Beyond BRCA: Promising results with multigene testing for breast cancer

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Results of a multicenter observational study suggest that testing for a panel of genes may be the wave of the future for hereditary breast and ovarian cancer and alter the course of clinical management. The findings, published in *JAMA Oncology*, showed that detection of mutations in genes beyond *BRCA1* and *2* would be likely to lead to recommendations for more cancer screening and/or preventive measures for women at risk of breast cancer and their family members unaffected by the disease.

Recommended: Surgical options for risk reduction in BRCA mutation carriers

The goal of the research, conducted between 2001 and 2014 at 3 academic centers, was to determine the potential clinical effect of multigene panel testing for hereditary breast and ovarian cancer in a clinical representative cohort. A total of 1046 women were enrolled and testing at Stanford University and Massachusetts General Hospital was with the 29-gene Hereditary Cancer Syndromes test (Invitae) and at Beth Israel Deaconess Medical Center was with the 25-gene MyRisk test (Myriad Genetics).

Forty of the women who were *BRCA1/2*-negative (3.8%; 95% CI, 2.8%-5.2%) harbored deleterious genetic mutations, most often in moderate-risk breast and ovarian cancer genes (*CHEK2*, *ATM*, and *PALB2*) and Lynch syndrome genes. Most of the mutations (92%) found in these women and 23 other mutation-positive women enrolled from the clinics were consistent with the spectrum of cancer(s) observed in those patients or their families, leading the authors to conclude that their results were clinically significant.