Semantic Biomedical Services

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Proposition

With the growing trends towards multi-organizational and inter-disciplinary nature of research and practice in Biomedical domain, there is a strong need for unifying the information resources in a coherent fashion. The information heterogeneity problem ranges from gene and molecular databases to public health and governmental resources. Semantic Web services provide a key to solve such problems. Semantic Web service is a technology that enables wrapping Web services with Semantic Web based ontological abstractions.

In Biomedical Informatics domain, researchers have been articulating and using ontologies for decades. The goal then was to perform better information indexing and retrieval. However, the current trends are towards generating OWL (Web Ontology Language) based descriptions for existing Biomedical ontologies [1][2].

The next logical step is to use these ontologies as "mediators" to allow various Biomedical applications to interoperate smoothly and seamlessly via Semantic Web services (referred from now on as Semantic Biomedical Services (SBS) in this paper).

Skepticism

One of the problems often pointed out by Semantic Web skeptics is that of "ontology heterogeneity". With the open democratic environment such as Web, it is quite difficult to achieve standardization of conceptualizations. However, if considering specialized domains such as Biomedicine, it becomes evident that despite of existence of several different standards and ontologies (HL7, CPT, SNOMED etc) there is an increased degree of uniformity, essentially through integration and mapping efforts via an upper ontology (UMLS).

Hence, given the current positive strides in OWL standardization, Web service maturity and Biomedical ontology integration and respective OWL conversions, the wide large-scale realization of these technologies in real-world health-care and research enterprises is very much a reality.

SBS Sketch

Semantic Biomedical Services would not be a new genre of "magical" applications that would solve all the heterogeneity and interoperability problems, however, these services would merely provide intelligent abstraction layer to allow machine interpretation and reasoning. The kind of problems solved by such Web service and ontology based solutions are

- Uniform access: Given that SBS would be based on open Web standards having wide implementations, we can envision seamless integration, irrespective of programming languages or platform.

- Ambiguity: A common agreed-upon ontology across SBS would allow effective matching via explicit semantics and hence ease the process of chaining several applications/services in a workflow fashion.

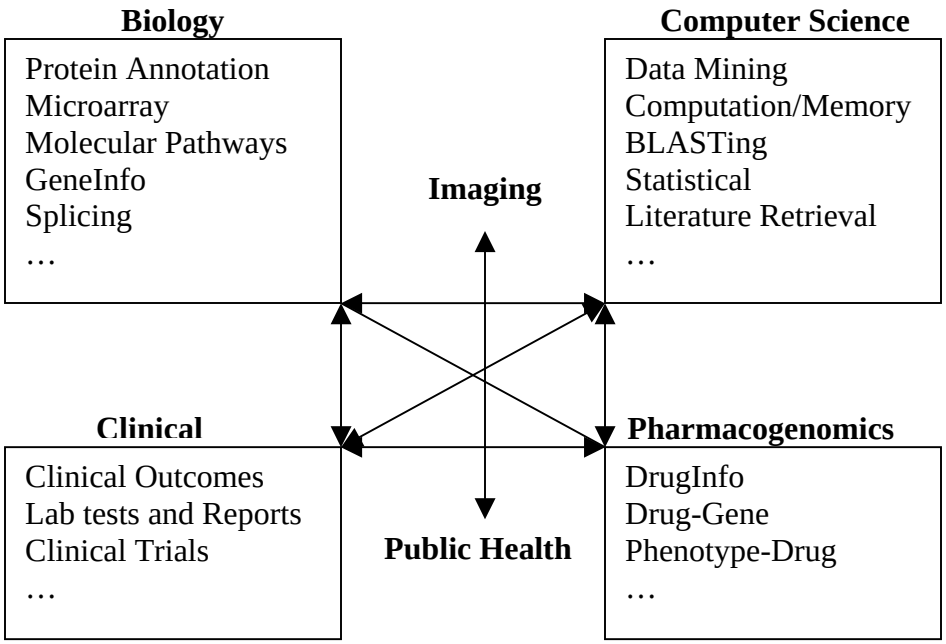

Fig 1. "Information" services in Biomedical research settings – Potential semantic Biomedical services

In current Biomedical research settings, several types of information services exist, however, they are highly localized and customized as per their respective needs. Figure 1. sketches at an abstract level, the possible kind of services that could be provisioned by a given Biomedical research facility. The current trends are towards NxN kind of cross-discipline research and investigation, the impact of discovery in one field is much wider. Most of the interesting discoveries are happening at the intersection of these fields.

