Title: Identification of High-Risk Hereditary Cancer Syndrome Patients in a Primary Care Cohort Using a Patient Facing Risk Stratification Tool

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The increasing utility of genetics in medicine has placed responsibility onto primary care clinicians as gatekeepers for genetics referrals and testing. However, physicians and nurses have reported a lack of formal education in genetics, which is made worse by frequently evolving testing guidelines. While research on innovative service delivery interventions and patient educational programs are limited, a variety of digital tools have been shown to be effective in identifying individuals at high-risk for hereditary cancer syndromes. This study aims to evaluate the effectiveness of a digital, patient-facing risk stratification tool in identifying patients who would benefit from genetic testing and counseling in comparison to clinician-based risk assessment performed in a rural primary care practice.

This IRB-exempt retrospective study evaluated a patient-facing digital tool (CARE platform™) designed to collect personal and family history to determine genetic testing eligibility based on NCCN criteria. We compared the testing eligibility rate of 896 patients undergoing cancer risk assessment with the tool during visits made from September 2022-February 2023 (cases) to clinician-based hereditary risk assessment data obtained from electronic medical records of 1039 patients from the same primary care sites from September 2018-July 2022 (controls).

A total of 594 patients completed assessment via the risk stratification tool (66.4%), suggesting an acceptance of the digital assessment approach. Compared to clinician-based assessment, in which 0.8% of patients were recognized as meeting criteria for testing, the digital tool was significantly more effective in identifying high-risk patients (11.1% of 896 patients seen [$p<0.001$; OR 16.0], and 16.6% of 594 who completed assessment [$p<0.001$, OR 25.7]). Extrapolation of the proportion of patients meeting criteria in the case group suggests that up to 89.5% of patients that could have benefitted from genetic counseling and testing in the control group were missed.

The implementation of the digital patient-facing risk stratification tool enhanced the identification of individuals who may benefit from genetic testing and subsequent cancer risk management in a rural primary care setting. The platform can standardize the process of finding eligible patients across clinics thereby improving access to genetic testing and counseling. The program can identify patients at risk for hereditary cancer syndromes and alleviate primary care clinician burden in staying up to date with testing recommendations.