CURRENT ONCOLOGY
A Canadian Cancer Research Journal

REVIEW ARTICLE

What characterizes cancer family history collection tools? A critical literature review

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ABSTRACT

Background Many tools have been developed for the standardized collection of cancer family history (FH). However, it remains unclear which tools have the potential to help health professionals overcome traditional barriers to collecting such histories. In this review, we describe the characteristics, validation process, and performance of existing tools and appraise the extent to which those tools can support health professionals in identifying and managing at-risk individuals.

Methods Studies were identified through searches of the MEDLINE, EMBASE, and Cochrane CENTRAL databases from October 2015 to September 2016. Articles were included if they described a cancer FH collection tool, its use, and its validation process.

Results Based on seventy-nine articles published between February 1978 and September 2016, 62 tools were identified. Most of the tools were paper-based and designed to be self-administered by lay individuals. One quarter of the tools could automatically produce pedigrees, provide cancer-risk assessment, and deliver evidence-based recommendations. One third of the tools were validated against a standard reference for collected FH quality and cancer-risk assessment. Only 3 tools were integrated into an electronic health records system.

Conclusions In the present review, we found no tool with characteristics that might make it an efficient clinical support for health care providers in cancer-risk identification and management. Adequately validated tools that are connected to electronic health records are needed to encourage the systematic identification of individuals at increased risk of cancer.

Key Words Family history, hereditary cancers, collection tools, screening, risk assessment, tools validation

Curr Oncol. 2018 Aug;25(4):e335-e350 www.current-oncology.com

INTRODUCTION

The role that heredity and familial exposure to nongenetic risk modifiers (lifestyle, environmental factors, health behaviours) play in cancer occurrence is well recognized[1,2]. With the development of genomic technologies, it has become easier to identify genetic mutations conferring an increased risk for developing malignancies. Eligibility for genetic testing is based on personal and familial health history criteria[3,4]. Collection of personal and family history (FH) is a noninvasive and relatively affordable way to perform a preliminary cancer-risk assessment and to identify individuals eligible for thorough genetic assessment[5,6]. Individuals found to be at increased risk might benefit from preventive and health promotion strategies[7]; those at average risk might be reassured[8]. Despite its essential role in cancer-risk prevention, FH is not systematically or adequately collected in clinical settings[9,10]. Hence, at-risk individuals remain unidentified[11,12]. Furthermore, when FH is collected, followed-up recommendations are not always provided[13,14]. Thus, at-risk individuals might not be referred to the appropriate resources[9,15] or might be falsely reassured[16].

Barriers to collecting FH in clinical settings include poor reimbursement, provider's lack of time and expertise, lack of guidelines and adequate tools, and limited functionality of electronic health information systems to capture and interpret FH data[17]. The use of adapted FH collection tools could potentially alleviate some of those barriers and assist providers in collecting and interpreting FH. Many

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tools have been described in the literature, and several systematic reviews have attempted to group them according to analytic perspective[10,18–20]. However, it is unclear which tools have the potential to help health professionals overcome traditional barriers to collecting cancer FH.

According to de Hoog *et al.*[20] and Taylor *et al.*[21], an ideal tool for collecting FH should be self-administered by patients, computerized, easy to use, and preferably linked or integrated into an electronic health record (EHR). An ideal tool should also allow for the FH to be easily updated over time. It should be designed to draw pedigrees, perform a criteria-based cancer-risk assessment, and deliver tailored, evidence-based management recommendations.

In the present review, we describe the characteristics, validation process, and performance of existing FH tools to determine which ones meet the criteria of the ideal tool and to assess the extent to which they can help clinicians in cancer-risk assessment and management. In contrast to prior works, our review focuses on cancer FH and considers all types of cancer in the adult population. It takes into account validated and non-validated tools to produce a broad picture of available tools used in both primary care and specialized clinics.

METHODS

Data Sources and Inclusion Criteria

We searched the MEDLINE, EMBASE, and Cochrane CENTRAL databases using combinations of the words "family history," "taking," "collection," "assessment," "cancer risk," "tools," "cancer," "questionnaire," "instrument," and "validation." We applied the PubMed function "similar articles" to articles meeting our inclusion criteria and searched the lists thus generated by PubMed. We also manually searched bibliographic references of retrieved articles and systematic reviews. No time limit was applied to the search. The final literature search took place on 1 September 2016.

To be included in the analysis, articles had to meet these inclusion criteria: publication in English or French, description of a tool used to collect cancer FH in adults, primary focus on the collection of FH, and evaluation of the potential benefits and psychological impacts of a FH collection tool. Articles mentioning a FH collection tool as part of the methods without describing the instrument were excluded. Pertinent references were first identified by scanning titles. Index terms and available abstracts were subsequently reviewed to determine whether articles met the inclusion criteria. Final inclusion was based on a full-text review of selected articles. Relevant information was extracted from the articles retained at that final step.

Data Extraction and Analysis

A descriptive approach was used to summarize the features and validation process of the retained tools. The tools were described based on characteristics previously reported in three systematic reviews[18–20], but user experience was also included in our analysis. An Excel database (Microsoft Corporation, Redmond, WA, U.S.A.) containing 51 variables was created to describe study and tool characteristics, including tool name, first author name, year of publication, country, and setting in which the tool was tested or used. Properties and attributes of the tools were grouped to build the framework used to conduct our analyses and to present results. Components of that framework included diseases or cancers targeted, tool format, and the tool's capacity to produce pedigrees, perform risk assessment and stratification, deliver recommendations. Data were processed and aggregated using the SAS software application (version 9.4: SAS Institute, Cary, NC, U.S.A.). Article screening, data extraction, and analysis were performed by the first author (JEC).

Analysis of Tool Performance

Two approaches were used to summarize and interpret tool performance. First, we combined papers in which tools were partly validated using the ACCE framework criteria (analytic validity; clinical validity; clinical utility; and ethical, legal, and social issues; Table I). The ACCE framework, commissioned by the U.S. Centers for Disease Control and Prevention, is dedicated to the assessment of the benefits and risks of genetic tests[23,24]. As proposed by Qureshi *et al.*[22], the framework can be used to evaluate FH collection tools.

