Towards a Standard Ontology Metadata Model

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Abstract— Bio-ontologies are becoming increasingly important in semantic alignment for data integration, information exchange, and semantic interoperability. Due to the large number of emerging bio-ontologies, it is challenging for naïve ontology users to search, select, and adopt a "right" ontology for their applications. Therefore, it is important to have a consistent terminology metadata model and a resource for discovering appropriate ontologies or other resource for use in annotating data. This paper aims to seek a common, shareable, and comprehensive method to create, disseminate, and consume metadata about terminology resources.

Keywords—Ontology; Metadata, Ontology Metadata Model

I. INTRODUCTION

Ontologies, thesaruri, termiologies, classifications and coding systems can be referred to collectively as 'terminology resources." Ontologies and other terminology resources are becoming increasingly important in semantic alignment for semantic data integration, information exchange, and interoperability [1, 2] especially in the biomedical field. New bio-ontologies continue to emerge. For example, the number of terminology resources published in the National Center for Biomedical Ontology (NCBO) BioPortal increased from 72 in 2008 [3] to 508 in April 2016. The explosion of available terminology resources makes it challenging for naïve users to search, select, and adopt the "right" terminology resource for their applications [4]. Core issues in adoption include: (1) Lack of advertisement: Users such as software developers often do not know about the existence of standards, vocabularies, or ontologies; (2) Lack of description: Users do not know which standard, terminology resource, or ontology would be best for their particular use; and (3) Lack of a rating system: Users do not know the quality of a selected ontology.

An ontology metadata model provides a method to richly characterize terminology resources. It therefore has the potential to mitigate the issue of discovery and improve ontology adoption. It also would facilitate comparison as well as coordination relating to standards and best practices. Structured metadata describes key aspects of a terminology resource such as scope, structure, provenance, availability, and usage statistics. It helps users to identify, evaluate, select, and deploy a resource more effectively and efficiently. The primary goal for this paper is to seek a common, shareable, and comprehensive method to create, disseminate, and consume metadata about terminology resources.

II. BACKGROUND

The NCI Center for Biomedical Informatics and Information Technology (CBIIT), NCBO, and National Cancer Research Institute (NCRI) in the U.K. support the fundamental concept that adoption of ontologies by the research and education communities is essential to data sharing, interoperability and reuse. NCI, NCBO and NCRI had a common interest in representing terminology resources using a standard ontology metadata model. Although there are several existing models and standards efforts for an ontology metadata profile, the NCI CBIIT, NCBO, and NCRI realized that no single metadata model could meet the requirements collected from those three institutions in 2012. Therefore we proposed to contribute to a possible standard ontology metadata model by harmonizing existing models.

A. NCI CBIIT

Major activities of the NCI CBIIT include developing, coordinating, and/or deploying biomedical-informatics and scientific-management information technology systems, infrastructure, open-source applications, semantics, and data resources in support of the NCI's research agenda [5]. NCI Enterprise Vocabulary Services (EVS) publishes a number of vocabularies needed and used by the NCI community [6]. It also provides tools and services to accurately code, analyze and share cancer and biomedical research, clinical and public health information. Though EVS publishes metadata with terminologies, challenges remain as to how to make descriptive information for those vocabularies available to the end-user communities in a comparable way to terminologies hosted by other organizations.

B. NCBO

The NCBO is one of the National Institutes of Health Centers for Biomedical Computing (NCBCs). The goal of the NCBO is to support biomedical researchers by providing online tools, a web-based BioPortal, and programming interfaces, enabling researchers to access, review, and integrate disparate ontological resources in all aspects of biomedical investigation and clinical practice [7]. BioPortal enables community participation in the evaluation and evolution of ontology content by allowing users to submit their ontologies to BioPortal, providing mappings, managing ontology versions, and collecting user feedback through structured notes and reviews [3]. The metadata model of the NCBO BioPortal is based on an extended version of the Ontology Metadata Vocabulary (OMV) [8].

C. NCRI

The U.K.'s NCRI focused on the technical and cultural aspects of data sharing [9]. The NCRI developed an online tool called "Cancer InfoMatrix" for the visualization and discovery of three types of standards: vocabularies, exchange formats, and reporting guidelines. The dilemma NCRI faced was how to promote a consistent set of "core" standards to the community in order to produce a coherent pattern of use of these "core" standards.

