SNOMED CT and Basic Formal Ontology - Convergence or Contradiction between Standards?

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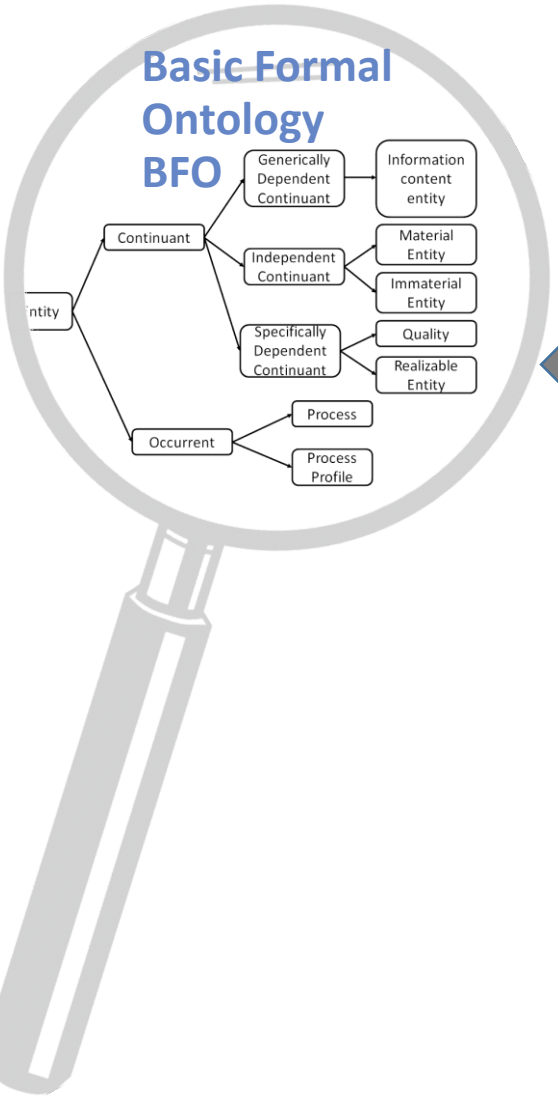
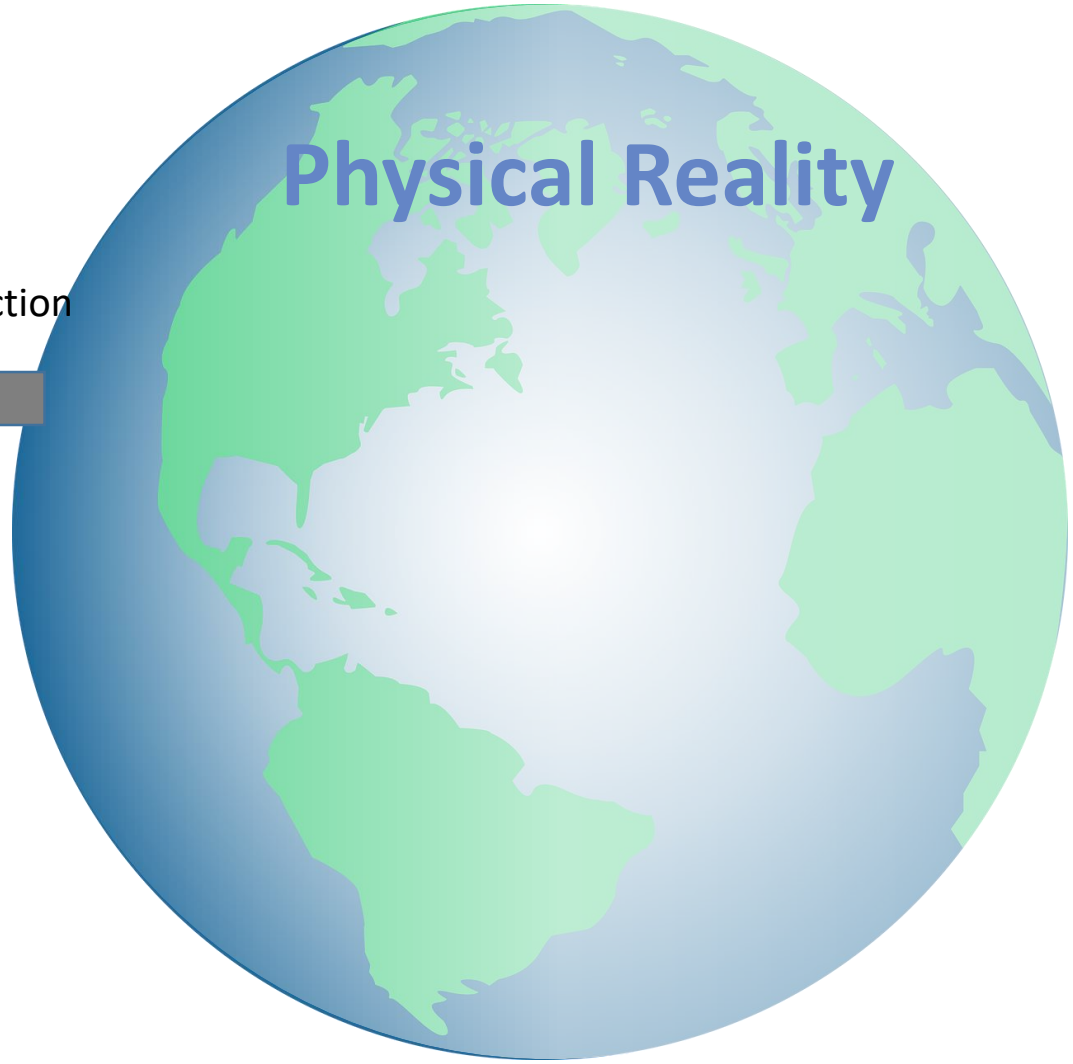
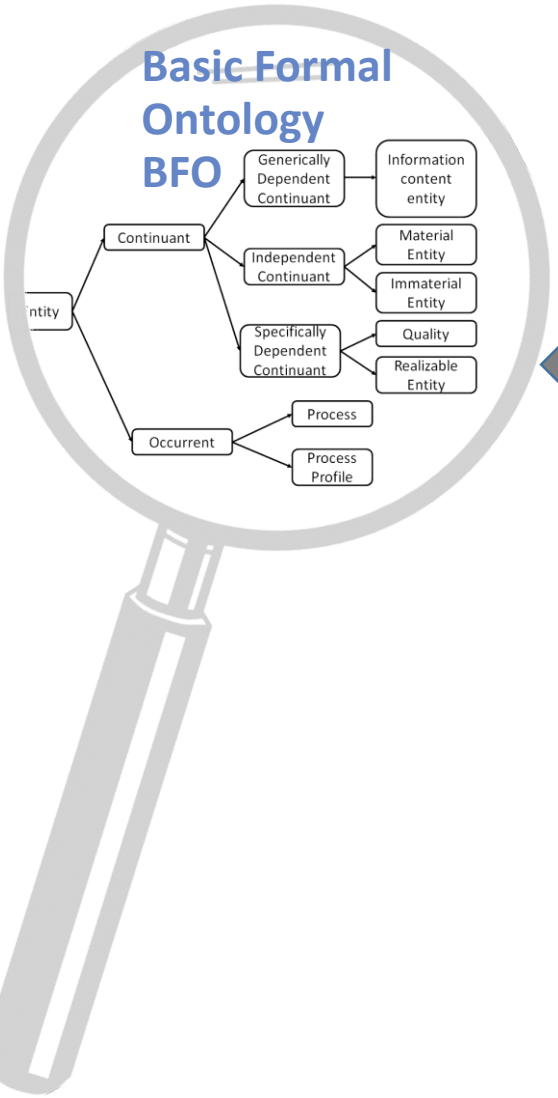
Modelling Advisory Group, April 19th, 2021


