

# VIP

Virtual Imaging Platform



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[purl.org/steschu](http://purl.org/steschu)



# Does **Medical Image Simulation** Require Formal Ontologies?

VIP – Virtual Imaging Workshop  
Lyon, France, Dec 14<sup>th</sup>, 2012

# What are (formal) Ontologies?

# What are (formal) Ontologies?

- Computer science view
  - Ontologies are purpose-oriented formal models of meaning (conceptualizations)
- Cognitive / linguistic view
  - Ontologies are concept systems or systems of semantic reference (no clear distinction from thesauri)
  - Also adopted by parts of the Semantic Web community
- Philosophy view (scientific realism)
  - Ontology is the study of what there is
  - Formal Ontologies give precise mathematical formulations of the properties and relations of certain entities.

# Formal ontology in a nutshell

- Formal ontology = logic based ontology
- Description logics: subset of first order logic
- Common standard: OWL (Ontology Web Language)
- Ontologies are taxonomies of classes
- Ontologies can define classes in terms of (Aristotelian) definitions

**Subclass (aka is-a):**

$A \text{ subClassOf } B$

iff

$$\forall a: A(a) \rightarrow B(a)$$

**Vertebrate**

subClassOf

**Primate**

subClassOf

**Homo Sapiens**

**Equivalence:**

$X \text{ equivalent to } Y \text{ and some } r Z$

iff

