

Overview of the NCBO workshop on ontology of clinical phenotypes

(<http://bioontology.org/wiki/index.php/DallasWorkshop>)

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Why did I attend the workshop?

- Invited
- Development of a pilot database called “BrainPharm” as part of the SenseLab’s Semantic Web effort
- BrainPharm captures pathological/pharmacological mechanism at the neuronal level for neurological diseases such as Alzheimer’s disease
- It represents our first steps to bridge data between molecular/neuronal level and disease level
- This effort relates to the theme of the workshop

Goals of the Workshop

- Develop a clear understanding and representation of the distinction between clinical and pre-clinical manifestations of signs, symptoms and findings.
- Take first steps towards harmonizing the ontological representation of disease signs and symptoms and clinical and laboratory findings with existing and emerging standards in knowledge representation from the health informatics and bioinformatics communities

Symptoms vs. Signs

- A symptom is a manifestation of a disease, indicating the nature of the disease, which is noticed by the patient. This is contrasted to signs which are observed by a medical practitioner.
- A symptom is subjective, observed by the patient, while a sign is observed and measured objectively by the doctor.

Quiz (sign or symptom)

- Chest pain
- Cyanosis (bluish discoloration of the skin)
- A baby cries

- Sometimes the context is important

Talks (Day 1)

- Signs, symptoms and laboratory findings from a health care perspective
 - rheumatology
 - neonatology
- Ontology approaches to describing the clinical phenotype
 - Reasoning with clinical exam and laboratory findings: The case of myocardial infarction
- Standardization of clinical and laboratory data in the context of clinical and translational research
 - Representation of clinical and laboratory findings in the CDISC SDTM

Talks (Day 2)

- Proposals for unification of standard terminologies for the representation of 'sign', 'symptom', 'finding' and related terms
- How consistent ontological design and development principles can lead to enhanced representation of signs, symptoms, and clinical and laboratory finding
 - PATO and Phenote: From model organism phenotypes to clinical medicine
- Strategies to achieve convergence of ontologies, vocabularies and data structures in representing signs, symptoms, and clinical and laboratory findings

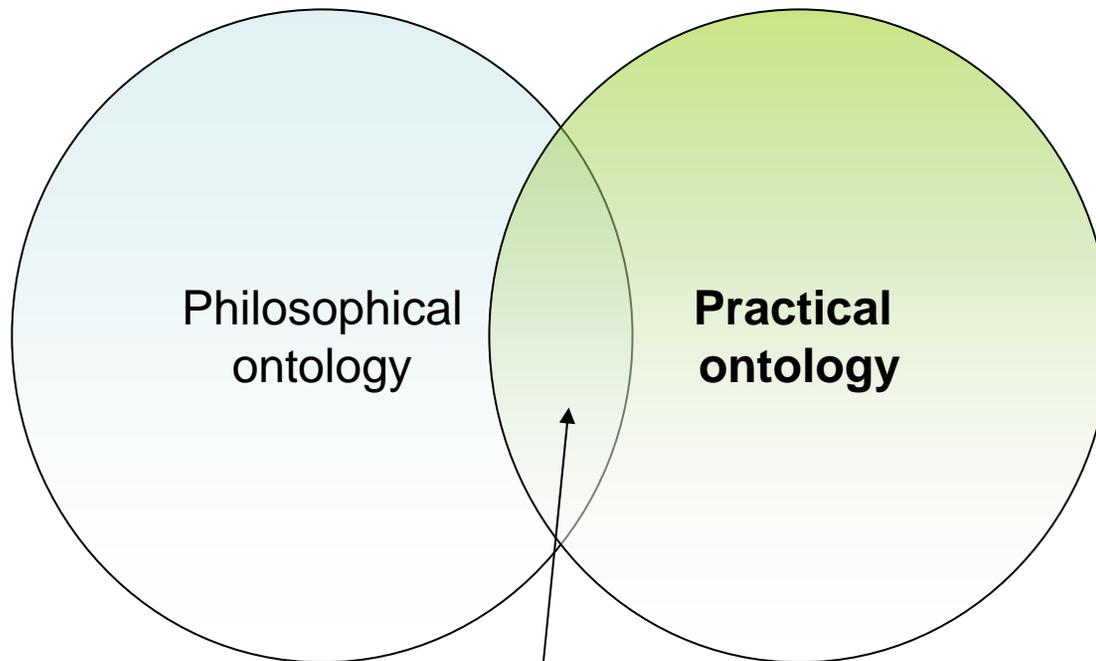
Genotype/Phenotype

- Mutant gene → mutant or missing protein → disease
- Gene ontology (GO)
 - Learning or memory (biological process)
 - Long-term potentiation or LTP (biological process) – synaptic plasticity
 - GABA-A receptor activity (molecular function)
 - Gene products associated with GABA-A
- Links GO to other databases (e.g., drug databases and phenotype ontologies such as PATO and PO)

How might it relate to HCLS IG?

- Terminology
- COI
- BioRDF (translational bioinformatics)
- Other task forces (LODD, SWAN-SIOC)

Semantic Web Meets Ontology



Can their intersection make
Semantic Web more intelligent
(GO may be a model)

The End