SNOMED CT's Problem List: Ontologists' and Logicians' Therapy Suggestions

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SNOMED's Ontogenetics

Principles of Formal Ontology

Fusion with CTV 3

Context Model

Logic-based descriptions

SNOMED im UMLS

multiaxial nomenclature of medicine

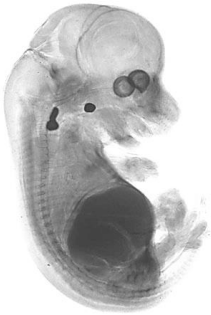
IHTSDO

Nomenclature / Pathology

1965 1970 1975 1980 1985 1990 1995 2000 2005

SNOP SNOMED SNOMED II SNOMED 3.0 SNOMED 3.5 SNOMED RT SNOMED CT

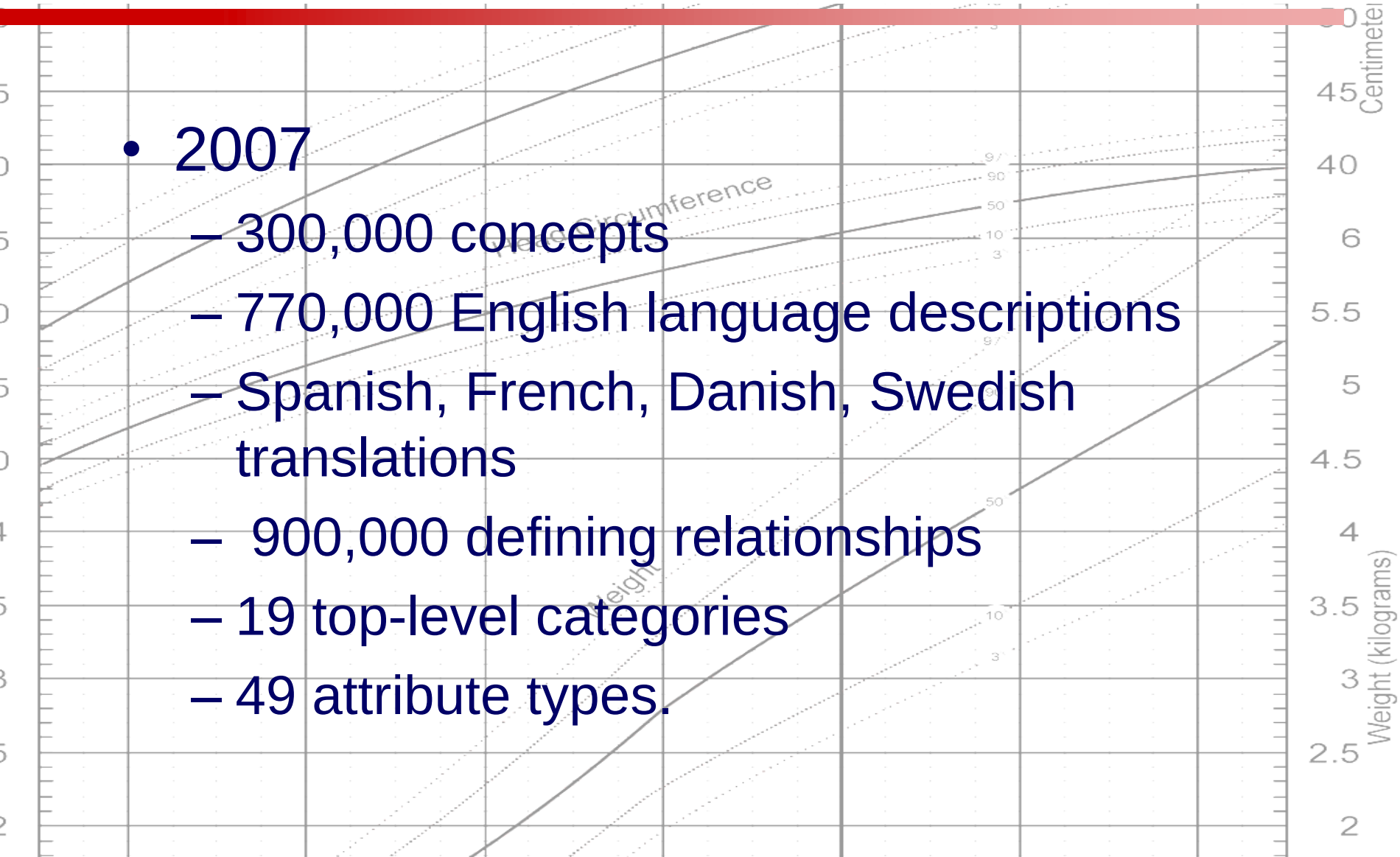
Embryo *Fetus* *Infant* *Child* *Adolescence*



