Introduction to ontologies

Semantic similarity

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Overview

1. Ontologies and graphs
2. Structural similarity
3. Information theoretic approaches
4. Set-based
5. Applications

Some examples

- Are cyclin dependent kinases *functionally* more similar to lipid kinases or to riboflavin kinases? How about *phenotypically*?

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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 defined in OBO Relation Ontology
  - in first order logic
  - needs to translate them into OWL

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:` $X \overset{\equiv}{\leftrightarrow} Y, \{X, Y\}$

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What properties do we want in a similarity measure?

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- sim is a *normalized* similarity measure if it has values in $[0, 1]$

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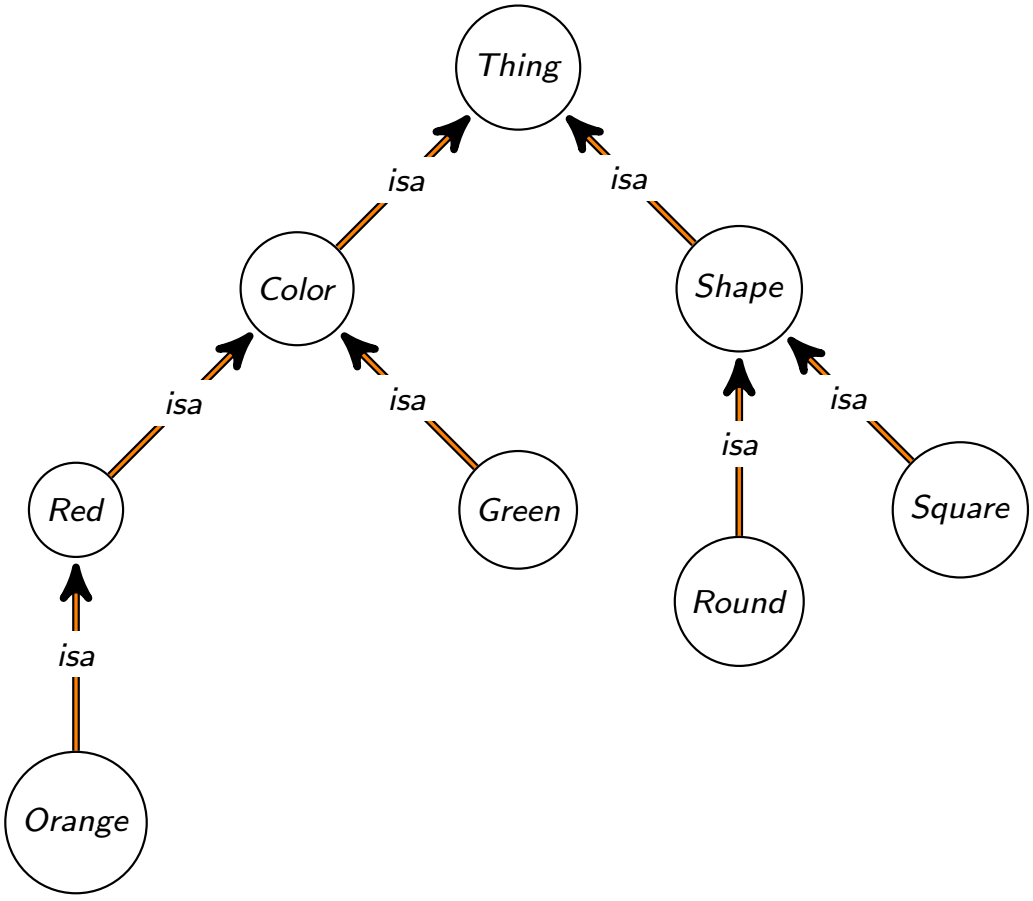

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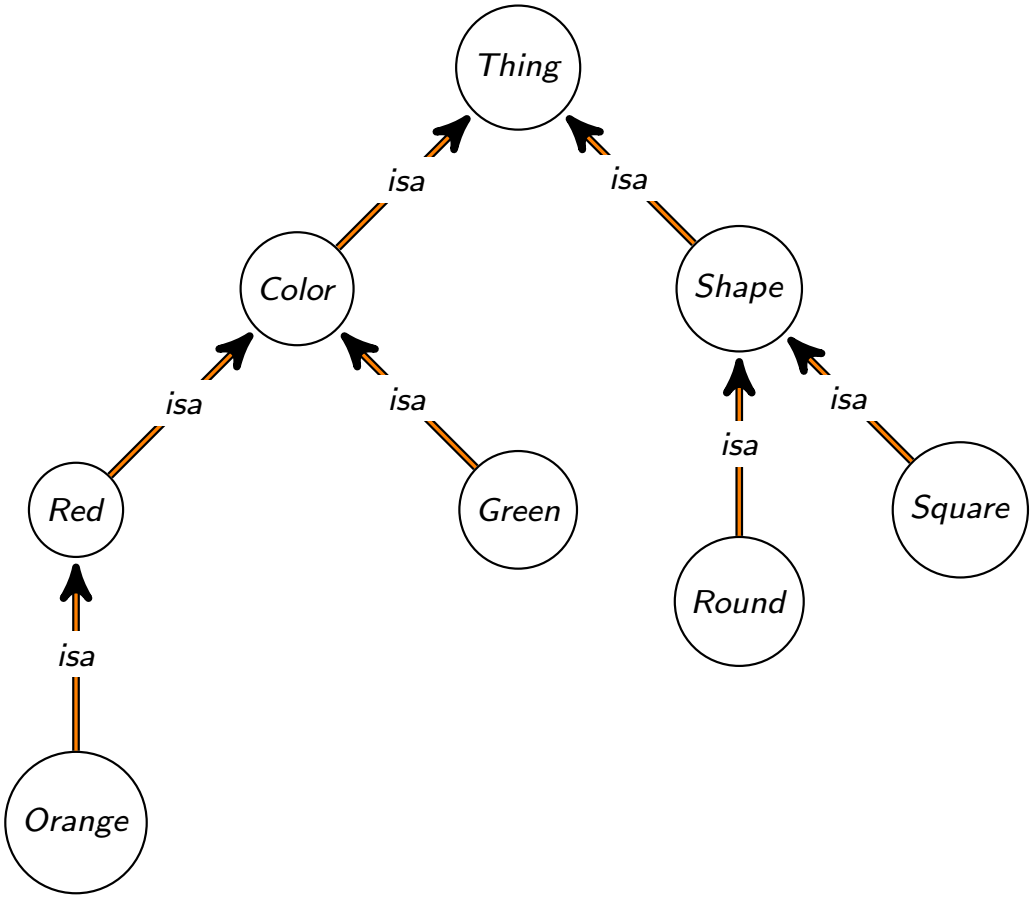

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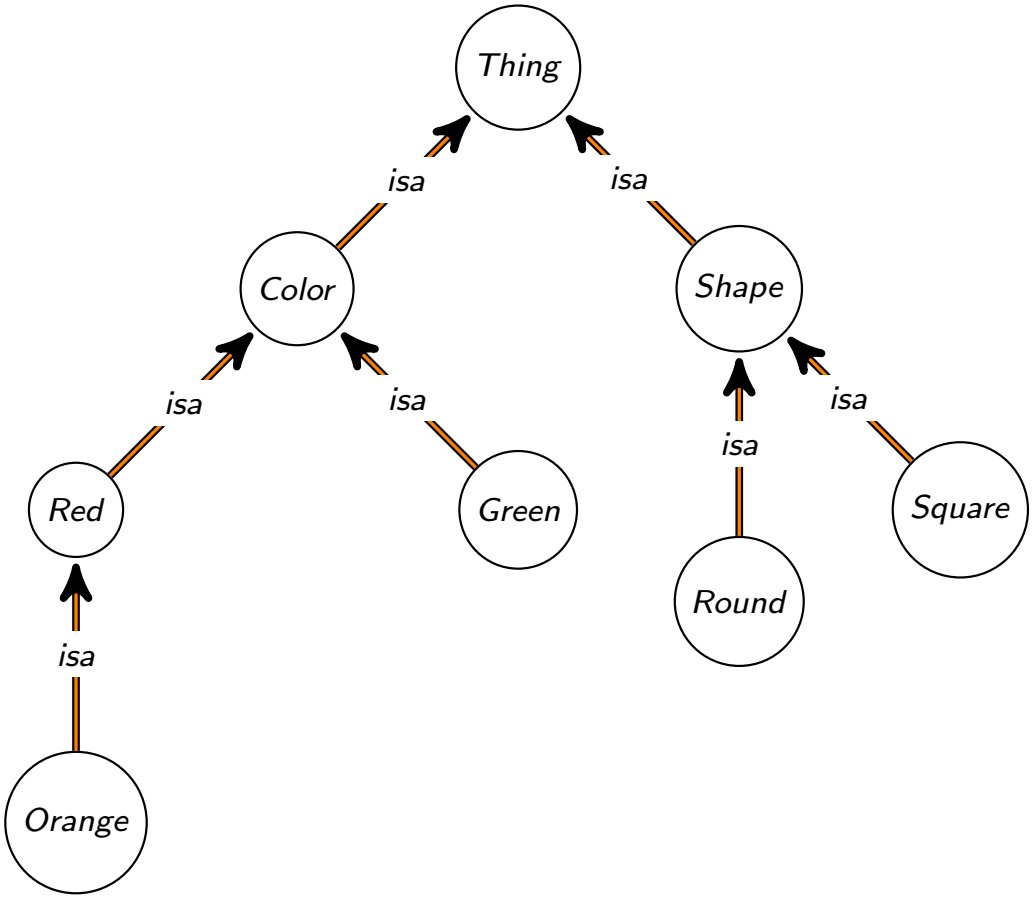

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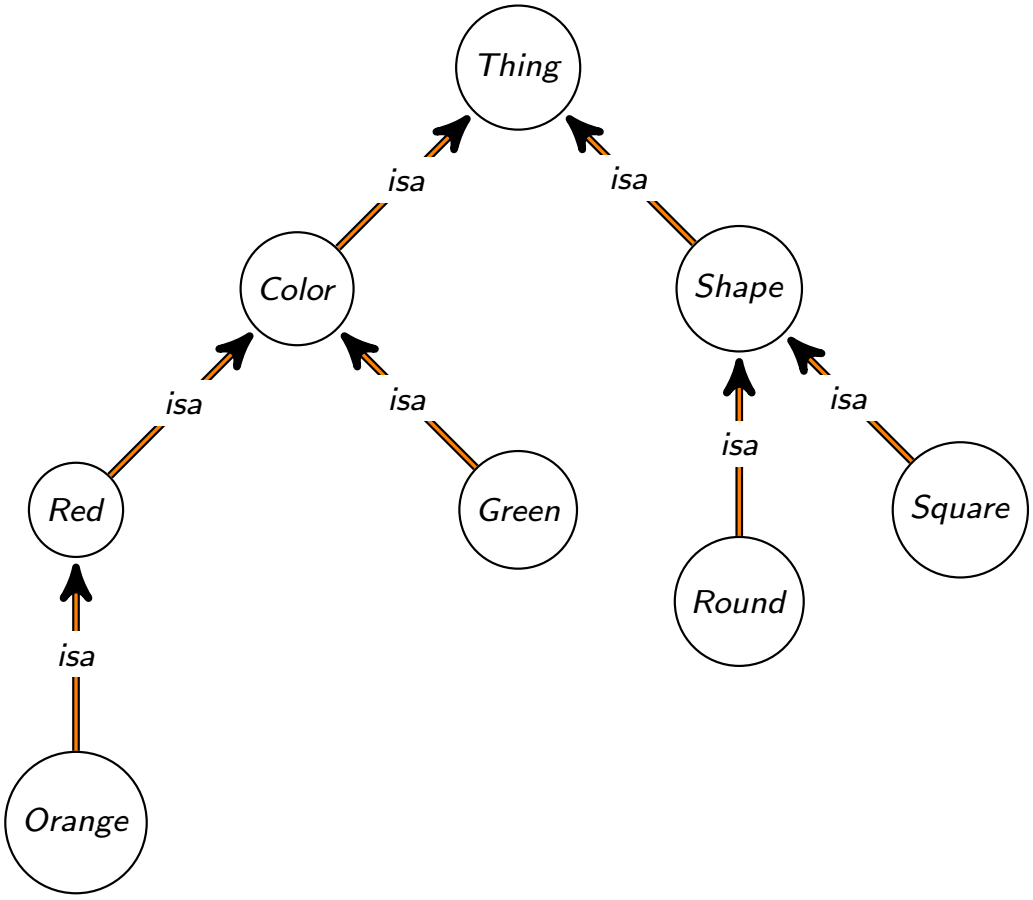

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- $dist_{Rada}(u, v) = sp(u, isa, v)$
- $sim_{Rada}(u, v) = \frac{1}{dist_{Rada}(u,v)+1}$

