GENETICS AND GENOMICS

Leveraging Health Information Technology to Collect Family Cancer History: A Systematic Review and Meta-Analysis

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abstract

PURPOSE Collection of family cancer histories (FCHs) can identify individuals at risk for familial cancer syndromes. The aim of this study is to evaluate the literature on existing strategies whereby providers use information technology to assemble FCH.

METHODS A systematic search of online databases (Ovid MEDLINE, Cochrane, and Embase) between 1980 and 2020 was performed. Statistical heterogeneity was assessed through the chi-square test (ie, Cochrane Q test) and the inconsistency statistic (I^2). A random-effects analysis was used to calculate the pooled proportions and means.

RESULTS The comprehensive search produced 4,005 publications. Twenty-eight studies met inclusion criteria. Twenty-seven information technology tools were evaluated. Eighteen out of 28 studies were electronic surveys administered before visits (18, 64.3%). Five studies administered tablet surveys in offices (5, 17.8%). Four studies collected electronic survey via kiosk before visits (4, 14.3%), and one study used animated virtual counselor during visits (1, 3.6%). Among the studies that use an FCH tool, the pooled estimate of the overall completion rate was 86% (CI, 72% to 96%), 84% (CI, 65% to 97%) for electronic surveys before visits, 89% (CI, 0.74 to 0.98) for tablet surveys, and 85% (CI, 0.66 to 0.98) for surveys via kiosk. Mean time required for completion was 31.0 minutes (CI, 26.1 to 35.9), and the pooled estimate of proportions of participants referred to genetic testing was 12% (CI, 4% to 23%).

CONCLUSION Our review found that electronic FCH collection can be completed successfully by patients in a time-efficient manner with high rates of satisfaction.

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INTRODUCTION

In the United States, approximately four million individuals carry a pathogenic mutation in a cancer-associated gene.[1,2] Guided personalized medicine, with a focus on genetics, leverages information about one's unique genetics to tailor cancer-preventative strategies.[3,4] However, fewer than 20% of affected individuals are aware of their underlying genetic condition.[2] Specifically, significant barriers to genetic testing and educational opportunities exist for patients with hereditary breast and ovarian cancer syndrome and Lynch syndromes, despite their lifetime elevated risk for gynecologic and nongynecologic cancers.[5,6] The significance of a missed diagnosis is measured in lives lost, as medical and surgical interventions can often decrease mortality.[7] Collection of an accurate family cancer history (FCH) can help to identify individuals with cancer-associated pathogenic mutations.

Despite the known benefits of screening and testing for hereditary cancer syndromes, it has historically been difficult to execute and there is wide variability in the collection and accuracy of family health history across medical systems. Patients often cannot recollect the pertinent details about their relatives' cancer history in real time during a physician visit without assistance from other family members. Additionally, collecting a family health history is time-consuming, sometimes requiring more than 30 minutes. Limited appointment time and the lack of provider training with consistent documentation in the electronic medical record (EMR), therefore, can result in an incomplete or imprecise history.[8,9] Ideally, an effective tool to collect FCH should be self-administered, easily updated over time, and allow for automatic cancer risk assessment, referral to genetic counseling and cascade testing.

A potential solution is health information technology (IT). Health IT has been shown to successfully improve clinical documentation, clinical workflows, quality of care, patient safety, communication, and clinical decision support compared with handwritten paper documentation and in-person collection during a

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CONTEXT

Key Objective

The purpose of this study was to determine whether the utilization of health information technology to collect personal and family history improves the identification and care of individuals at risk for hereditary cancer syndromes.

Knowledge Generated

As the first large-scale, systematic review that focuses on the collection of family health histories via electronic or online tools, this study demonstrates that electronic family cancer history tools have high patient completion rates, acceptable time requirements, and high levels of user satisfaction and collection of complete health information.

Relevance

While health information technology for family cancer history collection holds the potential to improve detection rates of inherited cancer syndromes, additional studies are needed to measure the clinical efficacy through acceptance by patients and health care providers to increase genetic testing uptake, adaptation of the same tools in different clinical settings, and incorporation of the tools in electronic medical records.

medical appointment.[10] For cancer-associated syndromes, health IT can be used to collect personal and family history, identify high-risk patients, and calculate disease risk. Additionally, following genetic assessment, IT can coordinate care with other providers, communicate results with patients, and assist patients in sharing their test results with relatives at risk for carrying the same familial mutation, thus allowing for cascade testing.[10-13] With the growing recognition that it is critically important to identify individuals with cancer-associated pathogenic mutations and use health IT, we sought to systematically review the literature on health IT for family history collection as it is easily accessible and highly comprehensive.

METHODS

Inclusion Criteria

The current study was conducted in accordance with the Preferred Reporting Items for Systematic Reviews and Meta-Analyses guidelines and was preregistered with PROSPERO (registration no.: CRD42020193024).[14] The literature search strategy was designed around the PICO format: Is there a difference in FCH completion rate and time, genetic counseling referral rate, and testing (outcome) in all patients presenting to outpatient clinics and hospitals (population of interest) following collection of FCH using IT tools (intervention) versus traditional paper and in-person FCH collection methods (comparison)? This study protocol received exemption status from institutional review board committee approval. The final literature search was conducted on July 1, 2020, and assessed online publications between 1980 and 2020. We searched the following bibliographic databases: Ovid MEDLINE, Ovid Embase, and the Cochrane Library (Wiley). There were no language, publication date, or article-type restrictions included in the search, with the exception that all articles had to be original. Inclusion criteria for the review included original research papers; human focus; manuscript in peer-reviewed journal; and primary focus on the use of health IT to capture, collect, and/or collate information on FCH. For all identified references, two independent investigators reviewed titles, index terms, and available abstracts to determine whether the articles appeared to meet inclusion criteria. If insufficient information was available to make a decision at this stage, the article was included for full-text retrieval. Each full-text article was then reviewed to determine final inclusion status, which was included in a data collection form. Any discrepancies were resolved by discussion between the investigators to reach consensus.

