Formal ontologies and multilingual terminologies as tools for knowledge level interoperability in the biomedical domain

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Problem

- Babylonian language confusion in biomedical semantics & knowledge representation

Knowledge

- What kinds of knowledge need to be represented?
- Is a more principled framework possible?
- How do biomedical formal ontologies and multilingual terminologies fit in this picture?

Universals

Symbols

Knowledge map

Individuals

"perro", "dog", "canino", “Hund” "canis", "dog" NCBI:txid9615 "Marley"

C. K. Ogden and I. A. Richards (1923) The Meaning of Meaning
Marley lives in Florida
Marley is a dog
"dog" is a noun
"canis familiaris" and "dog" are synonyms
dogs are vertebrates
dogs are possible vectors of rabies
Ontological “knowledge”:
Axioms that are universally true

Contingent knowledge:
typical, likely, possible

Symbolic knowledge:
Statements about properties and meaning of signs of language

Factual knowledge:
Statements about concrete entities and their relationships
Factual knowledge:
Statements about concrete entities and their relationships

- Marley is a dog
- Marley lives in Florida

Individuals
Statements about individuals

<Subject> <Predicate> <Object>
:Florida rdf:type :state
:Marley rdf:type :dog
:Marley :lives :Florida

Syntax TURTLE : https://www.w3.org/TR/turtle/
Ontological knowledge:
Axioms that are universally true

Contingent knowledge:
Typical, likely, possible

Symbolic knowledge:
Statements about properties and meaning of signs of language

Factual knowledge:
Statements about concrete entities and their relationships
Symbolic knowledge

Statements about properties and meaning of signs of language
Representations SKOS / Linked Data

:ex:Dog rdf:type skos:Concept
:ex:Dog skos:prefLabel "dog"@en;
:ex:Dog skos:prefLabel "perro"@es;

:ex:Animal rdf:type skos:Concept
:ex:Animal skos:broader ex:Dog

wr:dog lemon:sense wr:dog-English-Noun-1
wr:dog lemon:sense wr:dog-English-Verb-1
wr:dog-English-Noun-1 wt:hasPoS wt:Noun

Syntax TURTLE : https://www.w3.org/TR/turtle/
Wiktionary: http://wiki.dbpedia.org/wiktionary-rdf-extraction
Ontological knowledge: Axioms that are universally true

Contingent knowledge: typical, likely, possible

Symbolic knowledge: Statements about properties and meaning of signs of language

Factual knowledge: Statements about concrete entities and their relationships
Ontological knowledge:
Axioms that are universally true

dogs are vertebrates
Representation OWL

Dog subclassOf Vertebrate
Vertebrate subclassOf Animal
Vertebra subclassOf Bone
Vertebrate equivalentTo Animal and has-part some Vertebra

There is no dog that has no bones

OWL Manchester Syntax: https://www.w3.org/TR/owl2-manchester-syntax/
HermiT reasoner: http://www.hermit-reasoner.com/
Fact++ reasoner: http://owl.man.ac.uk/factplusplus/
Ontological knowledge:
Axioms that are universally true

Contingent knowledge:
Typical, likely, possible

Symbolic knowledge:
Statements about properties and meaning of signs of language

Factual knowledge:
Statements about concrete entities and their relationships
Contingent knowledge: typical, likely, possible

6 dogs are possible vectors of rabies

Universals
Triple representation

- No formal semantics!
- Different, mostly complex interpretations
- Don’t use formal languages for this

<table>
<thead>
<tr>
<th>Subject</th>
<th>Predicate</th>
<th>Object</th>
</tr>
</thead>
<tbody>
<tr>
<td>:Dog</td>
<td>:vector-of</td>
<td>:Rabies</td>
</tr>
<tr>
<td>:Tobacco</td>
<td>:causes</td>
<td>:Cancer</td>
</tr>
<tr>
<td>:Aspirin</td>
<td>:treats</td>
<td>:Pain</td>
</tr>
<tr>
<td>:Fever</td>
<td>:suggests</td>
<td>:Malaria</td>
</tr>
<tr>
<td>:Bird</td>
<td>:capable-of</td>
<td>:Flying</td>
</tr>
</tbody>
</table>

Dog subclassOf vector-of some Rabies

Tobacco subclassOf causes some Cancer

Aspirin subclassOf treats some Pain
Ontological knowledge:
Axioms that are universally true

Contingent knowledge:
typical, likely, possible

Symbolic knowledge:
Statements about properties and meaning of signs of language

Factual knowledge:
Statements about concrete entities and their relationships
Ontological knowledge
Axiomatic layer of clinical terminology systems

Ontological knowledge examples:
- SNOMED CT
- HL7 FHIR

Lexical layer of terminology systems
Symbolic knowledge

Contingent knowledge
Clinical guidelines

Contingent knowledge examples:
- Research databases
- Clinical guidelines

Symbolic knowledge examples:
- UniProtKB - Q71M42 (PC11X_PANTR)
- Condition (DomainResource)