

PODD: An Ontology-driven Data Repository for Collaborative Phenomics Research

Yuan-Fang Li, Gavin Kennedy, Faith Davies, and Jane Hunter

School of ITEE, The University of Queensland
{uqyli4,g.kennedy1,f.davies,j.hunter1}@uq.edu.au

Abstract. Phenomics, the systematic study of phenotypes, is an emerging field of research in biology. It complements genomics, the study of genotypes, and is becoming an increasingly critical tool to understand phenomena such as plant morphology and human diseases. Phenomics studies make use of both high- and low-throughput imaging and measurement devices to capture data, which are subsequently used for analysis. As a result, high volumes of data are generated on a regular basis, making storage, management, annotation and distribution a challenging task. Sufficient contextual information, the metadata, must also be maintained to facilitate the dissemination of these data. The challenge is further complicated by the need to support emerging technologies and processes in phenomics research. This paper describes our effort in designing and developing an ontology-driven, open, extensible data repository to support collaborative phenomics research in Australia.

Keyword: OWL, ontology, repository, phenomics, data management

1 Introduction

An organism’s phenotype is an observable or quantifiable trait of the organism as a consequence of its genetic makeup combined with its developmental stage, environment and disease conditions. Phenomics is the systematic and comprehensive study of an organism’s phenotype and is determined through a combination of high-throughput and high-resolution imaging- and measurement-based analysis platforms. Phenomics research, together with genomics research, represents a holistic approach to biological study [3, 11, 7].

Unlike genomics, phenomics research emphasizes physical, observable traits of the subject under study. Like genomics, vast amounts of data are produced by imaging and measurement platforms and analysis tools. The storage, management, analysis and publication of these data is a challenging problem.

Specifically, there are three key challenges for data management in phenomics research.

- The ability to provide a data management service that can manage large quantities of heterogeneous data in multiple formats (text, image, video) and not be constrained to a finite set of imaging and measurement platforms and data formats.

- The ability to support metadata-related services to provide context and structure for data within the data management service to facilitate effective search, query and dissemination.
- The ability to accommodate evolving and emerging technologies and processes as phenomics is still a rapidly developing field of research.

The Phenomics Ontology Driven Data (PODD) repository¹ is being developed to meet the above challenges facing the Australian phenomics research community, aiming at providing efficient and flexible repository functionalities for large-scale phenomics data. An important goal of PODD is to provide a mechanism for maintaining structured and precise *metadata* around the raw data so that they can be distributed and published in a reusable fashion.

Differences in research project reporting, organisms under study, research objectives, research methodologies and imaging and measurement platforms may result in differences in the *models* of data. In order to accommodate as wide a variety of biological research activities as possible, we have constructed the domain model using OWL [2] ontologies, instead of the traditional UML class diagrams and database schemas. The OWL domain model is at the core of the PODD repository as it drives the creation, storage, validation, query and search of data and metadata. In contrast to traditional data repositories that use database schemas as the underlying model, the employment of OWL ontologies as the domain model makes PODD highly extensible.

In this paper, we present our work in addressing the above challenges and highlight the OWL-based modeling approach we take. The rest of the paper is organized as follows. In Section 2 we present some related work and give a brief overview of PODD and relevant technical background. Section 3 presents the high-level design of the repository. In Section 4, we discuss the PODD domain ontology in more detail and show how the ontology-based modeling approach is used in the life cycle of domain objects. Finally, Section 5 concludes the paper and identifies future directions.

2 Overview

2.1 Related Work

In biological research, a large number of databases have been developed to host a variety of information such as genes (Ensembl²), proteins (UniProt³), scientific publications (PubMed⁴) and micro-array data (GEO⁵).

These databases are generally characterized by the fact that they specialize in a particular kind of data (protein sequences, publications, etc.) and the conceptual domain model is relatively well understood and stable. As a result, the need for extensibility and flexibility is not very high.

