

## Scientists Discover New Breast Cancer Risk Gene, XRCC2

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<http://www.asianscientist.com/health-medicine/breast-cancer-risk-gene-xrcc2-2012/>

*AsianScientist* (Apr. 12, 2012) - An international team of researchers has used new technology to fast track the discovery of a breast cancer risk gene, XRCC2.

The study, published in *The American Journal of Human Genetics*, was led by Professor Melissa Southey of the Genetic Epidemiology Laboratory, Department of Pathology at the University of Melbourne.

XRCC2 is the first breast cancer risk gene to be discovered using the latest genetic sequencing technology called massively parallel sequencing, which enables sequencing of large amounts of human DNA at high speed.

“The mutations in the newly identified gene XRCC2, although rare, explain another proportion of breast cancers that run in families where there is no known genetic cause and that particularly occur at an early age,” said Southey.

“Due to these results and our methodology we believe that further breast cancer risk genes will be identified at a faster rate than before and potentially for other cancers such as colorectal and prostate cancers,” she said.

Initially, using massively parallel sequencing, researchers identified XRCC2 mutations in two families (in Melbourne and the Netherlands).

This was followed by a larger series of studies in Melbourne and at the International Agency for Research on Cancer (IARC) in France. The studies used DNA from blood samples of 689 families with multiple members affected by breast cancer, and from 1,308 women who were affected at an early age by breast cancer and recruited from the general population, as well as 1,120 controls. More XRCC2 mutations were detected in the breast cancer cases but not in the controls.

Professor Southey said the discovery could help manage the risk of breast cancer for families with a strong history of the disease and no known genetic cause. Specific treatments could also be developed for patients whose breast cancer is associated with the XRCC2 mutation.

“Unaffected relatives of people with a mutation in this gene could also be offered predictive testing, subsequent genetic counseling and ongoing clinical management on the basis of their mutation status,” she said.

Currently, only about 30 percent of the familial risk for breast cancer has been explained, leaving the substantial majority still unaccounted for.

“Research indicates that no single gene is likely to account for a large proportion of the remaining unexplained genetic susceptibility to breast cancer,” said Southey.

“Although mutations in XRCC2 are rare, it is most likely that the remaining unknown breast cancer susceptibility genes will account for similar small proportions of the disease.”

The article can be found at: [Park DJ et al. \(2012\) Rare Mutations in XRCC2 Increase the Risk of Breast Cancer](#).

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Source: [University of Melbourne](#).

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