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ORIGINAL RESEARCH

Review and Comparison of Electronic Patient-Facing Family Health History Tools

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Abstract

Family health history (FHx) is one of the most important pieces of information available to help genetic counselors and other clinicians identify risk and prevent disease. Unfortunately, the collection of FHx from patients is often too time consuming to be done during a clinical visit. Fortunately, there are many electronic FHx tools designed to help patients gather and organize their own FHx information prior to a clinic visit. We conducted a review and analysis of electronic FHx tools to better understand what tools are available, to compare and contrast to each other, to highlight features of various tools, and to provide a foundation for future evaluation and comparisons across FHx tools. Through our analysis, we included and abstracted 17 patient-facing electronic FHx tools and explored these tools around four axes: organization information, family history collection and display, clinical data collected, and clinical workflow integration. We found a large number of differences among FHx tools, with no two the same. This paper provides a useful review for health care providers, researchers, and patient advocates interested in understanding the differences among the available patient-facing electronic FHx tools.

Keywords Family health history · Health IT · Patient engagement · Family history tools · Pedigree

Introduction

Family health history (FHx) is an important resource available to help clinicians diagnose and prevent disease (Doerr and Teng 2012; Guttmacher et al. 2004). Knowing a patient's FHx allows clinicians to identify disease risks and to initiate risk-reducing strategies such as early and frequent screening, prophylactic surgery, risk-reducing therapeutics, and lifestyle changes at an earlier or more treatable stage. Additionally, FHx is a powerful, inexpensive way to assess initial genetic risk for disease and reduce genomic variant uncertainty. Finally, FHx has been shown to generate greater attention to one's disease risk than genetic test results alone (Do et al. 2012; Tarini et al. 2008).

Despite its value, FHx is often underutilized in the clinical setting (Acheson et al. 2000). Primary care providers typically discuss FHx with half of their new patients and less than a quarter of their returning patients (Acheson et al. 2000; Medalie et al. 1998). In one study, fewer than 4% of patient charts included a FHx that was informative enough to accurately assess risk for common complex diseases (Powell et al. 2013). One of the primary barriers to clinicians better utilizing

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family history in clinical practice is time (or lack of reimbursement for time taken to collect family history) (Doerr and Teng 2012). It often takes 20–30 min to create a complete FHx but clinicians typically only have a few minutes to both collect and discuss resulting clinical recommendations with their patients (Waters et al. 1994; Acheson et al. 2000). There is simply not enough time during a busy clinic schedule to collect and analyze a three-generation family pedigree and appropriately refer the patient for genetic testing or counseling (Tyler and Snyder 2006). Even if clinicians had sufficient time to record a detailed FHx, a patient's self-reported FHx is often considered suboptimal (Ozanne et al. 2012). While the specificity of self-reported FHx is generally high (> 90%), the sensitivity ranges widely from 30 to 90% depending upon the degree of family relatedness and the reported disease (Murff 2009; Qureshi et al. 2009; Tehranifar et al. 2015). The majority of patients with increased disease risk remain unrecognized or potentially mismanaged (L. A. Baumgart et al. 2013; Sweet et al. 2002; Wood et al. 2014). Finally, it is often challenging for clinicians to keep up with increasing breadth and complexity of FHx guidelines and risks (Bellcross et al. 2011; Rolnick et al. 2011; Shields et al. 2008).

To overcome these challenges, several groups have developed paper-based and/or electronic FHx tools and/or questionnaires to help patients gather and organize FHx information outside the clinic (Arar et al. 2011; Baumgart et al. 2015; Cohn et al. 2010; Giovanni and Murray 2010; Hulse et al. 2011; Orlando et al. 2013; Rubinstein et al. 2011; Welch et al. 2015b). The benefits of this approach for clinical care are twofold. First, patients have better access to FHx information outside of a clinic visit, with time to contact relatives and consult family records at their leisure, allowing them to collect potentially more complete and accurate FHx. Second, the clinical encounter begins with a completed FHx, allowing the clinician to spend precious visit time utilizing family history to guide care rather than collecting it.

While paper-based FHx tools have been around for many years, electronic FHx tools provide added value in the new era of health IT (Hunt et al. 2003). First, collected FHx data can be interoperable with other health IT systems that manage patient information, such as electronic health records (EHRs). Second, it is possible to effectively provide patients and clinicians with clinical decision support (CDS) with risk assessment and recommendations based upon the data (Owens et al. 2011). Third, it allows data to be aggregated and used for clinical research. The U.S. Surgeon General set the stage in 2004 by releasing *My Family Health Portrait*, the first publicly available electronic FHx tool. Since then, several academic and commercial organizations have developed, released, evaluated, and/or commercialized electronic FHx tools for patients. With so many electronic FHx tools now available, a review and analysis would be helpful to prospective users. Therefore, the aims of this study are to (1) understand what FHx tools are available, (2) compare and contrast them to each other, and (3) highlight features of various programs. This review will help clinicians, researchers, patient navigators, cancer support groups, and patients decide which FHx tool best meets their needs and workflow. Furthermore, this review will provide a foundation for future evaluation and comparisons across FHx tools.

