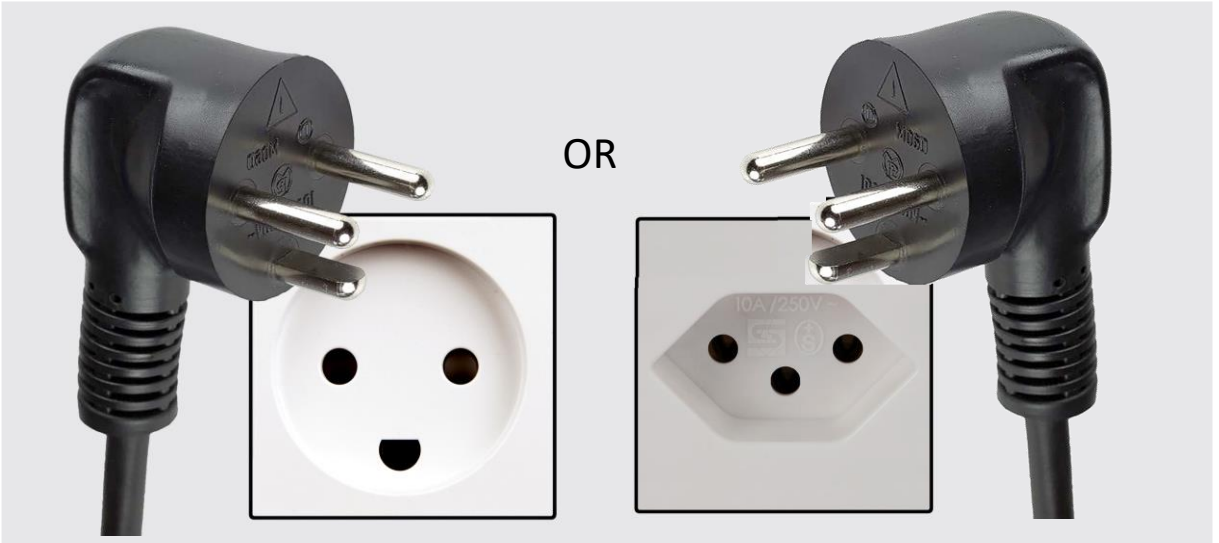
SNOMED CT and Basic Formal Ontology

Convergence or Contradiction?



SNOMED International Business Meetings
Modelling Advisory Group Oct 8, 2020

Stefan Schulz & Peter Hendler

Background

- ICBO 2019 conference Buffalo NY
- Discussing value of Cooperation
  - National Center for Ontological Research in the University at Buffalo (Barry Smith) – Interest in SNOMED CT as the largest biomedical ontology
  - SNOMED International (Jim Case) – Interest in academic cooperation and more principled content development obeying Applied Ontology principles
- Identification of four problem areas
- Further discussed and elaborated on behalf of the MAG by a subgroup led by Stefan Schulz
- Paper draft "SNOMED CT and Basic Formal Ontology - Convergence or Contradiction? "

SNOMED CT and Basic Formal Ontology - Convergence or Contradiction?

SNOMED International Modelling Advisory Group Draft

Stefan Schulz, Jim Case, Yongsheng Gao, Peter Hendler, Daniel Karlsson, Ronald Cornet, Robert Hausam, Harold Solbrig, Catalina Martínez-Costa…

Last revision: October 8th, 2020

1. Introduction

Ontologists categorise, describe and partially or fully define entities of a domain, using formal approaches rooted in a metaphysical context. Terminologists collect human language expressions and relate them to each other. Standards developers propose shareable descriptions and codes and provide them with definitional criteria that help decide whether a random entity does or does not belong to a given code / description. Computer scientists and health informaticians develop and implement software to manage ontology and terminology artefacts and use those artefacts to solve practical information processing problems.

