A User Centered Approach for Evaluating Biomedical Data Integration Ontologies

Gilbert Maiga

Department of Information Technology, Faculty of Computing and Information Technology
Makerere University, Kampala, Uganda
E-mail: gmaiga@cit.mak.ac.ug

Ddembe Williams

Department of Information Technology, Faculty of Computing and Information Technology Makerere University, Kampala, Uganda E-mail: d.williams@cit.mak.ac.ug

Abstract

There has been an emergence of various biomedical ontologies that seek to integrate data from the clinical and biological domains. These ontologies are attempts to relate clinical and biological data using various strategies to overcome issues of scope, process and differing levels of granularity. However, lack of knowledge about user needs for such biomedical ontologies, and absence of a general framework to assess their suitability for specific applications remain obstacles to their reuse and wide adoption in distributed computing environments. This paper describes a study that seeks to bridge this gap by proposing a framework to assess the suitability of a biomedical ontology for a user task. The framework draws on existing ontology evaluation approaches in relating user needs to biomedical ontology characteristics. Systems theory is used to explain the dynamics of a biomedical environment. The framework therefore includes feedbacks from the evaluation process to user characteristics of the integrated systems. This framework was validated by a study using structured interviews and questionnaires in a survey. The results indicate that it is sufficiently flexible for evaluating ontologies for biomedical data integration, while taking into account the conflicting needs of different users interested in accessing complex libraries of biomedical data.

Keywords: Biomedical Data Integration, Ontology Evaluation, User Evaluation Framework, Biological data, Clinical data, Systems theory

1. Introduction

The challenge of evaluating ontologies has become an important research and development endeavor for selecting an ontology to suit a given task, especially in distributed computing applications of the semantic web (Kalfoglou and Schorlmer, 2006). However, comparing ontologies remains challenging due to lack of standard frameworks for evaluating them, given that they differ by function, their expected input and outputs (Natalya and Musen, 2002). Existing frameworks evaluate ontologies largely on the basis of its taxonomy (Guarino and Welty, 2002). These find little use for assessing

biomedical ontologies that model function, processes and differing levels of granularity presented by biological and clinical data integration.

Philosophical Ontology is the science of what is, the kinds of structures and objects, properties, events, processes and relations in every area of reality, and broadly provides a definitive and exhaustive classification of entities in all spheres of being (Smith, 2003). Information systems ontology, to which biomedical ontology (BO) belongs, extends philosophical Ontology. Information systems ontology is an agreement about a shared, formal, explicit and partial account of a conceptualization consisting of classes, instances, relations, functions and axioms (Uschold and Gruuninger, 1996).

Biomedical ontologies integrate various sources of heterogeneous and rapidly changing biological and clinical data (Kohler *et al.*, 2003; Kumar *et al.*, 2006; Yugyung *et al.* 2006; Rey-Perez *et al.*, 2006; Sioutos *et al.*, 2006). They provide reusable models for integrating biomedical data in order to achieve interoperability between different sources, applications and groups. However, the reuse and wide adoption of BO for data integration in distributed computing environments remains constrained by: 1) lack of a general framework to assess their suitability for specific applications; and 2) lack of knowledge about user needs for such BO, since ontologies are subjective knowledge artifacts in terms of time, place and cultural environment, as reflected in their design (Alani and Brewster, 2006; EON, 2006). This underlines the difficulty in articulating specific properties to use in ranking ontologies since selection may depend on personal preferences of user requirements (Alani and Brewster, 2006).

In this study, a mixed method research strategy combining quantitative and qualitative deductive approaches was applied to identify user requirements for biomedical data integration systems. A deductive descriptive approach helped to identify theory and general properties of biomedical ontology evaluation. These were validated using structured interviews and questionnaires in a survey. The results are applied to the design of a framework for user assessment of biomedical ontologies. The framework is seen as a dynamic system, with feedbacks from the evaluation process guiding improvement of the integration ontology and re-specification of user requirements.

The rest of this paper is organized as follows. Section 2 discusses approaches to evaluation of information systems and ontologies. Section 3 presents a field study, its results the framework and its supporting theories. Conclusions and future work are given section 4.

2. Approaches to Information Systems Evaluation

The theory of evaluation is rooted in the twin ideas of accountability and social enquiry (Alkin and Christie, 2004). Thus Cronholm and Goldkuhl (2003) classify information systems evaluation approaches into formal rational, interpretive or criteria based, using respectively goal based, goal free and criteria based strategies. The formal rational evaluation approaches are largely quantitative processes, usually concerned with technical and economic aspects, employing goal based strategies. These strategies focus on intended services and outcomes, to achieve goals which can be phrased in quantitative or qualitative terms.

The interpretive evaluation approaches view information systems as social systems with embedded information technology. Similarly, Walsham (1993) argues that information systems evaluation should consider not only the purpose for the evaluation and associated factors, but also social context and process, and stress the need to consider evaluation as a learning process for all involved. Goal free evaluation strategies are appropriate in an interpretive approach, performed with limited evaluator involvement. In contrast criteria based evaluation use selected general qualities for evaluation where scores from multiple criteria are combined into an overall weighted sum (ibid).