Our second approach consisted in combining articles that had used performance indicators different from those suggested by the ACCE framework to evaluate the thoroughness of the FHS collected and the cancer-risk assessment ability of the tool. Concordance between tools and chosen references (for example, genetic counsellor, medical chart) was measured according to the various aspects of FH and cancer-risk assessment. Evaluation strategies and results were summarized in a table.

RESULTS

Tools Identified, Country and Setting of Use, and Target Users

Tables II and III present 62 FH collection tools that matched our criteria, identified from seventy-nine publications[11,12,14,16,25–99]. Most were developed in the United States and the United Kingdom (73%), almost half were used in primary care settings (47%), and more than three quarters were devised to be self-administered by lay individuals (78%).

Types of Tools Based on the Diseases Targeted

The identified tools could be classified as generic or cancer-specific. Generic tools (n = 17) allow for the collection of FH for several medical conditions, including cancers (Table II). Specific tools (n = 45) focus on the FH for one or several types of cancer or cancer syndromes (Table III). Most frequently, FH is assessed for breast, ovarian, colon, and prostate cancers.

Format of the Identified Tools

Tools for FH collection could be divided into three categories: paper-based (n = 31), interview-based (n = 10), and electronic (n = 21). Paper-based questionnaires are intended to be completed at home or in the clinic. They consist of structured, open-ended, and closed-ended questions; tables[28,33,53,65,78,79,82,84,85,96]; organigrams[66]; or pedigrees[26,27]. Interview-based tools consist of automated

TABLE I Application of the ACCE framework[a] to family history as a screening tool

	Framework element	Definition	Components
A	Analytic validity	An indicator of how a family history tool measures the characteristic (family history) that it intends to measure	■ Analytic sensitivity and specificity
C	Clinical validity	A measure of the accuracy with which a risk assessment tool based on family history information predicts disease risk	■ Clinical sensitivity and specificity ■ Positive and negative predictive values
C	Clinical utility	The degree to which benefits are provided by using a clinically valid risk assessment tool based on family history information	■ Availability of effective preventive and clinical interventions ■ Health risks and benefits of preventive and clinical interventions ■ Health risks and benefits of family history and risk assessment tools ■ Economic assessment
E	Ethical, legal, and social implications	Issues of data collection and interpretation that might negatively affect individuals, families, and societies	■ Stigmatization ■ Discrimination ■ Psychological harm ■ Risks to privacy and confidentiality

[a] Qureshi *et al.,* 2007[22]; adapted from: Yoon *et al.,* 2003[23].

telephone-based interviews[55,75], structured computer-assisted telephone interviews[87], and face-to-face interviews[54,56,61,76,80,93,98]. Non-automated telephone-based and face-to-face interviews are conducted with the support of structured questionnaires or pedigree information sheets[54,56,61,76,80,87,98,93].

Electronic tools allow for interactive question-answering in a logical process that uses dialog boxes[30–32,63,64], drop-down windows[62,81], or diagrams from which pedigrees can be built[97]. Three electronic tools display lists of possible or preformulated answers[45,46,51,95]. Three others have blank spaces[49,81] or empty pedigrees[40] that have to be completed. One electronic tool is a question prompter intended to be used by physicians during patient interviews[74]. Electronic questionnaires are available for use on a digital assistant[74], a tablet[51,86], a laptop[47,86], or a computer in the clinic[41,47,62,63,70,86]. They can be made accessible through the Internet[11,30–32,34,39,41–44,47,81,89,91,95,97], an online patient portal[40,45,46,49], or an intranet[71,72]. Updates to the FH are possible with 3 of the electronic tools[42,86,89]. Another 3 tools are incorporated into or linked back to EHRs[40,47,92].

Degree of Kinship Covered and Pedigree Production

Information about the degree of kinship covered was available for 53 tools (85%). Of those 53 tools, 13% ask respondents only about 1st-degree relatives; 87% and 49% include 2nd- and 3rd-degree relatives respectively. Almost half the tools ($n = 29$, 47%) are geared toward production of a pedigree, with 14 of them (23%) automatically producing a pedigree after the entry of FH data. Except for 1 automated telephone interview[75], all of those tools are electronic. Another 15 tools (24%) allow for the detailed collection of FH in a way that a pedigree can subsequently be constructed. Nevertheless, 32 tools (52%) are disease-oriented, seeking only a positive FH of cancer among relatives.

Cancer-Risk Assessment and Recommendation Delivery

Of 20 tools (32%) that provide a preliminary cancer-risk assessment, 80% do so automatically, including 15 electronic tools and 1 automated telephone interview[55]. Of the paper-based tools, 4 allow for a preliminary cancer-risk assessment and communication to respondents[66,79,84,85]. Tools providing cancer-risk assessments are mostly cancer-specific (70%). Tailored follow-up recommendations for patients can be delivered by 15 tools (24%), with 9 of them offering advice on risk-reducing strategies (healthy lifestyle, cancer surveillance, preventive interventions). Six tools propose management recommendations to clinicians. When appropriate, 9 tools refer respondents to genetic counselling. Respondents are invited or prepared by 8 tools to talk about their cancer risk with their health care professionals. In 5 tools that include a cancer-risk assessment component[49,66,74,84,85], no recommendations for follow-up are issued.

Respondent Experience and Appreciation of the Tools

User appreciation and experience were reported for 19 tools (31%). Overall, respondents expressed positive attitudes toward the tools, judging them as simple[71,91,96], easy to use[11,30,34,43,49,51,62,71,72,79,81,85,89,91], easy to understand[11,43,47,79,91,96], worthy of recommendation to peers[43,79], worthy of definitive incorporation into EHRs[92], or highly acceptable[75]. Fair or negative appreciations were reported for only 4 tools. Respondents felt "fairly satisfied" about the cancer-risk information provided[70], required assistance for completing the tool[43], and at times, considered them too long[49] or "brittle and clunky"[81]. The time required to complete the questionnaire was reported for 26 tools (42%). The completion time was 30 minutes or less for 20 tools. For 6 tools, completion time varied from 33 minutes to 120 hours.

