Universität Rostock Traditio et Innovatio

SEMS simulation experiment management system

Examples: Ontologies in bio-medical and neuroscience research

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In fact, bio-ontologies are beginning to proliferate in step with accruing biological data. The myriad of ontologies being created enables researchers not only to solve some of the problems in handling the data explosion but also introduces new challenges. One of the key difficulties in realizing the full potential of ontologies in biomedical research is the isolation of various communities involved: some workers spend their career developing ontologies and ontology-related tools, while few researchers (biologists and physicians) know how ontologies can accelerate their research.

(Quotation: **Biomedical ontologies: a functional perspective** by Rubin et al., 2008)

Outline

Ontologies ...

- in Life Sciences and Medicine
- in Computational Biology modeling
- in Neuroscience

Ontologies for Life Science data

OBI
Gene Ontology
ChEBI

http://vimeo.com/5344028

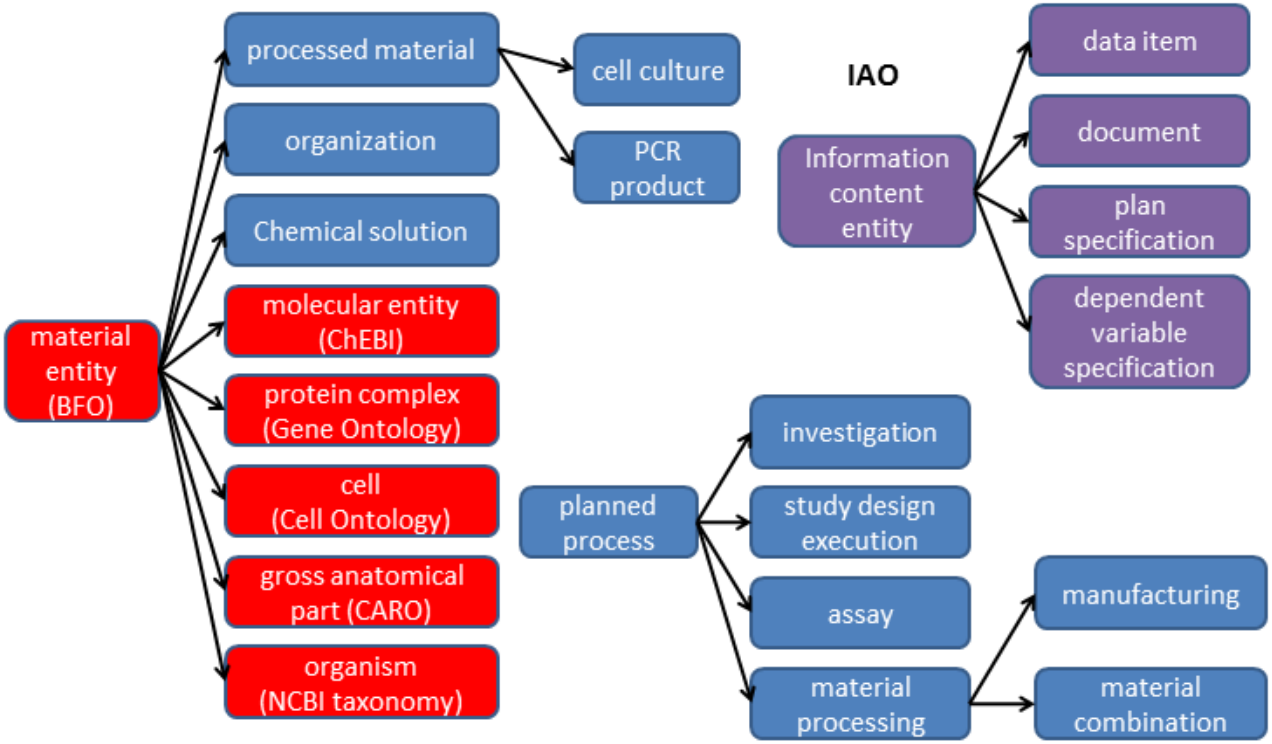
Ontology of Biomedical Investigations (OBI)

Facts

- Goal: consistent annotation of biomedical investigation
- International & collaborative project
- Integrated ontology for the description design of an investigation, the protocols and instrumentation used, the material used, the data generated and the type analysis performed
- Including universal terms and domain-specific terms
- Built on the Basic Formal Ontology (BFO)

Ontology of Biomedical Investigations (OBI)

Overview: High-level concepts



(Source: **Applications of Bio-Ontologies in Large-Scale Data-Driven Science: A Practical Introduction** by Janna Hastings, ECCB 2012)

Gene Ontology (GO)

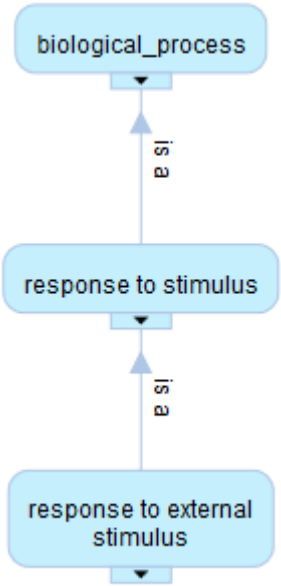
Facts

- Goal: collaborative effort to generate consistent descriptions of gene products in different databases
- Original resources: FlyBase, SGF, MGD
- three structured CVs of gene products in terms of their
  - Associated biological function (biological process)
  - Cellular component
  - Molecular function

Gene Ontology (GO)

Branch: Biological process

- series of events accomplished by one or more ordered assemblies of molecular functions
- Example: response to external stimulus



(Figure: response to external stimulus visualization in Bioportal, http://bioportal.bioontology.org/)

Gene Ontology (GO)

Branch: Biological process

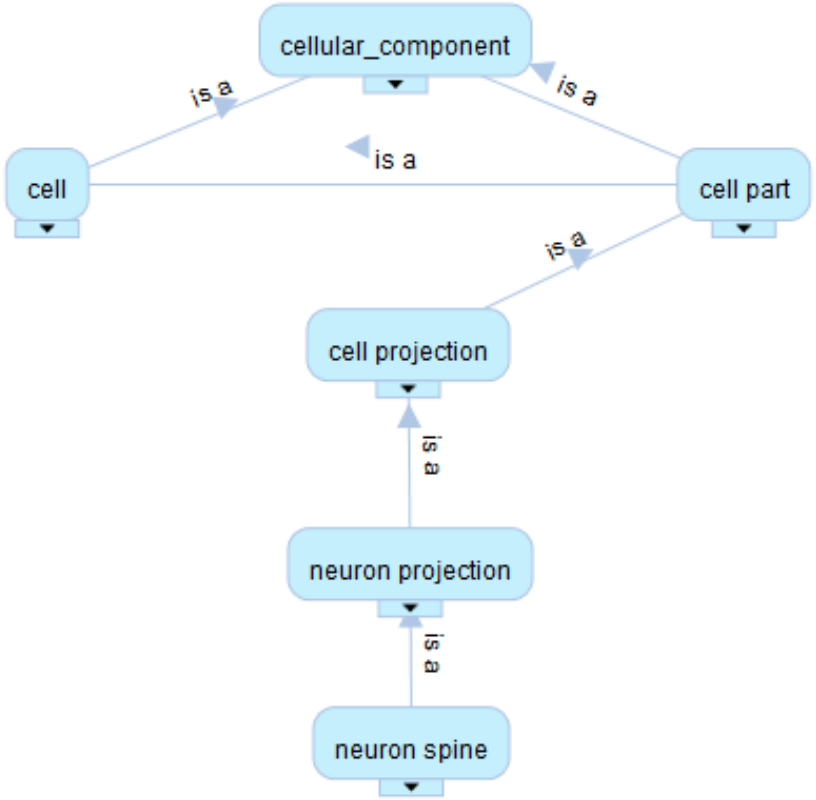
- series of events accomplished by one or more ordered assemblies of molecular functions
- Example: response to external stimulus

Preferred Name	response to external stimulus
Synonyms	response to environmental stimulus
Definitions	Any process that results in a change in state or activity of a cell or an organism (in terms of movement, secretion, enzyme production, gene expression, etc.) as a result of an external stimulus.
ID	GO:0009605
Full Id	http://purl.obolibrary.org/obo/GO_0009605
Comment	Note that this term is in the subset of terms that should not be used for direct gene product annotation. Annotations to this term will be removed during annotation QC.
EXACT SYNONYM	response to environmental stimulus
subset	goslim_plant gosubset_prok high_level_annotation_qc
xref_definition	GOC:hb
is_a	response to stimulus

Gene Ontology (GO)

Branch: Cellular component

- A component of a cell that is part of some larger object
- E.g., neuron spine as part of a cell



(Figure: Neuron spine visualization in Bioportal, http://bioportal.bioontology.org/)

Gene Ontology (GO)

