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Title: Expanding the reach of germline genetic testing: Use of web-based risk assessment to inform medical management amongst patients at breast and imaging centers

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Background

Developing an effective approach to the identification of individuals at increased cancer risk is key to preventing and/or providing early diagnosis of cancer. However, outside of targeted genetics clinics, under identification of individuals with hereditary cancer risk is well recognized, due in part to ever evolving complexity of germline genetic testing criteria and lack of systematic framework to perform robust risk assessment on all patients. In contrast, breast and imaging centers are ideally positioned to maximize the impact of positive genetic test results due to immediate availability of surveillance and diagnostic tools. Here we present data from breast and imaging centers using a patient-facing digital platform offered universally to all patients before their scheduled appointment designed to collect personal and family health information and assess cancer risk and genetic testing eligibility based on current guidelines.

Methods

We conducted a retrospective observational study of patients in breast and imaging centers who used a web-based risk stratification tool before standard ambulatory appointments to assess their lifetime risk for breast cancer based on the Tyrer-Cuzick (version 8.0) risk algorithm and eligibility for National Comprehensive Cancer Network (NCCN®) genetic testing criteria at the time of assessment. Testing criteria included hereditary breast, ovarian, pancreatic, and prostate cancers, Lynch syndrome, and familial adenomatous polyposis (FAP). Data was pulled for patients seen from June 2020 through May 2022 at participating breast and imaging centers throughout the United States. Outcome measures included percentage of individuals who completed the risk-assessment, met testing criteria, pursued germline genetic testing, received a positive germline result, and/or had a Tyrer-Cuzick breast cancer risk ≥20%.

Results

A total of 251,492 individuals completed assessments; 250,011 (99%) were females aged 18 years or older. Overall, at the time of assessment 80,814/251,492 (32.1%) met genetic testing criteria and 24.4% (19,694) of those meeting criteria opted to proceed with germline genetic testing. An additional 1,561 individuals who did not meet criteria pursued genetic testing. Of the 18,532 completed genetic tests, 1,507 (8.1%) had positive genetic test results. The majority of positive individuals (93%) met testing criteria. 40.7% (613/1,507) of positive results had an impact on breast cancer risk management options. In addition to individuals identified as high-risk through germline genetic testing evaluations, 13.1% (28,108/214,269) of individuals assessed using the Tyrer-Cuzick algorithm had ≥20% lifetime risk of breast cancer and met the threshold for modified medical management.

Conclusion

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In this study, the web-based assessment tool provided a standardized workflow that enabled individuals interested in receiving cancer risk assessment and germline testing an opportunity to do so. When offered to all patients, this digital platform can offer a scalable opportunity for breast and imaging centers to identify individuals eligible for modified medical management for breast cancer risk and other inherited cancer syndromes, which may ultimately improve the prevention and early treatment of individuals with cancer predisposition.