The BioTop Family of Upper Level Ontological Resources for Biomedicine

Stefan Schulz[1,2], Martin Boeker[2],
Catalina Martínez-Costa[1]

[1]Institute for Medical Informatics, Statistics and Documentation,
Medical University of Graz, Austria
[2]Medical Biometry and Statistics,
Medical Center, University of Freiburg, Germany

What formal ontologies are and do…

Why upper-level ontologies are necessary…

The role of BioTop in an ontology ecosystem…

What formal ontologies are and aren't

Formal ontologies are

"…theories that attempt to give precise mathematical formulations of the properties and relations of certain entities"

(Hofweber 2011)

Formal ontologies aren't

"…Swiss army knives for knowledge representation"

(Brochhausen et al., 2011)

Hofweber T. Logic and Ontology, Stanford Encyclopaedia of Philosophy (2011).
Brochhausen, M.; Burgun, A.; Ceusters, W.; Hasman, A.; Leong, T. Y.; Musen, M.; Oliveira, J. L.; Peleg, M.; Rector, A. and Schulz, S. (2011). Discussion of biomedical ontologies: Toward scientific debate. Methods Inf Med, 50:217-236.

What formal ontologies do and don't represent

They represent…

… what is universally true about entities of a domain

"all cell membranes contain lipids"

"all fetuses were embryos"

"cholecystectomy is the surgical removal of a gallbladder"

"fungi are not plants"

They don't represent…

… contingent characteristics, default knowledge, probabilistic associations

"Ebola infections are rare"

"adult humans have typically 32 teeth"

"Jaundice may indicate pancreas CA"

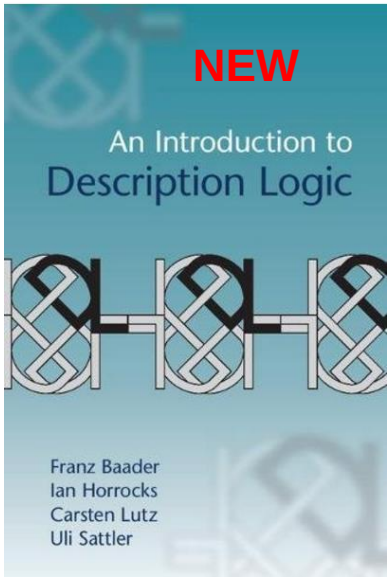
" Lmn-2 interacts with Elf-2 "

"most plant cells have cell walls"

Alan Rector (2008): "…very few interesting items of knowledge that are truly ontological…"

Formal ontologies use logics

- Formal ontologies are constituted by sets of axioms in some logic
- Most popular: description logics
- Expressiveness restricted to
  - Equivalence statements:
    class A is equivalent to class B
  - Subclass hierarchies:
    all members of A are members of B
  - Class disjointness: *no member of A is a member of B*
  - Existential restrictions:
    class of entities related to some A via a relation r
  - Relation hierarchies: *r is a subrelation of s*
  - (Universal restrictions, negations, …)
- Semantic Web standard OWL
  - (RDF linearization useful but not mandatory)



What are upper-level ontologies?

- Definition
  - Theories of highly general (domain-independent) categories*
- Examples
  - Basic Formal Ontology (BFO)
  - OBO Relation Ontology (RO)
  - General Formal Ontology (GFO)
  - Descriptive Ontology for Linguistic and Cognitive Engineering (DOLCE)
  - GALEN upper level ontology
  - UMLS Semantic Network
  - BioTop / BioTopLite

"domain upper level ontologies"

* Wolfgang Degen and Heinrich Herre. What is an upper level ontology? *Workshop on Ontologies*. 2001.

Why upper-level ontologies?

Import statement in program code

```
import re

(…)

Q = re.search(r'(.*) are (.*?) .*', text, re.IGNORECASE)
R = re.search(r'(.*) have (.*?) .*', text, re.IGNORECASE)
```

