Will Family Health History Tools Work for Complex Families? Scenario-Based Testing of a Web-Based Consumer Application

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Abstract

Objective: *Identify challenges that people from complex families may encounter when using traditional consumer family health history (FHH) applications and examine assumptions on which the applications are based.*
Method: *Scenario-based testing was employed in which three evaluators used a consumer FHH application guided by four scenarios, recording the challenges they encountered and time required. Challenges identified were analyzed through qualitative content analysis of field notes.*
Results: *Several types of FHH information deemed important in the scenarios could not be entered into the traditional FHH. Evaluators reported frustration at being unable to enter some information and perceived the resulting FHH as less accurate than it could be. These observations challenge certain implicit assumptions about families and consumers on which FHH applications are based.*
Conclusion: *Current consumer FHH applications mirror clinical FHH tools, which may not be the most appropriate approach for consumers, especially for people from complex and diverse families.*

Introduction

Clinicians have long elicited and analyzed patients' family health histories in order to predict risk for familial diseases. As consumer health informatics tools become more widely available, responsibility for documenting family health history (FHH) is moving from the clinician to the consumer. Concurrently, families are becoming increasingly diverse and complex, with more families being formed by step, foster, adoptive, and other non-genetic relationships. Because of these changes, practices associated with FHH warrant examination and updating. The purpose of this project was to identify potential challenges that people from complex and diverse families may encounter with traditional FHH tools.

Background

Family health history is a familiar clinical risk assessment tool. Traditionally, the FHH is a record of the health of genetically related family members, often depicted as a pedigree or family tree. The FHH usually includes information about three or four generations of family members [1, 2], including medical diagnoses, psychiatric diagnoses, known genetic mutations, general medical conditions or symptoms, complications of pregnancy, and cause of death for deceased family members [1, 3-11]. Although FHH has long been considered an important tool for risk assessment, its importance has been increased by genomic discovery and the resulting realization that most diseases are the result of complex interactions between multiple genes and environmental factors, all of which are reflected in the FHH [2, 5]. By analyzing a patient's FHH, a clinician can identify diseases for which the patient is at increased risk. The patient can then be counseled to reduce modifiable risk factors for those diseases. This approach is an important component of personalized medicine and has the potential to reduce the incidence of serious and chronic disease [5, 11].

Because collecting and analyzing FHH requires significant time and resources, responsibility for collecting and documenting FHH is being shifted to the consumer [5, 12]. Electronic consumer health tools for this purpose are in development and production; examples include Family Healthware [12] and OurFamilyHealth [13, 14]. The Surgeon General's Family Health History Initiative is a well-known national FHH project. It provides an online FHH application for consumer use, My Family Health Portrait (available at https://familyhistory.hhs.gov), hosted by the U.S. Department of Health and Human Services (HHS). This application enables a consumer to enter information about his own health and that of his genetic family members, save the information in a file on his computer, and create a printout to share with his health care providers [15]. (A more complete description of My Family Health Portrait can be found in [15]).

Preliminary evaluations of consumer FHH tools have demonstrated both strengths and areas for improvement. For example, a validation study of My Family Health Portrait demonstrated that the data consumers entered about family risk of diabetes and certain cancers were similar to data obtained by a genetic counselor, though genetic counselors obtained more complete and accurate information about family risk of coronary artery disease and stroke [16].

Analysis of OurFamilyHealth use demonstrated that users who elected to use the application were willing to spend a reasonable amount of time with it and entered information about a variety of diseases, but researchers noted that the number of family members in the histories was lower than ideal, and that a number of consumers arrived at the application and navigated away without using it [13]. Additionally, consumers used the FHH to track different diseases than clinicians; for example, consumers tracked allergies and arthritis while clinicians were more likely to track FHH of cancer [13].