Methods

To create a set of FHx tools to review, we identified potential FHx tools from National Institutes of Health (NIH)-sponsored conference proceedings, published articles, online information resources, and solicited recommendations from FHx experts from May through June 2017 (Catalogue of Global Activities 2018a; The NIH Family Health History Tool Conference 2016). For this analysis we included electronic tools—either web-based or software—used by patients to record their FHx information in English. For this study, a "patient" includes an individual acting in conjunction with a health care provider or independently to collect his/her FHx. We excluded EHRs and/or personal health records that have the capacity to collect FHx from patients (e.g., Epic MyChart questionnaire or Microsoft HealthVault), because such a service is not the primary purpose of the software. We excluded pedigree drawing tools intended to be used by only clinicians. If a pedigree drawing tool included a patient-facing questionnaire or component, we included and analyzed the component used by patients.

To extract information for our analysis, where available and as necessary, we used the FHx tool; watched video demonstrations; interviewed tool representatives; reviewed websites, news articles, and presentation slides; and reviewed published literature and reports. Extracted information was entered into an abstraction form for each FHx tool, and abstracted information was reviewed and confirmed by at least two study investigators. In addition, representatives for each FHx tool were given the opportunity to review and correct information about their FHx described in this manuscript.

For each FHx tool that met the inclusion criteria, we abstracted general data on its history, funding and financial support, developing organizations, contacts, and research publications and presentations. We primarily focused on features that affect how patients use the tool, the value they receive, and other aspects that could affect the usability of the FHx tool. We explored FHx tool features around three axes: *family history collection and display* (e.g., pedigree, FHx data entry approach, and collaboration), *clinical data collected* (e.g., type and number of diseases, approach to entering disease info, and genetic information), and *clinical workflow integration* (e.g., access by patients, clinician interface within tool, and Health Level 7 (HL7) interoperability).

Findings from this comparison are summarized below in tables and narrative discussion. In addition, notable themes and trends are identified and discussed. A quantitative analysis of FHx tools to identify features predictive of FHx tool success in terms of accuracy and completeness of data and utilization was considered; however, due to the limited sample size, variability of FHx features, and a lack of outcome data for the tools, such a quantitative analysis of potential success factors was not possible in this analysis.

Results

Of the approximately 30 FHx tools we considered, we included and abstracted the following 17 FHx tools summarized below (in alphabetical order). Notable points for each tool are summarized below. For screenshots of each FHx tool, see Appendix A.

AncestryHealth AncestryHealth (AH) is a patient-facing, web-based FHx tool developed by Ancestry.com, Inc. The beta version of their tool was launched in 2015; however, it is now closed to new users. The tool includes a "Family History Effect" feature that provides the user's relative risk ratios for certain disease based on the entered family history. It also includes information about the disease and known risk factors as well as recommendations for how to decrease risk. There is no data linkage between with the Ancestry.com genealogy database and the AncestryDNA genetic testing service (website URL: https://health.ancestry.com/).

CancerGene Connect CancerGene Connect (CGC) is a web-based platform that helps clinicians collect, analyze, and manage FHx information. The tool includes a questionnaire for the patient to complete prior to a clinical visit. Once the patient submits the FHx information, the platform helps clinicians run risk assessments, generate reports, track genetic test results, and support patient follow-up. Initially developed and validated at the University of Texas Southwestern from 2009 to 2015, CancerGene Connect was licensed to OMMDOMM Inc., which was recently acquired by Invitae Corporation (Pritzlaff et al. 2014) (website URL: http://cagene.com/).

Cancer IQ Cancer IQ (CIQ) is clinician-facing, web-based platform not only to help clinicians collect and organize FHx but also to manage clinical workflows related to risk assessment and genetic test ordering. Prior to a clinical visit, the patient completes the "CancerIQ Self-Assessment," which uses a series of questions and answers to collect a patient's FHx, medical history, screening, and lifestyle information. Since 2013, the platform has been developed and funded through accelerators and investment (website URL: http://www.canceriq.com/).

