A Framework for Evaluation of Biomedical Ontologies based on

Structural Components

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Submitted to the Department of Computer Engineering in fulfillment of the requirements for the degree of

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COLLEGE OF ELECTRICAL & MECHANICAL ENGINEERING NATIONAL UNIVERSITY OF SCIENCES AND TECHNOLOGY ISLAMABAD JULY, 2015

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This thesis has been read by an English expert and is free of typing, syntax, semantic, grammatical and spelling mistakes. Thesis is also according to the format given by the university.

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ABSTRACT

Biomedical ontologies are being developed in a large amount since the last decade. Recently, due to abrupt expansion in biological data and knowledge in the form of biomedical ontologies, the attention of their usefulness increases in the research activities. Meanwhile, there exist some challenges of accurately building and maintaining ontologies so that their benefits of reusability in the respective knowledge fields and applications can be exercised. Further on due to the complex structure of ontologies and difficult terminologies of biomedical domain, the evaluation of these ontologies turns out to be a challenging task. It is utmost need of current ontology researchers and developers to evaluate the quality of these biomedical ontologies so that the applicability and reuse of these ontologies will be improved. In motivation of this need we have proposed a methodology of evaluating the quality of biomedical ontologies with respect to basic ontology structural building blocks like concepts, classes including subclasses, superclasses, instances/individuals of the classes, triple components including subjects, predicates and objects, properties including object properties, data properties, annotation properties, functional properties, symmetric properties, asymmetric properties and reflexive properties. When domain knowledge is combined with these mentioned above structural components, it gives rise to a well structured and elaborative ontology. The experimental results explain obvious variations of structural components the candidate biomedical ontologies of same domain.

Key Words: *Biomedical ontology, ontology evaluation, SPARQL queries, Protégé, Ontology Web Language (OWL).*

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CHAPTER 1: INTRODUCTION

1.1: Ontology

Ontology is a precise formal specification of domain conceptualization [1]. In history ontology means the metaphysical study of the nature, and fundamental characteristics and relations of all individuals [2]. In the area of information sharing, ontologies play a significance role since 1990s [23]. Ontology can be considered as a declarative graphical model that illustrates the domain concepts, their characteristics and the affiliation among them [20]. These actually grasp the real world information. Guarino [3] describes ontology as it is approximate domain specifications and the degree of approximation is depicted by ontology evaluation as depicted by Figure 1.1.



Figure 1.1: Notion of Real World, Ontology and Ontology Evaluation

Ontologies can be interpreted as graphs which represent domain conceptualization and its additional information depicts their usage perspective [5]. Ontology serves as a data structure for knowledge abstraction in various fields of study. In the area of biomedical classifications, ontologies symbolize the basic concepts which are used in a specified biomedical domain along with their inter-relationships. The key perspective of ontologies is their structure and their capability to act accordingly for the desired purpose [23].

1.2: Structure of Ontologies

Ontologies are fundamentally taxonomic tree of conceptualizations in which generic concepts are placed at the peak or highest level of hierarchical tree and specialized concepts reside at the ground level [24]. When more information in the form of relations, entities/classes, concepts, individuals/instances gets fastened with taxonomies, these taxonomies grow into ontology [23]. Consequently one can build any domain specific ontology which comprises of concepts, relationship between concepts, concept taxonomies and properties of concepts [25].

1.3: Application of Ontologies

The usage of ontologies spans through various knowledge areas such as information extraction, knowledge management, data integration and most important one in semantic web [1]. Ontology is generally employed as structure knowledge about a specific field by presenting closely related concepts and relations among them. The notion of putting across semantics through ontologies awake the significance of software industry [5]. Moreover ontologies help in the construction of clinical decision support system (CDSS) and these CDSS progress the reprocessing and usage of data [22]. Biomedical Ontologies are utilized by medical specialist, hospitals and related bodies for different purposes including information gathering for decision support systems, interpretation of biomedical data for better data assimilation and data findings [21].

1.4: Importance of Ontology Evaluation

As concerned to evaluation of ontologies, it is an important task due to their usage in various fields. In literature significance of ontologies as engineering artifacts has been justified, so evaluation of ontologies is antecedent to their reuse. Up till now various ontology evaluation techniques have been proposed which are directed and useful for different intended purposes. Among other evaluation techniques, techniques and procedures for evaluating structural characteristics of ontologies are necessary as it guarantee that what is fabricated satisfies the application requirements. On the other hand ontology evaluation helps in reaching a decision whether to use the examined ontology or not [4]. Unfortunately due to the lack of well-

understood conceptions of ontology evaluation, there exists sluggish conversion of ontologies from specialized representing appearance into engineering and business components [5].



Figure 1.2: Evaluation and Selection of Appropriate Ontology

So there is a need to evaluate the anticipated structure and functionality of ontologies. By doing structural evaluation of ontologies, each candidate ontology can shows its density of expressivity in terms of structural constructs

1.5: Biomedical Ontologies

In biomedical domain, numerous ontologies are endeavoring to merge biomedical facts through a range of strategies [6]. Biomedical ontologies are used by researchers for maintaining and creating biomedical domain knowledge in the form of entities and relations. While biomedical scientists integrate and annotate domain knowledge using biomedical ontologies. Various biomedical ontologies have been constructed, for instance SNOMED [15], GO [16], NCI Thesaurus [17], MGED Ontology [18] and FuGO [19]. From these sources research communities can extract their required data.

1.6: Motivation: Structural Evaluation of Biomedical Ontologies

Due to extensive application of ontologies in knowledge demonstration, knowledge engineering and reuse, it gives rise to the notion of evaluation of ontologies [23]. While

searching ontologies in an endeavor to locate the appropriate ontology, it is not simple for user to do it by simply looking at the resulting ontologies. This task gets easy for user if he or she is given some quantitative or qualitative criteria against which ontology gives its quality score.

Structural measures for ontology evaluation have a number of benefits [53]:

- 1. They simply generate numerical figures. This makes easier the tracing the ontology evolution as medication in numerical figure shows clear evolution.
- 2. Structural characteristics and components can be effectively measured from ontology either directly from ontology graph ore any other query mean.
- 3. Structural components can be plainly reported and visualized.
- 4. Results of extracted structural properties can be easily checked and verified.

Different evaluation techniques that are based on quantitative and qualitative metrics have been proposed. As the ontologies fluctuate in their purpose, domain, language, domain, may be engineered by domain experts or scholars, and may be acquired by semi-automatic process or fully automatic process, consequently the evaluation of ontologies becomes a complex endeavor, and so lacks the standard evaluation method [23].

The existing frameworks of evaluating biomedical ontologies do not provide information of processes and low level detail as these frameworks lack the knowledge about structural metrics like concepts and relations among them [2]. That's why biomedical ontologies are not yet taken up for usage by the industry to a significant extent. To overcome this constraint the proposed methodology evaluates the domain specific biomedical ontologies on the basis of structural entities like classes, their instances, data properties, object properties and other structural metrics. Applying this approach it can be easily determined which ontology is suitable with respect to structural dimensions. For experimentation purposes Protégé 4.3 tool is used and SPARQL queries are applied on the stored biomedical ontologies.

Furthermore section 2 presents the existing approaches of ontology evaluation approaches with emphasis on the biomedical domain. Section 3 discusses the proposed methodology. In section 4 we reveal the results of proposed evaluation methodology and

comparison with other related approaches. In the last section conclusion including advantages of the proposed technique, its limitations and future directions are discussed.

CHAPTER 2: RELATED WORK

2.1 Basic Methodology of Constructing Ontologies

For the purpose of evaluating ontologies, first step is to go into the process of building ontology; how ontology can be build. As stated in [37] ontology is a way of organizing knowledge information. The structure of ontology is described in a way that it a combined set of categories (concepts, classes), relationships, attributes (properties), constraints and individuals (instances) [37]. Figure 2.1 shows the structure of ontology.



Figure 2.1: Structure of Ontology

Keeping in the mind the structure of ontology, according to [37] the first step of constructing ontology is to fabricate the ontology frame. In the second step essential concepts and terms are extracted from domain. Then concepts are interlinked with each other through relations. Also instances or individuals of concepts are elaborated. Afterwards ontology is formalized and coded. In the last step ontology is evaluated and verified. These steps are shown in figure 2.2.



