Tutorial: SNOMED CT Concept Model

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Delivering

SNOMED CT

The global
language of
healthcare

Content of tutorial

- Introduction to concept model
  - Concept model
    - Top level categories
    - Attributes
      - IS A – Subtype relationship
      - Defining attributes and values
    - Domains and Ranges
    - Primitive vs. Fully defined
    - Description Logic and auto-classification
    - Stated view vs. Inferred view, Normalised form
  - Expression and syntax
    - Pre - coordinated expression
    - Post - coordinated expression
    - Compositional grammar
    - OWL
- Clinical finding/disorder concept model
- Procedure concept model
- Situation with explicit context concept model

Audience and Objectives

- Audience
  - All standards and terminology leaders, implementers and users

- Objective
  - To understand the importance and the key aspects of concept model for SNOMED CT content development and advanced implementation.

Concept model

- The model for specifying **logical definition** of concepts in SNOMED CT

- The concept model is based on formal description logic

- The editorial rules for the permitted attributes and values

Top level categories

- [Clinical finding](#)
- [Procedure](#)
- [Situation with explicit context](#)
- [Observable entity](#)
- [Pharmaceutical/biologic product](#)
- [Physical object](#)
- [Staging and scales](#)

- Body structure
- Organism
- Substance
- Specimen

- Physical force
- Event
- Environment or geographical location
- Social context

- Qualifier value
- Record artifact
- Special concept
- SNOMED CT Model Component

Attribute – IS A - subtype relationship

- ## All hierarchies are based on true subtype relationship specified explicitly by IS A attribute
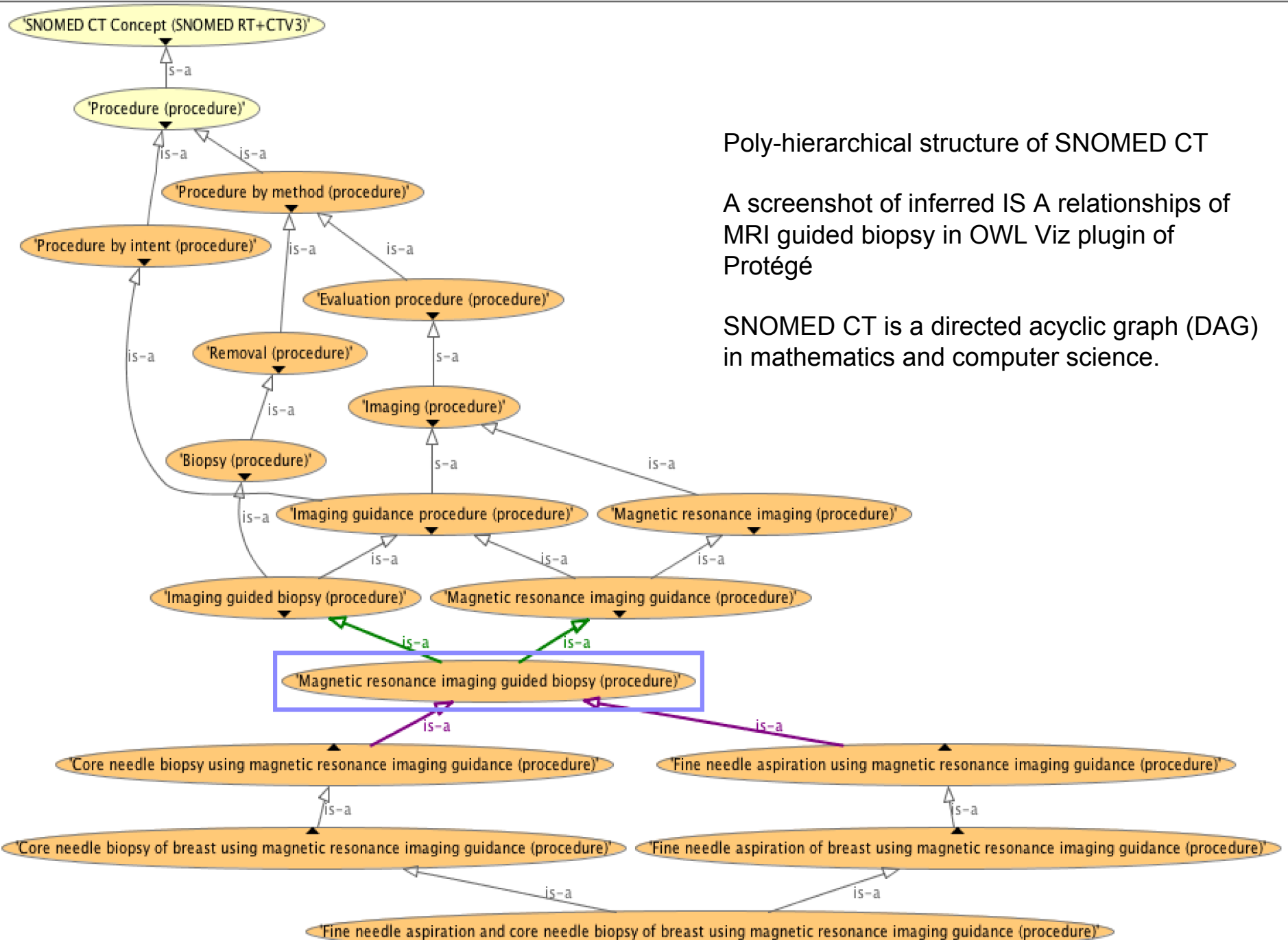 - Not member of
  - Singular vs. plural
  - Not part of

    Joint pain, Pain in multiple joints, ankle pain
    SEP model for anatomy:
      Entire hand, entire thumb
      Hand structure, thumb structure

- ## Poly-hierarchical structure – concept can have more than one parent

Poly-hierarchical structure of SNOMED CT

A screenshot of inferred IS A relationships of MRI guided biopsy in OWL Viz plugin of Protégé

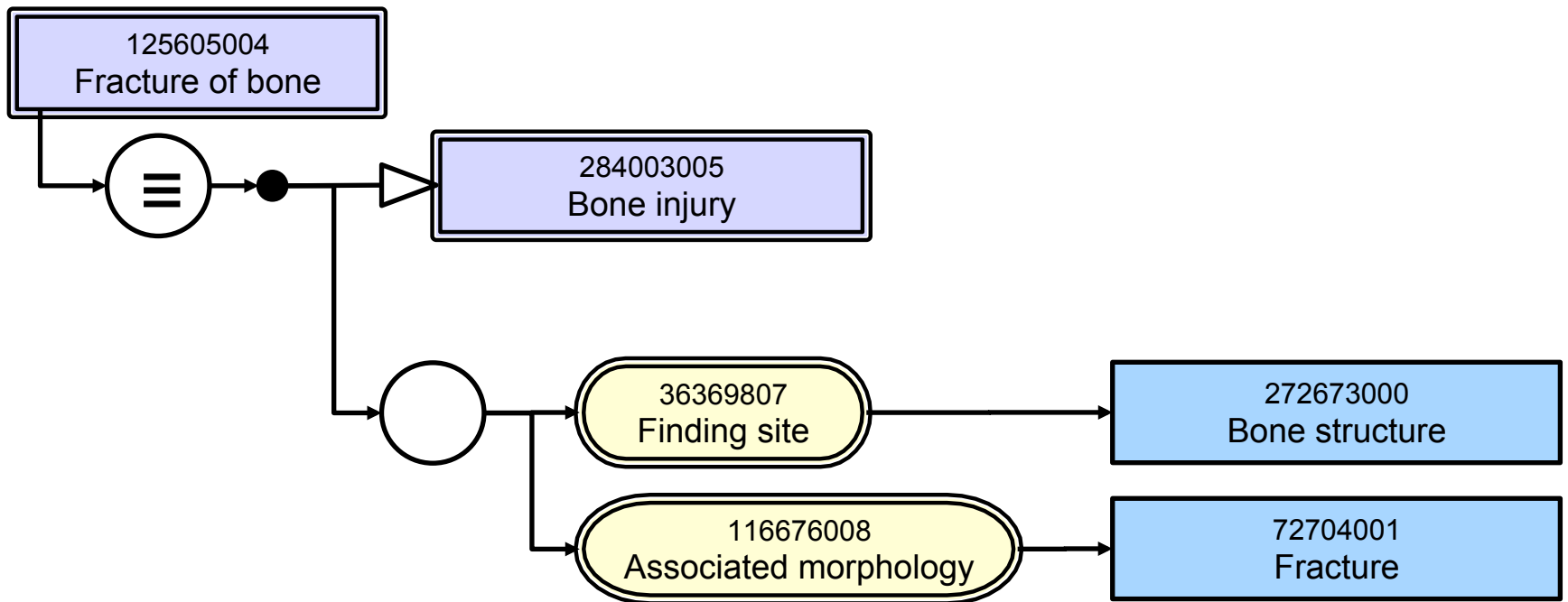SNOMED CT is a directed acyclic graph (DAG) in mathematics and computer science.

Defining attributes and values

IHTSDO standard diagram for representing concept definition
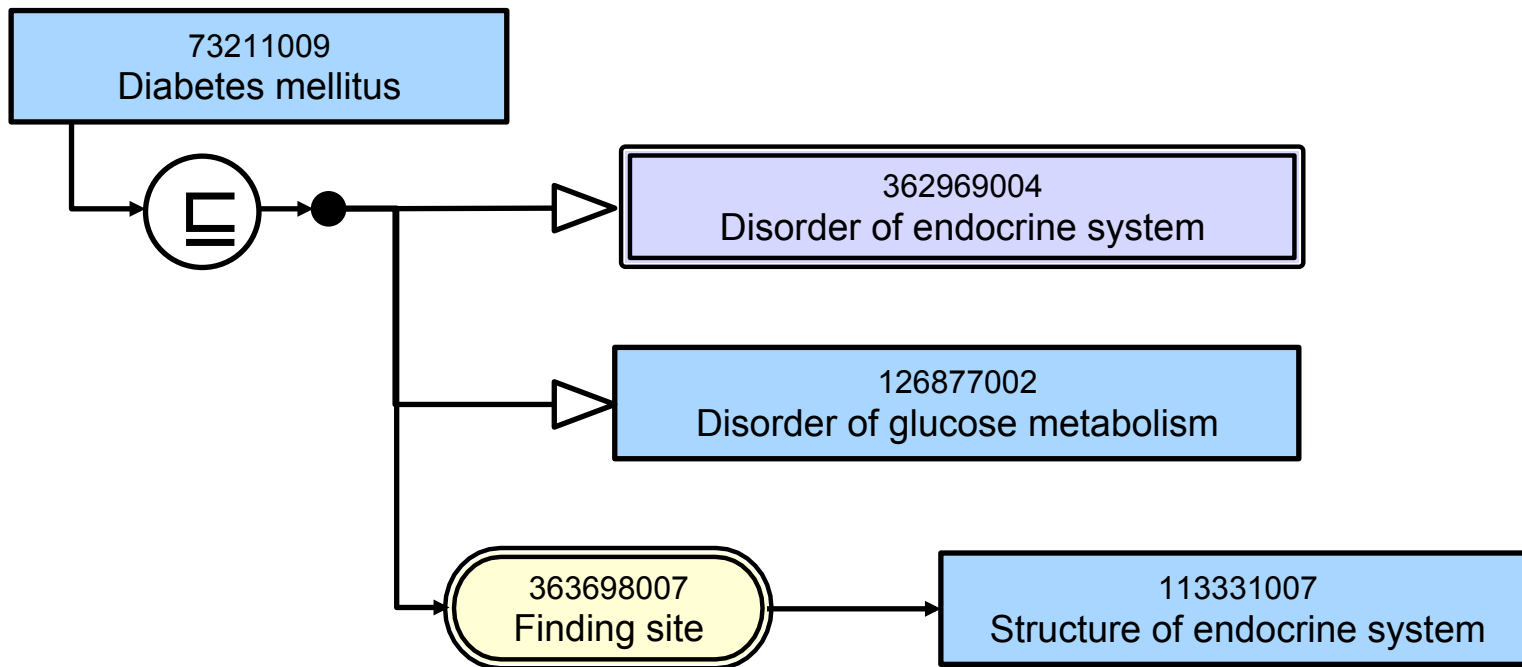
Defining attributes and values

