Study: Risk of cancer remains high for women over 50 with genetic BRCA1 or BRCA2 mutation

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Although genetic mutations in BRCA1 or BRCA2 are associated with a younger onset of breast and ovarian cancer, women with these genetic

mutations continue to face a high risk of cancer incidence after age 50, even if they have not been previously diagnosed with cancer. This is according to a new study led by Kelly Metcalfe, a professor at the Lawrence S. Bloomberg Faculty of Nursing.

The study published recently in the journal *Cancer*, followed over 2000 women between the ages of 50 to 75, from 16 countries, who were aware they had a BRCA mutation and had no previous diagnosis of cancer. The study found that the cumulative risk of these women developing any type of cancer after the age of 50 was 49 percent for those with a BRCA1 mutation and 43 percent for those with a BRCA2 mutation.

For those in the study cohort who had not undergone a cancer risk-reduction surgery, the risk was even greater at 77 percent for those with a BRCA1 mutation and 67 percent for those with a BRCA2 mutation.

"What is striking about our results is that breast and ovarian cancers were the most frequently observed cancers occurring and that is concerning, considering we know how to reduce the risk of cancer in women who have these genetic risk factors," says Metcalfe who is also a Senior Scientist at Women's College Hospital.

Of the women included in the study only 15 percent underwent a preventative bilateral mastectomy, and 43 percent a bilateral salpingo-oophorectomy (BSO)—removal of both ovaries and fallopian tubes—before the age of 50. The study found that these women had the lowest risk of any occurrence of cancer at just 9 percent.

"Our analysis highlights the effectiveness of these risk reduction surgeries, and emphasizes the need for individuals as well as health care providers, to consider clinical guidelines and recommendations for their cancer risk, including how their genetics might impact them even at a

later age," says Metcalfe.

She acknowledges that there are some limitations to the study including the fact that in some countries access to risk reduction surgeries may be limited, and knowledge of how often or whether these women received genetic counseling was not assessed.

"We are not aware of whether the participants in our study received additional counseling about their elevated risk of cancer as they age and we are also unable to determine why some of these women chose to forgo preventative surgery before the age of 50," says Metcalfe. "However, it is important to point out that screening alone only reduces mortality risk by increasing the chances of detecting the cancer early, it does not reduce the risk of cancer occurring."

Understanding the limitations of screening is of particular importance for women with either a BRCA1 or BRCA2 mutation especially in assessing their risk of ovarian cancer, as no good screening method exists to detect the cancer early enough. Metcalfe's study references the National Comprehensive Cancer Network (NCCN) guidelines where it is recommended that women with a BRCA1 mutation undergo a BSO between ages 35-40 and those with BRCA2 mutation between 40-45.

Future studies says Metcalfe, will look at exploring women's decision-making around whether or not to undergo surgery and their own understanding of risk. For now, Metcalfe is hopeful that advances in genetic testing will help make it more accessible to all women who want to know their risk status.

The Screen Project Canada, housed at Women's College Hospital, is one example of a consumer-based model of testing where individuals can pay to access genetic testing that allows anyone, regardless of family history, to find out if they carry a BRCA1 or BRCA2 mutation. While it

currently operates at a small cost to the patient, Metcalfe believes that lower barriers to genetic testing will not only save lives, but eventually become a mainstay of cancer care in Canada.

"It has been over 25 years since clinical testing for BRCA1 and BRCA2 began in Canada and the United States, we have come a long way in reducing cancer incidence, but not far enough," says Metcalfe. "The majority of cancers resulting from these two genes are preventable, we need to be offering women the best chance at a cancer-free life."

More information: Kelly A. Metcalfe et al, The risks of cancer in older women with BRCA pathogenic variants: How far have we come?, *Cancer* (2022). DOI: 10.1002/cncr.34615

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