

Vanderbilt University
Data to Knowledge (Clinical Data Standards) [ACI 6130]

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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

- ◆ Bodenreider O, Stevens R. [Bio-ontologies: current trends and future directions](#). *Brief Bioinform.* 2006 Sep;7(3):256-74.
- ◆ Cimino JJ, Zhu X. [The practical impact of ontologies on biomedical informatics](#). *Yearb Med Inform.* 2006:124-35.
- ◆ Bodenreider O. [Biomedical ontologies in action: role in knowledge management, data integration and decision support](#). *Yearb Med Inform.* 2008:67-79.



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



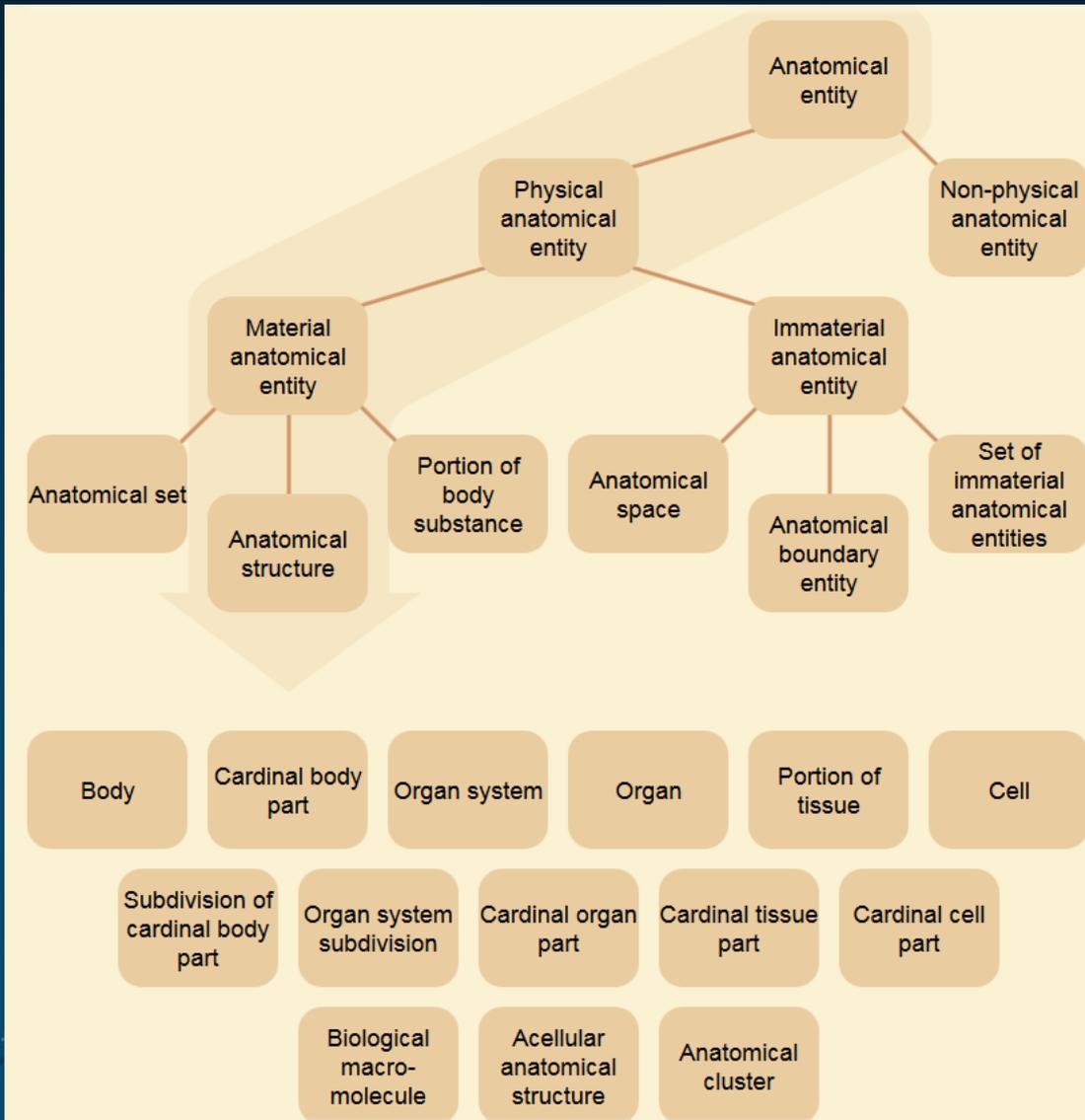
Names for Addison's disease