CRA Health CRA Health (CRA), formerly called HughesRiskApps, is a software-based FHx platform developed at Massachusetts General Hospital (MGH) in 2006 to provide FHx risk assessments to health care providers for cancer patients. Prior to a clinical visit, patients complete a FHx questionnaire on a tablet in the waiting room or at home (Ozanne et al. n.d.). A prenatal version also has been developed but is not yet commercially available (Edelman et al. 2014; Lin et al. 2013). Of note, the founders have done extensive work to establish FHx standards for interoperability and decision support (Chipman et al. 2013; Health Level 7 Clinical Genomics Work Group 2013). In 2015, HughesRiskApps secured investment and changed its name to CRA Health (website URL: https://www.crahealth.com/).

Family Healthware Family Healthware (FH) is a patient-facing, web-based FHx tool developed and validated by the Centers for Disease Control and Prevention (CDC) to help patients assess their familial risk for common chronic diseases (O'Neill et al. 2009; Rubinstein et al. 2011; Yoon et al. 2009). The tool provides a personalized risk assessment, health score, screening recommendations, and prevention plan to patients based upon the entered personal and FHx. Sanitas, Inc., licensed the tool in 2013 and began marketing the tool directly to patients. It is the only FHx tool that has received Food and Drug Administration (FDA) 510(k) clearance (website URL: https://www.familyhealthware.com).

Health Heritage Health Heritage (HH) is a patient-facing, web-based FHx tool developed at the University of Virginia and the Northshore University Health System. HH not only collects FHx of cancer, cardiovascular, neurological, and endocrine conditions but also creates personalized risk reports for patients (Cohn et al. 2010). Accessible to Northshore University Health System patients only through the patient portal, HH is able to pull medical diagnosis from Northshore's EHR and send risk assessment reports back to the EHR (Baumgart et al. 2015). HH was acquired by NantHealth in 2015 (website URL alias: https://tinyurl.com/HealthHeritage).

Inherited Health Inherited Health (IH) is a patient-facing, web-based FHx tool originally developed in 2008 by AccessDNA, Inc., to provide genetic health risks and personalized genetic test recommendations to users. InformedDNA, Inc., acquired AccessDNA, Inc., in 2014 and re-launched InheritedHealth to provide users with a personalized health guide with action steps and recommendations for genetic counseling. InformedDNA provides genetic counseling by telephone (website URL: https://inheritedhealth.com/).

Invitae FHx Tool Invitae Corporation, a company that provides genetic testing, developed and released a clinician-facing,

web-based tool for health care providers to identify their patients' genetic risks and to order the most appropriate genetic test(s). This tool includes a FHx questionnaire for patients to complete prior to their FHx clinical visit and integrates with Invitae FHx Tool (IFT)'s existing online ordering system, allowing FHx to be included in the test ordering and interpretation process (website URL: https://familyhistory.invitae.com).

ItRunsInMyFamily ItRunsInMyFamily (IRMF) is patient-facing, web-based FHx tool developed at the University of Utah and the Medical University of South Carolina (Welch et al. 2015b, c). IRMF uses artificial intelligence (chatbots) and social networking to improve both the completeness and accuracy of FHx information as well as to facilitate communication of health information among family members (Welch et al. 2015a). IRMF is currently NIH research funded (website URL: https://itrunsinmyfamily.com).

MeTree MeTree (MT) is a patient-facing, web-based FHx tool developed at Duke University to collect FHx from the primary care population (Orlando et al. 2016, n.d.). The tool provides risk assessment and decision support and supports integration with EHRs (Epic). Currently, MT is deployed at primary care clinical sites that are part of the IGNITE trial to assess impact of routine FHx collection on risk assessment and referral rates (Wu et al. 2015). MT is currently NIH research funded (website URL alias: https://tinyurl.com/Duke-MeTree).

My Family Health Portrait My Family Health Portrait (MFHP), created by the Surgeon General, is a patient-facing, web-based FHx tool supported, maintained, and promoted by the US government (Berger et al. 2013; Facio et al. 2010; Kanetzke et al. 2011; Owens et al. 2011). The first version was released in 2004, and enhancements have been made since then (Feero et al. 2015). MFHP is an open-source project distributed under the BSD-3 Clause License (*FHH* n.d.) (website URL: https://familyhistory.hhs.gov).

MyFamilyHealth MyFamilyHealth (MFH) is a patient-facing, web-based FHx tool developed in 2008 by Genseq Ltd., based in Malaysia. The current and future status of the project is unclear because several features, such as the ability to invite family members or to receive a personalized report, no longer function properly. Attempts to contact company representative have been unsuccessful (website URL: https://myfamilyhealth.com).