TABLE II Characteristics of 17 generic family history collection tools

Reference	Country	Tool name (when specified)	Format	Primary care	Main cancer or cancers investigated	Highest degree of kinship covered	Intended users	Pedigree production	Automatic risk stratification output	Recommendations generated
Cole *et al.*, 1978[25]	U.S.A.		Paper	No	Colon, ovary, prostate, breast	3rd	Patients	Yes, subsequently	No	No
Williams *et al.*, 1988[26], and Johnson *et al.*, 2005[27]	U.S.A.	Health Family Tree	Paper	No	Colon, breast	3rd	Students, parents	Yes, subsequently	No	No
Qureshi *et al.*, 2001 and 2005[28,29]	U.K.		Paper	Yes	Colon, ovary, prostate, breast	3rd	Patients	No	No	No
Colombet *et al.*, 2002, 2003, and 2003[30–32]	France	EsPeR	Electronic	Unspecified	Colon, prostate, breast	2nd	HPs	Yes, automatically	Yes	Yes: Cancer screening guidelines
Frezzo *et al.*, 2003[33]	U.S.A.		Paper	Yes	Colon, ovary, prostate, breast	Unspecified	Patients	Unspecified	No	No
Volk *et al.*, 2007[14]	U.S.A.		Paper	Yes	Colon, breast	Unspecified	Patients	No	No	No
Yoon *et al.*, 2009[34], Rubinstein *et al.*, 2011[35], Ruffin *et al.*, 2011[36], and O'Neill *et al.*, 2009[37]	U.S.A.	Family Healthware	Electronic	Yes	Colon, ovary, breast	2nd	Patients	Yes, subsequently	Yes	Yes: Risk-reducing strategies, preparation for risk discussions with HPs, referral to GCT
Cohn *et al.*, 2010[11], and Baumgart *et al.*, 2016[38]	U.S.A.	Health Heritage	Electronic	Yes	Colon, ovary, breast	2nd	Patients	Yes, automatically	Yes	Yes: Risk-reducing strategies, referral to GCT
Facio *et al.*, 2010[39]	U.S.A.	My Family Health Portrait	Electronic	No	Colon, ovary, breast	3rd	Patients	Yes, automatically	No	No
Hulse *et al.*, 2011[40]	U.S.A.	OurFamilyHealth	Electronic	Yes and specialized	Colon, breast	3rd	Patients	Yes, automatically	No	No
Orlando *et al.*, 2011[41], Orlando *et al.*, 2013[42], and Wu *et al.*, 2013 and 2014[43,44]	U.S.A.	MeTree	Electronic	Yes	Hereditary cancer syndromes	3rd	Patients	Yes, automatically	Yes	Yes: Referral to GCT, guidelines for clinicians
Slack *et al.*, 2011 and 2012[45,46]	U.S.A.		Electronic	Yes	All cancers	Unspecified	Patients	No	No	No
Baer *et al.*, 2013[47]	U.S.A.	Your Health Snapshot	Electronic	Yes	Colon, prostate, breast	2nd	Patients	No	Yes	Yes: Risk-reducing strategies
Walter *et al.*, 2013[48]	U.K.		Paper	Yes	Colon, breast	2nd	Patients	No	No	No
Doerr *et al.*, 2014[49]	U.S.A.	MyFamily	Electronic	Yes and specialized	Colon, ovary, breast	Unspecified	Patients	Yes, automatically	Yes	No
Emery *et al.*, 2014[50]	Australia		Paper	Yes	Colon, ovary, prostate, breast, melanoma	2nd	Patients	Yes, subsequently	No	No
Wang *et al.*, 2015[51]	U.S.A.	VICKY	Electronic	Yes	Colon, breast	2nd	Patients	Yes, automatically	No	No

EsPeR = Estimation Personnalisée de Risques [Personalized Risk Estimate]; VICKY = Virtual Counselor for Knowing Your Family History; HP = health care professional; GCT = genetic counselling and testing.

TABLE III Characteristics of 45 cancer-specific family history collection tools

Reference	Country	Tool name (when specified)	Format	Primary care	Main cancer or cancers investigated	Highest degree of kinship covered	Intended users	Pedigree production	Automatic risk stratification output	Recommendations generated
Morrison *et al.*, 1987[52]	U.S.A.	CPQ	Paper	No	All types	1st	Patients	No	No	No
Aitken *et al.*, 1996[53]	Australia		Paper	Unspecified	Melanoma	3rd	Patients	No	No	No
de Bock *et al.*, 1997[54]	Netherlands		Face-to-face interview	Yes	Breast, ovary	2nd	HPs	Yes, subsequently	No	No
Kadison *et al.*, 1998[55]	U.S.A.	Breast Cancer Telephone Risk Assessment System	Telephone	No	Breast	1st	Patients	No	Yes	Yes: Risk-reducing strategies, preparation for cancer-risk discussions with HPs
Mussio *et al.*, 1998[56]	Italy and Switzerland		Interview	No	All types	1st	Patients	No	No	No
House *et al.*, 1999[57], and Rose *et al.*, 2004[58]	U.K.		Paper	Yes	Breast, colon, ovary, prostate, uterus	1st	Patients	No	No	No
Leggatt *et al.*, 1999 and 2000[59,60]	U.K.		Paper	Yes	Breast, CRC	3rd	Patients	No	No	No
Church and McGannon, 2000[61]	U.S.A.		Face-to-face interview	No	Colon	3rd	HPs	Yes, subsequently	No	No
Westman *et al.*, 2000[62], Sweet *et al.*, 2002[63], and Kelly *et al.*, 2008[64]	U.S.A.	Jameslink	Electronic	No	27 Types	3rd	Patients	No	Yes	Yes: Advice about lifestyle and GCT
Hurt *et al.*, 2001[65]	U.S.A.		Paper	No	Breast	2nd	Patient	Yes, subsequently	No	No
Benjamin *et al.*, 2003[16]	U.K.		Paper	No	Breast	2nd	Patients	No	No	No
Fisher *et al.*, 2003[66]	Australia		Paper	Yes	Breast, ovary	3rd	Patients	No	No	No
Hughes *et al.*, 2003[67]	U.S.A.		Paper	Yes	Breast, ovary	2nd	Patients	Yes, subsequently	No	No
Grover *et al.*, 2004[68]	U.S.A.		Paper	No	Breast, ovary, uterus, brain, bladder, kidney, and GI cancers	3rd	Patients	Yes, subsequently	No	No
Wallace *et al.*, 2004[69]	U.K.		Paper	Yes	Breast, ovary, colon	2nd	Patients	No	No	No
Braithwaite *et al.*, 2005[70]	U.S.A.	GRACE	Electronic	No	Breast	3rd	Patients	Yes, automatically	Yes	Yes: Advice about lifestyle, GCT, breast cancer surveillance