IHTSDO standard diagram for representing concept definition

Role group

- Role grouping had been introduced in SNOMED CT to have clear semantics and correct inferences for complex concepts which 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 **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 example
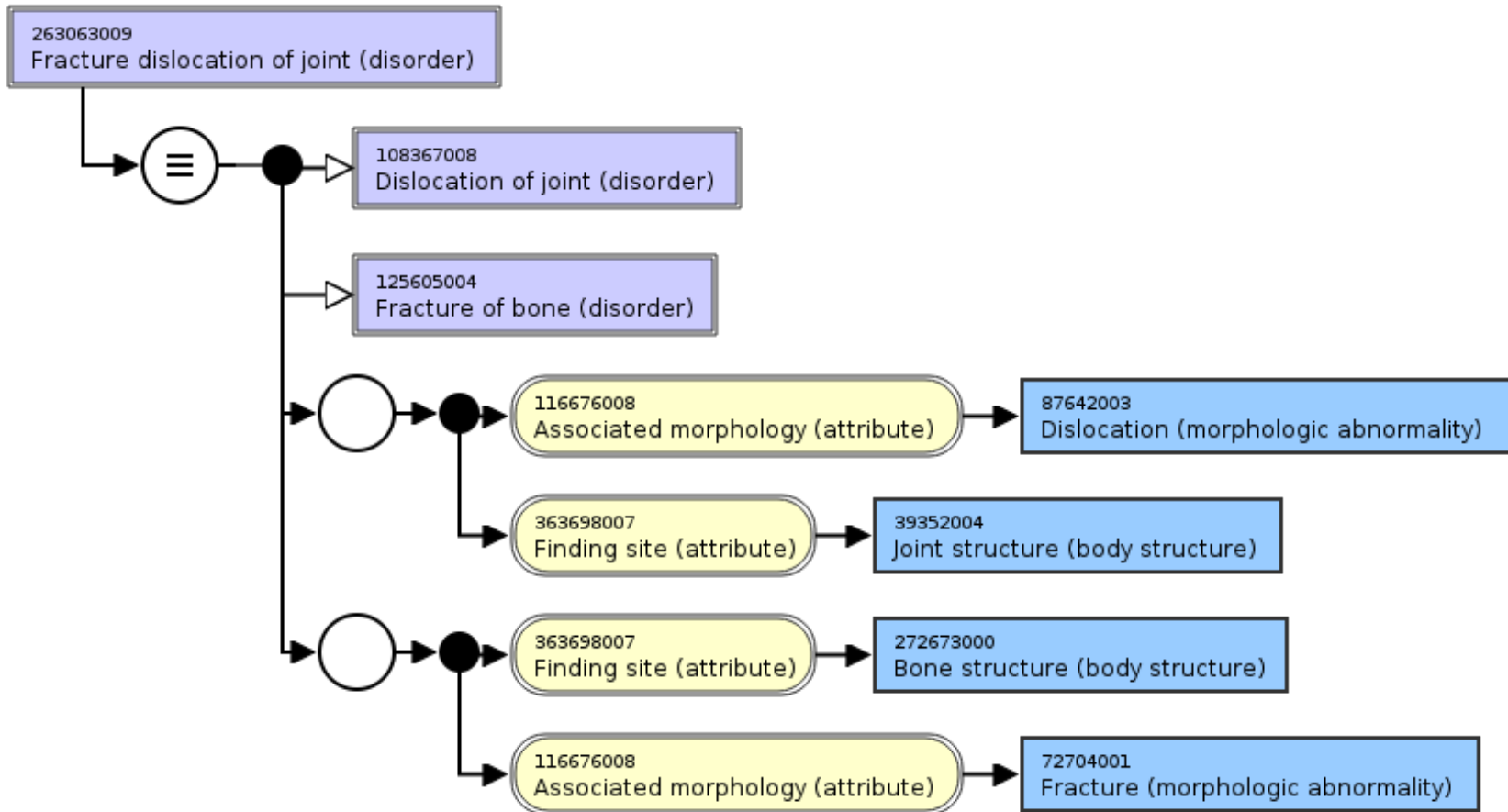
Domain and Range

Specify constraints for concept model
- Domain – an attribute can be applied to
  - Domain: Clinical finding
    - Attributes: finding site, associated morphology
  - Domain: Procedure
    - Attributes: method, procedure site

- Range - an attribute can take values from
  - Attribute: finding site
    - Range: Anatomical or acquired body structure (<<)
  - Attribute: associated morphology
    - Range: Morphologically abnormal structure (<<)
  - Attribute: method
    - Range: Action (<<)

Primitive vs. Fully defined

- ## Primitive
  - ### 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
  - ### Concepts are defined by necessary and sufficient conditions
    - #### Fracture of bone
    - #### MRI guided biopsy

Description Logic and auto-classification

- ## Description logic
  - A family of knowledge representation formalisms that define meaning of terms
  - Extensions of AL (attribute language) distinguished by the implemented constructs

- ## Benefits for using DL for terminology
  - Formal logic based semantics
  - Auto-classification provides inference for equivalence
  - Auto-classification provides inference for subsumption relationships
  - Consistency checking

Description logic – OWL 2 EL profile

- Designed for large bio-health ontologies
- Less expressive but more efficient for computing very large amount data
- SNOMED CT concept model follows EL profile
- Axioms/constructs in EL profile
  - Existential quantification, SOME
  - Intersection of classes, Conjunction, AND
  - Class inclusion – IS A
  - Domain restrictions
  - Range restrictions
  - Class equivalence
  - Object property inclusion – sub-attribute

Description logic – OWL 2 EL profile

- Constructs are not implemented in SNOMED CT
  - Class disjointness
  - Property equivalence
  - Transitive object properties
  - …

- Constructs are not supported by EL profile
  - Universal quantification, ALL, ONLY
  - Disjunction, OR
  - Class negation, NOT
  - Inverse object properties

Incomplete list of DL reasoners

- CEL - http://lat.inf.tu-dresden.de/systems/cel/
- FaCT++ - http://owl.cs.manchester.ac.uk/tools/fact/
- HermiT - http://hermit-reasoner.com
- Pellet - http://clarkparsia.com/pellet
- Racer - https://github.com/ha-mo-we/Racer
- RacerPro - http://www.racer-systems.com
- Snorocket – http://aehrc.com/research/health-data-management-and-semantics/clinical-terminology-tools/snorocket

Expressions

- **Pre-coordinated expression**
    - A concept is represented by a single code
        - 73211009 |Diabetes mellitus|
        - 169069000 |Computed tomography of chest|
        - 7246002 |Kidney biopsy|
        - **12676007 |Fracture of radius|**

- **Post-coordinated expression**
    - A concept is represented by combination of codes

    Expression with role group refinement represents the same concept 12676007.
        - 64572001 |Disease|:
            {116676008 |Associated morphology| =
            72704001 |Fracture|,
            363698007 |Finding site| = 62413002 |Bone
            structure of radius|}

Syntax: Compositional grammar

- ## Simple expression
  73211009 |Diabetes mellitus| or 73211009

- ## Multiple focus concepts
  217724009 |accident caused by blizzard| + 297186008 |motorcycle accident|

- ## 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

Syntax: OWL

- The OWL Web Ontology Language is a standard from W3C
- SNOMED CT in OWL can be generated by Perl scripts that has been included in the international release

DL Syntax	OWL Constructs	Manchester OWL Syntax
$\exists R.C$	someValuesFrom	R some C
$C \sqcap D$	intersectionOf	C and D
$\forall R.C$	allValueFrom	R only C
$\neg C$	complementOf	not C