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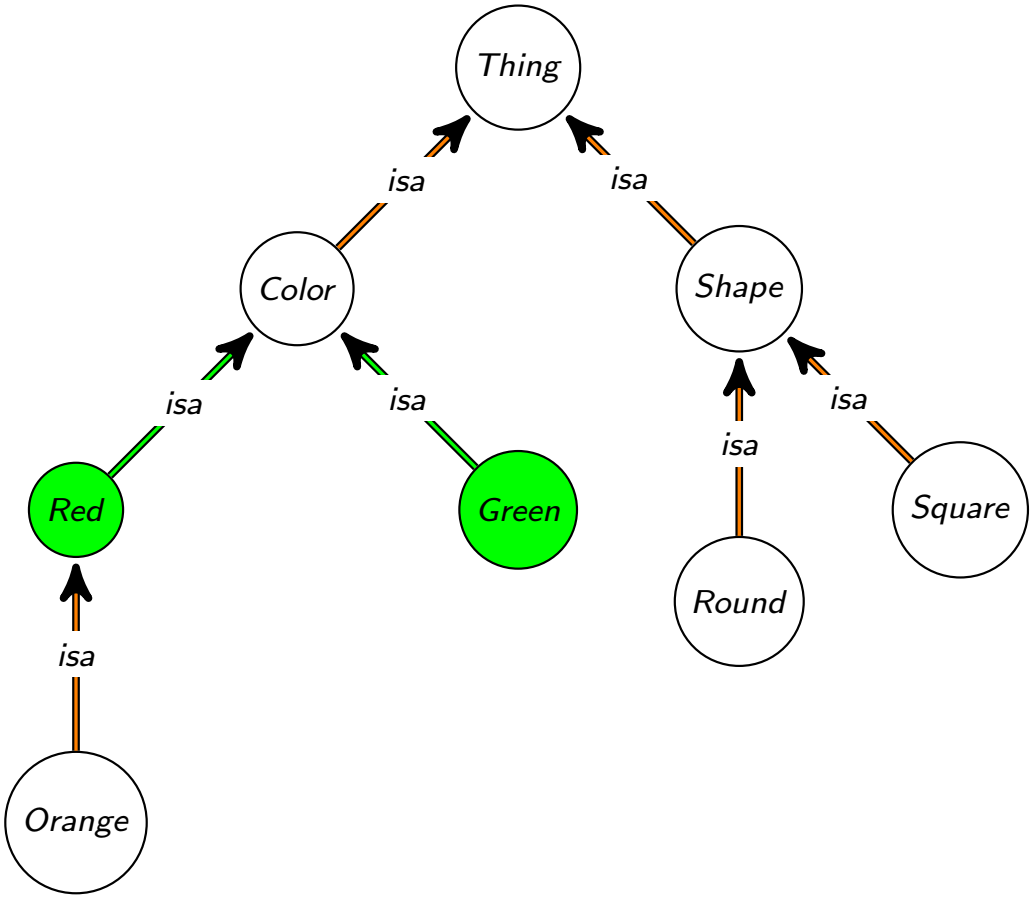

- distance on shortest path
- distance(green, red) $= 2$
- $sim_{Rada}(green, red) = \frac{1}{3}$

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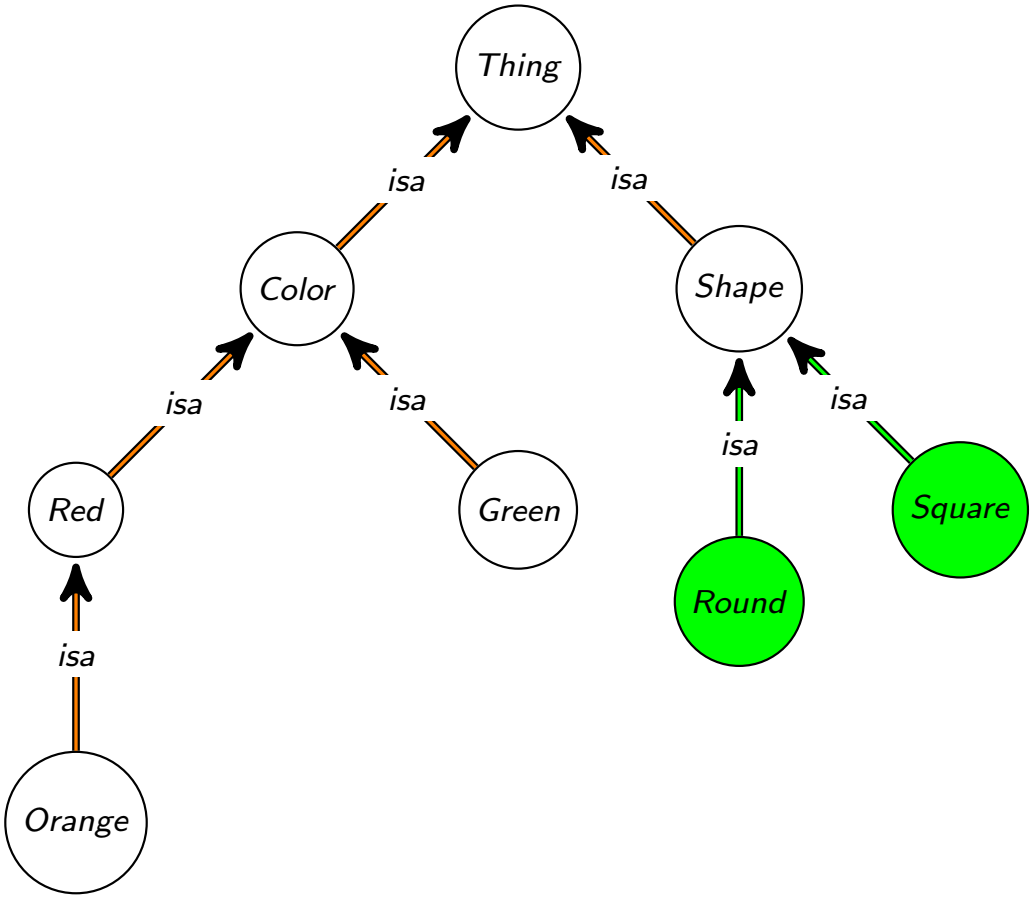

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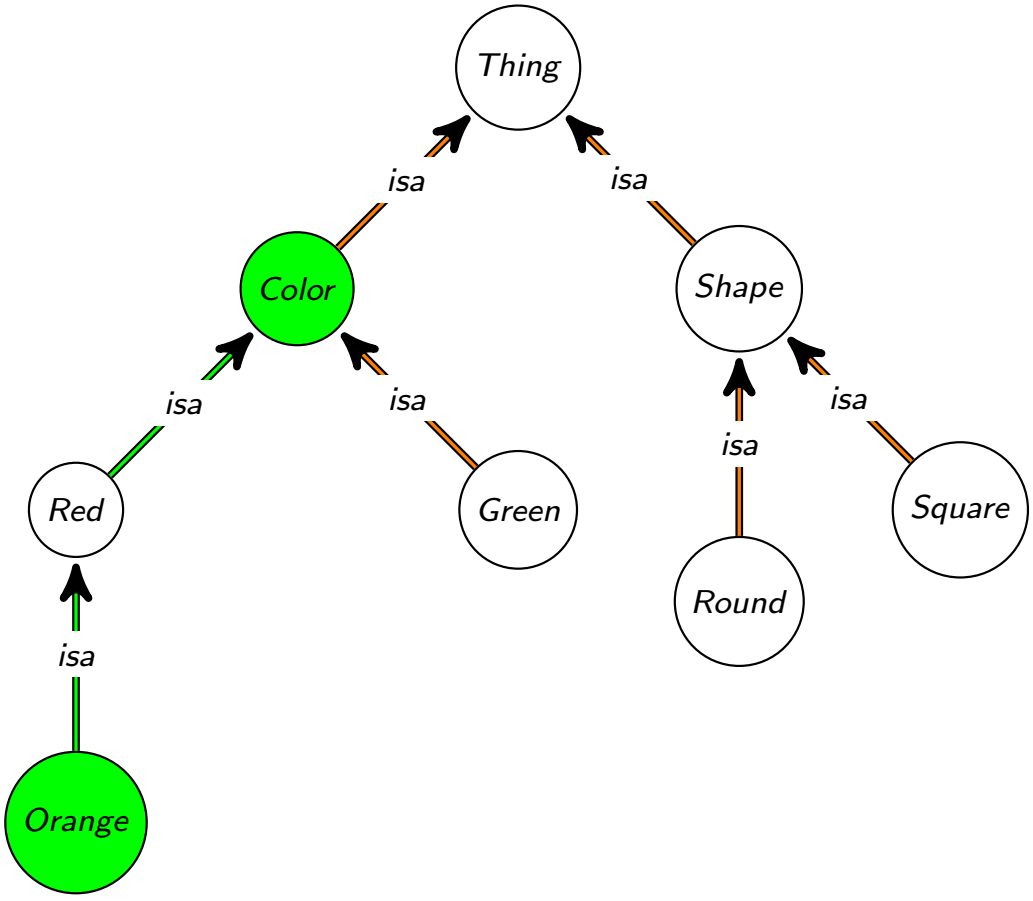

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- account for different edge types
  - non-uniform edge weighting

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- intrinsic:
  - $\sigma(x) = \mathit{depth}(x)$
  - $\sigma(x) = f(A(x))$ (for ancestors $A(x)$)
  - $\sigma(x) = f(D(x))$ (for descendants $D(x)$)
  - many more, e.g., Zhou et al.:
    $\sigma(x) = k \cdot \left(1 - \frac{\log |D(x)|}{\log |C|}\right) + (1 - k)\frac{\log \mathit{depth}(x)}{\log \mathit{depth}(G_T)}$

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- 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: $eIC_{Resnik}(x) = -\log p(x)$ (with $p(x) = \frac{|I(x)|}{|I|}$)
    - in biology, the most popular specificity measure when annotations are present

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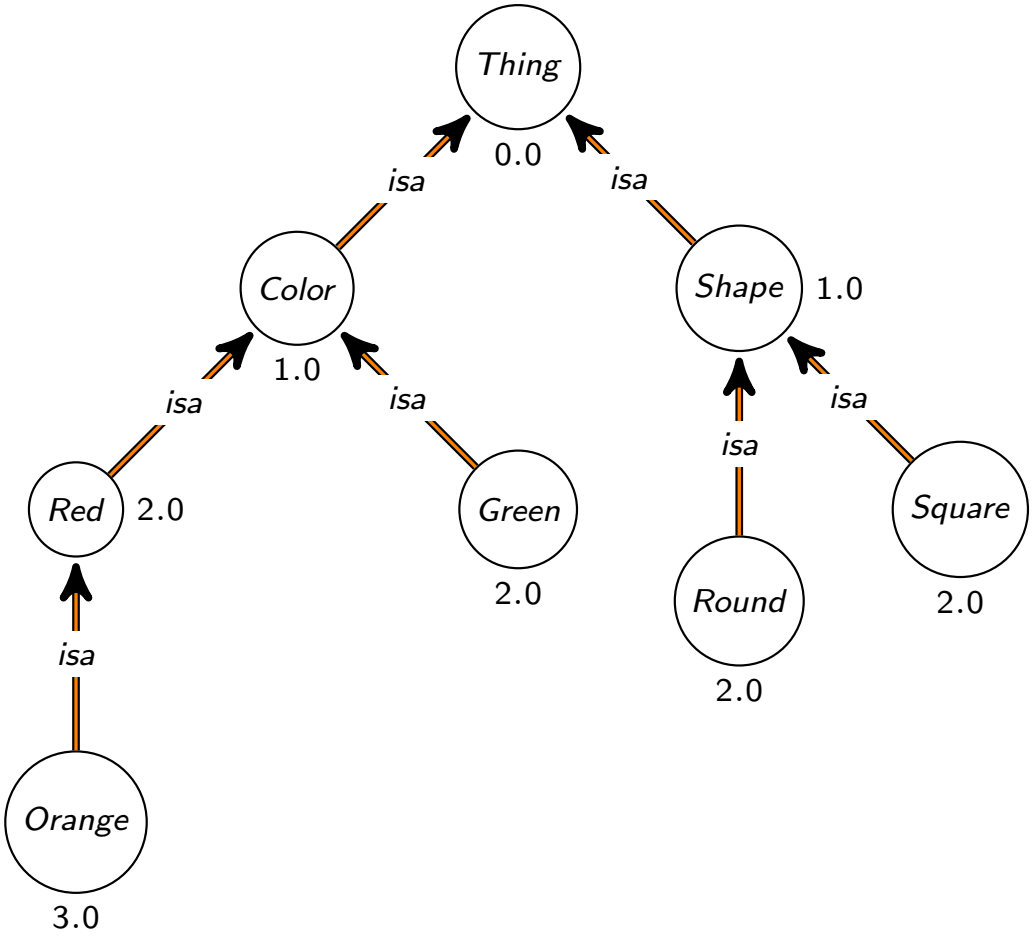

