Study examines genetic testing in diverse young breast cancer patients over a decade

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Breast cancer patients diagnosed under age 50 represent 18 percent of new invasive breast cancer cases in the United States. Compared to postmenopausal women, younger women are more likely to develop

aggressive subtypes of breast cancer, have a worse prognosis with increased risk of recurrence, and have higher overall mortality. Young breast cancer patients also are more likely to be diagnosed with triple-negative breast cancer, which is associated with a higher frequency of BRCA1 genetic mutations.

Although breast cancer mortality has steadily declined over the past decade, disparities in both incidence and mortality persist across racial/ethnic and socioeconomic groups. Among women younger than 40 years old, non-Hispanic black women have a higher incidence of breast cancer compared to non-Hispanic white women. Although Hispanic women are at lower risk for breast cancer than non-Hispanic White women, they have the second highest prevalence of BRCA1/2 gene mutations after Ashkenazi Jewish women and breast cancer is the leading cause of cancer death among Hispanic women. The frequency of variants of uncertain significance remains high among Asian women as they are underrepresented in genomic research.

There has been increasing awareness of multigene panel testing, including BRCA1 and BRCA2 (BRCA1/2) genetic testing for breast and ovarian cancers in recent years. Despite this increase in public awareness, few studies in the U.S. have addressed germline genetic testing in young racially/ethnically diverse breast cancer patients in an era of multigene panel testing.

Tarsha Jones, Ph.D., a researcher at Florida Atlantic University's Christine E. Lynn College of Nursing, collaborated with researchers from Columbia University Irving Medical Center to examine racial and ethnic differences in genetic testing frequency and results (pathogenic/likely pathogenic, variants of uncertain significance, negative) among diverse breast cancer patients diagnosed at age 50 or younger from January 2007 to December 2017.

Results of the study, published in the *Journal of Cancer Education*, showed that among 1,503 diverse young breast cancer patients, less than half (46.2 percent) completed hereditary breast and ovarian cancer genetic testing. However, the percentage of women who completed genetic testing increased over time from 15.3 percent in 2007 to a peak of 72.8 percent in 2015. The study also found racial/ethnic differences in genetic testing results with non-Hispanic blacks and whites having the highest frequency of pathogenic/likely pathogenic variants (18.2 percent and 16.3 percent, respectively), whereas Asians and Hispanics had the highest frequency of variants of uncertain significance (21.9 percent and 19 percent, respectively).

Since the burden of breast cancer is particularly high among young black women, with a mortality rate that is two times greater among young women of European ancestry, there is a need to engage more young black breast cancer patients in genetic counseling education and to highlight the importance of performing hereditary breast and ovarian cancer genetic testing for minority women.

"Although multigene panel testing offers more comprehensive cancer risk assessment, there is greater uncertainty in clinical decision-making due to increased likelihood of variants of uncertain significance, particularly among racial/ethnic minorities who have been found to have more frequent variants of uncertain significance results compared to non-Hispanic whites," said Jones, lead author and an assistant professor in FAU's Christine E. Lynn College of Nursing.

Participants who completed genetic testing were more likely to be younger, be married, have a family history of breast cancer, have stage 1 breast cancer, and be diagnosed after 2013. There were no significant differences in the completion of genetic testing based upon race/ethnicity or primary health insurance status. Compared to patients diagnosed before 2008, women diagnosed after 2013 were more than 10

times more likely to have genetic testing.

"Our study provides insights into pathogenic/likely pathogenic variants and variants of uncertain significance in breast and ovarian cancer susceptibility genes among young breast cancer patients and highlights the need to increase genetic testing completion among racially/ethnically diverse populations," said Jones.

In addition, the study found that women with metastatic breast cancer were more than 60 percent less likely to undergo genetic testing compared to women with stage 1 disease. Women who were older at breast cancer diagnosis also were less likely to have genetic testing. The odds of a young woman completing genetic testing increased nearly three-fold if she had a family history of breast cancer. Compared to patients with stage 1 breast cancer, those with stage 0 or stage 4 disease at diagnosis were less likely to complete genetic testing.

According to Jones, next steps include conducting a future study that examines the impact of receiving pathogenic/likely pathogenic genetic testing results and promoting family risk communication since pathogenic variants can be inherited.

"This study underscores the importance of increasing awareness of the importance of genetic testing among young minority women with breast cancer," said Safiya George, Ph.D., dean of FAU's Christine E. Lynn College of Nursing. "It also signals that these patients' health care providers and future health care providers of this population need to be reminded and/or made aware of the critical need for them to receive this important multigene panel testing."

Among 1,503 evaluable patients, the mean age was 42.7 years, 42.4 percent were non-Hispanic white, 13.3 percent were non-Hispanic black, 25.5 percent were Hispanic, 9.9 percent were Asian, and 8.9 percent

were other or unknown race/ethnicity. The majority (60.5 percent) of patients had private insurance, 22.8 percent had Medicaid, 7.2 percent had Medicare (either due to disability or existing co-morbidities), and 9.4 percent had other insurance or were uninsured.

More information: T Jones et al, Racial and Ethnic Differences in BRCA1/2 and Multigene Panel Testing Among Young Breast Cancer Patients, *Journal of Cancer Education* (2019). DOI: 10.1007/s13187-019-01646-8

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