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The ERN-LUNG Population Registry: Aims, Software-Implementation and First Results

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Abstract. The ERN-LUNG Population Registry is a new European-wide collection of patients with rare lung diseases, allowing patients to register online in the registry. Medical experts can recruit patients in the registry for disease-specific registries and care options. The Population Registry was implemented on the basis of the open source software OSSE and extended by functions for the self-registration of patients. Patients were invited through patient organizations between May and November 2022. 115 patients registered online in the registry, whereas 60 of them provided full data in the registry form. After first months of usage, further dissemination of the registry is necessary to reach more patients, e.g. by recruiting them via medical centres directly. Improvements of the registry should be conducted to achieve a higher number of fully completed forms.

Keywords. Rare Diseases, Registries, Patient Registry

1. Introduction

With 400 million people affected worldwide, rare diseases (RDs) are not an isolated phenomenon. Much more, RDs are often complex or multimorbide, with a significant negative impact on life expectancy and quality of life [1]. With approximately 8000 different RDs existing and a prevalence of less than 1.3 in 2000 affected people, research and care of these patients is challenging [2]. Therefore, the European Commission funds so called European Reference Networks (ERNs) to improve healthcare and research in RDs. ERNs provide researchers and clinicians a network to share their knowledge and experiences about RDs, as well as providing patient's diagnosis, care and treatments. Currently 24 networks for different rare and complex diseases or disease groups exist. One of these networks is the European Reference Network for Respiratory diseases (ERN-LUNG), which focuses on a number of rare lung diseases (RLD) affecting the lungs and airways [3, 4].

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One goal of the ERNs is to develop and provide registries and databases. Registries are core elements for epidemiologic, public health or clinical studies and collect information on individual patients [4]. Due to the small number of patients in RDs, it is even more necessary than in common diseases to collect and share patient data across the national level [5]. Therefore, ERN-LUNG develops the Population Registry, which is a software-platform where patients can self-register and enter their data in the system. The goal is to reach as many patients as possible affected by different RLDs in Europe.

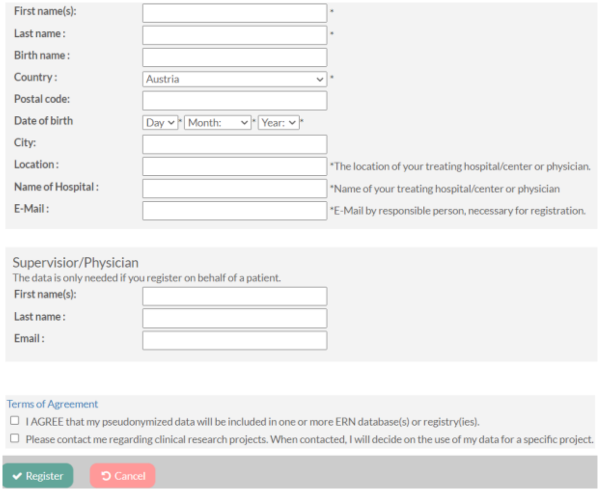
In this manuscript we present the aim and software-implementation of the Population Registry, as well as results after first months of usage.

2. Methods

2.1 Aim of the registry and software-implementation

The aim of the Population Registry is to provide patients with RLDs access to research and care by registering themselves in the registry. These patient-centred European-wide collection about RLDs is intended to strengthen research and to improve international cooperation between experts. Physicians/researchers, who are members of ERN-LUNG, can access the registry. These experts are specialized in one RLD and run a disease specific registry. In the Population Registry, experts can view data from a defined subset of patients regarding a specific disease, as well as inform and recruit them for possible studies in their own disease specific registry or possible care options. A key objective here is that patients can enter data online via a website, while ensuring data protection.