Physical Reality



abstraction





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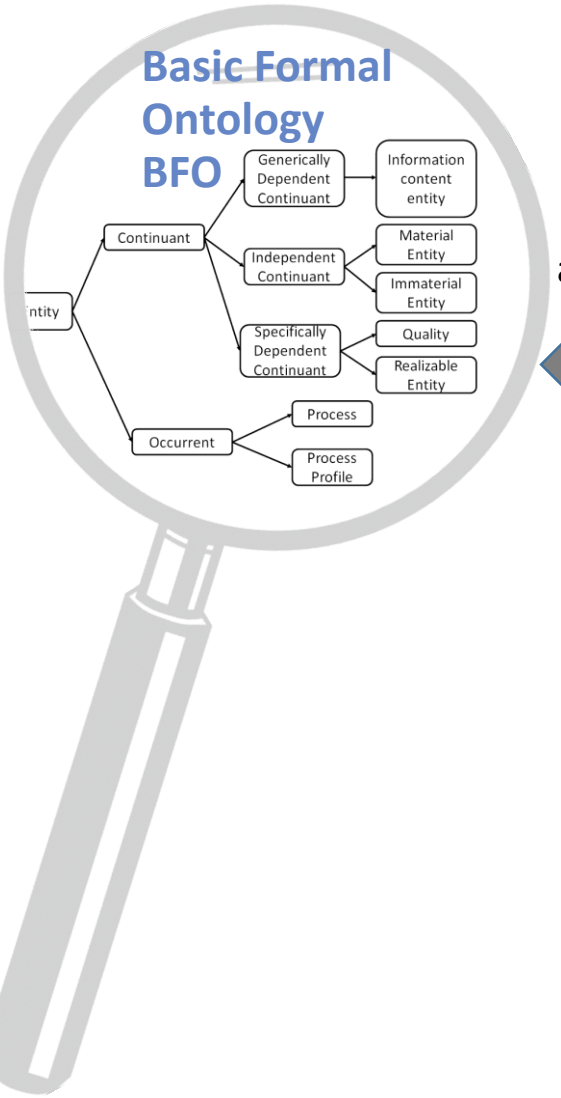