$$\forall x: X(x) \leftrightarrow Y(x) \wedge \exists z: C(z) \wedge r(x,z)$$

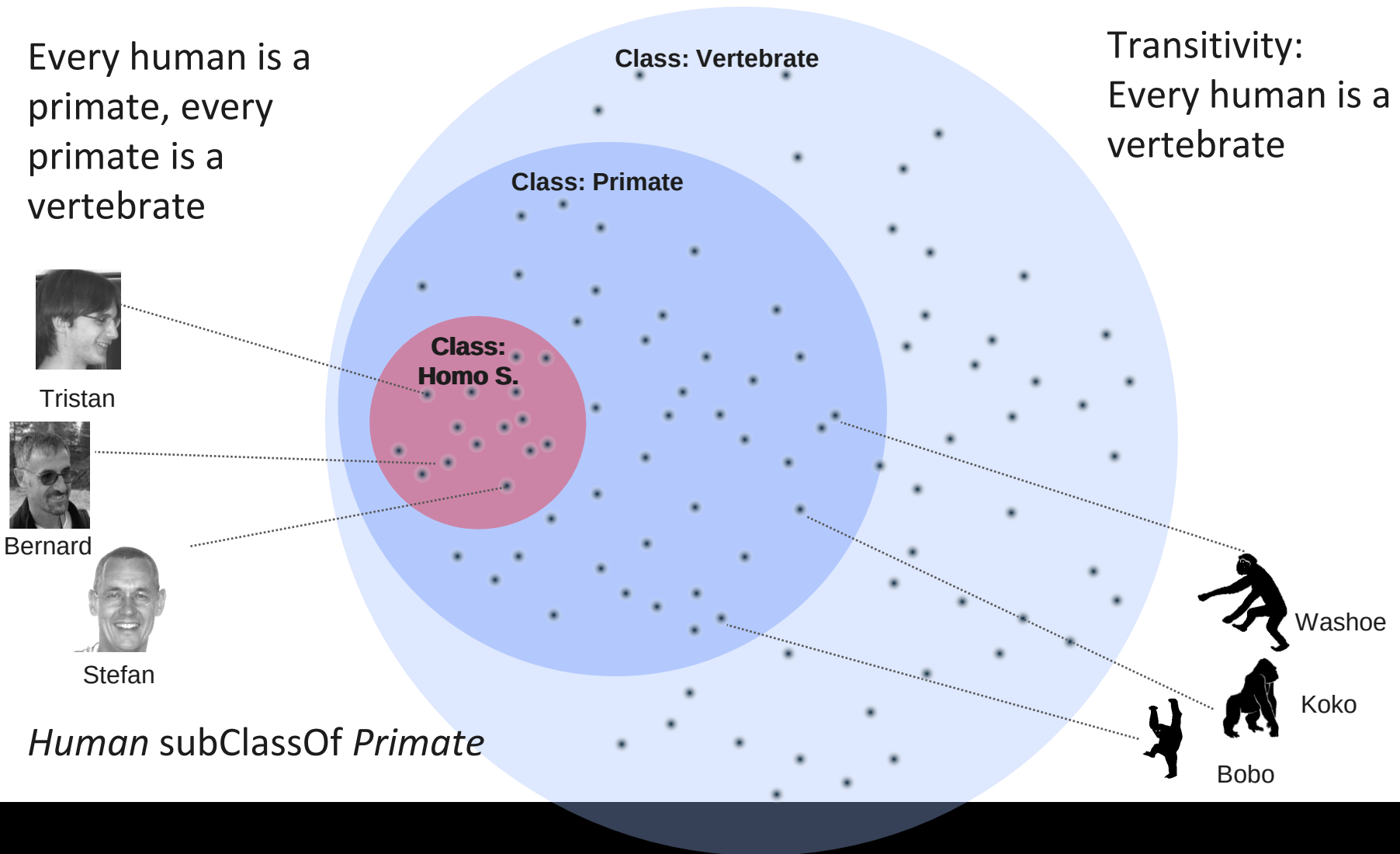
*Primate subClassOf Vertebrate*

*Vertebrate equivalentTo Animal  
and hasPart some Vertebra*

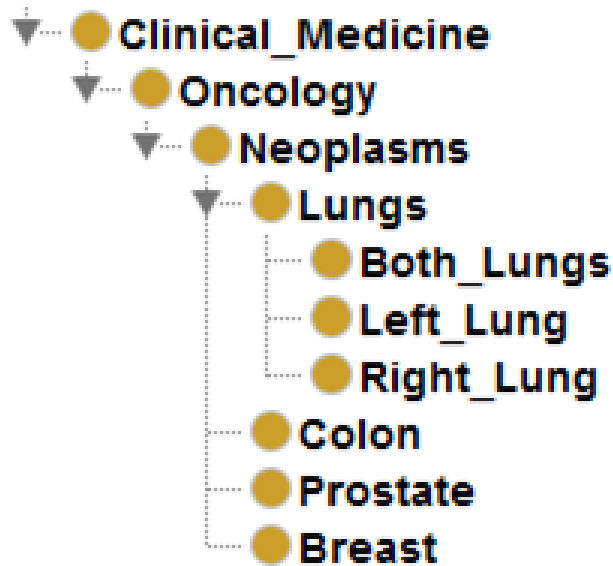
# Taxonomy as Venn diagram

Every human is a primate, every primate is a vertebrate

Transitivity:  
Every human is a vertebrate



# Doing taxonomy right

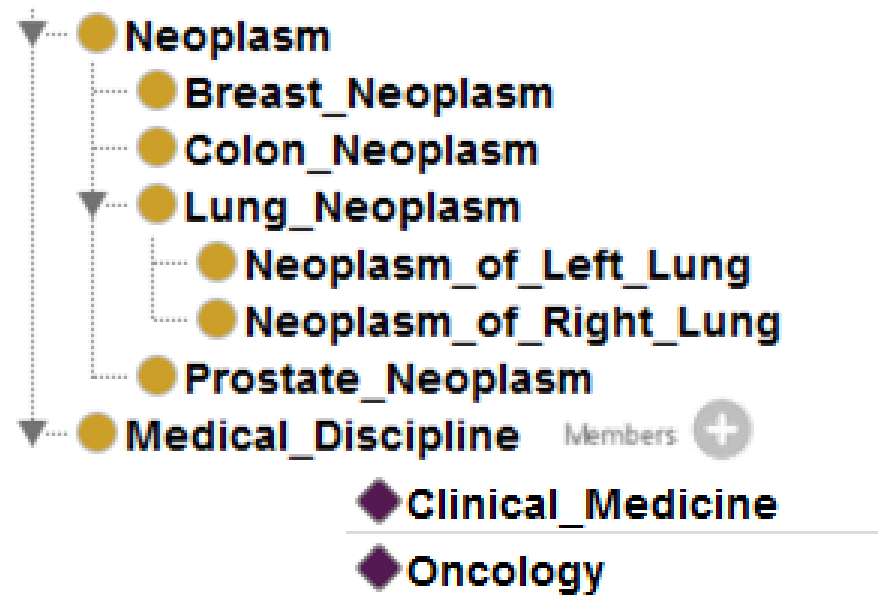
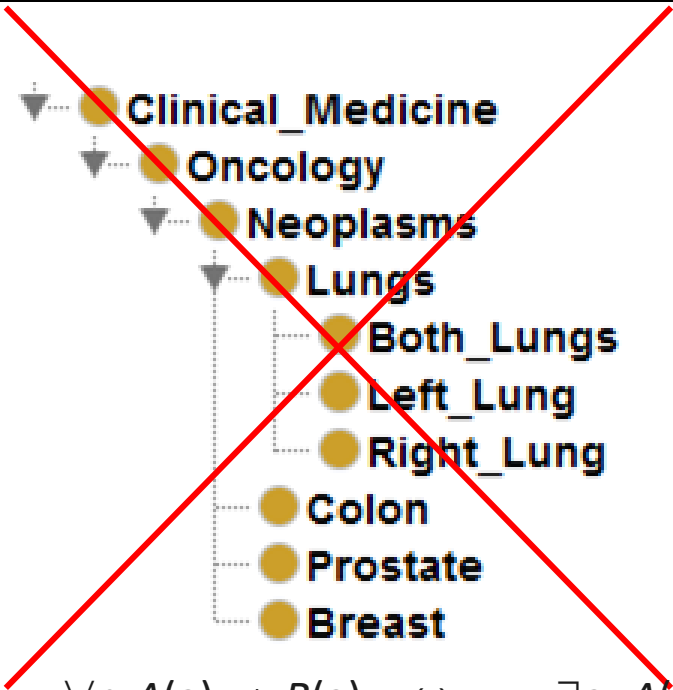


$$\forall a: A(a) \rightarrow B(a) \Leftrightarrow \neg \exists a: A(a) \wedge \neg B(a)$$

Test :

- there is no neoplasms that is not an oncology
- there is no prostate that is not a neoplasm
- there is no oncology that is not a clinical medicine

# Doing taxonomy right



$$\forall a: A(a) \rightarrow B(a) \quad \Leftrightarrow \quad \neg \exists a: A(a) \wedge \neg B(a)$$

Test :

- oncology is an instance of a medical discipline
- there is no prostate neoplasm that is not a neoplasm

**Labelling !**

# Formal ontology in a nutshell

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**Vertebrate**

subClassOf

**Primate**

subClassOf

**Homo Sapiens**

*Primate subClassOf Vertebrate*

**Equivalence:**

$X$  equivalent to  $Y$  and some  $r$   $Z$

iff

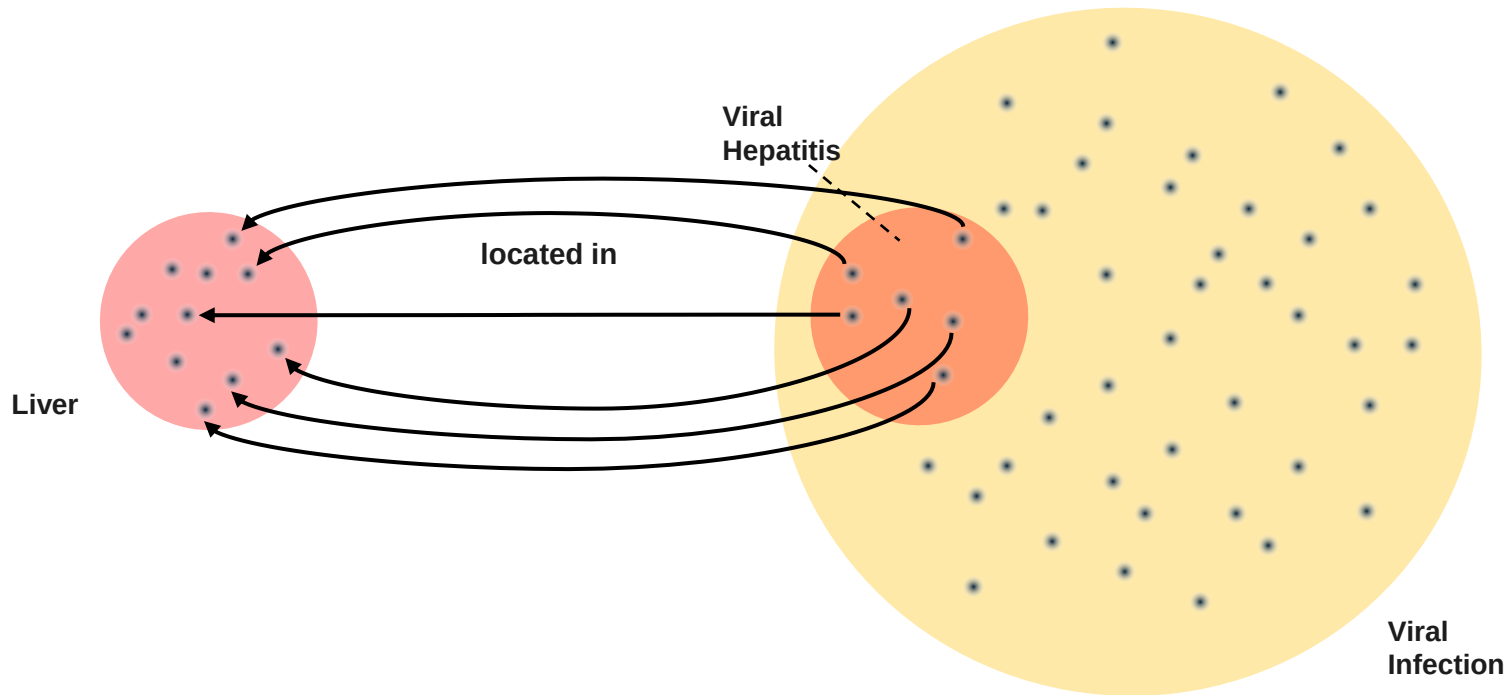
$$\forall x: X(x) \leftrightarrow Y(x) \wedge \exists z: C(z) \wedge r(x,z)$$