Import statement in ontology code

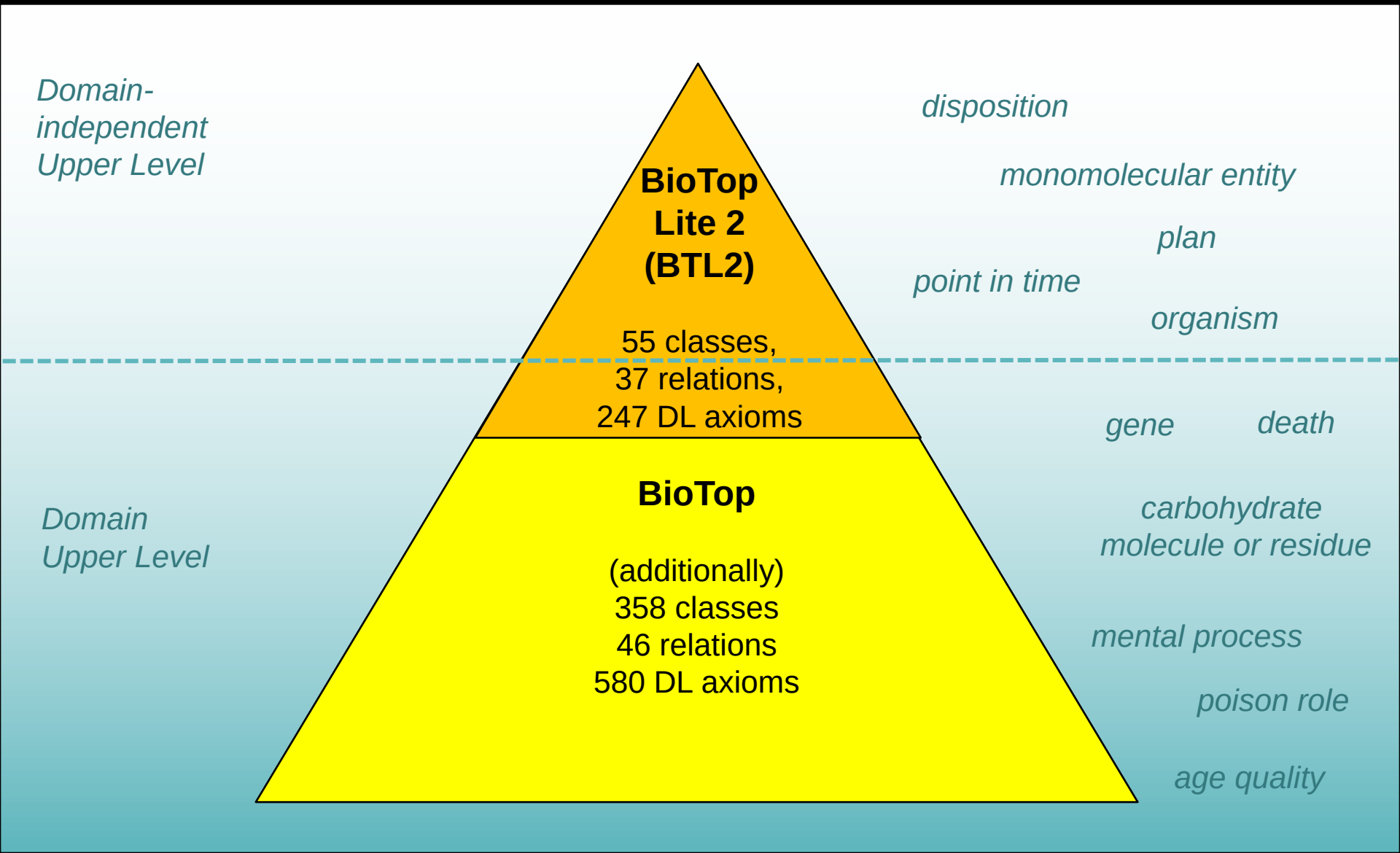
```
Import: http://purl.org/biotop/btl2.owl

Class: DigitalEntity
SubClassOf:
<http://purl.org/biotop/btl2.owl#InformationObject>

Class: IntellectualProduct
SubClassOf:
<http://purl.org/biotop/btl2.owl#InformationObject>,
<http://purl.org/biotop/btl2.owl#isPatientIn> some
(<http://purl.org/biotop/btl2.owl#Process>   and
(<http://purl.org/biotop/btl2.owl#hasAgent>
  some Human))
```

- Modular design: fundamental principle in SW engineering
  - Advantages: sharability, standardisation, interoperability
  - Challenges: interface management, versioning, performance

