IMPLEMENTATION OF A DIGITAL CANCER RISK ASSESSMENT TOOL AND GENETIC TESTING PROGRAM IN GASTROENTEROLOGY PRACTICES THROUGHOUT THE U.S. IDENTIFIES INHERITED CANCER SUSCEPTIBILITY SYNDROMES IN 15.6% OF PATIENTS TESTED

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Introduction:

Identifying patients at high-risk for cancer due to an inherited cancer predisposition is important to ensure proper management, but consistent, accurate, and scalable risk assessment is challenging. A web-based cancer risk assessment tool and genetic testing program has been available to ACG members since 2021 as means to streamline family history risk assessment, genetic education, genetic testing, and post-test genetic counseling by telehealth. This tool is designed to systematically assess all patients in a practice regardless of age or GI condition.

Methods:

Seven practices consisting of 17 GI providers have implemented this service between Sept 2021-May 2023. Depending on the practice all patients or patients considered high-risk were invited via text or email to complete a risk assessment via the web-based program (Figure 1). In some practices, only patients considered high-risk received the invitation.

Results:

Out of 25,349 patient appointments, 9,340 (36.8%) assessment invitations have been sent and 5,830 (62.4%) completed. 1,470 of 5,830 (25.2%) met NCCN criteria for genetic counseling and testing, 403 had genetic testing ordered (27.4%) and 302 (74.9%) completed testing. Of those tested, 47 (15.6%) were found to have 49 pathogenic/likely pathogenic variants (P/LPV) in a cancer susceptibility gene, 71 (23.5%) were found to have variants of uncertain significance, and 185 (61.3%) tested negative (Table 1). 12 individuals were diagnosed with high-risk cancer susceptibility syndromes; 7 with Lynch syndrome, 4 with Hereditary BreastOvarian cancer syndrome, and 1 with Li Fraumeni syndrome. There were 13 individuals with P/LPVs in moderate-risk genes (APC p.I1307K, ATM, CHEK2, HOXB13, and SDHA). 24 individuals were carriers of autosomal recessive disorders not affecting their cancer risk but important for reproductive risk counseling.

Discussion:

Systematic implementation of a digital risk assessment tool can identify patients with high-risk cancer susceptibility syndromes. Here we used such a tool to screen nearly 6,000 patients leading to testing of 302 and the identification of 47 individuals with management and/or reproductive implications. Most of these individuals may not have been diagnosed otherwise. Syndrome identification allows implementation of intensive surveillance, including earlier colonoscopy depending on pathogenic variant, to prevent or diagnose cancer earlier. Cascade testing can be offered to relatives.