MURINET

Multidisciplinary Research Network on Health and Disability in Europe

EUROPEAN PROJECT

Stefan Schulz

Medical Informatics
Research Group
University
Medical Center

Freiburg, Germany

Ontological Developments of the International Classification of Functioning, Disabilities and Health (ICF)
28-29 May 2010, Centro Culturale Don Orione
Artigianelli, Venezia, Italy

Biomedical Classifications and Ontologies

UNIVERSITÄTS KLINIKUM FREIBURG

Purpose of this talk

- To give an overview of terminological system in biology and medicine
- To clarify the distinctions between
  - Terminologies / Thesauri
  - Ontologies
- To promote good ontological practice
- To contrast ontologies with classifications
- To address ontology aspects in ICF

Purpose of this talk

- To give an overview of terminological system in biology and medicine
- To clarify the distinctions between
  - Terminologies / Thesauri
  - Ontologies
- To promote good ontological practice
- To contrast ontologies with classifications
- To address ontology aspects in ICF

Examples of Terminology Systems

- Medical Subject Headings (MeSH)
- International Classification of Diseases (ICD)
- Systematized Nomenclature of Medicine Clinical Terms (SNOMED CT)
- Open Biomedical Ontologies (OBO)

Medical Subject Headings (MeSH)

Medical Subject Headings (MeSH)

1. Anatomy [A]
2. Organisms [B]
   - Animals [B01] +
   - Algae [B02] +
   - Bacteria [B03] +
   - Viruses [B04] +
   - Fungi [B05] +
   - Plants [B06] +
   - Archaea [B07] +
   - Mesomycetozoea [B08] +
3. Diseases [C]
4. Chemicals and Drugs [D]
5. Analytical, Diagnostic and Therapeutic Techniques and Equipment [E]
6. Psychiatry and Psychology [F]
7. Biological Sciences [G]
8. Physical Sciences [H]
9. Anthropology, Education, Sociology and Social Phenomena [I]
10. Technology and Food and Beverages [J]
11. Humanities [K]
12. Information Science [L]
13. Persons [M]
14. Health Care [N]
15. Publication Characteristics [V]
16. Geographic Locations [Z]

Medical Subject Headings (MeSH)

Bacteria [B03]

- Atypical Bacterial Forms [B03.110] +
- Bacteria, Aerobic [B03.120]
- Bacteria, Anaerobic [B03.130]
- Bacteroidetes [B03.140] +
- Biofilms [B03.150]
- Blood-Borne Pathogens [B03.165]
- Chlorobi [B03.250] +
- Chloroflexi [B03.275] +
- Cyanobacteria [B03.280] +
- Endospore-Forming Bacteria [B03.300] +
- Fusobacteria [B03.370] +
- Gram-Negative Bacteria [B03.440] +
- Gram-Positive Bacteria [B03.510]
  - Actinobacteria [B03.510.024] +
  - Gram-Positive Cocci [B03.510.400] +
  - Gram-Positive Endospore-Forming Bacteria [B03.510.415] +
  - Gram-Positive Rods [B03.510.460] +
- Proteobacteria [B03.660] +
- Spirochaetales [B03.851] +
- Spores [B03.867] +
- Sulfur-Reducing Bacteria [B03.900] +

Hierarchical principle: broader term / narrower term (not a taxonomy)

Return to Entry Page

Bacteria [B03]
Endospore-Forming Bacteria [B03.300]
Gram-Positive Endospore-Forming Bacteria [B03.300.390]
Gram-Positive Endospore-Forming Rods [B03.300.390.400]
Staphylococcaceae [B03.300.390.400.800]
Staphylococcus [B03.300.390.400.800.750]
Staphylococcus aureus [B03.300.390.400.800.750.100]
Methicillin-Resistant Staphylococcus aureus [B03.300.390.400.800.750.100.500]
Staphylococcus epidermidis [B03.300.390.400.800.750.343]
Staphylococcus haemolyticus [B03.300.390.400.800.750.400]
Staphylococcus hominis [B03.300.390.400.800.750.425]

MeSH Trees

Bacteria [B03]
Gram-Positive Bacteria [B03.510]
Bacillales [B03.510.100]
Staphylococcaceae [B03.510.100.750]
Staphylococcus [B03.510.100.750.750]
Staphylococcus aureus [B03.510.100.750.750.100]
Methicillin-Resistant Staphylococcus aureus [B03.510.100.750.750.100.500]
Staphylococcus epidermidis [B03.510.100.750.750.343]
Staphylococcus haemolyticus [B03.510.100.750.750.400]
Staphylococcus hominis [B03.510.100.750.750.425]

Bacteria [B03]
Gram-Positive Bacteria [B03.510]
Gram-Positive Cocci [B03.510.400]
Staphylococcaceae [B03.510.400.790]
Staphylococcus [B03.510.400.790.750]
Staphylococcus aureus [B03.510.400.790.750.100]
Methicillin-Resistant Staphylococcus aureus [B03.510.400.790.750.100.500]

MeSH Metadata

MeSH Heading	Staphylococcus aureus
Tree Number	B03.300.390.400.800.750.100
Tree Number	B03.510.100.750.750.100
Tree Number	B03.510.400.790.750.100
Annotation	infection = STAPHYLOCOCCAL INFECTIONS & do not bother to coord with S. aureus unless particularly discussed (index IM); DF: STAPH AUREUS
Scope Note	Potentially pathogenic bacteria found in nasal membranes, skin, hair follicles, and perineum of warm-blooded animals. They may cause a wide range of infections and intoxications.
Allowable Qualifiers	CH CL CY DE EN GD GE IM IP ME PH PY RE UL VI
Entry Version	STAPH AUREUS
Previous Indexing	Staphylococcus (1966-1974)
Online Note	use STAPHYLOCOCCUS AUREUS to search MICROCOCCUS PYOGENES 1975-91; use STAPHYLOCOCCUS 1966-74
History Note	76; was MICROCOCCUS PYOGENES see under STAPHYLOCOCCUS 1963-75; MICROCOCCUS PYOGENES was see STAPHYLOCOCCUS AUREUS 1976-91
Date of Entry	19750721
Unique ID	D013211

International Classification of Diseases (ICD)

International Classification of Diseases (ICD)

International Statistical Classification of Diseases and Related Health Problems
10th Revision
Version for 2007

Tabular List of inclusions and four-character subcategories

Chapter List

Chapter List

Chapter	Blocks	Title
I	A00-B99	Certain infectious and parasitic diseases
II	C00-D48	Neoplasms
III	D50-D89	Diseases of the blood and blood-forming organs and certain disorders involving the im
IV	E00-E90	Endocrine, nutritional and metabolic diseases
V	F00-F99	Mental and behavioural disorders
VI	G00-G99	Diseases of the nervous system
VII	H00-H59	Diseases of the eye and adnexa
VIII	H60-H95	Diseases of the ear and mastoid process
IX	I00-I99	Diseases of the circulatory system
X	J00-J99	Diseases of the respiratory system
XI	K00-K93	Diseases of the digestive system
XII	L00-L99	Diseases of the skin and subcutaneous tissue
XIII	M00-M99	Diseases of the musculoskeletal system and connective tissue
XIV	N00-N99	Diseases of the genitourinary system
XV	O00-O99	Pregnancy, childbirth and the puerperium
XVI	P00-P96	Certain conditions originating in the perinatal period
XVII	Q00-Q99	Congenital malformations, deformations and chromosomal abnormalities
XVIII	R00-R99	Symptoms, signs and abnormal clinical and laboratory findings, not elsewhere classifie
XIX	S00-T98	Injury, poisoning and certain other consequences of external causes
XX	V01-Y98	External causes of morbidity and mortality
XXI	Z00-Z99	Factors influencing health status and contact with health services
XXII	U00-U99	Codes for special purposes

Disjoint categories

International Classification of Diseases (ICD)

Chapter VII:

Diseases of the eye and adnexa
(H00-H59)

H00-H06	Disorders of eyelid, lacrimal system and orbit
H10-H13	Disorders of conjunctiva
H15-H22	Disorders of sclera, cornea, iris and ciliary body
H25-H28	Disorders of lens
H30-H36	Disorders of choroid and retina
H40-H42	Glaucoma
H43-H45	Disorders of vitreous body and globe
H46-H48	Disorders of optic nerve and visual pathways
H49-H52	Disorders of ocular muscles, binocular movement, accommodation and refraction
H53-H54	Visual disturbances and blindness
H55-H59	Other disorders of eye and adnexa

Disjoint subcategories

Glaucoma (H40-H42)

H40 **Glaucoma**

Excludes: absolute glaucoma (H44.5)
congenital glaucoma (Q15.0)
traumatic glaucoma due to birth injury (P15.3)

H40.0 **Glaucoma suspect**
Ocular hypertension

H40.1 **Primary open-angle glaucoma**
Glaucoma (primary)(residual stage):
· capsular with pseudoexfoliation of lens
· chronic simple
· low-tension
· pigmentary

H40.2 **Primary angle-closure glaucoma**
Angle-closure glaucoma (primary)(residual stage):
· acute
· chronic
· intermittent

H40.3 **Glaucoma secondary to eye trauma**
Use additional code, if desired, to identify cause.

H40.4 **Glaucoma secondary to eye inflammation**
Use additional code, if desired, to identify cause.

H40.5 **Glaucoma secondary to other eye disorders**
Use additional code, if desired, to identify cause.

H40.6 **Glaucoma secondary to drugs**
Use additional external cause code (Chapter XX), if desired, to identify drug.

H40.8 **Other glaucoma**

H40.9 **Glaucoma, unspecified**

H42* **Glaucoma in diseases classified elsewhere**

H42.0* **Glaucoma in endocrine, nutritional and metabolic diseases**
Glaucoma in:
· amyloidosis (E85.-+)
· Lowe's syndrome (E72.0+)

H42.8* **Glaucoma in other diseases classified elsewhere**
Glaucoma in onchocerciasis (B73+)

Exclusions

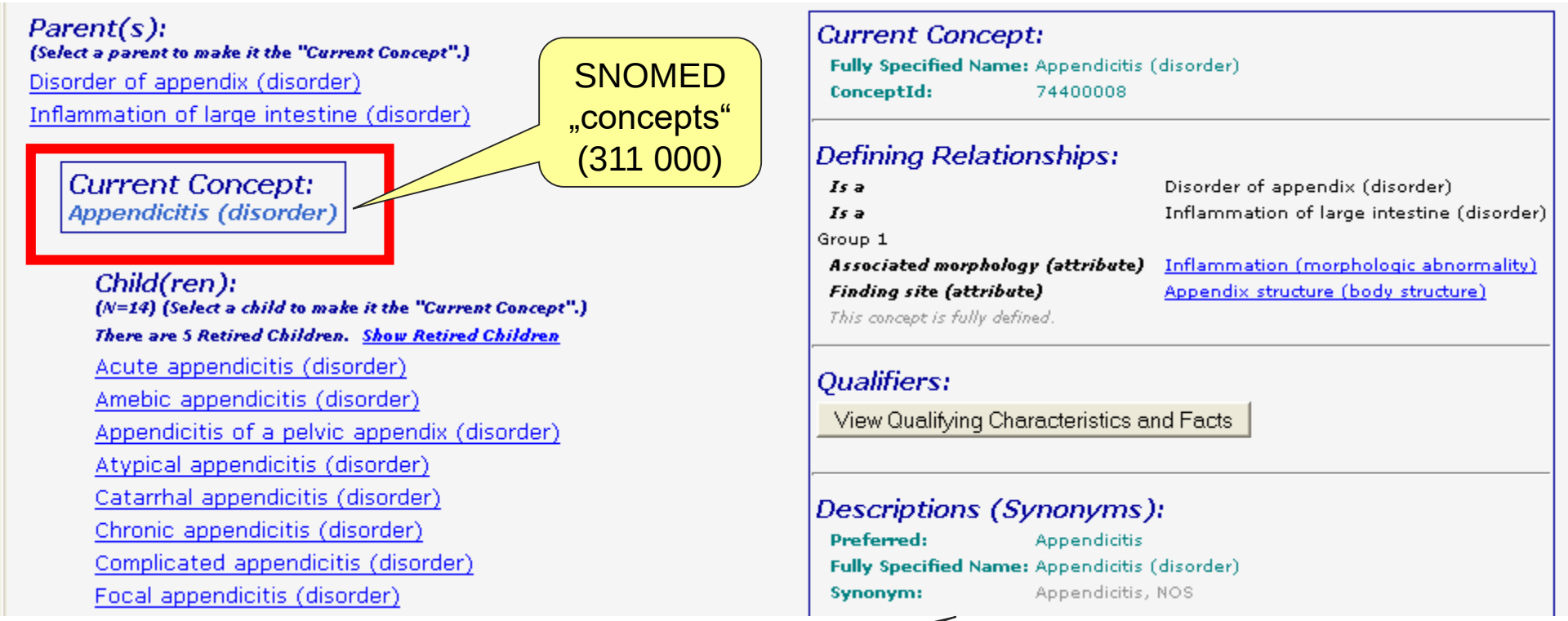
Disjoint classes at three and four-digit - level

Residual classes

Optional secondary classes

Systematized Nomenclature of Medicine Clinical Terms (SNOMED CT)