BioTop and BioTopLite



BioTop Design principles

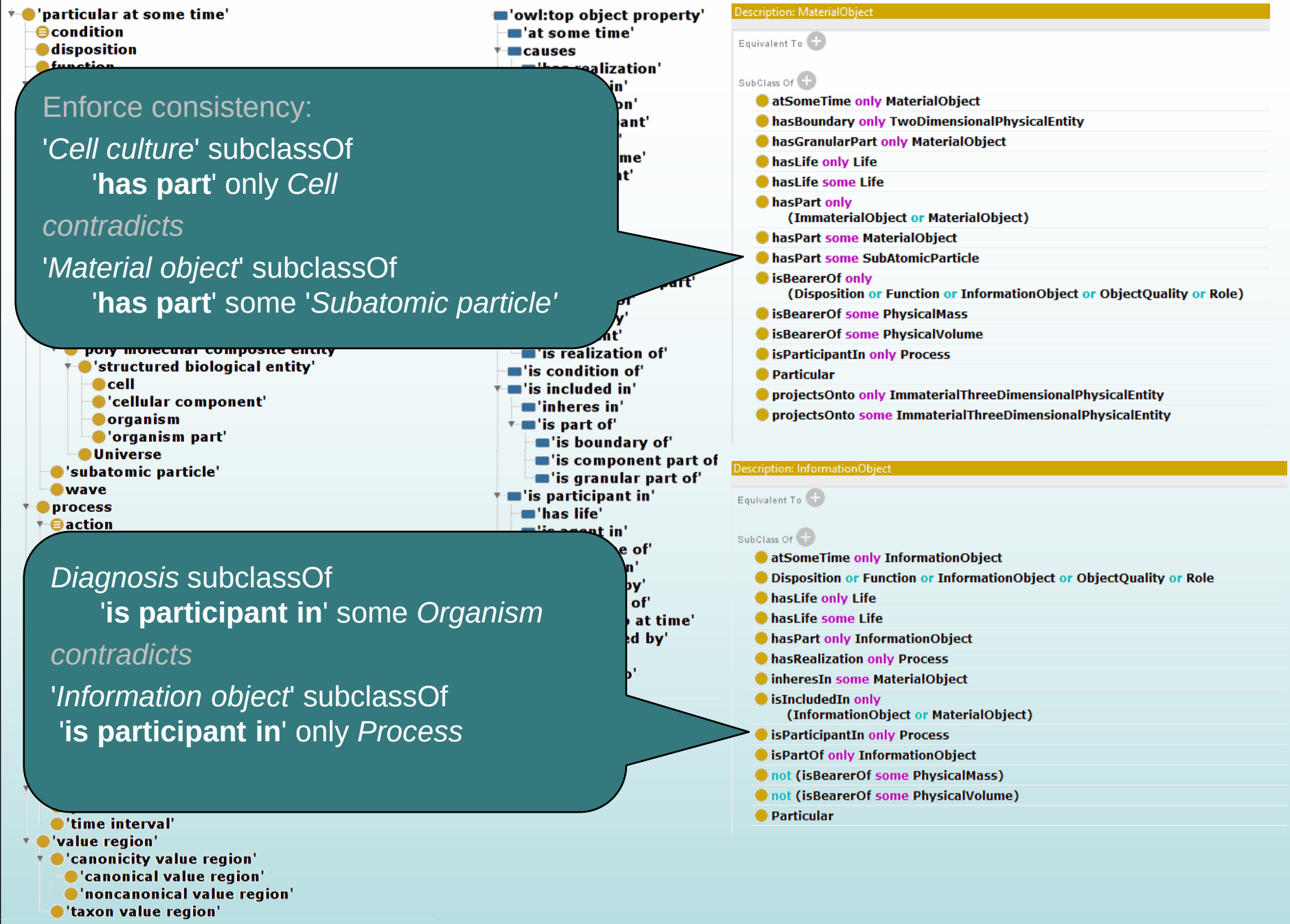
- Following the GoodOD "Good Ontology Design" guideline*
- Pragmatic realist view, admission of defined classes, e.g. "condition"
- Flat hierarchy
- Intuitive naming
- Textual elucidations with examples as annotation properties
- Expressiveness: OWL-DL
- Small set of relations (object properties)
- All instances are considered to be *temporally qualified* (ternary relations like **part of**(a, b, t) are not supported by OWL)

* Schulz et al. Guideline on Developing Good Ontologies in the Biomedical Domain with Description Logics http://www.iph.uni-rostock.de/fileadmin/PHF_Philosophie/media/goodod/GoodOD-Guideline_v1_2012.pdf.