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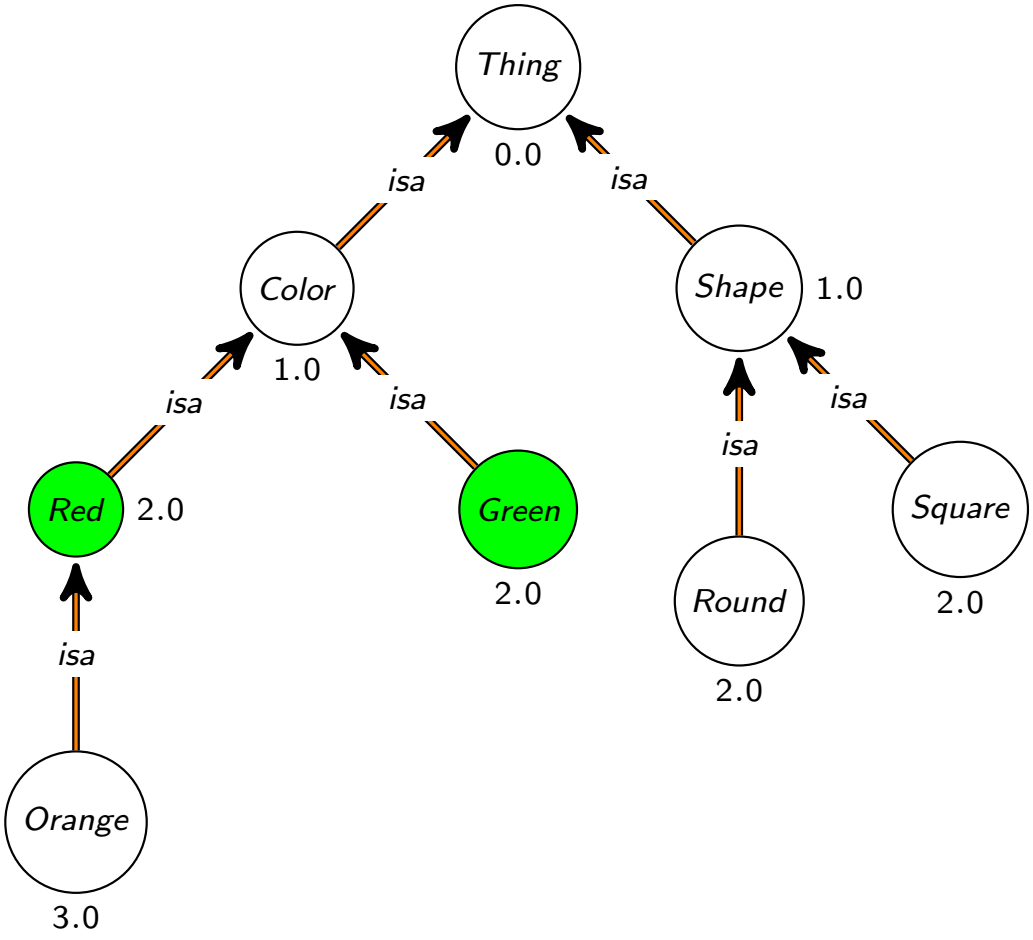

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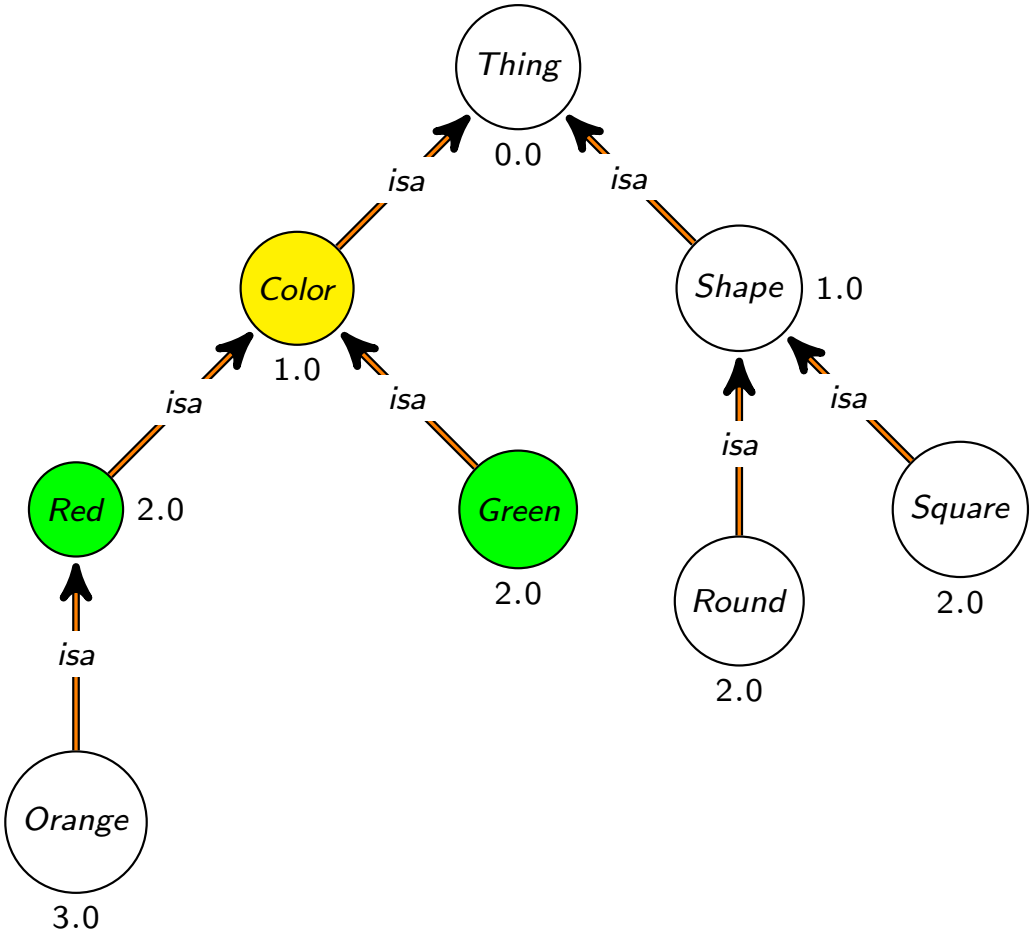

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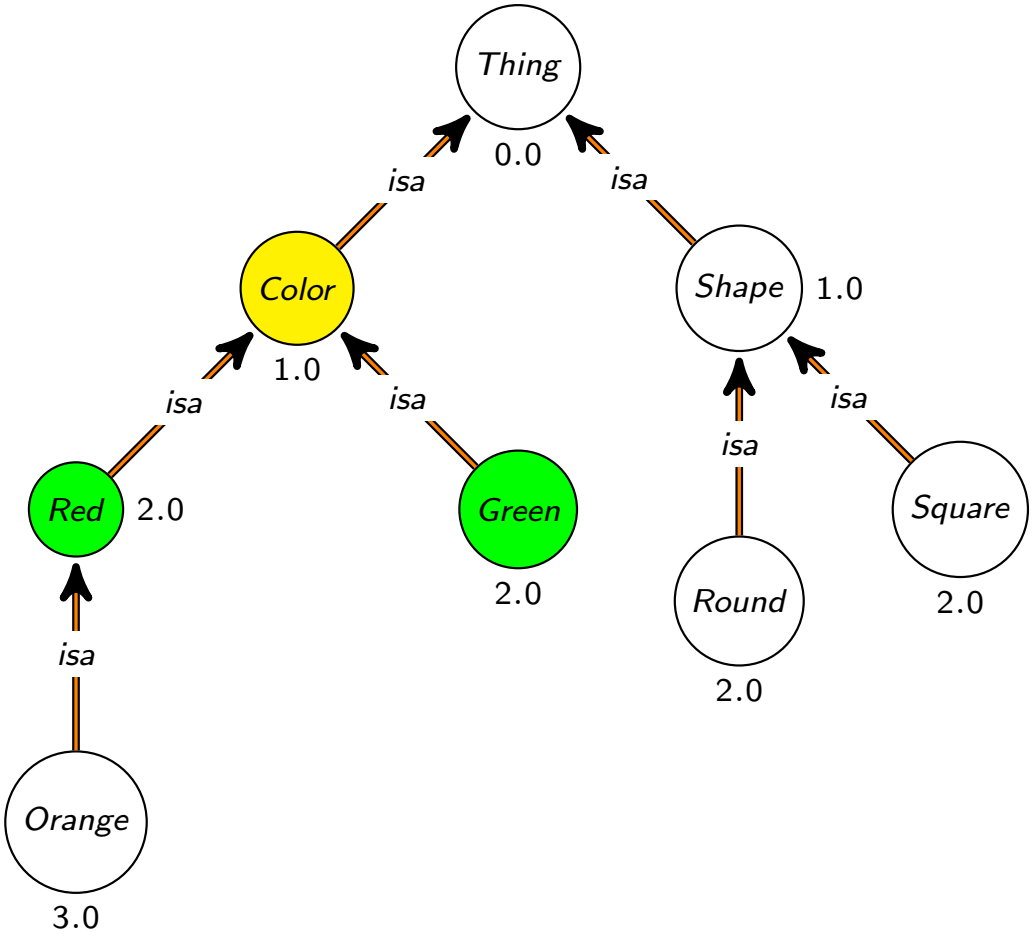

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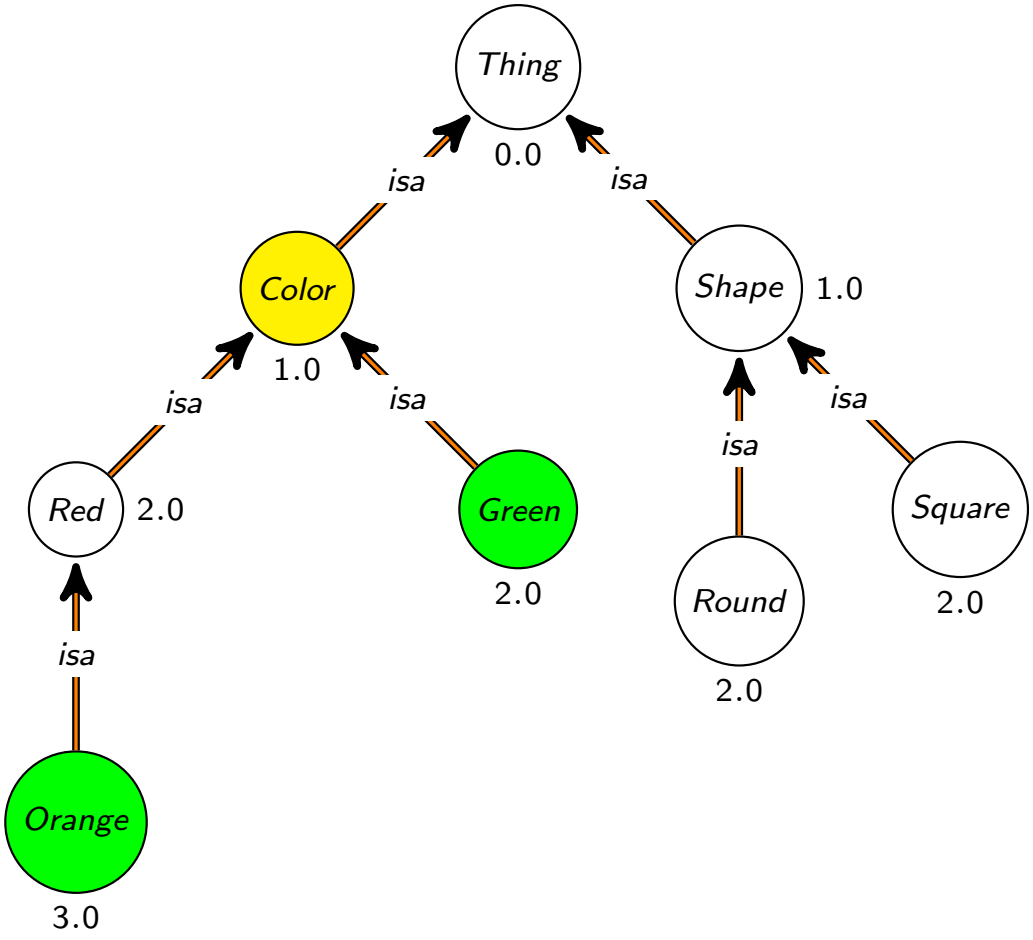

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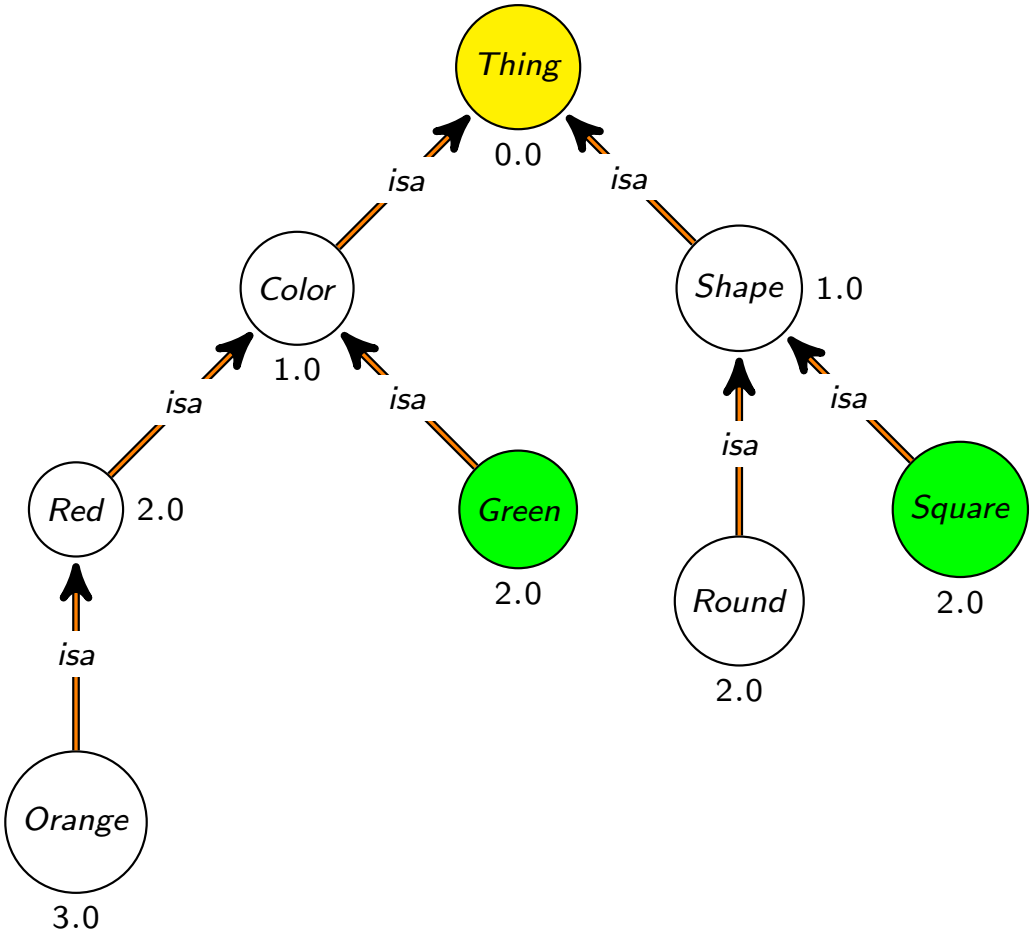

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- $sim_{Resnik}(Square, Orange)$ 0.0

How to measure similarity?

