1 year)                | 0.92                   | 0.91 to 0.94 | < .001 | 0.91                     | 0.89 to 0.93 | < .001 |
| Race/ethnicity                             |                        |              | .133   |                          |              | .012   |
| White                                      | 1.00                   |              |        | 1.00                     |              |        |
| Black                                      | 0.67                   | 0.43 to 1.03 |        | 0.45                     | 0.28 to 0.73 |        |
| Latina, English speaking                   | 1.00                   | 0.65 to 1.55 |        | 0.75                     | 0.45 to 1.22 |        |
| Latina, Spanish speaking                   | 1.02                   | 0.65 to 1.62 |        | 0.65                     | 0.39 to 1.10 |        |
| Other                                      | 0.33                   | 0.10 to 1.06 |        | 0.30                     | 0.07 to 1.19 |        |
| Disease type                               |                        |              | .036   |                          |              |        |
| In situ                                    | 1.00                   |              |        |                          |              |        |
| Invasive                                   | 1.51                   | 1.03 to 2.23 |        |                          |              |        |
| Family history of breast or ovarian cancer |                        |              | < .001 |                          |              | < .001 |
| No   | 1.00                   |              |        | 1.00                     |              |        |
| Yes  | 2.75                   | 1.94 to 3.90 |        | 3.11                     | 2.14 to 4.53 |        |
| Education                                  |                        |              | .012   |                          |              |        |
| High school or less                        | 1.00                   |              |        |                          |              |        |
| At least some college                      | 1.58                   | 1.11 to 2.26 |        |                          |              |        |
| Insurance                                  |                        |              | .023   |                          |              |        |
| None                                       | 1.00                   | 0.57 to 1.75 |        |                          |              |        |
| Private                                    | 1.00                   |              |        |                          |              |        |
| Medicaid                                   | 1.06                   | 0.58 to 1.94 |        |                          |              |        |
| Medicare                                   | 0.40                   | 0.23 to 0.73 |        |                          |              |        |

Abbreviation: OR, odds ratio.

strongest predictors of deleterious mutation carriage. Women diagnosed with breast cancer younger than age 40 years have a 10% frequency of *BRCA1/2* mutation carriage even in the absence of family history,<sup>33</sup> which substantially exceeds the general population frequency (1 in 400).<sup>34-37</sup> Prior studies reported higher uptake of genetic testing among those with first-degree relatives affected by cancer and those diagnosed at younger ages.<sup>8,20,28,38,39</sup>

Although this study has substantial strengths, including a high response rate and a racially/ethnically diverse sample of patients with breast cancer from population-based registries, it also has limitations. First, it was limited to two geographic areas; women from other areas, particularly rural areas, may have different experiences and concerns. Second, like most studies of this topic, we relied on patient self-report. Although we tried to explain the concept of genetic testing clearly and carefully evaluated our questions with intensive pretesting, it is possible that some respondents misconstrued the questions or remembered experiences inaccurately. Future research should consider complementary methods, such as analysis of taped interactions between providers and patients, to confirm these observations. Third, it is possible that associations observed were not causal. For example, the association between unmet need for discussion and worry might be confounded by an unmeasured variable (such as personality predisposition). Finally, we included only women; men with breast cancer may have different experiences with genetic testing that merit additional exploration in future work.<sup>40</sup>

In conclusion, we observed significant racial/ethnic variation in experiences with genetic evaluation and discussions of hereditary risk in this diverse sample of patients with breast cancer drawn from two population-based registries. Notably, we found a concerning unmet need for discussion that was more common among minorities. Although clinicians may fear that discussion of genetic risk will amplify

the stress of a breast cancer diagnosis,<sup>41</sup> many patients, especially minorities, appear likely to benefit from discussion to clarify their true risks. Public awareness of genetic testing has increased rapidly, in relation to recent judicial opinions, celebrity testing disclosures, and direct-to-consumer marketing.<sup>42-44</sup> Therefore, it is critical to recognize that patients with breast cancer, even those lacking recognized risk factors for deleterious mutation carriage, may nevertheless desire and benefit from an explicit discussion of genetic risk and its implications for their care.

Testing itself cannot and should not take the place of considered discussions of risk between physician and patient. This study suggests that discussions regarding the actual risk of a hereditary syndrome are critical, particularly in vulnerable populations. Physicians must take care to explain the difference between the risk of new primary cancer and recurrence of the incident cancer. Such discussions are essential to help patients at higher risk to access testing while also helping patients at lower risk to appropriately avoid testing without leaving lingering worry.

#### AUTHORS' DISCLOSURES OF POTENTIAL CONFLICTS OF INTEREST

Disclosures provided by the authors are available with this article at [www.jco.org](http://www.jco.org).

#### AUTHOR CONTRIBUTIONS

**Conception and design:** Reshma Jagsi, Monica Morrow, Ann S. Hamilton, John J. Graff, Steven J. Katz, Sarah T. Hawley

**Financial support:** Sarah T. Hawley

**Administrative support:** Steven J. Katz

**Collection and assembly of data:** Kent A. Griffith, Ann S. Hamilton, John J. Graff

**Data analysis and interpretation:** Reshma Jaggi, Kent A. Griffith, Allison W. Kurian, Monica Morrow, Steven J. Katz, Sarah T. Hawley

**Manuscript writing:** All authors

**Final approval of manuscript:** All authors

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## GLOSSARY TERMS

**BRCA1:** a tumor suppressor gene 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:** 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. Also known as the breast cancer 2 early onset gene.

**germline mutation:** an inherited variation in the lineage of germ cells. Germline mutations can be passed on to offspring.

