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Many Breast Cancer Patients Not Receiving Genetic Evaluation

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A substantial number of breast cancer patients who could benefit from genetic testing are not being tested, and many others are not being counseled.

A new study shows that in a large cohort of more than 1700 early breast cancer patients, 47.4% were not tested. Although the majority of patients did report having some type of genetic discussion, only half of those who were not tested received any discussion about genetics.

The study was published online March 12 in the *Journal of Clinical Oncology*.
"Patients diagnosed with breast cancer need comprehensive patient-centered communication and decision making," said study author Steven J. Katz, MD, MPH, professor of general medicine and of health management and policy at the University of Michigan, Ann Arbor.
He explained that the "whole process needs to be slowed down, as too often its considered a medical emergency, especially by patients.

"Both patients and doctors need time to collect all the information, including genetic predisposition, in those at higher pretest risk of mutations," Katz told *Medscape Medical News*. "Doctors need to be better trained at counseling and integrating genetic counseling into treatment decisions. Treatment of cancer is largely focused on the biological subtype of the diagnosed cancer, while genetic predisposition plays a much smaller role in surviving cancer."
The authors note that genetic counseling is indicated for breast cancer patients who have an elevated pretest risk of harboring a pathogenic mutation. About one third of newly diagnosed patients do have a higher risk for a genetic mutation, as determined on the basis of their having a family history of cancer, their ancestry, and/or tumor characteristics.
But as testing is becoming more extensive, genetic risk evaluation — including counseling and genetic testing — is currently chaotically deployed into practice, Katz noted.
"Cost is not generally a problem, because the cost of testing is decreasing very quickly. The problem is the clinical utility of the testing today for patients with breast cancer, as there is legitimate clinical uncertainty about its role in treatment decision making," he said.

Testing and Counseling Uneven

Katz and his colleagues note that ideally, counseling should take place prior to surgery, because bilateral mastectomy is one of the options for risk reduction in this population. But putting genetic counseling into practice can be challenging, they note.

Information about integrating genetic counseling into community practices for newly diagnosed breast cancer patients is limited. In this study, they examined the patterns and correlates of discussion along with patient assessments about the information they received.
Surveys were sent to a large, diverse population of women aged 20 to 79 years with favorable-prognosis breast cancer who were identified from the SEER database of Georgia and Los Angeles County as having newly diagnosed ductal carcinoma in situ or invasive breast cancer. The surveys were linked to SEER

clinical data and genetic test results. The cohort available for analysis included 1711 women with indications for formal genetic risk evaluation.
Of the women who were tested, 29.7% only received testing for *BRCA1/2*; 22.9% underwent a multigene panel test (representing 43.5% of those tested).
Of the patients who underwent testing, 14.0% received results indicating "variant of unknown significance (VUS)" only, and 8.6% were found to have a pathogenic mutation. The remainder (77.4%) received negative results.
Overall, nearly three quarters (74.6%) of the cohort received some type of genetic counseling: 43.5% received formal counseling, and 31.1% had a physician-directed discussion.
Genetic counseling was far less prevalent among those who were not tested. Only 22.6% received some type of formal counseling, and 28.0% had a physician-directed discussion. Conversely, almost all patients who were tested reported that they had received some form of genetic counseling (96.4% of those whose test results were negative, and 94.9% of those whose results indicated pathogenic mutations or VUS). About two thirds reported having received formal counseling (60.5% of those with negative test results, and 67.9% of those with pathogenic mutations or VUS).

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Breast Cancer

Gaps in Receipt of Clinically Indicated Genetic Counseling After Diagnosis of Breast Cancer

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Abstract

Purpose

Little is known about the extent to which genetic counseling is integrated into community practices for patients newly diagnosed with breast cancer. We examined the receipt of clinically indicated genetic counseling in these patients.

Patients and Methods

We surveyed 5,080 patients between the ages of 20 and 79 years, diagnosed from July 2013 to August 2015 with early-stage breast cancer and reported to the SEER registries of Georgia and Los Angeles County. Surveys were linked to SEER clinical data and genetic test results. The study sample (N = 1,711) comprised patients with indications for formal genetic risk evaluation.

Results

Overall, 47.4% did not get tested, 40.7% tested negative, 7.4% had a variant of uncertain significance only, and 4.5% had a pathogenic mutation. Three quarters (74.6%) received some form of genetic counseling (43.5%, formal counseling and 31.1%, physician-directed discussion).

Virtually all tested patients (96.1%) reported some form of genetic discussion (62.2%, formal counseling and 33.9%, physician-directed discussion). However, only one half (50.6%) of those not tested received any discussion about genetics. Younger women were more likely to report some type of counseling, controlling for other factors: odds ratio, 4.5 (95% CI, 2.6 to 8.0); 1.9 (95% CI, 1.1 to 3.3); and 1.5 (95% CI, 1.0 to 2.3) for women younger than 50 years of age, 50 to 59 years of age, and 60 to 69 years of age versus those 70 years of age and older. Patients' assessments of the amount of information they received about whether to get tested were similarly high whether they were counseled by a genetics expert or by a physician only (80.8% *v* 79.4% stated information was just right, $P = .59$).

Conclusion

Less than one half (43.5%) of patients with clinical indications received formal genetic counseling. There is a large gap between mandates for timely pretest formal genetic counseling in higher-risk patients and the reality of practice today.