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The breast cancer genetic testing experience: probing the potential utility of an online decision aid in risk perception and decision making

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Abstract

Background Despite the association of pathogenic variants (PVs) in cancer predisposition genes with significantly increased risk of breast cancer (BC), uptake of genetic testing (GT) remains low, especially among ethnic minorities. Our prior study identified that a patient decision aid, *RealRisks,* improved patient-reported outcomes (including worry and perceived risk) relative to standard educational materials. This study examined patients' GT experience and its influence on subsequent actions. We also sought to identify areas for improvement in *RealRisks* that would expand its focus from improved GT decision-making to understanding results.

Methods Women enrolled in the parent randomized controlled trial were recruited and interviewed. Demographic data was collected from surveys in the parent study. Interviews were conducted, transcribed, and coded to identify recurring themes. Descriptive statistics were generated to compare the interviewed subgroup to the original study cohort of 187 women.

Results Of the 22 women interviewed, 11 (50%) had positive GT results, 2 (9.1%) with a *BRCA1/2* PV, and 9 (40.9%) with variants of uncertain significance (VUS). Median age was 40.5 years and 15 (71.4%) identified as non-Hispanic. Twenty (90.9%) reported a family history of BC, and 2 (9.1%) reported a family history of *BRCA1/2* PV. The emerging themes included a preference for structured communication of GT results and the need for more actionable knowledge to mitigate BC risk, especially among patients with VUS or negative results. Few patients reported lifestyle changes following the return of their results, although they did understand that their behaviors can impact their BC risk.

Conclusions Patients preferred a structured explanation of their GT results to facilitate a more personal testing experience. While most did not change lifestyle behaviors in response to their GT results, there was a consistent call for further guidance following the initial discussion of GT results. Empowering patients, especially those with negative or VUS results, with the context to internalize the implications of their results and form accurate risk perception represents a powerful opportunity to optimize subsequent risk management strategies. Informed by this study, future work will expand *RealRisks* to include the return of results and decision support to navigate concrete next steps.

Keywords Genetic testing, Breast cancer, BRCA 1/2, Pathogenic variant, Decision aid

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Background

One of the primary bottlenecks to efficacious and inclusive breast cancer (BC) prevention is the identification of high-risk patients. Specifically, identifying women with pathogenic variants (PVs) in *BRCA1* and *BRCA2* (*BRCA1/2*) can inform risk management and prevention strategies to reduce the risk of developing breast and ovarian cancer [1, 2].Women with hereditary breast and ovarian cancer syndrome (HBOC) attributable to *BRCA1/2* PVs have a lifetime BC risk of 40% to 60% and a lifetime ovarian cancer risk of 20% to 40% [3–5]. Additionally, up to 10% of breast cancers and 15% to 20% of ovarian cancers are attributed to PVs in HBOC predisposition genes [6]. Risk management strategies, including enhanced BC screening with mammography and breast MRI, chemoprevention, and risk-reducing surgeries such as prophylactic mastectomy or bilateral salpingo-oophorectomy (BSO) can significantly decrease a *BRCA1/2* carrier's cancer risk (up to 90% with risk-reducing surgeries) once the patient is identified [6–11].

The United States Preventive Services Task Force (USPSTF) recommends that primary care providers (PCPs) screen asymptomatic women at increased risk of carrying *BRCA1/2* PVs [12, 13]. Pertinent risk factors include early onset of breast or ovarian cancer, multiple cases of breast or ovarian cancer in the family, bilateral breast cancer, male breast cancer, Ashkenazi Jewish descent, or a previously identified *BRCA1/2* PV in the family [13]. Although the indications for and availability of genetic testing (GT) continue to expand, many women at an elevated risk of carrying *BRCA1/2* PVs are never identified [14–17]. Racial/ethnic minorities, along with patients of lower education and income levels, are less likely to be referred for GT, further perpetuating disparities in clinical outcomes [14, 18–20].

Given the expanding criteria for GT, the need for genetic risk assessment continues to increase. One study found that less than 20% of patients had their genetic test ordered by a genetic counselor, and only approximately 50% of patients who underwent GT then discussed their results with a genetic counselor [18]. Given the limited accessibility of genetic counselors, decision support tools may offer an alternative for average-risk patients by providing similar information to that communicated in counseling sessions while preserving counseling resources for higher-risk patients [18]. Establishing the effectiveness of alternative strategies for both pretest counseling and return of results will be crucial to addressing the increased need for GT services as more patients are identified for BC genetic risk assessment.

Various studies have shown that risk perception, potentially more so than the GT result itself, may significantly influence patients' medical decision-making and associated clinical outcomes [21, 22]. However, it is crucial to recognize that the sole delivery of information does not equate to neither knowledge retention nor accurate risk perception [21]. These trends suggest that the way forward is to meet the patients at this level of discrepancy and introduce tools that help facilitate more accurate risk perception and provide support at every step of the diagnostic process.

