Abstract

Several ontologies attempting to combine biomedical data using various strategies in order to overcome issues of scope and differing levels of granularity have emerged. However, these Ontologies are little used in distributed applications. This is attributed to both the lack of knowledge about user needs for such systems and the absence of a general framework to assess their relative suitability for specific applications. In this paper, a framework to address this problem by relating user needs to ontology characteristic is proposed. Systems theory is used to explain the dynamics of a biomedical environment by providing feedbacks from the evaluation process to the user needs for integration systems. The framework has been validated using a descriptive field study. The results indicate that it is sufficiently flexible for evaluating ontology based biomedical integrated systems, taking into account the conflicting needs of different users interested in accessing complex libraries of biomedical data.

KEYWORDS: Biomedical ontology integration; ontology evaluation; evaluation framework; Systems approach; User evaluation

1. Introduction

The challenge of evaluating and selecting a suitable ontology for a given integration task, especially in distributed computing applications of the semantic web has become an important research and development endeavor. Comparing the effectiveness of ontologies remains challenging due to lack of standard frameworks for evaluating them, given that they differ in function, and in their expected input and outputs [5][21]. Existing frameworks evaluate ontologies largely on the basis of the taxonomy. These are therefore not suited for assessing granularity and processes presented by biomedical ontologies. There is also lack of knowledge about properties users require when selecting an ontology for a task [1] [14] [19]. As a result, reuse and wide adoption of biomedical ontologies by industry is not yet realized [6] [15].

Biomedical ontologies (BO) bring together disparate sources of heterogeneous biomedical data [16] [18] [26] [23]. They provide reusable models for integrating biomedical data in order to achieve interoperability between different sources. However, the wide use of BO’s in distributed applications is constrained by: 1) lack of general frameworks to assess their suitability for specific applications; 2) lack of knowledge about user needs for BO’s since ontologies are subjective knowledge artifacts in terms of time, place and cultural environment, as reflected in their design [1]. This underlines the difficulty in articulating specific properties to use in ranking ontologies since selection can depend on personal preferences of user requirements (ibid).

This study used a mixed method research strategy combining quantitative and qualitative deductive approaches to derive requirements for the framework. A deductive descriptive approach helped to identify theory and general properties of biomedical ontology evaluation. These were investigated using structured interviews in a survey. The results are used to derive a framework for assessment of biomedical ontologies. The framework is seen as a dynamic system, with feedbacks from the evaluation process guiding improvement of the integration ontology.

In this paper, section 2 discusses work on evaluating biomedical ontologies. Section 3 presents a field study and its results. Multi- criteria evaluations and systems theory are then presented as the underpinning concepts for deriving the framework. Conclusions from the study are drawn in section 4.

2. Evaluating Biomedical Ontologies

Biomedical ontologies integrate clinical and biological data so as to achieve interoperability between different sources [23] [16] [26] [18] [24]. Despite these efforts, integrating biomedical objects
across structure and processes using ontologies remains challenging. There is no unifying approach for user assessment of biomedical ontologies.

Ontology evaluation is a criterion based technical judgment for both the integrated and resulting ontologies [22]. Evaluation uses assessment criteria for any ontology namely: completeness, conciseness, consistency, expandability, robustness and clarity [9] [10]. Ontologies may also be assessed by user ratings and reviews, and for general ontological properties. Evaluation is done during design, development and prior to use [15]. It is important if ontologies are to be widely reused in distributed applications (ibid). The lack of a unifying framework for biomedical ontology evaluation therefore remains an obstacle for their reuse and adoption by industry and in web applications [1]. Despite its importance, little documented work on evaluating biomedical ontologies exists.

2.1. Existing Approaches to Ontology Evaluation

Based on type and purpose [2] provide the following ontology evaluation approaches:
1) Golden standard approaches. Evaluation is against a gold standard ontology or other representation of the problem domain for which an appropriate ontology is needed [9] [12].
2) Task based approaches. These assess the quality of results after using the ontology in an application [28].
3) Data or corpus driven approaches. Evaluate the ontology’s congruence with domain texts [3].
4) Assessment by humans to show how well the ontology meets a set of predefined criteria [20].

These approaches evaluate an ontology based on its taxonomy. They find little use for assessing biomedical ontologies that present both static and dynamic processes and relations. There is also scarcity of knowledge about properties users require when judging the suitability of a biomedical ontology [1] [14] [19]. As a result, adoption and reuse of biomedical ontologies by industry is not yet realized [6] [15]. This work is motivated by the need to contribute to evaluating biomedical ontologies, so they can gain wide use in web applications.