SNOMED's Ecological Niche and Natural Selection

- Changing Habitat:
  - Initially: Pathology
  - Present: Patient Care
  - Future: Whole Life Sciences ?
- Advantages for selection
  - need for globally standardized clinical terminology
  - flood of clinical and scientific data
  - emergence of applied ontology
  - availability of machine reasoning

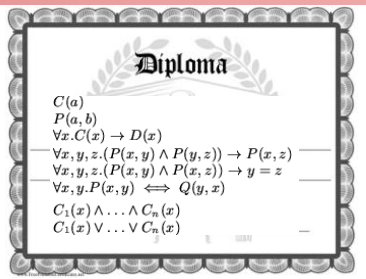
SNOMED's Growth Chart and Vital Parameters



- 2007
  - 300,000 concepts
  - 770,000 English language descriptions
  - Spanish, French, Danish, Swedish translations
  - 900,000 defining relationships
  - 19 top-level categories
  - 49 attribute types.

SNOMED's Health Check

Ontology Consultant



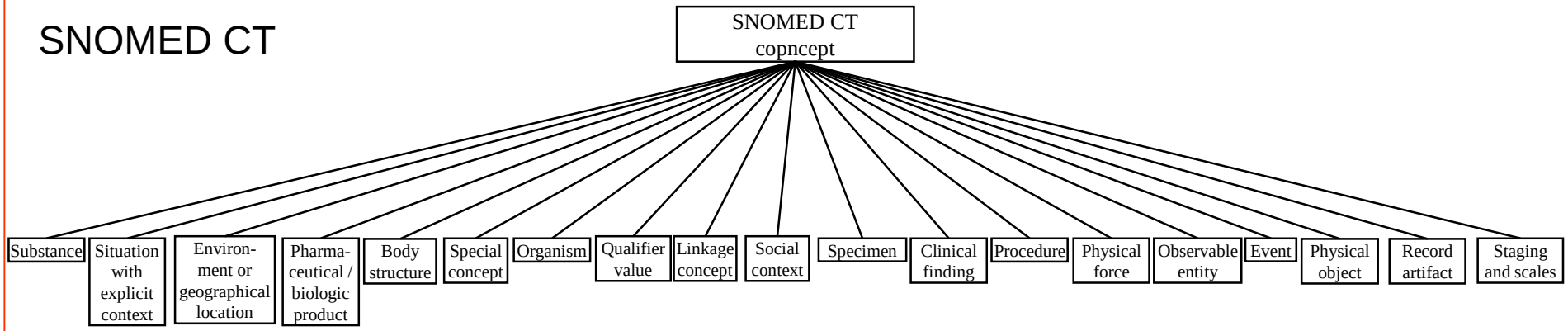
Logics Consultant

SNOMED's Problem List

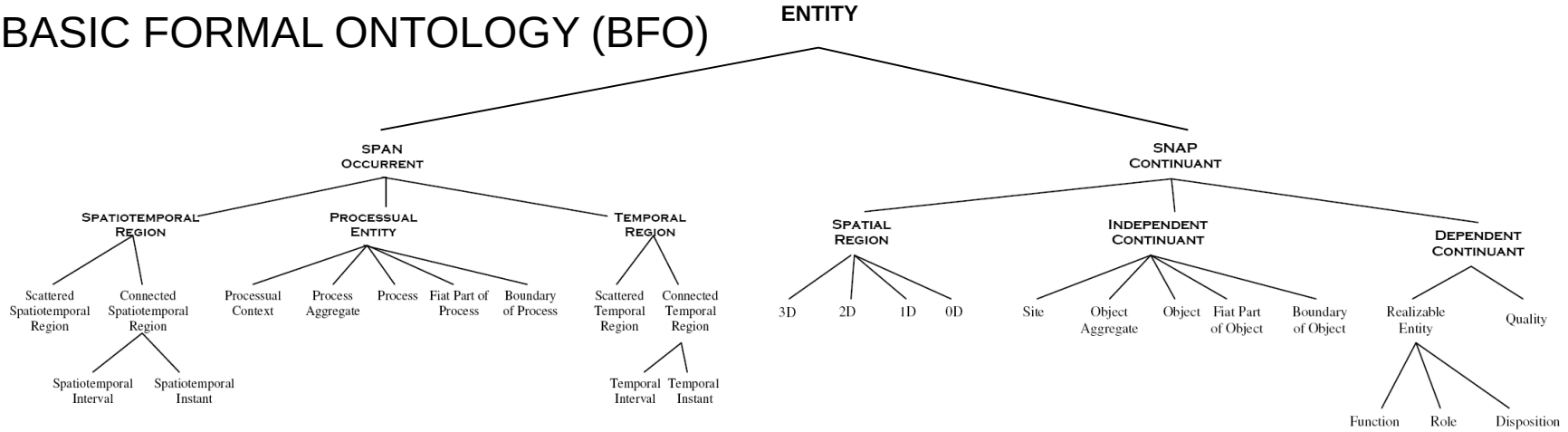
#1	Dystrophic Upper Level	
#2	Concept Borderline Disorder	
#3	Infestation by Individuals	
#4	Relation Idiosyncrasy	
#5	Taxonomic Dystrophy	
#6	SEP Implants	
#7	Partition Agenesis	
#8	Description Asthenia	
#9	Qualifier Syndrome	

Problem #1 Dystrophic Upper Level

SNOMED CT



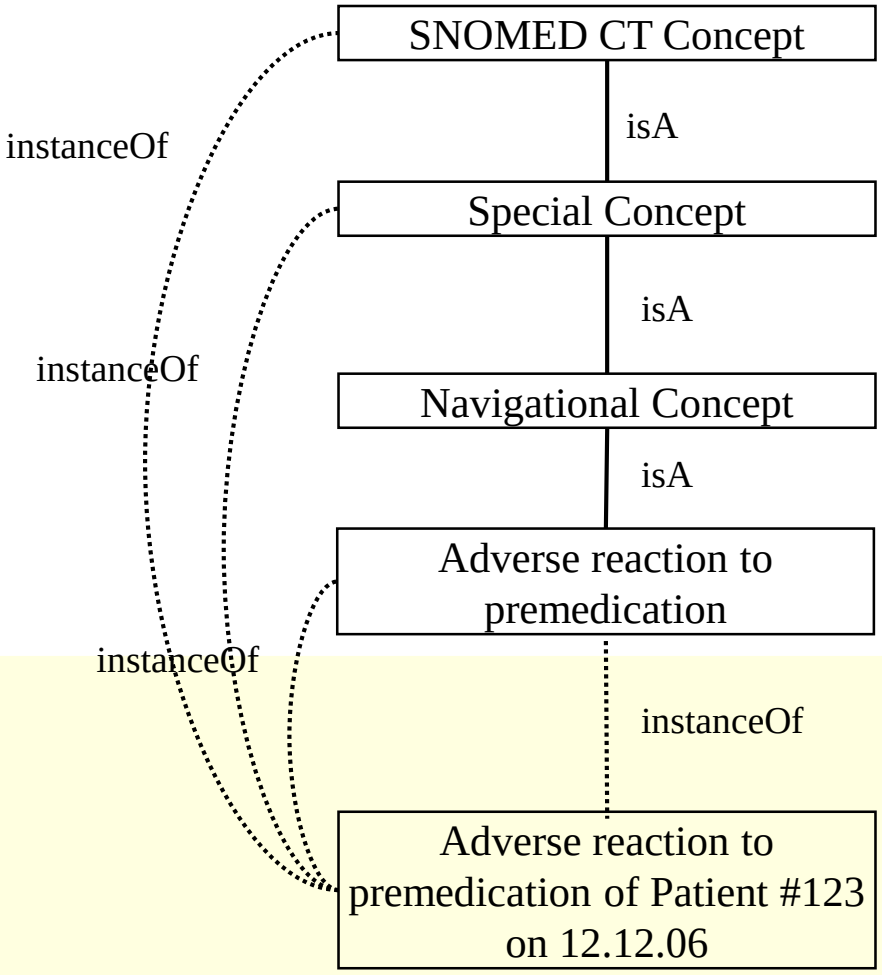
BASIC FORMAL ONTOLOGY (BFO)

