SNOMED CT – Tutorial

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Please open the SNOMED CT browser at:

https://browser.ihtsdotools.org

SNOMED CT Tutorial @JOWO 2019

Everything Ontologists* Always Wanted to Know about SNOMED CT** (**But Were Afraid to Ask)

* and potential SNOMED implementers / content creators

https://www.maxpixel.net/Paws-French-Bulldog-Curious-Vigilant-Dog-Expectant-4372435

SNOMED CT Tutorial @JOWO 2019

Everything Ontologists* Always Wanted to Know about SNOMED CT** (**But Were Afraid to Ask)

- Why SNOMED CT is out there?
- Where does it come from?
- Which are its scenarios of use
- Who owns SNOMED CT?
- How is its content maintained?
- Is SNOMED CT an ontology?
- Will SNOMED CT ever become an ontology?
- For who doesn't not care about ontologies…
  - Why should I care about SNOMED CT?
  - How can I implement and use it?
  - Which is the benefit of using it?
- For all: how can I learn more about it?

* and potential SNOMED implementers / content creators

https://www.maxpixel.net/Paws-French-Bulldog-Curious-Vigilant-Dog-Expectant-4372435

Schedule

10:30 – 11:00	SNOMED CT history and foundations
11:00 – 11:30	COFFEE BREAK
11:30 – 12:30	SNOMED CT content
12:30 – 14:00	LUNCH BREAK
14:00 – 15:00	Breakout sessions: 1. SNOMED CT in Austria 2. Ontological aspects of SNOMED CT

Type of tutorial

- Interactive
- Hands-on
  (Web browser + Internet)

Participants' profile

- Who are you?
- Why are you attending this tutorial?
- What do you want to learn from this tutorial?

Objectives of the Tutorial

- Objective

  To understand

  - The need of standardizing the electronic health record (EHR)
  - SNOMED's legacy
  - Key ontological aspects of SNOMED CT content
  - SNOMED content development and advanced implementation

  To discuss

  Ontological aspects of SNOMED CT

  SNOMED CT in Austria

Context: The electronic health record (EHRs)

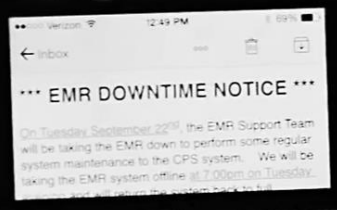
- Documentation is a crucial task in all health care process – and a duty of health care professionals
- Paper documentation in health care is being rapidly substituted by EHRs
- Clinicians are more and more flooded by information that are crucial for decision-making
- There is increasing awareness that EHR content are a precious resource to be exploited for
  - Decision making
  - Observational research
  - Quality assurance
  - Prediction
- There are high expectations that new AI techniques revolutionize the way how health data are used

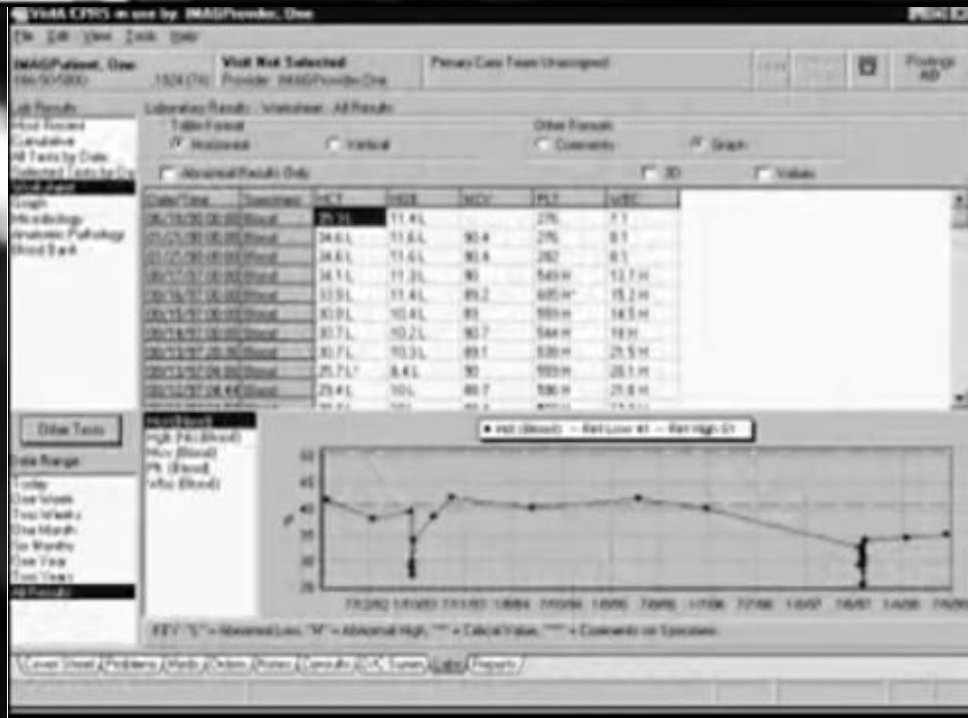
"I'm treating the computer screen"

"Innovation all around but it ain't in healthcare, Internet and apps for you, but we get ancient software"

"Just a glorified billing platform
with some patient stuff tacked on…"

"some be sayin' it's epic we sayin it's epic fail"



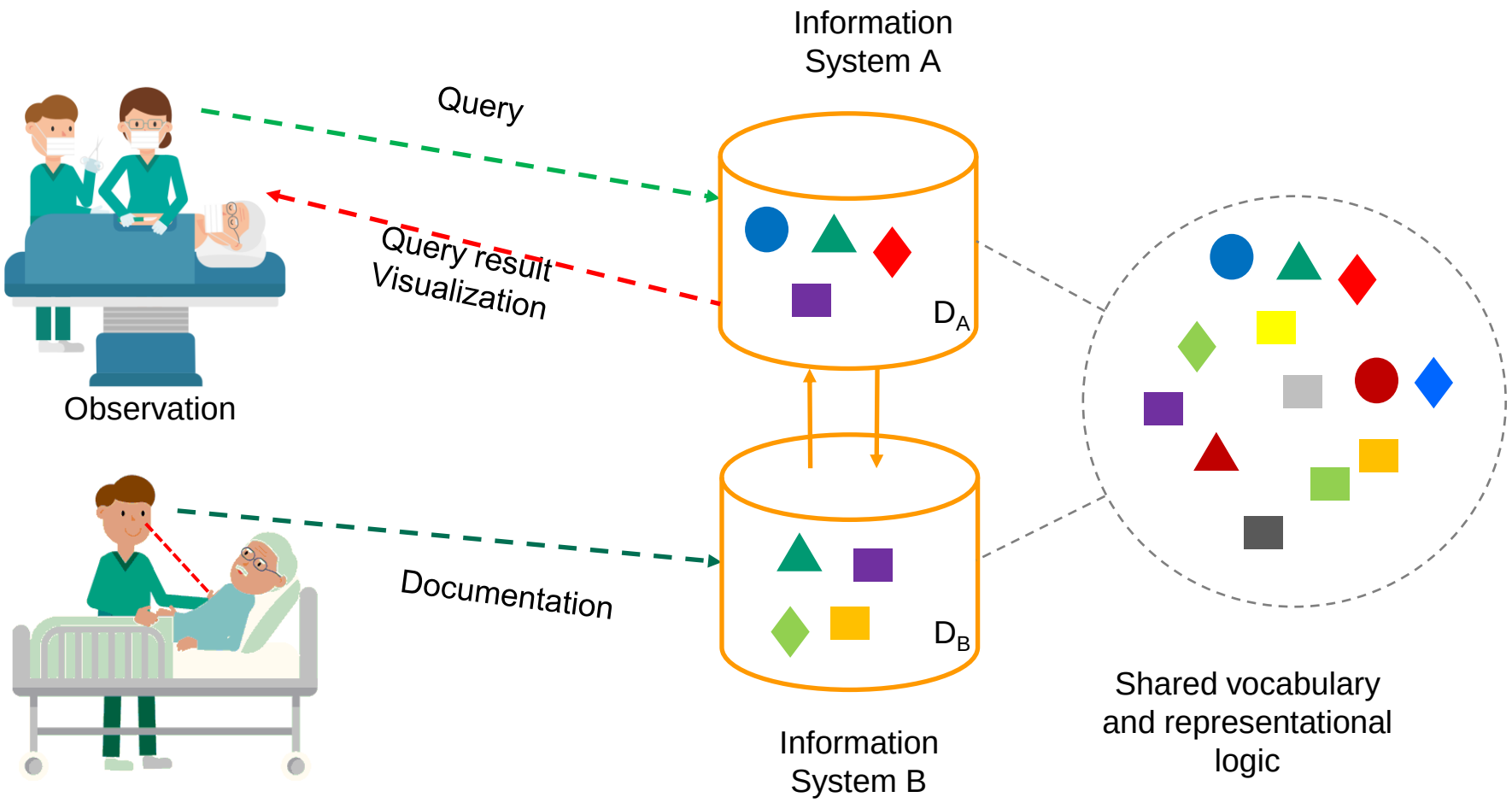


Current EHR shortcomings

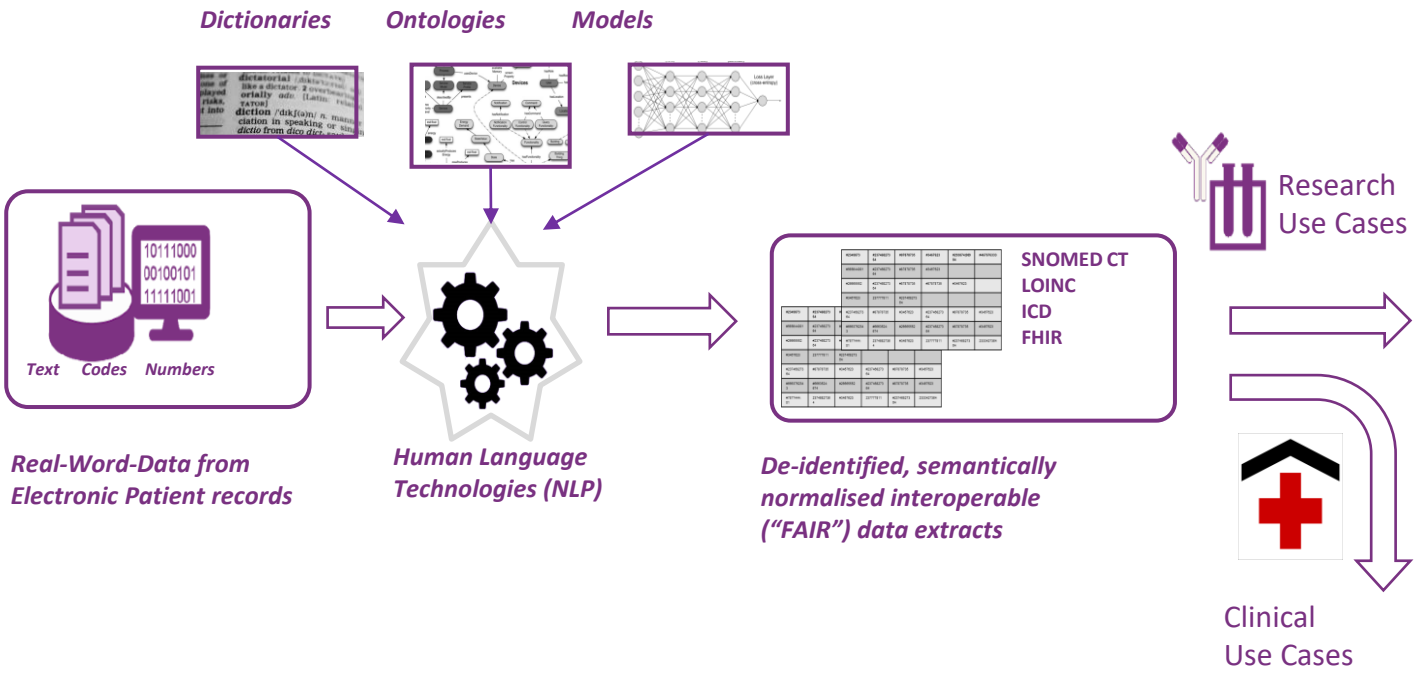
- Many commercial EHR systems are little mo[illegible]han substitutes of paper charts with 199[illegible]el
- Most content in EHR systems i[illegible]osed of local language, Englis[illegible]
- Much structur[illegible]ters) is not standar[illegible]
- A[illegible]n (and biased) by its use for b[illegible] codes, local procedure codes)
- EH[illegible] is also affected by modality of data entry
  - [illegible]yped by clinicians
  - Dictated and typed by typist
  - Speech recognition
  - Codes entered by clinicians
  - Codes entered by coders
  - Forms with picklists, checkboxes
  - Data produced by devices

Semantic Interoperability ?

Semantic interoperability?



EHR interoperability project in Graz



Medizinische Universität Graz

CBmed BIOMARKER RESEARCH

averbis text analytics

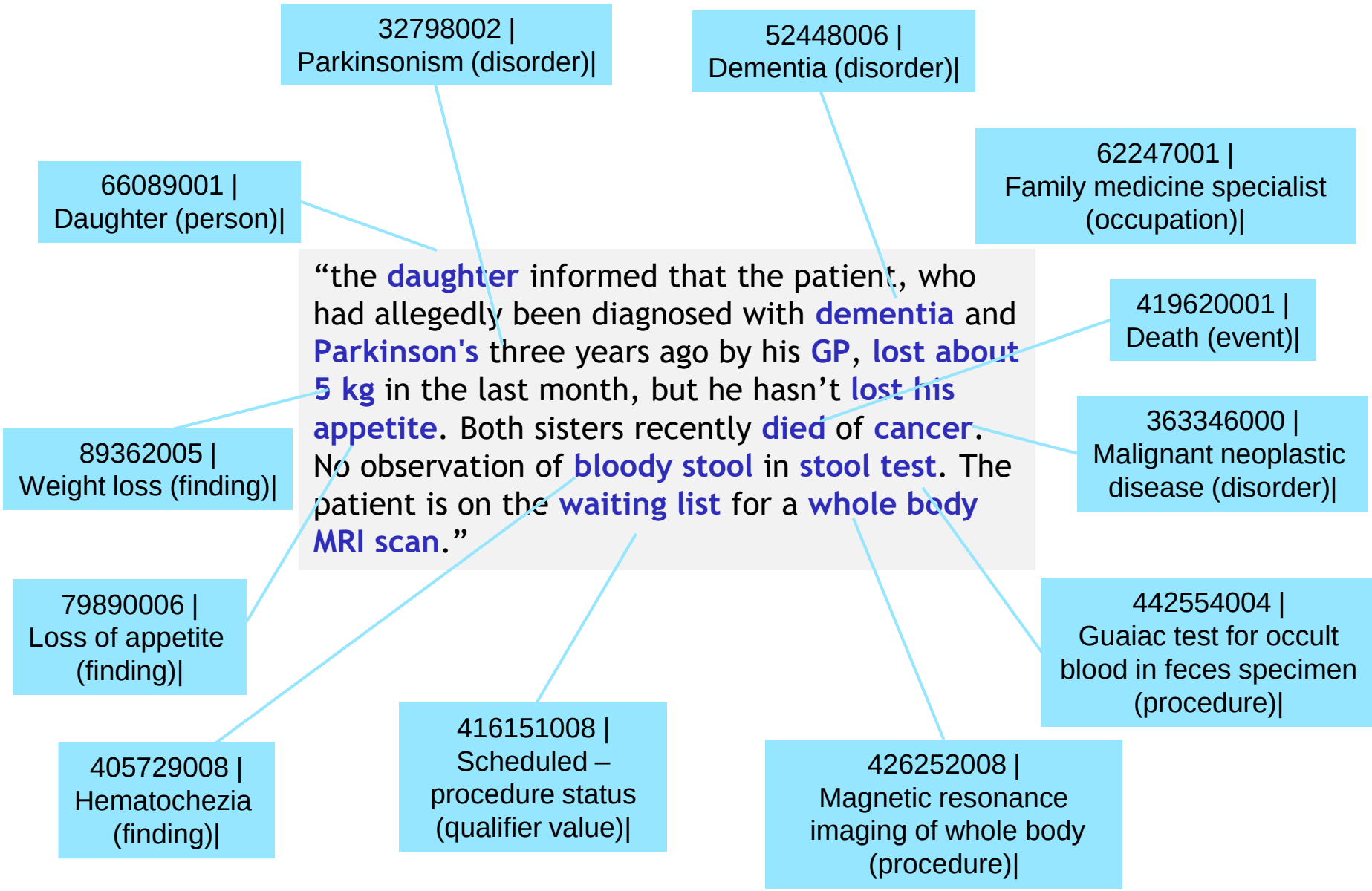
KAGes

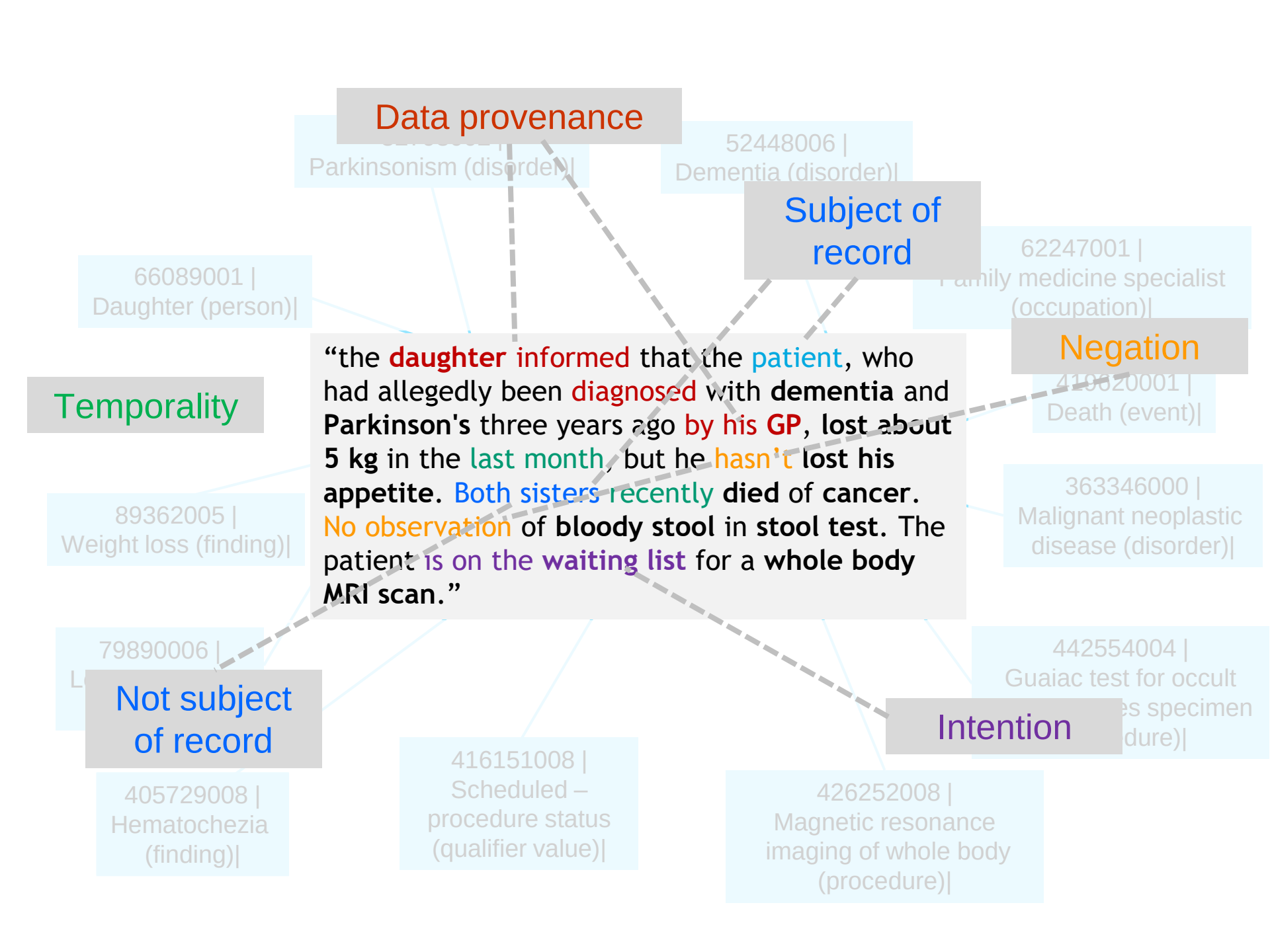
Roche Diagnostics

Desiderata

- Domain terms and expressions in different languages are related to some system of internationally standardised meaning (representational units in a formal model of meaning)
- The contexts in which these representational units are used in the EHR are made explicit in terms of temporality, intentionality, certainty, polarity, etc.

> “the daughter informed that the patient, who had allegedly been diagnosed with dementia and Parkinson's three years ago by his GP, lost about 5 kg in the last month, but he hasn’t lost his appetite. Both sisters recently died of cancer. No observation of bloody stool in stool test. The patient is on the waiting list for a whole body MRI scan.”


32798002 | Parkinsonism (disorder)|
52448006 | Dementia (disorder)|
66089001 | Daughter (person)|
62247001 | Family medicine specialist (occupation)|
"the daughter informed that the patient, who had allegedly been diagnosed with dementia and Parkinson's three years ago by his GP, lost about 5 kg in the last month, but he hasn't lost his appetite. Both sisters recently died of cancer. No observation of bloody stool in stool test. The patient is on the waiting list for a whole body MRI scan."
419620001 | Death (event)|
89362005 | Weight loss (finding)|
363346000 | Malignant neoplastic disease (disorder)|
79890006 | Loss of appetite (finding)|
442554004 | Guaiac test for occult blood in feces specimen (procedure)|
405729008 | Hematochezia (finding)|
416151008 | Scheduled – procedure status (qualifier value)|
426252008 | Magnetic resonance imaging of whole body (procedure)|


Data provenance
Parkinsonism (disorder)|
52448006 |
Dementia (disorder)|
Subject of record
62247001 |
Family medicine specialist (occupation)|
66089001 |
Daughter (person)|
Negation
Temporality
419620001 |
Death (event)|
"the daughter informed that the patient, who had allegedly been diagnosed with dementia and Parkinson's three years ago by his GP, lost about 5 kg in the last month, but he hasn't lost his appetite. Both sisters recently died of cancer. No observation of bloody stool in stool test. The patient is on the waiting list for a whole body MRI scan."
89362005 |
Weight loss (finding)|
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Hematochezia (finding)|
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Scheduled – procedure status (qualifier value)|
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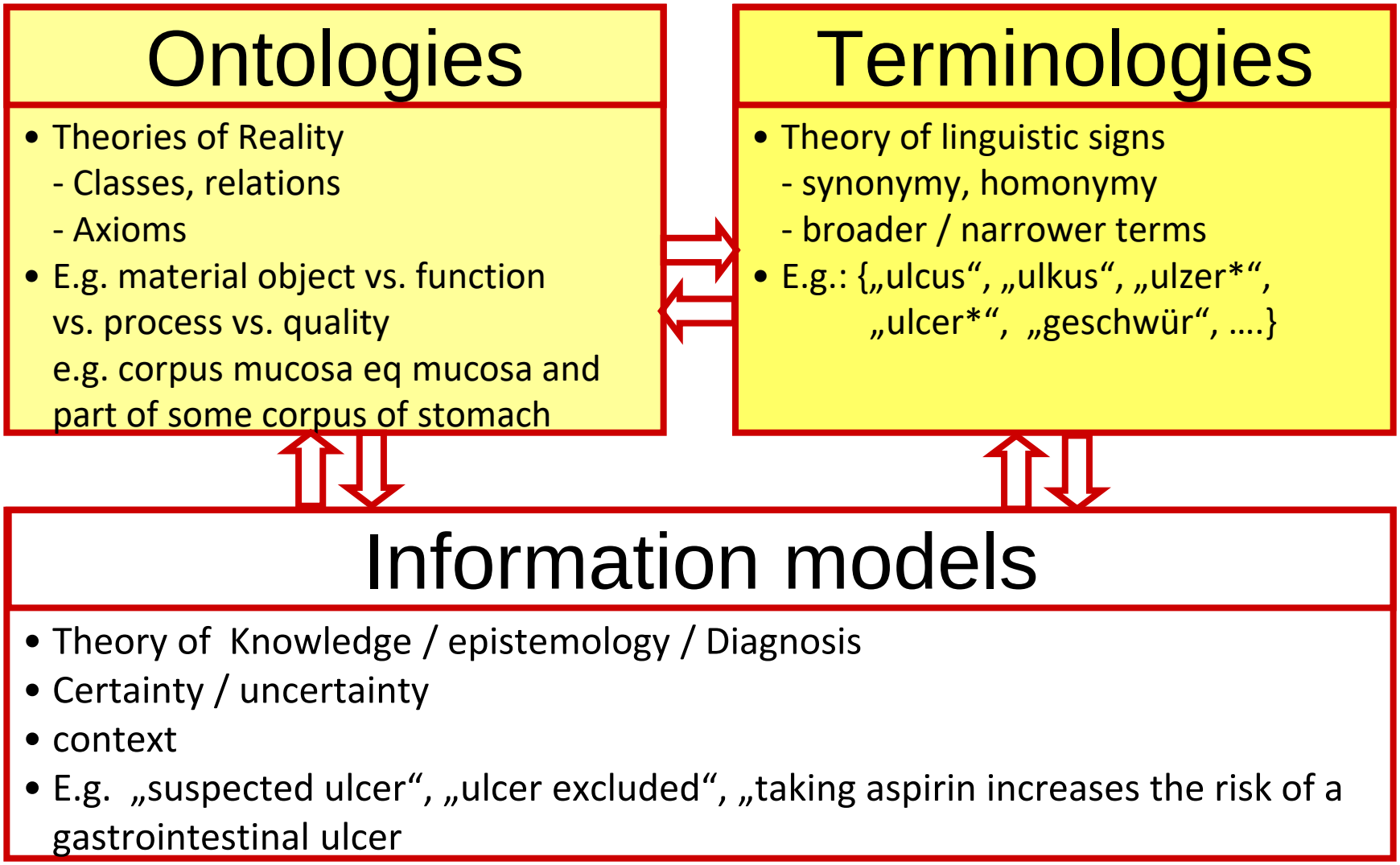

Ontologies
• Theories of Reality
- Classes, relations
- Axioms
• E.g. material object vs. function vs. process vs. quality
e.g. corpus mucosa eq mucosa and part of some corpus of stomach
Terminologies
• Theory of linguistic signs
- synonymy, homonymy
- broader / narrower terms
• E.g.: {„ulcus“, „ulkus“, „ulzer*“, „ulcer*“, „geschwür“, ….}
Information models
• Theory of Knowledge / epistemology / Diagnosis
• Certainty / uncertainty
• context
• E.g. „suspected ulcer“, „ulcer excluded“, „taking aspirin increases the risk of a gastrointestinal ulcer

Health and biomedical vocabularies

- Classifying causes of death: Bills of Mortality, 1592 to 1595
- International Statistical Classification of Diseases, Paris 1900, WHO 1946 (ICD-6), 1994 (ICD-10), 2017 (ICD-11 MMS)
- Thousands of domain and purpose specific vocabularies (terminologies, classifications) around the globe
- UMLS Metathesaurus systematises and maps currently 214 of them https://www.nlm.nih.gov/research/umls/sourcereleasedocs/index.html

9th Conf. int. Soc. geogr. Pathol., Leiden 1966
Path. Microbiol. *30:* 826–827 (1967)

1966

Columbia University, College of Physicians and Surgeons, New York, N.Y.