The NCRI worked to achieve consensus with and across NCI and NCBO in defining the way forward in promoting standards in order to facilitate international interoperability. A joint group, the Ontology Representation Working Group (ORWG), was founded to serve this purpose. The major goal for the ORWG was to seek a common, shareable and comprehensive method to create, disseminate, and consume metadata about ontologies.

Beside the three institutions that were involved in this activity, the OBO Foundry [10], BioSharing [11], the Research Data Alliance (RDA) [12], the Monarch Initiative [13], and Elixir in Europe [14] are also active in this area, e.g. Biosharing provides usage data about terminologies and other resources, and obofoundry.org provides high quality metadata for a selective collection of terminology resources. Work towards a common model for terminology resources metadata would benefit all.

III. METHODS

The development of an ontology metadata model contains four major steps: (A) Review the existing metadata models, (B) Identify important metadata based on a user survey of the three organizations that collaborated on the ORWG, (C) Propose and ballot new extensions and/or modifications toward a new normative edition of an ontology metadata model, and (D) Adoption of the new version by the community.

A. Review Existing Models

We reviewed existing ontology metadata models including ISO/ IEC 19763-3 [15], Ontology Definition Metamodel [16], Open Provenance Model [17], Open Ontology Repository [18], and Ontology Metadata Vocabulary (OMV) [19]. Our evaluation showed that ISO/IEC 19763-3, Ontology Definition Metamodel, and Open Provenance Model do not support the breadth of profile representation required by our three institutions. Specifically, these models have some limitations in their support of several use-cases important to this group, for example, the intended scope of the content of a terminology resource, its type, language or syntax, and its fitness for use for a given purpose. The Open Ontology Repository initiative also realized the importance of the ontology metadata and was willing to collaborate with the ORWG. Based on our evaluation, OMV was the most complete and thoughtfully developed metadata model among these models for the use cases. One of the three institutions, NCBO, already uses OMV as the supporting model for its BioPortal. And given that it contained the best metadata for the use cases, the ORWG decided to choose the OMV version 2.4.1 as its base model.

B. Identify a List of Important Metadata

A survey to the NCI, NCBO, and NCRI was created and conducted by the ORWG. The content of the survey was designed based on OMV core. Authors from the three institutions filled out the survey based on their own needs and requirements. Each piece of metadata from the OMV was prioritized as "Low, Medium, or High" and classified as "Required or Optional". A list of important metadata was finalized based on group consensus. The detailed survey can be found in the Results section. Usability testing was done by applying OMV to four relevant biomedical vocabularies: LOINC, SNOMED CT, RxNorm, and NCI Thesaurus. A summary of the outcome of usability testing results is also described in the Results section.

C. OMV Extensions or Modifications

Based on the results from our survey, review, and OMV usability tests, the ORWG group suggested that further extensions and/or modifications to OMV are an ideal approach to enable OMV to serve as a single model designed to support the requirements of the entities involved in this paper and potentially the rest of the biomedical community.

D. Adopt the OMV Extensions

The OMV extensions influenced the final selection of metadata in the CTS2 standard. A set of standardized terminology metadata allows applications using terminology services to build on a common infrastructure, and improve interoperability across applications. For example, NCI's LexEVS CTS2 server implements a portion of the model, as does a recent pilot project for implementing a federated network of terminology service nodes [20]. Recommendations were generated by the ORWG with a broad range of issues including publishing, distribution, implementation, maintenance licensing, provenance, and community input.

IV. RESULTS

The results of this study include (1) a list of important metadata data elements, (2) usability testing results, and (3) a set of recommendations consisting of changes to existing properties and proposed extensions in support of a newly revised version of OMV.

A. A list of important metadata

A list of important terminology metadata elements was extracted from the survey results. The content of this survey was derived from the OMV Ontology class. The structure of the OMV contains classes, elements, and relationships. The OMV has 15 classes such as Ontology, Ontology Type, Ontology Language, Ontology Engineering Methodology, Party, etc. Elements are properties or characteristics that are used to describe the class. For example, the Ontology class has 34 elements including URI, name, acronym, version, etc. The relationships link the OMV classes together.

Metadata elements were tagged with an "Occurrence Constraint" (whether an element should be Required or Optional) and "Ranking" (Importance of the element to utility - High, Medium, or Low) by the three institutions to judge the importance of the metadata based on their own requirements and needs. Eight out of the 34 OMV metadata elements were identified as "Required" to describe a source (e.g., name, description, and creationDate). Ten out of the 34 metadata elements were identified as "High" priority (e.g., URI, name, and description). Some metadata were classified as both "Required" and "High" (e.g., ontology name and description). Some were ranked as a "High" priority but were also recommended as "Optional". For example, not all ontologies in the NCBO BioPortal have a URI and some ontologies have more than one URI. Table 1 shows the group's determination of metadata elements identified as 'core'.