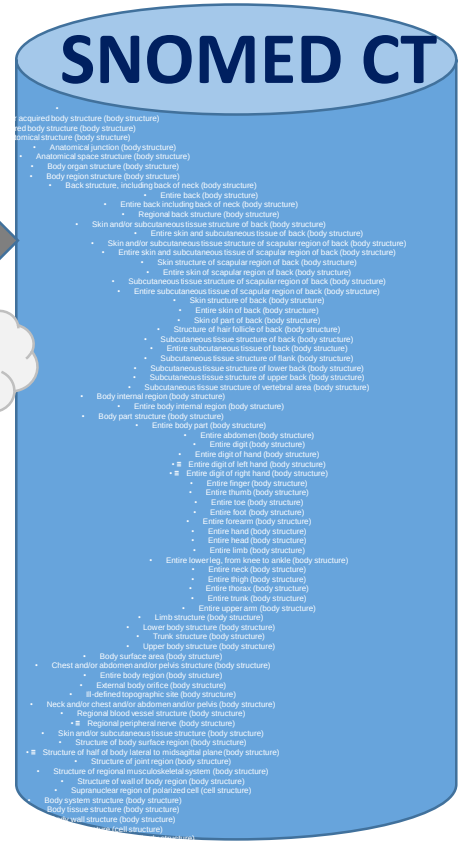
abstraction

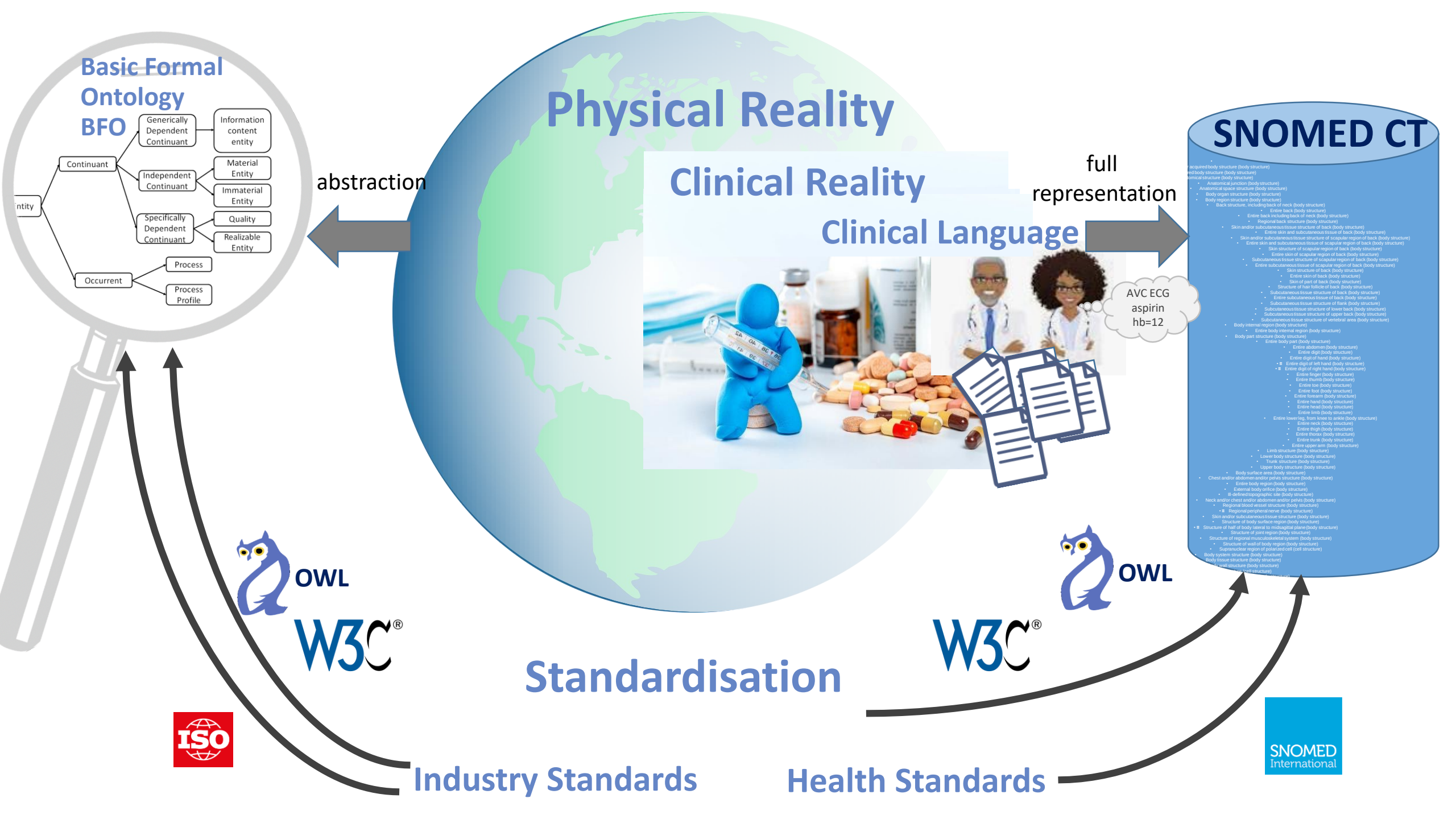
Physical Reality

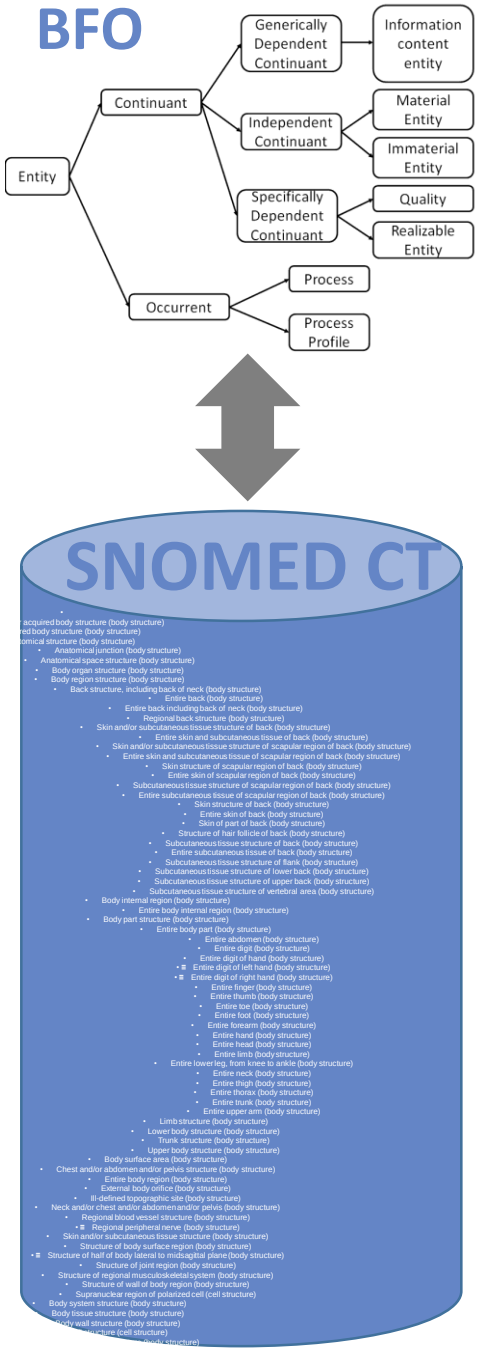
Clinical Reality

Clinical Language

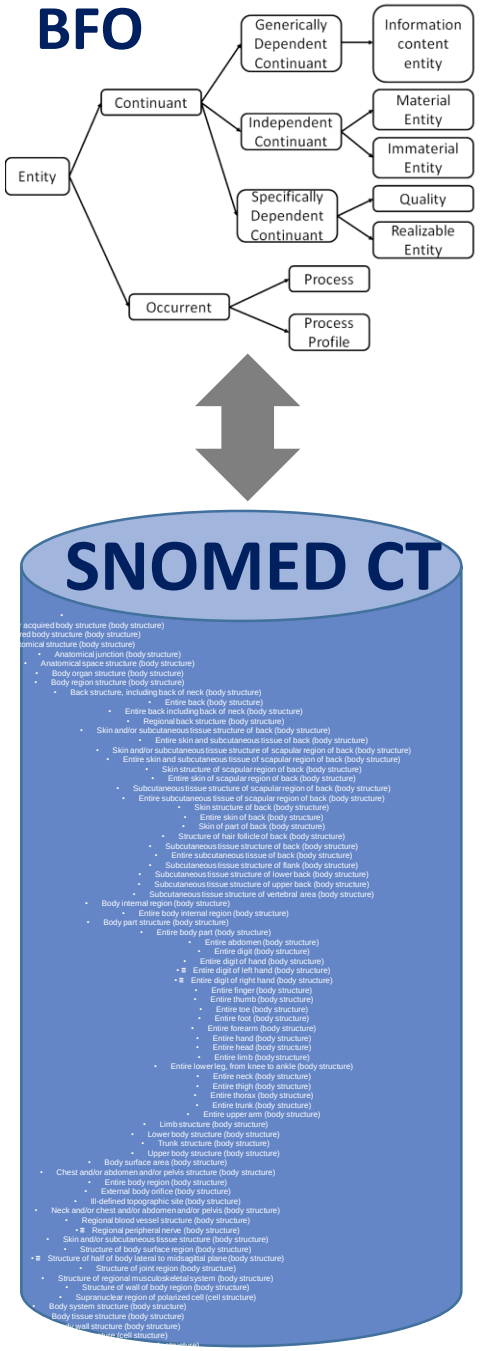
full representation




Basic Formal Ontology BFO
Generically Dependent Continuant
Information content entity
Continuant
Material Entity
Independent Continuant
Immaterial Entity
Specifically Dependent Continuant
Quality
Realizable Entity
Occurrent
Process
Process Profile
abstraction
Physical Reality
Clinical Reality
Clinical Language
full representation
AVC ECG
aspirin
hb=12
SNOMED CT
OWL
W3C®
Standardisation
ISO
Industry Standards
Health Standards
SNOMED International