BFO (Basic Formal Ontology) vs. SNOMED

	SNOMED CT (October 2020)	BFO Version 2
Scope	Clinically relevant entities in a broad range of abstractions	Most general “categories of being”
Focus	Clinical medicine, health care, social care, biomedical research	Physical reality, scientific research
Size	Very huge	Very tiny
Top level divisions	Flat, mostly disjoint top level concepts under “SNOMED CT concept”. However, things like (material entities, processes) can be found in several upper level hierarchies	Uppermost node “entity” split into “continuant” and “occurrent”. Everything in the world is either a continuant or an occurrent
Nodes represent	“Clinical ideas”, i.e. intensional meanings that extend to classes of potentially clinically relevant entities	Universals (which only exist in their instances), but which also extend to non-empty and very general classes
Relations	Binary relations (“linkage concepts ”),	Binary and ternary relations. The latter ones raise problems when creating an OWL version (require reification). Relations made their way into BFO only recently (Version 2); they are largely based on the OBO Relation Ontology
Formal representation	Description logics OWL EL	First order logic (Description logics representation not straightforward due to time-indexed relations), in its several OWL approximations use of OWL DL
Naming	Artificial, self-explaining labels (EN, ES, ...) called Fully Specified Names, real-world terms (quasi-synonyms) and numeric concept IDs	Artificial labels (EN, ES), no synonyms
Textual scope notes	Low coverage of textual definitions, underspecification of many primitive concepts due to lack of textual scope notes	Highly elaborated definitions and elucidations, refined in numerous iterations
References to external sources / standards	Standards, clinical literature for the curation of terminology content, e.g, Gray's anatomy, TA, FMA, and others for the body structure. Other examples, such as classifications for many clinical conditions, e.g. fracture, ulcers, etc.	Four-category ontology defended by E. J. Lowe (2006)
Hierarchies	Multiple (is-a, interpreted as OWL subclasses). Top-level categories, classes directly under SNOMED CT root node, are considered as disjoint classes (except 'physical object' and 'pharmaceutical / biologic product'). The rest are multiple but still following the disjointness from the top-level categories.	Single hierarchies. All divisions and subdivisions are strictly disjoint
Equivalence axioms	32 % (varying from ~100 % to 0 % by top hierarchy, see Appendix 1)	0%

Four problem areas

- "SNOMED should be about *things* in the real world and not about *concepts*"

  Solved: it is about both

- "SNOMED should have most of its terms fully defined"

  Solved: all ontologies have undefined primitives

- "SNOMED should have single inheritance"

  Solved: the stated version has single inheritance

- "SNOMED should be based on BFO and there should be Continuants and Occurrents and they should be mutually exclusive"

  Unsolved: the continuant – occurrent bipartition vs. current state of Findings hierarchy

Based on BFO with continuants and occurrents

- Top bipartition :
  - Continuants: always exist in their entirety as long they exist. Example: a person, a device, a quality (my body mass), a disposition (e.g. propensity to stroke)
  - Occurrents: have temporal parts Example: a year (spring, summer, fall, winter), a heart transplant, a person's life

… have a place, and material ones a volume and a mass

… have temporal parts and a duration

- Nothing can be both continuant and occurrent

BFO about diseases

- Ontology of General Medical Science (OGMS under BFO):
  - ogms:disorder: material continuants (a wound, a gene defect)
  - ogms:disease: dispositions (stroke risk, an allergic disposition)
  - ogms:disease course: occurrents (a stroke, an asthma attack)
- Main criticism
  - Redefinition of terms, not considering use of language
  - More than one entity for the same thing, e.g.
    tumor as an occurrent (growth process), tumor as a mass (material continuant)
  - Or are many things just both, continuants and occurrents?
    (…in four-dimensional ontologies, but not in BFO)

SNOMED CT finding hierarchy

- Boundary issues
  - between diseases and findings (hierarchy)
  - No clear boundary between disease and disorder (wording)
- Ontological bipartition:
  - morphological abnormalities as material (?) continuants
  - “other things” in Finding / Disorder hierarchies
- Findings hierarchy

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  - “other things” in Finding / Disorder hierarchies
- Findings hierarchy could be accepted as syncretistic
  - Because there are dispositions subsuming processes etc.
  - But then it would never fit under any formal ontology
- Findings hierarchy could be remodelled
  - High cost and unclear benefit
- Findings hierarchy could be re-interpreted
  - No disruption if interpreted as “clinical processes or states”

