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The Patient as Genomic Data Manager – Evaluation of the PROMISE App

Lena GRIEBEL[a,1], Marc HINDERER[a], Ali AMR[b], Benjamin MEDER[b], Marc Schweig[c], Dominic Deuber[d], Christoph Egger[d], Claudia Kawohl[e], Annika Krämer[e], Isabell Flade[f], Dominique Schröder[d], Hans-Ulrich PROKOSCH[a]

[a] *Chair of Medical Informatics, Friedrich-Alexander-University Erlangen-Nürnberg, Erlangen*

[b] *Department of Medicine III, University of Heidelberg, Heidelberg*

[c] *Backes: SRT GmbH, Saarbrücken*

[d] *Chair of Applied Cryptography, Friedrich-Alexander-University Erlangen-Nürnberg, Nürnberg*

[e] *CISPA Helmholtz Center for Information Security, Saarbrücken*

[f] *CeGaT GmbH, Tübingen*

Abstract. PROMISE (Personal Medical Safe) was a German research project which aimed to provide the responsibility of genomic data to the patient via a mobile app. The patient should accept or decline study requests to use his/her genomic data via the app. In the evaluation of the app the experiences with mobile health as well as the opinion on being the genomic data manager were measured. Furthermore, the test patients were asked about their opinion and their concerns on the PROMISE app. Most of the 19 test patients were aware of the high sensibility of genomic data and thought that the PROMISE app was a suitable solution. The largest part found it good that they were the responsible data owner. However, several participants also found it important to have a permanent contact person when it comes to questions on inquiries or the app.

Keywords. Personalized medicine, genomic sequencing, patient empowerment, technology acceptance

1. Introduction

In today's healthcare there is a shift to empower the patient from being a mere recipient of medical treatments to being an active partner in the own healthcare process [1]. It has been shown that patients that are willing to actively participate potentially can achieve better health outcomes than more passive patients [2-4]. In Germany statutory health insurances are obligated to provide electronic health records for their patients starting in the year 2021; and consumer-based health apps can be prescribed by physicians to their patients in the future [6].

A big trend in current healthcare is the so-called personalized medicine which takes individual factors into account and offers therapeutic measures individually tailored for each specific patient [7]. This includes the use of genomic data to develop

1 Corresponding Author: E-mail: lena.griebel@fau.de

the best therapy options. Different institutions also access and use anonymized genomic patient data for research purposes. Until now patients however mostly have no or very limited access to their genomic data which have been collected in the clinical setting. In the German BMBF funded (German Federal Ministry of Education and Research) research project PROMISE DS (Personal Medical Safe) the patient should be an active partner in this genomic medicine assuming that he or she is indeed able and willing to. Studies have shown that patients in general want to be involved in new health technologies [8, 9] but that it is necessary to assess the patients' preferences towards engagement strategies beforehand [10].

PROMISE DS developed and provided an infrastructure and patient app, which allows encrypted storage of genomic data in the background and patient management of access to this data in the foreground. The objective of our companion study was to evaluate patients´ attitude towards and acceptance of this innovative technology. We wanted to gain insights into the potential genomic data managers' point of view and thus learn for the further development and deployment of such a new and innovative technology. The main research questions were:

1. How much experiences have the patients with apps and health apps?
2. What is the knowledge of the patient regarding genomic testing and what are the patients' concerns?
3. What do patients think of the PROMISE app?
4. What is the patients' general opinion on being the genetic data manager?

2. Methods

We selected test patients to use the PROMISE app over four months. All the test patients were cardiomyopathy patients recruited at Heidelberg university hospital. Cardiomyopathy is a disease known for its genetic predisposition. The genomes of all patients in our study have been sequenced before.

