



A New Challenge for Sanoia.com: Orphan Diseases

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Abstract: The European Organization for Rare Diseases (EURORDIS) has listed yet up to 7,000 distinct rare diseases affecting more than 5% of the European population. Many patients suffering these rare diseases have first experienced a lack of information about both the disease itself and about where to obtain help from qualified professionals or online resources. Nowadays, on the contrary, concerns about security, privacy and ubiquitous access have emerged and represent now true limitations for on-line use of personal data. To address this situation, AIMSU and La Conception Hospital Internal Medicine department have decided to collaborate and provide a dedicated SANOIA.COM access to patients with a rare disease: Immune Thrombocytopenic Purpura (ITP). This system should allow patients to have a permanent access to their important medical data and to specific online resources, but also to self-report about symptoms, medical analysis or environmental factors. Ultimately, data-mining could offer a non-biased approach to discover unanticipated correlations and help redefine the definition of these rare diseases from patients' point of view.

Introduction

Orphan diseases, also referred as rare diseases, affect a small percentage of the population (less than 50 for 100 000). However, the European Organization for Rare Diseases (EURORDIS) has listed yet up to 7,000 distinct rare diseases affecting more than 5% of the European population [1]. These patients have first experienced a lack of information about both the disease itself and about where to obtain help from qualified professionals. To address this situation, several actions have been conducted like the implementation in 1997 of the online information server Orphanet [2], and recently, the development of online communities for rare diseases by EURORDIS [1]. Aiming to develop new ways to use the Web 2.0 resources in order to improve the daily lives of ITP patients, we hypothesized that SANOIA might be very useful by many aspects for patients living with this very rare disease. Indeed, SANOIA has demonstrated at the MED-E-TEL 2010 edition its relevance based on patient driven medical information, anonymity and ubiquitously accessible through a unique and secured number [3], and would allow them to directly and concretely participate to it via self-reporting about symptoms, medical analysis but also the influence of environmental factors (food, toxics, ...).

The ITP experimentation: aims and evaluations

We aim to know how the use of a version of SANOIA dedicated to ITP impact the health related quality of life (HRQoL) of patients with ITP. Sixty consecutive ITP patients will be enrolled from the department of Internal Medicine (Universitary Hospital la Conception, Marseille, France). Demographic and medical data of these patients will be recorded at inclusion. The study received local Institutional Review Board approval and each patient will provide written informed consent before enrolling. Patients will have to meet the following inclusion criteria: primary autoimmune thrombocytopenia according to American society guidelines; age between 18 and 65 years; permanent web access. Exclusion criteria include neuropsychiatric severe conditions and/or language problems. These patients will be randomized in 3 different groups: a control group (patients without SANOIA interface), and two other groups of patients having SANOIA, respectively with or without medico-technic hotline (phone and email). HRQoL will be assessed using the ITP-PAQ, that consists of 44 questions organized into 10 scales measuring Physical Health (Symptoms, Bother, Fatigue and Activity Scales), Emotional Health (Fear and

Psychological Health Scales), Work, Social Activity, Women's Reproductive Health (which includes menstrual symptoms and fertility) and Overall QoL [4]. These 44 questions are designed to quantify feelings, such as fear of bleeding or hospitalization, both due to ITP symptoms and limitations on physical, social or work function due to ITP symptoms or treatment side effects. Items are scored on Likert-type Scales of various sizes. ITP-PAQ scores will be evaluated for all patients at baseline, month 1, month 3 and month 6. Change in ITP-PAQ scores from baseline to month 6 is the primary endpoint. Additional considerations about the "real life" usefulness of SANOIA during medical or paramedical consultations will also be assessed, as well as the access via SANOIA to specific resources available for patients and their caregivers (ie, link to the emergency therapeutic protocol for ITP, open self-reporting).

A new way of improving medical knowledge about orphan diseases

We imagine the generation of a large and evolving database including symptoms, medical events and treatments, intercurrents events, environmental parameters self-reported by patients diagnosed with one or more of the 7000 orphan diseases. The self-reporting will be possible because of the wide implementation of an informatic interface (web and/or mobile access) all across Europe countries (diffused with the help of associations of patients, medical community and general medias), and will be performed by voluntary patients, already diagnosed for one of these diseases. Outputs from this first-round analysis should be shared with patients, medical community but also epidemiologic or environmental disciplines to determine parameters that are relevant enough to be implemented in the ongoing self-reporting (Fig. 1)

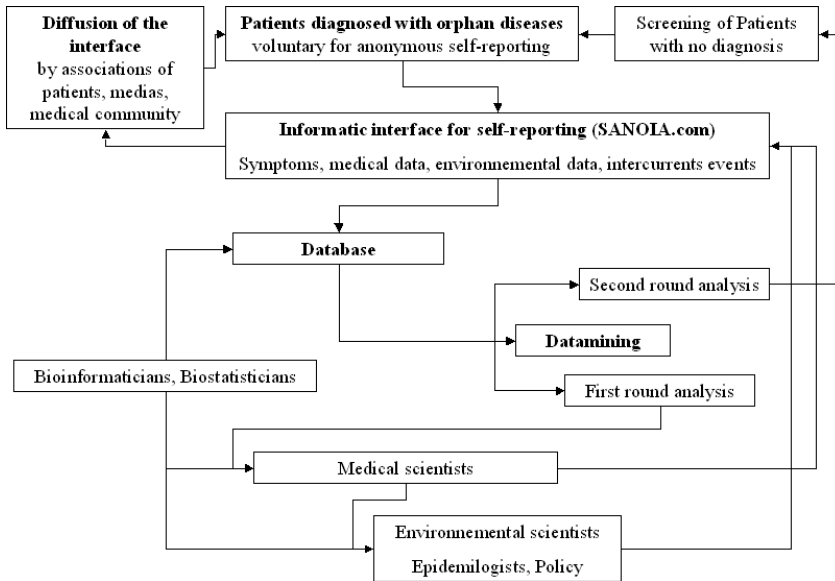


Figure 1: Patients' data driven research.

Conclusion

Data-mining could offer a non-biased (non-hypothesis driven) approach to discover unanticipated correlations and help redefine the definition (or “signature”) of these rare diseases from patients point of view. If the ITP experimentation proves successful, we aim to diffuse this interface to many other orphan diseases. As orphan drugs meet even more needs in pharmacovigilance than more common medications [5], we could design modules to help to patient to directly report to the European agencies and/or to be alerted immediately about new side-effects of their ongoing or past medications. Finally, because they are naive from standard medical knowledge, “expert” of their own disease with often a strong motivation to improve its knowledge, empowerment of patients with orphan diseases might provide a unique new source of large scale scientific data.

Acknowledgment

Laurent Chiche thanks Dominique Donnet-Kamel (INSERM, Mission Associations), Viviane Willis-Mazzichi (European Commission, Scientific Culture and Gender Issues), and Afonso Ferreira (CNRS, INS2I, Institute for Computer Sciences) for their valuable advice and encouragement. Sponsor : Orange Healthcare (co-development of a mobile access version).

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