*Vertebrate equivalentTo Animal  
and **hasPart** some *Vertebra**



# Aristotelian Definitions:

## *Genus proximum and differentia specifica*

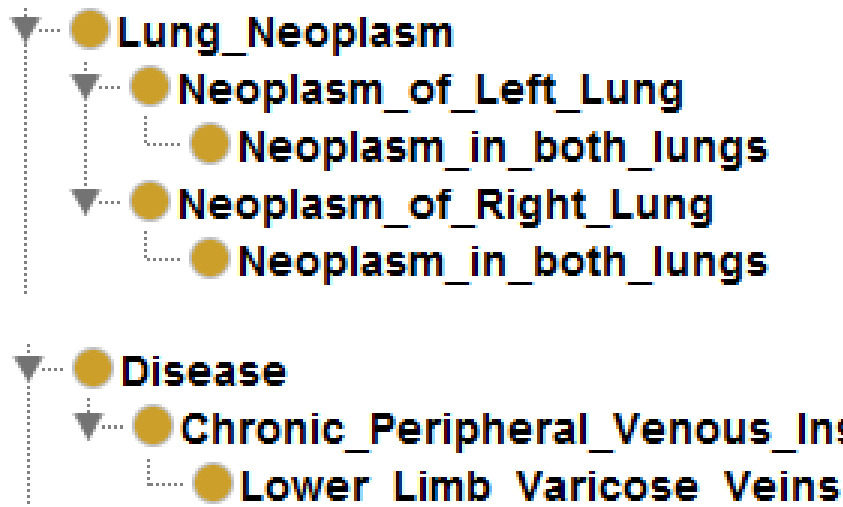


*ViralHepatitis* equivalentTo *ViralInfection* and **locatedIn** *Liver*

Test :

- There is no viral hepatitis that is not located in a liver
- There is no viral hepatitis that is not a viral infection

# Ontological Commitment

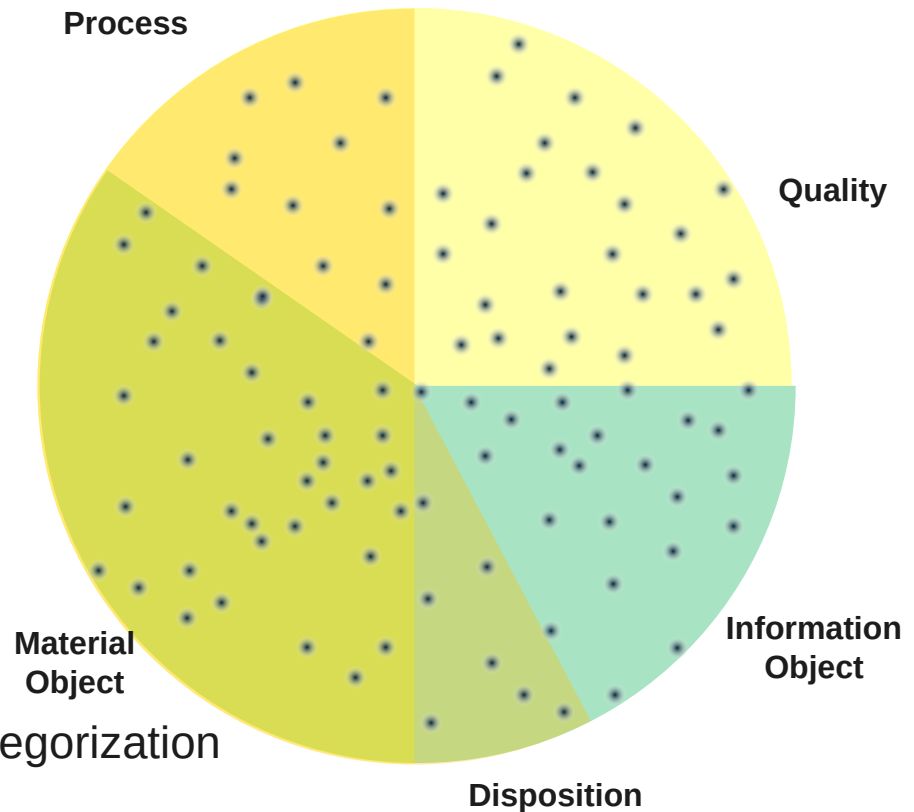
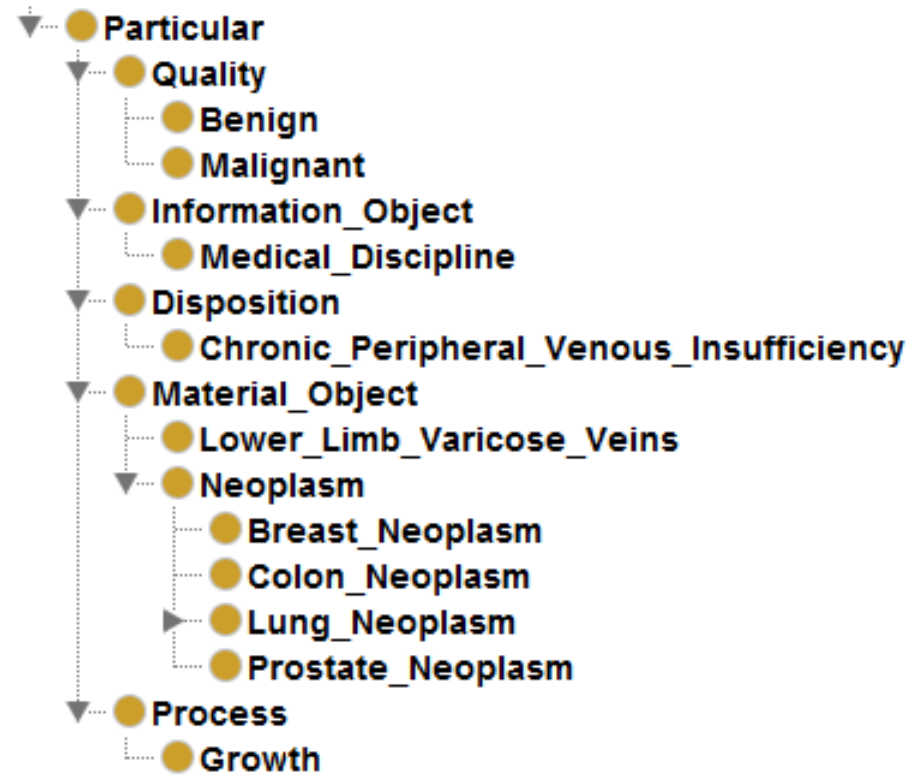


- Which are the instances?
- Does the label tell us what is meant?
- Is there an implicit context?

