5. Ontologies and languages in the biomedical semantic web

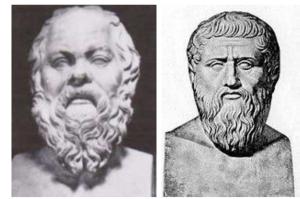
Dr. Isabella Hinterleitner hinterleitner.ih@gmail.com

Agenda

- Ontologies
 - Definition
 - Examples
 - Ontology Types
 - Ontology Management & Design
 - Connecting Ontologies
- Semantic Web
 - Standards
 - XML Examples
 - OWL and RDF Triples

What is an Ontology? (1)

- Philosophy: The word "ontology" comes from the philosophical discipline which deals with being (dem Sein), the being as a person and fundamental types of entitites.
- Separation between
 - Concept / class (= idea)
 - Instance (object of the real world, "shadow" of ideas)
 - Hierarchy of concepts
- (Sokrates, Platon)

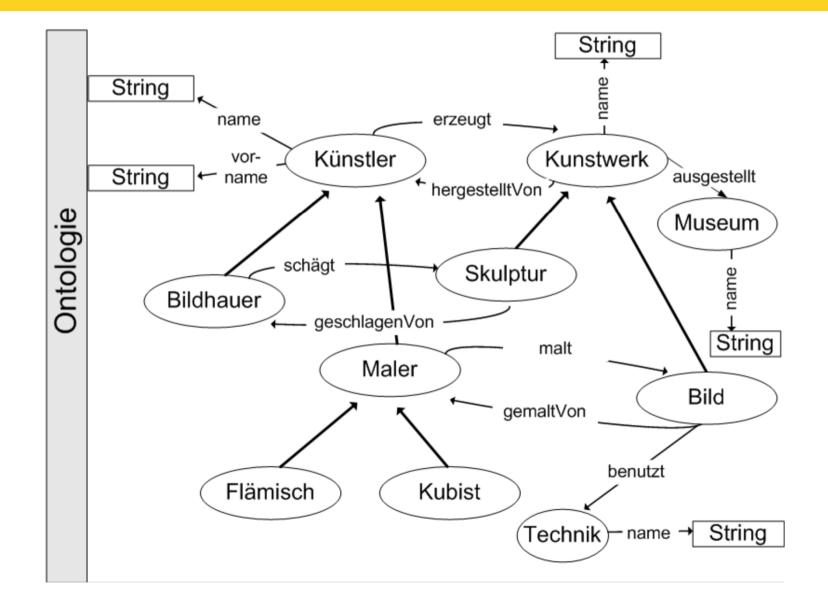


What is an Ontology? (2)

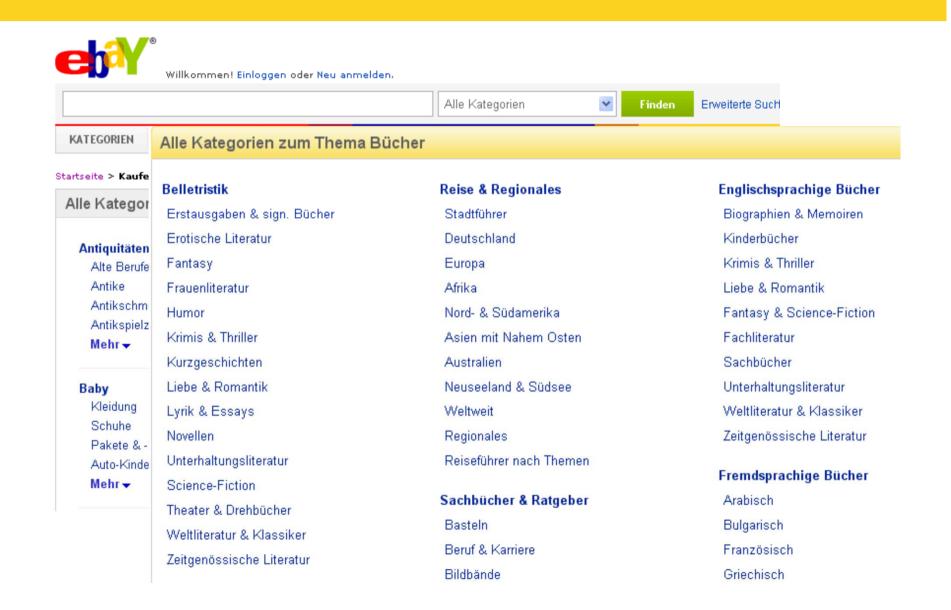
An ontology is an explicit, formal specification of a shared conceptualization. The term is borrowed from philosophy, where an ontology is a systematic account of Existence. For knowledge-based systems, what "exists" is exactly that which can be" represented. *(Thomas R. Gruber, 1993)*