¹ <http://www.itee.uq.edu.au/~eresearch/projects/podd/>

² <http://www.ensembl.org/>

³ <http://www.uniprot.org/>

⁴ <http://www.ncbi.nlm.nih.gov/pubmed/>

⁵ <http://www.ncbi.nlm.nih.gov/geo/>

Phenomics is a fast growing discipline in biology and new technologies and processes are evolving and emerging rapidly. As a result, the domain model must be flexible enough to cater for such changes. Currently there are a number of related domain models available.

Functional Genomics Experiment Model (FuGe) [4] is an extensible modeling framework for high-throughput functional genomics experiments, aiming at increasing the consistency and efficiency of experimental data modeling for the molecular biology research community. Centered around the concept of experiments, it encompasses domain concepts such as protocols, samples and data. FuGe is developed using UML from which XML Schemas and database definitions are derived. The FuGe model covers not only biology-specific information such as molecules, data and investigation, it also defines commonly-used concepts such as audit, reference and measurement.

Extensions in FuGe are defined using inheritance of UML classes. We feel that the extensibility we require is not met by FuGe as any addition of new concepts would require the development of new database schemas and code. Moreover, the concrete objects reside in relational databases, making subsequent integration and dissemination more difficult.

The Ontology for Biomedical Investigations (OBI)⁶ is an on-going effort of developing an integrative ontology for biological and clinical investigations. It takes a top-down approach by reusing high-level, abstract concepts from other ontologies. It includes 2,600+ OWL [2] classes and 10,000+ axioms (in the import closure of the OBI ontology). Although OBI is very comprehensive, its size and complexity makes reasoning and querying of OBI-based ontologies and RDF graphs computationally expensive and time consuming.

2.2 The PODD Repository

Under the National Collaborative Research Infrastructure Strategy (NCRIS), the Australian Government has funded two major phenomics initiatives: The Australian Plant Phenomics Facility (APPF), specializing in phenotyping crop and model plant species; and the Australian Phenomics Network (APN), which specializes in the phenotyping of mouse models. Both facilities have common requirements to gather and annotate data from both high- and low-throughput phenotyping devices. The scale of measurement can be from the micro or cellular level, through the level of a single organism, and up to (in the case of the APPF) the macro or field level.

An organism's phenotype, observable and quantifiable traits, is often the product of the organism's genetic makeup, its development stage, disease conditions and its environment. Any measurement made against an organism needs to be recorded in the context of these other data. The opportunity exists to create a repository to record the data, its contextual data (metadata) and data classifiers in the form of ontological or structured vocabulary terms. The structured nature of this repository would support manual and autonomous data discovery

⁶ <http://obi-ontology.org/>

as well as provide the infrastructure for data based collaborations with domestic and international research institutions. Currently there are no such integrated systems available to the two facilities.

The National eResearch Architecture Taskforce (NeAT) Australia initiated the PODD project to fill this gap. In PODD, we have engaged in the design and development of the Phenomics Ontology Driven Data (PODD) repository. The goal of PODD is to capture, manage, annotate and distribute the data generated by phenotyping platforms. It supports both Australian and international biological research communities by providing repository and data publication services.

2.3 The OWL Ontology Language

The Web Ontology Language (OWL) [2] is one of the cornerstone languages in the Semantic Web technology stack. Based on description logics [6], OWL DL (a sub species of OWL) defines a precise and unambiguous semantics, which is carefully crafted so that it is very expressive yet core reasoning tasks can be fully automated.

Information in OWL, as in RDF [5], is modeled in triples: $\langle \textit{subject}, \textit{predicate}, \textit{object} \rangle$, where *subject* is the entity of interest, *predicate* represents a particular characteristic/property of the entity and *object* is the actual value of that property. *Classes* are first-class citizens in OWL. They represent abstract concepts in a particular domain. Concrete objects are represented by OWL *individuals*. OWL *predicates* are used to relate OWL entities (classes, individuals, predicates) to their attributes or other entities.