For each of the articles meeting inclusion criteria, primary outcomes of interest consisted of patient population and setting, level of kinship collected, completion rate of all questions in the tool, completion time, incorporation into the EMR, resulting referral to genetic assessment, and patient satisfaction. We classified kinship as follows: first-degree relatives (FDR)—parents, siblings, and children; second-degree relatives (SDR)—grandparents, grandchildren, uncles, aunts, nephews, nieces, and half-siblings; and third-degree relatives (TDR)—great grandparents, great grandchildren, great uncles and aunts, and first cousins. Risk of bias in each study was evaluated using the Newcastle-Ottawa Scale.

Statistics

Meta-analysis models were used to estimate the proportion of individuals completing the FCH survey, the mean time required for survey completion, and the proportion of individuals referred for genetic testing as a result of information obtained via the survey. Statistical analyses were conducted with R software (Version 3.6.1 [July 5, 2019]; R Foundation for Statistical Computing, Vienna, Austria). Meta-analyses were conducted by tool type (electronic survey before medical visit, electronic survey via a tablet in the medical office, and electronic survey via a kiosk before visit). Statistical heterogeneity was tested through the chi-square test (ie, Cochrane Q test) and a P value $\leq .20$ was used to indicate the presence of heterogeneity. Statistical

heterogeneity was also assessed by the inconsistency statistic (I^2), with values > 50% considered as substantial heterogeneity. A random-effects analysis was used to calculate the pooled proportions and means. The random-effects analysis allows for more variability in the individual study proportion estimates when generating the pooled proportion and is more conservative. The pooled proportion was calculated using the Freeman-Tukey double arcsine transformation, and the 95% CI was calculated using the Clopper-Pearson interval, also called the exact binomial interval. We used the Freeman-Tukey double arcsine transformation because of its ability in stabilizing the variances. The studies included in this meta-analysis have a reported proportion of > 0.80, and when using other transformations, they grossly overstate the effects of proportions close to 0 and close to 1. The pooled mean time to completion was calculated by using the inverse variance method with untransformed means. To estimate the between-study variance, the DerSimonian-Laird estimator was used. For the outcome proportions of interest, the results of each study were expressed as binary proportions with exact 95% CIs.

Some of the included studies used both standard patient interview for collection of FCH and a health IT tool for FCH collection. For these studies, agreement between the two methods of FCH collection was assessed with Cohen's κ coefficient, reported in selected studies. Based on the Landis-Koch guidelines,[15] a κ value < 0.20 was regarded as poor agreement; 0.21-0.40 as fair agreement; 0.41-0.60 as moderate agreement; 0.61-0.80 as good agreement; and > 0.81 as very good concordance between the two tool methods of FCH collection.

RESULTS

Electronic Family Health History Collection Tools

The initial literature search identified 4,005 potentially relevant articles. Following review of titles and abstracts and based on inclusion and exclusion criteria, 103 articles were selected for full-text review and 28 articles were selected for inclusion (Fig 1). The included articles were published from 2000 to 2019, with the majority published after 2010 (25 studies). The studies included 27 unique tools for collection of FCH (Table 1). Seven of the FCH collection tools were developed outside of the United States. From the 28 studies included, the following health IT methods were used for FCH collection (Fig 2): electronic survey administered before visit (18, 64.3%), electronic survey via tablet administered in the medical office (5, 17.8%), electronic survey via kiosk in the hospital or medical office (4, 14.3%), and animated virtual counselor in the medical office (1, 3.6%). The virtual counselor VICKY is an animated computer character that helps to collect FCH using a touchscreen in the office. From 74 patients who were invited to the study, 70 patients completed FCH collection with VICKY (94.6%).

Patient Population

The 28 studies included 188,994 patients. The median patient age was 51.2 years (range 18-75 years). The percentage of female patients varied among publications (range 33%-100%). Studies were conducted in the inpatient or outpatient settings depending on the IT methods used. Targeted familial cancers differed based on the study design, patient population, and setting. Studies included a focus on cancer of the uterus, ovaries, stomach, GI tract, genitourinary system, hepatobiliary system, pancreas, and brain. Studies included variable levels of kinship, ranging from first degree to third degree.

Completion Rate

Among the 28 studies that used an FCH tool, the pooled estimate of the overall completion rate for health IT FCH tools was 86.0% (CI, 0.72 to 0.96; Fig 3A). The pooled estimated completion rate for health IT FCH collection via electronic survey using a tablet in the medical office was 89% (0.74 to 0.98), for health IT FCH collection completed via electronic survey before visit was 84.0% (CI, 0.65 to 0.97), and for health IT FCH collection completed via electronic survey using a kiosk was 85% (CI, 0.66 to 0.98); Figs 3B-3D). I^2 values were 99%-100% across all pooled analyses. Animated virtual counselor in the medical office was not pooled via meta-analysis because of the small sample size of the studies that used the tool.

Completion Time

Thirteen of the 28 studies (46.4%) reported a time needed for patients to complete the FCH tool, estimating a mean time pooled via meta-analysis of 31.0 minutes (CI, 26.1 to 35.9; Fig 3E). The quantile estimation method was used to estimate the sample mean and sample deviation for studies where median and range were reported. No material differences in the pooled mean completion time were observed among strata of kinship collected (FDR *v* SDR *v* TDR) as there were not enough data collected to quantitatively test for associations.