Test :

- There is no neoplasm in both lungs that is not a neoplasm in the left lung
- There is no varicose vein in the lower limb that is not a chronic peripheral venous insufficiency
- There is no patient with neoplasm in both lungs that is not a patient with the neoplasm in the left lung
- There is no patient with varicose lower limb veins that is not a patient with a chronic peripheral venous insufficiency

# Upper level ontologies: partition of the domain into disjoint and exhaustive categories



- Upper level ontologies enforce a strict categorization
- Constraints on upper-level categories
- Upper level ontology for the biomedical domain **BioTop**

# How formal ontologies challenge human cognition

- Built around taxonomies of classes
  - ATTENTION: our intuitive way of hierarchically organize terms is not strictly taxonomic
- State what is true for all individual members of a class (instances of a type)
- Requires to distinguish between classes and individuals
  - ATTENTION: human language is often misleading, e.g.  
Lyon is a big city vs. The liver is a big organ
- Individuals commit to upper-level categories
  - ATTENTION: our thinking fuses mutually dependent entities that belong to different categories, e.g. Cancer (growth process vs. mass of malignant tissue)
- Upper level categories should be made explicit
  - Explicit upper level ontology – common understanding
  - Implicit upper level ontology of each of us – misunderstanding

# What formal ontology is not

- Ontology  $\neq$  Knowledge representation
  - *"There are very few interesting items of knowledge that are truly ontological in this strict sense"* (Alan Rector)
  - antinomy: ὄντος (being) vs. ἐπιστήμη (knowledge)
- Ontology is not appropriate for
  - Default knowledge
    - "The hand has 5 fingers" (unless otherwise stated)
  - Probabilistic knowledge
    - Mesothelioma is a rare cancer
  - Contingent knowledge
    - Aspirin prevents myocardial infarction
    - Jaundice is a typical symptom of hepatitis

# Why formal ontology at all ??

- Formal definitions create maximum consensus on the meaning of terms
  - Ontologies as standards
  - Reusable terms and axioms
- Formal axioms encode statements about what is considered to be universally true in a domain
  - in contrast to knowledge proper
- Formal axioms permit logic-based reasoning
  - Consistency checking
  - New entailments
  - Equivalence of syntactically heterogeneous expressions can be computed: semantically interoperable systems

# Ontologies in life sciences and health care

# Ontologies in life sciences and health care

- Bottom-up ontology development:  
OBO (Open biomedical Ontologies) Foundry
- Top-down ontology development:  
SNOMED CT  
(Systematized Nomenclature of Medicine – Clinical terms)



# OBO Foundry: Orthogonality by Upper-level, and Granularity divisions

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

Smith, B.; Ashburner, M.; Rosse, C.; Bard, J.; Bug, W.; Ceusters, W.; Goldberg, L. J.; Eilbeck, K. et al. (2007). "The OBO Foundry: Coordinated evolution of ontologies to support biomedical data integration".Nature Biotechnology 25 (11): 1251–1255. doi:10.1038/nbt1346

# The OBO Foundry

- Collaborative bottom up initiative, driven by the success of the Gene Ontology
- Rooted in upper ontologies (BFO + RO)
- Goal of creating a suite of orthogonal interoperable reference ontologies in the biomedical domain
- Moving from semi-formal OBO syntax to OWL-DL
- Cross-ontology definitional axioms:
  - *Calcitonin secreting cell* (Cell Ontology) can be defined as a *Secretory cell* which secretes *Calcitonin* (ChEBI)
  - *Heart development* (Gene Ontology) can be defined as a *Developmental process* which has *Heart* (FMA) as participant

# SNOMED CT (Systematized Nomenclature of Medicine - Clinical Terms)

INTERNATIONAL HEALTH TERMINOLOGY  
STANDARDS DEVELOPMENT ORGANISATION



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## SNOMED CT

SNOMED CT (**S**ystematized **N**omenclature of **M**edicine-**C**linical **T**erms) is considered to be the most comprehensive, multilingual clinical healthcare terminology in the world. Each year, avoidable deaths and injuries occur because of poor communication between healthcare practitioners, or because busy practitioners forget or neglect to follow their own criteria for best practices. The delivery of a standard clinical terminology for use across the world's health information systems can therefore make a significant contribution towards improving the quality and safety of healthcare.

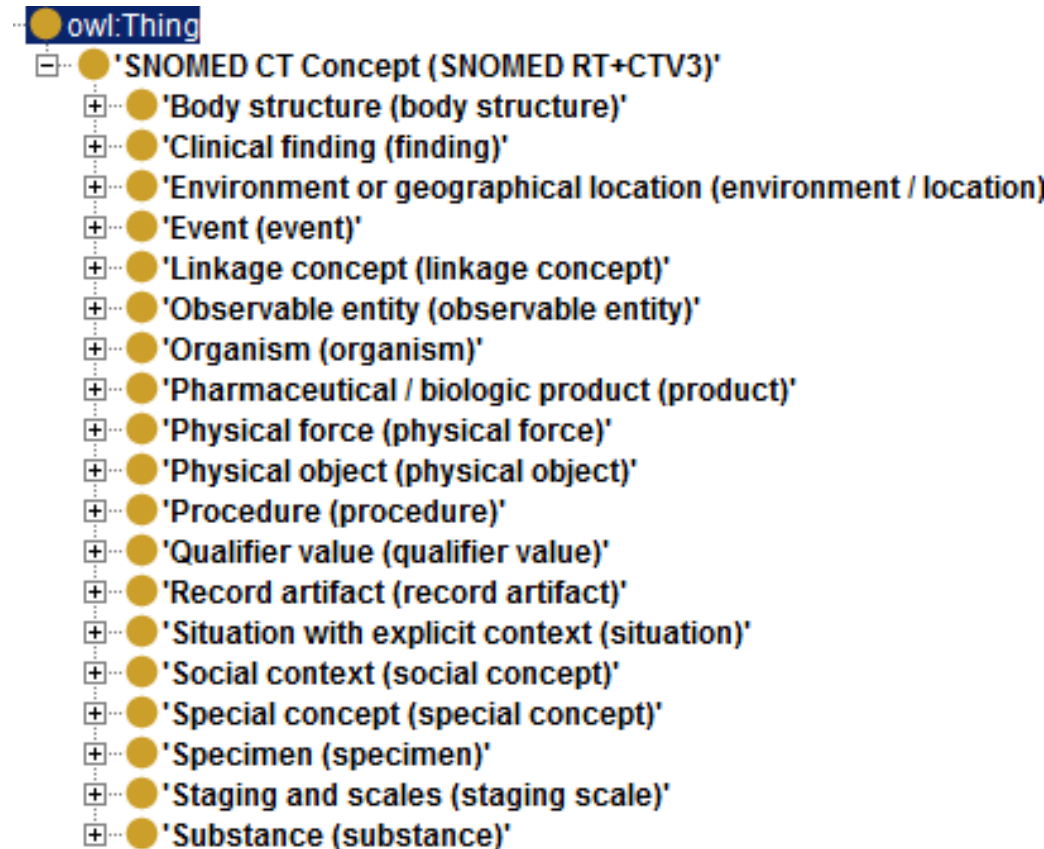
SNOMED CT aims to contribute to the improvement of patient care through underpinning the development of systems to accurately record health care encounters and to deliver decision support to health care providers. Ultimately, patients will benefit from the use of SNOMED CT to more clearly describe and accurately record their care, in building and facilitating better communication and interoperability in electronic health record exchange, and in creating systems that support health care decision making.