2.2. Multi Criteria Ontology Evaluation

These approaches help to select a good ontology from a given set based on several decision attributes. For each attribute, the ontology is evaluated and given a numerical score and an overall score computed as a weighted sum of its per-criterion scores [2] [20]. The approach however requires a lot of manual involvement by human experts [20].

Ontometric [20] is an example of a multi-criteria decision making method used by knowledge engineers to select an appropriate ontology among various alternatives using dimensions of cost, content, language, methodology and software development environment used. The selection criteria is based on a four step analytic hierarchy process (AHP) [27] namely: 1) Decide the criteria for selection; 2) Rate the relative importance of these criteria using pair-wise comparisons; 3) Rate the potential choice relative to the other based on each selection criterion, achieved by pair wise comparisons of the choices; 4) Combine the ratings derived in 2 and 3 to obtain an overall relative rating for each potential choice. However, Ontometric [20] is not sufficiently flexible for evaluating biomedical ontologies that take into account the conflicting needs of different users interested in accessing complex libraries of biomedical data.

3. Deriving the Evaluation Framework

The framework is derived in the following steps: 1) Determining requirements from the literature search and field study; 2) Selecting an ontology evaluation approach and theories that explains most requirements; 3) Extending the selected ontology evaluation approach using the requirements.

3.1. Determining Requirements (Field Study)

A mixed method research strategy combining quantitative and qualitative deductive survey was used to identify requirements for the evaluation framework. Existing literature was used to determine possible scope, functions, inputs, outputs for such a framework. These properties were tested for agreement by potential users in a field survey.

In the survey 580 doctors and 50 biologist in Uganda were randomly selected from the study population. Structured interviews were used to pretest the questionnaire using twenty doctors and biologists before distribution to the selected sample. The questionnaire tested for the level of agreement with proposed characteristics of the framework. It collected data on scope, inputs, processes and outputs for such evaluations. Correctly filled questionnaires were returned by 404 doctors and 46 biologists. The collected data was used to clarify user requirements for biomedical evaluation systems.

3.2. The Results
The statistical package for social sciences (SPSS) was used to analyze the collected data for the level (%) of agreement with the proposed user requirements. The requirements are used in the framework design.

3.2.1. Scope and Requirements for the Framework

The study determined scope (uses & users) of the framework. Table 1 indicates the level of agreement with the proposed users of the framework evaluation.

Table 1.
Users of a biomedical evaluation framework

<table>
<thead>
<tr>
<th>User category</th>
<th>Molecular biologist</th>
<th>Medical doctors</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Respondents</td>
<td>Agree (%)</td>
</tr>
<tr>
<td>Biologist</td>
<td>46</td>
<td>100</td>
</tr>
<tr>
<td>Medical clinicians</td>
<td>46</td>
<td>93.9</td>
</tr>
<tr>
<td>H/care manager</td>
<td>46</td>
<td>81.8</td>
</tr>
<tr>
<td>Systems develop</td>
<td>46</td>
<td>72.7</td>
</tr>
<tr>
<td>Legal Practice</td>
<td>46</td>
<td>54.5</td>
</tr>
<tr>
<td>Policy Makers</td>
<td>46</td>
<td>51.5</td>
</tr>
</tbody>
</table>

The results (table 1) reveal that biologists, medical practitioners, public health and information systems managers are potential beneficiaries/users of such evaluation system. Legal practitioners (54%) and policy makers (51%) are less likely to use the system.

The results of the study revealed requirements for the evaluation framework. To determine requirements, the survey tested for agreement on the need for the following aspects during evaluation:
1) Visualization of ontology structure
2) Compare the overlap and fit between user needs and the ontology.
3) Provide user feedback to improve upon existing models and help re-specify user requirements
4) Generic to cover the entire biological and clinical domains.
5) Reusable across research groups and disciplines
6) Flexibility in order to accept input of new data from various research groups and disciplines.
7) Sufficient details for users to easily identify useful properties and select a model suitable for their particular task.

Table 2 shows the level of agreement with proposed processing and output requirements of the framework.

The results reveal that most respondents (>70%) agree that as a requirement, a framework for evaluating a biomedical integration system should allow users to: Visualize the ontology structure; determine relevance of ontology for a given task; Compare the degree of overlap and fit between user needs and the ontology; provide feedback to improve existing models and help re-specify user requirements; have wide scope covering the entire clinical and biological 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.