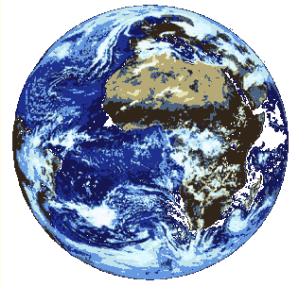


Upper level Ontologies

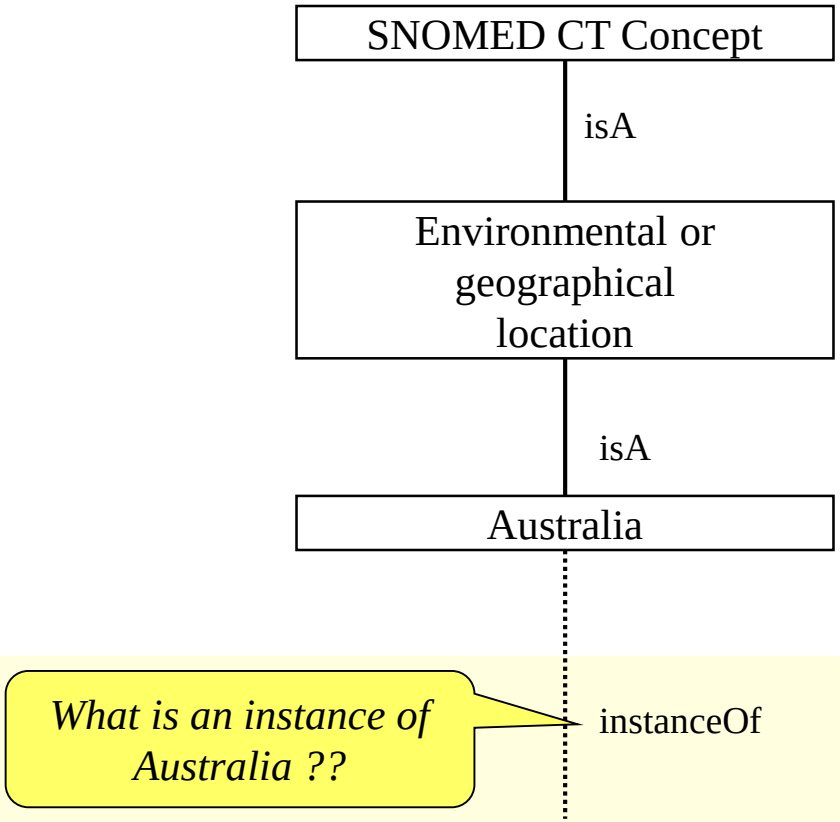
- Provide clearly defined categories that do not overlap
- The meaning of SNOMED CT´s toplevel categories is often unclear and fuzzy
- META-categories (that refer to the concepts) are not clearly distinguished from concepts proper

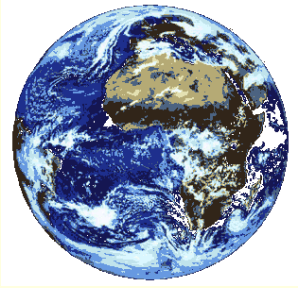
Problem #2 Concept Borderline Disorder





Problem #3 Infestation by Individuals





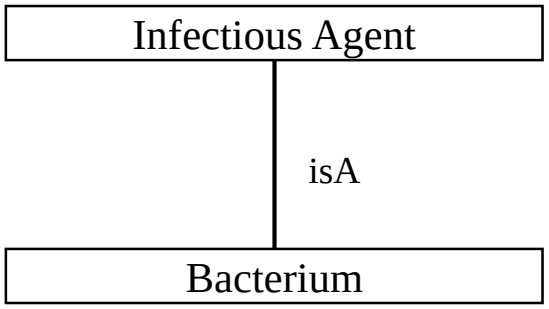
Problem #5:Relation Idiosyncrasy

- Desideratum: use few, clearly defined relations (e.g. OBO relations):
  instance_of, part_of, located_in, adjacent_to, earlier, derives_from, has_participant, has_agent
- SNOMED CT: relations not formally defined, fuzzy ("*Subject Relationship Context*"), obscure ("*Relationship Group*")

Smith B, Ceusters W, Klagges B, Kцhler J, Kumar A, Lomax, J et al. Relations in biomedical ontologies, Genome Biology. 2005;6(5).
Schulz S, Hanser S, Hahn U, Rogers, J. The semantics of procedures and diseases in SNOMED CT, Methods Inf Med. 2006; 45(4): 354-358.