The most appropriate evaluation approach depends largely on its context (Cronholm and Goldkuhl, 2003). Whatever the approach adopted, Alkin and Christie (2004) argue that evaluation models must consider *methods*, *valuing* and *use*. *Methods* deal with how the evaluation is to be done, and so focus on knowledge construction. *Valuing* concerns the role of the evaluator in assigning

criteria while *use* focuses on the purpose of the evaluation. These factors can be seen as the *process*, *people and purpose* that Ballantine *et al.* (2000) identify as drivers of information systems evaluation.

Information systems evaluation literature and practice have been closely linked with the technical project development cycle. For example, Beynon-Davis *et al.* (2004) propose a model for information systems evaluation that distinguishes between the strategic, formative, summative approaches, and post mortem analysis. Strategic evaluation is conducted as part of the planning process for an information system. Formative evaluation is a continuous, iterative informal process aimed at providing systematic feedback to developers of an information system (Remenyi and Smith, 1999). Summative evaluation assesses the worth of a project or program outcome in light of initially specified success criteria (*ibid*).

Ontology based biological and clinical data integration systems can also be seen as a type of information system, with systemic features that can be variously evaluated against user requirements. What distinguishes them from general information systems is arguably the dynamics of the environment in which they are used. Integrating the vast amounts of biological and clinical data require ontologies that can capture and represent new structure and processes that emerge with new data in the two domains. In a comparative evaluation of biomedical ontologies, the evaluator is the proposed user. Neither an interpretative nor a formal rational approach seems as appropriate as a criteria based approach in which the user's requirements motivate the evaluation criteria. Such an approach can be undertaken in a strategic, formative or summative evaluation framework.

2.1. The Challenge of Evaluating Biomedical Ontologies

There is no single unifying definition of what constitutes ontology evaluation (Gangemi *et al.*, 2005). Evaluation determines the quality and adequacy of an ontology for use in a specific context, and for a specific goal (Fernández, Cantador and Castells, 2006). It is a technical judgment of the contents of an ontology with respect to requirements specifications, competency questions, or a meta ontology as a frame of reference (Gangemi *et al.*, 2005; Gomez-Perez, 2004; Guarino and Welty, 2002).

Biomedical ontologies seek to integrate clinical and biological data so as to achieve interoperability (Kohler *et al.* 2003; Kumar *et al.*, 2006; Yugyung *et al.*, 2006; Rey-Perez *et al.*, 2006; Sioutos *et al.*, 2006). However, integrating biological and clinical objects across structure, function, processes and granularity using ontologies remains challenging, with no unifying approach against which they can be assessed by users (Pinto and Martins, 2000). There is also lack of knowledge about properties users require to select a suitable ontology for a task (Alani and Brewster, 2006; Kalfoglou and Schorlmer, 2003; Lambrix and Tan, 2006). This lack of a unifying framework for evaluating ontologies remains an obstacle for their reuse and adoption by industry and the wider web community (Alani and Brewster, 2006; Gangemi *et al.*, 2005; Kalfoglou and Schorlmer, 2006).

2.2. Current Approaches to Ontology Evaluation

Existing approaches to ontology evaluation use various contexts and conduct evaluation at different levels of complexity. A taxonomy of evaluation approaches based on type and purpose that adopts levels of vocabulary, taxonomy, semantic relations, application, syntax, structure and design is provided by Brank *et al.* (2005). This level based taxonomy categorizes existing ontology evaluation approaches (table 1) as follows:

- 1) Golden standard approaches that compare an ontology to a gold standard (Gomez-Perez 1994; Hovy, 2001).
- 2) Task based approaches that assess results after using the ontology in an application (Porzel and Malaka, 2004).
- 3) Data or corpus driven approaches that compare the fit of an ontology to domain texts (Brewster et al., 2004).
- 4) Assessment by humans to show how well the ontology meets a set of predefined criteria (Lozano-Tello and Gomez-Perez, 2004; Supekar, 2005).

	Evaluation Approach			
Evaluation level	Golden	Application	Data Driven	Human
L'aduation ic ver	Standard	Based	Data Dilven	Assessment
Lexical, vocabulary	X = used	X	X	X
Hierarchy	X	X	X	X
Semantic relations	X	X	X	X
Content application	X	X	Not used	X
Syntactic	X	Not used	X	X
Structure, architecture, design	Not used	Not used	Not used	X
Process, function, granularity (in biomedical ontology)	Not applied	Not applied	Not applied	Not applied

Table 1: A Taxonomy of Ontology Evaluation approaches (Brank et. al., 2005)

These approaches (table 1) are used by knowledge engineers for technical evaluations of the static knowledge of a domain represented by its taxonomy. Evaluating biomedical ontology requires determining the quality and adequacy of both static and dynamic functions, processes and granularity that they present (table 1). There is no unifying frame of reference against which such an evaluation can be conducted. This renders existing approaches inadequate. The proposed framework is motivated by the need to contribute towards developing a user focused evaluation framework for biomedical integration ontologies in such dynamic environments.

