Cancer Advances: Variation in CHEK2 Gene May Triple Breast Cancer Risk

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A study of more than 9,000 Danish residents shows that a specific genetic variation in the CHEK2 gene may triple a woman's risk of developing breast cancer in her lifetime. The findings will be published online July 31 in the *Journal of Clinical Oncology*.

CHEK2 is a gene responsible for repairing DNA damage and preventing the uncontrolled division of cells, which can lead to cancer. In this study, researchers looked for a specific mutation, CHEK2*1100delC, which prevents the gene's ability to fix damage to DNA.

Overall, researchers found that 12% of women who carried the mutation developed breast cancer, compared to 5% of non-carriers. Adjusting for other factors that determine the risk of breast cancer, such as age, body mass index, and use of hormone replacement therapy, they found that women with the CHEK2 mutation were 3.2 times as likely as women who had normal CHEK2 genes to develop the disease. In determining which women were at greatest breast cancer risk, the researchers found that mutation carriers on hormone replacement therapy who were more than 60 years old and overweight had nearly a one in four chance of developing the disease within 10 years.

Screening for CHEK2 could be a useful tool to identify women who are predisposed to breast cancer and would benefit from heightened screening for the disease?much in the way screening for BRCA 1/2 is used among women with family history of the disease today. BRCA 1/2 mutations are slightly more common than CHEK2 (1% vs. 0.5% of the overall population) but are associated with a very high risk of breast cancer and particularly aggressive forms of the disease.

According to the study's authors, a key limitation of the research was that it included only white Danish women; it is not known to what extent CHEK2 mutations are found among black, Hispanic or other women, or whether the breast cancer risk associated with CHEK2 mutations among these women is as great as the risk for those involved in this study.

What does this mean for patients?

Women with a family history of breast cancer should talk to their doctor or a cancer genetics specialist to determine whether genetic testing for breast cancer susceptibility is appropriate, and if so, what type of genetic tests would be most beneficial. Screening for CHEK2 is a feasible option, particularly because the test is non-invasive.

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