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More gene links to breast cancer risk

by [Melissa Stam m \(http://news.vanderbilt.edu/author/melissamarino/\)](http://news.vanderbilt.edu/author/melissamarino/) | Posted on Friday, Oct. 19, 2012 — 7:00 AM

The well-known *BRCA1* and *BRCA2* mutations linked to breast cancer are rare and have a strong effect on cancer risk. But since these mutations only contribute to a small percentage of breast cancers, much of the genetic risk is thought to come from common gene variants with weaker effects.



(iStock)

To identify these, [Jeffrey Smith \(http://dna.mc.vanderbilt.edu/genetics/\)](http://dna.mc.vanderbilt.edu/genetics/), M.D., Ph.D., associate professor of Medicine, [William Dupont \(http://biostat.mc.vanderbilt.edu/wiki/Main/WilliamDupont\)](http://biostat.mc.vanderbilt.edu/wiki/Main/WilliamDupont), Ph.D., professor of Biostatistics, and colleagues searched for single nucleotide polymorphisms, or SNPs – single “letter” changes in the genetic code – associated with breast cancer in four independent breast cancer study populations. The investigators evaluated more than 1,100 SNPs and validated a series of previously identified breast cancer risk variants. Additionally, they identified two previously unreported variants associated with breast cancer risk in three of the four study populations.

The findings, reported in the September *Cancer Epidemiology, Biomarkers and Prevention* (<http://cebp.aacrjournals.org/content/21/9/1565.abstract>), may help improve breast cancer risk prediction models to identify high-risk patients who may benefit from enhanced screening and prevention strategies.

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CA050468, CA068485), the National Center for Research Resources/[National Center for Advancing Translational Sciences](http://www.ncats.nih.gov/) (<http://www.ncats.nih.gov/>) (RR024975) of the National Institutes of Health, the Department of Defense (<http://www.defense.gov/>), and the US Department of Veterans Affairs (<http://www.va.gov/>).

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