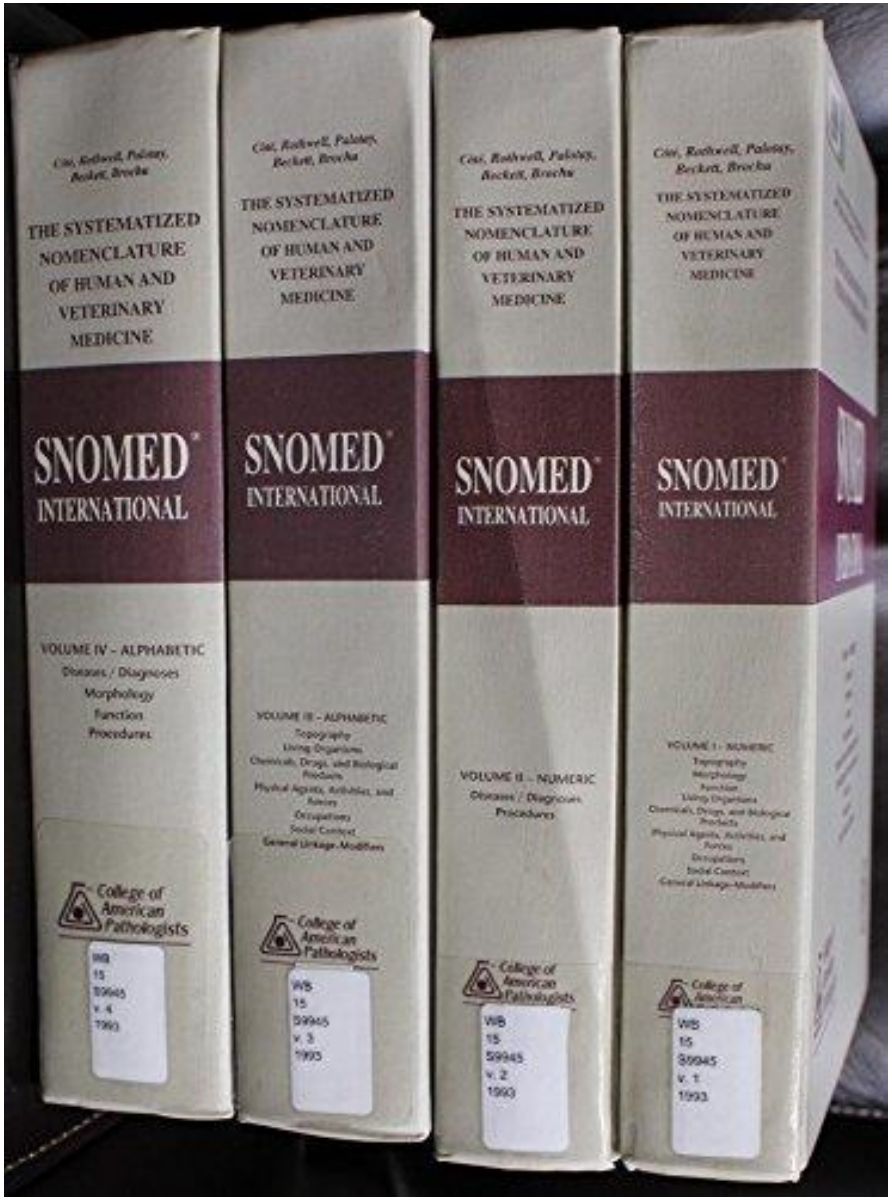
Systematized Nomenclature of Pathology

S. C. SOMMERS, New York, N.Y.

The Systematized Nomenclature of Pathology (SNOP) was developed for pathologists and other medical workers, to provide a convenient modern method for the storage and retrieval of laboratory data. Under the auspices of the Committee on Nomenclature and Classification of Disease, the system was developed and published by the College of American Pathologists in 1965.

SNOP was intended to serve the major reasons for storing medical information, such as (1) routine orderliness, (2) statistical reports, (3) case finding for a) teaching, b) conferences, and c) research. Before its publication SNOP was field-tested by 147 pathologists, who used it to store 234 408 surgical diagnoses and 4888 autopsy diagnoses. Of those testing the code, 94 per cent reported intending to continue using SNOP.

The organization of SNOP involves four fields of information: Topography (T), the anatomical site; Morphology (M), the morbid anatomical lesion; Etiology (E), the causative agent; and Function (F), the pathophysiological change. Any field may be used alone, or any combination of fields may be employed. The topographic field exists in two forms: (1) a two-digit numerical system with a duodecimal base, permitting simplified assignments of anatomic sites, such as T43 for coronary artery; and (2) a compatible expanded four-digit system permitting more anatomical detail, for



SNOMED

Systematisierte Nomenklatur der Medizin

Band I Numerischer Index

Herausgeber der amerikanischen Ausgabe
Roger A. Côté

Deutsche Ausgabe bearbeitet und adaptiert von
Friedrich Wingert

Springer-Verlag Berlin Heidelberg GmbH

SNOMED's history - From a nomenclature to an ontology-based terminology

Nomenclature for pathology

Multiaxial nomenclature for the whole medicine

Description logics framework

Fusion mit CTV3

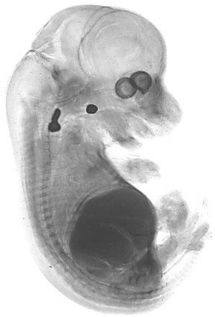
SNOMED Context model

IHTSDO Standard

SNOMED International

1965 1970 1975 1980 1985 1990 1995 2000 2005 2010 2015 2020

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



Cornet R, de Keizer N. Forty years of SNOMED: a literature review. *BMC Med Inform Decis Mak*. 2008;8 Suppl 1(Suppl 1):S2

SNOMED's history - From a nomenclature to an ontology-based terminology

- SNOP – "Standardised Nomenclature of Pathology"
- SNOMED – "Standardised Nomenclature of Medicine"
- SNOMED CT – "Standardised Nomenclature of Medicine – Clinical terms"
- SNOMED CT - "~~Standardised Nomenclature of Medicine~~ – Clinical terms“
- Typology
  - Nomenclature: system of naming
  - Terminology: collection of technical words and expressions
  - (Formal) Ontology: formal account of characteristics of (classes) of entities

Original idea:
Nomenclature: standardised names

D5-46210	Acute appendicitis, NOS

D5-46100	Appendicitis, NOS
G-A231	Acute

M-41000	Acute inflammation, NOS
G-C006	In
T-59200	Appendix, NOS

G-A231	Acute
M-40000	Inflammation
G-C006	In
T-59200	Appendix, NOS

- The same meaning can be expressed by different expressions
- No formal-sematic foundation of co-ordination of codes

Systematized NOmenclature of MEDicine V3.5 CTV3 (Clinical Terms v3 – ex UM NHS Read Codes)

D5-46210 Acute appendicitis, NOS

D5-46100 Appendicitis, NOS
G-A231 Acute

M-41000 Acute inflammation, NOS
G-C006 In
T-59200 Appendix, NOS

G-A231 Acute
M-40000 Inflammation
G-C006 In
T-59200 Appendix, NOS

RCGP/NHS Diabetes Classification of diabetes category and sub-categories	Read code Acronym	Clinical Terms version 3 (CTv3)	Rubric
RCGP/NHS Diabetes Classification of diabetes			
Type 1	T1DM	X40J4	Type 1 diabetes mellitus
Type 2	T2DM	X40J5	Type 2 diabetes mellitus
Genetic		X40JG	Genetic syndromes of diabetes mellitus
		X40JI	Diabetes mellitus autosomal dominant
Maturity-onset DM in the young	MODY	XSETH	Maturity-onset diabetes in youth (when used with term code 11, or 12)
		X40JJ	Diabetes mellitus autosomal-dominant type 2
		XaOPt	Maternally inherited DM
Other			
Generic		X40JA	Secondary diabetes mellitus
		XaMzI	Cystic-fibrosis-related diabetes mellitus
		XSETK	Drug-induced diabetes mellitus
Steroid induced		C11y0	Diabetes mellitus induced by steroids
		XaJUI	Diabetes mellitus induced by non-steroid drugs
		X40JB	Secondary pancreatic diabetes mellitus
Unknown			
Suspected		XaXPB	Suspected diabetes mellitus
When diagnosis excluded		XaFvt	Use this code when diabetes excluded
Non-diabetic hyperglycaemia			
Impaired glucose tolerance and impaired fasting glucose			
Impaired glucose tolerance	IGT	X40Jh	Impaired glucose tolerance
		X40JI	Impaired glucose tolerance in pregnancy
Glycated haemoglobin defined IGT	HbA1c defined IGT	XaPbt XaERp	IFCC Range 42–47 mmol/mol** DCCT Range 6.0–6.49%**
Impaired fasting glucose	IFG	XaIRY	Impaired fasting glycaemia
Gestational diabetes			
Gestational	GDM	L1808	Diabetes mellitus arising in pregnancy

** Subject to confirmation in NICE guidance.

SNOMED RT

SNOMED CT

Description logics

Source: de Lusignan et al. Call for consistent coding in diabetes mellitus using the Royal College of General Practitioners and NHS pragmatic classification of diabetes February 2012The Journal of Innovation in Health Informatics 20(2):103-113

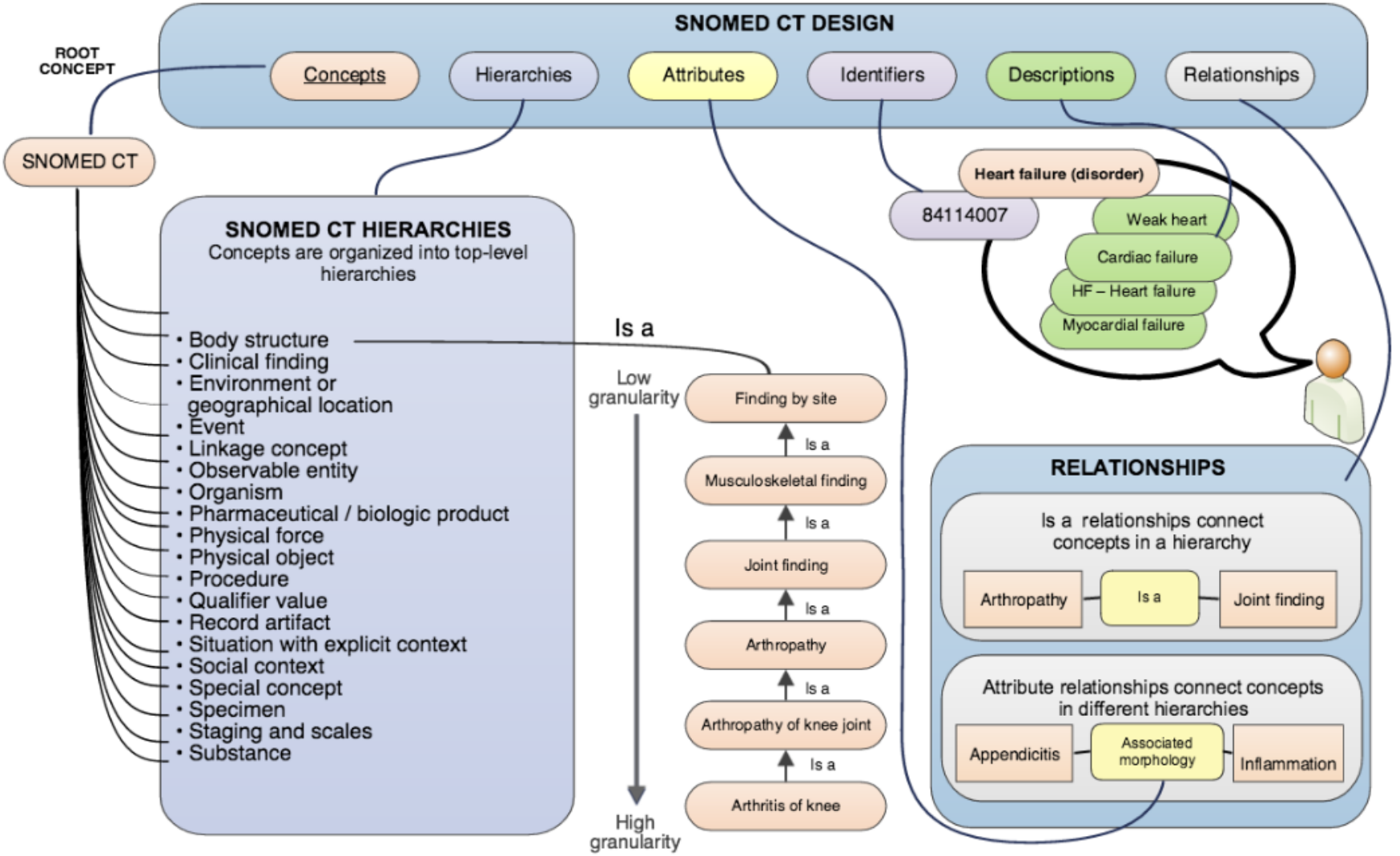
Key components of SNOMED CT

- Concepts
  - represent clinical meanings (intensions)
  - Have a unique SNOMED CT identifier
  - Are embedded in (multiple) subsumption (is-a) hierarchies
  - Relation types are concepts, too, called linkage concepts
- Description and term
  - Term: a string of characters in a particular human language
  - Description: a term in connection to a particular concept
  - Description type:
    - Fully Specified Name (FSN): precise, often artificial
    - Synonym: represents clinical language in use, sometimes ambiguous
- Relationship
  - Represents an association between two concepts via a linkage concept
- Common Features of components
  - History

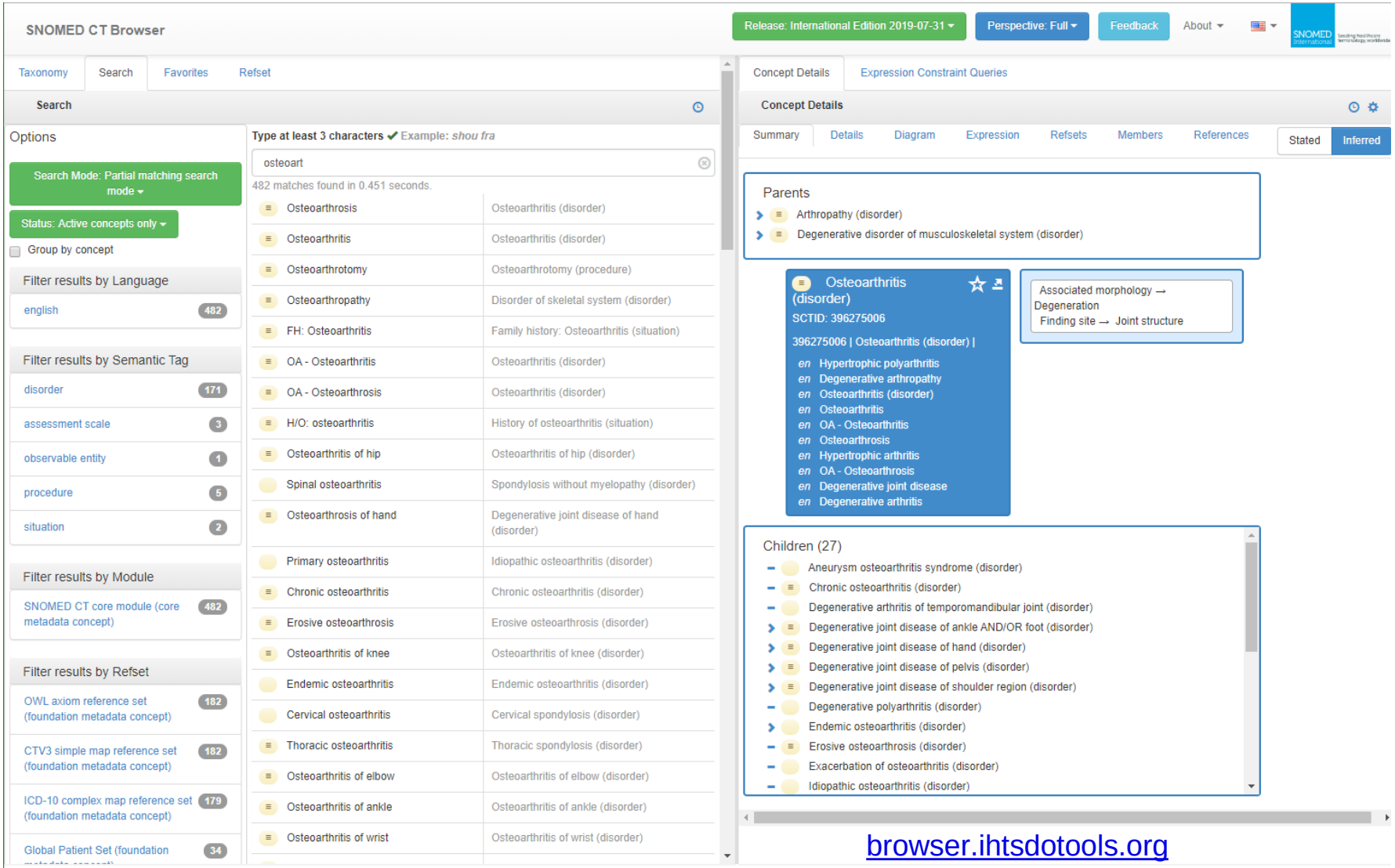
Size of SNOMED CT

Concepts	350,830
English descriptions	1,207,013
Spanish descriptions	979,112
English text definitions	6,236
Relationships (triples)	596,420

SNOMED CT design



The SNOMED CT browser



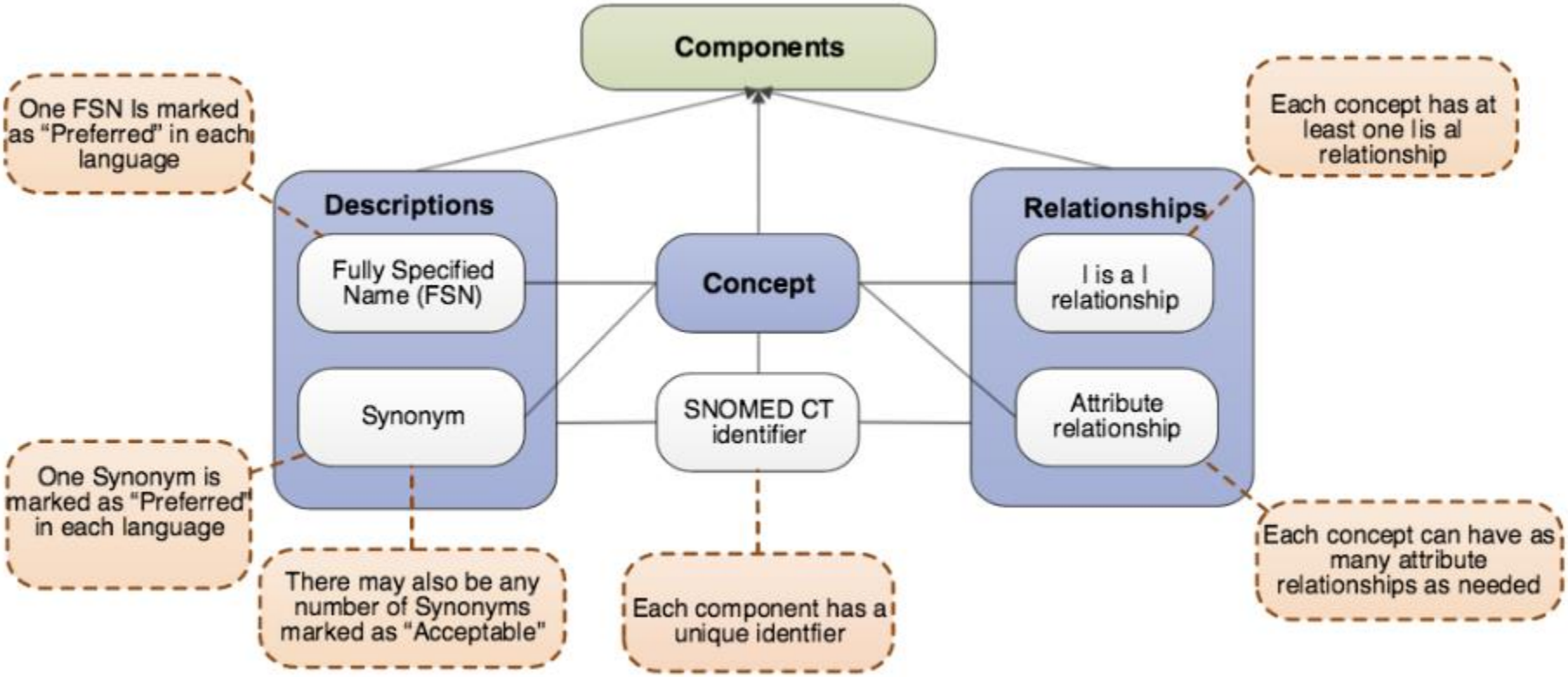
browser.ihtsdotools.org

Knowing the SNOMED CT browser

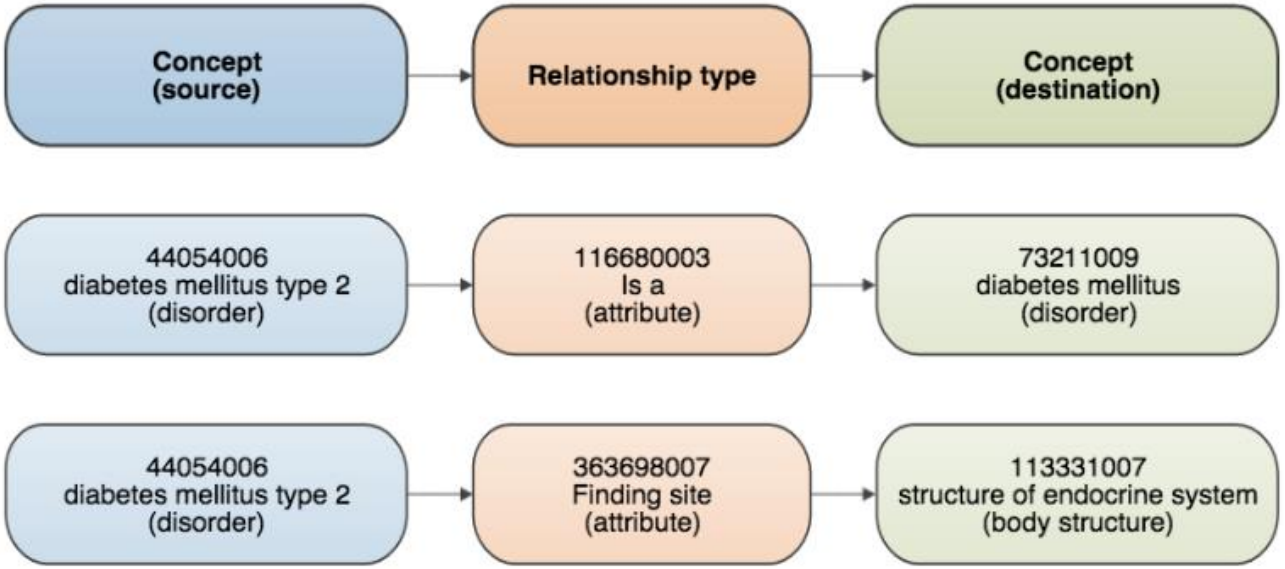
- https://browser.ihtsdotools.org/

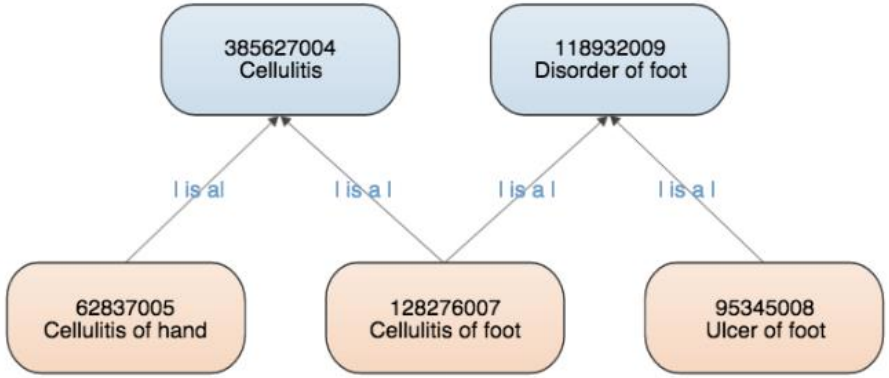
- Exercise:
  - Open the SNOMED CT browser
  - Identify e.g. "Fracture of bone"
    - Concept and their ID
    - Terms that belong to concepts
    - Fully specified names and related synonyms
    - The relations to other concepts
  - Check
    - Toplevel hierarchies
    - Semantic Tags
    - Differences?

