Genomic screening can ID undetected BRCA1/2 cancer risk

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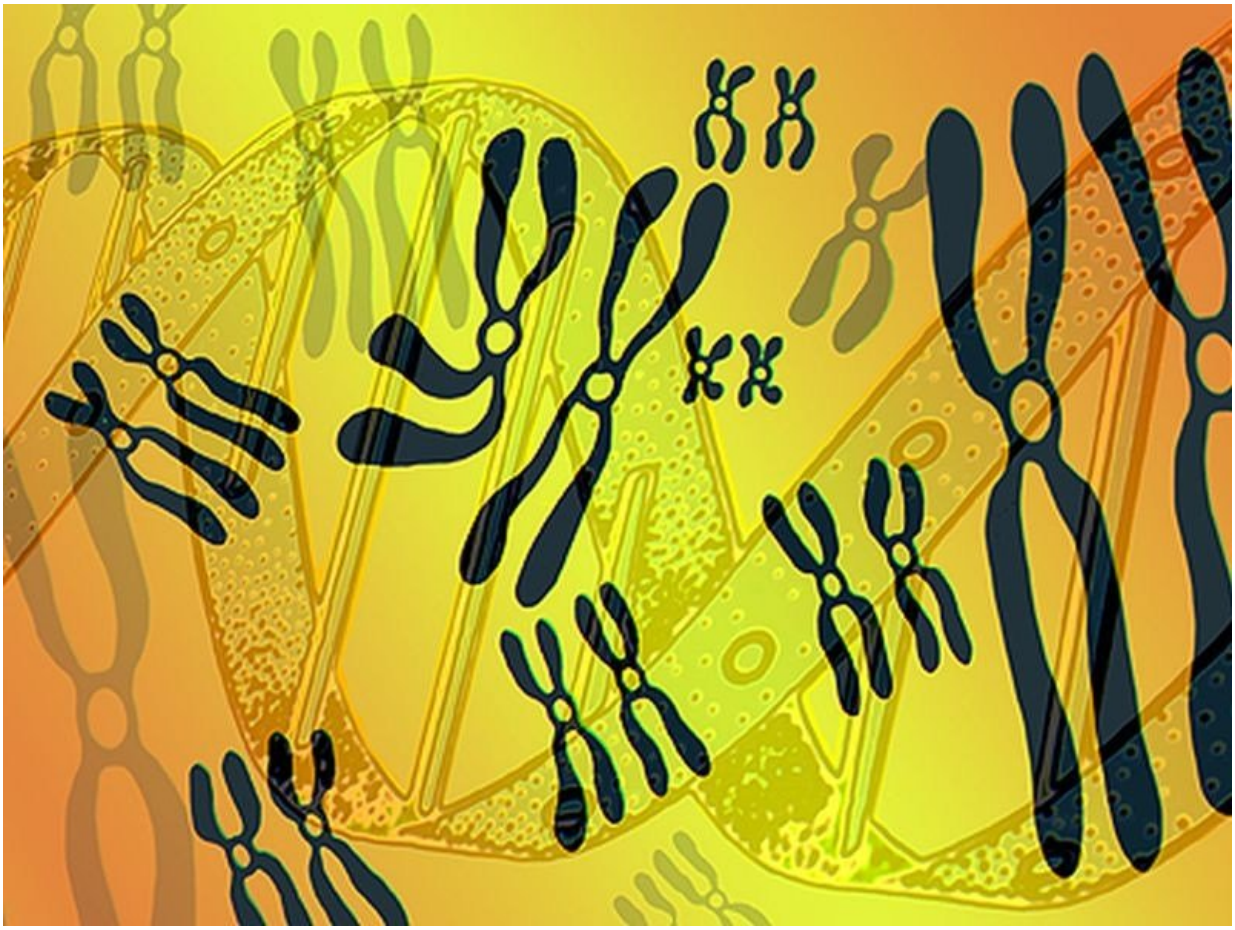

(HealthDay)—Exome sequencing-based screening may identify

pathogenic and likely pathogenic (P/LP) *BRCA1/2* variants that might otherwise remain undetected, according to a study published online Sept. 21 in *JAMA Network Open*.

Kandamurugu Manickam, M.D., M.P.H., from Nationwide Children's Hospital in Columbus, Ohio, and colleagues compared clinical data from individuals with (cases) and without (controls) P/LP variants in *BRCA1/2*. Participants were 50,726 adult volunteers who underwent exome sequencing at a single health care system from Jan. 1, 2014, to March 1, 2016.

The researchers found that 99.5 percent of those tested had no expected pathogenic *BRCA1/2* variants, while 0.5 percent were *BRCA1/2* carriers. Of the 267 cases, 55.4 percent were women and 44.6 percent were men; 35.6 percent had *BRCA1* variants, and 64.4 percent had *BRCA2* variants. The vast majority of cases (82 percent) had undergone no prior clinical testing. Just less than half (47.8 percent) of the 23 deceased *BRCA1/2* carriers and 20.9 percent of all *BRCA1/2* carriers had syndromic cancer diagnoses. Among the female variant carriers, 20.9 percent had a personal history of breast cancer versus 5.2 percent of 29,880 noncarriers (odds ratio, 5.95). Similarly, 10.1 percent of variant carriers had an ovarian cancer history versus 0.6 percent of the noncarriers (odds ratio, 18.3). Roughly half (49.4 percent) of the *BRCA1/2* carriers without prior testing but with comprehensive personal and family history data did not meet published guidelines for clinical testing.

"These findings suggest that genomic screening may identify *BRCA1/2*-associated cancer risk that might otherwise remain undetected within health care systems and may provide opportunities to reduce morbidity and mortality in patients," the authors write.

Several authors disclosed financial ties to the pharmaceutical industry.

More information: Abstract/Full Text