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

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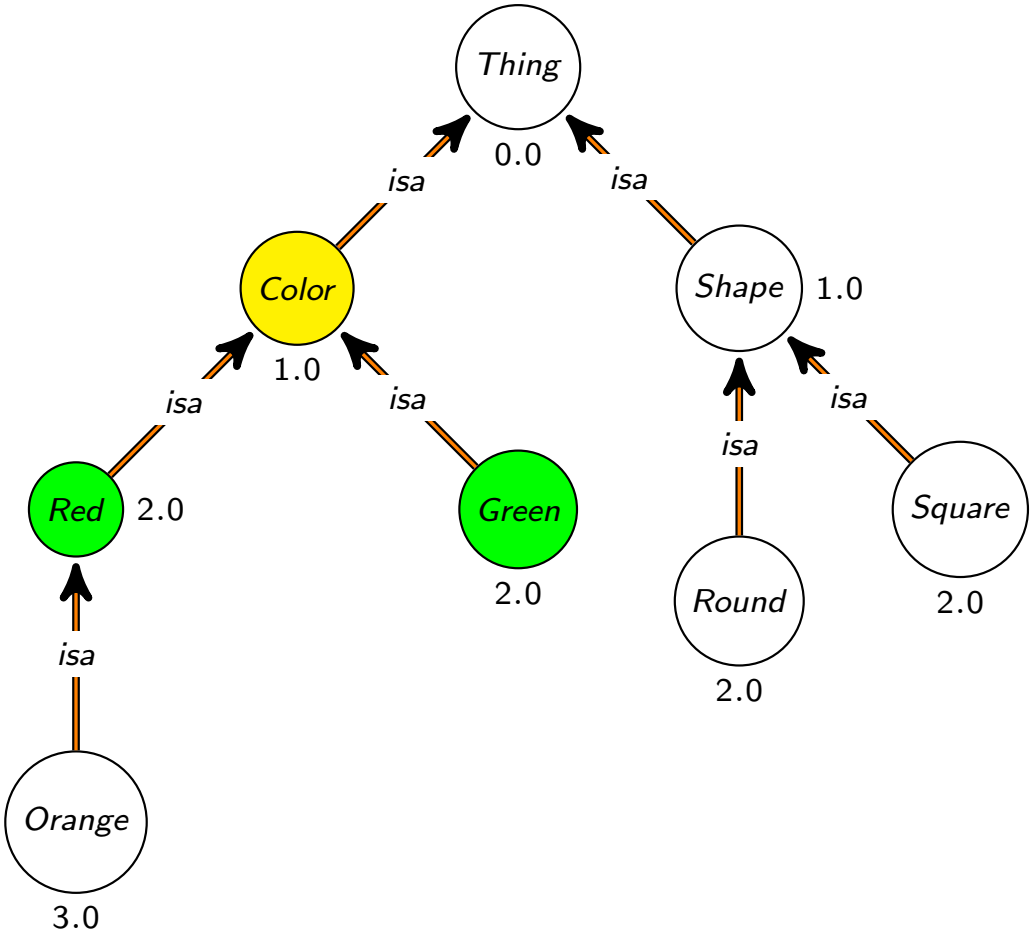

- Lin 1998:
  $sim_{Lin}(x, y) = \frac{2 \cdot IC(MICA(x,y))}{IC(x)+IC(y)}$

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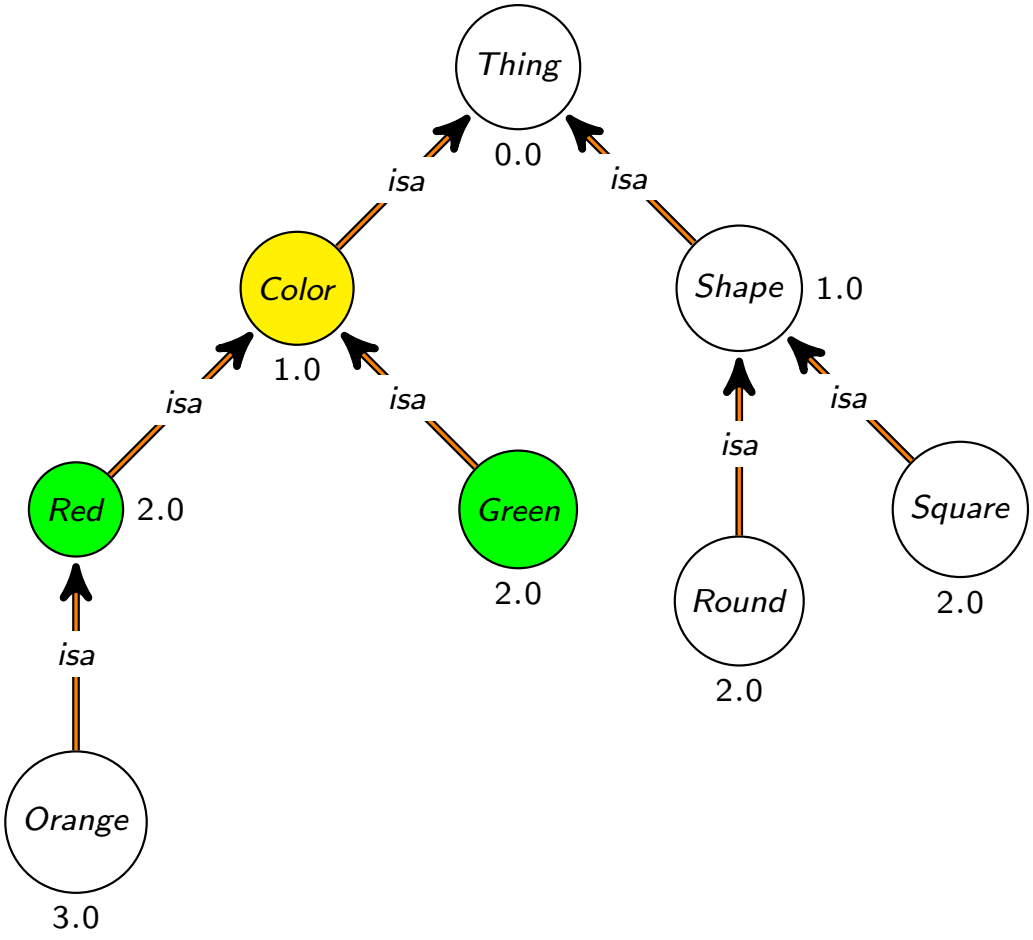

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- $sim_{Lin}(Green, Red) = 0.5$

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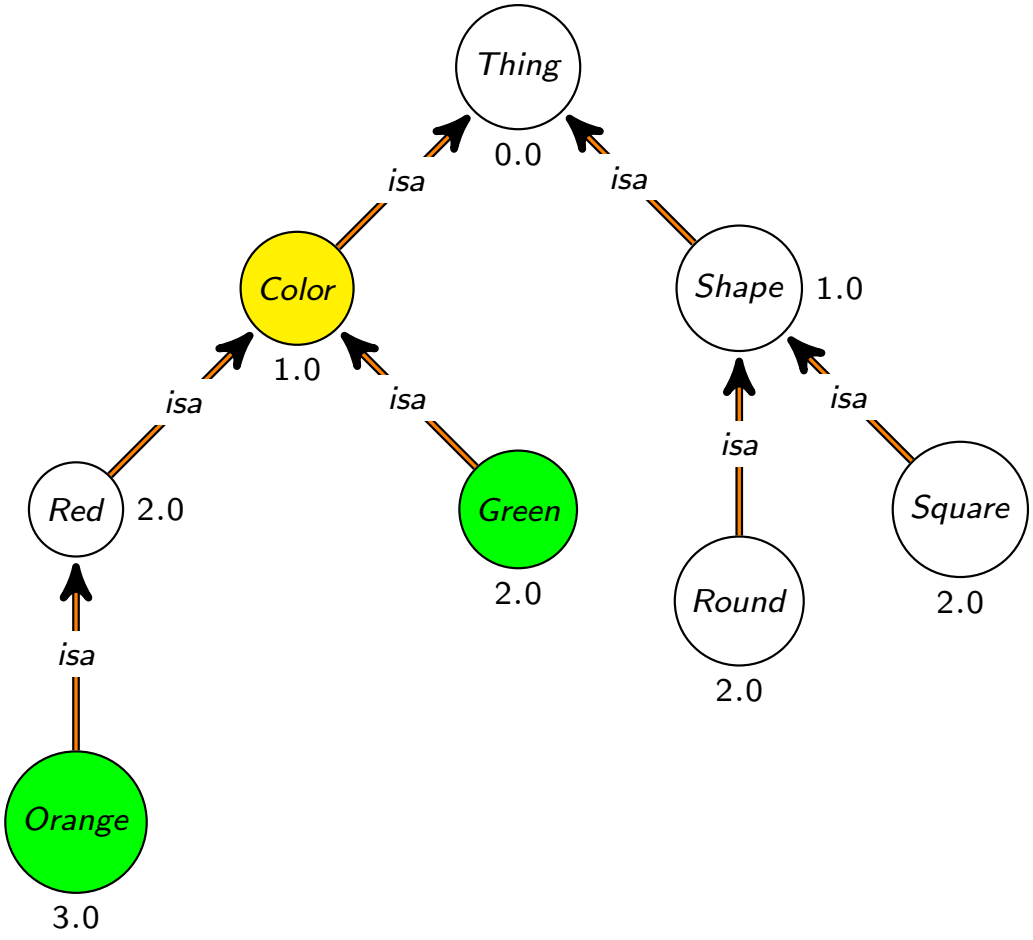

- Lin 1998:
  $sim_{Lin}(x, y) = \frac{2 \cdot IC(MICA(x,y))}{IC(x)+IC(y)}$
- $sim_{Lin}(Green, Orange) = 0.4$