MyLegacy MyLegacy (ML), originally called MyFamily, is a patient-facing, web-based FHx tool that was created at Cleveland Clinic Genomic Medicine Institute in 2011 (Doerr et al. 2014). The tool collects FHx for 12 diseases and provides risk assessment and recommendations for these conditions. In 2016, the product was licensed to Family Care Paths, Inc., which also offers genetic counseling to high-risk patients via telemedicine (website URL: http://www.familycarepath.com).

Myriad FHx Tool Myriad FHx Tool (MFT) is a patient-facing, web-based FHx tool developed in 2015 by Myriad Genetics, a genetic testing company. When patients enter their FHx prior to a clinical visit, the tool generates a hereditary cancer risk assessment and recommendations report that can be printed and shared with their health care provider. Clinicians can request a free customized version of the FHx tool to use with their patients (website URL: https://fht.myriad.com).

OurFamilyHealth OurFamilyHealth (OFH) is patient-facing, web-based FHx tool developed by Intermountain Healthcare (IHC) in 2011 (Hulse et al. 2008; Hulse et al. 2010; Ranade-Kharkar et al. 2013; Taylor et al. 2008). The FHx tool was available for IHC patients to access through their patient portal (Hulse et al. 2011). A summary report of entered FHx information can be printed by patient and shared with his/her clinician. Due to IHC's recent EHR transition, it is not currently available (website URL: https://tinyurl.com/OurFamilyHealth).

Family History Questionnaire Progeny is a pedigree drawing, risk assessment, and management software platform for health care providers. In 2011, Progeny added a web-based Family History Questionnaire for patients to complete their FHx prior to clinical visit. The Family History Questionnaire (FHQ) can be customized to meet specific workflow needs. In 2015, Progeny was acquired by Ambry Genetics, a genetic testing laboratory (website URL: http://www.progenygenetics.com/clinical/fhq).

Vicky Vicky (VKY), a patient-facing, web-based FHx tool developed at Boston University in 2014, uses a virtual agent interface to collect FHx from patient (Wang et al. 2015). VICKY is currently under evaluation in a randomized clinical trial among underserved populations and available only to these participants. VICKY is supported by funding from the NIH (website URL: https://ragserver.ccs.neu.edu/fhWeb/).

Comparison of FHx Tools' Developing Organizations

We found that two FHx tools were created by the government (FH and MFHP), eight were started in academic settings (CGC, CRA, HH, IRMF, ML, MT, OFH, and VKY), and seven started in commercial settings (AH, CIQ, IH, IFT, MFH, MFT, and FHQ). Four of the academically developed (CGC, CRA, HH, and ML) and one (FH) of the government-developed FHx tools have now been fully commercialized.

Eight FHx tools were developed, acquired, managed, or affiliated with a genetics company, six are genetic testing companies (AH, CGC, HH, IFT, MFT, and FHQ), and two are genetic counseling companies (IH, ML; see Table 1).

When plotting the FHx tools on a timeline, notable patterns emerge. The first FHx tools (FH and MFHP) were supported by the government in the mid-2000s. The years around 2010 saw the development of several FHx tools from academic settings (CRA, CGC, MT, OFH, and HH). The peak of commercialization occurred from 2014 to 2015 with five new commercial FHx tools (CIQ, IFT, IH, AH, and MFT) and three acquisitions (CRA, FHQ, and HH; see Fig. 1).

Family History Collection and Display

FHx tools collect and display patient-entered FHx information in a variety of ways (see Table 2). Ten FHx tools (CGC, CIQ, CRA, FH, HH, IH, IFT, ML, MFT, and FHQ) collect FHx information through a series of screens with questions and input fields. Two (MFHP and MT) use a table of listed relatives as a central point for FHx data entry, and two (IRMF and VKY) use a conversational entity to collect FHx from users. Five tools (AH, IRMF, MFH, OFH, and VKY) use the pedigree as a user interface for FHx data entry. Nine FHx tools (CGC, FH, HH, IFT, MFHP, MT, ML, MFT, FHQ, and VKY) generate a pedigree for the user to view after entering their FHx information, two (CIQ and CRA) display a pedigree to the clinician only, and one (IH) does not display a pedigree.

Six FHx tools support some aspect of family collaboration. Two (IRMF and OFH) support multi-directional sharing that allow multiple family members to share and edit the same FHx together (OFH not currently functional). Two (FH and HH) support bi-directional sharing, which entails only sending and receiving a user's own health history with another user. One (FHQ) supports unidirectional data entry, allowing a relative to contribute to the user's FHx. One (MFHP) supports unidirectional sharing of a FHx file, which can repositioned around the receiving relative.