Branch: Cellular component

- E.g., neuron spine as part of a cell

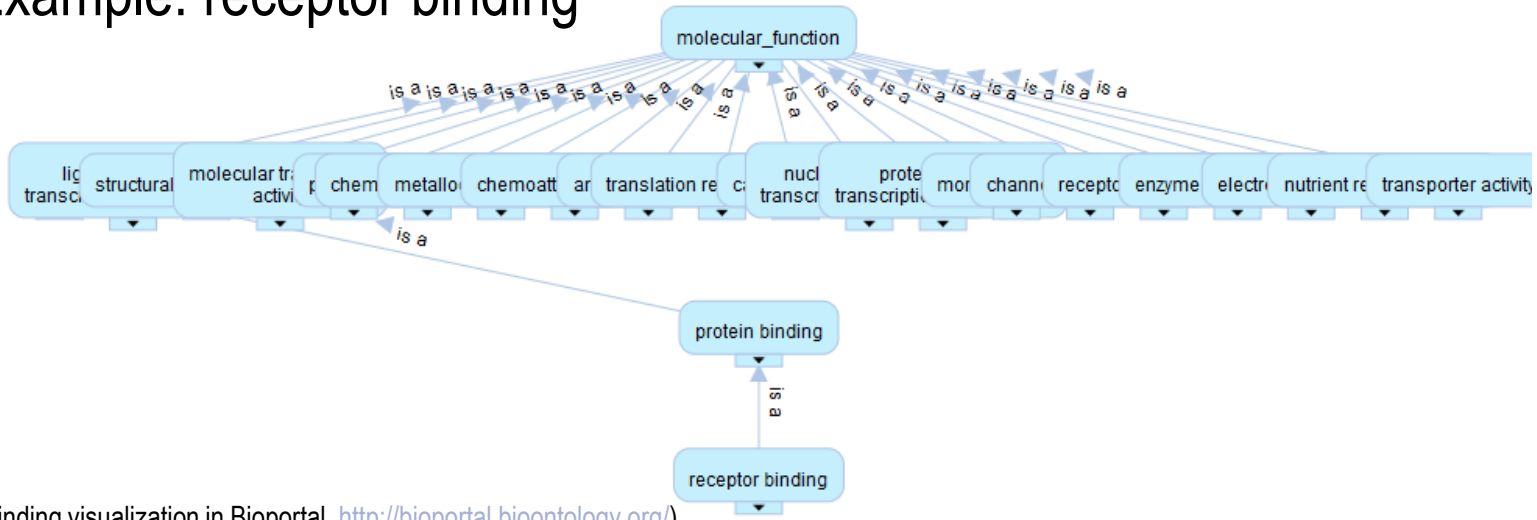
Preferred Name	neuron spine
Definitions	A small membranous protrusion, often ending in a bulbous head and attached to the neuron by a narrow stalk or neck.
ID	GO:0044309
Full Id	http://purl.obolibrary.org/obo/GO_0044309
created_by	jane
creation_date	2010-02-05T04:25:10Z
xref	NIF_Subcellular:sao1145756102
xref_definition	NIF_Subcellular:sao1145756102 ISBN:0198504888
is_a	neuron projection

(Figure: GO definition for Neuron spine in Bioportal, http://bioportal.bioontology.org/)

Gene Ontology (GO)

Branch: Molecular function

- Activity that occurs at the molecular level
- Including catalytic or binding activities
- Example: receptor binding



(Figure: receptor binding visualization in Bioportal, http://bioportal.bioontology.org/)

Gene Ontology (GO)

Branch: Molecular function

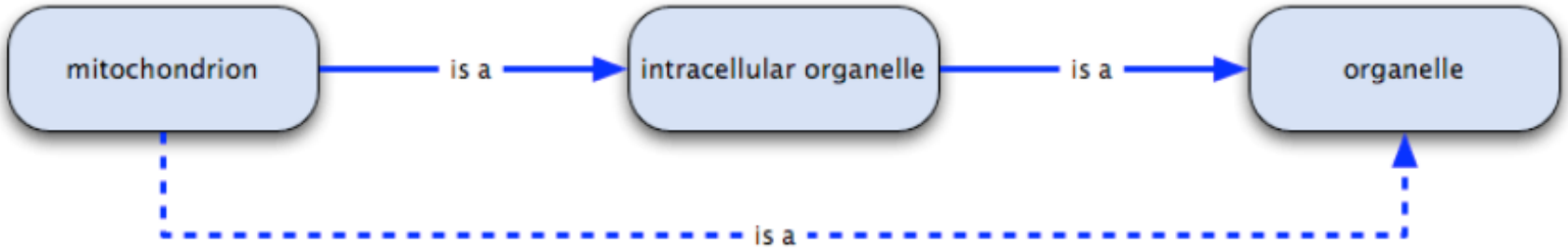
- Example: receptor binding

Preferred Name	receptor binding
Definitions	Interacting selectively and non-covalently with one or more specific sites on a receptor molecule, a macromolecule that undergoes combination with a hormone, neurotransmitter, drug or intracellular messenger to initiate a change in cell function.
ID	GO:0005102
Full Id	http://purl.obolibrary.org/obo/GO_0005102
NARROW SYNONYM	receptor ligand
RELATED SYNONYM	receptor-associated protein activity
subset	goslim_plant gosubset_prok
xref	Wikipedia:Ligand_(biochemistry)
xref_definition	ISBN:0198506732 "Oxford Dictionary of Biochemistry and Molecular Biology" GOC:bf GOC:ceb
is_a	protein binding

Gene Ontology (GO)

Relations: is_a

- describes that a class is a subtype of another
- "Is an instance of the child process an instance of the entire parent process?"
- Transitive relation

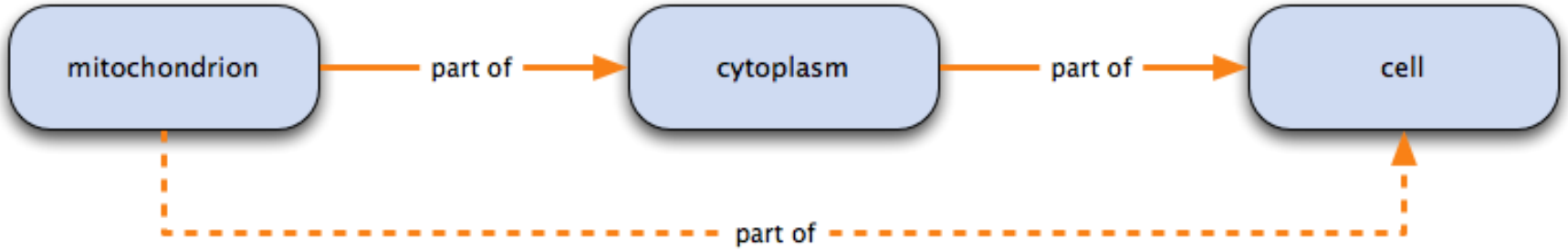


(Source: http://www.geneontology.org/GO.ontology.relations.shtml)

Gene Ontology (GO)

Relations: part_of

- Part-whole relationship (A part_of B)
- Wherever A exists, it is as part of B
- Given the occurrence of B, we cannot say for certain that A exists
- Transitive relation

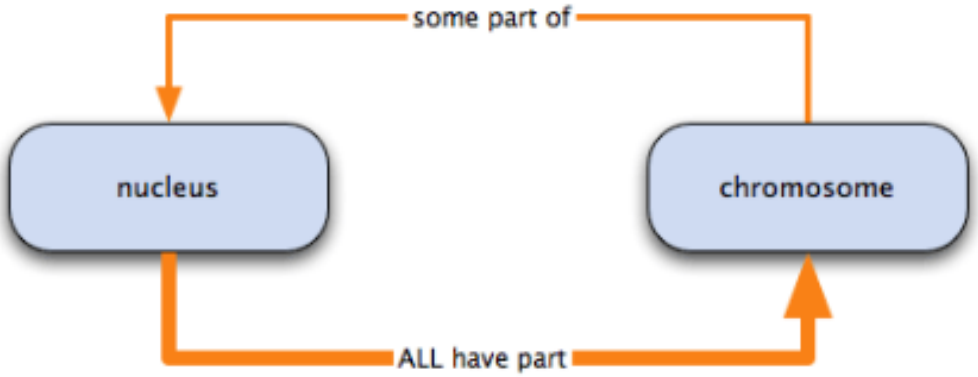


(Source: http://www.geneontology.org/GO.ontology.relations.shtml)

Gene Ontology (GO)

Relations: has_part

- Part-whole relationship from parent perspective (A has_part B)
- A always has B as a part
- If A exists, B will always exist; however, if B exists, we cannot say for certain that A exists.
- Transitive relation

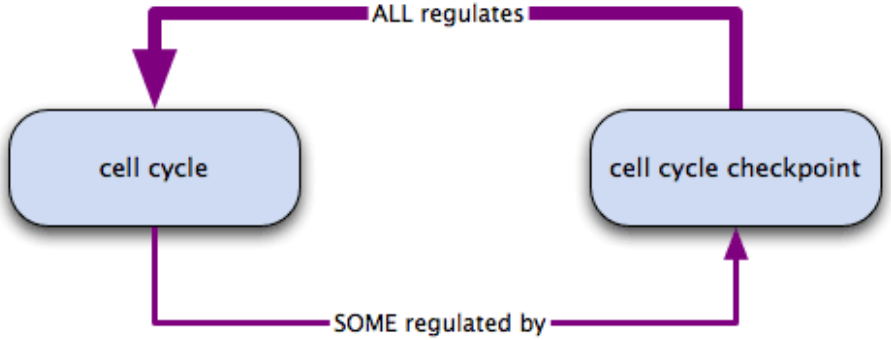


(Source: http://www.geneontology.org/GO.ontology.relations.shtml)

Gene Ontology (GO)

Specific relations: regulates

- One process directly affects the manifestation of another process or quality, i.e. the former regulates the latter
- Positively/negatively regulates



(Source: http://www.geneontology.org/GO.ontology.relations.shtml)

Gene Ontology (GO)

Mappings

- GO terms are regularly mapped on other classifications
  - Enzyme Commission (EC) enzyme numbers
  - Interpro
  - KEGG
  - Reactome events
  - Uniprot KB

Gene Ontology (GO)

Use cases: GO annotations

= assignments of ontology terms to genes, proteins ...