SNOMED CT intellectual property rights were transferred to the SNOMED SDO® in the formal creation of the IHTSDO.

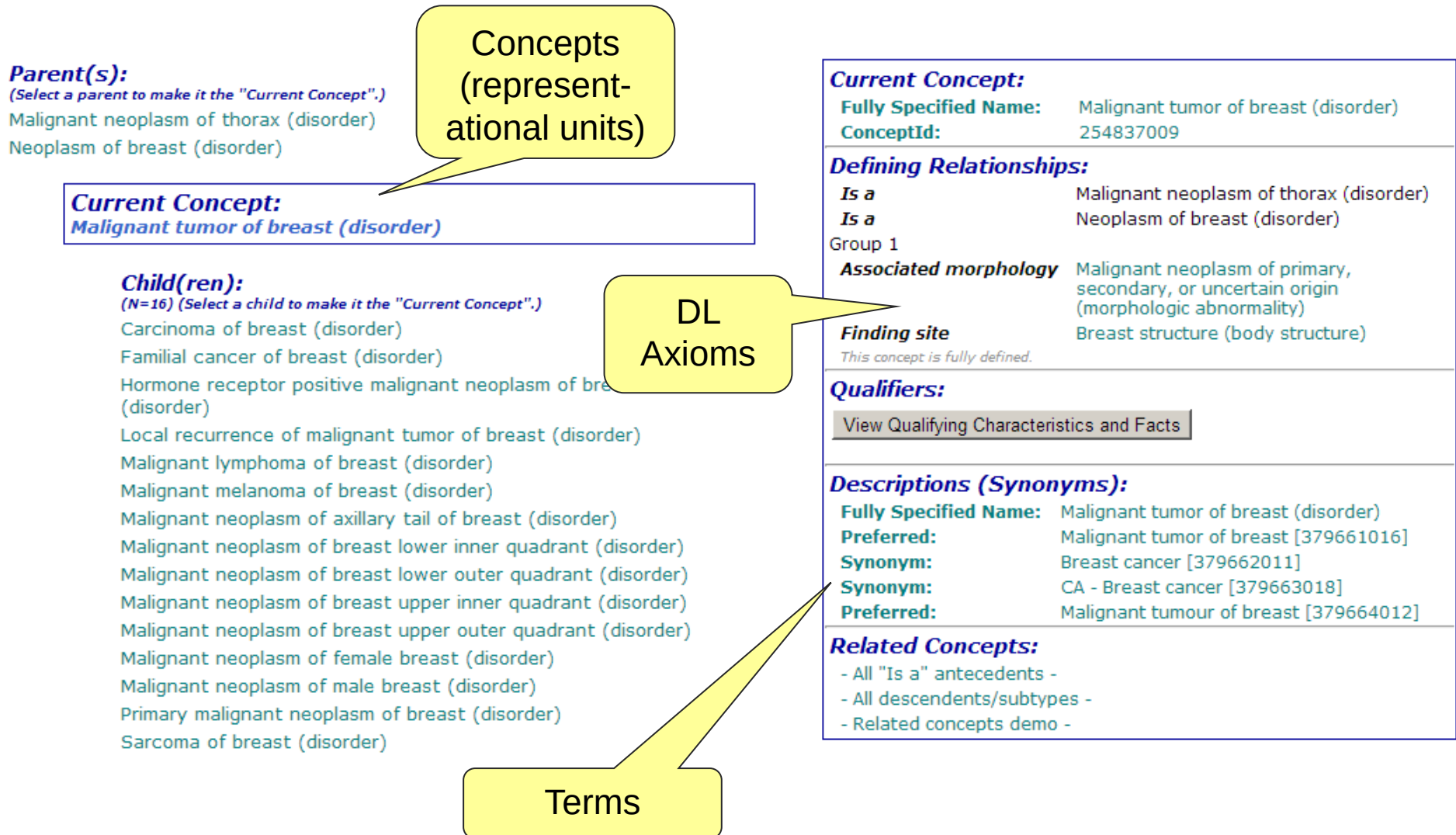
SNOMED CT was originally created by the College of American Pathologists by combining SNOMED RT and a computer based nomenclature and classification known as Clinical Terms Version 3, formerly known as Read Codes Version 3, which was created on behalf of the UK Department of Health and is Crown copyright.

# SNOMED CT - clinical terminology with ontological foundations

- Terminology for clinical data covering diseases, findings, procedures, organisms, substances etc.
- 311, 000 concepts, connected by 1,360,000 relational expressions
- Definitions with DL axioms
- Promoted as an international terminological standard"



# SNOMED CT: Terminology + Ontology



# Bioportal – repository for biomedical ontologies

## Term Search

Search for a term in multiple ontologies ?

[advanced options](#)

Search

### [Myocardium - NCI Thesaurus](#)

<http://ncicb.nci.nih.gov/xml/owl/EVS/Thesaurus.owl#Myocardium>

The striated muscle tissue of the heart enveloped by the epicardium and the endocardium.

[details](#) - [visualize](#) - [2 more from this ontology](#)

### [myocardium - Mouse adult gross anatomy](#)

[http://purl.obolibrary.org/obo/MA\\_0000164](http://purl.obolibrary.org/obo/MA_0000164)

[details](#) - [visualize](#) - [3 more from this ontology](#)

### [myocardium - RadLex](#)

<http://purl.bioontology.org/ontology/RID/RID1398>

[details](#) - [visualize](#) - [2 more from this ontology](#)

### [Myocardium - Medical Subject Headings \(MeSH\)](#)

<http://purl.bioontology.org/ontology/MSH/D009206>

The muscle tissue of the HEART. It is composed of striated, involuntary muscle cells (MYOCYTES, CARDIAC) connected to form the contractile pump to generate blood flow.