Figure 2.2: Steps on Constructing Ontology by [37]

2.2 Role of Ontology Evaluation in Ontologies Life Cycle

As discussed earlier the evaluation of validity and characteristics of ontology is highly necessary because of the fact that this evaluation assures what is constructed (i.e. ontology) fulfils the requirement of end user or any software application. Further on validation and evaluation of ontologies are of significance importance as numerous ontologies with same domain area are common and end user has to select the best among them. In [38] life cycle of ontologies is presented. The presented life cycle consist of this phases namely management phase which deals with quality control, technical phase which deals with processes for building ontology and support phase. In this ontology life cycle ontology evaluation gets into practice in the phase of support and management. Ontology evaluation can also be helpful in the cases where ontology is occupied by redundant data as these may lower the ontology usefulness [38].

2.3 Major Methodologies of Evaluating Ontologies

In current era of information overflow, sound structured knowledge bases and ontologies play a significant task in simplifying the access to knowledge and their processing [21]. A diversity of research efforts has been performed on evaluation of ontologies.

2.3.1 Golden Standard Evaluation

Among the different approaches of ontology evaluation Golden Standard Evaluation is famous [1]. This gold standard ontology is deemed to be well-constructed to act as a reference. In this approach the selected ontology is compared against a gold standard for a benchmark comparison. Although it put forward a way to evaluate ontologies but gold standard itself requires to be assessed, hence it is hard to set up its quality.

2.3.2 Task based Evaluation

Task based or application based approaches [7] evaluate the quality of ontology with respect to its usage in a certain application. Task based approach has major issue that as there are A Framework for Evaluation of Biomedical Ontologies based on Structural Components 7

different applications having different context, so what is applicable in one application context may contrast with other application.

2.3.3 Data driven Evaluation

Another approach named data driven evaluation approach, ontology's correspondence with the knowledge available in the domain is measured [8]. This includes the matching ontologies in opposition to existing domain knowledge. In other words degree of fit is measured between the domain data set and ontology. As domain knowledge keeps on evolving so data driven evaluation approach is not as effective as it considered knowledge to be constant. Also this methodology needs to have traceability mechanisms for showing basic relations between domain data set and ontology entities [50].

2.3.4 User based Evaluation

In user-based evaluation approach ontology is assessed through user's experiences [9]. In other words user-based evaluation estimate the quality of ontology with respect to predefined criteria set by humans. The problem with this approach is instituting the objective metrics for evaluation, and also it is difficult to determine who the right users are [4]. All the above mentioned approaches calculate empirical quality scores to depict the corresponding quality level of ontologies.

2.3.5 Multi-Criteria Evaluation

There exists multi-criteria ontology evaluation approach which assesses the ontologies on multiple dimensions. This type of evaluation collects various types of statistics about the domain knowledge available in ontology by inspecting the ontology. Some multi-criteria based evaluation techniques consider the population of ontology components.

Ontometric [9] is based on multi-criteria methodology. It uses the dimensions of content, development methodology, cost, language and used software development environment. This method requires the application to be provided by many values which are utilized to express the appropriateness of ontology. Ontometric is not flexible enough to take into consideration the variety of conflicting needs of users for ontology evaluation.

In [39] the authors advise to evaluate the ontology on quantifiable and non-quantifiable characteristics. This methodology is based on importing ontologies through web crawling and A Framework for Evaluation of Biomedical Ontologies based on Structural Components 8

saving them in the database. After wards the most appropriate ontology is extracted by providing weights and domains by user.

Another approach named AKTiveRank [40] locates related ontologies against the term entered by user. This technique used four metrics for evaluating ontologies. The metrics include semantic similarity, density, class match and betweenness. Each ontology is evaluated against these four evaluation metrics, and in result most suitable ontology is selected.

Corocho et al. establish a tool named ODEval which automatically identifies the syntactical problems in ontologies [41]. The problems detected are incompleteness, presence of cycles in the class inheritance hierarchy, redundancy of instances and classes, and finally inconsistency.

OntoClean [43], a methodology of evaluating and validating ontology, is proposed which is based on features including Unity, Rigidity, Identity and Dependence. These features are assigned by user to each class of ontology. Set of rules are generated based on these four features against which classes are inspected, whether these classes violate the rule or not. Based on these rules classes can be added or removed to correct the discovered problems.

Obrst et al. evaluated the ontologies on the metrics of validity, soundness, coverage of domain including granularity, richness and complexity of coverage, completeness, consistency, reusability and adaptability, inference ability, mappability to other ontologies and finally evaluation against requirements, use cases, data sources and applications [51].

In [52] ontologies are evaluated against the criteria of transparency of analyzing ontology in detail, cognitive ergonomics of easily comprehension and manipulation of ontology, metalevel integrity through quality indicators, computational efficiency and integrity processed by classifier or inference engine, flexibility of being adoptive to multiple views, compliance to procedures for integration, extension and adaption, organizational context fitness, compliance to expertise and generic accessibility for effective applications.

Florian and Patrick organize ontology evaluation process with respect to complexity, evaluation method and approach strategy [56]. Complexity refers to the evaluation layers/levels of ontology including structure/design/syntax, taxonomy/hierarch, vocabulary/lexical/data layer

and application/context level. Evaluation methods refer to feature/metric based, rule/logical based and evolution based. Evaluation methods also discuss the tools used for ontology evaluation including OntoMetric, EvaLexon, OntoClean, OntoManager, S-OntoEval and WebCore. Finally approach referes to application based, user based, golden standard and data driven.

2.3.6 Popularity – based Evaluation of Ontologies

Some approaches are based on the popularity of ontologies from a set on ontologies. For instance OntoKhoj [11] use OntoRank algorithm which is based on semantic links between ontologies. These links include subsumption and instantiation. OntoKhoj acts as a keyword-based search engine for ranking ontologies. It puts up word sense approach, followed by selection of synonyms and hypernyms from WordNet.

Swoogle [12] search engine queries its database containing semantic data, and applies quality metrics on ontologies for ranking. Its search facility is based on topic coverage. When a search word is submitted to Swoogle search engine, it brings into the ontologies that contain lexically equivalent relation or class to given search word. OntoSelect [13] considers semantic connectedness between imports of ontologies and named it ontology imports.

2.3.7 Evolution – based Evaluation of Ontologies

By nature ontologies evolve over time [38]. This evolution or modification is due to addition of more knowledge and this knowledge addition requires proper reorganization of content. So evolution based evaluation approaches generally detect modifications of domain knowledge to track quality of ontology. Also invalid modifications made to ontology are also detected and recovered in this approach. In literature three reasons are described for evolution based evaluation of ontologies; changes in conceptualization, changes in domain and changes in the explicit specification.

Plessers et.al proposed an evolution based technique for evaluating ontologies. In this technique a request of change is added to a change log when a change is required on the ontology [44]. After the addition change request, change is incorporated in the ontology. Finally the real change is compared with the change request from log. If both are identical, the modification is deemed as legitimate.

Another evolution based ontology evaluation technique is proposed by Haase et.al in [45]. In their work they track two kinds of inconsistencies while modification process takes place in ontology. Language based inconsistencies and user-defined inconsistencies are detected. Then inconsistencies are repaired across the various versions of the ontology by removing the components which create inconsistencies in the ontology.

2.3.8 Rule – based Evaluation of Ontologies

These evaluation methodologies are based on rules built in the ontology languages. These rules fundamentally track conflicts in the ontologies [38]. For instance if two instances have property of owl:differentFrom then according to rules these two instances cannot have a property owl:sameAs. In [46] a rule based approach is described which detects the conflicts in ontologies. Conflicting rules are discovered by users through RuleML. Another approach called Swoop identifies unsatisfiable concepts in OWL ontologies [47]. Ontology designers utilize this approach for assessing the ontology quality and problem indication.

2.3.9 Layered (Level) – based Evaluation of Ontologies

According to [1], ontology is a complex structure so because of this fact it is easier to evaluate ontology level wise as compared to whole ontology. Estimated layers of ontologies are syntactic, structural (or design), context (or application), taxonomy (or hierarchy), lexical (concept, data) and other semantic relation.

2.3.10 Structure – based Evaluation of Ontologies

Some work has been done in the dimension of structural evaluation of ontologies. These methodologies are based on the richness of knowledge which these put across. Among these is ActiveRank algorithm [10]. It does not depend on ontology library. It merges together a collection of metrics based on ontology structure to evaluate and rank ontologies. ActiveRank uses three major metrics i.e. Density Measure (DEM) metric, Centrality Measure (CEM) metric and Semantic Similarity Measure (SSM) metric. DEM metric depicts how well a concept is explained in the target ontology by adding up the superclasses, subclasses, relations and instances. CEM metric depends on the notion that if the concept exists in the middle of ontology then it has the highest representation level. Middle of the ontology is calculated as the distance from the root node to the given concept. SSM metric measures the closeness of concepts and links among them.

