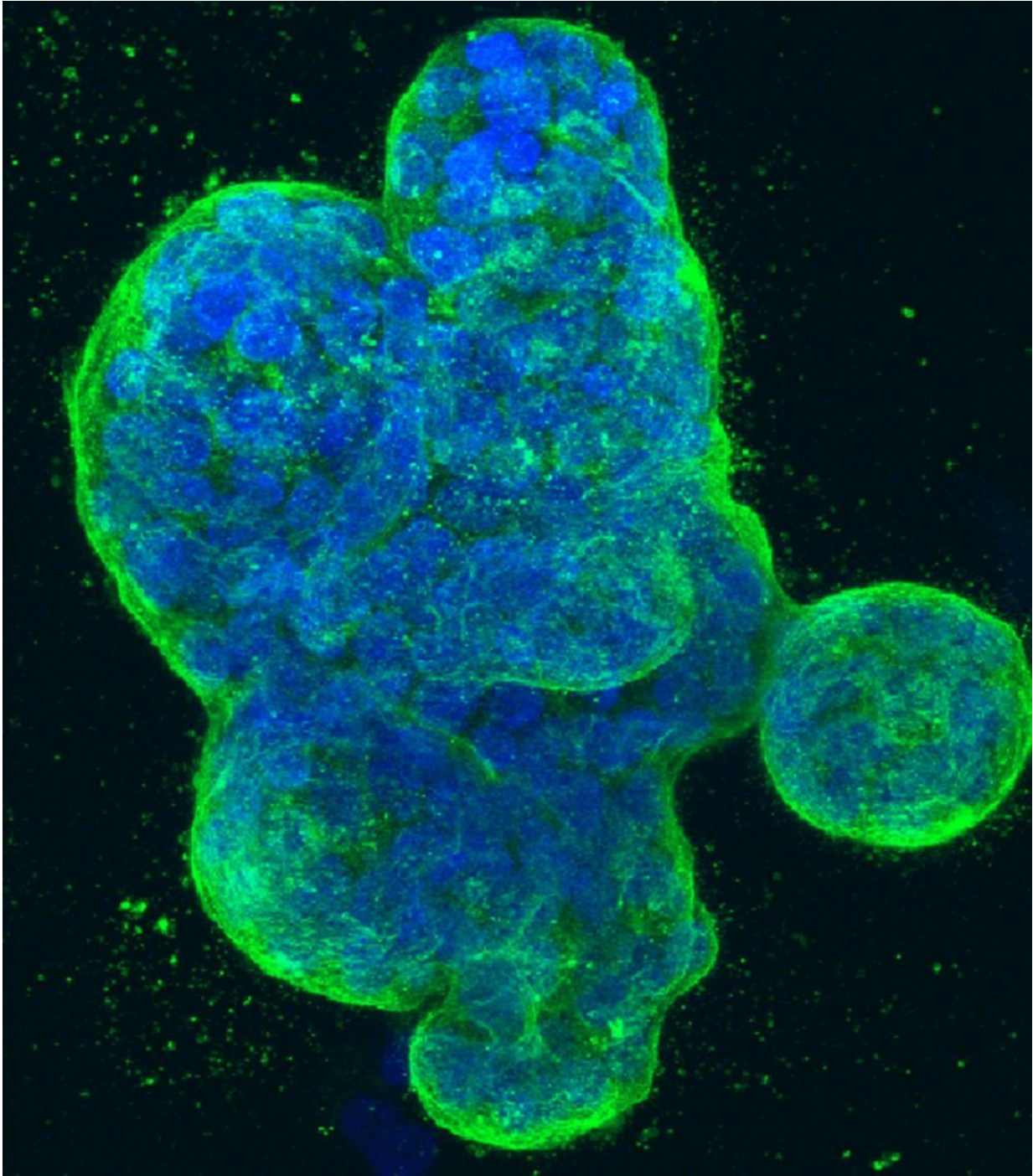


# **Paper identifies genes associated with risk for aggressive breast cancer**

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Three-dimensional culture of human breast cancer cells, with DNA stained blue and a protein in the cell surface membrane stained green. Image created in 2014 by Tom Misteli, Ph.D., and Karen Meaburn, Ph.D. at the NIH IRP.

A new study published in the *Journal of the National Cancer Institute* has identified specific genes associated with increased risk for triple-negative breast cancer, providing the basis for better risk management.

Triple-negative breast cancer is an aggressive type of cancer that cannot be treated using the most common therapies. It accounts for 15 percent of breast cancer in the Caucasian population and 35 percent in the African American population. It is also associated with high recurrence risk and poor five-year survival rate.

Germline genetic testing can identify women at increased risk of breast cancer by evaluating if there are genetic changes, often inherited from a parent, that increase the risk of certain cancers. However, it has been difficult to identify women at elevated risk of triple-negative breast cancer because only inherited mutations in BRCA1 (a human tumor suppressor gene discovered in 1990) have been linked to this subtype of breast cancer.

Here researchers performed genetic testing on 10,901 triple-negative breast cancer [patients](#) from two studies. For 8753 patients, 21 [genes](#) were tested, and for the remaining 2148 patients, 17 genes were tested.

Among the genes tested, researchers found that alternations in BARD1, BRCA1, BRCA2, PALB2, and RAD51D genes were associated with high risk for triple-negative breast cancer and greater than 20 percent lifetime risk for overall breast cancer among Caucasians. Researchers observed a similar trend among African Americans. In addition, mutations in BRIP1 and RAD51C were linked to more moderate risks of triple-negative disease.

This study is the first to establish which genes are associated with high lifetime risks of triple-negative breast cancer. While previous studies have found genetic variants in BARD1, BRIP1, PALB2, and RAD51C

more frequently in triple-negative breast cancer patients than in patients with other breast cancer subtypes, the current study shows this in more detail and identifies a new specific and strong association between the susceptibility gene RAD51D and triple-negative breast cancer risk. The study's findings will now enable expanded [genetic testing](#) to identify women at risk of this disease, potentially leading to better prevention strategies.

The new finding also suggests potential revisions to the National Comprehensive Cancer Network guidelines, which currently recommend only BRCA1/2 testing when a patient has a family history of breast cancer or someone is diagnosed at age 60 or younger. Based on a better understanding of gene-specific risks for triple-negative breast cancer from the study, it will be possible to establish guidelines for testing of other susceptibility genes; and women with genetic changes these genes should be able to receive better risk assessment.

"The results from our study showing that multiple genes cause increased risk of triple-negative breast [cancer](#) should help in the clinical management of women found to have inherited mutations in these genes," said Fergus J. Couch, Ph.D., one of the authors of the study.

**More information:** Hermela Shimelis et al, Triple-Negative Breast Cancer Risk Genes Identified by Multigene Hereditary Cancer Panel Testing, *JNCI: Journal of the National Cancer Institute* (2018). [DOI: 10.1093/jnci/djy106](https://doi.org/10.1093/jnci/djy106)

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