Stated/inferred views, normalised form

- ## Stated view
  - Attributes and values of a concept definition are stated by a modeler
  - 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
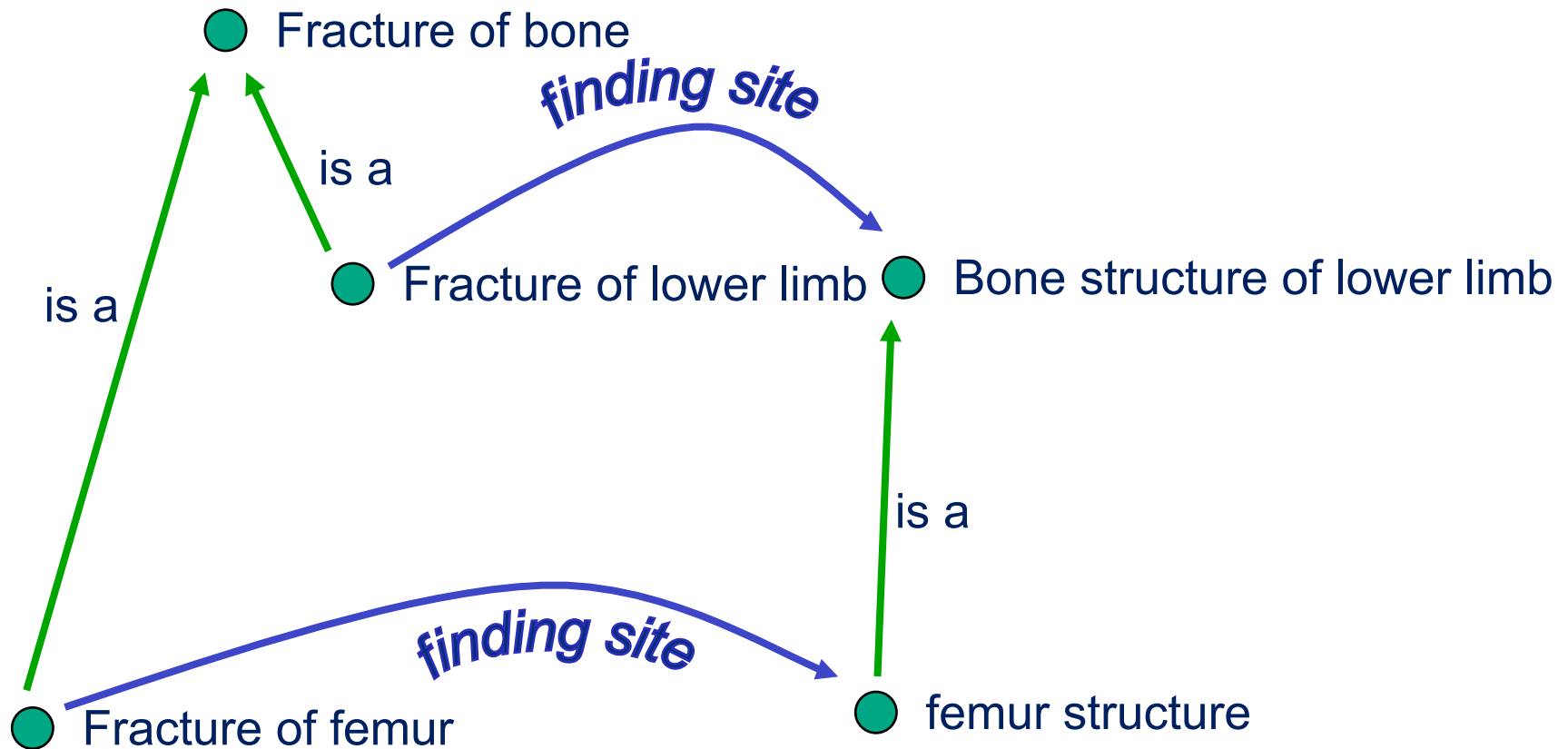
Normal form – Fracture of femur
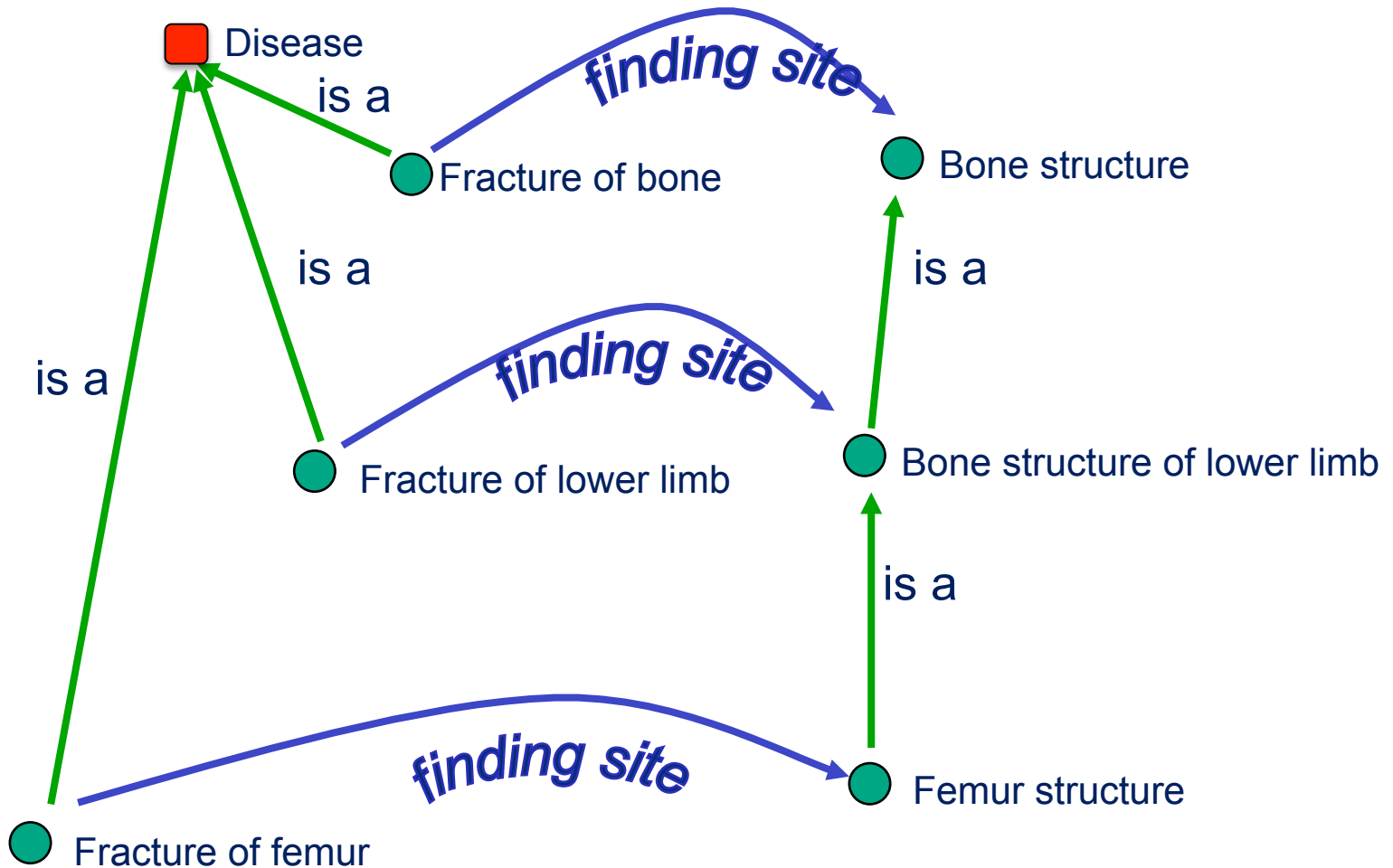
Inferred view – Fracture of femur
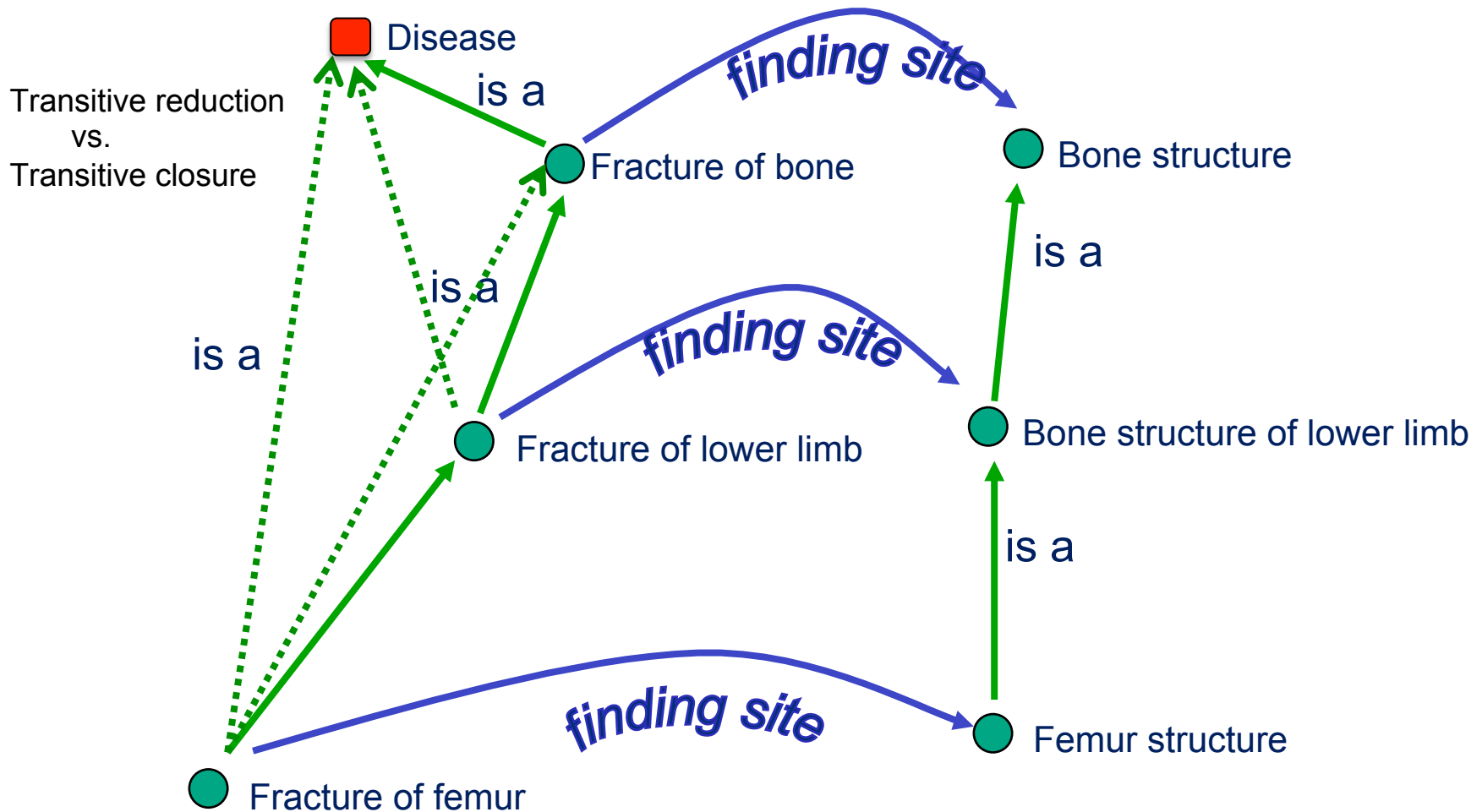
Clinical finding/disorder