TABLE III Continued

Reference	Country	Tool name (when specified)	Format	Primary care	Main cancer or cancers investigated	Highest degree of kinship covered	Intended users	Pedigree production	Automatic risk stratification output	Recommendations generated
Emery, 2005[71], and Emery *et al.*, 2007[72]	U.K.	GRAIDS	Electronic	Yes	Breast, ovary, colon, endometrial	2nd	HPs	Yes, automatically	Yes	Yes
Jones *et al.*, 2005[73]	U.S.A.		Paper	Yes	Breast, ovary, colon	3rd	Patients	No	No	No
Schroy *et al.*, 2005[74]	U.S.A.		Electronic	Yes	Colon	2nd	HPs	No	Yes	No
Acheson *et al.*, 2006[75]	U.S.A.	GREAT	Telephone	No	24 Types	3rd	Patients	Yes, automatically	No	No
Bravi *et al.*, 2007[76]	Italy		Face-to-face or telephone interview	No	Respiratory, GI cancers	1st	Trained interviewer	No	No	No
Kelly *et al.*, 2007[77]	U.S.A.		Paper	No	All types	1st	Patients	No	No	No
Murff *et al.*, 2007[78]	U.S.A.		Paper	Yes	Colon, breast, ovary	2nd	Patients	Yes, subsequently	No	No
Cohn *et al.*, 2008[79]	U.S.A.	*Are you at risk for hereditary breast cancer?*	Paper	Yes	Breast, ovary	3rd	Patients	No	No	Yes: Advice about GCT
Yip *et al.*, 2008[80]	U.S.A.	6Q	Face-to-face interview	No	Multiple endocrine neoplasia	Unspecified	HPs	No	No	No
Zimmerman *et al.*, 2008[81]	U.S.A.	ChMP	Electronic	Unspecified	Breast	2nd	Patients	Yes, automatically	Yes	No
Armel *et al.*, 2009[82]	Canada		Paper	No	Breast, ovary	3rd	Patients	Yes, subsequently	No	No
Ashton-Prolla *et al.*, 2009[83]	Brazil	FHS-7	Paper	Yes	Colon, breast, ovary	3rd	Patients	No	No	No
Bellcross *et al.*, 2009[84]	U.S.A.	RST	Paper	Yes	Breast, ovary	2nd	HPs	No	No	No
Dudley-Brown and Freivogel, 2009[85]	U.S.A.		Paper	No	HNPCC, FAP, MAP	3rd	Patients	No	No	No
Ozanne *et al.*, 2009[86]	U.S.A.	Hughes Risk App	Electronic	Yes	HBOC	Unspecified	Patients or HPs	Yes, automatically	Yes	Yes: Risk-management plan, GCT
Wideroff *et al.*, 2010[87]	U.S.A.	CATI	Computer-assisted telephone interview	No	All types	2nd	Patients or overall population	Yes, subsequently	No	No
Pieper *et al.*, 2012[88]	Germany		Paper	No	GI cancers, endometrial	1st	Patients	No	No	No

TABLE III Continued

Reference	Country	Tool name (when specified)	Format	Primary care	Main cancer or cancers investigated	Highest degree of kinship covered	Intended users	Pedigree production	Automatic risk stratification output	Recommendations generated
Vogel *et al.*, 2012[12]	U.S.A.		Paper	No	Lynch syndrome, breast, ovary	3rd	Patients	Yes, subsequently	No	No
Rupert *et al.*, 2013[89]	U.S.A.	Cancer in the Family	Electronic	Yes	HBOC	2nd	Patients or HPs	Yes, automatically	Yes	Yes: GCT, preparation for cancer-risk discussions with HPs
Koeneman *et al.*, 2014[90]	Netherlands		Paper	No	Lynch syndrome, breast, prostate	Unspecified	HPs	No	No	No
Pritzlaff *et al.*, 2014[91]	U.S.A.	CGC	Electronic	No	Breast, ovary, colon, pancreas, melanoma	Unspecified	Patients or HPs	Yes, automatically	Yes	Yes: Management plan based on NCCN guidelines and literature reviews
Scheuner *et al.*, 2014[92]	U.S.A.	Multicomponent cancer genetics toolkit	Electronic	Yes	HBOC, HNPCC	3rd	HPs	No	No	Yes: Criteria for GCT referral
Son *et al.*, 2014[93]	Korea		Interview	No	All types	3rd	HPs	Yes, subsequently	No	No
Eiriksson *et al.*, 2015[94]	Canada	Brief Family History Questionnaire	Paper	No	Lynch syndrome	Unspecified	Patients	No	No	No
Kallenberg *et al.*, 2015[95]	Netherlands		Electronic	Yes	All types	2nd	Patients	No	No	No
Schiavi *et al.*, 2015[96]	Canada	SCGS	Paper	No	Li-Fraumeni syndrome	3rd	Patients	No	No	No
Schultz *et al.*, 2015[97]	New Zealand		Electronic	No	Colon	2nd	Patients	No	Yes	Yes: Risk-reducing strategies, orientation to PCPs for cancer-risk discussion
Floria-Santos *et al.*, 2016[98]	Brazil		Interview	Yes	All types	3rd	Trained interviewer	No	No	No
Niendorf *et al.*, 2016[99]	U.S.A.	FHQ	Paper, telephone interview	Yes and specialized care setting	All types	3rd	Patients or trained interviewer	Yes, subsequently	No	No