- Characterize genomic regions associated with some function
- Infer protein complexes from interaction networks
- Identify up- and down-regulated genes
- Categorize genes by certain characteristics
- Assign appropriate GO functional annotations to proteins of study
- Identify associated pathways
- Annotate computational models

(Source: **Searching for Uses and Users in Gene Ontology Research** by MacMullen, 2008)

Chemical Entities of Biological Interest (ChEBI)

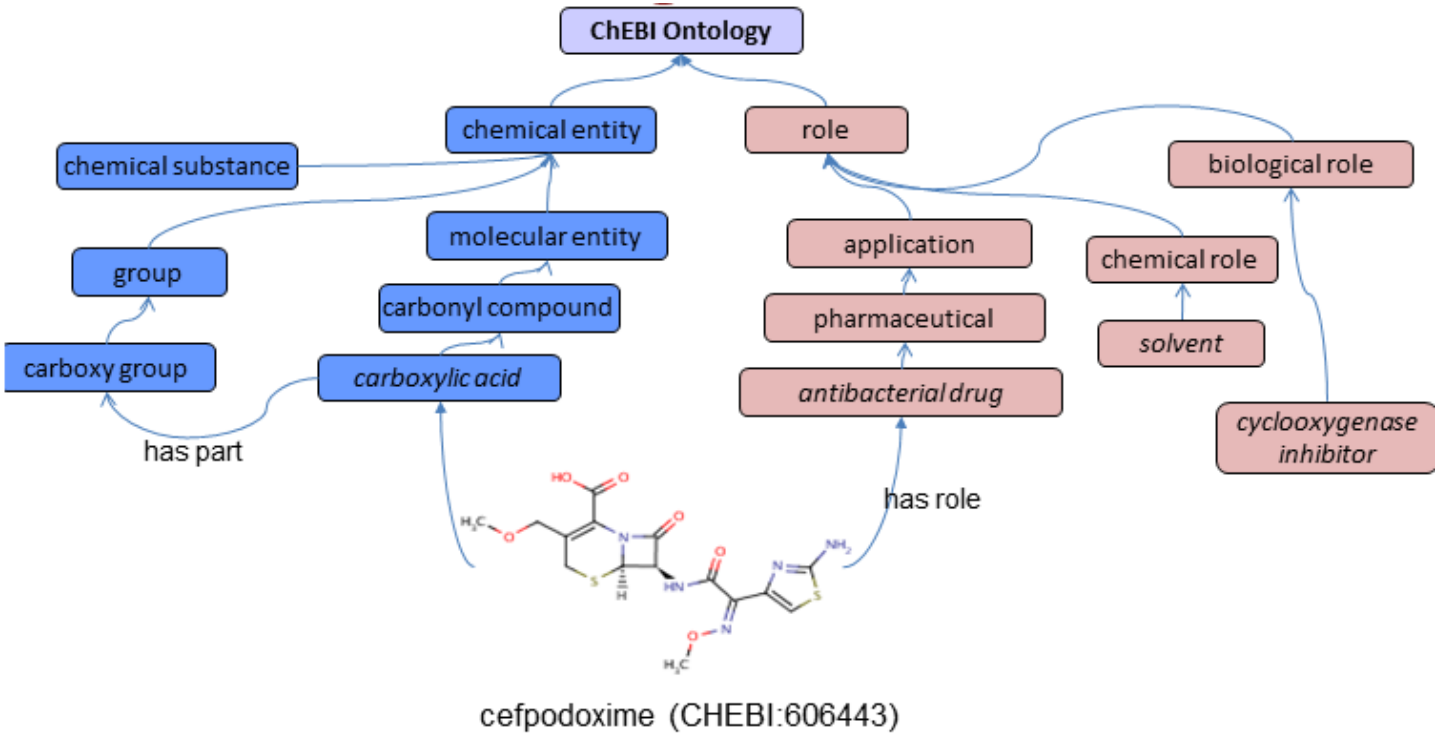
Facts

- Goal: standardized description of molecular entities/groups (parts of molecular entities)
- freely available dictionary of molecular entities focused on 'small' chemical entities
- Focus: manual annotation, non-redundancy, and provision of a chemical ontology
- Orthogonal to other ontologies within the OBO foundry, aligned to closely related ontologies, eg. Gene Ontology
- Examples: penicillin (ChEBI:17334)3 , caffeine (ChEBI:27732), cocaine (ChEBI:27958) and adrenaline (ChEBI:33568)

(Source: ftp://ftp.ebi.ac.uk/pub/.../chebi/tutorial/chebi_tutorial_block1.d)

Chemical Entities of Biological Interest (ChEBI)

Example



(Figure: Courtesy Janna Hasting, ECCB 2012)

Chemical Entities of Biological Interest (ChEBI)

Relations

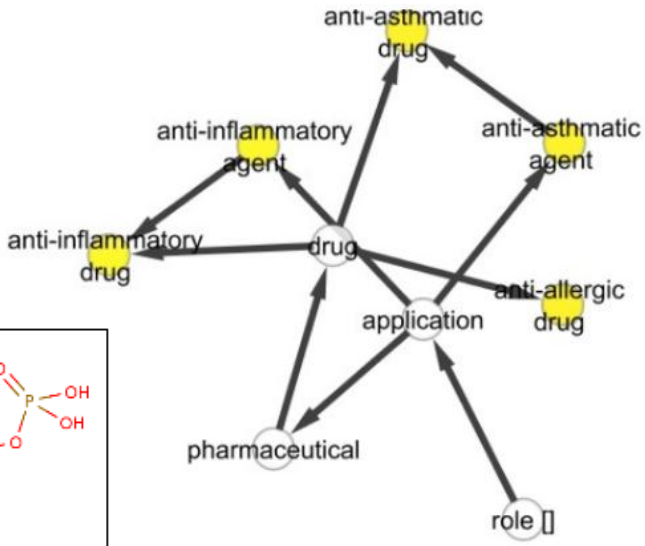
- Is_a: used for most chemical properties: Entity A' is a subtype of 'Entity B
- Has_part: relationship between part and whole
- Has_role: biological properties: particular behavior which an entity may exhibit, either naturally or by human application
- Other (rarely used) relations
  - Is_enantiomer_of: two entities are mirror images of and non-superposable upon each other
  - Is_tautomer_of:
  - Is_conjugate_acid_of / is_conjugate_base_of: connect acids with conjugate bases
  - ...

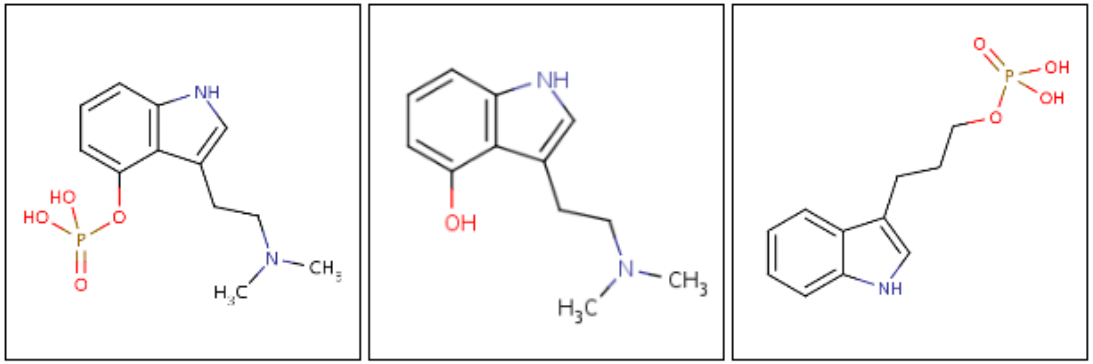
(Source: ftp://ftp.ebi.ac.uk/pub/.../chebi/tutorial/chebi_tutorial_block1.d)

Chemical Entities of Biological Interest (ChEBI)

Use cases

- Explorative drug discovery (enrichment analysis)
- Semantic similarities of chemical structures





(1) psilocybin (CHEBI:8614), (2) psilocin (CHEBI:8613), and (3) 3-(indol-3-yl)propyl phosphate (CHEBI:28162)