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

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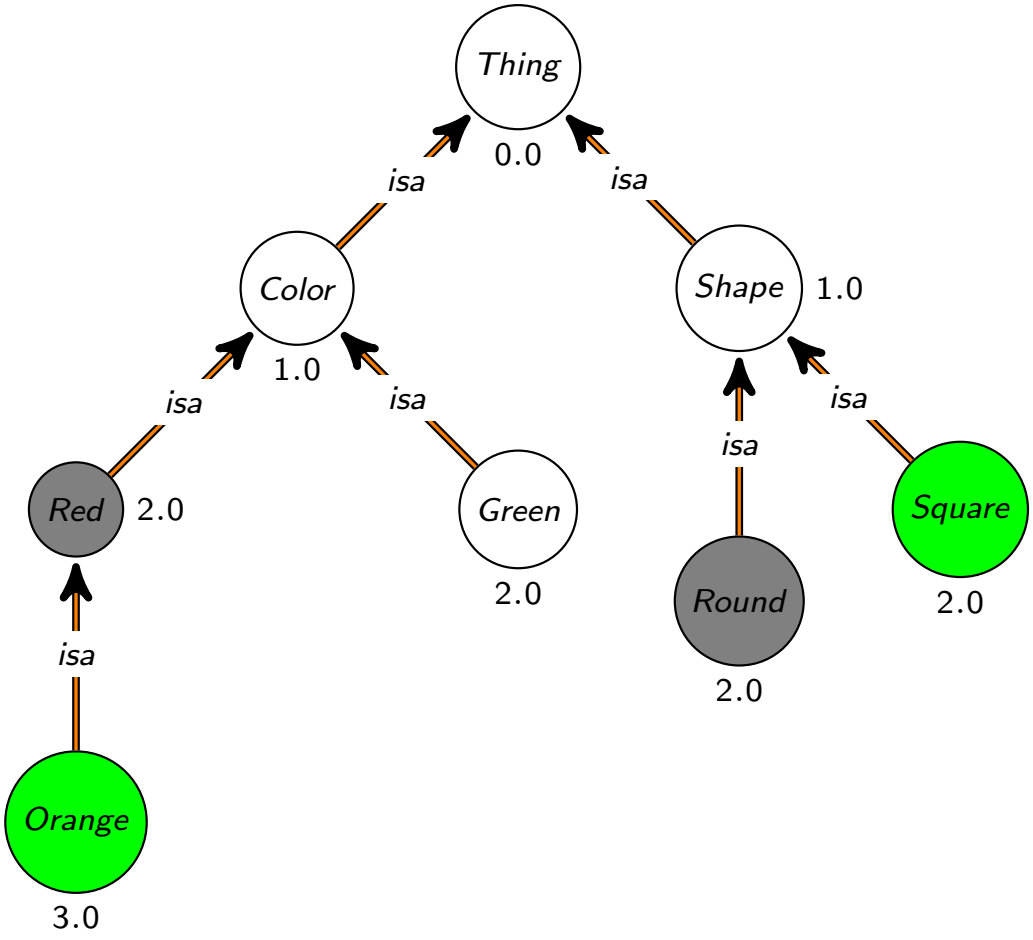

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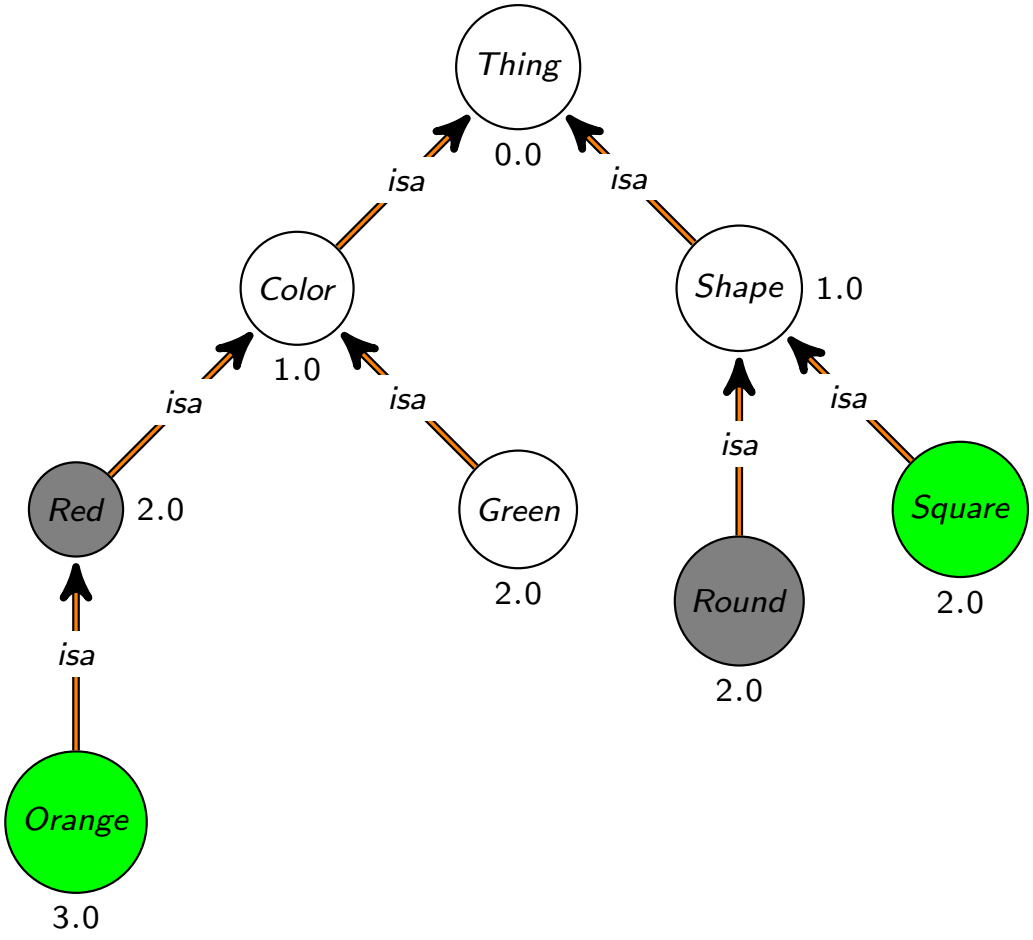

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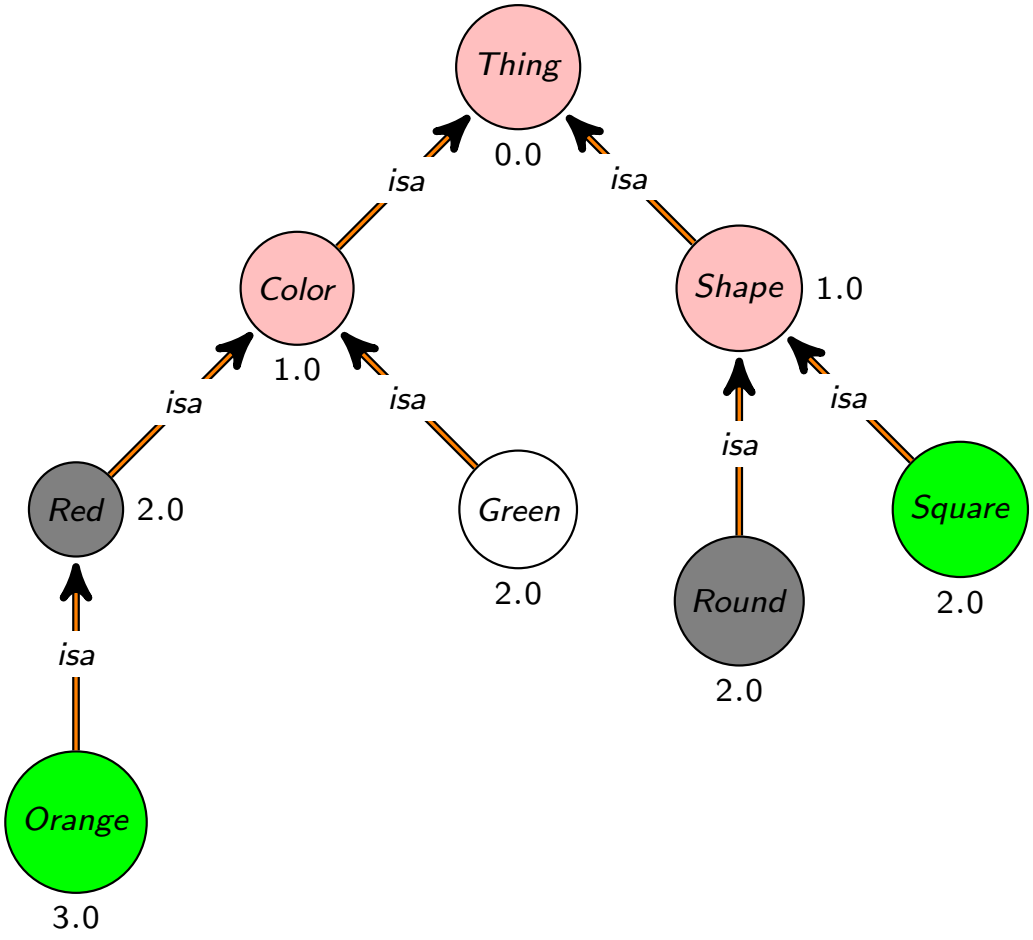

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- $simGIC(so, rr) = \frac{2}{11}$

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)

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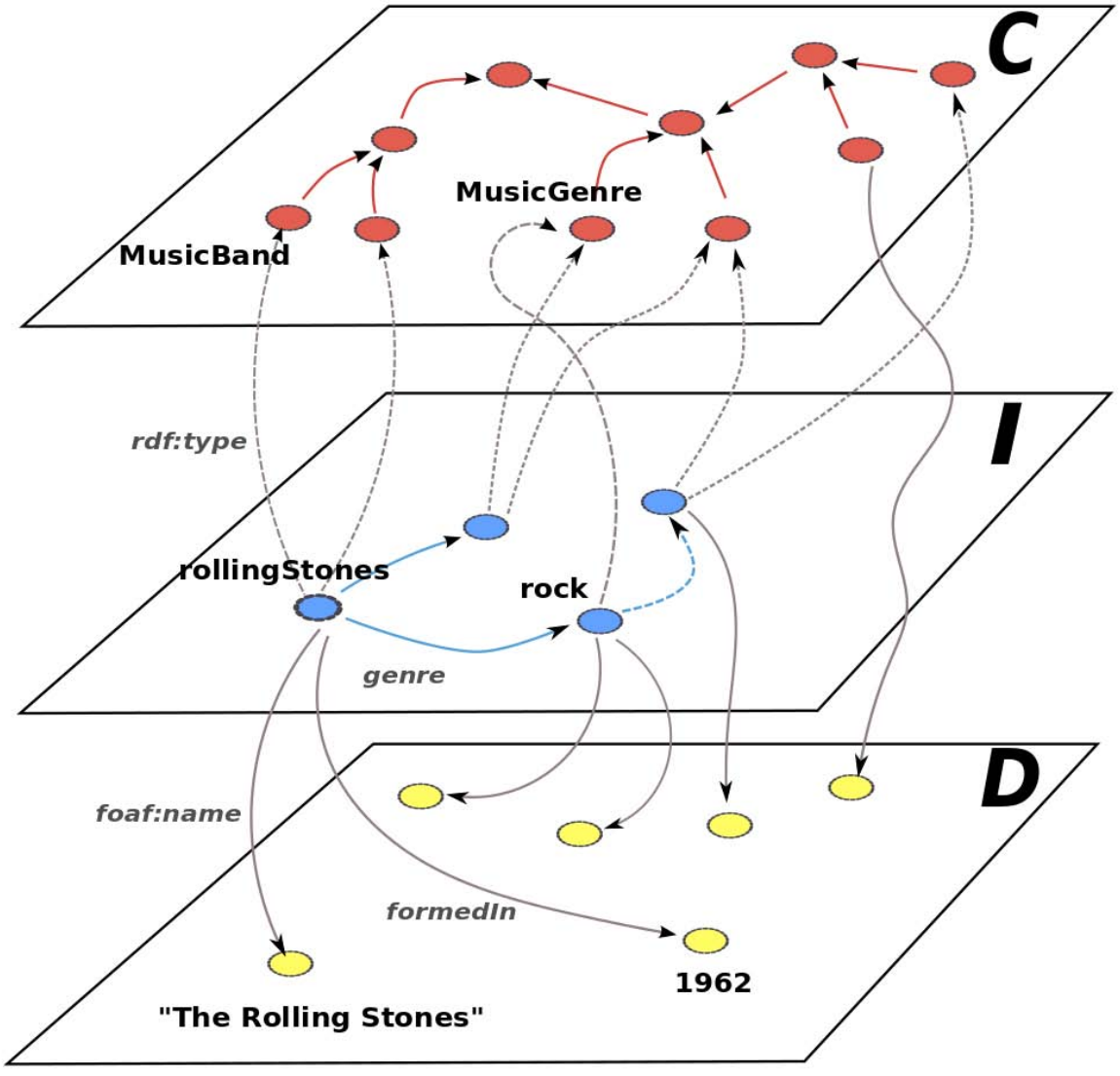

From Harispe et al., Semantic Similarity From Natural Language And Ontology Analysis, 2015.

