Semantic Web Technologies

Lecture 9: SWT for the Life Sciences 2: Successes and challenges for ontologies

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Outline

Introduction

Successes
- Exploiting the classification reasoning services
- Scalable querying of ontologies and data

Challenges
- Representation
- Reasoning issues

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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
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Examples in different application areas, using different features

- Data integration
- Instance classification (example today)
- Matchmaking and services
- Querying, information retrieval
  - Ontology-Based Data Access (example today)
  - Aid in browsing large ontologies
  - Ontologies to improve NLP (more tomorrow)

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- 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
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- The setting:
  - Lots of sequence data in data silos that needs to be enriched with biological knowledge
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- 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)

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- 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
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- Develop ontology for the subject domain, in OWL
  - Extract knowledge from peer-reviewed literature
  - Protein phosphatases; e.g.

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Class R5Phosphatase Complete
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  - Transform into OWL
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  - Identification of additional p-domains, refined the classification into further subtypes
- *A. fumigatus* phosphatates:
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- 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
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- Web-based, like most other bioinformatics resources
- Graphical querying to make it usable by the domain expert
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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/`), further manual improvements to create a proper conceptual data model
    - Simplify this conceptual data model into the appropriate OWL language (*DL-Lite*$_{\mathcal{A}}$, 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
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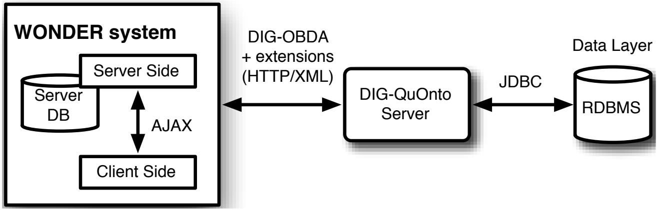
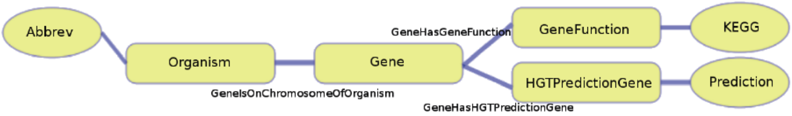
Architecture



Example: Diagram – *DL-lite*$_{\mathcal{A}}$ correspondence



$\delta(Abbrev) \sqsubseteq Organism$	An abbreviation is for an organism
$\rho(Abbrev) \sqsubseteq$ `xsd:string`	An abbreviation is of type string
$Organism \sqsubseteq \delta(Abbrev)$	Each organism has an abbreviation
(funct $Abbrev$)	Each individual has a single abbreviation
$\exists GeneIsOnChromosomeOfOrganism \sqsubseteq Gene$	Domain of object property
$\exists GeneIsOnChromosomeOfOrganism^{-} \sqsubseteq Organism$	Range of object property
$Gene \sqsubseteq \exists GeneIsOnChromosomeOfOrganism$	Each gene belongs to some organism
$Organism \sqsubseteq \exists GeneIsOnChromosomeOfOrganism^{-}$	Each organism has some gene
(funct $GeneIsOnChrOfOrganism$)	Each gene belongs to at most one organism
...	

Figure 2: Section of the HGT application ontology.

Formalisation of the graphical elements

Element name	Graphical Representation	Semantics of ontology elements	Semantics of building blocks of query graphs
Class node	C	C	$C(x)$
	C, D		$C(x), D(x)$
Is-a link	C D	$C \sqsubseteq D$	
Attribute node and link	C A	$\delta(A) \sqsubseteq C$ $\rho(A) \sqsubseteq \top_d$	$C(x),\ A(x,y)$
Role link	C P D	$\exists P \sqsubseteq C$ $\exists P^{-} \sqsubseteq D$	$C(x),\ R(x,y),\ D(y)$

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

`SELECT id, abbrev FROM organism JOIN genes ON abbrev = idorganism`	$\leadsto$	$\mathit{OrganismHasGene}(\mathbf{gene}(id), \mathbf{organism}(abbrev))$
`SELECT id, kegg FROM genes`	$\leadsto$	$\mathit{GeneHasGeneFunction}(\mathbf{gene}(id), \mathbf{function}(id))$ $\mathit{KEGG}(\mathbf{function}(id), kegg)$

Figure 3: Extract of the mapping from the HGT-DB database to the *DL-Lite*$_{\mathcal{A}}$ application ontology.

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-Lite*$_{\mathcal{A}}$ conjunctive query
  - Result obtained by:
    i. computing the certain answers for the CQ $q(\vec{y}) \leftarrow conj(\vec{z})$ (with $conj(\vec{z})$ the conjunction of atoms, and $\vec{y}$ a vector comprising the variables in $\vec{x}$ and in $\vec{w}$),
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- 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
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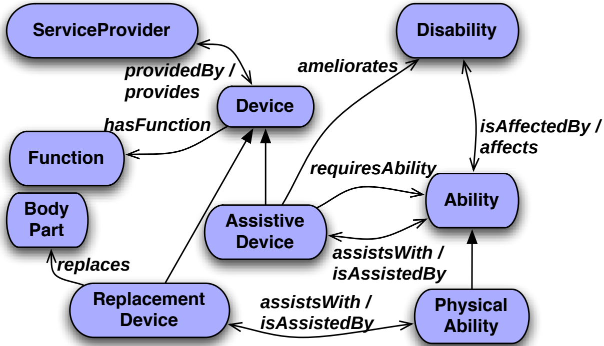
Additional features