SNOMED CT
Thesaurus aspects



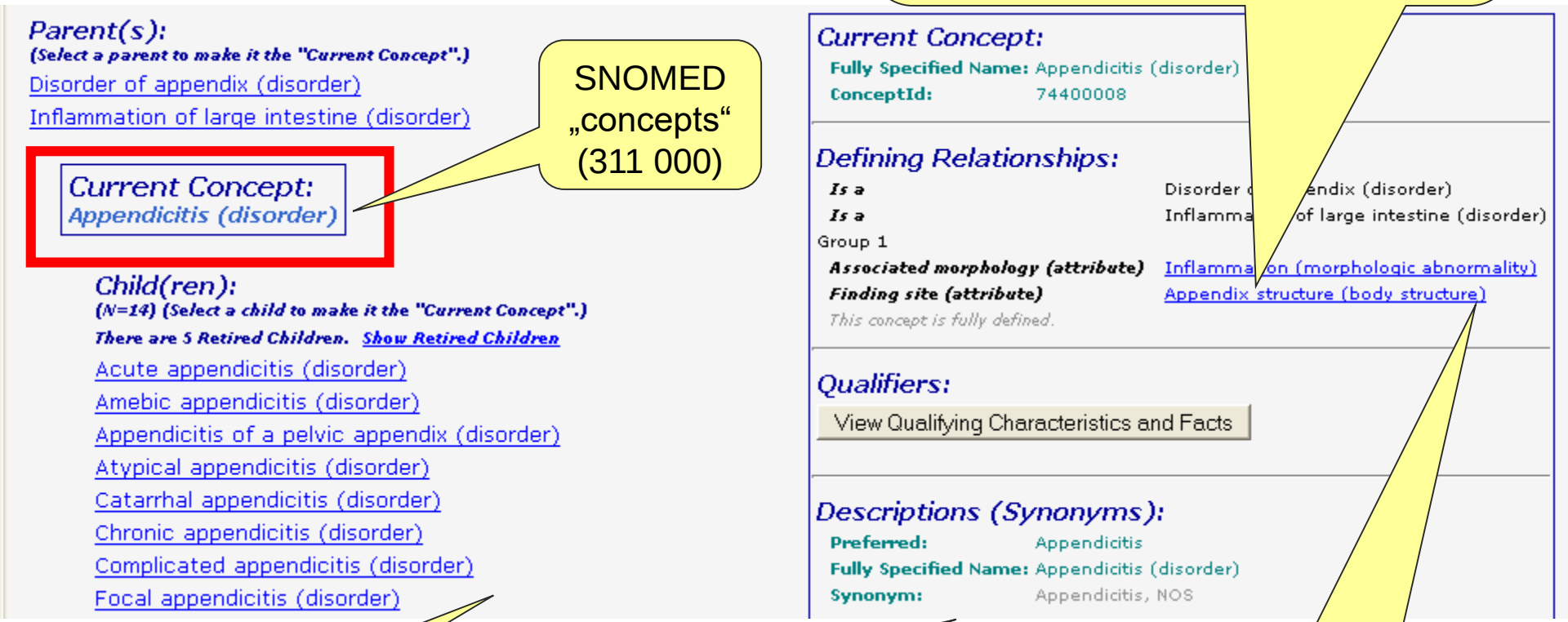
SNOMED „concepts“ (311 000)

732 000 engl. terms

SNOMED CT
Ontology aspects

restrictions based on
simple description logics:

$C1 - Rel - C2$ interpreted as:
$\forall x: instanceOf(x, C1) \Rightarrow$
$\exists y: instanceOf(C2) \wedge Rel(x,y)$



SNOMED „concepts“ (311 000)

specialization hierarchy (is-a) (taxonomy)

732 000 engl. terms

Relations (Attributes): z.B.
Associated morphology
Finding site

Open Biomedical Ontologies (OBO)

OBO Foundry

The Open Biological and Biomedical Ontologies

Ontologies | Resources | Participate | About

The OBO Foundry is a collaborative experiment involving developers of science-based ontologies who are establishing a set of principles for ontology development with the goal of creating a suite of orthogonal interoperable reference ontologies in the biomedical domain. The groups developing ontologies who have expressed an interest in this goal are listed below, followed by other relevant efforts in this domain.

In addition to a listing of OBO ontologies, this site also provides a statement of the OBO Foundry principles, discussion fora, technical infrastructure, and other services to facilitate ontology development. We welcome feedback and encourage participation.

Click any column header to sort the table by that column. The s link to the term request trackers for the listed ontologies.

OBO Foundry ontologies

Title	Domain	Prefix	File	Last changed
Biological process	biological process	GO	gene_ontology_edit.obo	2010/05/26
Cellular component	anatomy	GO	gene_ontology_edit.obo	2010/05/26
Chemical entities of biological interest	biochemistry	CHEBI	chebi.obo	2010/05/14
Molecular function	biological function	GO	gene_ontology_edit.obo	2010/05/26
Phenotypic quality	phenotype	PATO	quality.obo	2010/05/15
PRotein Ontology (PRO)	proteins	PRO	pro.obo	2010/05/18
Xenopus anatomy and development	anatomy	XAO	xenopus_anatomy.obo	2009/12/02
Zebrafish anatomy and development	anatomy	ZFA	zebrafish_anatomy.obo	2010/04/12

OBO Foundry: vision

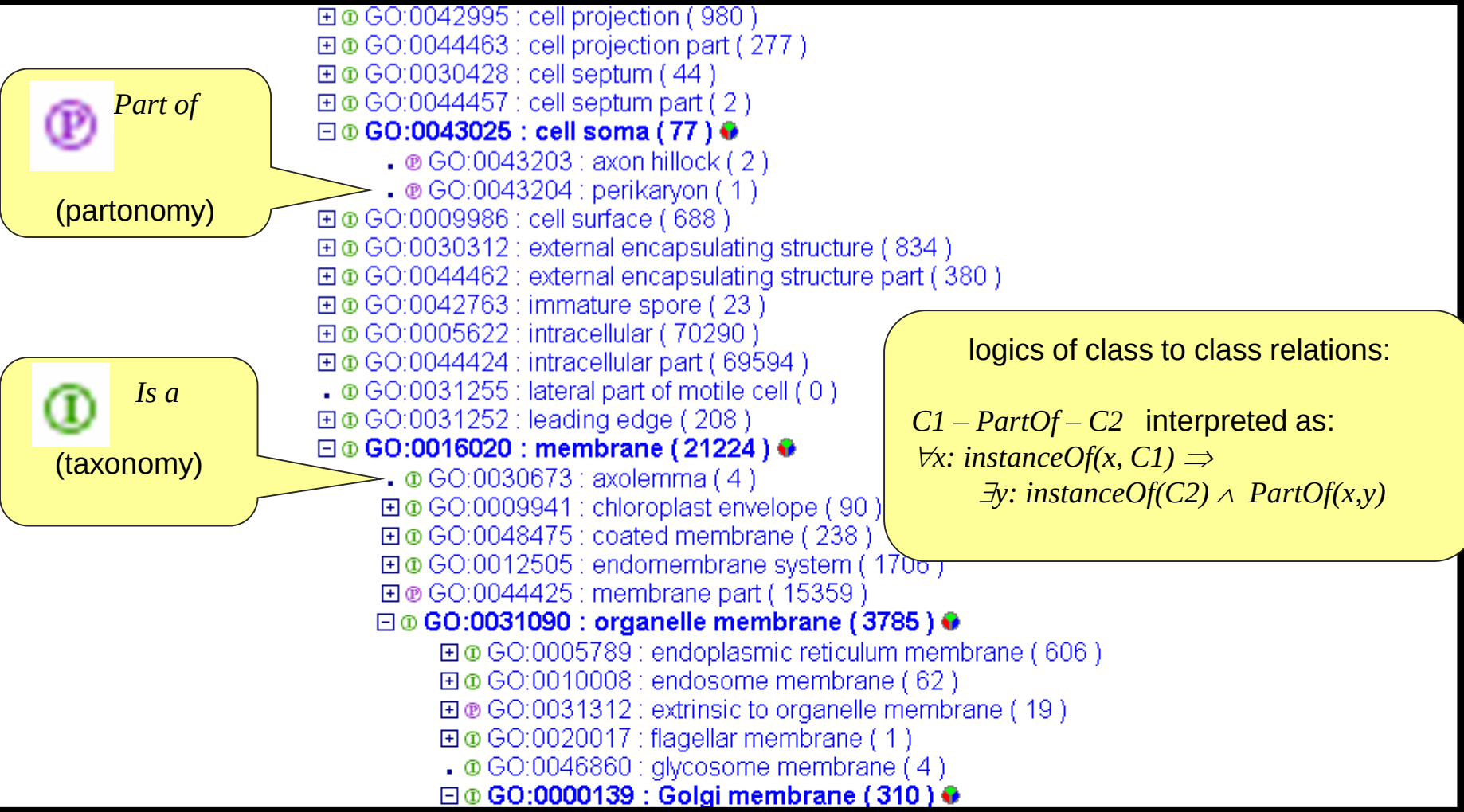
RELATION TO TIME / GRANULARITY	CONTINUANT				OCCURRENT
	INDEPENDENT		DEPENDENT		
ORGAN AND ORGANISM	Organism (NCBI Taxonomy)	Anatomical Entity (FMA, CARO)	Organ Function (FMP, CPRO)	Phenotypic Quality (PaTO)	**Biological Process** (GO)
CELL AND CELLULAR COMPONENT	Cell (CL)	**Cellular Component** (FMA, GO)	Cellular Function (GO)		
MOLECULE	Molecule (ChEBI, SO, RnaO, PrO)		**Molecular Function** (GO)		Molecular Process (GO)

Smith B et al. The OBO Foundry: coordinated evolution of ontologies to support biomedical data integration. Nat Biotechnol. 2007 Nov;25(11):1251-5.

OBO Foundry example: Molecular function hierarchy from Gene Ontology

- GO:0003674 : molecular_function [374855 gene products]
  - GO:0016209 : antioxidant activity [2323 gene products]
    - GO:0045174 : glutathione dehydrogenase (ascorbate) activity [9 gene products]
    - GO:0004362 : glutathione-disulfide reductase activity [56 gene products]
    - GO:0004601 : peroxidase activity [1299 gene products]
    - GO:0004784 : superoxide dismutase activity [477 gene products]
    - GO:0050605 : superoxide reductase activity [11 gene products]
    - GO:0004791 : thioredoxin-disulfide reductase activity [113 gene products]
  - GO:0005488 : binding [171370 gene products]
    - GO:0000035 : acyl binding [5 gene products]
    - GO:0043178 : alcohol binding [176 gene products]
      - GO:0033265 : choline binding [13 gene products]
      - GO:0035240 : dopamine binding [58 gene products]
      - GO:0051379 : epinephrine binding [10 gene products]
      - GO:0035276 : ethanol binding [9 gene products]
      - GO:0043533 : inositol 1,3,4,5 tetrakisphosphate binding [6 gene products]
      - GO:0070679 : inositol 1,4,5 trisphosphate binding [5 gene products]
      - GO:0000822 : inositol hexakisphosphate binding [40 gene products]
      - GO:0051380 : norepinephrine binding [30 gene products]
      - GO:0031210 : phosphatidylcholine binding [20 gene products]
    - GO:0033218 : amide binding [0 gene products]
      - GO:0033219 : urea binding [0 gene products]
    - **GO:0043176 : amine binding [1095 gene products]**
      - GO:0033226 : 2-aminoethylphosphonate binding [0 gene products]
      - GO:0042166 : acetylcholine binding [111 gene products]
      - GO:0016597 : amino acid binding [804 gene products]
      - GO:0033265 : choline binding [13 gene products]

OBO Foundry example: Gene Ontology partonomies and taxonomies



Different Purposes – Heterogeneous Approaches

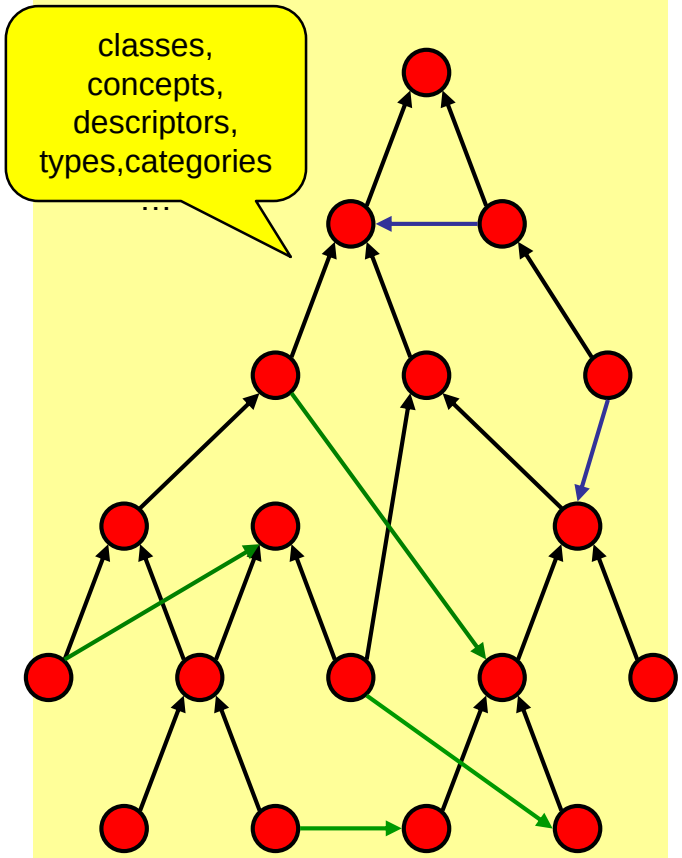
- **Terminology: MeSH** [Medical Subject Headings]:
  Hierarchy (broader / narrower) of *descriptors*, used for indexing biomedical publications for retrieval support
- **Classification: ICD** [International Classification of Diseases]:
  Strict taxonomy of non-overlapping classes for classifying statistically relevant health conditions
- **Ontology+Terminology: SNOMED CT**
  [Systematized Nomenclature of Medicine – Clinical Terms]:
  Hierarchical system of concepts with (partially) logic-based definitions for encoding medical records
- **Ontology: OBO Foundry** [Open Biomedical Ontologies]:
  Collection of orthogonal biomedical ontologies, mainly used for annotation of scientific data

What Biomedical Terminologies have in common

Natural language Terms / Labels

- Benign neoplasm of heart
- Benign tumor of heart
- Benign tumour of heart
- Benign cardiac neoplasm
- Gutartiger Herzumor
- Gutartige Neubildung am Herzen
- Gutartige Neubildung: Herz
- Gutartige Neoplasie des Herzens
- Tumeur bénigne cardiaque
- Tumeur bénigne du cœur
- Neoplasia cardíaca benigna
- Neoplasia benigna do coração
- Neoplasia benigna del corazón
- Tumor benigno do corazón

Hierarchically ordered Nodes and Links



Formal or informal Definitions

domain or region of DNA [GENIA]:
A substructure of DNA molecule which is supposed to have a particular function, such as a gene, e.g., c-jun gene, promoter region, Sp1 site, CA repeat. This class also includes a base sequence that has a particular function.