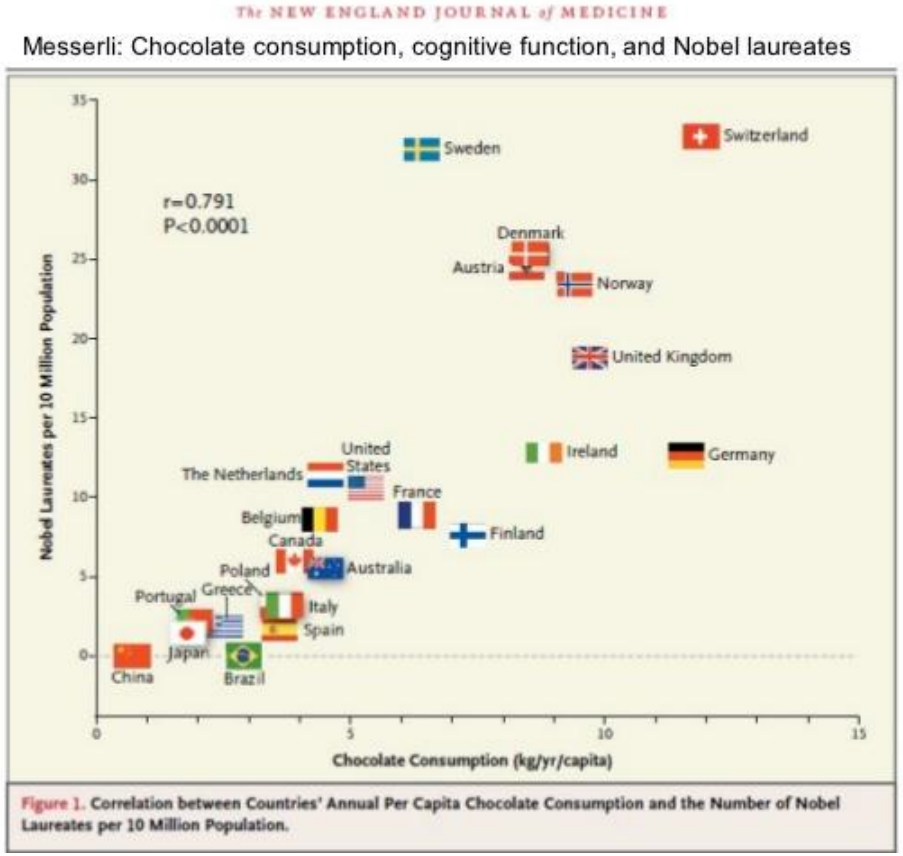
76% 75%

(Sources:http://metabolights.blogspot.de/2012/08/applications-using-chebi-semantic.html, **Exploring the biological impact of chemicals with the ChEBI Ontology** by Janna Hastings, 2012)

Chemical Entities of Biological Interest (ChEBI)

Use cases

- Exploring the chemical impact on metabolism, e.g. Metabolight project



(Source:**Exploring the biological impact of chemicals with the ChEBI Ontology** by Janna Hastings, 2012)

Bioclinical ontologies

Pros and cons

- Ontologies for powerful and interoperable information systems in healthcare
- support the need of the healthcare process to transmit, re-use and share patient data
- provide semantic-based criteria to support different statistical aggregations for different purposes
- support the integration of knowledge and data

- But: Also criticism on the impact that ontologies may have on the design and maintenance of real-world healthcare information systems

(Source: http://www.openclinical.org/ontologies.html)

Systemized Nomenclature Of Medicine Clinical Terms (SNOMED-CT)

Facts

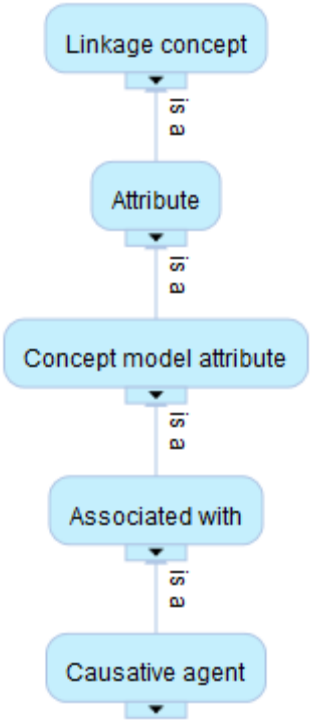
- comprehensive, multi-lingual clinical healthcare terminology
- core general terminology for the electronic health record (EHR) and contains more than 311,000 active concepts (classes) with unique meanings and formal logic-based definitions organized into hierarchies
- cross-map to other international standards
- owned, maintained and distributed by the International Health Terminology Standard Development Organisation (IHTSDO)

Source: http://www.ihtsdo.org/snomed-ct

Systemized Nomenclature Of Medicine Clinical Terms (SNOMED-CT)

Relations

- Is_a: relates a concept to the its more general concepts (standard relation to define concepts in SNOMED-CT)
- About 1 million further relationships in SNOMED-CT
- E.g., “causative agent“ or “finding site“



(Source: http://www.ihtsdo.org/snomed-ct)

Systemized Nomenclature Of Medicine Clinical Terms (SNOMED-CT)

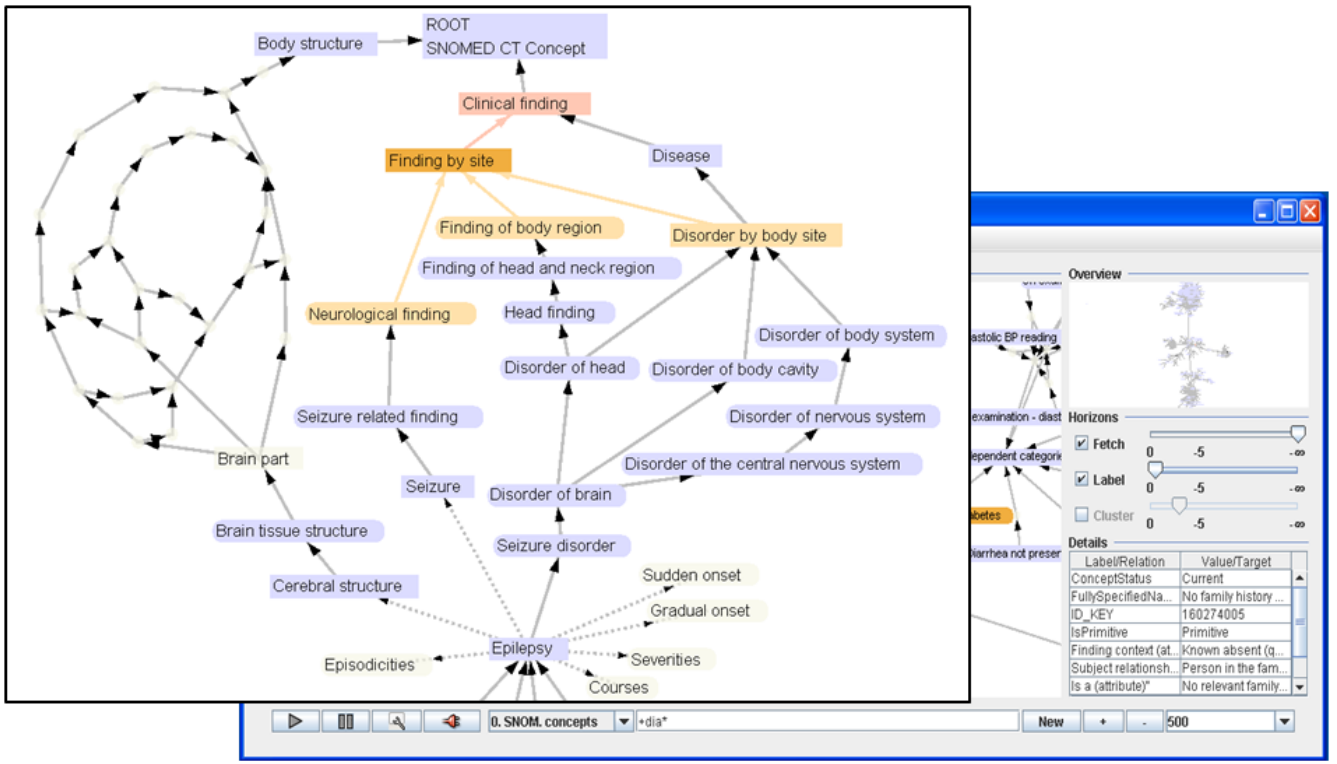
Uses

- capture clinical information at appropriate level
- Integration of patient data
- Improved data interpretation due to common vocabulary
- reduce the need to repeat health history due to data sharing
- Efficient search for patient data
- Provision of large sets of consistent data for medical research

(Source: http://www.ihtsdo.org/snomed-ct)

Systemized Nomenclature Of Medicine Clinical Terms (SNOMED-CT)

Example



(Source: **Interactive Visualization and Navigation of Complex Terminology Systems, Exemplified by SNOMED CT** by Sundvall et al., 2006)

Foundational Model of Anatomy Ontology (FMA)

Facts

- Knowledge source for biomedical informatics
- Goal: representation of the phenotypic structure of the human body; domain ontology of explicit knowledge about human anatomy
- Makes available anatomical information in symbolic form
- Extends the BFO

- 75.000 classes; > 120.000 terms, 168 relationship types
- One of the largest computer-based knowledge sources in biomedical sciences

(Source: http://sig.biostr.washington.edu/projects/fm/)

Foundational Model of Anatomy Ontology (FMA)

Components

Anatomy taxonomy (At):