- **Conceptualization** abstract modell (domain, identified relevant expressions, relationships)
- Explicit meaning of all expressions is defined
- Formal machine-readable
- Shared consense regarding ontology

Simple Ontology



Ontology in e-business



Existing Ontologies in Science

• NCI Metatheasaurus

Cancer Biology

Gene Ontology

Gene Products & Biological Processes

• BioPAX

Biological Pathways

• TAMBIS

Molecular Biology

• MGED

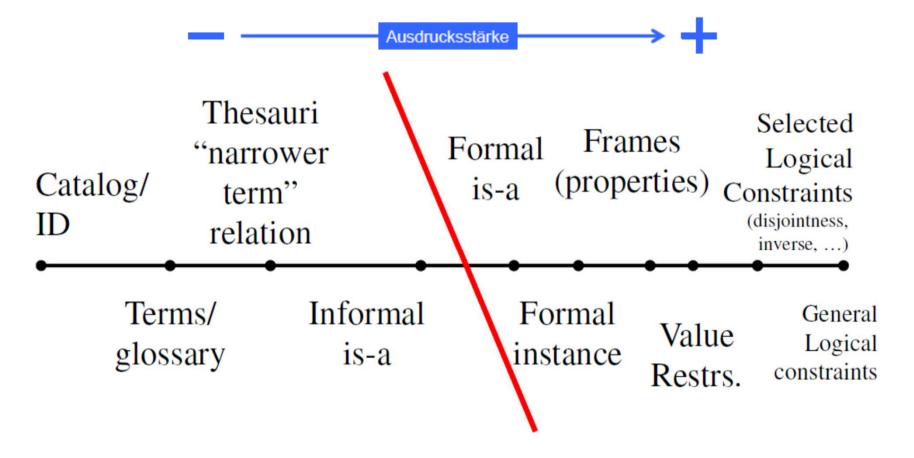
Microarray Data

NCIthesaurus

NCI Thesaurus Hierarchy

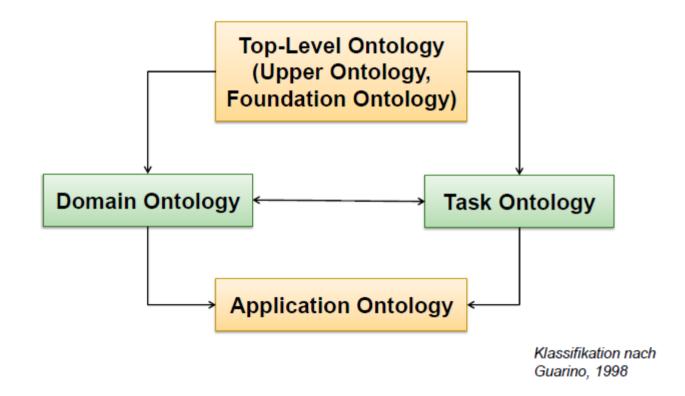
🗄 Abnormal Cell
🗄 Anatomic Structure, System, or Substance
🖹 Body Cavity
Abdominal Cavity
Cranial Cavity
- Endometrial Cavity
Nasal Cavity
- Oral Cavity
Orbit
Pelvis
Pericardial Cavity
Peritoneal Cavity
····Pleural Cavity
Thoracic Cavity
🗄 Body Fluid or Substance
⊡ Body Part
🖽 Body Region
🗄 Embryologic Structure or System
Body Fluid or Substance Body Part Body Region Embryologic Structure or System Microanatomic Structure Organ
⊞ Organ
🕀 Organ System
🖽 Other Anatomic Concept
±∵Tissue
🗄 Biochemical Pathway
H-Diological Process