Table 1 Developing organization information

FHx tools (listed alphabetically)	Year first available	Founding organization	Organization type	Affiliation with genetic services company
AncestryHealth (AH)	2015	Ancestry, Inc.	Commercial	Shares parent company with AncestryDNA, which provides genetic (ancestry) testing
CancerGene Connect (CGC)	2009	UT Southwestern	Academic*	Acquired by Invitae, which provides genetic testing
CancerIQ Self-Assessment (CIQ)	2014	CancerIQ, Inc.	Commercial	None
CRA Health (CRA)	2007	Mass General Hospital	Academic*	None
Family Healthware (FH)	2004	CDC	Government*	None
Health Heritage (HH)	2012	University of Virginia; Northshore	Academic*	Acquired by Nanthealth, which provides genetic testing
Inherited Health (IH)	2014	InformedDNA, Inc.	Commercial	Developed by InformedDNA, which provides genetic counseling
Invitae FHx Tool (IFT)	2014	Invitae Corp.	Commercial	Developed by Invitae, which provides genetic testing
ItRunsInMyFamily (IRMF)	2017	University of Utah and MUSC	Academic	None
MeTree (MT)	2009	Duke University	Academic	None
My Family Health Portrait (MFHP)	2004	Surgeon General	Government	None
MyFamilyHealth (MFH)	2008	Genseq Ltd.	Commercial	None
MyLegacy (ML)	2011	Cleveland Clinic	Academic	Licensed by FamilyCarePaths, which provides genetic counseling
Myriad FHx Tool (MFT)	2015	Myriad Genetics	Commercial	Developed by Myriad Genetics, which provides genetic testing
Our Family Health (OFH)	2011	Intermountain Healthcare	Academic	None
Progeny FHQ (FHQ)	2011	Progeny Genetics, Inc.	Commercial	Acquired by Ambry Genetics, which provides genetic testing
VICKY (VKY)	2014	Boston University and Northwestern University	Academic	None

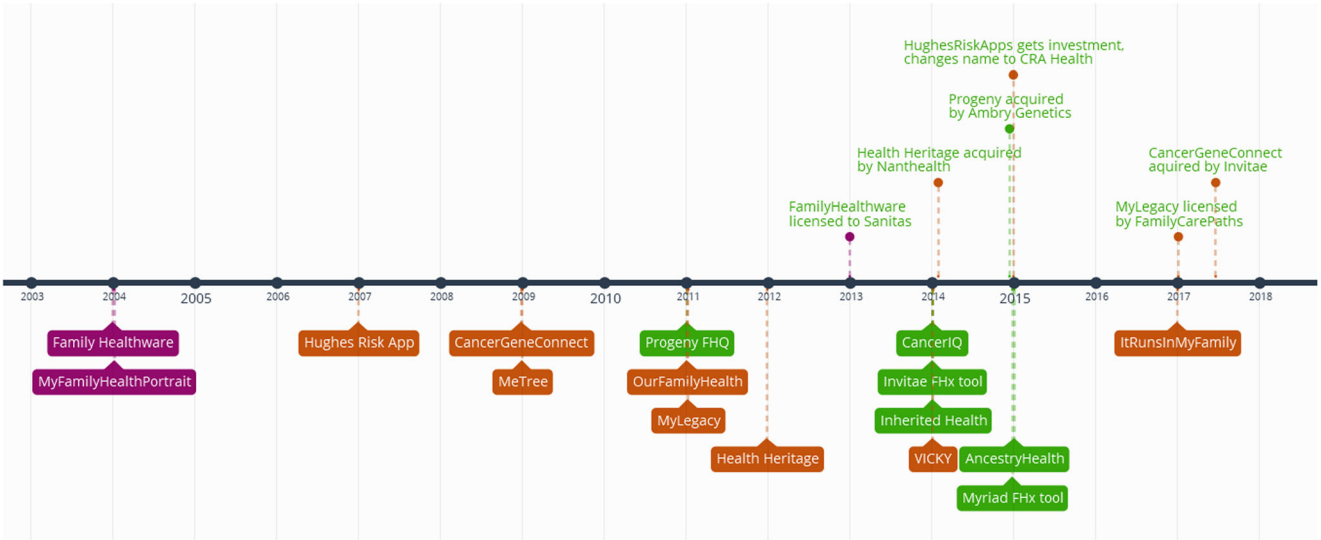

Fig. 1 Timeline of FHx tool development and acquisitions

Clinical Data Collected

With regards to diseases collected, cancer is collected (by default) by every FHx tool in our inventory. Seven (AH, HH, IH, MT, MFHP, OFH, and FHQ) collect diagnosis from several disease categories (e.g., cardiovascular and neurological), ranging from 47 to 533 diseases. Six FHx tools collect only cancer (CGC, CIQ, CRA, IFT, IRMF, and MFT) ranging from 18 to 112 disease. Four (FH, MFH, ML, and VKY) collect major diseases only (e.g., type-2 diabetes and stroke), ranging from 6 to 20 diseases.

