Many women with early-stage breast cancer who test positive for an inherited genetic variant are receiving cancer treatment that does not follow current guidelines, according to a study published online Feb. 6 in JAMA Oncology.

Allison W. Kurian, M.D., from Stanford University School of Medicine in California, and colleagues evaluated the association of germline genetic testing results with locoregional and systemic therapy use in women diagnosed with breast cancer. Deviation of treatment from practice guidelines was assessed. Data from the Surveillance, Epidemiology, and End Results registries of Georgia and California were used to identify 20,568 women (aged ≥20 years) who were diagnosed with stages 0 to III breast cancer between 2014 and 2016.

The researchers found that compared with women whose test results were negative, those with BRCA1/2 pathogenic variants were more likely to receive bilateral mastectomy for a unilateral tumor (61.7 versus 24.3 percent; odds ratio [OR], 5.52), less likely to receive postlumpectomy radiotherapy (50.2 versus 81.5 percent; OR, 0.22), and more likely to receive chemotheraphy for early-stage estrogen and/or progesterone receptor-positive disease (38 versus 30.3 percent; OR, 1.76). For pathogenic variants in other breast cancer-associated genes (ATM, CDH1, CHEK2, NBN, NF1, PALB2, PTEN, and TP53), similar patterns were seen, but not with variants of uncertain significance.

"Women with pathogenic variants in BRCA1/2 and other breast cancer-associated genes were found to have distinct patterns of breast cancer treatment; these may be less concordant with practice guidelines, particularly for radiotherapy and chemotherapy," the authors write.

More information: Abstract/Full Text (subscription may be required)

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