

Findings suggest a gap between need, availability of genetic counseling

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Physicians often fail to recommend genetic testing for breast cancer patients at high risk for mutations associated with ovarian and other cancers, according to a large study by researchers at the Stanford University School of Medicine and five other U.S. medical centers.

Asian-Americans and older women were particularly likely to be "undertested." Not testing these women represents a critical missed health care opportunity, the researchers said.

"We found that genetic counseling and testing are not well-matched to medical need," said Allison Kurian, MD, associate professor of medicine and of health research and policy at Stanford. "Women are very interested in genetic testing but many fail to receive it. This is particularly worrisome because it means that doctors are missing the opportunity to prevent cancers in mutation carriers and their family members."

Kurian is the lead author of the study, which will be published online Feb. 7 in *JAMA*. University of Michigan researchers Reshma Jagsi, MD, DPhil, and Steven Katz, MD, MPH, share senior authorship.

Surveying more than 2,500 women

Genetic testing can identify the presence of mutations in the BRCA1 and BRCA2 genes, which are strongly linked to the development of breast

and ovarian cancers, as well as the presence of other cancer-related mutations.

Kurian and her colleagues surveyed 2,529 women with stage-0 to stage-2 breast cancer two months after surgery. The women were asked whether they wanted genetic testing and, if so, whether they had received it.

Although about two-thirds of the women said they wanted to be tested, only about one-third said they had been tested. About eight in 10 of those at highest risk for BRCA mutations wanted testing, but just over half of them were actually tested. About 56 percent of the women who were not tested attributed the lack of testing to the fact that it was not recommended by their physicians.

Genetic counseling often unavailable

The survey also found that genetic counseling, either to help the patients decide whether to seek testing or to help them understand the results of their tests, often did not occur. Only about 40 percent of all high-risk women, and 60 percent of those high-risk women who were tested, reported having a genetic counseling session.

"Genetic testing results can affect what sort of surgery a woman may choose to treat her existing breast cancer, as well as what treatments she should pursue to reduce the risk of forming new cancers in the future," said Jagsi, who is professor and deputy chair of radiation oncology at Michigan. "We don't have a crystal ball, but genetic testing can be a powerful tool for certain women. It is worrisome to see so many of those women at highest risk for mutations failing even to have a visit focused on genetic counseling."

Overall, the survey's results indicate that women are often not learning of genetic mutations that could lead to the development of additional

cancers in them or in family members who may carry the same mutation. Women who know they carry a cancer-associated mutation may opt for more frequent or stringent screening, or sometimes even surgery to remove their breasts or ovaries before a cancer develops.

"The fact that many women are not offered genetic testing after a diagnosis of breast cancer is an important illustration of the challenges of driving advances in precision medicine into the exam room," said Katz, who is a professor of medicine and of health management and policy at Michigan.

"It is likely that some doctors don't realize the benefit that genetic testing provides," said Kurian. "They may also lack the ability to explain the testing process and results clearly with patients. Priorities for the future should include strategies to expand the genetic counselor workforce and interventions to improve physicians' skills in communication and cancer risk assessment."

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