The Population Registry is based on the Open Source Registry System for Rare Diseases (OSSE) [5]. OSSE provides a web-based electronic data capture system to manage users and permission, create and fill forms, as well as validate entries. Moreover, it supports to fill patient reported outcomes (PROs), which allow patients to enter their data on their own. To implement the Population Registry, we extended OSSE with its functionality and necessary features regarding self-registration and users notifications. Patients or their relatives can register themselves on the website (www.popreg-ern-lung.eu). During the registration, various information are inquired (see Figure 1).


First name(s): *
Last name : *
Birth name :
Country : Austria *
Postal code:
Date of birth Day * Month: * Year: *
City:
Location : *The location of your treating hospital/center or physician.
Name of Hospital : *Name of your treating hospital/center or physician
E-Mail : *E-Mail by responsible person, necessary for registration.

Supervisior/Physician
The data is only needed if you register on behalf of a patient.
First name(s):
Last name :
Email :

Terms of Agreement
☐ I AGREE that my pseudonymized data will be included in one or more ERN database(s) or registry(ies).
☐ Please contact me regarding clinical research projects. When contacted, I will decide on the use of my data for a specific project.

Figure 1. Registration form in the Population Registry

After providing this information, the consents need to be agreed. Afterwards, the patient account is checked and activated by an administrator within 48 hours to prevent unsolicited accounts. Then, the user receives an activation link via email to initialize his account, set a password and fill the PRO. The PRO contains various details about the patient such as symptoms and their first occurrence, diagnosis, year of diagnosis etc. As soon as patients has entered their data, experts of ERN-LUNG are informed automatically via email that a new patient with a certain disease registered. They can login to the registry and view and contact patients with customizable e-mail messages.

2.2 Patient recruitment, data collection and analysis

All patients of all ages with any kind of RLD were able to register in the Population Registry. The registry was launched online on 28th of February 2021. For recruitment, patient organizations of RLDs were contacted via email with an online flyer attached. They were asked to share the information about the Population Registry with their members. The patient organisations were identified in a previous online search for each EU member state according to the nine disease groups of ERN-LUNG [6]. Afterwards, 72 patient organizations were contacted via e-mail in May, August and November 2021. In addition, media-dissemination was performed via social and print media.

In the following, we show information about total and complete registrations, patient origin and diagnosis, gender and age, as preliminary results of the patient registry.

3. Results

A total of 115 patients from 15 different countries registered in the Population Registry. Most patients were from United Kingdom (n=45), Germany (n=22), Austria (n=15), Spain (n=13), France (n=9) and others (n=11), as shown in Figure 2.

Figure 2. Distribution of patients in Europe

78 of the patients activated their account, with only 60 of those have entered their data completely and therefore provided full information. Most of these patients are suffering from sarcoidosis (n=29), interstitial lung diseases (n=13), primary ciliary dyskinesia

(n=9), pulmonary hypertension (n=6), non-CF bronchiectasis (n=2) and cystic fibrosis (n=1). The patients are female (n=31) or male (n=29). The average age is 51 years.

4. Discussion

In this work, we presented the aims, implementation and first results of the Population Registry, which enables the registration of patients with RLDs by their own. Compared to other registries for RDs, the registry is not a disease-specific collection of data on a disease to answer a specific medical question, but it is intended to create the data basis for networking patients with RLDs and the associated experts throughout Europe [7].

In total, 115 patients registered in the registry. It is noticeable that patient accounts are available, who have not entered any data or completed their registration. Further investigations are necessary to determine whether the registration is too complex for patients and to identify potential barriers [8]. Furthermore, it should be noted that a large number of patients could not be reached by pure online recruitment. The recruitment should be extended to medical centres and hospitals with the involvement of physicians, since not every patient is involved in a patient organization. Then the success of such a registry can be determined, above all, if there is an acceptance by the patients. Moreover, further studies are needed whether such kind of registries effect public health in general, e.g. in providing health information or giving therapy pathways.

5. Conclusions

In this publication, we presented the ERN-LUNG Population Registry, allowing RLDs patients to self-register and experts to recruit them for disease-specific registries and core options. Further work is needed to expand recruitment and improve usability.

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