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Setting the Scene to Link SNOMED CT to Realism-Based Ontologies

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AIDH IMIA

SNOMED CT

Parents

- Malignant adenomatous neoplasm (disorder)
- Malignant tumor of biliary tract (disorder)

Cholangiocarcinoma of biliary tract (disorder)

SCTID: 312104005

312104005 | Cholangiocarcinoma of biliary tract (disorder) |

en Cholangiocarcinoma of biliary tract (disorder)

en Cholangiocarcinoma of biliary tract

Finding site → Biliary tract structure

Associated morphology → Cholangiocarcinoma

Children (4)

- Cholangiocarcinoma of intrahepatic biliary tract (disorder)
- Cholangiocarcinoma of perihilar bile duct (disorder)
- Klatskin's tumor (disorder)
- Primary cholangiocarcinoma of extrahepatic bile duct (disorder)

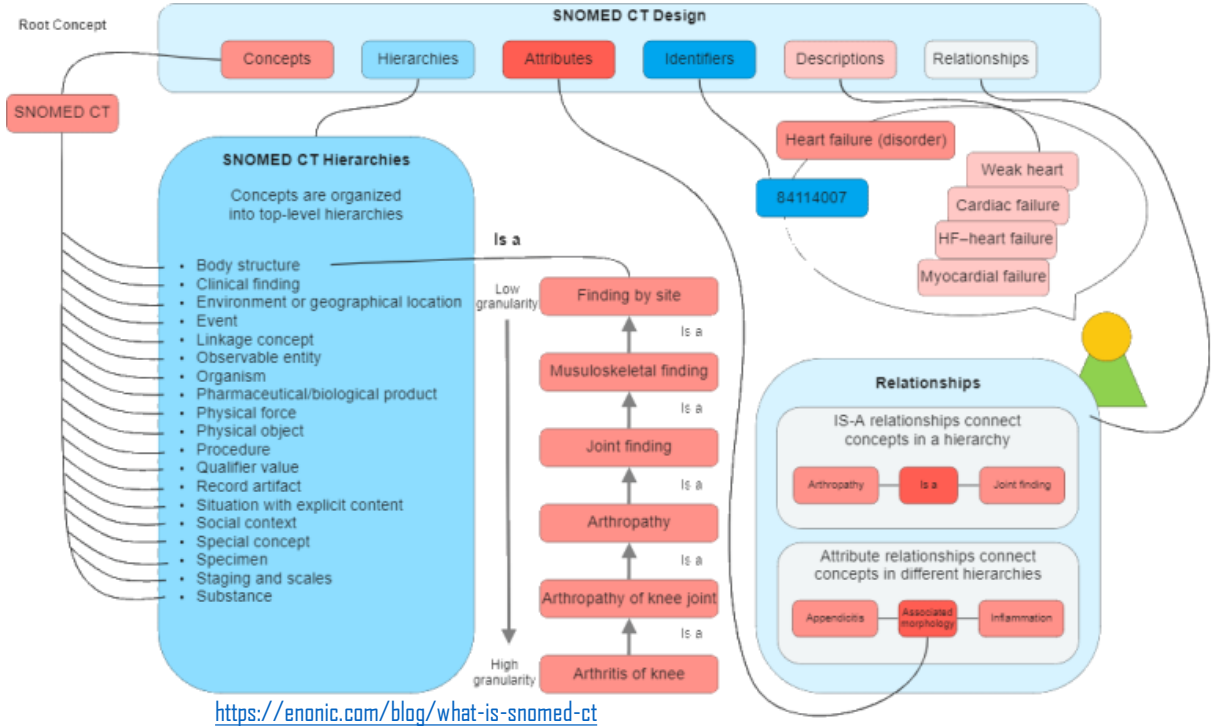
SNOMED CT's Description Logic: EL++

```
SubClassOf( :312104005 |Cholangiocarcinoma of biliary tract (disorder)|
            :443961001 |Malignant adenomatous neoplasm (disorder)|)

SubClassOf( :312104005 |Cholangiocarcinoma of biliary tract (disorder)|
            :363415003 |Malignant tumor of biliary tract (disorder)|)

EquivalentClasses(
        :312104005 |Cholangiocarcinoma of biliary tract (disorder)|
        ObjectIntersectionOf(:64572001 |Disease (disorder)|
            ObjectSomeValuesFrom(:609096000 |Role group (attribute)|
            ObjectIntersectionOf(
                ObjectSomeValuesFrom(
                    :116676008 |Associated morphology (attribute)|
                    :70179006 |Cholangiocarcinoma (morphologic abnormality)|)
                ObjectSomeValuesFrom(
                    :363698007 |Finding site (attribute)|
                    :34707002 |Biliary tract structure (body structure)|)))))
```