To this end, Kukafka et al. developed and evaluated the web-based *RealRisks* decision aid (DA) for women to screen for GT eligibility and a complementary decision support tool called Breast Cancer Risk Navigation Tool (*BNAV*) for their primary care providers (PCPs) [23]. *RealRisks* includes interactive educational modules on breast cancer risk assessment, genetic testing, screening, and chemoprevention. A key feature of *RealRisks* is the ability to calculate a personalized breast cancer risk score via Fast Healthcare Interoperability Resources (FHIR). Within *RealRisks*, a user interface displays their electronic health record (EHR) data to patients, allowing them to review, add and modify their data before running the risk assessments to calculate 5-year, 10-year, and lifetime absolute invasive breast cancer risk according to the Breast Cancer Surveillance Consortium (BCSC) risk calculator [24, 25]. The Six-Point Scale (SPS) family history screener, as well as a detailed family pedigree, have also been incorporated into *RealRisks* to assess candidacy for GT and provide a foundation for follow-up discussions with healthcare providers [26].

In a cluster randomized controlled trial (RCT) of standard educational materials alone vs. in combination with *RealRisks* and *BNAV* among 187 women and 67 clinicians, respectively, there was a significant decrease in BC worry and perceived lifetime BC risk in the intervention compared to the control arm [23]. However, there was no significant increase in the primary endpoint of genetic counseling uptake at 6 months in the intervention vs control arm (19.8% vs. 11.6%, $p=0.14$) [23]. Standard materials included a brochure from our institution's breast cancer prevention clinic as well as materials from the Susan G. Komen Foundation on genetics and breast cancer in either English or Spanish [23].

Research to understand the experiences of women who underwent HBOC GT following exposure to DAs such as *RealRisks* is limited. Therefore, we conducted semi-structured interviews to identify how women enrolled in the RCT who opted for GT understood, interpreted, and acted upon their GT results. The aims of this study include: 1) examining patients' GT experience and its influence on subsequent actions pertaining to BC prevention and follow-up; and 2) identifying areas for improvement in *RealRisks* that would expand its focus from improved GT decision-making (*e.g.,* the decision

to test) to understanding and interpreting results from HBOC GT (*e.g.*, return of results).

Methodology

Recruitment

Individuals were recruited from the cohort of the parent study, described previously [23]. Eligible patients were aged 21–75 years, without a personal history of breast or ovarian cancer, no history of genetic counseling or testing for HBOC, eligible for *BRCA1/2* GT based on a validated family history screener [18], and ability to provide informed consent in English or Spanish [23]. Of 187 evaluable patients (101 in the intervention group and 86 in the control group), a total of 58 patients had received GT confirmed by the electronic health record (EHR) at 24 months following study enrollment and were thus eligible for this qualitative nested study. Contact information from enrollment in the prior study was utilized for recruitment. Baseline clinical and demographic information were also extracted from databases used in the parent study. Outreach was conducted by email and phone. Study procedures were approved by the Columbia University Irving Medical Center (CUIMC) Institutional Review Board. Patient eligibility and recruitment are detailed in Fig. 1.

Data collection

Two versions of the interview guide were developed corresponding to the participant's original assignment to the control or intervention arm in the parent study (Appendix A). Participants were further grouped based on their GT results: negative, positive, or variants of uncertain significance (VUS). Patients included in the parent study only underwent testing for *BRCA 1* and *BRCA 2*. Patients were categorized as having "unknown" results if there was no available documentation of their results and they were unable to corroborate their results themselves. Interviews were conducted in Spanish and English by a bilingual interviewer. The interviews took place over Zoom video conference and were audio recorded and transcribed. Demographic and BC risk factor data for each participant were collected from surveys administered during the parent study. Regarding clinical factors, having an "established healthcare provider" referred to patients having a healthcare provider whom they regularly saw for outpatient care.

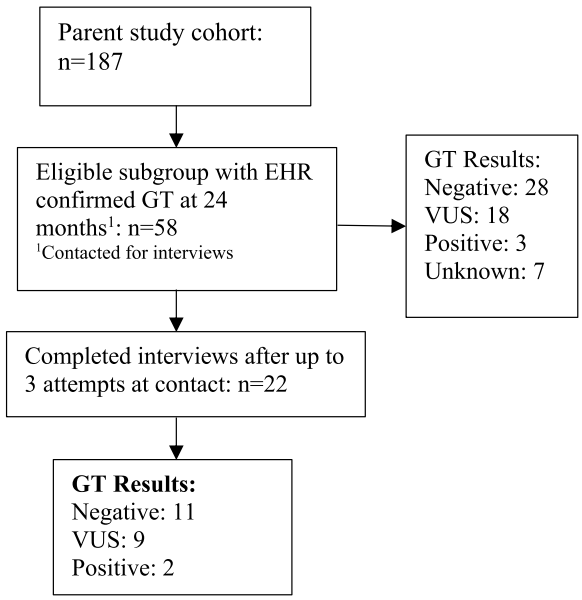

Fig. 1 Cohort selection flow diagram. EHR: electronic health record; GT: genetic testing; VUS: variant of uncertain significance