Perhaps some of the unexpected consumer use patterns with FHH tools can be understood by examining certain implicit assumptions that underlie current consumer FHH tools. Because the tools are generally based on a clinical definition of FHH and clinicians' needs regarding FHH, their successful use relies on consumers sharing the clinical understanding, knowledge, and value of FHH. Specifically, use of these tools assumes that (1) consumers know or have access to genetic FHH information; (2) consumers value FHH for the same reasons clinicians do, that is, for examining genetic inheritance; and (3) the approach and format for FHH used in clinical practice, that is, collecting and documenting information about each individual family member to create a pedigree or family tree, is a reasonable approach and format for consumers to use.

Preliminary evaluations of these tools have been conducted with volunteer populations that tend to be motivated, young, and well educated. The suitability of consumer FHH tools for more diverse populations is yet to be determined. People from complex and diverse families are a group that may especially be expected to have difficulty with traditional genetic FHH tools, because their experience of family is markedly different than the genetic family of clinical FHH interest. Complex and diverse families are non-genetically defined families, including families formed by adoption; single parent families; same sex couples with and without children; and blended families in which the mother, father, or both have had children with more than one partner, resulting in multiple step parent, step sibling, and half sibling relationships. These complex and diverse families make up a substantial and growing proportion of American families [17-20]. Therefore, meeting the FHH needs of this group is an important priority.

This project employed lab-based usability testing to identify potential challenges that people from complex and diverse families may have when attempting to use a typical genetic FHH consumer application. Identifying these challenges may provide baseline knowledge for future FHH research with complex and diverse families, and may help provide insight about inaccuracies in the assumptions about consumers and FHH that are implicit in consumer FHH tools.

Methods

Scenario-based usability testing as described by Zayas-Caban and colleagues [21] was employed, in which testers use a tool guided by information and instructions contained in a scenario, recording their time on task and challenges encountered. Four FHH scenarios were developed by the first author based on deidentified family health histories collected in a previous study of healthy young adults [22, 23]. The scenarios were developed specifically to evaluate usability for people from non-genetically defined families, and described four complex families:

1. a family formed by adoption by a biological relative,
2. a family with step and half-sibling relationships,
3. a family in which in the index person received an organ donated by a biological relative, and

Scenario 1: Don

You are Don, a 21-year-old male with a history of kidney disease due to a congenital kidney disorder. You received a kidney transplant from a living donor who is your cousin. You have two siblings: an older sister (she is 25) and a younger brother (he is 17), both of whom are healthy. Your sister is the surviving twin of a premature birth; her identical twin sister died in infancy of complications related to prematurity.

Your mother was diagnosed with hypertension in her 40s. Your mother and siblings are all physically active. Your father is deceased, having died from liver failure related to alcohol abuse. He and your mom divorced when you were an infant, and you didn't really know your father and don't know much about him.

Your maternal grandmother is living and well, but she has smoked cigarettes since her teens. Your maternal grandfather has prostate disease. You have 2 maternal aunts. The older one smokes and is physically active; the younger aunt is sedentary. You have one maternal uncle who smokes and is physically active. Your paternal grandfather is deceased, and you don't know anything about his health or his cause of death. Your paternal grandmother has heart valve disease.

You have one nephew, the son of your sister, and he is healthy. You have a male cousin, the son of your younger maternal aunt, who is an alcoholic. You have another cousin, the son of your older maternal aunt, who is age 26, healthy, and who donated his kidney to you. You don't really know him well, though, and don't know much about him.

You also think of your sister's husband as an important member of your family. He is healthy and active. Your neighbor has been a "dad" figure for you. He is obese, had hypertension diagnosed at age 60, and is physically active. Your neighbor's wife is close to you too, and she was diagnosed with hypertension in her 50s, used to smoke but quit smoking at age 40, and she is physically active.

