

Ontologies in computational biology

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Overview

Ontologies and the Semantic Web

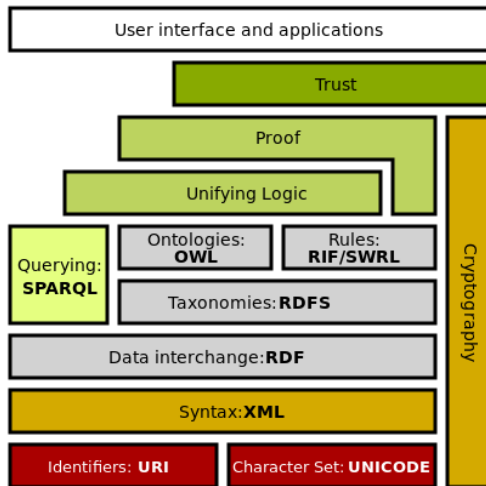
Ontologies and graphs

Semantic Similarity

Machine learning and ontologies

Applications

The Semantic Web



Description Logics: overview

- ▶ TBox: defines the terminology of the domain
- ▶ ABox: states facts (assertions) about the world
- ▶ Reasoning: derive implicitly represented knowledge (e.g., subsumption)

Description Logic: ALC

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:
 - ▶ $C \sqcap D$
 - ▶ $C \sqcup D$
 - ▶ $\neg C$
 - ▶ $\forall r.C$
 - ▶ $\exists r.C$
- ▶ We use \perp as abbreviation of $A \sqcap \neg A$, \top as abbreviation of $A \sqcup \neg A$

Description Logic: ALC

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:

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

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: terminologies

- ▶ Also possible to use more general constraints for the definition of concepts
- ▶ A *generalized concept inclusion* (GCI) is of the form $C \sqsubseteq D$ where C, D may be concept descriptions
- ▶ An interpretation \mathcal{I} is a model of a set of GCIs \mathcal{T} (a general TBox) if it satisfies all its concept inclusion axioms: $C^{\mathcal{I}} \subseteq D^{\mathcal{I}}$ for all $C \sqsubseteq D \in \mathcal{T}$

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: ALC

- ▶ \mathcal{ALC} can be seen as a fragment of FOL:
 - ▶ concept names are unary predicates, role names binary predicates
 - ▶ concept descriptions are formulas with one free variable
- ▶ the formulas resulting from transformation to FOL are known to be decidable (two-variable fragment)

Web Ontology Language (OWL)

- ▶ OWL 2 is based on the Description Logic $\mathcal{SROIQ}(\mathcal{D})$
- ▶ \mathcal{ALC} with
 - ▶ complex role inclusions: $r \circ s \subseteq r$
 - ▶ role hierarchy: $r \subseteq s$
 - ▶ role transitivity $r \circ r \subseteq r$
 - ▶ nominals: $\{a_1, \dots, a_n\}$ as concept constructor
 - ▶ qualified number restrictions: $(\leq nr.Q)$
 - ▶ datatype properties: $\exists r.[\geq n(Integer)]$

Terminology

- ▶ Instances
- ▶ Properties
 - ▶ Object properties
 - ▶ Datatype properties
- ▶ Classes
- ▶ Meta-classes
 - ▶ OWL Full
 - ▶ Punning
- ▶ Axiom
 - ▶ Class axioms: Subclass, Equivalent class, Disjoint class
 - ▶ Property axioms
- ▶ Ontology
- ▶ OWL: Web Ontology Language

Syntax

- ▶ originally an extension of RDF and RDF Schema
- ▶ several different syntaxes

Consider the axiom $Parent \equiv Human \sqcap \exists hasChild.\top$

Functional Syntax

```
EquivalentClasses(:Parent  
  ObjectSomeValuesFrom(:hasChild owl:Thing))
```

RDF/XML Syntax

```
<owl:Class rdf:about="http://example.com/demo-ontology.owl#Parent">
  <owl:equivalentClass>
    <owl:Restriction>
      <owl:onProperty rdf:resource="http://example.com/demo-ontology.owl#hasChild"/>
      <owl:someValuesFrom rdf:resource="&owl;Thing"/>
    </owl:Restriction>
  </owl:equivalentClass>
</owl:Class>
```

RDF Turtle Syntax

```
:Parent rdf:type owl:Class ;  
    owl:equivalentClass [ rdf:type owl:Restriction ;  
                            owl:onProperty :hasChild ;  
                            owl:someValuesFrom owl:Thing  
                        ] .
```


OWL/XML Syntax

```
<EquivalentClasses>
  <Class IRI="#Parent"/>
  <ObjectSomeValuesFrom>
    <ObjectProperty IRI="#hasChild"/>
    <Class abbreviatedIRI="owl:Thing"/>
  </ObjectSomeValuesFrom>
</EquivalentClasses>
```

Manchester OWL Syntax

```
Class: Parent
  EquivalentTo:
    hasChild some owl:Thing
```

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
$\{a\} \sqcup \{b\} \sqcup \dots$	{a b ...}	{John Robert Mary}

OWL classes and namespaces

- ▶ \perp is owl:Nothing
- ▶ \top is owl:Thing
- ▶ owl: is a *namespace* (<http://www.w3.org/2002/07/owl#>)
- ▶ owl:Thing expands to <http://www.w3.org/2002/07/owl#Thing> (a class IRI)
- ▶ all OWL entities (ontologies, classes, properties, instances) are referred to by an IRI
- ▶ namespaces define a common (IRI-)prefix, e.g.,
 - ▶ rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
 - ▶ rdfs: <http://www.w3.org/2000/01/rdf-schema#>
- ▶ can define own namespaces:

Namespace: mynamespace <<http://www.kaust.edu.sa#>>

Class: mynamespace:Student # <http://www.kaust.edu.sa#Student>

Object properties

- ▶ Object property characteristics:
 - ▶ transitive
 - ▶ symmetric, asymmetric
 - ▶ reflexive, irreflexive
 - ▶ functional, inverse functional
 - ▶ inverse of
- ▶ Domain and range

Annotation properties

- ▶ OWL entities (classes, properties, axioms, ontologies, etc.) can have *annotations*
- ▶ outside of OWL semantics (unless for OWL Full)
- ▶ useful to add labels, synonyms, explanation, (textual) definitions, authoring information, versions, etc.
- ▶ predefined: `rdfs:label`, `owl:versionInfo`, `rdfs:comment`, `rdfs:seeAlso`, `rdfs:isDefinedBy`
- ▶ Dublin Core

OWL Reasoning

- ▶ Classification: compute the most specific sub- and super-classes for each named class in an OWL ontology
- ▶ Subsumption: find all sub-, super- or equivalent classes of an OWL class description
- ▶ Consistency: find contradictions in OWL knowledge base
- ▶ Instantiation: is a an instance of C ?

Complexity of reasoning in OWL

- ▶ OWL 2 (*SROIQ*) is 2NEXPTIME-complete
- ▶ OWL (1) (*SHOIN*) is NEXPTIME-complete
- ▶ OWL Lite (*SHIF*) is EXPTIME-complete

OWL profiles

- ▶ OWL 2 EL: PTIME-complete
- ▶ OWL 2 RL: PTIME-complete
- ▶ OWL 2 QL: AC^0 w.r.t. data size

OWL 2 EL

- ▶ Class axioms:
 - ▶ subclass, equivalent class, disjoint class
- ▶ Object property axioms:
 - ▶ domain and range restrictions, property inclusion, property chains, property equivalence, transitive and reflexive properties
- ▶ Class descriptions:
 - ▶ intersection, existential quantification, enumerations to a single individual
- ▶ Assertions: all

Why OWL?

- ▶ OWL exploits 20+ years of research on Description Logic
- ▶ well-defined semantics
- ▶ complexity and decidability well understood
- ▶ known algorithms
- ▶ scalability demonstrated in practise

Why OWL?

Major benefit is the large number of tools and infrastructure:

- ▶ Editors: Protege, WebProtege
- ▶ Reasoners: HermiT, Pellet, FaCT++, **ELK**, KAON2, RACER,...
- ▶ Explanation, justification
- ▶ Modularization
- ▶ APIs (esp. the OWL API)

OWL vs Databases

Database	OWL Ontology
Closed World Assumption	Open World Assumption
Unique Name Assumption	No UNA
Schema constraints data structure	Axioms behave like inference rules

Examples: OWL vs Databases

Based on slides by Ian Horrocks

- ▶ hasPet some owl:Thing SubclassOf: Human
- ▶ Phoenix SubclassOf: petOf only Wizard
- ▶ HarryPotter: Wizard
- ▶ DracoMalfoy: Wizard
- ▶ HarryPotter hasFriend RonWeasley
- ▶ HarryPotter hasFriend HermioneGranger
- ▶ HarryPotter hasPet Hedwig

Query: Is Draco a friend of Harry Potter?

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Query: Is Draco a friend of Harry Potter?

