HHS Public Access

Author manuscript

Health Policy Technol. Author manuscript; available in PMC 2022 June 01.

Published in final edited form as:

Health Policy Technol. 2021 June ; 10(2): . doi:10.1016/j.hlpt.2021.100511.

Usefulness of mobile apps for communication of genetic test results to at-risk family members in a U.S. integrated health system: a qualitative approach from user-testing

Cameron B. Haas[1,2,*], **Aaron Scrol**[1], **Chethan Jujjavarapu**[3], **Gail P. Jarvik**[4], **Nora B. Henrikson**[1]

[1]Kaiser Permanente Washington Health Research Institute, Seattle, WA

[2]Department of Epidemiology, School of Public Health, University of Washington, Seattle, WA

[3]Department of Biomedical Informatics and Medical Education, School of Medicine, University of Washington, Seattle, Washington, USA

[4]Departments of Medicine (Medical Genetics) and Genome Sciences, University of Washington School of Medicine, Seattle, WA

Abstract

Objective: To assess the usefulness a mobile based application to send genetic test results to at-risk family members in a U.S. integrated health system.

Methods: We conducted semi-structured in-person interviews with members of Kaiser Permanente Washington who had enrolled in a prospective study and received genetic test results. Participants were given the task to use the app and comment on the experience. The moderator asked participants to share perspectives on the usefulness of a mobile based app and their lived experiences of sharing their test results with family members.

Results: Fourteen study participants who had undergone genetic testing were interviewed. Four primary themes emerged as relevant to the use of mobile-based apps as a tool for communicating genetic test results to at-risk family members: (i) Participants felt a sense of obligation to share positive test results with relatives; (ii) Participants felt that the advantages of using email were similar to those of the app; (iii) Participants felt that younger individuals would be more comfortable with an app; and, (iv) Participants felt they could use the app independently and in their own time.

Conclusion: A mobile based app could be used as a tool to improve cascade screening for pathogenic/likely pathogenic test results. The benefits of such a tool are likely greatest among

[*]Corresponding author: Mailing address: 1730 Minor Ave., Ste. 1600, Seattle, WA 98101.

Conflict of interest

Cameron B. Haas, Aaron Scrol, Chethan Jujjavarapu, Gail P. Jarvik, and Nora B. Henrikson declare that they have no conflicts of interest.

relatives still at the stage of family planning, as well as among family members with strained relationships. There would be minimal burden on the system to offer a mobile based app as a tool.

Public interest summary

It is important that when an individual receives genetic testing they inform their family members of the results so that their relatives may also benefit. However, this is not commonly done. We created a mobile app that would help facilitate this communication between family members. In this study, we talked with patients about how they felt about the mobile app and if they would use it for sending their genetic test results to any family members. We found that most patients would use a mobile app for their younger relatives, such as nieces or nephews, or with family members with whom they have little contact. While a mobile app might not be the best option in all circumstances, we show that it could be offered as an additional tool to help increase the number of relatives who are informed about their need for genetic testing.

Keywords

Cascade testing; Mhealth; Genetic testing; Mobile app; Communication

Introduction

Cascade testing programs, where relatives of patients who test positive for pathogenic or likely pathogenic (P/LP) variants are contacted and invited to receive clinical follow-up and testing, are an important strategy for precision medicine[1–3]. Such programs have the potential to identify patients with high penetrance genetic diseases for intervention and treatment, improving health and reducing mortality[4]. However, the success of cascade screening depends on the communication of pathogenic findings to at-risk family members[5]. Presently, patient referral rates for genetic counseling and testing for at-risk family members remain low[6,7].

Once a patient has received a genetic test result, they are responsible for contacting and informing at-risk relatives to provide the information[8]. However, up to a third of at-risk family members go un-notified and do not benefit from genetic counseling, testing, clinical follow-up, and continued detection of P/LP carriers through cascade screening[4,9–12]. Previous research suggests that more distant relatives (i.e., second or third degree relatives) are less likely to be informed, as well as those with emotionally strained relationships[4,9–11,13–15]. Variant carriers have expressed a feeling of duty to inform relatives, suggesting that more ways to facilitate the communication of test results to extended family are warranted[4,9–11]. However, trials attempting to increase this communication through skill-building have not been effective[16,17]. Providing educational material for patients to share with their relatives has been shown to increase the likelihood that informed family members undergo genetic testing[18].

Our objective was to evaluate the usefulness of a mobile application (app) as a communication tool for sending genetic test results to at-risk family members among a sample of Kaiser Permanente Washington (KPWA) members from the Electronic Medical Records and Genomics (eMERGE) Network, phase III study for which genetic testing had

been conducted for all study participants. We used these patient interviews to also understand where and when in the clinical workflow the app could be presented and to gauge the level of support needed in order to successfully send the information.

Materials and Methods

In conjunction with a user-testing study of the mobile app described below, we conducted semi-structured in-person interviews with individuals who had undergone genetic testing and had received their test results using the eMERGEseq panel, which included sequencing of 68 genes and 14 single nucleotide variants, including 59 ACMG recommended genes[19]. We sought to follow the COREQ guidelines during study design and analysis[20]. Our research team consisted of three qualitative researchers and program managers at KPWHRI (CH, AS, NH) and two researchers based at the University of Washington (CJ, GJ) with experience in human centered design and clinical implementation of genome sciences. The application programming interface of the ShareDNA app was conducted by the team at the University of Washington, while qualitative work around the conceptual use of apps for communicating genetic test results was conducted and financed by grants for researchers at KPWHRI in an effort to minimize bias towards incentivizing use of an app during interviews.

