eHealth Services for the European Reference Network on Rare Anaemias (eENERCA)

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Abstract. This paper presents an electronic registry system for the purposes of the eENERCA for rare congenital conditions that require lifelong follow up and treatment. The main objective of the eENERCA project focuses on the prevention of major rare anaemias (RAs) by facilitating the access, at a European level, to the best genetic counselling, diagnosis and clinical management of the patients with RA independently of their country of origin. This can be achieved by promoting an extension of the full Electronic Health Record system and specifically the electronic registries for RAs, across Europe for the purposes stated hence promoting service development for the benefit of patients. The proposed eRegistry will serve as an epidemiological tool to improve the management of patient services and ultimately improve patient care.

Keywords. Rare anaemias, eRegistry, eHealth services, Electronic Health Record

Introduction

Rare congenital conditions require lifelong follow up and treatment. This is particularly so for the congenital anaemias which over time, due to complications of the disease but also of the treatment, become multi-organ disorders, requiring the involvement of several medical and paramedical specialties [1,2]. The need for complex and expert services and the promotion of such services is the reason why the ENERCA\textsuperscript{2} project was initiated along with other European initiatives for rare disease both general and disease specific. Patients and healthcare providers will benefit from this Registry, since they can be provided with valuable sources of information on their disease. The development of services however cannot progress without epidemiological information being made available to health authorities. In the case of rare diseases this is particularly important since medical consultation and expertise is very difficult to be available in all health units that need to treat rare conditions of patients. The proposed eRegistry will serve as an epidemiological tool to improve the man-

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agement of patient services and ultimately improve patient care by storing patient numbers and location, diagnosis and proof of diagnosis and the treatments offered. Thus, a physician will have the ability to monitor a patient. Likewise, the eRegistry will provide some important outcome measures resulted from the stored data, facilitating the treatment research. This will include annual deaths, morbidity and complication rates, adverse events and age distribution.

1. Information needed for patient and doctor services

To design and develop an RA registry system basic information is needed that includes: patient numbers, patient locations, confirmed diagnosis, the tests used to confirm the diagnosis, and other patient’s characteristics. Each diagnosis is classified according to a recognised coding system e.g. ICD10, SNOMED or OrphaCode.

Epidemiology however cannot be completed without the inclusion of patient outcomes, linked with some additional personal and clinical information [3]. Furthermore the medical data contained in the registry, apart from the planning of services, can be used for research purposes [4]. In this respect data may be extracted from an electronic patient record therefore, the registry and the electronic medical record must be designed in agreement with fundamental protocols and standards in order to achieve interoperability between the two systems for direct communication.

Furthermore, the inclusion of personal data makes requires that confidentiality and security of the medical data is established. This is an issue that is specifically underlined within the eENERCA project for the promotion of electronic registries for RAs across Europe.

2. Functionality of the proposed eRegistry

A patient registry is an organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease [5]. For the purposes of major RAs and other epidemiological health records, the eRegistry must meet several clinical, ethical and technical requirements.

Poorly designed human-machine interfaces can lead to many unexpected problems, thus for our system’s purposes, the structure of the front end of the database will be designed following Human Computer Interaction (HCI) [6] principles with the expectation to make the system user friendly.

A central characteristic of an eRegistry is interoperability, which means to use a generalized approach to represent every conceivable kind of health record data structure in a consistent way allowing communication and exchange of data between systems [7]. The Institute of Electrical and Electronics Engineers defines interoperability as the “ability of two or more components to exchange information and to use the information that has been exchanged” [8]. The absence of consensus on data standards in terminology, messaging, data structures, and data recording remains a primary barrier to an interoperable infrastructure [9, 10]. However, several standards and technical specifications are currently under development within the European Union as well as International standards (such as HL7, IHE, CEN/tC 251, 13940 etc) that ensure interoperability and patient-centered philosophy.
Knowledge of the epidemiological situation across Europe will be a major factor in policy development, service location, networking for the better diagnosis and case management and will also contribute in the understanding of the quality of both laboratory and clinical standards.

The database model of the eRegistry will include a list of tests required to firmly establish the diagnosis of the type of anaemia, for discriminating these rare anaemias from one another. After consultation with partners, an initial structure was decided which includes the following modalities:

2.1. Demographics

This section includes all necessary information to identify and locate the patient including the hospital or centre where the patient is receiving regular treatment. The list of data is based on the minimal dataset recommended by the Health Ministerial Conference which was held in Dublin in 2013 [11]. The EPIRARE dataset [12] was also consulted. This information, which is essential for epidemiological purposes, may also be hidden in cases where anonymisation is necessary.

2.2. Diagnosis

This is the most important single item and most essential for the epidemiological survey. The diagnosis must be correct and presented in a way that can be utilized for epidemiological purposes. For an interoperable system, it is important to follow the ICD 10 code, which is an international code, in recording patient’s diagnosis.

2.3. Clinical data

This section includes a yearly summary of clinical data. There is a compelling need for the creation of a centralized resource of clinical data on patients diagnosed with a rare disease in order to increase the efficacy of diagnosis, treatment and follow up of patients.

3. Discussion

The system will be available by the end of the project via a web interface through a password protected, tiered login process. The front-end is being developed using Django [14], which is an open-source high-level Python Web framework that encourages rapid development and clean, pragmatic design.

Currently, the design of a pilot study for eRegistry system for testing purposes is being designed that comprises some sections from the system analysis described above. A major issue for rare anaemias that will be solved with this registry is when the need arise for the collection of bio-samples. More research is focused on the generic origin of rare diseases, and researchers are increasingly interested in obtaining and storing bio-samples from patients for future analysis. Our future plans include expansion of the eRegistry to an EHR system, a tool assisting physicians to follow standards. The EHR will support an auditing tool as well.
4. Conclusion

Patients and healthcare providers will benefit from this eRegistry, since they can be provided with valuable sources of information on their disease. Rare anaemias eRegistries offer many advantages for research, due largely to their flexible design and ability to follow a broad group of patients for long periods. These registries will play an important role in improving understanding of rare anaemias, developing guidelines for disease treatment and management, and providing information to support the development of new treatments. This builds a foundation for a consensus and evidence-based disease management approach. Last by not least it demonstrates how eHealth funded initiatives supported by EU can benefit through applied research the citizen in need and the medical profession.

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References