- ▶ DB: No
- ▶ OWL: Don't know

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Query: How many friends has Harry Potter?

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Query: How many friends has Harry Potter?

- ▶ DB: 2
- ▶ OWL: At least 1

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Query: How many friends has Harry Potter?

- ▶ DB: 2
- ▶ OWL: 2

Examples: OWL vs Databases

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Adding new facts:

- ▶ Dumbledore: Wizard
- ▶ Fawkes: Phoenix
- ▶ Fawkes isPetOf DumbleDore
- ▶ DB: Update rejects, constrain violation
- ▶ OWL: infer that Dumbledore is Human; infer that Dumbledore is a Wizard

Ontology-based information systems

Ontology like DB schema, instances like data

Advantages:

- ▶ Relatively easy to maintain and update schema
- ▶ Query answers reflect both schema and data
- ▶ Can deal with incomplete information
- ▶ Answer intensional and extensional queries

Disadvantages:

- ▶ Semantic can seem counter-intuitive (OWA, UNA)
- ▶ Query answering (logical entailment) much more difficult

Hands-on part 1

- ▶ start Jupyter and open the Notebook
 - ▶ <https://github.com/bio-ontology-research-group/ontology-tutorial/blob/master/ontology-analysis.ipynb>
- ▶ run the “Reasoning with ontologies” part
 - ▶ then modify the code and retrieve all superclasses of “increased B cell apoptosis”
 - ▶ and then the superclasses of “B cell apoptotic process”

Ontologies, axioms, and bioinformatics

- ▶ ontologies are ubiquitous
- ▶ rich formal characterization (axioms)
- ▶ can they also be used for data analysis?

Some examples

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- ▶ 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?

Ontologies and graphs

- ▶ semantic similarity measures can be graph-based, feature-based, or model-based
- ▶ we may need to generate graphs from ontologies
 - ▶ *is-a* relations are easy
 - ▶ how about *part-of*, *regulates*, *precedes*, etc.?
- ▶ relational patterns are implicit in OWL axioms
 - ▶ in first order logic
 - ▶ needs to translate them into OWL
 - ▶ defined in OBO Relation Ontology

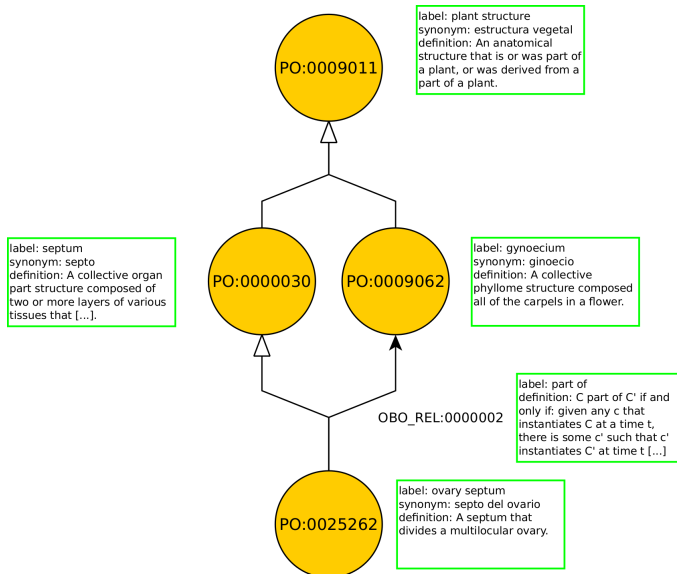
Relations as patterns

- ▶ OBO Relation Ontology (RO):
 - ▶ <https://github.com/oborel/obo-relations>
- ▶ Basic Formal Ontology (BFO):
 - ▶ provides top-level classes
 - ▶ Continuant, Process, Function, Material object, etc.
 - ▶ used for some OBO Foundry ontologies
- ▶ RO and BFO provide a top-level system of classes and relations shared across many biomedical ontologies
 - ▶ even GO, although somewhat hidden!
- ▶ this system defines patterns used to generate graphs

Relations as patterns

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

Relations as patterns



Semantic similarity

- ▶ We want to use *background knowledge* in ontologies to
 - ▶ determine similarity between classes,
 - ▶ instances,
 - ▶ and entities with ontology annotations

How to measure similarity?

- ▶ semantic similarity measures similarity between classes
- ▶ semantic similarity measures similarity between instances of classes
- ▶ semantic similarity measures similarity between entities *annotated* with classes
- ▶ \Rightarrow reduce all of this to similarity between classes

How to measure similarity?

What properties do we want in a similarity measure?

A function $sim : D \times D$ is a similarity on D if, for all $x, y \in D$, the function sim is:

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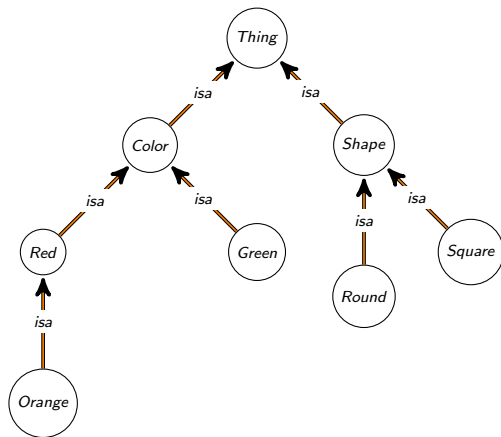
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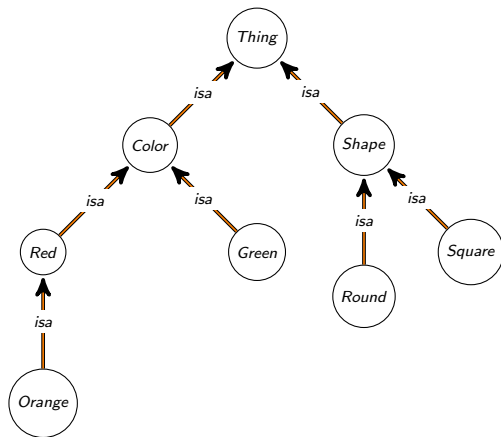
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- ▶ $sim(x, x) > sim(x, y)$ for $x \neq y$
- ▶ sim is a *normalized* similarity measure if it has values in $[0, 1]$

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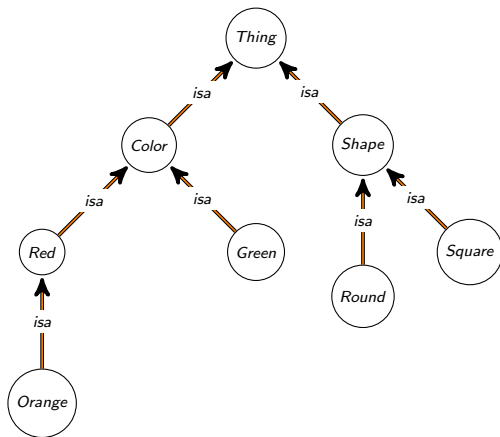


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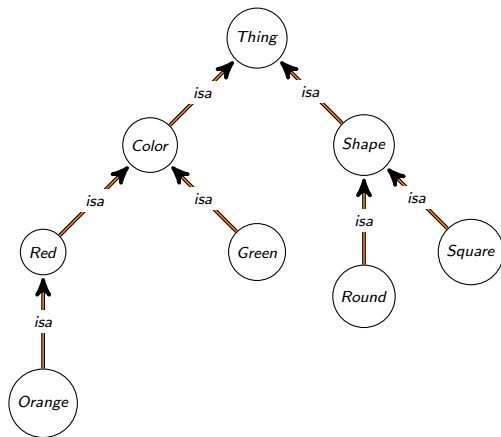
- distance on shortest path (Rada *et al.*, 1989)

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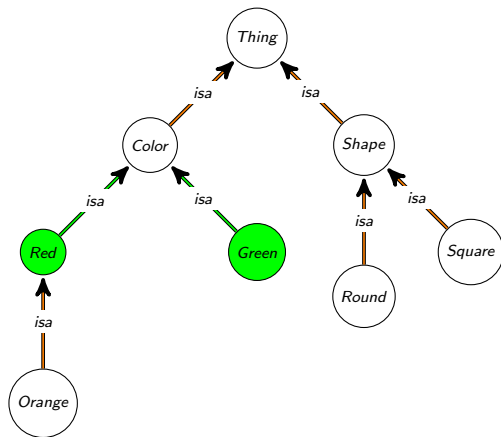
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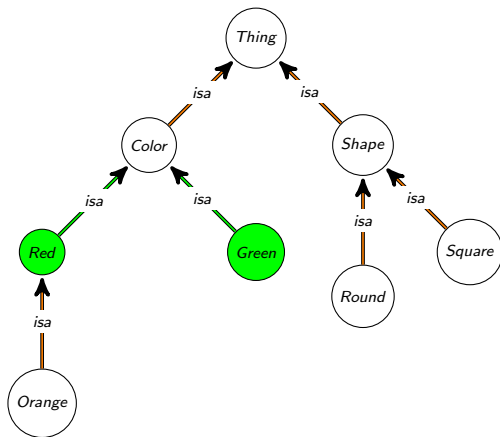
- ▶ distance on shortest path (Rada *et al.*, 1989)
- ▶ $dist_{Rada}(u, v) = sp(u, isa, v)$
- ▶ $sim_{Rada}(u, v) = \frac{1}{dist_{Rada}(u, v) + 1}$

How to measure similarity?