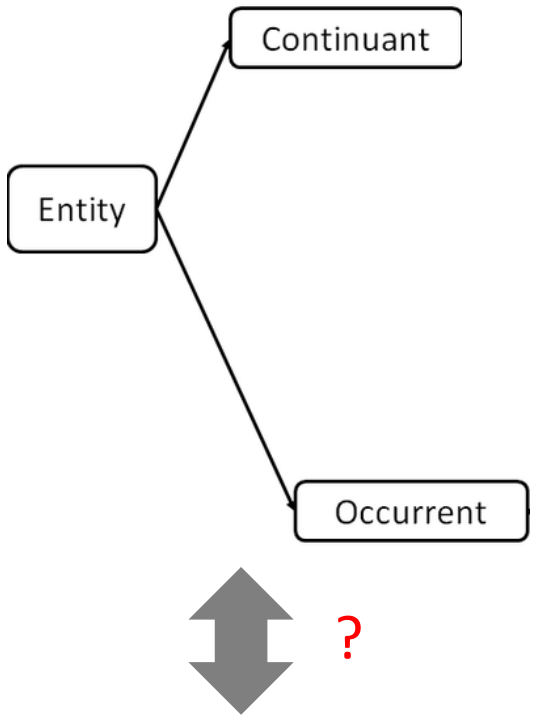
	SNOMED CT	BFO
Type	Domain ontology (+ terminology)	Foundational (upper-level) ontology
Scope	EHR relevant entities, "bottom-up"	Most general “categories of being”, relevant for natural science, "top-down"
Intended use	Standardise clinical terms and ground them ontologically	Provide foundational system of categories and axioms as upper level for domain ontologies
Intended users	Developer of clinical systems	Ontology and terminology developers
Size	Very huge (350 k concepts)	Very tiny (29 classes, 21 relations)
Top level divisions	Divisions rooted in legacy, following clinical and linguistic criteria	Uppermost node “entity” split into “continuant” and “occurrent”
Nodes represent	“Clinical ideas” (intensional meanings ?)	Universals, i.e. types of entities
Relations	Binary relations (“linkage concepts”),	Binary and ternary relations.
Formal representation	Description logics OWL EL	First order logic, approximated by Description logics OWL DL
Text definitions	For small part of concepts	For each class and relation
Hierarchies	Multiple hierarchies	Single hierarchy
Standardisation	Established health standard (IHTSDO → SNOMED Intl.)	Under development ISO/IEC PRF 21838-2.2 Information technology — Top-level ontologies (TLO) — Part 2: Basic Formal Ontology (BFO)



Integration SNOMED CT - BFO

- Rationale
  - Alignment between standards strengthens standardisation efforts
  - Foundational ontologies enforce consensus of meaning of domain ontology content
  - BFO is referred to as an upper level in many other ontologies used in biomedical sciences
- Feasibility
  - Find solution that is non-disruptive for SNOMED CT
    - Harmonisation between SNOMED CT high-level concepts and BFO classes
    - Harmonisation between SNOMED CT and BFO relations
    - Harmonisation between SNOMED CT concept model and BFO axioms
  - Starting with one particularly contentious area: findings / disorder
    - Create model
    - Align examples to the model
    - Discuss results and make decisions

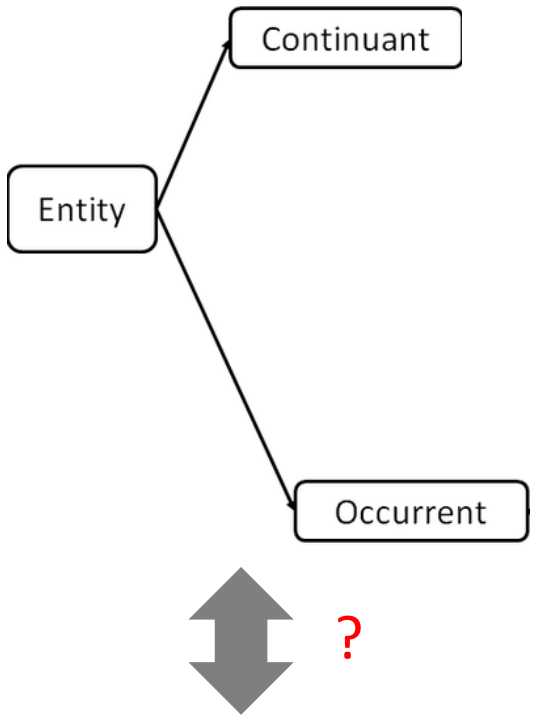
Findings / disorder hierarchy



- Problem 1
  - BFO splits the world into continuants and occurrents
    - continuants are static entities (which are already there in their entirety)
    - occurrents are dynamic entities (which have temporal parts / phases)
  - SNOMED CT does not commit to either continuants or occurrents
    - Argument: there are continuant and occurrent aspects of the same thing, e.g. tumours have a size (→ continuants) but also a growth process (→ occurrent).
    - According to BFO these are two different things, according to SNOMED, duplication of content would be laborious and of no practical values
    - Most foundational ontologies have similar distinctions
- Problem 2
  - BFO rejects the notion of "concept"
  - Many diseases are, in fact, constructs that are ill-defined and repeatedly re-defined. SNOMED CT must not blind out such content, because it is part of clinical discourse

Finding / disorder hierarchy : Problem 1

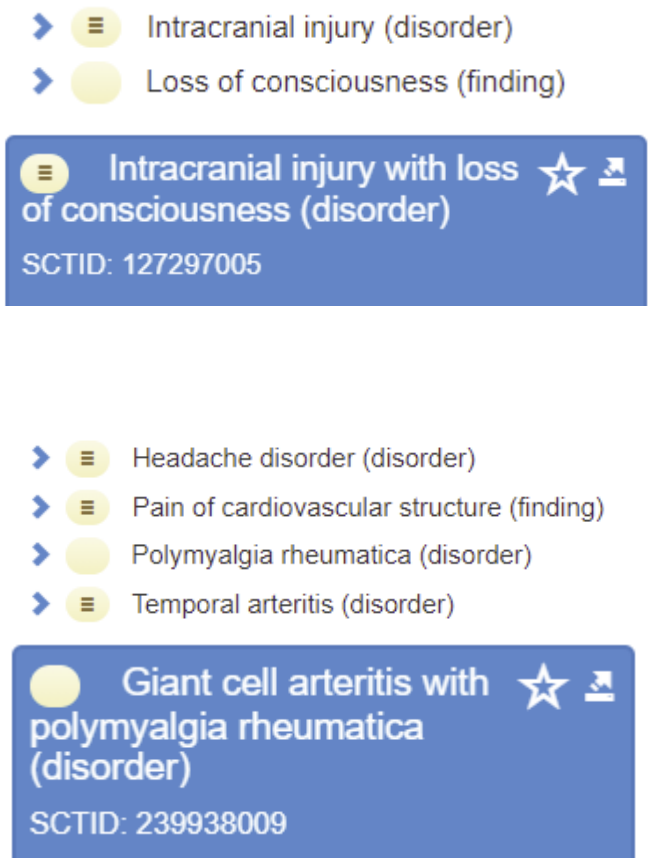
"BFO splits the world into continuants and occurrents"



Evidence why concepts under *Clinical finding* already have an implicit **occurrent** meaning:

1. Definitions relate to some other similar concepts:
   - via *associated morphology* to continuants
     e.g. *sarcoma (disorder)* to sarcoma *(morphological abnormality)*
   - This shows that there are separate concepts for the "continuant" aspect already
2. Single role grouping
   - encloses links to (1.)
   - endorses the "having something" semantics, e.g.:
     fracture of radius and ulna is-a *fracture of radius*
     fracture of radius and ulna is-a *fracture of ulna*
   - only plausible with the meaning:
     "having a combined radius/ulna fracture" is "having a radius fracture" and "having an ulna fracture" (otherwise "part of")

Finding / disorder hierarchy : Problem 2



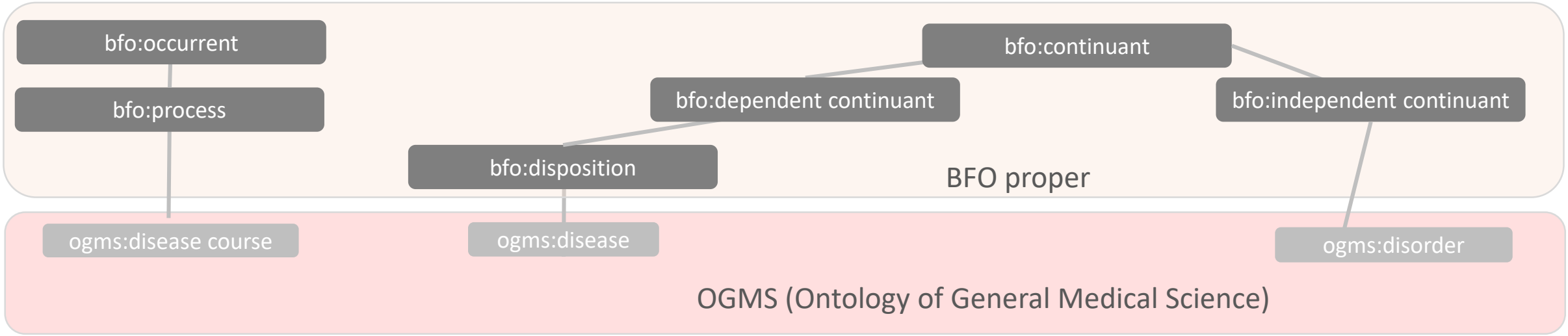
"Diseases are rather concepts than real objects"

Why the **occurrent** interpretation of finding / disorder concepts defuses the problem of representing ill-defined "construct-like" diseases like RA (rheumatoid arthritis)

1. It privileges the "having something" semantics
   - in the sense of "having RA" as "having a diseased named RA", does not require fully defining RA
2. Again, it supports expressing the co-occurring of different things by subclassing
   Examples (left): - injuries with functional disorders
   - signs, symptoms and diseases

Conclusion: only the interpretation of findings, sign, symptoms, diseases, injuries, material and behavioural phenomena as occurrents is compatible with the current hierarchy, which expresses combination as subclassing

Analysis of diseases / disorders / findings according to BFO



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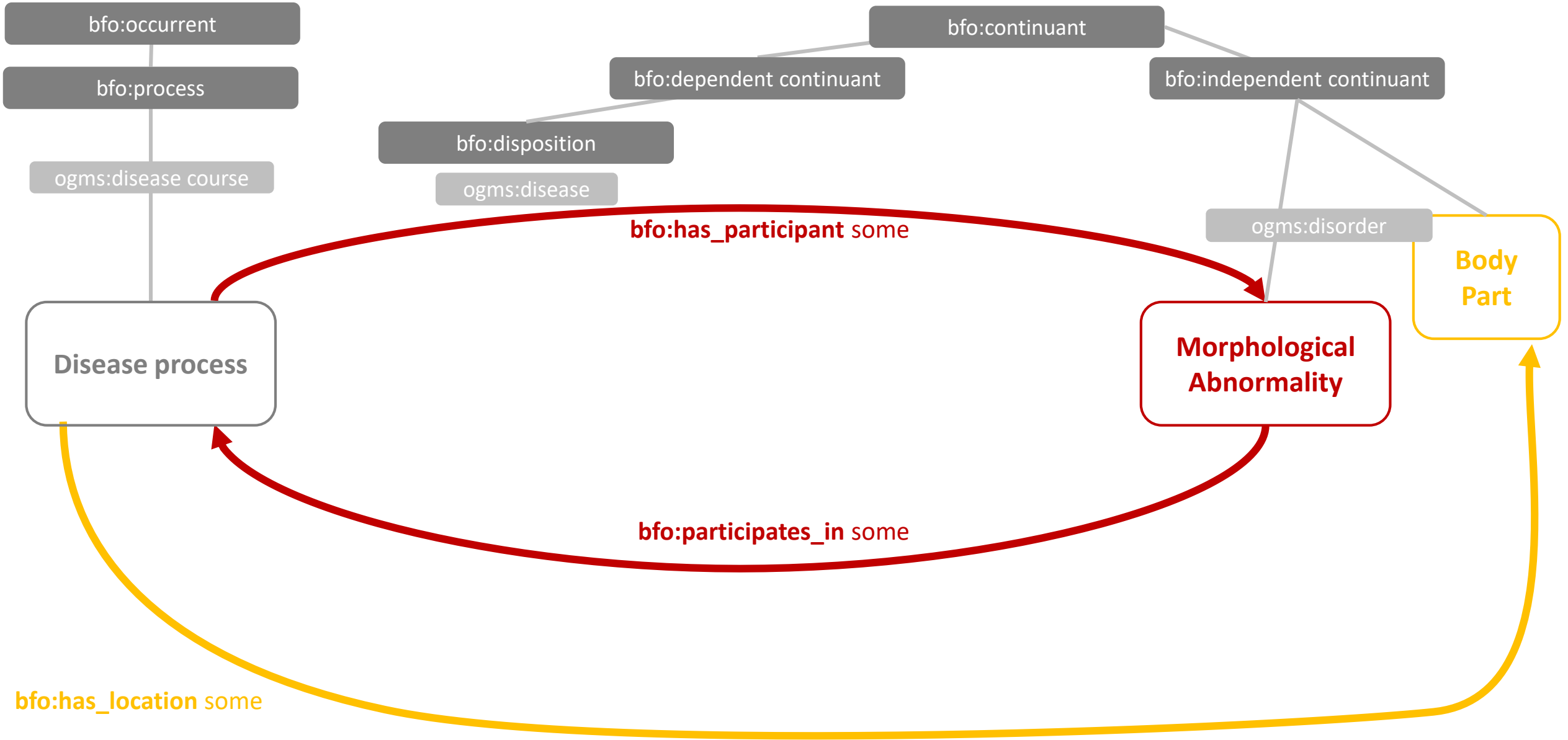

