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Vision and Challenges of a Cartographic Representation of Expert Medical Centres for Rare Diseases

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Abstract. In Germany, many highly specialized facilities for the diagnosis and treatment of rare diseases exist. However it is quite difficult for patients to find the required specialists because of the fact that information on the internet is scattered and of variable quality. The German Federal Ministry of Health initiated several activities to address this issue. This paper describes the project *se-atlas* which aims at presenting the medical care options for people with rare diseases on an interactive map and in a list format. Potential users of this resource will be patients and their relatives, doctors, non-medical personnel and the general public. Most information derived from the data sets is already listed in ORPHANET. The project's primary goals are to steadily increase the data set and to ensure its quality. This paper presents the goals and measures taken in this project. It gives an overview of the challenges implementing such a new service and the visions behind.

Keywords. Rare Diseases, Cartographic Visualization, Patient Empowerment, Patient Involvement, Patient Information.

Introduction

Diseases with a particularly low prevalence are called rare diseases. The European Union considers diseases to be rare when they affect not more than 5 per 10,000 persons. It is estimated that there are between 5,000 and 8,000 different rare diseases, affecting nearly 29 million people in the European Union and 4 million people in Germany alone [1], [2].

It is often difficult for patients and other healthcare professionals to know where to find these specialists as the diseases experts are also rare. To overcome this difficulty, the French Ministry of Health and the National Institute of Health and Medical

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Research (INSERM) jointly launched ORPHANET (www.orpha.net) in 1997, a multi-lingual information portal on rare diseases and orphan drugs. Since 2001 Germany became a partner and provides data for the international dataset.

In 2009 the Council of Ministers of the European Union recommended to the EU countries to identify rare disease expert centres and to make the information available to the end users. Recommendations on the definition of what is a centre of expertise have been published by the EU Committee of Experts on Rare Diseases [3]. Following the European Council recommendations, Germany published a National Plan for Rare Diseases (RD) in August 2013 which includes 52 policy proposals to guide and structure actions in the context of rare diseases within their health and social systems [4]. One of these proposed action fields focus on information management and has the goal to improve the level of information available to and information procurement by those afflicted, their relatives, doctors, therapists and caretakers. This action field led to the recommendation to establish a central information portal with an overview of experts in a map format which motivates the project *se-atlas* (section 3.5.3 [4]). Funded by the Federal Ministry of Health (Germany), the goal of the project *se-atlas* is to focus on an innovative representation of medical care options for people with rare diseases in Germany derived from the ORPHANET data set. In addition to the name and address of an institution, also information like the rare disease treated, individual contact data including telephone numbers, opening hours, etc. are stored. Data will be presented in both the format of an interactive map and a detailed listing. The project is intended to help completing the existing data set and to improve the accessibility by enhancing search functions and by testing new ways of interaction in order to keep the data relevant and up to date. Potential users of the information platform are patients and their relatives, practitioners, non-medical personnel and the general public.

The project *se-atlas* started in June 2013 and is scheduled to be completed by the end of May 2015. In the following we will give an overview about the goals and measures taken in this project, the intended realization and some of the challenges.

1. Goals and Measures

Based on the entered search term and the estimated or provided location of the user, relevant expert centres will be presented by the website in form of a map and a detailed listing. The user shall be able to filter the presented health care providers by different criteria such as distance, specification and quality. The goal is to provide a fast and intuitive overview about the different facilities. *se-atlas* intends to incorporate both categorization concepts described in the following and will provide a visualization of the different categories. The achievement of these goals should be supported by an early inclusion of representatives of the target group in the development process. The results have been summarized in a requirement specification document and discussed within the project partners. For the end of the project exemplary usability and acceptance tests are planned.

1.1. Quality Criteria

It is important that the listed institutions and experts fulfil defined quality criteria. ORPHANET Germany has implemented for this purpose an advisory board of experts.

Additionally a criteria checklist provided by the European Union Committee of Experts on Rare Diseases (EUCERD) has been in use since June 2013 [5].

Besides the so-called EUCERD-criteria, which are used for quality control by ORPHANET, a new German criteria catalogue, called NAMSE-criteria, has been developed differentiating between three types of centres for rare diseases, namely types A, B and C [4]. Distinctions are made among others between facilities which provide out-patient or in-patient services, have a focus on specific disease groups, and conduct or participate in research.

In addition to these official criteria new ideas based on the idea of Web2.0 shall be implemented, tested and evaluated within the project. The user shall have the opportunity to easily report if an entry or a search result is misleading or missing. Additionally, centres of rare diseases and patient organizations will be empowered to grant affirmations of quality by approving list entries in an internal area of the website.

The concept considers that the *se-atlas* map can be included as a web widget to any other webpage; for example to a page of a patient organization, which only visualizes expert centres covering one or more specific diseases. This way higher visibility of the project can be reached and the interest in keeping the data up-to-date by the corresponding community is increased.

1.2. Classifying address data and linkage to ORPHANET

To find the relevant expert centres, the address data have to be classified and linked with the corresponding diseases and their synonyms. *se-atlas* uses the ORPHANET classification to classify address data [6], [7]. The ORPHANET classification has a hierarchical structure and contains 8.945 diseases and synonyms grouped in 31 categories, e.g. *rare cardiac diseases*, *developmental anomalies during embryogenesis*, *inborn errors of metabolism*, r*are gastroenterological diseases*, *rare neurological diseases*, etc. [8]. Additionally, further classifications and listings such as the German modification of ICD10 and the German translation of MeSH will be integrated for the classifying of the centres for the operating medical experts. A project run by the German Institute of Medical Documentation and Information (DIMDI) currently maps the alpha-ID of the ICD10GM to the ORPHANET classification, so that the ICD10GM codes could be considered for classification in an easier way in future. The linkage of medical expert centres to multiple diseases is very complex and time consuming. *se-atlas* will offer new tools to support this process technically.