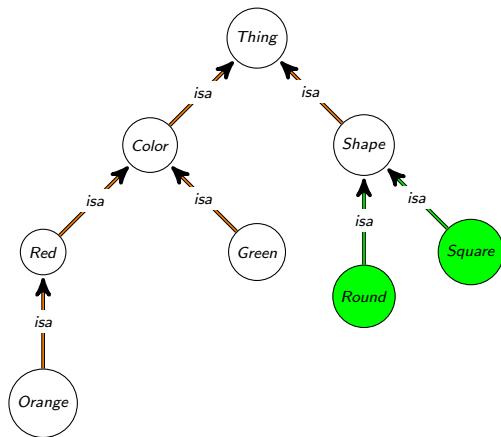
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How to measure similarity?



- ▶ distance on shortest path
- ▶ $\text{distance}(\text{green}, \text{red}) = 2$
- ▶ $\text{sim}_{\text{Rada}}(\text{green}, \text{red}) = \frac{1}{3}$

How to measure similarity?



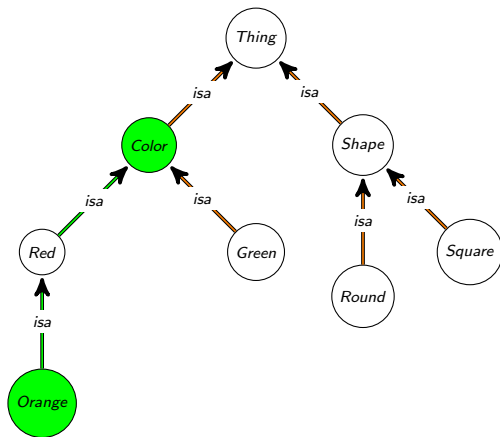
► distance on shortest path

► $\text{distance}(\text{square}, \text{round}) = 2$

►

$$\text{sim}_{\text{Rada}}(\text{square}, \text{round}) = \frac{1}{3}$$

How to measure similarity?



► distance on shortest path

► $\text{distance}(\text{orange}, \text{color}) = 2$

►

$$\text{sim}_{\text{Rada}}(\text{orange}, \text{color}) = \frac{1}{3}$$

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 - ▶ number of children
 - ▶ information content
- ▶ *density* of a branch in the ontology
 - ▶ number of siblings
 - ▶ information content
- ▶ account for different edge types
 - ▶ non-uniform edge weighting

How to measure similarity

- ▶ term specificity measure $\sigma : \mathcal{C} \mapsto \mathbb{R}$:
 - ▶ $x \sqsubseteq y \rightarrow \sigma(x) \geq \sigma(y)$

How to measure similarity

- ▶ term specificity measure $\sigma : C \mapsto \mathbb{R}$:

- ▶ $x \sqsubseteq y \rightarrow \sigma(x) \geq \sigma(y)$

- ▶ intrinsic:

- ▶ $\sigma(x) = f(\text{depth}(x))$

- ▶ $\sigma(x) = f(A(x))$ (for ancestors $A(x)$)

- ▶ $\sigma(x) = f(D(x))$ (for descendants $D(x)$)

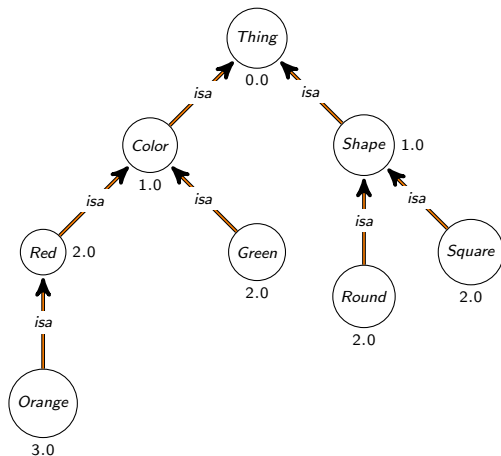
- ▶ many more, e.g., Zhou et al.:

$$\sigma(x) = k \cdot \left(1 - \frac{\log |D(x)|}{\log |C|}\right) + (1 - k) \frac{\log \text{depth}(x)}{\log \text{depth}(G_T)}$$

How to measure similarity

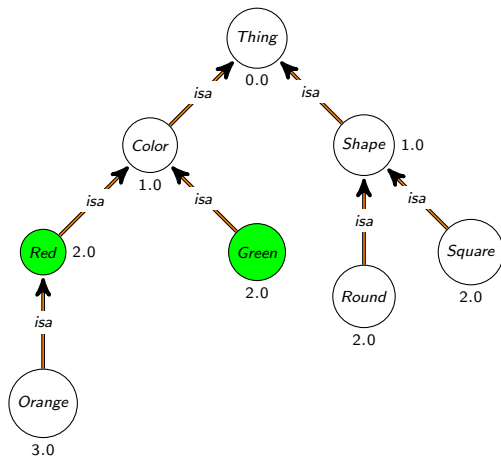
- ▶ term specificity measure $\sigma : C \mapsto \mathbb{R}$:
 - ▶ $x \sqsubseteq y \rightarrow \sigma(x) \geq \sigma(y)$
- ▶ intrinsic:
 - ▶ $\sigma(x) = f(\text{depth}(x))$
 - ▶ $\sigma(x) = f(A(x))$ (for ancestors $A(x)$)
 - ▶ $\sigma(x) = f(D(x))$ (for descendants $D(x)$)
 - ▶ many more, e.g., Zhou et al.:
$$\sigma(x) = k \cdot \left(1 - \frac{\log |D(x)|}{\log |C|}\right) + (1 - k) \frac{\log \text{depth}(x)}{\log \text{depth}(G_T)}$$
- ▶ extrinsic:
 - ▶ $\sigma(x)$ defined as a function of instances (or annotations) I
 - ▶ note: the number of instances monotonically decreases with increasing depth in taxonomies
 - ▶ Resnik 1995: $eIC_{\text{Resnik}}(x) = -\log p(x)$ (with $p(x) = \frac{|I(x)|}{|I|}$)
 - ▶ in biology, one of the most popular specificity measure when annotations are present

How to measure similarity?



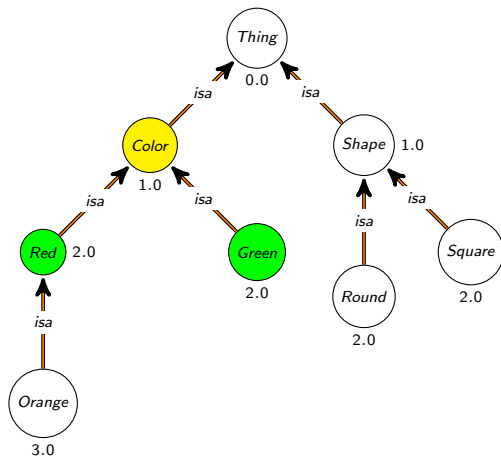
- Resnik 1995:
similarity between x and y is the information content of the *most informative common ancestor*

How to measure similarity?



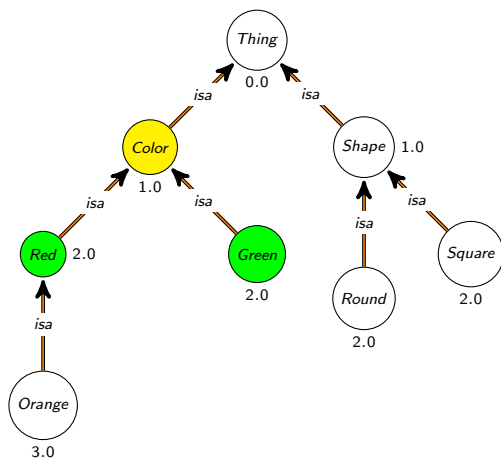
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How to measure similarity?



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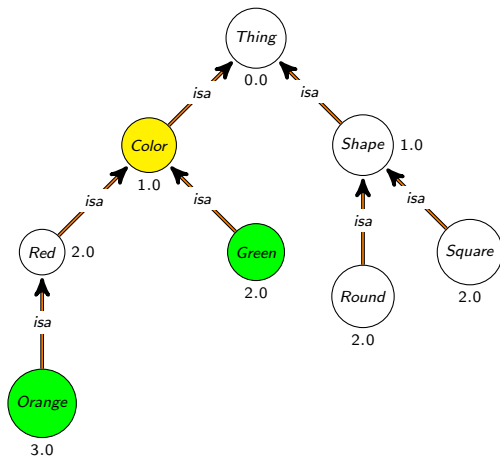
How to measure similarity?