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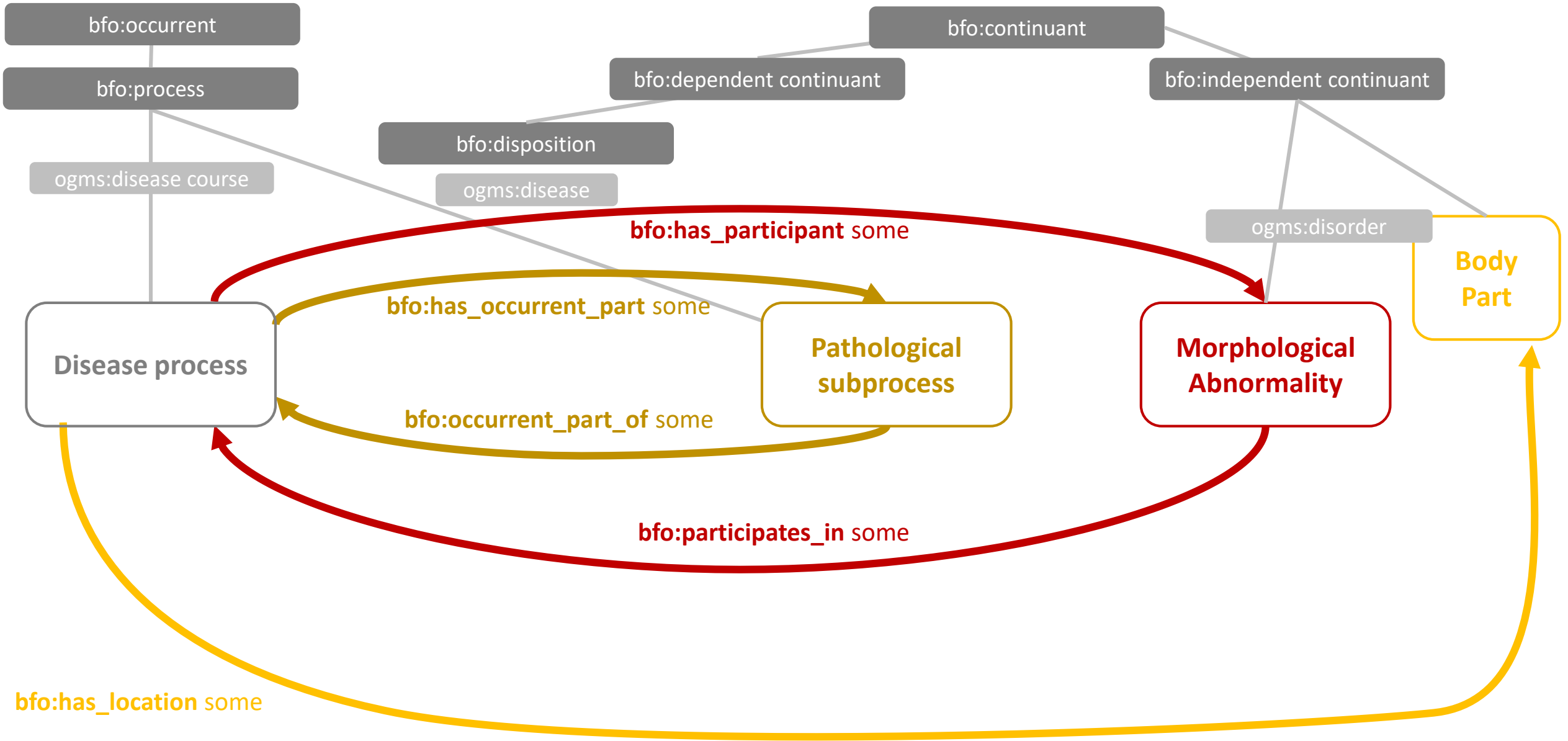