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Another evaluation approach named oQual [42] that is based on dimensions of functional, structural and usability profiling. In structural mode of evaluation, ontology is evaluated against features based on both semantic and syntax of an ontology. Theses structural features include fan-outness, density, depth, breadth, specific differences, distribution degree, logical adequacy, modularity and meta-adequacy. Functional measures include accuracy, recall and precision. Usability profiling includes interfacing efficiency and recognition.

OntoCAT approach uses comprehensive collection of metrics for evaluating ontology for the purpose of their reuse in other applications [48]. The metrics used by this approach comprises of structural properties, hub, size and root properties.

In [49] Full Ontology Evaluation (FOEval) is proposed which is based on four ontology features. These features include coverage, richness, detail-level and comprehensiveness. Coverage includes relation coverage and class coverage. Class coverage means that in the ontology how many class names matched the search keyword provided by user. Similarly relation coverage means that in the ontology how many relations matched the search keyword provided by user. Second feature that is Richness also has two facets e.g. attribute richness and relation richness. Attribute richness is measure of average number of attributes per class and relation richness is measure of total relations in the ontology. Detail level feature is further divided into specific detail level and global detail level. Specific detail level shows the significance of searched terms in the ontology while global detail level differentiates vertical ontology from horizontal ontology. Comprehensiveness comprises of average number of annotated relations.

FOEval approach [49] also partitions available ontology evaluation approaches into four groups: what should be evaluated (that is partial ontology is evaluated or whole ontology), when it should be evaluated (that is during ontology constructing process, before ontology construction, during ontology maintenance, or before ontology use) and lastly based on what it should be evaluated (that is gold standard evaluation, task based evaluation, corpus based evaluation, user based evaluation and multi-criteria based evaluation).

Fernandez-Beris et al. [54] express ontology evaluation from the perspective of correctness, ranking, quality and software engineering. Correctness and general ranking of ontologies are verified against the structural properties while quality of ontologies is evaluated against the dimensions of reliability, usability and functionality. Under the perspective of software evaluation, it comprises of internal measures (that are related with software itself), external measures (that are assessed while operating process or testing) and in-use quality measures.

OntoQA (Ontology Quality Assurance) [38] is a feature-based ontology evaluation tool. The metrics of OntoQA are divided into two categories: instance metrics and schema metrics. Instance metrics deals with the organization of instances within the ontology. Under the category of schema metrics, the sub-metrics are relationship richness, inheritance richness and attribute richness. Whereas in instance metrics, the sub-metrics are class richness, class connectivity, class importance, cohesion and relationship richness. It is quite simple evaluation tool as it requires less user involvement. As an input OntoQA considers set of user supplied search terms or crawled populated ontology and grade them according to metrics mentioned above. The experimentations are conducted on three ontologies; Glyco, TAP and SWETO. Glyco is ontology for the domain of glycomics, TAP is a general purpose ontology while SWETO is also general purpose ontology having emphasis on scientific publications. Summarized result of these three ontologies is as below shown in table 2-1.

Ontology	Classes	Relations	Instances
Glyco	361	56	660
TAP	6959	25	85637
SWETO	44	101	813217

Table 2-1: Summary of OntoQA Results

2.4 Evaluation of Biomedical Ontologies

Being enormous expansion of biomedical data that is being produced by various experimental techniques, it has generated a lot of opportunities for biomedical data discovery. At the same time challenges have been faced in application of ontologies in biomedical research as

sphere of knowledge covered by biomedical ontologies are intermingled along with their different modeling and building techniques [14].

Though there present a lot of work in biomedical ontologies but towards evaluation of these ontologies no significant efforts have been taken. Fernandez et al [26] evaluated the ontologies correctness and ranking according to structural property. They also proposed ontology evaluation framework which is based on the aspects of functionality, structural, usability, reliability, quality in use and maintainability. Netzer et al. [27] evaluate ontologies on intrinsic methods (based on structural properties) and extrinsic methods (based on data corpus, task and expert view).

One approach found in literature with title Ontobee [36] extract the statistics of ontologies. It is web-based system (http://www.ontobee.org) that retains the statistical record of about 128 ontologies. In Ontobee repository ontologies are presented as Linked Data. It only focuses on four components namely classes, object properties, datatype properties and annotation properties. Web interfaces of Ontobee are shown in figure 2.3 and figure 2.4

Our proposed work is directed towards structural evaluation of similar biomedical domain ontologies. We include metrics based on density or coverage of classes or concepts, individuals, object properties, data properties, annotation properties, functional properties, symmetric properties, asymmetric properties and reflexive properties.



Figure 2.3: Ontobee main Interface

Keywords:	Search terms
Number of Terms (including imported terms) (Detailed 9	Statistics)
number of Terms (including imported terms) (<u>Detained</u>	
 <u>Class</u> (40296) <u>ObjectProperty</u> (113) 	
<u>AnnotationProperty</u> (103)	

Figure 2.4: Ontobee Interface of showing Statistics

CHAPTER 3: GATHERING DATA

3.1: Candidate Biomedical Ontologies

For experimental purposes total of eleven biomedical ontologies are imported in Protégé 4.3 from The Open Biological and Biomedical Ontologies (<u>http://www.obofoundry.org/</u>). Three of the imported ontologies are from the domain of anatomy, two of them are from neuroscience domain, two from the field of medicine, two ontologies from biomedical experimental domain and two from medical statistics. Their detail is described in table 3-1.

Sr. No	Ontology Title	Domain	File
1	Uber Anatomy Ontology	Anatomy	ext.owl
2	Porifera Ontology	Anatomy	poro.owl
3	Hymenoptera Anatomy Ontology	Anatomy	hao.owl
4	NIF Cell	Neuroscience	NIF-Cell.owl
5	NIF Dysfunction	Neuroscience	NIF-Dysfunction.owl
6	Ontology for General Medical Science	Medicine	ogms.owl
7	Ontology for Medically Related Social Entities	Medicine	omrse.owl
8	NMR-Instrument Specific Component of Metabolomics Investigations	Experiments	NMR.owl
9	Microarray Experimental Conditions	Experiments	MGEDOntology.owl
10	STATistics Ontology	Statistics	stato.owl
11	Ontology of Biological and Clinical Statistics	Statistics	obcs.owl

3.2: Procedure of Importing Biomedical Ontologies in Protégé 4.3

Following are the key steps of importing ontologies in Protégé 4.3.

i. Open Protege 4.3 and click on Direct Import under the tab of Ontology Imports as shown in figure 3.1.

ii. In the Import Ontology Wizard, there are four ways of importing ontologies as depicted in figure 3.2. First one is to import ontology directly from any location in a hard disk. Second one is a way to import any desired ontology directly from web source. Other two ways are related to import ontologies from loading ontology from workspace and loading ontologies from ontology libraries. In our current experimental work we have used first two methods of importing ontologies.

🝕 untitled-ontology-96 (http://www.semanticweb.org/texitech/ontologies/2015/4/20/untitled-ontology-96) : [http:/ 👝 💷 💌			
File Edit View Reasoner Tools Refactor Window Help			
 			
Active Ontology Classes Object Properties Annotation Properties Individuals OntoGraf SPARQL Query			
Ontology header:			
Ontology IRI http://www.semanticweb.org/texitech/ontologies/2015/4/20/untitled-ontology-96			
Ontology Version IRI e.g. http://www.semanticweb.org/texitech/ontologies/2015/4/20/untitled-ontology-96/1.0.0			
Annotations 🛨			
Ontology imports Ontology Prefixes General class axioms			
Indirect Imports			
To use the reasoner click Reasoner->Start reasoner Show Inferences			

Figure 3.1: Direct Importing of Ontologies in Protege 4.3

🍕 Import ontology wizard		×
	Import type	
	Please choose an option:	
	 Import an ontology contained in a specific file. Import an ontology contained in a document located on the web. Import an ontology that is already loaded in the workspace. 	
	Go Back Continue C	ancel

Figure 3.2: Option of Importing Ontologies in Protege 4.3

iii. By selecting the second option, the wizard requires Uniform Resouce Identifier (URI) of candidate ontology to be entered in the text box in front of URI as shown in figure 3.3. For instance URI of NIF-Cell ontologies is <u>http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.owl.</u>

🍕 Import ontology wizard		x
	Import from URL	
	Please specify the URL that points to the file that contains the ontology. (Please note that this should be the physical URL, rather than the ontology URI)	
	URI Thttp://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.owl	
	Bookmarks Bookmarked URIs + http://adverse-event-reporting-ontology.googlecode.com/svn/trunk/src/ontology/aero.owl	i
Y	Manually specify import declarations. This is generally not needed as Protege will choose a reasonable default.	
	Go Back Continue Ca	ancel

Figure 3.3: Importing Ontology using URI

Mark Import ontology wizard		
Import verification		
	Please wait. Verifying import	

Figure 3.4: Verification of Import Ontology

iv. After pressing Continue button in the prevous wizard, verification of importing ontologies is done as shown in figure 3.4.

v. After successful verification of importing ontologies, wizard confirms the successful import of the given ontology as shown in figure 3.5.