Name or Properties	Occurrence Constraint (Required or Optional)	Ranking (High, Medium, or Low)	
URI	Optional	High	
name	Required	High	
description	Required	High	
keywords	Optional	High	
hasDomain	Optional	High	
creationDate	Required	Medium	
knownUsage	Optional	High	
naturalLanguage	Required	Medium	
hasOntologyLanguage	Required	High	
resourceLocator	Required	Medium	
version	Required	High	
hasLicense	Required	High	
useImports	Optional	High	

B. Usability Test Results for the OMV

The OMV was tested with four widely used vocabularies: LOINC [21], SNOMED CT [22], RxNorm [23], and NCI Thesaurus [24]. The reasons for choosing these four include (1) the purpose and scope of these vocabularies are different. LOINC is a coding system for laboratory and clinical observations. SNOMED CT is a systematically organized computer processable ontology of medical terms. RxNorm, published by NLM, and included in the UMLS, provides normalized names and a model for clinical drugs available in the US. Finally, the NCI Thesaurus includes broad coverage related to the cancer research domain. (2) The native presentation and structure of these terminologies vary; LOINC is a coding system with pre-coordinated terms constructed from elements in six axes, while SNOMED CT and the NCI Thesaurus are based on description logics, and RxNorm is a terminology with a sophisticated model but not a Description Logic (DL) ontology. A sample application of the metadata elements to RxNorm was presented at [25].

C. OMV Extensions/Modifications

A revision to the OMV was recommended by both refinement of existing OMV ontology class elements [21] and via extensions. Modifications included (a) Rename, (b) Refine, (c) Relocate, (d) Remove, and (e) Harmonization. The purpose of these modifications was to make the OMV easier to understand and use. Modifications and changes are described below and summarized in Table 2.

a) Rename – To solve the ambiguity problem: The element name was changed to the widely accepted term. For

example, the "name" and "acronym" were changed to the fullName and shortName respectively.

b) Refine definition – To solve the ambiguity problem: Some textual definitions for elements were modified. For example, the definition of the "fullName" was defined as "the name by which an ontology is known". Sample values for the element were also included in the definition. For example, the full name of an ontology can be "Logical Observation Identifiers Names and Codes".

c) Relocate – To provide a better way to organize the information: Elements were relocated to other classes if they did not belong to the OMV Core class. For example, "creationDate" and "modificationDate" were removed from the Core Ontology class and relocated into a newly created Version class.

d) Remove – To simplify the model: Some elements were identified as not necessary according to group consensus. These unimportant elements were removed from the new model (e.g., "hasPriorVersion").

e) Harmonization – To refine/extend the model: OMV's single "endorsedBy" element represents an important concept that should be expanded into more atomic elements for greater expression. The "endorsedBy" is a relation between "Ontology" to a "Party" as used in the OMV to represent either a person or organization. Therefore it is difficult to disambiguate the types of entities supporting the ontology. New properties such as "certifiedBy" and "mandatedBy" were added to the OMV extensions in order to capture these details with finer granularity. For example, "certifiedBy" is done under formal reviews by CBIIT or "mandatedBy" for requirements for use by regulatory or governmental agencies such as under the Meaningful Use criteria for deployment of electronic health records in the United States.

The OMV extensions were developed by harmonizing existing well-known metadata standards such as Dublin Core [26]. The extensions for the OMV Ontology class are presented in Table 2. The last column in Table 2 shows the comparable terms from Dublin Core Metadata.

D. Recommendations/Next Steps

The ORWG developed a set of recommendations relating to issues for ontology distribution, implementation, and maintenance as well as licensing, provenance, and community participation. A summary of those recommendations are as follows.

a) Core Ontology Metadata: An iterative approach should be adopted to implement consistent ontology metadata beginning with core metadata and moving outward to more comprehensive metadata as it proves beneficial. The scope of the metadata should include core areas such as content, structure, provenance, documentation, and certification. A revised version of OMV core metadata should be evaluated or audited against several ontologies relevant to the biomedical community. Future updates or revisions to the core ontology profile should be created based on the feedback from the evaluation. b) Adoption of Prevailing Standards: Reuse of portions of widely adopted models would improve the opportunities for broader adoption, ease of use of existing tools, and therefore the potential for federation and semantic interoperability. To the authors' knowledge, OMV deployment so far has been within stand-alone environments, although API's (e.g., the REST API in BioPortal) have been used to facilitate query and retrieval. The impact for the replacement of OMV elements would affect NCBO's modeling in BioPortal. Continued observation of emerging models important to the community such as updates to the CTS2 was recommended.

