Title: Digital cancer genetic risk assessment in a gynecologic oncology clinic

Emily Webster, Muhammad Danyal Ahsan, Luiza Perez, Leslie Bull, Laura Keenahan, Sarah R. Levi, Shannon M. Kieran, Evelyn Cantillo, Eloise Chapman-Davis, Kevin Holcomb, Ravi N. Sharaf, Melissa Kristen Frey; Weill Cornell Medicine, New York, NY; Ambry Genetics, Aliso Viejo, CA; Weill Cornell Medical College, New York, NY

Background: Population-based screening for hereditary cancer syndromes and referral of those who screen positive for genetic testing is a cost effective, evidence-based national health priority. However, under-recognition of hereditary cancer syndromes remains a critical concern, with fewer than 20% of individuals with Hereditary Breast and Ovarian Cancer Syndrome and Lynch Syndrome identified. Here, we evaluate the Ambry Genetics Comprehensive, Assessment, Risk, and Education (CARE) Program™, a risk-stratification tool to assess eligibility for National Comprehensive Cancer Network (NCCN®) genetic testing criteria and lifetime risk for breast cancer based on the Tyrer-Cuzick (version 8.0) risk algorithm.

Methods: Between November 2022 and February 2023, new patients in a gynecologic oncology clinic were prompted by text message to complete CARE™ prior to their appointment. Those using CARE™ then completed a System Usability Scale (SUS), a validated 10-item questionnaire for the assessment of perceived usability.

Results: Sixty patients presented for a new patient gynecologic oncology appointment and were prompted to complete CARE™. The median patient age was 55 years (interquartile range, 40-68). The patients self-identified as Non-Hispanic White (44, 73%), Non-Hispanic Black (5, 8%), Asian (5, 8%), Hispanic (3, 5%), other (2, 3%), and unknown (1, 2%). All identified as female. Forty-three patients (72%) completed CARE™. Among CARE™ users, 11 (26%) were identified as meeting criteria for genetic testing; of these, 9 (82%) proceeded with point-of-care same day genetic testing. Nine (21%) patients were eligible for enhanced breast cancer screening based on an estimated lifetime breast cancer risk of 20% or greater; of these, 100% were referred for high-risk breast screening. There were no patient demographics associated with completion of CARE™. Forty-one patients (72%) completed the SUS survey; the median SUS score was 80 (interquartile range, 60-92.5), which corresponded to 85-89th percentile and a letter grade of A-.

Conclusions: Implementation of a digital tool for cancer genetic risk assessment identified a quarter of patients to be eligible for genetic testing, among whom 82% completed same-day genetic testing. Additionally, the tool was reviewed favorably by patient users. Large randomized controlled trials of health information technology tools are needed to move closer to the goal of population-based hereditary cancer risk assessment.