[details](#) - [visualize](#) - [1 more from this ontology](#)

### [myocardium - Experimental Factor Ontology](#)

[http://www.ebi.ac.uk/efo/EFO\\_0000819](http://www.ebi.ac.uk/efo/EFO_0000819)

Muscle layer of organ which has as its parts the myocardium proper and the conducting system of the heart.

[details](#) - [visualize](#) - [2 more from this ontology](#)

### [Myocardium - Logical Observation Identifier Names and Codes](#)

<http://purl.bioontology.org/ontology/LNC/LP101987-8>

[details](#) - [visualize](#) - [6 more from this ontology](#)

### [Myocardium - Foundational Model of Anatomy](#)

<http://sig.uw.edu/fma#Myocardium>

Muscle layer of organ which has as its parts the myocardium proper and the conducting system of the heart.

[details](#) - [visualize](#) - [1 more from this ontology](#)

### [Myocardium - Cell line ontology](#)

[http://purl.obolibrary.org/obo/FMA\\_9462](http://purl.obolibrary.org/obo/FMA_9462)

[details](#) - [visualize](#) - [6 more from this ontology](#)

### [Myocardium - Read Codes, Clinical Terms Version 3 \(CTV3\)](#)

<http://purl.bioontology.org/ontology/RCD/XC01s>

[details](#) - [visualize](#) - [5 more from this ontology](#)

### [myocardium - Zebrafish anatomy and development](#)

[http://purl.obolibrary.org/obo/ZFA\\_0001319](http://purl.obolibrary.org/obo/ZFA_0001319)

[details](#) - [visualize](#) - [2 more from this ontology](#)

### [myocardium - BRENDA tissue / enzyme source](#)

[http://purl.obolibrary.org/obo/BTO\\_0000901](http://purl.obolibrary.org/obo/BTO_0000901)

The middle and thickest layer of the heart wall, composed of cardiac muscle.

[details](#) - [visualize](#)

### [myocardium - Teleost Anatomy Ontology](#)

[http://purl.obolibrary.org/obo/TAO\\_0001319](http://purl.obolibrary.org/obo/TAO_0001319)

[details](#) - [visualize](#) - [2 more from this ontology](#)

### [myocardium - CRISP Thesaurus, 2006](#)

<http://purl.bioontology.org/ontology/CSP/1390-2774>

muscle tissue of the heart composed of striated, involuntary muscle known as cardiac muscle.

[details](#) - [visualize](#) - [1 more from this ontology](#)

### [myocardium - eVOC \(Expressed Sequence Annotation for Humans\)](#)

[http://purl.obolibrary.org/obo/EV\\_0100022](http://purl.obolibrary.org/obo/EV_0100022)

The middle layer of the heart wall that consists of cardiac muscle.

[details](#) - [visualize](#) - [2 more from this ontology](#)

### [Myocardium - Cardiac Electrophysiology Ontology](#)

[http://purl.org/obo/owl/FMA#FMA\\_9462](http://purl.org/obo/owl/FMA#FMA_9462)

[details](#) - [visualize](#) - [1 more from this ontology](#)

### [myocardium - Minimal anatomical terminology](#)

[http://purl.obolibrary.org/obo/MAT\\_0000453](http://purl.obolibrary.org/obo/MAT_0000453)

[details](#) - [visualize](#)

### [Myocardium - BioModels Ontology](#)

[http://purl.org/obo/owlapl/fma#FMA\\_9462](http://purl.org/obo/owlapl/fma#FMA_9462)

[details](#) - [visualize](#) - [1 more from this ontology](#)

Submit your ontology to  
<http://bioportal.bioontology.org>

# Formal ontologies and beyond...

 BioPortal

BrowseSearchMappingsRecommenderAnnotatorResource IndexProjects

Medical Subject Headings (MeSH)

Summary

Details

|                     |                                                                                                                                                                                                                              |
|---------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| ONTOLOGY ID:        | 1351                                                                                                                                                                                                                         |
| BIOPORTAL PURL:     | <a href="http://purl.bioontology.org/ontology/MSH">http://purl.bioontology.org/ontology/MSH</a>                                                                                                                              |
| STATUS:             |                                                                                                                                                                                                                              |
| FORMAT:             | RRF                                                                                                                                                                                                                          |
| CATEGORIES:         | Health                                                                                                                                                                                                                       |
| GROUPS:             | Unified Medical Language System                                                                                                                                                                                              |
| CONTACT:            | Stuart Nelson, M.D., <a href="mailto:nelson@nlm.nih.gov">nelson@nlm.nih.gov</a>                                                                                                                                              |
| HOME PAGE:          | <a href="http://www.nlm.nih.gov/mesh/meshhome.html">http://www.nlm.nih.gov/mesh/meshhome.html</a>                                                                                                                            |
| PUBLICATIONS PAGE:  | <a href="http://www.nlm.nih.gov/mesh/meshhome.html">http://www.nlm.nih.gov/mesh/meshhome.html</a>                                                                                                                            |
| DOCUMENTATION PAGE: | <a href="http://www.nlm.nih.gov/mesh/meshhome.html">http://www.nlm.nih.gov/mesh/meshhome.html</a>                                                                                                                            |
| DESCRIPTION:        | Medical Subject Headings (MeSH);National Library of Medicine; 2011                                                                                                                                                           |
| LICENSE INFORMATION | This ontology is made available via the UMLS. Users of all UMLS ontologies must abide by the terms of the UMLS license, available at <a href="https://uts.nlm.nih.gov/license.html">https://uts.nlm.nih.gov/license.html</a> |

ReviewsAdd your review

REVIEW BY STESCHU ON 12/14/2012

★★★★★ Usability

★★★★★ Domain Coverage

★★★★★ Correctness

★★★★★ Quality Of Content

★★★★★ Degree Of Formality

★★★★★ Documentation And Support

MeSH is not an ontology. It has never claimed to be one. Its concepts are not classes in the sense of OWL. Its hierarchical links are not subclass relations. If you interpret them as such you get strange inferences such as "Every thumb is a hand". This would do injustice to MeSH , which is a great resource, which fulfils it goals without subscribing to OWL semantics.