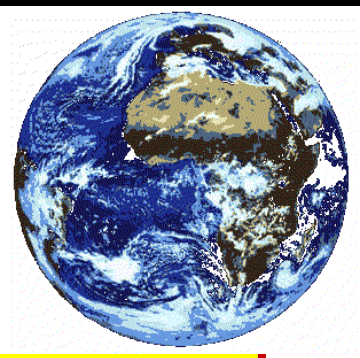
Peptides [MeSH]:
Members of the class of compounds composed of AMINO ACIDS joined together by peptide bonds between adjacent amino acids into linear, branched or cyclical structures. OLIGOPEPTIDES are composed of approximately 2-12 amino acids. Polypeptides are composed of approximately 13 or more amino acids. PROTEINS are linear polypeptides that are normally synthesized on RIBOSOMES.

```
19429009|chronic ulcer of skin|
116680003|is a|=64572001|disease|
    {116676008|associated morphology|=
        405719001|chronic ulcer|
     363698007|finding site|=
        39937001|skin structure|}
```

Purpose of this talk

- To give an overview of terminological system in biology and medicine
- To clarify the distinctions between
  - Terminologies / Thesauri
  - Ontologies
- To promote good ontological practice
- To contrast ontologies with classifications
- To address ontology aspects in ICF

Organizing the world



Terminology

Set of terms representing the system of concepts of a particular subject field. (ISO 1087)

Ontology

Ontology is the study of what there is. Formal ontologies are theories that attempt to give precise mathematical formulations of the properties and relations of certain entities. (Stanford Encyclopedia of Philosophy)

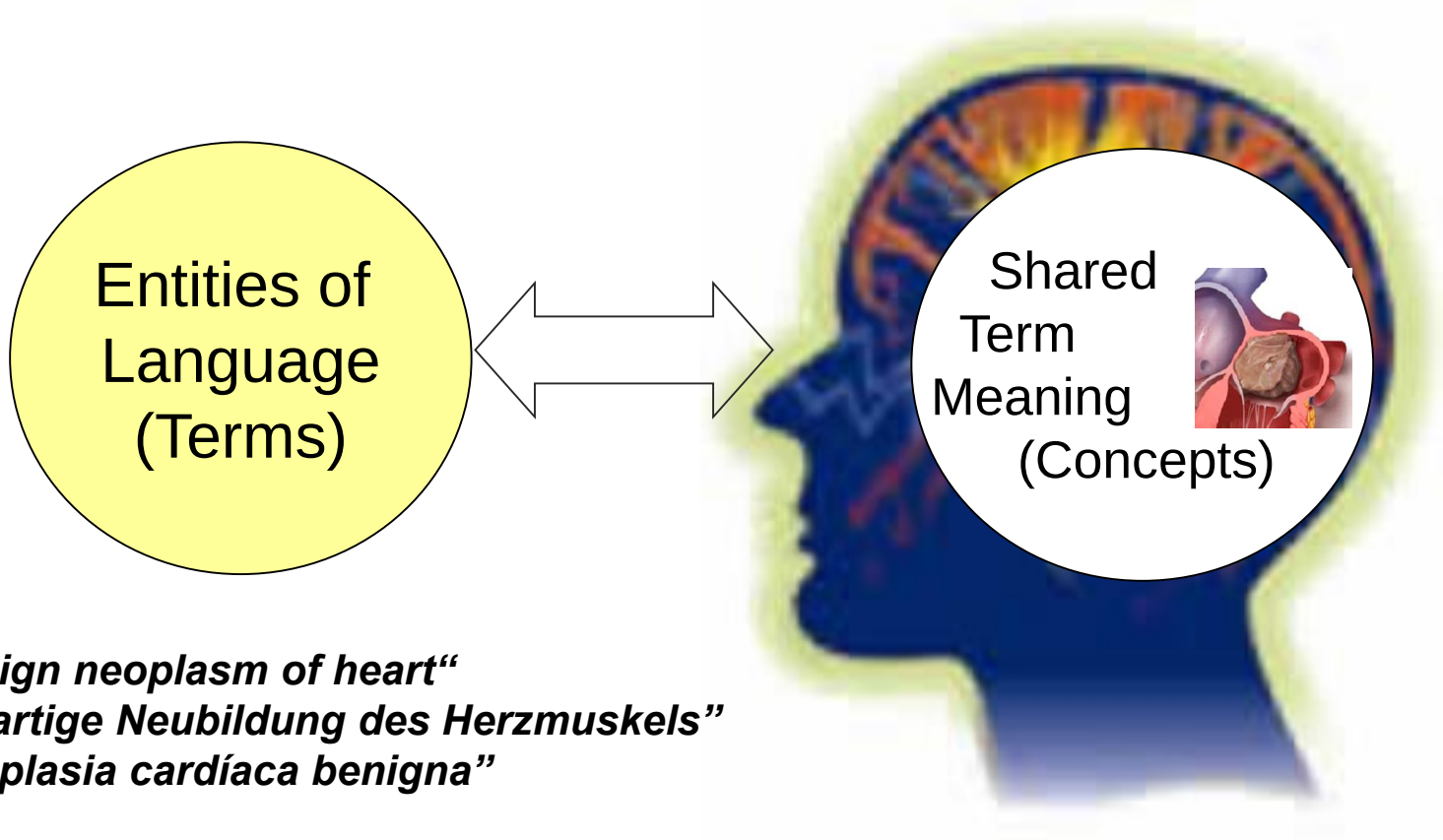
Terminologies start with human language

Terminology

Ontology

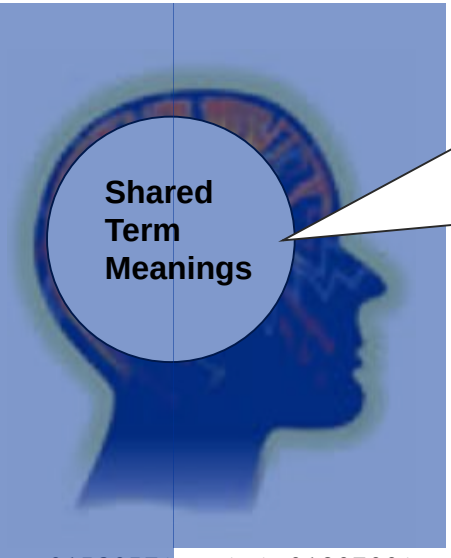
Set of terms representing the system of concepts of a particular subject field. (ISO 1087)

Ontology is the study of what there is. Formal ontologies are theories that attempt to give precise mathematical formulations of the properties and relations of certain entities. (Stanford Encyclopedia of Philosophy)



„benign neoplasm of heart“
„gutartige Neubildung des Herzmuskels”
“neoplasia cardíaca benigna”

Example: UMLS (mrconso table)

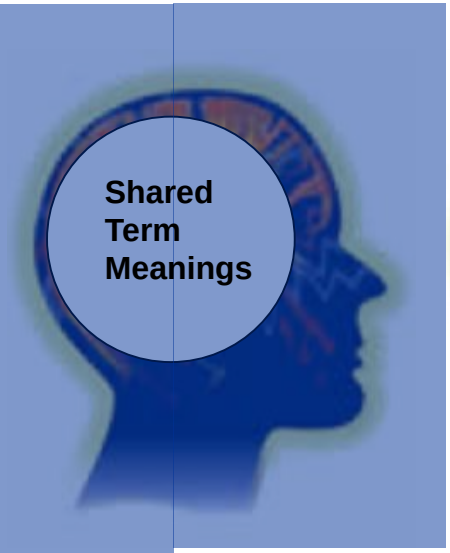


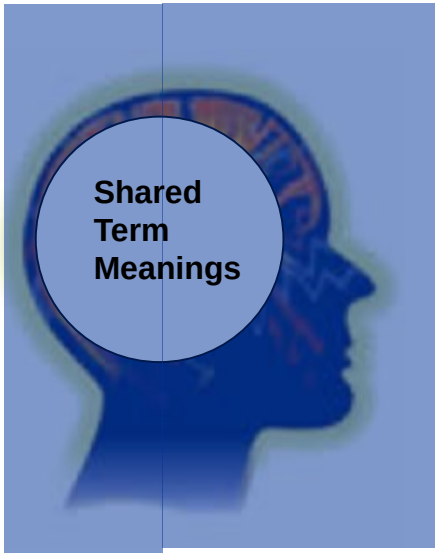
Entities of Language (Terms)

```
C0153957|ENG|P|L0180790|PF|S1084242|Y|A1141630||||MTH|PN|U001287|benign neoplasm of heart|0|N||
C0153957|ENG|P|L0180790|VC|S0245316|N|A0270815||||ICD9CM|PT| 212.7|Benign neoplasm of heart|0|N||
C0153957|ENG|P|L0180790|VC|S0245316|N|A0270817||||RCD|SY|B727.| Benign neoplasm of heart|3|N||
C0153957|ENG|P|L0180790|VO|S1446737|Y|A1406658||||SNMI|PT|    D3-F0100|Benign neoplasm of heart, NOS|3|N||
C0153957|ENG|S|L0524277|PF|S0599118|N|A0654589||||RCDAE|PT|B727.|Benign tumor of heart|3|N||
C0153957|ENG|S|L0524277|VO|S0599510|N|A0654975||||RCD|PT|B727.| Benign tumour of heart|3|N||
C0153957|ENG|S|L0018787|PF|S0047194|Y|A0066366||||ICD10|PS|D15.1|Heart|3|Y||
C0153957|ENG|S|L0018787|VO|S0900815|Y|A0957792||||MTH|MM|U003158|Heart <3>|0|Y||
C0153957|ENG|S|L1371329|PF|S1624801|N|A1583056|||10004245|MDR|LT|10004245|Benign cardiac neoplasm|3|N||
C0153957|GER|P|L1258174|PF|S1500120|Y|A1450314||||DMDICD10|PT| D15.1|Gutartige Neubildung: Herz|1|N||
C0153957|SPA|P|L2354284|PF|S2790139|N|A2809706||||MDRSPA|LT| 10004245|Neoplasia cardiaca benigna|3|N||
```

Unified Medical Language System, Bethesda, MD: National Library of Medicine: http://umlsinfo.nlm.nih.gov/

Example: UMLS

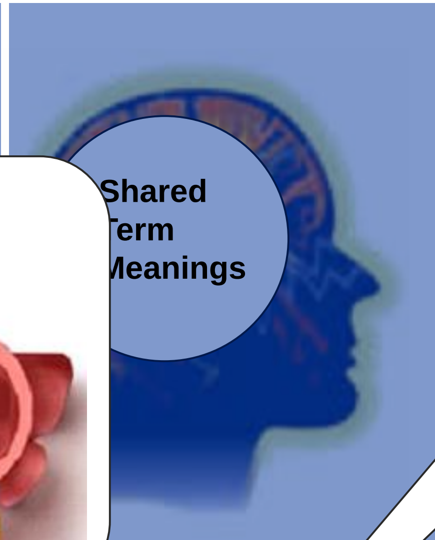


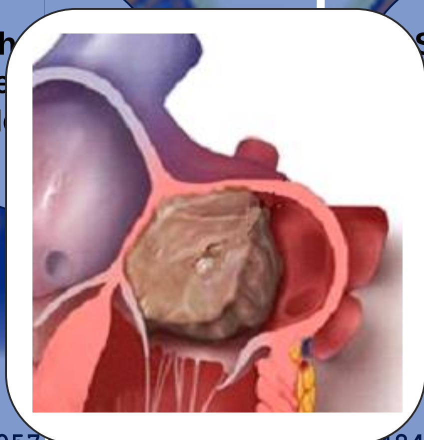


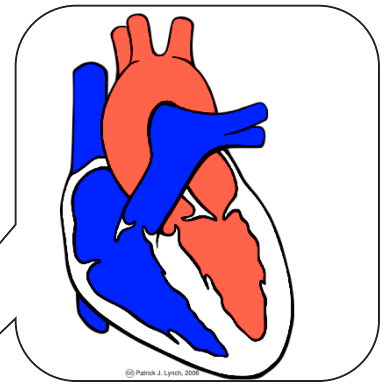

```
C0153957|A0066366|AUI|PAR|C0348423|A0876682|AUI  |                    |R06101405||ICD10|ICD10|||N||
C0153957|A0066366|AUI|RQ |C0153957|A0270815|AUI  |default_mapped_ from|R03575929||NCISEER|NCISEER|||N||
C0153957|A0066366|AUI|SY |C0153957|A0270815|AUI  |uniquely_mapped_ to |R03581228||NCISEER|NCISEER|||N||
C0153957|A0270815|AUI|RQ |C0810249|A1739601|AUI  |classifies          | R00860638||CCS|CCS|||N||
C0153957|A0270815|AUI|SIB|C0347243|A0654158|AUI  |                    |R06390094          || ICD9CM|ICD9CM||N|N||
C0153957|A0270815|CODE|RN|C0685118|A3807697|SCUI |mapped_to           | R15864842||SNOMEDCT|SNOMEDCT||Y|N||
C0153957|A1406658|AUI|RL |C0153957|A0270815|AUI  |mapped_from         | R04145423||SNMI|SNMI|||N||
C0153957|A1406658|AUI|RO |C0018787|A0357988|AUI  |location_of         | R04309461||SNMI|SNMI|||N||
C0153957|A2891769|SCUI|CHD|C0151241|A2890143|SCUI|isa                |R19841220|47189027|SNOMEDCT|SNOMEDCT|0|Y|N||
```

