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Genetic counseling, testing and family communication into survivorship after diagnosis of breast cancer

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Abstract

Purpose: To examine receipt of genetic testing and communication with relatives about results into survivorship after diagnosis of breast cancer.

Patients and methods: Women aged 20–79 diagnosed with early-stage breast cancer in 2014–2015 and reported to the Georgia and Los Angeles County SEER registries were surveyed approximately seven months and six years after diagnosis (N=1412). We asked about genetic counseling, testing, and communication with relatives about results. We categorized women into indications for testing based on clinical guidelines at time of diagnosis and at the time of the follow-up survey.

Results: 47.4% had indications for genetic testing at any time: 28.0% at baseline and an additional 19.4% at time of the follow-up survey (FUPs only); 71.9% (95% CI 67.4%−76.4%) of those with a baseline indication reported genetic testing vs. 53.3% (95% CI 47.3% - 59.2%) with an indication at FUPs only and 35.0% (95% CI 31.6% - 38.4%) with no indication (p<.001). There were no significant racial or ethnic differences in receipt of testing, controlling for age and clinical indications (p=0.239); results for genetic counseling were similar. Only 3.4% of survivors had

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direct-to-consumer testing (DTCt) for cancer. Testers who reported a pathogenic variant (n=62) were much more likely to have talked to most or all their first-degree adult relatives about genetic testing than those with a variant of unknown significance (n=49) or a negative finding (n=419): 62.7% vs 38.8% and 38.0%, respectively (p<.001).

Conclusion: Many women with indications for genetic counseling and testing into survivorship do not receive it. But those tested reach out to family members based on the clinical relevance of their results. Very few patients obtained DTCt, which suggests that these tests do not substitute for clinical testing in breast cancer survivors.

Introduction

Support for universal germline genetic testing after a breast cancer diagnosis is growing because of concerns that targeted guidelines[1–3] fail to identify many patients who could benefit from genetic counseling and testing. Studies show that about one-third of patients with breast cancer meet clinical practice guidelines for genetic counseling and testing at the time of diagnosis, but many do not receive it[4–8]. Access to genetic counseling and testing for the 280,000 people diagnosed with breast cancer in the U.S. each year is increasingly important because results influence locoregional and systemic treatment decisions and inform the risk of second primary cancers over time[9, 10]. Additionally, genetic testing results in patients have important implications for cancer risk stratification and prevention for relatives. Thus, genetic counseling and testing is important for the nearly four million survivors of breast cancer living in the U.S and their family members. However, virtually nothing is known about its uptake in the years after a diagnosis of breast cancer. Untested patients with clinical indications for genetic counseling and testing at the time of diagnosis continue to benefit from counseling and testing in the survivorship period, which we consider as the period after completion of first-course therapy with surgery, radiation and/or chemotherapy. Additionally, patients who did not have an indication at time of initial treatment management may meet criteria later due to a diagnosis of a new primary cancer or metastatic recurrence, a change in pertinent family history, or a change in testing guidelines.

We examined patient report of genetic counseling, clinical genetic testing, and use of direct-to-consumer genetic testing (DTCt) from diagnosis through the first six years of survivorship in a diverse, population-based cohort of women diagnosed with breast cancer in 2014–15 as reported to the Surveillance, Epidemiology and End Results (SEER) registries of Georgia and Los Angeles County. We hypothesized that a substantial proportion of survivors who did not receive genetic counseling and testing at the time of diagnosis did so during the survivorship years. We further hypothesized that a considerable number of survivors who did not receive clinical testing may have utilized DTCt instead.

Methods

The iCanCare Study is a population-based, longitudinal survey study of women with early-stage breast cancer and their clinicians. As detailed previously[7] women ages 20–79 who were newly diagnosed with early-stage breast cancer (stages 0-II) in 2014–2015 as reported to the SEER registries of Georgia and Los Angeles County were surveyed. African American, Asian and Latina women were oversampled. Women were ineligible if they had

stage III or IV disease, had tumors larger than five centimeters, or could not complete a questionnaire in English or Spanish (N=258). A total of 2,502 women completed surveys, resulting in a 68.0% baseline response rate. The median time from diagnosis to completion of the initial baseline survey was 7.8 months (25–75% range 5.6 – 10.1 months) and 83.6 months (25–75% range 53.1 – 86.8 months) for the follow-up survey.