# **Alternative to formal ontologies: INFORMAL terminologies / thesauri**



# Alternative to formal ontologies: INFORMAL terminologies / thesauri

- Group together words / terms according to similarity in meaning
- Basic relations:
  - Synonymy
  - Broader / Narrower (ordering relations)
- Concept = Group of (quasi)synonyms
- Multiple hierarchies
- Mainly designed for retrieval
- Text definitions / explanations (scope notes) if required
- No formal semantics

# MeSH - Medical Subject Headings

## Neoplasms [C04]

### Neoplasms by Site [C04.588]

- Abdominal Neoplasms [C04.588.033] +
- Anal Gland Neoplasms [C04.588.083]
- Bone Neoplasms [C04.588.149] +
- Breast Neoplasms [C04.588.180]
  - Breast Neoplasms, Male [C04.588.180.260]
  - Carcinoma, Ductal, Breast [C04.588.180.390]
  - Hereditary Breast and Ovarian Cancer Syndrome [C04.588.180.483]
  - Inflammatory Breast Neoplasms [C04.588.180.576]
- Digestive System Neoplasms [C04.588.274] +
- Endocrine Gland Neoplasms [C04.588.322] +
- Eye Neoplasms [C04.588.364] +
- Head and Neck Neoplasms [C04.588.443] +
- Hematologic Neoplasms [C04.588.448] +
- Mammary Neoplasms, Animal [C04.588.531] +
- Nervous System Neoplasms [C04.588.614] +
- Pelvic Neoplasms [C04.588.699]
- Skin Neoplasms [C04.588.805] +
- Soft Tissue Neoplasms [C04.588.839] +
- Splenic Neoplasms [C04.588.842]
- Thoracic Neoplasms [C04.588.894] +
- Urogenital Neoplasms [C04.588.945] +

## Skin and Connective Tissue Diseases [C17]

### Skin Diseases [C17.800]

#### Breast Diseases [C17.800.090]

##### Breast Cyst [C17.800.090.249]

##### ► Breast Neoplasms [C17.800.090.500]

##### Breast Neoplasms, Male [C17.800.090.500.260]

##### Carcinoma, Ductal, Breast [C17.800.090.500.390]

##### Hereditary Breast and Ovarian Cancer Syndrome [C17.800.090.500.483]

##### Inflammatory Breast Neoplasms [C17.800.090.500.576]

##### Fibrocystic Breast Disease [C17.800.090.750]

##### Gynecomastia [C17.800.090.875]

##### Lactation Disorders [C17.800.090.937] +

##### Mastitis [C17.800.090.968] +

|                             |                                                                                                                                                                                                              |
|-----------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>MeSH Heading</b>         | Breast Neoplasms                                                                                                                                                                                             |
| <b>Tree Number</b>          | <a href="#">C04.588.180</a>                                                                                                                                                                                  |
| <b>Tree Number</b>          | <a href="#">C17.800.090.500</a>                                                                                                                                                                              |
| <b>Annotation</b>           | human only; <a href="#">BREAST NEOPLASMS, MALE</a> is also available; for animal use <a href="#">MAMMARY NEOPLASMS, ANIMAL</a> <a href="#">24.6+</a> ; coordinate IM with histological type of neoplasm (IM) |
| <b>Scope Note</b>           | Tumors or cancer of the human <a href="#">BREAST</a> .                                                                                                                                                       |
| <b>Entry Term</b>           | Breast Cancer                                                                                                                                                                                                |
| <b>Entry Term</b>           | Breast Tumors                                                                                                                                                                                                |
| <b>Entry Term</b>           | Cancer of Breast                                                                                                                                                                                             |
| <b>Entry Term</b>           | Cancer of the Breast                                                                                                                                                                                         |
| <b>Entry Term</b>           | Human Mammary Carcinoma                                                                                                                                                                                      |
| <b>Entry Term</b>           | Mammary Carcinoma, Human                                                                                                                                                                                     |
| <b>Entry Term</b>           | Mammary Neoplasm, Human                                                                                                                                                                                      |
| <b>Entry Term</b>           | Mammary Neoplasms, Human                                                                                                                                                                                     |
| <b>Entry Term</b>           | Neoplasms, Breast                                                                                                                                                                                            |
| <b>Entry Term</b>           | Tumors, Breast                                                                                                                                                                                               |
| <b>Allowable Qualifiers</b> | <a href="#">BL BS CF CH CI CL CN CO DH DI DT EC EH EM EN EP ET GE HI IM ME MI MO NU PA PC PP PS PX RA R</a>                                                                                                  |
| <b>Entry Version</b>        | BREAST NEOPL                                                                                                                                                                                                 |
| <b>Date of Entry</b>        | 19990101                                                                                                                                                                                                     |
| <b>Unique ID</b>            | D001943                                                                                                                                                                                                      |

# Example: Medical Subject Headings (MeSH)

## Literature search: monoclonal antibodies and cancer therapy

**PubMed.gov**  
US National Library of Medicine  
National Institutes of Health

PubMed  **MeSH terms**

[RSS](#) [Save search](#) [Advanced](#) [Help](#)

[Show additional filters](#) [Clear all](#)

**Text availability**  
Abstract available  
Free full text available  
Full text available

**Publication dates**  
5 years  
10 years  
Custom range...

**Species**  
Humans  
Other Animals

**Article types**   
Clinical Trial  
Meta-Analysis  
Practice Guideline  
Randomized Controlled Trial  
✓ **Review**  
Systematic Reviews  
more ...

**Display Settings:** ☒ Summary, 20 per page, Sorted by Recently Added [Send to:](#) ☒ [Filters:](#) [Manage Filters](#)

**Results: 2021 to 2040 of 5979**   Page

**Filters activated:** Review, English [Clear all](#)

☐ [Trastuzumab as the lead monoclonal antibody in advanced breast cancer: choosing which patient and when.](#)  
2021. Oliver IN.  
Future Oncol. 2008 Feb;4(1):125-31. **Review.**  
PMID: 18241007 [PubMed - indexed for MEDLINE]  
[Related citations](#)

☐ [Predicting outcome to VEGF-targeted therapy in metastatic clear-cell renal cell carcinoma: data from recent studies.](#)  
2022. Golshayan AR, Brick AJ, Choueiri TK.  
Future Oncol. 2008 Feb;4(1):85-92. **Review.**  
PMID: 18241003 [PubMed - indexed for MEDLINE]  
[Related citations](#)

☐ [Rituximab: beyond simple B cell depletion.](#)  
2023. Kessel A, Rosner I, Toubi E.  
Clin Rev Allergy Immunol. 2008 Feb;34(1):74-9. **Review.**  
PMID: 18240027 [PubMed - indexed for MEDLINE]  
[Related citations](#)

**Results by year**

**Find related data**  
Database:

**Search details**  
"Antibodies, Monoclonal"[Mesh] AND  
"Neoplasms"[Mesh] AND  
(Review[ptyp] AND English[lang])  
 [See more...](#)

# "Nontologies"

- Use OWL syntax, which should not be interpreted according to description logics semantics
- Formal reasoning would lead to incorrect entailments
- Examples: NCI thesaurus, Radlex
- Many other ontologies contain problematic axioms that contradict the intended meaning
- Example (NCI thesaurus):

*Calcium-Activated\_Chloride\_Channel-2* subclassOf

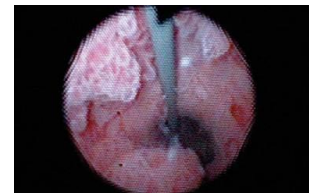
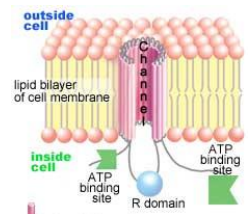
**Gene\_Product\_Expressed\_In\_Tissue** some *Lung* and