Semantic relations

Example: UMLS







```
C0153957|A...|AUI|PAR|C0348423|A0876682|AU...  |                    |R06101405||ICD10|ICD10|||N||
C0153957|...66|AUI|RQ |C0153957|A0270815|...I  |default_mapped_ from|R03575929||NCISEER|NCISEER|||N||
C0153957...6366|AUI|SY |C0153957|A02708..|AUI  |uniquely_mapped_ to |R03581228||NCISEER|NCISEER|||N||
C015395...0270815|AUI|RQ |C0810249|A173..01|AUI |classifies          | R00860638||CCS|CCS|||N||
C01539..|A0270815|AUI|SIB|C0347243|A0..4158|AUI  |                    |R06390094          || ICD9CM|ICD9CM||N|N||
C0153..7|A0270815|CODE|RN|C0685118|..3807697|SCUI |mapped_to          | R15864842||SNOMEDCT|SNOMEDCT||Y|N||
C015.957|A1406658|AUI|RL |C015395.|A0270815|AUI  |mapped_from         | R04145423||SNMI|SNMI|||N||
C0153957|A1406658|AUI|RO |C0018787|.0357988|AUI  |location_of         | R04309461||SNMI|SNMI|||N||
C0153957|A2891769|SCUI|CHD|C0151241|A2890143|SCUI|isa                 |R19841220|47189027|SNOMEDCT|SNOMEDCT|0|Y|N||
```

INFORMAL

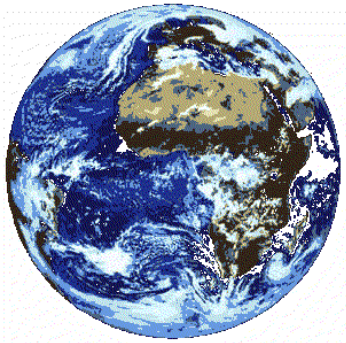
Semantic relations

Formal Ontology represents the world

ology

Set of terms representing the system of concepts of a particular subject field. (ISO 1087)

Ontology

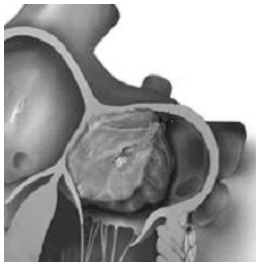


Ontology is the study of what there is (Quine).
Formal ontologies are theories that attempt to give precise mathematical formulations of the properties and relations of certain entities.
(Stanford Encyclopedia of Philosophy)

Ontology

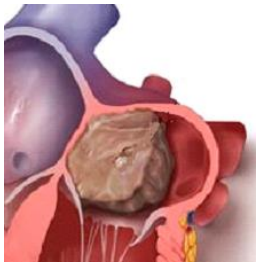
Entity Types

The type “benign neoplasm of heart”

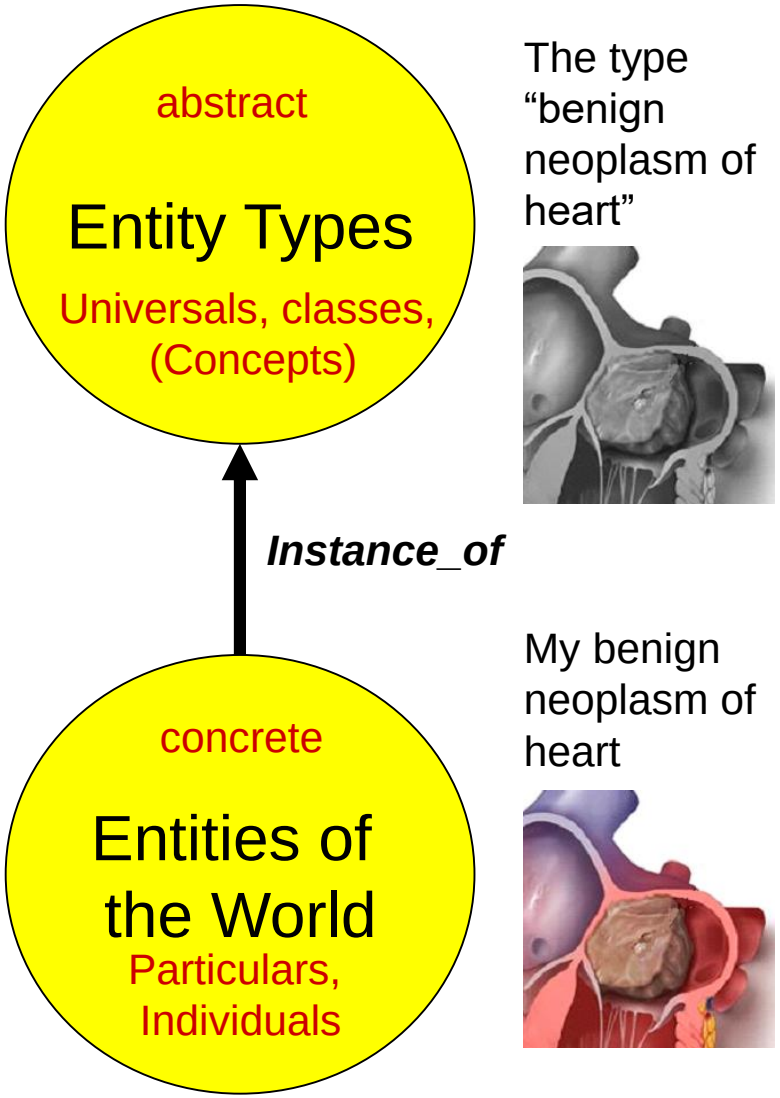


Entities of the World

My benign neoplasm of heart



Ontology

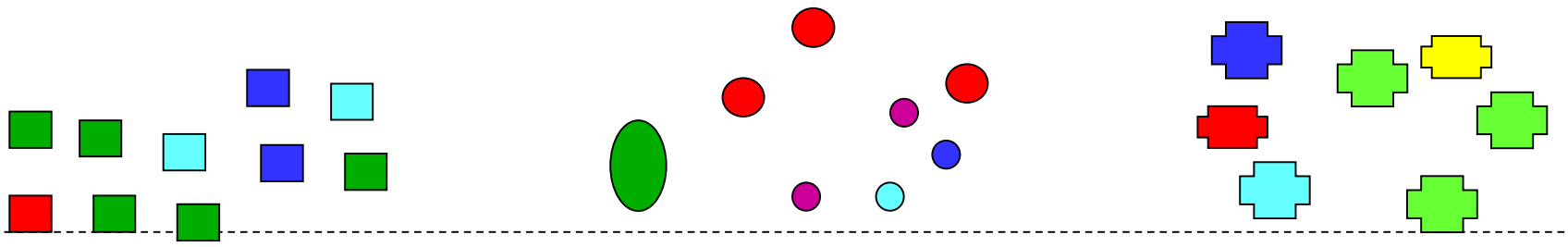

abstract
Entity Types
Universals, classes, (Concepts)
The type "benign neoplasm of heart"
Instance_of
concrete
Entities of the World
Particulars, Individuals
My benign neoplasm of heart

Hierarchical framework for ontologies

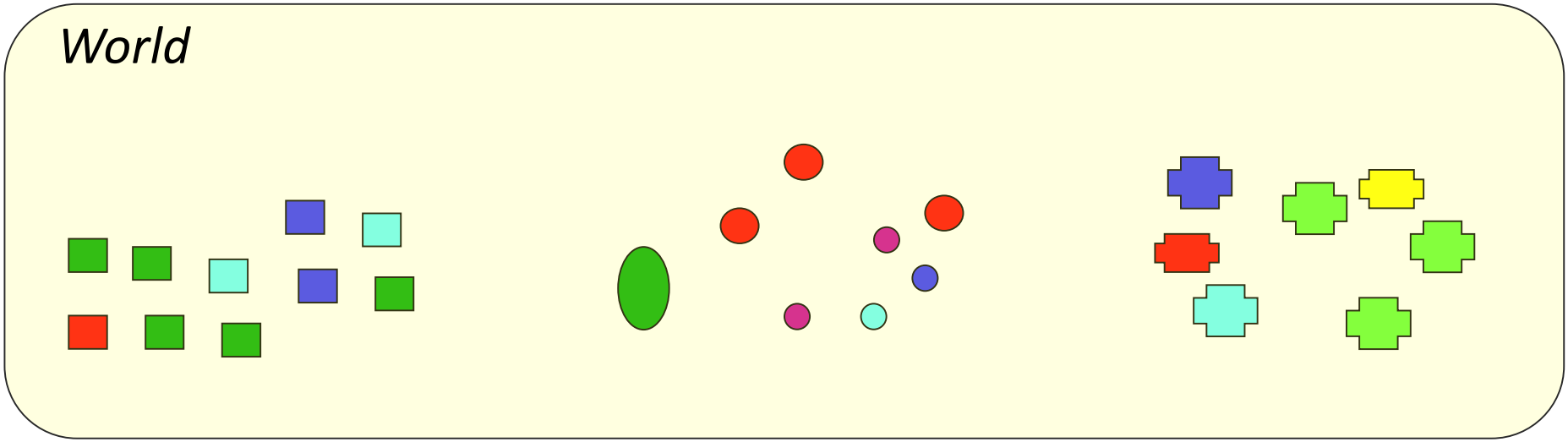
- Taxonomy: relates types and subtypes:
  - *Tumor of Heart subClassOf Tumor* equivalent to:
  - All instances of *Tumor of Heart* are instances of *Tumor* (without exceptions)
- Relations:
  - *instance_of* relates individuals with types, all others relate individuals (e.g. *part_of*) or are derived from them (e.g. *is_a*)
- Definitions: describe what is <u>always</u> true for <u>all</u> individuals that instantiate a type
  - *Tumor of Heart subClassOf has_location some Heart* :
    All instances of *Tumor of Heart* are located in some *Heart*

Hierarchies, Types, Classes, Individuals

World



Hierarchies, Types, Classes, Individuals

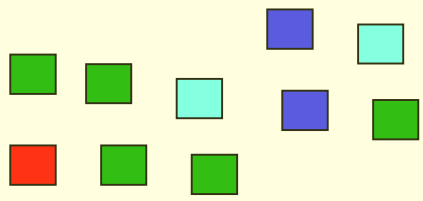


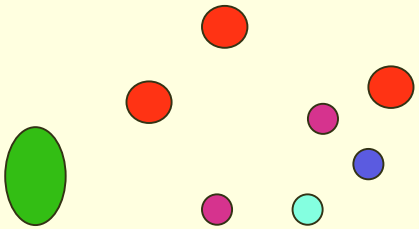
Hierarchies, Types, Classes, Individuals