Expressiveness of Ontologies



Ontologie-Spektrum nach McGuinness, 2001

Type of Ontologies



Ontology Types

Top-Level Ontology

- General, interdisciplinary ontology
- Describes general concepts (Zeit, Raum, Vorgang) independen of
- A certain domain or problem statement

Domain Ontology

• General concepts regarding a generic domain

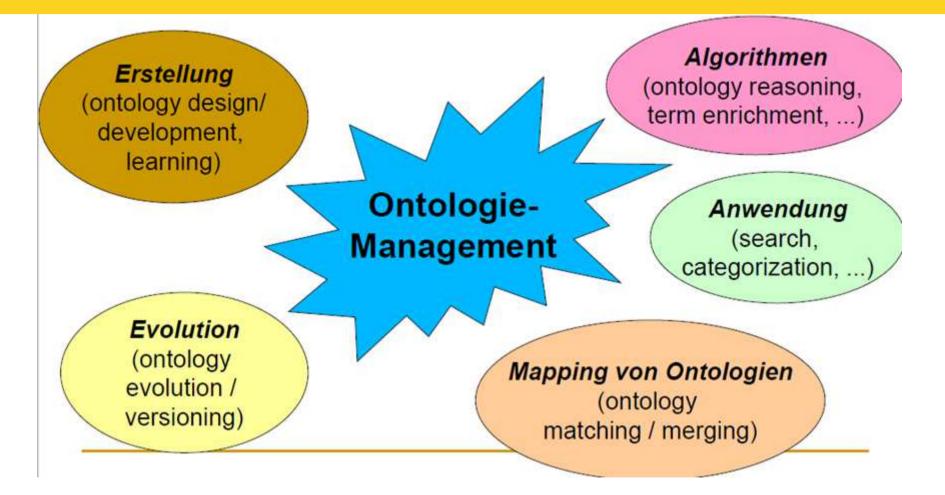
Task Ontology

• Basic concepts regarding tasks

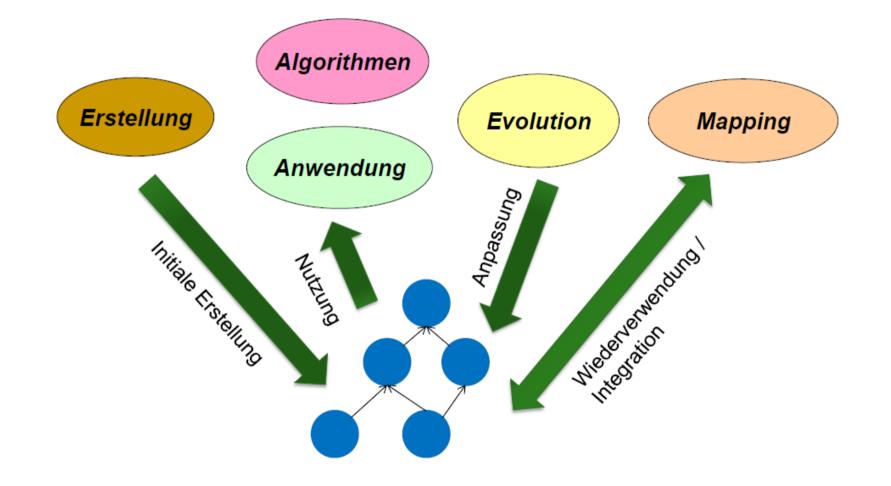
Application Ontology

- Especially regarding a concrete focussed domain or tasks
- is focussed in general on a domain or task ontology

Ontology Management



Ontology Management



Design and Development

Methods of Design

Includes all activities that are needed for the design of ontologies

Why?

