PROCEEDINGS OF SCIENCE

SelEe- Rare diseases citizen science research

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The citizen science project SelEe ('Researching rare diseases in a citizen science approach') aims to research rare diseases in collaboration with citizens by using digital applications. SelEe adopts a participatory approach as it is important to recognise the active role of patients and their families as informed partners. They are often the experts on their own disease. Similarly, involving citizens beyond those affected can increase the visibility and understanding of rare diseases. Citizens can contribute their strengths, engage socially and learn new knowledge about digital apps, the diseases and their research. Engaging citizens in a continuous exchange of information as well as developing digital applications for research can make research more accessible to citizens while strengthening research and its relevance.

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1. Introduction

In the European Union, a disease is declared rare if no more than 5 in 10,000 people are affected [1,2]. Currently, there are more than 6000 known rare diseases (RDs), 80% with genetic origin [3,4]. Low prevalence, complex symptomatology, limited expertise and limited availability of health services, require special efforts to obtain a specific diagnosis and initiate appropriate treatment [5,6]. There is an increase in late, incorrect or unclear diagnoses, regardless of socioeconomic status or gender/age [7]. Therefore, research and patient care in RDs need to be improved by closing research gaps including a spatially more widely distributed set of affected persons [8], including them as informed and active patients [9]. As affected people often have great expertise regarding their disease and are organized in patient organizations, the citizen science approach seems promising. The use of modern technic, e.g. in means of communication like social networks, but also dedicated web platforms and smartphone apps - allows superregional projects to reach the essential number of people affected by the disease [10].

The project SelEe ('Researching rare diseases in a citizen science approach') is a citizen science project on RDs in Germany by the Institute for Information Systems at Hof University of Applied Sciences and the Institute of Medical Informatics at Goethe University Frankfurt. It is supported by the Alliance of Chronic Rare Diseases, representing the non-profit alliance of RD patient organizations in Germany [11]. In SelEe, citizens, especially patients, are involved in the project and determine the research questions and topics of the project from the beginning to support the data processing and provision of the research results in the further course of the project.

In this publication, the SelEe project is presented with a focus on the objectives of the SelEe project and the methodological involvement of citizens. Furthermore, the expected results as well as the relevance and benefits for RDs and Citizen Science research are discussed.

The project was also presented on this topic at the Austrian Citizen Science Conference 2022 with a poster and a short presentation.

2. Methods

The SelEe project aims to research RDs by developing and using digital applications in cooperation with citizens. Citizens are to co-decide and co-discuss which topics are to be researched, which research questions are to be asked in the project and with which methodology these questions can be answered using a digital application.

The project is organised into different phases, which are strongly related to each other and contain iterations (see Figure 1). In the first phase "specification of the topic", which started at the beginning of the project, citizens are recruited, using different media dissemination. Furthermore, research questions and topics, which should be addressed in SelEe, are collected and discussed using quantitative and qualitative methods (focus groups, workshops and questionnaires). Citizens, especially the group of patients with RDs and their relatives, were able to describe information about their respective diseases and already formulate the first research hypotheses to be investigated.

Based on the results of this phase, the requirements for the digital application were defined by patients and their relatives in the first year of the project. They suggested and discussed which functions the digital application should have and which data should be collected.

In the subsequent implementation phase, the digital application is designed and programmed by both professionals (research associates as well as software developers) and citizens (e.g. interested students). The progress of the implementation is monitored, discussed and validated by a team that we call the "citizen research team" (9-12 teammembers). It consists of motivated citizens (predominantly patients and relatives of RDs) who want to participate in the whole

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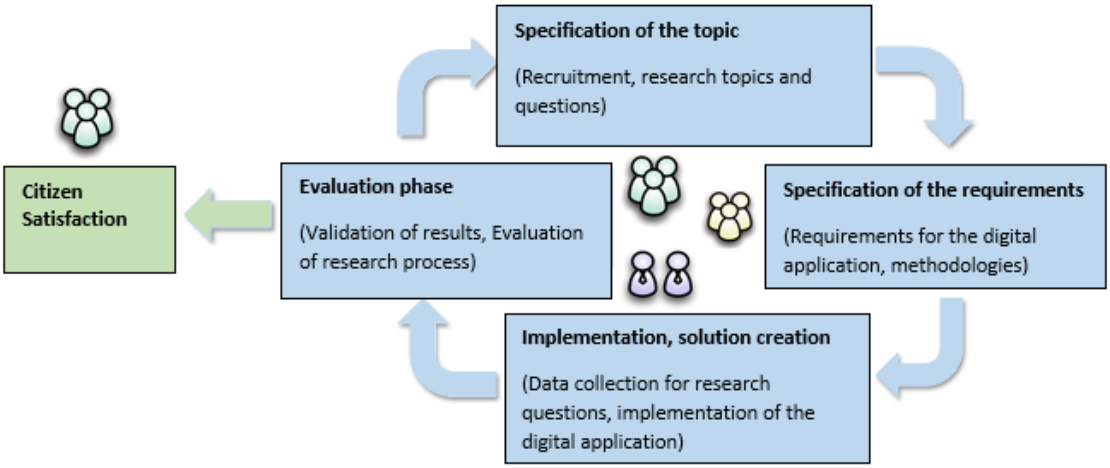
research process until the end of the project. However, permanent participation is not mandatory and can be terminated at any time.

In the following "evaluation" phase, the data collected by the digital application will be analyzed according to the research questions. The results are published on the SelEe website (www.selee.de) and made available anonymously, as well as published in international journals and presented at Citizen Science conferences. In addition, the use of the digital application will be analysed in terms of usability and acceptance. This opportunity allows us to improve the application in the future. Furthermore, the regular evaluation of the citizen research process by the citizens should be performed to reach high citizen satisfaction.

Figure 1: Research process in the SelEe project



3. Expected Results

The SelEe project aims to increase the visibility of RDs in society through citizen participation and create added value for research and those affected. Citizens can contribute their strengths, engage socially and gain new knowledge about digital apps, rare diseases (e.g. from the group of neurological diseases) and their research. A very important aspect in the field of rare diseases remains public awareness and acceptance, which SelEe aims to support. The increasing amount of research data could make it easier for people affected to be recognised in the health system. Likewise, researchers can learn directly from citizens by engaging in direct dialogue with the citizen research team, for example. Together with predominantly patients and their relatives, the project topic ‘Conception of a documentation support for a patient-managed record‘ was successfully defined in the first phase of the project, requirements for the digital application were collected and a citizen research team was established for further intensive cooperation. With the development of the digital application, a sustainable concept is to be created that can also be used after the end of the project and possibly be continued by citizens.

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