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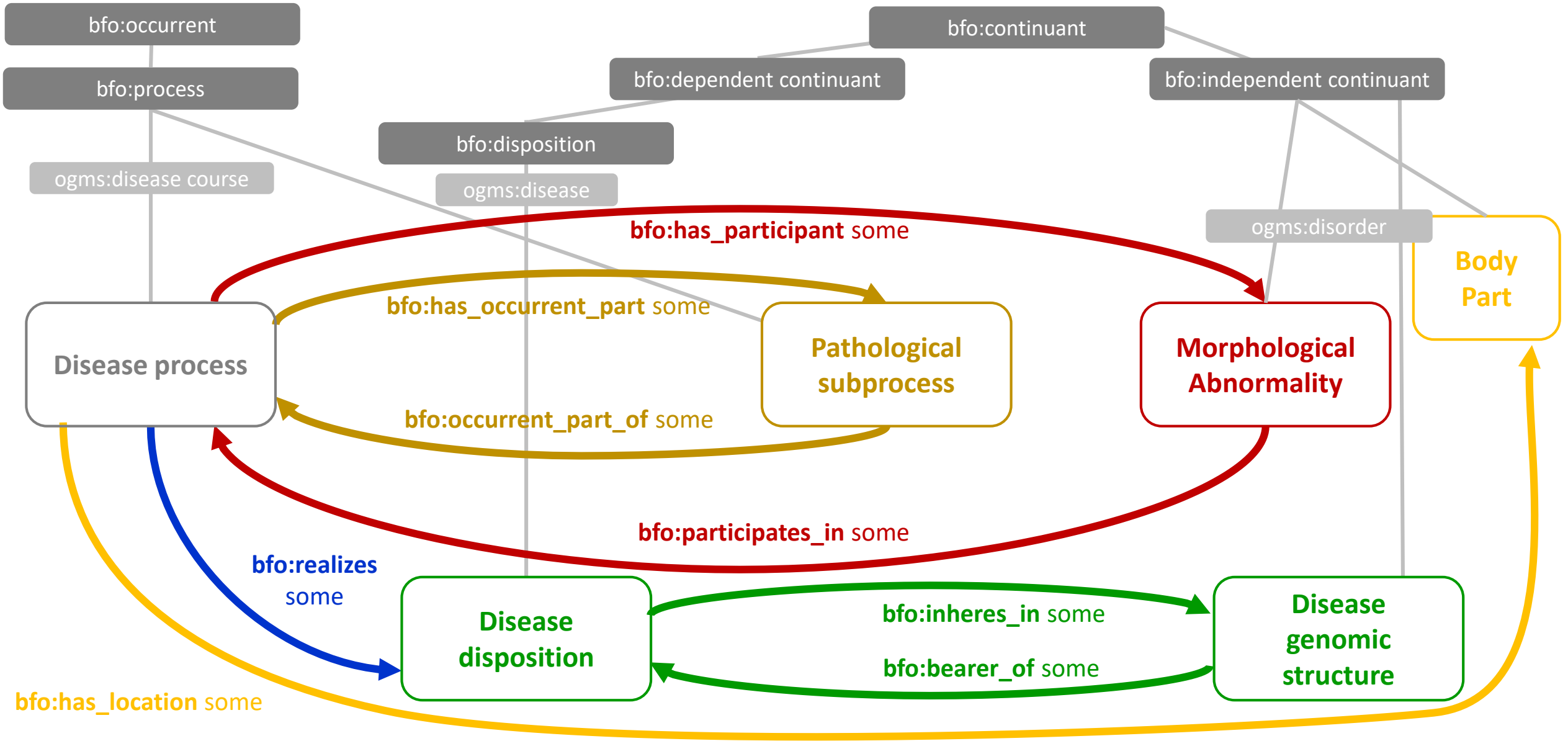
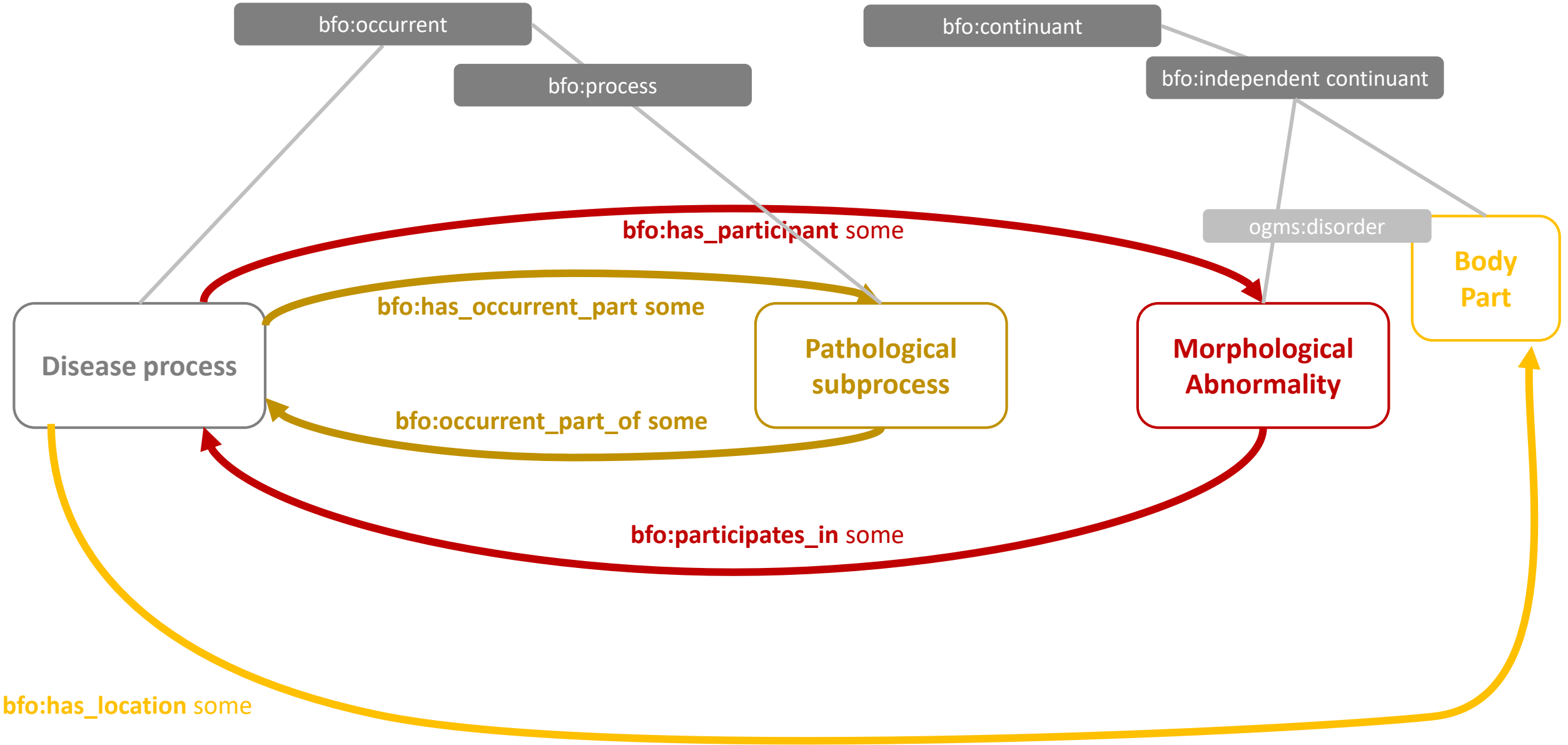