- Development of consistent ontologies
- Efficient development of complex ontologies
- Distributed development of ontologies

Support by means of Tools

- Partly Semi(automatic) solutions
- Ontology Learning

Ontology Design

1) There is no one correct way to model a domain there are always viable alternatives. The best solution almost always depends on the application that you have in mind and the extensions that you anticipate.

2) Ontology development is necessarily an iterative process.

3) Concepts in the ontology should be close to objects (physical or logical) and relationships in your domain of interest. These are most likely to be nouns (objects) or verbs (relationships) in sentences that describe your domain.

N.F. Noy, D. McGuinness: Ontology Development 101: A Guide to Creating Your First Ontology. Stanford Knowledge Systems Laboratory, 2001.

Tools





www.cntoknowledge.org/oil



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Dynamics of Ontologies

Consistent Evolution

- Implementation of a new requirement/adaption of the ontology
- consistency: After the modification the ontology should be consistent again
- Example: Deleting/Adding a concept
- Versioning
- Accessing different versions of ontology
- How is versioning realized?
- Differences between two ontology versions
- How did version 2 develop from version 1

Connecting Ontologies

Ontologie-Mapping

- Mapping between two ontologies **A** and **B**
- Which Ontologieconcepts from **A** resemble the concepts in **B**?
- Ontology Matching → (semi) automatic comparison of ontologies
- Different techniques can be applied
- Integration (merging) of different ontologies
- Support new analysis methods for ontologies
- Reuse of knowledge!

The Semantic Web Vision

"The Semantic Web is a vision: the idea of having data on the Web defined and linked in such a way that it can be used by machines not just for display purposes, but for automation, integration and reuse of data across various applications."

Tim Berners - Lee

Semantic Web Standards

- XML Extensible Markup Language
- RDF Resource Description Framework
- OWL Web Ontology Language





- Extensible Markup Language
- Formatting documents and data by assigning

Metadata

(Data about data)

• Syntax resembles HTML



SBML (Systems Biology Markup Language)

Systems Biology ML Markup Language

CellML





Examples for XML Formats

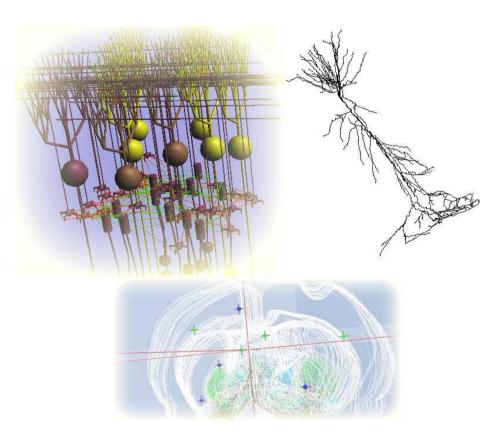
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CellML Model: Current Systems in Ventricular Cells

Examples for XML Formats

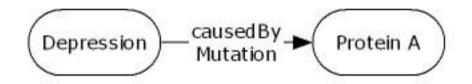
- Many other domain specific formats
- e.g. BrainML, NeuroML, MorphML...





- XML gives structure to data and documents
- RDF and OWL add logic, meaning and connectivity





Subject - Predicate - Object



<Depression> <causedByMutation> <Protein A>

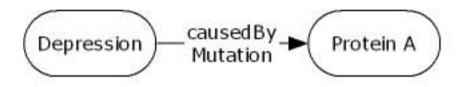
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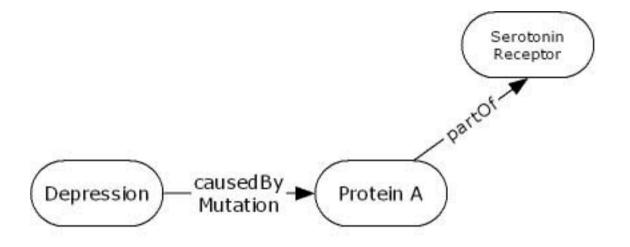


