Semantic similarity and machine learning with ontologies

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Preliminaries: ontologies

- Specific artifacts expressing the intended meaning of a vocabulary in terms of primitive categories and relations describing the nature and structure of a domain of discourse
  - in order to account for the competent use of vocabulary in real situations (such as annotations in databases, etc.)
- the intended meaning of *primitive* categories and relations is expressed through axioms (axiomatic method, Tarski)

Preliminaries: axioms

- *classes* represent kinds of things in the world
  - *Arm*, *Apoptosis*, *Influenza*, *Homo sapiens*, *Drinking behavior*, *Membrane*
- *instances* of classes are individuals satisfying the classes' intension
  - my arm, the influenza I had last year, one ethanol molecule, etc.
- *relations* between instances arise from interactions, configurations, etc., of individuals
  - my arm is **part of** me, the **duration of** my influenza was 10 days
- *axioms* specify the conditions that instances of a class must satisfy
  - every instance of *Hand* is a **part of** an instance of *Arm*

Description Logics: overview

- TBox: axioms pertaining to the terminology of the domain (classes)
- ABox: axioms stating facts (assertions) about the world
- RBox: axioms holding for relations
- Reasoning: derive implicitly represented knowledge (e.g., subsumption)
- NB: a “knowledge graph” is an ABox + RBox

Manchester OWL Syntax

DL Syntax	Manchester Syntax	Example
$C \sqcap D$	C and D	Human and Male
$C \sqcup D$	C or D	Male or Female
$\neg C$	not C	not Male
$\exists R.C$	R some C	hasChild some Human
$\forall R.C$	R only C	hasChild only Human
$(\geq nR.C)$	R min n C	hasChild min 1 Human
$(\leq nR.C)$	R max n C	hasChild max 1 Human
$(= nR.C)$	R exactly n C	hasChild exactly 1 Human
$\{a\} \sqcup \{b\} \sqcup \ldots$	{a b ...}	{John Robert Mary}

Description Logic ALC: syntax

Definition

Let N_C be a set of concept names and N_R be a set of relation names, $N_C \cap N_R = \emptyset$. $\mathcal{ALC}$ concept descriptions are inductively defined as:

- If $A \in N_C$, then A is an $\mathcal{ALC}$ concept description
- If C, D are $\mathcal{ALC}$ concept description, and $r \in N_R$, then the following are $\mathcal{ALC}$ concept descriptions:
  - $C \sqcap D$
  - $C \sqcup D$
  - $\neg C$
  - $\forall r.C$
  - $\exists r.C$

- Use $\bot$ as abbreviation of $A \sqcap \neg A$, $\top$ as abbreviation of $A \sqcup \neg A$

Examples of concept descriptions, dl1.pdf, p8

Description Logic ALC: semantics

Definition

An interpretation $\mathcal{I} = (\Delta^{\mathcal{I}}, \cdot^{\mathcal{I}})$ consists of a non-empty domain $\Delta^{\mathcal{I}}$ and an interpretation function $\cdot^{\mathcal{I}}$:

- $A^{\mathcal{I}} \subseteq \Delta^{\mathcal{I}}$ for all $A \in N_C$,
- $r^{\mathcal{I}} \subseteq \Delta^{\mathcal{I}} \times \Delta^{\mathcal{I}}$ for all $r \in N_R$

The interpretation function is extended to $\mathcal{ALC}$ concept descriptions as follows:

- $(C \sqcap D)^{\mathcal{I}} := C^{\mathcal{I}} \cap D^{\mathcal{I}}$
- $(C \sqcup D)^{\mathcal{I}} := C^{\mathcal{I}} \cup D^{\mathcal{I}}$
- $(\neg C)^{\mathcal{I}} := \Delta^{\mathcal{I}} - C^{\mathcal{I}}$
- $(\forall r.C)^{\mathcal{I}} := \{d \in \Delta^{\mathcal{I}} | \text{for all } e \in \Delta^{\mathcal{I}} : (d, e) \in r^{\mathcal{I}} \text{ implies } e \in C^{\mathcal{I}}\}$
- $(\exists r.C)^{\mathcal{I}} := \{d \in \Delta^{\mathcal{I}} | \text{there is } e \in \Delta^{\mathcal{I}} : (d, e) \in r^{\mathcal{I}} \text{ and } e \in C^{\mathcal{I}}\}$