The basis for the underlying data set of the eligible facilities will be provided by the project partner ORPHANET Germany. The project's primary goals are to consistently increase the data set and to ensure its quality. If address data or the linkage to the corresponding diseases changes in one system, these changes will be adapted in the other database. Full correspondence between these two services is intended.

2. Realization and Challenges

The website is realized by using common technologies like JSP/JSF, JavaScript, HTML5, and CSS. The underlying database is MySQL on a Tomcat Server. First functionalities are implemented as a prototype already. For the cartographic visualization Google Maps, Nokia Here and OpenStreetMap (OSM) [9] have been tested. The quality of the data provided by OSM for Germany is very high and without charge, so

the decision was made to use OSM in *se-atlas*. Coming along with this decision is that an own OSM-server has to be hosted and a higher effort for familiarization is necessary.

One important challenge is to classify the registered specialists with their forte. The basis for the underlying data set of the eligible facilities as well as the classification of rare diseases will be provided by the project partner ORPHANET Germany (see section 1.2). For better understanding and research in the classification a java-based browsing tool was developed for internal use.

The number of diseases per category varies considerably. The category *rare genetic diseases* contains the most diseases, *rare intoxications* the fewest. Some diseases of the ORPHANET classification cannot be allocated to a distinct parent disease but fit different categories. For better search results, in consultation with ORPHANET International, a functional classification is provided, which contains a subset of the original classification and which links those diseases to their main categories.

In discussion with clinical experts and representatives of the users, scenarios and mock-ups describing the overall functionality of the website have been created. Figure 1 shows the mock-up for the cartographic view of the search results for the rare *Gaucher disease*. The different building symbols at the map represent the different categories (see figure 1). On the right side a short overview of the results in a list format is provided. The results can be limited to a maximum value of distance, which can be calculated with the help of the Geolocation API [9] or a minimum level of specificity. The specificity is calculated based on the relation between the diseases typed in and the assigned diseases.

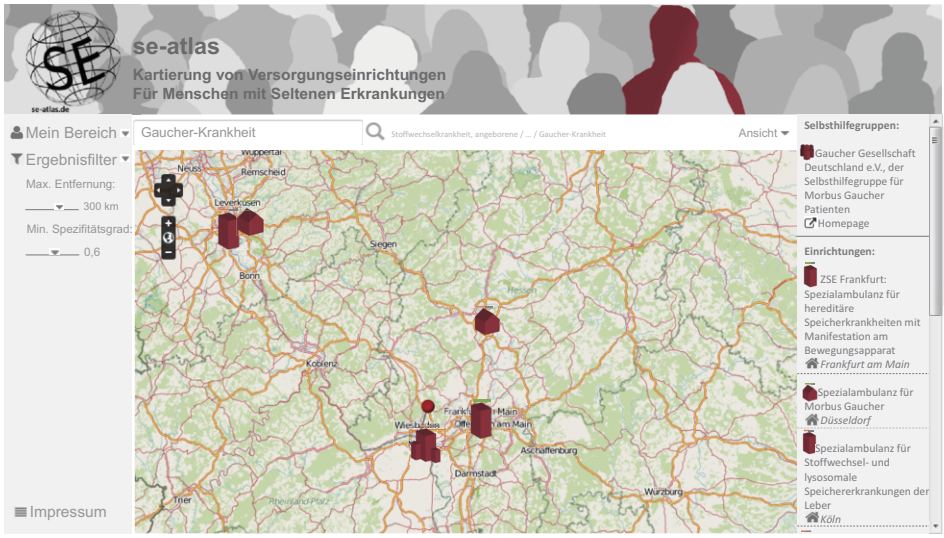

Figure 1. Mock-up of the website, not the final design.

Because names of diseases are often difficult to spell and not very common, errors like interchanges of letters or left outs should be covered by the search engine. For example for the disease *ichthyosis*, also the entry *ichtyosis*, *ichthoysis*, or *ichtiosis* should be possible. For this example, algorithms like Soundex, (Double-) metaphone, Kölner Phonetic, Levenshtein-Distance, or N-gramme (Dice-coefficient) have been tested and analysed. Based on terms extracted from the different classifications and listings (ORPHANET classification, ICD10GM, and MeSH) using autocomplete in combi-

nation with these algorithms, we obtained useful results in form of dropdown listings. A combination of Levenshtein and Kölner Phonetic seemed most promising. Even with over 20,000 terms in our database, feasibility und usability were satisfying.

3. Conclusion and Outlook

In the previous sections, we described the actual status and the emerging challenges of the project *se-atlas*. For this project it is very important that a consequent exchange of experiences between the interdisciplinary partners takes place. New concepts, for example representing data or including patient feedback are valuable contributions from other project partners. A close cooperation within the consortium will be necessary, but more importantly a conclusion as to who will operate the system after the funding period will need to be made.

In general, there are already several options for expansion of the system. The technical concepts and quality assurance strategies developed could be transferred to other European countries.

The next steps in the project *se-atlas* will be to complete the first running prototype of the system in spring 2014. Access will initially be restricted to a limited number of test persons and the available number of searchable expert centres will be small in this phase. In parallel the process of generating new data and the updating of existing data will start with the help of the partner organizations.

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