Interface With EMR

Seven (25.9%) of the 27 unique FCH collection tools had the capacity to automatically incorporate the health IT FCH tool results into the patient's individual EMR. In MeTree, provider reports were integrated into the medical record for use at the patient visit.[16] Risk factors from family history addressed by Health Heritage (ie, adenomatous polyps, *BRCA1* mutation, and elevated body mass index) were translated to the patient's record to enhance the geneticist's risk assessment through the EMR.[17,18] The Your Health Snapshot tool as mentioned in Baer et al[19] is a web-based risk appraisal tool that incorporates both family history and lifestyle factors in a patient's EMR. OurFamily Health stores family history simultaneously in a patient's EMR as the application is launched. Similarly, MyFamily delivers the patient's family history via a patient-specific report through

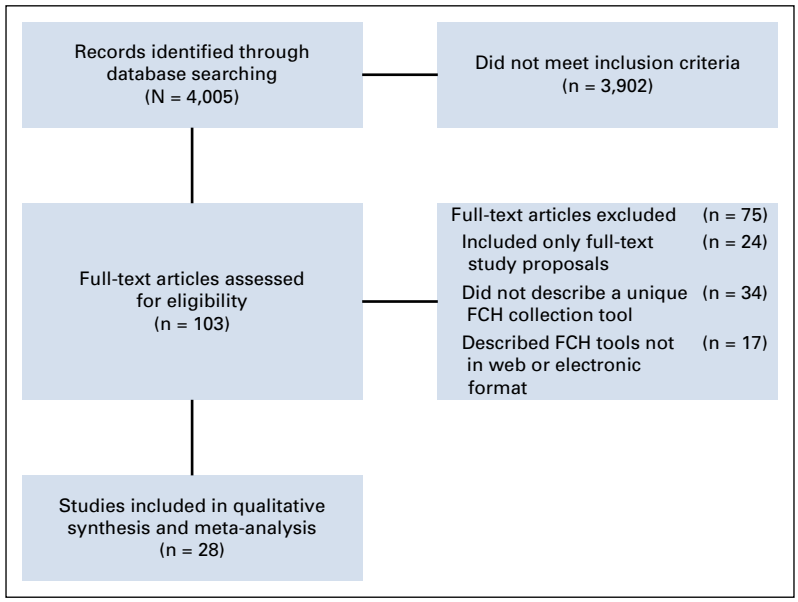

FIG 1. Preferred Reporting Items for Systematic Reviews and Meta-Analyses flow diagram of search strategy and study selection. FCH, family cancer history.

the EMR. However, with all seven studies, the incorporation was limited because of the different structures of EMRs available, making compatibility a main challenge.

Referral to Genetic Counseling and Testing

Nine studies (32.1%) captured information on referral to genetic counseling following the use of an IT FCH tool. After undergoing collection of FCH, pooled estimate of the proportion of participants referred to genetic testing was 12% (CI, 0.04 to 0.23; Fig 3F). Seven FCH collection tools (The GREAT survey, Cancer in the Family, ChMP, Colorectal Cancer Risk Prediction Tool, Genetic Risk Assessment in the Clinical Environment, MyFamily, and Ontario-FHAT) generated a report following data input that could be shared with patients and their relatives detailing their cancer risk assessment.

Correlation Coefficient

Five studies (17.8%) used both standard patient interview for collection of FCH and a health IT tool for FCH collection. These studies compared results from each method of information collection to assess for concordance using Cohen's κ coefficient. Overall, there was almost perfect agreement between the health IT FCH tools and previously used modalities in FCH collection. Four studies (80%; Guivatchian et al,[20] Kallenberg et al,[21,22] Baumgart et al,[17,18] and Facio et al[23]) demonstrated very good agreement ($\kappa > 0.81$; Table 2). When comparing agreement between the IT FCH and standard patient interview via simple pooled κ values, the electronic tools administered before the medical visit were equally concordant compared with electronic tool administered at the visit via tablet (0.92, 0.70-1.00 respectively).

Patient Satisfaction

Eight studies (28.6%) included a qualitative assessment of patient satisfaction with the health IT FCH tool. For each of these studies, the majority of patients completing the IT tools (51%-100%) found the tool easy to use and expressed satisfaction with the experience. Patients described the electronic survey via tablet in the medical office to be user friendly, the time for completion to be appropriate,[20] and the questions easy to understand.[24] Patients stated that the electronic survey before the medical visit was easy to use and stated that they were satisfied with being able to answer the questions at one's own pace.[16,21,22,25,26] For patients who used a kiosk system in the medical office, participants stated that it was conveniently available before the appointment with medical personnel present to help with troubleshooting.[27] The animated virtual counselor was described as interactive and participants rated it as stress-free and comprehensive.[28]

Bias Assessment

The risk of bias assessed using the Newcastle-Ottawa Scale was found to be high in 19 studies and medium in 9 studies studies. Funnel plots were generated to test for the presence of publication bias, demonstrating a slight asymmetry on visual inspection (Figs 4A-4F).

DISCUSSION

In this systematic review and meta-analysis, we have evaluated the literature on health IT strategies used to capture FCH. We identified 28 peer-reviewed studies using 27 unique FCH collection tools. The most commonly reported strategy of health IT FCH tool was an electronic survey administered either before the office visit or via a tablet in the office. Other reported strategies included kiosk-based interface in an outpatient clinic and an animated virtual counselor.

