Is *BRCA* testing causing women to undergo unnecessary prophylactic mastectomy?

Yes. According to this cross-sectional analysis of an ongoing prospective cohort study of the trends in *BRCA1* and *BRCA2* mutation testing, 82 (51.2%) of 160 non–*BRCA* mutation carriers underwent bilateral mastectomy from 2006 through 2014.

FAST TRACK

Women who are at high risk for breast cancer should be referred to a cancer genetics counselor for formal *BRCA* testing and counseling

Rosenberg SM, Ruddy KJ, Tamimi RM, et al. BRCA1 *and* BRCA2 *mutation testing in young women with breast cancer [published online ahead of print February 11, 2016]. JAMA Oncol. doi:10.1001/jamaoncol.2015.5941.*

EXPERT COMMENTARY

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Because the prevalence of *BRCA1* and *BRCA2* mutations is elevated among young women diagnosed with breast cancer, guidelines recommend carrier testing for women diagnosed with this disease at age 50 years or younger.[1] Are women being tested, however, and what are their treatment decisions surrounding those test results? The Young Women's Breast Cancer Study (YWBCS) seeks to answer such questions.

Details of the study

Study investigators recruited women diagnosed with breast cancer at age 40 or younger from 11 academic and community hospitals in the United States and Canada beginning in 2006. There were 897 evaluable participants who were recruited between 2006 and 2014. Their mean age at diagnosis was 35.5 years and 86.1% of them were white non-Hispanic. A respective 84.5% and 99.8% of women had at least a college education and were insured.

Overall, *BRCA* testing was performed within 1 year of breast cancer diagnosis in 87% of participants, with rates rising from 77% in 2006 to 95% in 2013. Among participants tested, 7.6% had a *BRCA1* mutation, 4.5% had a *BRCA2* mutation, 4.6% had an indeterminate result of unknown clinical significance, and 81.3% had a negative test result.

A total of 86.4% of women found to be mutation carriers proceeded with risk-reducing bilateral mastectomy; 51.2% found not to be mutation carriers had this same prophylactic surgery.

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WHAT THIS EVIDENCE MEANS FOR PRACTICE

Although it is encouraging to see that the proportion of young women with breast cancer who are receiving counseling and genetic testing is rising, the findings from this study of highly educated, largely white and affluent women is not generalizable to all US women diagnosed with breast cancer at a young age.

That more than half of *BRCA*-negative women in this study chose bilateral prophylactic mastectomy, a procedure not recommended in this population, is concerning, and reflects nationwide trends.[2] The increasing use of next-generation sequencing (which yields information on moderate- and low-penetrance genes in addition to *BRCA* status) means that women and their providers increasingly are being confronted with genetic testing results that call for formal genetics expertise. Unfortunately, genetics counselors remain in short supply and many clinicians without specific genetics training are offering these tests. As editorialists appropriately point out, these trends may further increase the number of relatively low-risk women proceeding with unwarranted bilateral mastectomy.[3] In my practice, I continue to refer women whose family or personal histories indicate high-risk status to a cancer genetics counselor for formal counseling and possible testing.

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