🍕 Import ontology wizard		×
Confirm imports		
	The system will import the following ontologies. Press Finish to import these ontologies, or Cancel to exit the wizard without importing any ontologies.	
	http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.owl	
\langle) prot	Ė
	Go Back Finish	Cancel

Figure 3.5: Confirmation of Imported Ontologies

3.3: Description of Imported Biomedical Ontologies in Protégé 4.3

i. Imported Ontology get visible under the tab of Ontology Imports which confirm that the candidate ontology is now active and any operation can be performed on it. This is shown in figure 3.6.

ii. To view the classes of candidate ontology, class tab is pressed to view classes. Classes are view as hierarchical view as shown in figure 3.7.

iii. To view the object properties of candidate ontology, object property tab is pressed to view them as shown in figure 3.8.

iv. To view the annotation properties of candidate ontology, annotation property tab is pressed to view them as shown in figure 3.9.

v. To view the individuals of candidate ontology, individuals tab is pressed to view these as shown in figure 3.10.

vi. To view the classes of candidate ontology graphically, OntoGraf tab is pressed to view these as shown in figure.

🍕 untitled-ontology-96 (http://www.semanticweb.org/texitech/ontologies/2015/4/20/untitled-ontology-96) : [http:/ 👝 💷 📧				
File Edit View Reasoner Tools Refactor Window Help				
 Image: search for entity 				
Active Ontology Classes Object Properties Annotation Properties Individuals OntoGraf SPARQL Query				
Ontology header:				
Ontology IRI http://www.semanticweb.org/texitech/ontologies/2015/4/20/untitled-ontology-96				
Ontology Version IRI e.g. http://www.semanticweb.org/texitech/ontologies/2015/4/20/untitled-ontology-96/1.0.0				
Ontology imports Ontology Prefixes General class axioms				
Imported ontologies:				
Direct Imports + NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.owl)				
To use the reasoner click Reasoner->Start reasoner Show Inferences				

Figure 3.6: Visibility of Imported Ontology in Protégé 4.3

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.owl) : [http://ontology.neuinfo.org/NIF					
File Edit View Reasoner Tools Refactor Window Help					
🗢 🖒 NF-Cell 🗸 Search for entity					
Active Ontology Classes Dindividuals OntoGraf SPARQL Query Object Properties					
Class his					
Class hierarchy: Thing III III Annotations: Thing IIII III					
🐮 🔄 🕱					
▼•● Thing					
The second secon					
DIRECTED-BINARY-RELATION					
SubClass Of	SubClass Of 🛨				
AbbrevSource					
BirnlexCurator	Indus Andestor)				
BirnlexDefinitionSource					
BirnlexExternalSource Members 🐨					
🖳 😑 CurationStatus 🖉					
To use the reasoned	click Reasoner->Start reasoner 🗌 Show Inferences				

Figure 3.7: Viewing Classes of Active ontology in Protégé 4.3

A Framework for Evaluation of Biomedical Ontologies based on Structural Components



Figure 3.8: Viewing Object Properties of Active ontology in Protege 4.3



Figure 3.9: Viewing Annotation Properties of Active ontology in Protege 4.3



Figure 3.10: Viewing Individuals of Active ontology in Protege 4.3



Figure 3.11: Graphical View of Classes in Protege 4.3
CHAPTER 4: PROPOSED METHODOLOGY

We have used the biomedical ontologies imported from Open Biological and Biomedical Ontologies (http://www.obofoundry.org/). In this chapter proposed methodology is briefly explained.

4.1: System Architecture



Figure 4.1: Evaluation Architecture of Biomedical Ontologies

Figure 4-1 shows the architecture of proposed evaluation technique of biomedical ontologies. The initial step "*Gathering Domain Specific Biomedical Ontologies from Ontology Libraries*". Here *Domain Specific* means to search the related ontologies specific to single domain i.e. NeuroScience Ontologies, Anatomy Ontologies, Medicine Ontologies etc. In the second step of "*Importing Biomedical Ontologies in Protégé 4.3 in XML/RDF Format*" domain

A Framework for Evaluation of Biomedical Ontologies based on Structural Components

specific biomedical ontologies are imported in Protégé 4.3. in XML/RDF format from Ontology Libraries available on web like OBO BioPortal (http://www.obofoundry.org/). In the next step of "*Executing SPARQL Queries on Selected Ontologies for Extracting Structural Constructs*", SPARQL queries are applied to evaluate the quality of candidate ontologies so that structural constructs can be enumerated. In the last step, overall score of each candidate ontology is calculated so that the appropriate biomedical ontology can be selected.

4.2 Ontology Web Language (OWL), Protégé and SPARQL

Protégé is one of the convenient and practical approach of building an ontology from the scratch. Protégé also acts as an ontology editor with OWL and RDFS and OWL languages as a representation mean. Protégé 4.3 version is used in our research experimentations. It offers the facility of composing various types of ontologies using simple graphical user interfaces (GUI) including options for adding classes, subclasses, entities, object properties, data properties, annotation properties and individuals. For extracting different aspects of ontologies SPARQL Protocol and RDF Query Language (SPARQL) queries can be executed in Protégé. In addition Protégé also provide the facility of viewing the ontologies in different hierarchal styles like grid, radial, spring, vertical tree, horizontal tree, vertical directed and horizontal directed. Any OWL based ontology with extension .exe can be imported in different ways. One way is to directly import ontology from web providing its uniform resource identifier (URI). An alternate way is to save manually .owl file of ontology and import it manually by giving its location in disk.

Ontology Web Language (OWL) now a day's become a standard formal language for representing Semantic Web knowledge. OWL is standardized by World Wide Web Consortium (W3C), which is a standards body for managing web development standards. OWL facilitates automated reasoning as it is based on description logic. One important aspect of OWL is that it can characterize both the reasoning knowledge and domain knowledge by using the constructs of class definitions and axioms [31].

SPARQL is an extremely useful querying and reasoning language that gives effortless access to general classes/concepts, attributes/properties and annotations of dataset i.e. ontologies [32]. In [53] SPARQL queries are applied over OWL ontology for the purpose of extracting

structural patterns and anti-patterns. Due to immense power of SPARQL queries, it best exposes the structural characteristics of OWL based biomedical ontologies.

4.3 Research Methodology or Evaluation Procedure

Among SNOMED, GO, OBO, NCI Thesaurus, MGED Ontology and FuGO, we have chosen OBO because it is the only knowledge source which contains comprehensive biomedical ontologies of related fields. These biomedical ontologies can be accessible through OBO BioPortal. We have chosen the biomedical ontologies that are .owl extension (Ontology Web Language).

4.4 Rationale of Structural Evaluation

While looking for a best biomedical ontology that exclusively illustrates the particular domain concept, one would look forward to find certain extent of detail in the illustration of the data about that concept [10]. This notion leads towards how fine a certain concept is stated in detail in terms of its sub-classes, its properties, relationships, annotations and triples. In ActiveRank [10] structural constructs of direct relations, indirect relations, super-classes, siblings, subclasses and instances are used. In [5] the evaluation framework employs the structural dimensions such as density, consistency, depth, breadth, modularity, leaf and sibling distribution, etc are used for evaluating the ontologies. Similarly from different research efforts in biomedical domain like [21] and [28] the importance of concepts/class, instances, triples and relations/properties are obvious in biomedical ontologies as these are the core elements for determining their quality and significance. Collection of data vocabularies can be considered as hierarchal graph which fundamentally comprises of terms or concepts which acts as nodes in the graph connected together with edges known as relations [29]. In [30] classes and relation among them are taken as core feature and major functional perspective of biomedical ontologies.

4.5 Selected Criteria

According to [33] an Ontology Web Language (OWL) based ontology comprises of Classes/concepts, individuals/instances and properties/relations as a major building blocks. So in our proposed evaluation methodology of biomedical ontologies, we have chosen these three

major components along with the triple density. In table 1 structural building blocks are used against which all candidate ontologies are evaluated.

4.5.1. Classes

Classes are collection of individuals or entities [33]. The word class and concept are used interchangeably. These may be arranged hierarchy of superclasses and subclasses. Subclasses are child classes of superclasses. In Protégé 4.3 class hierarchy of every ontology starts from 'Thing' class, which is superclass of everything. Major classes of Uber Anatomy Ontologies are *anatomical entity, body cavity, immaterial anatomical entity, material anatomical entity, behavior process, biological_process* and so on.

