Aiding the Data Integration in Medicinal Settings by Means of Semantic Technologies

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Outline

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Introduction to the Problem

- context of our work – ontology evolution (Knowledge Web EU NoE)
- development of a simple methodology of ontology lifecycle scenario
- implementation of a respective framework, unifying all phases of the ontology lifecycle – DINO (Dynamics, INtegration or Data, INtensive; Ontologies)
- universal application, however, life-sciences and bio-medicine are our primary concern (due explicit needs for semantic solution and other domain specialties)
- critical and non-trivial task not covered by the state-of-the-art – ontology integration

Desired Features of the Integration

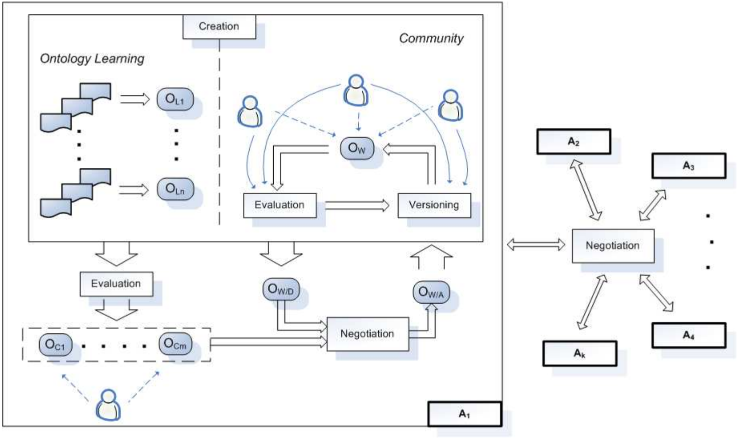
1. process new knowledge semi-automatically in dynamic domains
2. automatically compare the new and current knowledge
3. resolve and/or mark possible major inconsistencies between the new and current knowledge
4. automatically order the new knowledge according to user-defined preferences
5. transform the new knowledge into a form of sorted suggestions in simple natural language, alleviating human efforts in the task of the final incorporation of new knowledge

Why Healthcare?

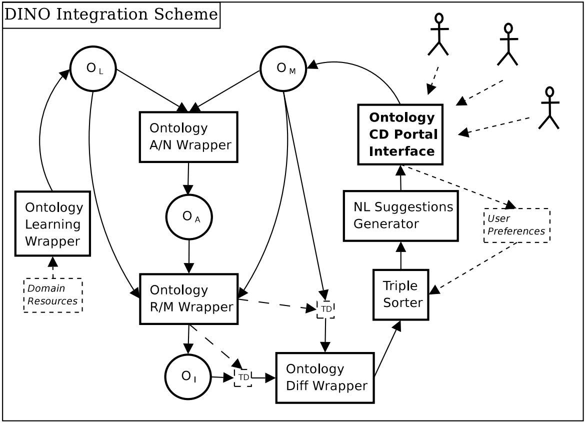
- explicit need for semantic solutions:
  - data stored in unstructured or disparate repositories, multiple formats hampering interoperability
  - cannot be queried to the full potential efficiently within traditional solutions (e.g. databases)
- dynamic nature of the domain
- new, sometimes even critical, knowledge continually appears and has to be efficiently processed and integrated
- emphasis on easy-to-use solutions, since medical experts are generally not experts in data or ontology engineering

Outline

Ontology Lifecycle Scenario



Ontology Integration



Outline

1. Introduction
   - Introduction to the Problem
   - Motivation
2. Architecture of the Framework
   - Broader Context
   - Ontology Integration
3. Using the Framework
   - Usage Example
   - Evaluation
4. Selected Life Science Use Cases
5. Conclusions and Future Work
   - Conclusions
   - Future Work

Text

...while cerebellar astrocytoma is usually discovered by means of CT... using a diagnostic procedure of scanning... GVHD, an immune dysfunction... GVHD, a disease being a type of dysfunction...

Learned Ontology

```
<owl:ObjectProperty rdf:ID="discovered-by"/>
<owl:Thing rdf:ID="CT"/>
<owl:Thing rdf:ID="cerebellar-astrocytoma">
  <discovered-by rdf:resource="#CT"/>
</owl:Thing>
<owl:Class rdf:ID="diagnostic-procedure"/>
<owl:Class rdf:ID="immune-dysfunction"/>
<owl:Class rdf:ID="dysfunction"/>
<owl:Class rdf:ID="scanning">
  <rdfs:subClassOf rdf:resource="#diagnostic-procedure"/>
</owl:Class>
<immune-dysfunction rdf:ID="GVHD"/>
<owl:Class rdf:ID="disease">
  <rdfs:subClassOf rdf:resource="#dysfunction"/>
</owl:Class>
```

Master Ontology

```
<owl:ObjectProperty rdf:ID="InstrumentalProperty"/>
<owl:ObjectProperty rdf:ID="DiscoveredUsing">
  <rdfs:subPropertyOf rdf:resource="#InstrumentalProperty"/>
  <rdfs:range rdf:resource="#Manifestation"/>
  <rdfs:domain rdf:resource="#DiagnosisProcedure"/>
</owl:ObjectProperty>
<owl:Class rdf:ID="Manifestation"/>
<owl:Class rdf:ID="Procedure"/>
<owl:Class rdf:ID="DiagnosisProcedure">
  <rdfs:subClassOf rdf:resource="#Procedure"/>
</owl:Class>
<owl:Class rdf:ID="SoftTissueCytoma"/>
<owl:Class rdf:ID="AstroCytoma">
  <rdfs:subClassOf rdf:resource="#SoftTissueCytoma"/>
</owl:Class>
<owl:Class rdf:ID="Disease"/>
<owl:Class rdf:ID="Dysfunction">
  <rdfs:subClassOf rdf:resource="#Disease"/>
</owl:Class>
```

