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Brief Communications

Recall of Genomic Testing Results Among Patients with Cancer

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Disclosures of potential conflicts of interest may be found at the end of this article.

Abstract

Background. Genomic testing of somatic and germline DNA has transformed cancer care. However, low genetic knowledge among patients may compromise care and health outcomes. Given the rise in genomic testing, we sought to understand patients' knowledge of their genetic test results.

Materials and Methods. We conducted a survey-based study with 85 patients at a comprehensive cancer center. We compared self-reported recall of (a) having had somatic/germline testing and (b) their specific somatic/germline results to the genomic test results documented in the medical record.

Results. Approximately 30% of patients did not recall having had testing. Of those who recalled having testing, 44% of patients with pathogenic/likely pathogenic germline mutations and 57% of patients with reported somatic alterations did not accurately recall their specific gene or variant-level results.

Conclusion. Given significant knowledge gaps in patients' recall of genomic testing, there is a critical need to improve patient-directed education and return-of-results strategies.

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Introduction

Despite the importance of somatic and germline genetics in oncology, genetic knowledge among the general population [1, 2] and patients with cancer is limited [3, 4]. Genetic knowledge gaps are important as they are associated with suboptimal patient outcomes. Low genetics knowledge is associated with lower screening uptake [5]. Furthermore, in a treatment context, patients who know their own tumor testing results are more involved in therapeutic decision-making and more likely to receive hormonally targeted therapy [6, 7]. Finally, because knowledge is disproportionately low in individuals of lower socioeconomic status and racial/ethnic minorities, understanding and addressing knowledge gaps may help ameliorate care disparities [7, 8]. Because a lack of knowledge about genomics generally (e.g., genetic concepts) and a patient's own test results ("personal genomic knowledge") may influence the quality of care that patients with cancer receive, we sought to better understand somatic, germline, and general genomic knowledge gaps among patients with solid tumors who have received genomic testing.

Materials and Methods

We surveyed patients at the City of Hope (COH) Comprehensive Cancer Center in Duarte, CA, between 2018 and 2019. English-speaking adult patients were eligible if they received genomic testing and had an Eastern Cooperative Oncology Group performance status ≤2. Eligible patients were referred to the study by collaborating clinicians and then enrolled by study staff. Standard practice at COH is for the oncology team to return somatic results to patients and for geneticists and genetic counselors to return germline results to patients. All study activities were approved by the City of Hope Institutional Review Board, including a waiver of documentation of informed consent.

Survey domains include demographics, disease characteristics (i.e., somatic and germline results), receipt of

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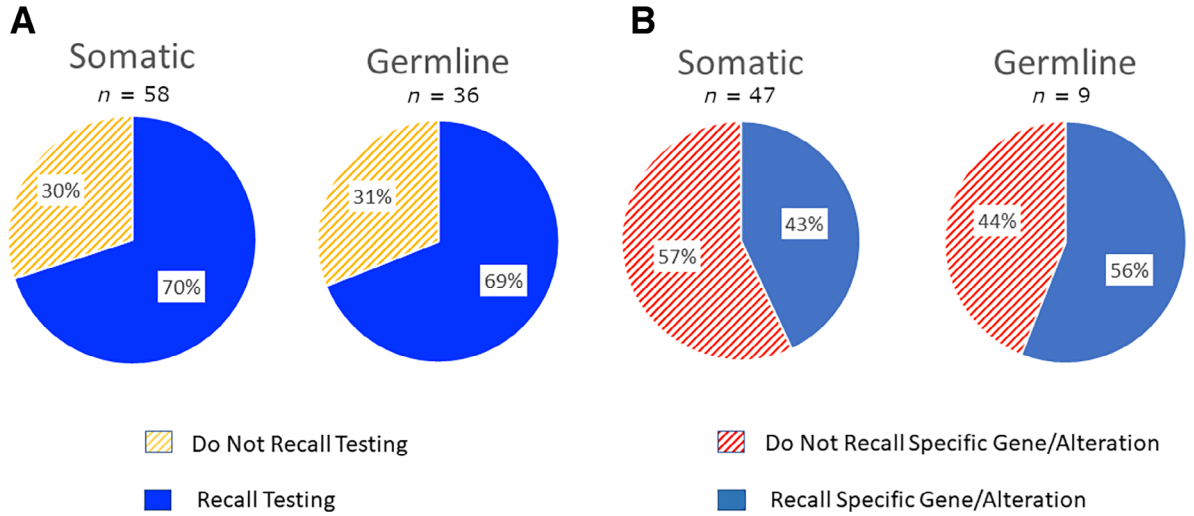

Figure 1. Overall recall of testing history and specific gene/alterations among patients who tested positive. **(A):** Recall of having had genomic testing. **(B):** Among patients who recall testing positive, recall of specific gene/alteration.

genetic counseling, cancer genomic knowledge (adapted from Blanchette et al. [9]), and germline genetic knowledge (Supplemental online survey) [10]. Patients could complete surveys on paper or via RedCap. We obtained genetic testing information (e.g., somatic and germline results) and health care use (e.g., targeted therapies) from the medical record. To assess patients' personal genomic knowledge, we compared their self-reported recall of their testing results with results in the medical record. Patients were considered to have "positive" results if they had (a) somatic findings on their test report (excluding variants of uncertain significance) and/or (b) germline pathogenic/likely pathogenic findings.

