When can SWT considered to be successful?

- Only if Berners-Lee’s vision of the SemWeb (as in the SciAm 2001 paper) has been realised?
- Absolute measures? e.g.,
  - User’s (browsing and buying) usage of Amazon’s recommender system with and without SWT
  - Information retrieval: compare precision and recall between a statistics-based and a SWT-based implementation of a document system
  - Feasibility and performance of a set of user queries posed to a RDBMS and its RDF-ised version
- Relative measures
  - According to whom is it a success?
    - philosopher, logician, engineer, domain expert, CEO...
  - What was taken as baseline material? e.g.,
    - from string search in a digital library to ontology-annotated sorting of query answer
    - no or clustering-based instance classification to a SWT one with OWL-based knowledge bases
SWT challenges or failures?

- **Challenge**: solution to problem y not possible yet (or very difficult to achieve) with current SWT, but in theory is (expected to be) feasible
- **Failure**: technology x claims to solve problem y but it does not and will not do so, or technology x is developed for a non-existing problem but does not solve real problems
  - Is y one that, at least in theory, can be solved with SWT?
  - Was y described too broadly, so that it solves only a subset of the cases?
  - Were there perhaps additional requirements put on a solution?
- Are disconnected technologies with ad-hoc patches a challenge to solve or a failure in devising a generic suite?
- A failure according to one may be considered a challenge by another
- Offer and demand, perceptions, perspectives, expectations

---

Instance classification with protein phosphatases (Wolstencroft et al, 2007)

- **The setting**:
  - Lots of sequence data in data silos that needs to be enriched with biological knowledge
  - Need to organise and classify genes and proteins into functional groups to compare typical properties across species
- **The problems**:
  - There is no proper, real life, use case that demonstrates the benefits of DL reasoning services such as taxonomic and instance classification
  - Limitations of traditional similarity methods, and automated protein motif and domain matching
  - Automation of p-domain analysis, but not for its interpretation (i.e., detects presence but not consequences for sub-family membership)

---

Idea

- Maybe OWL reasoning can help with the interpretation of the analysis results:
  - That it does the classification of the (family of) proteins as good as a human expert for organisms x (in casu, human)
  - That the approach is ‘transportable’ to classification of the (family of) proteins in another organism of which much less is known (in casu, Aspergillus fumigatus), hence make predictions for those instances by means of classifying them
- Use taxonomic classification and instance classification reasoning services

---

How it can be done

- Develop ontology for the subject domain, in OWL
  - Extract knowledge from peer-reviewed literature
  - Protein phosphatases; e.g., Class R5Phosphatase Complete
    - (Protein and (hasDomain two TyrosinePhosphataseCatalyticDomain) and (hasDomain some TransmembraneDomain) and (hasDomain some FibronectinDomain) and (hasDomain some CarbonicAnhydraseDomain) and hasDomain only (TyrosinePhosphataseCatalyticDomain and TransmembraneDomain and CarbonicAnhydraseDomain))
  - Obtain instance data
    - Process protein sequences by InterProScan
    - Transform into OWL
- Put it together in some system with a reasoner
  - InstanceStore
  - Racer reasoner
Results

• Human phosphatases:
  • The reasoner as good as human expert classification
  • Identification of additional p-domains, refined the classification into further subtypes
• A. fumigatus phosphatases:
  • Some phosphatases did not fit in any class, representing differences between the human and A. fumigatus protein families
  • Identification of a novel type of calcineurin phosphatase (has extra domain, like in other pathogenic fungi)
• Overall: demonstration that ontology-based approach with automated reasoning has some advantages over (in addition to the) existing technologies & human labour, and resulted in discovery of novel biological information

Web-based, graphical, ontology-based querying of lots of data (Calvanese et al, 2010)