◆ 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

*Precise
textual
definition*

is regional part of
Upper urinary tract

is constitutional part of

has superclass (is a)
Corticomedullary organ

Kidney

Add to list

has regional part
Anterior inferior renal segment
Anterior superior renal segment
Inferior renal segment
Posterior renal segment
Superior segment of kidney

has constitutional part
Fibrous capsule of kidney
Parenchyma of kidney
Renal pelvis
Renal sinus
Stroma of kidney

Details of current class

Kidney

FMA ID

7203

definition

Corticomedullary 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

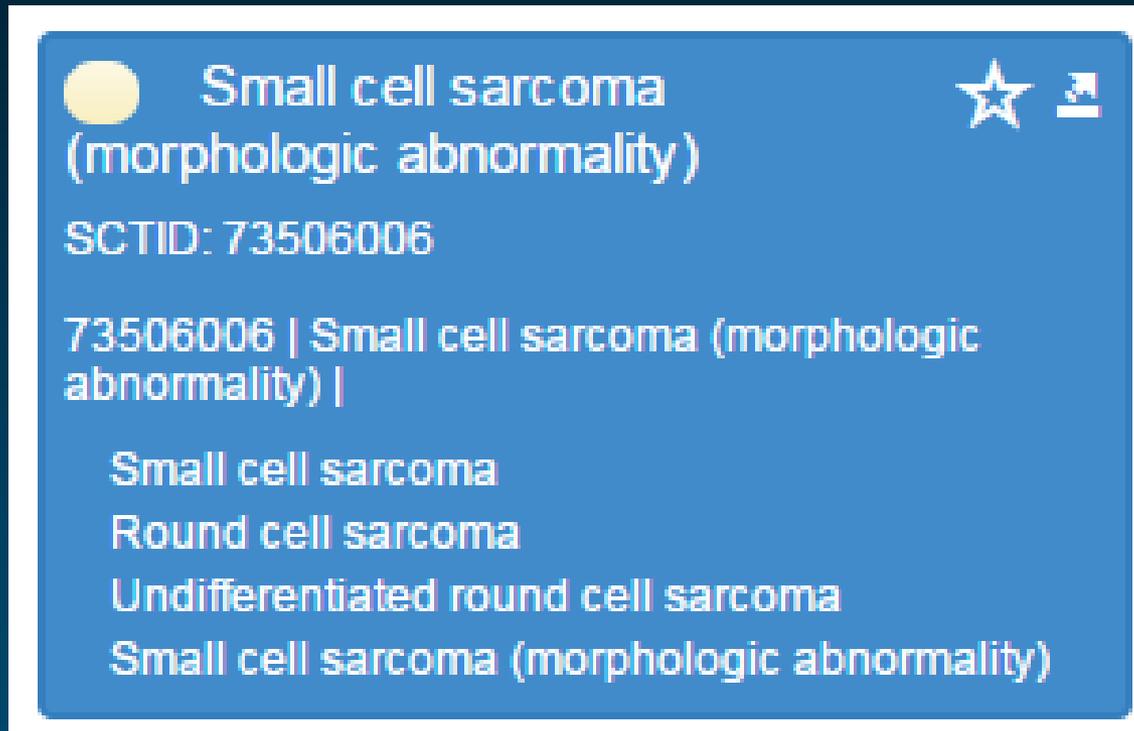
In practice

Terminology vs. ontology (revisited)

