

Polygenic risk score helpful for women with familial breast CA

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Image courtesy of Blausen Medical

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(HealthDay)—For women affected by familial breast cancer, a polygenic risk score based on 22 genomic variants can identify women at high-risk of breast cancer, according to a study published in the Dec. 10 issue of the *Journal of Clinical Oncology*.

Sarah Sawyer, M.D., of the Peter MacCallum Cancer Centre in Melbourne, Australia, and colleagues identified 1,143 women with [breast cancer](#) from a larger screening of women at high risk for hereditary breast cancer who underwent *BRCA1* and *BRCA2* mutation screening. Genotyping was performed for 22 breast cancer-associated genetic variants, and a polygenetic [risk score](#) was calculated as the sum of the log odds ratios for each allele. Scores were compared with those of 892 controls.

The researchers found that, compared with controls, women with a family history of breast cancer had a highly significant increase of risk alleles. Compared with *BRCA*-mutation carriers, women who were *BRCA1*- or *BRCA2*-negative had a significantly increased polygenic risk. Compared with women with low polygenic risk, non-*BRCA1/2*

carriers in the top quartile of polygenic risk were significantly more likely to have had early-onset breast cancer (before age 30: odds ratio, 3.37) and had a higher rate of second breast cancer (odds ratio, 1.96).

"Genetic testing for common risk variants in women undergoing assessment for familial breast cancer may identify a distinct group of high-risk women in whom the role of risk-reducing interventions should be explored," the authors write.

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