Codebook and qualitative data analysis

The research team developed a codebook using a deductive approach to identify themes that could provide insight into the experiences of women who underwent HBOC GT. The codebook was defined and organized prior to conducting interviews. The codebook further underwent a round of iterative refinement in which three transcripts coded using this system were reviewed by the coders to identify if additional codes were needed. Each code was assigned a definition with instructions regarding code application. The coding team was comprised of two experienced coders. Scott's Pi was tabulated as a measure of intercoder reliability at 0.624, indicating substantial agreement [27]. The qualitative data analysis software ATLAS.ti. was used to code transcripts by the research team. Interviews were analyzed using the following nine codes: 1) behavioral changes based on GT results, 2) communication of GT results, 3) experience receiving results, 4) initial reaction to GT results, 5) understanding/lack of understanding of GT results, 6) method of receiving GT results, 7) *RealRisk* references, 8) recommendations regarding GT, and 9) understanding of risk factors.

Quantitative statistical analysis

To compare the interviewed subgroup to the total eligible cohort, comparison of demographic characteristics was conducted using R Studio (Version 1.4.1717). Normality was determined using the Shapiro-Wilks normality test. Data for continuous variables are presented as medians (with interquartile range) and counts and percentages for categorical variables. The Kruskal–Wallis test and ANOVA were used to compare variables as appropriate. Categorical variables were compared using the Pearson chi-squared test. A p-value less than 0.05 was deemed significant for all statistical analyses.

Results

Of the 58 patients recruited from the parent trial who were eligible for this study, 22 (38%) were interviewed. Two interview transcripts were not included in the analysis due to data missingness. As shown in Table 1, a comparison of the interviewed patients to the total cohort revealed minimal inter-group differences except for family history of *BRCA1/2* pathogenic variants (PVs) (58.3% vs 18.2% with no family history, $p = 0.01$).

Of the 58 eligible patients, 29 (50%) had negative GT results, 19 (33%) had a variant of unknown significance (VUS), 3 (5%) had a PV, and 7 (12%) had unknown results (Table 1). Of the 22 participants who completed the interview, 6 (27%) had VUS results, 2 (9%) had a PV, and 11 (50%) had negative results. Half of those interviewed ($n = 11$; 50.0%) identified as Jewish. Self-reported racial identities of the interviewed participants included White ($n = 16$; 72.7%), Black/African American ($n = 5$; 22.7%), and Asian ($n = 1$; 4.5%). All participants had completed their high school education at a minimum. There were no significant differences between those who received the intervention and those in the control group among those interviewed (Table 2).

Qualitative results

Table 3 documents the frequency with which each code was applied across the interview transcripts. Five themes emerged from the transcripts: 1) preferences regarding communication of GT results; 2) lifestyle changes influenced by GT; 3) understanding and emotional reception of GT results; 4) utility and role of *RealRisks* in deciding to pursue GT; and 5) recommendations on how to improve the GT process. The organization of these codes into the resulting themes is documented in Fig. 2. Of note, there were no consistent patterns in themes reported between women in the intervention vs control groups.

Theme 1: preferences regarding communication of GT results

Regardless of the GT result, patients frequently expressed appreciation for the role of genetic counselors and particularly the opportunity to discuss their GT results in a face-to-face encounter. Among patients who provided a direct answer to "who was most helpful" ($n = 9$), genetic counselors and medical professionals were considered to be the "most helpful" in explaining their GT results (33.3% and 55.6%, respectively). In contrast, receiving results, even from a genetic counselor, over the phone was viewed as a stressful event. Comments associated with understanding or having few questions after receiving results were frequently associated with having worked with a genetic counselor in person in the setting of a structured visit. As one patient summarized:

> *"Well, I did follow up, you know, when I met with the counselor and they also suggested seeing somebody at the hospital, which I did. They were very informative."*

Patients who received PV results specifically cited that delivery of results over the phone was not favorable (n = 2). One of the participants expressed the following:

> *"It was pretty bad. I think hearing that by phone was definitely not what I was expecting. I was in the middle of a workday, and I got this random call, and this person started tellingme all these things. That was a little disappointing, I think for me."*

Another patient who had received notification of a PV over the phone similarly noted:

> *"…I think if I had an appointment where this would be told to me in person and I had like time to process the information and go over things, it would have been much easier."*

As one patient with a negative result alluded to, the negative emotions surrounding receiving PV results may be overwhelming, suggesting that an in-person approach may be favorable:

> *"I do understand that some people can get so clouded by the fact they possibly could have cancer, they may not even digest what they're telling you but my recommendation for sure is definitely keep doing the in-person, sitting down with the person and maybe holding their hand through the process of really understanding what that genetic test can be."*

Whereas patients with PVs noted dissatisfaction with the brevity and lack of support in receiving their results, especially by phone, patients with VUS and negative results more often expressed appreciation of the depth and time taken to fully explain their results to them. Moreover, those with VUS and negative results reported an overall more positive experience in receiving their GT results. One patient who had received the result of VUS explained:

> *"I think again having a counselor explain what they did, what the results mean, where does that put you in a risk bracket, it's highly helpful for me as a patient to know that okay, I know that maybe I don't need to take additional drastic changes or actions, but I should continue to do what I need to do, to go to the doctor to check every year, mammograms, and*

Table 1 Baseline participant characteristics stratified by interview participation

Characteristic	Interview not completed ($n=36$)	Interview completed ($n=22$)	p-value
Intervention (%)[a]	22 (61.1)	15 (68.2)	0.79
Demographics			
Age (median [IQR])	41.50 [31.75, 54.00]	40.50 [32.50, 47.00]	0.90
Race (%)			0.81
American Indian or Alaska Native	1 (3.3)	0 (0.0)	
Asian	1 (3.3)	1 (4.5)	
Black or African American	6 (20.0)	5 (22.7)	
White	21 (70.0)	16 (72.7)	
Multiracial	1 (3.3)	0 (0.0)	
Non-Hispanic (%)	19 (52.8)	15 (71.4)	0.27
Jewish ancestry (%)	9 (25.0)	11 (50.0)	0.10
Education			0.44
8–11 years (without graduating high school)	3 (8.3)	0 (0.0)	
High school graduation or GED	2 (5.6)	0 (0.0)	
Some college or university classes (but no degree)	3 (8.3)	3 (14.3)	
Associate or bachelor's degree	13 (36.1)	7 (33.3)	
Graduate degree, post-graduate degree, or professional degree	15 (41.7)	11 (52.4)	
Marital status (%)			0.41
Divorced/Separated	5 (13.9)	2 (9.1)	
Engaged	3 (8.3)	0 (0.0)	
Married	14 (38.9)	13 (59.1)	
Single, never married	13 (36.1)	7 (31.8)	
Widowed	1 (2.8)	0 (0.0)	
Clinical Factors			
Established healthcare provider (%)	35 (97.2)	21 (95.5)	1
Primary health insurance (%)			0.48
Medicaid	11 (30.6)	5 (22.7)	
Medicare	3 (8.3)	1 (4.5)	
Other	2 (5.6)	0 (0.0)	
Private	20 (55.6)	16 (72.7)	
Preferred language = Spanish (%)	9 (25.0)	1 (4.5)	0.10
Reproductive Factors			
Menarche age (%)			0.91
7–11 years	7 (19.4)	4 (18.2)	
12–13 years	21 (58.3)	14 (63.6)	
14 years or older	8 (22.2)	4 (18.2)	
Menopausal status (%)			0.55
Pre-menopausal	22 (61.1)	15 (68.2)	
Currently going through menopause	4 (11.1)	1 (4.5)	
Post-menopausal (i.e., not had a period for over 2 years)	9 (25.0)	4 (18.2)	
Unknown	1 (2.8)	2 (9.1)	
Age at first birth (%)			0.72
< 20 years	5 (13.9)	2 (9.1)	
20 to 24 years	7 (19.4)	2 (9.1)	
25 to 29 years	4 (11.1)	4 (18.2)	
30 years or older	7 (19.4)	6 (27.3)	
No births	13 (36.1)	8 (36.4)	
Family History of Cancer			
Family history of BC (%)			0.88

Table 1 (continued)

Characteristic	Interview not completed ($n=36$)	Interview completed ($n=22$)	p-value
Don't Know	1 (2.8)	1 (4.5)	
No	1 (2.8)	1 (4.5)	
Yes	34 (94.4)	20 (90.9)	
BC in mother (%)	15 (42.9)	12 (57.1)	0.45
BC in sister (%)	5 (14.3)	2 (9.5)	0.92
BC in grandmother (%)	9 (25.7)	6 (28.6)	1
BC in aunt (%)	25 (71.4)	8 (38.1)	**0.03**
BC in male family member (%)	0 (0.0)	5 (23.8)	**0.01**
Family history of OC (%)	16 (44.4)	6 (27.3)	0.30
Family history of BRCA variant (%)			**0.01**
Don't Know	12 (33.3)	16 (72.7)	
No	21 (58.3)	4 (18.2)	
Yes	3 (8.3)	2 (9.1)	
Genetic Testing Results			
GT Result (%)			0.11
Negative	18 (50.0)	11 (50.0)	
VUS	10 (27.8)	9 (40.9)	
Positive	1 (2.8)	2 (9.1)	
Unknown	7 (19.4)	0 (0.0)	

GT genetic testing, *BC* breast cancer, *OC* ovarian cancer

[a] Those randomized to the intervention group in the parent randomized control trial

I know that hopefully the genetics company is going to update me if new information or new technology changes the view of the results."

Overall, patients with PVs consistently reported a perceived benefit with face-to-face genetic counselor sessions, and patients with VUS or negative results were more often satisfied with the communication they received relative to patients who received PV results.