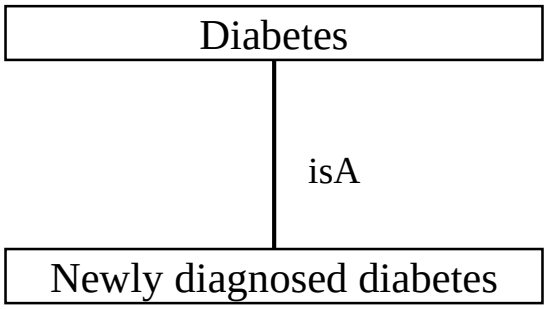
Problem #5 Taxonomic Dystrophy

“Is-A Overloading”



not every instance of bacterium is an infective agent!

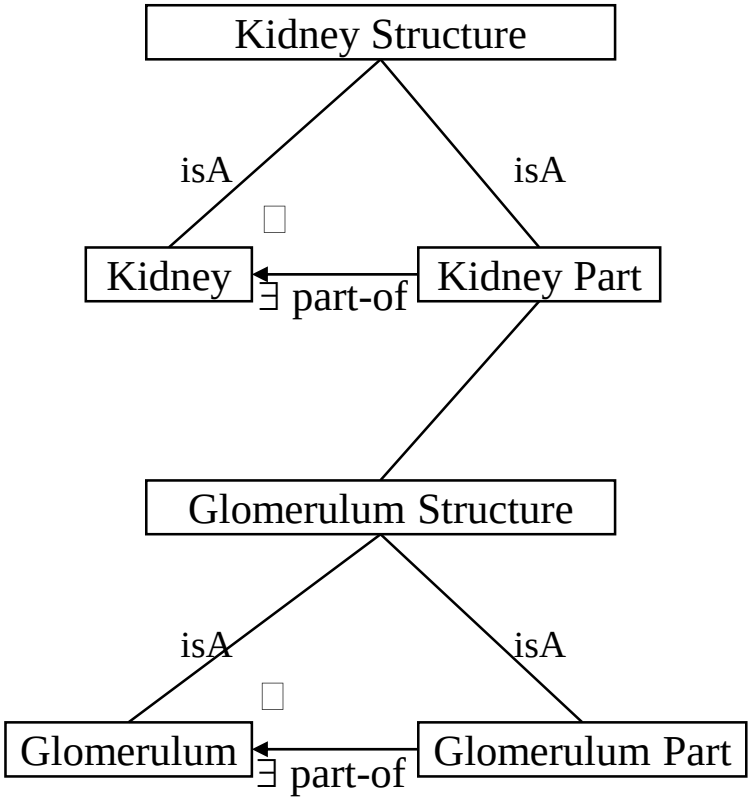
“Epistemological Intrusion”



diabetes as such is not of a different type by the fact that is has recently been diagnosed

Problem #6 SEP Implants

- “Prostheses” for expressing anatomical part-of relations as taxonomies
- Reason: to enable part-of reasoning with 2nd generation terminological reasoners