- anatomical, canonical entities according to the characteristics they share

Anatomical Structural Abstraction (ASA):

- part-whole and spatial relationships that exist between entities

Anatomical Transformation Abstraction (ATA):

- morphological transformation of the entities represented in At

Metaknowledge (Mt):

- principles, rules and definitions according to which classes and relationships in the other three components of FMA are represented

(Source: http://sig.biostr.washington.edu/projects/fm/)

Some Metrics for bio-medical ontologies (bioportal)

NUMBER OF CLASSES:	83281
NUMBER OF INDIVIDUALS:	7
NUMBER OF PROPERTIES:	186
MAXIMUM DEPTH:	22
MAXIMUM NUMBER OF SIBLINGS:	219
AVERAGE NUMBER OF SIBLINGS:	54
CLASSES WITH A SINGLE SUBCLASS:	73
CLASSES WITH MORE THAN 25 SUBCLASSES:	3384
CLASSES WITH NO DEFINITION:	82134

FMA

NUMBER OF CLASSES:	395036
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	41
MAXIMUM DEPTH:	32
MAXIMUM NUMBER OF SIBLINGS:	10010
AVERAGE NUMBER OF SIBLINGS:	1
CLASSES WITH A SINGLE SUBCLASS:	29002
CLASSES WITH MORE THAN 25 SUBCLASSES:	2053
CLASSES WITH NO DEFINITION:	394352

SNOMED-CT

NUMBER OF CLASSES:	38860
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	8
MAXIMUM DEPTH:	18
MAXIMUM NUMBER OF SIBLINGS:	711
AVERAGE NUMBER OF SIBLINGS:	2
CLASSES WITH A SINGLE SUBCLASS:	4652
CLASSES WITH MORE THAN 25 SUBCLASSES:	281
CLASSES WITH NO DEFINITION:	0

GO

NUMBER OF CLASSES:	38551
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	7
MAXIMUM DEPTH:	33
MAXIMUM NUMBER OF SIBLINGS:	4241
AVERAGE NUMBER OF SIBLINGS:	2
CLASSES WITH A SINGLE SUBCLASS:	3323
CLASSES WITH MORE THAN 25 SUBCLASSES:	358
CLASSES WITH NO DEFINITION:	11701

ChEBI

NUMBER OF CLASSES:	3689
NUMBER OF INDIVIDUALS:	163
NUMBER OF PROPERTIES:	114
MAXIMUM DEPTH:	41
MAXIMUM NUMBER OF SIBLINGS:	138
AVERAGE NUMBER OF SIBLINGS:	27
CLASSES WITH A SINGLE SUBCLASS:	911
CLASSES WITH MORE THAN 25 SUBCLASSES:	459
CLASSES WITH NO DEFINITION:	1218

OBI

Some Metrics for bio-medical ontologies

Statistics on OBO foundry types

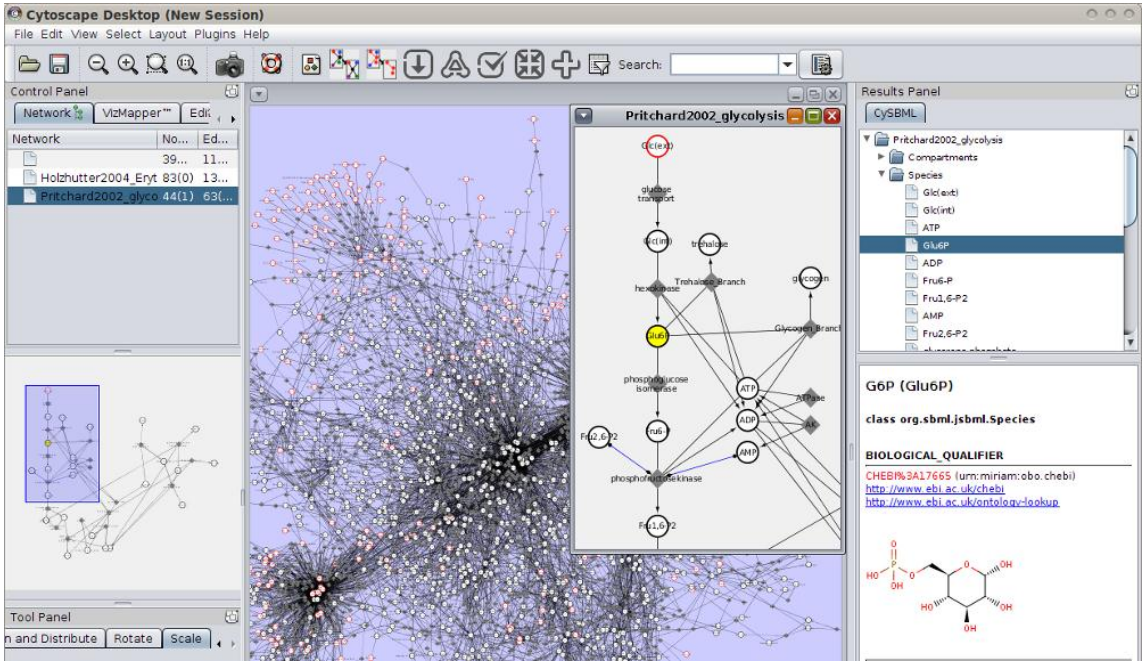
OBO foundry ontologies
library as of NOV 2010

Relation name	Number of times used in ontologies	Number of times used in formal definitions
located_in	294126	58
part_of	97113	6383
partial_overlaps	39381	0
regional_part_of	39329	0
constitutional_part_of	24808	0
tributary_of	18428	0
branch_of	14351	0
regulates	8586	3654
hasMapping	7263	0
negatively_regulates	7063	2980
positively_regulates	6966	2946
has_rank	6028	0
sequence_of	5906	0
systemic_part_of	5340	0
develops_from	5129	175
start	5019	0
end	5011	0
has_functional_parent	4363	0
has_role	4145	0
surrounded_by	2444	2

(Figure: Hoehndorf R, Dumontier M, Oellrich A, Rebholz-Schuhmann D, et al. (2011) Interoperability between Biomedical Ontologies through Relation Expansion, Upper-Level Ontologies and Automatic Reasoning. PLoS ONE 6(7): e22006. doi:10.1371/journal.pone.0022006 http://www.plosone.org/article/info:doi/10.1371/journal.pone.0022006)

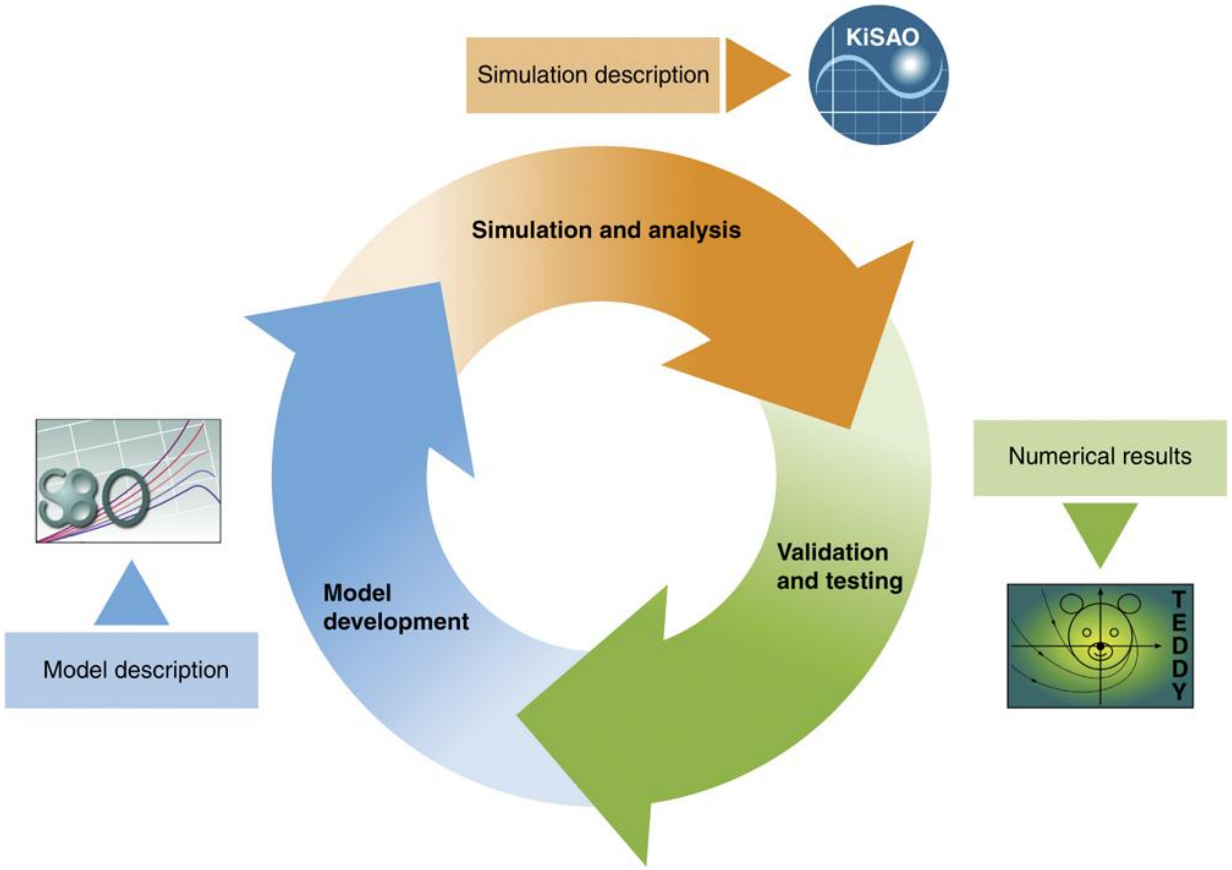
Model annotation

- Re-use of GO, ChEBI, Uniprot-KB, KEGG, Taxonomy … to annotate models
- SBML2OWL