$\mathcal{C} ::= C$	- Class name	$\mathcal{AX} ::= C \sqsubseteq C$	- Class subsumption
\top	- Top class	$C = C$	- Class equivalence
\perp	- Bottom class	$C \sqcap C = \perp$	- Class disjointness
$C \sqcup C$	- Class union	$P \sqsubseteq P$	- Property subsumption
$C \sqcap C$	- Class intersection	$P = P$	- Property equivalence
$\neg C$	- Class negation	$\geq 1 P \sqsubseteq C$	- Property domain
$\forall P.C$	- Universal quantification	$\top \sqsubseteq \forall P.C$	- Property range
$\exists P.C$	- Existential quantification	$\top \sqsubseteq \leq 1 P$	- Functional property
$P: o$	- Value restriction	$P = (\neg P)$	- Inverse property
$\geq n P$	- At least number restriction		
$\leq n P$	- At most number restriction		
$\{a_1, \dots, a_n\}$	- Enumeration		

Fig. 1. OWL expressions

Fig. 2. OWL class and predicate axioms

Class descriptions can be used to construct (anonymous) complex class expressions from existing ones, as shown in Figure 1. \mathcal{C} represents (possible complex) class expressions; C is a class name; P stands for a predicate; n is a natural number and a_i 's are individuals. OWL uses *axioms* to place restrictions

on OWL classes and predicates. These axioms include class subsumption, equivalence, disjointness; predicate domain, range, etc. Figure 2 shows some of the axioms.

The OWL language has been widely used in life sciences and biotechnology [10, 12, 1] as a modeling language for its expressivity and extensibility. There is also growing tool support for tasks such as reasoning, querying and visualization, making it a viable option for the modeling and representation of domain concepts and objects in phenomics.

3 High-level Design of PODD

PODD is intended to be an open platform that allows any user to access data that is either published or explicitly shared with them by the data owners. Moreover, PODD has been envisioned to provide data management services for a wide variety of research projects and phenotyping platforms.

The key design considerations of PODD include:

- Data storage and management. It is estimated that several TB of data will be generated by PODD clients per year. Hence, the ability to efficiently manage large volumes of data is crucial.
- Repository reusability. Data generated by a wide range of projects and phenotyping platforms will be stored in the repository. Hence, the domain model needs to be flexible enough to cater for the administrative, methodological and technical differences across projects and platforms.
- Data persistence and identification. In order to support the dissemination of scientific findings, data in the repository needs to be publicly accessible after being published. Hence, a persistent naming scheme is required.

In the development of PODD we employ a number of core technologies to meet the above requirements.

- We use Fedora Commons⁷, a digital repository for the management, storage and retrieval of domain objects.
- We use iRODS [9], a distributed, grid-based storage software system, for the actual storage solution of domain objects across a virtual data fabric.
- We incorporate the Sesame⁸ triple store for the storage and query of RDF triples (ontology definitions of concrete objects).
- We use the Lucene⁹ open-source search engine for the full-text index and search of repository contents, including values in the RDF triple store.

The high-level architecture of the PODD repository can be seen in Figure 3.

⁷ <http://www.fedora-commons.org/>

⁸ <http://www.openrdf.org/>

⁹ <http://lucene.apache.org/>

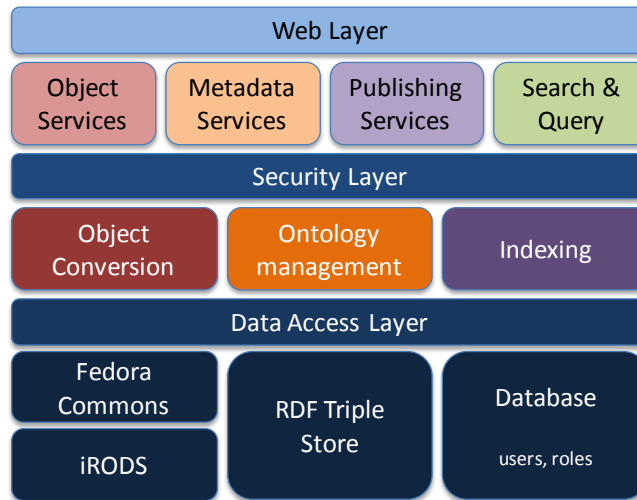


Fig. 3. The high-level architecture of the PODD repository.