- ▶ Resnik 1995:
similarity between x and y is the
information content
of the *most informative common ancestor*

- ▶
$$\text{sim}_{\text{Resnik}}(\text{Green}, \text{Red}) = 1.0$$

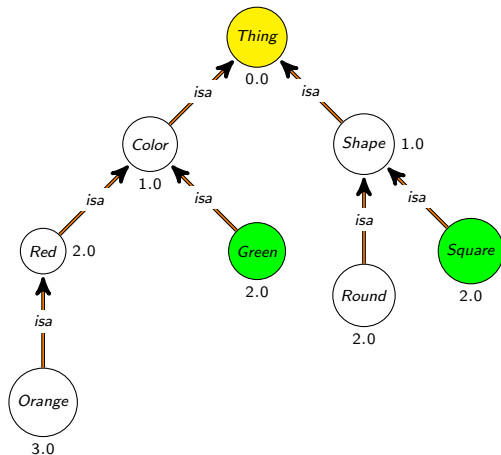
How to measure similarity?



- ▶ Resnik 1995:
similarity between x and y is the
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- ▶
$$\text{sim}_{\text{Resnik}}(\text{Green}, \text{Orange}) = 1.0$$

How to measure similarity?



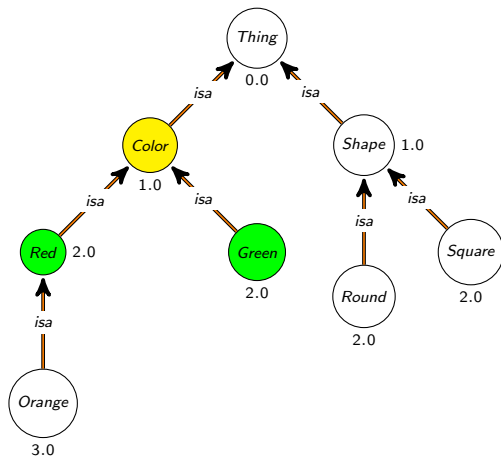
- ▶ Resnik 1995:
similarity between x
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- ▶
 $sim_{Resnik}(Square, Orange)$
0.0

How to measure similarity?

- ▶ (Red, Green) and (Orange, Green) have the same similarity
- ▶ need to incorporate the specificity of the compared classes

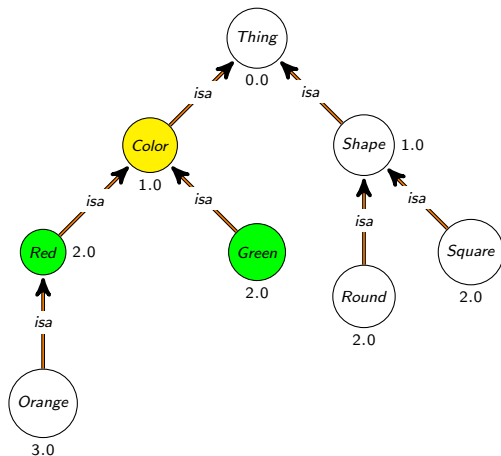
How to measure similarity?



► Lin 1998:

$$sim_{Lin}(x, y) = \frac{2 \cdot IC(MICA(x, y))}{IC(x) + IC(y)}$$

How to measure similarity?

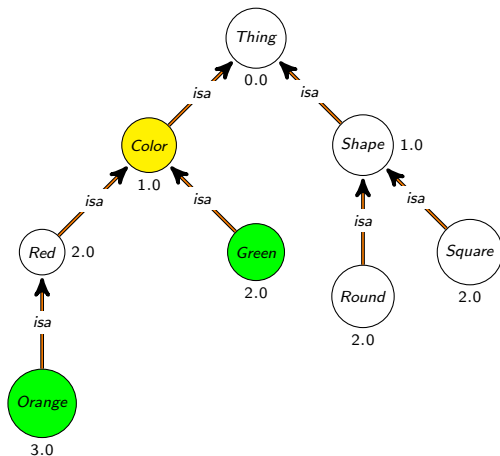


► Lin 1998:

$$sim_{Lin}(x, y) = \frac{2 \cdot IC(MICA(x, y))}{IC(x) + IC(y)}$$

► $sim_{Lin}(Green, Red) = 0.5$

How to measure similarity?



► Lin 1998:

$$sim_{Lin}(x, y) = \frac{2 \cdot IC(MICA(x, y))}{IC(x) + IC(y)}$$

►

$$sim_{Lin}(Green, Orange) = 0.4$$

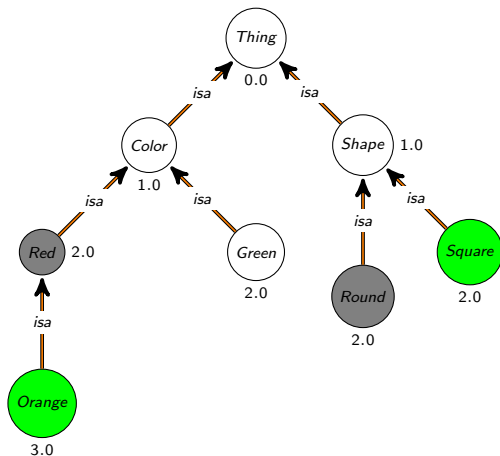
How to measure similarity?

- ▶ many(!) others:
 - ▶ Jiang & Conrath 1997
 - ▶ Mazandu & Mulder 2013
 - ▶ Schlicker et al. 2009
 - ▶ ...

How to measure similarity?

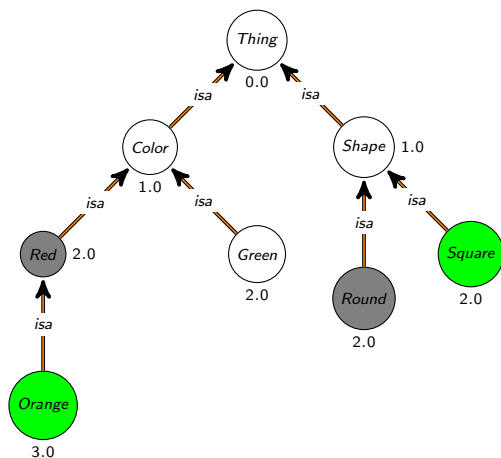
- ▶ we only looked at comparing pairs of classes
- ▶ mostly, we want to compare *sets* of classes
 - ▶ set of GO annotations
 - ▶ set of signs and symptoms
 - ▶ set of phenotypes
- ▶ two approaches:
 - ▶ compare each class individually, then merge
 - ▶ directly set-based similarity measures

How to measure similarity?



- similarity between a square-and-orange thing and a round-and-red thing

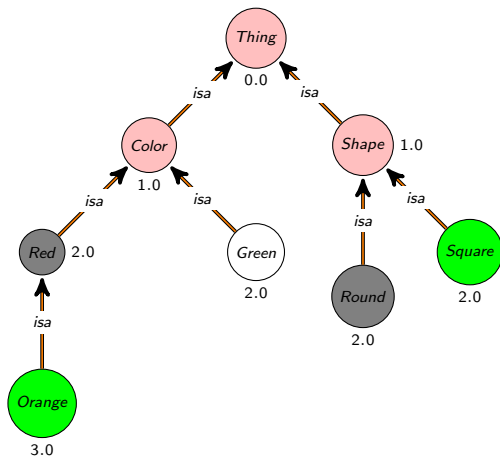
How to measure similarity?



- ▶ similarity between a square-and-orange thing and a round-and-red thing
- ▶ Pesquita et al., 2007:

$$\text{simGIC}(X, Y) = \frac{\sum_{c \in A(X) \cap A(Y)} IC(c)}{\sum_{c \in A(X) \cup A(Y)} IC(c)}$$

How to measure similarity?



- ▶ similarity between a square-and-orange thing and a round-and-red thing
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$$\text{simGIC}(X, Y) = \frac{\sum_{c \in A(X) \cap A(Y)} IC(c)}{\sum_{c \in A(X) \cup A(Y)} IC(c)}$$
- ▶ $\text{simGIC}(so, rr) = \frac{2}{11}$

How to measure similarity?

- ▶ alternatively: use different merging strategies
- ▶ common: average, maximum, **best-matching average**
 - ▶ Average: $sim_A(X, Y) = \frac{\sum_{x \in X} \sum_{y \in Y} sim(x, y)}{|X| \times |Y|}$
 - ▶ Max average: $sim_{MA}(X, Y) = \frac{1}{|X|} \sum_{x \in X} \max_{y \in Y} sim(x, y)$
 - ▶ Best match average: $sim_{BMA}(X, Y) = \frac{sim_{MA}(X, Y) + sim_{MA}(Y, X)}{2}$

How to measure similarity?

