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An international group of investigators led by scientists at Memorial Sloan Kettering Cancer Center (MSKCC) and the [National Cancer Institute](#) has identified a new genetic marker of risk for [breast cancer](#). Women with this DNA variation are at a 1.4 times greater risk of developing breast cancer compared to those without the variation. The findings are to be published online on March 3, 2008 in the journal *Proceedings of the National Academy of Sciences*. [\[PubMed Abstract\]](#)

"These results are exciting because they point us to new molecular pathways that may be associated with breast cancer," said the head of the research team and the study's senior author, [Kenneth Offit, MD, MPH](#), Chief of the Clinical Genetics Service at MSKCC.

The study used a methodology called genome-wide association mapping, which looks at genetic variations across the entire genome that alter the individual building blocks of DNA makeup. These alterations may occur more frequently in individuals who have certain types of disease than in carriers without such disease. In this study, a new gene locus, a specific place on a chromosome where a gene is located, was associated with breast cancer risk. That gene locus is on the long arm of chromosome 6.

“These research findings are of great interest because of the method of genome- wide association used to discover this new locus as well as others in recent months,” said Bert Gold, PhD, a Staff Scientist at the National Cancer Institute in Frederick, MD., and first author of the current study.

While the risk associated with this genetic marker is much lower than that of *BRCA* genetic mutations for example, this discovery will increase the understanding of the genetic variants that contribute to breast cancer.

These results are exciting because they point us to new molecular pathways that may be associated with breast cancer.

Kenneth Offit, MD, MPH, study's senior author and Chief of the Clinical Genetics Service at MSKCC

Researchers used samples largely from MSKCC, but also from other sites in the US, Canada, and Israel. Participants were all of Ashkenazi (Eastern European Jewish) ancestry. The study used a three-phase design centered on 249 families with multiple cases of breast cancer and no mutations of the *BRCA* genes.

“This newly identified genetic marker will not have any immediate clinical implications or impact on current screening guidelines for familial breast cancer,” said Dr. Offit. “As such, a test for these markers is not available to the general public and these tests should be performed only as part of research studies.”

Dr. Offit’s research team is now confirming that this risk marker is observed in other populations, and is studying possible changes in two genes in the chromosome 6q region.

The study was funded in part by federal funds from the Intramural Research Program of the National Cancer Institute, Center for Cancer Research, the Breast Cancer Research Foundation, the Susan Komen Foundation, the [Lymphoma](#) Foundation, and the Niehaus, Southworth, Weissenbach Cancer Research Fund.

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