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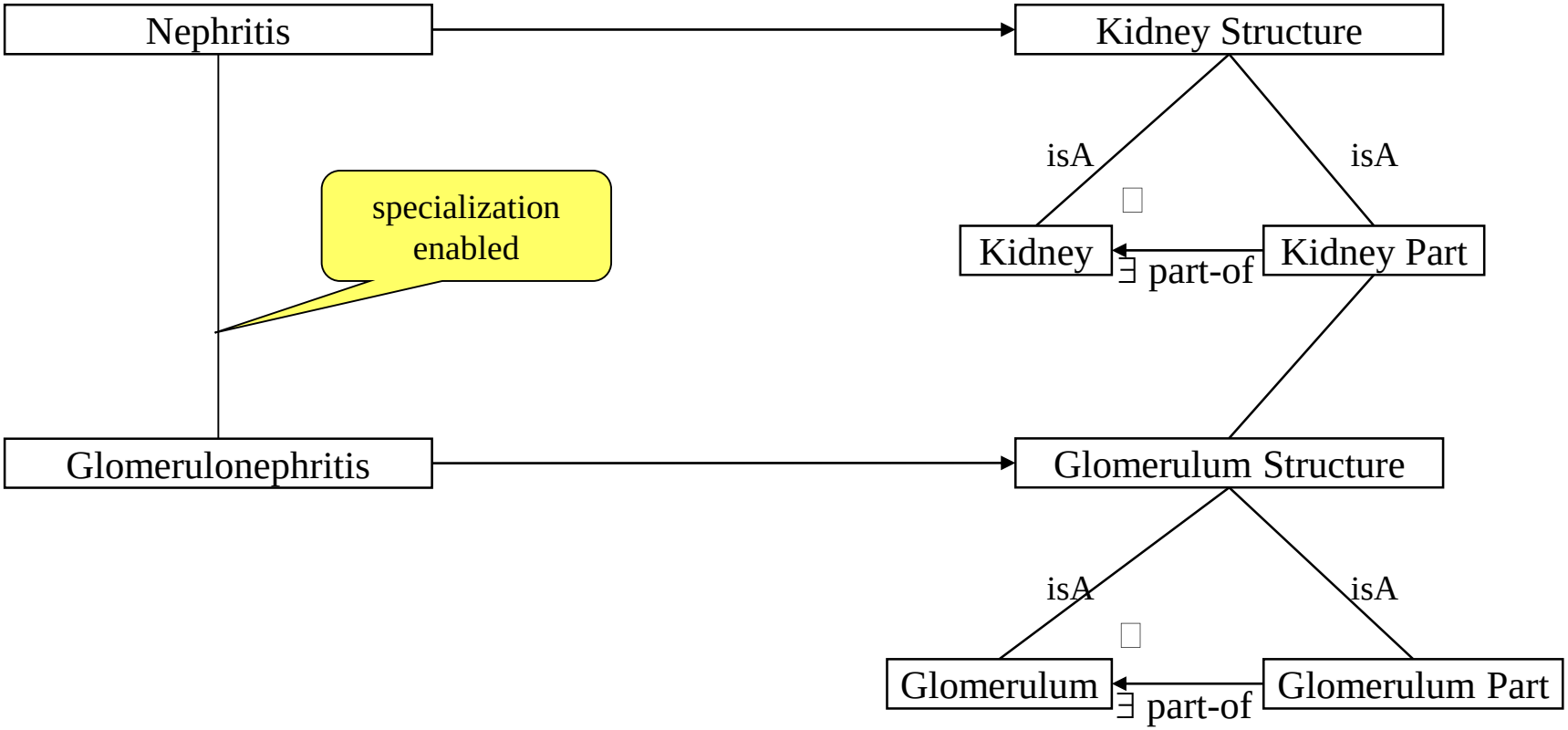