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
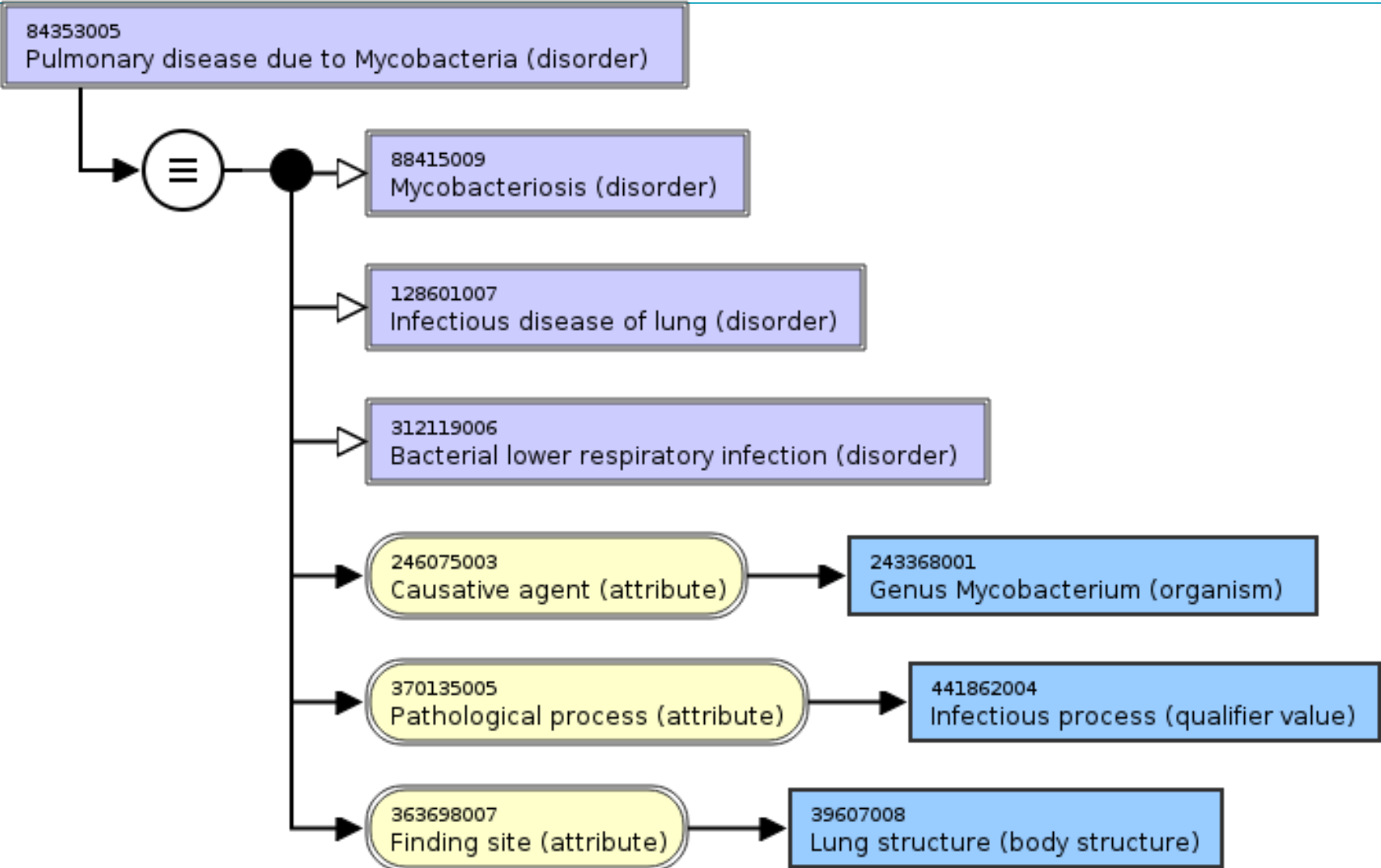
Penicillamine nephropathy
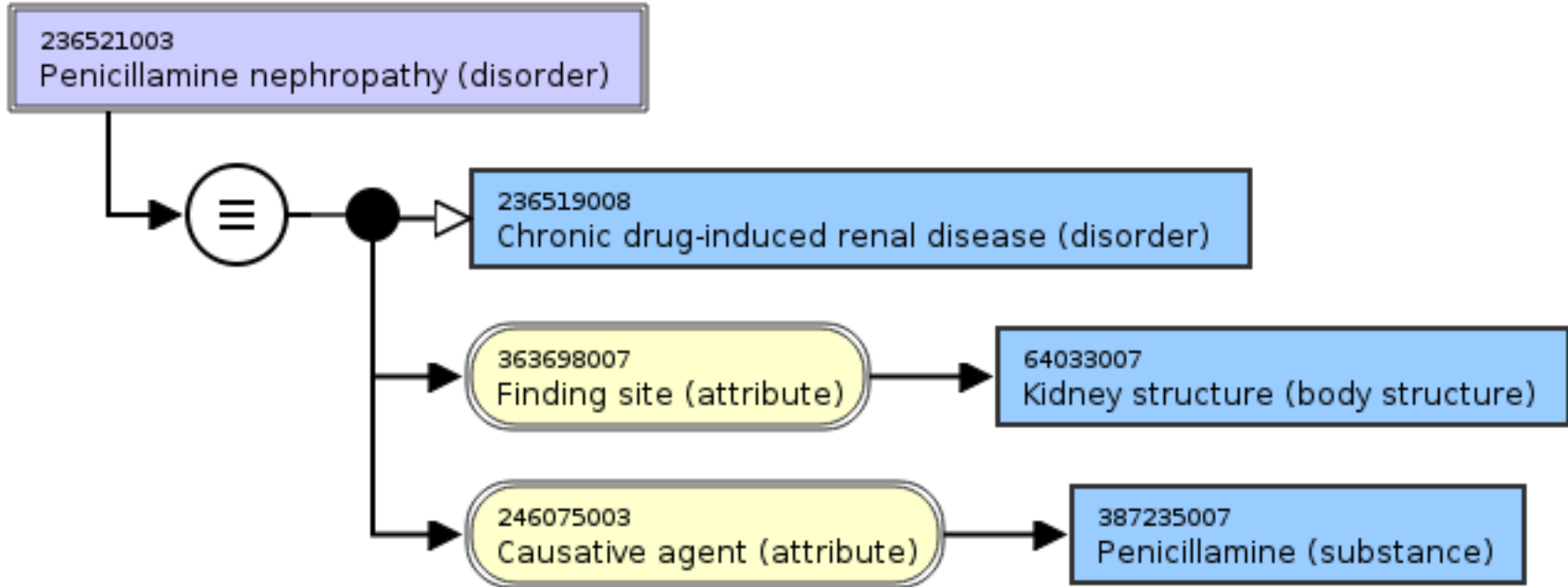
Procedure

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
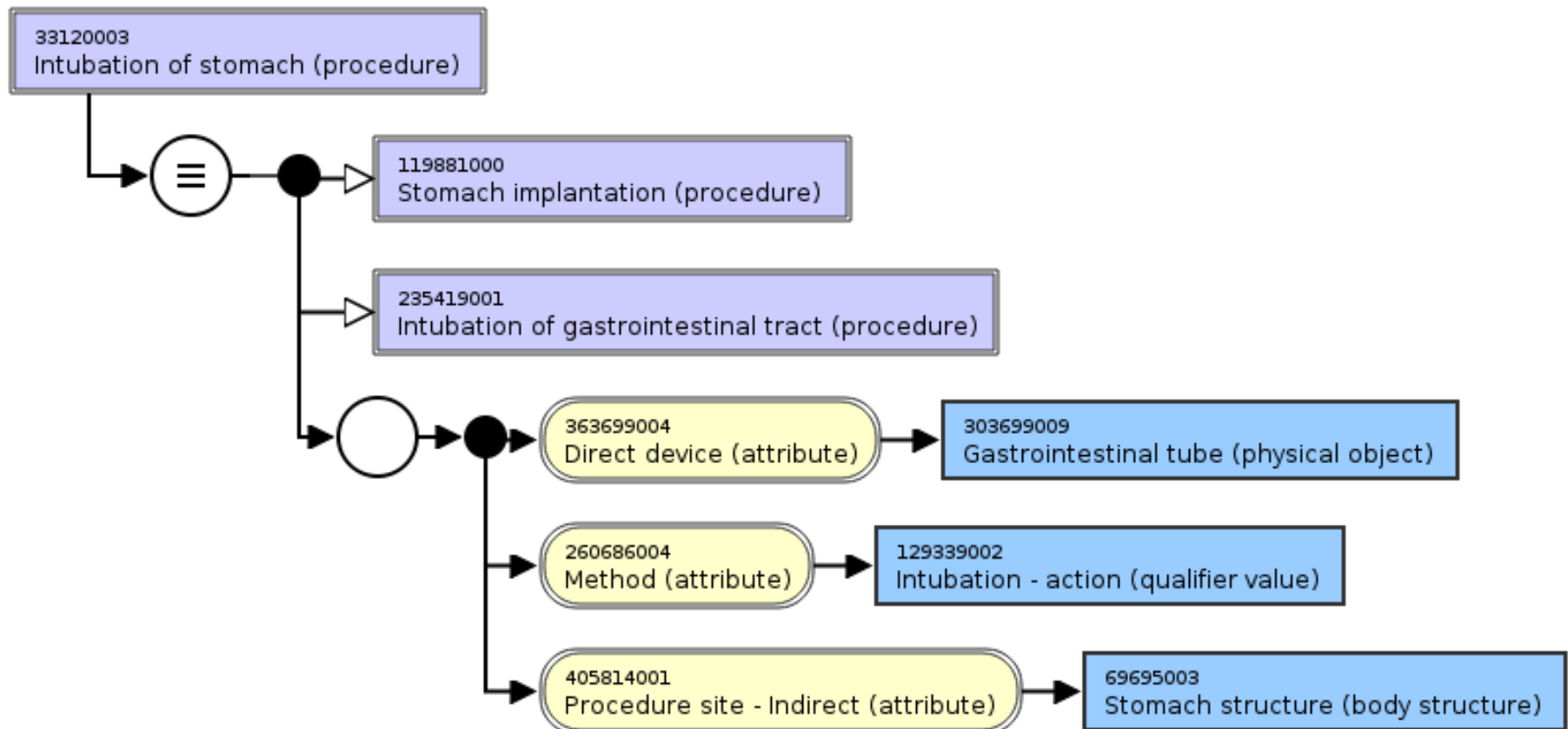
Endoscopic biopsy
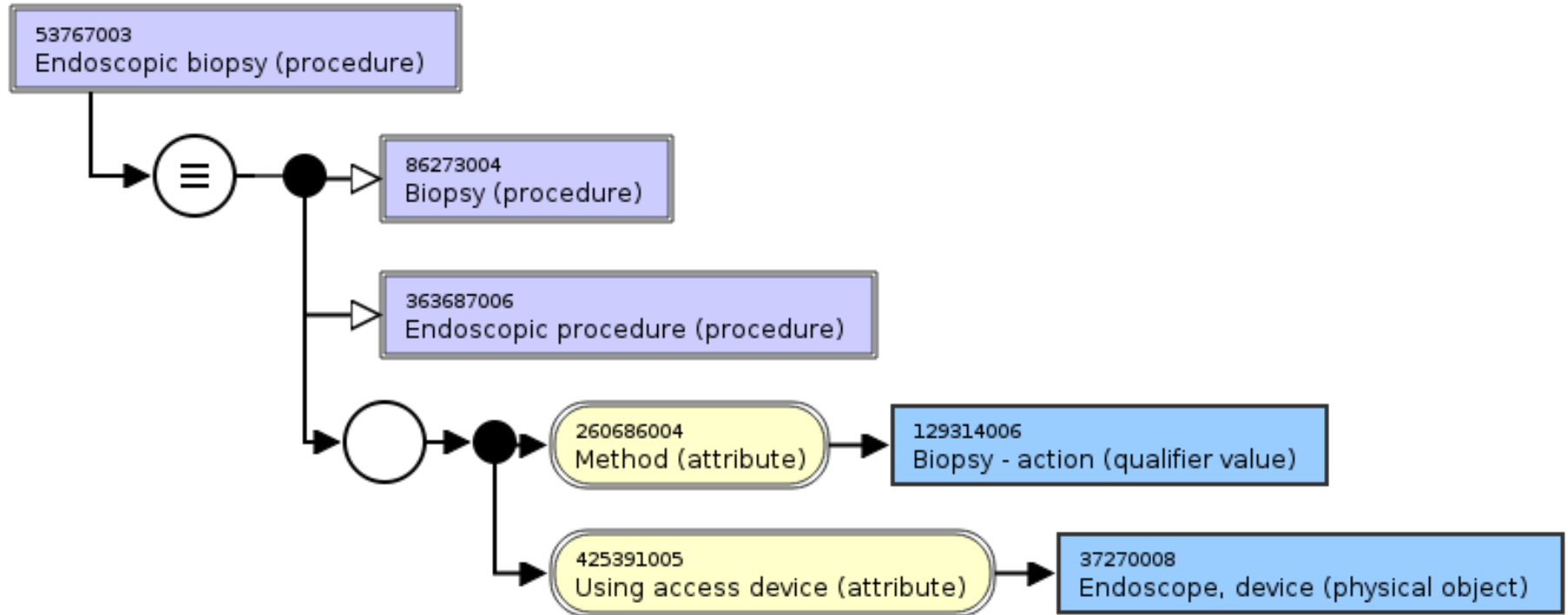
Procedure

