SelEe – Researching Rare Diseases through Citizen Science

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Introduction

- In the European Union, a disease is considered a rare disease (RD) if it affects no more than 5 out of 10,000 people.
- At present, there are about 6,000 rare diseases, the majority of which are genetic.
- Due to the low prevalence, the knowledge of the treating physicians is often insufficient, which leads to the fact that affected persons wait a very long time for the correct diagnosis and thus also for an appropriate treatment.
- On the other hand, those affected often know their diseases very well; they are experts on their illnesses. It therefore makes sense to use this existing expert knowledge in a citizen science project to gain new insights into the diseases.

Citizen Science

- As a citizen science project, the project is designed to involve researching citizens in all phases of the project.
- This applies in particular to the definition of the research questions to be addressed and the determination of the research methods to be used.

Project Partners

- The Institute for Information Systems is the central research facility for computer science at Hof University. The research group Analytical Information Systems has been conducting citizen science projects in the field of health research since 2011, especially around the research of seizure-like headaches such as migraine and cluster headache (3-7).
- The IMI was founded in January 2016 at the University Hospital Frankfurt. With its expertise, it acts as a mediator between medical/clinical issues and solutions from informatics. Its activities focus on applied research and development projects in various areas of medical informatics, especially in the field of rare diseases (8-11).
- ACHSE is a network of self-help organizations that exist for numerous rare diseases. Its goal is to promote knowledge about rare diseases among those affected, their relatives and medical staff.

Project Flow

- In the first phase, (online) workshops were held with people affected and representatives of various self-help organizations. They worked out which options are possible and useful for supporting those affected with digital applications.
- It turned out that most of the participants prefer a system for documenting the disease state, which creates an individual data collection that can be presented to medical professionals. In addition, the data collections of several people affected by a specific rare disease can be used to answer specific research questions.

- A citizen science core team was formed to design a digital application to be implemented, preference was given to a smartphone app for Android and iOS platforms.
- In monthly workshops, the requirements for such an app were collected and recorded in wireframes, which did then serve as the basis for programming the apps. Special emphasis is placed on the possibility of barrier-free operation of the apps, for example by blind participants.
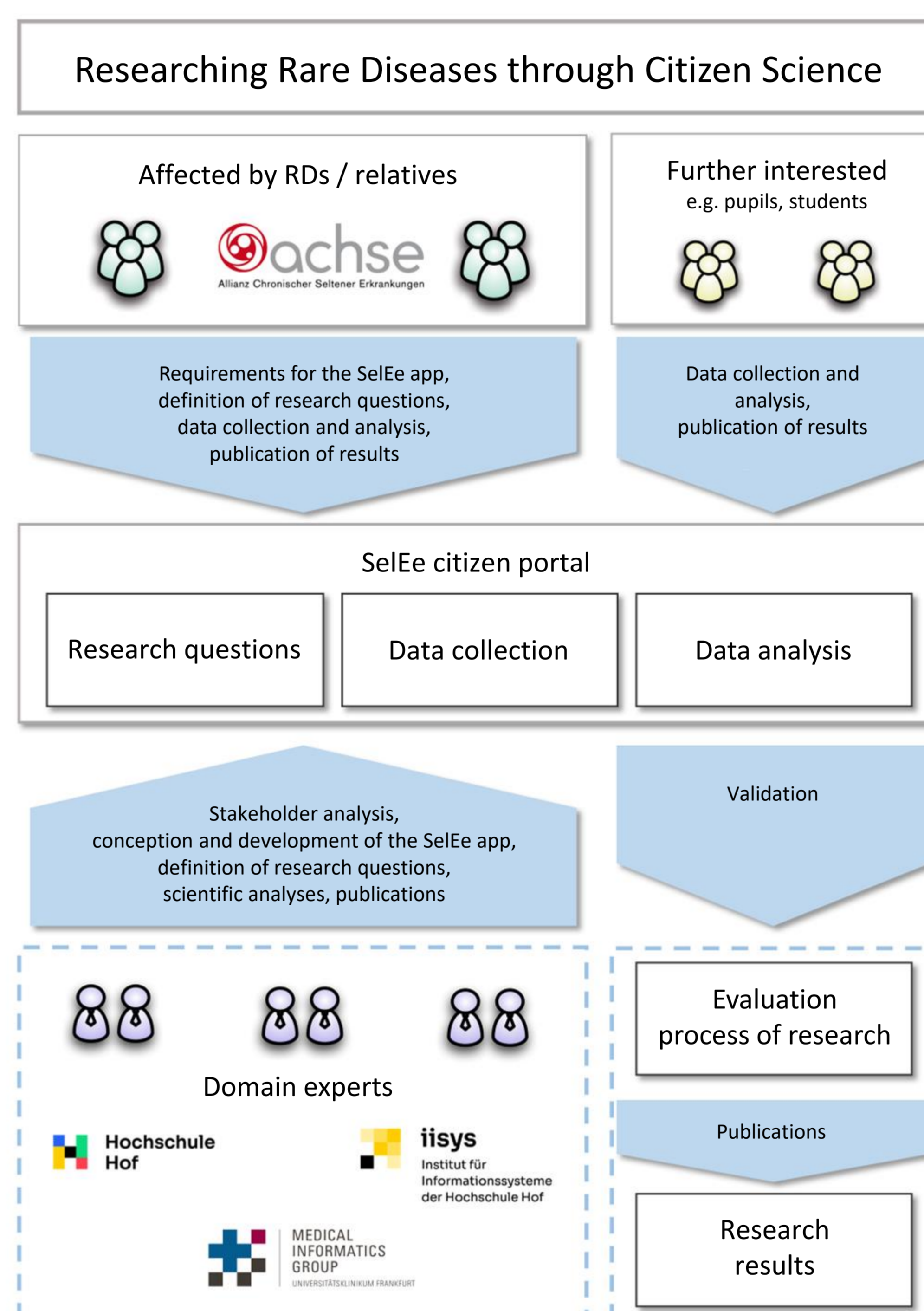

Figure 1: Overview of the planned project sequence.

- After programming and testing the apps, the data collection phase on the apps is then expected to start in the winter of 2022.
- This is followed by the evaluation of the data, where interested pupils and students as well as citizens with an affinity for research can also participate.
- The research results obtained are published in specialist journals and at specialist conferences.
- At the end of the project, the entire research process is evaluated, especially the degree of participation of the citizen researchers.
- Figure 1 shows the planned project flow in the project.

Current Status

- Currently, the programming of the apps is done and the first versions of the apps are nearly ready for testing. Besides that, workshops and discussions with the core research team take place on an ongoing basis.
- In addition, the team is working intensively on the challenges arising from the planned, barrier-free usability of the apps.

Publications

Project SelEe

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