SNOMED CT logical model (I)

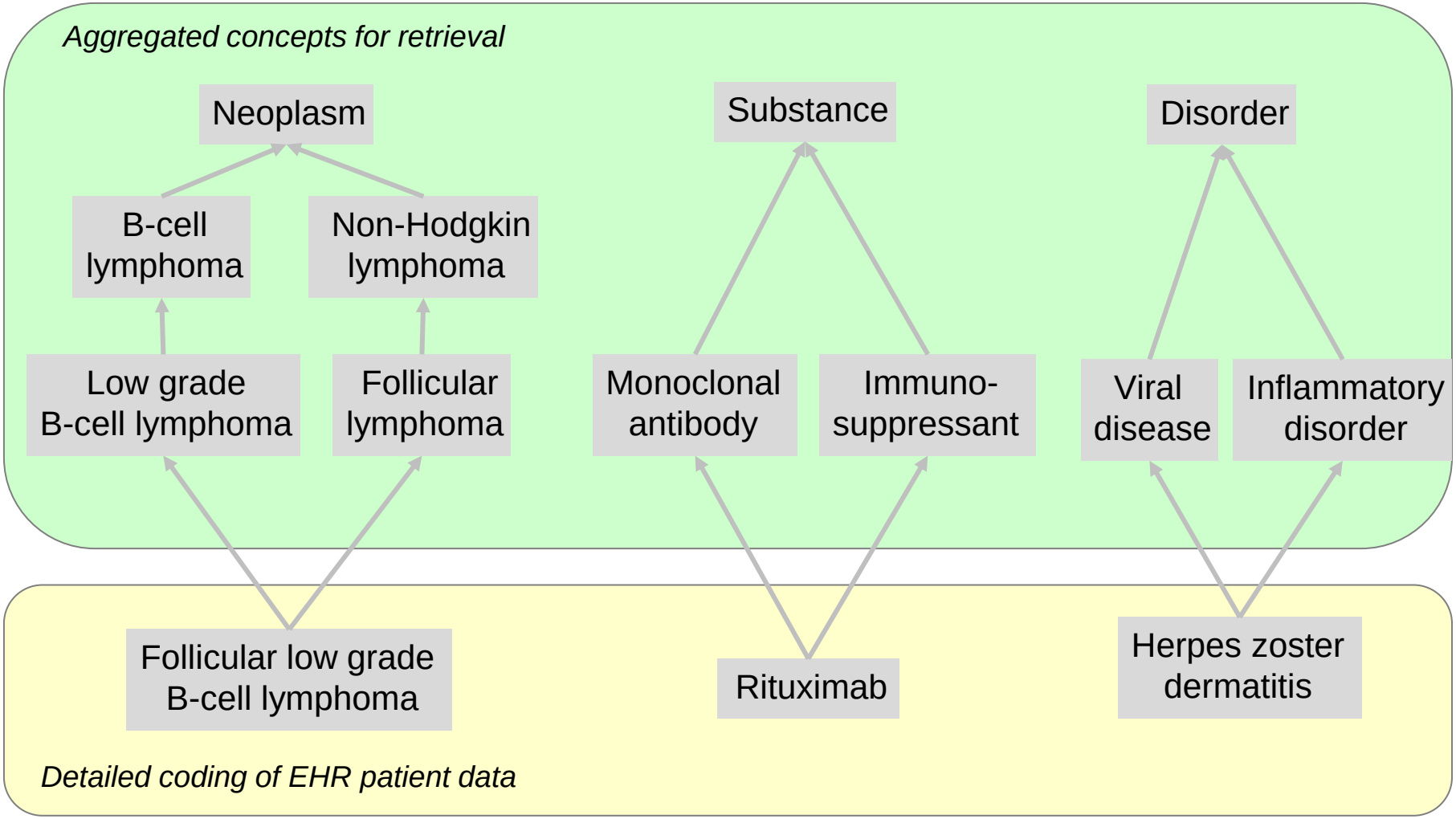


SNOMED CT logical model (III)





Purpose of multiple hierarchies



Polyhierarchical structure

- Exercise
  - Analyse the concept
    Fine needle aspiration and core needle biopsy of breast using magnetic resonance imaging guidance (procedure)
    (SCTID: 433685008)
    - Draw its subclass hierarchy
    - Have a look on its defining characteristics
    - What is the difference between the "inferred" and "stated" view?

Example: "Fracture of Bone"

Fracture of bone (disorder)

SCTID: 125605004

125605004 | Fracture of bone (disorder)
|

en Broken bone
en Fracture of bone (disorder)
en Fracture
en Fracture of bone

Finding site → Bone structure
Associated morphology → Fracture

Tabular representation in SNOMED CT release

Fracture of bone (disorder)

SCTID: 125605004, Defined, Active

TERMINOLOGY

Term	Acceptability (en)
F ☆ Fracture of bone (disorder)	Preferred
S ★ Fracture of bone	Preferred
S ✓ Broken bone	Acceptable
S ✓ Fracture	Acceptable

Término	Aceptabilidad (es)
F ☆ fractura de hueso (trastorno)	Preferido
S ★ fractura de hueso	Preferido

ONTOLOGY

Axiom

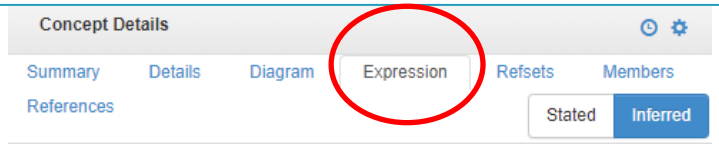
Type	Destination	Group
Is a (attribute)	Disease (disorder)	0
Finding site (attribute)	Bone structure (body structure)	1
Associated morphology (attribute)	Fracture (morphologic abnormality)	1

Fracture of Bone: Logic-based concept definitions

- Example:
- Text definition:
  - A fracture of bone is a disease with a fracture morphology located at some bone structures
  - A disease with a fracture morphology located at some bone structure is a Fracture of bone
- More "ontological"
  - For every instance of the type *Fracture of bone* the following applies:
    - It has some morphology of the type *Fracture* (i)
    - It has some location of the type *Bone structure* (ii)
  - Every entity for which (i) and (ii) applies is an instance of the type / concept *Fracture of Bone*

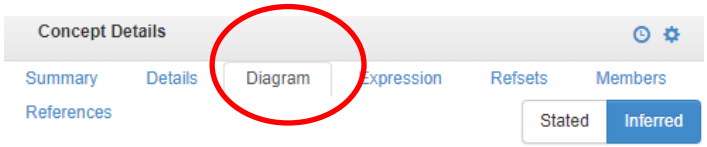
Fracture of Bone:
Logic-based concept definitions

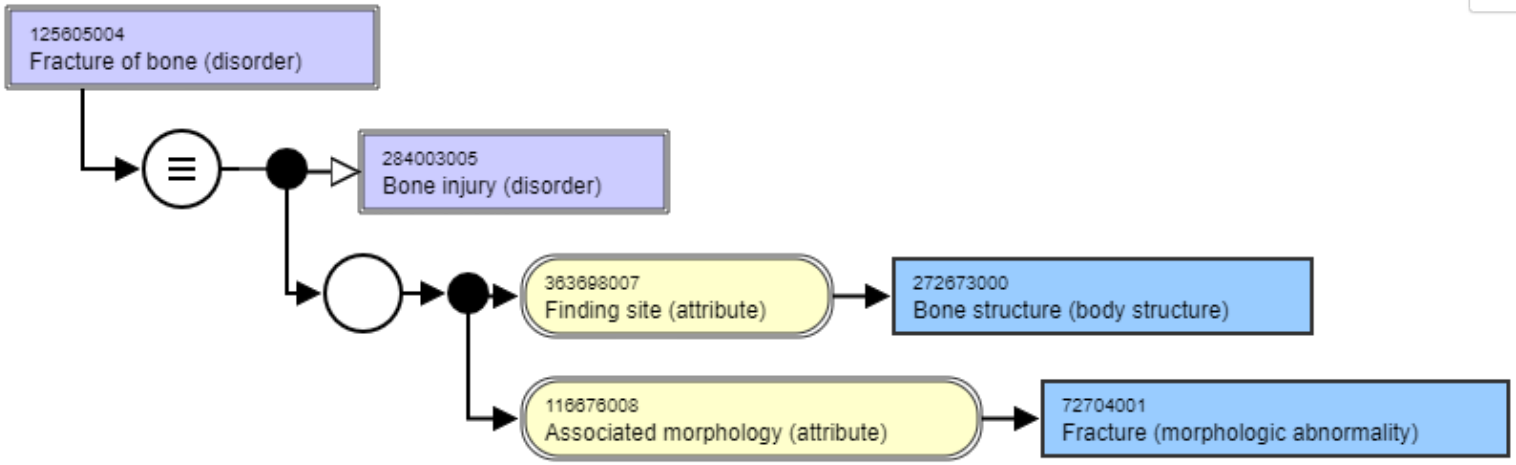
- SNOMED compositional grammar



284003005 |Bone injury (disorder)| :
{ 363698007 |Finding site (attribute)| = 272673000 |Bone structure (body structure)|,
116676008 |Associated morphology (attribute)| = 72704001 |Fracture (morphologic abnormality)| }

- SNOMED diagram





Syntax: Compositional grammar

- Simple expression

  73211009 |Diabetes mellitus| or 73211009

- Multiple focus concepts

  7246002|Kidney biopsy| + 129249002 |Needle biopsy17724009 |

- Expression with refinement

  182201002 |hip joint|:

  272741003 |laterality| = 24028007 |right|

- Expression with nested refinement

  397956004 |prosthetic arthroplasty of the hip|:

  363704007 |procedure site| =

  (24136001 |hip joint structure|:

  272741003 |laterality| = 7771000 |left|)

- Expression with role group refinement

Fracture of Bone: Logic-based concept definitions

- FOL:

$\forall$x: instance-of(x, Fracture_of_bone) $\Leftrightarrow$
instance-of(x, Disease) $\wedge$
$\exists$y,z: instance-of(y, Fracture) $\wedge$ instance-of(z, Bone_Structure) $\wedge$
associated-morphology(x, y) $\wedge$ finding-site(x, z)

- DL (Manchester Syntax):

Fracture_of_bone equivalentTo
Disease and
(associated_morphology some Fracture) and
(finding_site some Bone_Structure)

Description logic – OWL 2 EL profile

- Constructs used for SNOMED CT
  - Equivalence: EquivalentTo
  - Subsumption: SubclassOf
  - Conjunction: and
  - Exist. quantification: some
- Constructs not (yet) implemented in SNOMED CT
  - Property chains
  - Transitive and reflexive object properties
  - Property equivalence
  - Concrete domains (numbers)
- Constructs not supported by EL profile
  - Universal quantification: all, only
  - Disjunction: or
  - Class negation: not
  - Inverse object properties

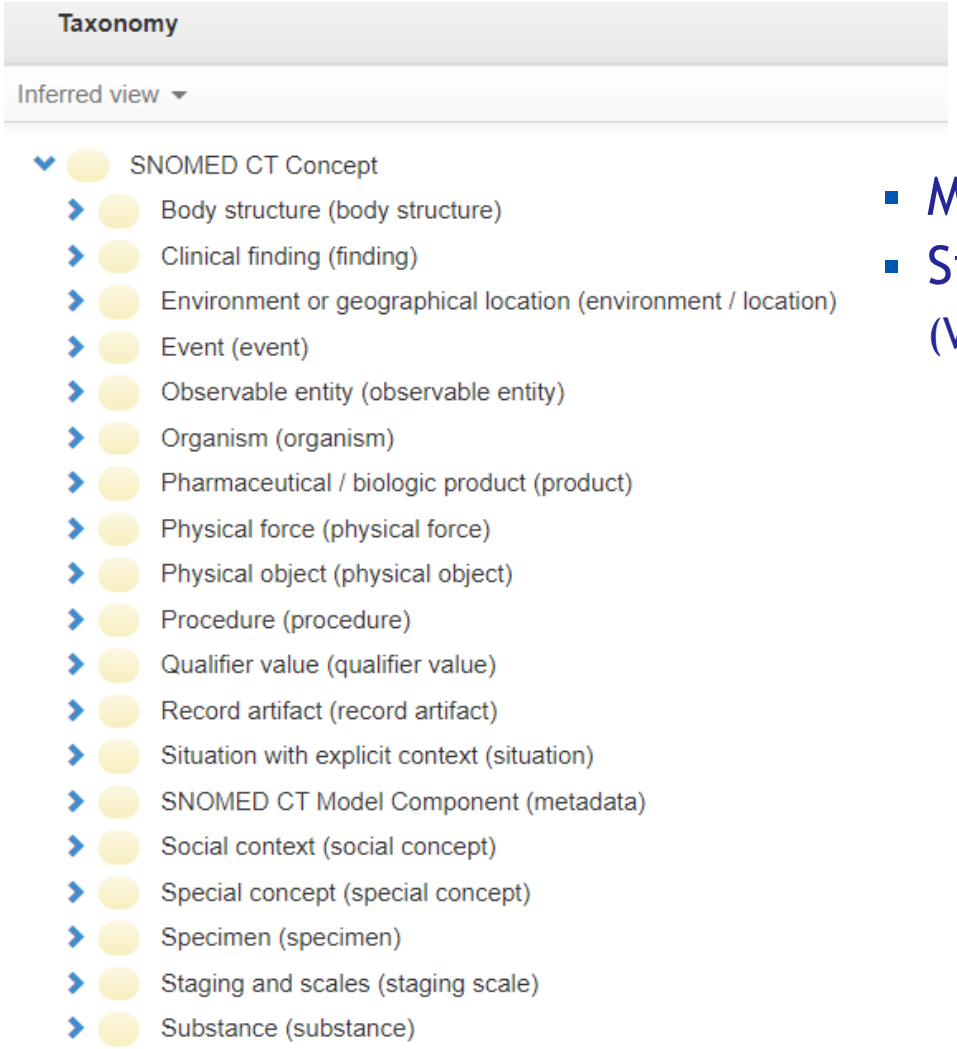
makes DL reasoning intractable

Concept model

- Based on the logical model
- The editorial rules for the permitted attributes and values in **logical definitions** of concepts in SNOMED CT
- Particularly, which relationships are permitted between concepts in particular branches of the hierarchy
  - Domain / range restrictions for attributes
  - Attribute hierarchy
- Examples:

|**Finding site**| specifies the body site affected by a condition.
|**Associated morphology**| specifies the morphologic changes seen at the tissue or cellular level that are characteristic features of a disease.
|**Procedure site**| describes the body site acted on or affected by a procedure.
|has active ingredient| indicates the active ingredient of a drug product, linking the |pharmaceutical / biologic product| hierarchy to the |substance| hierarchy.

Top level categories



- Mostly disjoint
- Still reflect SNOMED's legacy

  (Version 3.5)
  - T (Topography) – Anatomic terms
  - M (Morphology) – Changes found in cells, tissues and organs
  - L (Living organisms) – Bacteria and viruses
  - C (Chemical) – Drugs
  - F (Function) – Signs and symptoms
  - J (Occupation) – Terms that describe the occupation
  - D (Diagnosis) – Diagnostic terms
  - P (Procedure) – Administrative, diagnostic and therapeutic procedures
  - A (Physical agents, forces, activities) – Devices and activities associated with the disease
  - S (Social context) – Social conditions and important relationships in medicine
  - G (General) – Syntactic linkages and qualifiers

SNOMED CT Top Level

- Exercise
  - Choose examples under your top-level category
  - Identify concepts with the same or similar name under a different hierarchy
  - Discuss
    - Whether the example shown fits under the top level
    - Whether the top-level concept is well-chosen and understandable
    - Whether the top-level concept is well-chosen under ontological criteria
    - How are part-of hierarchies modeled in SNOMED CT?
    - Where do ambiguities arise and why?

Attribute (Relationship Types)

- Are considered special type of SNOMED CT concepts
  - Linkage concepts: Concept model attributes and concept history attribute
- Examples:
  - has-ingredient, associated morphology, finding site, finding informer, has disposition, …
- Exercise
  - Browse through of attributes
  - Compare to known relation ontologies
  - Inspect concept history attributes

Pre-coordination vs. postcoordination

Pre-coordination

2nd degree burn of a single finger

```
211908006 |Deep partial thickness burn of a single finger (disorder)|
                                    ≡
<<< 29673001 |Second degree burn of single finger, not thumb (disorder)| :
{ 116676008 |Associated morphology| = 262588000 |Deep partial thickness burn
(morphologic abnormality)|,363698007 |Finding site| = 56213003 |Skin of
finger (body structure)| }
```

Post-coordination

2nd degree burn of the back of the right index finger

```
<<< 29673001 |Second degree burn of single finger, not thumb (disorder)| :
{ 116676008 |Associated morphology| = 262588000 |Deep partial thickness burn
(morphologic abnormality)|,363698007 |Finding site| = 37314006 | Skin
structure of dorsal surface of index finger (body structure) |, 272741003
|Laterality| = 24028007 |Right (qualifier value)| }
```

Role grouping

- To allow nesting of axioms and therefore correct inferences for complex concepts, e.g. that involve more than one site, or more than one morphology.
- The attribute-value pairs are logically associated with each other by grouping them together (nesting) to indicate that certain roles must go together, e.g. which site goes with which morphology.
- Role group can be interpreted as (reflexive) **has-part** to take conditions or procedures expressed by expressions as values

Spackman KA, Dionne R, Mays E, Weis J. Role grouping as an extension to the description logic of ONTYLOG, motivated by entity modeling in SNOMED. AMIA Annual Symposium Proceedings; Washington, DC; 2002. pp. 712–716
Stefan Schulz, Alan Rector, Jean-Marie Rodrigues, Kent Spackman. Competing Interpretations of Disorder Codes in SNOMED CT and ICD, AMIA Annu Symp Proc. 2012; 2012: 819–827.

Role group (cont.)

WITHOUT ROLE GROUP

Fracture_of_bone equivalentTo
Fracture and
(associated_morphology some Fracture) and
(finding_site some Bone_Structure)

WITH ROLE GROUP

Fracture_of_bone equivalentTo
Fracture and role_group some
((associated_morphology some Fracture) and
(finding_site some Bone_Structure))

EXPLICIT ROLE GROUP

Fracture_of_bone equivalentTo
Fracture and has_part some
((associated_morphology some Fracture) and
(finding_site some Bone_Structure))

Primitive vs. Fully defined

- Primitive (subclassOf)
  - Concepts are defined by necessary conditions only
    - Disease
      - –Diabetes mellitus
        - »is a disorder of endocrine system
        - »Is a disorder of glucose metabolism
    - Procedure
      - – Percutaneous transluminal angioplasty (procedure)
        - »Is a Transluminal angioplasty
        - »Is a Catheter procedure
        - »Surgical repair procedure by device
- Fully defined (equivalentTo)
  - Concepts are defined by necessary and sufficient conditions
    - Fracture of bone
    - MRI guided biopsy

Stated/inferred views, normalised form

- Stated view
  - Attributes and values of a concept definition are stated by a modeller
  - Distributed in “stated relationship table” in release
- Inferred view
  - Attributes and values of concept definition are generated by description logic reasoner
    - Includes relationships inferred from the stated view
    - Redundant relationships removed

  The relationship table in release is based on inferred view
- Normalised form
  - Only presents proximal primitive super-concepts and non-redundant defining relationships
  - Suitable for comparing expressions

Fracture of femur example

- Stated view

Fracture
and
RoleGroup some
(Finding site some
femur
and
Associated
morphology some
fracture)

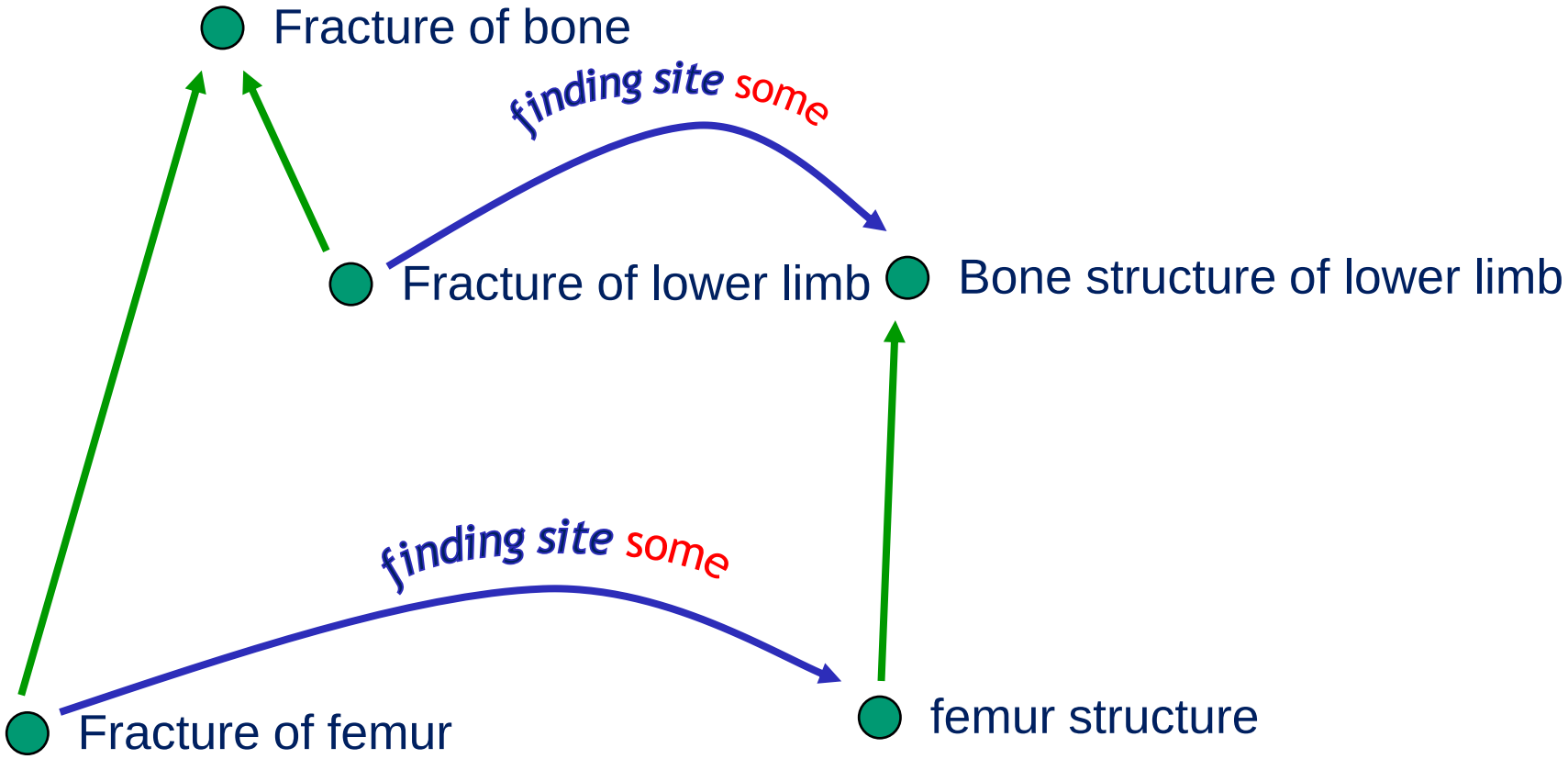
- Inferred view

Fracture of lower limb
and
RoleGroup some
(Finding site some
femur
and
Associated
morphology some
fracture)

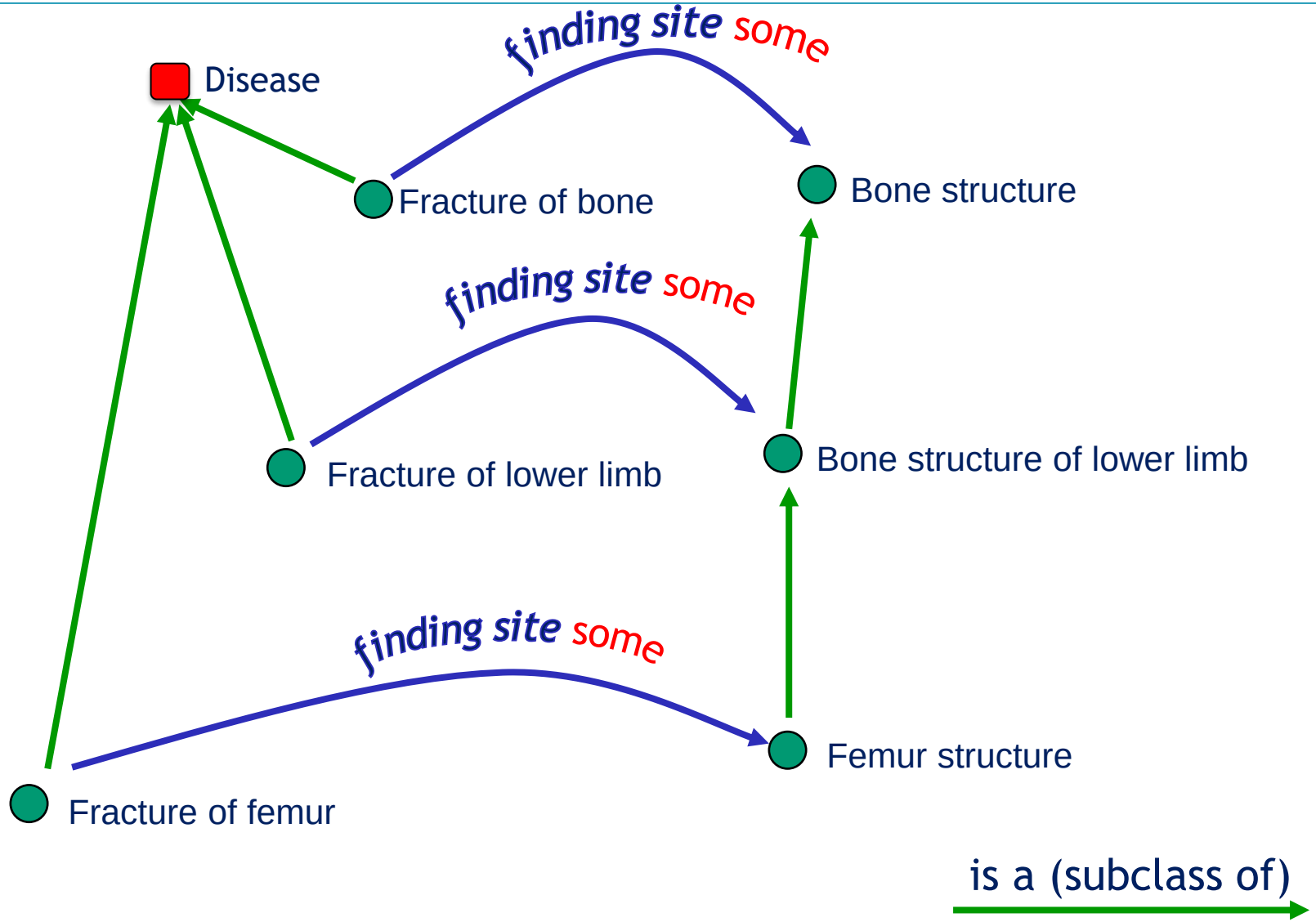
- Normal form

Disease
and
RoleGroup some
(Finding site some
femur
and
Associated
morphology some
fracture)