3. Deriving the Biomedical Evaluation Framework

A framework is a reusable design for all or part of a software system that is represented by a set of abstract classes and the way their instances interact (Fayad *et. al.*, 1999). It is structure within which something can be built. *Frameworks* with underlying ontological principles to support knowledge reuse are said to be ontology-based. In this study, framework refers to the interaction between user requirements and biomedical concepts in the process of selecting an ontology to fit an integration task. The evaluation framework is derived as follows: 1) identifying requirements for the framework from literature sources; 2) examine and select an ontology evaluation approach and theories that fit and explain such requirements; 3) validating requirements using a field study; 4) extending the selected ontology evaluation approaches and theories with requirements to derive a new evaluation framework.

3.1. Multi Criteria Ontology Evaluation

Multiple criteria approaches deal with the problem of selecting a good ontology from a given set based on defining several decision criteria or attributes. For each criterion, the ontology is evaluated and given a numerical score and an overall score computed as a weighted sum of its per-criterion scores (Brank *et al.*, 2005; Lozano-Tello and Gomez-Perez 2004). While this approach requires a lot of manual involvement by human experts, it allows a combination of criteria at many levels (Brank *et al.*, 2005). Ontometric is a multi criteria decision making method that helps knowledge engineers to determine the suitability of a particular ontology for their system requirements (Lozano-Tello and Gomez-Perez, 2004). It is a multilevel framework of 160 characteristics organized around a taxonomy with five basic dimensions of ontology content, implementation language, development methodology, the software tools used, and the cost for the project (ibid).

Ontometric is based on the analytic hierarchy process (AHP), a flexible decision making tool for complex multi criterion problems (Saaty, 1977). Its selection process has four steps namely; 1) decide upon the criteria for selection; 2) rate the relative importance of these criteria using pair-wise comparisons; 3) rate each potential choice relative to each other on the basis of each selection criterion by performing pair wise comparisons of the choices; 4) combine the ratings derived in steps 2 and 3 to obtain an overall relative rating for each potential choice. AHP allows users of Ontometric the flexibility to derive a suitable metric to use when selecting an ontology for a particular task. For each task, users compute a suitable quantitative metric against which ontology assessment and selection may

be conducted. This is an advantage as it provides flexibility when used to calculate a metric in a dynamic environment like biomedicine where new objects, processes and relations continuously emerge.

3.2. Contributions from Systems Theory

General systems theory (GST) is here adopted to explain the emergent properties from the complex and dynamic nature of data in biomedical integration systems (Huang *et al.* 2004). GST explains structure and properties of systems in terms of relationships from which new properties of wholes emerge that are not found among those of elements, and the corresponding behavior of the whole cannot be explained in terms of the behavior of the parts (Von Bertalanffy 1962). Concepts from the process of self organization (SO) may be used to extend systems theory. In SO, the internal organization of a system increases in complexity without being guided or managed by an outside source and displays emergent properties which do not exist if the lower level is removed (Gershenson 2006). In SO the environment is unpredictable and the elements interact to achieve dynamically a global function (*ibid*).

Engineered systems self organize by adaptation, anticipation, robustness or a combination of these features (Gershenson 2006). Self-organizing systems, rather than being a type of systems, are a perspective for studying, understanding, designing, controlling, and building systems; the crucial factor being the observer, who has to describe the process at an appropriate level and aspects, and to define the purpose of the system; SO can therefore be everywhere, it just needs to be observed (Heylighen and Gershenson 2003). Organization is structure that has a purpose. The observer has to focus their viewpoint, set the purpose of the system to see the attractor as an organized state at the right level and in the right aspect in order to observe self-organization - a perspective used to design, build, and control artificial systems. A key characteristic of an artificial self-organizing system is that structure and function of the system emerge from interactions between the elements (*ibid*).

3.3. The Field Study

A field study was conducted to validate user requirements for evaluating a biomedical integration system. A mixed method research strategy combining quantitative and qualitative deductive survey was used to determine the scope, inputs, processes, outputs for such a framework, from existing literature. For the survey 580 doctors and 50 biologists in Uganda were randomly selected from the study population. Structured interviews were used to pretest the questionnaire with 20 doctors and biologists before distribution to the selected sample. The questionnaire tested for the level of agreement with proposed properties of the evaluation framework among biologists and doctors. Correctly filled questionnaires were returned by 404 doctors and 46 biologists. The collected data was used to clarify user requirements for biomedical evaluation systems.

The statistical package for social sciences (SPSS) was used to analyze data for the level (%) of agreement with the proposed requirements of the framework. The validated requirements are applied in the design of the evaluation framework

3.4. The Results

3.4.1. The Users of a Biomedical Evaluation Framework

Scope refers to the users and uses of the evaluation framework. What are its uses? Who are its possible users and beneficiaries? The data collected from the field survey was used to provide answers for these questions. Table 2 indicates the level of agreement with the proposed users of such biomedical evaluation.

