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Genetic testing for breast or ovarian cancer risk may be greatly underutilized

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Although a test for gene mutations known to significantly increase the risk of hereditary breast or ovarian cancer has been available for more than a decade, a new study finds that few women with family histories of these cancers are even discussing genetic testing with their physicians or other health care providers. In a report in the *Journal of General Internal Medicine*, which has been released online, investigators from the Massachusetts General Hospital (MGH) Institute of Health Policy and Dana-Farber Cancer Institute note that their findings illustrate the challenges of bringing genetic information into real-world clinical practice.

"Testing for BRCA1 and 2 mutations has been around a long time and should be a good indicator of whether genetic testing is making its way into regular medical practice," says Douglas Levy, PhD, of the MGH Institute for Health Policy, the study's lead author. "When a well-established genetic test is not being incorporated into clinical practice when appropriate, we are a long way from meeting the promise of personalized, genetically-tailored medical care."

Most women's lifetime risk of breast cancer is about 13 percent, and the risk for ovarian cancer is less than 2 percent. But women with mutations in the BRCA1 or BRCA2 genes may be 3 to 7 times more likely to develop breast cancer and 9 to 30 times more likely to develop ovarian cancer than women with unaltered forms of the genes. Several organizations have issued clinical guidelines designating who should be screened for BRCA1/2 mutations, and while there have been

discrepancies among the guidelines, all of them include a history of breast or ovarian cancer in close relatives among the criteria indicating elevated risk. The authors note that most U.S. health insurers cover at least part of the cost of BRCA1/2 testing for at-risk women.

The current study analyzed data from the 2000 and 2005 National Health Interview Surveys, both of which included supplementary questions assessing cancer control. More than 35,000 women participating in those surveys did not have a personal history of breast or ovarian cancer, and around 1 percent of them were determined to be at high risk because a mother, sister or daughter had such a tumor. Among these high-risk women, about half were aware that genetic testing was available, but only 10 percent had discussed it with a physician, less than 5 percent had been advised to have the test, and only 2 percent had done so.

"A patient's personal preferences are paramount in her decision to undergo genetic testing, but at the very minimum high-risk patients should have a discussion with their clinician." Levy explains. "Although we didn't examine factors that may impede these discussions, I believe that improvements won't come until it becomes easier and more routine to collect, update and interpret family history information, which could come through the availability of decision support tools based on electronic medical records.

"Right now we can stress that women should be aware of their family history of breast and ovarian cancer - on both sides of the family - communicate that information with their physicians and update it as necessary. Any woman concerned about her family history should not hesitate to raise the topic of testing with her physician," adds Levy, who is an assistant professor of Medicine at Harvard Medical School. "In addition, organizations dedicated to cancer prevention and treatment should help disseminate the guidelines - which have been updated in recent years - particularly to primary care physicians."

Source: Massachusetts General Hospital (news : web)