CPQ = Cancer Patient Questionnaire; HP = health care professional; CRC = colorectal cancer; GCT = genetic counselling and testing; GI = gastrointestinal; GRACE = Genetic Risk Assessment in the Clinical Environment; GRAIDS = Genetic Risk Assessment in an Intranet and Decision Support; GREAT = Genetic Risk Easy Assessment Tool; 6Q = 6-Question panel; ChMP = Collaborative Medical History Portal; FHS-7 = simple 7-question instrument about family history of breast, ovarian, and colorectal cancer; RST = Referral Screening Tool; HNPCC = hereditary nonpolyposis colorectal cancer; FAP = familial adenomatous polyposis; MAP = *MYH*-associated polyposis; HBOC = hereditary breast and ovarian cancer; CATI = computer-assisted telephone interview; CGC = CancerGene Connect; NCCN = U.S. National Comprehensive Cancer Network; SCGS = Sarcoma Clinic Genetic Screening; PCP = primary care physician; FHQ = family history questionnaire.

Tool Evaluation and Performance Using the ACCE Framework

Analytic validity was measured for 5 tools and involved various FH parameters (Table IV). Analytic sensitivity varied from 33% to 100%, and specificity varied from 76% to 97%. Clinical validity was calculated for 6 tools (Table IV). Sensitivity for identifying increased risk varied from 0% to 100%; specificity, from 54% to 92%; positive predictive value, from 24% to 80%; and negative predictive value, from 92% to 100%.

A formal evaluation of the clinical utility of the tool was not performed in any publication. However, based on study results, we identified potential benefits for respondents and their relatives. Assessment of FH helped to identify cancer patients for whom a referral to a genetics clinic would be warranted because of the pattern of cancer occurrence in their family[12,68,80,94,96,99]. The tool allowed for increased and improved-quality referrals to genetics clinics[12,69,71,72,90,92]; for FH-based decision-making in primary care[41,43,44,47]; and for efficient[26,28,45,46,49,88,92], efficacious[11,39–44,51,54,62–64,68,73,75,76–78,81,82], and exhaustive[25] collection of FH and updates[41,86,89]. Increased compliance with cancer screening[55,61] and changes in health behaviours[34] were also noted. By collecting the FH before clinical

TABLE IV Analytic and clinical validity among the retrieved family history (FH) collection tools

Reference	Tool name (where specified)	Comparator or validation strategy	Validation outcomes
Analytic validity			
Mussio *et al.,* 1998[56]		Cancer registries	Sensitivity: 83%; specificity: 97% (for information on malignant tumour occurrence in 1st-degree relatives)
Church and McGannon, 2000[61]		Registry staff's detailed family history	Sensitivity: 72%; specificity: 77%; negative predictive value: 87%; positive predictive value: 59% (for the occurrence of colon cancer)
Yip *et al.,* 2008[80]		Medical records (clinical, anatomic, histologic, biochemical, and radiologic information)	Sensitivity: 83%; specificity: 76% (for detection of *MEN1* in patients with apparent sporadic primary hyperparathyroidism)
Facio *et al.,* 2010[39]		Genetic counsellor's supplemented pedigree	Sensitivity and specificity varied from 99.7% to 99.9% and from 80.9% to 90% respectively (for the occurrence of cancers considered in the study)
Wang *et al.,* 2015[51]		Genetic counsellor	Sensitivity: 40% (for colon cancer identified) and 33% (for breast cancer identified)
Clinical validity			
Ashton-Prolla *et al.,* 2009[83]		Genetic counselling risk assessment	Sensitivity: 88%; specificity: 56%; positive predictive value: 24%; negative predictive value: 97% (for identification of women at high risk of breast and colon cancer)
Bellcross *et al.,* 2009[84]	RST	Risk stratification provided by 4 validated risk assessment models: BOADICEA, BRCAPRO, Myriad II, FHAT	Clinical sensitivity, specificity, positive predictive value, and negative predictive value of 81%, 92%, 80%, and 92% respectively (for increased risk identification)
Cohn *et al.,* 2010[11]	Health Heritage	Genetic assessment team	Sensitivity: 0%–100% (for increased risk identified)
Walter *et al.,* 2013[48]		Risk stratification from a standard 3-generation pedigree	Sensitivity: 81%–96%; specificity: 83%–88% (ability of the questionnaire to identify individuals at increased risk for breast and colon cancer)
Emery *et al.,* 2014[50]		Genetic counsellor's risk assessment	Sensitivity: 95%; specificity: 54% (for the identification of individuals potentially at increased risk for conditions searched in the questionnaire)
Eiriksson *et al.,* 2015[94]	Brief Family History Questionnaire	Tumor assessed with immunohistochemistry for mismatch repair proteins and microsatellite instability, and germline testing for Lynch syndrome	Sensitivity: 100%; specificity: 76.5%; positive predictive value: 26%; negative predictive value: 100% (for presence of mutation)

RST = Referral Screening Tool; BOADICEA = Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm; FHAT = Ontario family history assessment tool.

appointments, clinicians had more time to assess and discuss cancer risk, resulting in enhanced-quality counselling and improved individual management[49,91].

The fourth component of the ACCE framework—ethics, legal, and social issues—were not discussed in the retained publications, except for psychological effects. After FH documentation and cancer-risk assessment, psychological evaluations for respondents showed scores for distress and depressive symptoms that were, on average, within normal limits[29,65,70,77].

Tool Performance According to Indicators Other Than the ACCE Framework

The performance of 30 cancer FH collection tools was assessed using indicators different from those proposed in the ACCE framework. Table v summarizes the strategies and comparators used by the research teams to assess the thoroughness of the FH collected and the appropriateness of risk stratifications and referrals to genetics clinics. Validation outcomes were reported as narratives supplemented with quantitative data, simple frequencies or proportions (or both) not related to intrinsic validity, correlation coefficients, concordance scores, and percentages of agreement. Sensitivities, specificities, and odds ratios were calculated to assess the appropriateness of referrals to genetics clinics. Overall, those tools found a good level of concordance for FH collection and risk assessment with their respective comparators. The tools outperformed medical charts in FH collection.