We described the distribution of patient characteristics and tested their association with genomic test result recall using Fisher's exact tests. We used Wilcoxon rank sum tests to compare personal genomic knowledge with patients' general and cancer-specific genetic knowledge scores. The p values <.05 were considered statistically significant. All analyses were conducted using SAS 9.4.

Results

Of the 97 patients who enrolled, 85 completed the survey and are included in the analyses. Two received germline testing only, 49 received somatic testing only, and 34 received both (Table 1). Most patients had lung (29%) or breast cancer (32%), were White (88%), were female (71%), and had completed at least some college education (69%). In total, 97% of patients with germline tests received genetic counseling, and 59% of all patients received targeted therapy.

Among patients who received germline and/or somatic testing (as documented in the medical record), approximately 30% did not recall having had genomic testing (Fig. 1). Among somatic test recipients, patients were more likely to recall that they had been tested if they had a positive result (74%) relative to those with negative results (23%; $p = .0004$). Among those with positive results who recalled having been tested, 44% did not accurately recall their specific germline test results (i.e., gene and/or alteration) and 57% did not accurately recall their specific somatic test results. We did not detect a difference in the recall of results for either testing type based on positive/negative test status. Germline result recall was better among patients with breast cancer and early-stage cancer, but this difference was not statistically significant ($p = .05$ *and .06, respectively*; Supplemental online Table 1). Somatic result recall was best among patients with lung cancer and more highly educated patients ($p = .03$ and .001, respectively). Somatic result recall was better among patients who received of a copy of their test results ($p = .01$), but no difference was detected by germline result recall ($p = .41$). No recall differences were detected by participants' sex or racial/ethnic group, receipt of genetic counseling, targeted therapy use, or clinical trial enrollment. Receipt of targeted therapy was higher among those who recalled their positive test results versus those who did not; however, this was not significantly different (73% vs. 47%, $p = .09$; data not shown).

Genomic knowledge was variable. Of nine questions asked about cancer genomics, patients correctly answered an average of five questions (SD = 2). Of 19 questions asked about germline genetics, patients correctly answered an average of 10 (SD = 5). Patients who accurately recalled their results were more likely to have a higher germline genetic knowledge scores (germline mean difference = 4, $p = .04$; somatic mean difference = 3, $p = .02$) but not a higher cancer genomic knowledge score (Supplemental online Fig. 1).

Discussion and Conclusion

Study strengths include an assessment of personal genomic knowledge across multiple cancer types in patients

Table 1. Participant characteristics ($n = 85$)

Characteristics	*n* (%)
Test type	
Germline only	2 (2)
Germline mutation positive	0 (0)
Somatic only	49 (58)
Somatic mutation positive	27 (55)
Both germline and somatic	34 (40)
Germline mutation positive	9 (26)
Somatic mutation positive	20 (59)
Cancer type	
Lung	25 (29)
Breast	28 (33)
Ovarian	11 (13)
Other	21 (25)
Stage	
I	4 (5)
II	15 (18)
III	16 (19)
IV	48 (56)
Unknown	2 (2)
Age, years	
30–39	10 (11)
40–49	10 (11)
50–59	19 (22)
60–69	28 (34)
70–79	16 (18)
80–89	2 (2)
Gender	
Female	61 (72)
Male	24 (28)
Hispanic ethnicity	19 (22)
Race	
White	75 (88)
Black	2 (2)
Asian	7 (8)
Other	1 (1)
Marital status	
Married	60 (71)
Divorced or widowed	16 (19)
Other	7 (8)
Unknown	2 (2)
Education	
High school or less	15 (18)
College	43 (51)
Graduate school	15 (18)
Unknown	12 (14)
Occupation	
Employed	34 (40)
Unemployed/ Student/ Homemaker	25 (29)
Retired	20 (23)
Unknown	6 (7)

who had somatic and/or germline testing. However, findings from our single-site study of largely White, well-educated, female patients cannot be generalized to other populations. Additionally, with our sample size, we could not sufficiently power subgroup analyses by cancer type or testing indication.

We demonstrate substantial knowledge gaps among patients with cancer who underwent somatic and/or germline genomic testing as part of their care. Our data suggest that patients frequently fail to recall that they received genomic testing and may not readily recall their specific test results even if they know that testing occurred. This finding is concerning for the 26% of patients who had positive results but were unaware that testing occurred, particularly given that our study was conducted at an National Cancer Institute–designated comprehensive cancer center with specialized oncology care and a large genetics program. Given the underuse of targeted therapy and indicated care for germline mutations carriers, increasing patients' personal genomic knowledge may be an effective strategy to increase patient care engagement and uptake of genomically guided care. Genetic counseling was nearly ubiquitous among germline test recipients, so expansion of genetic services alone may be insufficient to fully inform patients. More work is needed to determine whether these knowledge gaps are demonstrated in other care settings and populations and if they are associated with poor quality care. Finally, novel direct-to-patient pretest education and return-of-results strategies should be explored. Leveraging multimedia, lay-oriented videos, and web tools could improve the transmission of genomic information to patients and help scale genomic care delivery.

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