4.5.2. Super-Classes and Sub-Classes

These are the major parent classes in the candidate ontology. In our proposed methodology the top first level of classes are interpreted as super-classes under the root class of Thing. All other classes are interpreted as subclasses. For instance the major super-classes in Uber Anatomy Ontology *anatomical entity, body cavity, immaterial anatomical entity, material anatomical entity, behavior process, biological_process, CARO_000003, cellular_component, material entity, molecular_function, processual_entity, quality and root.*

Sub-classes of Anatomical Entity include anatomical cluster, body cavity or lining, immaterial anatomical entity and material anatomical entity. Anatomical cluster posses further two subclasses which are abdominal scute series and accessory articulation.

4.5.3. Individuals/Instances

Individuals correspond to objects or instances in the domain under discussion[33]. Instances of classes are also termed as Individuals. Individuals of Uber Anatomy Ontology are: *bulbo-urethral gland, epididymis, epoophoron, major vestibular gland, paraurethral gland, prostate duct* and *prostate gland*.

4.5.4. Triples

OWL ontologies are collection of triples [34]. Triple can be interpreted as a form of sentence that states a single event or fact about a resource [35]. Triple consist of subject, predicate and object.

Subject is first part that describes the resource in the form of Uniform Resource Identifier (URI). This URI refers to that resource which identifies it. Predicate follows Subject. In triple second part is predicate which illustrate the type of relationship in triple. It is a phrase from some domain ontology showing some property attached with the subject. It also can be represented in URI form. Object is the last part of triple. It describes something that is associated to the subject through some property or relationship. It is of two categories: URIs and character strings (literal) [35]. On executing the query of extracting subjects, predicate and object, we obtain following under mentioned subjects, predicate and objects.

Table 4-1:	Triples	Compone	ents
------------	---------	---------	------

Subjects	Predicates	Objects
Average depth of sequence	Source	NAID GSCID-BRC
coverage		
Subgraph modularity calculation	Definition	A network subgraph quality
		calculation
B Transformation	Туре	Class
Dye Swap Merge	Has curation status	Ready for release



Figure 4.2: Triple Components

4.5.5. Properties

Properties are intermediate relationship between two individuals [33]. Other interchangeable names used for properties are relations, roles and attributes. In other words it connects together the two individuals. In our experimentations we have chosen object property, data property, annotation property, inverse property, functional property, symmetric property, asymmetric property and reflexive property. These all are explained below.

4.5.5.1. Object Properties

Object properties denote the association between two instances / individuals [33]. These connect an individual to other individual. There exists support for establishing object properties between two individuals in Protégé 4.3 while building ontology. Its examples includes *has category level, has member, has function, has part, is preceded by* and many more. Inheritance relationship also lies under this category.

4.5.5.2. Data Properties

These properties express the relationship between an object (individual) and data values. In a more specific sense these properties associate an object to an XML Schema Datatype value or an RDF literal [33]. There exists support for establishing data properties between one individuals and some data value in Protégé 4.3 while building ontology. Its examples includes has *measurement value, has specified value, has x coordinate value, has y coordinate value, has_feature_value* and many more.

4.5.5.3. Annotation Properties

Annotation properties explain properties, classes, ontology itself and individuals with different bits of information. This information (meta-data) includes creation date, comments, reference to resources or author [33]. OWL comprises of five annotation properties: rdfs:label, owl:versionInfo, rdfs:seeAlso, rdfs:comment and rdfs:isDefinedBy. One special feature of Annotation properties is that these have or be the not the sub-property. Like other properties there also exists support for establishing annotation properties to individuals, classes, properties and ontology itself in Protégé 4.3 while building ontology.

4.5.5.4. Inverse Properties

Inverse properties are associated with object properties as each object property may possess a analogous inverse property [33]. For instance if some object say M is connected to object N, then according to definition of inverse property object N is connected to object M via some other property whose logic is totally opposite. For Example, *isIngredientOf* and *hasIngredient* are inverse property of each other. Similarly *hasBase* is inverse property of *baseOf*. Inverse property adds some characteristics to object property. It is of less importance that's why its weightage or priority of existence in ontology is considered as low. There also exists support for establishing inverse properties to object property in Protégé 4.3 while building ontology.

4.5.5.5. Functional Properties

Through functional property, there can be at most one object/instance which is linked to some other object through this property [33]. These properties are also recognized as single valued properties in literature. Its examples include has time stamp, has measurement value, is immediately preceded by, inheres in and many more. Like inverse property, it also adds characteristics to Object property. For Example *legally revokes, legally transfers, has time stamp, inheres in,* and *has measurement value.*

4.5.5.6. Symmetric Properties

Symmetric property relates the two objects in a way that if object M is linked to object N via property P then N is also linked to M via property N [33]. It is one of the characteristics properties of Object property. For Example, *extends_fiber_into, continuous with, adjacent to, homologous_to, attached to* and *adjacent to.*

4.5.5.7. Asymmetric Properties

Asymmetric property relates the two objects in a way that if object M is linked to object N via property P then N is cannot be linked to M via property P [33]. As obvious from the previous statement it is inverse of symmetric property. It is one of the characteristics properties of Object property. For Example *administrates, administered by, is enrolled in school, legally transfers* and *legally invokes*.

4.5.5.8. Reflexive Properties

Through this property say P an object M relate to itself via P. It is one of the characteristics properties of Object property.

For the sake of simplicity and understandability examples from real world have been taken.

Sr. No	Criteria Title	Description	
1	Individuals	Class instances or object. Like if Country is a Class then Pakistan, Iran,	
		France are individuals/instances.	
2	Properties	Properties are said to be relation between two instances/individual of	
		classes	
2a	Object Property	Relationships between two instances/individuals of classes. Has_Part,	
		isChildOf, is_a.	
2b	Data Property	A data property connects the instance to some data value with certain	
		relation. For instance Umer to the data literal '25', with hasAge relation	
2c	Annotation	Annotation property appends information to instances, classes, data	
	Property	properties and object properties like imported_from, editor_note,	
		description, definition, references to resources such as web pages etc. An	
		annotation property, linking the class 'JetEngine' to the data literal	
1	T D	(string) "Matthew Horridge".	
2d	Inverse Property	It is acts as a reverse property like hasPart, isPartOf, isIngredientOf,	
2e	Functional	Functional properties are single valued properties. If a property is	
	Property	that is related to that instance via the property is headingham before the	
Of	Transitiva	Transitive property relates individual A to individual B, and also	
21	Property	individual D to individual C then it can be said that individual A is	
	roperty	related to individual C, i.e. bestneester	
20	Symmetric	If a property P is symmetric, and the property relates individual X to	
2g	Properties	individual V then individual V is also related to individual V via	
	Toperties	property P i.e. hasSibling hasBrother	
2h	Asymmetric	If a property P is asymmetric and the property relates individual M to	
	properties	individual N then individual N cannot be related to individual M via	
	FF	property P. Example: If the individual John is related to the individual	
		Micheal via the isChildOf property, then it can be inferred that Micheal	
		is not related to John via the isChildOf property.	
2i	Reflexive	It is self-related property. For example of this: using the property knows,	
	properties	an individual George must have a relationship to itself using the property	
		knows. In other words, George must know himself.	
3	Classes	Classes consist of a set of individuals .i.e. Country, Person	
3a	Sub Classes	Child or derived Classes	
3c	Super Classes	Parent Classes	
4	Triples	OWL axioms are mapped into triples. For Example, if C1 and C2 are	
		classes, then C1 rdfs:subClassOf C2 is a triple, Similarly C1 rdfs:domain	
		C2 is another triple showing mapping of Object PropertyDomain	

 Table 4-2: Selected Criteria for Biomedical Ontologies

4a	Subject	In the above stated examples of triple Class C1 is Subject	
4b	Predicate	In the above stated examples of triple rdfs:subClassOf and rdfs:domain	
		are predicates which join Subject with Object	
4c	Object	In the above stated examples of triple Class C2 is Object	

CHAPTER 5: RESULTS

5.1: Extraction of Results

We have applied SPARQL queries on the imported biomedical ontologies in Protégé 4.3. For the sake of readability, we restrict ourselves to present some of the queries.

5.1.1: Query Interfaces of Triple Components

a. Enumeration of Subject Density

To extract subject density from the given ontology, following SPARQL query will be executed as shown in figure 5.1.

