Risk of breast cancer and a single-nucleotide polymorphism

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The single-nucleotide polymorphism (SNP) known as 2q35-rs13387042 is associated with increased risk of estrogen receptor (ER) -positive and -negative breast cancer, according to a study published online July 1 in the *Journal of the National Cancer Institute*.

This study was undertaken to confirm previous research that identified this SNP as a marker of susceptibility to ER-positive breast cancer.

Roger L. Milne, Ph.D., of the Centro Nacional de Investigaciones Oncológicas in Madrid, and colleagues used data from 25 case-control studies in the Breast Cancer Association Consortium to study the genetic association of breast cancer risk and the SNP. The studies included more than 31,000 women with invasive breast cancer, more than 1,000 women with ductal carcinoma in situ, and almost 36,000 women who served as controls. Participants were from Europe and Asia.

The researchers found that carrying one of the two alleles of SNP 2q35-rs13387042 was associated with increased risk of breast cancer. The magnitude of the association, however, was lower than previously noted. This association was also observed in both ER-positive and ER-negative breast cancer in white women of European origin.

"SNP 2q35-rs13387042 lies in a 90-kb region of high linkage disequilibrium that contains neither known genes nor noncoding RNAs. The causal variant (or variants) in this region has not been determined, and it is possible that it may confer a higher risk than rs13387042," the

authors write. "Elucidating the causal mechanism may improve our understanding of the etiology of breast cancer."

In an accompanying editorial, Kenneth Offit, M.D., MPH, of the Memorial Sloan-Kettering Cancer Center in New York, acknowledged the study's "highly statistically significant" findings. But, like many genome-wide association studies, he said, it requires further research.

According to Offit, however, the study is a "good example of the promises and challenges of current genetic epidemiologic approaches to SNP genotyping for breast cancer risk."

Source: Journal of the National Cancer Institute (news : web)