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Acceptability and Outcomes of Multigene Panel Testing among Young Black Breast Cancer Survivors

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Keywords

breast cancer; survivorship; genetic testing; update testing

Background

The majority of inherited breast cancer (BC) is attributed to pathogenic variants in *BRCA1* and *BRCA2*.¹ The remaining are due to pathogenic variants in other individually rarer “non-*BRCA*” genes (e.g., *PALB2*, *CHEK2*, *ATM*).² “Multigene panel testing” (MGPT) assesses for variants in both *BRCA* and non-*BRCA* genes and is the gold standard for BC genetic testing (GT).³

MGPT with inclusion of the *BRCA* genes became widely available in 2013. Women tested prior to 2013 who were found not to carry pathogenic *BRCA* variants are eligible for “update” MGPT.

MGPT may be particularly important for young Black women; statewide data from Florida demonstrate a higher than expected 12.4% *BRCA* mutation frequency in this group.⁴ The contribution of non-*BRCA* mutations to BC disparities is less established.

Prior research demonstrates high levels of interest in MGPT.⁵ However, less than 1% of the study sample identified as Black. Furthermore, MGPT outcomes were not examined. To our knowledge, no studies report on MGPT acceptability or outcomes in Black BC survivors. This study addresses these gaps.

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Methods

Procedures and Participants

Participants were recruited from a parent study of early onset BC in premenopausal Black women.⁴ Recruitment methods for the parent study are detailed elsewhere.⁴ Eligible participants were: (1) Black women; (2) diagnosed with BC $\leq$ 50 years old; (3) diagnosed 2009–2012; and (4) living in Florida at diagnosis. Parent study participation included (1) telephone-based genetic counseling and (2) *BRCA* GT with mail-based saliva collection.

Of 882 eligible individuals contacted, 480 (54%) consented to participate, and 396 (83%) completed *BRCA* testing. Pathogenic *BRCA* variants were identified in 49 women (12%), 86 (22%) received a variant of uncertain significance (VUS) result, and 261 (66%) tested negative.⁴

Update MGPT with a 37-gene panel was conducted for all parent study participants with available DNA. Of the 347 women with negative or VUS results on original *BRCA* GT, MGPT identified pathogenic variants in 12 known breast and ovarian cancer genes for 20 women (6%). For this pilot study, these 20 women were mailed a letter informing them about availability of MGPT results via telephone-based genetic counseling. If no response was received after three weeks, a study team member telephoned the participant up to five times.

Participants completed self-report surveys at baseline and 1-month post-genetic counseling; upon completion, participants received a \$20 gift card.

Measures

Knowledge about MGPT: Five items; each correct answer received one point (range: 0–5).

Perceived risk of BC recurrence: Rated on a 100 point scale (0=no chance to 100=definitely).

Cancer-related distress: Impact of Event Scale-Revised⁶ (higher scores=more BC-related distress).

Satisfaction with update testing: Genetic Counseling Satisfaction Scale (GCSS)⁷(range 0–4; scores $\geq$ 2 indicated satisfaction with MGPT).

Analytic Strategy

Descriptive statistics characterized the acceptability of MGPT, defined a priori as: (1) $\geq$ 60% receipt of MGPT results and (2) $\geq$ 75% satisfaction with MGPT. Wilcoxon signed-ranks tests examined pre- to post-MGPT changes in knowledge, perceived BC risk, and distress. All analyses were conducted using SPSS (version 25, IBM).

Results

Eleven women (55%) were unable to be contacted, 1 (5%) declined, and 8 (40%) consented and completed the baseline assessment (Table 1).

Six (67% of contacted participants) received MGPT results. Of them, five (83%) completed the 1-month follow-up. All participants (100%) reported satisfaction with MGPT (GCSS 2).

There were no pre-to-post differences in knowledge ($p=0.581$) or perceived BC risk ($p=0.317$) (Table 2). There was a trend towards a pre-to-post increase in distress ($p=0.066$).