• The setting:
  • Large amounts of data available on the Web, which can be accessed by canned or precomputed queries presented via web forms, or SQL
  • Domain expert wants more flexibility in data analysis and hypothesis testing, and independence from the sysadmin to do the queries for them
• The problems:
  • There is no proper, real life, use case that demonstrates the benefits of scalable, user-usable, Ontology-Based Data Access
  • That one has to know how the data is stored, instead of concerning oneself with what kind of information is in the database
  • Domain expert-unfriendly query mechanisms (SQL, SPARQL)

Idea

• Ontology-Based Data Access, to achieve data access at the ‘what-layer’, i.e., adding a semantic layer to the database
• Web-based, like most other bioinformatics resources
• Graphical querying to make it usable by the domain expert
• Usage of, mainly, reasoning services for querying the ontology and the data

How it can be done

• Develop ontology of the subject domain, in OWL
  • Reverse engineering existing database HGT-DB ([http://genomes.urv.cat/HGT-DB/](http://genomes.urv.cat/HGT-DB/)), further manual improvements to create a proper conceptual data model
  • Simplify this conceptual data model into the appropriate OWL language (DL-LiteA, which is roughly OWL 2 QL)
• Create mappings between the terms in the ontology to SQL queries over the database
  • Using the OBDA Plugin for Protégé
  • Oracle database (can also be PostgreSQL, DB2, …), 4GB genomics database (HGT-DB), tables with 16-46 columns
• Connect this to an OBDA-enabled reasoner
  • In this case: QUONTO (but can be others)
Introduction

Successes

Challenges

Summary

Architecture

Formalisation of the graphical elements

Example: Diagram – DL-lite_A correspondence

Example: mapping concepts & relations of the Ontology to SQL query over the relational database
Queries

- SPARQL queries for conjunctions and equalities
- Epistemic queries in *EQL-Lite* for constraints involving inequalities and string matching
  - Imposes constraints on top of the certain answers retrieved by a *DL-LiteA* conjunctive query
  - Result obtained by:
  i. computing the certain answers for the CQ \( q(\vec{y}) \leftarrow \text{conj}(\vec{z}) \)
     (with \( \text{conj}(\vec{z}) \) the conjunction of atoms, and \( \vec{y} \) a vector comprising the variables in \( \vec{x} \) and in \( \vec{w} \)),
  ii. filtering the resulting tuples according to the constraint expression \( \text{cons}(\vec{w}) \), and
  iii. projecting onto \( \vec{x} \) (a vector comprising the variables corresponding to the highlighted nodes in the query pane)

Results

- Demo of the *Wonder* system (Web-ONtology baseD Extraction of Relational data)
  - Builds upon the theory, technology, and implementation developed for Ontology-Based Data Access
  - Graphical ontology browsing, query formulation, and query execution in a Web browser
  - Rigorous formal characterisation and uses a coupling with an OWL file
  - (U)CQs (in SPARQL syntax) and EQL-Lite queries managed by the DIG-QuOnto reasoner
  - Performance good, GUI insignificant influence on performance
  - Usability testing: usable, and domain experts came up with a range of new queries to analyse the data

Additional features

- *Wonder* currently focuses on querying one database
- OBDA architecture allows for querying incomplete data (data integration scenario\(^1\))
- Querying of the application ontology itself, as well as a combination of querying the ontology and the data\(^2\)
  - in certain settings, possible to include queries that use the knowledge in the ontology for which there is no data in the database, and still retrieve the right results

---

Sample query in OBDA Plugin

\[
q(x) := \text{Device}(x), \text{assistsWith}(x, y), \text{UpperLimbMobility}(y)
\]

A few general issues

- RDF triple stores vs. RDBMSs vs OWL ABoxes in memory; more generally:
  - Making ‘legacy’ (operational) systems ‘Semantic Web compliant’
  - Add a ‘wrapper’ over the legacy system so that from the outside it looks like it uses SWT
- How to integrate rules other than at instance level
- Modularization
- Semantics-based language transformations
- Coordination among tools with different functionalities