**Gene\_Product\_Expressed\_In\_Tissue** some *Mammary\_Gland* and

**Gene\_Product\_Expressed\_In\_Tissue** some *Trachea*

*Ureter\_Small\_Cell\_Carcinoma* subclassOf

**Disease\_May\_Have\_Finding** some *Pain*

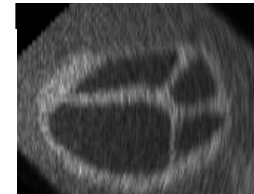


# (N)ontologies of interest for imaging

- Radlex
  - 24800 classes covering anatomy, procedures, diseases, substances, devices, relevant for radiologic imaging
  - all classes are also individuals (punning)
  - Relations ('is a', 'part of') asserted are at the level of individuals
  - On classes no other axioms than subclass axioms
- Foundational model of anatomy
  - Complete model: Protégé Frames (no formal semantics)  
Parts of it available as OWL
  - All assertions at class level
  - Logical entailments only true for "canonical" anatomy
- SNOMED CT

# Challenges of "correct" ontology for image representation including simulation

- Same terms (e.g. "cardiac motion") can be used for different things
  1. A real cardiac motion in a patient (process)
  2. Part of an image (information entity) that represents a real cardiac motion
  3. A simulation artifact (information entity), which does not refer to any specific cardiac motion
  4. The "concept" cardiac motion (cognitive entity)
- If you prefer 4. or if the distinction between 1. – 4. does not matter, then you shouldn't use formal ontologies



# Where ontologies or thesauri are sufficient

- Provision of controlled terms
  - Good text definitions should be available
- Hierarchy expansion for retrieval
  - hierarchical links at the level of broader term / narrower terms
  - is-a → is narrower than
  - part-of → is narrower than
- "Hand-crafted" inference rules
  - no use of description logics classifiers
- Possible standard SKOS



# Own experiences with ontologies in large projects

- EU funded projects with multiple partners
  1. @neurlST: Data integration (clinical, genomic, simulation) on cerebral aneurysms
  2. DebugIT: Decision support system for infectious diseases
  3. SemanticHealthNet: Semantic interoperability between heterogeneous semantic representations in the EHR
- Experiences:
  - in 1. and 2. much effort put in formal ontology
  - Mostly used as a controlled vocabulary (1.)
  - DL reasoning only for computing inferred ontology, which then used with production rules
  - 3. Formal foundation seems fundamental to reach the interoperability goal. However, intellectual input considerable and scalability still open

# **Current state of the art of Applied Ontology as a discipline**

# Current state of the art of Applied Ontology as a discipline

- Applied Ontology – still emerging discipline
- Prevalence of makeshift ontology artifacts
- Ontology engineering required to be more principled
- Necessary resources
  - Standards (Semantic Web – OWL )
  - Good practice guidelines (e.g. **GoodOD** Guideline)
  - Quality management
  - Best-of-breed examples
  - Industry-standard tools
    - Editors
    - Reasoners

# Current state of the art of Applied Ontology as a discipline

## Guideline on Developing Good Ontologies in the Biomedical Domain with Description Logics

URL: <http://www.purl.org/goodod/guideline>

Version 1.0  
December 2012

Send feedback to:  
[martin.boeker@uniklinik-freiburg.de](mailto:martin.boeker@uniklinik-freiburg.de)  
[ludger.jansen@uni-rostock.de](mailto:ludger.jansen@uni-rostock.de)

Schulz S<sup>1,3</sup>, Seddig-Raufie D<sup>1</sup>, Grewe N<sup>2</sup>, Röhl J<sup>2</sup>,  
Schober D<sup>1</sup>, Boeker M<sup>1</sup>, Jansen L<sup>2</sup>

<sup>1</sup>: Institute of Medical Biometry and Medical Informatics,  
University Medical Center Freiburg

<sup>2</sup>: Institute of Philosophy, University of Rostock

<sup>3</sup>: Department of Medical Informatics, University of Graz

11th December 2012

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# Decision on using formal ontology in life science research projects

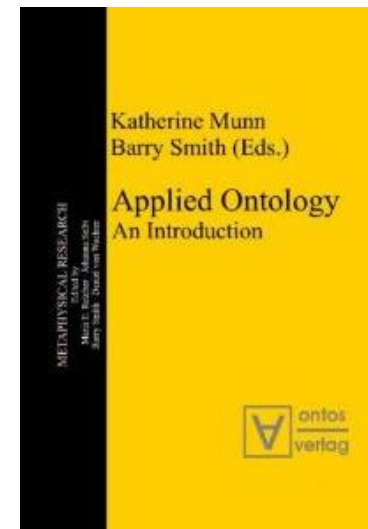
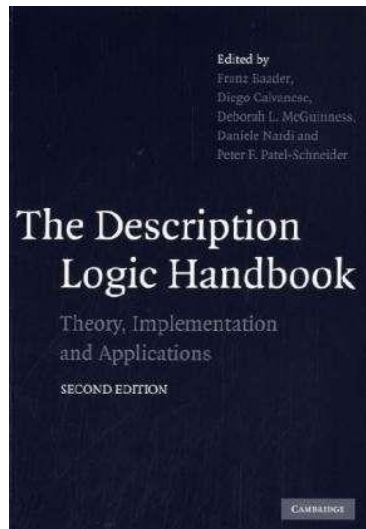
- Functional requirements
  - Controlled terminology
  - Query expansion for retrieval
  - Precise definitions of terms
  - Precise classification of domain entities
  - Reasoning to establish semantic equivalence
  - Representation of contingent knowledge
  - Default reasoning
  - Probabilistic reasoning

# Decision matrix

## Thesauri / Ontologies / KR formalism

|                                             | thesauri | ontology | KR |
|---------------------------------------------|----------|----------|----|
| Controlled domain language                  | x        |          |    |
| Query expansion for retrieval               | x        | x        |    |
| Precise definitions of terms                |          | x        |    |
| Precise classification of domain entities   |          | x        |    |
| Reasoning to establish semantic equivalence |          | x        |    |
| Representation of contingent knowledge      |          |          | x  |
| Non-monotonic reasoning                     |          |          | x  |
| Probabilistic reasoning                     |          |          | x  |

# Further readings





# Ontology on the Web

- Description Logics: <http://dl.kr.org/>
- Protégé: <http://protege.stanford.edu/>
- Bioontology: <http://www.bioontology.ch/>
- Buffalo Ontology Site: <http://ontology.buffalo.edu/smith/>
- OBO Foundry: <http://obofoundry.org/>
- Bioportal: <http://bioportal.bioontology.org/>
- SNOMED CT: <http://www.ihtsdo.org/snomed-ct/>  
<http://terminology.vetmed.vt.edu/sct/menu.cfm>
- CO-ODE (Pizza ontology): <http://www.co-ode.org/>
- GoodOD Guideline: <http://www.iph.uni-rostock.de/GoodOD-Guideline.1299.0.html>