Offering genetic testing at the point of care may increase uptake

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Genetic testing for hereditary cancers, such as breast, colon, pancreatic, and ovarian cancer, helps at-risk individuals understand their familial risk for these diseases and make informed decisions about next steps in

care. But fewer than 20 percent of at-risk patients utilize this testing, and even fewer engage in genetic counseling after referral, often due to clinical workflow challenges or barriers to care.

Amid national efforts to increase access to genetic testing, a new study led by a Boston University School of Public Health researcher has identified a streamlined approach in clinical settings that may help advance these efforts by simplifying the process of identifying hereditary cancer risk and determining subsequent care.

Published in the journal *Genetics in Medicine*, the study measured patient uptake of genetic testing in clinical practices that implemented a digital cancer risk assessment across different clinical workflow models, including a traditional referral, point-of-care scheduling, point-of-care counseling, and point-of-care genetic testing.

The findings showed that clinical practices that combined a digital cancer risk assessment along with point-of-care testing more than doubled the average uptake of genetic testing. While this streamlined strategy shows promise for cancer prevention and detection, the researchers say more work needs to be done to increase utilization of these valuable services.

"This study is one of the largest to show the possible advantages of a scalable mainstreamed approach to facilitating greater uptake of genetic testing across a variety of clinical settings," says study lead and corresponding author Dr. Catharine Wang, associate professor of community health sciences at BUSPH. "In spite of this advantage, however, overall testing rates varied widely among sites deploying this approach, suggesting that there is still much room for improvement."

For the study, Dr. Wang and colleagues from BUSPH and digital healthcare company CancerIQ, Inc. analyzed data among approximately

33,000 high-risk patients (out of more than 100,000 screened) who met genetic testing criteria for breast and ovarian cancer, and/or Lynch syndrome, an inherited disorder that increases risk for many types of cancer. Providers in 27 health centers or primary care/specialty offices conducted a hereditary cancer risk assessment using the CancerIQ digital precision prevention platform that optimizes clinical workflow through a range of automated processes, including gathering data and categorizing patients into different risk tiers, streamlining the genetic counseling and testing process, and creating personalized care plans.

In the traditional referral workflow, patients were referred to a genetic specialist who called to schedule an appointment; with point-of-care scheduling, they scheduled testing with a specialist immediately during their appointment; with point-of care counseling, the patients were offered an immediate consult by with a specialist; and with point-of-care testing, they were offered immediate genetic testing once they were determined to be at risk.

On average, 16 percent of high-risk patients opted for genetic testing overall, from 35 percent of patients at offices that implemented the point-of-care testing workflow, to just 6 percent of patients at offices utilizing the referral process. The point-of-care testing model enables doctors to educate and discuss the testing with patients directly, rather than necessitate an additional pretest appointment with a genetic counselor.

The researchers say further work should address continued challenges with this workflow, such as physicians' hesitation to order tests or counsel patients, and patients' concerns about insurance coverage.

"We still have a lot of learn about best practices for increasing access to cancer genetic services, particularly among patients who are medically underserved and face numerous logistical and structural barriers to accessing care," Dr. Wang says. "Given these current constraints, it is

important to examine alternate models of care delivery, and this study suggests that point-of-care testing is an effective delivery model for improving genetic testing outcomes."

The senior author of the study is Dr. Ziming Xuan, professor of community health sciences at BUSPH. The study was coauthored by Haibo Lu, cofounder and chief data officer of Cancer IQ, and the late Dr. Deborah Bowen, who was a professor in the Department of Bioethics and Humanities at the University of Washington School of Medicine.

More information: Catharine Wang et al, Implementing digital systems to facilitate genetic testing for hereditary cancer syndromes: An observational study of 4 clinical workflows, *Genetics in Medicine* (2023). DOI: 10.1016/j.gim.2023.100802

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