We sent respondents a paper follow-up survey approximately six years after their initial diagnosis in 2021–2022, with an option to complete the survey online. As in prior work with this study, we utilized a modified Dillman approach to patient recruitment, including reminders to non-respondents and a $20 up front cash incentive. Patients were deemed ineligible for the follow-up study if they were deceased (N=108) or were too ill (N=33) to participate. The follow-up survey was completed by 1,412 of the 2,361 eligible women (follow-up survey response rate of 59.8%, see Supplemental Figure 1). Responses to the surveys were merged with SEER clinical data and a de-identified analytic dataset was created. The study was approved by the University of Michigan Institutional Review Board (IRB) and the state and institutional IRBs of the SEER registries. We obtained informed consent from each participant.

We asked women in the follow-up survey about the occurrence of new primary cancers by cancer type (including breast), recurrence of breast cancer (including anatomic location), their family history of cancer, receipt of genetic counseling, receipt of clinical germline genetic testing and results, communication with relatives about cancer genetic testing, and use of DTCt after diagnosis. We categorized women into indications (yes/no) based on the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) at time of diagnosis (genetic counseling and testing indication baseline) and at the time of the follow-up survey (genetic counseling and testing indication at follow-up survey (FUPs) only).

Outcome Measures

All outcome measures were derived from the follow-up survey:

- Patients were first asked "*How long has it been since you last had a counseling session with a genetic counseling expert – that is, an appointment where the whole discussion is about genetic cancer risk?*";
- The next question was *"How long has it been since you last had a blood or saliva genetic test for future cancer risk that was ordered by a doctor or genetic counselor*?" Response categories were: Never, within the past two years, two to five years ago, more than five years ago.
- Patients who reported receipt of testing were then asked: *What were the results of the* ***most recent*** *genetic test that was ordered by a doctor or genetic counselor? Please mark* ***ALL*** *that apply (*negative, positive for a gene mutation, uncertain (variant of unknown significance (VUS)). Patients who reported a result of positive for a gene mutation were coded as positive, regardless of other responses checked. Patients who reported a VUS result and who did not report a positive result were coded as VUS. Patients who reported negative results and no positive or VUS results were coded as negative.

- Patient report of DTCt followed the questions above. We framed these questions on tests sold by *"companies that offer genetic tests for cancer risk on the internet, without the need to involve your doctor. Anyone can buy these tests online, get a testing kit in the mail, collect their spit in a special cup or tube, and mail the test kit back to the company for analysis. Examples of companies offering this "direct-to-consumer" testing include* ***23andMe***, ***AncestryDNA***, *and* ***Color***."
  - We first asked "How much have you researched these types of tests online? Response categories used a five-point Likert scale from not at all to a lot.; We then asked, "Have you ever taken a direct-to-consumer genetic test for cancer risk that you ordered on the internet?" Examples included 23andMe, AncestryDNA, Color, and Other and for each example, response categories were yes and no.
- Patient report of communication with family members about their test results: "*Have you talked with your immediate adult blood relatives (parents, brothers and sisters, children) about getting clinical genetic testing to learn more about their own future cancer risk?"* Response categories were: Yes, I have talked to most or all of my adult family members; Yes, I have talked to some of my adult family members (but not all); No, I haven't talked to any adult family members.

Independent variables: Supplemental Table 1 shows the criteria for genetic counseling and testing indications based on NCCN guidelines at the time of baseline[11] and follow-up surveys[12]. Indications at baseline were derived from the baseline survey and the SEER data (presence of triple-negative breast cancer (TNBC) subtype). Indications at time of the follow-up survey were derived from that survey and the SEER data (presence of TNBC subtype). Other covariates included age at diagnosis, race and ethnicity, education level, and annual household income all derived from the baseline survey; clinical stage and grade at time of diagnosis and geographic site were derived from the SEER data.