Mobile application

ShareDNA is a mobile app designed to help patients communicate genetic test results to their at-risk relatives. Development of the mobile app was funded by a Research Innovation Award from the Institute of Translational Health Sciences at the University of Washington. The app uses encryption software for safety and privacy of users. The free mobile app is currently available on both iOS and Android operating systems. As part of our qualitative study, we assessed the perceived ease of use (usability) of the tool by observing the participants walk through the procedures and functions for sending family members their genetic test results, followed by the 16-item Post Study System Usability Questionnaire (PSSUQ) version 3[21], a validated instrument designed for usability evaluations. Results from the PSSUQ component of the interview and app development were generally favorable and are reported elsewhere[22] and were used to improve the usability of the ShareDNA app. Details regarding the improvements made to the app as a result of participant feedback are also reported in previously published work on the development of the ShareDNA app[22].

Following introductory text, the user is first required to create an account by providing an email address and creating a password. They then upload a document containing their genetic test results by either uploading a file from documents saved on their device or taking a picture of the document. The app allows the user to send a message through email or text message. Generic language auto-populates the subject line (if sending via email) and the body of the text, with the ability to alter and customize as desired. Finally, the user may add contacts directly from the device's list of contacts or enter new contact information, along with identifying their relationship to the recipient(s). Multiple recipients can be added to a single message. Within the app, links are provided that direct users to a local medical genetics clinic at the University of Washington or to national providers listed by the National

Society of Genetic Counselors. Links to more educational material are also available for from Medline and the National Cancer Institute for general genetic conditions (Medline) and specific information for hereditary cancers (National Cancer Institute). Recipients receive the message from the phone number or email of the sender, directing them to download and enter the ShareDNA app. Once the recipient has created their own account, they are able to access the senders' documents through their own list of files. Informational pages and links are embedded into the app which describe the study team and directs users to resources for genetic testing and counseling.

Participant selection and setting

The eMERGE study is a national network, organized and funded by the National Human Genome Research Institute (NHGRI), that combines DNA biorepositories with electronic medical record (EMR) systems for genetic research in support of implementing genomic medicine.

Our original sampling frame was participants in the KPWA/University of Washington site of the eMERGE study who had received genetic testing as part of population-based genetic screening and received P/LP results. We sent invitation letters to all 48 participants with a P/LP genetic test result, of which 8 enrolled, 6 called for more details but declined participation, and 34 did not respond. Based on this limited response, we expanded the sampling frame to also include people with benign/likely benign (B/LB) results. With the goal of reaching a near equal sample size between P/LP and B/LB participants, we then mailed invite letters to 100 members with B/LB results with oversampling for women and racial/ethnic minorities, of which 6 enrolled, 5 responded but declined, and 89 did not respond. We used collaborative team analysis meetings to assess when we had reached data saturation, or extensive repetition of themes without novel themes emerging. After the second round of invitations, we concluded we were close to data saturation and we did not seek to expand the sampling frame further. All participants provided written consent before beginning the interview and received $50 as a thank you for participation. The expected length of each interview was approximately one hour.

Data collection and interviews

We used the Technology Acceptance Model (TAM), an information systems theory created to assess how users come to accept and use a device, to develop the interview guide[23]. Under the TAM, two components, perceived usefulness and perceived ease of use, capture the attitudes of participants and the behavioral intention to use the tool[23]. The perceived usefulness is intended to assess the individual's perception of how a tool will enhance the desired task, in this case communicating pathogenic results. Open-ended questions were used to capture the perceptions of participants on the usefulness of the app in delivering genetic test results to at-risk family members.

We developed an interview guide through an iterative process to include questions that would reflect the primary research objective. Interview domains included: (a) the potential benefits of using apps for family communication, (b) timing of using the app relative to

getting test results, and (c) predicted reactions from relatives when using the app. We conducted internal pilot testing for clarity and phrasing of the questions, and to limit respondent perception of socially desirable responses[24]. The full interview guide is available in Appendix A. Language at the beginning of each interview was included to clarify the independence of the qualitative interviewer (CH) from app development to reduce the impact of social desirability bias on the participants.

All interviews were conducted in-person between October and December, 2019. Interviews consisted of two stages: (1) usability testing of app functionality, and (2) a semi-structured interview. We conducted all interviews with the aid of a study electronic tablet (Apple iPad) onto which the app was preloaded and hypothetical results and contact information were provided. We provided participants with dummy email addressed to create accounts for the app and then to share fabricated genetic test results with two hypothetical family members using the app. The iPad screen recorded their interactions with the app and was paired with audio recordings which were professionally transcribed.

Data Analysis

Each interview resulted in five documents: 1) a demographic survey describing their age, gender, cancer history, race/ethnicity, and general mobile device use; 2) a video recording of the mobile device while performing the requested task using the ShareDNA app; 3) the PSSUQ; 4) the audio recording of the interviews; and, 5) the transcription of the interview. Interview documents were imported and analyzed using ATLAS.ti 8.4.