Table 2:	Users of a Biomedical Evaluation Framework

	Molecular Biologist		Medical Doctors		
User Category	No. of	Agreement Level	No. of	Agreement Level	
	Responses	(%)	Responses	(%)	
Molecular biologist	46	100	404	98	
Medical practitioner	46	93.9	404	100	
Health care managers	46	81.8	404	68	
Information systems developers.	46	72.7	404	57	
Legal practitioners	46	54.5	404	54	
Policy Makers	46	51.5	404	52	

Table 2 reveals that biologists, medical practitioners, public health and information systems managers as potential users of such a framework. Legal practitioners (54%) and policy makers (51%) are less likely to use the system.

3.4.2. Requirements for a Biomedical Evaluation Framework

The survey also tested the need for a biomedical evaluation system to enable users to: 1) visualize ontology structure and determine its relevance to for a task; 2) compare overlap between an ontology and a reference meta model; 3) determine the fit between an ontology and a task; 4) provide feedback to improve on existing ontologies or respecify requirements; 5) have a scope covering the entire clinical and biological fields; 6) be reusable across research groups and disciplines; 7) be flexible to accept data from various research groups and disciplines; 8) detailed for users to identify and select properties that fit a task; 9) relate concepts that require aggregation; 10) have a reference ontology. Table 3 shows the level of agreement with the proposed requirements of the framework.

Table 3: Requirements for the evaluation Framework

Requirement	Molecular Biologist		Medical Doctors		
Requirement	No. of responses	Agreement Level (%)	No. of responses	Agreement Level (%)	
Visualize structure	46	91	404	74	
Relevance of concepts	46	91	404	85	
Compare to meta ontology	46	68	404	72	
Fit ontology to task	46	90	404	81	
Provide Feedback	46	90	404	91	
Reusability	46	94	404	77	
Flexible input	46	94	404	78	
Wide Scope (Generic)	46	85	404	73	
Adequate details	46	88	404	75	
Model aggregation (granularity)	46	88	404	68	
Reference meta model	46	72	404	53	

The results in table 3 show that a biomedical integration system needs to be flexible enough to cater for differences in data formats and reporting requirements across research groups and disciplines. It also needs to contain sufficient detail for its properties to be easily recognized. A meta-language or theory models an unbounded universe of discourse. The integration model needs to be based on a meta-language with an unbounded universe of discourse (UOD). Granularity is a measure of size of components in a system. Levels of granularity are synonymous with levels of complexity. The results of this study indicate preference for biomedical integration systems that are able to bridge data across levels of granularity for biological and clinical sources. The results also reveal that evaluations of biomedical data integration systems should allow users to: visualize the ontology structure; compare the degree of overlap between user needs and the ontology; determine relevance of an ontology for a given task and provide feedback to improve existing models and help re-specify user requirements. It should also have a wide scope across biological and clinical fields; be reusable and cater for data integration across biomedical research groups; provide the flexibility to accept input of new biological

and clinical data across research groups and disciplines; and have sufficient detail users to easily recognize the important properties that make it suitable for their particular task.

3.4.3. A Reference Meta Model for Biomedical Data Integration

The requirements (Table 3, rows 3 and 11) indicate a need for a meta model (ontology) to represent biomedical knowledge during evaluation. Such a meta model acts as a frame of reference against which evaluations are conducted. In the field study, data was collected on the uses of biomedical integration systems. The results from the analyzed data (Table 4) were then used to informally specify requirements for such a meta model, as presented in Table 5. Deriving the informal specification of the meta model is guided by methodologies for ontology construction that emphasize the requirement for flexibility (Table 3, row 7) in formalizing knowledge and use of competency scenarios (Gruninger and Fox, 1995; Uschold and Gruninger, 1996). Competence scenarios (Table 4 and 5) help to re scope the meta ontology by identifying its major motivating applications as given in Table 5. Competence scenarios are used to extract the main concepts and relations of the meta ontology (Gomez-Perez, 2004). Competence scenarios and the resulting biomedical meta model act as requirements specification against which an ontology can be evaluated (ibid).

Table 4: Competence areas (Uses) for a Biomedical Integration System

	Molecular Biologist		Medical Doctors	
Competence Scenario	Number of	Agreement	Number of	Agreement
	Respondents	Level(%)	Respondents	Level
Relate a genetic profile to a patient	46	91	404	91
Relate clinical history to a patient	46	97	404	97
Relate patient gene profile to a disorder	46	91	404	91
Relate a persons trait to a genetic profile	46	91	404	91
Relate genetic profile to characteristics	46	91	404	91
Relate genes to development of proteins tissues and organs	46	91	404	91
Relate organs to a genetic disorder	46	85	404	85
Determine gene prevalence in population	46	88	404	88
Determine disorder prevalence in population	46	90	404	90

The competence scenarios, concepts and relations identified for such a Meta-ontology are given in table 5. The concepts and relations are the main features of the derived meta-model for a biomedical ontology for integrating biological and clinical data.

