Online Population-Based Patient Registry
to Collect and Share Health-Related Data of Rare Disease Patients

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Abstract

Rare diseases, those affecting few patients, are in fact *neglected diseases* that demand special concern and care. We are constructing an online system to collect health-related data and complaints from rare-disease patients to share that information with stakeholders, thereby facilitating the development of new or improved drugs, treatments, and policies and enhancing the quality of life (QOL) for patients with such diseases. The system will also track and monitor the physical states and conditions of patients, which will facilitate the management of their QOL and improve communication with health professionals.

The number of cancer patients across the world in 2008 was reported as 12.7 million [1]. Also worldwide, some diseases affect fewer than 1,000 patients. These are called *rare diseases*.

Drugs for rare diseases are called *orphan drugs* because they demand special concern and care to be brought into the market. Why is such special treatment required? Development of an orphan drug is sometimes not promoted, even though the seeds of the drug exist, because of commercial reasons. The low number of disease patients dictates a small drug market, which might be judged as financially insufficient to support development of an orphan drug. Therefore, many national governments have programs to promote the development of orphan drugs, which are required by laws or regulations (e.g., Orphan Drug Act in the US, Pharmaceutical Affairs Act, Article 77-2 in Japan, and EC Regulation No 141/2000 in the EU).

Promotion of such drugs is apparently necessary not only in industry but also in academia. Many researchers in various fields have undertaken cancer research, but have neglected rare disease research. Researchers being active in an extensive field might have many chances to publish papers and to obtain research grants and academic posts, and enhance their own reputations. Plenty of information related to major diseases can be accumulated at one facility. In contrast, rare disease patients are sparsely distributed, so physicians and researchers would lack sufficient information to promote studies of the disease. Putting together scattered information related to a specific rare disease can be expected to spur studies to unravel the causes of such diseases and to establish treatments.

Regarding the problem differently, almost all patients with rare diseases cannot be permanently cured at this time, which requires that patients live with chronic diseases during daily activities. For progressive diseases (e.g. nervous disorders), performing self-tracking of physical states would help to estimate prognosis and to manage important choices related to issues such as treatments. In the case of non-progressive diseases with frequent exacerbations and remissions (e.g. immune system diseases), self-monitoring of daily physical conditions would better support health maintenance.

As one methodology to overcome these stresses, creating a system to gather, archive, and share data and information related to rare diseases and their patients can be expected to have important results. Collected data can encourage new entry into rare disease research and orphan drug development. Data might also be useful as part of a personal health record (PHR) system for patients.

Patient registry

A framework to collect patient records is called the *patient registry* (or *patient register*). Although such a registry has traditionally been a pile of papers on a physician's desk at a clinic or hospital, it can constitute a collection of data and information for promoting studies, developing drugs, designing policies, and operating PHR for a certain disease.
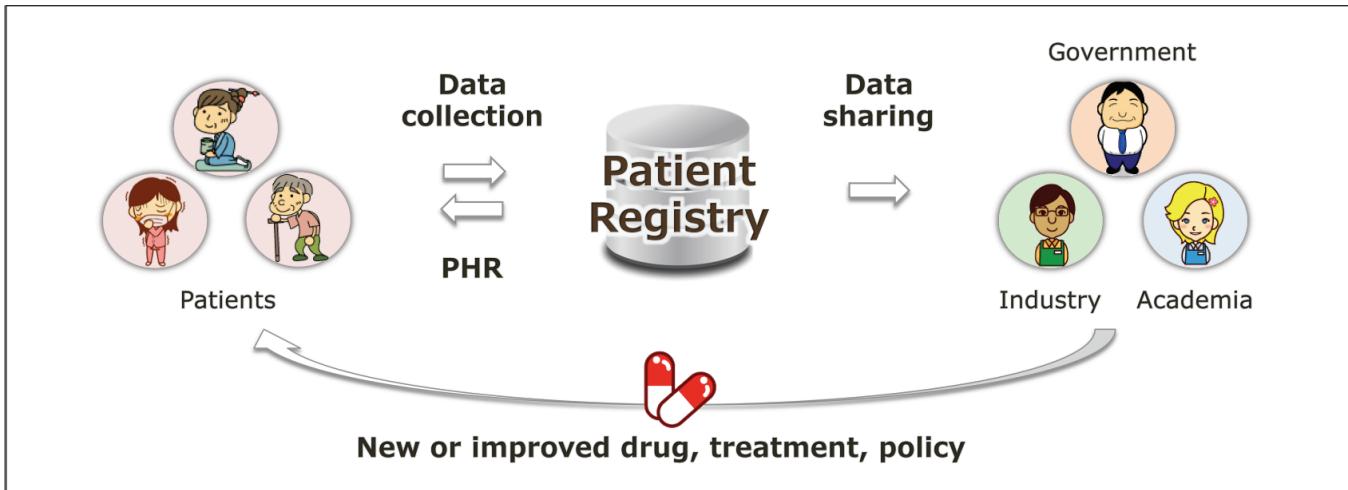
Fig. 1 Schematic illustration of our patient registry.

A new record is added to the patient registry by the patient, a family member of the patient (especially when the patient is a child or has communication difficulty), or an attending physician of the patient.

Types of patient registries

Roughly two kinds of patient registry exist: those established by researchers in academia and those developed by patient groups. Most existing patient registries are of the former type [2]. Patient registries established in academia collect clinically more precise and less noisy data related to patient conditions, although they are likely to be limited to clinical data. Patient registries established by patient groups collect data that are clinically less important but more closely connected with patients' daily life. True endpoints associated with medical treatment are not merely getting improved laboratory test results, but giving patients a better life. Therefore, collecting data and information related to effects of a disease on a patient's daily life is truly important.

We can also collect patient data from blogs, microblogs, and social networking services. Online community services specifically targeting disease patients exist already on the internet. Although such resources have great potential to gather valuable data, they do include a large quantity of noise. Especially, it is not a trivial task to confirm whether a person who posts is indeed a patient or not: even the person often does not know or believe it without correct diagnosis.

Our challenge

We, a team consisting of a patient umbrella organization (JPA; Japanese Patients Association) and researchers in academia, are striving to construct an online population-based patient registry for rare disease patients in Japan.

JPA, established in 1986 (initially as JPC; Japan Patients Council), is now the largest and the most influential patient organization for rare diseases in Japan.

We have been evaluating a test version of the registry, devoting attention to the following four criteria:

• Credibility of registered patient information

• Incentives for patients to register

• Sustainability of the patient registry

• Collaboration with patient registries overseas

To fulfill these criteria, various techniques must be combined from broad fields such as natural language processing (NLP), human–computer interface (HCI), lifelogging, self-quantification, and gamification. We will kick off this patient registry by providing important information related to four rare diseases.

Our ultimate goal is to improve the quality of life (QOL) of rare disease patients by supporting rare disease studies, drug development and marketing, patient self-care, patient–physician communication, and policy planning.

Acknowledgements

References

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