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

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- feature learning on knowledge graph

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- e.g., iterated, edge-labeled random walk
  - over instances and classes
  - walks form *sentences*
  - sentences form a *corpus*
  - feature learning on corpus through Word2Vec (or factorization of co-occurrence matrix)
  - RDF2Vec: `http://data.dws.informatik.uni-mannheim.de/rdf2vec/`
    - with support for reasoning: `https://github.com/bio-ontology-research-group/walking-rdf-and-owl`

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

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- generates (dense) feature vectors for nodes (classes, instances) and relations

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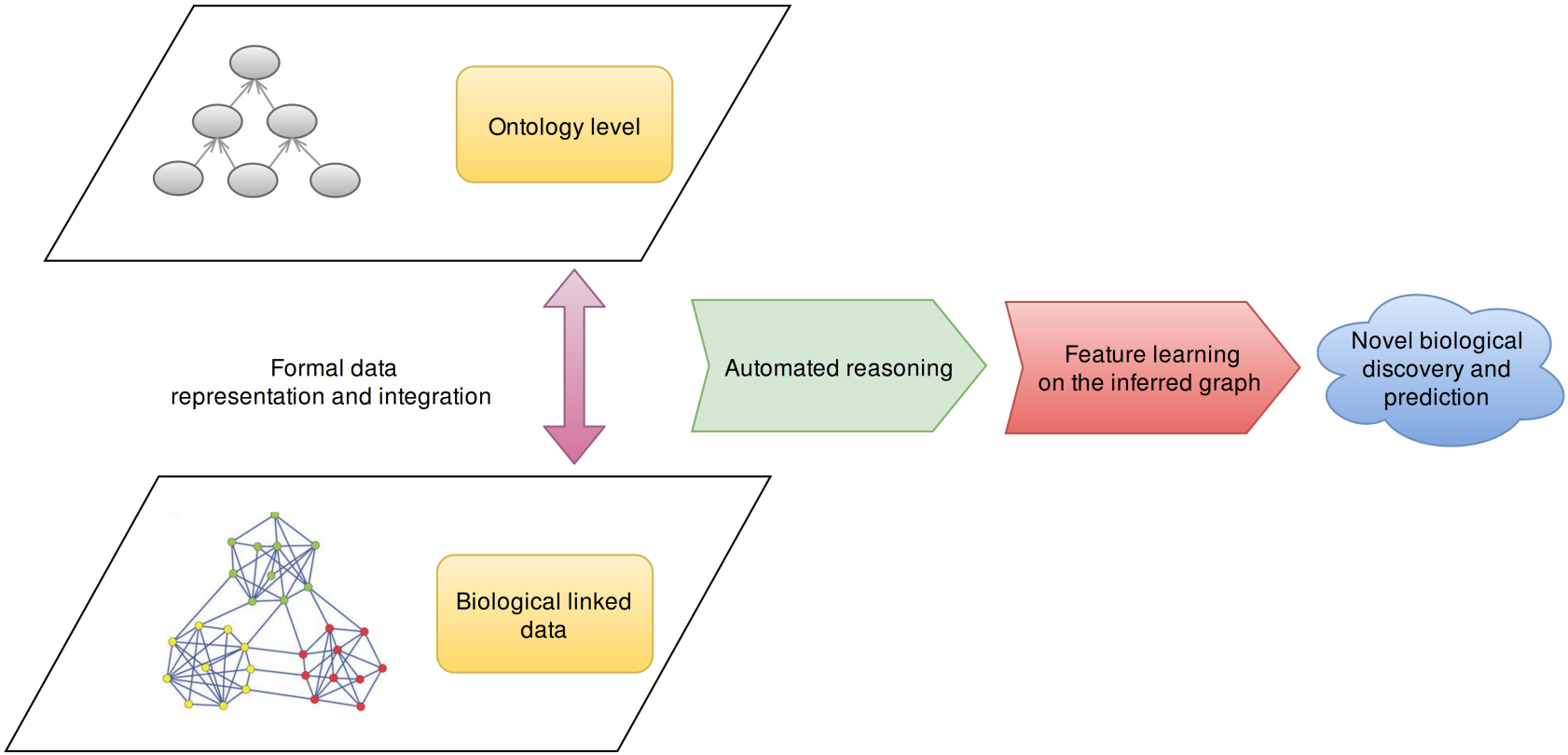

How to measure similarity?

- vector-based similarity measure
- cosine similarity: $sim(X, Y) = \frac{\sum_{i=1}^{n} X_i Y_i}{\sqrt{\sum_{i=1}^{n} X_i^2}\sqrt{\sum_{i=1}^{n} Y_i^2}}$
  - bounded between $[-1, 1]$
- Euclidean distance: $sim(X, Y) = \sqrt{\sum_{i=1}^{n}(X_i - Y_i)^2}$
  - not bounded (and rarely used)

How to measure similarity?

- many graph based semantic similarity measures for comparing two classes
- several set-based measures
  - directly set-based
  - merging pair-wise comparison
- most useful when comparing instances/annotations
- other approaches consider relations between instances:
  - path-based
  - random-walk
- very recent: knowledge graph embeddings
  - and any vector-based similarity measure

How to measure similarity?

Recommended reading:

- recommended, comprehensive overview: Sebastian Harispe et al. Semantic Similarity from Natural Language and Ontology Analysis. Morgan & Claypool Publishers, 2015
- Catia Pesquita et al. Semantic Similarity in Biomedical Ontologies. PLoS CB, 2009.
- Maximilian Nickel et al. A Review of Relational Machine Learning for Knowledge Graphs, Proceedings of the IEEE, 2016.

Applications of semantic similarity

- ontologies are used *almost everywhere* in biology
- many applications of semantic similarity:
  - predicting interacting proteins
  - predict candidate genes
    - using the guilt-by-association principle, or without
  - predict drug targets and indications
  - as features in machine learning models

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?

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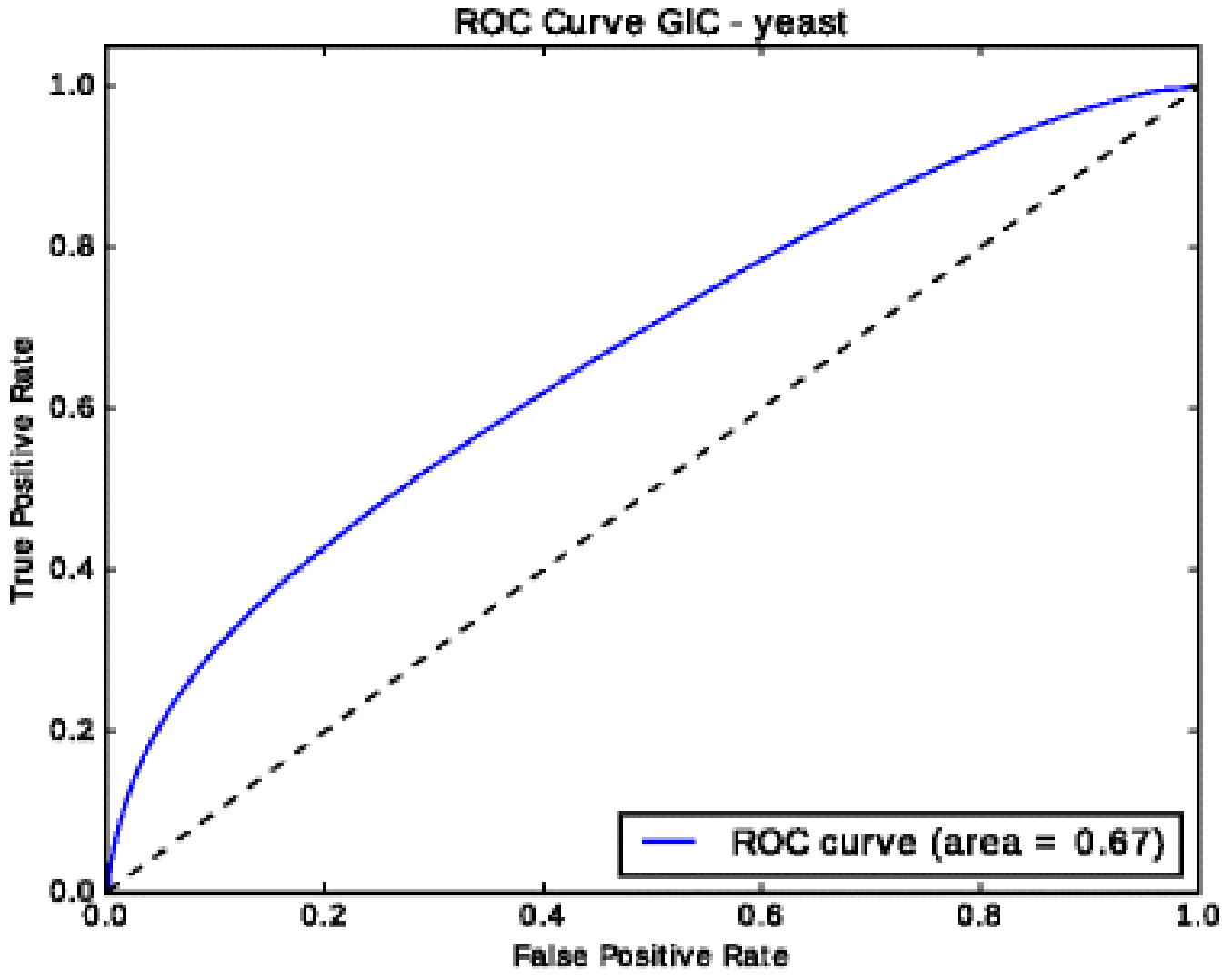
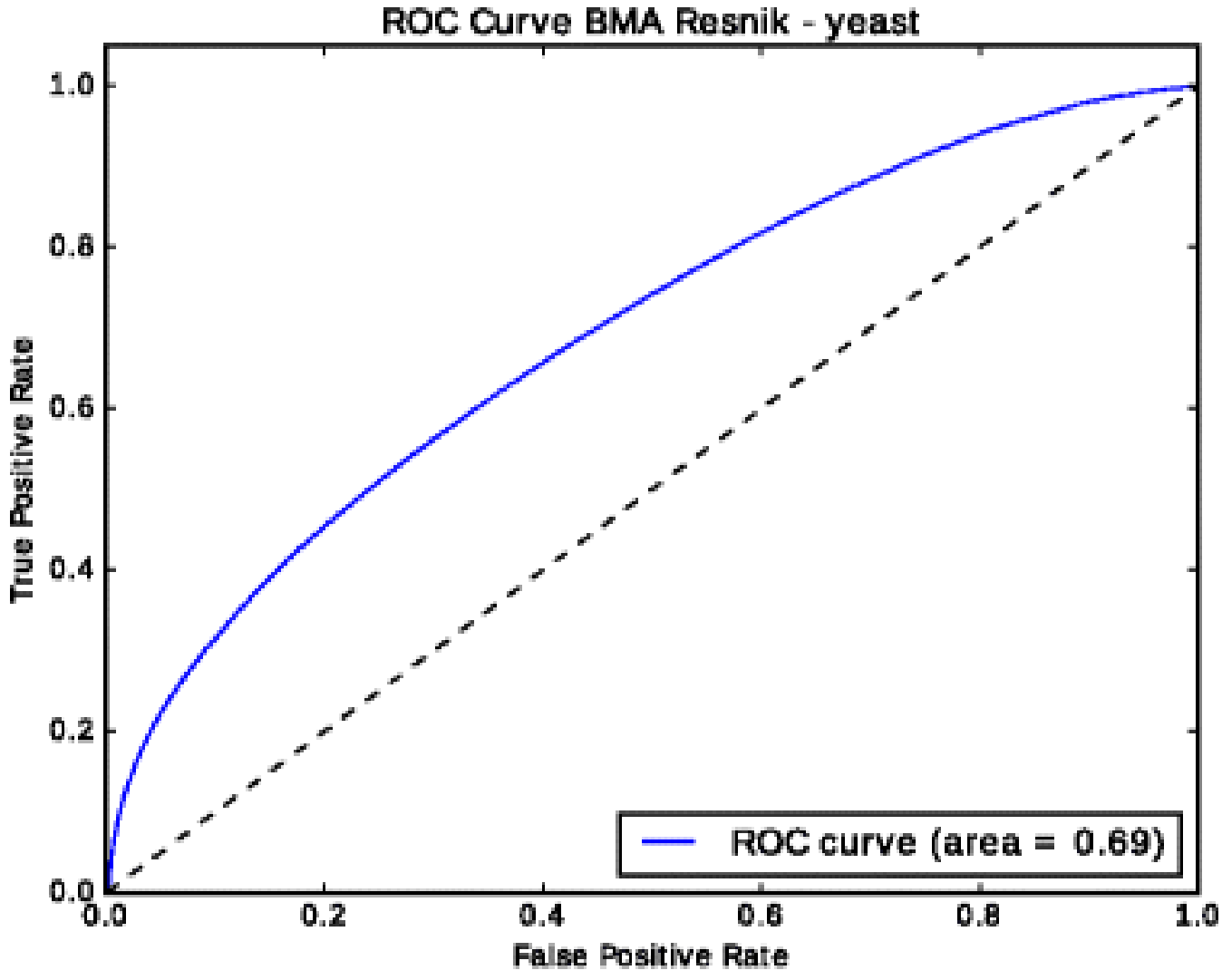
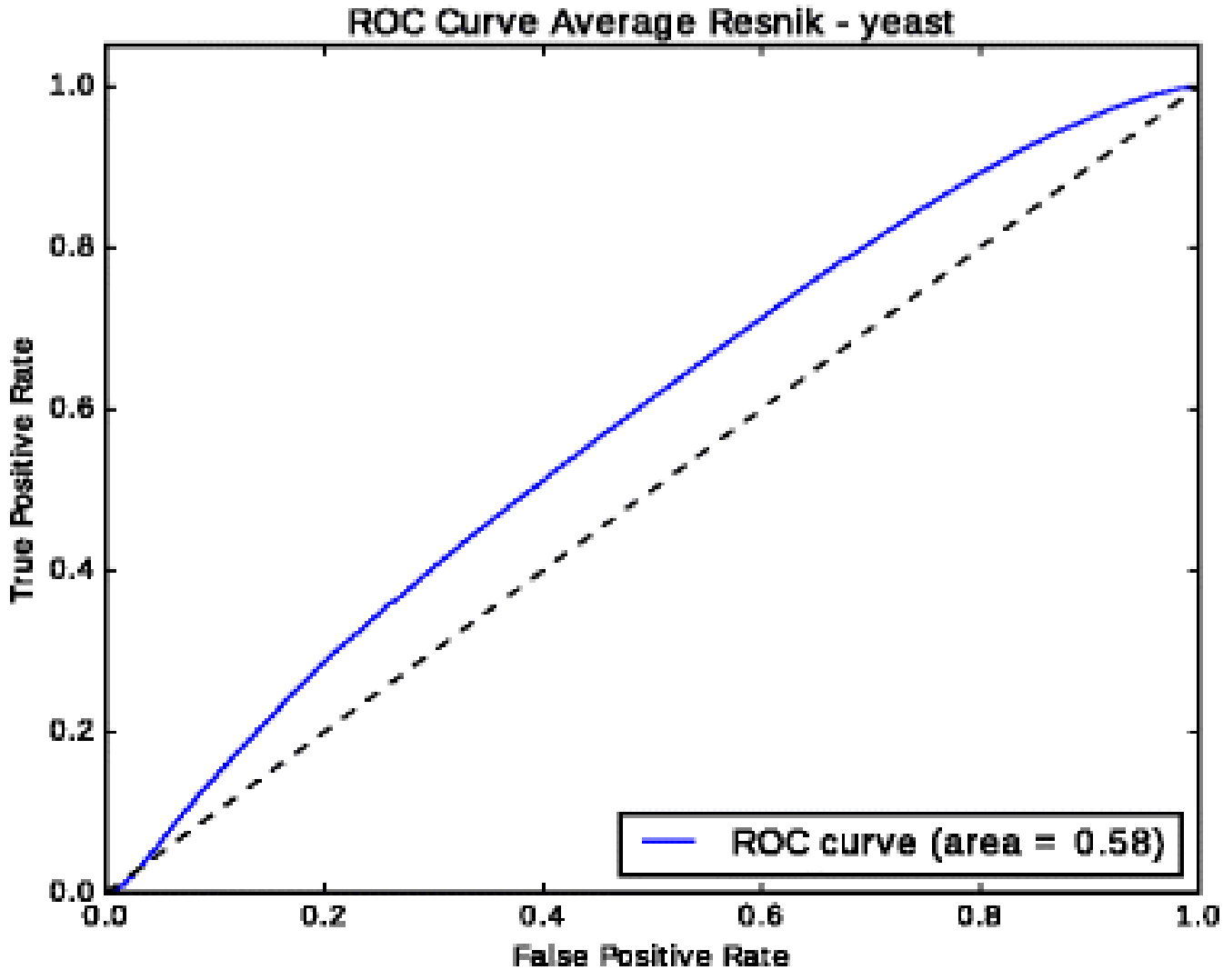