- ▶ Semantic Measures Library:
 - ▶ comprehensive Java library
 - ▶ <http://www.semantic-measures-library.org/>
- ▶ R packages: GOSim, GOSemSim, HPOSim, LSAfun, ontologySimilarity,...
- ▶ Python: sematch, fastsemsim (GO only)

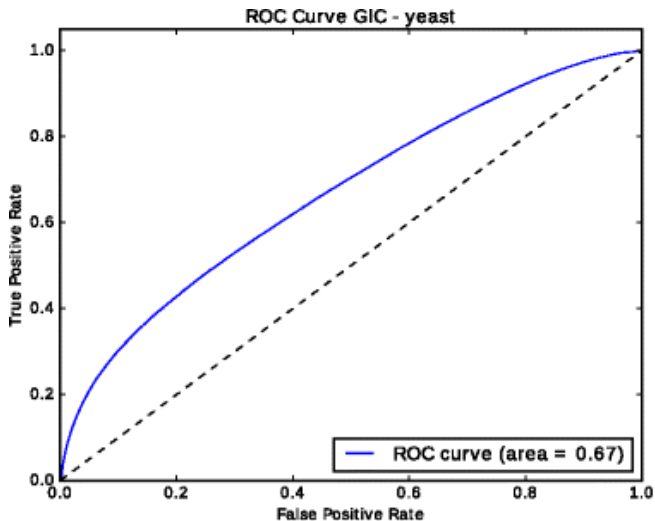
Applications of semantic similarity

Hypothesis

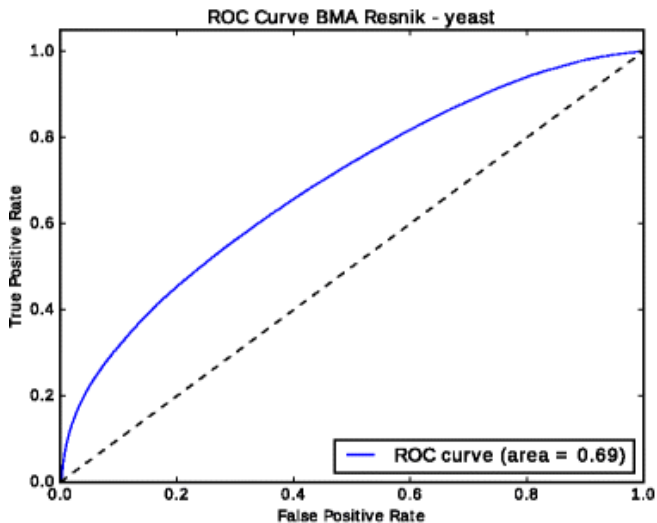
Interacting proteins have similar functions.

- ▶ relies on background knowledge about functions (encoded in GO)
- ▶ “similarity” can mean:
 - ▶ part of the same pathway
 - ▶ siblings of a common super-class
 - ▶ located in the same location
- ▶ set-based comparison of GO functions
 - ▶ single GO hierarchy or all?
 - ▶ which similarity measure?

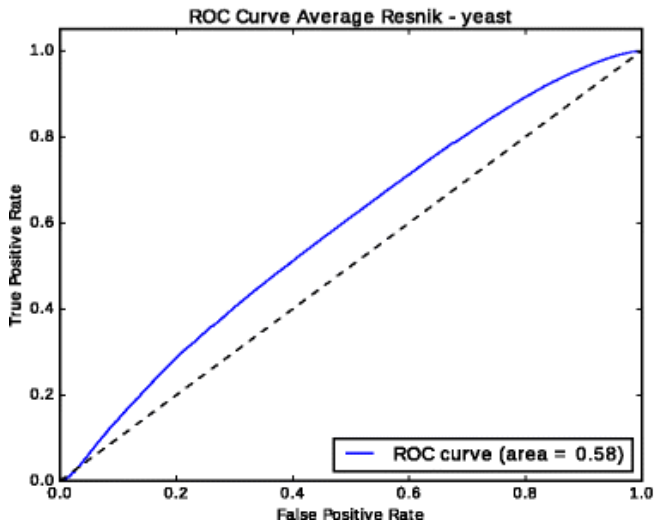
Applications of semantic similarity



Applications of semantic similarity



Applications of semantic similarity



Applications of semantic similarity

- ▶ no obvious choice of similarity measure
- ▶ depends on application
 - ▶ predicting PPIs in different organisms may benefit from a different similarity measure!
- ▶ different similarity measures may react differently to biases in data
- ▶ needs some testing and experience

Applications of semantic similarity

Recommendations:

- ▶ use Resnik's information content measure
- ▶ use Resnik's similarity
- ▶ use Best Match Average
- ▶ use the full ontology
- ▶ classify your ontology using an automated reasoner before applying semantic similarity
 - ▶ although many ontologies come pre-classified
- ▶ \Rightarrow but there are many exceptions
 - ▶ similar location \Rightarrow use location subset of GO
 - ▶ developmental phenotypes \Rightarrow use developmental branch of phenotype ontology

Applications of semantic similarity

- ▶ choice of ontology determines the kind of similarity
- ▶ functional similarity: Gene Ontology
- ▶ anatomical, structural similarity: anatomy ontologies (Uberon, MA, FMA, etc.)
- ▶ phenotypic similarity: phenotype ontology (HPO, MP, etc.)
- ▶ chemical structural similarity: ChEBI

Applications of semantic similarity

- ▶ phenotypic similarity used to:
 - ▶ diagnosis: similarity between patient phenotypes and disease phenotypes
 - ▶ also between patient phenotypes and gene–phenotype associations
 - ▶ Phenomizer: <http://compbio.charite.de/phenomizer/>
 - ▶ disease modules: similarity between disease and disease
 - ▶ clustering/stratification: similarity between patient and patient
 - ▶ disease gene discovery: similarity between patient/disease phenotypes and gene–phenotype associations
 - ▶ in humans
 - ▶ in model organisms
 - ▶ drug repurposing: side-effect similarity; similarity between side effect profile and gene–disease associations

Applications of semantic similarity

- ▶ comparing entities annotated with *different* ontologies/vocabularies of the *same* (or related) domains
 - ▶ medical: UMLS, HPO, DO, ORDO, NCIT, ICD, SNOMED CT, MeSH, ...
 - ▶ phenotype: HPO, MP, CPO, WBPhenotype, FBCV, MeSH, ...
 - ▶ chemical: ChEBI, MeSH, DrOn, RXNorm, DrugBank, ...
- ▶ needs mapping, alignment, or integration
 - ▶ mapping: given a term t , find corresponding class in ontology O
 - ▶ can be 1:1, 1:n, n:1, n:m
 - ▶ t can be from ontology, vocabulary, database, or text
 - ▶ use O for analysis
 - ▶ alignment: given two ontologies or vocabularies O_1 and O_2 , find all mappings between classes/terms in O_1 and O_2
 - ▶ applicable to ontologies and vocabularies
 - ▶ use O_1 or O_2 for analysis
 - ▶ integration: given two ontologies O_1 and O_2 , combine both ontologies into a single ontology O
 - ▶ maintain meaning of classes
 - ▶ use O for analysis

Applications of semantic similarity

- ▶ lexical mappings: use class labels (and synonyms) to find matches
 - ▶ hypertension (HP:0000822) and hypertension (MP:0000231)
- ▶ semantic mappings: use class axioms to find matches
 - ▶ pulmonary valve stenosis (MP:0006182) and Pulmonic stenosis (HP:0001642)
 - ▶ both definitions based on constricted (PATO:0001847) and pulmonary valve (UBERON:0002146)
- ▶ hybrid: combine lexical and semantic mappings

Applications of semantic similarity

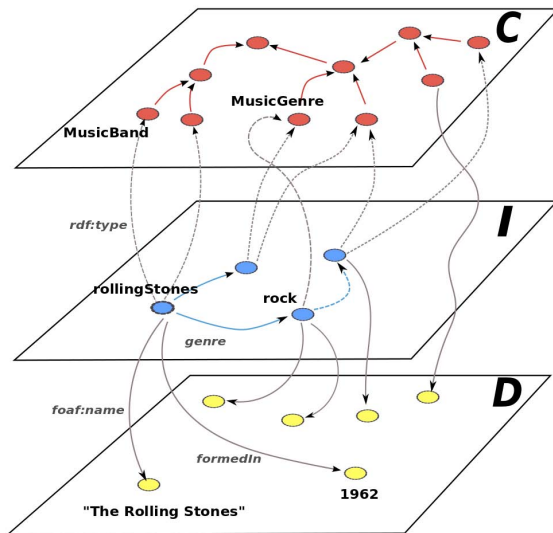
tools for ontology mapping, matching, integration:

- ▶ AgreementMaker Light:
<https://github.com/AgreementMakerLight/AML-Jar>
 - ▶ structural (semantic) and lexical matches
 - ▶ can use domain-specific background knowledge
- ▶ LogMap: <https://github.com/ernestojimenezruiz/logmap-matcher>
 - ▶ structural (semantic) and lexical matches
 - ▶ biology-themed versions
- ▶ NCBO Annotator:
<https://bioportal.bioontology.org/annotator>
 - ▶ lexical matches only
 - ▶ can annotate full text
- ▶ recent tools and comprehensive ongoing evaluation:
 - ▶ OAEI: <http://oei.ontologymatching.org/>

Hands-on part 2

- ▶ back to Jupyter...
- ▶ run the “Semantic Similarity” part
 - ▶ then find the mouse genotype with the most similar set of phenotypes to “Tetralogy of Fallot” (OMIM:187500)
 - ▶ or: use the data from <https://hpo.jax.org/app/download/annotation> to add more diseases and query by disease (hint: a disease is really just a set of phenotypes)

How to measure similarity?