DISCUSSION

In the present review, we identified 17 generic and 45 cancer-specific tools developed for cancer FH collection. Most of the retrieved tools were paper-based and designed to be self-administered by patients and family members. One third of the tools identified were available electronically, and one quarter were able to automatically produce a pedigree, provide cancer-risk assessment, and deliver evidence-based recommendations. The validation process showed that the performance of the tools varied depending on the disease or diseases being investigated, the FH parameters, and the comparators considered. One third of the tools were partly validated against a standard reference.

To our knowledge, our review is the first to focus on FH collection tools developed to report on all types of cancer in the adult population. It is also the first to assess the strategies used to validate tools according to the ACCE framework. It represents an important update concerning the progress made over time in developing FH collection tools. Our findings about the greater number of cancer-specific tools, the preference for paper-based and self-administered instruments, the inconsistent validation, and the lack of functionality, are similar to those in earlier reviews[18–20].

We did not find any tool that met all the characteristics of an ideal tool to support clinicians in decision-making and cancer-risk management effectively, but 6 were considered promising: GRACE[70], MeTree[42], Health Heritage[11], Hughes Risk App[86], Cancer in the Family[89], and CancerGene Connect[91]. All 6 tools are electronic and self-administered; all draw pedigrees and provide cancer-risk assessment and management recommendations. However, updates to the FH are possible in only 3 of them (MeTree, Hughes Risk App, Cancer in the Family)[42,86,89]. Only 2 (Health Heritage, MeTree)[11,42] were evaluated for FH accuracy and completeness, and risk-assessment accuracy or compliance with proposed genetics referral guidelines. Moreover, only 3 were deemed easy to use (Health Heritage, CancerGene Connect, Cancer in the Family)[11,89,91]. More importantly, none are embedded into an EHR system. Hickey *et al.*[100] and Feero *et al.*[101] made a case for integrating, into EHRs, a common FH core dataset that would allow for the standardized collection and exchange of FH throughout health information systems, ensuring a continuum of patient care. Although none of these 6 proposed tools are "ideal" cancer FH questionnaires, they can still help health care providers to identify at-risk individuals and families. They could be used in medical clinics to screen patients requiring a genetic counselling referral or in genetics clinics to document FH and to conduct a preliminary risk assessment before a formal genetic counselling interview.

Nevertheless, non-electronic tools still have their place in cancer FH collection, given that not every clinical setting is equipped with an EHR system and not every health care provider has access to and can make use of the Internet and electronic devices. Validated paper-based questionnaires, automated telephone interviews, and telephone and face-to-face interviews can play a significant role in identifying at-risk individuals if they can guide health care providers in assessing risk and managing decisions, and if the data can be easily retrieved and updated. Otherwise, their contribution will remain partial and will continue to require additional human resources.

Lu *et al.*[17], on behalf of the American Society of Clinical Oncology, advocated for the use of a minimum cancer FH, including 1st- and 2nd-degree relatives in both the maternal and paternal lineages. For nearly 70% of retrieved tools, cancer FH was collected up to 2nd-degree relatives. However, it remains unclear how that minimum FH affects cancer-risk assessment. It would be worthwhile to compare the performance of the tools according to the degree of kinship covered and the perspective taken (pedigree-oriented vs. disease-oriented). Cost-effectiveness analyses should also be undertaken to determine whether an evidenced-based benefit accrues to the use of one type of tool over another.

Automatic production of pedigrees by an electronic or an automated collection tool can be beneficial for health care providers. Three-generation pedigrees allow for an appreciation of family size, a determination of the pattern of medical condition inheritance within the family, and easier identification of at-risk individuals[102]. Of the 23 electronic or automated FH collection tools identified in this review, only 14 were able to generate a pedigree automatically. Thus, improvements are needed in this regard.

Tools for FH collection that estimate individual risk of cancer and propose management strategies would be valuable to health care providers and would facilitate provider–patient risk communication. Only one third of the tools identified here can provide risk assessment, and one quarter can issue management recommendations. Indeed, we identified 5 tools that provide a preliminary

TABLE V Validation of collected family history (FH), risk stratification, and referral decisions