Applications of semantic similarity



Applications of semantic similarity



Applications of semantic similarity

Recommendations:

- use Resnik's information content measure
- use Resnik's similarity
- use Best Match Average
- use the full ontology
- $\Rightarrow$ but there are many exceptions
  - similar location $\Rightarrow$ use location subset of GO
  - developmental phenotypes $\Rightarrow$ use developmental branch of phenotype ontology

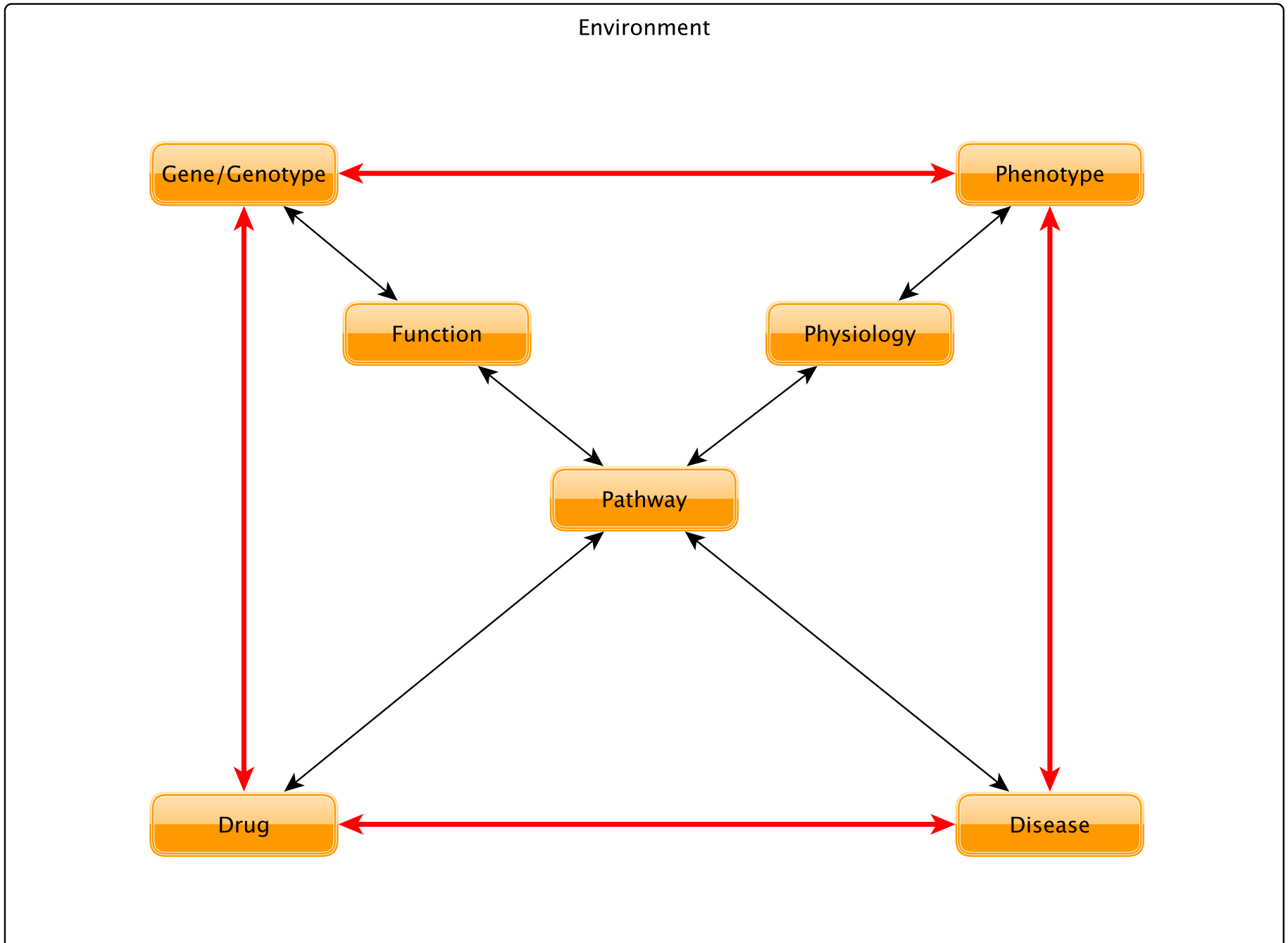
Applications of semantic similarity

- 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

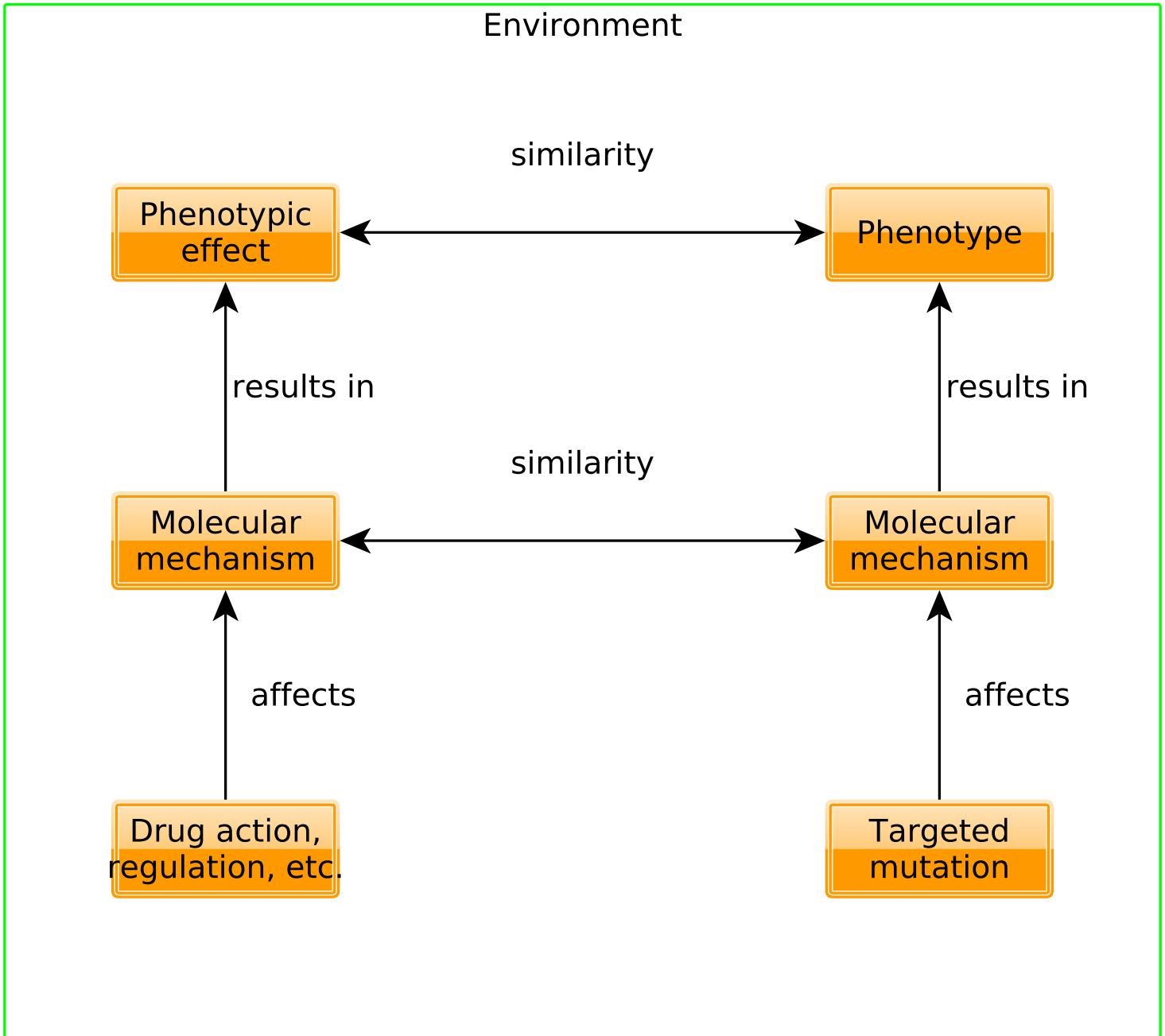
Applications of semantic similarity

- 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



Applications of semantic similarity



Applications of semantic similarity

- Guilt-by-association:
  - x is associated with y
  - z is similar to x
  - therefore: z may 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

- No guilt-by-association:
  - x causes a
  - y has b
  - a similar to b
  - therefore: b is caused by x
- candidate genes (monogenic and polygenic disease):
  - Phenomizer: gene x causes phenotypes a; patient y has symptoms b; a is similar to b; therefore: gene 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

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://oaei.ontologymatching.org/`

Applications of semantic similarity

semantic similarity and text mining:

- find all occurrences of classes of one (or more) ontologies in text
  - using lexical matching or semantic annotations of text
  - TextPresso (`http://www.textpresso.org/`), NCBO Annotator (`https://bioportal.bioontology.org/annotator`), WhatIzIt (`http://www.ebi.ac.uk/webservices/whatizit/info.jsf`)
  - ontology-specific text normalization tools
    - DNorm (diseases), GNorm (gene names), OSCAR (chemicals), ...
- use for database construction (automatic annotation), relation extraction, network construction (co-occurrence network), etc.