3.3. Summary of Requirements

The analysis points to the following as key user requirements for evaluating a biomedical ontology:
1) Reusability and wide scope. The framework is useful for assessing and selecting ontologies for integrating data useful to different users namely: biologists, doctors, and public health care and information systems managers.
2) Flexibility. The evaluation process needs the flexibility to accept input of new biological and clinical data, across research groups and disciplines.
3) The process should enable visualization of ontology structure in order to determine its fit to requirements.
4) Provide user feedback to improve upon existing models and help to re-specify requirements.
5) Be detailed enough for users to easily recognize importance properties that make it suitable for their particular task.
These requirements are captured in the design of the evaluation framework. The framework requires theories that combine and represent these requirements. The choice of approaches for representing these requirements is thus informed by its underlying principles.

3.4. Contributions from Systems Theory

Systems theory 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 [25]. 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 [8]. SO 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 [7] [11]. The observer sets 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 SO. A key characteristic of an artificial self-organizing system is that structure and function of the system emerge from interactions between elements (ibid). This perspective of SO systems is adopted for this study to support a framework for evaluating BOIS. Such a framework needs to account for the conflicting needs of different users interested in accessing complex libraries of biomedical data using ontology based systems.

3.5. The Framework

Existing ontology evaluation approaches are standards for structuring and integrating knowledge in a domain of discourse using specified criteria. Such criteria are often combined and used for user assessment and selection of an ontology appropriate for a given task [20]. In biomedicine, where new data objects emerge and new user requirements emerge with the rapidly increasing data, flexible models that enable users to iteratively search through an ontology library using multiple criteria are 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. This framework is an attempt to evaluate biomedical ontology integration systems with the evaluator as the proposed user. A criteria-based approach in which the user’s requirements motivate the assessment criteria in a formative evaluation [17] framework is found appropriate as it aims to provide systematic feedback to designers and implementers, influencing the process of development and the final integration ontology.

This framework (figure 1) extends the Ontometric method for deciding the suitability of a particular ontology for a project [20]. Ontometric, based on the AHP framework [27] allows ontology engineers and domain experts the flexibility to select the hierarchy for the decision criteria to be used in evaluations. It however offers no specific features that support evaluating systems in dynamic environments like biomedicine that require reusing, extending and modifying existing ontologies to accommodate new types of data objects and avoid the huge effort of starting or building entirely new ontologies [1]. The evaluation framework as proposed in this study (figure1) mitigates this shortcoming by adopting a systems approach so that domain or user needs are considered when pruning, extending or modifying an existing ontology.

Ontology scope, processes and are used to define the inputs to framework. These are used to assess the fit between user requirements and the selected ontology. Visualization of biomedical structure, comparing the fit between user needs and an ontology are done during assessment. Systems theory is used to explain flexibility of this framework via the feedbacks to improve upon existing models or redefine user requirements (to be captured in an evaluation tool). The overlap between user needs and the ontology determines the fit between the two. This expresses how well the biomedical integration system conforms to user requirements (i.e. completeness). Completeness is adopted as a metric for the fit between requirements and a selected ontology.

3.6. Using the Framework

The framework guides users to select an appropriate biomedical ontology for a task via the following steps: 1) Determine task requirements ; 2) Rate the relative importance of these requirements; 3) Select the ontology to be assessed; 4) match the ontology to requirements; 5) Identify emergent requirements ; 6) Use emergent properties to re-specify an ontology or user needs, else matched ontology is recommended for the integration process.
4. Conclusions and Future Work

The literature shows the important need for evaluating ontologies used in building distributed computing applications. The paper identifies the challenges faced by existing frameworks in evaluating biomedical ontologies. Requirements for a flexible new framework to evaluate such ontologies are identified using a field study. Flexibility, reusability, wide scope, multiple criteria and the need for iterative feedbacks during evaluation are identified as key requirements for the framework. These requirements are used to extend OntoMetric [20], in deriving the new framework, using systems theory. The framework enables users to assess and select a suitable biomedical ontology for their particular use case when accessing ever-increasing libraries of biomedical data. Steps for the utility of such a framework are also given. The novelty of this framework lies in the ability to relate ontology structure and user derived objectives in order to derive requirements used to iteratively and incrementally improve on existing biomedical ontologies. This is achieved in environments where the amounts of data are ever increasing and user needs are changing.

Work is ongoing to build a target biomedical Meta ontology as a frame of reference for use when using this framework. A tool with metrics is also to be developed as an application of this framework.

References