Analytic plan

We first described patient characteristics for the analytic sample of 1,412 respondents who completed the baseline and follow-up surveys after diagnosis (Table 1). We then showed patient report of genetic risk evaluation by indication for genetic counseling and testing and by timing of testing and counseling during the study period. We then examined patient report of testing by race and ethnic groups, controlling for age, education, and clinical stage. Next, we described engagement and communication with relatives about genetic test results by self-reported results outcomes (pathogenic variant (PV), variant of uncertain significance (VUS), negative). Finally, we described patient report of use of DTCt during the study period. To account for the effects of differential response rates, we repeated all analyses using analytic weights based on covariates with significantly different response rates and examined the results for differences from the unweighted analyses.

Results

Population characteristics:

Table 1 shows characteristics of the study population. The median age was 55.3; about half were of minoritized race or ethnicity (18.2% Black, 18.0% Latina, and 9.5% Asian); 27.1% had a high school education or less; and 31.8% reported annual income of less than $40,000. The distribution of clinical stage and grade at time of diagnosis reflected the selection of patients, with more favorable disease in the inception cohort. Patient report of a second primary breast cancer (3.5%) or metastatic recurrence (1.5%) was uncommon. The patient sample was nearly equally distributed between the two state registries (51.6% from Georgia vs. 48.4% from Los Angeles County). Less than half of the respondents (47.4%) had indications for GRE over the study period: 28.0% at baseline and an additional 19.4% at time of the follow-up survey (FUPs only).

Genetic testing and counseling:

Figure 1 shows patient-reported genetic testing and genetic counseling at FUPs by indication category (baseline, FUPs only, no indication). Nearly three-quarters (71.9%, 95% CI 67.4% −76.4%) of those with a baseline indication reported genetic testing over the observation period, vs. 53.3% (95% CI 47.3% - 59.2%) with an indication at FUPs only and 35.0% (95% CI 31.6% - 38.4%) of those with no indication ($p<.001$). A substantial proportion of those who reported testing received the test during the survivorship period: 13.0% of testers with baseline indications tested in the prior two years of the survey vs 19.5% of those with indications in FUP only ($p<.001$). Assessment of confounding showed no substantial effects of age, education, or clinical stage and thus these descriptive results are not adjusted. Figure 2 shows that there were no significant racial or ethnic differences in receipt of testing during survivorship, controlling for age and clinical indications ($p=.239$). Results for report of genetic counseling were very similar (Supplemental Figures 2 and 3).

Communication of test results with family members:

Testers who reported a PV result (n=62) were much more likely to have talked to most or all their first-degree adult relatives about genetic testing than those with a VUS (n=49) or a negative finding (n=419): 62.7% vs 38.8% and 38.0%, respectively ($p<.001$).

Interest and receipt of DTCt:

Overall, there was very little interest in DTCt for cancer risk: 5.1% researched DTCt online somewhat to a lot and only 3.4% had DTCt.

Follow-up response rates differed by income, employment, education, race, receipt of hormonal therapy, and having subsequent non-breast cancer. To account for possible bias related to differential response, we generated weights and re-ran all analyses using the weights. There were no meaningful differences in our results.

Discussion

We performed a large, SEER-based longitudinal survey study of patients diagnosed with early-stage breast cancer in 2014–15 at two time points: seven months after diagnosis and then six years into survivorship. We found that a substantial proportion of women met NCCN guidelines for genetic counseling and testing that were published during the initial diagnosis and treatment periods. Additionally, many of those who were not candidates for genetic counseling and testing at time of diagnosis became eligible over the course of the survivorship period because of pertinent new cancers or additional family history, and the somewhat broader indications promulgated by NCCN guidelines over the course of the study period. Yet, many women eligible for genetic counseling and testing did not receive it. We observed this gap uniformly across race and ethnic groups, with no significant differences across subgroups.

In response to growing evidence for the clinical utility of testing and studies suggesting under-testing, several professional organizations have expanded the criteria for genetic counseling and testing[13, 14] and there is growing advocacy for near-universal germline testing after diagnosis of breast cancer[15–17]. It has become even more important to increase testing after diagnosis, as evidence continues to grow about the need for germline test results for both locoregional and systemic management[10]. Additionally, germline genetic testing after diagnosis of breast cancer is an essential strategy to close the unacceptable gap in cascade testing of families with hereditary cancer risk[18–21].