BTL2 Classes

BTL2 Relations

BTL Axioms (examples)



Enforce consistency:

'*Cell culture*' subclassOf
'has part' only *Cell*

contradicts

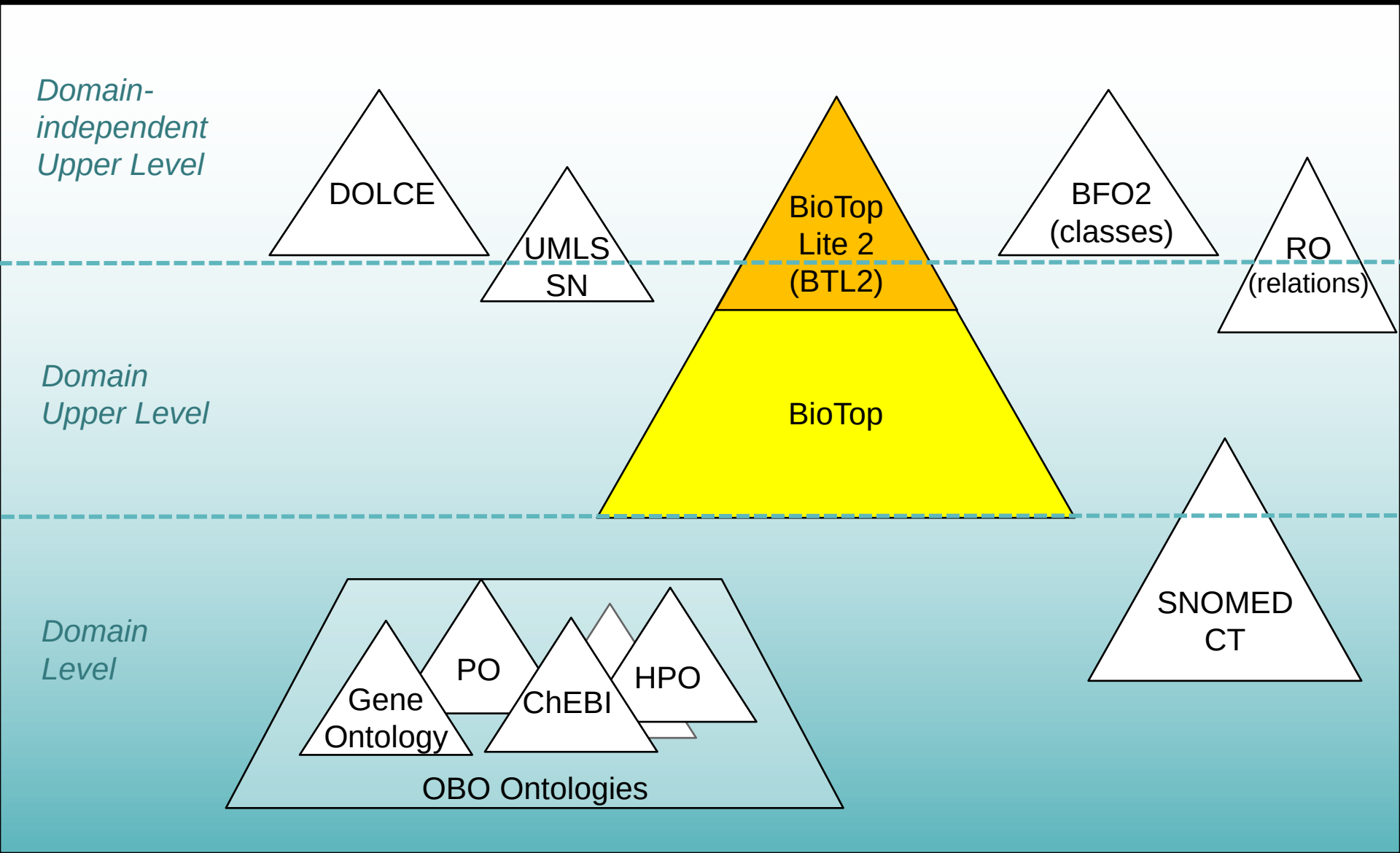
'*Material object*' subclassOf
'has part' some '*Subatomic particle*'