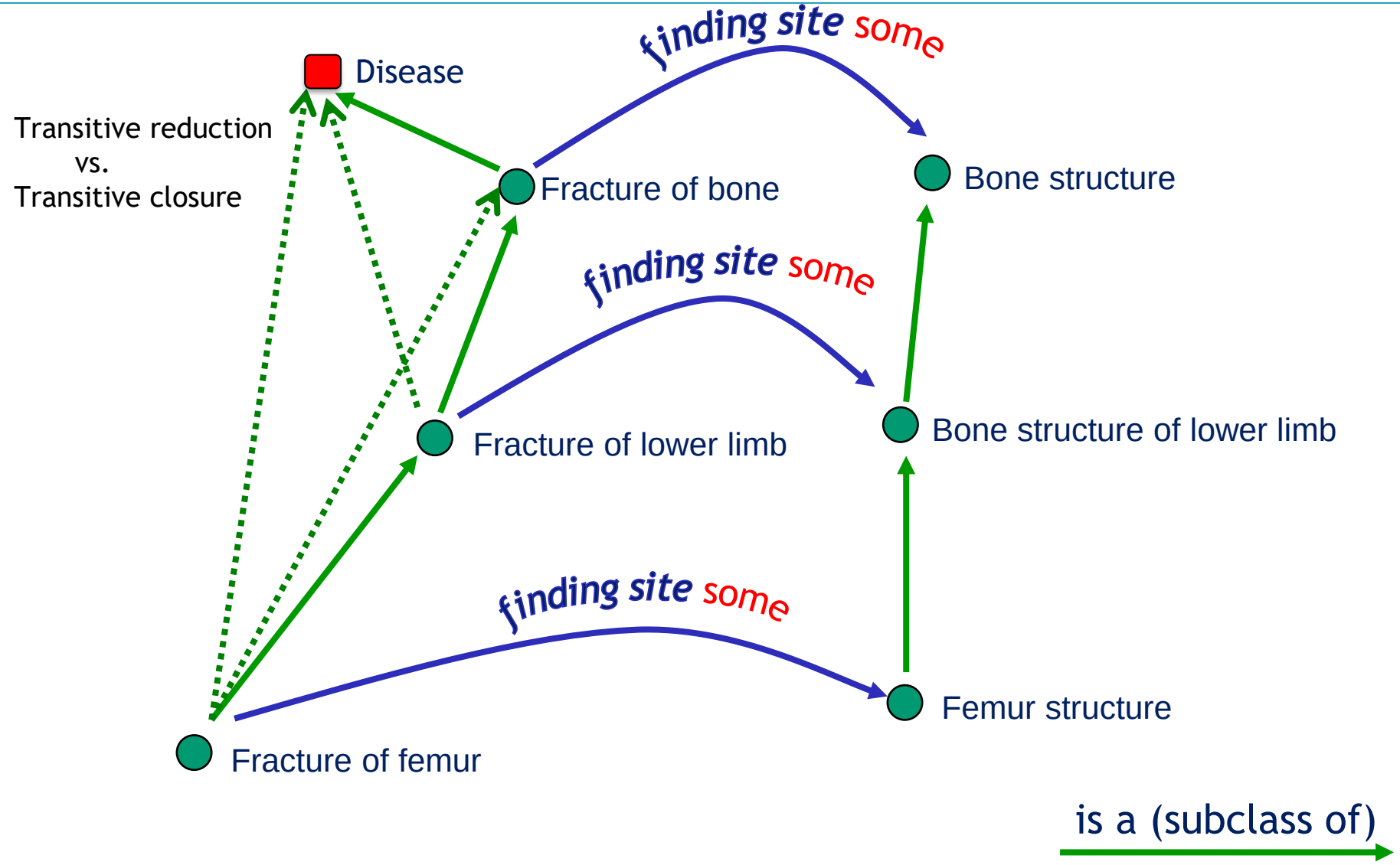
Stated view – fracture of femur



Normal form - Fracture of femur



Inferred view – after classified by DL reasoner



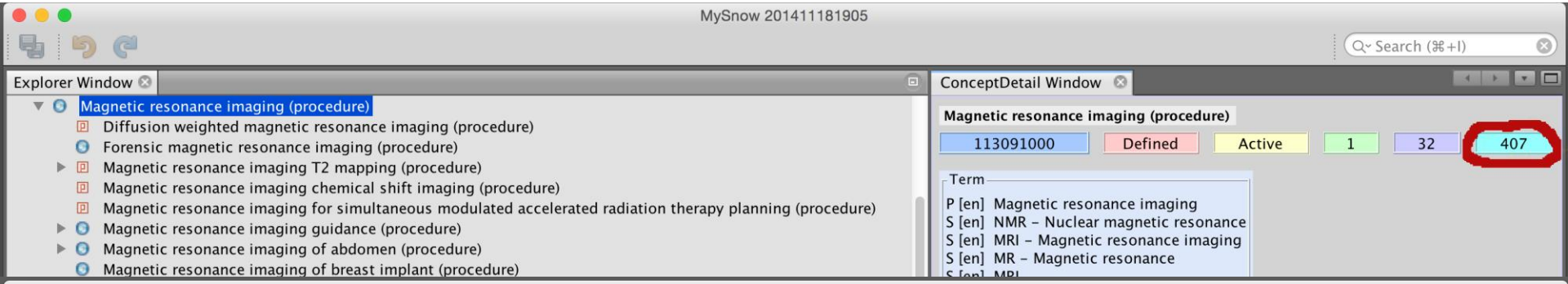
Transitive reduction vs. closure

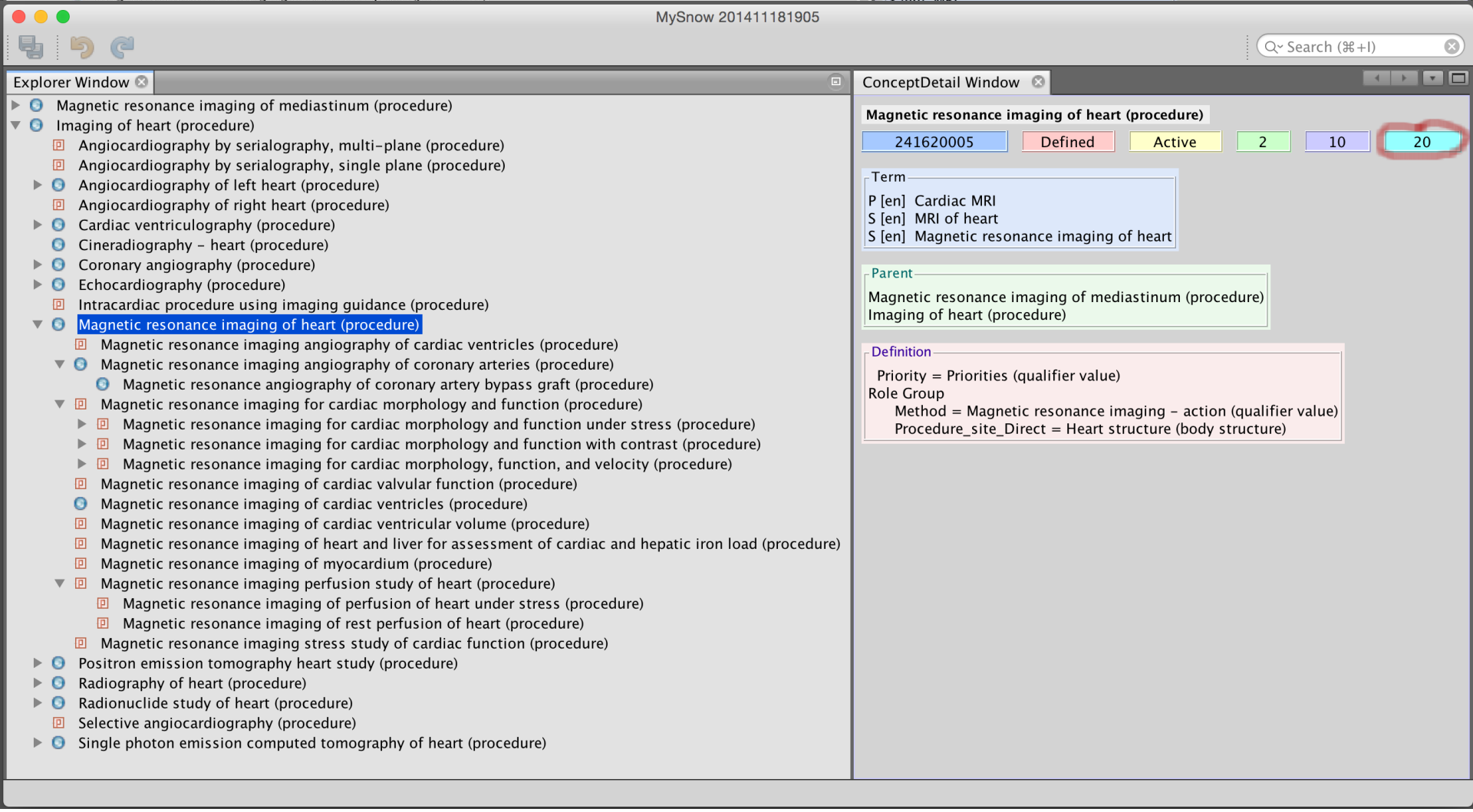
- **Transitive Reduction**

1. Fracture of bone -> Disease
2. Fracture of lower limb -> Fracture of bone
3. Fracture of femur -> Fracture of lower limb

- **Transitive Closure**

1. Fracture of bone -> Disease
2. Fracture of lower limb -> Disease
3. Fracture of femur -> Disease
4. Fracture of lower limb ->Fracture of bone
5. Fracture of femur -> Fracture of bone
6. Fracture of femur -> Fracture of lower limb


MySnow 201411181905
Search (⌘+I)
Explorer Window
Magnetic resonance imaging (procedure)
Diffusion weighted magnetic resonance imaging (procedure)
Forensic magnetic resonance imaging (procedure)
Magnetic resonance imaging T2 mapping (procedure)
Magnetic resonance imaging chemical shift imaging (procedure)
Magnetic resonance imaging for simultaneous modulated accelerated radiation therapy planning (procedure)
Magnetic resonance imaging guidance (procedure)
Magnetic resonance imaging of abdomen (procedure)
Magnetic resonance imaging of breast implant (procedure)
ConceptDetail Window
Magnetic resonance imaging (procedure)
113091000
Defined
Active
1
32
407
Term
P [en] Magnetic resonance imaging
S [en] NMR - Nuclear magnetic resonance
S [en] MRI - Magnetic resonance imaging
S [en] MR - Magnetic resonance


MySnow 201411181905
Search (⌘+I)
Explorer Window
Magnetic resonance imaging of mediastinum (procedure)
Imaging of heart (procedure)
Angiocardiography by serialography, multi-plane (procedure)
Angiocardiography by serialography, single plane (procedure)
Angiocardiography of left heart (procedure)
Angiocardiography of right heart (procedure)
Cardiac ventriculography (procedure)
Cineradiography - heart (procedure)
Coronary angiography (procedure)
Echocardiography (procedure)
Intracardiac procedure using imaging guidance (procedure)
Magnetic resonance imaging of heart (procedure)
Magnetic resonance imaging angiography of cardiac ventricles (procedure)
Magnetic resonance imaging angiography of coronary arteries (procedure)
Magnetic resonance angiography of coronary artery bypass graft (procedure)
Magnetic resonance imaging for cardiac morphology and function (procedure)
Magnetic resonance imaging for cardiac morphology and function under stress (procedure)
Magnetic resonance imaging for cardiac morphology and function with contrast (procedure)
Magnetic resonance imaging for cardiac morphology, function, and velocity (procedure)
Magnetic resonance imaging of cardiac valvular function (procedure)
Magnetic resonance imaging of cardiac ventricles (procedure)
Magnetic resonance imaging of cardiac ventricular volume (procedure)
Magnetic resonance imaging of heart and liver for assessment of cardiac and hepatic iron load (procedure)
Magnetic resonance imaging of myocardium (procedure)
Magnetic resonance imaging perfusion study of heart (procedure)
Magnetic resonance imaging of perfusion of heart under stress (procedure)
Magnetic resonance imaging of rest perfusion of heart (procedure)
Magnetic resonance imaging stress study of cardiac function (procedure)
Positron emission tomography heart study (procedure)
Radiography of heart (procedure)
Radionuclide study of heart (procedure)
Selective angiocardiography (procedure)
Single photon emission computed tomography of heart (procedure)
ConceptDetail Window
Magnetic resonance imaging of heart (procedure)
241620005
Defined
Active
2
10
20
Term
P [en] Cardiac MRI
S [en] MRI of heart
S [en] Magnetic resonance imaging of heart
Parent
Magnetic resonance imaging of mediastinum (procedure)
Imaging of heart (procedure)
Definition
Priority = Priorities (qualifier value)
Role Group
Method = Magnetic resonance imaging - action (qualifier value)
Procedure_site_Direct = Heart structure (body structure)

CONCEPT MODELS OF COMMONLY USED HIERARCHIES

Findings, Procedures, and Situations

- 22298006|Myocardial infarction (disorder)
  - 129574000|Postoperative myocardial infarction (disorder)
    - After = Surgical procedure (procedure)

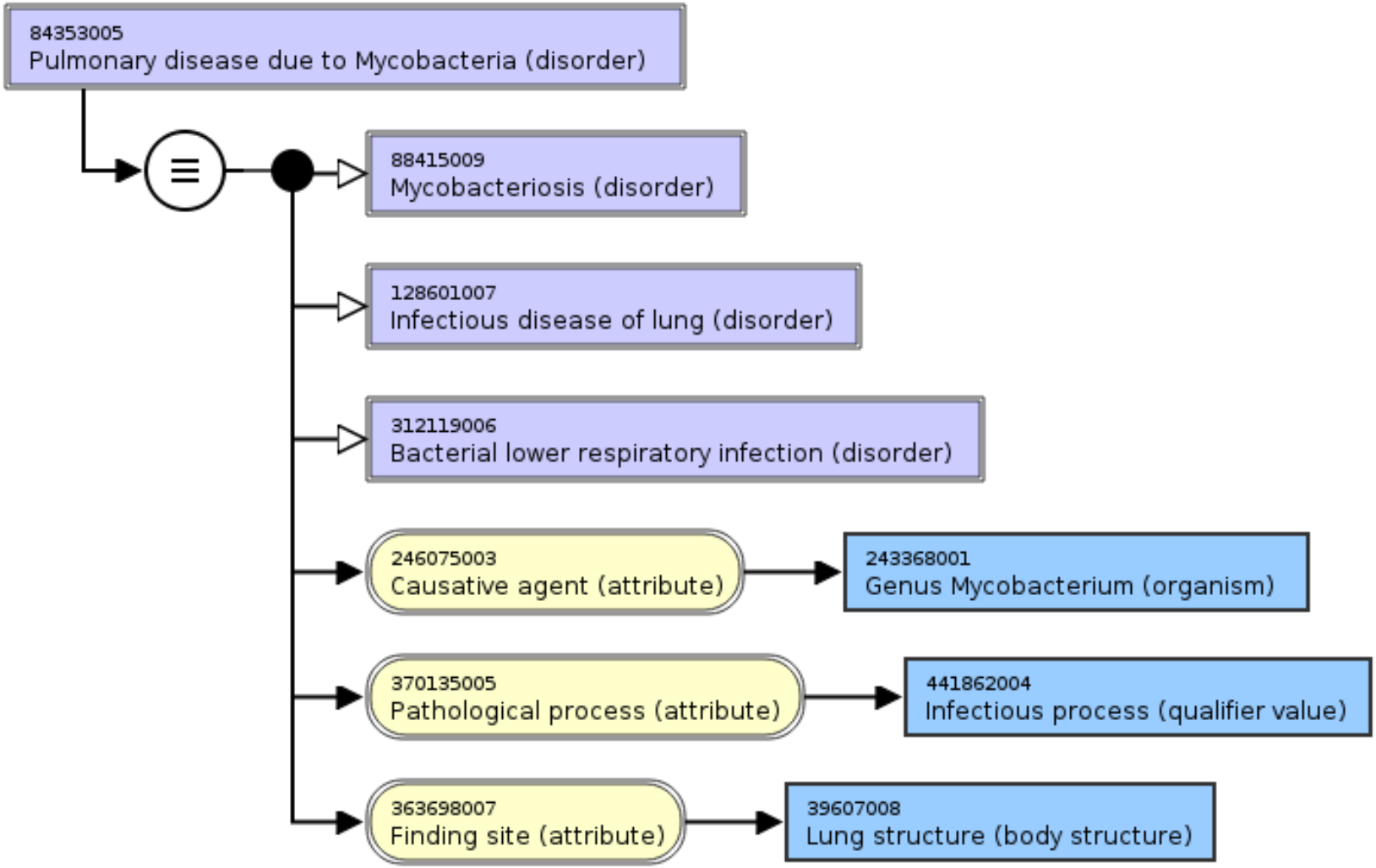
- 399211009|History of myocardial infarction (situation)
  - Temporal context = In the past
  - Associated finding = Myocardial infarction (disorder)

- 266897007|Family history: Myocardial infarction (situation)
  - Subject relationship context = Person in family of subject (person)
  - Associated finding = Myocardial infarction (disorder)

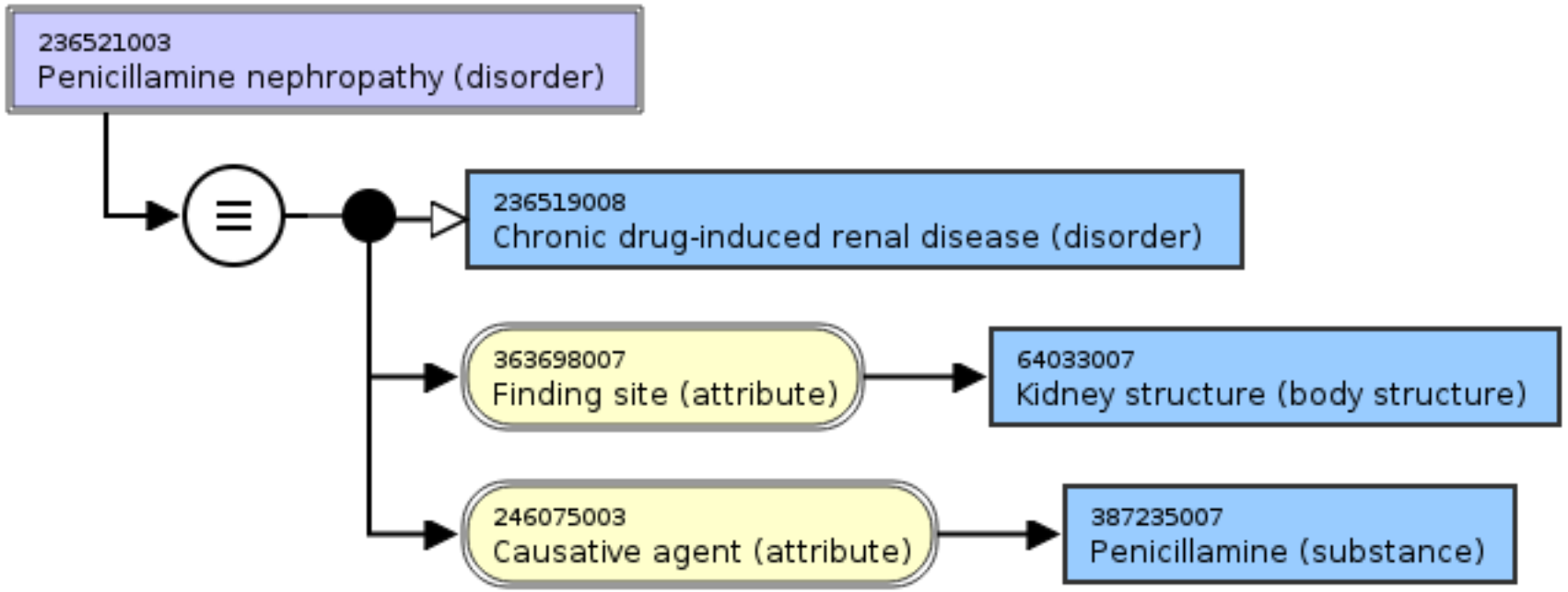
Clinical finding/disorder

Attributes	Range of allowable values
Finding site	Anatomical or acquired body structure (head, kidney, artery, bone)
Associated morphology	Morphologically abnormal structure (fracture, stenosis, inflammation)
Associated with	Clinical finding, Procedure, Event …
Due to	Clinical finding, Event
After	Clinical finding, Procedure
Causative agent	Organism, Substance, Physical object, Physical force, Pharmaceutical/biologic product
Pathological process	Infectious process, Hypersensitivity process, Autoimmune
Clinical course	Courses (chronic, acute)
Occurrence	Periods of life (congenital, fetal period, childhood, adulthood)
Severity	Severities (mild, moderate, severe)

Pulmonary infection due to mycobacteria



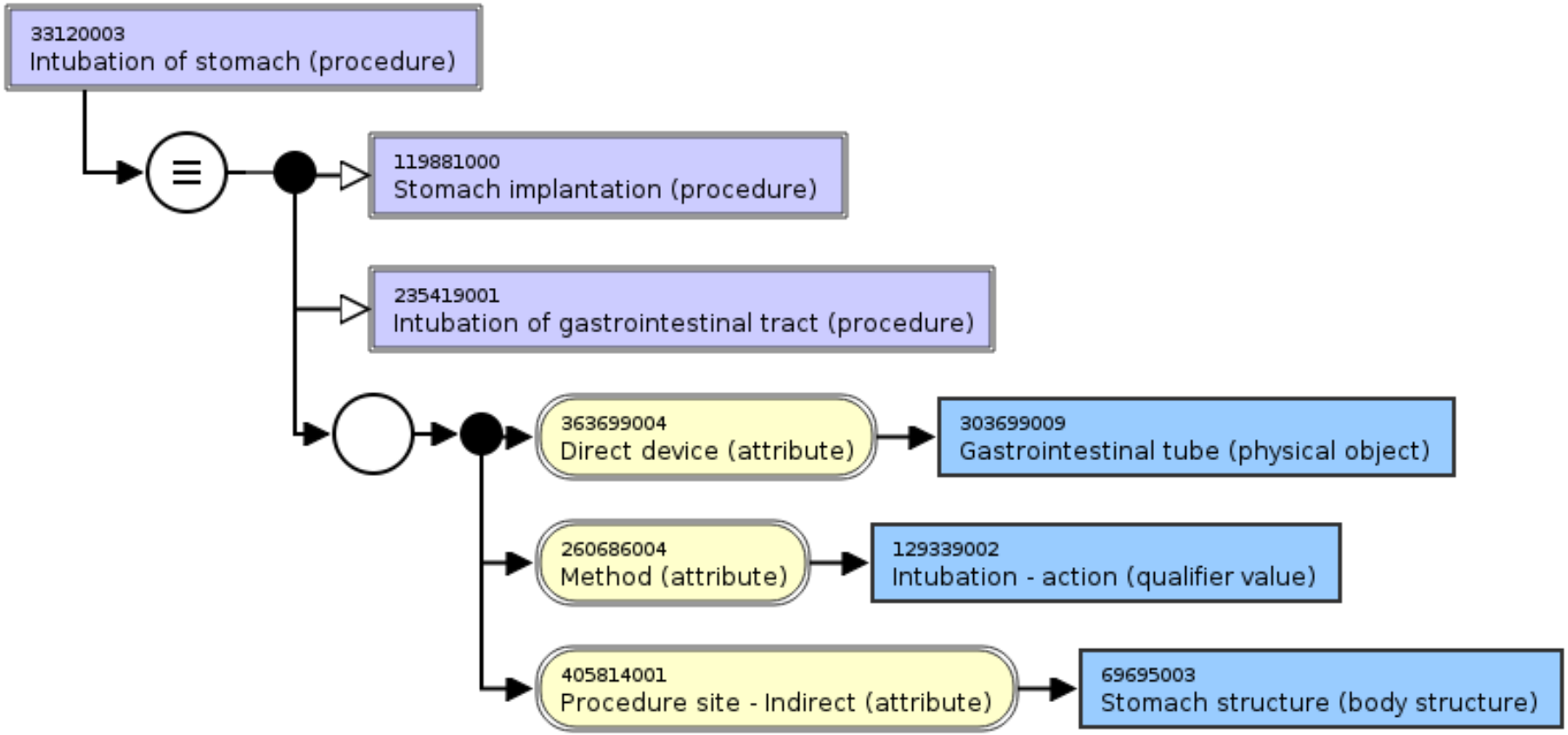
Penicillamine nephropathy



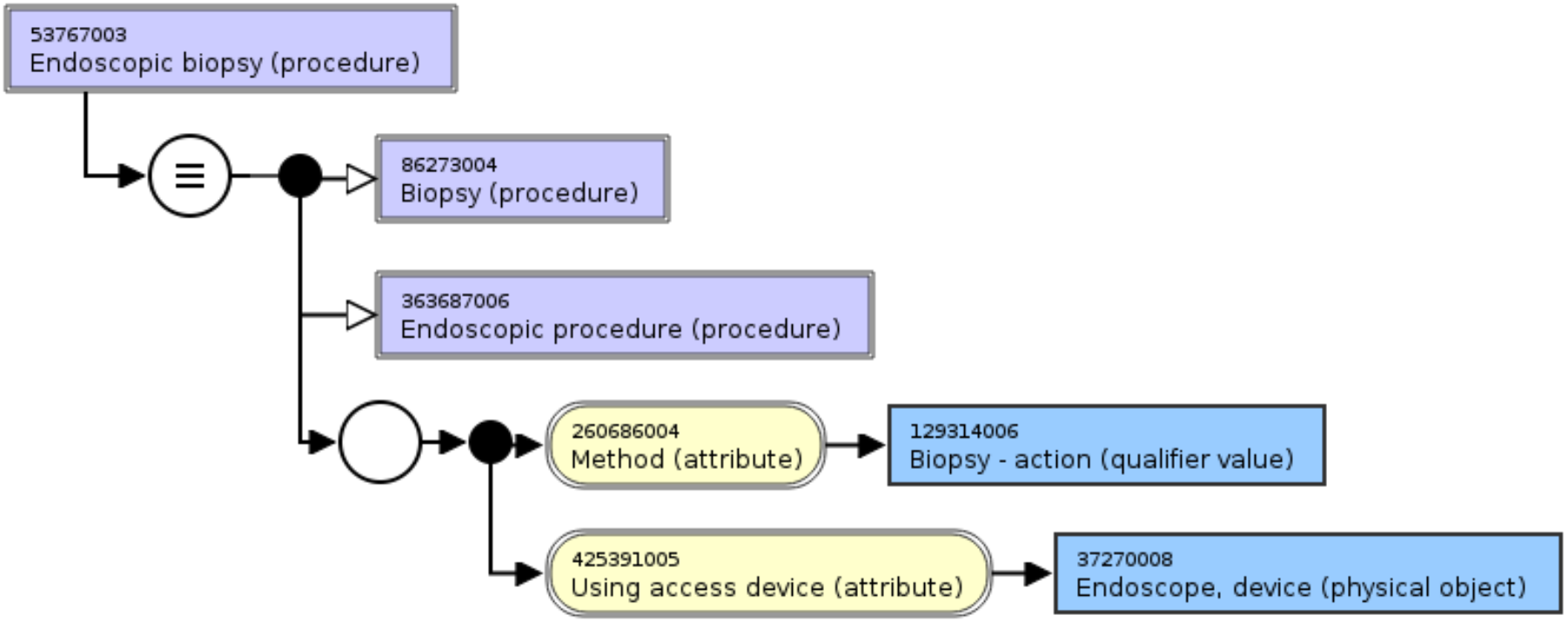
Procedure

Attributes	Range of allowable values
Procedure site	Anatomical or acquired body structure
Procedure site - direct	Anatomical or acquired body structure
Procedure site - indirect	Anatomical or acquired body structure
Method	Action (insertion, imaging action, evaluation)
Procedure morphology	Morphologically abnormal structure
Direct morphology	Morphologically abnormal structure
Indirect morphology	Morphologically abnormal structure
Procedure device	Device
Direct device	Device
Indirect device	Device
Using device	Device
Using access device	Device