We used a template analysis approach for data analysis[24]. Using a deductive approach based on the research questions and the interview guide, two researchers trained in qualitative research developed a set of analysis templates. One study team member (CH) piloted the templates on a subset of interviews and emerging themes were added as needed. A second team member (NH) piloted the subset of interviews using the revised analysis template then coding was checked for general concordance. One study team member (CH) then coded all interviews using the final version of the analysis template and developed initial coding memos for recurring themes. Two study team members (CH, NH) collaboratively synthesized the themes, with illustrative quotes, across interviews through development of a final coding memo. We evaluated coding memos and quotes for differences in codes by participants with P/LP results versus B/LB, which allowed for stratified thematic analysis. Using content analysis as a theoretical framework, we organized major and minor themes with supporting quotes and selected exemplar quotes to reflect responses from different participants.

Results

After invitations to 149 individuals (in two waves) our response rate was 9% (14/149), 8 from among 49 P/LP test results and 6 more from 100 participants with B/LB test results. Our final sample included 14 participants. There were 2 participants with self-reported cancer diagnoses, and another 5 had a history of multiple colonic polyps without a cancer

diagnosis. Most interviewees were male (57%) and self-identified as White (64%), with a mean age of 69 (range=60–93) years (Table 1).

Participants reported having received their genetic test results within the last 2 years at the time of interview. Among the 8 participants with P/LP test results, 7 had already shared their results to some extent with at-risk family members. Among those 7 who had informed their family members, all reported having done so over phone for at least one relative and 4 mentioned the use of email for communicating with extended family members. Among the 6 participants B/LB test results, none reported having formally shared their genetic test results with family members beyond their significant other.

Following user-testing of the app, 13 of 14 participants felt that they would use the app with at least one relative to communicate P/LP test results. When asked whether they would use an app to share genetic test results some had responses similar to: “I think the app is easy -- I think the app is a way to share information” (B/LB participant 111). However, another participant said that they “… probably would have gotten frustrated and just -- made a Xerox as opposed to sending a file” (P/LP participant 106).

Four main themes emerged as most relevant to evaluating the feasibility of using a mobile based app for communicating pathogenic test results to at-risk family members: (i) participants felt a sense of obligation to share positive test results with relatives; (ii) participants felt that the advantages of using email were similar to those of the app; (iii) participants felt that younger individuals would be more comfortable with an app; and, (iv) participants felt they could use the app independently and in their own time. Table 2 presents these major themes with exemplar quotes.

Participants report a sense of obligation to share positive test results with relatives

Of the 8 P/LP, all participants brought up that when they were informed that they had tested positive for a heritable condition they felt a sense of responsibility to inform their relatives. Of the 6 B/LB, 5 participants expressed that had their tests been positive they would have felt a greater duty to inform relatives. One participant mentioned this feeling of responsibility throughout the interview: “Because I felt like I was sort of on the spot, suddenly responsible. And I think it's right that it's sort of up to that person or me to pass it along or not” (P/LP participant 104). Especially in cases when preventive measures can be taken, participants said that providing more information, such as actual test results and links to resources, to their relatives is seen as beneficial.

Participants expressed greater responsibility to inform younger relatives, particularly those in the stages of family planning.

All quotes related to this minor theme came from participants who had themselves received a P/LP test result. Communicating the information to nieces and nephews appeared to present more urgency than for older relatives: “… yes, I think I would use it with my nephew, and he has a child” (P/LP participant 106). Some of participants placed particular importance on relatives with progeny: “And my siblings too. Because I was concerned about if my brother had it, his son might have it. One sister had a son, so I told her” (P/LP participant 103). Family planning for younger relatives was an important consideration when

describing the urgency of communicating with at-risk family members: “One prompt, probably the most severe, was something where I knew my nephew was thinking of having children, and I thought it would be very important for him to know that he should do a special test. And so in that case, I definitely knew I wanted to do it” (P/LP participant 107).

Participants acknowledged complexities in sharing results with relatives with whom they experienced strained relationships

Among all 14 participants, 7 discussed scenarios in which communicating with an estranged family member may be challenging. While some participants expressed that this feeling of obligation extended even in cases in which the relationship was “strained,” there was an acknowledgement of the complexity of some familial relationships that they could see that might justify not communicating. When asked about why they would choose to use text messaging as the means of informing their sister, one participant said: “Because the relationship that I have with my sister is a little bit strained anyway. And I feel like it's easier for her to approach a conversation with me, regardless of the subject, if it's not necessarily by phone” (B/LB participant 114). A similar scenario was used as an example for when “it may be better for the message to be a little more anonymous and sterile […] For example, if there's a distant cousin that you have a bad relationship with but you still want to inform them” (P/LP participant 107).

Participants felt that the advantages of using email were similar to those of the app.