From Harispe et al., Semantic Similarity From Natural Language And Ontology Analysis, 2015.

How to measure similarity?

▶ Shortest Path

- ▶ applicable to arbitrary knowledge graphs
- ▶ does not capture similarity well over all edge types, e.g., *disjointWith*, *differentFrom*, *opposite-of*, etc.

▶ Random Walk

- ▶ with or without restart
- ▶ iterated
- ▶ does not consider edge labels \Rightarrow captures only adjacency of nodes
- ▶ scores whole graph with *probability* of being in a state
- ▶ can take multiple seed nodes
 - ▶ widely used to find disease genes

How to measure similarity?

- ▶ feature learning on knowledge graph

How to measure similarity?

- ▶ feature learning on knowledge graph
- ▶ e.g., iterated, edge-labeled random walk
 - ▶ walks form *sentences*
 - ▶ sentences form a *corpus*
 - ▶ feature learning on corpus through Word2Vec (or factorization of co-occurrence matrix)

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 - ▶ with support for reasoning over bio-ontologies:
<https://github.com/bio-ontology-research-group/walking-rdf-and-owl>
 - ▶ Onto2Vec: <https://github.com/bio-ontology-research-group/onto2vec/>

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- ▶ Translational knowledge graph embeddings: TransE, TransR, TransE, HolE, etc.
 - ▶ analogy-based
 - ▶ <https://github.com/thunlp/KB2E>

How to measure similarity?

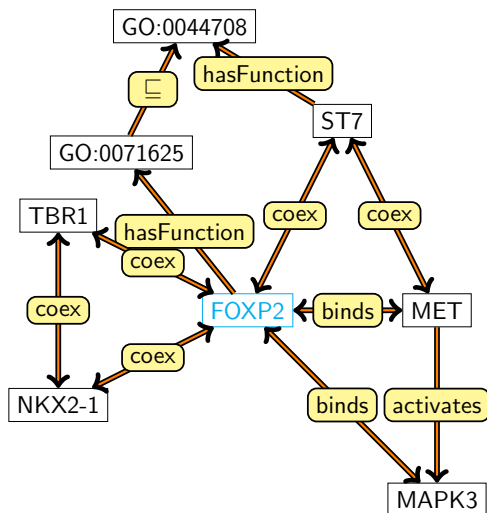
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 - ▶ <https://github.com/thunlp/KB2E>
- ▶ generates (dense) feature vectors for nodes (classes, instances) and relations

Knowledge graph embeddings

Definition

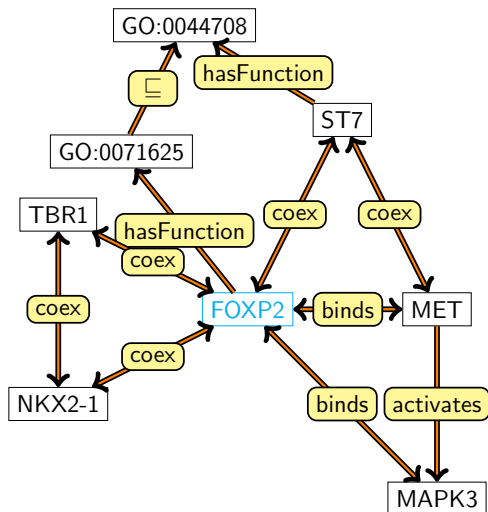
Let $KG = (V, E, L; \vdash)$ be a knowledge graph with a set of vertices V , a set of edges $E \subseteq V \times V$, a label function $L : V \cup E \mapsto Lab$ that assigns labels from a label set Lab to vertices and edges, and an inference relation \vdash . A knowledge graph embedding is a function $f_\eta : KG \mapsto \mathbf{R}^n$ (subject to certain constraints).

Neuro-symbolic feature learning

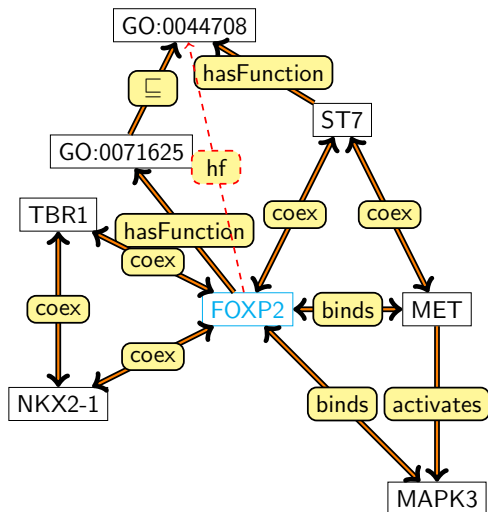


- task: predict if FOXP2 is involved in disease D
- task: what chemicals could (directly or indirectly) affect FOXP2's function?
- which features are relevant?

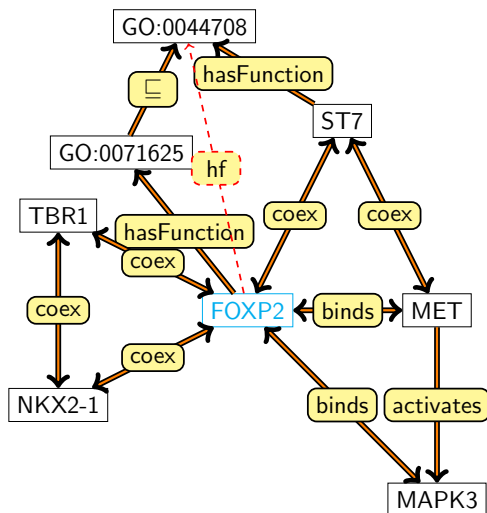
Neuro-symbolic feature learning



Neuro-symbolic feature learning

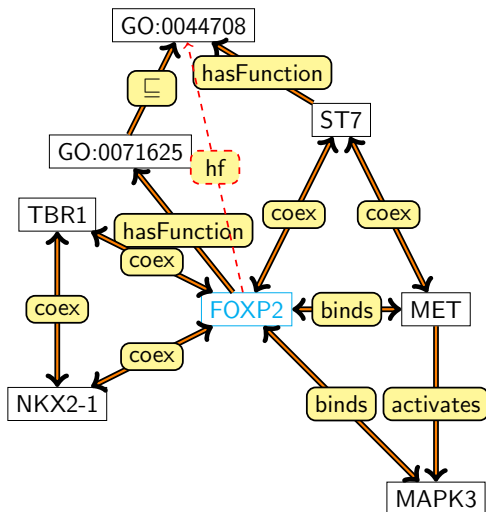


Neuro-symbolic feature learning



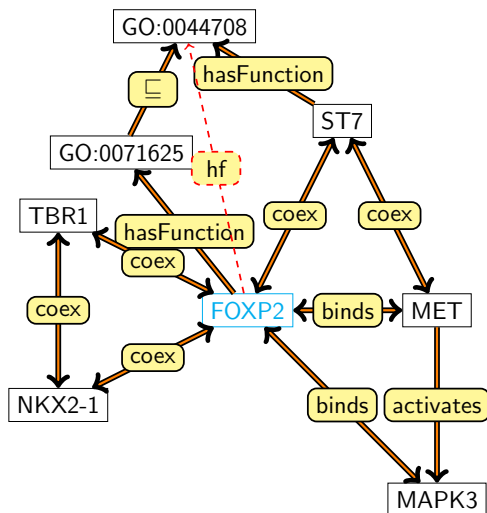
- :FOXP2 :binds :MET
- :coex :ST7
- :hasFunction
- GO:0044708

Neuro-symbolic feature learning



- :FOXP2 :binds :MET
:coex :ST7
:hasFunction
GO:0044708
- :FOXP2 :hasFunction
GO:0071625
subClassOf
GO:0044708