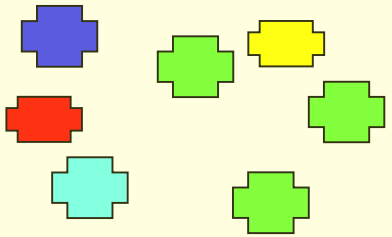
Ontology

Type 1

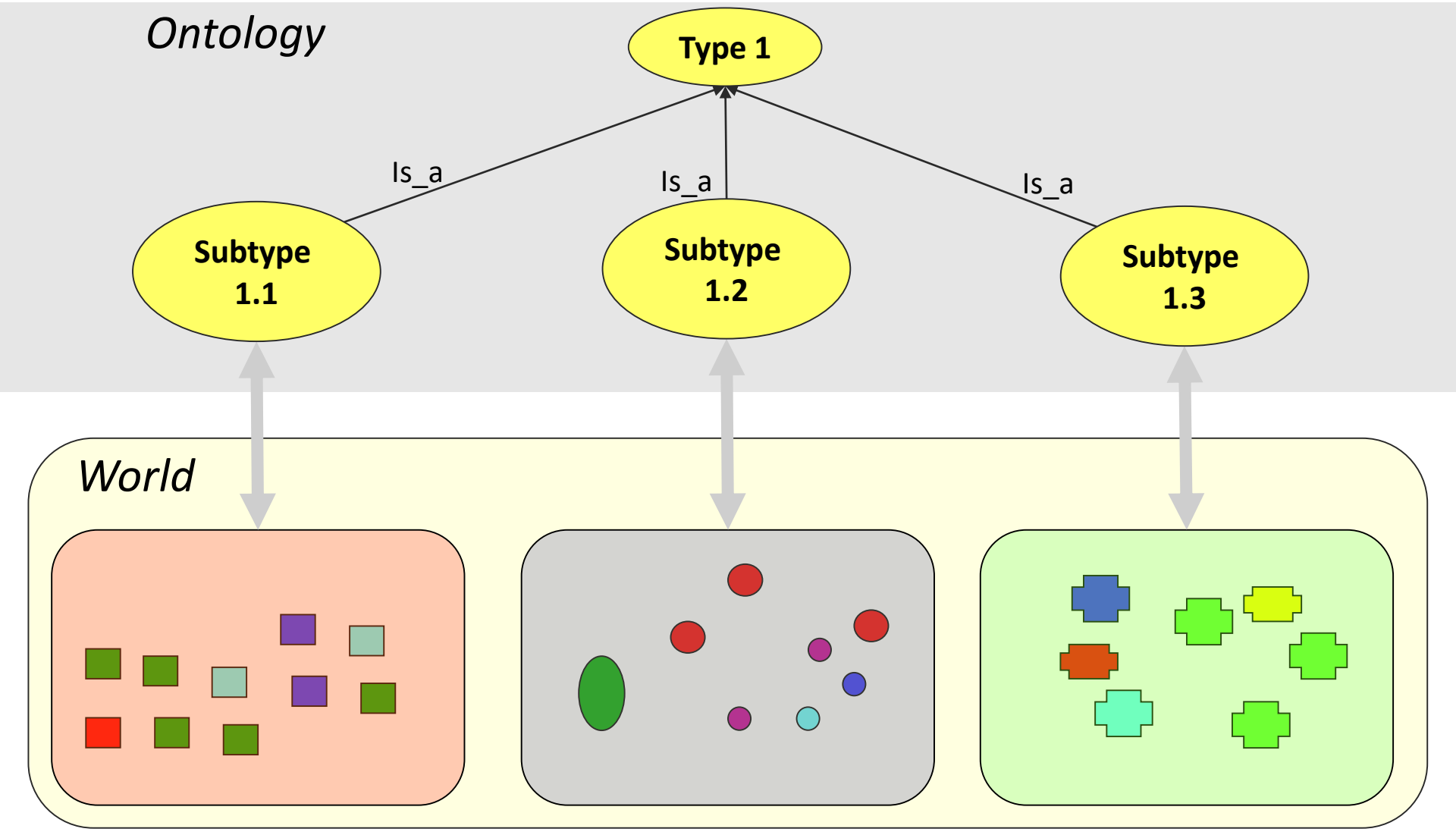
World



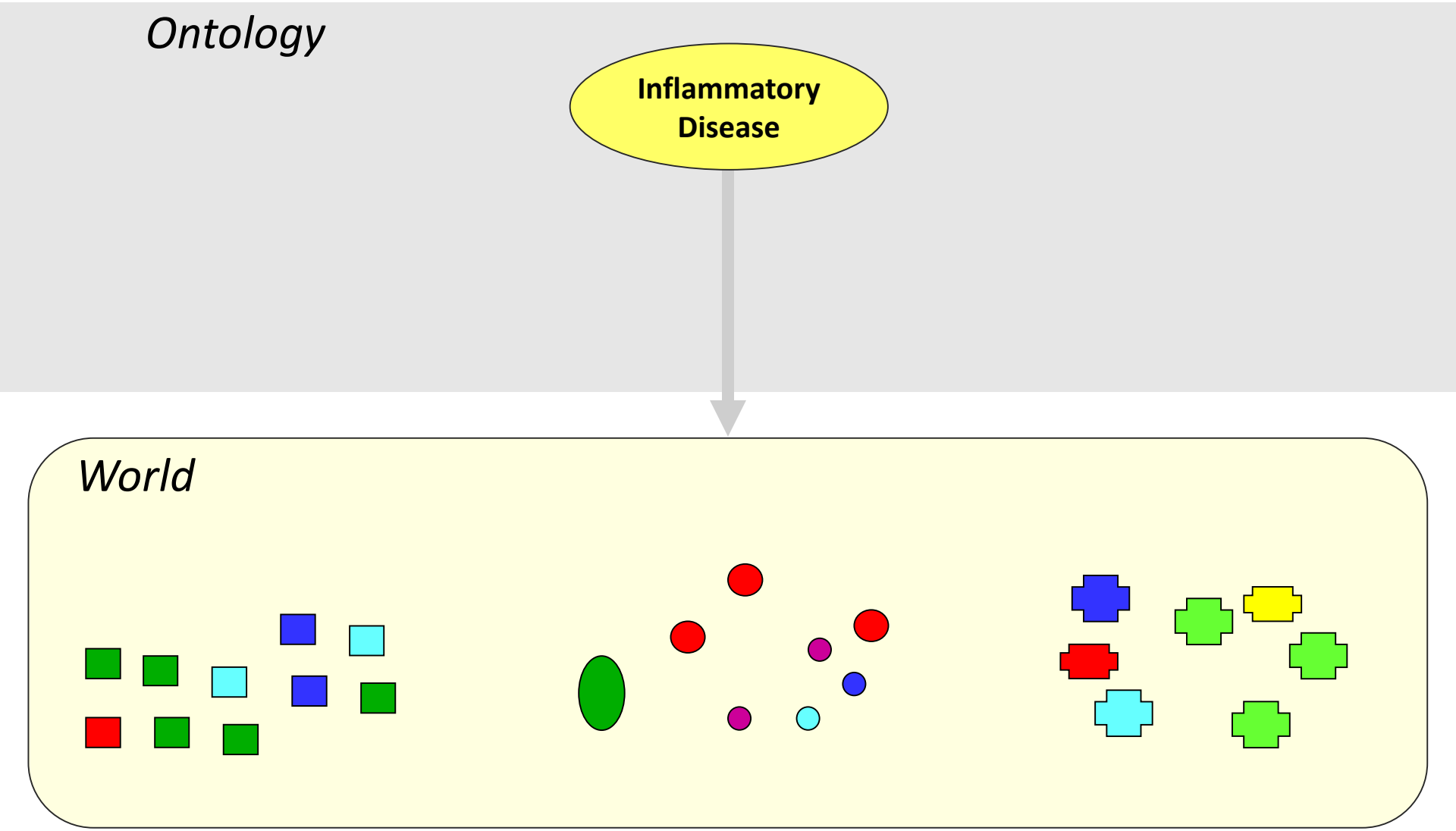




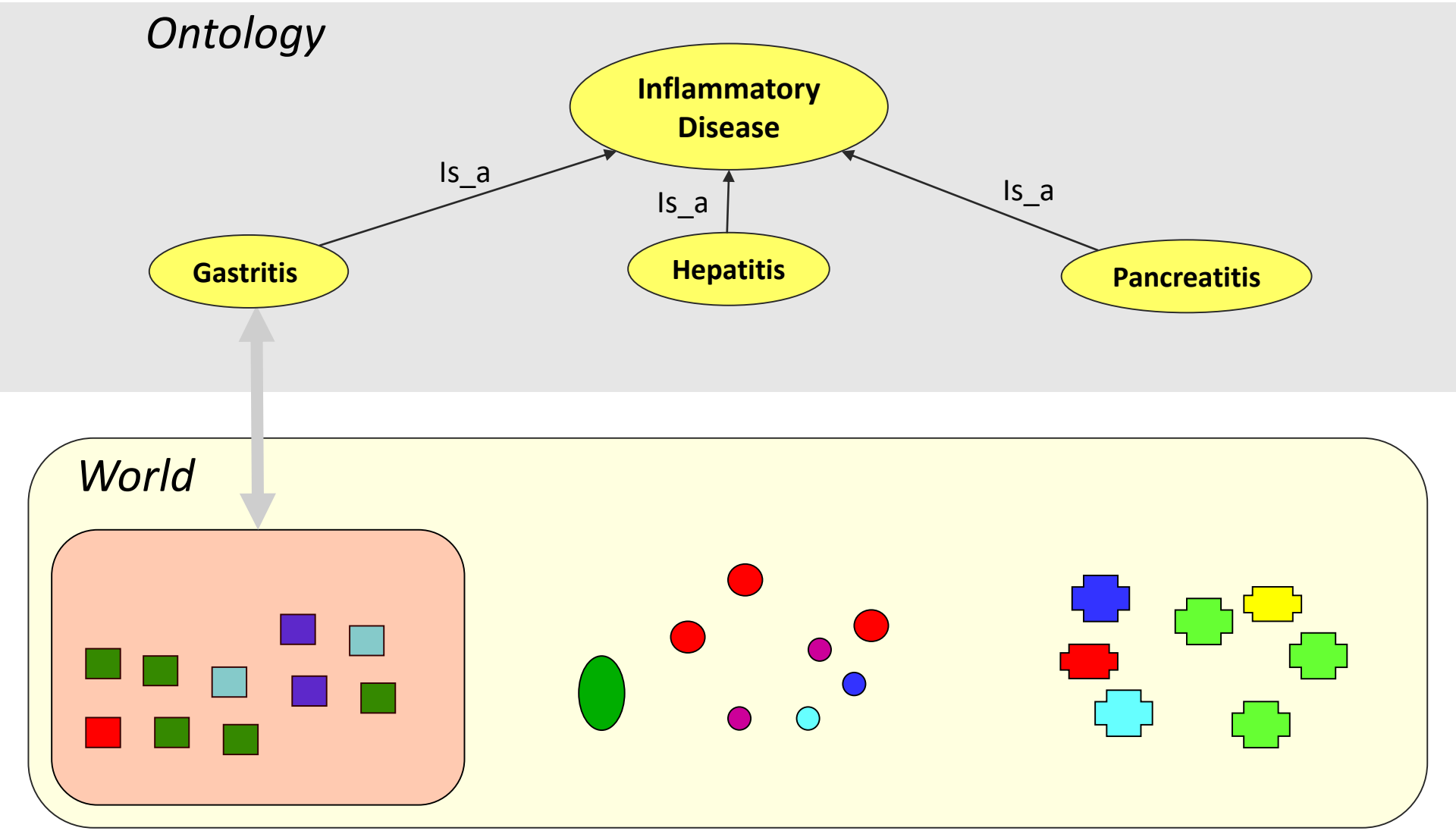
Hierarchies, Types, Classes, Individuals



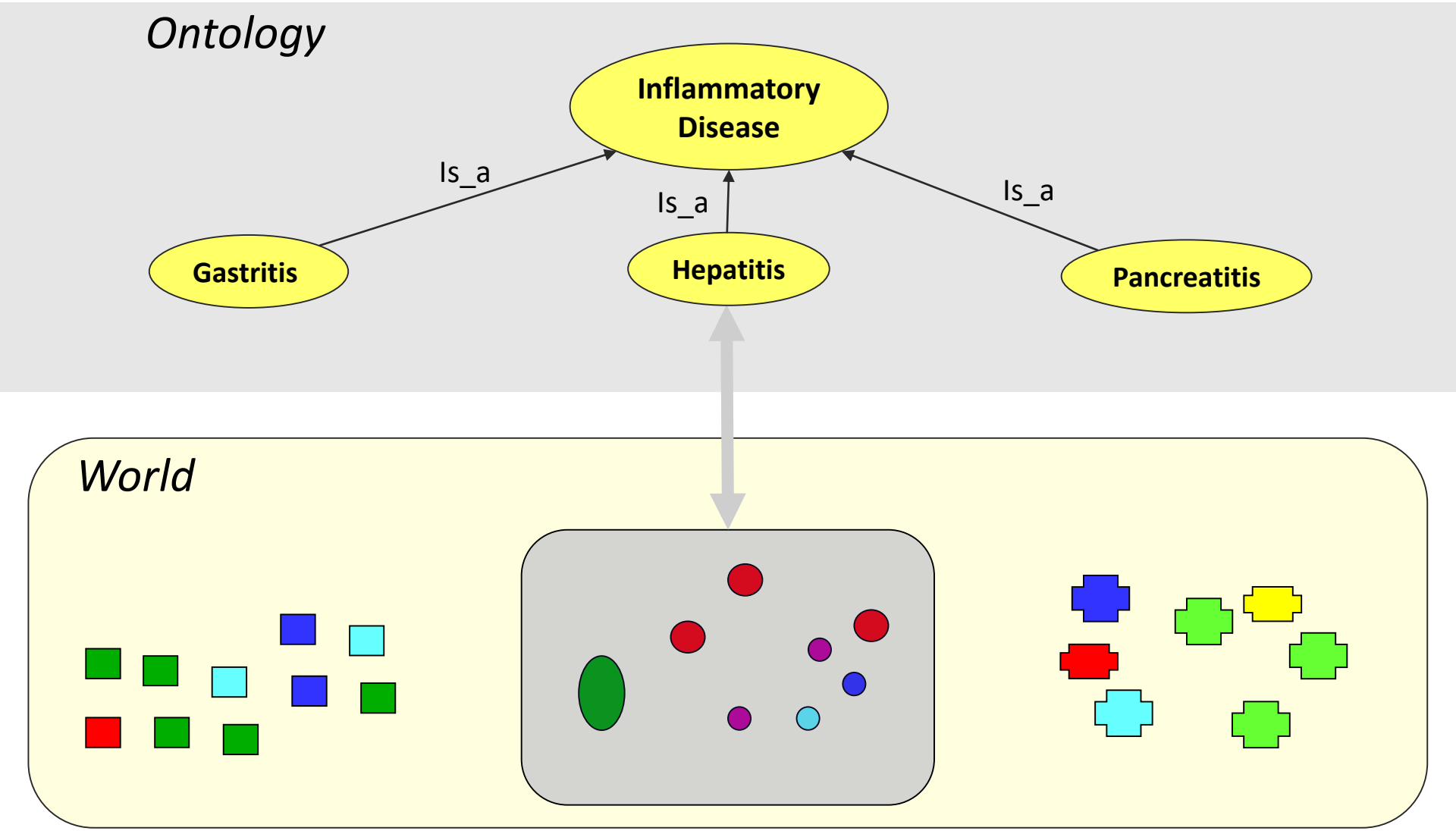
Hierarchies, Types, Classes, Individuals



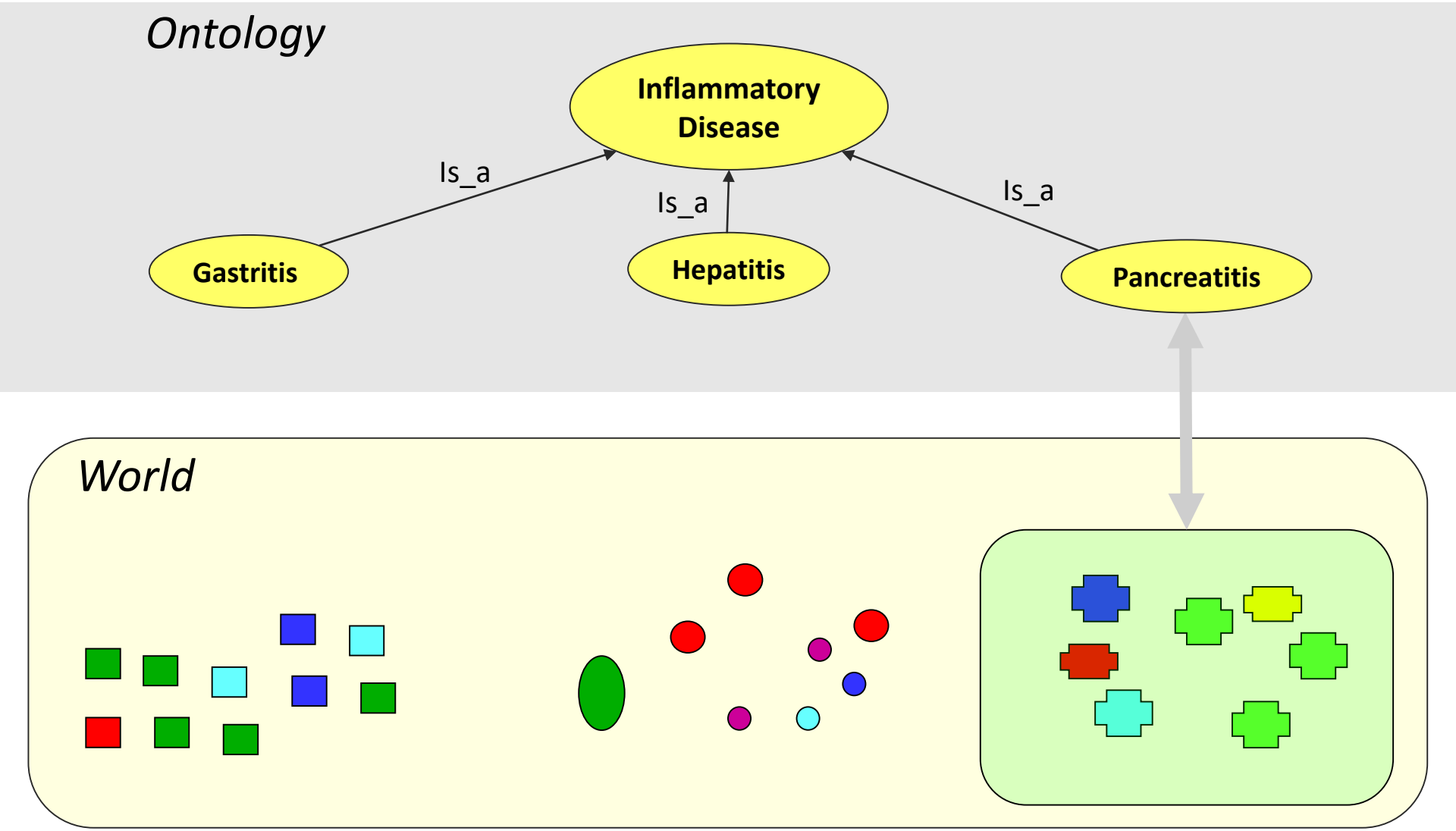
Hierarchies, Types, Classes, Individuals