Competence Scenario	Biological Objects	Clinical Objects	Object Relations	
Genetic profile to a patient	Genetic profile	Patient	Patient has genetic profile	
Clinical history to a patient		Clinical history, patient	Patient has clinical history	
Patient gene profile linked to	Gene	Patient, Disease, Disorder	Patient <i>has</i> gene profile	
disorder			Gene cause of disorder	
Persons trait linked to a genetic	Genetic trait, genetic	Person	Person may have trait	
profile	profile		Genetic profile cause of trait	
Role of genes to development	Gene, Protein dev,		Gene cause of Prot. Dev.	
of proteins tissues and organs	Protein, tissue, organ		Protein <i>part of</i> tissue	
			Tissue part of organ	
Tissues or organs affected by a	Tissues, organs	Disorder, disease	Tissues may have disorder	
genetic disorder			Organ may have disorder	
Prevalence of gene in	Gene	Population	Population may have gene	
population		_	prevalence	
Prevalence of disorder in		Population, disorder,	Population may have disorder	
population		prevalence	prevalence	
Prevalence of gene in	Gene	Population	Population may have gene	
population			prevalence	
Prevalence of disorder in		Population, disorder,	Population may have disorder	
population		prevalence	prevalence	

Table 5: Informal Specification of the Meta-ontology using Competence Scenarios

The informal specification in Table 5 helps to reveal properties important for a target biomedical Meta ontology. *Processes* (e.g. disease, disorder) and *non-prosessual* (static) entities (e.g. organ) are shown as distinct types useful for structuring knowledge in the biomedical domain. *Intra* and *trans* domain relationships within and between biological and clinical classes used to structure knowledge are also revealed. *Intra* domain relationships are between either biological objects (e.g. Gene cause_of Protein dev.), or clinical objects. Trans domain relations are between clinical and biological objects. *Trans* domain relationships may be used to relate dynamic objects (e.g. Population may_have disorder prevalence). The results also reveal that these trans domain relations may be used to model biomedical objects at different granular levels (e.g. Patient has gene). Relations between processes and non-processual types are also revealed (e.g. Organ may_have Disorder). The informal specification of the meta-ontology using competence scenarios in table 5 results therefore helps to reveal that a biomedical integration meta model should be able to:

- i. Represent and relate biomedical structure (i.e. biological and clinical objects and their relations).
- ii. Represent and relate dynamic biomedical processes and functions associated with clinical and biological data.
- iii. Model the intra and trans domain relationships between biological and clinical data
- iv. Relate biological and clinical data across levels of granularity (aggregation).

Structure (objects and relations), processes, functions and representation of granularity are properties that need to be captured in the design of a biomedical integration meta model (ontology). The meta model acts as a frame of reference for assessing other biomedical integration systems. Its representation of structure, granularity and processes are therefore important properties to be considered during evaluation.

3.4.4. Summary of Requirements for the Framework

The analysis points to the following as key user requirements for a framework to evaluate a biomedical ontology:

1) Reusability and wide scope. The framework is useful for assessing and selecting ontologies for integrating data useful to biologists, doctors, and public health care and information systems managers.

- 2) Flexibility. The framework should enable users to assess and select a biomedical ontology that relates existing and emergent genetic to corresponding patient clinical data and processes. The evaluation process also requires the flexibility to accept input of new biological and clinical data, across research groups and disciplines
- 3) The process of evaluation should enable a user to visualize ontology structure, determine preference for a given task and compare the degree of overlap between the integration ontology and user needs. It should be detailed enough for users to easily recognize the important properties that make it suitable for their particular task.
- 4) It should provide users with feedback to improve existing models and help re-specify user requirements where necessary.
- 5) Evaluation needs to be conducted against a Meta model representing structure, process and granularity as a frame of reference.

In the design of the evaluation framework for biomedical ontology integration, these requirements are captured.

3.5. The Evaluation Framework

This section brings together the requirements for biomedical data integration into one coherent evaluation framework by extending existing theories on systems and evaluation. The choice of approach and theory to extend is informed by their fit to requirements. For a comparative evaluation of ontology based biomedical data integration systems with the evaluator as proposed user (as is the case with the present study), a multi criteria evaluation approach is adopted for the design of the evaluation framework. Systems theory is used to explain the dynamics of the evaluation environment.

