

Brain: Biomedical Knowledge Manipulation

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Received on XXXXX; revised on XXXXX; accepted on XXXXX

Associate Editor: XXXXXXXX

ABSTRACT

Summary: This section should summarize the purpose/novel features of the program in one or two sentences.

Availability and Implementation: This section should state software availability if the paper focuses mainly on software development or on the implementation of an algorithm. Examples are: 'Freely available on the web at <http://www.example.org>. Website implemented in Perl, MySQL and Apache, with all major browsers supported'; or 'Source code and binaries freely available for download at URL, implemented in C++ and supported on linux and MS Windows'. The complete address (URL) should be given. If the manuscript describes new software tools or the implementation of novel algorithms the software must be freely available to non-commercial users. Authors must also ensure that the software is available for a full TWO YEARS following publication. The editors of Bioinformatics encourage authors to make their source code available and, if possible, to provide access through an open source license see www.opensource.org for examples.

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Relational databases and ontologies hold most of the available structured biomedical information. The content of these repositories is often extracted from scientific literature by manual curation with the help of text-mining tools. The transformation from raw text into structured data is most important, as the curated information can then be classified, managed and queried more easily. Databases facilitate the re-use of previous work in a computer-friendly manner and support the biomedical knowledge to scale-up. The availability of structured data enables the formulation of queries with the Structured Query Language (SQL) in order to retrieve the desired information over relational databases. For instance, it is possible to quickly find the cellular location of a protein just by looking at its entry on Uniprot and without having to read several scientific articles on the topic. The query language used and the structure of the database are the factors limiting the complexity of the questions that can be formulated over the biomedical information. For example, relational databases are powerful to store values about instance data such as the sequence or the name of a gene product, but are daunting to use to represent hierarchical structures (ref) such as the Gene Ontology. Therefore queries resolving around

hierarchies are going to be complicated to formulate using SQL and will not necessarily deliver a complete answer. For instance, simple queries such as retrieving the list of all the mammals from the NCBI taxonomy involves retrieving first the direct descendants of the mammals and for each of them recursively retrieving their descendants. This type of construct is not trivial to implement and is not yet supported by open-source solution like MySQL or PostgreSQL.

Ontologies and databases in biology are somehow used differently than in other fields. They essentially serve as read-only resources where the researchers come to browse or download the content. The majority of the time, no transactions or updates are happening as opposed to services such as Amazon or Ebay.

- The future of biomedical databases (ELIXIR) is leverage of their content via interoperability with other resources. OWL provide the means to do this, databases don't.
- Features: Creation and storage of OWL knowledge-bases. Import of external knowledge-bases/ontologies. Simplification of interaction in regards to OWL-API. Fast classification time (Elk reasoner). Support complex queries via inference. Fast and suitable to be used in production.
- Evaluation: MySQL build of Go is compared versus OWL build of GO (identical content - different representation). A series of biomedical questions will be answered in SQL and OWL respectively on the MySQL and on the OWL ontology. Comparison of performances – Brain is fast and scalable (thread friendly implementation).

Problems: - Need to be able to formulate complex queries – hidden/implicit knowledge - inference. - Need for interoperability among resources – semantic web technologies. - Need to be able to leverage resources – benefit from integration - Relational databases are struggling with biomedical hierarchical representations. Instance versus classes - Databases in biology are used as read-only resources - Databases don't support inference natively but it could be overcome by a pre-processing step (similar to a classification for a knowledge base).

Solution: - Semantic Web technologies could be slow – OWL 2 EL. - Advantages of triple -stores – none, they still are relational flat data. - Simplifying interaction on OWL-API, tracktable problem. Focusing on biology, not computer science. - More and more ontologies are represented in OWL.

OWL 2 EL building blocks: - Individual, properties, classes - Axioms - Reasoner

- relation with description logic - trackability argument + definition - OWL – query and representation language - table comparison query + times

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ACKNOWLEDGEMENT

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