Applications of semantic similarity

http://aber-owl.net/aber-owl/diseasephenotypes/

- find phenotypes (signs and symptoms) associated with common diseases
  - no resource available for comparison
- pattern-based mining of literature with Aber-OWL: PubMed
- evaluation (of genetically based disease phenotypes) with experimentally validated disease genes

Applications of semantic similarity

http://aber-owl.net/aber-owl/diseasephenotypes/

Phenotypes for 'bubonic plague'

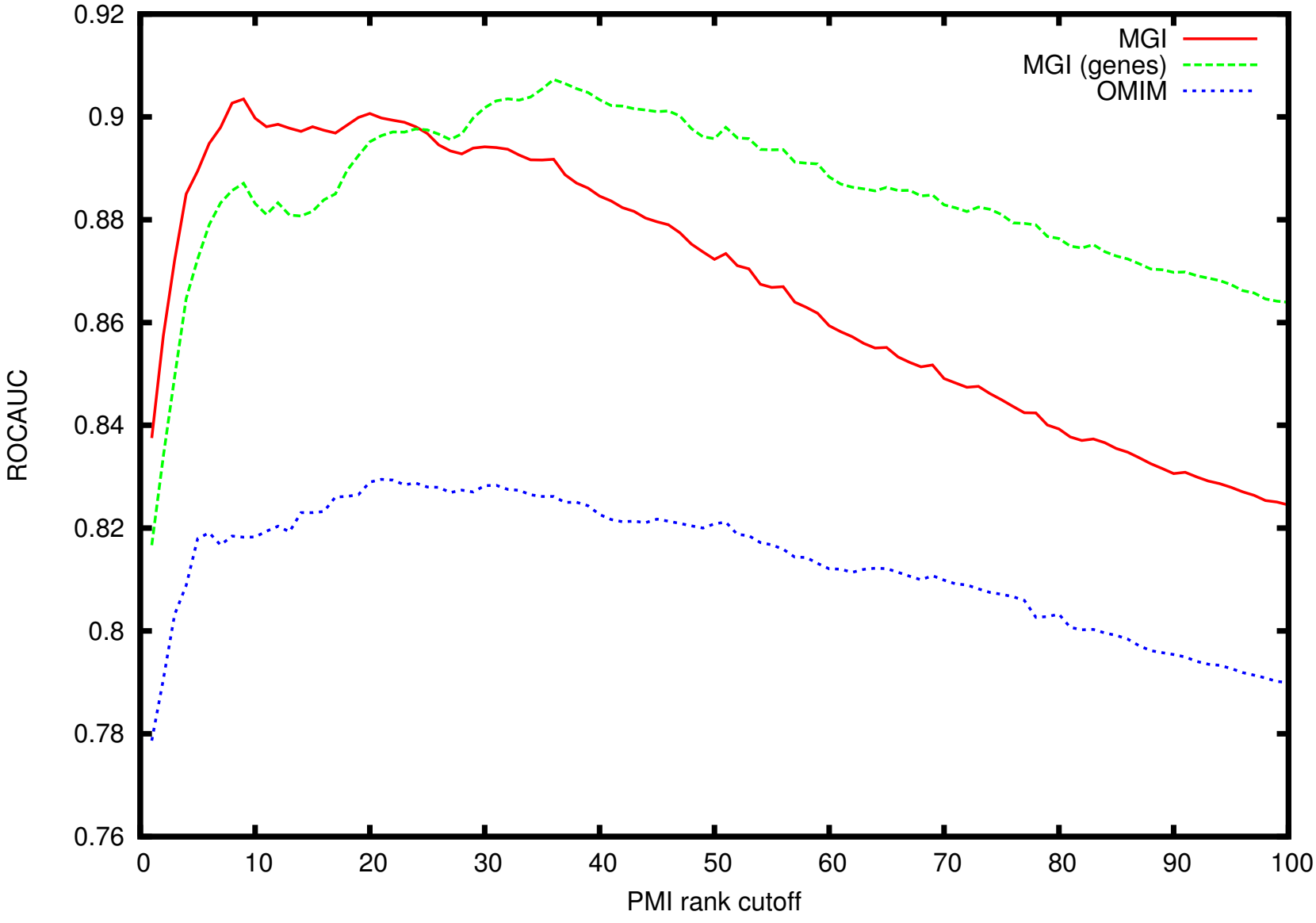
- Mouse models
- Network

Show 100 entries

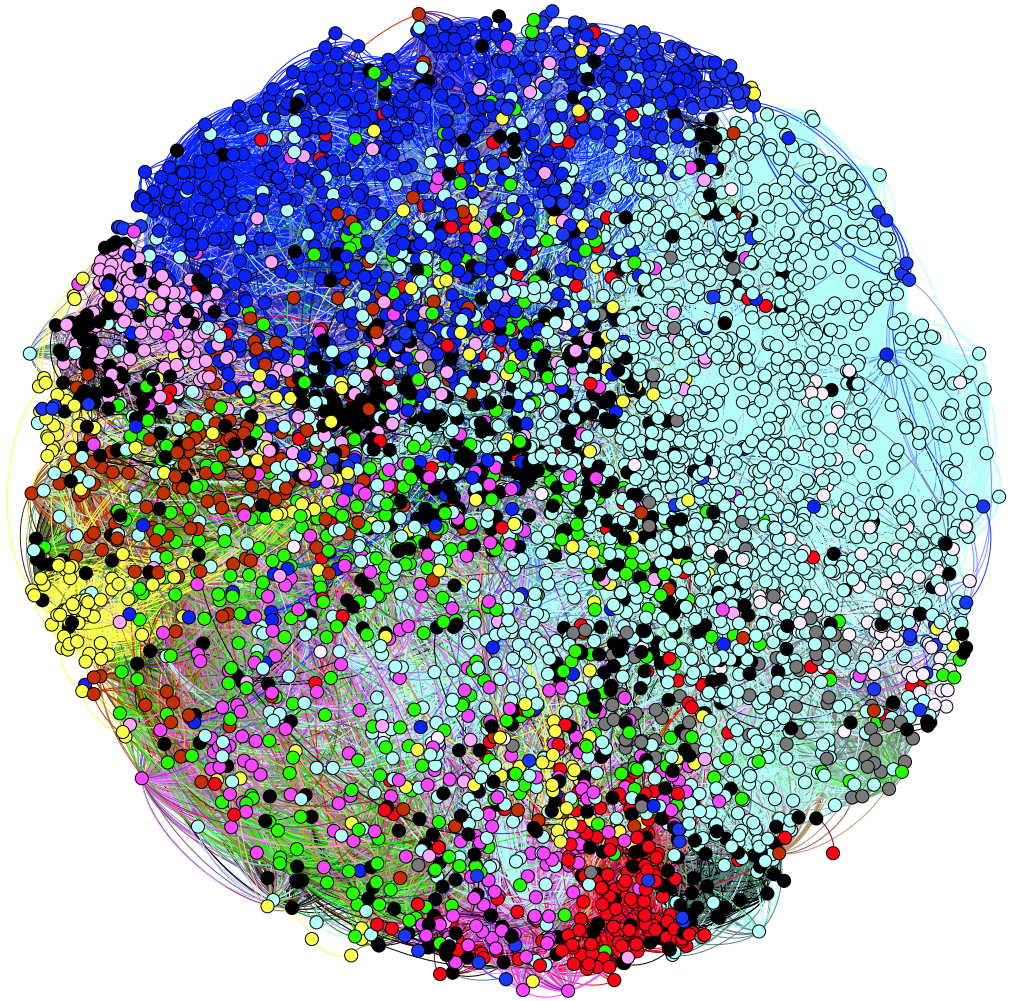
Phenotype	ID	Pointwise Mutual Information
death	MP:0001641	0.22837393990989352
Increased IgA level	HP:0003261	0.21745157416550895
eye lesions	MP:0013146	0.21110960811807034
Lymphadenitis	HP:0002840	0.20726444631165528
lymph node inflammation	MP:0003865	0.2070583004125834
Mediastinal lymphadenopathy	HP:0100721	0.16667331911036262
Abnormality of the lymph nodes	HP:0002733	0.14851361059698084
decreased susceptibility to infection	MP:0002409	0.1442019284611426
Pustule	HP:0200039	0.10968881998477503
tachypnea	MP:0005426	0.10850084511186676
Atrioventricular canal defect	HP:0006695	0.10364210532782134
Tachypnea	HP:0002789	0.10273022349597967
increased pulmonary respiratory rate	MP:0005573	0.09848893732226834

Applications of semantic similarity

http://aber-owl.net/aber-owl/diseasephenotypes/



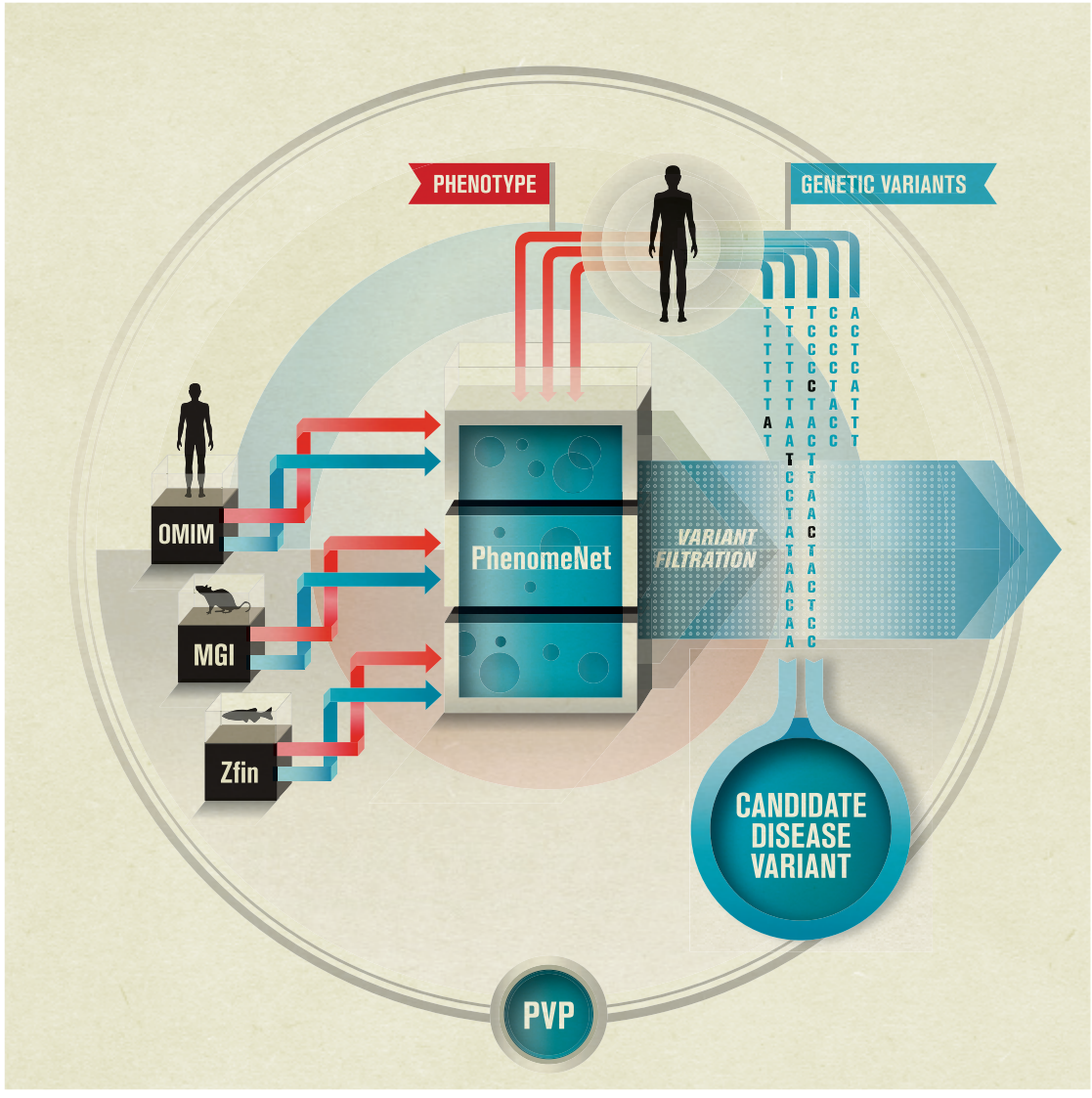
Applications of semantic similarity



Applications of semantic similarity

- semantic similarity can be used as features in machine learning models
  - when annotation space is too large
    - e.g., GO: 50,000 classes
    - replace binary representation
  - to incorporate background knowledge
    - semantic similarity encodes *implicitly* for ontology structure and axioms
    - encodes for *specificity* of classes
  - negative: reduce all annotations to single value
    - leads to loss of information
    - but is easier to use by many machine learning methods

Applications of semantic similarity



PVP Evaluation

- Synthetic data
  - 8,746 ClinVar pathogenic variants appended to WEX from 1000 Genome Project
  - 11,251 ClinVar pathogenic variants appended to WGS from 1000 Genome Project
- Real patient data
  - **19 exomes** from **UK10K_RARE_THYROID** study sample dataset.
  - 43 exomes from Addenbrooke's Hospital at Cambridge, UK with diseases of diverse phenotypes (e.g Bardett-Biedl Syndrome)
  - 1 patient's exome (Glutaric aciduria)
  - 5 genomes from Personal Genome Project

Synthetic WEX patients

	Top Hit	Top 10	ROC AUC
GWAVA	1.41%	6.32%	0.711
DANN	6.06%	26.69%	0.928
eXtasy	14.85%	42.99%	0.782
CADD	15.15%	32.05%	0.946
Exomiser	24.65%	58.56%	0.89
Phevor	28.25%	64.70%	0.912
PVP	**78.80%**	**89.50%**	**0.978**

Synthetic WGS patients

	Top Hit	Top 10	ROC AUC
GWAVA	0.46%	0.70%	0.949
DANN	4.95%	18.99%	0.99
CADD	5.76%	24.30%	0.962
Genomiser	31.35%	77.98%	0.948
PVP	**76.47%**	**88.61%**	**0.995**

Summary

- many semantic similarity measures
  - graph-based
  - feature-based
- useful for similarity-based prediction
  - similar entities $\Rightarrow$ guilt-by-association
  - different entities
- combine with data and text mining
- features in machine learning methods

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 follow in our demonstration
- Beaker Notebook
  - like iPython Notebook, or Jupyter, with lots of kernels
  - Python, Groovy, Javascript, ...
- `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
- you can build on this and extend for your own research!