We included patients with a minimum age of 18 years with a medical indication for genetic testing and regular access to an Android-based smartphone. Patients who fulfilled the inclusion criteria were given an information sheet about the study and had to give their informed consent. The treating physician helped them to install the PROMISE app on their personal smartphone. It was made clear that the app would not offer medical advantages for the patient but the focus of the project was to learn about possible ways of designing an app with the patient as genomic data manager. Over the course of four months the patients received 12 simulated queries through the mobile application. There were no real customers; the requests have been developed by project members. The inquiries were designed to look as if they originated from hospital researchers, university researchers and commercial companies. The patient had the options 1) to accept, 2) to decline or 3) to ignore the request. Before and after the use of the PROMISE app the patients each fulfilled a questionnaire consisting out of closed questions in a pen-and-paper-approach. The questionnaires were designed on the basis of self-developed questions and already existing tools such as the questionnaire on patients' perspective on pharmacogenetics testing by Rogausch et al [11]. Supplementary to the paper-based questionnaires interviews were performed at the second time point to further assess the opinion toward genetic testing. The evaluation

was performed with 19 patients at both time points – the interviews with 11 participants.

3. Results

3.1 App Using Experience and Knowledge on Genomic Testing

We asked the participants about their current use of apps and health apps.

The largest part said that they used apps on a daily basis (14 of 19). Nevertheless, ten of 19 participants stated that they never use specific health apps, five used health apps very rarely and only each two persons stated to use health apps on a daily or weekly basis.

The test patients were asked to self-assess their knowledge regarding technical terms such as "gene", "genetics", "genome" or "pharmacogenomics". Whereas the knowledge on the terms "gene" and "genetics" was rather good, the patients assessed their knowledge on "pharmacogenomics" very low. 15 out of 19 test patients said after the test use of the PROMISE app that they are aware or very aware that genetic data are particularly sensitive; before they used the app also 13 patients agreed or fully agreed on that. In the interviews data security was as well revealed as a major concern regarding genetic testing.

Also the major part of the participants agreed or fully agreed that genetic information should only be stored when high safety precautions are met (15 agreed/fully agreed before the test use; 18 agreed/fully agreed after the test use). Most participants (14 before the test use, 18 after the test use) stated that they thought that the safety concept of PROMISE protected their privacy well or very well.

3.2 Opinion on the PROMISE App

After the test use of PROMISE most participants said that they found the (simulated) study inquiries very interesting or interesting (79%), 21% were unsure about it. None of them did find the inquiries disruptive in their daily life.

Also after testing PROMISE, 10 persons were afraid to falsely accept inquiries due to misunderstanding them, only three to falsely decline inquiries. In the interviews nearly every participant stated that the PROMISE app was a good idea. Several participants also mentioned that it was very important to have a permanent contact person to ask when it comes to difficulties or uncertainties when using the PROMISE app.

3.3 Opinion on Being the Data Manager

Most of the test persons found it good or very good (15 before and 18 after the test use) to own a safely stored copy of their own genetic data. Before the test use four persons were unsure, after the test use only one person was unsure about this question. Also in both time points most participants stated that they were satisfied or highly satisfied with being in control over the use of the genetic sequence data (13 before, 17 after).

Nevertheless, 5 resp. 4 persons said that they were afraid of being the responsible person for the genetic sequence data.

4. Discussion

Even though the participants stated that they used apps in general very frequently, almost no one used health apps frequently. The general self-assessed knowledge on genomics and related terms was relatively low. Most test persons saw genetic data as especially sensitive and in need of high protection but also found the security concept of PROMISE convincing. Most participants were interested in the simulated inquiries and found them non-disruptive. They were more afraid to falsely accept study inquires than to falsely decline study inquiries.

Owning a personal copy of the genomic data was found good or even very good by most of the participants. Also, most persons were satisfied with being in control over the use of the data. Besides the fact that the largest part of participants was not afraid of it, it has to be taken into account that several persons indeed were afraid of being the only data owner.

5. Conclusion

In our small study setting it got clear that data security plays an important role for the data-managing users. Further we might conclude that the layperson users would need support when it comes to the usage of technical/medical terms and scientific concepts in study inquiries, e.g. by a permanent contact person such as their treating physician. Also the test users were not familiar with the use of health apps. In general one might say that the users should not feel overtaxed by their responsibilities.

Although the findings of our study are not representative due to the small number of participants they nevertheless show that medical laypersons are indeed interested and willing to be the data manager of personal genetic data but that electronic services supporting them should be designed very thoroughly regarding their comprehensibility and data security and that a “real” contact person would be important.

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