Study shows expanded cancer gene testing feasible, beneficial for patients

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A new University of Kentucky Markey Cancer Center study published in *JCO Precision Oncology* demonstrates a feasible way to expand germline genetic testing for patients with cancer.

Germline testing analyzes an individual's DNA to identify inherited genetic mutations that increase the risk for cancer and other diseases. For patients with cancer, the tests can determine potential therapies and identify family members that would benefit from risk reduction strategies and early screening opportunities.

Current guidelines recommend germline testing for patients with certain cancers including ovarian, pancreatic, and metastatic prostate cancer. While patients with other types of cancer may also receive germline testing as part of research initiatives, those test results are not routinely reported.

The study led by UK Markey Cancer Center researcher Jill Kolesar, Pharm.D., aimed to bridge this gap by sharing research-based germline test findings with oncologists.

In the study, Markey Cancer Center's Molecular Tumor Board (MTB) reviewed germline research test results from a group of patients with various cancers. Findings were then communicated to patients' oncologists, along with recommendations for genetic counseling and confirmatory testing.

Among 781 participants, more than 4% had genetic mutations related to inherited cancers. More than a third of those cases would have been missed by the existing guidelines. The team also identified 14 patients at risk for other hereditary diseases not typically covered by standard germline testing for cancer risk.

"These findings not only demonstrate the feasibility of expanded germline testing but also its potential to bring lifesaving benefits to even more patients and improve personalized cancer care," said Kolesar, a professor in UK's College of Pharmacy who directs Markey's Precision Medicine Clinic and co-directs the MTB.

The study's findings also suggest a role in expanding germline testing for patients with other cancers including endometrial, lung and head or neck cancer.

More information: Megan L. Hutchcraft et al, Feasibility and Clinical Utility of Reporting Hereditary Cancer Predisposition Pathogenic Variants Identified in Research Germline Sequencing: A Prospective Interventional Study, *JCO Precision Oncology* (2024). DOI: 10.1200/PO.23.00266

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