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
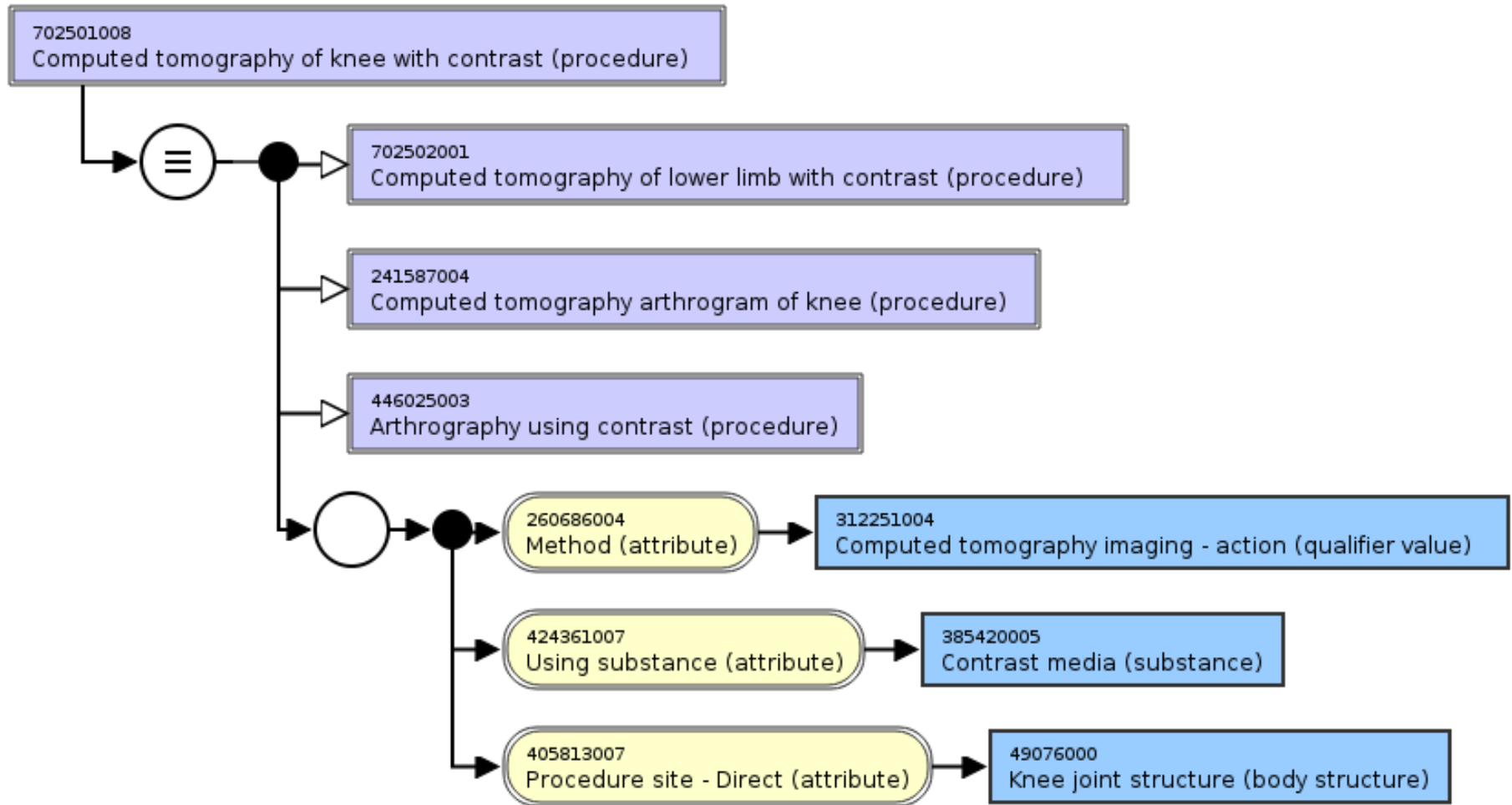
Situation with explicit context

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
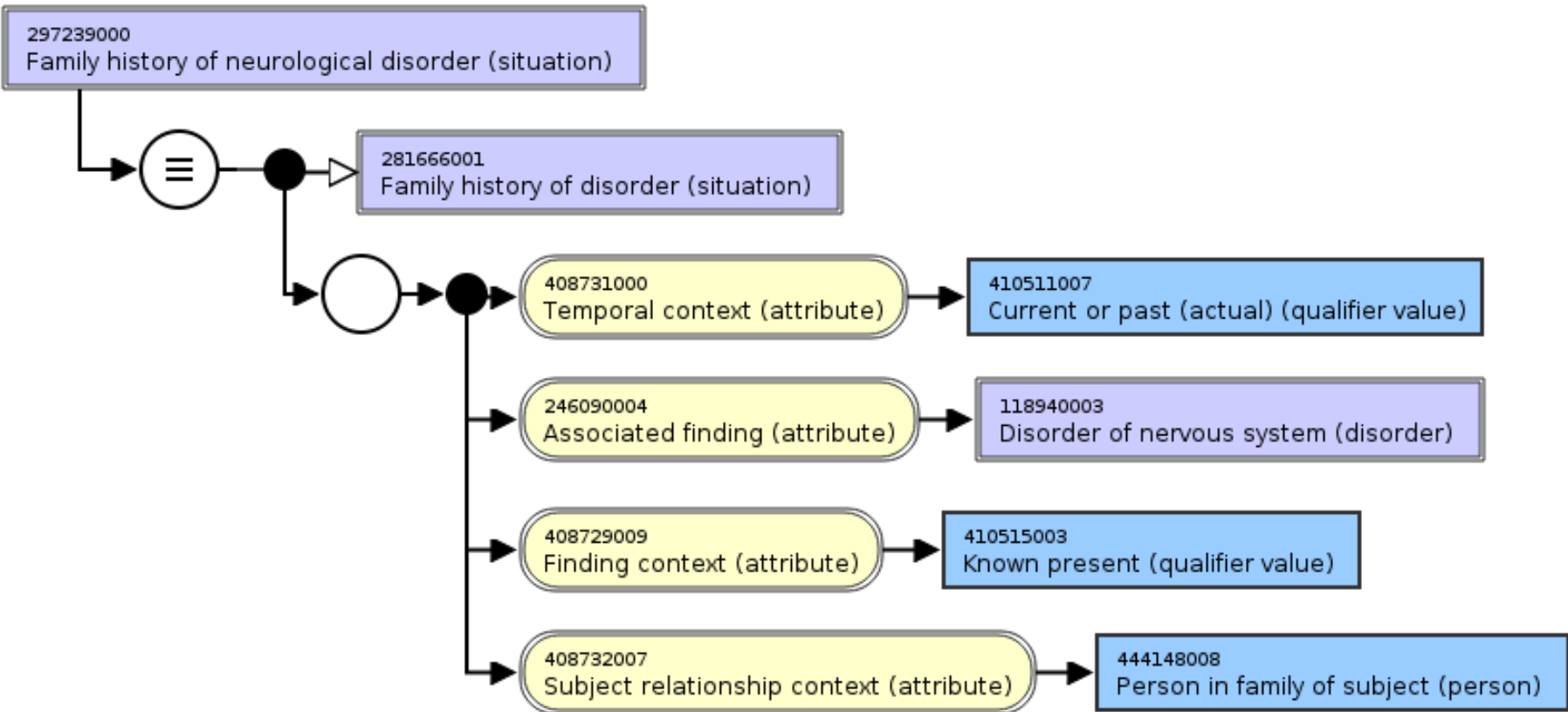
Temporal context values and examples