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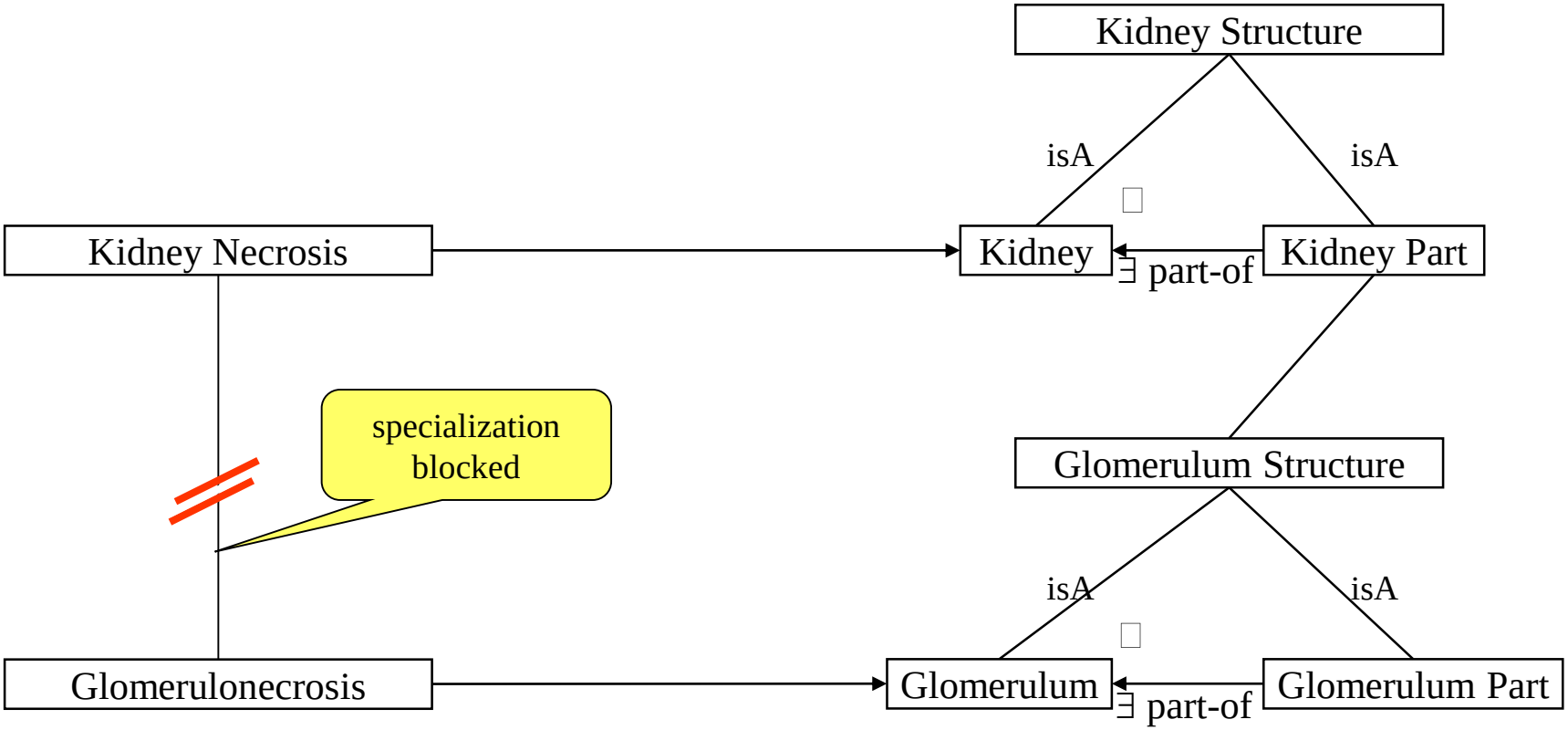
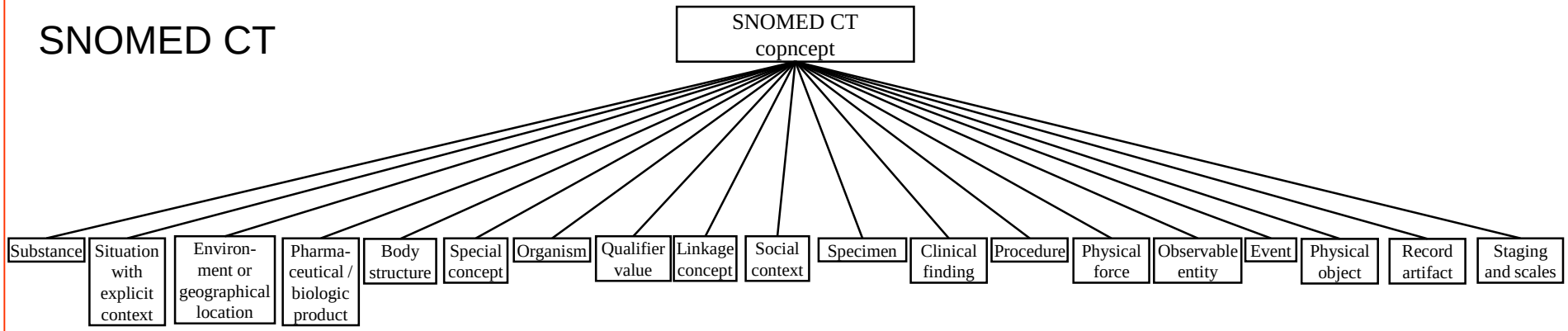


Problem #6 SEP Implants

- SNOMED CT uses the “specialization mode” even where incorrect
- SNOMED CT attaches the same term to the concepts and their “structure” sibling:
  - The term “Kidney” is attached to Kidney and Kidney-Structure. According to the extended SEP hierarchy, Glomerulum is a kind of Kidney structure. Consequently, a Glomerulum is a “Kidney”