There are four different approaches to adding disease information about relatives in the FHx. The most common involves adding relatives first and then adding a disease to the relative—a process used by 11 FHx tools (AH, CGC, CIQ, FH, IRMF, MT, MFHP, MFH, MFT, OFH, and FHQ). Slight variation to this approach is used by two FHx tools (ML and VKY) by adding relatives and then choosing which relative had a particular disease. Two FHx tools (CRA and IFT) first collect relatives who have a disease and then add other relatives later. The last approach, used by two FHx tools (HH and IH), involves selecting a disease in the family and then adding

Table 2 Family history features

FHx tools (listed alphabetically)	FHx data entry	Pedigree display	Collaboration with family
AncestryHealth (AH)	Pedigree to new screen	Primary	None
CancerGene Connect (CGC)	Series of screens	Displayed at end	None
CancerIQ Self-Assessment (CIQ)	Series of screens	To clinician only	None
CRA Health (CRA)	Series of screens	To clinician only	None
Family Healthware (FH)	Series of screens	Displayed at end	Bi-directional
Health Heritage (HH)	Series of screens	Displayed at end	Bi-directional
Inherited Health (IH)	Series of screens	None	None
Invitae FHx Tool (IFT)	Series of screens	Displayed at end	None
ItRunsInMyFamily (IRMF)	Chatbot and pedigree with side panel	Primary	Multidirectional sharing
MeTree (MT)	Family table to new screen	Displayed at end	None
My Family Health Portrait (MFHP)	Family table with pop up screens	Displayed at end	Unidirectional as data file to relative
MyFamilyHealth (MFH)	Pedigree with pop up	Primary	Multidirectional sharing (not functional)
MyLegacy (ML)	Series of screens	Displayed in report	None
Myriad FHx Tool (MFT)	Series of screens	Displayed at end	None
Our Family Health (OFH)	Pedigree with pop up	Primary	None
Progeny FHQ (FHQ)	Series of screens	Displayed at end	Unidirectional from relative
VICKY (VKY)	Virtual agent with side panel; interactive pedigree	Displayed at various steps	None

a relative to the disease. Eight FHx tools (CIQ, CRA, HH, IH, IFT, IRMF, MFT, and FHQ) collect some form of genetic test information, ranging from discrete individual test results to whether or not genetic testing was performed (see Table 3).

Comparison of Clinical Workflow Integration

The availability of FHx tools to patient users varied. Six FHx tools are available for patients to access directly, four (IRMF, MFHP, MFH, and MFT) are free, and two (FH and IH) are available for a fee. Seven FHx tools (CGC, CIQ, CRA, HH, IFT, ML, and FHQ) are accessible to patients through their health care provider typically through e-mail invitation, patient portal, and/or waiting room tablet. Two are only accessible to participants in research studies (MT and VKY), and two are no longer accessible (AH and OFH).

Eight FHx tools (AH, FH, HH, IH, IRMF, MFHP, MFH, and MFT) provide some form of decision support (e.g., risk assessment and/or recommendations) to the patient. In most FHx tools, the patient is encouraged to share their FHx information with a care provider, or the sharing process is greatly simplified for the patient (e.g., single button to e-mail report to care provider). Five FHx tools (CGC, CIQ, CRA, IFT, and FHQ*) provide decision support to clinicians only. Two (MT and ML) provide decision support to both patient and clinician. Two (OFH and VKY) do not provide decision support. Seven FHx tools (CGC, CIQ, CRA, IFT, MT, ML, and FHQ) support the ability for a clinician to login to access their patients' FHx results through the software. Eleven FHx tools (CGC, CIQ, CRA, HH, IFT, IRMF, MT, MFHP, ML, FHQ, and VKY) support some form of HL7 interoperability, either to import and/or to export using HL7 pedigree model and/or HL7 FHIR (see Table 4).

