

## New study finds black women have higher frequency of BRCA mutations than previously reported

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Women who have inherited mutations in the BRCA1 or BRCA2 genes are more likely to develop breast cancer or ovarian cancer, especially at a younger age. Approximately 5 percent of women with breast cancer in the United States have mutations in BRCA1 or BRCA2 based on estimates in non-Hispanic white women. Moffitt Cancer Center researchers recently conducted the largest U.S. based study of BRCA mutation frequency in young black women diagnosed with breast cancer at or below age 50 and discovered they have a much higher BRCA mutation frequency than that previously reported among young white women with breast cancer.

Young black women are more likely to have aggressive types of breast cancer compared to non-Hispanic white women, yet the reason for this disparity remains uncertain. Moffitt researchers wanted to assess if mutations in the BRCA gene could help account for this higher rate of aggressive breast cancers among young black women in the U.S. They analyzed the BRCA mutation frequency and family history of 396 black women in Florida who were diagnosed with invasive breast cancer under the age of 50. They discovered that 12.4 percent of the participants had mutations in either BRCA1 or BRCA2. Furthermore, over 40 percent of those with a mutation had no close relatives with breast or ovarian cancer, which suggests that family history alone, may not identify those at risk for carrying a BRCA mutation.

As personalized medicine becomes more integrated into clinical care, it is becoming increasingly important for physicians to be aware of potential BRCA mutations at the time of diagnosis to be able to recommend the best therapy for their patients. "Our results suggest that it may be appropriate to recommend BRCA testing

in all black women with invasive breast cancer diagnosed at or below age 50," said Tuya Pal, M.D., a clinical geneticist at Moffitt, who led this effort.

However, many minorities do not undergo recommended genetic testing and counseling. According to an earlier report by the same researchers published in the journal Breast Cancer Research and Treatment, only about half of the black women were referred for or received genetic counseling or testing. They discovered that healthcare providers tend to refer patients for genetic counseling more frequently if the patients have a college education, are 45 years of age or younger, or have triple negative breast cancer. Additionally, black patients are more likely to seek genetic services if they receive a physician's referral, have private health insurance, and higher incomes. "Overall, our results suggest that there is a great need to improve access to genetic services among high-risk black women." says lead author of this report, Deborah Cragun, Ph.D., researcher and genetic counselor at Moffitt.

Patients who become aware of BRCA mutations can choose to have more <u>breast cancer</u> screening, or preventative mastectomies or oophorectomies. Additionally, this knowledge can be shared with family members so they can also be more proactive about cancer prevention. "Women who are identified with a mutation have an opportunity to be proactive about their health through cancer preventive options," explained Susan Vadaparampil, Ph.D., M.P.H., behavioral scientist at Moffitt, who co-led this effort.

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