

African-American and white women share genes that increase breast cancer risk

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The same genes that greatly increase the risk of breast cancer in U.S. white women, including women of Ashkenazi Jewish descent, also greatly increase breast cancer risk among African American women. These genes include the BRCA1, BRCA2 and PALB2 genes, each of which was associated with a more than seven-fold risk of breast cancer, as well as four other genes associated with a more moderate increase in risk. Previous studies of women of African ancestry were too small to assess genes other than BRCA1 and BRCA2.

"This means that the multi-gene panels that are currently available to test women diagnosed with [breast cancer](#) or women at high risk due to their family history will be useful for African American women," explained corresponding author Julie Palmer, ScD, director of BU's Slone Epidemiology Center and the Karin Grunebaum Professor in Cancer Research at Boston University School of Medicine.

Researchers at Boston University and the Mayo Clinic collaborated with members of the CARRIERS consortium to conduct sequencing of germline DNA from 5,054 African American women with breast cancer and 4,993 age-matched African American women without cancer for mutations in 23 cancer predisposition genes. They then estimated the risks of developing breast cancer associated with having a mutation in any of the genes.

More than seven percent of women with breast cancer had a mutation in one of the genes, as compared with two percent of the controls. Among women with breast cancers that lacked estrogen receptors, termed [estrogen receptor](#) negative breast cancer, more than 10 percent had a mutation, as compared with five percent in women with estrogen

receptor positive breast cancer. "We also found that mutations in PALB2, RAD51C and RAD51D confer increased risks of estrogen receptor negative breast cancer in the African American population," said Fergus Couch, Ph.D., co-author of the study and the Zbigniew and Anna M. Scheller Professor of Medical Research at Mayo Clinic.

According to the researchers, testing for breast cancer predisposition genes can prevent breast cancer deaths, both in women who have never had breast cancer and in women with breast cancer. "Depending on results of the testing and an individual's own weighing of pros and cons, a woman with a mutation in any of these genes may choose more aggressive screening for cancer, and women with mutations in the high risk BRCA1 and BRCA2 genes may choose removal of her breasts and/or ovaries as a way to prevent initial breast cancer or recurrence," added Palmer who is also a professor of epidemiology at Boston University School of Public Health.

Currently, rates of breast cancer genetic testing are substantially lower in African American women with breast [cancer](#) than in white patients of the same ages. Differences in recommendations given to African American women has been identified as one of the drivers of this disparity. "To the extent that the differences in recommendations are the result of misconceptions among clinicians about the prevalence of genetic mutations and associated risks in African American women, awareness of our findings may serve to increase the proportion of African American women who are offered testing," added Palmer.

These findings appear online in the *Journal of the National Cancer Institute*.

Provided by Boston University School of Medicine

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