Meta-analyses were conducted for all patients as well as for the most commonly reported IT tools (electronic survey

TABLE 1. Characteristics of 27 Electronic FCH Collection Tools

Studies	FCH Collection Tool	Setting	Cancer Risk Investigated	Degree of Kinship	Incorporated Into EMR	Collection Tool Details
Everett et al[29]	Tablet-based survey	Tablet-based survey before appointment at a pancreatic tumor clinic	Pancreatic	Third	Yes	Comparison of FCH entry in EMR with tablet-based surveys Development of eight genetic counseling and testing referral criteria for pancreatic adenocarcinoma
Hulse et al[30]	Our Family Health, web-based patient-facing health history tool	Our Family Health online via patient portal	Colon and breast (among other chronic conditions)	Third	Yes	Query of detailed demographic, disease-related, or health behavior information of any member of the pedigree Users can add members to the tree in any direction, spanning outward to include up to TDR
Guivatchian et al[20]	Tablet survey	Waiting room before outpatient colonoscopy visits	Uterine, ovarian, stomach, GI, GU, hepatobiliary, pancreatic, and brain	Second	No	Collection of FCH for FDR and SDR Creation of a full family pedigree Identification of patients warranting genetic evaluation for hereditary cancer syndromes
Kallenberg et al[21,22]	Online survey	Outpatient colorectal clinic	Colorectal	Second	No	Collection of family history of CRC and Lynch syndrome–associated tumors in FDR and SDR
Baumgart et al,[17,18] Cohn et al[25]	Health Heritage, patient-facing web-based tool	Online survey before appointments at genetics' clinic	Melanoma breast, colorectal, ovarian, uterine pancreatic, and prostate	Second	Yes	Extraction of detailed clinical data from a patient's EMR Permission to exchange family history information to provide comprehensive risk Inclusion of risk assessment for hereditary cancer syndromes
Facio et al,[23] Thompson et al[31]	MFHP	Online survey before visit	Ovarian, breast, and colon	Second	No	Collection of family history of FDR and six conditions: heart disease, stroke, diabetes, colon, breast, and ovarian cancers
Pritzlaff et al[24]	CancerGene Connect (web-based program)	Four primary outpatient clinics	Breast, ovarian, colon, pancreas, and melanoma	Second	No	130 questions with branching logic covering family medical history Cancer risk assessment, psychosocial assessment, a result tracking system, and a patient follow-up system were included
Wu et al,[16] Orlando et al[32]	MeTree (patient-facing collection tool)	Kiosk at the hospital or primary care clinic	Breast, ovarian, colon, and thrombosis	Second	Yes	Collection of FCH into MeTree Risk stratification of patients for BC, ovarian cancer, CRC, thrombosis, and hereditary cancer syndromes Recommendation for risk-guided prevention strategies
Zimmerman et al[26]	ChMP (web-based history collection service)	Online surveys before visit	Breast	Second	No	Generation of a family tree using a pedigree drawing algorithm Risk assessment models provided quantitative and qualitative risk assessment for BC

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TABLE 1. Characteristics of 27 Electronic FCH Collection Tools (Continued)

Studies	FCH Collection Tool	Setting	Cancer Risk Investigated	Degree of Kinship	Incorporated Into EMR	Collection Tool Details
Westman et al[27]	FCH survey	Kiosks in the lobby of the cancer hospital and clinic	Not specified	Second	No	Collection of FCH by branched point decision making screens Decision tree mimicked face-to-face genetic counseling
Wang et al[28]	VICKY (Animated Virtual Counselor for Knowing Your Family History)	A large safety net hospital in New England	Breast and colorectal	Second	No	Collection of family health history on heart disease, diabetes, hypertension, stroke, and cancers
O'Neill et al,[33] Wang et al,[34] Rubinstein et al[35]	Family Healthware (self-administered, web-based tool)	Online or on a computer in office	Colorectal, breast, and ovarian	Second	No	Completion of baseline survey assessing health behaviors, lifestyle choices, risk perceptions for oneself, FDR, and SDR Generation of personalized risk assessments
Schultz et al[36]	Online survey	A science festival event	Colorectal	Second	No	Identification of patient's risk of developing CRC based on her family history up to SDR
Acheson et al[37]	GREAT	Online survey before outpatient appointments at the Center for Human Genetics	Not specified	Third	No	Record of age of diagnosis and cancer sites of FDR and SDR Collection of personal risk factors for cancer for the patients
Baer et al[19]	YHS, web-based risk appraisal tool	Primary care practices during annual examinations	Colorectal, lung, breast, and prostate	Second	Yes	Collection of cancer histories of FDR and SDR Risk estimates for cancers (colon cancer, lung cancer, BC, and prostate cancer)
Sweet et al[38]	JamesLink kiosk	JamesLink kiosk with touchscreen at an oncology ambulatory clinic	Breast, ovarian, prostate, melanoma, lung, and colorectal	Second	No	Branched-point decision-making screens for patient to enter familial cancer histories Classification of patients as high risk (hereditary cancer syndrome or early-onset cancer) received genetic counseling
Rupert et al[39]	Cancer in the Family (Online clinical decision support tool)	Online survey before wellness examinations	Breast and ovarian	Second	No	Internet-based tool with separate interfaces for patients and providers with information on hereditary breast and ovarian cancer Personalized risk results for patients and providers
Ozanne et al[40]	HughesRiskApps (open-source family history collection)	Tablet PCs in waiting rooms and examination rooms	Breast and ovarian	NA	No	Completion of family history on breast and ovarian cancers BRCAPRO and the myriad models estimated risks of carrying a *BRCA1* or *BRCA2* mutation
Slack et al[41]	Web-based family history tool	Seven primary care clinics	NA	NA	No	History collection using PatientSite portal with 24 modules and a total of 233 questions
Doerr et al[42]	MyFamily (a history collection and clinical decision support tool)	Web-based MyFamily survey via the portal	Colorectal, ovarian, and breast	NA	Yes	Construction of a family tree Generation of a patient-specific report: *MyFamily* report, which included risk assessment and education contents

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TABLE 1. Characteristics of 27 Electronic FCH Collection Tools (Continued)

