

Digital risk assessment tool can ID risk for cancer susceptibility syndromes, according to study

October 27 2023, by Elana Gotkine



Implementation of a digital risk assessment tool can identify patients with high-risk cancer susceptibility syndromes, according to a study

presented at the annual meeting of the American College of Gastroenterology, held from Oct. 20 to 25 in Vancouver, British Columbia, Canada.

Heather Hampel, from the City of Hope National Medical Center in Duarte, California, and colleagues examined implementation of a digital cancer risk assessment tool and [genetic testing](#) program in gastroenterology practices. Seven practices of 17 gastrointestinal providers implemented the service between September 2021 and May 2023.

Overall, 5,830 of 9,340 assessment invitations were completed out of 25,349 patient appointments. The researchers found that 25.2 percent of the invitations completed met the National Comprehensive Cancer Network criteria for [genetic counseling](#) and testing; 27.4 percent had genetic testing ordered and 74.9 percent completed testing.

Overall, 15.6 percent of those tested were found to have 49 pathogenic/likely pathogenic variants (P/LPVs) in a cancer susceptibility gene, while 23.5 and 61.3 percent had variants of uncertain significance and tested negative, respectively.

Twelve individuals were diagnosed with high-risk cancer susceptibility syndromes: seven, four, and one with Lynch syndrome, hereditary breast-ovarian cancer syndrome, and Li Fraumeni syndrome, respectively. Thirteen individuals had P/LPVs in moderate-risk genes; 24 [individuals](#) carried autosomal recessive disorders that did not affect their [cancer risk](#) but were important for reproductive risk counseling.

"Patients should possess awareness and knowledge regarding hereditary genetic cancer predispositions, as well as the significance of undergoing suitable genetic screening and counseling for themselves and their families," presenting author Daniel J. Pambianco, M.D., of Gastro

Health in Charlottesville, Virginia, said in a statement. "

Numerous cancers and conditions of this nature can either be prevented or identified in their early stages."

More information: [Press Release](#), [Abstract](#)

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Citation: Digital risk assessment tool can ID risk for cancer susceptibility syndromes, according to study (2023, October 27) retrieved 29 April 2024 from <https://medicalxpress.com/news/2023-10-digital-tool-id-cancer-susceptibility.html>

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