- ◆ Lexical and terminological resources
 - Mostly collections of names for biomedical entities
 - Often have some kind of hierarchical organization (e.g., relations)
- ◆ Ontological resources
 - Mostly collections of relations among biomedical entities
 - Sometimes also collect names



Small cell carcinoma Terminology



● Small cell sarcoma (morphologic abnormality)  

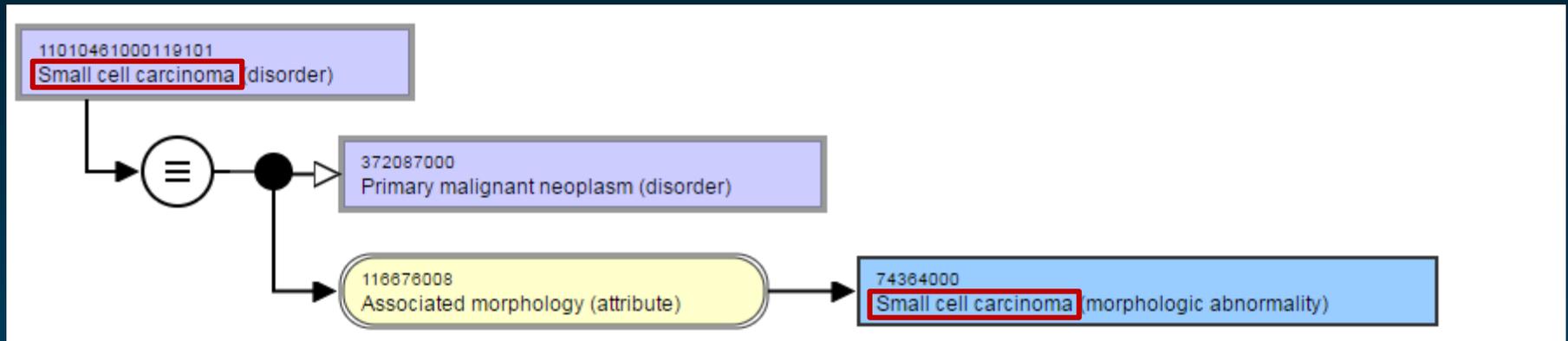
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73506006 | Small cell sarcoma (morphologic abnormality) |

- Small cell sarcoma
- Round cell sarcoma
- Undifferentiated round cell sarcoma
- Small cell sarcoma (morphologic abnormality)

