Rare Diseases in Citizen Science – Preliminary Experiences in Developing a Personal Health App

Jannik SCHAAF[a,1], Andreas KHOURI[b], Thomas ZERR[b], Jörg SCHEIDT[b], Michaela NEFF[a] and Holger STORF[a]

[a] *Goethe University Frankfurt, University Hospital, Institute of Medical Informatics, Frankfurt, Germany*
[b] *Institute of Information Systems, University of Applied Sciences Hof, Hof, Germany*
ORCiD ID: Jannik Schaaf https://orcid.org/0000-0002-0058-155X

Abstract. SelEe is a German citizen science project aiming to develop a smartphone app for a patient-managed record. The goal is to study rare diseases with the support of interested citizens and people affected by rare diseases. We established a core research team, including professional researchers (leading the project) and citizens. Citizens have the opportunity to discuss the progress, make suggestions regarding the app's design and data entry and contribute to the dissemination of the project. To gather feedback and experiences from the core research team, we performed an online questionnaire regarding the topics "influence and communication", "improvements and learning effect", and "satisfaction". Finally, 9 citizens of the core research team participated. The results show that the citizens are very satisfied with the design of the app, their participation opportunities and the communication in the project.

Keywords. Rare diseases, citizen science, mobile health

1. Introduction

A disease is defined as rare, if it affects no more than 1.3 out of 2000 people [1]. Due to the low prevalence, research on rare diseases (RDs) is challenging. To close the knowledge gap, citizen science (CS) projects can be an opportunity to strengthen research [2]. SelEe 'Seltene Erkrankungen bürgerwissenschaftlich erforschen' (engl. Researching rare diseases in a citizen science approach') is a German CS research project. In the initial project phase, people affected by RDs or their relatives participated in different workshops to define research topics and questions. As a result, it was decided to develop a smartphone app to collect data from patients with RDs. The goal is to support research and provide patients with a patient-managed record as a personal overview of their data (e.g., every day symptoms or medication) [2]. Since the objective of CS is to involve citizens in every phase of a project [3], we created a core research team, which works closely with the professional researchers. This team includes researchers and software developers from the Goethe University Frankfurt and the

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[1] Corresponding Author: Dr. Jannik Schaaf, Institute of Medical Informatics, Goethe University Frankfurt, University Hospital, Theodor-Stern Kai 7, 60590 Frankfurt; E-mail: jannik.schaaf@kgu.de

University of Applied Sciences Hof, who lead the project and program the app, as well as citizens who provide feedback during the development.

However, according to Schaefer et al., most CS projects focus on the evaluation of the scientific output [4]. Questioning citizens about their experience with the project is limited in most project evaluations [2,5]. Therefore, it has been suggested to include the so called "citizen scientist dimension" as part of the evaluation [6]. This includes topics like target group alignment, degree of involvement, communication, or collaboration. We are not aware of any CS health projects with a focus on digital applications that have studied these aspects. After approximately 1.5 of 3 years of SelEe, it is important to examine these aspects in order to learn for the further course of the project. Hence, the aim of this work is to describe the core research team and to evaluate the citizen scientist dimension. To this end, we derived the research topics "influence and communication", "improvements and learning effect", and "satisfaction".

2. Methods

2.1 Definition and work of the core research team

In the initial project phase, participants were invited via different channels (see Fig.1, step 0) [2]. Interested citizens were able to register for the project via an online questionnaire. All registered persons (n = 69) were invited to three workshops to gather topics and requirements for the implementation of the app (see Fig. 1, step 1). The first workshop had 9 participants, the second 10 and the third 11 [2]. Once the requirements were identified, the core research team was established (see Fig. 1, step 2). All 69 people were invited to join the core research team. Eventually, 9 people formed the core research team and participated in 6 workshops for the app design up until September 2022.
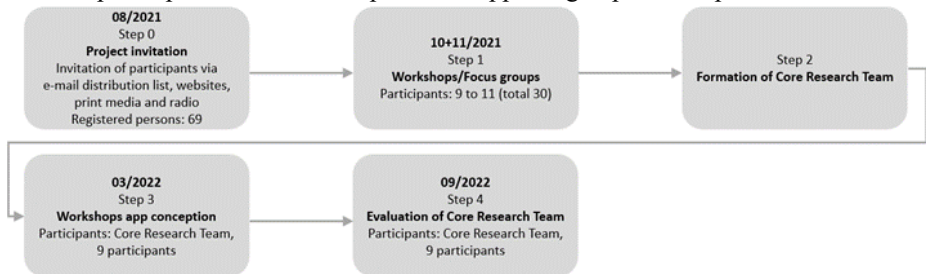

Figure 1. Citizen involvement in the SeLEe project in the first project year.

The monthly workshops were conducted via a video conference application and lasted approximately 90 minutes. Like the workshops in step 1, they served to gather the citizens' opinions about the future SelEe-app. This includes the definition of specific functionalities to design a first prototype version. The functionalities were translated into mockups by the professional researchers and discussed with the citizens. This was done iteratively until the whole core research team was satisfied. In addition, the data fields for data entry were defined by the citizens to create a basis for the patient-guided documentation. Each citizen had the opportunity to present data that they felt should be collected on an RD. Based on this, we identified commonalities in the data elements and created a basic data set that can be entered in the app and can be expanded with user-specific individual elements. Additionally, citizens of the core research team were also

involved in the dissemination of the project (e.g., content of the monthly newsletter or posters for conferences).