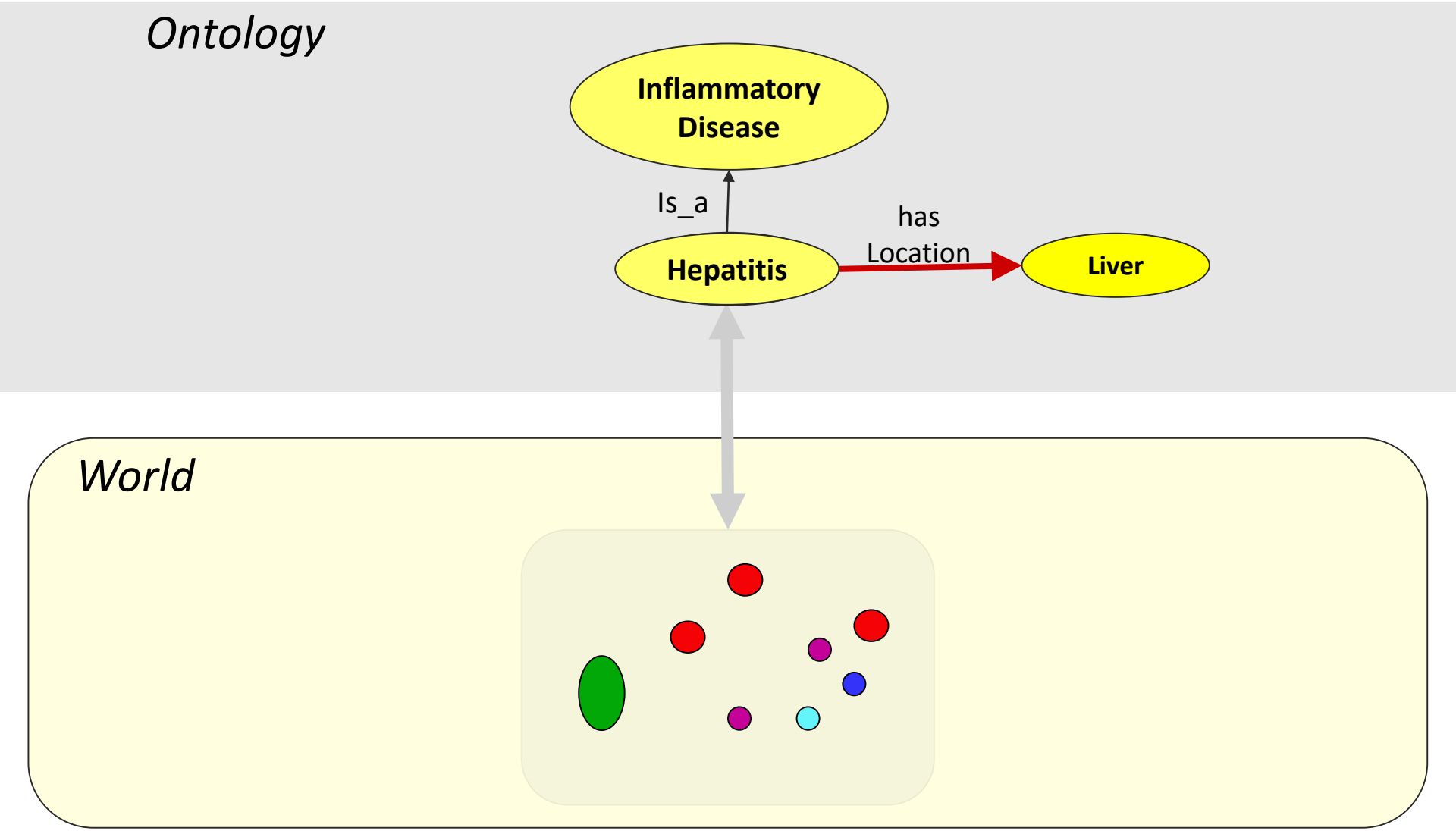
Hierarchies, Types, Classes, Individuals



Hierarchies, Types, Classes, Individuals

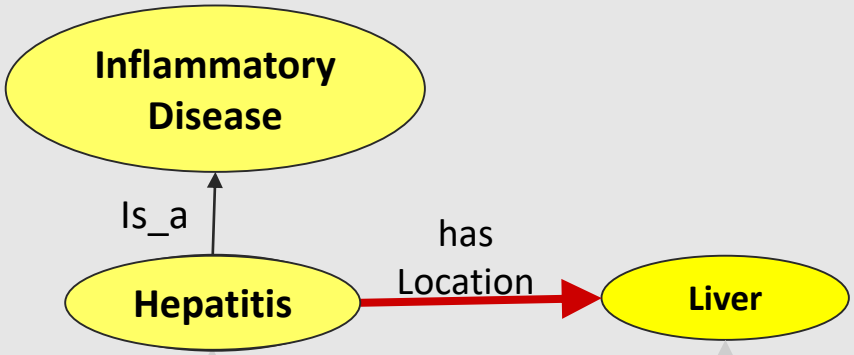


Relations and Definitions

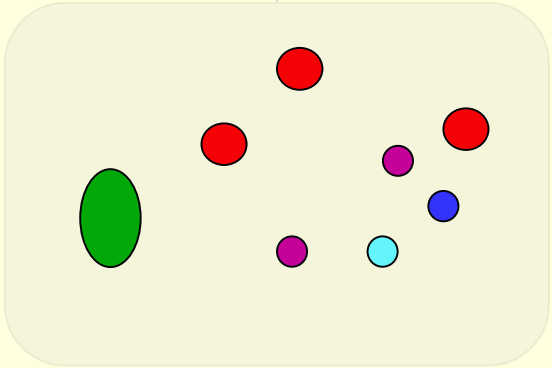


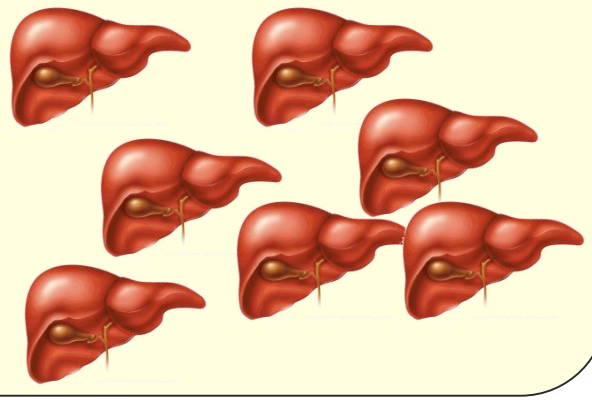
Relations and Definitions

Ontology



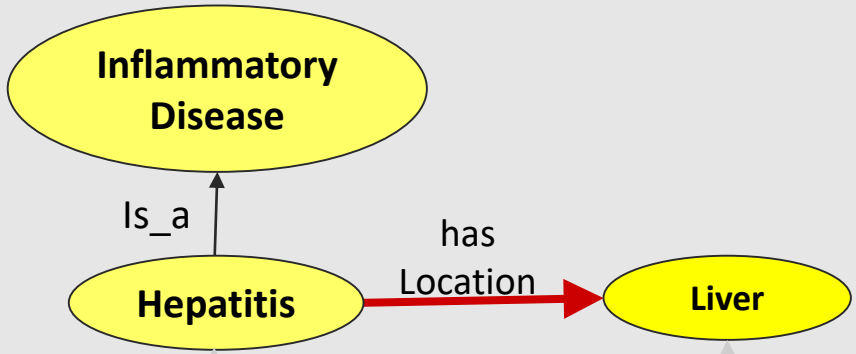
World



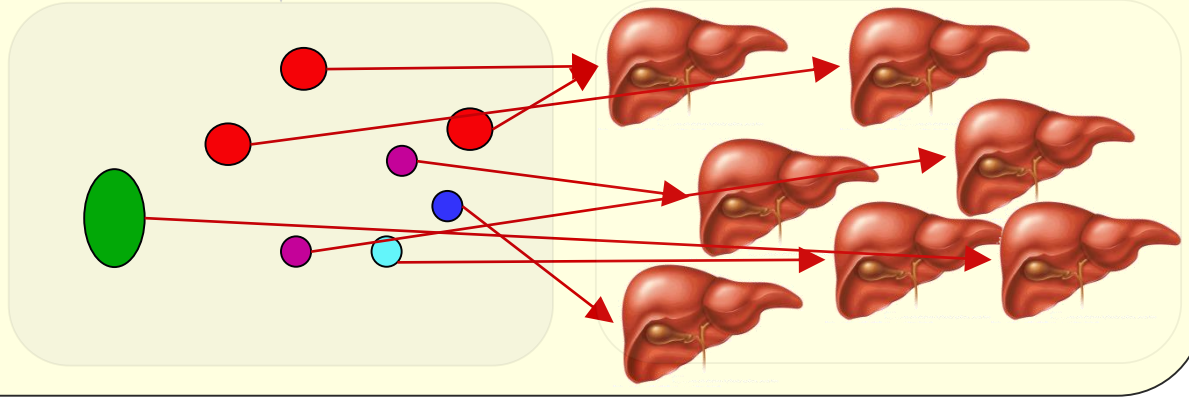


Relations and Definitions

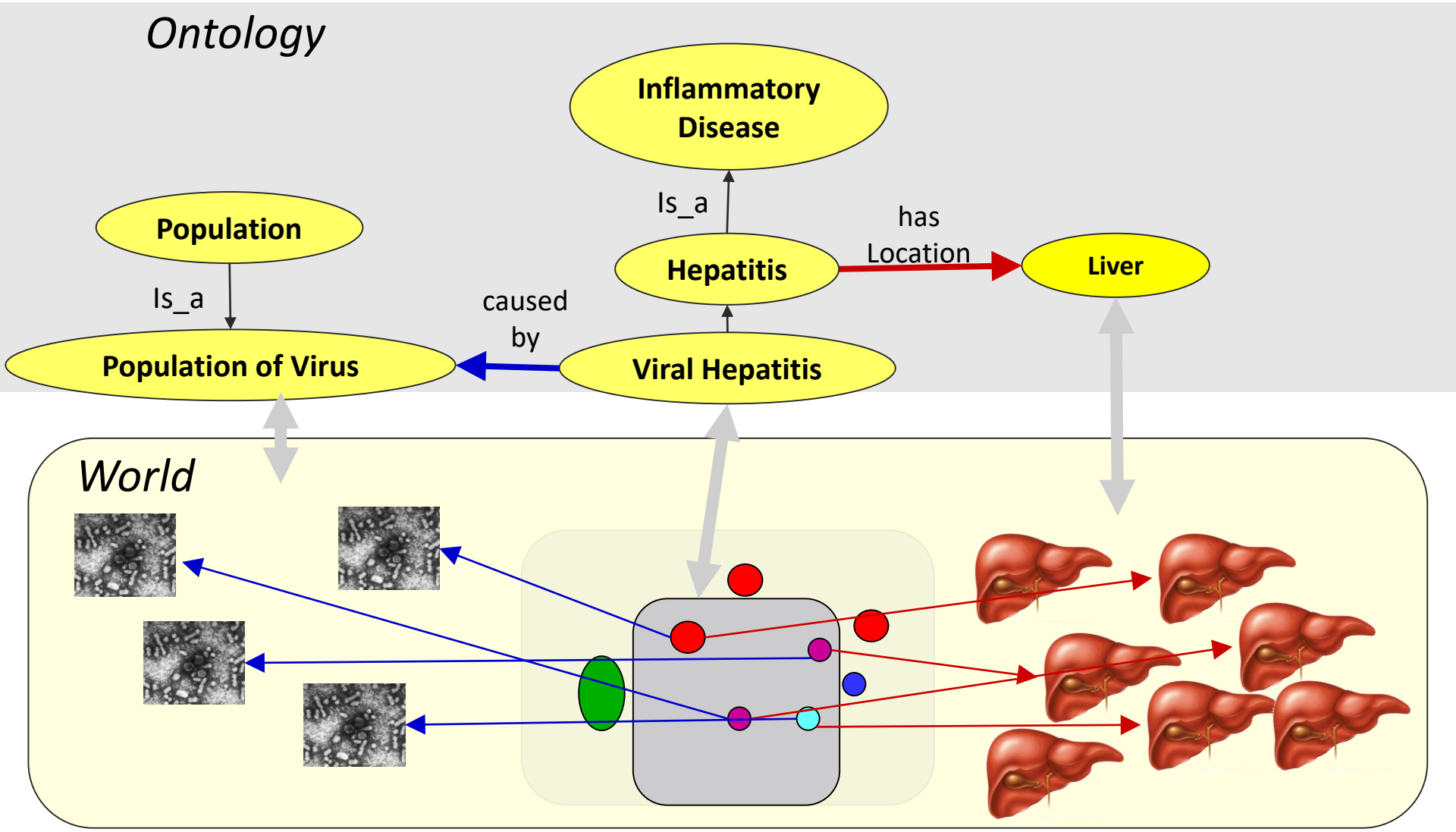
Ontology



World



Relations and Definitions



Languages for formal ontologies

- Natural Language:

  "Every hepatitis is an inflammatory disease that is located in some liver"
  "Every inflammatory disease that is located in some liver is an hepatitis"

- First Order Logic:

$$\forall x: instanceOf(x, Hepatitis) \Leftrightarrow instanceOf(x, Inflammation) \wedge \exists y: instanceOf(y, Liver) \wedge hasLocation(x,y)$$

- Description Logics:

$$Hepatitis \equiv Inflammation \sqcap \exists hasLocation.Liver$$

Logic is computable: it supports machine inferences but…

it only scales up if it has a very limited expressivity

OWL – Ontology Web Language

- Semantic Web standard for ontologies
  - OWL 2.0 provides three different levels of expressiveness
- Based on Description Logics
- Popular editing tools available (Protégé)
- Classifiers: Fact++, Racer, Pellet, HermiT
- Increasingly used in OBO Foundry ontologies as a primary format (already available as export format)
- Most SNOMED CT expressible in OWL

OWL – What can sensibly be expressed

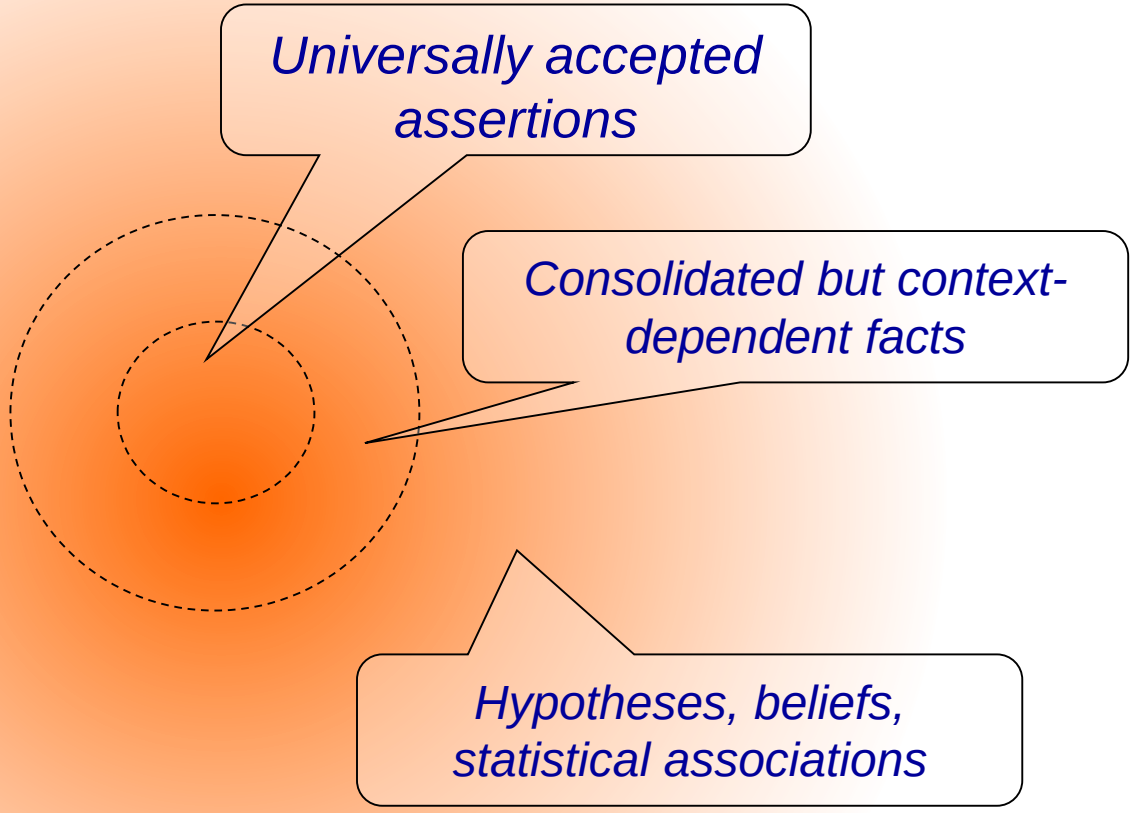
- Only suitable to represent shared, uncontroversial meaning of a domain vocabulary
- Supports universal statements about instances of a type:
  - All Xs are Ys
  - For all Xs there is some Y
- Properties of types are properties of all entities that instantiate these types (strict inheritance)

OWL – What cannot be expressed

- Context dependent knowledge
  - *„Allergic Rhinitis is a common disorder (in Europe)“*
- Probabilistic knowledge
  - *„95% of people infected with viral hepatitis recover “*
  - *“Smoking is a cardiovascular risk factor”*
- Default / canonic knowledge
  - *„Adult humans have 32 teeth“*
- Meta-classes (instances of instances), e.g.
  - *Clyde subClassOf Elephant subClassOf Species*
    (“punning” not expressible in description logics)
- Non quantified relations between classes
  - *Treats(Aspirin, Headache)*

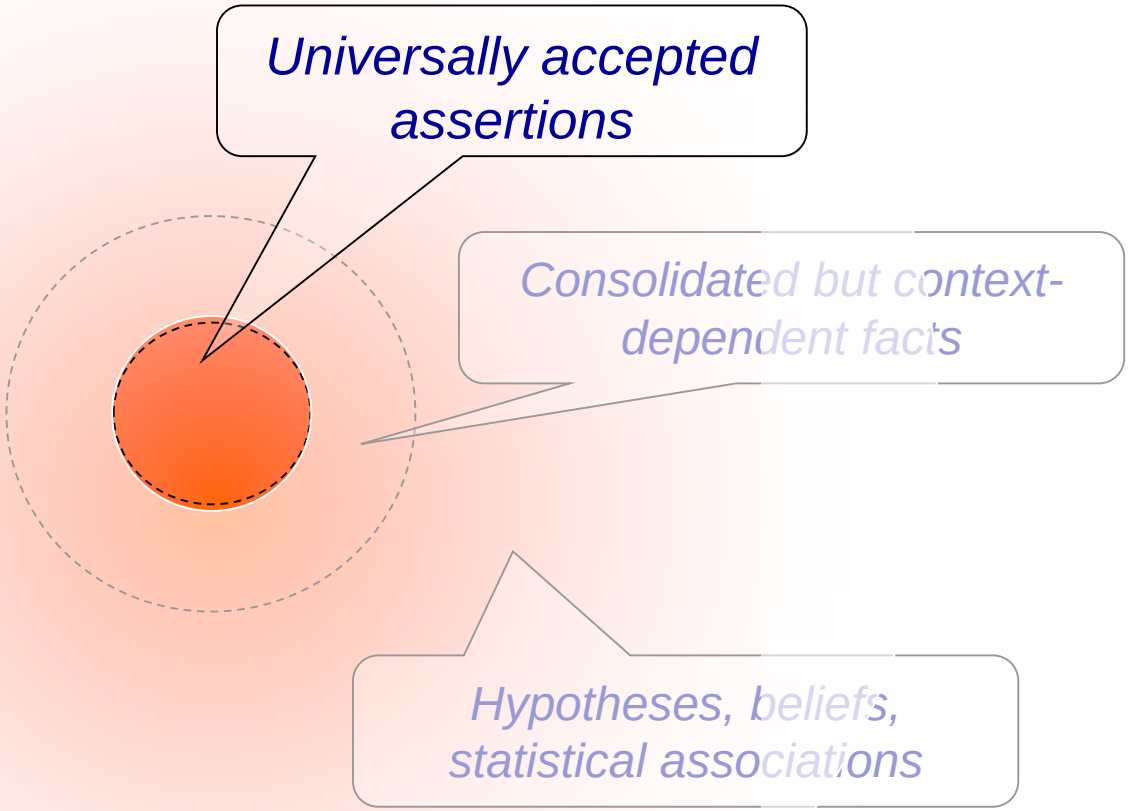
Ontology ⊂ Knowledge Representation

Continuum of knowledge



Domain Knowledge

Ontology !

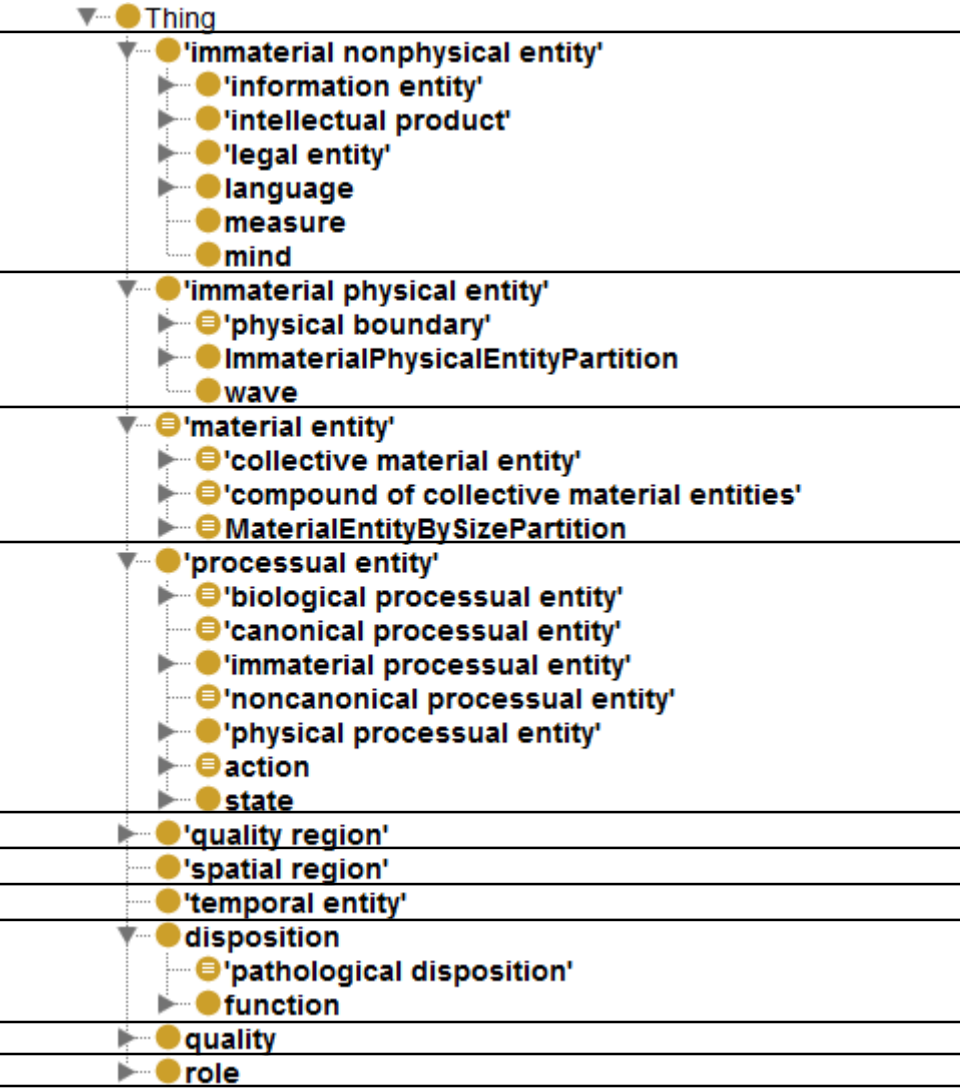


Domain Knowledge

Purpose of this talk

- To give an overview of terminological system in biology and medicine
- To clarify the distinctions between
  - Terminologies / Thesauri
  - Ontologies
- To promote good ontological practice
- To contrast ontologies with classifications
- To address ontology aspects in ICF

Partition the ontology by principled upper level categories



Mutually disjoint Upper Level Categories in BioTop

http://purl.org/biotop

Other (domain independent) toplevel ontologies:
- DOLCE
- BFO
- GFO

Beisswanger E., Stenzhorn H., Schulz S., Hahn U; BIOTOP: An upper domain ontology for the life sciences. A description of its current structure, contents, and interfaces to OBO ontologies; Applied Ontology; 2008; 3(4): 205-212

Limit to a parsimonious set of semantically precise Basic Relations

First version of the OBO Relation Ontology

Foundational relations

is_a

part_of

Spatial relations (connecting one entity to another in terms of relations between the spatial regions they occupy)

located_in

contained_in

adjacent_to

Temporal relations (connecting entities existing at different times)

transformation_of

derives_from

preceded_by

Participation relations (connecting processes to their bearers)

has_participant

has_agent

Barry Smith, Werner Ceusters, Bert Klagges, Jacob Köhler, Anand Kumar, Jane Lomax, Chris Mungall, Fabian Neuhaus, Alan L Rector and Cornelius Rosse. Relations in biomedical ontologies. *Genome Biology*, 6(5), 2005.

Don’t use superclasses to express roles

- *Is_a (Fish, Animal)*
- *Is_a (Fish, Food)* ??

- *Is_a (Acetylsalicylic Acid, Salicylate)*
- *Is_a (Acetylsalicylic Acid, Analgetic Drug)* ??