c) Ontology Usage: "knownUsage" is one of the most important and yet difficult metadata elements to capture, measure, or share. "Usage" can be described by a simple declarative example, by actual use case, case study, or by reference to a project, activity, or data. Since it is difficult to define a formal and detailed schema for this element, the ORWG suggested that it should be carefully described with best practices or guidelines to capture information from the community. Language should be succinct, but should emphasize relatively verbose sharing of details of how an ontology is used including successes, failures, challenges, innovations, activities such as mapping or merging, level of effort and so on. The goal is to capture a wide range of instances of how, where and why an ontology is being used, including those outside the primary scope or intent published by the developer. All usage examples could be helpful, even if they are novel or represent outlying use cases.

d) Intellectual Property: Licensing or Rights Expression Description of licensing rights in the current metadata models (i.e., "hasLicense") is fairly general. Additional information about attribution, reuse, distribution, and guidance for variances in licenses should be provided by ontology owners/submitters. These variances include licensing of artifacts as creative works (e.g., Creative Commons), open source licensing of source code, hybrid or dual licensing (e.g., commercial open source), and complexities in licensing by geographic region (e.g., affiliate licensing under IHTSDO for SNOMED-CT).

e) Creation, Maintenance. and Distribution of Terminology Metadata Profiles: Terminology owners are the ideal source to initially populate profiles as well as to curate subsequent revisions. Although users are valuable in providing more atomic updates, annotations, reviews, and unique perspectives, the level of detail in a profile and the difficulty in assuring accuracy requires dependence on a more authoritative and familiar source. Biosharing, for instance recognizes this as well, and attempts to get resource owners to 'claim' and update the metadata record. Source information must also be accurate and timely and even with initial population by the ontology owners, continued curation by the manager of the repository or registry remains a significant burden and at risk for obsolescence. The ORWG recommended a common approach where owners or community members may maintain a verified or validated profile locally by populating a "standard" metadata profile

based on the revision of the OMV for publication using a unique identifier and namespace. A profile service would be able to periodically query and archive updated ontology profiles or the service could ping the service with the URL to the resource at the moment it is modified or updated.

V. DISCUSSION

The lack of a standard terminology metadata schema to comprehensively describe biomedical ontologies is a challenge and a barrier to users looking to identify terminology resources appropriate for their use. Even though a tool such as the Ontology Recommender [27] can help steer people to resources that contain terms in specific areas of interest, choosing an appropriate resource is also a matter of understanding its characteristics, how widely it is used and for what purposes, how it is supported, and so on. This study focused on identifying and evaluating the requirements of terminology metadata profiles for various levels of semantic precision. It also investigated the extensible framework (including a terminology metadata profile, an effective federated collaboration platform, and the capability of engaging the community) for representing important ontology resources.

Of all the available metadata models, the OMV Version 2.4.1 was selected as the best foundation for developing a shared, harmonized, and standard metadata model. However the OMV Consortium has no active governance structure to accept community contributions and there has been no published update since 2009. It is currently insufficient to meet the representational needs of a richer community-based framework. This study identified gaps as well as recommendations to extend and refine features of the OMV.

A. Challenges to Populate Values for a Terminology Metadata Model

It is difficult to populate values for non-ontologies since they do not have a more formal/explicit ontological representation. Some "Required" elements in the OMV have to be left blank. For example, LOINC has a NULL value for the required element "hasOntologyLanguage". Therefore, the occurrence constraints should be loosened to allow the model to fit knowledge representation resources that fall along the various points along the semantic spectrum; from thesauri to controlled vocabularies to taxonomies and to ontologies.

Another prevailing observation was that it was sometimes difficult to find the right values to populate the metadata model for a terminology resource. For example it took one of the authors a full work day to populate values from LOINC for the whole OMV model. In some cases, the information was not readily available (e.g., not published or difficult to find). In other cases, although the information seemed to be reasonably aligned with the metadata description, it was divergent enough to lead to uncertainty about how appropriate a value was for a given metadata element. Perhaps better data was available elsewhere, or a specific value was different enough to suggest the creation of a new metadata element to satisfy a perceived gap in the model. As mentioned earlier, the ontology owner or developer is best suited to seed and maintain the metadata information. This mitigates much of the concern about gaps in knowledge by a curator or the appropriateness of populated values.