(Source: http://apps.cytoscape.org/apps/cysbml)

Ontologies to support the modeling life cycle

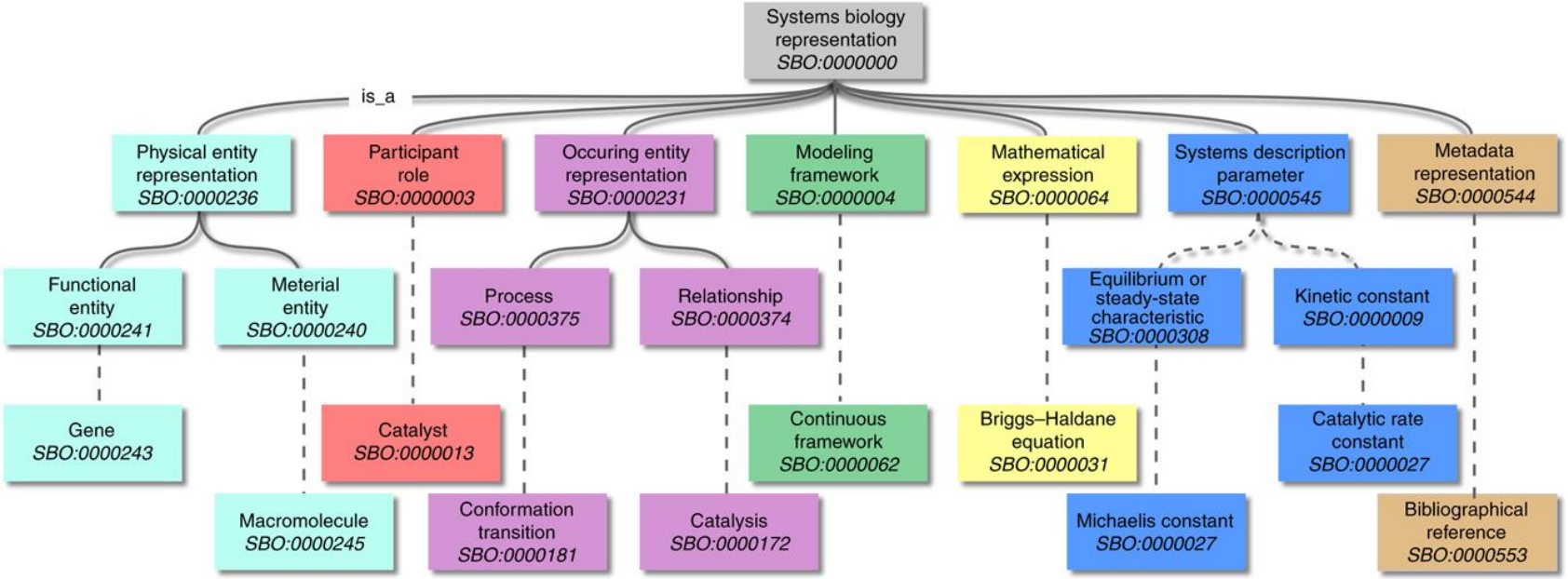


(Source: **Controlled vocabularies and semantics in systems biology** by Courtot et al., 2011)

Systems Biology Ontology (SBO)

Facts

- describes the entities used in computational modeling



(Source: **Controlled vocabularies and semantics in systems biology** by Courtot et al., 2011)

Systems Biology Ontology (SBO)

Facts

- describes the entities used in computational modeling
- Formats: OBO, OWL
- Goal: Annotation of model encodings

```
<reaction id="E1" reversible="false" sboTerm="SBO:0000182" >
    <listOfReactants>
        <speciesReference species="S" sboTerm="SBO:0000015" />
    </listOfReactants>
    <listOfProducts>
        <speciesReference species="P" sboTerm="SBO:0000011" />
    </listOfProducts>
    <listOfModifiers>
        <modifierSpeciesReference species="E" sboTerm="SBO:0000013" />
    </listOfModifiers>
    <kineticLaw sboTerm="SBO:0000029">
        <math xmlns="http://www.w3.org/1998/Math/MathML" >[...]</math>
        <listOfParameters>
            <parameter id="K" value="1" sboTerm="SBO:0000027" />
```

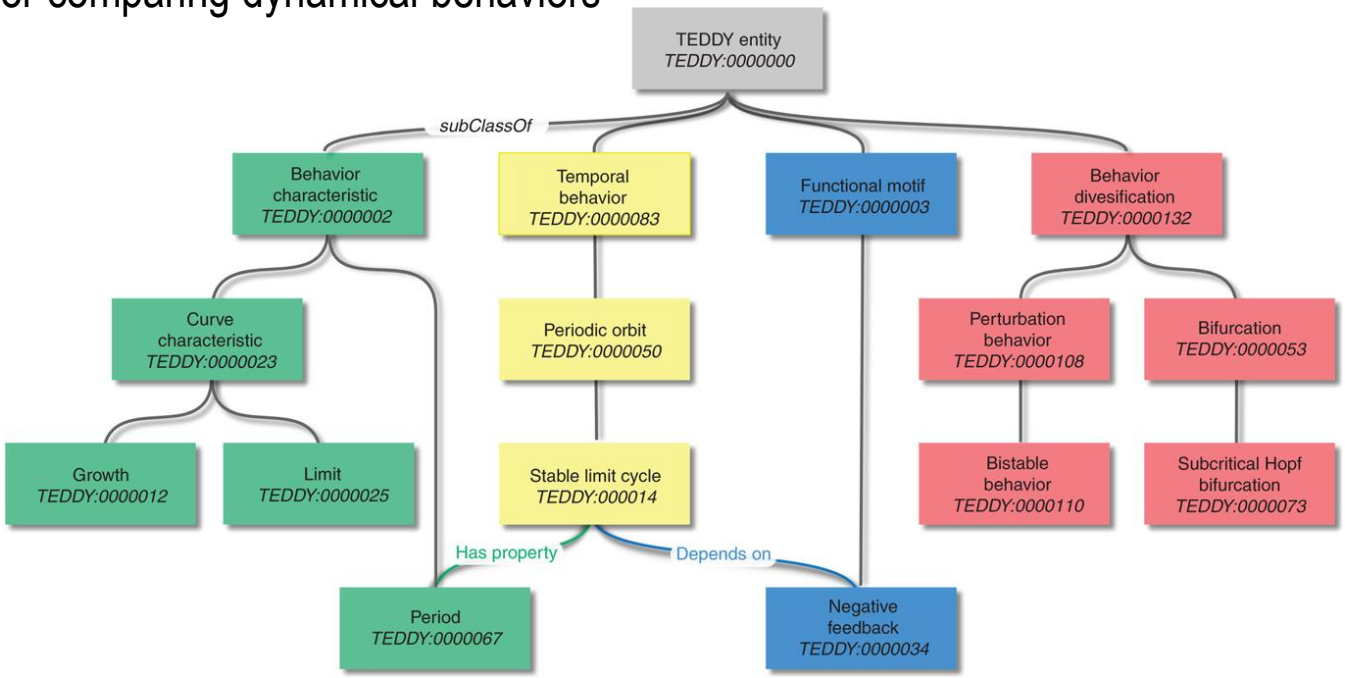
Conversion
Substrate
Product
Catalyst
Henri–Michaelis–Menten rate law
Michaelis constant

(Source: **Controlled vocabularies and semantics in systems biology** by Courtot et al., 2011)

Terminology for the description of dynamics (TEDDY)

Facts

- Terminology for comparing dynamical behaviors



(Source: **Controlled vocabularies and semantics in systems biology** by Courtot et al., 2011)

Terminology for the description of dynamics (TEDDY)