The survivorship period that immediately follows an often-arduous initial course of therapy is an essential time of recovery for patients with breast cancer. However, there are important clinical issues during survivorship that warrant close engagement and continuity with medical oncology, including treatment related side-effects, medication management for patients on longer-term therapies, and assessment and management of future cancer risk in patients and in their families through genetic counseling and testing. Oversight by medical oncologists and other clinicians including primary care during the survivorship period may result in missed opportunities to optimize patient and family outcomes. A particular challenge is the need to record an accurate and up-to-date family history of cancer, which may be under-ascertained in follow-up encounters with patients.

Our results suggest optimism in engaging more patients in genetic counseling, testing and family communication during survivorship. During the study period, rates of testing and counseling after a diagnosis of breast cancer increased in the geographic regions of our study[22] and our findings demonstrate high rates of testing and counseling over time, especially in patients who had indications at time of cancer diagnosis. We did not observe significant race and ethnic disparities in counseling or testing. Additionally, our results suggest that patients reach out to family members based on the clinical relevance of their results. Finally, a reassuring finding from our study is that very few patients reported any interest in DTCt and fewer obtained it across the long survivorship period. There has been concern that a substantial number of patients may seek DTCt and fail to differentiate DTCt from clinical-grade testing[23–27]. Our findings suggest that DTCt is not substituted for clinical testing by breast cancer survivors.

Limitations: Although the response rate for the two surveys in this longitudinal study was high, our results could have been biased by differential response rates. We accounted for this possibility by performing weighted analyses based on measured covariates, but there may have been differential response rates by unmeasured ones. We may have misclassified indications for genetic counseling and testing because some pertinent patient personal or family history cancer history was not ascertained. Outcomes were derived from patient reports which may be prone to recall bias. Reassuringly, in our prior work with this cohort, we found good concordance between patient self-report of genetic testing in the baseline survey (29%)[6] and linkage to genetic testing data obtained directly from laboratories (26%) near the time of cancer diagnosis[28]. Additionally, survey face and construct validity were high. Finally, the lower rate of testing observed in patients with indications at follow-up only vs. baseline is partly related to the shorter observation period between the two groups.

Conclusion: Germline genetic testing is increasingly important after a diagnosis of cancer, for treatment management and for cancer risk reduction in families with hereditary cancer syndromes. While it is ideal to obtain genetic testing results during treatment planning, the survivorship period remains a major missed opportunity to engage patients and family members who may be at risk for hereditary cancer susceptibility and may be candidates for effective risk reduction and treatment strategies. Proponents of universal testing of all breast cancer patients argue that it would increase the detection of clinically meaningful results, reduce disparities in receipt of testing, and facilitate a clear focus on cascade testing and cancer risk reduction for survivors and family members[14]. However, a potential adverse outcome is more detection of meaningless results - particularly VUS, which are more frequently detected in racially and ethnically minoritized groups. Indeed, broadening clinical guidelines in conjunction with the advent of larger multi-gene test panels has already markedly increased the rate of clinically less meaningful results, particularly VUS[22, 29]. Proponents of broadening guidelines argue that clinically non-contributory findings such as VUS can be managed successfully by clinicians and that the failure to detect meaningful PVs is a much bigger problem. More research is needed on the potential adverse consequences of less clinically meaningful test results on management of breast cancer and engagement with families regarding cascade genetic risk evaluation. Additionally, more testing of patients diagnosed with cancer yearly, and of the growing number of cancer survivors, should motivate more research to evaluate and implement multi-pronged strategies to facilitate genetic counseling, testing and outreach to family members in clinical practice.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Context Summary

Key objective:

What is the uptake of genetic testing and counseling in a cancer-registry based cohort of survivors of breast cancer up to 6 years after diagnosis?

Knowledge generated:

Clinical indications for genetic testing increased into survivorship but many eligible women did not receive it. Tested patients reach out to family members based on the clinical relevance of their results. Few patients reported interest in direct-to-consumer testing and fewer obtained it, which suggests that these test options are not substituted for clinical testing in breast cancer survivors.

Relevance (written by Stephanie Wheeler):

Germline genetic testing is increasingly important after a diagnosis of cancer, for treatment management and for cancer risk reduction in families with hereditary cancer syndromes, yet many survivors and their family members do not receive it, representing an important area for future inquiry.