Existing approaches to ontology evaluation define standards used to assess static knowledge in a domain of discourse using specified criterion (Lozano-Tello and Gomez-Perez, 2004). In biomedicine, the environment is dynamic with new user requirements, processes and relations emerging with the rapidly increasing data. As revealed by the results of the survey, evaluation of biomedical data integration system requires a flexible and reusable system able to accept input of new across research groups and disciplines. A meta-ontology is required as frame of reference for evaluation, with feedbacks to improve existing models and help re-specify user requirements. Visualization of ontology structure helps to compare and determine the fit between an ontology and a task. To integrate these requirements, the proposed framework adopts a criteria-based approach in which user needs motivate the assessment criteria in a formative evaluation. This is found appropriate as it aims to provide systematic feedback to ontology designers and implementers, influencing the process of development and the final integration system, as earlier pointed out by Kumar (1990).

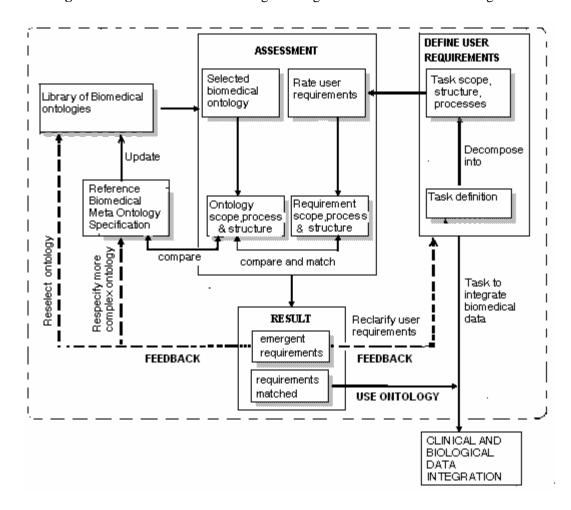


Figure 1: Framework for Evaluating Ontologies for Biomedical Data Integration

A framework for biomedical data integration should be flexible, reusable, and have a metaontology as frame of reference for evaluation. To realize these requirements, the proposed framework (Figure 1) extends the Ontometric evaluation method (Lozano-Tello and Gomez-Perez, 2004). Ontometric, based on the analytic hierarchy process (AHP), gives users the flexibility to select the hierarchy for the decision criteria (metric) to be used in evaluations (Saaty, 1977). Such flexibility enables the framework to be reused by different categories of users selecting ontologies for different tasks. Flexibility of the evaluation framework allows different categories of users to iteratively search through an ontology library using multiple criteria. This is more likely to result into: 1) selection of an appropriate ontology for a given task or 2) re-specification of new requirements for an ontology to fit the task. Reference ontology, specified in the requirements for this framework is also used by the Ontometric method. However the reference meta ontology in the new evaluation framework defines process and granularity, important properties for biomedical data integration systems. The framework is the basis for an evaluation tool (under development). To achieve this, the requirements are organized (figure) into dimensions of: user requirements definition, a biomedical ontology library, a reference meta ontology, assessment tool and feedbacks. Assessment involves visualization of ontology structure and comparing the overlap between an ontology and a task using parameters defined in the reference meta model.

The evaluation framework requires feedbacks to improve existing ontology models and to help re-specify user requirements. The dynamic environment of biomedicine requires reusing, extending and modifying existing ontologies to accommodate emerging types of data, processes and relations. This avoids the huge effort of starting or building entirely new ontologies (Alani and Brewster 2006). The proposed evaluation framework (Figure 1) adopts a systems approach so that domain or user needs are considered when pruning, extending or modifying an existing ontology. Systems theory is used to

explain the emergent requirements and feedbacks to improve upon existing models or redefine user requirements. Systems theory helps to explain the emergent properties from the evaluations that are iteratively used to re-specify, extend and modify existing biomedical ontologies into more complex ones or to redefine user requirements.

3.6. Using the framework

The framework guides users to select an appropriate ontology for a biomedical data integration task via the following steps:

- 1. Define the task that requires biological and clinical data integration
- 2. Decompose the task into a set of requirements
- 3. Define the biological and clinical objects, and relations for the task
- 4. Rate the relative importance of these objects and relations for the integration task
- 5. Select and Visualize the ontology to be assessed
- 6. Compare the ontology to a reference biomedical ontology
- 7. Compare the integration ontology to requirements (selection criteria)
- 8. Identify unmatched (emergent) requirements for use in re-scoping the meta ontology or user needs.
- 9. If ontology fits requirements, recommend it for the integration task or process.

4. Conclusions and Future Work

The literature suggests an important need to evaluate ontologies for use in building distributed computing applications. The paper identified changing structure, processes and relations with increasing biological and clinical data as major challenges faced by existing frameworks in evaluating biomedical ontology. Requirements for a flexible new framework to evaluate such ontologies are identified using a field study. Flexibility, generecity, reusability, a reference Meta model, and iterative feedbacks during evaluation are identified as key requirements for such a framework. The requirements are used to articulate a new framework for evaluating ontologies for biomedical data integration based on biomedical process, relations and treatment of granularity.