Description Logic: terminologies

- A concept definition is of the form $A \equiv C$ where
  - A is a concept name
  - C is a concept description
- A TBox is a finite set of concept definitions such that it
  - does not contain multiple definitions,
  - does not contain cyclic definitions
- A *defined concept* occurs on the left-hand side of a definition
- A *primitive concept* does not occur on the left-hand side of a definition
- An interpretation $\mathcal{I}$ is a model of a TBox $\mathcal{T}$ if it satisfies all its concept definitions: $A^{\mathcal{I}} = C^{\mathcal{I}}$ for all $A \equiv C \in \mathcal{T}$

Description Logic: assertions

- An assertion is of the form $C(a)$ (concept assertion) or $r(a, b)$ (role assertion), where C is a concept description, r is a role, a, b are individual names from a set N_I of such names
- An ABox is a finite set of assertions
- An interpretation $\mathcal{I}$ is a model of an ABox $\mathcal{A}$ if it satisfies all its assertions:
  - $a^{\mathcal{I}} \in C^{\mathcal{I}}$ for all $C(a) \in \mathcal{A}$
  - $(a^{\mathcal{I}}, b^{\mathcal{I}}) \in r^{\mathcal{I}}$ for all $r(a, b) \in \mathcal{A}$

Description Logic: Reasoning

- Subsumption: Is C a subconcept of D?
  - $C \sqsubseteq_{\mathcal{T}} D$ iff $C^{\mathcal{I}} \subseteq D^{\mathcal{I}}$ for all models $\mathcal{I}$ of $\mathcal{T}$
- Satisfiability: Is the concept C non-contradictory?
  - C is satisfiable w.r.t. $\mathcal{T}$ iff $C^{\mathcal{I}} \neq \emptyset$ for some model $\mathcal{I}$ of $\mathcal{T}$
- Consistency: Is the ABox $\mathcal{A}$ non-contradictory?
  - $\mathcal{A}$ is consistent w.r.t. $\mathcal{T}$ iff it has a model that is also a model of $\mathcal{T}$
- Instantiation: Is e an instance of C?
  - $\mathcal{A} \models_{\mathcal{T}} C(e)$ iff $e^{\mathcal{I}} \in C^{\mathcal{I}}$ for all models $\mathcal{I}$ of $\mathcal{T}$ and $\mathcal{A}$.

Offtopic: knowledge graphs

My favorite definition of "knowledge graph":
A knowledge graph is an ABox + RBox.

- ontologies are (mostly) the TBox!

Ontologies provide background knowledge

Annotation	Value
label	T cell aggregation
definition	The adhesion of one T cell to one or more other T cells via adhesion molecules.
class	http://purl.obolibrary.org/obo/GO_0070489
ontology	GO-PLUS
Equivalent	leukocyte aggregation and (has participant some T cell)
SubClassOf	lymphocyte aggregation, has participant some T cell
has_obo_namespace	biological_process
id	GO:0070489
synonyms	T-cell aggregation, T lymphocyte aggregation, T-lymphocyte aggregation

Ontologies provide background knowledge

Annotation	Value
label	T cell activation
definition	The change in morphology and behavior of a mature or immature T cell resulting from exposure to a mitogen, cytokine, chemokine, cellular ligand, or an antigen for which it is specific.
class	http://purl.obolibrary.org/obo/GO_0042110
ontology	GO-PLUS
Equivalent	cell activation and (has input some T cell)
SubClassOf	has input some T cell, lymphocyte activation
has_obo_namespace	biological_process
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Using background knowledge

Problem statement (first attempt):

Given a set of entities (instances) within an ontology (DL theory). Can we discover/predict *new* relations between the entities, or between entities and classes in the ontology?

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- what features are relevant?
  - depends on the relation!

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Given a set of entities (instances) within an ontology (DL theory). Can we discover/predict *new* relations between the entities, or between entities and classes in the ontology?

- what relations, and when is a fact "new"?
- what features are relevant?
  - depends on the relation!
- finding new facts is only one (minor?) use case
  - other uses: encode background knowledge for machine learning models; add new classes; expand definition; constrained learning; etc.
  - computing "similarity"

Semantic similarity: some examples

- Are cyclin dependent kinases *functionally* more similar to lipid kinases or to riboflavin kinases? How about *phenotypically*?
- Which protein in the *mouse* is functionally most similar to the zebrafish *gustducin* protein?
- Which mouse knockout resembles *Bardet-Biedl Syndrome 8*?
- Are there mouse knockouts that resemble the side effects of diclofenac?
- Which genetic disease produces similar symptoms to ebola?
- Does functional similarity correlate with phenotypic similarity?