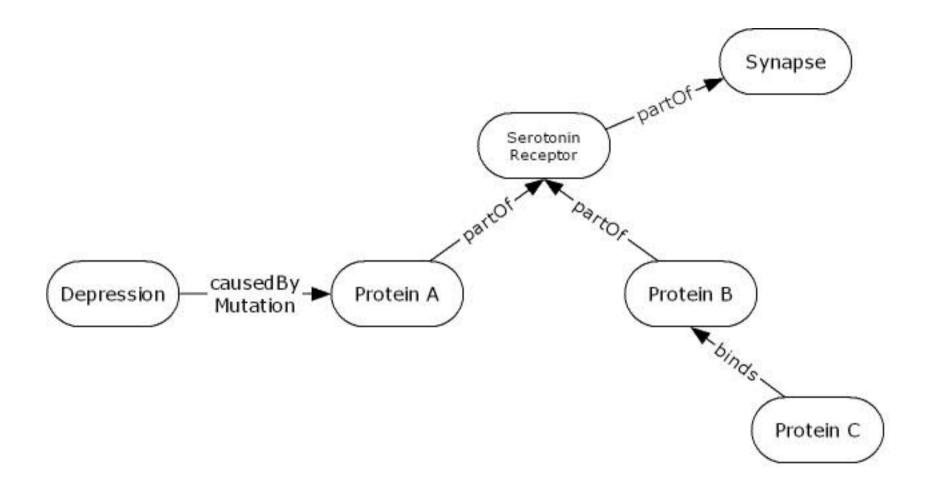
URIs (Unified Resource Identifieres) are used to identify resources

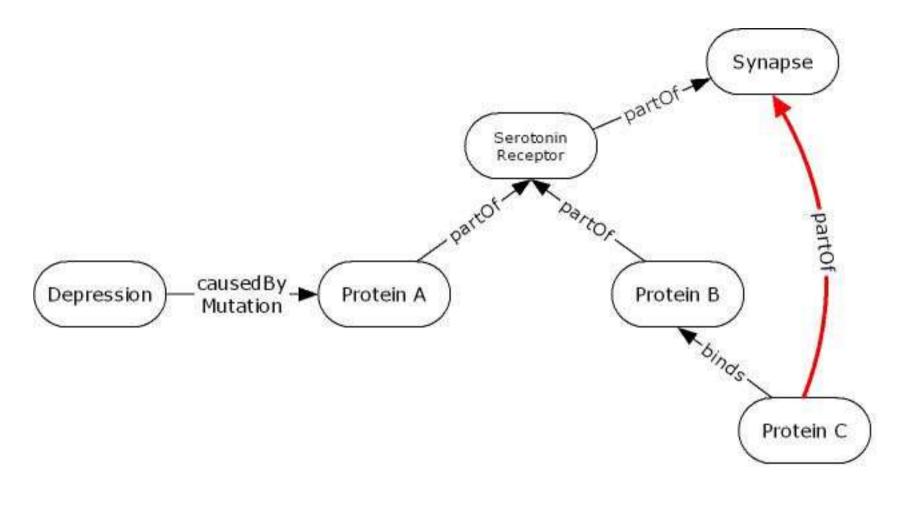
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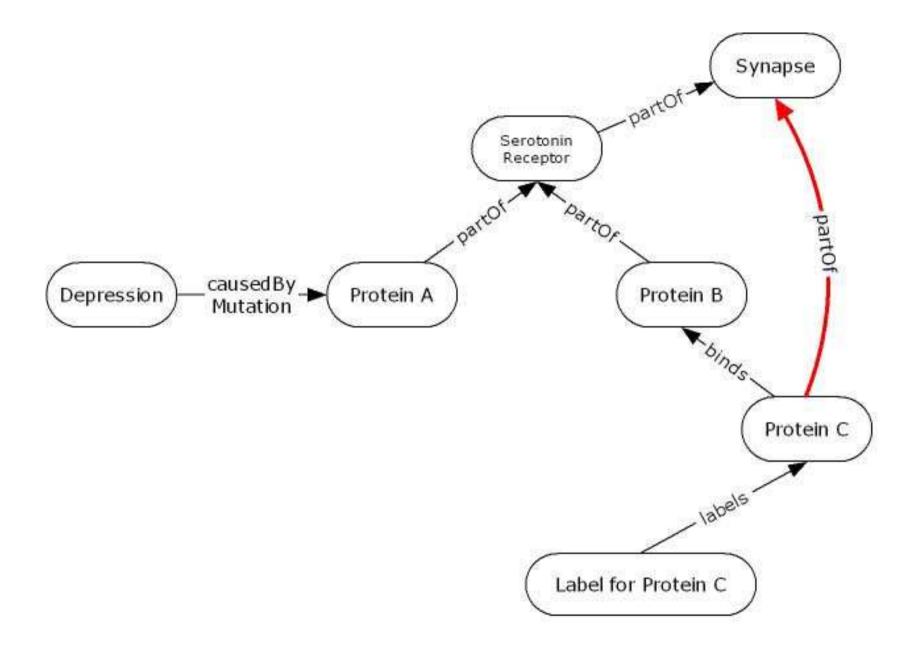