Facts

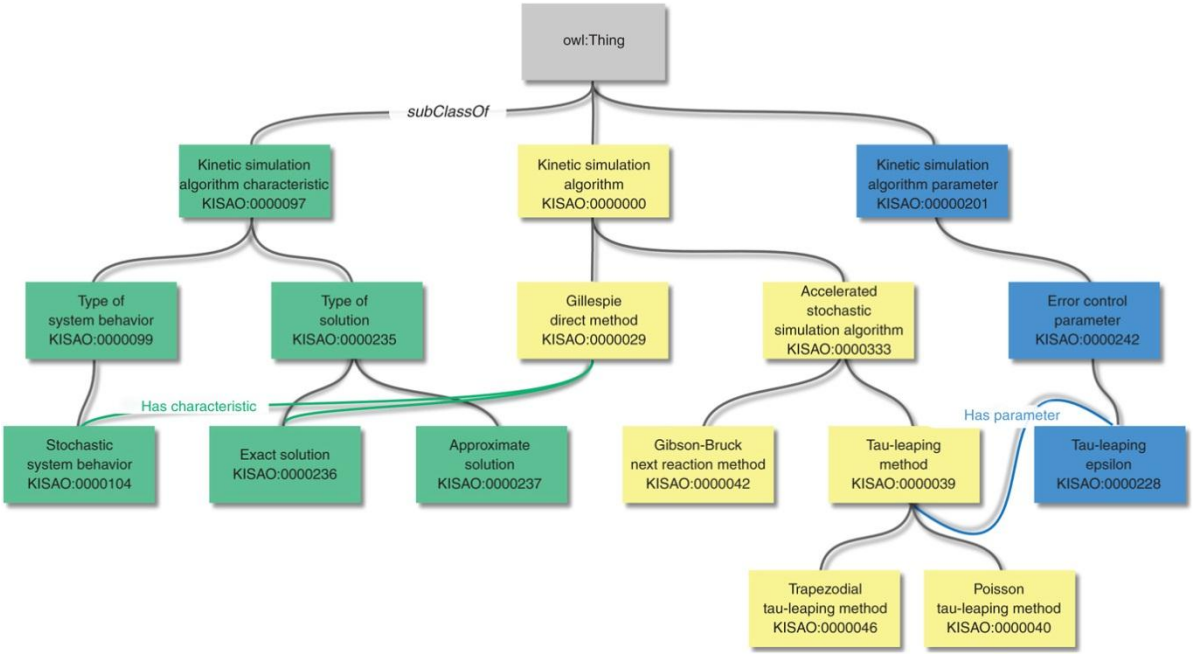
- Terminology for comparing dynamical behaviors
- Format: OWL
- Goal: naming the critical dynamical features of models, and relating them within one set of numerical results

(Source: **Controlled vocabularies and semantics in systems biology** by Courtot et al., 2011)

Kinetic Simulation Algorithm Ontology (KiSAO)

Facts

- Characterization and classification of simulation algorithms used to solve kinetic models



(Source: **Controlled vocabularies and semantics in systems biology** by Courtot et al., 2011)

Kinetic Simulation Algorithm Ontology (KiSAO)

Facts

- Characterization and classification of simulation algorithms used to solve kinetic models
- Format: OBO, OWL
- Goal: To unambiguously identify a simulation algorithm in an experiment

SED ML

```
<listOfSimulations>
    <uniformTimeCourse id="sim1"
                       initialTime="0"
                       outputStartTime="0"
                       outputEndTime="5"
                       numberOfPoints="51">
        <algorithm kisaoID="KISAO:0000019" />
    </uniformTimeCourse>
```

CVODE

(Source: **Controlled vocabularies and semantics in systems biology** by Courtot et al., 2011)

Metrics for systems biology ontologies (bioportal)

NUMBER OF CLASSES:	557
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	6
MAXIMUM DEPTH:	9
MAXIMUM NUMBER OF SIBLINGS:	19
AVERAGE NUMBER OF SIBLINGS:	1
CLASSES WITH A SINGLE SUBCLASS:	41
CLASSES WITH MORE THAN 25 SUBCLASSES:	
CLASSES WITH NO DEFINITION:	1

SBO

NUMBER OF CLASSES:	161
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	17
MAXIMUM DEPTH:	8
MAXIMUM NUMBER OF SIBLINGS:	18
AVERAGE NUMBER OF SIBLINGS:	6
CLASSES WITH A SINGLE SUBCLASS:	9
CLASSES WITH MORE THAN 25 SUBCLASSES:	
CLASSES WITH NO DEFINITION:	13

TEDDY

NUMBER OF CLASSES:	229
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	11
MAXIMUM DEPTH:	7
MAXIMUM NUMBER OF SIBLINGS:	16
AVERAGE NUMBER OF SIBLINGS:	8
CLASSES WITH A SINGLE SUBCLASS:	8
CLASSES WITH MORE THAN 25 SUBCLASSES:	
CLASSES WITH NO DEFINITION:	11

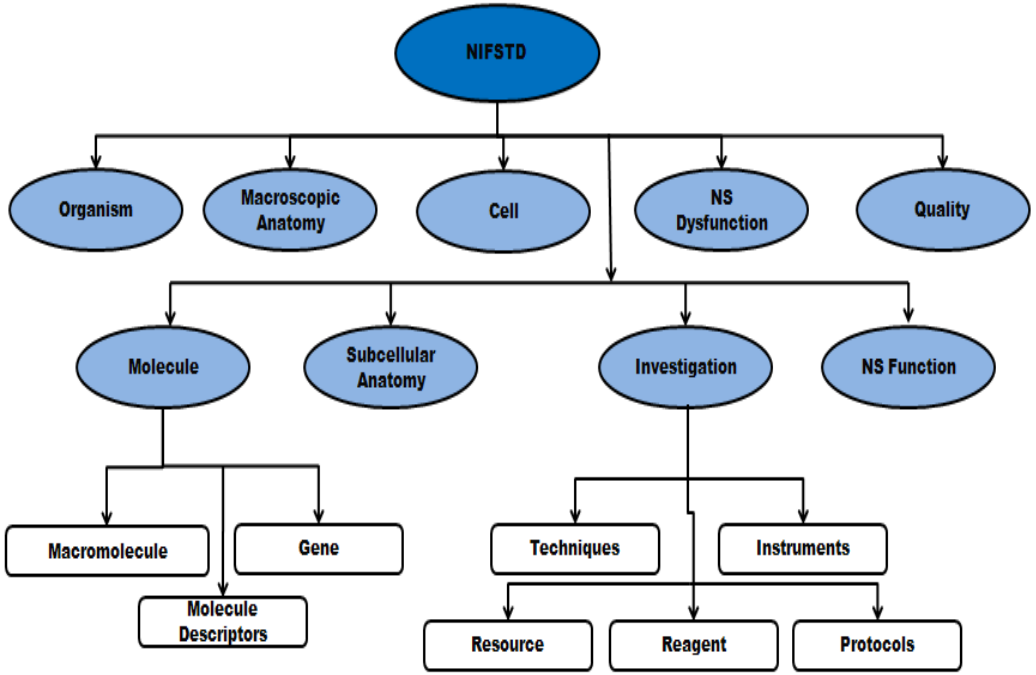
KiSAO

(Source: **bioportal**)

NIF Standard Ontology (NIFSTD)

Facts

- Core component of the Neuroscience Information Framework for access and integration of neuroscience
- Set of modular ontologies
- ~60, 000 distinct concepts and synonymes
- Format: OWL-DL



NIF Standard Ontology (NIFSTD)

Facts

- Terminology to describe neuroscience data and resources
- Goal/Wish:
  By utilizing the core classes of the NIFSTD, ontologies for neuroscience can be built covering almost any domain. Because they all reference the same core classes, we can aggregate information together through knowledge networks

NIF Standard Ontology (NIFSTD)

Facts

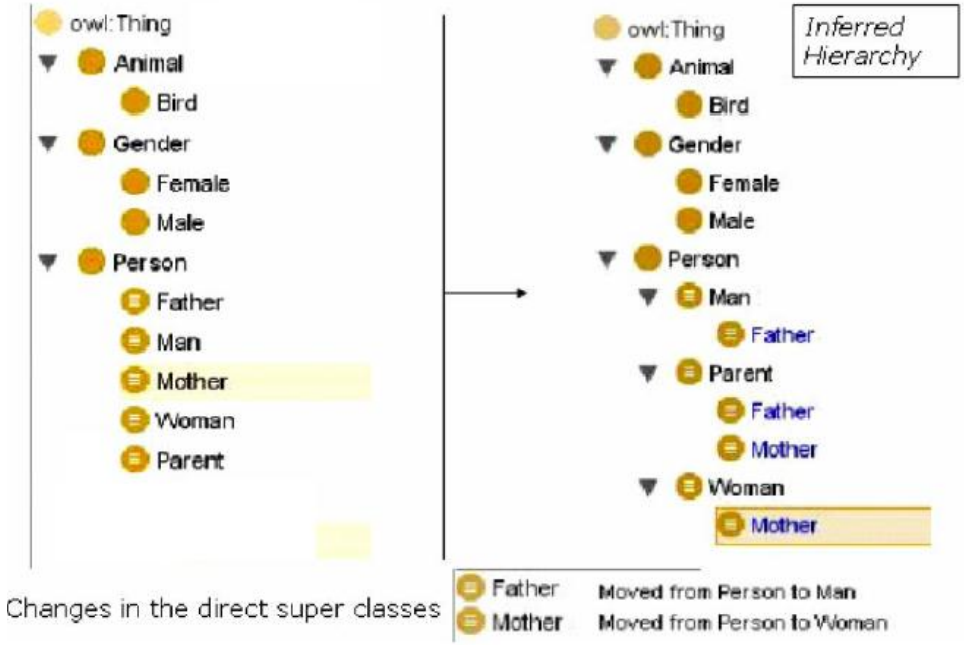
- Terminology to describe neuroscience data and resources
- Example: neuron definitions
  - Neurons classified by
    - **Soma location** in different brain regions - e.g., Hippocampal neuron, Cerebellum neuron
    - **Neurotransmitter** - e.g., GABAergic neuron
    - **Circuit roles** - e.g., Intrinsic neuron, Projection neuron
    - **Morphology** - e.g., Spiny neuron
    - **Molecular constituents** - e.g., Pervalbumin neuron, Calretinin neuron