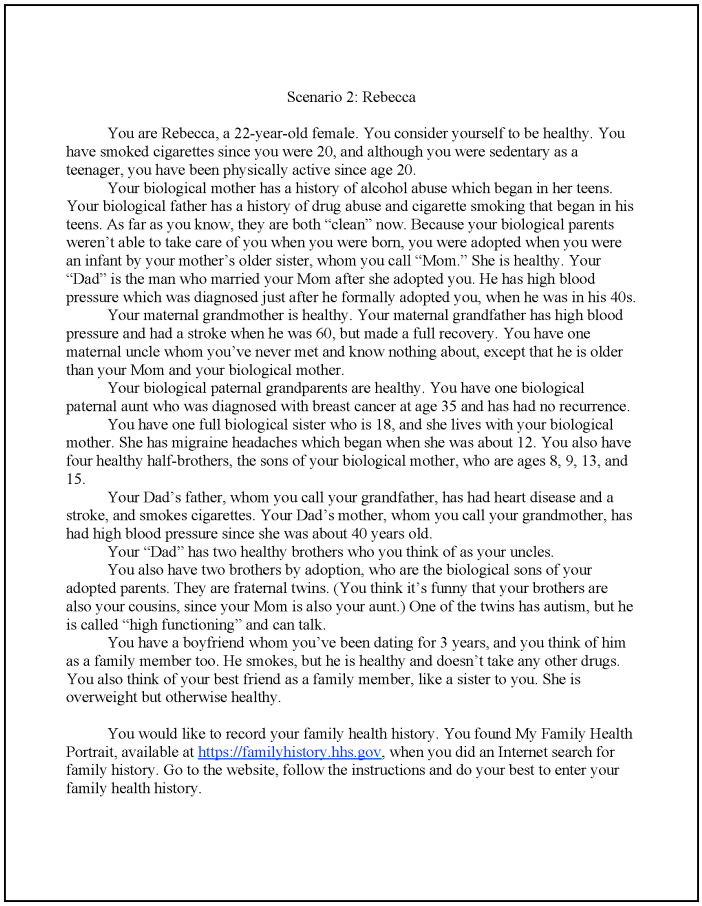
You want to record your family health history. You decide to use some software you found doing a Google search, My Family Health Portrait, available at https://familyhistory.hhs.gov. Go to the website, follow the instructions and do your best to enter your family health history.

Figure 1. Scenario 1, organ transplant recipient with estranged father.

4. a family headed by a lesbian couple with children.

The scenarios described the family and the health of family members, and instructed the tester to create a FHH using My Family Health Portrait (see example scenarios, figures 1 and 2). My Family Health Portrait was selected for the evaluation because it is widely available, hosted by HHS, and is representative of traditional genetic FHH consumer applications.

The evaluation was completed by three health informatics graduate students, all of whom were registered nurses, in the context of a directed research experience for course credit. The project was reviewed and exempted from further review by the organization's Institutional Review Board for the Protection of Human Subjects. Each student evaluator independently completed each of the four scenarios, using My Family Health Portrait to enter the FHH as instructed in each scenario. The evaluators maintained written field notes of the time required and challenges encountered while attempting to complete the tasks in each scenario.



Scenario 2: Rebecca

You are Rebecca, a 22-year-old female. You consider yourself to be healthy. You have smoked cigarettes since you were 20, and although you were sedentary as a teenager, you have been physically active since age 20.

Your biological mother has a history of alcohol abuse which began in her teens. Your biological father has a history of drug abuse and cigarette smoking that began in his teens. As far as you know, they are both "clean" now. Because your biological parents weren't able to take care of you when you were born, you were adopted when you were an infant by your mother's older sister, whom you call "Mom." She is healthy. Your "Dad" is the man who married your Mom after she adopted you. He has high blood pressure which was diagnosed just after he formally adopted you, when he was in his 40s.

Your maternal grandmother is healthy. Your maternal grandfather has high blood pressure and had a stroke when he was 60, but made a full recovery. You have one maternal uncle whom you've never met and know nothing about, except that he is older than your Mom and your biological mother.

Your biological paternal grandparents are healthy. You have one biological paternal aunt who was diagnosed with breast cancer at age 35 and has had no recurrence.