Theme 2: lifestyle changes influenced by GT

As indicated, most patients ($n=14$, 65%) did not change lifestyle behaviors in response to their negative and VUS GT. Those who did report modifications in daily behaviors as a response to their GT result most commonly described changes in diet, exercise, screening, and smoking. These lifestyle changes were reported by patients who received positive and negative results. Commonly identified risk factors among participants, regardless of GT result, were alcohol use, smoking, and family history. As one patient with a PV result described:

"I feel like there are so many epigenetic factors, so that comes into daily life and it'sreally made me more conscious about everything from eating well to sleeping well toexercising to making sure I follow through on some of the screening recommendations,so yeah I think it has had an impact on my daily life."

Both patients with PVs reported that receiving their results influenced family planning, with one patient stating:

"...we did go through IVF to select for non-BRCA genes in the embryos so yeah it had an impact on my family".

Regardless of if behavioral modifications were or were not implemented, most patients did understand that lifestyle factors, such as sleeping, exercise, and diet, can impact their BC risk.

Theme 3: understanding and emotional reception of GT results

In discussing the implications of their GT results, women expressed an understanding that a negative GT result or the absence of a PV does not guarantee a cancer-free lifetime ($n=15$, 75%). Patients who had received negative or VUS results commonly reported more positive emotions receiving their results compared to those with PVs. Nearly half of the VUS patients (44.4%) explicitly reported feeling relief on receiving their results. All

Table 2 Baseline participant characteristics stratified by parent study arm within subgroup that completed interviews

Characteristic	Control Arm ($n=7$)	Intervention Arm[a] ($n=15$)	*p*-value
Demographics			
Age (median [IQR])	41.00 [35.00, 42.50]	40.00 [33.00, 52.00]	1
Race (%)			0.60
American Indian or Alaska Native	0 (0.0)	0 (0.0)	
Asian	0 (0.0)	1 (6.7)	
Black or African American	1 (14.3)	4 (26.7)	
White	6 (85.7)	10 (66.8)	
Multiracial	1 (3.3)	0 (0.0)	
Non-Hispanic (%)	5 (71.4)	10 (66.7)	0.78
Jewish ancestry (%)	3 (42.9)	8 (53.3)	1
Education			0.39
8–11 years (without graduating high school)	0 (0.0)	0 (0.0)	
High school graduation or GED	0 (0.0)	0 (0.0)	
Some college or university classes (but no degree)	1 (14.3)	2 (14.3)	
Associate or bachelor's degree	1 (14.3)	6 (42.9)	
Graduate degree, post-graduate degree, or professional degree	5 (71.4)	6 (42.9)	
Marital status (%)			0.51
Divorced/Separated	0 (0.0)	2 (13.3)	
Engaged	0 (0.0)	0 (0.0)	
Married	4 (57.1)	9 (60.0)	
Single, never married	3 (42.9)	4 (26.7)	
Widowed	0 (0.0)	0 (0.0)	
Clinical Factors			
Established healthcare provider (%)	7 (100.0)	14 (93.3)	1
Primary health insurance (%)			0.73
Medicaid	2 (28.6)	3 (20.0)	
Medicare	0 (0.0)	1 (6.7)	
Other	0 (0.0)	0 (0.0)	
Private	5 (71.4)	11 (73.3)	
Preferred language = Spanish (%)	0 (0.0)	1 (6.7)	1.0
Reproductive Factors			
Menarche age (%)			0.68
7–11 years	1 (14.3)	3 (20.0)	
12–13 years	4 (57.1)	10 (66.7)	
14 years or older	2 (28.6)	2 (13.3)	
Menopausal status (%)			0.58
Pre-menopausal	6 (85.7)	9 (60.0)	
Currently going through menopause	0 (0.0)	1 (6.7	
Post-menopausal (i.e., not had a period for over 2 years)	1 (14.3)	3 (20.0)	
Unknown	0 (0.0)	2 (13.3)	
Age at first birth (%)			0.6
< 20 years	0 (0.0)	0 (0.0)	
20 to 24 years	1 (14.3)	0 (0.0)	
25 to 29 years	1 (14.3)	15 (100.0)	
30 years or older	3 (42.9)	0 (0.0)	
No births	2 (28.6)	0 (0.0)	
Family History of Cancer			
Family history of BC (%)			0.10
Don't Know	1 (14.3)	0 (0.0)	

Table 2 (continued)

Characteristic	Control Arm ($n=7$)	Intervention Arm[a] ($n=15$)	*p*-value
No	1 (14.3)	0 (0.0)	
Yes	5 (71.4)	15 (100.0)	
BC in mother (%)	3 (42.9)	9 (60.0)	0.3
BC in sister (%)	1 (14.3)	1 (6.7)	0.26
BC in grandmother (%)	3 (42.9)	3 (20.0)	0.13
BC in aunt (%)	1 (14.3)	7 (46.7)	0.15
BC in male family member (%)	1 (14.3)	4 (26.7)	0.29
Family history of OC (%)	3 (42.9)	3 (20.0)	0.544
Family history of BRCA variant (%)			0.53
Don't Know	4 (57.1)	12 (80.0)	
No	2 (28.6)	2 (13.3)	
Yes	1 (14.3)	1 (6.7)	
Genetic Testing Results			
GT Result (%)			0.81
Negative	3 (42.9)	8 (53.3)	
VUS	3 (42.9)	6 (30.0)	
Positive	1 (14.3)	1 (6.7)	
Unknown	0 (0.0)	0 (0.0)	