(Source: **NEUROSCIENCE INFORMATION FRAMEWORK STANDARD ONTOLOGIES (NIFSTD)** by Imam et al., 2011)

NIF Standard Ontology (NIFSTD)

Facts

- Single inheritance principle
  - Simple tree structure
  - But: inferred structure may lead to multiple parents
  - NIFSTD relies on automated reasoning
- Format: OWL-DL



(Source: https://confluence.crbs.ucsd.edu/download/attachments/16875650/NIF_Webinar_Feb_9_slides.pdf)

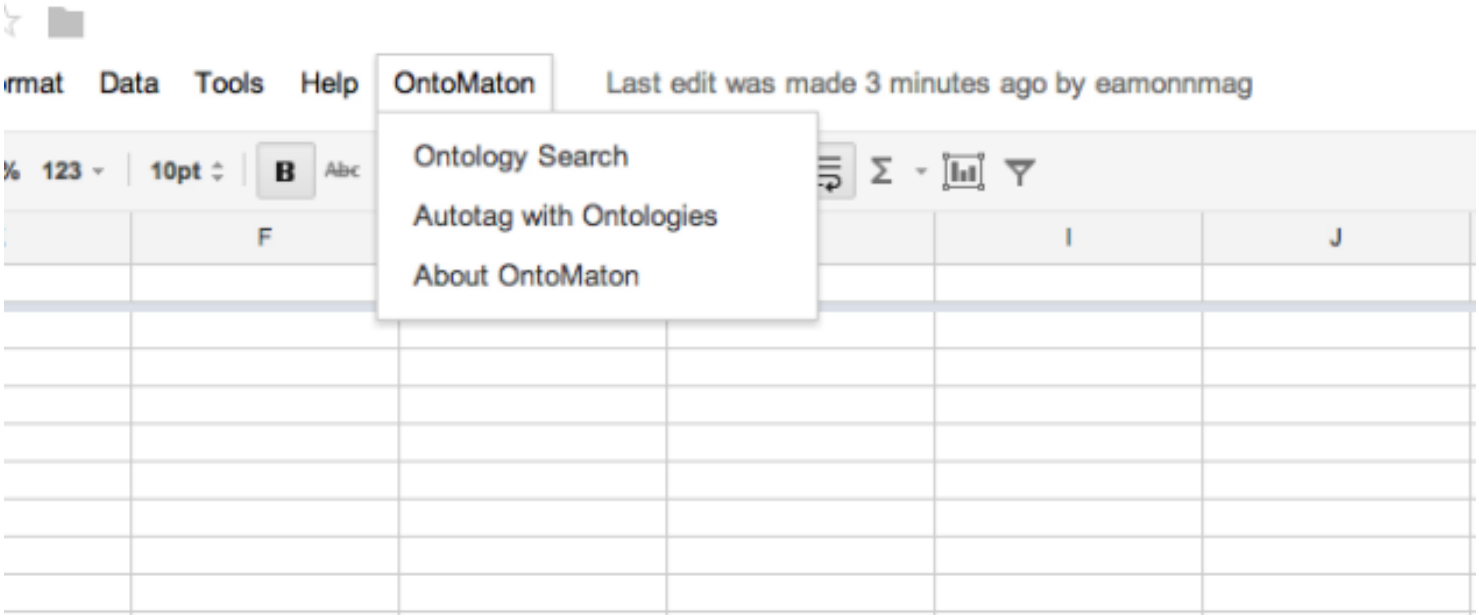
NIF Standard Ontology (NIFSTD)

Uses

- Neuroscience Information framework (Web-based neuroscience resources)
- NCBO Annotator (tagging free text with ontology concepts)
- OntoCAT (High level abstraction API for interacting with ontology resources)
- Eagle-i (a searchable network of research resources)
- OntoMaton (ontology search and tagging directly within google spreadsheets)
- Gemma (meta-analysis, re-use and sharing of genomics data)

NIF Standard Ontology (NIFSTD)

Uses



http://isatools.wordpress.com/tag/ncbo-bioportal/

Neuro Behavior Ontology (NBO)

Facts

Two-fold ontology

- ontology of behavioral processes
  - behavior processes complementing and extending GO's process ontology
- ontology of behavioral phenotypes
  - classification of both normal and abnormal behavioral characteristics of organisms
- Format: OWL

Neuro Behavior Ontology (NBO)

Uses

- OntoCAT
- OntoMaton

The neurobehavior ontology: an ontology for annotation and integration of behavior and behavioral phenotypes.

Georgios V Gkoutos, Paul N Schofield, and Robert Hoehndorf
Int Rev Neurobiol (2012), PMID 23195121

Ontology of subcellular anatomy (SAO)

Facts

- Goal: describe data from light and electron microscopic imaging and provide the conceptual bridge between whole brain anatomy and macromolecular scales
- Format: OWL
- Notion of instances to answer questions statistically, e.g., “How many primary dendrites does a Purkinje cell have?” can be answered based on the number of instances of that cell available in the knowledge base

Reviews on ontologies in neuroscience

- Neuroscience Information Framework (NIF) project (Gardner et al., 2008a)
- Bota and Swanson (2008a),
- Gardner et al. (2008b)
- Bowden et al. (2007)
- Full review: Bug et al. (2008)

Metrics for ontologies in neuroscience

Bioportal

NUMBER OF CLASSES:	59110
NUMBER OF INDIVIDUALS:	4612
NUMBER OF PROPERTIES:	264
MAXIMUM DEPTH:	1
MAXIMUM NUMBER OF SIBLINGS:	6212
AVERAGE NUMBER OF SIBLINGS:	189
CLASSES WITH A SINGLE SUBCLASS:	5704
CLASSES WITH MORE THAN 25 SUBCLASSES:	21759
CLASSES WITH NO DEFINITION:	57320

NIFSTD

NUMBER OF CLASSES:	813
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	6
MAXIMUM DEPTH:	10
MAXIMUM NUMBER OF SIBLINGS:	21
AVERAGE NUMBER OF SIBLINGS:	1
CLASSES WITH A SINGLE SUBCLASS:	64
CLASSES WITH MORE THAN 25 SUBCLASSES:	
CLASSES WITH NO DEFINITION:	813

NBO

NUMBER OF CLASSES:	821
NUMBER OF INDIVIDUALS:	0
NUMBER OF PROPERTIES:	85
MAXIMUM DEPTH:	12
MAXIMUM NUMBER OF SIBLINGS:	97
AVERAGE NUMBER OF SIBLINGS:	16
CLASSES WITH A SINGLE SUBCLASS:	62
CLASSES WITH MORE THAN 25 SUBCLASSES:	2
CLASSES WITH NO DEFINITION:	821

SAO

The Open Biological and Biomedical Ontologies format (OBO format)

- Commonly used format in biomedical ontologies
- Collaborative approach to developing principles for ontology development for creating a suite of **orthogonal** and **interoperable** reference ontologies for the biomedical domain
- Core ontologies, ~60 candidates

Title	Domain	Prefix
Biological process	biological process	GO
Cellular component	anatomy	GO
Chemical entities of biological interest	biochemistry	CHEBI
Molecular function	biological function	GO
Phenotypic quality	phenotype	PATO
PRotein Ontology (PRO)	proteins	PR
Xenopus anatomy and development	anatomy	XAO
Zebrafish anatomy and development	anatomy	ZFA

(Source: http://www.obofoundry.org/)

The Open Biological and Biomedical Ontologies format (OBO format)

- Covers a subset of OWL DL
- Additionally defines standard syntax for representing important classes of meta-data including as synonyms, references to publications and deprecated IDs

[Term]

id: GO:0001101

name: response to acid namespace: biological_process

def: "Any process that results in a change in state or activity of a cell or an organism (in terms of movement, secretion, enzyme production, gene expression, etc.) as a result of an acid stimulus." [GOC:rn]

is_a: GO:0042221 ! response to chemical stimulus

Example: GO OBO flat file

The NCBO bioportal

- access to commonly used biomedical ontologies and to tools for working with them
- 333 ontologies
- browse the library of ontologies
- search for a term across multiple ontologies
- annotate text with terms from ontologies
- search biomedical resources for a term

http://bioportal.bioontology.org/

Ambiguity of terminology

Look at the different representations of blood in existing ontologies (FMA, SNOMED, GO, WordNet, UMLS Metasaurus, MENELAS).

How do these descriptions differ and why?

Contributing to Ontology development

Commentary

Nature Biotechnology **23**, 1095 - 1098 (2005)
doi:10.1038/nbt0905-1095

Are the current ontologies in biology good ontologies?

Larisa N Soldatova[1] & Ross D King[1]

The failure of many bio-ontologies to follow international standards for ontology design and description is hampering their application and threatens to restrict their future use.

Summary Part 2

- Find the right ontology for your domain
- Use tools to do so
  - Bioportal
  - Ontology Lookup Service
- Don’t reinvent the wheel (“meta-silos”)

References Part 2

tbw