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New breast cancer gene identified

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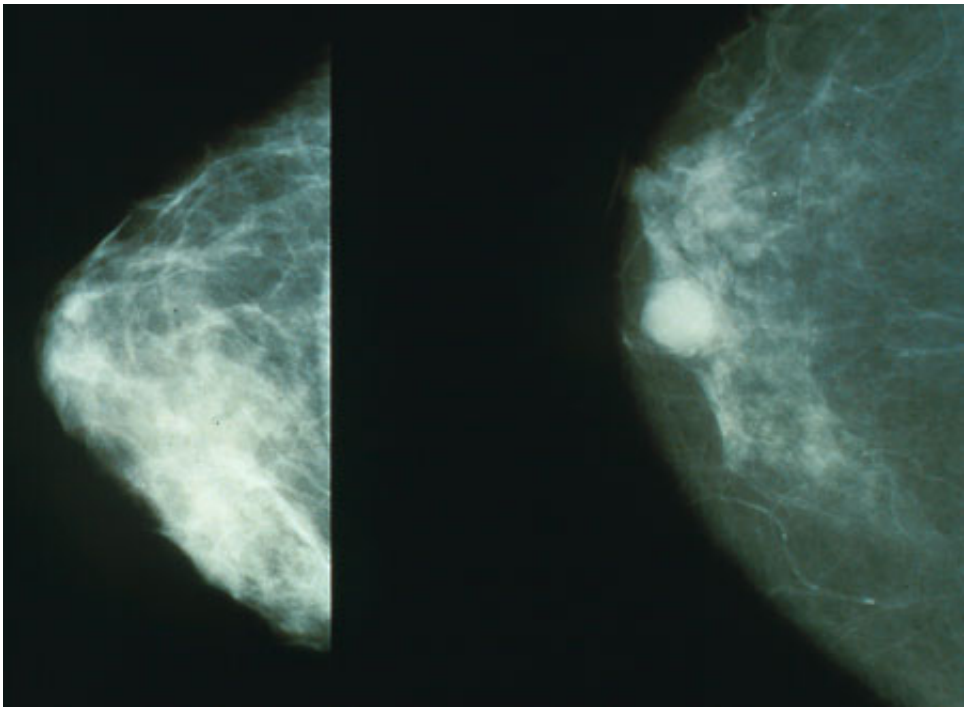

Mammograms showing a normal breast (left) and a breast with cancer (right). Credit: Public Domain

A new breast cancer gene has been identified in a study led by Women's College Hospital (WCH) researcher Dr. Mohammad Akbari, who is also an assistant professor with the Dalla Lana School of Public Health at the University of Toronto. The study, which was published online today in *Nature Genetics*, describes how mutations in a gene called RECQL are strongly linked to the onset of breast cancer in two populations of Polish and French-Canadian women.

"Our work is an exciting step in identifying all of the relevant genes that are associated with inherited breast cancer," said Dr. Akbari, who collaborated with fellow WCH senior researcher Dr. Steven Narod, as well as with Drs. Cezary Cybulski and Jan Lubi?ski from Pomeranian Medical University in Poland and Dr. William Foulkes from McGill University. The Toronto and Polish groups are longstanding research collaborators.

Approximately 10 per cent of all breast cancer cases are hereditary, though it's believed that only half of all breast cancer susceptibility genes are known.

In this study, about 20,000 different genes were studied, among 195 breast cancer patients with strong family histories of breast cancer who did not have a mutation in BRCA1 or BRCA2. The patients came from two populations—a Polish group and a French-Canadian group—who are genetically very homogenous. Dr. Akbari then led the team that confirmed the association of the identified gene, RECQL, with breast cancer by studying 25,000 more patients and unaffected people from these two populations.

"This study showed that studying specific founder populations like Polish and French-Canadian women is an excellent approach for identifying disease-associated genes," explained Dr. Narod, director of the familial breast cancer research unit at Women's College Research Institute and a professor with the Dalla Lana School of Public Health at the University of Toronto and coauthor on the study.

Specific, recurrent RECQL mutations within both the Polish and French-Canadian populations were identified in this study. Within the Polish group, one type of RECQL mutation showed a five-fold increased risk for developing breast cancer compared to individuals without a mutation. Meanwhile, within the French-Canadian population, another

type of RECQL mutation occurred 50 times more frequently among familial breast cancer patients, compared to population controls.

Though RECQL mutations appeared to be quite rare, the authors observed a very high penetrance rate - that is, in the populations studied they estimate that up to one-half of women who have a mutation are destined to get breast cancer. They expect that women from other countries will be studied shortly.

Dr. Akbari also supports the value of screening all women with breast cancer for genetic mutations, such as those occurring in RECQL. Based on a woman's genetic profile, appropriate targeted therapies could be selected."In the future, we might be able to select or develop treatments that can work around or correct relevant genetic mutations that are linked to breast cancer," explained Dr. Akbari. "This opens the door for new and better ways of approaching treatment."

More information: Germline RECQL mutations are associated with breast cancer susceptibility, *Nature Genetics*, nature.com/articles/doi:10.1038/ng.3284

Provided by Women's College Hospital