When discussing participant preferences for mode of communicating test results to family members, 6 of the participants drew comparisons to the mobile app. Participants discussed the various modes of communicating P/LP results to family members, including doing so in-person, over the phone, through email, or via text message. No single method appeared to be the dominant preference for all relatives. The 4 participants who had P/LP results and used emailed to communicate the information to relatives mentioned that one reason they did so using email was the ability to attach the test results and have a permanent record to show a doctor. Several participants described their own experience using email to inform their relatives: “I also attached [the test results]-- I'm glad you mentioned that. The genetic counselor here at Group Health or Kaiser put together a very nice little -- sort of boilerplate summary of what the test results meant. And that was the best thing to have, because -- and the sort of generic template that I just looked at is similar, but not as focused on the particular test result that I had” (P/LP participant 107). The participant had included the language provided by their genetic counselor in the body of an email they sent to family members, along with a copy of their test results attached to the email. After using the app the participant felt that the process would be similar to using email, “But [the app] would be -- this would be much more convenient” (P/LP participant 107).

Participants also noted that email allowed for a single correspondence to be broadly distributed. When large numbers of people need to be contacted, participants described the appeal of email: “I mean, I have a huge family. And the thing I like about email is they can just forward it to their kids or their parents” (P/LP participant 101). Many participants had gone through the experience of passing along their test results to family members within the last couple of years. One participant described the use of multiple methods to communicate

the information with family members: “in some cases I called people. In most cases I sent emails because I could write it up and they would actually have something in front of them” (P/LP participant 105).

Email and text messaging were also seen as more plausible and advantageous for relatives who live further away: “most of my relatives live in Seattle. If I had more out of the area, I would think that would be -- sending an email or text would be faster or easier" (B/LB participant 112). Most individuals who lived in close proximity to their relatives seemed to prefer in-person communication for those relatives. “We have five kids in the house, no longer kids. So I spoke to them one at a time or together, however we could mix it up” (P/LP participant 102).

Email was also seen as almost a habitual form of communication among study participants, who felt a sense of familiarity in using email. Participants expressed somewhat of an innate appeal of email, as one participant explained: “because it's just what they've done all their lives” (P/LP participant 101). One participant described that “to be able to type something would have been very comforting” (B/LB participant 109), while another felt an inexplicable impulse: “I would prefer email. I don't know why” (B/LB participant 113).

The app used during the interview included data encryption software which required re-entering user passwords before sending files. We asked participants if this security measure was important to them and most felt that the re-entering of their password was more of an annoyance than a necessary feature. In general, this aspect was not seen as a concern for participants, but one did mention possible relatives who may have concerns: “Some of them are more sensitive to security on line, so they may or may not want electronic health information sent over the Internet and would rather have me print it out and mail it -- the hard copy” (B/LB participant 113).

Participants felt that younger individuals would be more comfortable with an app.

Nearly all participants, 12 out of all 14, expressed sentiment that an app may be easier to use for younger people and a more appropriate means of communicating their genetic test results to their younger relatives. Generational differences were highlighted as a determinant of user preference: “Now, two of them, my aunt and uncle, would be completely helpless with the app and downloading it off -- forget it. They're in their 80s. They would be helpless. But I would send them a hard copy. But the cousins, all of them, are very app -- you know, app users” (B/LB participant 109).

Participants felt that in the future app-based approaches to communicating this information would be more easily implemented: “Given the way things have changed with younger people than me, yeah. In fact, a whole bunch of them of my generation and older, they're just getting so facile with all this stuff and knowledgeable and -- mostly stimulated by grandkids probably and kids and all that” (P/LP participant 103). Considering the ages of our participants, some felt that even within their generation there would be variable aptitude for using an app, one participant felt that even “…10, 15 years [their] junior […] would find this much easier to use” (P/LP participant 106).

Participants felt they could use the app independently and in their own time.

Among the 7 participants who had informed relatives of their P/LP results, respondents reported taking varying amounts of time before informing their relatives of their risk. One participant told family members “Pretty quick, within a month” (P/LP participant 103) of receiving their own test results. Another participant “waited until after Christmas because it was roughly this time of year, and I just said I'm not going to screw with this before Christmas. I'm going to learn a little bit more and then after Christmas, probably January of that year, I started” (P/LP participant 105). After receiving P/LP test results, participants felt that they would need some time to process the information before starting to inform their relatives. When asked why they would wait before informing family members, one B/LB participant explained: “Because the last thing I would want at a time like that in my life was all of them coming back to me saying, oh, what are you going to do now? And I would have to say I don't know what I'm going to do now. I have no idea what's next. So I would wait” (B/LB participant 109).

Most participants, 11 of the 14, felt that they would be able to leave their genetic counselor or doctor’s office and successfully use the app to communicate with their family members. Some participants felt that instructional materials would be helpful, while other participants said they could figure out the app autonomously. Hypothetical scenarios were described by participants of how this might look in the clinic:: “And then using the app so you have the result or you can give the patient the option, now, what you could do is here's the information on this page here, go home and you can download it and think about who you want to share it with” (P/LP participant 103).

Several participants felt that the recipient should be contacted prior to sending genetic testing information via the app. These participants felt that a phone call or email would be necessary to prepare the relative for the information in order to avoid confusion or panic. While it may not be necessary for all relatives, some may want to be contacted before sending any information, “… you figure the average mom and dad in this case might be good -- it might even be a little much for them. You might want to start it with a phone call or something” (P/LP participant 104). Some participants felt that this it would be important to “have a pre-discussion with the family members that they are sending it to” (B/LB participant 111), especially if they were going to send the information using an app. Such pre-discussion could even entail the use of email: “I would almost want to send her an email before I sent her something from that explaining I got some test results that I'm going to share with you, and I'm going to send them to you through this app” (B/LB participant 114).