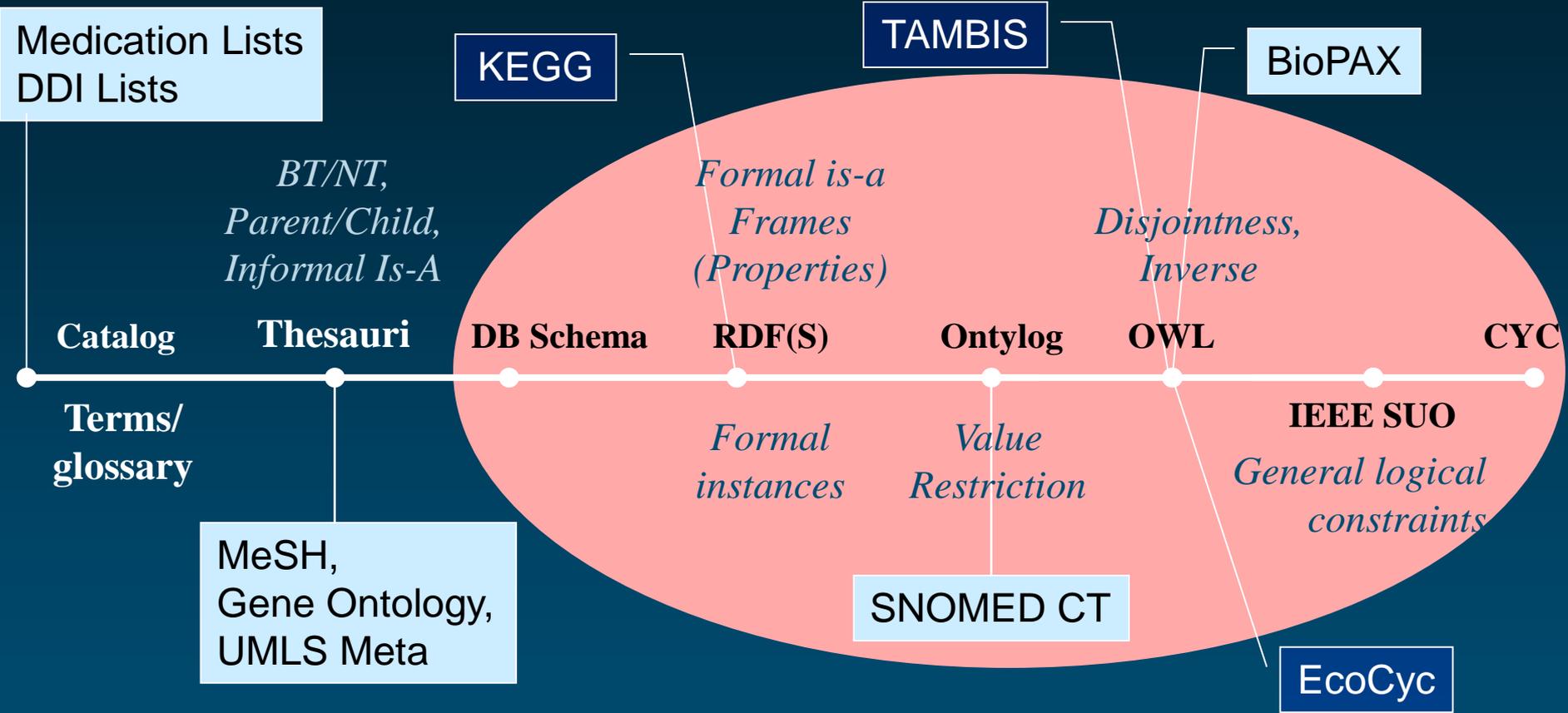
◆ 4 terms (1 fully specified name + 3 synonyms)