Table 3 Clinical data collected

FHx tools (listed alphabetically)	Disease categories	Number of diseases	Approach to add diseases to relatives	Genetic test information
AncestryHealth (AH)	Several categories	450	Add relatives, then add disease to relative	No
CancerGene Connect (CGC)	Cancer	112	Add relatives, then add disease to relative	No
CancerIQ Self-Assessment (CIQ)	Cancer	30	Add relatives, then add disease to relative	Yes. Checkbox if tested positive for BRCA1, BRCA2, Lynch, or other genes
CRA Health (CRA)	Cancer	18	Add relatives who have disease	Yes. Checkbox if tested positive for a genetic test (non-specific)
Family Healthware (FH)	Major diseases	6	Add relatives, then add disease to relative	No
Health Heritage (HH)	Several categories	47	Select disease in family, add relative to disease	Yes. Choose from 20 genes, specify result (pos, neg, VUS)
Inherited Health (IH)	Several categories	282	Select disease in family, add relative to disease	Yes. Checkbox for whether a relative had a genetic mutation that increases risk
Invitae FHx Tool (IFT)	Cancer	24	Add relatives who have disease, add other relatives later	Yes. Checkbox for whether a genetic test was performed for six genes
ItRunsInMyFamily (IRMF)	Cancer	97	Add relatives, then add disease to relative	Yes. Choose from 141 genes and specify result (pos, neg, VUS)
MeTree (MT)	Several categories	123	Add relatives, then add disease to relative	No
My Family Health Portrait (MFHP)	Several categories	87	Add relatives, then add disease to relative	No
MyFamilyHealth (MFH)	Major diseases	15	Add relatives, then add disease to relative	No
My Legacy (ML)	Major diseases	12	Add relatives, choose which relatives have disease	No
Myriad FHx Tool (MFT)	Cancer	26	Add relatives, then add disease to relative	Yes. Checkbox if tested positive for 23 genes
Our Family Health (OFH)	Several categories	533	Add relatives, then add disease to relative	No
Progeny FHQ (FHQ)	Several categories	387	Add relatives, then add disease to relative	Yes. Dropdown of test result and free text entry of gene
VICKY (VKY)	Major diseases	20	Add relatives, choose which relatives have disease	No

Table 4 Clinical workflow features

FHx tools (listed alphabetically)	Availability to patients	Decision support provided to	Clinician interface within tool	Supports HL7 interoperability
AncestryHealth (AH)	Closed to new users	Patient	No	No
CancerGene Connect (CGC)	Through a health care provider	Clinician only	Yes	Yes
CancerIQ Self-Assessment (CIQ)	Through a health care provider	Clinicians only	Yes	Yes
CRA Health (CRA)	Through a health care provider	Clinicians only	Yes	Yes
Family Healthware (FH)	Available to public for $9.99/month	Patient	No	No
Health Heritage (HH)	Through a health care provider	Patient	No	Yes
Inherited Health (IH)	Available to public for $39.95/year	Patient	No	No
Invitae FHx Tool (IFT)	Through a health care provider	Clinicians only	Yes	Yes
ItRunsInMyFamily (IRMF)	Available to public for free	Patient	No	Yes
MeTree (MT)	Research study access only	Patient and clinician	Yes	Yes
My Family Health Portrait (MFHP)	Available to public for free	Patient	No	Yes
MyFamilyHealth (MFH)	Available for free. Not fully functional	Patient (not functional)	No	No
MyLegacy (ML)	Through a health care provider	Patient and clinician	Yes	Yes
Myriad FHx Tool (MFT)	Available to public for free	Patient	No	No
Our Family Health (OFH)	Not currently functional	None	No	No
Progeny FHQ (FHQ)	Through a health care provider	Clinicians only[a]	Yes	Yes
VICKY (VKY)	Research study access only	None	No	Yes

[a] Patient possible if workflow customized

Discussion

We reviewed and summarized the features of 17 patient-facing FHx tools and found a large number of differences among FHx tools, with no two the same. This variation is likely due to their founding and developing organization types, intended users, and market strategies. Since electronic FHx tools are fairly new, best practices and proven approaches for collecting FHx from patients are still in the early phases of being understood. For example, we identified three different approaches to enter relatives and four different approaches to add disease information to relatives. To date, very few FHx tool usability evaluations have been conducted, a gap that warrants further exploration. Addressing this problem is particularly important as FHx tools incorporate new approaches to FHx collection, such as the use of virtual agents and social networking. It is important to understand which approaches, if any, are the most effective and efficient to collect FHx from users. Just because FHx tools are available, does not mean clinicians and patients will use them. This was confirmed in a 2015 study that showed that only 3% of US population had used a web-based FHx tool, despite 37% having collected their FHx (Welch et al. 2015b).