Neuro-symbolic feature learning

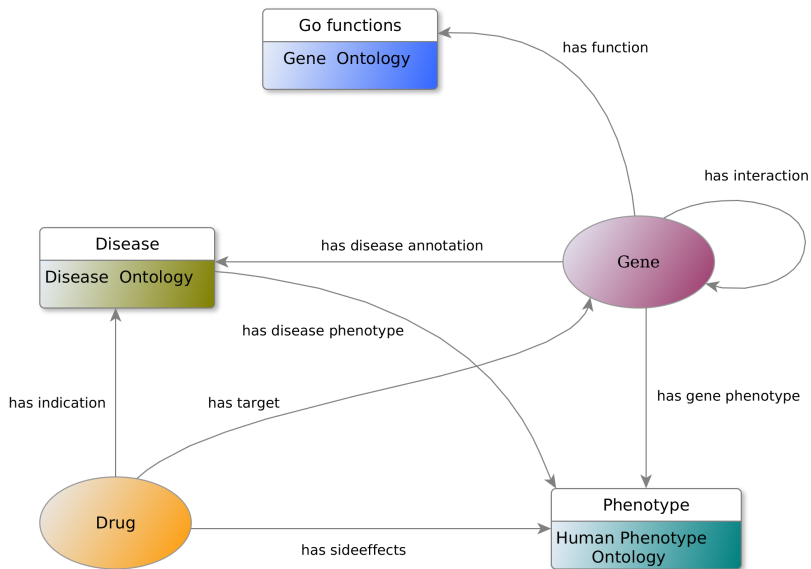


- ▶ :FOXP2 :binds :MET
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GO:0044708
- ▶ :FOXP2 :hasFunction
GO:0071625
subClassOf
GO:0044708
- ▶ :FOXP2 :coex :TBR1
:coex :NKX2-1 :coex
:TBR1 :coex ...

Neuro-symbolic feature learning

- ▶ skip-gram model learns representation/features for each node
 - ▶ Word2Vec model, given a word predicts context
 - ▶ use local and non-local information
- ▶ automated reasoning deductively closes the knowledge graph
 - ▶ making this a neuro-symbolic model
- ▶ useful for edge prediction, similarity, clustering, as feature vectors
 - ▶ edge prediction: analogy, classifier (e.g., SVM)

Neuro-symbolic feature learning



Neuro-symbolic feature learning

Object property	Source type	Target type	Without reasoning		With reasoning	
			F-measure	AUC	F-measure	AUC
has target	Drug	Gene/Protein	0.94	0.97	0.94	0.98
has disease annotation	Gene/Protein	Disease	0.89	0.95	0.89	0.95
has side-effect*	Drug	Phenotype	0.86	0.93	0.87	0.94
has interaction	Gene/Protein	Gene/Protein	0.82	0.88	0.82	0.88
has function*	Gene/Protein	Function	0.85	0.95	0.83	0.91
has gene phenotype*	Gene/Protein	Phenotype	0.84	0.91	0.82	0.90
has indication	Drug	Disease	0.72	0.79	0.76	0.83
has disease phenotype*	Disease	Phenotype	0.72	0.78	0.70	0.77

Alsharani et al. Neuro-symbolic representation learning on biological knowledge graphs. Bioinformatics, 2017.

Multi-modal feature learning

The forkhead-box P2 (FOXP2) gene polymorphism has been reported to be involved in the susceptibility to schizophrenia; however, few studies have investigated the association between FOXP2 gene polymorphism and clinical symptoms in schizophrenia.

Multi-modal feature learning

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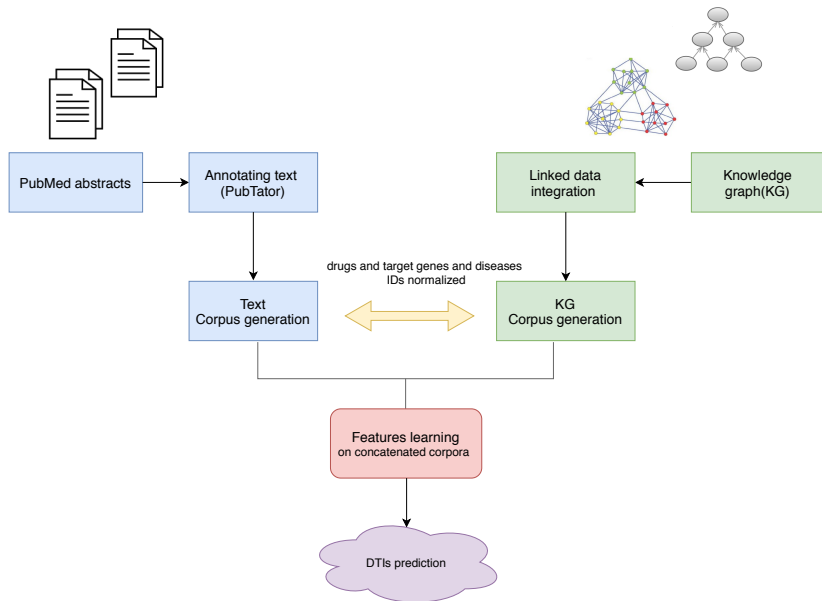
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- ▶ :FOXP2 :coex :TBR1 :coex :NKX2-1 :coex :TBR1 :coex ...

Multi-modal feature learning

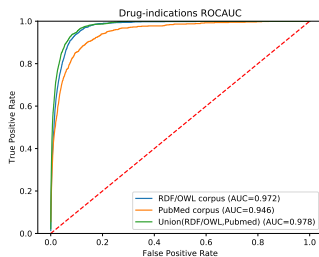
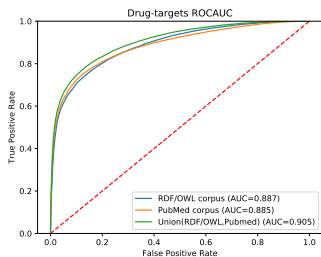
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- ▶ :FOXP2 :binds :MET :coex :ST7 :hasFunction GO:0044708
- ▶ :FOXP2 :hasFunction GO:0071625 subclassOf GO:0044708
- ▶ :FOXP2 :coex :TBR1 :coex :NKX2-1 :coex :TBR1 :coex ...

Multi-modal feature learning



Multi-modal feature learning: drug targets and indications



Alshahrani & H. Drug repurposing through multi-modal learning on knowledge graphs. BioRxiv, 2018.

Ontologies: axioms, not graphs!

Overview

Browse

DLQuery

Download

Annotation	Value
label	B cell apoptotic process
definition	Any apoptotic process in a B cell, a lymphocyte of B lineage with the phenotype CD19-positive and capable of B cell mediated immunity.
class	http://purl.obolibrary.org/obo/GO_0001783
ontology	GO-PLUS
Equivalent	apoptotic process and (occurs in some B cell)
SubClassOf	occurs in some B cell , lymphocyte apoptotic process
id	GO:0001783
has_obo_namespace	biological_process

Ontologies: axioms, not graphs!

Gene Ontology:

- ▶ behavior DisjointWith: 'developmental process'
- ▶ behavior SubclassOf: only-in-taxon some metazoa
- ▶ 'cell proliferation' DisjointWith: in-taxon some fungi
- ▶ 'cell growth' EquivalentTo: growth and ('results in growth of' some cell)
- ▶ ...

Ontology embeddings

Definition

Let $O = (C, R, I; ax; \vdash)$ be an ontology with a set of classes C , a set of relations R , a set of instances I , a set of axioms ax and an inference relation \vdash . An ontology embedding is a function $f_\eta : C \cup R \cup I \mapsto \mathbf{R}^n$ (subject to certain constraints).

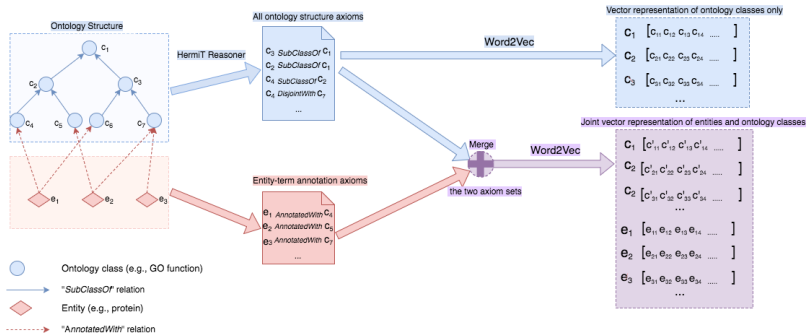
Ontology embeddings

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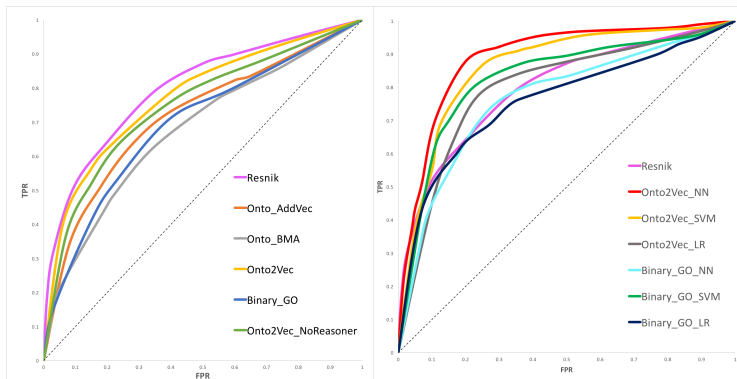
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We use co-occurrence within ax^\vdash to constrain the embedding function, where the constraints on co-occurrence are formulated using the Word2Vec skipgram model.

