Significant increase in number of women tested for BRCA gene, but many high-risk patients still missing out

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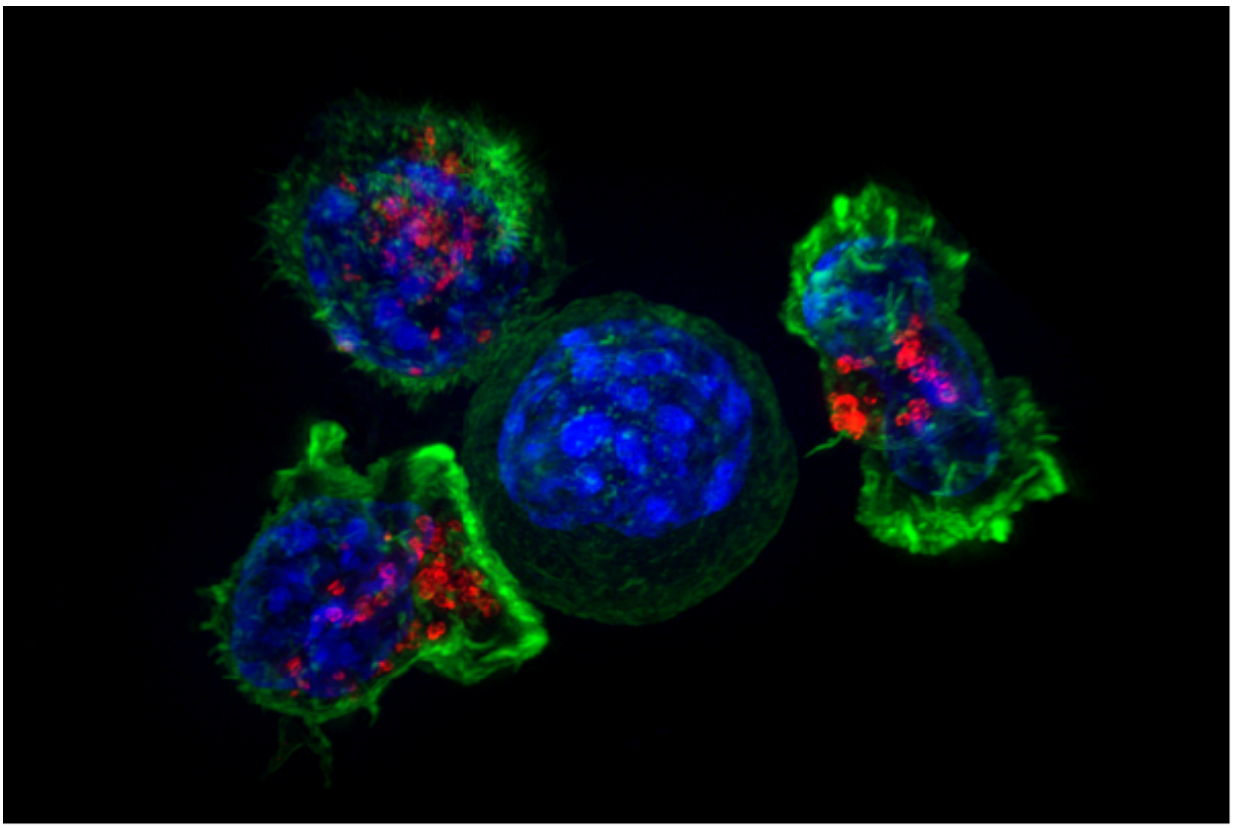

Killer T cells surround a cancer cell. Credit: NIH

Discovery of the *BRCA* genetic mutation in the mid-90s represented a breakthrough in breast and ovarian cancer prevention. About 5-10% of

breast cancer cases and 10-18% of ovarian cancer cases can be attributed to two *BRCA* mutations. Testing for these genetic abnormalities has risen steadily over the past decade. Previously, mainly women with a history of cancer were referred for genetic testing, but as awareness has grown, more low-risk women are undergoing *BRCA* testing. A new study in the *American Journal of Preventive Medicine* found that the proportion of women without a history of cancer who underwent *BRCA* testing rose sharply from 24.3% in 2004 to 61.5% in 2014.

"It is estimated that 60%-80% of patients referred for genetic counseling and testing do not meet the referral requirement based on family history. To optimize the infrastructure and medical resources allocated for genetic testing, it is important to understand the current use of *BRCA* mutation testing in the U.S. health system," commented lead investigator Fangjian Guo, MD, PhD, Assistant Professor, Department of Obstetrics and Gynecology, Center for Interdisciplinary Research in Women's Health, the University of Texas Medical Branch.

BRCA testing is groundbreaking because it can allow for very early screening and prophylactic treatments that can save lives. In this study, investigators found that *BRCA* testing has shifted from mainly breast or ovarian cancer patients and individuals at high risk to unaffected women. Although it has been recommended by the U.S. Preventive Services Task Force (USPSTF) since 2005 for women with a family history of breast and/or ovarian cancer, *BRCA* testing has been underutilized, especially among at-risk populations. According to current data, it is estimated that only 30% of breast cancer survivors with the *BRCA* mutation have been identified, and that number drops significantly to 10% for asymptomatic *BRCA* carriers.

In 2004, about 75% of women undergoing *BRCA* testing had a history of cancer, but by 2014, that number had dropped to approximately 40% of the total. "Currently, many women who do get tested are actually low

risk and do not have any personal or family history of breast or ovarian cancer. With low-cost genetic testing ($200 or $300, roughly the price of a three-dimensional mammogram) available, even more unaffected individuals and cancer patients may choose to receive these tests even when they have to pay out of pocket," explained Dr. Guo.

Over the past decade, widespread direct to consumer marketing for genetic tests has raised consumers' interest in *BRCA* testing, and increased women's self-referrals and referrals by their physicians to genetic services even when they are at low risk for mutations. According to Dr. Guo, "This may be the driving force for the shift in the role of *BRCA* testing during [the] last decade. However, this may not necessarily translate into a great improvement in identifying mutation carriers, as many of the tests are performed in women who do not carry harmful *BRCA* mutations. Policymakers may need to take this into consideration to promote proper use of the test and maximize the detection of mutation carriers."

Along with marketing and an increased awareness among patients and practitioners about the *BRCA* mutation, other mechanisms may have played a role in boosting the number of women getting tested. The Affordable Care Act, for example, mandates coverage for preventive services that are recommended by USPSTF, including *BRCA* testing. Also, the 2013 Supreme Court ruling against Myriad Genetics and their patent claim over the gene loosened the grip on who could perform *BRCA* testing, making it more affordable.

Despite the increase of *BRCA* testing, many high-risk women remain unidentified, while more and more low-risk women undergo screening. "Next-generation sequencing technologies are dramatically reducing costs for genetic testing and sequencing. However, current guidelines and clinical practice fail to efficiently identify women who carry harmful *BRCA* mutations. Effective testing strategies need to be

identified that promote equitable distribution and rational use of *BRCA* testing and maximize the detection of mutation carriers," concluded Dr. Guo.

More information: "Use of BRCA Mutation Test in the U.S., 2004-2014," by Fangjian Guo, MD, PhD, Jacqueline M. Hirth, PhD, MPH, Yu-li Lin. MS, Gwyn Richardson, MD, Lyuba Levine, MD, MMSci, Abbey B. Berenson, MD, PhD, Yong-Fang Kuo, PhD, *American Journal of Preventive Medicine*, volume 52, issue 6 (June 2017) dx.doi.org/10.1016/j.amepre.2017.01.027

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