- 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)
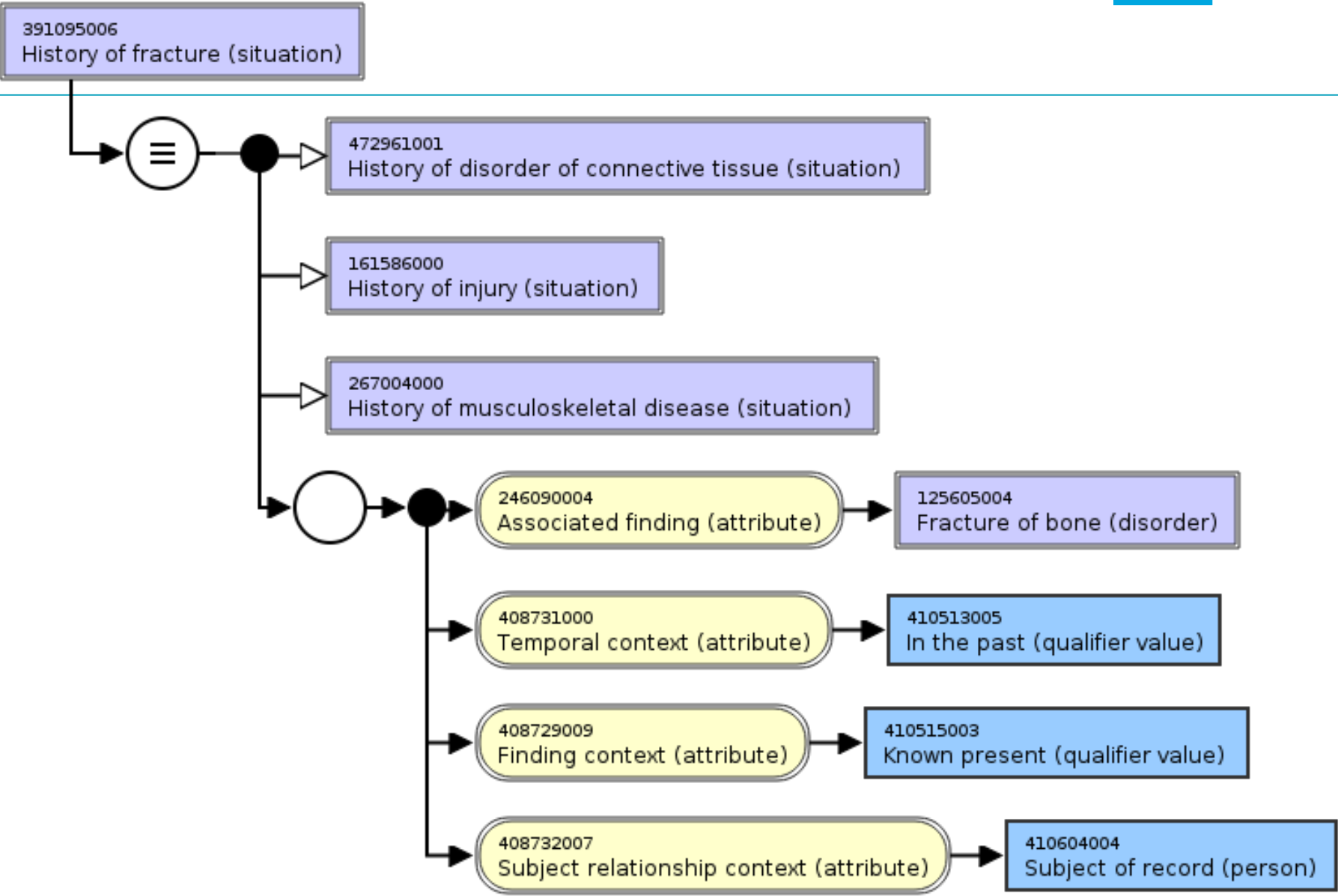
Finding context values and examples

- 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)
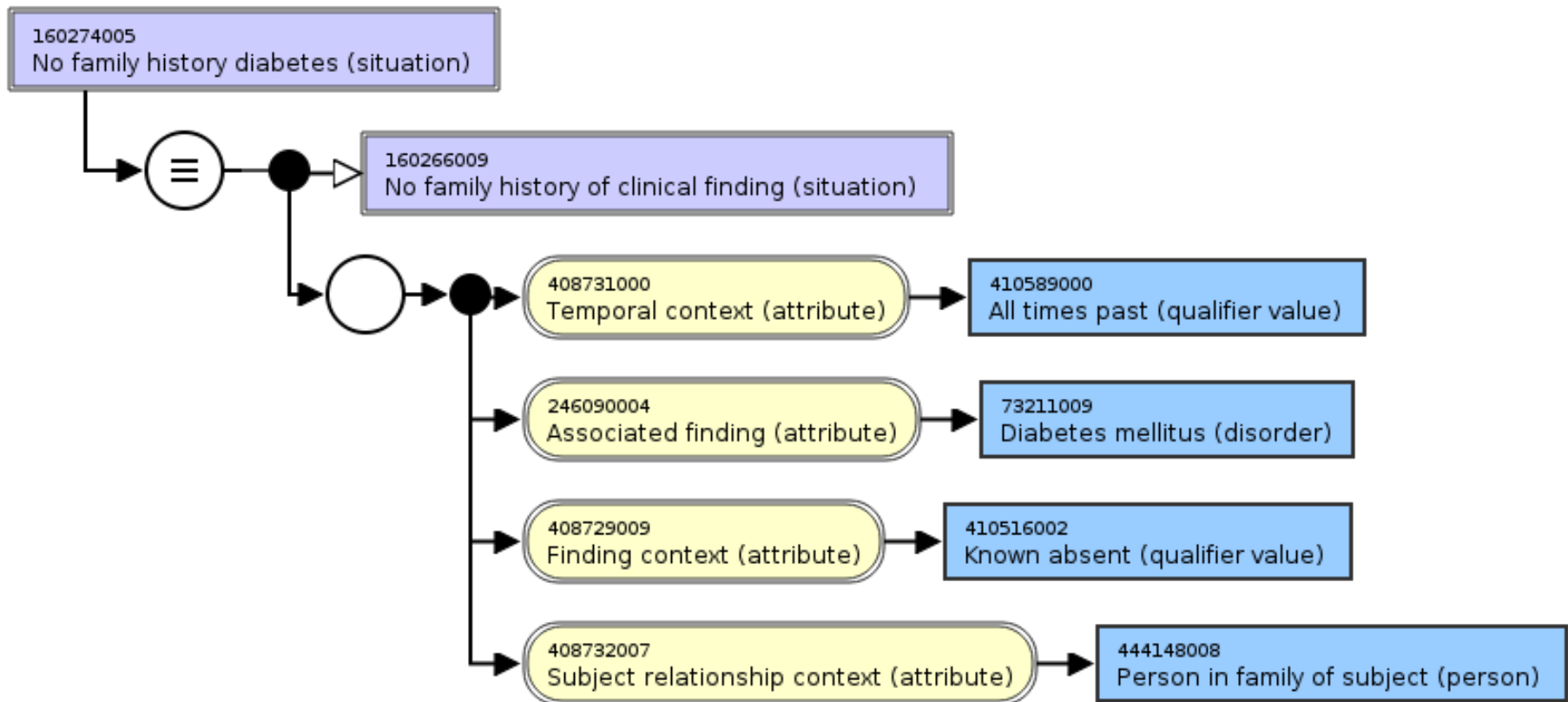   - No family history diabetes (situation)

Suspected clinical finding
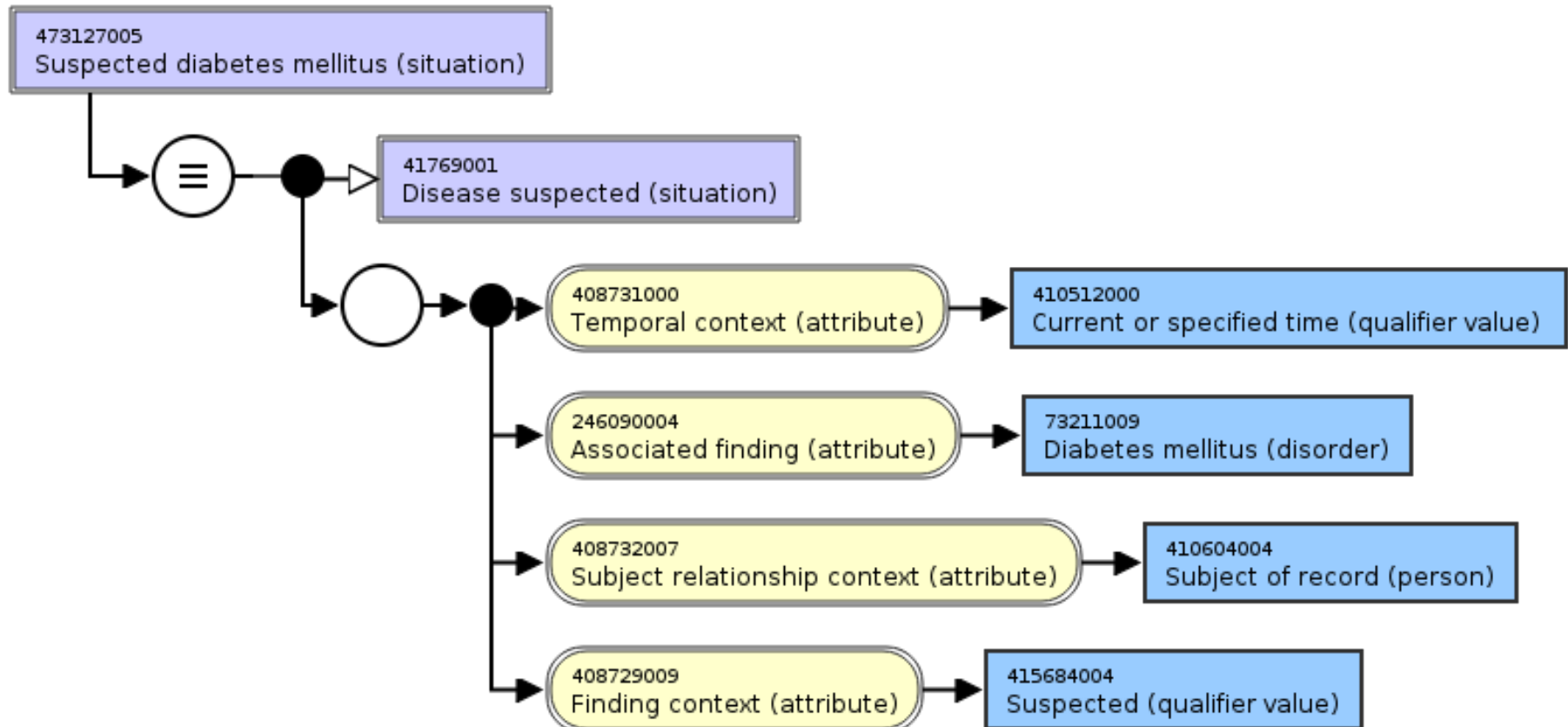
Clinical finding not suspected
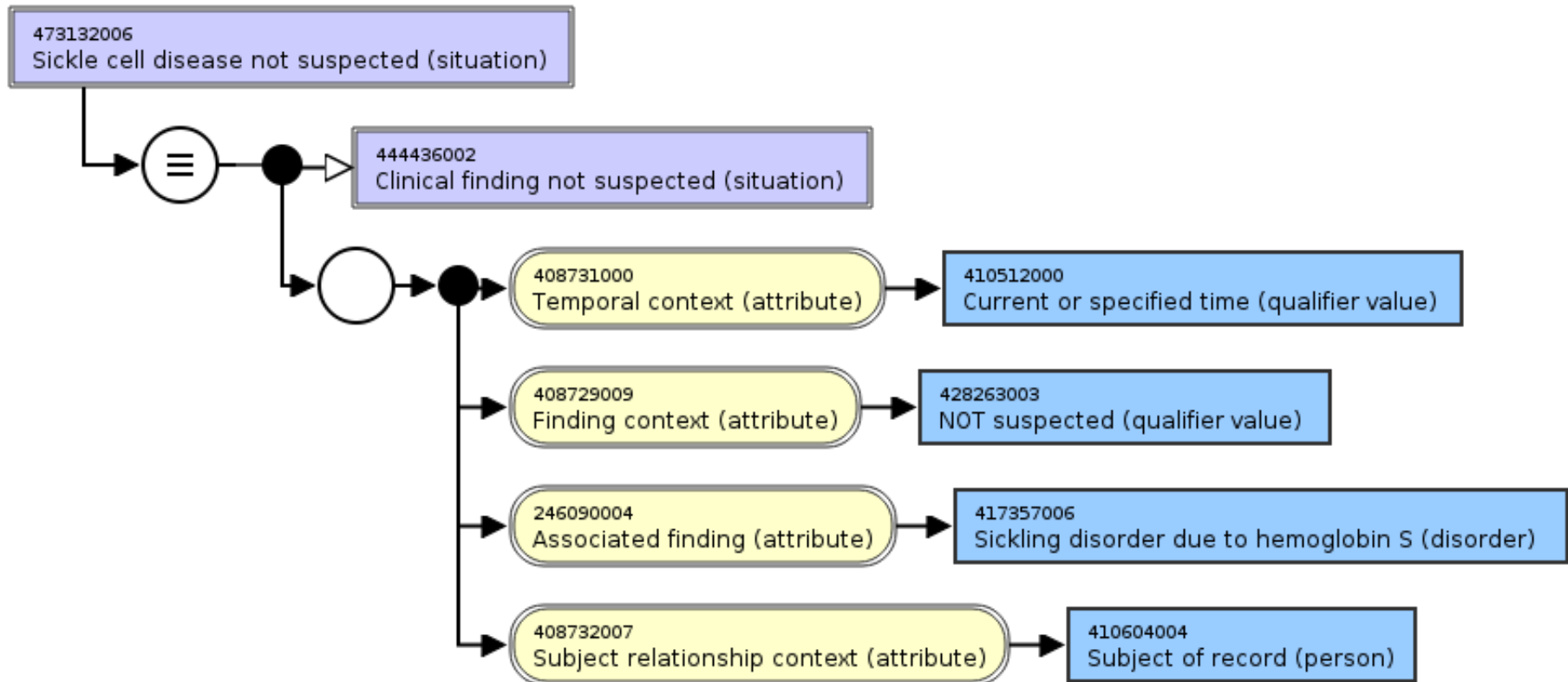
Finding absent