Language limitations considerations

- Known trade-offs between expressiveness and computational complexity
- Different ontology developers and their scopes (and purposes of the ontologies):
  - to some, there is more in OWL/OWL2 than needed and used (recollect slide 32 of lecture 8)
  - to some, there is not enough (some of the limitations and extensions discussed in lecture 2, 6 and 7)
- From a logician’s perspective, language limitations are not failures per sé, only challenges to find the more interesting and useful combinations of features
- From a modeller’s perspective, the trade-offs can be such that it is deemed a failure with respect to the expectations and application needs

Limitations as identified by users/modellers (Schulz et al, 2009)

- \(n\)-ary relations, where \(n > 2\)
- “Hepatitis hasSymptom Fever in most but not all cases”
  - What about doing it with probabilistic default knowledge (lecture 7)?
  - \((\psi | \phi)[l, u]\) as “generally, if an object belongs to \(\phi\), then it belongs to \(\psi\) with a probability in \([l, u]\)”
  - e.g., \((\exists \text{hasSymptom.Fever} | \text{Hepatitis})[1,1]\)
- “In 2000, worldwide prevalence of diabetes mellitus was 2.8%”
  - Probabilistic, or arithmetic, or what have we?
  - First, it assumes some class \text{Human} and a class \text{HumanDiabetesMellitus}, where some of the instances of the former have (are \text{bearerOf}) an instance of the latter
  - Second, we have some notion of prevalence, but what is it associated to (a property of)? of the human \text{population} in the world, not a property of an individual human
Limitations as identified by users/modellers (Schulz et al, 2009)

• ... Diabetes example continued
  • Authors’ proposal to put it in the ABox with arithmetic operators, e.g. \( |\text{DiabeticHuman}| = 0.028 \)
  • Another option: put in TBox with a data property, e.g., \( \text{HumanDiabetesMellitus} \sqsubseteq \exists \text{hasPrevalence} \cdot \text{real} \)
  • Yet another: represent the probability of a human having diabetes mellitus
  • What are the pros and cons of each option w.r.t. subject domain semantics, Ontology, and the ontology languages?

Problems with Drug Abuse Prevention (in SNOMED CT)

• DrugAbusePrevention \sqsubseteq \text{Procedure} \sqcap \exists \text{hasFocus.DrugAbuse}
• DrugAbusePrevention \equiv \text{Procedure} \sqcap \exists \text{hasParticipant.Person} \sqcap \exists \text{causes.} \left( \text{State} \sqcap \exists \text{hasParticipant.} \neg \right. \sqcap \text{DrugAbuse})

Scenarios

1. Supporting the ontology development process
2. Classification
3. Model checking (violation)
4. Finding gaps in an ontology & discovering new relations
   • Deriving types and relations from instance-level data
   • Computing derived relations at the type level
5. Comparison of two ontologies ([logical] theories)
6. Reasoning with part-whole relations
7. Using (including finding inconsistencies in) a hierarchy of relations
8. Reasoning across linked ontologies
9. Complex queries

Checking against instances

- Usual model checking
- Model checking against real instances in the ABox/Database
  - For each DL-concept in the OWL-formalised ontology (representing a universal), there has to be at least one ABox instance (as representation of the entity in reality)
  - To spot “redundant” DL-concepts w.r.t. the data-needs
- Model violation
  - Reducing the amount of instances to only those that do not violate the TBox (or: the more inconsistencies, the better)
  - For instance, to find a few candidate molecules that satisfy a given set of properties, out of a large pool of possibly suitable molecules; e.g., for drug discovery in pharmainformatics, tyre production

Discovering information

- The idea is that the combination of bio-ontologies, instances, and automated reasoning services somehow can find either the missing relations, or the types, or both
- How can one find what is, or may, not be in the ontology but ought to be there?
- At the TBox-level
  - computing derived relations (object properties)
  - find out where relations that are known by the developer have not yet been added to the ontology (finding ‘known gaps’)
  - add ‘ontological’ notions with top type ‘whole’ in a partonomy; e.g., 17 types of macrophage in the FMA each must be part of something
  - flag classes that have no relation (no or no is a) to anything else in the ontology