Studies	FCH Collection Tool	Setting	Cancer Risk Investigated	Degree of Kinship	Incorporated Into EMR	Collection Tool Details
Braithwaite et al[43]	GRACE	Survey on the computer	Breast	Second	No	Family history information was collected up to SDR Risk assessment was divided into two sections: your risk assessment and how to manage your risk
Weigl et al[44]	RAPS for CRC survey	Web-based survey via computer or smartphone at the German Cancer Consortium	Colorectal	First	No	Inclusion of 97 questions: the participants' history of diseases, uptake of health examinations, family history of CRC, and environmental and lifestyle factors
Bredart et al[45]	BOADICEA BC risk assessment model	Online survey through the BOADICEA website	Colorectal, ovarian, and breast	NA	Yes	Inclusion of familial, hormonal, reproductive, lifestyle risk factors, and sociodemographic background contributing to BC Knowledge or usage frequency of BC risk assessment tools Genetic counseling and testing for cancer predisposition
Bellcross et al[46]	The Breast Cancer Genetics Referral Screening Tool (B-RST)	Online survey before genetic counseling for HBOC	Breast	Third	No	Full three-generation cancer pedigrees and genetic test results were obtained for patients with family history of BC Each pedigree analyzed using B-RST 2.0 to determine the risks (positive, negative—moderate risk, or negative—average risk)
Skinner et al[47]	CRIS	Touchscreen computer program	Colorectal	First	No	CRIS algorithms determined whether elevated-risk patients were up-to-date according to guidelines associated with their risk factors Screening orders and test completion were retrieved from the EMR at 6 months after visit
Harty et al[48]	CRISP	CRISP tablet at outpatient appointments	Colorectal	First	No	Collections of behavioral, environmental, and risk factors A combination of these risk factors produced an absolute risk score and provided screening advice
Thomas[49] (2018)	FHL	Interactive website of FHL at a comprehensive BC center	Breast	Second	No	Collection of personal and family history on FDR and SDR Risk assessment were created using scoring algorithms for all known hereditary cancer syndromes

Abbreviations: BC, breast cancer; B-RST, Breast Cancer Genetics Referral Screening Tool; ChMP, Collaborative Medical History Portal; CRC, colorectal cancer; CRIS, Cancer Risk Intake System; CRISP, colorectal cancer risk prediction tool; EMR, electronic medical record; FCH, family cancer history; FDR, first-degree relatives; FHL, Family HealthLink; GRACE, Genetic Risk Assessment in the Clinical Environment; GREAT, The Genetic Risk Easy Assessment Tool; GU, genitourinary; HBOC, hereditary breast and ovarian cancer; MFHP, My Family Health Portrait; NA, not available; RAPS, risk-adapted prevention strategies; SDR, second-degree relatives; TDR, third-degree relatives; YHS, Your Health Snapshot.

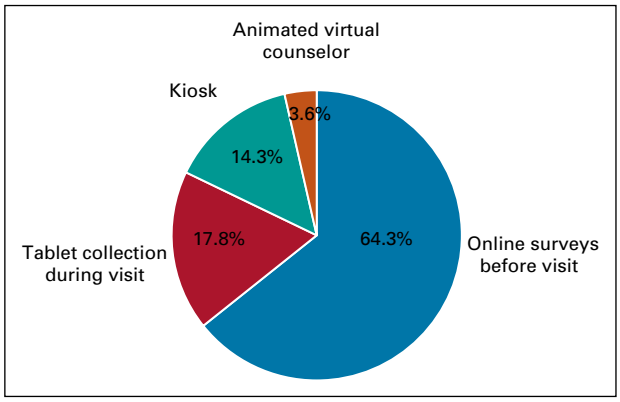

FIG 2. Four strategies for family cancer history collection.

before visit, and electronic survey via tablet in the medical office and via kiosk before visit). Across all IT tools, 86% (CI, 0.72 to 0.96) of patients completed FCH collection. Self-administered electronic surveys demonstrated a completion rate of 84% (CI, 0.65 to 0.97), tablet-based strategies had a completion rate of 89.0% (CI, 0.74 to 0.98), and kiosk-based strategies had a completion rate of 85% (CI, 0.66 to 0.98). Patients required approximately 31 minutes to complete the tool (CI, 26.1 to 35.9). Tools that collected FCH of higher degree of kinship did not always require more time for completion. Time potentially varied based on the branched-point decision-making tree: as higher degree of kinship is involved, the number of questions being asked would be affected. Completion of the FCH collection resulted in referral of 12% (CI, 0.04 to 0.23) of patients to genetic assessment. This result is similar to the national estimates of genetic testing in women with a history of breast or ovarian cancer: 15.3% underwent genetic testing with a history of breast cancer, and 10.5% underwent genetic testing with history of ovarian cancer.[2] All studies including a κ coefficient demonstrated at least good agreement and 80% demonstrated very good agreement between the electronic FCH collection tool with previously used FCH collection modality. Seven (25.9%) family history tools were designed to electronically incorporate family history information to the patient's EMR.

While previous systematic reviews have assessed different forms of family history collection tools,[50-52] this is the first large-scale, systematic review that focuses on the collection of family health histories via electronic or online tools. Paper-based questionnaires, telephone interviews, and face-to-face interviews during medical appointments can collect FCH and identify at-risk patients. However, there is a concern of standardization, exhausting of additional human resources, and whether these data can easily be retrieved and updated by all health care teams.[52] Compared with prior archaic FCH collection methods, our findings demonstrate that electronic and online tools are feasible methods for collecting family histories effectively with high completion rate, short completion time, greater patient satisfaction, capacity of incorporation into EMR, referral to genetic counseling, and almost perfect agreement between the health IT FCH tools and prior FCH collection methods.