**Surveillance, Epidemiology, and End Results (SEER):** a national cancer registry that collects information from all incident malignancies in multiple geographic areas of the United States.



**AUTHORS' DISCLOSURES OF POTENTIAL CONFLICTS OF INTEREST**

**Concerns About Cancer Risk and Experiences With Genetic Testing in a Diverse Population of Patients With Breast Cancer**

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**Reshma Jagsi**

**Employment:** University of Michigan

**Honoraria:** International Journal of Radiation Oncology Biology Physics

**Consulting or Advisory Role:** Eviti

**Research Funding:** AbbVie (Inst)

**Kent A. Griffith**

No relationship to disclose

**Allison W. Kurian**

**Research Funding:** Myriad Genetics, Invitae

**Monica Morrow**

No relationship to disclose

**Ann S. Hamilton**

No relationship to disclose

**John J. Graff**

No relationship to disclose

**Steven J. Katz**

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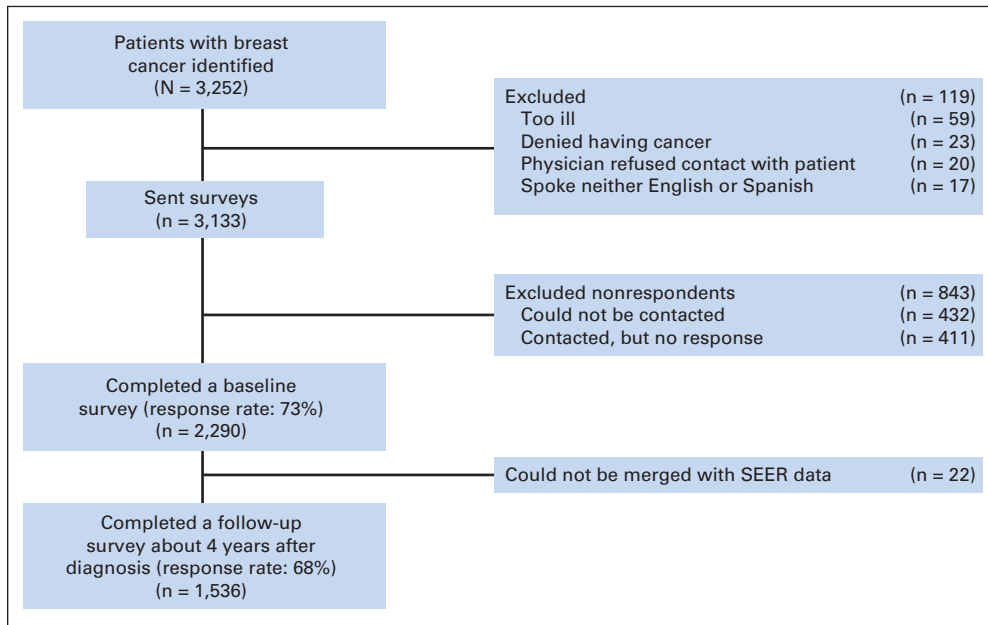
**Sarah T. Hawley**

No relationship to disclose

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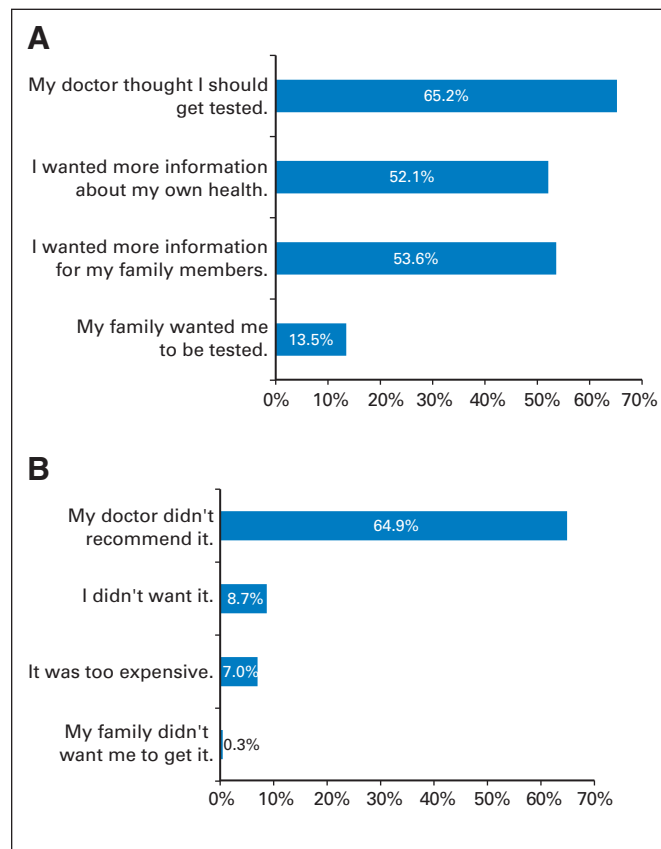
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**Appendix**



**Fig A1.** Patient flow into the study. This figure depicts the flow of patients into the study from those initially identified to the final analytic sample.

### Genetic Testing Experiences of Patients With Breast Cancer



**Fig A2.** The data in this figure derive from responses of a survey sample of 1,536 women diagnosed with early-stage breast cancer between 2005 and 2007 and reported to the population-based SEER registries of metropolitan Los Angeles and Detroit. (A) Weighted percentages for reasons for getting tested among the population who received testing (n = 269). (B) Weighted percentages for reasons for not getting tested among population who did not receive testing (n = 1,118). Weighting includes both design weights compensating for oversampling of minorities and disproportionate selection across SEER sites, as well as nonresponse weights, as detailed in Methods and the Data Supplement.