

Evaluating the Rate of Genetic Testing in Newly Diagnosed Breast Cancer Patients

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Background

Genetic testing has become an important tool for optimizing breast cancer management for newly diagnosed breast cancer patients as the result can influence surgical decision. In addition, identifying mutations in a breast cancer patient is important for surveillance of other cancers and has important implications for other family members¹. Studies have demonstrated a wide range of uptake of genetic testing varying between 27-70%. The aim of this study is to identify our institutional rate of genetic testing in newly diagnosed operative breast cancer patients who met the criteria for testing based on NCCN guidelines^{2,3}. We hypothesize our multidisciplinary clinic model with genetic counselor on-site at our NCI-designated Comprehensive Cancer Center would result in a higher rate of genetic testing than currently reported.

Methods

- Retrospective single institution chart review
- Inclusion Criteria: newly diagnosed patients with stage 0-III breast cancer between January 2018 and February 2019
- Exclusion Criteria: diagnosis of recurrent or metastatic breast cancer
- Evaluated patients' eligibility for genetic testing according to NCCN guidelines at the time of their diagnosis and the rate of genetic testing.

Results

Study Population:

- 229 newly diagnosed operative breast cancer patients included in study
- Mean age at diagnosis was 61.9 years

Genetic Testing:

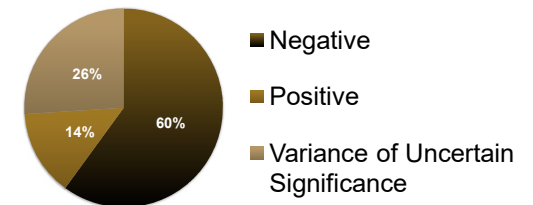
- 133 patients (58%) met criteria for genetic testing
- 116 patients (87%) who met criteria were offered genetic testing
- 93 patients (70%) who met criteria for testing, underwent genetic testing
- 20 patients (15%) who met criteria declined testing
- 4 patients who did not meet criteria for testing underwent genetic testing
- Testing Results: 57 patients (60%) tested negative, 13 (14%) positive, 24 (26%) Variance of uncertain significance.
- No variability between surgeons in genetic testing eligibility ($p=0.30$) or rate of testing ($p=0.72$)
- Patients who met criteria for testing were younger than those who did not meet eligibility (57.7 years vs. 67.7 years, $p < 0.001$)

References:

1. Chiba A, Hoskin TL, Hallberg EJ, et al. Impact that Timing of Genetic Mutation Diagnosis has on Surgical Decision Making and Outcome for BRCA1/BRCA2 Mutation Carriers with Breast Cancer. *Ann Surg Oncol*. 2016;23(10):3232-3238. doi:10.1245/s10434-016-5328-7
2. Knerr S, Bowles EJA, Leppig KA, Buist DSM, Gao H, Wernli KJ. Trends in BRCA test utilization in an integrated health system, 2005–2015. *J Natl Cancer Inst*. 2019; 111(8):djz008.
3. Childers CP, Childers KK, Maggard-Gibbons M, Macinko J. National Estimates of Genetic Testing in Women With a History of Breast or Ovarian Cancer. *J Clin Oncol*. 2017;35(34):3800-3806.

Rate of Genetic Testing				
	Total # Patients	Tested (%)	Positive Mutation (%)	VUS (%)
Eligible for Testing	133	93 (70%)	12 (13%)	23 (25%)
Non-Eligible for Testing	96	4 (4.2%)	1 (25%)	1 (25%)

Genetic Testing Results



Conclusions

Genetic testing has increasingly become an important part of breast cancer treatment. Testing was offered to 87% of patients who met criteria. Our rate of testing in those who met the criteria was 70%, which was well above average rate reported. In our clinic, we have a multidisciplinary clinic model where our genetic counselors are on-site and available to evaluate the patient at the time of the initial surgeon's visit. Streamlining the process of genetic counseling and testing at the time of surgical consultation may be a model to increase the rate of genetic testing.