ORIGINAL ARTICLE

Family history in colonoscopy patients: feasibility and performance of electronic and paper-based surveys for colorectal cancer risk assessment in the outpatient setting

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Background and Aims: Family history is crucial in stratifying patients' risk for colorectal cancer (CRC). Previous risk assessment tools developed for use in clinic or endoscopy settings have demonstrated suboptimal specificity for identifying patients with hereditary cancer syndromes. Our aim was to test the feasibility and performance of 2 family history surveys (paper and electronic) in individuals presenting for outpatient colonoscopy.

Methods: Patients presenting for outpatient colonoscopy at a tertiary care center were asked to complete a 5-question paper risk assessment survey (short paper survey) either alone or in conjunction with a second, comprehensive electronic family risk assessment survey (comprehensive tablet survey). Each subject's survey results, along with the electronic medical record, were reviewed, and 10 high-risk criteria and PREMM1,2,6 model scores (a predictive model for carrying a Lynch syndrome-associated gene mutation) were used to identify patients warranting genetic evaluation for suspected hereditary cancer syndromes.

Results: Six hundred patients completed the short paper survey (cohort 1), with an additional 100 patients completing both the short paper and comprehensive tablet survey (cohort 2). Using 10 high-risk criteria and/or a PREMM1,2,6 score $\geq$5%, we identified 10% and 9% of patients as high risk for CRC in cohorts 1 and 2, respectively. Of the 69 high-risk subjects, 23 (33%) underwent genetic evaluations and 7 (10%) carried germline mutations associated with cancer predisposition. Both patients and endoscopists reported the tools were user-friendly and helpful for CRC risk stratification.

Conclusions: Systematic assessment of family history in colonoscopy patients is feasible and can help endoscopists identify high-risk patients who would benefit from genetic evaluation. (Gastrointest Endosc 2017;■:1-8.)

Colorectal cancer (CRC) is the third most common cancer and the second leading cause of cancer death in the United States.[1] An individual's family history of cancer is a primary criteria used to determine both the age to start screening and the surveillance interval. Most CRCs are presumed to be sporadic; however, 30% of individuals with CRC have a family history of the disease and up to 5% to 6% of CRCs are linked to gene

Abbreviations: CRC, colorectal cancer; EMR, electronic medical record; FDR, first-degree relative.

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TABLE 1. Five-question paper risk assessment survey (short paper survey)

*1.	Do you have a first-degree relative (father, mother, siblings, child) with any of the following conditions?	
	• Colon or rectal cancer	Yes No
	• Cancer of the uterus, ovary, stomach, small intestine, urinary tract (kidney, ureter, bladder), bile ducts, pancreas, or brain	Yes No
	If the answer is yes to any of the above, were they diagnosed before age 60?	Yes No
*2.	Do you have three or more relatives (this includes grandparents, aunts, uncles, cousins) with a history of colon or rectal cancer?	Yes No
3.	Do you have a first-degree relative (father, mother, brother, sister, child) diagnosed with colon polyps before the age of 60?	Yes No
4.	Have *you* had any of the following conditions diagnosed before age 50?	
	• *Colon or rectal cancer	Yes No
	• *Colon or rectal polyps	Yes No
	• Cancer of the uterus, ovary, stomach, small intestine, urinary tract (kidney, ureter, bladder), bile ducts, pancreas, or brain	Yes No
5.	Have *you* had a total of 10 or more colon polyps removed in your lifetime?	Yes No

*Original question from Kastrinos et al.[15]

mutations associated with hereditary cancer syndromes, with Lynch syndrome (hereditary nonpolyposis CRC) and familial adenomatous polyposis among the most common.[2]

Multiple studies have shown that family history assessments performed in primary care and in oncology and gastroenterology clinical settings are incomplete or inaccurate.[3-7] In a review of screening colonoscopy records from a statewide colonoscopy registry, Butterly et al[7] found that 17% of patients with a known family history of familial adenomatous polyposis or Lynch syndrome were given inappropriate surveillance intervals of 10 years. As part of a quality improvement initiative conducted at the University of Michigan, a retrospective review of 200 patient records found incomplete documentation of family history of CRC in the endoscopy report and electronic medical record (EMR) in 25% of cases.[8]

A number of studies have demonstrated that family history information provided by patients is accurate for most common cancer diagnoses and is often significantly more complete than the information reflected in physician notes.[5,9-11] There has been interest in developing tools to help patients collect and enter their family history information for use in clinical care, such as the Centers for Disease Control's Family Healthware and the Office of the Surgeon General's "My Family Health Portrait" online family history tools[12,13]; however, these are not in general use in most healthcare systems.

There remains a need for targeted family history assessments to screen patients for hereditary cancer syndromes at point-of-care cancer screenings, such as colonoscopy. Calculation of PREMM1,2,6 model scores, a validated predictive tool for assessing likelihood of germline DNA mismatch repair mutations associated with Lynch syndrome, for all patients beginning at age 20 was shown to be cost-effective and to decrease the incidence of CRC by 43.9% compared with current practice.[14] Kastrinos et al[15] applied recursive partitioning analysis to family history surveys completed by colonoscopy patients and developed a 3-question survey to identify patients at higher risk for hereditary colon cancer syndromes. Validation of this tool in 6000 patients referred for direct-access outpatient colonoscopy categorized 15% to 20% as high risk for CRC and detected 95% of individuals known to carry germline mutations associated with CRC risk. Although this tool demonstrated excellent sensitivity for identifying germline mutation carriers in this and other studies,[16] the low specificity leaves room for improvement because it is not feasible for 20% of all colonoscopy patients to be referred for genetic evaluation.

Our hypothesis was that expanding the information collected regarding patients' family history of cancer would improve the specificity for identifying patients at highest risk for genetic syndromes. Our aim was to evaluate the feasibility and performance of 2 family history survey instruments, 1 paper-based and 1 electronic-based, for CRC risk assessment in the outpatient colonoscopy setting.

METHODS

Design of tools

Tool 1: paper risk assessment survey (short paper survey). We developed the short paper survey (Table 1) by modifying the questions previously validated by Kastrinos et al,[15] adding key components of current medical guidelines: (1) personal history of a Lynch syndrome–associated malignancy (a feature of the revised Bethesda criteria)[17]; (2) personal history of 10 or more polyps (a feature of the 2014 National Comprehensive Cancer Network criteria for genetic evaluation)[18]; and (3) a family history of colon polyps (American Gastroenterological Association guidelines).[1] Additional adjustments were made based on physician feedback received during the initial rollout of the survey at the University of Michigan, namely changing the ages at diagnosis of CRC and Lynch syndrome–associated cancers in first-degree relatives (FDRs) from age less than 50 to age less than 60 to coincide with the American Gastroenterological Association CRC surveillance guidelines.[1]