B. Ontology Evolution

Ontology evolution has been defined as the "timely adaptation of an ontology and consistent propagation of changes to the dependent artifacts" [28]. As terminology resources increase in size or complexity, management of dependencies becomes increasingly challenging. Metadata about and/or copies of a resource may occur in multiple registries and repositories. Changes to resource semantics can impact applications and other terminology resources that import and extend the changed resource. The formal change process needs to include a registry of known instances and dependencies. Additional elements such as "hasReferenceEntity" and "causeChange" (to identify likely impacts) were added to the OMV extensions to address this challenge. More work is needed to define metadata elements for representing resource semantics and granular changes to terminology content and enabling 'time travel'.

C. Future Work

NCBO and NCI's LexEVS CTS2 server implement a portion of the developed model. Future work should include (1) working with the wider community to validate the model for other terminologies and amend/extend as needed; and (2) developing a set of competency questions, e.g. for creating terminology search queries based on the OMV extension. A search engine based on this could allow users to search appropriate terminologies using a combination of the metadata elements, and perhaps even in combination with Ontology Recommender or a similar tool that suggests ontologies based on sample text input. This should be done in collaboration with the rest of the community, since resources like Biosharing are already working on a 'wizard' to help users find data standards that meet their needs.

VI. CONCLUSION

The ORWG conducted research to identify relevant terminology metadata models that could form the foundation for a standard ontology profile for use by NCI, NCBO, and NCRI. The OMV version 2.4.1 was selected as the base model. It was tested on LOINC, SNOMED-CT, RxNorm, and NCI Thesaurus, resulting in a revision of the OMV. The OMV extension, already partially implemented by NCBO and LexEVS/CTS2, could serve as the starting point for a terminology resource metadata standard in the biomedical research community, providing a framework for further work with other organizations active in this space such as OBO Foundry, RDA, or Biosharing. Even providing a standard set of basic metadata about terminologies would be of great value to help make terminology descriptions consistent across resources.

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OMV	Action	OMV Extensions	Comparable terms from Dublin Core Metadata
URI	N/A	URI	
name	Rename and Refine definition	fullName	dc:title
acronym	Rename and Refine definition	shortName	
description	Refine definition	description	dc: description
documentation	Refine definition	documentation	
reference	N/A	reference	
notes	N/A	notes	
keywords	N/A	keywords	dc:subject
status	Rename	developmentStatus	
creationDate	Remove and Addition (creationDate moves to Version.creationDate and versionDate is added here)	versionDate	dc:created
modificationDate	Remove (modificationDate moves to Version.modificationDate)		dc:modified
naturalLanguage	Refine definition	naturalLanguage	dc:language
numberOfAxioms	N/A	numberOfAxioms	
hasContributor	N/A	hasContributor	dc:contributor
hasCreator	Refine definition	hasCreator	dc: creator
usedOntologyEngineeringT ool	N/A	usedOntologyEngineeringTool	
usedOntologyEngineeringM ethology	N/A	usedOntologyEngineeringMethology	
conformsToKnowledgeRepr esentationParadigm	N/A	conformsToKnowledgeRepresentationParadigm	
endorsedBy	N/A	endorsedBy	
hasDomain	N/A	hasDomain	dc:hasDomain
isOfType	N/A	isOfType	
designedForOntologyTask	N/A	designedForOntologyTask	
hasFormalityLevel	N/A	hasFormalityLevel	
knownUsage	N/A	knownUsage	
hasOntologyLanguage	N/A	hasOntologyLanguage	
hasOntologySyntax	N/A	hasOntologySyntax	
resourceLocator	N/A	resourceLocator	
version	N/A	version	dc:hasVersion
hasLicense	N/A	hasLicense	dc:license dc:rights dc:rightsHolder
useImports	N/A	useImports	de.rightor10100
hasPriorVersion	Remove	*	
isBackwardCompatibleWith	N/A	isBackwardCompatibleWith	
isCompatibleWith	N/A	isCompatibleWith	
numberOfClasses	N/A	numberOfClasses	
numberOfProperties	N/A	numberOfProperties	
numberOfIndividuals	N/A	numberOfIndividuals	
	Addition	certifiedBy	
	Addition	mandatedBy	

Table 2: OMV Extensions