Discussion

We conducted a qualitative study about the usefulness of a mobile app for sharing genetic test results with relatives. We found that most participants felt comfortable using the app to send genetic test results to at-risk family members. Specific family relationships that may benefit from the option of a mobile app to communicate test results include younger relatives (e.g. nieces and nephews) and estranged family members. We found that most participants reported they could use a mobile app independently, which allows for patients proceed with sharing results in their own time. Many participants did not find the app to

have any advantage over emailing reports; suggesting that the advantage of encrypted data was not compelling to these participants.

Our study highlights that patients with P/LP genetic test findings feel a sense of responsibility to inform their at-risk family members, but that no single method of doing so is likely to be a panacea for delivering the appropriate information to all family members. Many of the advantages that the use of email provides translate easily to a mobile based app, namely the ability to attach documents (e.g. genetic test results) and as a form of mass communication (i.e. one message for numerous recipients)[18,25]. We anticipated that participants would prefer the security and encryption provided by the mobile app, but our study's participants did not appear to value these features[25]. Other studies have also noted that patients seem generally unconcerned that sharing genetic testing results with family members could compromise patient privacy[26,27], suggesting this may not be an important factor that would influence the use of an app for communicating genetic test results.

Our group of study participants were all older than 60 years of age and many felt that the appeal of using a mobile app to perform the task of informing relatives will likely increase in the coming years. Participants felt that a unique benefit of a mobile app was the curated links to resources, including genetic counseling and testing services[28]. Providing links to resources may have similar benefits as shown in a randomized trial in which dissemination of educational materials improved genetic testing and follow-up among recipients[18]. Access to as much health-related information as possible has been shown to be highly desirable for individuals considering genetic testing[29]. According to the responses from participants in our qualitative study, the additional burden on the health care system to offer a mobile app would be minimal, as most participants felt they could successfully use an app autonomously and that it allowed for the flexibility to do so when the timing felt right. Compared to previous clinical trials which have focused on skill-building and providing educational material, the time and labor for relying on an app would be fairly low[16,30–34].

The perceived duty to inform has been noted in previous studies[4,10,25,35,36]. Consistent with previous studies showing increased desire to disclose genetic information when relatives are still in family planning stages or the information affects multiple generations, a mobile app may fill the perceived need to prioritize relationship[37]. This observation has been observed previously in studies of the barriers and facilitators of family communication about genetic risk[4,9,10,15,38]. Consistently there was more interest in the utility of an app-based communication approach for younger relatives. Mobile devices have been an increasingly important tool for family communication, particularly in discussions regarding sensitive or complicated topics[39,40]. Together these two themes suggest that an app-based method for communicating genetic test results may be a preferred method to target relatives for which the risk perceived by the proband appears to be the greatest. Emotionally distant familial relationships has been recognized as a barrier in the communication chain and was mentioned as a possible issue by participants in our study[4,10,16,41]. While participants generally did not like the sterile and distanced approach of a mobile-based app, some noted that it may be an advantage when presenting the information in such circumstances because fulfills their perceived obligation to inform. This may suggest that a mobile app may be an

effective option for reaching family members who are the least likely to be contacted by more conventional methods, such as a phone call or seen in-person.

The sequence of events following a diagnosis of a pathogenic genetic variant should allow for patients to process the information and then allow for them to open communication with their relatives regarding the implications on their risk in their own time[42]. Participants did feel that the mobile app would allow for them to internalize the test results and then decide the timing for disseminating the information to their relatives. The advantage of location independence offered by m-health technologies has been a motivation for translational to clinical health care settings[23,40]. Participants generally felt a sense of self-efficacy in their ability to use the app without clinician assistance, an important psychometric measure related to disclosure of genetic cancer information[43]. Additionally, the ability to use an app independently and at home allows for the patient to initiate contact with relatives if they feel there should be precursory communication, as some participants suggested and as has been consistently shown in previous qualitative research[10,35,44]. In a clinical setting, genetic counselors could offer a mobile app as a means of communicating the information to at-risk relatives, recommending that they do so when they feel they are ready.

It is important to acknowledge the limitations of our study. Our sample size was small and reflects the constraints of a secondary study within a pre-defined sampling frame. Further, our sample size is consistent with many qualitative evaluations of eHealth[45]. However, the sample size and sampling frame did limit our ability to assess the experiences of younger, female, more diverse, and rural individuals. Our youngest participant was 60 years of age, so we cannot assess the perceptions of younger patients. Future work among younger participants will be important to confirm the assumptions described by participants in our study. Interviews were almost exclusively conducted in an urban area and possibly a limiting factor for individuals from more rural areas.

In our first round of recruitment, which was exclusively among those with P/LP results, only one female agreed to participate. While we oversampled for females in the following round of recruitment the perspective of males is likely overrepresented in our study, especially in stratified analysis by P/LP status. However, it may also be seen as an advantage as previous research has noted a gendered sense of responsibility to inform among women[46]. Future research should investigate gender-specific preference in forms of communicating genetic test results in order to understand how this may impact our findings.