SBS in Action

One of the requirements to represent the services semantically is to provide formal ontological descriptions for the service parameters and conditions. However, creating isolated local ontologies is not a good solution, given that the goal is to achieve wide scale interoperability, also such "stand-alone" ontologies defeats the purpose of using ontologies in first place.

Table 1. illustrates some of the widely-accepted ontologies in Biomedical domain that facilitates the creation of semantic descriptions for SBS parameters.

Ontology	Domain/Purpose	Possible use (Fig 1.)	OWL conversion status
Gene Ontology	Gene product annotation and uniform naming	GeneInfo Protein Annotation Microarray BLAST Data Mining	UP*
UMLS-Unified Medical Language System	Medical domain/Natural Language Processing	Clinical Trials/Outcomes Lab tests	Some parts of UMLS Meta-thesaurus have been ported to OWL
BioPAX	Biological Pathway modeling	Molecular Pathways	AV*
GALEN	Medical domain/automated reasoning and inference	EMRs Clinical Trials/Outcomes	AV*
FMA-Foundational Model of Anatomy	Anatomy/automated reasoning and visualization	EMRs Clincal Trials/Outcomes	UP*
SNOMED-Systematized Nomenclature of Medicine	Medical domain/terminology and coding	EMRs Clinical Trials/Outcomes	UP*
MGED- Microarray Gene Expression Data	Microarray data	Microarray	AV*

*UP= Under Progress
*AV=Available

Table 1. Biomedical ontologies, applications and corresponding OWL availability.

Challenges

Given the current state of technologies and standards, there are several additional challenges that need to be faced to realize the notion of Semantic Biomedical services.

- Ontology conversion: As easy as it sounds, the task of porting ontologies to OWL is not just writing an "XSLT" kind of syntactic conversion module. The first challenge is of understanding the limitations of what can and cannot be expressed in OWL or one of its flavors. The further challenges are providing explicit semantics to concepts, which are vaguely described in the ontologies (which essentially can have number of interpretations depending upon the context).
- Tool limitations: Given the recent standardization of OWL and Web services, the tools still aren't equally mature enough to be used for critical applications.

- Scalability is an important aspect with respect to reasoning with large Biomedical ontologies. Even with Web services technology, the issues of scalability and transaction support are still in research phase.
- Privacy/Security: One of the major concerns with Web wide application of these services is that of privacy and security. Despite of stringent healthcare privacy laws in countries like United States, we cannot assure that the data will be completely private and secure given the vulnerability of systems in real world.

Conclusion

The current Biomedical research settings tend to be highly interdisciplinary, requiring wide and possibly global collaborations. Semantic Web services and Biomedical ontologies provide a key to solve the interoperability and the information heterogeneity problem for Biomedical applications in such settings. The current efforts are underway towards realizing Semantic Biomedical services, however, several hard problems need to be resolved to achieve the grand vision of Web scale seamless interoperability for Biomedical applications.

References

[1] Vipul Kashyap, Alexander Borgida: Representing the UMLS Semantic Network Using OWL: (Or "What's in a Semantic Web Link?"). International Semantic Web Conference 2003: 1-16

[2] C.J. Wroe, R. Stevens, C.A. Goble, M. Ashburner, A Methodology to Migrate the Gene Ontology to a Description Logic Environment Using DAML+OIL Pacific Symposium on Biocomputing 8:624-635(2003).