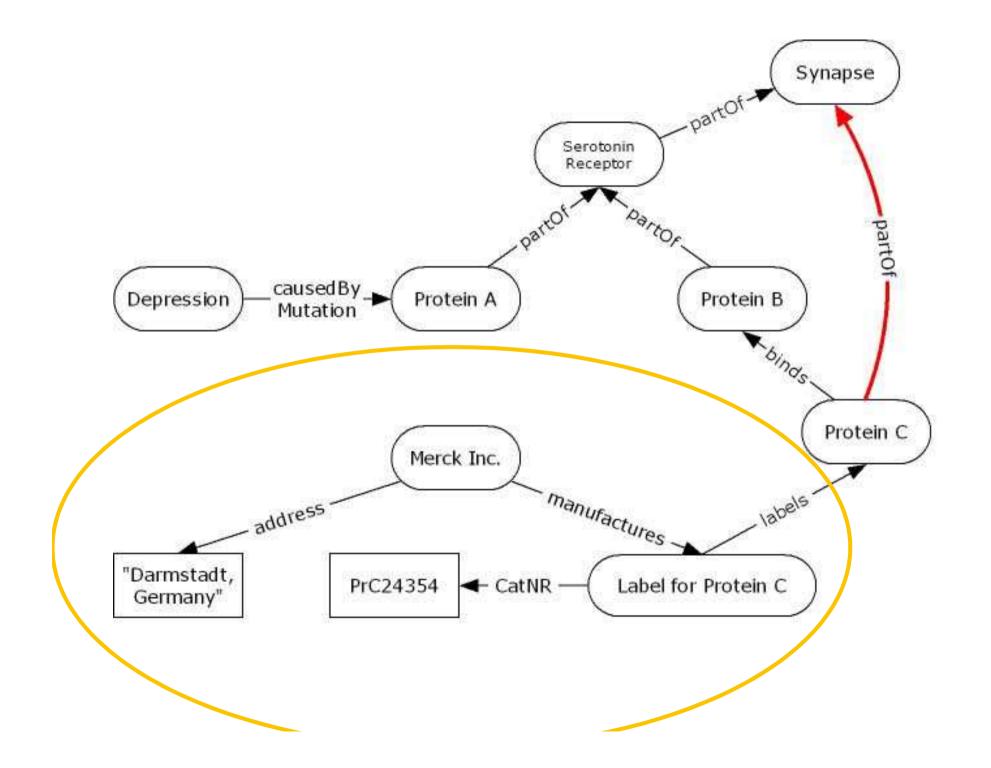






Inference





Thanks for your attention