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)
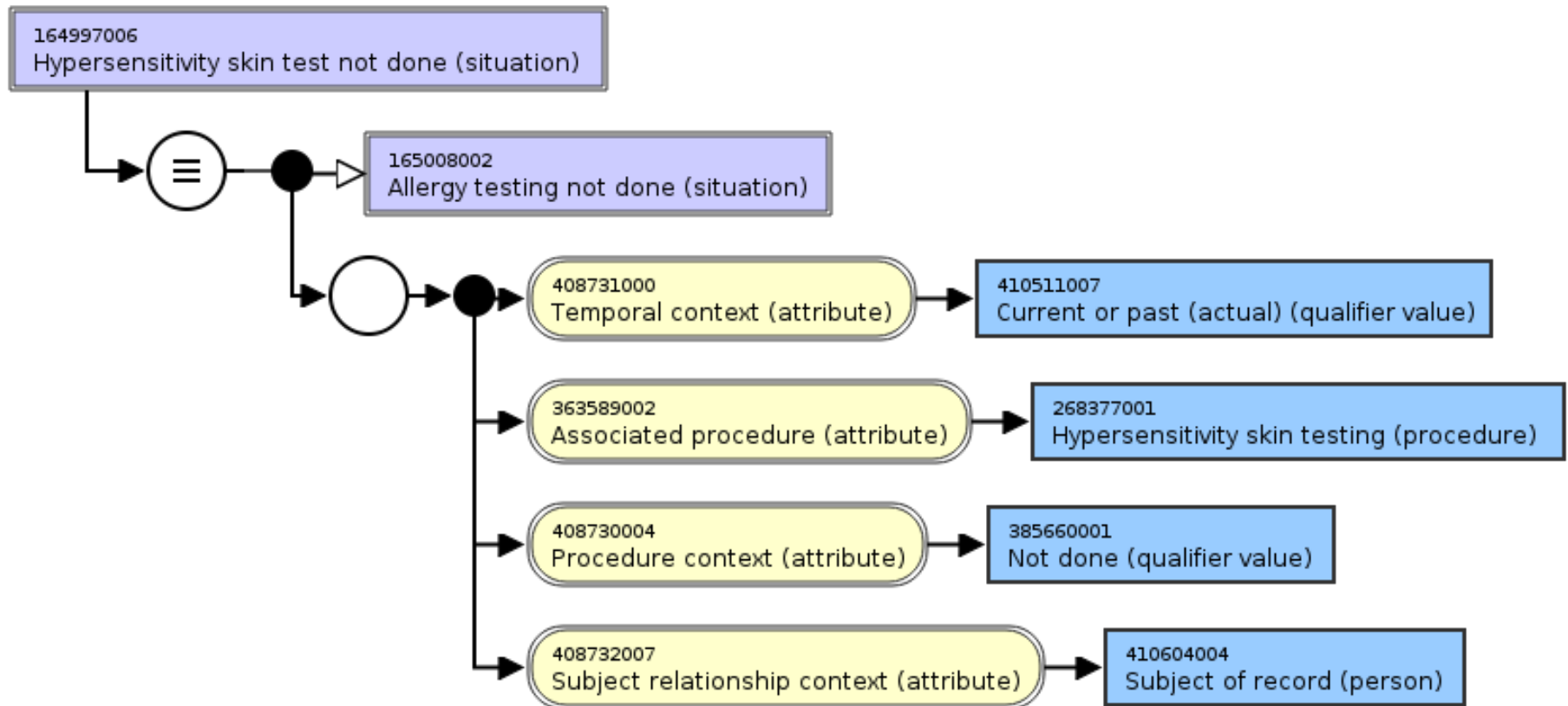 - Planned (qualifier value)
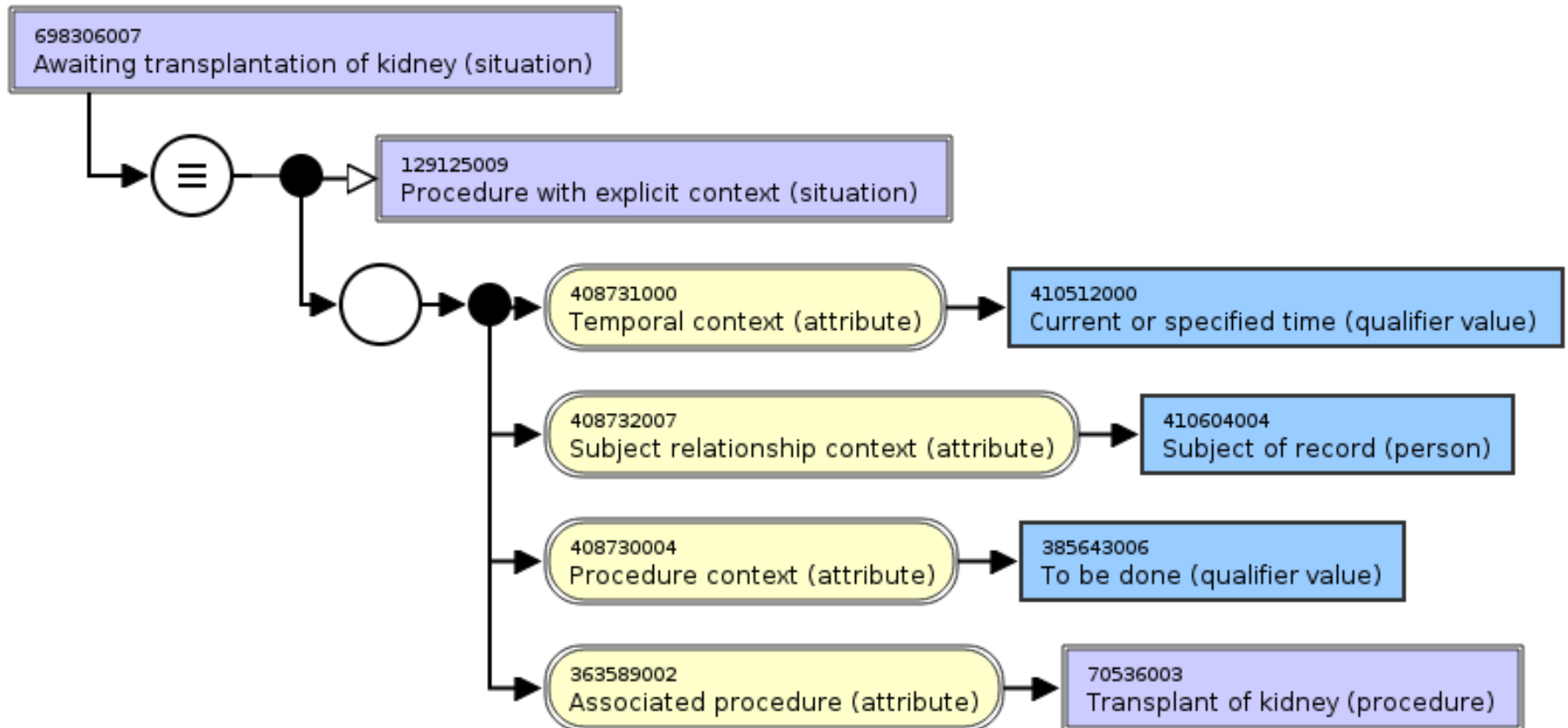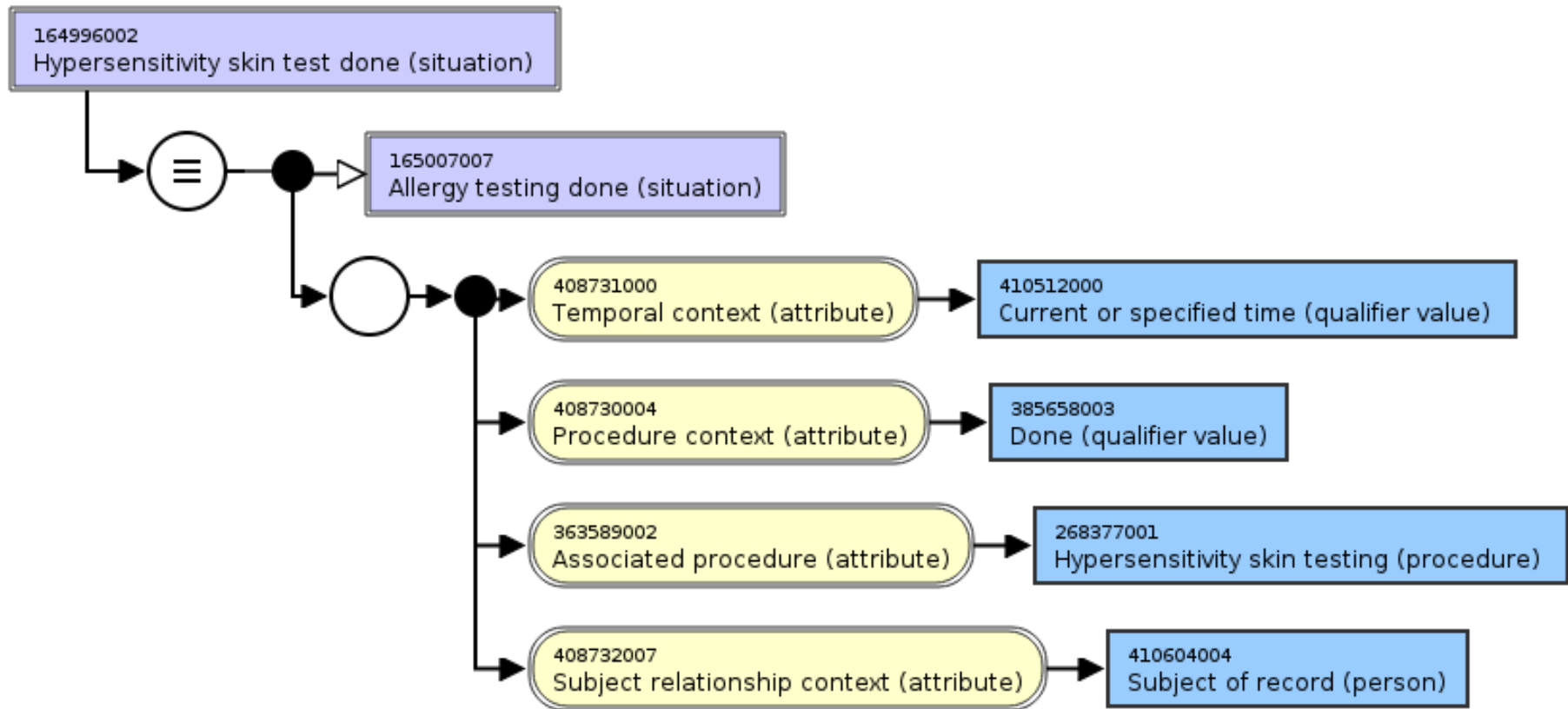
Procedure contraindicated