Problem #7: Partition Agenesis

SNOMED CT



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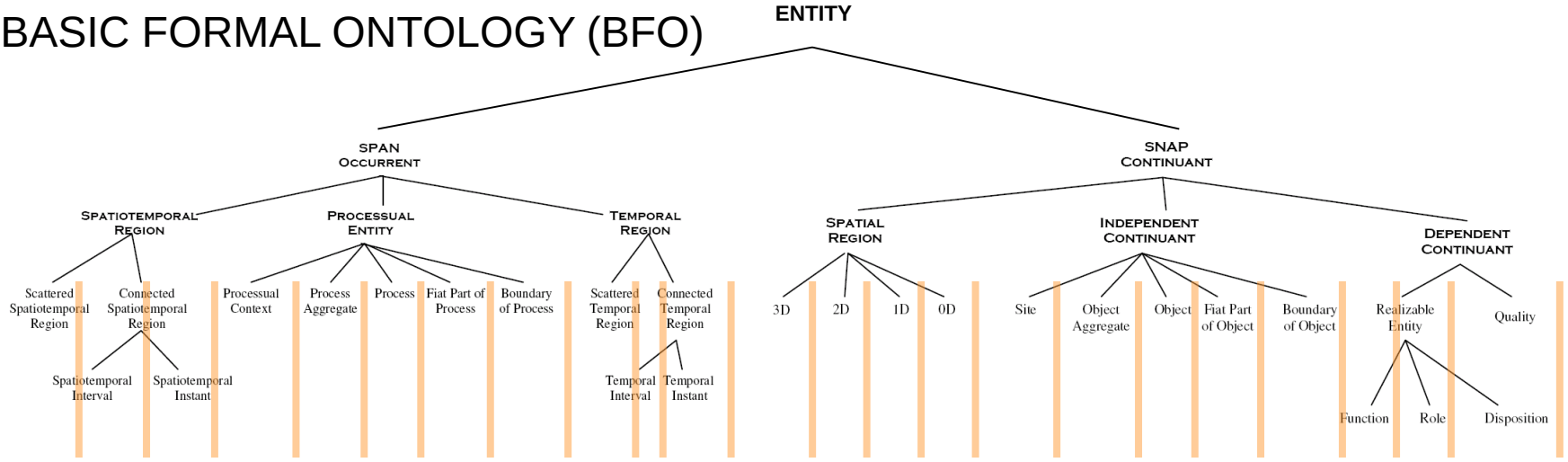

Problem #8: Description Asthenia

- Aristotelian Definitions:
  definiendum ≡ genus ⊓ differentia specifica
  Amputation of Foot ≡ Amputation ⊓ ∃ has-target.Foot
- In SNOMED CT unnecessary primitive Defs:
  Amputation of Foot ⊑ Amputation ⊓ ∃ has-target.Foot

Problem #9: The qualifier syndrome

- SNOMED CT qualifiers:
  laterality, severity, onset, …
- E.g., asthma allows the use of the qualifier severity with the value “severe”
- But the concept “severe asthma” is not defined in terms of this qualifier:
  Severe Asthma ≡ Asthma ⊓ ∃ has-quality.Severe

SNOMED's Treatment Plan

#1	Dystrophic Upper Level	Level Rectification
#2	Co	s Isolation
#3	Infe	olerance of Individuals
#4	Re	econstruction
#5	Tax	leansing
#6	SE	and Substitution
#7	Partition Agenesis	Taxonomy Partitioning Operation
#8	Description Asthenia	Definition Revitalization
#9	Qualifier Syndrome	Qualifier Transplant

For Details, see SNOMED CT's Health Record:

General Therapeutic Principles

- Adhesion to ontological standards: Upper-level Ontology (DOLCE, BFO), Open Biomedical Ontology (OBO) Relation Ontology
- Adhesion to logical standards: W3C Ontology Web Language (OWL-DL)
- Adoption of a computationally tractable description language (CEL++)
- Demarcate crisp interfaces
  - internally: between ontology and terminology components
  - externally: between SNOMED and information models
- Follow good classification principles

The Celestial Emporium of Benevolent Knowledge

Jorge Luis Borges

"On those remote pages it is written that animals are divided into:

a. those that belong to the Emperor
b. embalmed ones
c. those that are trained
d. suckling pigs
e. mermaids
f. fabulous ones
g. stray dogs
h. those that are included in this classification
i. those that tremble as if they were mad
j. innumerable ones
k. those drawn with a very fine camel's hair brush
l. others
m. those that have just broken a flower vase
n. those that resemble flies from a distance"