One in Five African American Women with Breast Cancer Have a Genetically Higher Risk of Breast Cancer [1]

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In a recent genetic study, researchers found that one in five African American women with breast cancer have an inherited (passed down in the family) mutation (change) in at least one of the 18 genes that are linked with a higher risk of breast cancer. Compared to the general population, African American women are more likely to be diagnosed with breast cancer at a younger age, die from the disease, and have triple-negative breast cancer. Triple-negative breast cancer is a fast-growing and difficult-to-treat cancer that does not have hormone receptors (for the hormones estrogen and/or progesterone) or HER2 receptors (a protein found on some breast tumors). Researchers have suspected that these differences are due to inherited genes linked to breast cancer, but this is the first study to look at all known breast cancer gene mutations, not just *BRCA* genes.

For this study, researchers used genetic information from 249 unrelated African American women with breast cancer to look for mutations in 18 genes using a new genetic test called BROCA. Overall, 56 out of 249 women (22%) had at least one mutation that increases the risk of breast cancer. They also found that the mutations most commonly occurred on *BRCA1, BRCA2, CHEK2, PALB2, ATM,* and *PTEN* genes. In addition, they found that mutations were most common for women with triple-negative breast cancer, with 30% of women with this type having a breast cancer gene mutation. Also, 27% of women diagnosed before age 45, 49% of women with a second breast cancer, and 30% of women with a family history of either breast or ovarian cancer had mutations in at least one breast cancer gene.

Researchers also found that most of the mutations were unique to each person, meaning that it varied between each family. Other groups of people with a higher inherited risk of breast cancer have specific mutations that are passed through many generations. For example, Ashkenazi Jewish women are known to carry three specific mutations in the *BRCA1* and *BRCA2* genes, which can be easily tested. This study shows that such an approach would not work as well for African American women, though, because multiple tests would be needed to find the variety of mutations in the breast cancer genes.

What this means for patients

?For many years, we?ve seen breast cancer take a heavy toll on African American women, and this study begins to resolve unanswered questions about what?s driving these disparities,? said lead author Jane E. Churpek, MD, Assistant Professor of Medical Oncology at the University of Chicago in Illinois. ?While larger studies are needed to confirm our results and compare them to other populations, we hope our findings will lead to life-saving genetic screening for African American women with a family history of more aggressive forms of breast cancer.? Breast cancer screening for women with a higher risk of breast cancer often differs from screening for the general population. Talk with your doctor about your risk of breast cancer, the screening methods recommended for you, and any steps you can take to lower your risk.

Dr. Churpek was a recipient of a Conquer Cancer Foundation of ASCO Young Investigator Award in 2011.

Questions to Ask Your Doctor

- What is my risk of breast cancer?
- Do I have a higher risk of the disease?
- Should I meet with a genetic counselor to assess my risk?
- If I do have a mutation in a breast cancer risk gene, what are my options to lower my risk?

For More Information

Guide to Breast Cancer [2]

Genetics [3]

Cancer Screening [4]

What to Know: ASCO?s Guideline on Drugs to Lower Breast Cancer Risk [5]

Links:
[1] http://www.cancer.net/one-five-african-american-women-breast-cancer-have-genetically-higher-risk-breast-cancer
[2] http://www.cancer.net/node/18618
[3] http://www.cancer.net/node/24864
[4] http://www.cancer.net/node/24972
[5] http://www.cancer.net/node/29926