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A Registry Framework Enabling Patient-Centred Care

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Abstract. Clinical decisions rely on expert knowledge that draws on quality patient phenotypic and physiological data. In this regard, systems that can support patient-centric care are essential. Patient registries are a key component of patient-centre care and can come in many forms such as disease-specific, recruitment, clinical, contact, post market and surveillance. There are, however, a number of significant challenges to overcome in order to maximise the utility of these information management systems to facilitate improved patient-centred care. Registries need to be harmonised regionally, nationally and internationally. However, the majority are implemented as standalone systems without consideration for data standards or system interoperability. Hence the task of harmonisation can become daunting. Fortunately, there are strategies to address this. In this paper, a disease registry framework is outlined that enables efficient deployment of national and international registries that can be modified dynamically as registry requirements evolve. This framework provides a basis for the development and implementation of data standards and enables patients to seamlessly belong to multiple registries. Other significant advances include the ability for registry curators to create and manage registries themselves without the need to contract software developers, and the concept of a registry description language for ease of registry template sharing.

Keywords. Disease registry, patient-centred care, data elements, registry description language

Introduction

Patient-centric information resources (or registries) are essential [1] [2] [3] [4] [5] [6]. With the focus of health care reform turning to patient-centred care, registries become even more critical. Patient-centred care is defined as care that is "respectful of and

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responsive to individual patient preferences, needs, and values, and ensuring that patient values guide all clinical decisions" [7]. The challenge then becomes the ability to dynamically assess to what extent patient-centred care is part of routine care in order to influence clinical decision making [8]. To achieve this, it is recognised that diagnosis begins with standardised data collection [9] and in this regard, appropriate patient registry design and implementation is crucial as a core enabler.

Registries come in many forms: disease-specific, recruitment, contact, clinical trial, post market and surveillance. In a rare disease context, patient registries must operate across jurisdictions and country borders. It is also expected that they interoperate with other registries, biobanks and most critically, electronic health records [6]. Unfortunately, the majority do not. Disease registries are typically stand alone, developed for different computer platforms, designed and implemented to different software development quality standards, implement varying levels of security, and are often times locked-in to proprietary data standards and system technologies rather than open standards. There are international efforts designed to harmonise these legacy systems such as the European Union Framework 7 project, RD Connect, which is an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research – http://rd-connect.eu).

Registries by their very definition become enduring information repositories that must be accessible and functional well beyond the life of funded initiatives. However, given project funding cycles and competitive research, registries are typically not interoperable, unnecessarily duplicated, and the data captured within these systems are not amenable to be linked easily to other important information resources. In a previous study, we proposed a checklist for stakeholders wishing to implement or deploy a registry system. In this checklist we identified key criteria for consideration such as technology choices, system design, security, sustainability, interoperability [4] to assist in strategic planning. We also discussed the term 'interoperability'. Unfortunately, with standalone registry systems, tedious manual and repetitive data exchange still occurs between them. NATO outlined four levels/Degrees of interoperability and we stated that patient registries must adopt Degree 3 and/or 4, seamless sharing of data, and seamless sharing of information, respectively [4]. Serious consideration must also be given to legacy systems to determine if the effort required to support, curate and extract information via manual methods weighs up against investment in migrating to a superior system with automated and interoperable processes.

It is recognised that registry requirements can evolve over time. For example, a registry may begin its life as a contact registry and then become a disease-specific one. If the software architecture cannot support this evolution, then this leads to separate registry creation and fragmentation [4]. We contend that designing and implementing an open source patient registry framework rather than just a single registry is a viable solution that can lead to achieving these higher degrees of interoperability [2] [4] [5] [6]. We provide an overview of this framework, its features and its development roadmap – ultimately to not just capture information but to become a useful knowledge management tool for patient-centred care.