PREFIX rdf:<http://www.w3.org/1999/02/22-rdf-syntax-ns#> PREFIX owl:< http://www.w3.org/2002/07/owl#> PREFIX xsd: < http://www.w3.org/2001/XMLSchema#> PREFIX rdfs: < http://www.w3.org/2000/01/rdf-schema#>

SELECT (count (DISTINCT ?s)

AS ?Subject_Count) { ?s ?p ?o }

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.owl) : [htt 👝 💷 🗨			
File Edit View Reasoner Tools Refactor Window Help			
Search for entity			
Individuals OWLViz DL Query OntoGraf SPARQL Query Ontology Differences			
Active Ontology Entities Classes Object Properties Data Properties Annotation Properties			
SPARQL query:			
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""></http:>			
PREFIX owl: <http: 07="" 2002="" ow#="" www.w3.org=""></http:>			
PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf_schema#="" www.w3.org=""></http:></http:>			
SELECT (count (DISTINCT ?s) as ?Subject_Count) { ?s ?p ?o }			
Subject Count			
"37470"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>			
Execute			
To use the second stick Deserves a Charlesson and Charlesson			
To use the reasoner click Reasoner->Start reasoner			

Figure 5.1: Enumeration of Subject Density

b. Enumeration of Predicate Density

To extract predicate density from the given ontology, following SPARQL query will be executed as shown in figure 5.2. Required prefixes are added.

SELECT (count(DISTINCT ?p)
AS ?Predicate _Count) { ?s ?p ?o }

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.owl) 💼 🔳 💌			
File Edit View Reasoner Tools Refactor Window Help			
 Image: Search for entity 			
Individuals OWLViz DL Query OntoGraf SPARQL Query Ontology Differences			
Active Ontology Entities Classes Object Properties Data Properties Annotation Properties			
SPARQL query:			
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count(DISTINCT ?p) as ?Predicate_Count) { ?s ?p ?o }</http:></http:></http:></http:>			
Predicate_Count			
"112"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>			
Execute			
To use the reasoner click Reasoner->Start reasoner 🗹 Show Inferences			

Figure 5.2: Enumeration of Predicate Density

c. Enumeration of Object Density

To extract objects density from the given ontology, following SPARQL query will be executed as shown in figure 5.3. Required prefixes are added.

SELECT (count (DISTINCT ?o)
AS ?Object_Count) { ?s ?p ?o }

🔏 NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Ce 👝 💿 💌			
File Edit View Reasoner Tools Refactor Window Help			
Annotation Properties Individuals OntoGraf SPARQL Query Active Ontology Entities Classes Object Properties Data Properties			
SPARQL query:			
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count (DISTINCT ?o) as ?Object_Count) { ?s ?p ?o }</http:></http:></http:></http:>			
Object_Count			
"45110"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>			
Execute			

Figure 5.3: Enumeration of Object Density

d. Enumeration of Triple Density

To extract triple density from the given ontology, following SPARQL query will be executed as shown in figure 5.4. Required prefixes are added.

```
SELECT (COUNT (DISTINCT *)
AS ?Triple_Count) { ?s ?p ?o }
```

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Ce			
File Edit View Reasoner Tools Refactor Window Help			
🗢 🖒 NF-Cell 🚽 Search for entity			
Annotation Properties Individuals OntoGraf SPARQL Query			
Active Ontology Entities Classes Object Properties Data Properties			
SPARQL query:			
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count (DISTINCT *) AS ?Triple_Count) { ?s ?p ?o }</http:></http:></http:></http:>			
Triple_Count			
"143332"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>			
Execute			
To use the reasoner click Reasoner->Start reasoner Show Inferences			

Figure 5.4: Enumeration of Triple Density

5.1.2: Query Interfaces of Classes and Individuals/Instances

a. Enumeration of Classes Density

To extract class density from the given ontology, following SPARQL query will be executed as shown in figure 5.5. Required prefixes are added.

SELECT (count (DISTINCT ?class) AS ?Class_Count) { ?class a owl:Class }

🝕 NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Ce 👝 💷 📧			
File Edit View Reasoner Tools Refactor Window Help			
Active Ontology Classes Individuals OntoGraf SPARQL Query			
SPARQL query:			
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count(DISTINCT ?class) as ?Class_Count) { ?class a owl:Class }</http:></http:></http:></http:>			
Class_Count			
"2986"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>			
Execute			
To use the reasoner click Reasoner->Start reasoner Show Inferences			

Figure 5.5: Enumeration of Classes Density

b. Enumeration of Individuals/Instances Density

To extract Individuals/Instances density from the given ontology, following SPARQL query will be executed as shown in figure 5.6. Required prefixes are added.

SELECT (count (DISTINCT ?s) AS ?Inividual_Count) { ?s a ?class }

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-C		
File Edit View Reasoner Tools Refactor Window Help		
🗢 🖒 🚺 🗸 Search for entity		
Active Ontology Classes Individuals OntoGraf SPARQL Query		
SPARQL query:		
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count(DISTINCT ?s) as ?Individuals_Count) { ?s a ?class }</http:></http:></http:></http:>		
"5616"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>		
Execute		
To use the reasoner click Reasoner->Start reasoner Show Inferences		

Figure 5.6: Enumeration of Individuals/Instances Density

5.1.3: Query Interfaces of Properties / Relation

a. Enumeration of Object Properties Density

To extract density of object properties from the given ontology, following SPARQL query will be executed as shown in figure 5.7. Required prefixes are added.

SELECT (count(DISTINCT ?x)

AS ?Object_Property_Count)

{?x a owl:ObjectProperty}

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-C		
File Edit View Reasoner Tools Refactor Window Help		
🗢 🗗 🚺 🕶 Search for entity		
Active Ontology Classes Individuals OntoGraf SPARQL Query		
SPARQL query: DBB 2		
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count(distinct ?x) as ?Object Property Count) {?x a owl:ObjectProperty} Object_Property_Count "85"^^<http: 2001="" www.w3.org="" xmlschema#integer=""></http:></http:></http:></http:></http:>		
Execute		
To use the reasoner click Reasoner->Start reasoner Show Inferences		

Figure 5.7: Enumeration of Object Properties Density

b. Enumeration of Data Properties Density

To extract density of data properties from the given ontology, following SPARQL query will be executed as shown in figure 5.8. Required prefixes are added.

SELECT (count(DISTINCT ?x)

AS ? Data_Property_Count)

{?x a o	wl:DataP	roperty}
---------	----------	----------



Figure 5.8: Enumeration of Data Properties Density

c. Enumeration of Annotation Properties Density

To extract density of annotation properties from the given ontology, following SPARQL query will be executed as shown in figure 5.9. Required prefixes are added.

SELECT (count(DISTINCT ?x)

as ? Annotation_Property_Count)

{?x a owl: Annotation Property}



Figure 5.9: Enumeration of Annotation Properties Density

d. Enumeration of Inverse Properties Density

To extract density of inverse properties from the given ontology, following SPARQL query will be executed as shown in figure 5.10. Required prefixes are added.

SELECT (count(DISTINCT ?x) AS ? Inverse_Property_Count) {?x a owl: Inverse Property}

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-C								
File Edit View Reasoner Tools Refactor Window Help								
♦ ♦ NIF-Cell Search for entity								
Active Ontology Classes Individuals OntoGraf SPARQL Query								
SPARQL query:								
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count(distinct ?x) as ?Inverse_Property_Count) {?x a owl:InverseProperty}</http:></http:></http:></http:>								
Inverse_Property_Count								
"0"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>								
Execute								
To use the reasoner click Reasoner->Start reasoner Show Inferences								

Figure 5.10: Enumeration of Inverse Properties Density

e. Enumeration of Functional Properties Density

To extract density of functional properties from the given ontology, following SPARQL query will be executed as shown in figure 5.11. Required prefixes are added.

SELECT (count (DISTINCT ?x) AS ? Functional_Property_Count) {?x a owl: Functional Property}



Figure 5.11: Enumeration of Functional Properties Density

f. Enumeration of Symmetric Properties Density

To extract density of symmetric properties from the given ontology, following SPARQL query will be executed as shown in figure 5.12. Required prefixes are added.

SELECT (count(DISTINCT ?x) AS ? Symmetric_Property_Count) {?x a owl:Symmetric Property}



Figure 5.12: Enumeration of Symmetric Properties Density

g. Enumeration of Asymmetric Properties Density

To extract density of asymmetric properties from the given ontology, following SPARQL query will be executed as shown in figure 5.13. Required prefixes are added.