Despite limitations, our study provided a unique opportunity to assess preferences concurrent with the use of a mobile app in people who had received genetic testing results. By conducting our interviews in conjunction with user-testing of a mobile app, we were able to explore the personal experiences of participants based on applied context of using an app rather than a hypothetical scenario. However, the hypothetical scenario of what our participants would have done had they had a mobile app when they originally received their genetic test results is a potential limitation. A randomized trial, as has been conducted for other interventions aimed to increase family communication of genetic test results and cascade screening[16,28,30,34,47], would be an important step to know the true impact such an app may have and may follow from this small usability study. We asked participants how

they would expect family members to react if they were to receive a message using the app and their likelihood of being tested, rather than asking the recipients themselves. Future work is needed to understand whether receiving such a message would affect the recipient's likelihood of seeking genetic testing. In our recruitment approach, patients with variants of unknown significance (VUS) were excluded. Patients with VUS genetic test results are a particularly important group, as family screening may be informative for understanding the clinical implications of a VUS. These patients are also an important area of future research.

Another important strength of our research was the use of participants who had been involved in population-based genetic testing versus individuals completely unfamiliar with genetic testing and may not have thought about communicating such information to their relatives. The mixed sample of P/LP and B/LB was chosen due to a saturated invitation pool among P/LP samples and a desire to balance these strata. We observed very little differences in themes between the two groups, but did note that the subtheme around informing younger relatives in the stages of family planning was exclusive to the P/LP participants and likely reflects the significant amount of time they've had to contemplate the generational impact of their specific genetic test results[44,48]. By stratifying our analyses, we were able to focus on the perspectives of those who had lived through relaying their genetic test results to at-risk family members. The lived experience of those who had themselves been faced with communicating their test results to family members was possibly reflected in their sense of responsibility to inform, having dealt with the task before.

Based on our data, clinic-provided mobile apps would likely be a supplemental tool rather than replace more traditional methods. Downloading and using an app does not appear to present a barrier in and may help patients fulfill their obligation to inform at-risk relatives, particularly among relatives who are in the stages of family planning and those who have difficult relationships. Mobile apps should be considered as a possible tool to realize the potential of cascade screening programs.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Funding

Research in this publication was supported by the National Human Genome Research Institute and National Cancer Institute of the National Institutes of Health, grant number 5U01HG008657-05 REVISED ("The Electronic medical Records and Genomics Network, Phase III") and award number T32 CA094880 ("Cancer Prevention Training: Epidemiology, Nutrition, Genetics & Survivorship"). The Institute of Translational Health Sciences is supported by grants UL1 TR002319, KL2 TR002317, and TL1 TR002318 from the NIH National Center for Advancing Translational Sciences through the Clinical and Translational Science Awards Program (CTSA).

References

1. Bennette CS, Gallego CJ, Burke W, Jarvik GP & Veenstra DL The cost -effectiveness of returning incidental findings from next-generation genomic sequencing. Genet. Med 17, 587–595 (2015). [PubMed: 25394171]
2. Gallego CJ et al. Next-generation sequencing panels for the diagnosis of colorectal cancer and polyposis syndromes: A cost-effectiveness analysis. J. Clin. Oncol 33, 2084–2091 (2015). [PubMed: 25940718]

3. Caswell-Jin JL et al. Cascade Genetic Testing of Relatives for Hereditary Cancer Risk: Results of an Online Initiative. J. Natl. Cancer Inst 111, 95–98 (2019). [PubMed: 30239769]
4. McGivern B et al. Family communication about positive BRCA1 and BRCA2 genetic test results. Genet. Med 6, 503–9. [PubMed: 15545746]
5. Forrest LE, Delatycki MB, Skene L & Aitken M Communicating genetic information in families–a review of guidelines and position papers. Eur. J. Hum. Genet 15, 612–618 (2007). [PubMed: 17392704]
6. Sharaf RN, Myer P, Stave CD, Diamond LC & Ladabaum U Uptake of genetic testing by relatives of lynch syndrome probands: a systematic review. Clin. Gastroenterol. Hepatol 11, 1093–100 (2013). [PubMed: 23669308]
7. Li S-T et al. Factors influencing the decision to share cancer genetic results among family members: An in-depth interview study of women in an Asian setting. Psychooncology 27, 998–1004 (2018). [PubMed: 29314485]
8. McConkie-Rosell A et al. Dissemination of genetic risk information to relatives in the fragile X syndrome: Guidelines for genetic counselors. Am. J. Med. Genet 59, 426–430 (1995). [PubMed: 8585560]
9. van den Nieuwenhoff HWP, Mesters I, Gielen C & de Vries NK Family communication regarding inherited high cholesterol: why and how do patients disclose genetic risk? Soc. Sci. Med 65, 1025–37 (2007). [PubMed: 17507128]
10. Wiseman M, Dancyger C & Michie S Communicating genetic risk information with in families: a review. Fam. Cancer 9, 691–703 (2010). [PubMed: 20852947]
11. Dancyger C et al. Communicating BRCA1/2 genetic test results with in the family: A qualitative analysis. Psychol. Health 26, 1018–1035 (2011). [PubMed: 21797732]
12. Landsbergen K, Verhaak C, Kraaimaat F & Hoogerbrugge N Genetic uptake in BRCA-mutation families is related to emotional and behavioral communication characteristics of index patients. Fam. Cancer 4, 115–119 (2005). [PubMed: 15951961]
13. MacDonald DJ et al. Selection of family members for communication of cancer risk and barriers to this communication before and after genetic cancer risk assessment. Genet. Med 9, 275–282 (2007). [PubMed: 17505204]
14. Stoffel EM et al. Sharing Genetic Test Results in Lynch Syndrome: Communication With Close and Distant Relatives. Clin. Gastroenterol. Hepatol 6, 333–338 (2008). [PubMed: 18258490]
15. Kang E et al. Communication with Family Members about Positive BRCA1/2 Genetic Test Results in Korean Hereditary Breast Cancer Families. J. Genet. Med 8, 105–112 (2011).
16. Montgomery S V et al. Preparing individuals to communicate genetic test results to their relatives: report of a randomized control trial. Fam. Cancer 12, 537–46 (2013). [PubMed: 23420550]
17. Daly MB, Montgomery S, Bingler R & Ruth K Communicating genetic test results with in the family: Is it lost in translation? A survey of relatives in the randomized six-step study. Fam. Cancer 15, 697–706 (2016). [PubMed: 26897130]
18. Dilzell K, Kingham K, Ormond K & Ladabaum U Evaluating the utilization of educational materials in communicating about Lynch syndrome to at-risk relatives. Fam. Cancer 13, 381–9 (2014). [PubMed: 24770865]
19. Green RC et al. ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genet. Med 15, 565–74 (2013). [PubMed: 23788249]
20. Tong A, Sainsbury P & Craig J Consolidated criteria for reporting qualitative research (COREQ): a 32 -item checklist for interviews and focus groups. Int. J. Qual. Heal. care J. Int. Soc. Qual. Heal. Care 19, 349–57 (2007).
21. Fruhling A Assessing the Reliability, Validity and Adaptability of PSSUQ. in AMCIS Proceedings 378(2005).
22. Jujjavarapu C et al. ShareDNA: A Smartphone App to Facilitate Family Communication of Genetic Results. BMC Med. Genet
23. Bagozzi R The Legacy of the Technology Acceptance Model and a Proposal for a Paradigm Shift. J. Assoc. Inf. Syst 8, 244–254 (2007).
24. Braun V & Clarke V Using thematic analysis in psychology. Qual. Res. Psychol 3, 77–101(2006).