When used either before the office visit or in the office via tablet, kiosk, or virtual-based system, FCH IT tools collected valuable family history that is often incomplete because of time constraints. By designating time before the physician evaluation for thorough collection, FCH IT tools allow for more time for face-to-face discussion once patients are with the physician.[16,27]

Many of the studies included in this analysis suggest similar characteristics that improve completion of FCH collection tools for detection of hereditary cancer syndromes. These tool characteristics include wide accessibility, self-administered by the patients, easy to use, presenting medical terms in an easy-to-understand format, displaying a pedigree, and including management plans to help patients understand hereditary cancer syndrome risk. As the COVID-19 pandemic continues to drive improved utilization of telemedicine and limited in-person face-to-face patient experiences, medical systems and physicians should keep these characteristics in mind to design electronic tools that will be well received and achieve the intended goal in their patient population.

This study has several limitations. Although this is a systematic review including studies of different patient populations and study designs, we were unable to control for selection bias and were not able to match patient characteristics. Because of the heterogeneity in findings across studies, the random-effects model was used. However, because the I^2 values are close to 100%, it is difficult to determine the generalizability of our results. Second, accessibility to an electronic device and internet connection from home can be a limitation in remote areas. While FCHs were promptly collected for FDR, SDR, and TDR, there was not a validation tool available among the different IT tools to confirm the histories collected besides the five studies that listed a Cohen's κ coefficient to demonstrate concordance (Table 2). Third, while missing data might be a significant issue, we do not have information regarding missing data available from the studies presented. Fourth, as the proportion estimates in this meta-analysis are heterogeneous across studies, the random-effects model is used with the assumption that each study's true transformed proportions follow the normal distribution. However, one limitation is that the Freeman-Tukey double arcsine transformation might violate this assumption because of bounded domains.[53] In terms of publication bias, while there does not appear to be strong publication bias present in the meta-analyses conducted in this study, there is a slight asymmetric nature to the funnel plots. Additionally, we were unable to obtain long-term data or follow-up results regarding patient compliance with genetic referrals or testing and therefore are unable to measure the clinical efficacy. However, a strength of this study is a large population size of

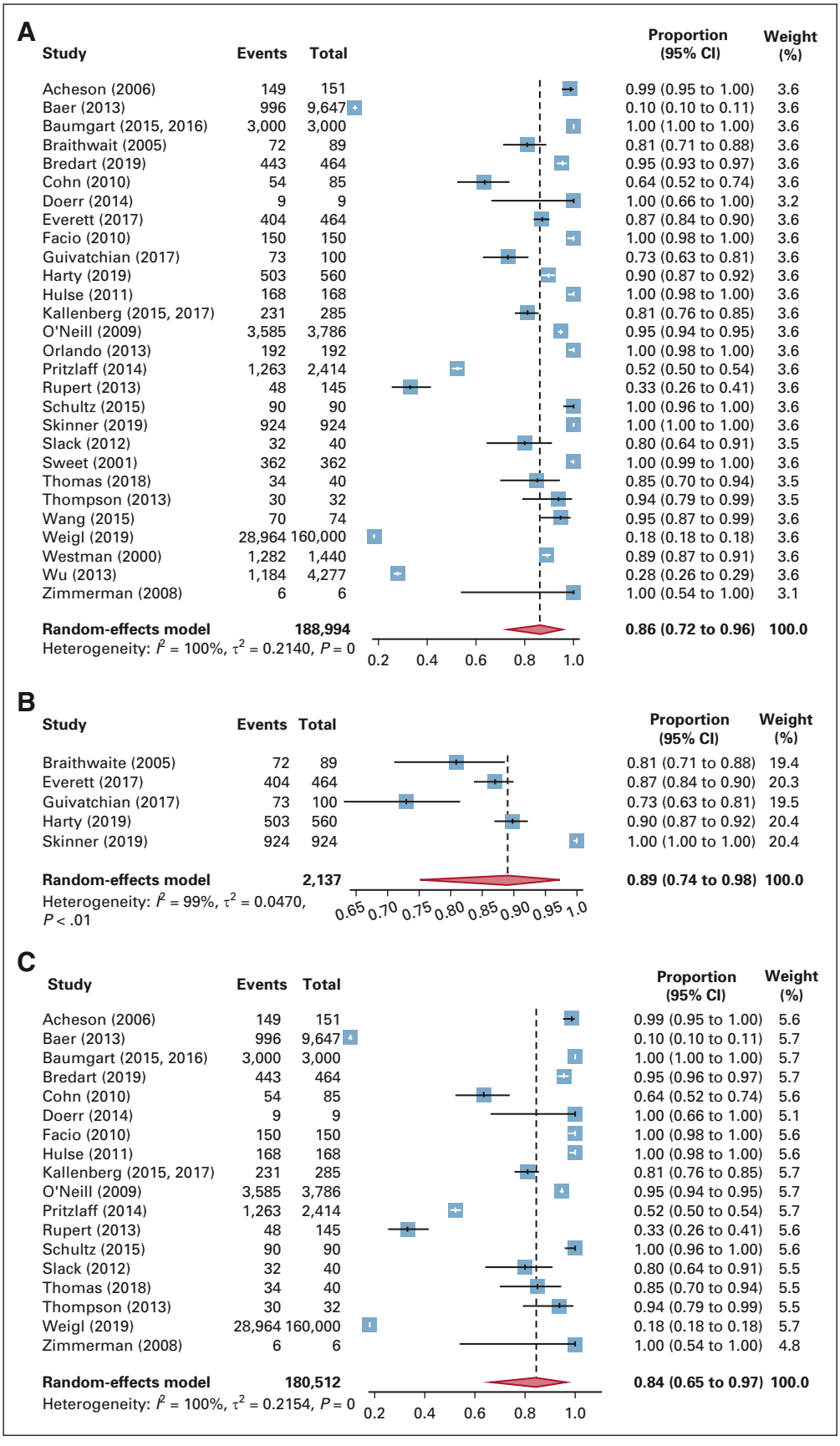

FIG 3. (A) Pooled proportion of patients completing any collection tool. (B) Pooled proportion of participants completing tablet collection tool. (C) Pooled proportion of participants completing online survey collection tool. (D) Pooled proportion of participants completing kiosk survey collection tool. (E) Pooled mean time to completion of family cancer histories collection tool. (F) Pooled proportion of participants referred to genetic testing. MRAW, pooled raw means.