Intubation of stomach



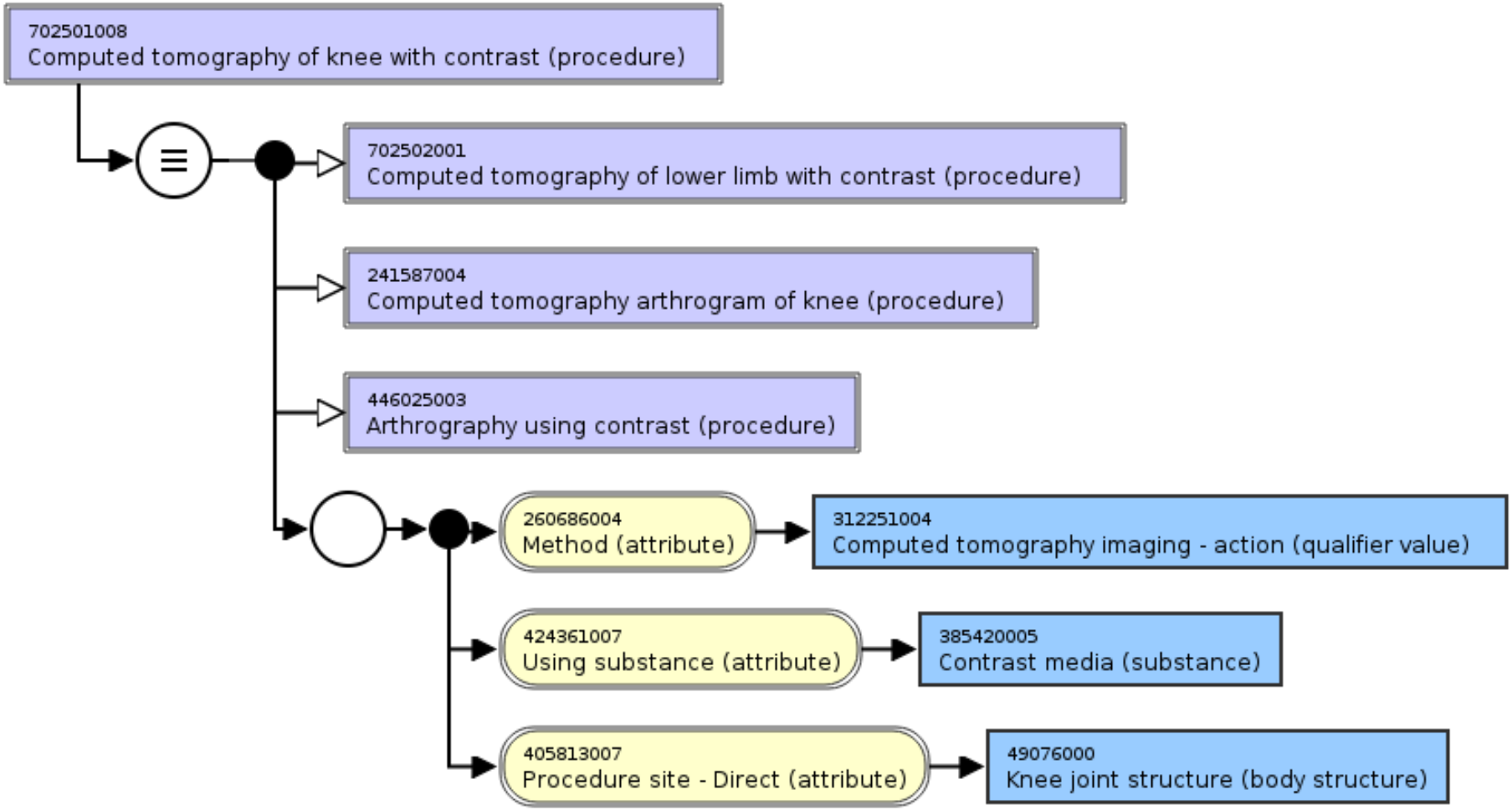
Endoscopic biopsy



Procedure

Attributes	Range of allowable values
Using substance	Substance (e.g. contrast media)
Direct substance	Substance, Pharmaceutical/biologic product
Has intent	Intents (e.g. guidance, diagnostic, therapeutic; Imaging guided procedure)
Access	Surgical access values (e.g.)
Surgical approach	Procedural approach ()
Route of administration	Route of administration value (e.g.)
Has focus	Clinical finding
Priority	Priorities (e.g.)
Revision status	Primary operation, Revision – value, Part of multistage procedure
Recipient category	Person, Family, Community, Donor for medical or surgical procedure, Group
Using energy	Physical force (e.g.)

CT of Knee with contrast



Situation with explicit context

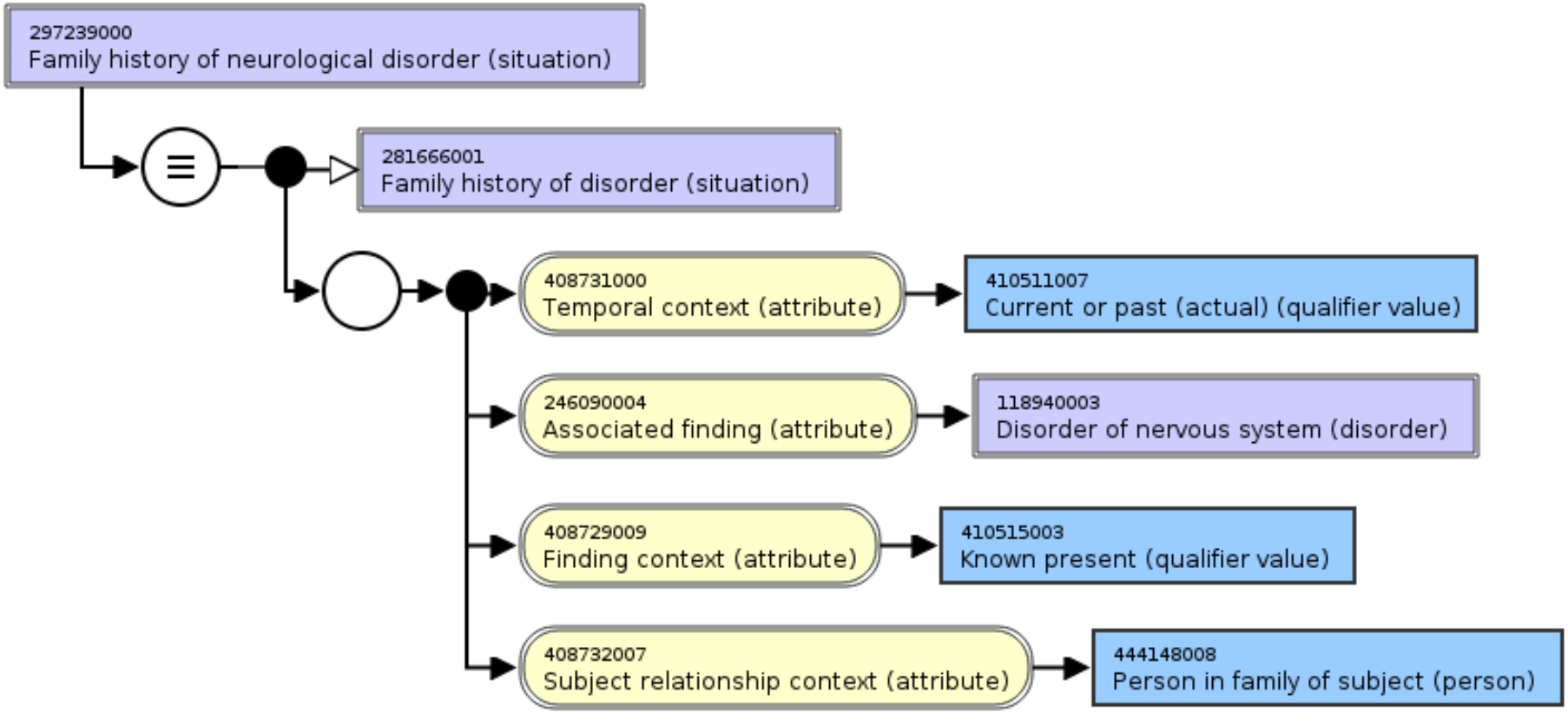
Attributes	Range of allowable values
Subject relationship context	Person
Temporal context	Temporal context value
Associated finding	Clinical finding, Event, Observable entity
Finding context	Finding context value
Associated procedure	Procedure, Observable entity
Procedure context	Context values for actions

Subject context values and examples

- Subject of record (person)
- Person in family of subject (person)
  - Grandparent of subject (person)
  - Parent of subject (person)
    - Mother of subject (person)
    - Father of subject (person)
  - Spouse of subject (person)
    - Wife of subject (person)
    - Husband of subject (person)
  - Sibling of subject (person)
  - Child of subject (person)

- Examples:
  - Wife pregnant (situation)
  - Father smokes (situation)
  - Family history of neurological disorder (situation)

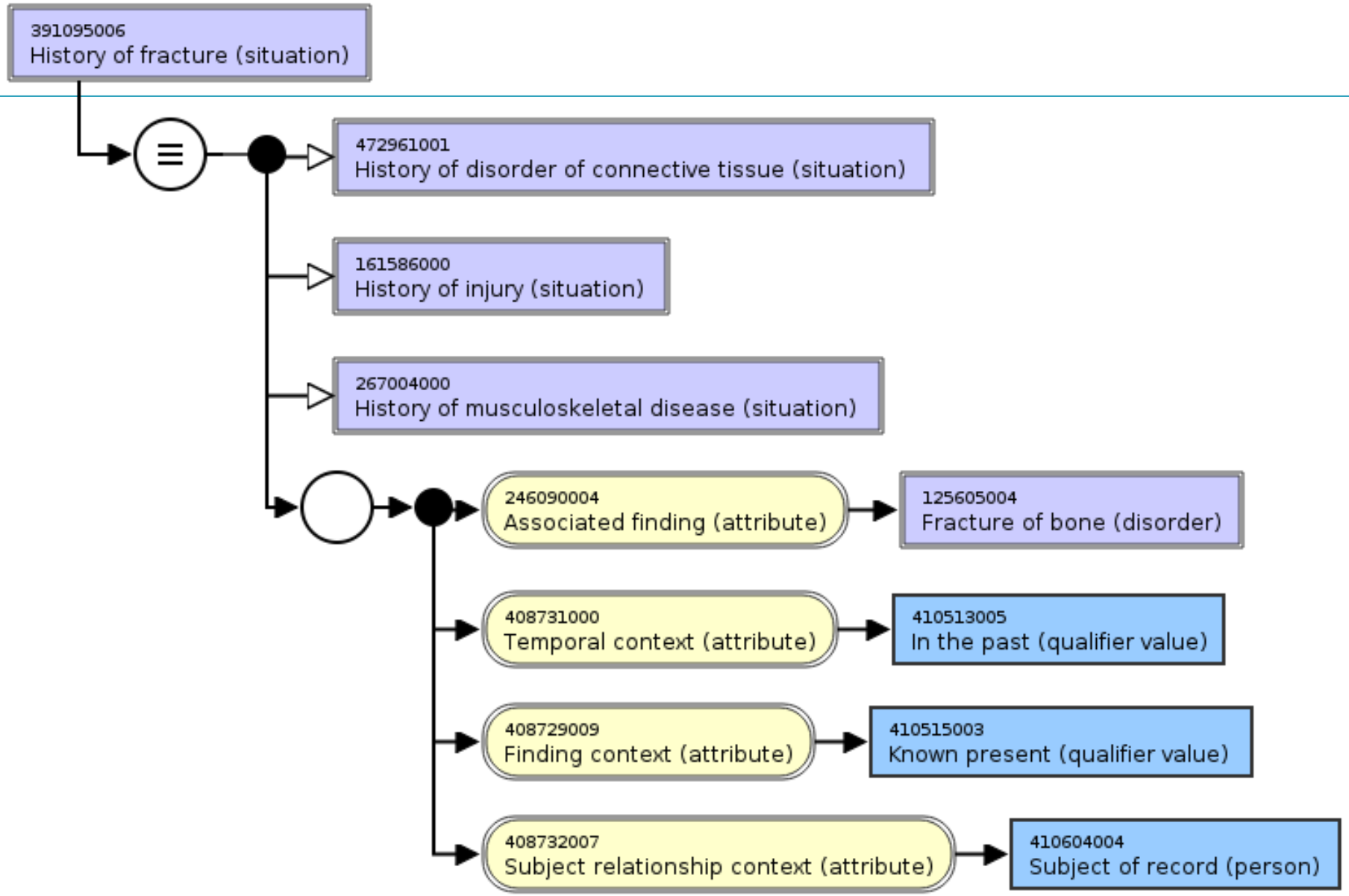
Family history of neurological disorder



Temporal context values and examples

- In the past (qualifier value)
  - Past - time unspecified (qualifier value)
  - Past - time specified (qualifier value)
    - All times past (qualifier value)
- Current or specified time (qualifier value)
  - Specified time (qualifier value)
  - Current (qualifier value)
- Current - time specified (qualifier value)
- Current - time unspecified (qualifier value)

- Example:
  - History of fracture (situation)

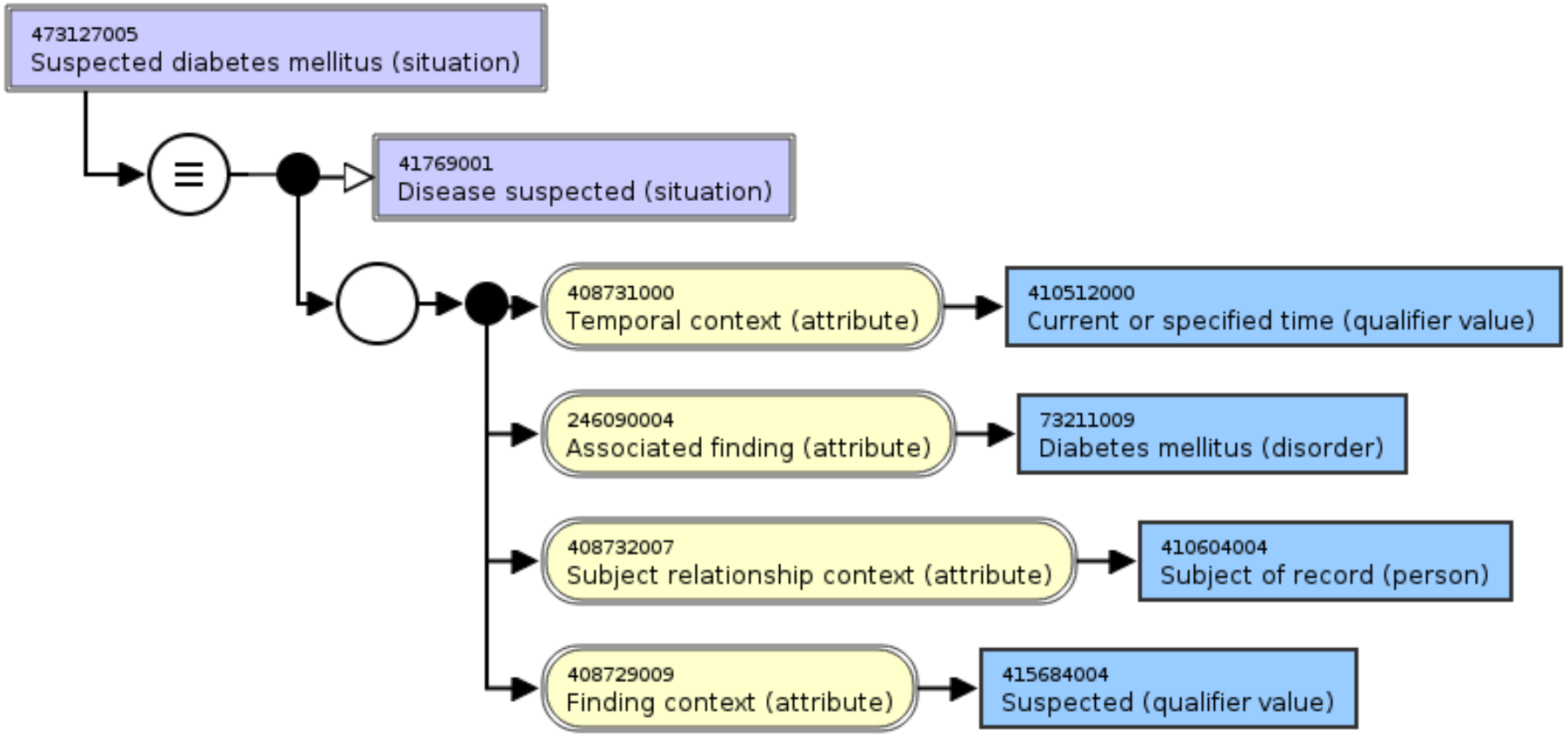

391095006
History of fracture (situation)
≡
472961001
History of disorder of connective tissue (situation)
161586000
History of injury (situation)
267004000
History of musculoskeletal disease (situation)
246090004
Associated finding (attribute)
125605004
Fracture of bone (disorder)
408731000
Temporal context (attribute)
410513005
In the past (qualifier value)
408729009
Finding context (attribute)
410515003
Known present (qualifier value)
408732007
Subject relationship context (attribute)
410604004
Subject of record (person)

Finding context values and examples

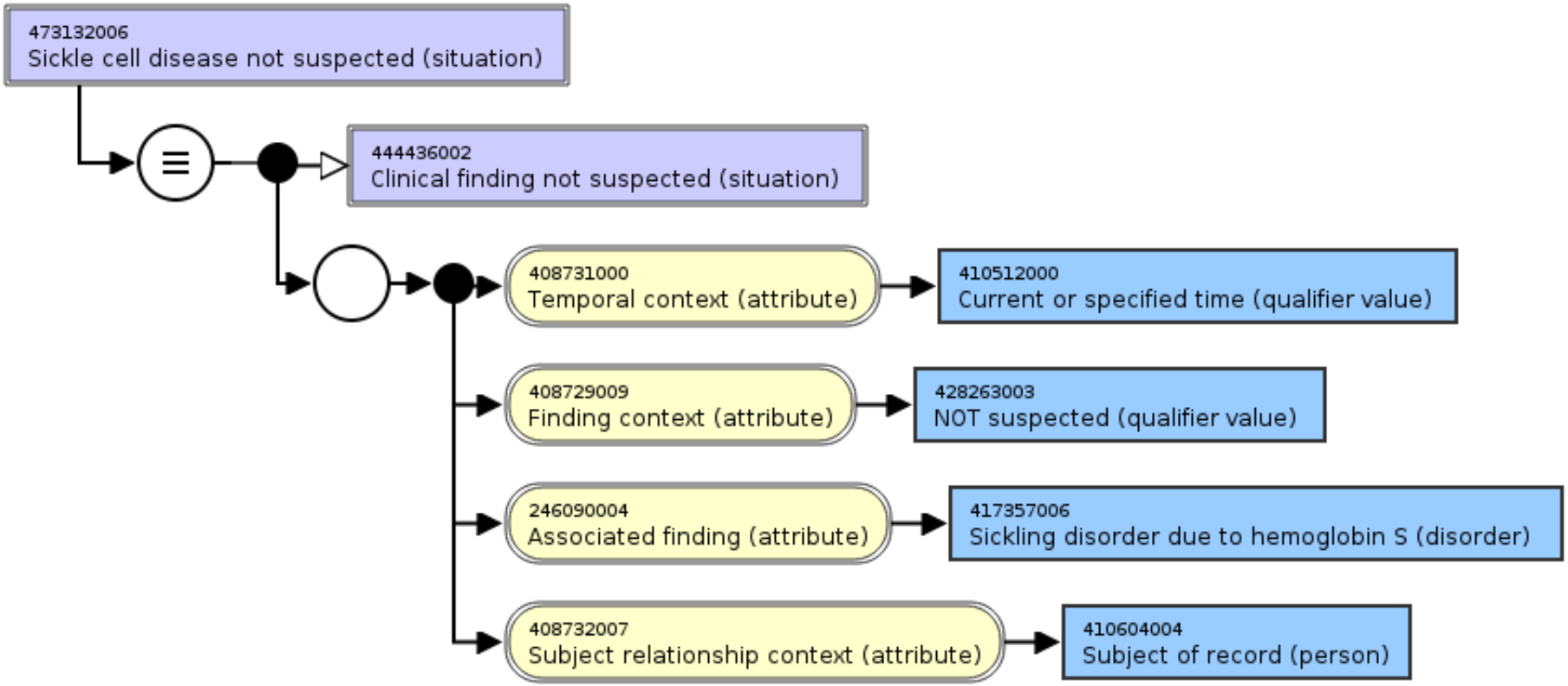
- Known (qualifier value)
  - Known present (qualifier value)
  - Known possible (qualifier value)
    - Suspected (qualifier value)
  - NOT suspected (qualifier value)
  - Known absent (qualifier value)
- Unknown (qualifier value)

- Example:
  - Suspected diabetes mellitus (situation)
  - Sickle cell disease not suspected (situation)
  - No family history diabetes (situation)