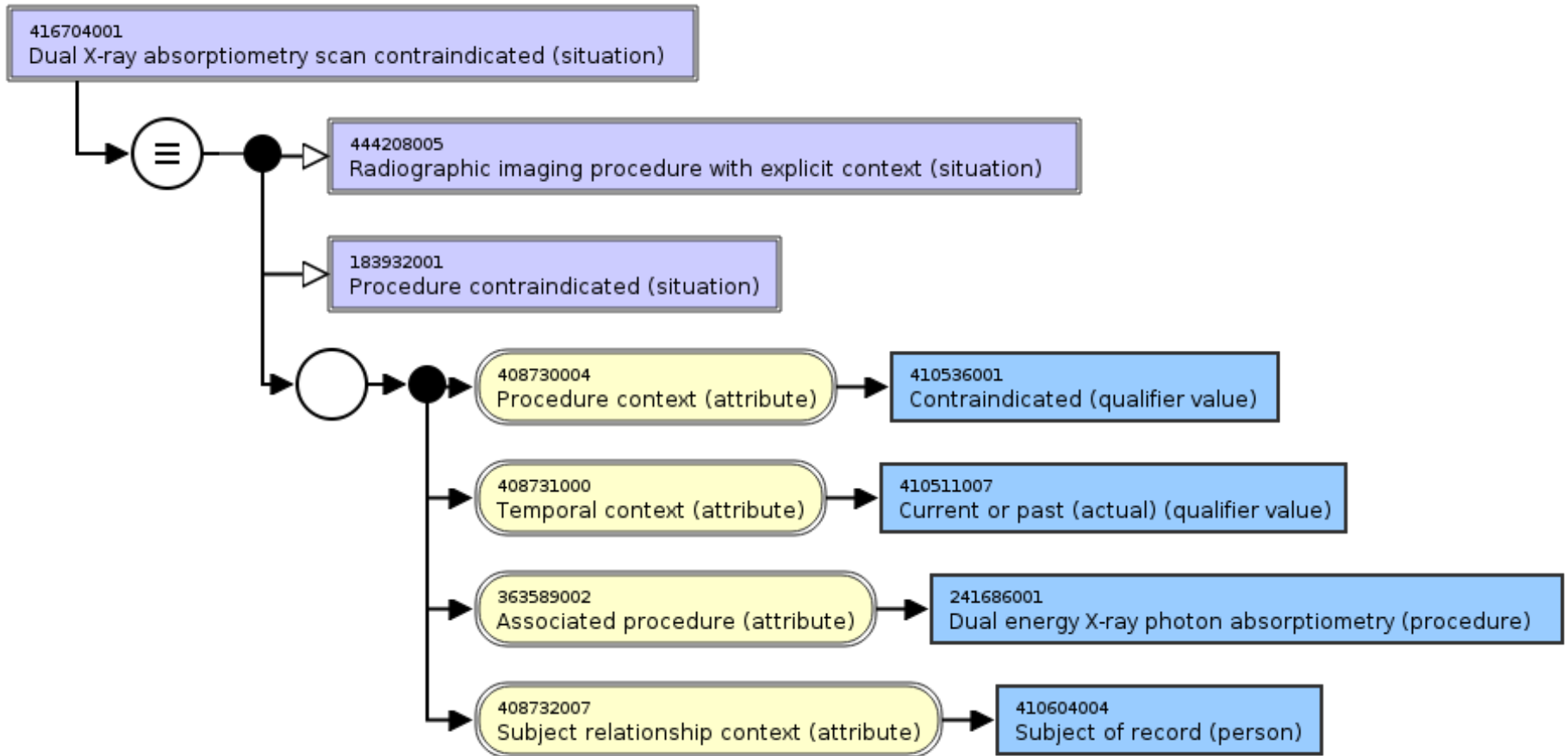
Procedure declined
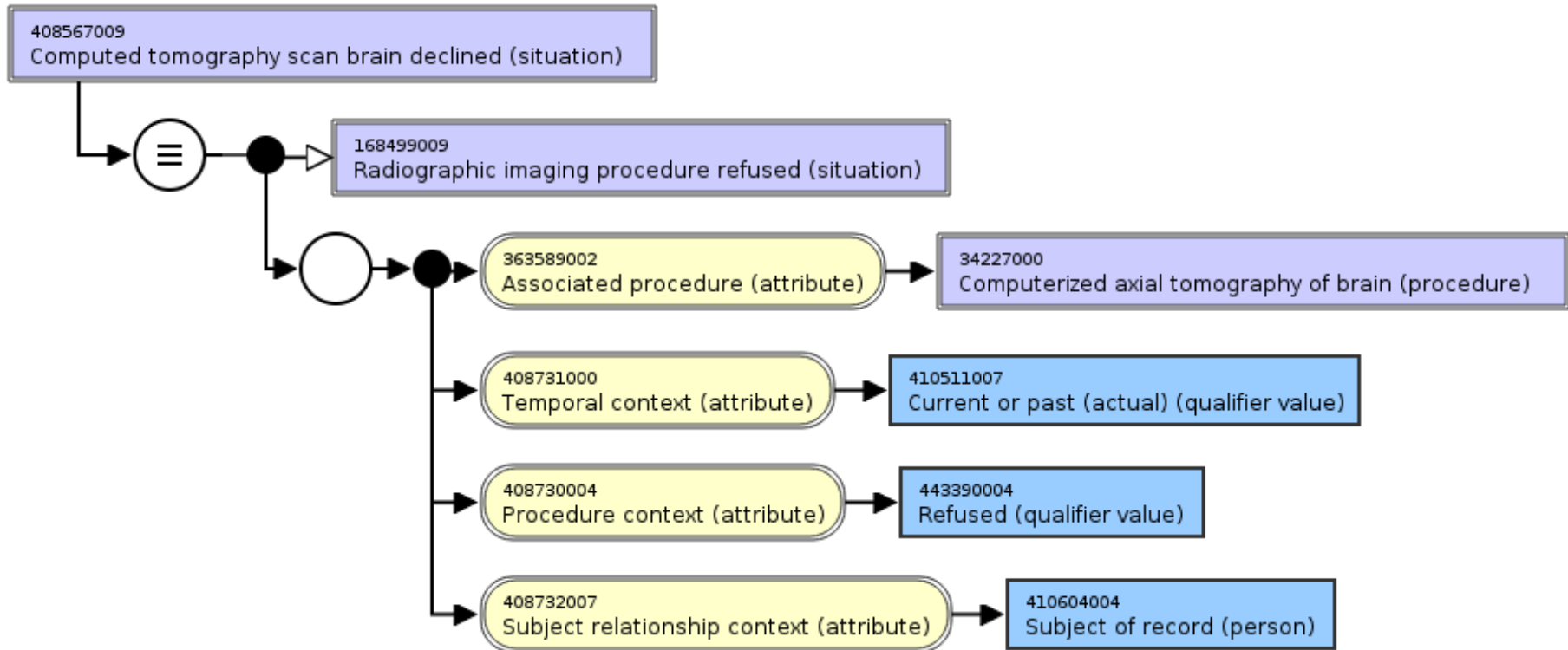
Procedure not done

Procedure to be done

Procedure done

QUESTIONS & DISCUSSION

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