4 Ontology-based Domain Modeling

One of the important design decisions to be made early in the development process is the domain model. Domain modeling aims at providing solutions for two important tasks. Firstly, efficient and flexible data organization; and secondly, data contextualization in the form of *metadata*, so as to provide meaning for the raw data (documents, publications, image files, etc.) to facilitate search, query, dissemination, and so on.

As we emphasized previously, the domain model should be flexible enough to accommodate the rapid changes and advancement of phenomics research. Inspired by FuGe and OBI, we created our own PODD ontology in OWL to define essential domain concepts and relations.

4.1 The PODD Ontology

As PODD is designed to support the data management tasks for phenomics research, a number of essential domain concepts and the relationships between these concepts need to be modeled. Domain concepts will be modeled as OWL classes; relationships between concepts and object attributes will be modeled as OWL object- and datatype-predicates. Concrete objects will be modeled as OWL individuals.

The Domain Concepts In our modeling, the top-level concept is *Project*, which is an administrative concept and contains essential meta information about the research project, such as the administering organization, principal investigator, project membership, project status, etc.

A number of concepts may be associated with the project concept.

Project Plan describing the current project plan at the core metadata element level.

Platform describing any single technical measurement platform used in the project. Technical measurement platform means any platform for which parameters and parameter values may be captured.

Genotype describing the genotype of the materials used in the investigations. Multiple genotypes are described here and then can become fields in the instances of the Material object.

Investigation describing a planned process within a project that executes some form of study design and produces a coherent set of results. It can be considered equivalent to an experiment.

The **Investigation** concept is of central importance. It captures the data and metadata of experiments under a project. A number of concepts are defined to assist in the modeling of investigations.

Experimental Design describing experimental design components, e.g. plant layouts, sampling strategies, etc.

Growth Condition describing growth conditions, such as growth chambers used, environmental settings, etc.

Process representing a planned component of an investigation. It is a description of a series of steps taken to achieve the objective of the investigation.

Protocol describing a step within a process that is a consistent whole. e.g. sterilize seeds, plant seedlings, image the plants.

Material describing the materials used in the investigation. Materials can be either inputs or outputs. They can be chemicals, substrates, whole organisms or samples taken from a whole organism. The meaning of the Material is usually derived from its position in the model (as well as core metadata elements such as Type).

Event capturing ad-hoc events and actions that occur against an individual material. In most instances the events and their timing are described in the process/protocol. An Event object can be utilized to either record fixed events in a form that allows for investigative analysis, or to record one off observations (e.g., the plant under observation died).

Measurement describing a single measurement against a single material. e.g. an image of a plant is a single measurement. Measurement objects can capture measurement variables (e.g. shutter speed, lighting, etc).

Analysis An Analysis object is a variation on a protocol object, in that it describes a step in a process. It is currently linked to the Project, Investigation objects as well as to specific Measurement objects since analyses can be performed on single measurements and also on the entire investigation or project based on multiple inputs.

Inter-concept Relationships The structures and workflows of phenomics research activities are captured using inter-concept relationships, which are defined using OWL predicates.

Different research projects will utilize different measurement platforms and have significantly different approaches and project designs.. As a result, different projects may have different structures. In OWL, there are a number of ways of defining the same relationship. In order to achieve high modeling flexibility and accommodate as many scenarios as possible, we have made the following design decisions:

- Use OWL restrictions to define inter-concept relationships. OWL restrictions impose constraints on the OWL classes they are defined in.
- Only define domain, or range, but not both, for predicates, so that the predicates can be used by different concepts.