SELECT (count(DISTINCT ?x) AS ? Asymmetric_Property_Count) {?x a owl:Asymmetric Property}

NIF-Cell (http://ontology.neuinfo.org/NIF/BiomaterialEntities/NIF-Cell.ow										
File Edit View Reasoner Tools Refactor Window Help										
Active Ontology Classes Individuals OntoGraf SPARQL Query										
SPARQL query:										
PREFIX rdf: <http: 02="" 1999="" 22-rdf-syntax-ns#="" www.w3.org=""> PREFIX owl: <http: 07="" 2002="" owl#="" www.w3.org=""> PREFIX xsd: <http: 2001="" www.w3.org="" xmlschema#=""> PREFIX rdfs: <http: 01="" 2000="" rdf-schema#="" www.w3.org=""> SELECT (count(distinct ?x) as ?Asymmetric_Property_Count) {?x a owl:AsymmetricProperty}</http:></http:></http:></http:>										
Asymmetric_Property_Count										
"0"^^ <http: 2001="" www.w3.org="" xmlschema#integer=""></http:>										
Execute										
To use the reasoner click Reasoner->Start reasoner Show Inferences										

Figure 5.13: Enumeration of Asymmetric Properties Density

h. Enumeration of Reflexive Properties Density

To extract density of reflexive properties from the given ontology, following SPARQL query will be executed as shown in figure 5.13. Required prefixes are added.

SELECT (count(DISTINCT ?x)

as ? Reflexive_Property_Count)

{?x a owl: Reflexive Property}



Figure 5.14: Enumeration of Reflexive Properties Density

5.2: Results in Tabular and Graphical Form

After executing respective SPARQL queries on ontologies, results are shown in table 5-1, 5-2 and 5-3. Graphical representation is shown from figure 5.15 to 5.29.

S N o	Domain	Ontology Title	Distinct Subject	Total Subject	Distinct Predicate	Total Predicat e	Distinct Object	Total Object	Distinct Triple	Total Triples
1		Uber Anatomy Ontology	380147	1752812	114	1752812	475374	1752812	1752812	1752812
2	Anatomy	Porifera Ontology	11066	45846	60	45846	13417	45846	45846	45846
3		Hymenoptera Anatomy Ontology	37919	153215	24	153215	39910	153215	153215	153215
4		NIF Cell	37470	143332	112	143332	45110	143332	143332	143332
5	Neuroscience	NIF Dysfunction	31287	132016	99	132016	39171	132016	132016	132016
6		Ontology for General Medical Science	3747	16467	51	16467	4689	16467	16467	16467
7	Medicine	Ontology for Medically Related Social Entities	5156	21172	61	21172	5944	21172	21172	21172
8	Experiments	NMR- Instrument Specific Component of Metabolomics Investigations	1403	4901	08	4901	1643	4901	4901	4901
9		Microarray Experimental Conditions	6049	23753	40	23753	7514	23753	23753	23753
10		STATistics Ontology	10996	45705	60	45705	13373	45705	45705	45705
11	Statistics	Ontology of Biological and Clinical Statistics	7983	33104	44	33104	36	33104	33104	33104

 Table 5-1: Comparative Triples Density of Biomedical Ontology



Figure 5.15: Subject, Object and Predicate of Anatomy Ontologies



Figure 5.16: Results of Subject, Predicate and Object of Neuroscience Ontologies



Figure 5.17: Results of Subject, Predicate and Object of Medicine Ontologies



Figure 5.18: Results of Subject, Predicate and Object of Experimental Ontologies



Figure 5.19: Results of Subject, Predicate and Object of Statistics Ontologies

Sr. No	Domain	Ontology Title	Distinct Super Classes	Distinct Subclass	Distinct Classes	Total Classes	Instances / Individu als	Total Instances
1		Uber Anatomy Ontology	12	24902	24914	144658	123704	676504
2	Anatomy	Porifera Ontology	8	970	978	4636	3031	18249
3		Hymenoptera Anatomy Ontology	4	2346	2350	16824	13991	65967
4	Nauroacianaa	NIF Cell	9	2977	2986	10041	5616	51926
5	Neuroscience	NIF Dysfunction	6	2961	2967	9933	5585	50719
6	Madising	Ontology for General Medical Science	6	207	213	806	1117	6312
7	Medicine	Ontology for Medically Related Social Entities	11	365	376	1431	1062	8253
8	Experiments	NMR-Instrument Specific Component of Metabolomics Investigations	11	290	301	870	306	1902
9		Microarray Experimental Conditions	6	230	236	1866	1302	10275
10		STATistics Ontology	7	969	976	4628	3002	18199
11	Statistics	Ontology of Biological and Clinical Statistics	8	751	759	3032	1556	13068

Table 5-2: Comparative Density of Concepts/Classes, Instances and Individuals





Figure 5.20: Comparative Density of Concepts/Classes, Instances and Individuals of Anatomy Ontologies

Figure 5.21: Comparative Density of Concepts/Classes, Instances and Individuals of Neuroscience Ontologies



Figure 5.22: Comparative Density of Concepts/Classes, Instances and Individuals of Medicine Ontologies



Figure 5.23: Comparative Density of Concepts/Classes, Instances and Individuals of Experimental Ontologies



Figure 5.24: Comparative Density of Concepts/Classes, Instances and Individuals of Statistics Ontologies

SN	Domain	Ontology Title	Object Property	Data Property	Annotati on Property	Inverse Property	Function al Property	Symmetr ic Property	Asymme tric Property	Reflexiv e Property
1		Uber Anatomy Ontology	183	0	199	0	0	6	0	0
2	Anatomy	Porifera Ontology	50	0	49	0	6	0	0	0
3		Hymenopte ra Anatomy Ontology	4	0	15	0	0	0	0	2
4		NIF Cell	85	0	178	0	3	4	0	9
5	Neuroscienc e	NIF Dysfunctio n	83	0	169	0	3	4	0	9
6		Ontology for General Medical Science	78	0	52	0	2	0	0	0
7	Medicine	Ontology for Medically Related Social Entities	70	0	59	0	10	0	5	0
8	Experiments	NMR- Instrument Specific Component of Metabolom ics Investigatio ns	11	0	34	0	0	0	0	0
9		Microarray Experiment al Conditions	85	42	44	0	8	0	0	0
10		STATistics Ontology	48	04	28	0	6	0	0	0
11	Statistics	Ontology of Biological and Clinical Statistics	36	06	19	0	9	0	0	0

Table 5-3: Comparative Density of Properties/Relations



Figure 5.25: Result of Properties of Anatomy Ontologies



Figure 5.26: Result of Properties of NeuroScience Ontologies



Figure 5.17: Result of Properties of Medicine Ontologies



Figure 5.28: Result of Properties of Experimental Ontologies



Figure 5.29: Result of Properties of Statistics Ontologies

5.3: Discussion on Experimental Results

We majorly generalize the attained experimental results into the major categories: triples, classes and properties. Giving each category equal weightages in triples total number of subjects, predicates and object are summed up. Similarly under the category of classes, total classes and individuals are added. Finally under the category of properties, all properties of specific biomedical ontology are added to get total properties count. At the end all classes, triples and properties are summed up to calculate total structural coverage of specific ontology and suggest the best one on structural basis.

Here we summarized the results with respect to categories of ontologies in tabular form. Firstly we concisely summarize the results of biomedical ontologies related to Anatomy including Uber Anatomy Ontology, Porifera Ontology and Hymenoptera Anatomy Ontology. In the second step we summarize the results of biomedical ontologies related to Neuroscience including NIF Cell and NIF Dysfunction. Then we summarize the results of biomedical ontologies related to Medicine including Ontology for General Medical Science and Ontology for Medically Related Social Entities. After summarizing the results of Medicine ontologies including NMR-Instrument Specific Component of Metabolomics Investigations and Microarray Experimental Conditions, results of Experimental medical ontologies are concisely discussed. Lastly we summarize the results of biomedical ontologies related to Statistic Ontology and Statistics including Ontology of Biological and Clinical Statistics.

5.3.1 Result of Anatomy Ontologies

As discussed earlier, the summarized result of biomedical ontologies related to Anatomy domain is shown in table below and in figure below. From the table and graph it is clear that Uber Anatomy Ontology possess the highest coverage of structural components among the candidate Anatomy Ontologies.

Sr.	Ontology Title	Triples	Classes/Individuals	Properties	Total
No					
1	Uber Anatomy Ontology	2608347	148618	394	2757359
2	Porifera Ontology	70389	4009	105	74503
3	Hymenoptera Anatomy	231068	16341	21	247430
	Ontology				

Table 5-4: Summarized Result of Anatomy Ontologies



Figure 2.30: Summarized Result of Anatomy Ontologies
5.3.2 Result of NeuroScience Ontologies

As discussed earlier, the summarized result of biomedical ontologies related to Neuroscience domain is shown in table below and in figure below. From the table and graph it is clear that NIF Cell Ontology possess the highest coverage of structural components between the candidate Neuroscience Ontologies.