The new framework extends OntoMetric (Lozano-Tello and Gomez-Perez, 2004), a flexible multi criteria ontology evaluation method. It therefore brings the flexibility offered by Ontometric, when defining a metric based on user requirements to biomedical ontology evaluation. Systems theory is used to explain the complexity of new ontologies when emergent requirements are used to re-specify and extend existing ones. It can therefore help to derive requirements for iteratively and incrementally extending and modifying existing biomedical ontology structure to suit changing user needs. This helps to avoid the huge effort of starting or building entirely new ontologies. Steps for using such a framework are also outlined.

Work is ongoing to build a target biomedical Meta ontology as a frame of reference for use by this framework. This framework is the basis for a tool being developed, as an application to enable users to assess and select a suitable biomedical data integration ontology for their particular use case from a library of biomedical ontologies.

References

- [1] Alani, H. And Brewster, C. 2006. Metrics for Ranking Ontologies. In *Proceedings of the 4th International Conference on Evaluation of Ontologies for the Web*. EON Workshop, Edinburgh International Conference Center. Edinburgh. May 22nd 2006.
- [2] Alkin, M.C. and Christie, C.A. 2004. An evaluation theory tree. In M.C. Alkin Ed., *Evaluation Roots*. Thousand Oaks, CA: Sage. Chapter 2, 12-54.
- [3] Avison, D., Horton, J., Powell, P., and Nandhakumar, J. 1995. Incorporating Evaluation in the Information Systems Development Process. In *Proceedings of the third evaluation of Information Technology Conference*. Henly, UK.
- [4] Ballantine, J., Levy, M., Martin, M., Munro, I., and Powell, P. 2000. An Ethical Perspective on Information Systems Evaluation. *International Journal of Agile Management Systems*. 23, 233-241.
- [5] Beynon-Davis, P., Owens, I. and Williams, D.M. 2004. Information systems Evaluation and the Information Systems Development Process. *The Journal of Enterprise Information Management*. 17, 276–282.
- [6] Brank, J., Grobelnik, M. and Mladenic, D. 2005. A Survey of Ontology Evaluation Techniques. *At the conference on Data Mining and Data Warehouses* SiKDD.
- [7] Brewster, C., Alani, H., Dasmahapatra, S., and Wilka, Y. 2004. Data Driven Ontology Evaluation. *Proceedings of the Language Resources and Evaluation Conference* LREC 2004, Lisbon, Portugal.
- [8] Cronholm, S. and Goldkuhl, G. 2003. Strategies for Information Systems Evaluation: Six Generic Types. *Electronic Journal of Information Systems Evaluation*. 62, 65 -74.
- [9] Crosby, P.B., 1988. *Quality is Free: The Art of Making Quality Certain*, New York: McGraw-Hill, 1988.
- [10] Davidson, S., Overton, C. and Bunan, P. 1995. Challenges in Integrating Biological Data Sources. *Journal of Computational Biology*. 24.
- [11] Ding, Y. and Foo, S. 2002 Ontology research and development, Part 2. A Review of Ontology mapping and evolving. *Journal of Information Science*, 28, 375 388.
- [12] EON. 2006. Evaluation of Ontologies for the Web, 4th International EON Workshop located at the 15th International World Wide Web Conference, Edinburgh International Conference Center, Edinburgh, United Kingdom, Available: http://km.aifb.uni-karlsruhe.de/ws/eon2006. Accessed May 25th, 2006.
- [13] Fayad, E.M., Schmidt, D.C., and Johnson, R. 1999. Chapter 1 in Building Application Frameworks: Object-Oriented Foundations of Framework Design. New York: John Wiley \& Sons, September 1999
- [14] Fernandez, M., Cantador, I., and P Castells. 2006. CORE: A Tool for Collaborative Ontology Reuse and Evaluation.
- [15] Gangemi, A., Catenacci, C., Ciaramita, M. and Lehmann, J. 2005. A theoretical framework for ontology evaluation and validation. *In Proceedings of SWAP 2005*.
- [16] Garvin, D.A., 1984. What does 'Product Quality' Really Mean?, *Sloan Management Review*, fall, 1984.
- [17] Gershenson, C. and Heylighen, F. 2003. When can we call a system self-organizing? *In Advances in Artificial Life, 7th European Conference*, and ECAL 2003 LNAI 2801
- [18] Gershenson, C. 2006. A General Methodology for Designing Self-Organizing Systems. *ECCO working paper*. Available: http://uk.arxiv.org/abs/nlin.AO/0505009
- [19] Gomez-Perez, A. 1994. Some ideas and Examples to Evaluate Ontologies. Technical Report KSL-94-65. Knowledge System Laboratory. Stanford University. Also in Proceedings of the 11 the Conference on Artificial Intelligence for Applications, CAIA94.
- [20] Gomez-Perez, A. 2004. Ontology Evaluation. Handbook on Ontologies. 251-274.
- [21] Gruber, T. 1995. Towards Principles for the Design of Ontologies Used for Knowledge Sharing. *International Journal of Human-Computer studies*, 43 5/6:907–928.