Semantic similarity

semantic similarity measures:

- for words, terms, classes
- role of background knowledge:
  - statistical/distributional semantics, large corpora
  - ontologies: (graph) topology
- similarity measures: hand-crafted or data-driven?

Semantic similarity or machine learning

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  - capture certain intuition about what constitutes "similarity"
  - different measures for different kinds of similarity
  - usually interpretable (and explainable)
- machine learning methods are mostly data-driven
  - the architecture of the model is still hand-crafted
  - usually hard to interpret

Ontologies and graphs

- semantic similarity measures *and machine learning models* on ontologies can be graph-based, feature-based, or model-based
  - graph-based: ontology as a graph
  - feature-based: extract (or obtain) features for classes/relations
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- we may need to generate graphs from ontologies
  - *is-a* relations are easy (this is just `owl:subClassOf`)
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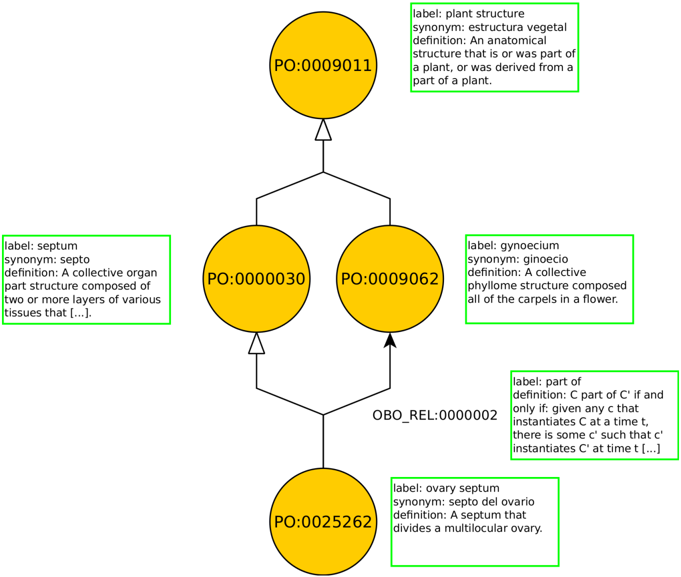
  - disjointness, universal vs. existential quantification, cardinality restrictions, intersection, union, negation?
- relational patterns are implicit in OWL axioms
  - design patterns as "relations" between classes

Relations as patterns



Relations as patterns

- `X SubClassOf: Y`: $X \xrightarrow{\text{is-a}} Y$
- `X SubClassOf: part-of some Y`: $X \xrightarrow{\text{part-of}} Y$
- `X SubClassOf: regulates some Y`: $X \xrightarrow{\text{regulates}} Y$
- `X DisjointWith: Y`: $X \xleftrightarrow{\text{disjoint}} Y$
- `X EquivalentTo: Y`: $X \overset{\equiv}{\leftrightarrow} Y$, $\{X, Y\}$
- ...

NB: in bio-ontologies, the OBO Relation Ontology defines these patterns

Asserted and inferred

- relation patterns can be asserted or inferred
- `X SubClassOf: part-of some Y`
- `Y SubClassOf: part-of some Z`
- `part-of o part-of SubPropertyOf: part-of`
- $\vdash$ `X SubClassOf: part-of some Z`
- Therefore: $X \xrightarrow{\text{part-of}} Z$
- $\Rightarrow$ we should use deductive inference to generate these patterns

Tree models

- some languages have the *finite model* and *tree model* properties
  - such as the Description Logic $\mathcal{ALC}$
  - generated through a tableaux algorithm
- nodes: individuals
  - node labels: concept names, concept descriptions
- edges: relations between individuals
- can be extended to more expressive languages (with blocking, cycles, etc.)

Methods and tools

- edges should be "meaningful": not merely syntax (why?)
  - the RDF serialization of OWL is a graph and contains all information but is a bad idea for semantic similarity or machine learning (more later)
  - conceptual graphs?
- OBO Format represents ontologies as graphs:
  - Protege/OWLAPI: OBO export
  - OBO toolsets (e.g., ROBOT)
  - `https://github.com/bio-ontology-research-group/Onto2Graph`

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  - `https://github.com/bio-ontology-research-group/Onto2Graph`
- but: a conversion of an ontologies into a graph will almost always lead to a loss of information