Solution to be proposed

Clinical findings / disorders as clinical processes or states

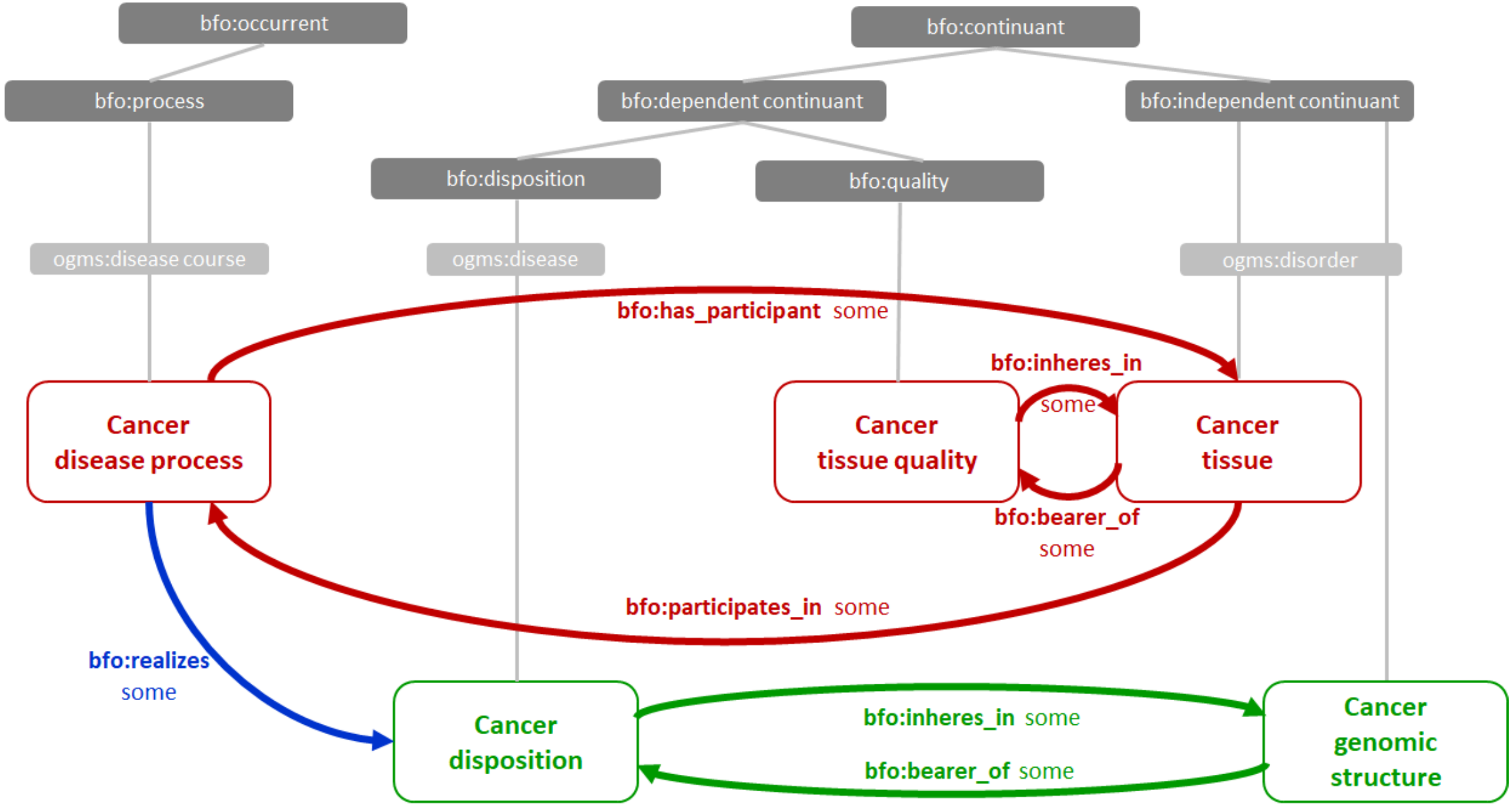
- Already reflected by the principle of self-grouping: concept X already means “having X”
  - Otherwise current modelling pattern of composite entities would be wrong, e.g. combined forearm fracture as child of both radius fracture and ulna fracture
  - In the new occurrent interpretation: state/process with a fracture
  - Also dispositions to be interpreted as "state with a disposition"

Morphological abnormalities as clinical continuants

- As being already placed under "body structure": obviously material continuants like all anatomical entities

Next steps?

- Finishing the manuscript
- Authors' conference call still this year?
- Sharing the agreed manuscript with BFO representatives
- Publication (Applied Ontology or Journal of Biomedical Semantics)
- Update SNOMED CT annotations and documentation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Cancer disease process
Cancer tissue quality
Cancer tissue
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bfo:realizes some
Cancer disposition
bfo:inheres_in some
bfo:bearer_of some
Cancer genomic structure