Reference	Tool name (where specified)	Comparator or validation strategy	Validation outcomes
Cole *et al.,* 1978[25]		Final pedigrees obtained from revision for accuracy and completeness of initial pedigrees built from answered questionnaires	■ Half the pedigrees (*n*=60) built from the questionnaire required minor or major changes or additional information
de Bock *et al.,* 1997[54]		Estimation of the degree of certainty about the FH information provided using a 4-point scale (from 1=very sure to 4=very unsure)	■ Degree of certainty varied from 1.1 (mean) for mother and sisters to 2.1 (mean) for grandmothers
Morrison *et al.,* 1997[52]	CPQ	Tumour registry data built from chart review	■ Mean number of affected relatives identified per cancer patient was 1.83 by the CPQ versus 1.38 by the tumour registry ■ Complete agreement with the registry for FH in 60% of cancer patients with cancer FH
House *et al.,* 1999[57]		General practitioner's risk classification of 250 respondents reviewed by a geneticist	■ Among 104 patients assigned to intermediate colon cancer risk by the general practitioner based on answers to the questionnaire, 5 were reassigned to the high-risk group
Sweet *et al.,* 2002[63], and Kelly *et al.,* 2008[64]	Jameslink	Medical charts	■ Among participants who completed Jameslink (*n*=362), only 69% had FH information available in their medical record ■ The tool assigned 101 patients to a high-risk category, with confirmation of their status by evidence in charts for 69 ■ Low chart documentation rate of high-risk status (14%) and low referral rate to genetic counselling (7%)
Fisher *et al.,* 2003[66]		Interview with a genetic counsellor and subsequent risk stratification	■ Agreement between the FH questionnaire and the genetic counsellor for risk stratification was 100% (*n*=89)
Frezzo *et al.,* 2003[33]		Chart review and interview pedigree, with subsequent risk stratification by a genetic counsellor or a medical geneticist	■ Of the 78 participants, 32 were identified at an increased risk by the questionnaire compared with 30 identified by the interview pedigree and 18 identified by their chart ■ Increased risk identified by the study questionnaire or the interview pedigree for 61% compared with 40% identified through charts (chi-square *p*=0.01)
Grover *et al.,* 2004[68]		Medical charts	■ Complete agreement observed between 77% of charts having a comprehensive cancer FH (*n*=184) and FH collected using the questionnaire
Wallace *et al.,* 2004[69]		Telephone or in-person interview with a genetic nurse or a fieldworker to check the consistency of the information collected	■ In a sample of 305 respondents, 7% had their initial risk stratification altered after the interview with the genetic nurse, based on information collected with the questionnaire
Emery, 2005[72]	GRAIDS	Cluster randomized controlled trial comparing practices using GRAIDS and those receiving an education session and guidelines for familial cancer-risk management	■ More referrals consistent with referral guidelines in practices using GRAIDS (OR: 5.2; 95% CI: 1.7 to 15.8; *p*=0.006)
Acheson *et al.,* 2006[75]	GREAT	Genetic counsellor' s pedigree	■ Agreement of 94% for 1st-degree relatives, 67% for 2nd-degree relatives, 38% for 3rd-degree relatives, and 63% for all cancers, with 90% agreement on the type of cancer ■ Good agreement on subsequent risk stratification: *K*=0.7; correlation: 0.77
Bravi *et al.,* 2007[76]		Answers to first interview (cases) versus answers to a second interview (controls) with the same questionnaire	■ Positive agreement for any cancer was 78%, *K*=0.7

TABLE V Continued

Reference	Tool name (where specified)	Comparator or validation strategy	Validation outcomes
Kelly *et al.,* 2007[77]		Comparison between written and interview reports of cancer FH with the same questionnaire	■ Total concordance for the identification of affected relatives ■ Among respondents with cancer FH, 57% agreement for age, and 70% agreement for the type of cancer
Murff *et al.,* 2007[78]		Medical charts	■ In a sample of 310 participants, 128 additional affected relatives identified ■ Age of cancer diagnosis recorded for 81% of affected relatives compared with 40% in the charts ■ More individuals at increased risk identified: 29 versus 19 in the charts
Volk *et al.,* 2007[14]		Electronic health records	■ The FH questionnaire alone identified 85% and 97% of patients with a positive FH of breast and colon cancer respectively ■ New information provided by patients using the FH questionnaire resulted in an increase in the patient's risk level for 50% and 32% of patients with a positive FH of colon and breast cancer respectively
Cohn *et al.,* 2008[79]	*Are you at risk for hereditary breast cancer?*	Content validity (development) and risk assessment by a genetic counsellor	■ Identification of 7 of 10 at-risk women by the genetic counsellor
Armel *et al.,* 2009[82]		Pedigrees created from FH questionnaire updated by a genetic counsellor	■ Of initial pedigrees (*n*=121), 92% were modified during genetic counselling ■ Probability for having a *BRCA1/2* mutation revised in 12%, alteration of eligibility for genetic testing revised in 5%
Bellcross *et al.,* 2009[84]	RST	Genetic counsellor's telephone interview	■ Concordance between initial and corrected FH: 0.89
Cohn *et al.,* 2010[11]	Health Heritage	Genetic assessment team	■ Completeness of the FH collected varied from 54% to 182% depending on the parameter considered
Wideroff *et al.,* 2010[87]	CATI	Original FH reviewed for accuracy in a second interview (consistency with malignancy and specificity for cancer sites)	■ Of 2657 cancer reports, 79% were consistent both for malignancy and site
Hulse *et al.,* 2011[40]	OurFamilyHealth	Electronic health records	■ Structured family history available in medical records for only 14% of patients (*n*=168) who used the tool, with a general discordance on the type of data collected
Orlando *et al.,* 2011[41], Orlando *et al.,* 2013[42], and Wu *et al.,* 2013 and 2014[43,44]	MeTree	Pre-implementation validation: stakeholder cognitive interviewing, genetic counsellor perception; quality assessment of collected FH based on purposed-devised criteria, assessment of genetic referral appropriateness based on guideline recommendations for genetic counselling referral (NCCN, CFHG)	■ Changes to the interface and the clinical decision support documents ■ Of the FHs collected, 99.8% were considered to be of high quality ■ Agreement with guidelines recommendations was 85% to 90% for genetic counselling referrals
Pieper *et al.,* 2012[88]		Telephone interview	■ Minor changes to initial FH
Vogel *et al.,* 2012[12]		Structured genetic interview, electronic medical record	■ Of the 26 respondents identified from the structured genetic interview as meeting criteria for referral to genetic counselling, 81% were identified by the FH questionnaire ■ In 76% of participants, more family members with cancer were identified by FH questionnaire than by the electronic medical record

TABLE V Continued

Reference	Tool name (where specified)	Comparator or validation strategy	Validation outcomes
Doerr *et al.,* 2014[49]	MyFamily	Estimation of clinicians' agreement score with tool-provided risk assessment	■ Agreement score varied from 1 to 2.5 among surveyed clinicians on a Likert scale of 1 (strongly agree) to 5 (strongly disagree)
Son *et al.,* 2014[93]		Pedigree completeness assessment in two telephone interviews after an initial face-to-face survey and an additional survey targeting missing information	■ Completion of the pedigree went from 79% at first interview to 86% at the third ■ Few corrections were needed in subsequent telephone interviews
Scheuner *et al.,* 2014[92]	Multicomponent Cancer Genetics Toolkit	Appraisal of FH documentation and cancer-risk assessment with or without the use by clinicians of a reminder questionnaire for FH collection Genetic counsellor's assessment of familial risk provided by referring clinicians	■ Significant increase in cancer FH documentation when the reminder was used, more significant change in familial risk assessment when reminder was not used by referring clinicians (38.5% vs 18%)
Kallenberg *et al.,* 2015[95]		Phase 1: Genetic referral decisions based on genetic counsellor's pedigree Phase 2: Genetic referral decisions based on telephone interviews data	■ Phase1: 90% sensitivity and 98% specificity in the identification of individuals deserving referrals to genetic specialists ■ Phase 2: 100% sensitivity and 97% specificity
Floria-Santos *et al.,* 2016[98]		Retaking of the FH with the same FH questionnaire, 5 years later, for a subsample of 14 families judged to be at moderate or high risk	■ Of initial pedigrees, 90% were confirmed
Niendorf *et al.,* 2016[99]		Genetic counselling	■ Agreement for increased-risk individuals identified by the screening questionnaire was 87% (*n*=500)