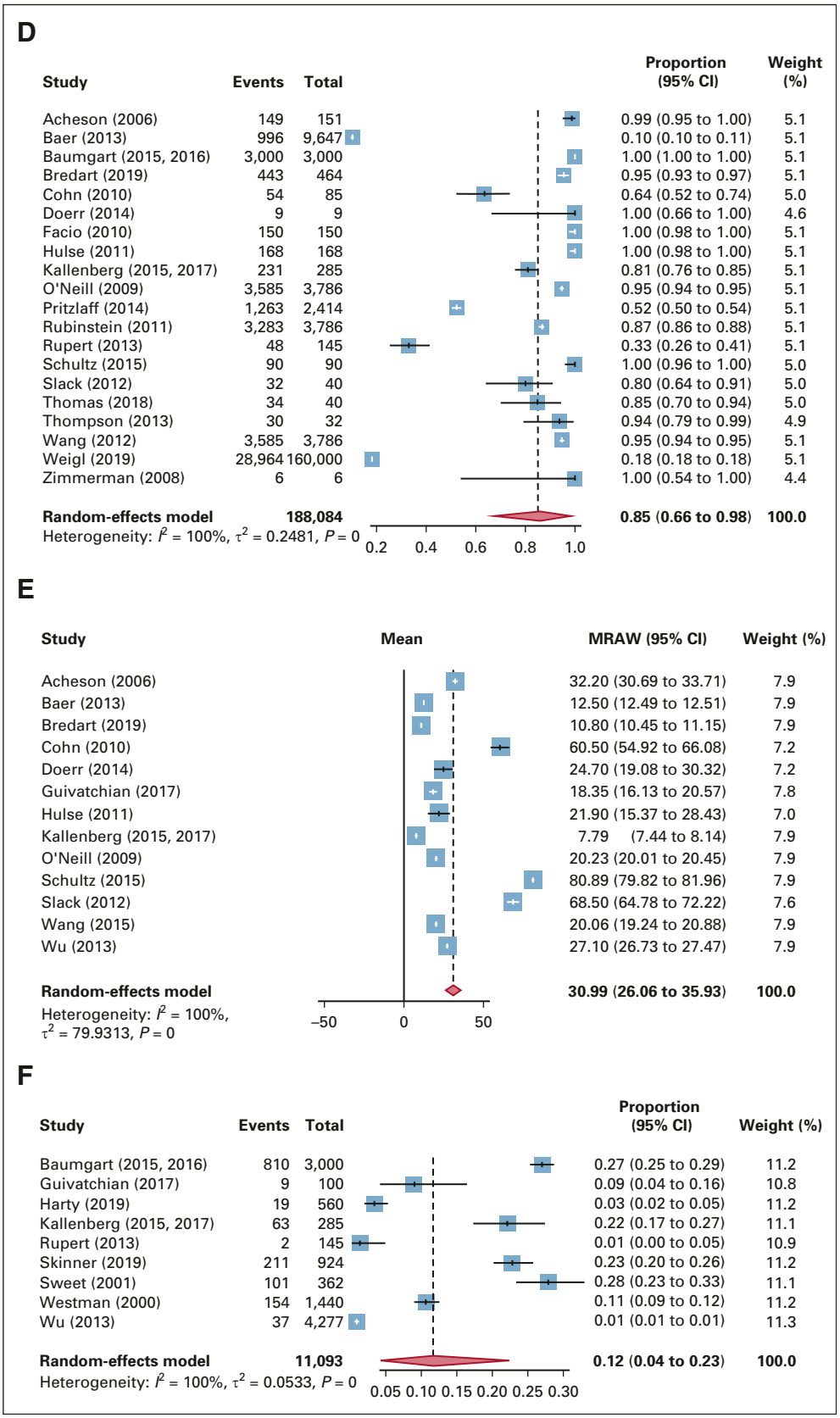

FIG 3. (Continued).