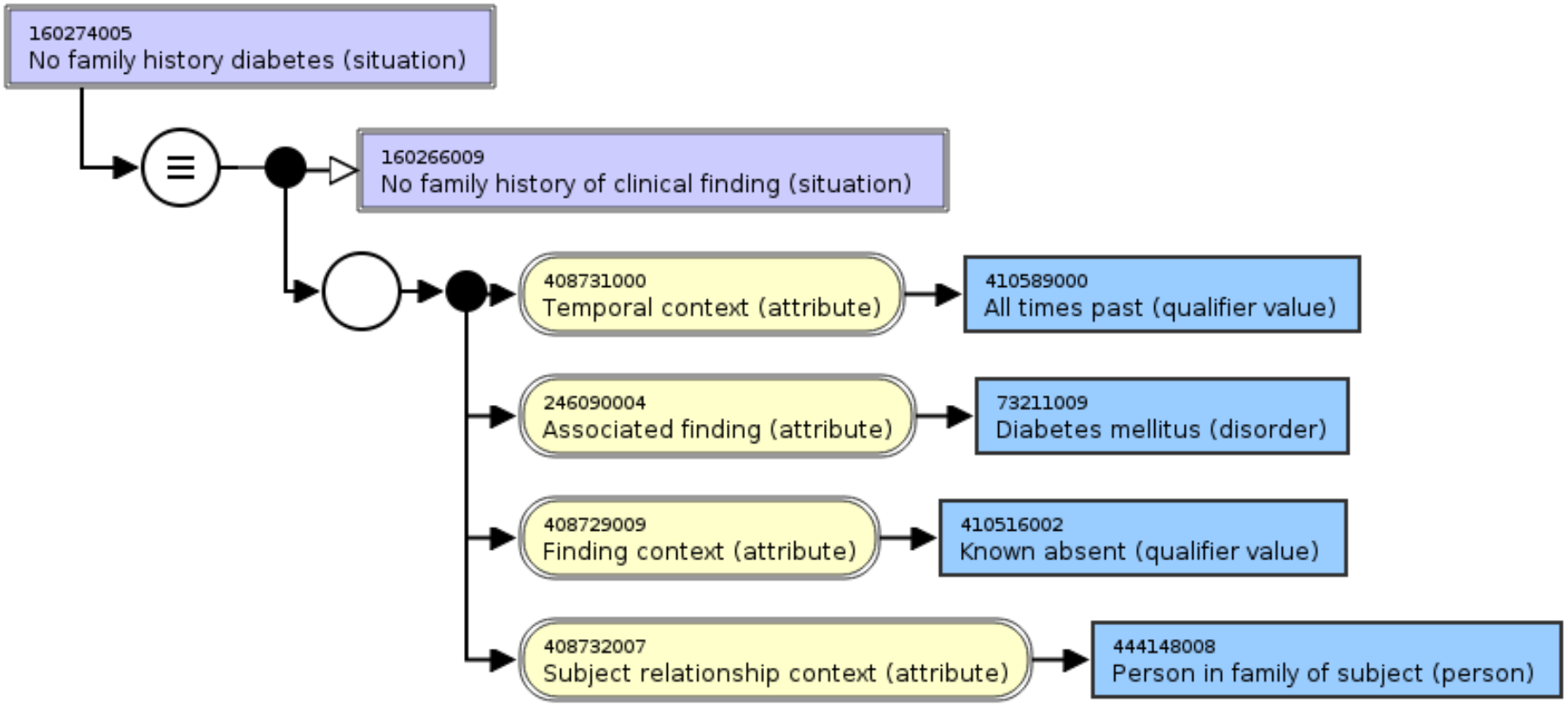
Suspected clinical finding



Clinical finding not suspected



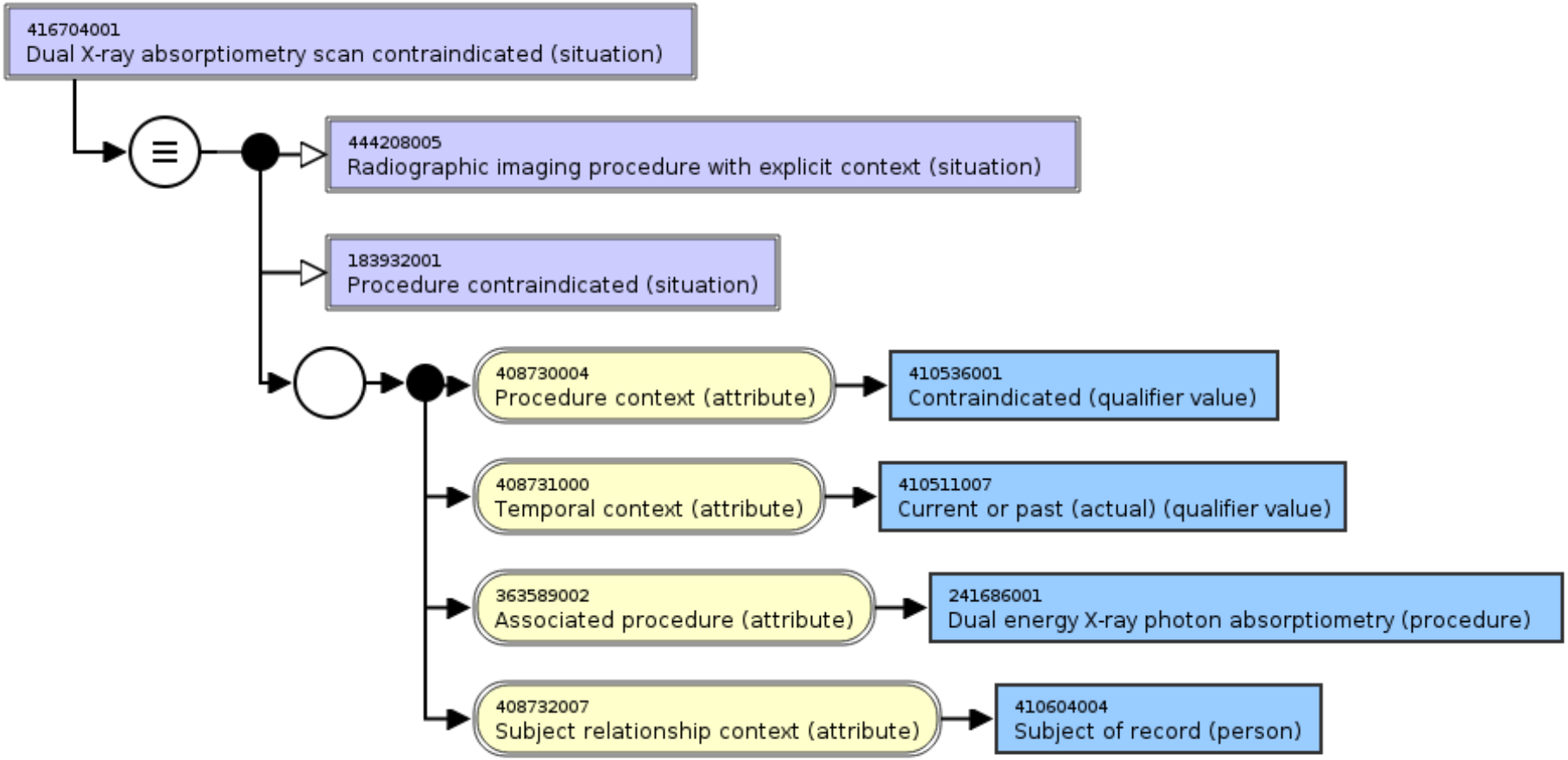
Finding absent



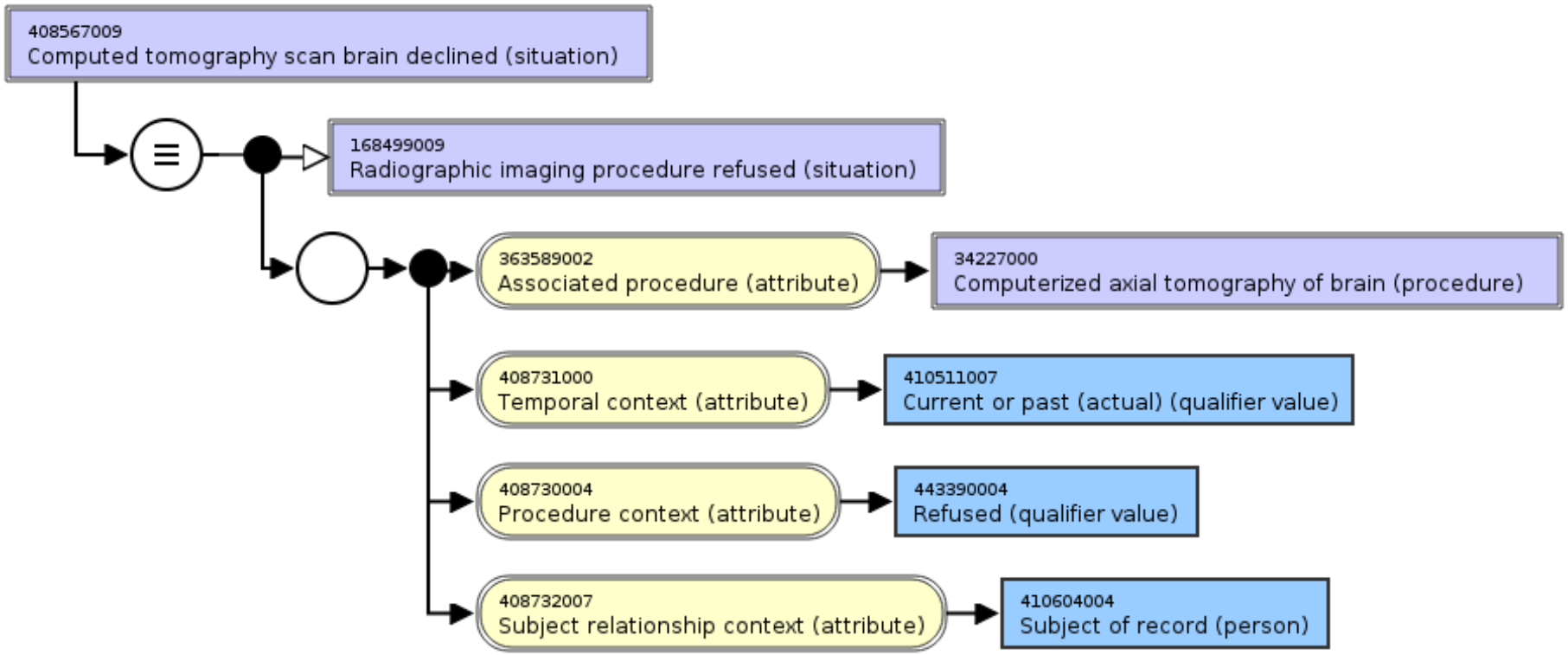
Procedure context values

- Contraindicated (qualifier value)
- Indicated (qualifier value)
- Not indicated (qualifier value)
- Not done (qualifier value)
- Post-starting action status (qualifier value)
  - In progress (qualifier value)
    - Suspended (qualifier value)
    - Started (qualifier value)
  - Ended (qualifier value)
    - Discontinued (qualifier value)
    - Done (qualifier value)
- Pre-starting action status (qualifier value)
  - Not to be done (qualifier value)
    - Refused (qualifier value)
    - Canceled (qualifier value)
  - Organized (qualifier value)
  - To be done (qualifier value)
  - Under consideration (qualifier value)
  - Planned (qualifier value)

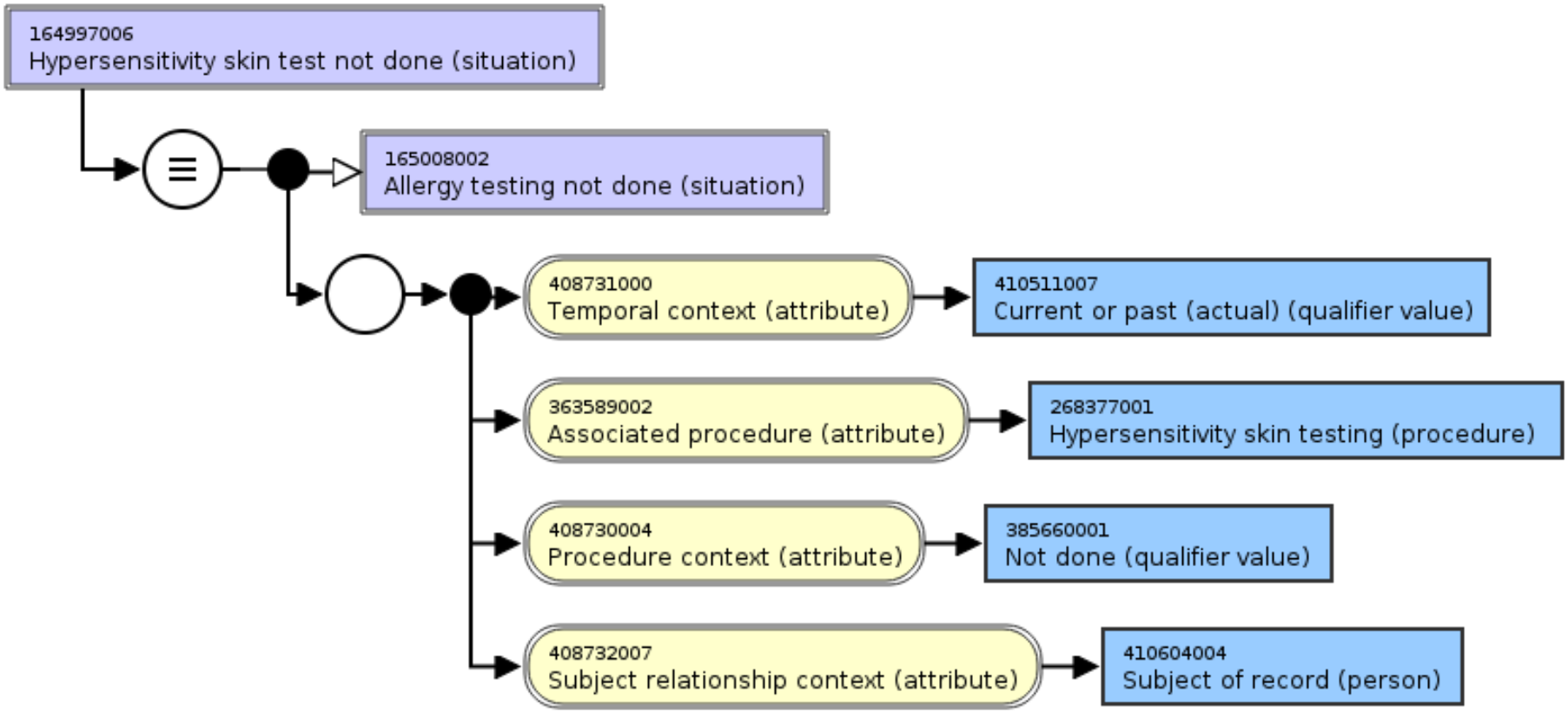
Procedure contraindicated



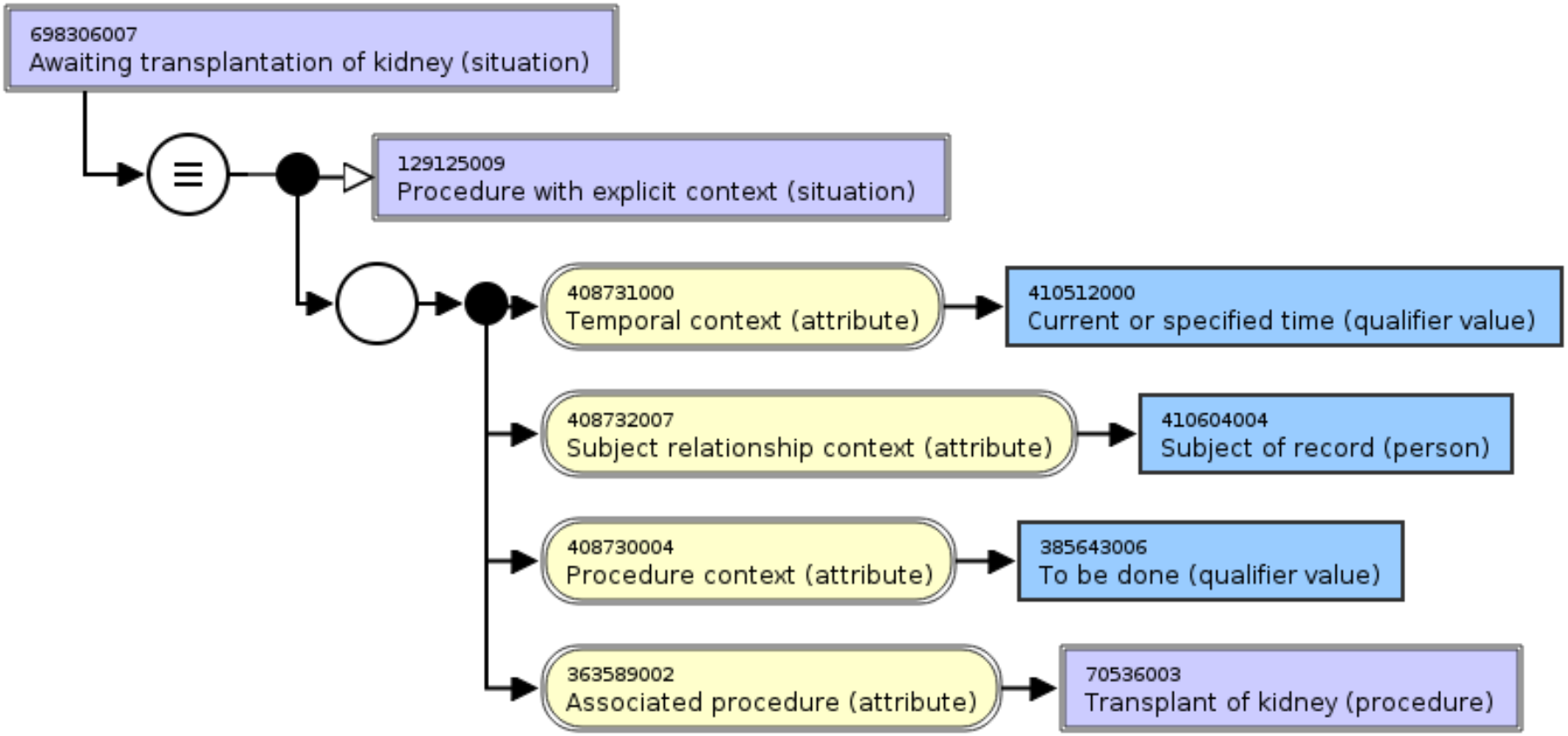
Procedure declined



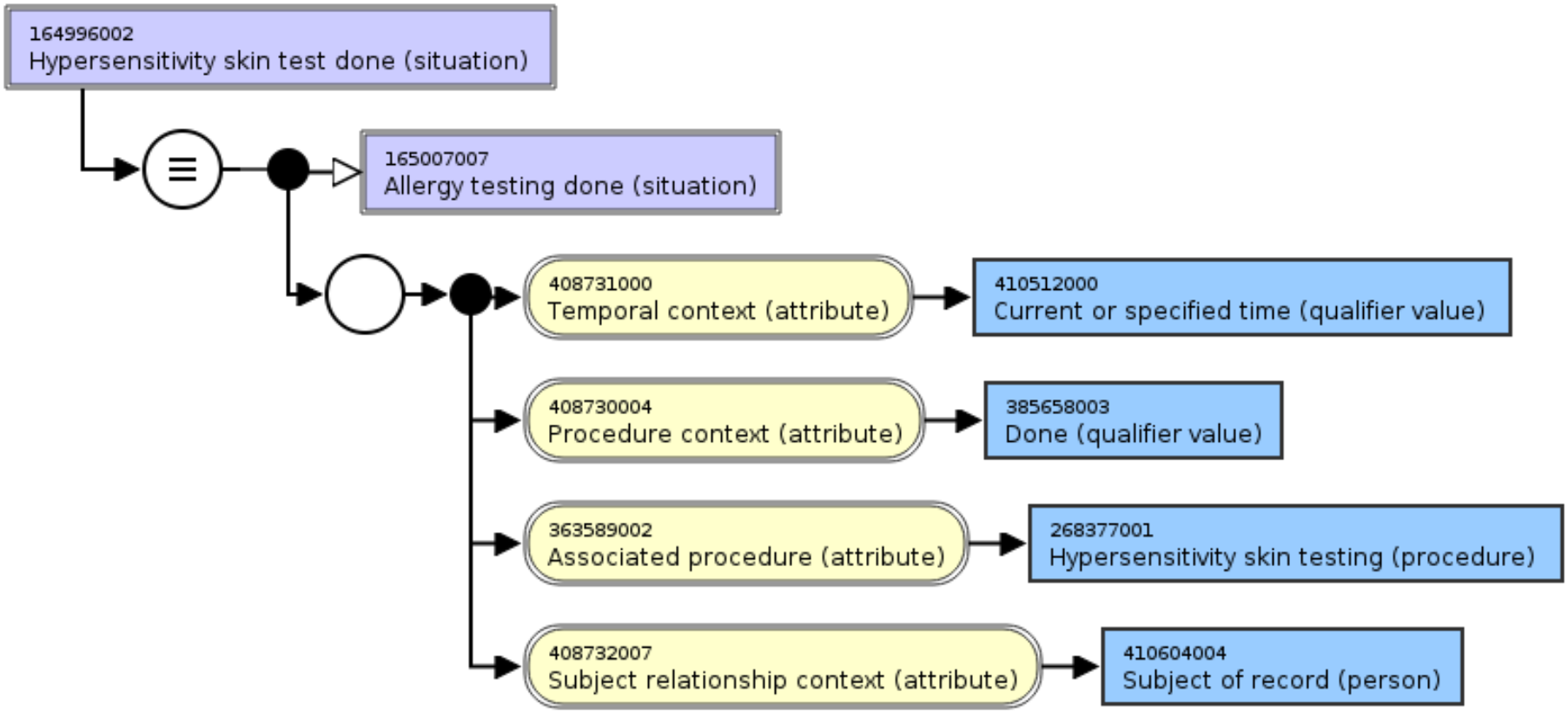
Procedure not done



Procedure to be done



Procedure done



Questions and Discussion

Delivering

SNOMED CT

The global language of healthcare

Contact: info@ihtsdo.org
Website: www.ihtsdo.org

SNOMED CT - Ontological aspects

- What scares some ontologists
  - “SNOMED CT is a terminology”
  - The use of the word “concept” in SNOMED CT
  - Upper level concept “SNOMED CT concept”
  - Compliance with foundational ontologies
  - Continuants (endurants) and occurrents (perdurants) in the same hierarchy, especially the finding / disorder hierarchy
  - Polysemy
  - Textual definitions / fuzzy terms
  - Multiple hierarchies
  - Addition vs. Conjunction
  - Epistemic intrusion
  - Hidden negation

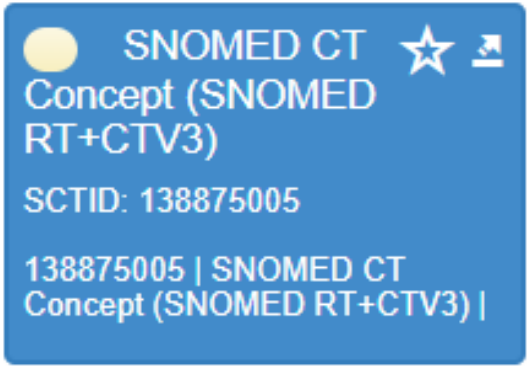
“SNOMED CT is a terminology”

- “Terms” = expression in human language (words, phrases), in opposition of some unfortunate use of the word “term” in ontology circles
- A terminology describes the language of a domain, normally without any clearly defined ontological basis
- SNOMED started as a “nomenclature” (conventions of naming), but dropped this categorization
- SNOMED CT now makes a clear distinction between a “conceptual” (ontological layer) and a terminological layer with terms (“descriptions”) linked to the former
- A good characterization would be the one of a “ontology-based terminology”

The use of the word “concept” in SNOMED CT

- Despite the idiosyncrasies of certain ontologists re “concept”, this word is ambiguous and problematic to translate
- “Concept” is, however, deeply rooted in the SNOMED community
- SNOMED CT adds a new flavour to it, by using it for OWL classes, as well as to OWL object properties (binary predicates)
- It’s just a naming issue, but in order to get closer to the use of words in the OWL specification, it could be wise to use it always modified “SNOMED CT concept”, and to use “class”, “object property” to avoid imprecision

Upper level class “SNOMED CT concept”



- The use of description logics requires a clear-cut type-token (class-member, concept-particular) distinction. Meta-classes are not supported .
- However, this is what the top class “SNOMED CT concept” suggests.
- If I am an instance of 337915000 |Homo sapiens (organism)|, which is subclass of 138875005 |SNOMED CT Concept (SNOMED RT+CTV3)|, then I am an instance of SNOMED CT concept (simple syllogism) !
- Clearly a misnomer, but harmless for the use of SNOMED CT
-

Compliance with foundational ontologies

- SNOMED CT's upper level bears the legacy of the 11 axes of SNOMED 3.5, and is has never been re(designed) following ontological principles
- The same can be said about its object properties ("linkage concepts"). Here, some new ones, e.g. "inheres in" were borrowed from foundational ontologies
- Some SNOMED CT hierarchies can be more easily mapped to foundation (procedures, organisms, substances), others are highly heterogeneous, like qualifier values, in which the role of a class (to serve as a "value") is taken for an ontological category

Subtypes of different upper-level categories in the same hierarchy

- The "finding" hierarchy (and its subhierarchy "disorder") contain
  - material objects (e.g. 1694004 |Accessory lobe of lung (disorder)|),
  - processes (25136009 |Ataxic gait (finding)|)
  - dispositions (394685004 |High suicide risk (finding)|)
- Ambiguous classes
  - (91936005 |Allergy to penicillin (finding)|) – Allergic disposition or allergic attack?
  - 254838004 |Carcinoma of breast (disorder)| - Mass of tissue or growth process or both?
- Solutions
  - Accept disjunction classes (Object or Process or Disposition)
  - "Dot objects" / logical polysemy (mutually dependent entities)
  - Ambiguity of language, disambiguation by context

Polysemy

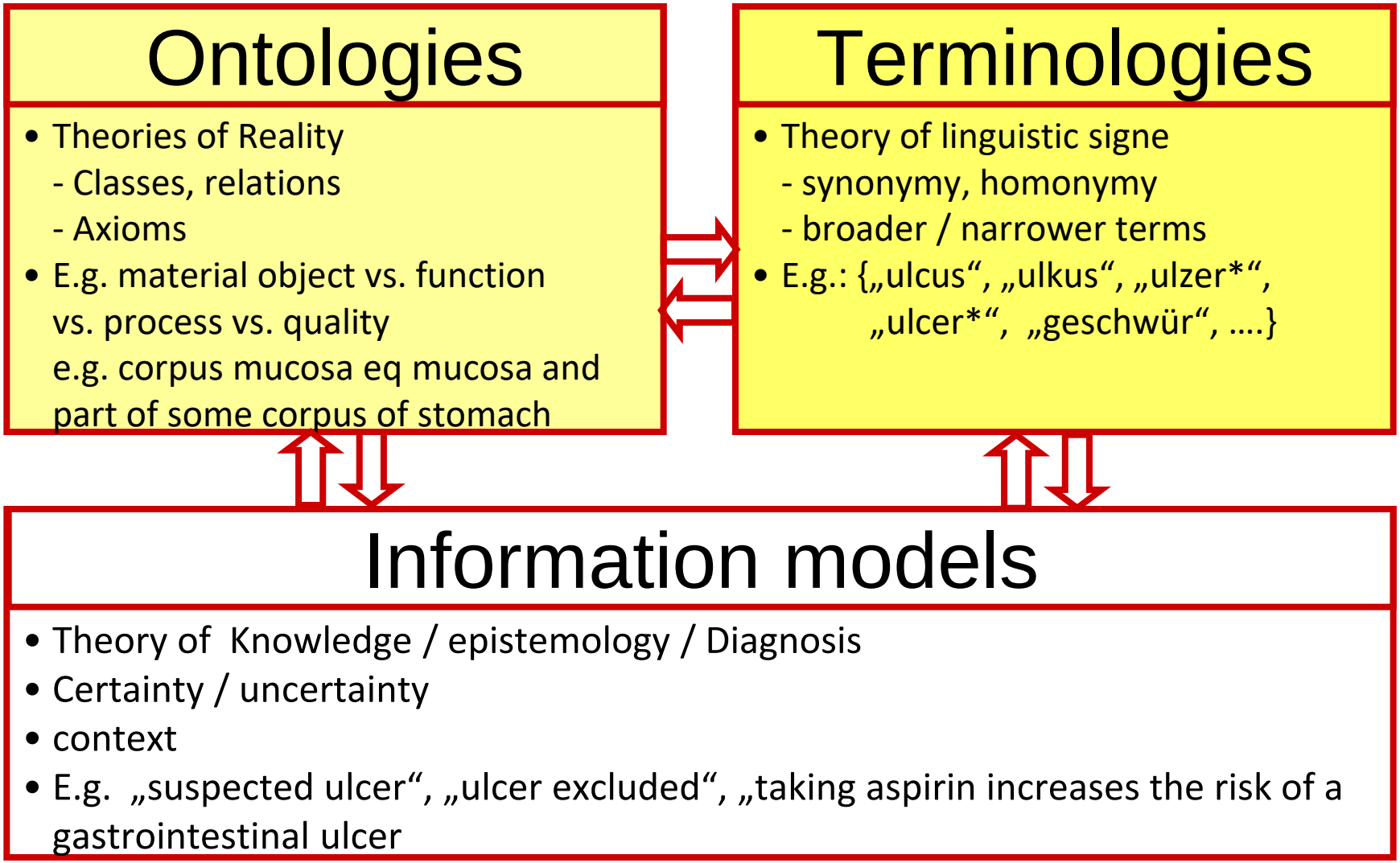
- 118605002 |Hodgkin lymphoma, nodular lymphocyte predominance (disorder)|
- 70600005 |Hodgkin lymphoma, nodular lymphocyte predominance (morphologic abnormality)|

- 18629005 |Administration of drug or medicament (procedure)|
- 419988009 |Action of drug administration (qualifier value)|

Ontology / Epistemology boundary issues

- Boundary between ontological content ("model of meaning") and information model (model of use)
- E.g. "blood pressure:
  - What is blood pressure
  - What is known / what is recorded / what should be recorded about blood pressure