Diagnosis subclassOf
'is participant in' some *Organism*

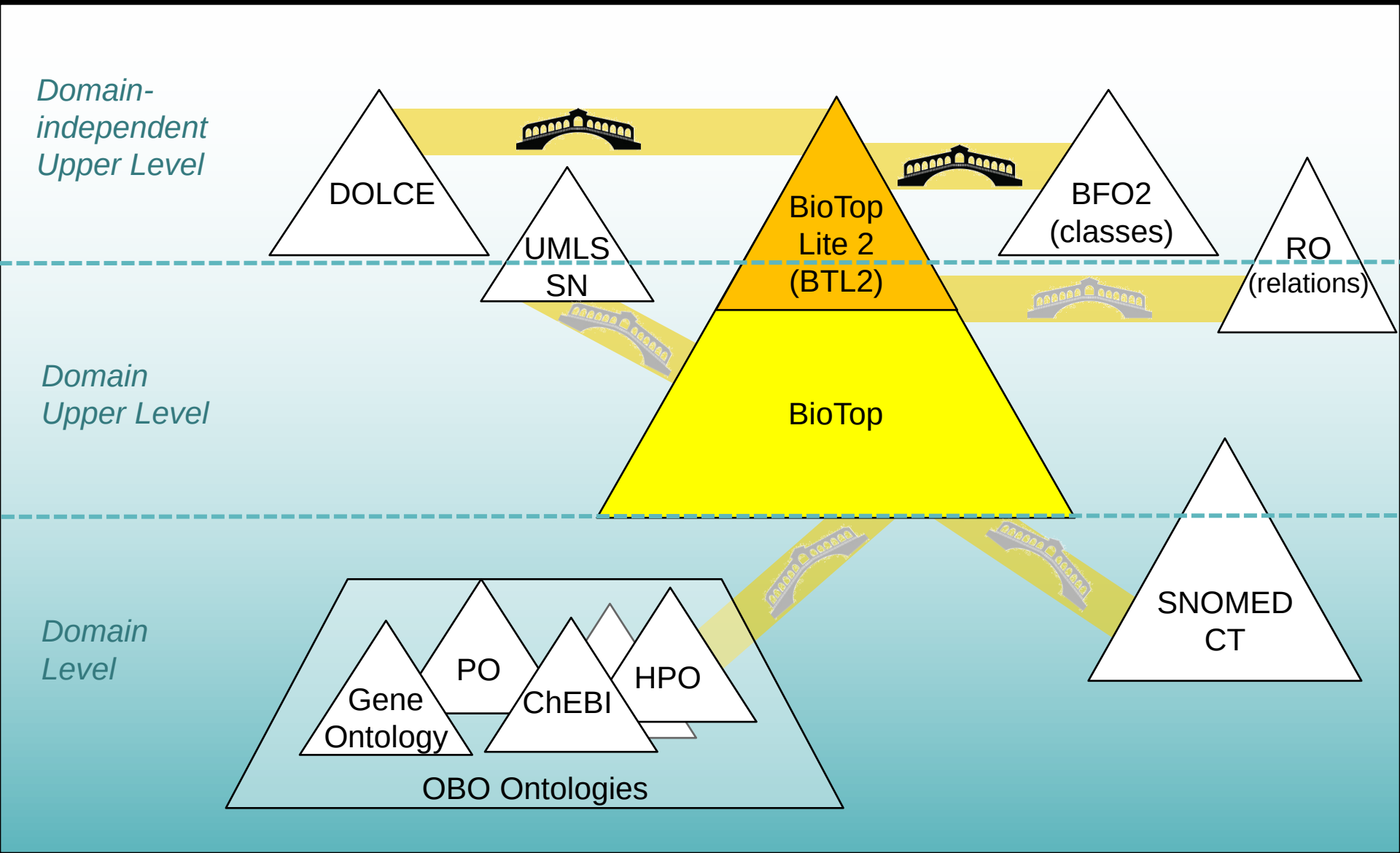
contradicts

'*Information object*' subclassOf
'is participant in' only *Process*

BioTop, BioTopLite and BioTop Bridges



BioTop, BioTopLite and BioTop Bridges



BioTop(Lite) access and references

- Access to BioTop, BioTopLite, and Bridging files
  - Repository: http://biotopontology.github.io/
  - URIs: BioTop: http://purl.org/biotop/biotop.owl BTL2: http://purl.org/biotop/btl2.owl
- Mailing list
  - https://groups.google.com/forum/#!forum/biotop
- References:
  - SemanticHealthNet EU Network of Excellence:
    Upper level for information model and clinical terminology
    http://www.semantichealthnet.eu
  - CELDA: ontology of cell types, in vitro as well as in vivo, based on species, anatomy, subcellular structures, developmental stages and origin
    http://cellfinder.org/about/ontology
  - International Health Terminology Standards Development Organization: in several experimental ontologies (event, condition, episode; observables)
  - TNM-O ontology support for staging of malignant tumours
    https://jbiomedsem.biomedcentral.com/articles/10.1186/s13326-016-0106-9
  - TeleHealth Ontology (TEON)
    http://journals.ukzn.ac.za/index.php/JISfTeH/article/view/143
  - IntegrativO Ontology
    http://integrativo.github.io/

EFMI EUROPEAN FEDERATION for MEDICAL INFORMATICS

Farr The Farr Institute of Health Informatics Research

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Stefan Schulz
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purl.org/steschu

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Contact:
stefan.schulz@medunigraz.at