Conclusions

Per clinical practice guidelines, women tested for single inherited cancer genes (i.e., *BRCA*) should receive update MGPT.⁸ This study provides insight into the acceptability and short term impact of MGPT among young, Black BC survivors.

Six women expressed interest in receiving MGPT results. This is 30% of eligible participants, but 67% of those contacted. The registry-based recruitment strategy presented challenges in contacting participants, as patients frequently relocate and are often lost to follow-up. Thus, the context in which this study was conducted may result in an underestimate of patients' interest in MGPT results.

Consistent with prior studies of *BRCA* GT,⁷ participants reported high levels of satisfaction with MGPT. There were no pre-to-post differences in knowledge or perceived risk; this may be due to lack of power, or reflect an actual lack of change. There was a trend toward increased cancer-related distress from pre-to-post.

The study is limited by the small sample size, limiting interpretation of findings. Adequately powered studies are needed in order to confirm or refute the observed results.

In conclusion, this pilot study is among the first to prospectively assess acceptability and early outcomes of MGPT. We were unable to establish contact with over 50% of eligible participants; this may present a challenge to widespread dissemination of MGPT in underserved populations. Most contacted patients elected to receive MGPT results and reported satisfaction with MGPT; thus, MGPT is acceptable to young, Black BC survivors.

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References

1. Chen S, Parmigiani G. Meta-analysis of BRCA1 and BRCA2 penetrance. *Journal of Clinical Oncology*. 2007;25(11):1329. [PubMed: 17416853]
2. Ellsworth RE, Decewicz DJ, Shriver CD, Ellsworth DL. Breast cancer in the personal genomics era. *Current genomics*. 2010;11(3):146–161. [PubMed: 21037853]
3. Slavin TP, Niell-Swiler M, Solomon I, et al. Clinical Application of Multigene Panels: Challenges of Next-Generation Counseling and Cancer Risk Management. *Front Oncol*. 2015;5:208. [PubMed: 26484312]
4. Pal T, Bonner D, Cragun D, et al. A high frequency of BRCA mutations in young black women with breast cancer residing in Florida. *Cancer*. 2015;121(23):4173–4180. [PubMed: 26287763]

5. Vicuña B, Delaney HD, Flores KG, et al. Preferences for multigene panel testing for hereditary breast cancer risk among ethnically diverse BRCA-uninformative families. *Journal of community genetics*. 2018;9(1):81–92. [PubMed: 28971318]
6. Weiss DS. The impact of event scale: revised In: *Cross-cultural assessment of psychological trauma and PTSD*. Springer; 2007:219–238.
7. DeMarco TA, Peshkin BN, Mars BD, Tercyak KP. Patient satisfaction with cancer genetic counseling: a psychometric analysis of the Genetic Counseling Satisfaction Scale. *Journal of genetic counseling*. 2004;13(4):293–304. [PubMed: 19736695]
8. National Comprehensive Cancer Network (NCCN). Genetic/Familial High-Risk Assessment: Breast and Ovarian (Version 2.2019). NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) 2018; https://www.nccn.org/professionals/physician_gls/pdf/genetics_screening.pdf. Accessed November 12, 2018.

Table 1.

Participant sociodemographic characteristics (N=8).

	Mean (SD)	N (%)
Age (years)	50.1 (8.5)	
Born in United States		7 (87.5)
Partnered		5 (62.5)
High School Diploma/GED		7 (87.5)
Employed		3 (37.5)
Income \$35k/year		4 (50.0)
Privately insured		4 (50.0)

Table 2.

Results of Wilcoxon signed-ranks tests (N=8).

Variable	Mean (SD)		Z	p
	Pre	Post		
MGPT knowledge	1.5 (1.2)	1.2 (0.4)	-0.552	0.581
Perceived BC risk	38.8 (35.6)	37.5 (25.0)	-1.000	0.317
Cancer-related distress	16.0 (15.2)	18.2 (15.4)	-1.841	0.066