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

Ontology spectrum



Simple Terminologies

Expressive Ontologies

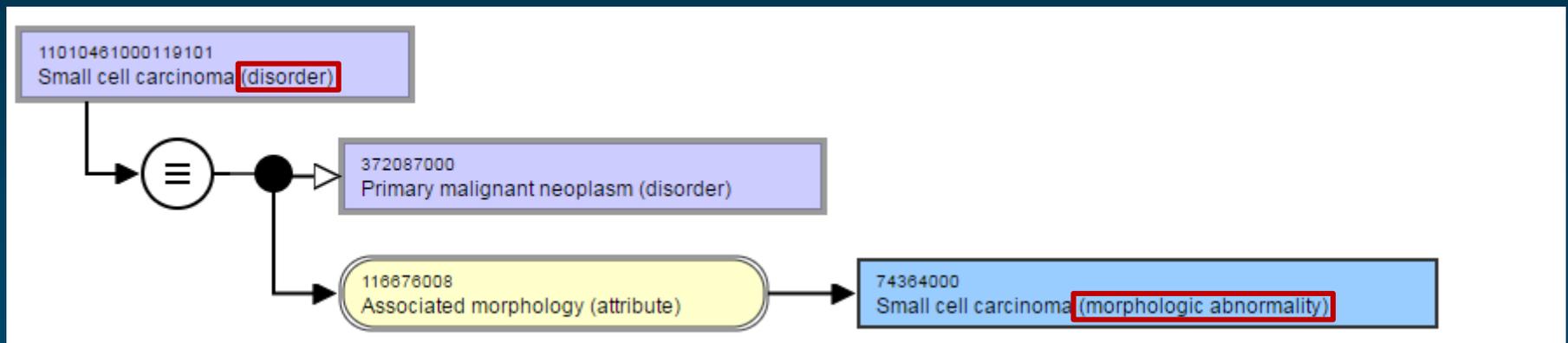


Ontology for terminology development

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

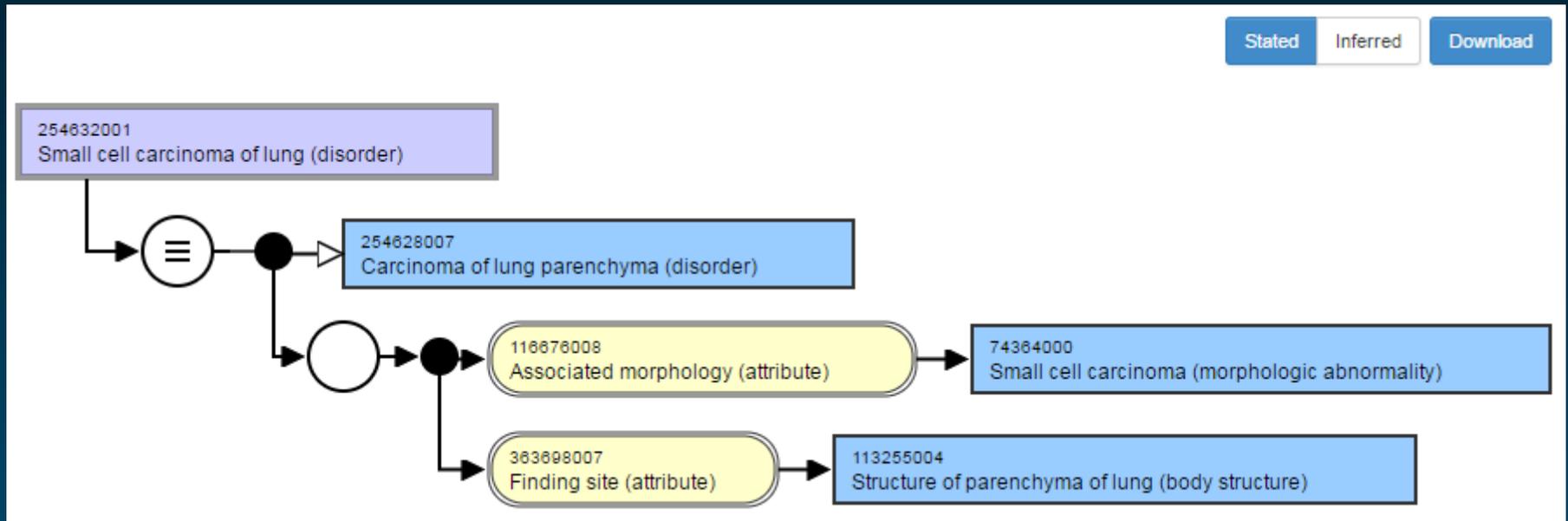
◆ Stated

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