You have one full biological sister who is 18, and she lives with your biological mother. She has migraine headaches which began when she was about 12. You also have four healthy half-brothers, the sons of your biological mother, who are ages 8, 9, 13, and 15.

Your Dad's father, whom you call your grandfather, has had heart disease and a stroke, and smokes cigarettes. Your Dad's mother, whom you call your grandmother, has had high blood pressure since she was about 40 years old.

Your "Dad" has two healthy brothers who you think of as your uncles.

You also have two brothers by adoption, who are the biological sons of your adopted parents. They are fraternal twins. (You think it's funny that your brothers are also your cousins, since your Mom is also your aunt.) One of the twins has autism, but he is called "high functioning" and can talk.

You have a boyfriend whom you've been dating for 3 years, and you think of him as a family member too. He smokes, but he is healthy and doesn't take any other drugs. You also think of your best friend as a family member, like a sister to you. She is overweight but otherwise healthy.

You would like to record your family health history. You found My Family Health Portrait, available at https://familyhistory.hhs.gov, when you did an Internet search for family history. Go to the website, follow the instructions and do your best to enter your family health history.

Figure 2. Scenario 2, Woman adopted by a biological relative

The lists of challenges were analyzed using qualitative analysis methods. Each of the three authors individually reviewed all challenges identified by the evaluators. Each author independently coded the challenges through a process of open coding for conventional qualitative content analysis. The authors distilled the coded themes into categories, and collaboratively reached consensus about themes, categories, and examples of each.

Results

Time on task. The time required to complete one scenario ranged from 45 minutes to 90 minutes, with an average of one hour spent completing a scenario. Evaluators were similar in their recordings of time on task.

Challenges. Evaluators recorded multiple challenges that were grouped into three categories by the authors: difficulty or inability to enter specific data; website functionality challenges; and negative user responses. Multiple examples were identified in each of these categories.

Difficulty or inability to enter specific data. A primary limitation noted was the difficulty or inability to enter specific data contained in the FHH scenarios. All evaluators found the website to be missing desired data entry options. The most common complaint in this area was the inability to document non-genetically related family members such as adopted family members, boyfriend/girlfriend, and mother- or father-in law. Evaluators noted that they could not enter both a biological mother and an adoptive mother for one individual. The inability to enter distant genetic relatives such as great grandparents was also identified as a limitation. Thus, evaluators noted their belief that the FHH recorded was not as accurate as it should have been. Evaluators also noted that several key family health factors contained in the scenarios were either not available options in the application, or were not sufficiently specific to enter in the application (see Table 1).

This inability to enter family health information limited evaluators' ability to enter more complex family characteristics. One evaluator noted "the site is not able to record health and family stories, just actual conditions." As a result, there was a mismatch between a user's storytelling style of FHH and the structured FHH of the website. In many cases, the evaluators were unable to enter specific information because the choices on the pick lists did not apply, and a lack of fields for free-text entry was apparent. In an example of the challenges associated with entering

complex family information, an evaluator noted "the assumption is that siblings are from the same parent although this is not always the case."

Table 1. Family health factors that could not be entered

Family health factor	Example(s)
Select causes of death	Motor vehicle accident Premature birth Unknown cause of death
Developmental conditions	Developmental delays
Select health conditions	Drug addiction Liver failure
Lifestyle factors	Smoking history Substance use Sedentary lifestyle
Severity of health conditions	Mild autism
Course of health conditions	Breast cancer treated without recurrence Stopped smoking
Non-specific information about family members' health	Muscle weakness Delayed speech Delayed walking

Website functionality challenges. Several technical issues were noted by the evaluators. Errors noted included an apparently accidental mixing of English and Spanish on the screen, and download instructions for Microsoft Internet Explorer that showed screenshots from Mozilla Firefox instead. One evaluator noted that when entering information about a half-brother, a persistent error occurred repeatedly which eventually caused the evaluator to give up. Browser pop-up blocker applications limited the ability to show FHH diagrams and data entry screens. One evaluator discovered that the Cancel button deleted the entire session rather than canceling out of the current screen as had been expected, and lost the complete session.

