Real-world adherence to germline genetic testing guidelines in breast cancer: patterns and disparities in a community oncology setting

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Introduction

- Germline genetic testing is essential in personalized breast cancer care, guiding surveillance, treatment decisions, and risk-reduction for patients and their families.
- Pathogenic variants in genes such as BRCA1/2, TP53, PTEN, STK11, CDH1, and PALB2 increase hereditary breast cancer risk and may signal susceptibility to additional cancers.
- The NCCN guidelines recommend testing patients with early breast cancer and specific high-risk features, including:
  - 1. Breast Cancer diagnosed at <50 years old
  - 2. Triple negative breast cancer, male breast cancer, multiple primary breast cancers
  - 3. Concerning family history
- However, adherence to these guidelines in real-world community settings, and the role of social determinants, such as race, remain incompletely understood.

Objectives

- We evaluated adherence to NCCN genetic testing guidelines and explored social determinants associated with testing completion in a Southeastern Virginia breast cancer population.

Methods and Materials

- A retrospective chart review was performed for patients diagnosed with breast cancer at Sentara Norfolk General Hospital between June 1, 2019 and June 1, 2024. Genetic testing eligibility was assessed according to NCCN Guidelines Version 1.2025.

Sample

- The final sample included 89 patients (88 female, 1 male) with a mean age of 55.5 years (SD = 7.1).
- Most patients were White (70%), followed by Black patients (25%).
- The majority presented with early-stage disease: 84% Stage IA, 11% Stage IB, and 5% Stage II.
- Most were married (64%), with 23% single and 9% divorced.
- Regarding NCCN testing eligibility, 28% qualified based on family history, 20% based on age, and 11% due to cancer subtype.

Results

- Genetic testing was ordered for most patients (75%, n=67), and 65 ultimately completed testing.
- Fisher's Exact tests showed no significant associations between social determinants of health and testing completion (all $p > .05$).
- Although subgroup comparisons suggested possible differences in completion rates among eligible patients (Table 2), none reached statistical significance.

Limitations

- Retrospective design limits causal inference and relies on accuracy of medical record documentation.
- Single-center study may reduce generalizability to
- Sample size limits statistical power
- Incomplete or unknown testing statuses for some patients may underestimate true disparities.
- Potential unmeasured confounders, including socioeconomic status, provider referral patterns, and patient-reported barriers, were not captured.

Results

Table 1. Percentage and total count of all individuals who received or did not receive testing, by eligibility (N = 89)

	Received testing	%	Did not receive testing	%	Unknown	%
Eligible	46	51.70	6	6.60	1	1.10
Ineligible	19	21.30	14	15.80	3	3.50
Total tested	65	73.00	20	22.40	4	4.60

Table 2. Testing rates by subgroup for eligible patients only (n = 53)

	Received testing	%	Did not receive testing	%	Unknown	%
Cancer stage						
IA	38	88.40	4	9.30	1	2.30
IB	6	75.00	2	25.00	0	0.00
II	2	100.00	0	0.00	0	0.00
Race						
Black	10	**76.90**	3	23.10	0	0.00
White	33	**91.70**	2	5.60	1	2.80
Asian	1	50.00	1	50.00	0	0.00
American Indian	1	100.00	0	0.00	0	0.00
Unknown	1	100.00	0	0.00	0	0.00
Marital status						
Married	31	**88.60**	3	8.60	1	2.90
Single	7	**70.00**	3	30.00	0	0.00
Divorced	5	100.00	0	0	0	0.00
Widowed	3	100.00	0	0	0	0.00
Criteria met						
Family history	23	**92.00**	2	8.00	0	0.00
Age	15	**83.30**	2	26.70	1	5.60
Cancer type	8	80.00	2	20.00	0	0.00

Discussion

- In this community-based breast cancer cohort, adherence to NCCN germline testing guidelines was strong, with most patients receiving or being offered appropriate testing. This demonstrates that high-quality genetic evaluation can be delivered reliably in non-academic settings.
- Subgroup patterns, however, suggest potential emerging disparities. Black patients completed testing at lower rates than White patients, and single patients were less likely to complete testing than married individuals. While these differences were not statistically significant, they may reflect barriers related to access, communication, or social support.
- These trends highlight the need for targeted efforts to ensure equitable uptake of genetic services as germline testing increasingly informs treatment, surveillance, and family risk assessment.

Conclusions

- Overall compliance with NCCN germline genetic testing recommendations was high in this cohort.
- No statistically significant associations between social determinants and test completion were found; however, subgroup patterns suggest important disparities.
- Black patients and single patients showed lower completion rates, pointing to potential inequities in access, navigation, or communication that warrant further investigation.
- These findings reinforce the need for ongoing quality-improvement initiatives to ensure equitable genetic counseling and testing across all patient populations.

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