2.2 Evaluation

To collect the experiences from citizens in the core research team, we performed a survey with 11 questions (shown in Table 1) at the end of the conception phase (see Fig. 1, step 4). In question 1, we asked whether the citizens have an RD. We sent an invitation mail to all 9 citizens of the core research team. The survey was conducted in September 2022 using an online questionnaire in German language (invitation required). For the data analysis of the survey and the synthesis of the results, we used absolute frequencies (N). To present the results of the open questions, the statements of the participants were summarized and exemplary quotations were selected.

3. Results

All invited participants completed the questionnaire. 6 of them are affected by an RD, whereas 3 are relatives of an affected person (question 1). Full results are shown in Table 1 with sample replies to questions 4 and 11 shown below.

Table 1. Results of the survey

No.	Concept	Question	Answer option	N
2	Satisfaction	How satisfied are you with the workshops of the core research team?	(6) Extremely satisfied	4
			(5) Very satisfied	4
			(4) Somewhat satisfied	0
			(3) Somewhat dissatisfied	0
			(2) Very dissatisfied	0
			(1) Extremely dissatisfied	1
3	Satisfaction	I liked the work in the core research team.	(5) Strongly agree	4
			(4) Agree	5
			(3) Neutral	0
			(2) Disagree	0
			(1) Strongly disagree	0
4	Improvements and learning effect	What kind of cooperation within the core research team would you suggest?	See section below	
5	Satisfaction	How satisfied are you with the course of the project so far, especially with the opportunities for participation?	(6) Extremely satisfied	5
			(5) Very satisfied	3
			(4) Somewhat satisfied	0
			(3) Somewhat dissatisfied	0
			(2) Very dissatisfied	1
			(1) Extremely dissatisfied	0
6	Influence and communication	How would you rate your personal influence on the implementation of the digital application?	(6) Extremely satisfied	3
			(5) Very satisfied	4
			(4) Somewhat satisfied	1
			(3) Somewhat dissatisfied	0
			(2) Very dissatisfied	0
			(1) Extremely dissatisfied	1

No.	Concept	Question	Answer option	N
7	Satisfaction	How satisfied are you with the first conception of the app?	(6) Extremely satisfied	5
			(5) Very satisfied	4
			(4) Somewhat satisfied	0
			(3) Somewhat dissatisfied	0
			(2) Very dissatisfied	0
			(1) Extremely dissatisfied	0
8	Influence and communication	How transparent do you feel our communication processes are?	(5) Very high transparency	2
			(4) High transparency	7
			(3) Medium transparency	0
			(2) Low transparency	0
			(1) No transparency	0
9	Influence and communication	How satisfied are you with the communication and summary of the respective workshops within the core research team?	(6) Extremely satisfied	4
			(5) Very satisfied	3
			(4) Somewhat satisfied	1
			(3) Somewhat dissatisfied	1
			(2) Very dissatisfied	0
			(1) Extremely dissatisfied	0
10	Improvements and learning effect	How do you assess your own learning effect in the project so far?	(6) Excellent	0
			(5) Very good	5
			(4) Good	4
			(3) Not good	0
			(2) Not good at all	0
			(1) Bad	0
11	Improvements and learning effect	Do you have any comments or suggestions for improvement?	See section below	

In question 4, the participants gave replies such as "The core research team is great" or "The interaction is enormously important for this work". Another particpant stated: "It would be even better if we had met in person once".

In question 11, participants gave suggestions for improvement of the participation. One participant stated: "All suggestions were readily taken up. I did not expect there to be so many opportunities to participate in the project". Another participant stated: "The suggestions were implemented well as far as they were possible".

4. Discussion

The motivation of this publication was to describe the work of the core research team in the SelEe project and to evaluate the citizen scientist dimension in this group.

The results show that citizens in the core research team are very satisfied with their participation opportunities as well as with the first conception of the app. These results are also reflected in the questions about influence, communication and learning effect. Even though there were some negative ratings, e.g. in question 6, no negative comments in question 11 were provided. In summary, our study indicates that a core research team working closely with the researchers, represents a good possibility for the realization of CS projects. However, it remains unclear why only 9 of 69 interested persons wanted to be part of the core research team. Future research might therefore be valuable in identifying barriers for people to participate in this way and how to overcome them to achieve greater representation.

Similar to our project, Hayen et al. implemented a transdisciplinary research team in a RD project on cystic fibrosis. The authors concluded that patient experience should be taken into account at every stage of a project but requires a significant effort. However, the additional effort is only justified if the patient's expertise is a major benefit for the project [6].

Regarding the methodology, we are not aware of any standards to measure the citizen science dimension. Therefore, we have created our own questionnaire in accordance with Kieslinger et al. [7]. Similar to our study, Lelie et al. evaluated a workplace health program in a CS project with topics like "communication", "participation" and "satisfaction" [8]. At the end of the SelEe project, we will continue our evaluation to gain further insights about the core research team.

5. Conclusions

We described the work of a core research team in a CS project, which has the goal to develop a smartphone app in the field of RDs. The survey with citizens of the core research team showed promising results in the categories satisfaction, influence and communication, and improvements and learning effect. In summary, the results show that forming a core research team can achieve a high level of satisfaction in CS.

Acknowledgments

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