Onto2Vec

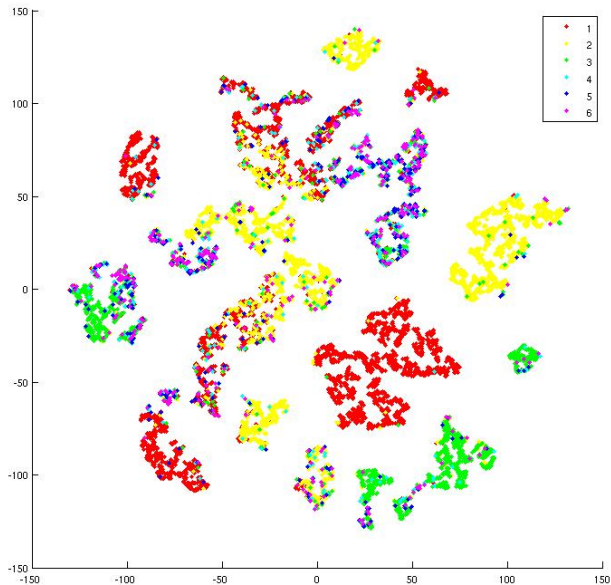


Predicting PPIs: trainable similarity measures



Smaili et al. Onto2Vec: joint vector-based representation of biological entities and their ontology-based annotations, Bioinformatics, 2018.

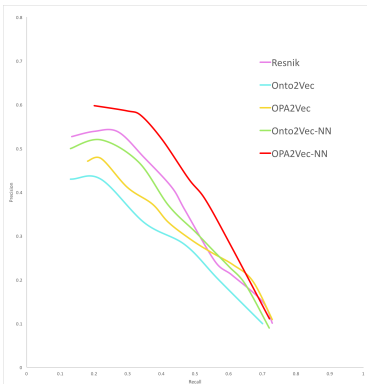
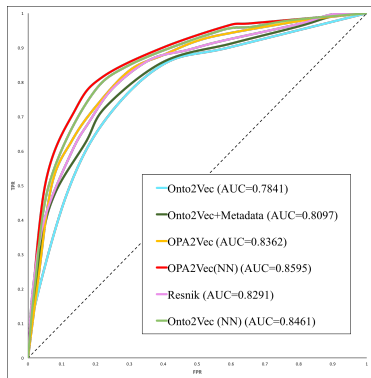
Visualizing embeddings



Ontologies Plus Annotations 2 Vec



Phenotype-based prediction of candidate genes



Hands-on part

- ▶ run the machine learning with ontologies part
- ▶ explore the different similarity measures
- ▶ repeat what you have done with semantic similarity using the ontology embeddings

How to measure similarity?

- ▶ vector-based similarity measure

- ▶ cosine similarity:
$$\text{sim}(X, Y) = \frac{\sum_{i=1}^n X_i Y_i}{\sqrt{\sum_{i=1}^n X_i^2} \sqrt{\sum_{i=1}^n Y_i^2}}$$

- ▶ bounded between $[-1, 1]$

- ▶ Euclidean distance:
$$\text{sim}(X, Y) = \sqrt{\sum_{i=1}^n (X_i - Y_i)^2}$$

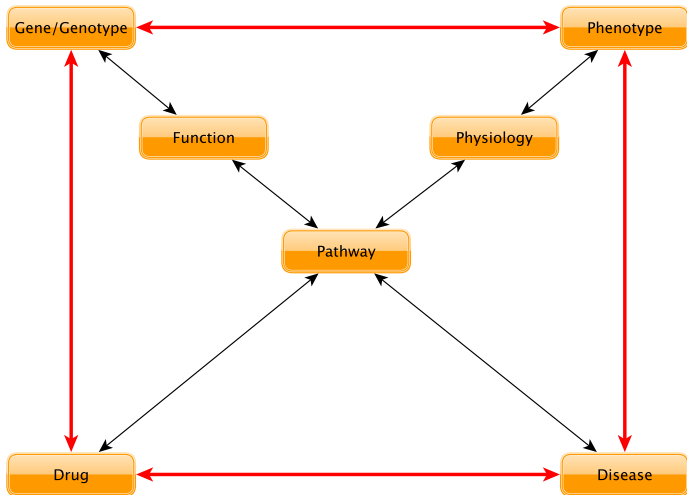
- ▶ not bounded (and rarely used)

- ▶ any other kind of function

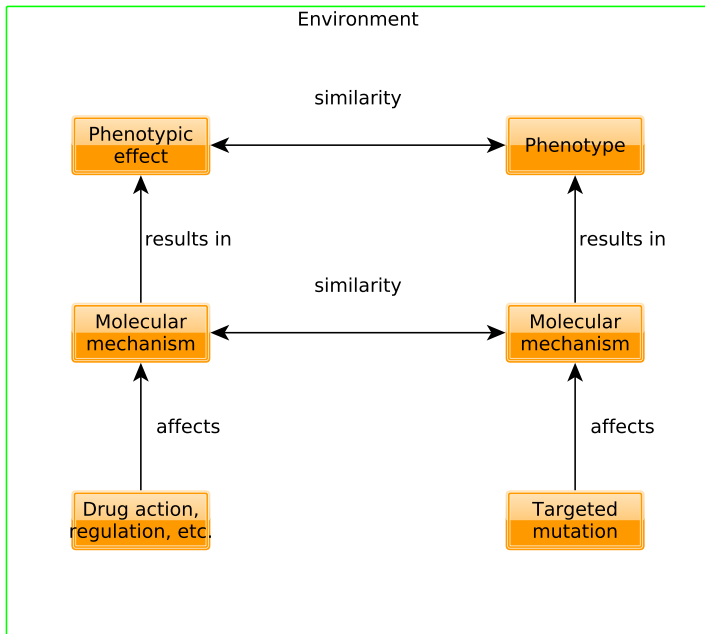
- ▶ Neural Networks can approximate *any* function (universal approximation theorem)
 - ▶ “trainable” semantic similarity measures
 - ▶ will need training data

Applications of semantic similarity

Environment



Applications of semantic similarity



Applications of semantic similarity

- ▶ same kind of entity, same ontology:
 - ▶ x is associated with y
 - ▶ z is similar to x
 - ▶ therefore: z may *also* be associated with y
- ▶ candidate genes (polygenic disease):
 - ▶ FunSimMat: similar function \Rightarrow similar/same disease
 - ▶ side effect similarity: similar side effects \Rightarrow similar targets/indications

Applications of semantic similarity

- ▶ different kind of entity, same ontology:
 - ▶ Phenomizer: genotype x associated with phenotypes a ; patient y has symptoms b ; a is similar to b ; therefore: x causes the symptoms in b
 - ▶ PhenomeNET: similar to Phenomizer but using model organism phenotypes (knockouts)
 - ▶ PhenomeDrug: knockout of gene x causes phenotypes a ; drug y causes side effects b ; a is similar to b ; therefore: drug y inhibits x (or: phenotypes b are caused by inhibition of x)
 - ▶ needs to compare model organism phenotypes and human phenotypes \Rightarrow ontology alignment/integration/mapping

Hands-on: semantic similarity

- ▶ if you have not done so *before* the tutorial, don't start now
 - ▶ you need to download *a lot* of data
 - ▶ you can just follow our demonstration and try later
 - ▶ (unless Internet is exceptionally fast for a conference Wifi, then just go ahead and do everything now)
- ▶ Jupyter Notebook
 - ▶ notebooks consist of code and rich text fragments
 - ▶ human readable (with nice figures) *and* executable
 - ▶ need to install the SciJava kernel (default: iPython)
 - ▶ very widely used
- ▶ <https://github.com/bio-ontology-research-group/ontology-tutorial>

Hands-on: semantic similarity

In the tutorial, we will

- ▶ download an ontology
- ▶ explore the ontology with OWLAPI
- ▶ classify the ontology with an OWL reasoner
 - ▶ and query using an OWL reasoner
- ▶ store the inferred version locally
- ▶ use the Semantic Measures Library to:
 - ▶ explore the ontology as graph
 - ▶ compute similarity between classes
 - ▶ use different similarity measures
 - ▶ compare patients to mice
- ▶ learn to use Onto2Vec and OPA2Vec
- ▶ you can build on this and extend for your own research!

Hands-on: semantic similarity

Do the tutorial...

Hands-on: semantic similarity

- ▶ now play with the Notebook:
 - ▶ look at the results list (check MGI)
 - ▶ try another disease (check OMIM)
 - ▶ or a drug effect (check SIDER)
- ▶ you can also test another ontology
 - ▶ GO for functional similarity
 - ▶ ChEBI for chemical (structural) similarity
 - ▶ or yeast phenotypes