GT genetic testing, *BC* breast cancer, *OC* ovarian cancer

[a] Those randomized to the intervention group in the parent randomized control trial

Table 3 Frequency of code use across interview transcripts

Code	Frequency
Behavioral changes based on GT result	109
Communication of GT results	64
Experience receiving GT results	137
Initial reaction to GT results	31
Understanding/ lack of understanding of GT results	75
Method of receiving GT results	39
RealRisk references	59
Recommendations regarding GT testing	37
Understanding of risk factors	84

GT genetic testing

patients with a negative result described their feelings as related to relief and joy ($n=11$ out of 11, 100%):

> *"It was great. I was ecstatic. She was very happy. I was happy.";*

> *"When you have a family history of cancer, it's scary to think about and you never know what's in your future, so just the fact that I'm not predisposed to it was a little bit of a relief."*

Family experience with either cancer or GT was noted as influencing the patient's experience of receiving and processing GT results by five patients (22.7%). There was no explicit pattern regarding the nature of this influence based on the GT result. Some patients described family experience as positive, pointing towards the ability to tap into a sense of familiarity, support, and perception of information access ("*…helpful in providing some information"*). Instances in which family experience contributed negatively centered around increasing anxiety due to either apprehension of risk or past negative healthcare experience. For one patient who received a positive result, prior family experience with cancer offered some reassurance:

> *"I think like the reason why I was not scared was technically because I knew I was young, my mom had cancer when she was young and they found it right away and she was treated. So, I felt empowered that I knew I had this predisposition but I was aware of it and I could act upon."*

In contrast, another patient with a negative result explained:

> *"I guess I was more suspicious that I had to come in, I guess. I had an experience withgenetic testing for another family member and it was kind of like if it was negative, youjust found out and if it was positive, you had to come in so I think it provoked moreanxiety than maybe just getting the test results however and following up."*

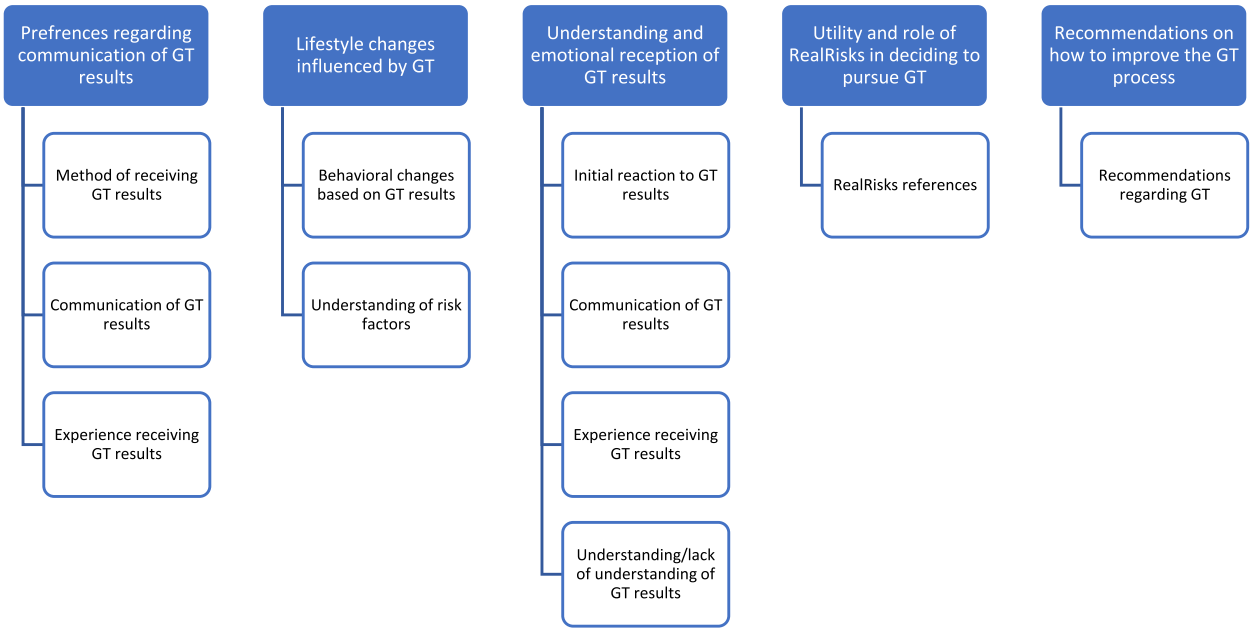

Fig. 2 Schematic illustrating the organization of deductively derived codes (column subheadings) that contributed to the elucidation of five distinct emergent themes (column headings)

Regardless of their results, patients expressed strong emotions, which varied from relief to anxiety, and it was evident that personal experience, especially with family members, can impact emotional responses.