Evaluators also noted that they wanted and needed types of functionality currently not available. Lack of free-text entry fields prevented evaluators from being able to describe relatives' comprehensive health characteristics. Evaluators also wished for more graphical manipulation and less typing. Several complaints were recorded about the inability to quickly document that nothing is known about a specific family member (e.g. a deceased grandparent). Evaluators noted that more specific age ranges would be useful; when entering an age for elderly family members, only an age range of "60 +" was available. One evaluator noted the need for better error correction functionality: upon finding an error after data entry, he had to delete a family member because he was unable to change the relationship between the family member and the index person, which had been incorrectly entered.

Evaluators were surprised that they had to save their work on a local computer or in a Microsoft HealthVault account rather than being able to create an account on the website that would allow them easy future access for editing the FHH. Finally, evaluators wished for more ability to share the FHH with electronic health records or third-party applications on computers or mobile devices.

Negative user responses. Though not specifically asked to record feelings or emotional responses to completing the scenarios, evaluators noted frustration several times, primarily associated with being unable to enter information that was available in the scenarios and appeared potentially important to the evaluator. A particularly frank comment, "I don't like this one," was recorded in response to the scenario in which the index person was adopted by a biological relative, along with "not user-friendly for people not having traditional family structure."

Positive feedback. Although evaluators were only asked to record challenges associated with completing the scenarios, they noted several positive features related to My Family Health Portrait, too. The evaluators noted that as an electronic FHH application designed for the general population, the resource offered a website that was relatively easy to use. The opening screen enabled users to create either a new FHH or upload a locally-saved XML file from a previous session. One evaluator commented that "the main page is easy to use." The legend for the family history diagram and FAQ were described as helpful.

One evaluator noted that "it's nice to have multilingual support," referring to the availability of English, Spanish, Portuguese, and Italian. Evaluators also noted that specific diseases were grouped in more easily understood terminology; after selecting a category of disease/condition, the user was then able enter more specifics if warranted. A glossary defined medical terminology. Evaluators also commented positively on apparent learning by the system: once a disease had been selected for a family member, it remained on the pick list for future use in that session. In addition, evaluators were impressed by the system correctly constructing a pick list of relatives who could be the parents of a family member being added.

Evaluators also responded positively to the option to import family health information from Microsoft HealthVault. Once a FHH had been completed, the user was able to save the information to their local computer or to HealthVault. Instructions for downloading family histories were described as "nice and easy." By storing the family history on the local computer or in Microsoft HealthVault, one evaluator noted "it is nice to have the ability to make a copy of your data and give it to another family member." Printing of FHH was described as "easy to do and then share with relatives."

Discussion

This scenario-based usability evaluation demonstrated several potential challenges that people from complex and diverse families might encounter when using traditional FHH consumer applications. These challenges may be evidence that the implicit assumptions described above underlying consumer FHH applications may be inaccurate, at least for these types of families.

Assumption 1: Consumers have access to genetic FHH information. In fact, people from families formed by adoption, single parent families, and blended families may not have access to health information about some or any genetic family members [23]. The evaluators discovered that the scenarios, based on actual FHHs, often did not contain the specific information required by the application. Additionally, evaluators noted that they had a wealth of information, which they believed was important FHH information, that could not be entered into the application, resulting in frustration. A consumer facing similar frustration might choose to not use the application.