1. Overview of Patient-Centric Registry Framework

The Registry Framework (RF) allows scientists and registry curators with standard computing skills to dynamically construct a complete patient registry from scratch, and

customise it for their specific needs, with little or no need to engage a software developer at any stage. New data elements for a diverse range of phenotypic and genotypic features can be defined at any time and can then be utilised and reused in any of the created registries. Fine grained, multi-level user and workgroup access can be applied to each data element to ensure appropriate access and data privacy. A number of key features of this framework are listed in Table 1. While this is not an exhaustive list, it includes desirable features, such as the ability to create multiple registries, patients being defined once but belonging to multiple registries, and the ability for curators to create data elements dynamically, well after the registry has been defined, enabling the registry to adapt to the evolving requirements of data capture.

Of particular interest is that within the RF, a registry created is defined by a description language. A standard patient registry can now be defined in a standardised, concise way. For example, the myotonic dystrophy (DM1) registry (excluding data elements) can now be encoded in just in just over 200 lines of computer-readable text [5] as opposed to the same registry implemented in a programming language (standalone) using 5000 lines of programming code [2] [4]. This definition file can be imported, exported, versioned and stored in a shared accessible environment [5]. Patient consent is captured through data elements, and while it can be customised as required, an example three level consent is currently in place: i) the patient consents to be part of the registry and have data retained and shared in accordance with the information provided to them; ii) the patient consents to be contacted about clinical trials or other studies related to their condition; and iii) the patient consents to be sent information on their condition.

1.1. Data Element Specification

Data element (DE) is a term used to define physiological measurements such as date of birth, body mass index, genotype, and so forth. Significant work has been undertaken to define data elements common to a class of diseases [1] [10]. While there are definitions for common data elements, for those that are common/specific to a given disease, a data element specification is required to be implemented. Not surprisingly, data elements currently implemented within patient registries are not sharable or reusable in other systems.

To date, the typical way to 'share' data elements (DE) is to share the names of the fields, usually captured in a spreadsheet. Unfortunately, this does not capture the DE specification details. If a DE specification existed, it would then be possible to share and exchange these definitions in a standardised way. In Table 1, we show that a data element can be an integer, float, string, date, range of values (permissible values), describe a file to be uploaded (e.g. a consent form) or be derived (calculated) from other data elements, referred to as derived data elements (DDE) [5]. It is possible to apply validation rules (min/max) for numeric fields, pattern validation for textual fields (such as health care card patterns) and to develop consistent graphical user interface (GUI) components for specific DEs. Within the RF, DEs and DDEs are described in a description file; they can be shared and most importantly, reused in other registries. In terms of the specification, a DE is made up of three sections, header, definition and ontology. A number of operations are possible as a result of this data element specification. For instance, a permissible value group (PVG) called *Size* can be specified with permissible values (PVs): (*large*, *medium*, *small*). Two different range DEs might use the same *Size* PVG. This level of abstraction ensures that both PVGs

and DEs are not tied to any single registry definition and can be reused easily in multiple registries.

1.2. Current Deployments

The RF is currently deployed for clinical-based, patient organisation-driven registries: DMD (live), SMA (live), DM1 [2] [4]. A number of other international and national disease registries driven by clinicians, patient advocate groups, patient wellbeing – surveillance and industry are in preparation for deployment.

2. Discussion: Future Directions of the Registry Framework

The framework design principles are to transform a registry to a knowledge management system, rather than merely an information capture system. We outline key directions of the RF development roadmap that facilitate this transformation.

2.1. Online Sharable Data Element Definition Resource

A registry consists of forms, sections and data elements contained within. It is possible to share and reuse forms and sections of previously created registries, as a definition file is generated for each registry. There will be a search capability to allow users to find previously defined registries, forms, sections and data elements. There is an upload section to allow data elements created by third parties to be shared. Data elements specification can now be shared. We are creating an online environment to store data elements that have already been developed. These data elements will be tagged in a fashion to enable them to be structured within a given data element ontology, such as according to NINDS common data element format [10].

2.2. Interoperability between Data Elements and Electronic Health Records

An important consideration for data elements is their interoperability, not just with other registries but with electronic health records (EHR). Fortunately, an ISO standard exists for data elements used within an EHR (CEN/ISO EN13606). Within an EHR context, the concept of a DE equivalent is referred to as Archetypes and we are defining our patient registry DE specification to be consistent with the Archetype model. Through this alignment, it is then possible to seamlessly exchange data between systems.