Theme 4: utility and role of realrisks in deciding to pursue GT

Just over one-third of patients enrolled in this study reported that they had formed intentions to undergo GT prior to engaging with *RealRisks*. Specifically, eight participants of those interviewed (36.4%) stated that *RealRisks* did not have any impact on their decision to pursue GT as they had already made the decision to do so prior to accessing *RealRisks*. Comments such as: "*it didn't change my decision because I was going into it with the intention of getting the actual test*" were common. This sentiment was consistent across GT results.

Theme 5: recommendations on how to improve the GT process

Across GT results, patients frequently cited the need for more guidance regarding next steps after receiving their results. Specifically, over one-third of patients felt more information was needed regarding modifiable lifestyle factors and action items that can be implemented to reduce risk. These patients described a lack of GT awareness, understanding of risk factors, and education regarding the next steps. One participant commented:

"I feel that I mean I probably should be able to describe in detail what the known risk factors are, but I don't know that I could."

Another patient described:

"A lot of questions opened up about what were the implications of that, like I knew the overall implications, but I had a lot of questions about the details about what would bethe changes to my sort of day-to-day life and when would I have to start preventative screening and those things."

One specific comment expressed that this need is especially underscored among the Latino/Hispanic community:

"..it would help if the genetic counselor, the website [RealRisks] and the doctors empower the patient with more information in terms of what they can do for their lifestyle, even if it's not proven, especially in the Latino/Hispanic community, they are looking for solutions on what they can do."

On a similar note, participants (n = 2 out of 22, 9.1%) recommended that there be an increase in awareness of testing availability among patients, in order to expand the accessibility of GT. Specifically, one Latinx patient stated that:

"I think that a lot of people, even my own peers and my own community, don't have an understanding of the importance of getting genetic testing, the access

to it, the fact that in addition to evolution in the medical community, insurance has improved and kind of financial accessibility to it. Although I don't know that I necessarily would benefit from it, and it almost feels like more publicity, but more global and communal awareness around genetic testing. I do believe I'm part of a community and my larger community would certainly benefit from that."

In summary, explanations of actionable follow-up and risk factors are among the most common points of feedback.

Discussion

This qualitative interview study aimed to explore the salient factors that influenced the genetic testing (GT) experiences of women in order to increase appropriate *BRCA1/2* GT in the primary care setting. It is important to note that our decision support tool, *RealRisks,* in its current iteration did not include a module on return of results, and an additional purpose of conducting these interviews was to inform the development of such a module. This interview study confirms the need for a decision support aid that informs every step of the GT experience, including risk assessment, education, decision support, return of results, follow-up care, and lifestyle modification. Additionally, translating intention to actual behavior (e.g., to pursue GT when indicated) requires favorable attitudes and perceived behavioral control, which are modifiable factors that could be targeted within *RealRisks* and similar applications to help at-risk individuals recognize their risk status, pursue appropriate GT, and engage in appropriate risk-mitigating actions [28].

After deciding to pursue or decline GT, both the factual understanding of test results and the emotional context in which results are received are critical influencers of subsequent action. Unlike the GT result itself—which has been found to have varying significance independently—the patient's risk perception has been consistently found to be directly associated with the patient's medical decision-making (including the decision to undergo prophylactic surgery) [21, 29]. Thus, empowering the patient with the knowledge and context with which to internalize the implications of their results and form accurate risk perception represents a powerful opportunity to mediate subsequent health behaviors. This finding is especially relevant given that certain women with negative *BRCA1/2* testing are still at high risk for breast cancer, and few studies have focused on providing guidance to this specific subgroup [30]. While this nested study was too small to extrapolate any larger conclusions about the utility of *RealRisks* in this regard, it provides helpful targeted guidance to inform the iterative refinement process.

Among patients who received negative test results, most understood that this result does not guarantee that they will never develop cancer. In this sense, patients with negative test results demonstrated an adequate understanding of lifetime risk and the role of non-genetic factors in contributing to risk. Nevertheless, the most frequently reported emotions surrounding variant of uncertain significance (VUS) and negative test results were joy and relief. Prior studies identified that BC history and prior experience with cancer at large in the healthcare setting were also associated with the emotional reception of GT results [31]. However, no significant trends in these factors influencing reactions to results were identified in this cohort.

Among patients dissatisfied with the communication of their GT results, specific complaints included feeling poorly supported, abrupt delivery of information, and requiring more guidance for subsequent steps in medical management. In contrast, those who had positive experiences specifically referenced the time taken to explain the patients' risk based on their personal information. While positive experiences tended to correlate with in-person encounters and negative experiences with phone encounters, these patterns may also be a testament to the nature of these discussions. Of note, patients who did not receive in-person communication of results in this study were only contacted over the phone, as opposed to a videoconferencing platform. This finding may support the need for additional patient support regardless of GT result [32]. The dissatisfaction with phone delivery of GT results noted in this also study highlights the need for ongoing evaluation of the efficacy of telemedicine, which has also become increasingly commonplace since the COVID-19 pandemic.

