Re-evaluation of Hereditary Cancer Screening and Testing at an Urban Breast Cancer Survivorship Program - A Pilot Study

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Introduction

Current national guidelines recommend consideration of genetic testing for all newly diagnosed breast cancer (BC) patients (pts). However, hereditary cancer predisposition genetic testing has been historically underutilized for many reasons, including limitations of prior National Comprehensive Cancer Network (NCCN) guidelines, insurance/financial barriers, and emotional burden of diagnosis and testing. Thus, many BC survivors did not receive current standard of care genetic testing at diagnosis. Benefits of germline genetic testing and identification of pathogenic variants (PV) in late BC survivorship include eligibility for enhanced screening for other malignancies and potential cascade testing of family members. We conducted a pilot study evaluating interest in and uptake of genetic testing among BC survivors.

Methods

Charts of pts seen at Montefiore Einstein Comprehensive Cancer Center BC survivorship program from January 1, 2023 to December 31, 2023 (n=551) were reviewed. All pts were at least 5 years from diagnosis and completed BC treatment. Pts who had not previously received multi-gene panel testing were contacted by telephone to determine interest in genetic counseling and testing. The benefits of genetic counseling were also formally discussed with pts by the clinician caring for them at their annual survivorship visit or person distributing survey.

References

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Results

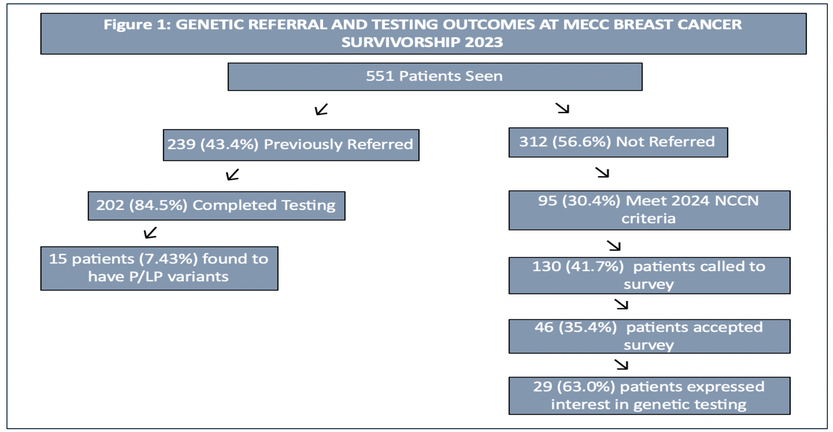


Figure 1: 130 with no prior testing were called for telephone survey, prioritizing those previously referred but not tested, and those who met current NCCN guidelines for testing. 46 pts completed the survey, 26 expressed interest in genetic testing.

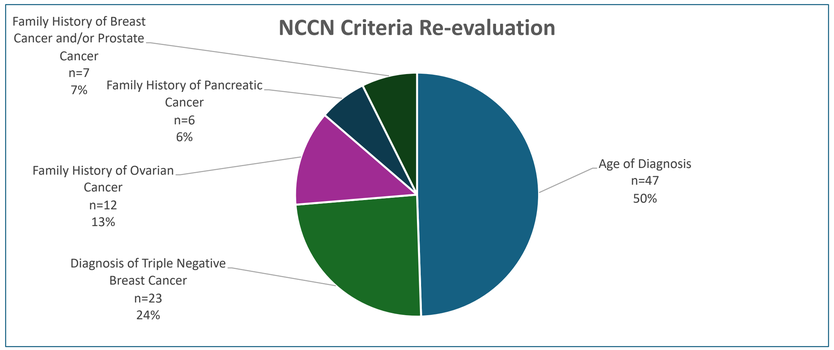


Figure 2: Of pts who did not previously undergo testing, 95 (30%) met 2024 NCCN criteria for testing by age at diagnosis, diagnosis of triple negative BC, family history (FH) of ovarian cancer, 6 by FH of pancreatic cancer, and 7 by FH of BC and/or prostate cancer.

Figure 3: Results of testing include *BRCA1* PV (n=1), *TP53* PV (n=1), MUTYH PV (n=1). *Age of diagnosis refers to age at first breast cancer diagnosis.

Gene	Genetic Variant	Age at diagnosis (years)*	Age at referral (years)	Personal History	Family History
MUTYH	p.G396D	54	64	- Breast Cancer: Stage IIB left IDC, ER-/PR-/HER2- - Endometrial cancer: Stage IA Uterine serous carcinoma	- Maternal grandmother: ovarian cancer - Maternal cousin: ovarian cancer
TP53	p.R213L	32	50	- Breast Cancer: Left Paget's disease and DCIS, ER-/PR-; Stage IIA right IDC, ER-/PR-/HER2-	- Daughter: breast cancer - Maternal aunt: pancreatic cancer
BRCA1	Del exon 20	62	63, 70	- Breast Cancer: Stage I left IDC, ER-/PR-/HER2-	- Sister, maternal half-sister, maternal half-niece, paternal aunt: breast cancer

Conclusions

Our study demonstrates feasibility of and interest in germline genetic testing in late BC survivorship. Clinicians caring for BC survivors who have not undergone genetic testing should discuss benefits of testing for patients and their families, as well as changes in guidelines and insurance coverage. Next steps include completing genetic consultation and testing for referred patients and continuing to reach out to all pts without prior genetic testing or with outdated testing.

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