CPQ = Cancer Patient Questionnaire; GRAIDS = Genetic Risk Assessment in an Intranet and Decision Support; OR = odds ratio; CI = confidence interval; GREAT = Genetic Risk Easy Assessment Tool; RST = Referral Screening Tool; CATI = Computer-Assisted Telephone Interview; NCCN = U.S. National Comprehensive Cancer Network; CFHG = Michigan Department of Community Health's Cancer Family History Guide.

risk assessment, but none issue follow-up recommendations[49,66,74,84,85]. The lack of follow-up represents a missed opportunity to empower respondents with choices concerning their health and providers with the ability to manage at-risk individuals.

Health care providers often state that lack of time precludes them from routinely collecting FH. However, the fact that 81% of the identified tools can be self-administered by lay individuals has the potential to help overcome that barrier. The tools allow patients and family members to provide FH information without lengthening in-office consultations. Moreover, answering the questionnaires at home offers patients the opportunity to contact family members for more precise information[102].

Tool completion time was not reported for more than half the tools. Unfortunately, the reasons for that non-reporting were not provided. Given the importance of completion time to the acceptability and usability of the tools, authors should document that aspect more thoroughly. Also, researchers should try to balance FH comprehensiveness and ease of questionnaire completion when developing new tools.

Systematic validation of FH collection tools is needed; 33% of the tools identified in our review did not benefit from validation against any comparator. The ACCE framework, the first publicly available analytic process for the evaluation of the risks and benefits of genetic testing, constitutes an important resource for validating cancer FH tools[103], and both Qureshi *et al.*[22] and Valdez *et al.*[6] advocated for its use. Few of the identified tools were validated in a way that complies with some components of the framework, which, when considered in its entirety, has the potential to allow for a standardized, comprehensive, and in-depth assessment of a tool's performance and effects. Wider use of the ACCE or an equivalent framework[104] should be encouraged when tools are being developed, especially if evidence-based recommendations are to be delivered to lay individuals and health professionals.

Interpretation of the results of the present review should be considered in light of several potential limitations. First, our search was limited to reports published in French and English, which might have resulted in publication bias. However, we did not find additional relevant articles in other languages. Second, only 1 reviewer analyzed the papers and extracted the data. However, the latter limitation was mitigated through data cross-checking and repeated readings of relevant article sections. Third, the literature search might have missed papers of interest given that it ended in September 2016. In that regard, we conducted an overview of the literature spanning

1 October 2016 through 3 January 2018. We found four additional relevant papers[105–108], but none reported any particular innovative tool characteristics. Including those papers in the present review would not have significantly changed the main results. Fourth, we could have used more precise search terms such as "pedigree production," "tablet and smartphone apps," and "laptop," which could have enriched the present work. Finally, ranking the tools by score could have been more insightful for readers. However, that approach would have required a purpose-designed and validated scale. To our knowledge, such a scale does not yet exist—but it is needed.

CONCLUSION AND PERSPECTIVES

Currently, there is no standard cancer FH collection tool. The tools identified here can help health professionals in the systematic collection of FH. They can facilitate the identification of individuals at increased risk of cancer while also saving time for the health care provider. However, most of the identified tools do not produce pedigrees, perform cancer-risk assessment, or deliver management recommendations, and few are integrated into EHRs, which limits the support that they provide to health care providers. Those areas are the ones that require improvement. Notably, information technology developments are needed to integrate electronic cancer FH collection tools into EHRs to promote secure sharing of health information.

Developing and making available multifaceted cancer FH collection tools is important. However, increasing the capability and willingness of health care providers to use the outcomes of FH assessment for preventive and, sometimes, therapeutic purposes is a challenge[14]. Research and new strategies are necessary to address that challenge. In the meantime, continuous effort should be made to upgrade the functionality of existing tools that will improve the ability of health professionals to identify and manage high-risk individuals. As has already occurred for hereditary cancer identification and genetic counselling referral guidelines in a pediatric population[109], smartphone and tablet applications are other avenues that can be explored to document FH in adults. Because of their widespread use and popularity, such devices have the potential to streamline FH information-sharing between patients, family members, and health care providers. Several Web-based initiatives for collecting cancer FH have also been reported[22,110]. However, those initiatives still have to be evaluated in scientific studies to gain acceptance.

The emerging role of the genetic counselling assistant in the field of genetic counselling[111] holds much promise and could potentially increase the efficiency of certified genetic counsellors and expand their patient volume. The effect on genetic counselling accessibility and uptake attributable to the use by genetic assistants of the tools recommended in the present review represents an interesting perspective for further research.

More studies of cancer FH collection tools—their validation, utility, social impacts, implementation, utilization, and user experience—are needed. Comparative studies evaluating the efficacy of generic and cancer-specific tools in collecting cancer FH are also needed. Cohort studies with populations of individuals at increased risk might also offer the possibility to assess, in real-world conditions, the clinical validity of cancer FH collection tools.

ACKNOWLEDGMENTS

JEC is a recipient of scholarships from the Fonds d'enseignement et de recherche (Faculty of Pharmacy, Université Laval) and the Centre de recherche sur le cancer–Université Laval. We thank Sue-Ling Chang for linguistic editing of the manuscript.

CONFLICT OF INTEREST DISCLOSURES

We have read and understood *Current Oncology*'s policy on disclosing conflicts of interest, and we declare that we have none.

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