

Brain: Biomedical Knowledge Manipulation

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ABSTRACT

Summary: Brain is a software library facilitating the creation and manipulation of ontologies and knowledge-bases represented in the Web Ontology Language (OWL).

Availability and Implementation: The Java source code and the library are freely available at <https://github.com/loopasam/Brain> and on the Maven Central repository (GroupID: uk.ac.ebi.brain). The documentation is available at <https://github.com/loopasam/Brain/wiki>.

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Supplementary information: Supported features and a comparative table are available at the journal's web site.

1 MOTIVATION

Knowledge bases, a concept from computer science (see Krötzsch *et al.*, 2012 for an introduction), could be a solution to improve the interoperability and the value of the large amount of biomedical information available online. At the time of writing, a few options are available to handle such knowledge bases: Complex libraries as the OWL-API (Horridge and Bechhofer, 2011) or didactic graphical user interfaces such as Protege (Stanford Center for Biomedical Informatics Research, 2012) or TopBraid (ref). An intermediary framework, OWLTools (Mungall, 2012), provides some methods to query biomedical knowledge bases, but the interaction with the library is mostly done via command-lines which limits the scale of projects that can be build with it. Brain - the software library presented in this manuscript addresses this matter and provides a comprehensive and simplified interface, dedicated to the programmatic creation and query of biomedical knowledge bases. The library aims at bridging the gap between graphical user interfaces and the OWL-API and is particularly usefull to develop web applications. Brain has a particular focus on the EL profile of OWL, as it covers the majority of biomedical use-cases and enables good performances and scalability.

2 SCALABLE KNOWLEDGE BASES

The Web Ontology Language (OWL) derives from Description Logic and has been designed to capture the knowledge of a domain of interest in the form of a structured vocabulary (W3C OWL Working Group, 2009). This feature makes it particularly interesting from the perspective of the life sciences, since a number

of ontologies and classification schemes have been developed from the origin of the discipline. Brain focuses on a particular profile of OWL, called EL which consists of a subset of the constructs available in the original language (Motik *et al.*, 2009). This profile is designed to be *tractable*, meaning that the axioms available have a polynomial complexity and are therefore easier to compute than the full version of OWL. Brain primarily supports the OWL 2 EL profile for its computational properties and suitability for real-life biomedical applications, where millions of axioms could be potentially extracted from complex repositories such as ChEMBL (Gaulton *et al.*, 2012). Moreover, the EL profile is expressive enough to cover a good portion of biomedical knowledge: Most of Open Biomedical Ontologies (OBO) such as the Gene Ontology (GO - Ashburner *et al.*, 2000) or the Chemical Entities of Biological Interest (ChEBI - De Matos *et al.*, 2010) are already included in this profile, opening doors to large-scale meaningful data integration. Brain builds on the top of Elk, a fast reasoner dedicated to EL ontologies (Kazakov *et al.*, 2011). Elk shows good performances at handling large datasets and offers the possibility to run some reasoning tasks in parallel; therefore clusters or multicore architecture can scale the speed of reasoning as more data are added to the knowledge base. Brain wraps and simplifies the interaction with Elk while still leaving the possibility to fine tune the configuration for advanced users.

3 PROGRAMMATIC FEATURES

Brain is implemented as a facade, leveraging the access to the OWL-API and providing a series of convenience methods for common use-cases encountered in the biomedical domain. In order to simplify the interaction with the OWL-API, Brain has been designed according to a series of concepts described below.

3.1 Unique ontology

An instance of a Brain object hold a reference to only one knowledge base. It is yet possible to import some external ontologies, either stored locally or via a network but Brain will always merge the added information to the existing ontology.

3.2 Unique names

The names of OWL entities handled by a Brain object have to be unique. This choice is motivated by the fact that Brain hides as much as possible the cumbersome interaction with prefixes, IRI and URIs.

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3.3 Typeless interaction

The interaction with the library relies on the user-friendly Manchester syntax entered as string (Horridge *et al.*, 2006). This choice permits to move away from the cumbersome creation of Java objects and is particularly suitable in a web server environment where requests are likely to be typeless. Using strings as input also speeds the production and flexibility of the code written to move to an OWL representation from a relational or flat-file database for example.

3.4 Error-handling

Because the interaction with Brain is built around strings rather than Java objects, a special care has to be put on exceptions handling in order to safely maintain the correct execution of the program. Brain throws different types of error tailored to the operation performed by the user. This feature is mandatory in large applications and helps to maintain the consistency of the underlying knowledge base.

Example of axiom implemented using Brain

Natural language: A nucleus is part of some cells.

Description Logic: $\text{Nucleus} \sqsubseteq \text{part-of}.\text{Cell}$

OWL (Manchester syntax): $\text{Nucleus} \text{ subClassOf } \text{part-of} \text{ some Cell}$

Brain implementation:

OWL expressions can be entered with the user-friendly Manchester syntax (Horridge *et al.*, 2006) and the name of the classes are displayed in a convenient way. More complex applications, requiring a deeper control over the ontology, can take advantage the OWL-API but require proficiency in Java and can be daunting for casual users. OWL-API deals with all aspects of ontology generation for semantic web purposes, nonetheless the level of granularity can be cumbersome for some biomedical applications. Brain aims at filling the gap between the OWL-API and graphical interfaces: It is implemented as a facade, providing a series of convenience methods for common use-cases encountered in the biomedical domain and leveraging the access to the OWL-API.

The full list of currently supported constructs is available in the supplementary material. Brain relies on the intuitive and explicit Manchester syntax to formulate OWL class expressions, just as in the Protege editor. The interaction with the OWL-API centers around strings handling rather than Java objects, making Brain suitable to parse and answer requests in the context of a web service or an OWL end-point for instance. Using strings as input also speeds the production and flexibility of the code written to move to an OWL representation from a relational or flat-file database for example. The library supports the loading and referencing of external ontologies in order to integrate and reason over data from different sources. An important feature of Brain is the query

mechanism, which is similar to that implemented in Protege. Powerful questions can be formulated over the knowledge-base using the Manchester syntax, abstracting away complex interaction with the Java object provided by the OWL-API. An example of question answering over the GO using Brain is compared against a traditional SQL query in the supplementary material.

4 CONCLUSION

Brain is an open-source Java library designed to build and query biomedical knowledge bases or OWL ontologies. The library is centered on the EL profile, and designed to be suitable and scalable for biomedical knowledge representation. The convenience methods provided by Brain should simplify the development of biomedical knowledge-bases and allow developers to increase their productivity while effectively dealing with data integration challenges.

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