What are ontologies and why do we need them for terminologies?

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Learning objectives

- Describe the differences in purpose between ontology and terminology
- Discuss specific aspects of ontologies vs. terminologies
- Describe how ontological principles can help build better terminologies
Suggested readings


Ontology vs. terminology

Theoretical distinctions
Ontology vs. terminology

- **Ontology**
  - Defining types of things and their relations

- **Terminology**
  - Naming things in a domain

- **Thesaurus**
  - Organizing things for a given purpose

- **Classification**
  - Placing things into (arbitrary) classes

- **Knowledge bases**
  - Assertional vs. definitional knowledge
Terminology vs. ontology

◆ Terminological resources
  - Collections of terms (e.g., controlled vocabularies)
  - Useful for indexing and annotation
  - MeSH, GO

◆ Ontological resources
  - Collections of
    - kinds of entities (substances, qualities, processes)
    - relations among them
  - Useful for reasoning
  - UMLS Semantic Network, SNOMED CT

Ontological gradient
Names for Addison’s disease

**Synonyms: list of terms**

- Addisonian syndrome
- Bronzed disease
- Addison melanoderma
- Asthenia pigmentosa
- Primary adrenal deficiency
- Primary adrenal insufficiency
- Primary adrenocortical insufficiency
- Chronic adrenocortical insufficiency

- eponym
- symptoms
- clinical variants
Foundational Model of Anatomy (FMA)

Formal distinctions to organize the top level
Various kinds of relations

FMA Example

Kidney

FMA ID
7203

definition
Corticomедullary organ which has as its part renal pelvis shared with and connected to ureter. Examples: There are only two, right kidney and left kidney.

arterial supply
Renal artery

bounded by
Surface of kidney

contained in
Retroperitoneal space
Ontology vs. terminology

In practice
Terminology vs. ontology (revisited)

- **Lexical and terminological resources**
  - Mostly collections of names for biomedical entities
  - Often have some kind or hierarchical organization (e.g., relations)

- **Ontological resources**
  - Mostly collections of relations among biomedical entities
  - Sometimes also collect names
Small cell carcinoma  Terminology

- 4 terms (1 fully specified name + 3 synonyms)

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Small cell carcinoma Ontology

- Same name, 2 types of things
  - Formal distinction between
    - SCC as a disorder
    - SCC as a morphologic abnormality
- Logical definition with necessary and sufficient conditions

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Ontology spectrum

Medication Lists
DDI Lists

BT/NT,
Parent/Child,
Informal Is-A

Catalog

Terms/
glossary

MeSH,
Gene Ontology,
UMLS Meta

Thesauri

KEGG

TAMBIS

BioPAX

DB Schema

RDF(S)

Ontylog

OWL

IEEE SUO

General logical constraints

Formal is-a
Frames
(Properties)

Disjointness,
Inverse

Formal instances

Value Restriction

SNOMED CT

Simple Terminologies

Expressive Ontologies

EcoCyc

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Ontology Dimensions based on McGuinness and Finin
Ontology for terminology development
Ontological principles help disentangle polysemy

- **Distinction among types**
  - Cancer **disorder** (diagnosed/treated by the oncologist) vs. cancer **morphology** (observed by the pathologist)
  - **Substance** (chemical entity) vs. **product** containing the substance (prescribable entity)
Ontology tools help build large ontologies

◆ Description logics (DL)
  ● Subset of first order logic that is computationally tractable
  ● Web Ontology Language (OWL)
  ● Various levels of expressivity
  ● Low expressivity (EL++) sufficient for terminological support

◆ DL reasoners support
  ● Consistency checking
  ● Automatic inference of subclass relations
Primitive vs. defined classes

- **Primitive**
  - Only necessary conditions (for an instance to belong to the class)

- **Defined**
  - Necessary and sufficient conditions for the class
  - Form the basis for inferring relations among classes
    - Equivalence with other defined classes
    - Subclass with other defined or primitive classes
Stated vs. inferred relations

◆ Stated
  - Relations as expressed by the developer
  - Mostly attributes + high-level ancestor

◆ Inferred
  - Relations as computed by the DL reasoned
  - May identify more precise parent classes than what was stated
  - May identify additional parent classes
Small cell carcinoma of lung

Stated

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Small cell carcinoma of lung

Inferred

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Summary
Summary

◆ Ontology and terminology have different purposes
  ● Defining types of things vs. naming things

◆ Gradient between terminology and ontology
  ● Different levels of formality and expressivity

◆ Ontology supports terminology development
  ● Principles for organizing terms consistently
    ■ Ontological distinctions
    ■ Logical/textual definitions (attributes)
  ● Hierarchies can be inferred automatically
  ● Consistency checking