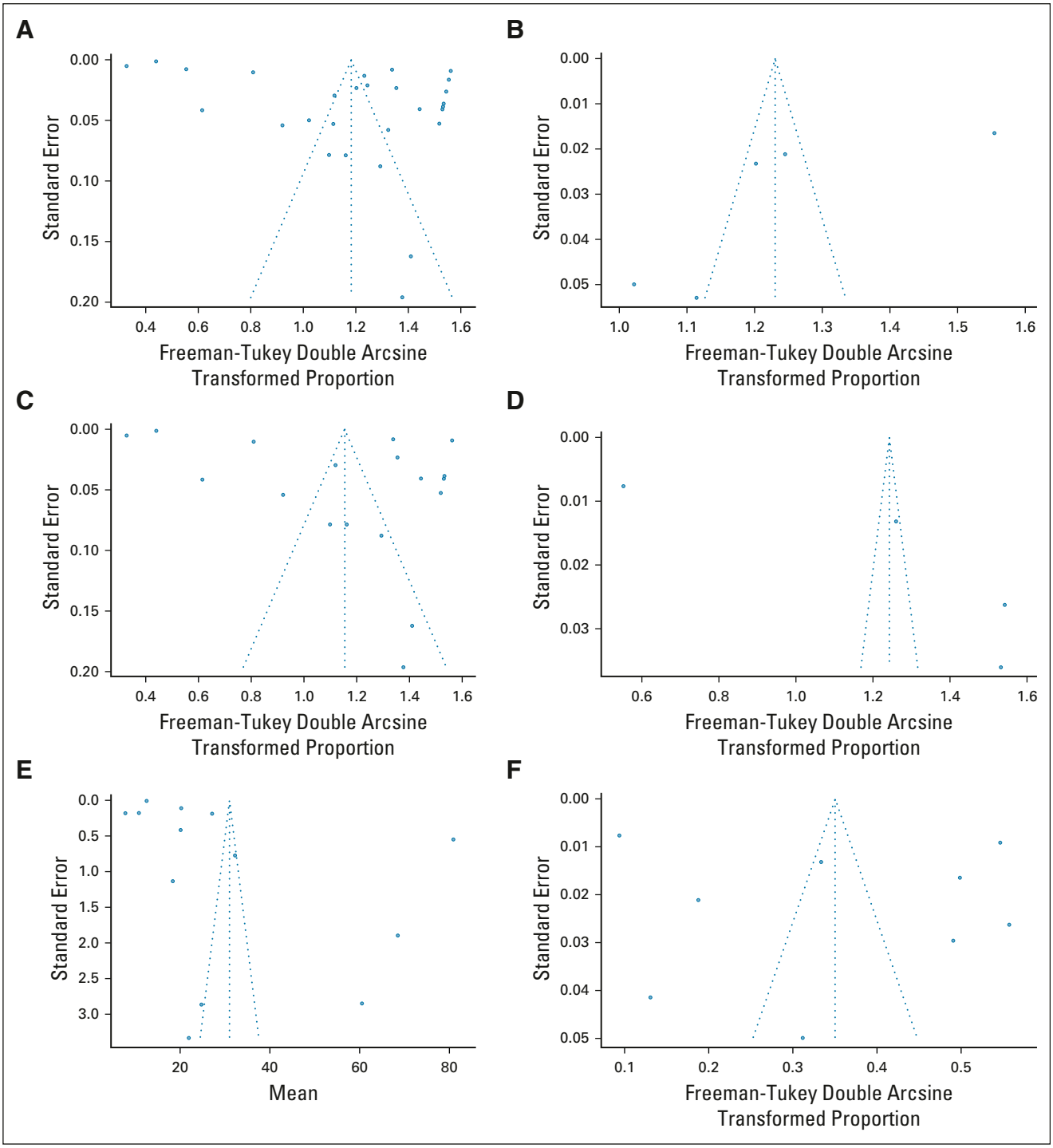

FIG 4. (A) Pooled proportion of patients completing any collection tool funnel plot. (B) Pooled proportion of participants completing tablet collection tool funnel plot. (C) Pooled proportion of participants completing online survey collection tool funnel plot. (D) Pooled proportion of participants completing kiosk survey collection tool funnel plot. (E) Pooled mean time to completion of family cancer histories collection tool funnel plot. (F) Pooled proportion of participants referred to genetic testing funnel plot.

mostly English-speaking participants, including close to 190,000 patients. This allowed for comprehensive review of using health IT to collect FCH and to increase awareness of hereditary cancer syndromes through subsequent genetic testing and counseling.

In conclusion, the collection of a patient's FCH is essential for triaging patients to genetic testing and counseling. There have been promising data in the literature regarding the use of electronic and online methods of family health history collection. However, this is, to our knowledge, the

TABLE 2. Concordance of the Electronic FCH Collection Tool With Previously Used Modality Using the Cohen's κ Coefficient

Study	Family History Collection Tool Platform	Concordance of the Electronic Tool With Previously Used Modality	Cohen's κ Coefficient
Guivatchian et al[20]	Tablet survey	Agreements between paper-based versus electronic comprehensive tablet survey modalities	Very good agreement (κ = 0.92)
Kallenberg et al[21,22]	Online questionnaire	Agreement in referral decision to genetic specialist between self-administered online questionnaire and a full pedigree data	Very good agreement (κ = 1)
Acheson et al[37]	GREAT, online-administered survey	Agreement between geneticist's BC risk assessments based on computerized tool versus counselor's pedigrees	Moderate agreement (κ = 0.70)
Baumgart et al[17,18]	Health Heritage, patient-facing web-based tool	Agreement between Health Heritage and NCCN guidelines	Very good agreement (κ = 0.81)
Facio et al[23]	MFHP online survey	Agreement between family history data collected by a genetics professional and semiautomated pedigree collection using MFHP	Very good agreement in three type of cancers: BC (κ = 0.95), ovarian cancer (κ = 0.91), and colon cancer (κ = 0.87)

NOTE. Based on the Landis-Koch guidelines: κ value < 0.20 was regarded as poor agreement; 0.21-0.40 as fair agreement; 0.41-0.60 as moderate agreement; 0.61-0.80 as good agreement; and > 0.81 as very good agreement between the two tool methods of FCH collection.

Abbreviations: BC, breast cancer; FCH, family cancer history; GREAT, The Genetic Risk Easy Assessment Tool; MFHP, My Family Health Portrait; NCCN, National Comprehensive Cancer Network.

first large systematic review and meta-analysis to address unique electronic FCH collection tools and encourage uptake of genetic testing and counseling through these tools. Our findings demonstrate that electronic FCH tools have high patient completion rates, acceptable time requirements, and high levels of user satisfaction and collection of complete health information. While Health IT for FCH collection holds the potential to improve detection rates of inherited cancer syndromes, additional studies are needed to measure the clinical efficacy through acceptance by patients and health care providers to increase genetic testing uptake, adaptation of the same tools in different clinical settings, and incorporation of the tools in EMRs. The long-term impact of such tools can be further evaluated by assessing uptake of cascade testing and risk-reducing surgeries.

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