Representational artefacts



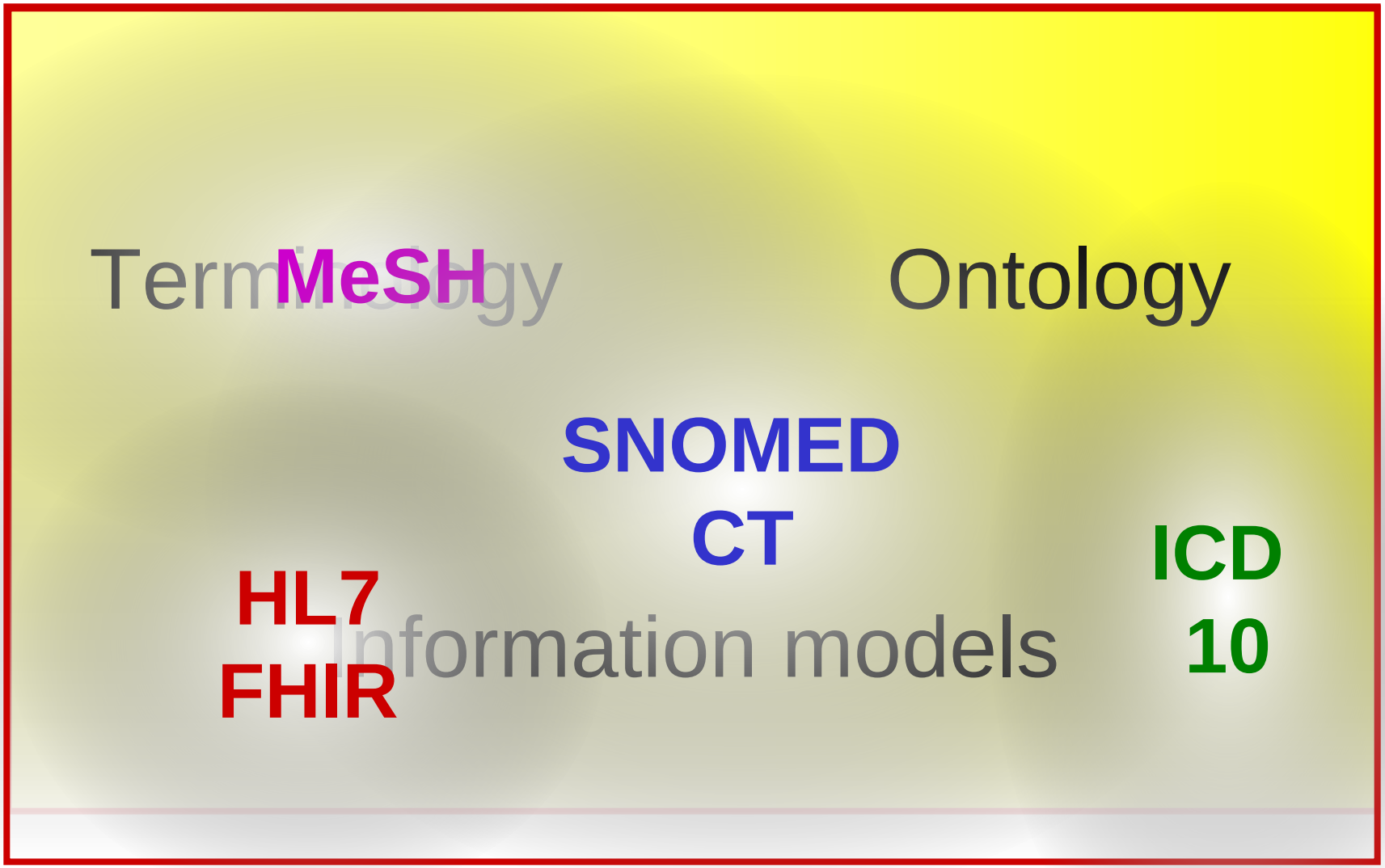
In reality often no clear division

Terminology

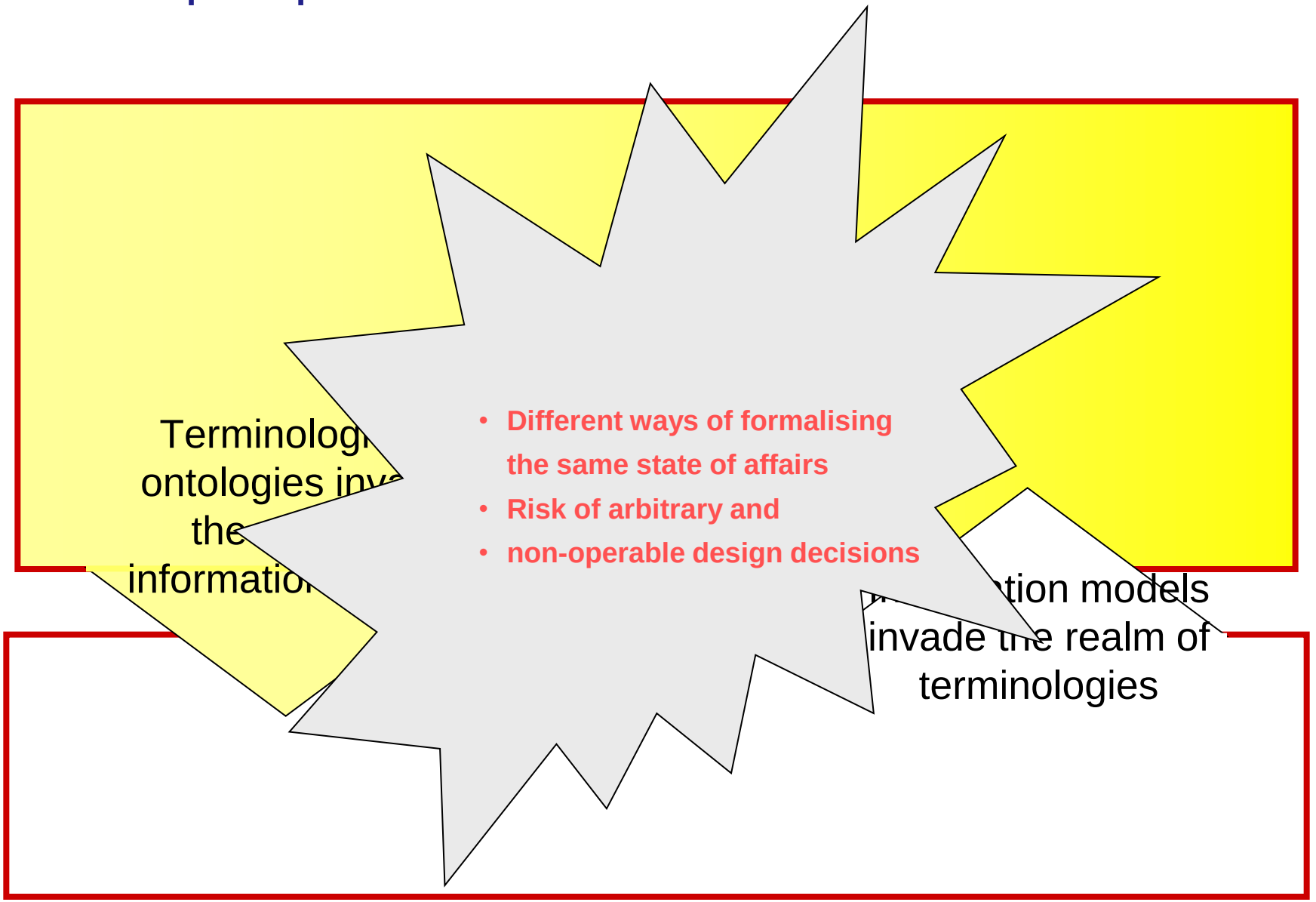
Ontology

Information models

In reality often no clear division



Overlap / epistemic intrusion



Textual definitions

Adolescent MeSH Descriptor Data 2019

Details | Qualifiers | MeSH Tree Structures | Concepts

MeSH Heading	Adolescent
Tree Number(s)	M01.060.057
Unique ID	D000293
Annotation	almost always check tag: NIM; see Manual Chapter 9
Scope Note	A person 13 to 18 years of age.
Entry Term(s)	Adolescence Adolescents Adolescents, Female Adolescents, Male Teenagers Teens Youth
See Also	Minors
Entry Combination	psychology:Psychology, Adolescent
Date Established	1966/01/01
Date of Entry	1999/01/01
Revision Date	2015/06/23

SNOMED CT

Parents

- Autosomal recessive hereditary disorder (disorder)
- ≡ Glycogen storage disease (disorder)

Glycogen storage disease, type II (disorder)

SCTID: 274864009

274864009 | Glycogen storage disease, type II (disorder) |

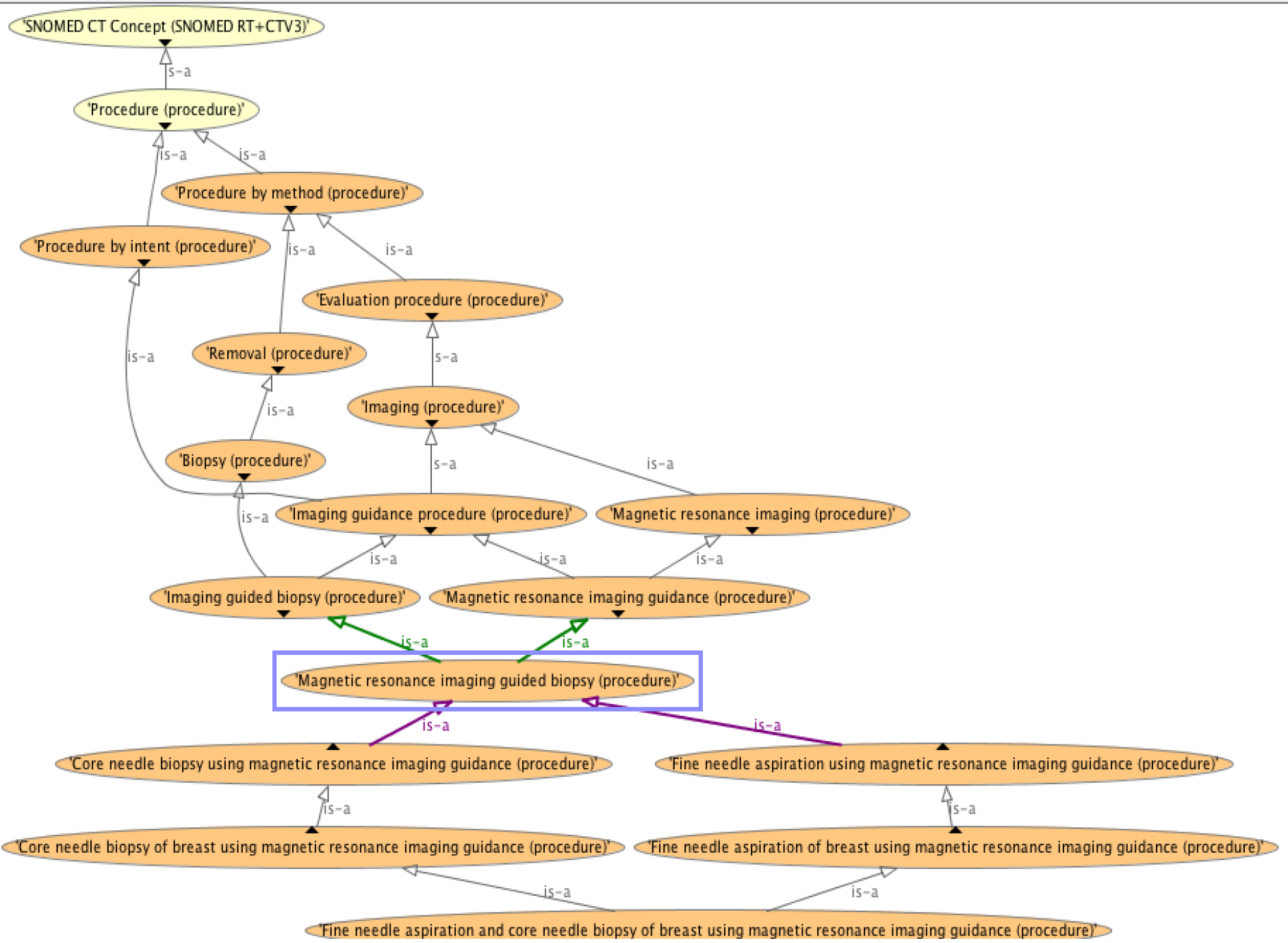
en Alpha-1,4-glucosidase acid deficiency
en Glycogen storage disease due to acid maltase deficiency
en Glycogen storage disease, type II
en Glycogen heart disease
en Pompe's disease
en Pompe disease
en Glycogenosis type II
en Glycogenosis due to acid maltase deficiency
en Glycogen storage disease due to acid maltase deficiency (AMD) is an autosomal recessive trait leading to metabolic myopathy that affects cardiac and respiratory muscles in addition to skeletal muscle and other tissues. AMD represents a wide spectrum of clinical presentations caused by an accumulation of glycogen in lysosomes.
en Glycogen storage disease, type II (disorder)

Occurrence → Congenital

Textual definitions

Multiple Hierarchies

- Some ontologists reject the idea of multiple hierarchies. This has created some misunderstanding.
- What is their concern:
  - A “pure” ontology corresponds to the Aristotelian principle of Genus + differentia, e.g.
    - A viral disorder is a disorder caused by some virus
    - A hepatic disorder is a disorder located in some liver
    - Viral hepatitis is a hepatic disorder caused by some virus
- However, by logic, these axioms entail that viral hepatitis is a viral disorder
- The principle "stated single hierarchies" → inferred multiple hierarchies is also pursued by SNOMED


'SNOMED CT Concept (SNOMED RT+CTV3)'
is-a
'Procedure (procedure)'
is-a
is-a
'Procedure by method (procedure)'
'Procedure by intent (procedure)'
is-a
is-a
'Evaluation procedure (procedure)'
is-a
'Removal (procedure)'
s-a
'Imaging (procedure)'
is-a
'Biopsy (procedure)'
s-a
is-a
is-a
'Imaging guidance procedure (procedure)'
'Magnetic resonance imaging (procedure)'
is-a
is-a
is-a
'Imaging guided biopsy (procedure)'
'Magnetic resonance imaging guidance (procedure)'
is-a
is-a
'Magnetic resonance imaging guided biopsy (procedure)'
is-a
is-a
'Core needle biopsy using magnetic resonance imaging guidance (procedure)'
'Fine needle aspiration using magnetic resonance imaging guidance (procedure)'
is-a
is-a
'Core needle biopsy of breast using magnetic resonance imaging guidance (procedure)'
'Fine needle aspiration of breast using magnetic resonance imaging guidance (procedure)'
is-a
is-a
'Fine needle aspiration and core needle biopsy of breast using magnetic resonance imaging guidance (procedure)'

Ontological Analysis, Ontological Commitment, and Epistemic Contexts

Stefan Schulz

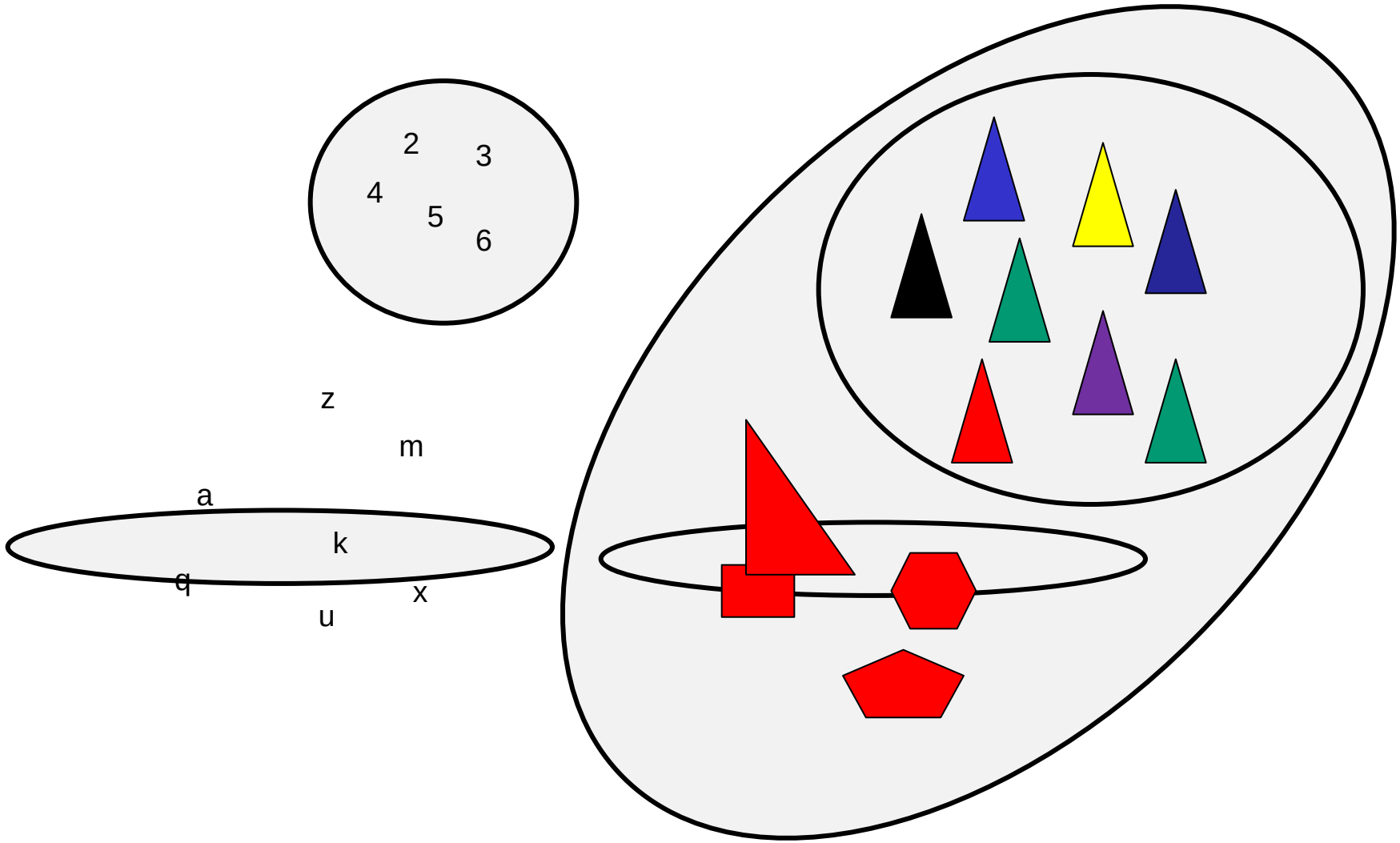
WHO – IHTSDO Joint Advisory Group
First Face-to-Face Meeting
Heathrow, London
15-16 Dcember 2010

Ontological analysis

- What do the representational units in a representational artifact represent?
  - members of classes
  - instances of concepts
  - denotation of terms
- What are the entities they are dep… they can't exist) ?
- In which upper level categories do they belong ?

From a description logics perspective most SCT concepts are classes

Classes and their extensions



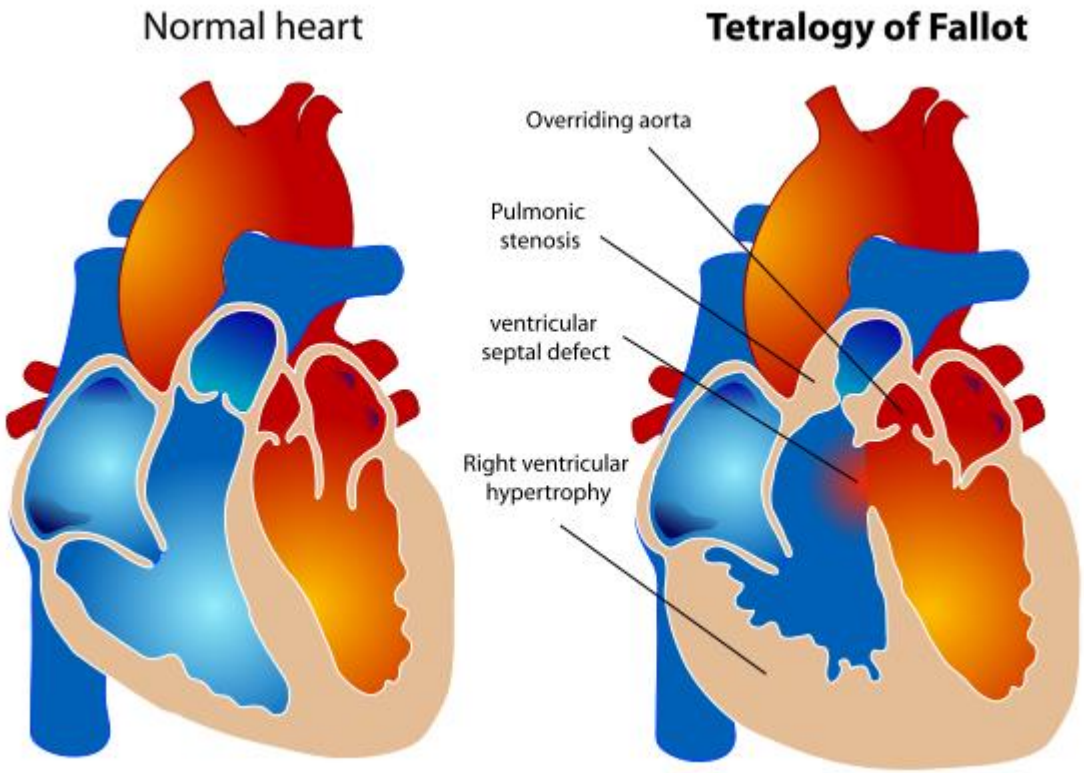
Example

- Representational unit (class, concept, term): Melanoma
- Member, instance: e.g. basal cell carcinoma at left cheek of patient #12334
- Dependencies: every basal cell carcinoma is located in some skin
- Upper level Categories:
  - Material entity?
  - Process?
  - both?
  - what is the ontological commitment of “basal cell carcinoma ”?

Ontological commitment

- “Agreement about the ontological nature of the entities being referred to by the representational units in an ontology” (modified definition following Gruber 93)
- Formal ontologies: subsumption and equivalence statements are either true or false
- Problem: change of truth-value of axioms and sentences according to resulting competing interpretations
- Example: Tetralogy of Fallot in SNOMED CT and ICD10

Tetralogy of Fallot



Q21 **Congenital malformations of cardiac septa**

Excludes: acquired cardiac septal defect (I51.0)

Q21.0 **Ventricular septal defect**

Q21.1 **Atrial septal defect**

Coronary sinus defect
Patent or persistent:
· foramen ovale
· ostium secundum defect (type II)
Sinus venosus defect

Q21.2 **Atrioventricular septal defect**

Common atrioventricular canal
Endocardial cushion defect
Ostium primum atrial septal defect (type I)

Q21.3 **Tetralogy of Fallot**

Ventricular septal defect with pulmonary stenosis or atresia, dextroposition of aorta and hypertrophy of right ventricle.

Q21.4 **Aortopulmonary septal defect**

Aortic septal defect
Aortopulmonary window

Q21.8 **Other congenital malformations of cardiac septa**

Eisenmenger's defect
Pentalogy of Fallot

Excludes: Eisenmenger's
· complex (I27.8)
· syndrome (I27.8)

Q21.9 **Congenital malformation of cardiac septum, unspecified**

Septal (heart) defect NOS

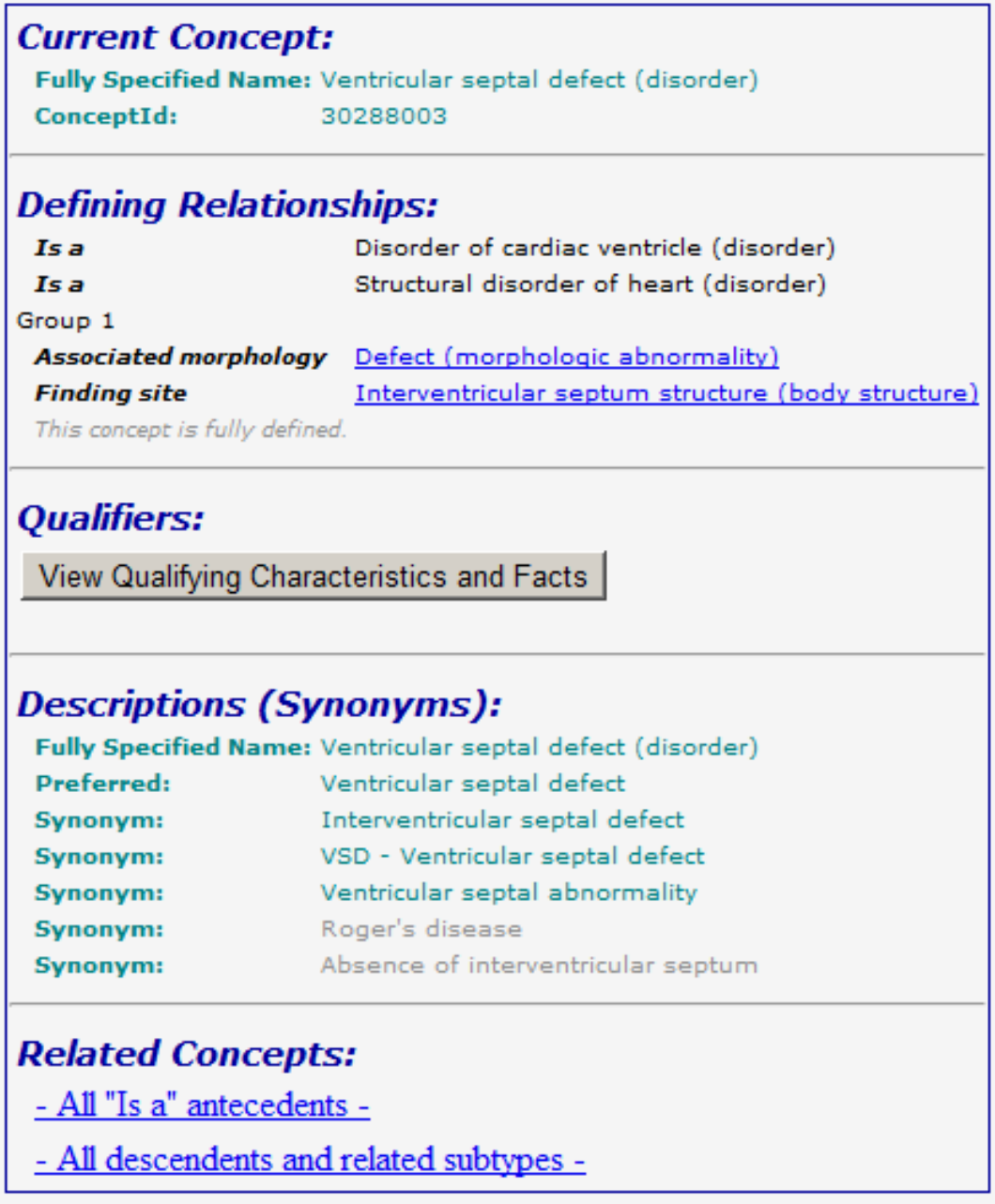
Current Concept:

Fully Specified Name: Tetralogy of Fallot (disorder)
ConceptId: 86299006

Defining Relationships:

Is a	Congenital abnormality of ventricles and ventricular septum (disorder)
Is a	Overriding aorta (disorder)
Is a	Pulmonic valve stenosis (disorder)
Is a	Right ventricular hypertrophy (disorder)
Is a	Ventricular septal defect (disorder)
Occurrence	Congenital (qualifier value)
Group 1	
Associated morphology	Congenital anomaly (morphologic abnormality)
Finding site	Cardiac ventricular structure (body structure)
Group 2	
Associated morphology	Defect (morphologic abnormality)
Finding site	Interventricular septum structure (body structure)
Group 3	
Associated morphology	Stenosis (morphologic abnormality)
Finding site	Pulmonary valve structure (body structure)
Group 4	
Associated morphology	Overriding structures (morphologic abnormality)
Finding site	Thoracic aorta structure (body structure)
Group 5	
Associated morphology	Hypertrophy (morphologic abnormality)
Finding site	Right ventricular structure (body structure)

This concept is primitive.



Every heart disorder that includes a defect of an interventricular septum structure is a ventricular septum defect.