In our study, patients with identified PVs tended to report a less thorough and supportive GT experience than those with negative/VUS results. This observation may indicate a paradoxical tendency to focus more on ensuring negative/VUS patients adequately understand the nuances of their results rather than the sensitivity of communicating positive results. This aligns with prior studies in which patients had a more accurate perception of positive results, perhaps due to their more apparent or ostensibly straightforward implications [33]. This study will serve to inform future work to address the persistent need for alternative personalized, accessible genetic service resources [34–38].

Another particularly relevant theme that emerged was the need for more guidance regarding the next steps after receiving GT results. In this group, uncertainty regarding next steps was primarily reported among participants

with negative results. Namely, a recurrent thread centered around patients voicing a need for both more information and clarity regarding medical follow-up and daily risk reduction practices, especially in the Latinx/Hispanic community. While the relationship between race/ethnicity and BC knowledge has been contested across literature, there have been indications that race/ethnicity is, at the very least, a relevant factor in predicting patient activation, testing, and follow-up [39]. A recent sequential mixed-methods study found that taken cumulatively, non-Hispanic Whites and individuals with greater health literacy at baseline had a more accurate understanding of their BC risk [40].

Among the few patients in our study who reported behavioral changes in response to their GT results, lifestyle modifications regarding diet, exercise, cancer screening, or substance use were most common. Accordingly, when asked to describe known BC risk factors, patients also commonly referenced weight, dietary habits, smoking/drug use, and family history. These patterns suggest that patients are willing to apply their understanding of risk factors to actionable lifestyle changes. Interestingly, both patients with positive and negative results reported being motivated to implement lifestyle changes where applicable, indicating that factors other than the GT result may serve as an impetus in behavior modification. Namely, this motivation may be driven by the experience of GT itself or the perception of being at higher risk for BC (regardless of GT result) given that these participants had received the recommendation to undergo GT. These findings underscore the importance of providing the knowledge required to implement these changes, especially since numerous respondents self-reported a worrisome knowledge gap.

Overall, the literature suggests that the problem of under-utilization of GT is complex, with barriers at multiple levels, and that there needs to be more widely implemented, long-term interventions to address this issue. While it may seem intuitive to focus on the breadth of genetic counseling/education services to increase GT uptake, various studies have shown that knowledge gain and retention are relatively limited after counseling [41, 42]. Thus, while there is a call for better communication of genetic risk via counseling, multiple studies have shown that it is the individual perception of risk, even given adequate risk communication, that more directly influences results [21, 29, 41]. Therefore, accounting for factors that impact risk perception in conjunction with information delivery through targeted decision support aids may promote health equity in GT and improve clinical outcomes.

We acknowledge several limitations to our study. Namely, the small size of the interviewed cohort limits the scope of experiences and perspectives solicited, thus limiting the generalizability of our findings. Additionally, all participants who were interviewed had decided to pursue GT prior to engaging with *RealRisks*, preventing us from obtaining an accurate understanding of the role of *RealRisks* in the initial decision to pursue GT. Moreover, the interviewed subgroup was enriched for patients with a family history of breast cancer (90.9%), potentially skewing the perspectives represented. Of note, while we were able to identify participants who did receive genetic counseling based on their reference to the experience in the interview, we do not have access to sufficient data to directly compare frequency of genetic counseling across groups. Strengths of our study include quantitative data to better understand the relevant clinical and demographic context of our patient population. Additionally, the qualitative analysis was conducted by an interdisciplinary team of researchers who lent their respective expertise in medicine, public health, and biomedical informatics to thoroughly analyze and parse the interview data.

Conclusion

Overall, we found that patients expressed preferences for verbal explanations of their GT results to facilitate a more personal, supportive testing experience. While most patients did not change lifestyle behaviors in response to their GT results, there was a consistent call for further guidance and navigation following the initial discussion of GT results. Regarding the applicability of *RealRisks*, interviews have elucidated the need for a return of results module, directly informed by the content of this interview study. This module will provide the patient with critical context as to the medical implications of the participant's GT result, and targeted action steps including risk reduction strategies, and specific steps for follow-up where necessary.

Supplementary Information

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Supplementary Material 1.

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Authors' contributions
AV wrote the primary manuscript text, completed quantitative analysis, produced corresponding tables and figures, and contributed to qualitative analysis of patient interviews. BS conducted patient interviews, coded interview transcripts, and contributed to qualitative analysis of patient interviews. YEF, HW, AM, and SU contributed to qualitative analysis and revision of the manuscript. KC and RK oversaw the completion of the study, guided development of methodology and results analysis, and provided comprehensive revisions for the manuscript.

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Data availability
The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate
Study materials and procedures were approved by the Columbia University Irving Medical Center (CUIMC) Institutional Review Board (IRB-AAAR3676). Individual informed consent was granted by interviewed study participants through an online informed consent form.

Consent for publication
Not applicable.

Competing interests
The authors declare no competing interests.

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