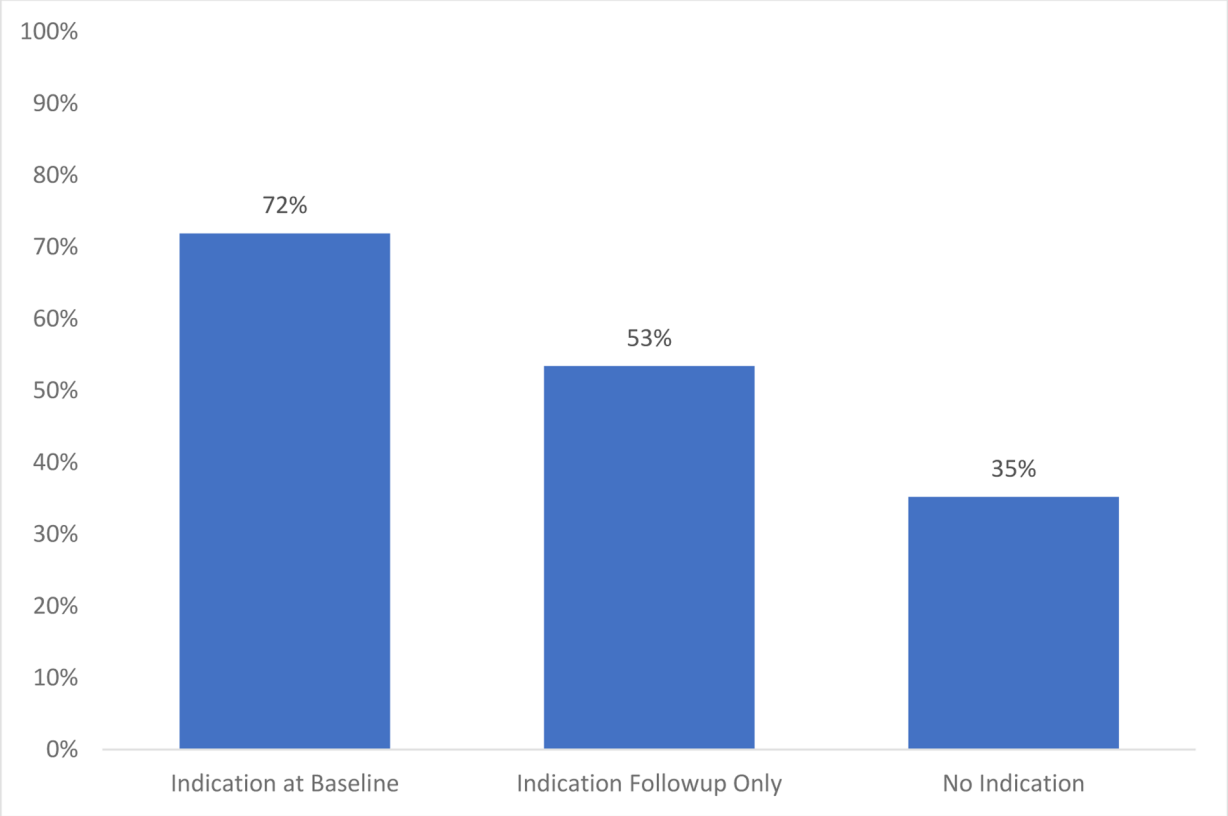

Figure 1.

Percent of respondents who reported germline genetic testing in the follow-up survey by clinical guidelines at time of the baseline survey (at time of diagnosis) and at time of the follow-up survey (approximately six years after diagnosis).