Therefore tetralogy of Fallot is a kind of ventricular septum defect

Tetralogy of Fallot definition

SNOMED CT:

TetralogyOfFallot equivalentClass

PulmonicValveStenosis and *VentricularSeptalDefect* and *OverridingAorta* and *RightVentricularHypertrophy*

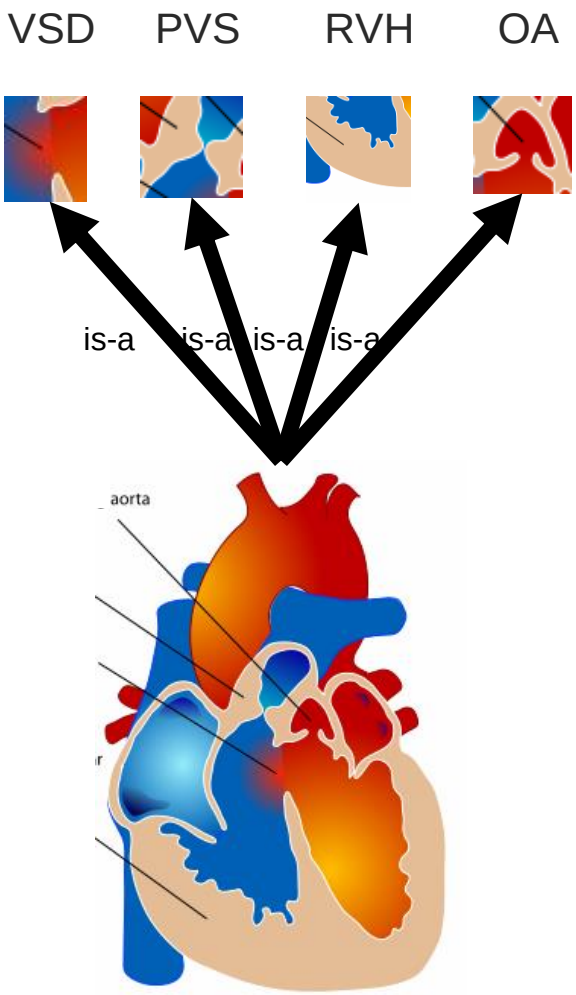
TetralogyOfFallot is a child concept of *VentricularSeptalDefect*

ICD10:

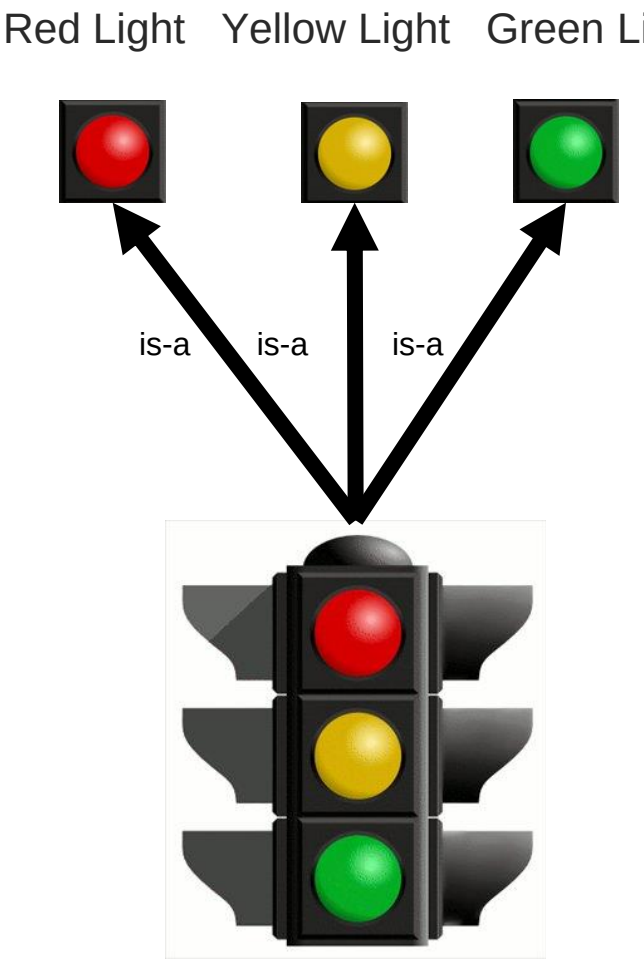
Tetralogy of Fallot is a child of “congenital malformations of cardiac septa” and a sibling of “ventricular septal defect”

Proper parts or taxonomic parents ?

Example from Harold Solbrig



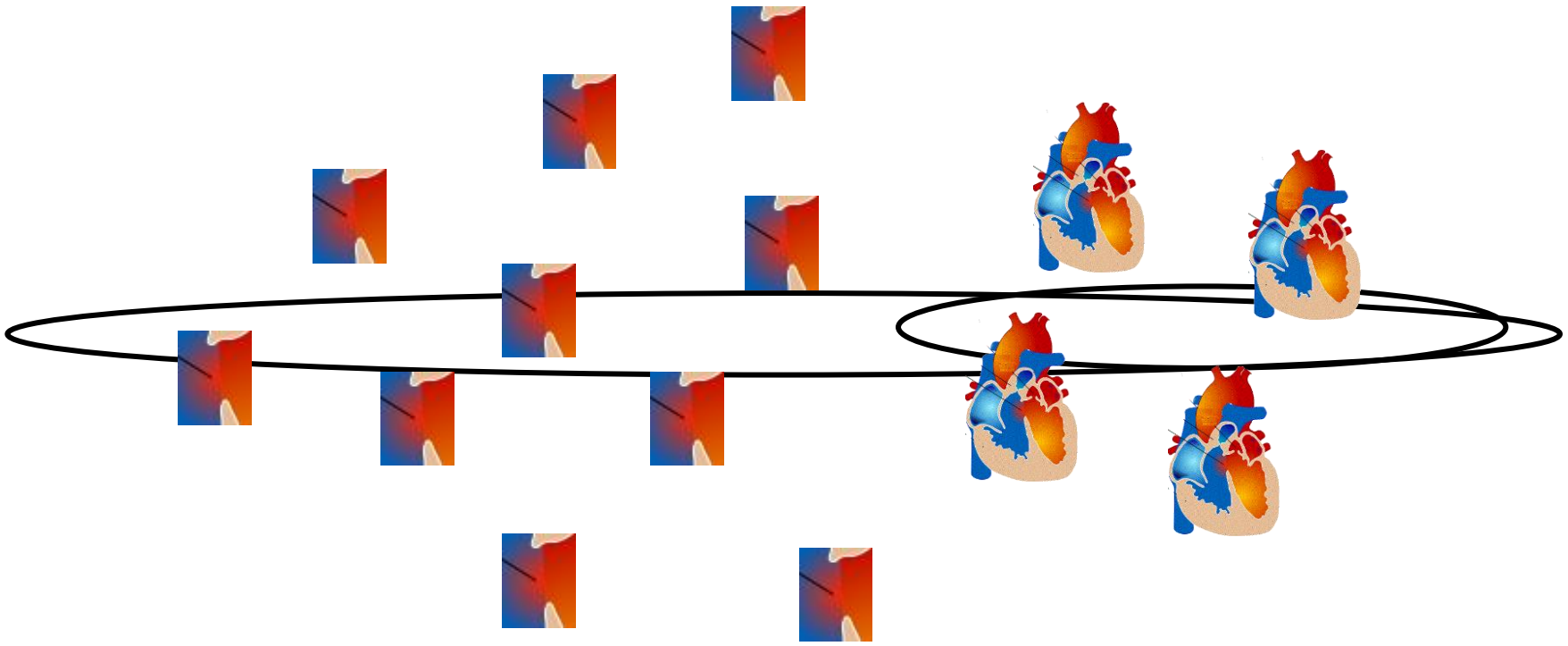
Tetralogy of Fallot



Traffic Light

Ontological Commitment 1

Extension of "*Ventricular Septal Defect* " includes extension of "Tetralogy of Fallot": **FALSE**



Ontological Commitment 2

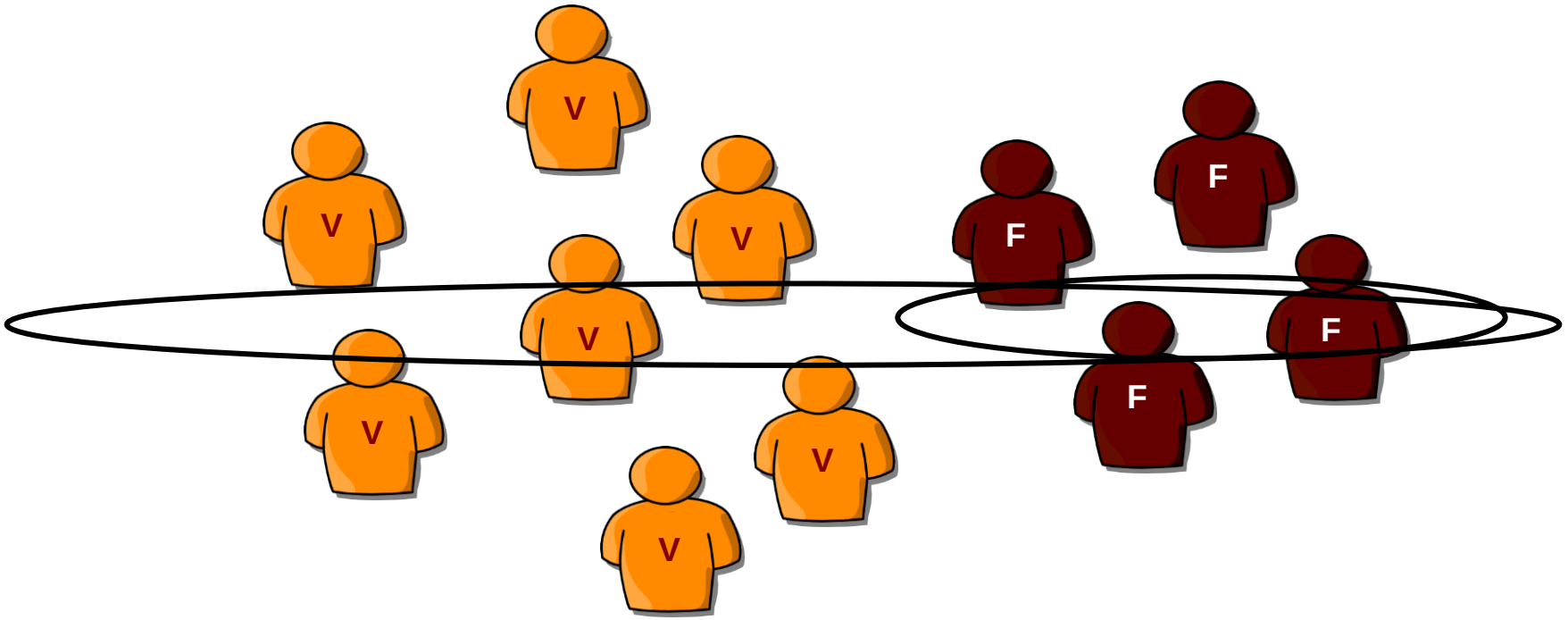
SNOMED CT concepts are instantiated by patients or clinical situations.

- *VentricularSeptalDefect* stands for “Patient with a ventricular septum defect”
- *Tetralogy of Fallot* stands for “Fallot Patient”
- All Fallot patients are also patients with ventricular septum defect because every instance of Tetralogy of Fallot (pathologic structure) has one instance of ventricular septum defect as part

- Consequence:
  - Finding and procedure concepts extend to classes of patients but not to classes of findings or procedures

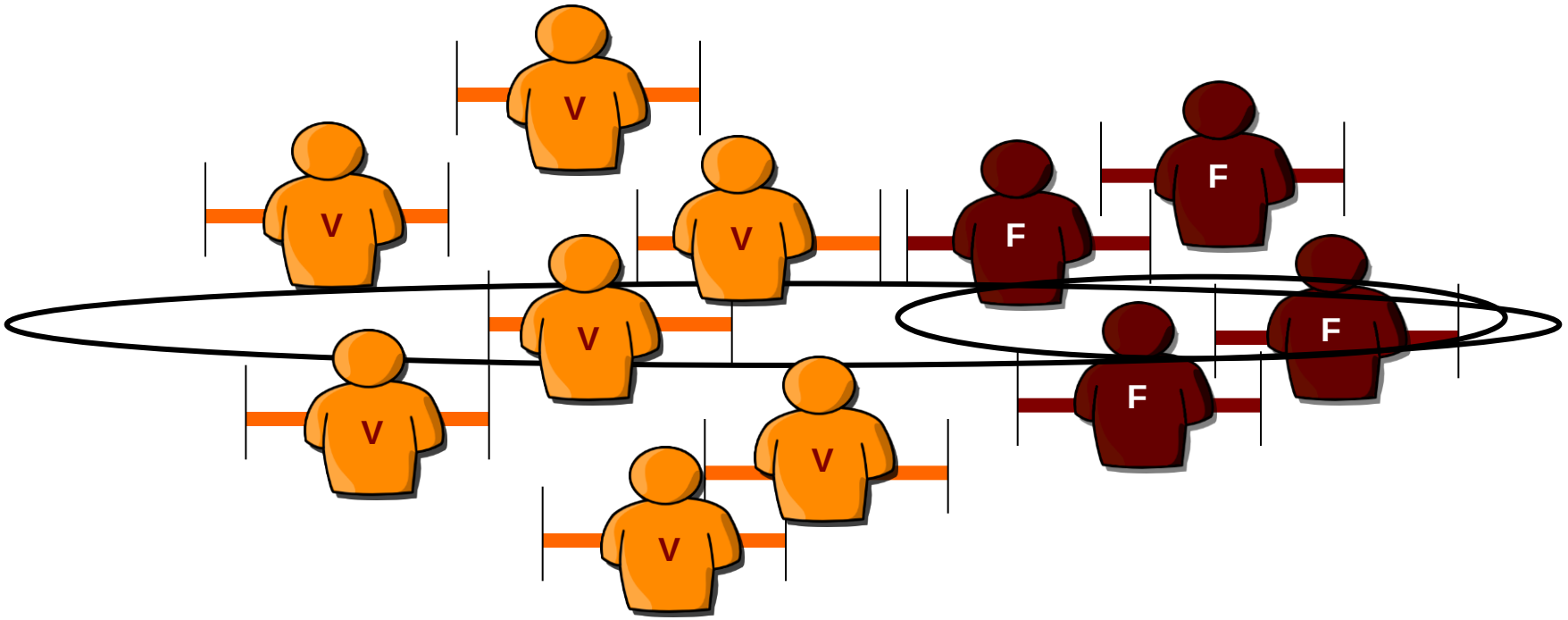
Ontological Commitment 1

Extension of "**Patient with** *Ventricular Septal Defect*" includes extension of "**Patient with** Tetralogy of Fallot": **TRUE**



Ontological Commitment 3

Extension of "**Situation with** Pulmonic Valve Stenosis" includes extension of "**Situation with** Tetralogy of Fallot": **TRUE**



Problem

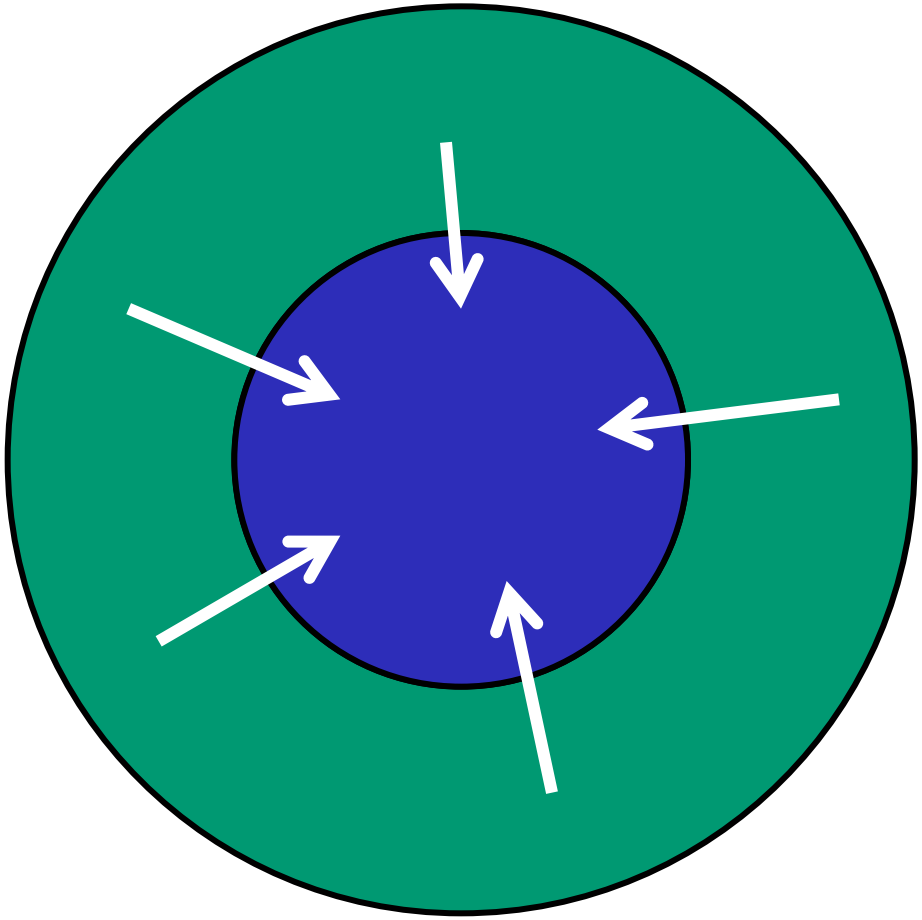
- The same term can be used to denote pathological structures, patients, or situations
- Difficulties with classes that have compositional objects as members
  To what extends “A with B”:
  - a mereological sum A + B?
  - A kind of A which is located in an organism which is also the location of some B ?
  - A kind of B which is located in an organism which is also the location of some A ?
  - the organism?
  - the situation?

Formal representation of complex SNOMED CT expressions.
Schulz S, Markó K, Suntisrivaraporn B.
BMC Med Inform Decis Mak. 2008 Oct 27;8 Suppl 1:S9.

Epistemic contexts

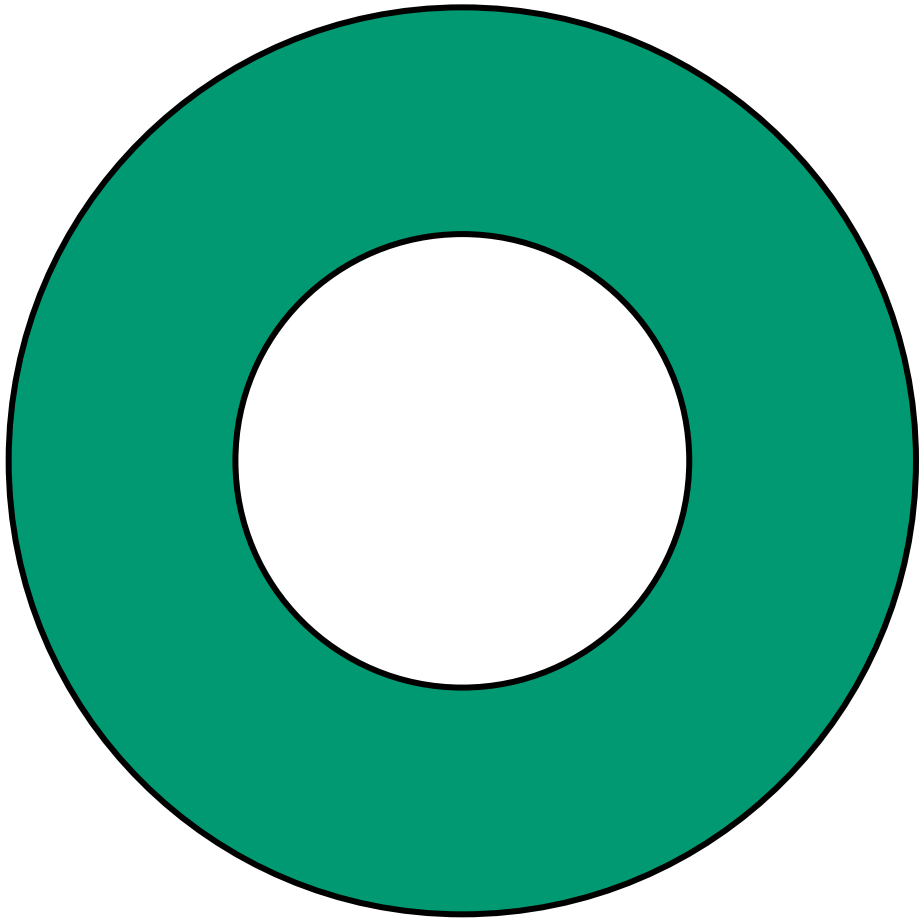
- Pregnancy, not (yet) confirmed
- Diarrhoea of presumed infectious origin
- Atypical squamous cells of uncertain significance, probably benign
- Natural death with probable cause suspected
- Family history of dementia
- Absent foot

both in SNOMED CT and ICD

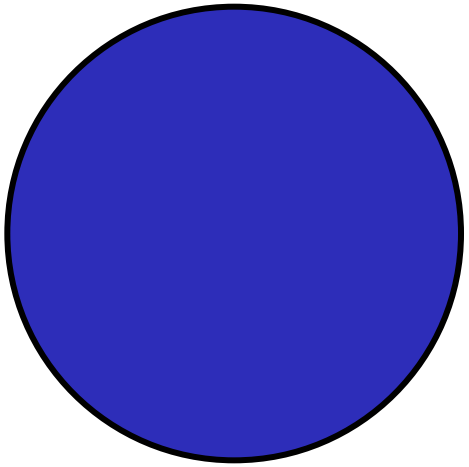


both in SNOMED CT and ICD

Information entities,
Diagnostic statements

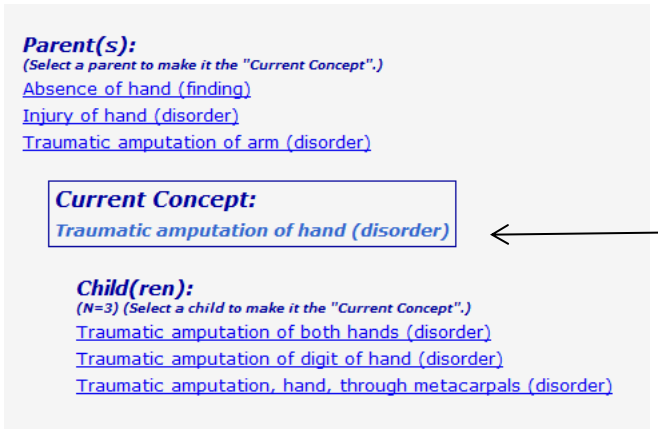


Context-free representation of
diseases,disorders

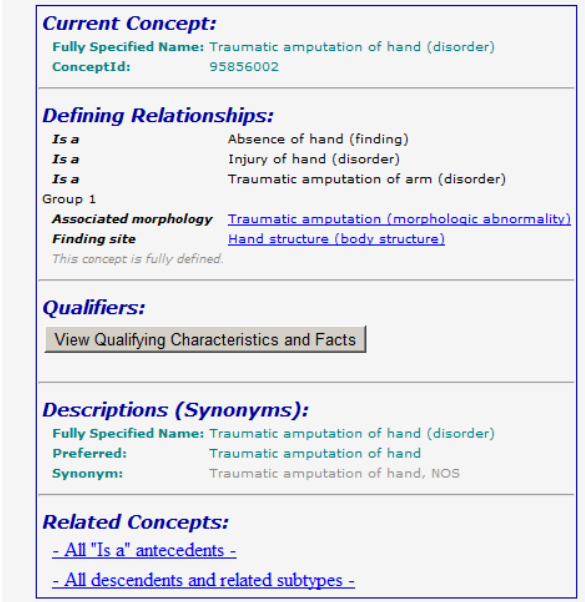


Ontological core

Other example of conflicting meanings



Child



Sibling

S68 **Traumatic amputation of wrist and hand**
S68.0 **Traumatic amputation of thumb (complete)(partial)**
S68.1 **Traumatic amputation of other single finger (complete)(partial)**
S68.2 **Traumatic amputation of two or more fingers alone (complete)(partial)**
S68.3 **Combined traumatic amputation of (part of) finger(s) with other parts of wrist and hand**
S68.4 **Traumatic amputation of hand at wrist level**
S68.8 **Traumatic amputation of other parts of wrist and hand**
S68.9 **Traumatic amputation of wrist and hand, level unspecified**

S58 **Traumatic amputation of forearm**
Excludes: traumatic amputation of wrist and hand (S68.-)
S58.0 **Traumatic amputation at elbow level**
S58.1 **Traumatic amputation at level between elbow and wrist**
S58.9 **Traumatic amputation of forearm, level unspecified**

Conclusions

- Many hierarchies and definitions SNOMED CT suggest that SNOMED CT's ontological commitment is heterogeneous
- SNOMED CT's alternative commitments are completely implicit, thus leaving burden of interpretation to the user.
- But the alternative interpretations shed light on clinicians' reasoning
- Both SNOMED CT and ICD10 mix elements of an ontology with elements of information models (information artifacts)

The Ontology-Epistemology Divide: A Case Study in Medical Terminology

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[4] *Laboratoire d'Informatique Médicale, Université de Rennes I, France*

Abstract. Medical terminology collects and organizes the many different kinds of terms employed in the biomedical domain both by practitioners and also in the course of biomedical research. In addition to serving as labels for biomedical classes, these names reflect the organizational principles of biomedical vocabularies and ontologies. Some names represent invariant features (classes, universals) of biomedical reality (i.e., they are a matter for ontology). Other names, however, convey also how this reality is perceived, measured, and understood by health professionals (i.e., they belong to the domain of epistemology). We analyze terms from several biomedical vocabularies in order to throw light on the interactions between ontological and epistemological components of these terminologies. We identify four cases: 1) terms containing classification criteria, 2) terms reflecting detectability, modality, uncertainty, and vagueness, 3) terms created in order to obtain a complete partition of a given domain, and 4) terms reflecting mere fiat boundaries. We show that epistemology-loaded terms are pervasive in biomedical vocabularies, that the "classes" they name often do not comply with sound classification principles, and that they are therefore likely to cause problems in the evolution and alignment of terminologies and associated ontologies.

Third Intl. Conference on Formal Ontologies in Information systems (FOIS 2004)
http://lhncbc.nlm.nih.gov/lhc/docs/published/2004/pub2004064.pdf

Consolidating SNOMED CT's Ontological Commitment

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Abstract

SNOMED CT is a clinical terminology that provides terms with meaning by logical axioms. This enforces precise agreements about the ontological nature of the entities referred to, commonly described as ontological commitment. We demonstrate that SNOMED CT implicitly supports at least three different kinds of commitments, viz. the reference to (i) independently existing entities,(ii) to representational artifacts, and (iii) to clinical situations. Our analysis shows how the truth-value of a sentence changes according to one of these perspectives. We argue that a clear understanding of to what kind of entities SNOMED CT concepts extend is crucial for the proper use and maintenance of SNOMED CT. We argue that the three kinds of commitment can co-exist but need to be clearly distinguished.

Forthcoming in Applied Ontology