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Physicians' misunderstanding of genetic test results may hamper mastectomy decisions

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A recent survey of over 2,000 women newly diagnosed with breast cancer found that half of those who undergo bilateral mastectomy after genetic testing don't actually have mutations known to confer increased risk of additional cancers, according to a study by researchers at the Stanford University School of Medicine and four other U.S. medical centers.

Instead the women had what are known as variants of uncertain significance, or VUS, that are often eventually found to be harmless. A bilateral mastectomy is a surgical procedure in which both of a woman's breasts are removed after a diagnosis of cancer in one breast.

The finding highlights the need for genetic counselors to help both patients and physicians better understand the results of genetic testing intended to determine a woman's risk for cancer recurrence or for developing a separate cancer in her ovaries or unaffected breast.

"Our findings suggest a limited understanding among physicians and patients of the meaning of genetic testing results," said Allison Kurian, MD, associate professor of medicine and of health research and policy at Stanford. "Clinical practice guidelines state that variants of uncertain significance should not be considered to confer high cancer risk, and that patients with these variants should be counseled similarly to a patient whose genetic test is normal. However, many of the physicians surveyed in our study stated that they manage these patients in the same way as they do patients with mutations known to increase a woman's risk."

Only about half of the surveyed women who received genetic testing ever discussed their test results with a genetic counselor, and between one-quarter and one-half of the surveyed breast cancer surgeons indicated they treat women with VUS no differently than women with known cancer-associated mutations, the researchers found. Furthermore, some women undergo surgery prior to receiving genetic testing or seeing the results.

Kurian is the lead author of the study, which will be published online April 12 in the *Journal of Clinical Oncology*. University of Michigan researchers Reshma Jagsi, MD, DPhil, and Steven Katz, MD, MPH, share senior authorship.

The need for genetic testing

The findings come on the heels of a February study by many of the same researchers showing that physicians often fail to recommend genetic testing for breast cancer patients at high risk for mutations in the BRCA1 or BRCA2 genes, which are strongly associated with ovarian and other cancers.

In this study, the researchers asked 2,502 women newly diagnosed with breast cancer whether they had received genetic testing, and if so, whether the testing and any discussion of results occurred before or after breast surgery.

They found that of the 666 women who had received testing, 59 percent were considered to have a high risk of a dangerous mutation in a cancer-associated gene. About one-quarter of these women had genetic testing only after surgery—meaning critical decisions were made about their care before information about their mutation status was available. Delays in testing were particularly pronounced in women who lacked private health insurance.

The researchers then polled the surgeons who treated the women in the survey. They found that, when compared with doctors who had treated 51 or more newly diagnosed breast cancer patients during the previous year, doctors who had treated fewer than 21 breast cancer patients were: less confident in discussing the results of genetic testing with patients, more likely to order the genetic test without referring women to a genetic counselor, less likely to delay surgery in order to have test results available for surgical decision-making and more likely to manage a patient with variants of uncertain significance in the same way they would manage patients with proven high-risk mutations in cancer-associated genes.

"Our findings suggest that we are not maximizing the benefit of genetic testing for our patients with breast cancer because of barriers related to timeliness of testing and lack of expertise necessary to incorporate results into treatment decisions," said Katz, who is a professor of medicine and of health management and policy at the University of Michigan.

Expertise to interpret the results

Although genetic testing has become more common and less costly, it's also become more confusing. The advent of multiplex gene panels that simultaneously test for mutations or variations in many different genes can render results that are difficult to interpret without the help of a trained genetic counselor. Uncertainties as to the meaning of test results may lead less-experienced surgeons to recommend aggressive treatment in the form of bilateral mastectomies, or cause women to opt for what they may feel is the safest option to manage their cancer.

Conversely, high-risk women who do carry dangerous mutations need this information to make informed decisions about their health care choices.

"The gaps identified in this study are striking," said Jagsi, professor and deputy chair of radiation oncology at the University of Michigan. "It is critical to ensure that patients at high risk for known cancer-associated mutations are fully informed of the potential benefits of genetic testing, and counseled accurately about the meaning of test results."

"We're learning that clinicians' knowledge of breast cancer genetics can be highly variable," said Kurian, who is a member of the Stanford Cancer Institute. "It's important for women at high risk of carrying a dangerous mutation to see someone with expertise in cancer genetics when planning their care. Unfortunately, in many cases genetic counselors may not be optimally integrated into the care of newly diagnosed cancer patients, making it difficult to rapidly triage these patients. Our study highlights the urgent need for improved patient access to cancer genetics experts, particularly genetic counselors, and for educating physicians about the appropriate use of genetic testing and interpretation of test results."

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