- 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

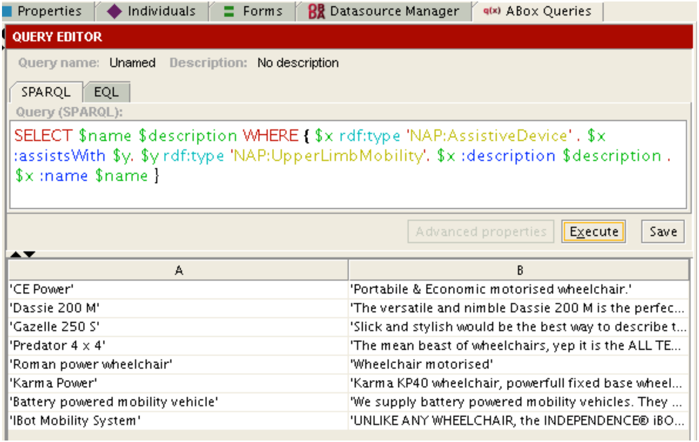
[1] A. Amoroso, G. Esposito, D. Lembo, P. Urbano, and R. Vertucci. Ontology-based data integration with MASTRO-I for configuration and data management at SELEX Sistemi Integrati. In Proc. of SEBD 2008, pages 8192, 2008.

[2] C. M. Keet, R. Alberts, A. Gerber, and G. Chimamiwa. Enhancing web portals with Ontology-Based Data Access: the case study of South Africa's Accessibility Portal for people with disabilities. In Proc. of OWLED 2008, 2008. CEUR-WS Vol 432

Informal overview of kind of knowledge in ADOLENA



Sample query in OBDA Plugin



A	B
'CE Power'	'Portabile & Economic motorised wheelchair.'
'Dassie 200 M'	'The versatile and nimble Dassie 200 M is the perfec...
'Gazelle 250 S'	'Slick and stylish would be the best way to describe t...
'Predator 4 x 4'	'The mean beast of wheelchairs, yep it is the ALL TE...
'Roman power wheelchair'	'Wheelchair motorised'
'Karma Power'	'Karma KP40 wheelchair, powerfull fixed base wheel...
'Battery powered mobility vehicle'	'We supply battery powered mobility vehicles. They ...
'IBot Mobility System'	'UNLIKE ANY WHEELCHAIR, the INDEPENDENCE® iBO...

```
q(x) :- Device(x), assistsWith(x, y), UpperLimbMobility(y)
```

Outline

Introduction

Successes

- Exploiting the classification reasoning services
- Scalable querying of ontologies and data

Challenges

- Representation
- Reasoning issues

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

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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 *challenge*s 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

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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)?
  - $(\psi \mid \phi)[l, u]$ as "generally, if an object belongs to ϕ, then it belongs to ψ with a probability in $[l, u]$"
  - e.g., (∃hasSymptom.Fever | Hepatitis)[illegible]
- "In 2000, worldwide prevalence of diabetes mellitus was 2.8%"
  - Probabilistic, or arithmetic, or what have we?
  - First, it assumes some class Human and a class HumanDiabetesMellitus, where some of the instances of the former have (are bearerOf) an instance of the latter
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  - It then intends to say that the human taking aspirin will not have a myocardial infarction *at all times in the future*, which can be represented in a suitable temporal logic with the $\Box^+$
  - e.g., $\texttt{AspirinIntake} \sqsubseteq \Box^+\texttt{prevents.MyocardialInfarction}$, or $\texttt{MyocardialInfarction} \sqsubseteq \Box^+\texttt{preventedBy.AspirinIntake}$, or $\texttt{AspirinIntake} \sqsubseteq \Box^+\texttt{hasPhysiologicalEffect.}\neg\texttt{MyocardialInfarction}$?

Outline

- The standard reasoning services (recollect lecture 5) are obviously sorted out
- Performance issues for the 'debugging' and explanation reasoning, and how to provide the 'best' explanation
- Querying OWL 2 DL, and any ABox data
- Additional reasoning scenarios

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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

explanation and examples in: Keet, C.M., Roos, M. and Marshall, M.S. A survey of requirements for automated reasoning services for bio-ontologies in OWL. Third international Workshop OWL: Experiences and Directions (OWLED 2007), 6-7 June 2007, Innsbruck, Austria. CEUR-WS Vol-258.

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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

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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

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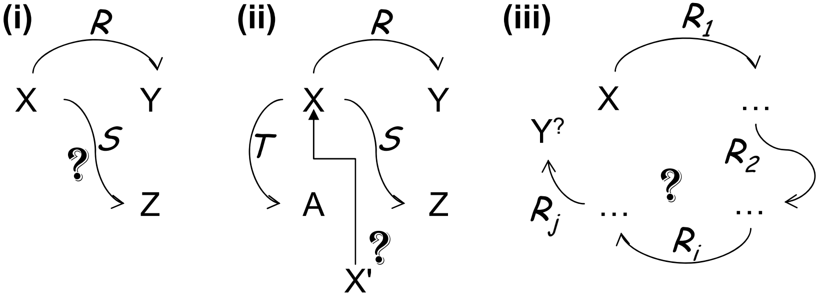
Discovering information

- For the TBox through querying the data (ABox, RDBMS)
  i. "for each x:X, y:Y, r:R, XRY, does there exist a z:Z, s:S, such that there exist ≥ 1 x and xsz?"
  ii. "for each x:X, y:Y, r:R, XRY, does there exist an xsz and an xta where z:Z, s:S, a:A, t:T hold?"
  iii. Find-me-anything-you-have: "for each x:X, return any $r_1, \ldots r_n$, their type of role and the concepts $Y_1, \ldots Y_n$ they are related to"



Summary

Introduction

Successes

- Exploiting the classification reasoning services
- Scalable querying of ontologies and data

Challenges

- Representation
- Reasoning issues