For each of the concepts described in the previous subsection except for *Project*, we define an OWL object-predicate with the range being the concept. For example, for concept *Analysis*, we define a predicate *hasAnalysis* and define its range to be *Analysis*.

Object Attributes Attributes are inherent properties of an object, such as the start date of a project, the timestamp of an event, etc. In our model, we use OWL datatype-predicates to model object attributes, similar to the modeling of inter-object relationships.

Figure 4 shows the partial definition of the OWL class *Project*, in OWL DL syntax [2]. Restriction 1 states that any *Project* instance must have exactly one *ProjectPlan* (through the predicate *hasProjectPlan*, the range of which is *ProjectPlan*). The other 3 restrictions are similarly defined.

$$Project \sqsubseteq = 1 \text{ hasProjectPlan} \tag{1}$$

$$\sqsubseteq \geq 1 \text{ hasInvestigation} \tag{2}$$

$$\sqsubseteq = 1 \text{ hasStartDate} \tag{3}$$

$$\sqsubseteq \leq 1 \text{ hasPublicationDate} \tag{4}$$

Fig. 4. Partial OWL Definition for the Project concept.

4.2 Ontology-based Model in Object Life Cycle

Concrete objects, instantiations of various concepts such as *Project* and *Investigation*, are stored in PODD and can subsequently be retrieved for different purposes. As stated in Section 1, the ontology-based domain model is at the center of the whole life cycle of objects. In this subsection, we briefly present the roles the ontology-based model perform at various stages of the object life cycle.

Ingestion When an object is created, the user specifies which type of object she intends to create and the repository will pull up all the ontological definitions for that type (from the OWL class corresponding to that type and its super classes). Such definitions will be used to (a) guide the rendering of object creation interfaces and (b) validate the attributes and inter-object relationships the user has entered before the object is ingested.

Retrieval & update When an object is retrieved from the repository, its attributes and inter-object relations are retrieved from its ontology definition, which is used to drive the on-screen rendering. When any value is updated, it is validated and updated in object's ontology definition.

Query & search An object's ontology definitions will be stored in an RDF [5] triple store, which can be queried using the SPARQL [8] query language. Similarly, ontology definitions can be indexed and searched by search engines such as Lucene.

In summary, ontology-based domain modeling enables us to build very expressive and extensible phenomics domain models. Ample tool support is also available to perform ontology-based tasks such as validation, querying and searching.

5 Conclusion

Phenomics is an emergent discipline that is poised to have a significant impact upon industrial-scale biological research. Phenomics presents a number of data management challenges, such as managing high volumes of data, integrating highly heterogeneous datasets and ensuring the data will exist in perpetuity.

To meet the data management needs of the Australian phenomics research community, the PODD repository is being developed to enable efficient storage, retrieval, contextualization, query, discovery and publication of large amounts of data.

Central to the design and development of PODD is the use of OWL ontologies as the domain model. Based on description logics and with an emphasis on the Web, the OWL language features a precise semantics, high expressivity and high extensibility. It also has mature and growing tool support. As a result, the OWL language has been widely used in bioinformatics and life sciences to mark up genetic, molecular and disease information.

In the PODD model, core domain concepts are defined as OWL classes. Their attributes and relationships with other domain concepts are defined as OWL class restrictions. Concrete domain objects are then initialized, conforming to the ontologies defined for their concepts. Such a modeling approach has a number of benefits.

- Firstly, concepts are defined with unambiguous syntax and precise semantics, enabling automated validation and rendering.
- Secondly, the extensible nature of OWL language ensures that new concepts can be easily added.
- Thirdly, as OWL and RDF are both open standards, interoperability between repositories is expected to be high.

In this paper, we introduce the high-level architecture of the PODD repository and the main technologies used in developing it. We focus on the ontology-based domain modeling approach, present the PODD domain model and discuss its role in the life cycle of concrete domain objects.

The development of the PODD repository will focus on enhancing ontology-based modeling, representation and processing of phenomics data. Ontology-based annotation services, automated data integration and data visualization will be part of the future directions.

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