

# Breast cancer gene mutation more common in Hispanic, young black women, study finds

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A genetic mutation already known to be more common in Ashkenazi Jewish breast cancer patients is also prevalent in Hispanic and young African-American women with breast cancer, according to one of the largest, multiracial studies of the mutation to date.

Researchers at the Stanford University School of Medicine and the Northern California Cancer Center reported the finding from a study of 3,181 breast cancer patients in Northern California. It revealed that although Ashkenazi Jewish women with breast cancer had the highest rate of the BRCA1 mutation at 8.3 percent, Hispanic women with breast cancer were next most likely, with a rate of 3.5 percent. Non-Hispanic whites with breast cancer showed a 2.2 percent rate, followed by 1.3 percent of African-American women of all ages and 0.5 percent in Asian-American women. Of the African-American breast cancer patients under age 35, 16.7 percent had the mutation.

The work, which will be published in the Dec. 26 issue of the *Journal of the American Medical Association*, marks the largest study to date to look at the prevalence of BRCA1 mutations among patients in the four ethnic and racial groups, said lead author Esther John, PhD, research scientist at the Northern California Cancer Center and consulting associate professor of health research and policy at Stanford.

The information could help doctors decide which patients to refer to genetic counseling, the researchers said. They added that they hope the information prompts genetic counselors to develop materials for discussing breast cancer risk in a culturally sensitive way and in languages other than English.

"If a woman has breast cancer she may ask the

question, 'Could I be a carrier for a BRCA1 mutation' If I am, my daughters and sons need to know it," said senior author Alice Whittemore, PhD, professor of health research and policy at Stanford. She said that until now, doctors knew only that Ashkenazi Jewish women were more likely to carry a mutation, and therefore frequently referred these women to genetic counseling. What they didn't know is how women of different ethnic groups needed to be treated in terms of their BRCA1 status.

"Traditionally studies have focused on white women," said John. "There is a great need to study racial minorities in the United States."

The risk of a woman developing breast cancer sometime during her life is about one in eight. Although death rates from the disease are dropping, the American Cancer Society estimates that 40,000 women will die from the disease this year.

All people have the BRCA1 gene, which makes a protein that helps the cell repair its DNA. Women who inherit a mutation in that gene from either parent are less able to fix DNA damage and tend to accumulate mutations that lead to cancer. They have a roughly 65 percent risk of developing breast cancer and 39 percent risk of ovarian cancer. If one family member tests positive for a mutation, it can alert other women in the family to also get tested and to take preventive measures.

Without the information from this study, doctors have treated all women other than Ashkenazi Jews as having the same risk level for the mutation. Now doctors who see Hispanic or young African-American breast cancer patients have more information to guide their decisions about referring those women to genetic counseling or testing.

"The message is that these minority breast cancer patients may need screening in ways that we hadn't appreciated before," Whittemore said. She noted that Hispanic women in Northern California, where this study was conducted, derive from different countries than Hispanic women from the East Coast. For that reason, the findings may not apply to Hispanic people in other parts of the country.

The research team found a few other surprises in the data. One is that although mutations can occur throughout the BRCA1 gene, the Hispanic women in the study were more likely to carry a particular mutation that's also common in Ashkenazi Jewish women. Other ethnic groups carried a wide range of different mutations.

John and Whittemore think the Hispanic women may have this mutation because of their Spanish ancestry. Spain was the home of Sephardic Jews who could have shared the mutation with Ashkenazi Jews of Eastern European origin.

The prevalence of the mutation in young African-American women with breast cancer also came as a surprise, given that the rate is low in the overall African-American population. The researchers say the finding is consistent with a long-known pattern that when young African-American women get breast cancer it tends to be a particularly aggressive form of the disease, which is a hallmark of tumors that arise from BRCA1 mutations. Whittemore said this information doesn't change how doctors treat those tumors, but it could help prompt more doctors to recommend genetic counseling for those young African-American breast cancer patients.

Source: Stanford University

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