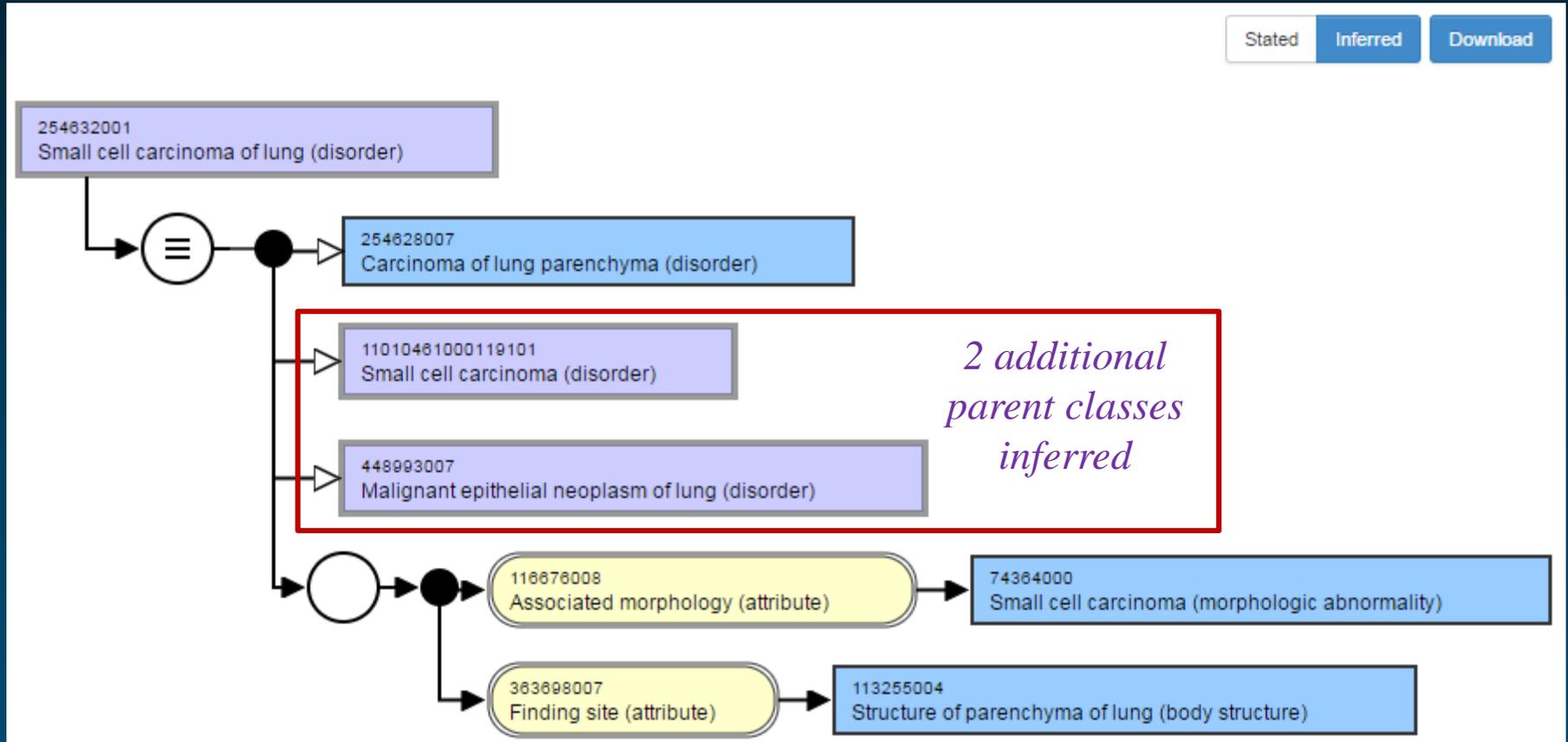
◆ Inferred

- 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



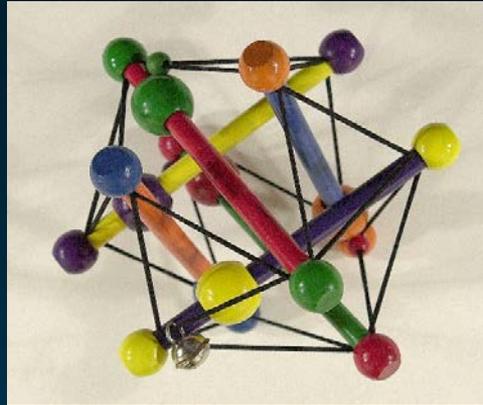
Small cell carcinoma of lung Inferred



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



Medical Ontology Research

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