Analysis of disorders / findings harmonised with SNOMED CT



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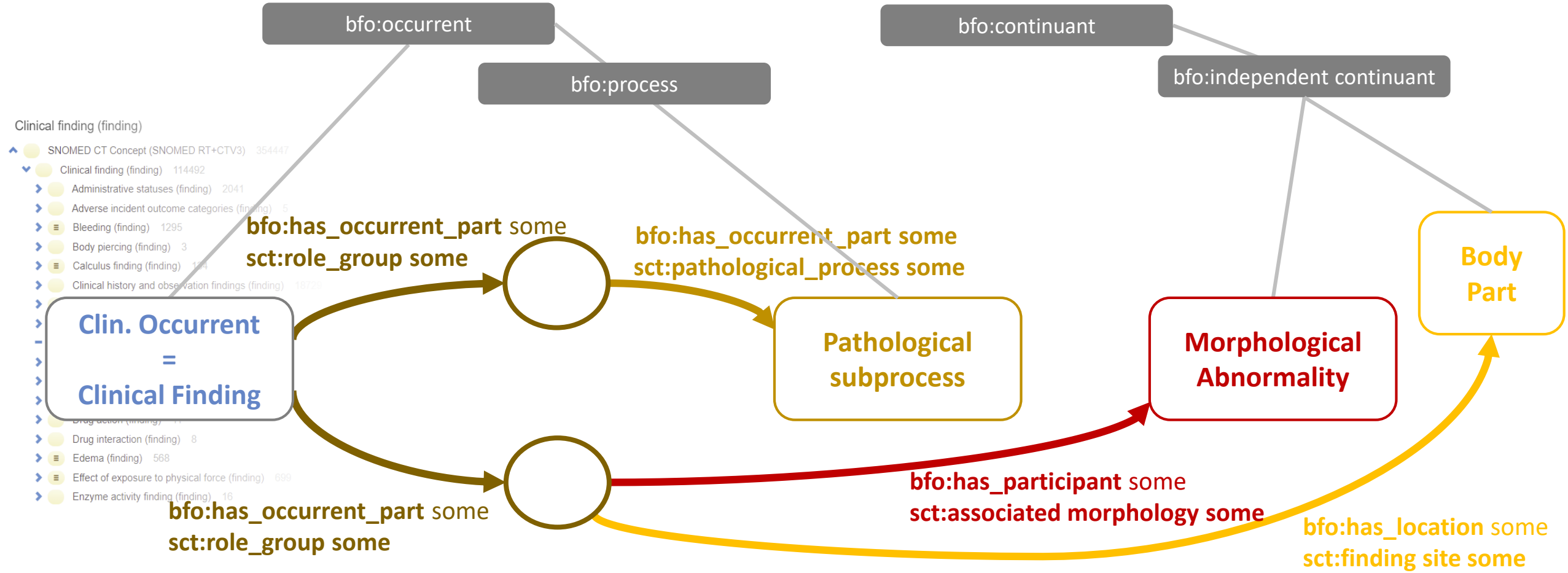
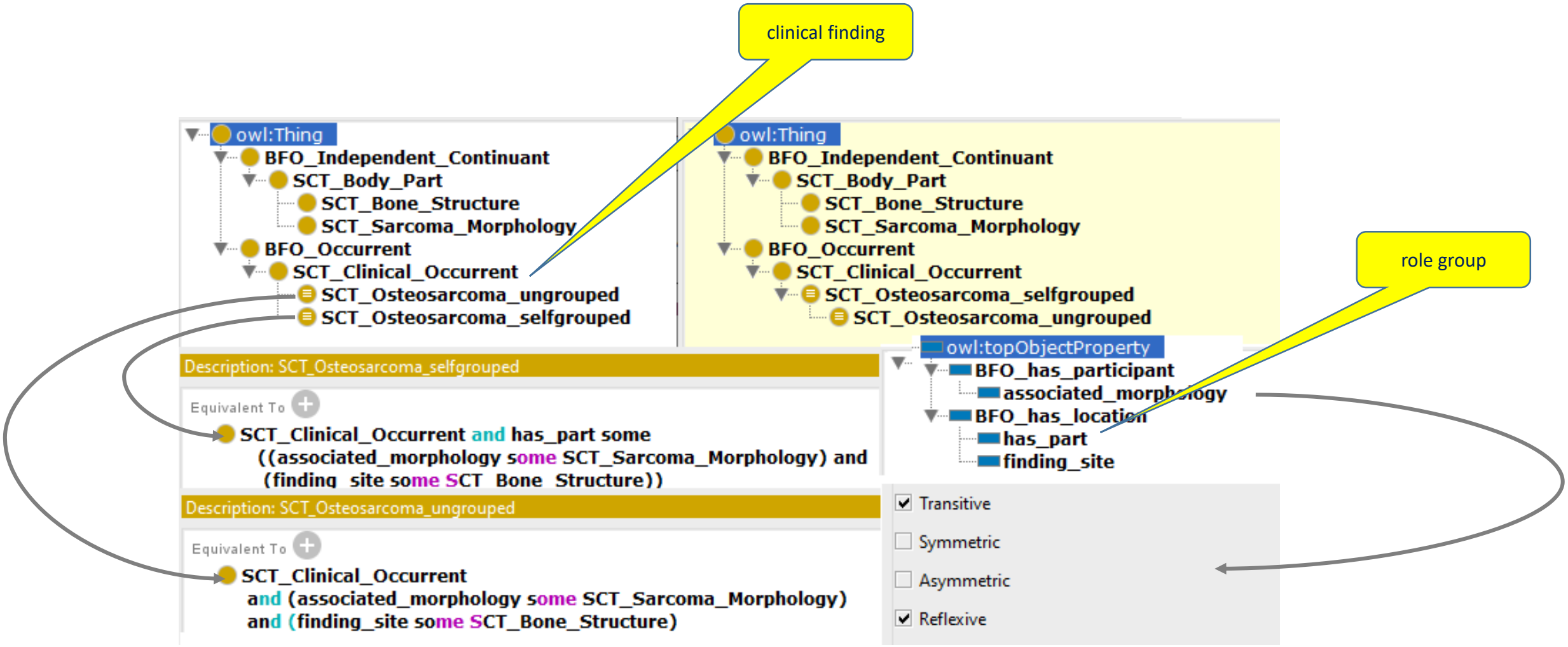
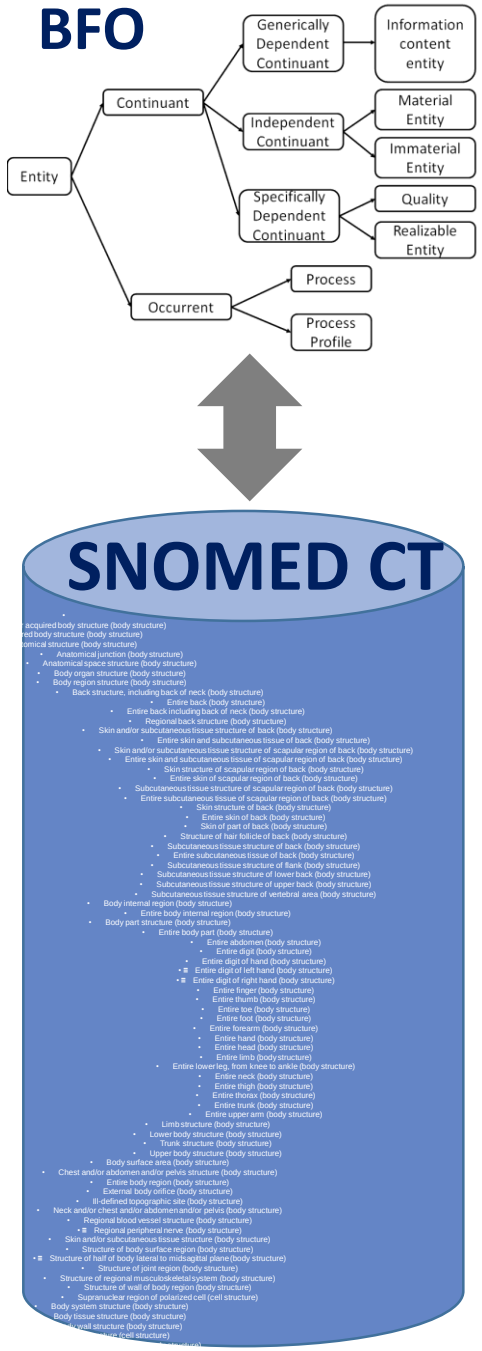


OWL model demonstrating re-interpretation and consistent reasoning





Intermediate conclusion – next steps

- Non-disruptive re-interpretation of SNOMED CT findings / disorders seem possible
  - *Clinical finding* as *Clinical occurrent*
  - *sct:role_group* aligned with transitive and reflexive BFO relation bfo:*has_occurrent_part*
  - *sct:associated morphology* under *bfo:has_participant*
  - *sct:pathological process* under *bfo:has_occurrent_part*
- Currently in progress
  - Elaborate on solution for clinical occurrents that relate to dispositions: less straightforward in terms of BFO but feasible
  - Collect frequent disease / finding terms and compare their representation in other ontology
  - Discuss proposed model in light of new insights
- Goal: finalise manuscript for Applied Ontology