25. Unger S, Simond E, Davoine E, Katapodi MC & Unger S Dissemination of Genetic Information in Swiss Families with Lynch Syndrome: A Qualitative Exploratory Study. Clin. Oncol. Res 1–5 (2020) doi:10.31487/j.COR.2020.01.01.
26. Suthers GK Letting the family know: balancing ethics and effectiveness when notifying relatives about genetic testing for a familial disorder. J. Med. Genet 43, 665–670 (2006). [PubMed: 16371501]
27. Schwartz MD Identification of BRCA1 and BRCA2 Mutation Carriers Through a Traceback Framework: Consent, Privacy, and Autonomy. J. Clin. Oncol 35, 2226–2228 (2017). [PubMed: 28463631]
28. Eijzenga W et al. How to support cancer genetics counselees in informing at-risk relatives? Lessons from a randomized controlled trial. Patient Educ. Couns 101, 1611–1619 (2018). [PubMed: 29789176]
29. Wakefield CE, Homewood J, Mahmut M, Taylor A & Meiser B Usefulness of the Threatening Medical Situations Inventory in individuals considering genetic testing for cancer risk. Patient Educ. Couns 69, 29–38 (2007). [PubMed: 17706910]
30. Hodgson J et al. Outcomes of a randomised controlled trial of a complex genetic counselling intervention to improve family communication. Eur. J. Hum. Genet 24, 356–360 (2016). [PubMed: 26130486]
31. Forrest LE, Burke J, Bacic S & Amor DJ Increased genetic counseling support improves communication of genetic information in families. Genet. Med 10, 167–172 (2008). [PubMed: 18344705]
32. Hodgson JM et al. Improving family communication after a new genetic diagnosis: a randomised controlled trial of a genetic counselling intervention. BMC Med. Genet 15, 33 (2014). [PubMed: 24628824]
33. Batte B et al. Family Communication in a Population at Risk for Hypertrophic Cardiomyopathy. J. Genet. Couns 24, 336–348 (2015). [PubMed: 25304619]
34. Frey MK et al. Prospective Feasibility Trial of a Novel Strategy of Facilitated Cascade Genetic Testing Using Telephone Counseling. J. Clin. Oncol 38, 1389–1397 (2020). [PubMed: 31922918]
35. Godard B, Hurlimann T, Letendre M, Egalité N & INHERIT BRCAs. Guidelines for disclosing genetic information to family members: from development to use. Fam. Cancer 5, 103–16 (2006). [PubMed: 16528614]
36. Offit K, Groeger E, Turner S, Wadsworth EA & Weiser MA The'duty to warn' a patient's family members about hereditary disease risks. JAMA 292, 1469–73 (2004). [PubMed: 15383518]
37. Whyte S, Green A, McAllister M & Shipman H Family Communication in Inherited Cardiovascular Conditions in Ireland. J. Genet. Couns 25, 1317–1326 (2016). [PubMed: 27271705]
38. Forrest K et al. To tell or not to tell: barriers and facilitators in family communication about genetic risk. Clin. Genet 64, 317–326 (2003). [PubMed: 12974737]
39. Devitt K & Roker D The Role of Mobile Phones in Family Communication. Child. Soc 23, 189–202 (2009).
40. Istepanian RSH & Lacal JC Emerging mobile communication technologies for health: some imperative notes on m-health. in Proceedings of the 25th Annual International Conference of the IEEE Engineering in Medicine and Biology Society (IEEE Cat. No.03CH37439) 1414–1416 (IEEE). doi:10.1109/IEMBS.2003.1279581.
41. Chivers Seymour K, Addington-Hall J, Lucassen AM & Foster CL What Facilitates or Impedes Family Communication Following Genetic Testing for Cancer Risk? A Systematic Review and Meta-Synthesis of Primary Qualitative Research. J. Genet. Couns 19, 330–342 (2010). [PubMed: 20379768]
42. Gaff CL et al. Process and outcome in communication of genetic information within families: a systematic review. Eur. J. Hum. Genet 15, 999–1011 (2007). [PubMed: 17609674]
43. deGeus E et al. Development of the Informing Relatives Inventory (IRI): Assessing Index Patients' Knowledge, Motivation and Self-Efficacy Regarding the Disclosure of Hereditary Cancer Risk Information to Relatives. Int. J. Behav. Med 22, 551–560 (2015). [PubMed: 25515913]