Be aware of the “rigidity” of entity types (distinguishing categories from roles)

Guarino, N. Welty, C.A. 2008) An overview of ONTOCLEAN (2008) In Staab, S. Studer, R. Eds. Handbook on Ontologies, International Handbooks on Information Systems

Don’t be mislead by natural language expressions

- *Is_a (right Hand, Hand)*
- *Is_a (planned Endoscopy, Endoscopy) ??*
- *Is_a (prevented Pregnancy, Pregnancy) ??*

Be aware of the “ontological commitment”

- *It must be clear whether “Endoscopy” means*
  - *a record about an endoscopy encompassing planning and execution: The record exists even if the plan is never executed*
  - *the endoscopy itself*

Schulz S, Cornet R: SNOMED CT’s Ontological Commitment. 2009: 111-114 (ICBO: International Conference on Biomedical Ontology, 2009, Buffalo, New York, USA): http://icbo.buffalo.edu/Proceedings.pdf

Be aware of ambiguities

- "*Institution*" may refer to
  1. (abstract) institutional rules
  2. (concrete) things instituted
  3. act of instituting sth.
- "*Tumor*"
  1. evolution of a tumor as a disease process
  2. having a tumor as a pathological state
  3. tumor as a physical object

The same term may have different meanings, which may require different (disjoint) classes in an ontology

Don‘t mix up ontology with epistemiology

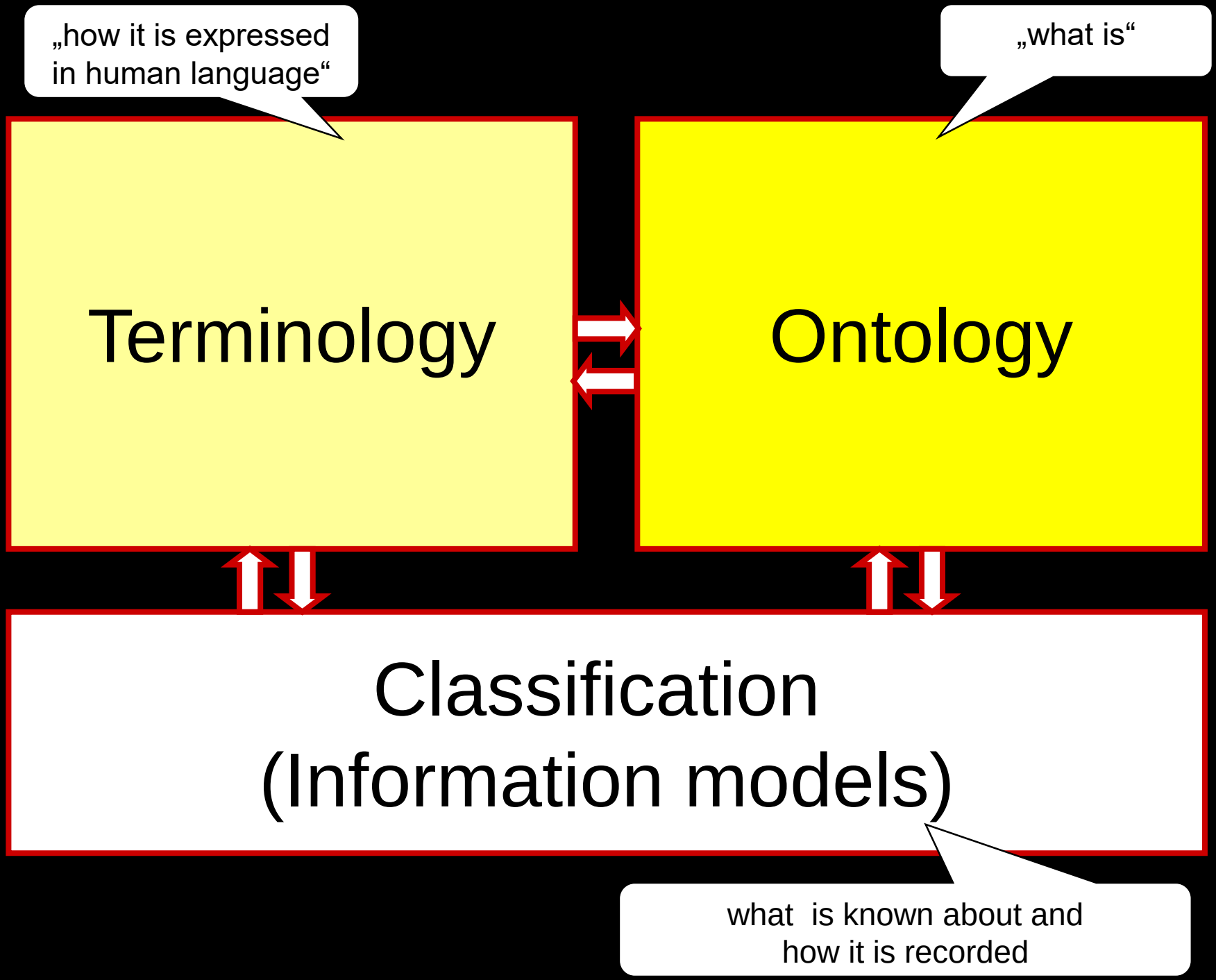
- *Is_a (Infection of unknown origin; Infection)*
- *Is_a (Newly diagnosed diabetes; Diabetes)*
- *Is_a (Family history of diabetes; Diabetes)*
- *Is_a (Diabetes NOS; Diabetes)*
- *Is_a (Gender, unknown; Gender)*

Ontology = what there is
Epistemology = what is known

It is important to record both things, but an ontology, in a strict sense, is not the right artifact. We need an information model linked to an ontology

Purpose of this talk

- To give an overview of terminological system in biology and medicine
- To clarify the distinctions between
  - Terminologies / Thesauri
  - Ontologies
- To promote good ontological practice
- To contrast ontologies with classifications
- To address ontology aspects in ICF


„how it is expressed in human language“
„what is“
Terminology
Ontology
Classification (Information models)
what is known about and how it is recorded

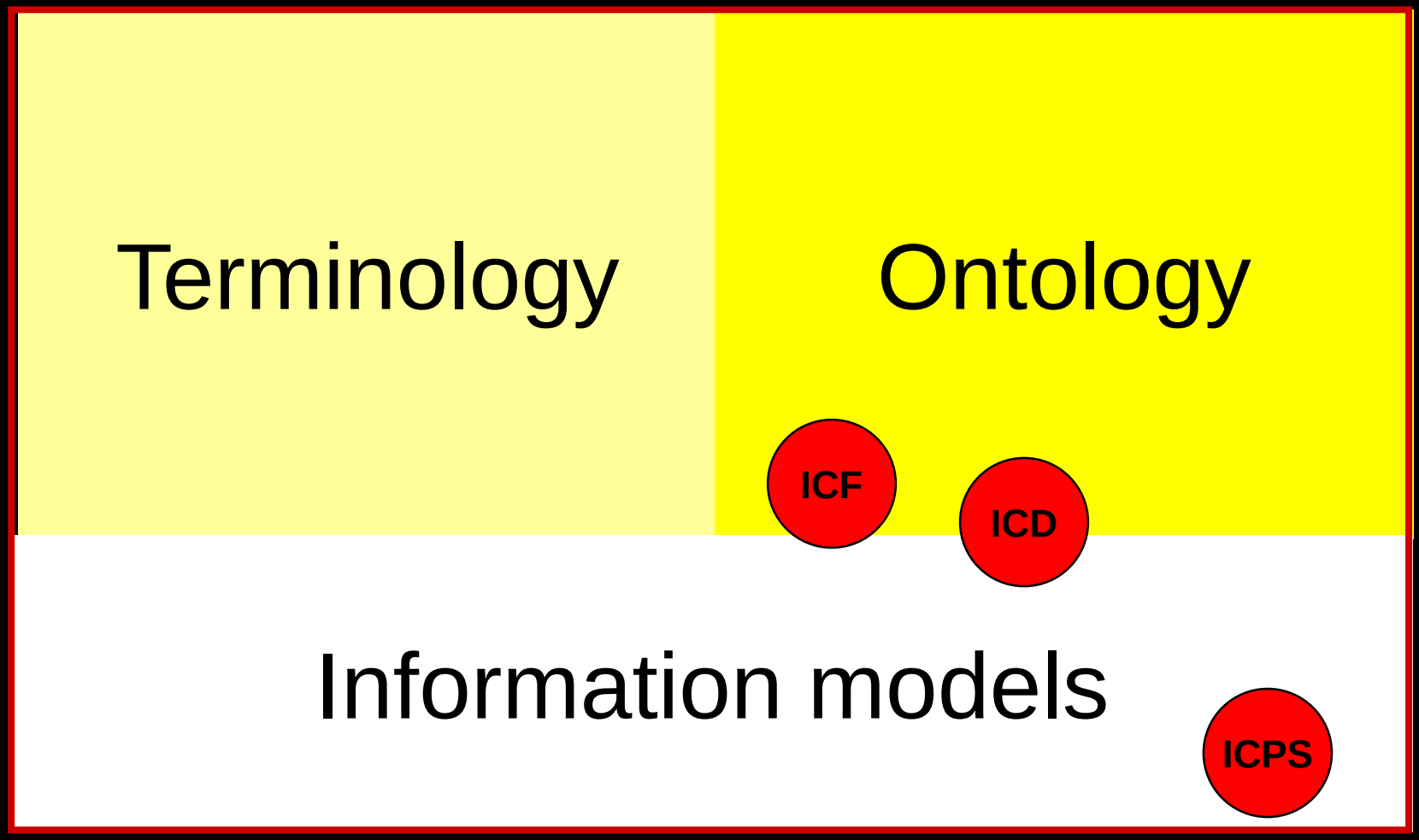
Ontologies vs. Classifications

Ontologies	**Classifications**
Nodes correspond to classes of individual entities	
Hierarchies are strict subclass hierarchies	
expressible in description logics	
Classes correspond (ideally) to natural kinds), multiple parenthood is natural (at least in the inferred ontologies) (e.g. Diabetes mellitus class in SNOMED classifies all diabetes mellitus individuals)	Classes are mutually disjoint, hence most classes with idiosyncratic delineations (e.g. Diabetes mellitus class in ICD-10 does not classify all diabetes mellitus individuals)
The definition of classes is (ideally) independent of the context of use	The meaning of class membership is highly independent on the context of use
Classes are context-independent and do not include epistemic aspects	Classes sometimes fuse the entity with the knowledge about the entity
Residual classes (NOS, NEC) not permitted	Residual classes (NOS, NEC) important for maintaining the disjointness principle

Ontologies vs. Classifications

- Open questions:
  - Are the abovementioned criteria for classifications still valid for WHO FIC classifications?
  - Are future classifications mainly information models, i.e. strict context-dependent linear data acquisition models?
  - Example: The International Classification of Patient Safety (ICPS) does not fulfill “traditional” classification principles

Schulz S, Karlsson D, Daniel C, Cools H, Lovis C: Is the "International Classification for Patient Safety" a classification? In: Adlassnig K-P, Blobel B, Mantas J, Masic I (Hrsg.): Medical Informatics in a United and Healthy Europe - Proceedings of MIE 2009 – The XXIInd International Congress of the European Federation for Medical Informatics Amsterdam: IOS Press Books Online, 2009; 502-506.


Terminology
Ontology
ICF
ICD
Information models
ICPS

Purpose of this talk

- To give an overview of terminological system in biology and medicine
- To clarify the distinctions between
  - Terminologies / Thesauri
  - Ontologies
- To promote good ontological practice
- To contrast ontologies with classifications
- To address ontology aspects in ICF

ICF under ontology scrutiny

- Resources: BioTop upper ontology, compatible with BFO, DOLCE, and OBO Relation Ontology
- Methods: Find appropriate upper level classes that subsume ICF classes

Body function

- *ICF:BodyFunction: subClassOf biotop:Disposition*
- Definition of *biotop:Disposition*:
  A realizable entity. Its manifestation is a process its bearer is involved in virtue of the bearer's physical make-up.
- The specific characteristic of disposition is that they exist even unrealized. E.g. an organism has a function to procreate even if this function is never realized
- The relation *has realization* (inverse *realization of*) links a function to a process
- The relation *inheres in* (inverse *bearer of*) links a function to the entity which has the function

Body function: problems found

- *ICF:Pain* is a subclass of *ICF:body function*. This is not correct, because pain is a process. A process cannot be a function: Processes have temporal parts, functions haven't. Processes happen, functions inhere. There could be a related function such as pain sensitivity but this is different from pain. It does not make sense to say that a pain is "realized"
- *ICF:Voice quality* is a subclass of *ICF:body function*. Qualities are different from functions because they are not realizables

Body structure

- Coarse-grained anatomy:
- Subsumed by *BioTop:Structured biological entity*
- Peculiarity: most body structure classes have the suffix "structure", similar to SNOMED CT: x_structure means x or any part of it. Thus, part-of relations are masked as taxonomies:

  Bones of hand subClassOf Hand structure

  means

  Bones of hand subClassOf part of some Hand

Activity and Participation

- Corresponds quite nicely to *BioTop:Processual entity* , which implies the existence of a participant (expressed by *Biotop:has participant*)
- Sometimes it is difficult to distinguish between Activity and Function
- Distinguishing criterion: Activities are Processes. They happen, functions don’t. However, a process can be the realization of a function / disposition

Environmental factors

- Products and Technology
  - Ontologically heterogeneous
  - Products are subsumed by *BioTop:MaterialEntity*
  - Technology is subsumed by *BioTop:InformationEntity*
  - Difference: products materially exist. technology can be implemented in products
- Support and relationship:
  - Persons and animals, bearer of a specific role
  - Attitudes: dispositions? They are realized by certain activities
- Services, systems, policies: again heterogeneous
  - e.g. *BioTop:LegalEntity* , *BioTop:Regulation or Law*
    Systems can also correspond to *BioTop:MaterialEntity*

Conclusions

- Ontologies have quite distinctive features from terminologies / thesauri
- Some common ground between Ontologies and classification system
- Good practice important – bad examples abound (OWL semantics must be understood)
- ICF has many features of an ontology and can partially be aligned with upper level ontologies
- Detailed scrutiny still to be done (e.g. delineation between function and process)
- Big biomedical ontology projects (OBO, SNOMED) should be considered in the ICF process

Open for participation

http://www.iaoa.org/