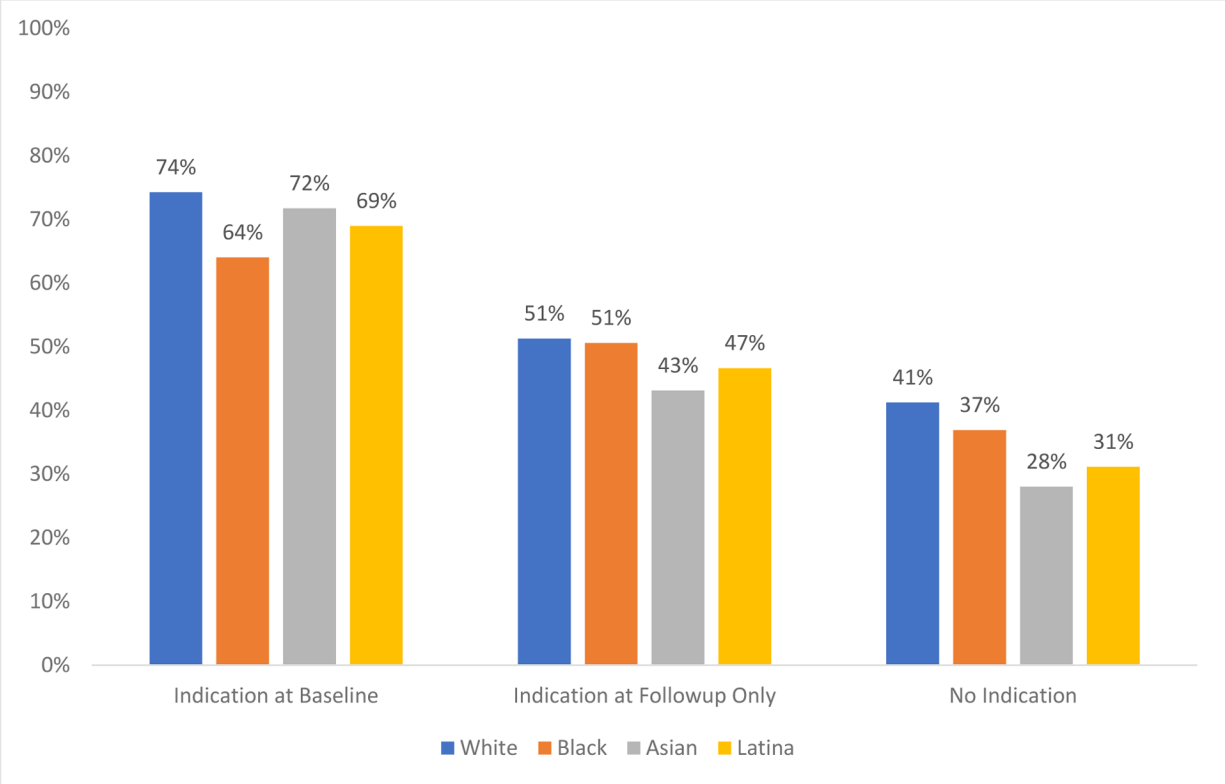

Figure 2.

Percent of respondents who reported genetic testing in the follow-up survey by guideline indication and race and ethnic group identity. Results are adjusted for age, education, and clinical stage at diagnosis.

Table 1.

Characteristics of the Study Population

	n	%
Age (missing = 1)		
<40	28	2.0%
40 – 49	168	11.9%
50 – 59	364	25.8%
60 – 69	522	37.0%
70+	329	23.3%
Race (missing = 29)		
White	750	54.2%
Black	252	18.2%
Latina	249	18.0%
Asian	132	9.5%
Education (missing = 33)		
High School/GED or less	373	27.1%
Some college or technical school	401	29.1%
College Graduate or higher	605	43.9%
Income (missing = 232)		
<20K	174	14.7%
20K – <40K	201	17.0%
40K – <60K	202	17.1%
60K – <90K	212	18.0%
90K+	391	33.1%
Stage (missing = 32)		
0	267	19.4%
I	773	56.0%
II	340	24.6%
Grade (missing = 63)		
1	381	28.2%
2	615	45.6%
3	353	26.2%
New Breast Cancer Since Diagnosis (missing = 31)		
No	1332	96.5%
Yes	49	3.5%
Breast Cancer Distant Recurrence Since Diagnosis (missing = 29)		
No	1362	98.5%
Yes	21	1.5%
Geographic Site		
Georgia	729	51.6%

	n	%
Los Angeles	683	48.4%
Indication for Genetic Risk Evaluation after Diagnosis		
Baseline	395	28.0%
Followup Only	274	19.4%
No Indication	743	52.6%