We noted many dissimilarities in the quantity and types of diseases collected by the FHx tools. These large discrepancies are likely due to the lack of a common disease terminology value set for FHx tools, which hampers the ability for FHx tools to be interoperable with each other or with EHRs. By comparison, the type of core FHx information collected is fairly consistent across FHx tools primarily because it has been standardized through the American Health Information Community and HL7 (Feero, Bigley, Brinner, and Family Health History Multi-Stakeholder Workgroup of the American Health Information Community 2008; Health Level 7 Clinical Genomics Work Group 2013). Without a standardized terminology value set for FHx tools, it is left up to the FHx tool creators' clinical focus, market opportunities, client's requests, and personal biases to define. Given the high number of FHx tools now available, it may be prudent for the government or another organizational body to convene a working group to define a core value set for diseases that a standard-based FHx tool collect. Despite the fact that 11 FHx tools support HL7 standards, integration with EHRs is still a challenge. Though, a few FHx tools have started successfully integrating with EHRs using the Fast Healthcare Interoperability Resources (FHIR) interface. Further exploration is needed regarding the native EHR FHx capabilities and the value added for incorporating third-party FHx tools.

We had planned to provide more detail about the CDS of each FHx tool in this paper. However, we discovered many diverse approaches of algorithms, guidelines, and risk models used that made such information impossible to compute. For

example, many FHx tools implement publicly available guidelines and validated risk models (e.g., USPSTF, NNC, and BRCAPro), but some developed their own proprietary CDS. These differences touch on the ongoing issue of FDA regulation and oversight of how CDS is developed, validated, and maintained. Clinical and analytical validity is an important step for ensuring that risk models and decision support are accurate and appropriate. It would be an important exercise to tease apart the differences in CDS among the tools; however, since this manuscript was focused on the features of the FHx tools themselves, rather that the risk models used, we felt that such an analysis was more appropriate for a future manuscript.

Early on, the government took a clear leadership role to encourage the use of electronic FHx tools, not only to develop and validate their own FHx tools but also to fund academic efforts to do the same. These efforts contributed evidence supporting importance of FHx tools, likely spurring a wave of commercial activities and investment around FHx tools in recent years. Moreover, in 2012, the Center for Medicare & Medicaid Services issued a measure for EHRs to collect FHx to meet Meaningful Use (MU) requirements. Though the nature of what an FHx tool collects is limited in scope (e.g., first-degree relatives only), the MU requirement precedes the spike in FHx tool releases and acquisitions between 2014 and 2015. Clearly, the government plays an influential role to facilitate the development and use of FHx tools.

Several FHx tools are not accessible or are no longer actively supported (AH, IH, MFH, OFH). It would be important to conduct interviews to understand the rationale for initially investing in the development of a FHx tool and the factors and decisions that led to its abandonment. Conversely, exploring the tools that have gained traction would also be worth investigating to understand successful attributes of FHx tools. However, it is unclear how the usage of these FHx tools compares. Such usage data is not readily available because FHx tool owners keep such information confidential. Nevertheless, it could be estimated through interviews with providers and web traffic monitors. However, such an investigation was outside the scope of the current research project. FHx tools that are associated with a consistent funding stream or commercial incentive are more likely to invest in broad dissemination and adoption of their FHx tool. While there does not appear to be much commercial opportunity to market a patient-centric FHx tool by itself, combining with a clinician-facing FHx tool (five FHx tools) or driving sales for genetic services (eight FHx tools) appears to be promising sustainability approach.

Finally, this assessment represents the features as they exist currently (mid-2017). Because new FHx tools will become available and current tools will continually improve, we will maintain a living inventory of FHx tools on the Global Alliance for Genomics and Health website (Catalogue of Global Activities 2018a).

Conclusion

There is a wide variety of FHx tools available to help patients collect their FHx. Tools vary by how they are organized, displayed, collected, and integrated into the clinical workflow. This paper provides a helpful summary for health care providers, researchers, and patient advocates interested in understanding the differences among the available FHx tools. Understanding the FHx tool that best meets the patient's and clinician's needs will ultimately lead to better clinical outcomes through more accurate identification of disease risk, appropriate genetic testing, and initiation of corresponding disease intervention and surveillance.

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Compliance with Ethical Standards

Conflicts of Interest BM, HM, and JS are founders and shareholders of ItRunsInMyFamily, Inc.

MD, during her time at Cleveland Clinic, was a co-inventor of the MyFamily (now called MyLegacy) intellectual property portfolio. The IP is licensed to Family Care Path, Inc. As part of this license, MD is entitled to a share in both royalties and returns on equity.

KW, RA, KB, and CH declare that they have no conflict of interest.

Human Studies and Informed Consent No human studies were carried out by the authors for this article.

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