- [22] Gruninger, M. and Fox, M.S. 1995. Methodology for the Design and Evaluation of Ontologies. *In: Proceedings of the Workshop on Basic Ontological Issues in Knowledge Sharing*, IJCAI-95, Montreal.
- [23] Guarino, N. and Welty, C. 2002. Evaluating Ontological Decisions with OntoClean. *Communications of the ACM*. 45, No. 2.
- [24] Heylighen, F. and C. Gershenson. 2003. The meaning of self-organization in computing. *IEEE Intelligent Systems*, 72–75.
- [25] Hongzhan, H., Cathy, H., Anastasia, N., Zhangzhi H., Winona, C., and Barker, B. 2004. The iProClass integrated database for protein functional analysis, *Computational Biology and Chemistry*, 28. 2004:87–96.
- [26] Hovy, E. 2001. Comparing sets of semantic relations in ontologies. In Rebecca Green, Carol A Bean, and Sung Hyon Myaengeds., *Semantics of Relationships, chapter 6*. Dordrecht, NL: Kluwer.
- [27] Huang, H., Hu, Z., Suzek, B.E. and Wu, C.H. 2004. The PIR integrated protein databases and data retrieval System. *Data science journal*, 3:163.
- [28] Kalfoglou, Y. and Schorlmer, M. 2003. Ontology Mapping: The State Of The Art. *The Knowledge Engineering Review*, 181: 1–31.
- [29] Kalfoglou, Y. and Schorlmer, M. 2006. Issues with evaluating and using publicly available ontologies. In *Proceedings 4th International EON Workshop, Evaluating Ontologies for the Web*, Edinburgh, Scotland.
- [30] Kohler, J., Philipi, S. and Lange, M. 2003 Semeda: Ontology based semantic integration of biological databases. *Bioinformatics*, 19, 2420 -2427.
- [31] Kumar, K.1990 Post Implementation Evaluation of Computer based Information Systems CBIS: current practices, *Communications of ACM*, 33, 203-12
- [32] Kumar, A., Linayip, Y., Smith, B. and Grenon, P. 2006 Bridging the gap between medical and bioinformatics: An ontological case study in colon carcinoma. *Computers in Biology and Medicine* 36, 694–711
- [33] Lambrix, P. and Tan, H. 2006. SAMBO. A system for aligning and merging biomedical ontologies *Web Semantics: Science, Services and Agents on the World Wide Web*, 4 196 206
- [34] Lozano-Tello, A. and Gomez-Perez, A. 2004. Ontometric: A Method to Choose the Appropriate Ontology". *Journal of Database Management*. Special Issue on Ontological analysis, Evaluation.
- [35] Natalya F. N. and. Musen, M.A. 2002. Evaluating Ontology-Mapping Tools: Requirements and Experience. *Available: http://seer.nj.nec.com.* Accessed 6 may 2006.
- [36] Pinto, H. S., and Martins, J. P. 2000. Reusing Ontologies. In *Proceedings of AAAI 2000 Spring Symposium Series, Workshop on Bringing Knowledge to Business Processes*, AAAI Press.
- [37] Porzel., R. and R. Malaka. 2004. A task-based approach for ontology evaluation. Proc. of the 16th European Conf. on Artificial Intelligence (ECAI 2004), Valencia, Spain, August 2004.
- [38] Remenyi, D. and M. Smith 1999. "Maximize Information Systems Value by Continuous Participative Evaluation." Logistics Information Management 12(1/2): 14-31.
- [39] Rey-Perez, D., Maojo, V., Garcia-Resal, M., Alonso-Calvo, R., Billhardt, H., Martin-Sánchez, F., and Sousa, A. 2006. ONTOFUSION: Ontology-based integration of genomic and clinical databases. *Computers in Biology and Medicine*, 36, 712-730.
- [40] Saaty, T. 1977. "A Scaling Method for Priorities in Hierarchical Structures". Journal of Mathematical Psychology, Vol.15, (1977) 234–281.
- [41] Smith, B. 2003. Ontology. *Blackwell Guide to the Philosophy of Computing and information*, Oxford: Blackwell, 55- 166.
- [42] Sioutos, N., Coronado, S., Haber, M., Hartel, F., Shaiu, W., and Wright, W. C. 2006. NCI thesaurus: A semantic model integrating cancer-related clinical and molecular information. *Journal of Biomedical Informatics*.

- [43] Supekar, K. 2005. A peer-review approach for ontology evaluation. *Proc.8th Intl. Protégé Conference*, Madrid, Spain, July 18–21, 2005.
- [44] Ushold, M. and M. Gruninger. 1996. Ontologies: Principles, methods and applications, in *The Knowledge Engineering Review*, 112, 93–55.
- [45] Von Bertalanffy, L. 1962. General system theory A Critical Review. *General Systems* 7, 1-20.
- [46] Walsham, G. 1993. Interpreting Information systems in organizations. Wiley, Chichester.
- [47] Yugyung, L., Supekar, K. and Geller, J. 2006 Ontology Integration: Experience with medical Terminologies. *Computers in Biology and Medicine*, 36, 893 919.