Sr. No	Ontology Title	Triples	Classes/Individuals	Properties	Total
1	NIF Cell	226024	8602	279	234905
2	NIF Dysfunction	202573	8552	268	211393

Table 5-5: Summarized Result of NeuroScience Ontologies



Figure 5.31: Summarized Result of NeuroScience Ontologies

5.3.3 Result of Medicine Ontologies

As discussed earlier, the summarized result of biomedical ontologies related to Medicine domain is shown in table below and in figure below. From the table and graph it is clear that Ontology for Medically Related Social Entities possess the highest coverage of structural components between the candidate Medicine Ontologies.

Sr.	Ontology Title	Triples	Classes/Individuals	Properties	Total
No					
1	Ontology for General	24954	1330	132	26416
	Medical Science				
2	Ontology for Medically	32333	1438	144	33915
	Related Social Entities				

Table 5-6: Summarized Result of Medicine Ontologies



Figure 5.32: Summarized Result of Medicine Ontologies

5.3.4 Result of Experimental Ontologies

As discussed earlier, the summarized result of biomedical ontologies related to Experimental domain is shown in table below and in figure below. From the table and graph it is clear that Microarray Experimental Conditions possess the highest coverage of structural components between the candidate Experimental Ontologies.

Sr.	Ontology Title	Triples	Classes/Individuals	Properties	Total
No					
1	NMR-Instrument Specific	7955	607	45	8607
	Component of				
	Metabolomics				
	Investigations				
2	Microarray Experimental Conditions	37356	1538	179	39073

 Table 5-7: Summarized Result of Experimental Ontologies

A Framework for Evaluation of Biomedical Ontologies based on Structural Components



Figure 5.33: Summarized Result of Experimental Ontologies

5.3.5 Result of Statistic Ontologies

As discussed earlier, the summarized result of biomedical ontologies related to statistics domain is shown in table below and in figure below. From the table and graph it is clear that STATistics Ontology possess the highest coverage of structural components between the candidate statistics ontologies.

Sr.	Ontology Title	Triples	Classes/Individuals	Properties	Total
No					
1	STATistics Ontology	70134	3978	86	74198
2	Ontology of Biological	41167	2315	70	43552
	and Clinical Statistics				

Table 5-8: Summarized Result of Statistics Ontologies



Figure 5.34: Summarized Result of Statistics Ontologies

5.4: Comparison of Proposed Technique with other Existing Approaches

5.4.1 Comparison with Ontobee Approach

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One approach found in literature with title Ontobee [36] extract the statistics of ontologies. It only focus on four components namely classes, object properties, datatype properties and annotation properties whereas our proposed approach as discussed earlier include these components along with triples including subject, object and predicate, superclasses, subclasses and individuals/instances. Furthermore the properties are include functional, symmetric, asymmetric and reflexive properties. So our framework describe the structural composition of ontologies in a greater extent as compared to Ontobee. Summary of this is shown in Table 5-9.

able 5-9: Comparison of Proj	posed Evaluation	Approach and O	пювее

	Subjects	Predicates	Objects	Classes	Individuals	Object Properties	Datatype Properties	Annotation Properties	Other Properties
Proposed Framework			\checkmark	\checkmark	\checkmark	\checkmark	\checkmark	\checkmark	\checkmark
OntoBee				\checkmark		\checkmark	\checkmark	\checkmark	

5.4.2 Comparison with OntoQA Approach

OntoQA (Ontology Quality Assurance) [38] is a feature-based ontology evaluation tool. The metrics of OntoQA are divided into two categories: instance metrics and schema metrics. Instance metrics deals with the organization of instances within the ontology. Under the category of schema metrics, the sub-metrics are relationship richness, inheritance richness and attribute richness. Whereas in instance metrics, the sub-metrics are class richness, class connectivity, class importance, cohesion and relationship richness. It is quite simple evaluation tool as it requires less user involvement.

Majorly OntoQA only focus on three components namely classes, relations and instances whereas our proposed approach as discussed earlier include these components along with triples including subject, object and predicate, superclasses, subclasses and individuals/instances. Furthermore the properties are include functional, symmetric, asymmetric and reflexive properties. So our framework describe the structural composition of ontologies in a greater extent as compared to OntoQA. Summary of this is shown in Table 5-10.

	Subjects	Predicates	Objects	Classes	Individuals	Object Properties	Datatype Properties	Annotation Properties	Other Properties	Relations
Proposed Framework	\checkmark	\checkmark	\checkmark			\checkmark	\checkmark	\checkmark	\checkmark	
OntoQA				\checkmark	\checkmark					

 Table 5-10: Comparison of Proposed Evaluation Approach and OntoQA

CHAPTER 6: CONCLUSION AND FUTURE RECOMMENDATIONS

This chapter summarizes the entire research effort of this dissertation. It comprises of concludes, pros and cons. Finally future recommendations have been advised at the end.

6.1: Conclusion

Ontologies are recognized as a valuable mean of representing domain knowledge. Especially it becomes the core component of Semantic Web. The ontologies which have been developed require to be evaluated against certain criteria to ensure their quality and appropriateness about domain representation. That is whether they represent the domain concepts properly. The need of ontology evaluation is highly necessary as users of ontologies are facing problems in selecting the appropriate ontology, which satisfies their requirement, among different sets of ontologies. The other factor which makes ontology evaluation a crucial task is ontology reuse in different applications and their further development.

As ontologies are core mean of formally representing and sharing domain knowledge, among other ontology evaluation frameworks, there should be certain basic structural building blocks in their composition. For the sake of this requirement quality of biomedical ontologies has been assessed in current work with respect to population of their structural components. The biomedical ontologies which have greater component density have high expressivity of domain concepts. These ontologies having high score demonstrate their potential and significance for reusability and interoperability with other existing ontologies and related application.

The criteria which we use comprises of triples (subjects, predicates and objects), classes (or concepts), individuals (or instances) and properties including object properties, data properties, annotation properties, functional properties, symmetric properties, asymmetric properties and reflexive properties. The biomedical ontologies which are selected are in .owl form that is written in Ontology Web Language (OWL). These ontologies are imported in Protégé 4.3, one of the ontology building and ontology editing tool. For extraction of results SPARQL Protocol and RDF Query Language (SPARQL) queries are applied on each biomedical ontology.

The ontologies are imported from The Open Biological and Biomedical Ontologies (http://www.obofoundry.org/). OBO is one of the major ontology repository containing various biomedical ontologies in .obo format and .owl format. Three of the imported biomedical ontologies are from the domain of anatomy, two of them are from neuroscience domain, two from the field of medicine, two ontologies from biomedical experimental domain and two from medical statistics.

The results of experiments are shown in both detailed and summarized form. Only distinct values of all structural components are considered. Three major categories are formed against which candidate ontologies are evaluated. These major categories include triples, classes and properties. Giving each category equal weightages in triples total number of subjects, predicates and object are summed up. Similarly under the category of classes, total classes and individuals are added. Finally under the category of properties, all properties of specific biomedical ontology are added to get total properties count. After describing detailed results in both tabular and graphical way, ontologies from same domain are compared with each other to get the appropriate and relative best ontology by summing up all classes, triples and properties to calculate total structural coverage of specific ontology and suggest the best one on structural basis.

6.2: Advantages of Proposed Work

Being considering the major structural components for evaluation purposes, it makes very clear and discrete ontology evaluation methodology. Experimentations depict comprehensive and understandable results which help in deciding among many ontologies which one is most appropriate and best with respect to structural components. These results are used by researchers and domain experts for importing and using the best ontologies in their applications.

6.3: Limitation of Proposed Work

One major limitation of our work is that or experimentation is done only on biomedical ontologies that are .owl (Ontology Web Language) based. But as the basic components of ontologies are about same, the proposed ontology evaluation framework can be applied on other formats of ontologies like .obo (The Open Biological and Biomedical Ontologies). Further on biomedical ontologies which are supported by Protégé 4.3 are selected for experimentations to extract results.

6.4: Future Recommendations

As our results show adequate improvements in ontological evaluation, this technique can be utilized in developing web-based intelligent system for evaluation of ontology. Further on the criteria can be applied to other domains than biomedical. As obvious the proposed work is based on structural constructs, it can be further integrated with other metrics based on both quality and quantity criteria. Furthermore this structural framework can be integrated with other semantic oriented evaluation techniques of biomedical ontologies to create such a comprehensive evaluation framework which scope every aspect of ontologies.

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