Assumption 2: Consumers will understand and value FHH for examining genetic inheritance. Consumers may value FHH for reasons that are different than clinicians'. For example, consumers may wish to emphasize the environmental and behavioral risk factors shared by family members more than is possible with traditional FHH tools [12, 23, 24]. Evaluators in this project noted frustration with being unable to enter potentially important lifestyle factors, like substance use and sedentary lifestyle, into the FHH. This is consistent with the findings of other researchers regarding consumers' desire to incorporate lifestyle factors related to health, and health conditions of less interest to clinicians, in FHH applications [12, 13]. It is possible that people from complex and diverse families would place even more value on lifestyle factors and the influence of the social family than is allowed with traditional FHH tools because that is the information available to them. In some cases, consumer FHH tools may simply not accept some of their FHH information, such as multiple partners, or a combination of adoptive and birth relatives as documented by the evaluators in this project. Dissonance between the consumer's own conceptualization of family and the genetic family of the traditional FHH, and negative emotional responses to FHH tools that are inconsistent with the personal experience of family [23, 25], are important considerations.

Assumption 3: The approach and format for FHH used in clinical practice, collecting and documenting information about each individual family member to create a pedigree or family tree, is a reasonable approach and format for consumers to use. Evaluators noted several challenges with this approach in constructing a consumer FHH. Information about FHH that was too general to be recorded, the lack of an easy method to indicate that nothing is known about a genetic relative, and difficulty entering specific types of information indicate that this approach may not be optimal for consumers. Requests for more graphical interfaces and opportunities for free text entry indicate potential improvements. The observation by an evaluator that the storytelling style of the scenario FHH did not match well with the structured FHH of the website may be a particularly important insight about the different ways that consumers and clinicians may conceptualize FHH.

Complex and diverse families constitute a substantial number of contemporary families, and therefore represent a consumer group that should not be overlooked in efforts to promote health and personalized medicine. In fact, attention to this group by the health care community is particularly warranted because complex families, especially those with multiple step parent, step sibling, and half sibling relationships, are more common among groups who are at increased risk for poor health outcomes, including populations who are of lower socioeconomic status [26, 27], less educated [28], have children at a younger age [27-29], and are African American [27-30]. Because people at

risk for poor health outcomes are more likely to live in complex families, attention to the health needs of these families is especially needed in order to address health disparities. Further exploration of the challenges identified in this project is vital to ensure that consumer FHH tools do not exclude these families.

The current project focused on only one consumer FHH tool, though that tool is relatively representative of currently available FHH tools and is the most widely available at this time. Additionally, this project used knowledgeable evaluators who are more familiar with electronic consumer health tools than many consumers, and who may have higher expectations about usability than typical consumers due to their interest and education in health informatics. Future research with consumers is needed to explore and validate the challenges discovered in this evaluation and to determine an optimal approach for consumer FHH applications. It is possible that inclusion of factors important to clinicians (e.g. traditional FHH elements) and factors important to consumers (possibly non-genetically related family members and lifestyle factors) will need to be blended in FHH applications in order to meet the needs of both groups and create truly useful tools. Such blending may prove useful for building knowledge about the genomic determinants of health because it may present a more complete picture of the combination of genetic and environmental factors leading to patterns of health and illness in families.

Some challenges identified in this study do not appear to be specific to complex, diverse families. Functionality challenges (e.g., mixing Spanish and English languages) could potentially affect any user. These issues were communicated with the website developers as appropriate. Other challenges identified may apply to consumer health applications in areas other than FHH. For example the desire for less typing and more graphical interface is likely to be applicable to other domains, as is the desire to store created files externally. It is interesting that evaluators considered saving the FHH file on their local computers rather than on the website to be a disadvantage; saving the file locally was envisioned as protecting privacy and promoting security. Perhaps as cloud computing models are becoming more widespread, users will be increasingly willing to store information externally, associated with the appropriate application, rather than preferring to maintain their confidential information on their own devices.

Conclusion

Consumer FHH applications are moving both ability and responsibility for collecting and managing FHH from the clinician to the consumer. As consumers become more involved in FHH, attention must be paid to usability of FHH tools for all types of families. Complex and diverse families make up an important and substantial number of contemporary families, and may have unique challenges when using traditional genetic FHH applications. The potential challenges identified by scenario-based testing in this project represent a starting point for further investigation in order to ultimately meet the FHH needs of all types of families.

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