3. Conclusion

Interoperable disease registries underpin patient centred care, as is evidenced for rare disease patient care. In this manuscript we provided an overview of a registry framework that enables seamless adherence to not only common data standards, but also outlines a standard registry definition description language. This standard definition language is used to define components of a registry, namely, forms, sections, data elements and permissible value groups, for ease of sharing and adoption. It is then

possible for new groups to utilise an existing definition file to create a new registry or add new components to an existing registry compatible with other international/national registries. All this can be achieved without the need to engage with software developers. Finally, the commitment to open standards enables extensions to the framework to incorporate workflow modules to support models of care, notifications/reminders for clinicians. We have prototyped a clinical adjudication workflow within the framework that has multiple applications. In this way, the registry framework becomes a modular knowledge management system.

Table 1. Key features of the Registry Framework.

Feature	Description
Dynamic Creation of Data Elements (reusable fields)	Users of the RDRF (typically assigned administrators) have the ability to add new Data elements.
Data element support for various "abstract data types"	Framework supports: String (allows pattern matching/restrictions to be imposed); integer (with max/min); range (list/permissible values); calculated (functions); file (upload/download); float (real/decimal numbers); alphanumeric; Boolean (true/false presented as a check box).
Dynamic creation of a Registry	More than one Registry per web site is possible.
Dynamic Creation of Registry Forms	A Registry is made up of many Forms and each form is made up of Form Sections.
Dynamic creation of Questionnaire page for a Registry	Nominating a form as a questionnaire exposes the form on a public URL. The data captured by the form is stored as a "questionnaire response" which when approved by a curator, creates the patient record and also updates the clinical data record for the new patient.
Export Registry Definition File	A Registry is defined in Registry Definition File. This file can be exported from one RF installation to another (YAML format).
Import Registry Definition File	Enabling another RF installation to duplicate a complete Registry (YAML format).
Support for user defined Derived Data Elements	A Derived Data Element with a calculated designation can dynamically generate a value based on the values of other defined Data Elements or data object model.
Access permissions of each data elements can be modified dynamically (at Runtime)	Current roles in RF are admin, curator, clinician, genetic staff, and patient. This is customisable.
Widgets can be assigned dynamically to data element fields	Widgets can be selected dynamically (at runtime). This allows different display components (e.g. Date fields) to be chosen at run time. For instance, a Date Picker presents a calendar widget.
Exposed REST web service	Allows for patient data to be updated/retrieved by any client that can create HTTP requests.
Longitudinal data snapshots	Storage of longitudinal snapshots of data.
Dynamic multiplicity of fields for some form sections	A form section marked as "multiple" allows its fields to be dynamically added or removed en block (e.g. a multiple contacts section could list contact name, email, relationship as three fields - by marking the section as multiple the framework adds an add/remove button to the page which allows multiple contacts to be added.
Molecular sequence	HGVS annotations can be captured.

Patients in multiple registries	Within the RDRF, patients can be in one or more registries without the need of duplicating patient information.
One RF = Multiple registries	Multiple registries can be managed in one installation.
IP address restrictions	Within RDRF it is possible to define external IP (Internet Protocol) address to ban or allow user(s) from accessing registries defined within the RF.
User login attempts auditing	Audit trails of all user login attempts.
Questionnaire validation	Moderated workflow for questionnaire submission.
19. Open source and RF Deployment	RF is open source (GNU GPL v3). Centos 6 via creation of RPMs which are uploaded to a YUM Repository. Docker image on docker registry.
User documentation	https://readthedocs.org/projects/rare-disease-registry-framework/
Registry landing page	It is possible to create a customisable landing page for each registry.
Consent	Within RF there are capabilities for multiple levels of patient consent.
Demo available	https://rdrf.ccgapps.com.au/demo/ (username and log in: admin | admin; curator | curator; clinical | clinical; genetic | genetic

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