44. McClaren BJ et al. Cascade carrier testing after a child is diagnosed with cystic fibrosis through newborn screening: investigating why most relatives do not have testing. Genet. Med 15, 533–540 (2013). [PubMed: 23348769]

45. Maramba I, Chatterjee A & Newman C Methods of usability testing in the development of eHealth applications: A scoping review. Int. J. Med. Inform 126, 95–104 (2019). [PubMed: 31029270]

46. d'Agincourt-Canning L Experiences of Genetic Risk: Disclosure and the Gendering of Responsibility. Bioethics 15, 231–247 (2001). [PubMed: 11700677]

47. Burns C, Yeates L, Semsarian C & Ingles J Evaluating a custom-designed aid to improve communication of genetic results in families with hypertrophic cardiomyopathy: study protocol for a randomised controlled trial. BMJ Open 9, e026627 (2019).

48. Graves KD et al. Communication of genetic test results to family and health-care providers following disclosure of research results. Genet. Med 16, 294–301 (2014). [PubMed: 24091800]

Highlights

- Patients feel a sense of responsibility to inform their at-risk family members
- Mobile apps could be a preferred method for younger relatives
- Mobile apps could be a supplemental tool to improve cascade testing
- Downloading and using a mobile app does not appear to present a barrier

Table 1.

Participant characteristics for mobile base app interview (total sample size=14)

P/LP Results	8 (57%)
Mean Age (Range)	69(60–93)
Men	10 (71%)
Race/Ethnicity	
White	9 (64%)
Asian	3 (21%)
Hispanic	1 (7%)
Black	1 (7%)
Cancer Dx	2 (14%)
Marital Status	
Married	11 (79%)
Single	1 (7%)
Divorced	1 (7%)
Widowed	1 (7%)

Table 2.

Major study themes, subthemes, and exemplar quotes

Theme	Subtheme	Quote(s)
Participants report a sense of obligation to share positive test results with relatives.		"It's not, but I'm the one telling them about it. I was thinking about this on the way down. It's similar to if I'm in my house on Queen Anne and maybe I'm thinking of selling my house. I have an inspector coming in. And he says, hey, you're on a - - this whole side of the hill is on a pile of sand, and there's a 20 percent chance that It's going to slide over the next five years. I mean, It's pretty dangerous. You might want to have this checked out in more depth, and you might want to shore it up. Do you tell your neighbors about that? He said the whole hillside is going to go. You know, you don't want to worry them, but you're responsible maybe for telling them." (ID-107)
	Participants expressed greater responsibility to inform younger relatives, particularly those in the stages of family planning.	"And so, you know, which -- you would be balancing all those different elements about whether to use this app -so, yes, I think I would use it with my nephew, and he has a child. So -- but I don't know the direction -- before I would alarm him or encourage it, I would like to know more so that would be on me to find that out." (ID-103)
	Participants acknowledged complexities in sharing results with relatives with whom they experienced strained relationships.	"[I would choose text message] Because the relationship that I have with my sister is a little bit strained anyway. And I feel like It's easier for her to approach a conversation with me, regardless of the subject, if It's not necessarily by phone." (ID-114)
Participants felt that the advantages of using email were similar to those of the app.		"Well, I think of all my siblings, they'll all computer literate, so that's why I would offer an app as an option. As far as cousins, I really don't know all of my cousins' level of comfort with apps. So that would be one reason to give them the option of telling them over the phone or emailing an attachment or something like that." (ID-113)
Participants felt that younger individuals would be more comfortable with an app.		"Given the way things have changed with younger people than me, yeah. In fact, a whole bunch of them of my generation and older, they're just getting so facile with all this stuff and knowledgeable and -- mostly stimulated by grandkids probably and kids and all that, social stuff, with pictures, pictures, pictures." (ID-103)
Participants felt they could use the app independently and in their own time.		"I mean, you don't ask anybody how to do the app. So most of the apps are self-explanatory. So I guess the task would be here to make this like -- make it explanatory like you would do any other app." (ID-111)