Agreed Mapping

```
<owl:ObjectProperty rdf:ID="DiscoveredUsing">
  <owl:equivalentProperty rdf:resource="#discovered-by"/>
</owl:ObjectProperty>
<AstroCytoma rdf:ID="cerebellar-astrocytoma"/>
<owl:Class rdf:ID="DiagnosisProcedure">
  <owl:equivalentClass rdf:resource="#diagnostic-procedure"/>
</owl:Class>
<owl:Class rdf:ID="immune-dysfunction">
  <owl:subClassOf rdf:resource="#Dysfunction"/>
</owl:Class>
<owl:Class rdf:ID="Dysfunction">
  <owl:equivalentClass rdf:resource="#dysfunction"/>
</owl:Class>
```

Refining the Merge by Inference

- inconsistency resolution:
  - **disease** and **dysfunction** are said to be subclasses of each other
  - the learned inconsistent assertion (**disease** < **dysfunction**) is therefore removed by default
- learned knowledge augmentation:
  - using range and domain of the **DiscoveredUsing** property in the master ontology, we can infer that:
    - **cerebellar astrocytoma** is an instance of **Manifestation**
    - **CT** is an instance of **DiagnosisProcedure**

Resulting Suggestions

```
Config::        w_c = 1.0       w_r = 1.0       rho = 0.2       t = 5
Pos    ::       Scanning discover cytoma
Neg    ::       subclass disease dysfunction

--------

+0.667::        CEREBELLAR ASTROCYTOMA is a new istance of ASTROCYTOMA.
+0.667::        CEREBELLAR ASTROCYTOMA is a new istance of MANIFESTATION.
+0.389::        CT is a new istance of DIAGNOSIS PROCEDURE.
+0.333::        GVHD is a new istance of IMMUNE DYSFUNCTION.
-0.444::        A new class SCANNING is a sub-class of DIAGNOSIS PROCEDURE.
-0.667::        CEREBELLAR ASTROCYTOMA is DISCOVERED USING CT.
-0.833::        A new class IMMUNE DYSFUNCTION is a sub-class of DYSFUNCTION.
```

Preliminary Evaluation and Current State

- **preliminary evaluation:**
  - sorting places 80.7% of triples correctly compared to an order given by a human user (on small artificial sample)
  - negotiation component has been evaluated using the *Ontology Alignment Evaluation Initiative* test suite, preliminary results promising
- **current state:**
  - testing, tuning and debugging of the full implementation of the framework presented in the paper
  - combination with new concept in MarcOnt Portal – MarcOnt Portal services – and collaborative Protégé initiated within implementation of the whole lifecycle framework

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Longitudinal Electronic Health Record

- **needs:**
  - platforms supporting creation and management of long-term EHR
  - integration of different data sources
  - population of common conceptual structure (once it has been created)
  - efficient and expressive querying
- **solutions:**
  - ontologies bound to patient data
  - means for dynamic population of patient records from diverse resources (using learning, alignment and integration)
  - querying for free – using the state of the art OWL reasoning tools

Epidemiological Registries

- **needs:**
  - population-wise health records
  - extension of the needs in longitudinal EHR
  - integration and selection of the knowledge in patient records
- **solutions:**
  - merging of patient records, filtering the knowledge within the integration
  - population of an epidemiological ontology
  - again, querying using the state of the art OWL reasoning tools, adding symbolic dimension to the traditional statistic processing

Public Health Surveillance

- **needs:**
  - ongoing collection, analysis and dissemination of health-related data in order to facilitate a public health action
  - needs more or less the same as in the previous case
  - however, emphasis on efficient dynamic processing of new data
- **solutions:**
  - generic ontology integration and population services
  - explicit support for efficient dynamic integration of new knowledge from textual resources (by ontology learning)

Management of Clinical Trials

- **needs:**
  - electronic representation of clinical trials data
  - heterogeneity and integration problems (usually, several different institutions involved)
  - cost-effective querying demanded
- **solutions:**
  - ontologies developed and/or mediated using the DINO framework
  - querying of different clinical trial data straightforward

Genomics and Proteomics Research

- **needs:**
  - bridging the research and clinical practice
  - integrate specific knowledge e.g. in GO or UMLS – medical controlled dictionaries
  - efficient symbolic querying
- **solutions:**
  - aiding semi-automatic ontology development
  - data mediation using ontology integration
  - even for not very well specified data, the mechanism of sorted suggestions generation can reduce the efforts in merging the knowledge

Outline

Conclusions

- DINO – a mechanism for dynamic ontology integration – introduced
- based on ontology learning, meaning negotiation, merging, refinement and generation of suggestions in natural language, sorted according to user-defined relevance
- preliminary evaluation, current state of the implementation reported
- importance of the ontology lifecycle framework for integration of more general healthcare data described using realistic use cases

Future Work

- improve the natural language generation mechanism
- finish the basic testing and tuning of the platform
- include the DINO integration into the broader context of the lifecycle platform implementation
- employ and test the whole framework in realistic settings in a healthcare industry, possibly in line with the presented use cases
- incorporate the feedback and challenges identified in the realistic evaluation within further improvement of the framework