

Eight of 10 people with cancer risk genes don't know it

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Source: Yale University

Summary: Genomic screening shows that more than 80 percent of those who carry an identifiable genetic risk for breast, ovarian, prostate, and pancreatic cancer don't know it.

FULL STORY



Genetic laboratory (stock image).

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Genomic screening of more than 50,000 people shows that more than 80% of those who carry an identifiable genetic risk for breast, ovarian, prostate, and pancreatic cancer don't know it despite frequent interaction with the healthcare system.

The findings were published Sept. 21 in the journal *JAMA Network Open*.

In the absence of routine screening, most people only discover they carry cancer-associated variants in the BRCA1 or BRCA2 genes when that person or family members receive a cancer diagnosis.

"As a colleague said, it usually takes a tragedy for people to get tested," said Michael Murray, professor of genetics at Yale School of Medicine and senior author of the study conducted at Geisinger Medical Center in Danville, Pennsylvania. "Our reliance on a documented personal or family history as a trigger to offer testing is not working. Hopefully, one day we can change that with effective DNA-based screening for everyone."

The mean age of subjects in the study was 60. Two hundred and sixty-seven of those screened possessed a BRCA risk variant, yet only 18% of them were aware they possessed this risk factor for cancer prior to being informed by the study.

Among the group of living BRCA-positive patients, 16.8% of them had a BRCA-associated cancer. In the small group of BRCA-positive patients who had died before the study's conclusion, 47.8% of them had a BRCA-associated cancer.

"Once risk is identified, we can apply proven tools for early diagnosis and prevention, and we believe that the 31% difference in cancer incidence in these two groups is a window into an opportunity to decrease cancer and cancer deaths through genomic screening approaches," Murray said.

Story Source:

Materials provided by **Yale University**. Original written by Bill Hathaway. *Note: Content may be edited for style and length.*

Journal Reference:

1. Manickam K, Buchanan AH, Schwartz MLB, et al. **Exome Sequencing–Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants.** *JAMA Network Open*, 2018; 1 (5): e182140 DOI: 10.1001/jamanetworkopen.2018.2140

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