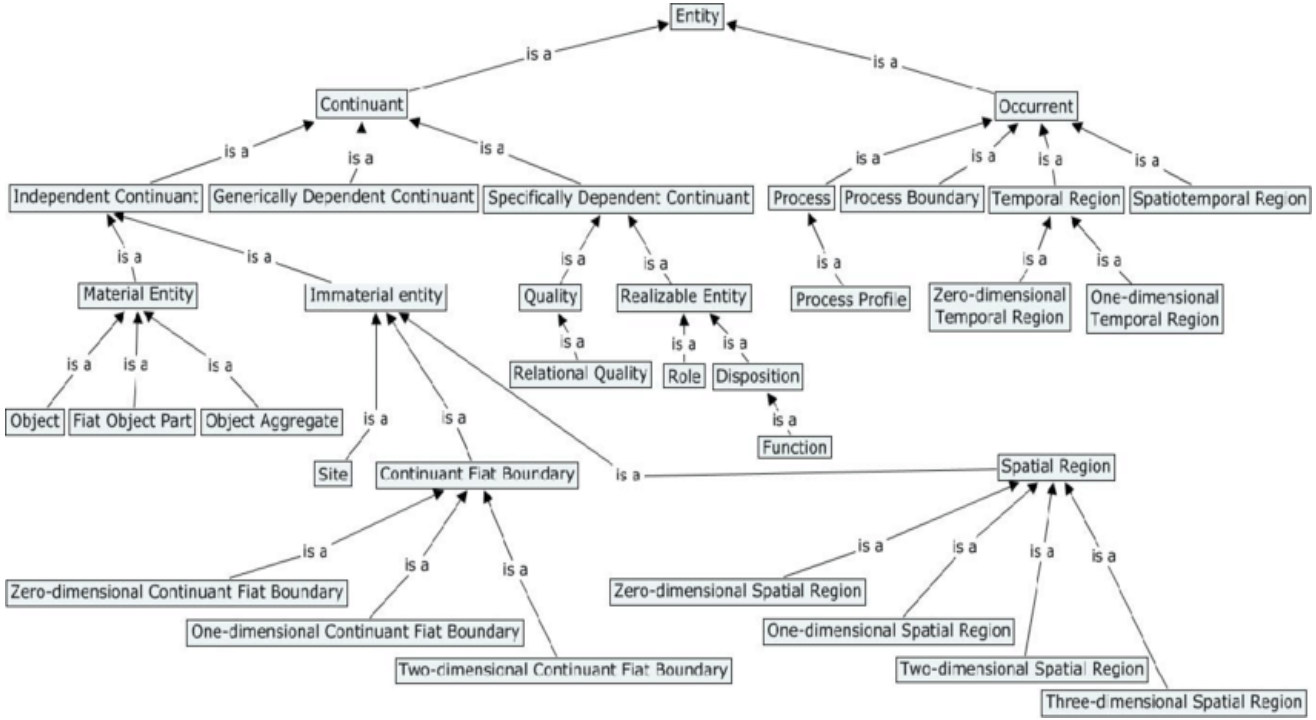
SNOMED CT hierarchies



https://enonic.com/blog/what-is-snomed-ct

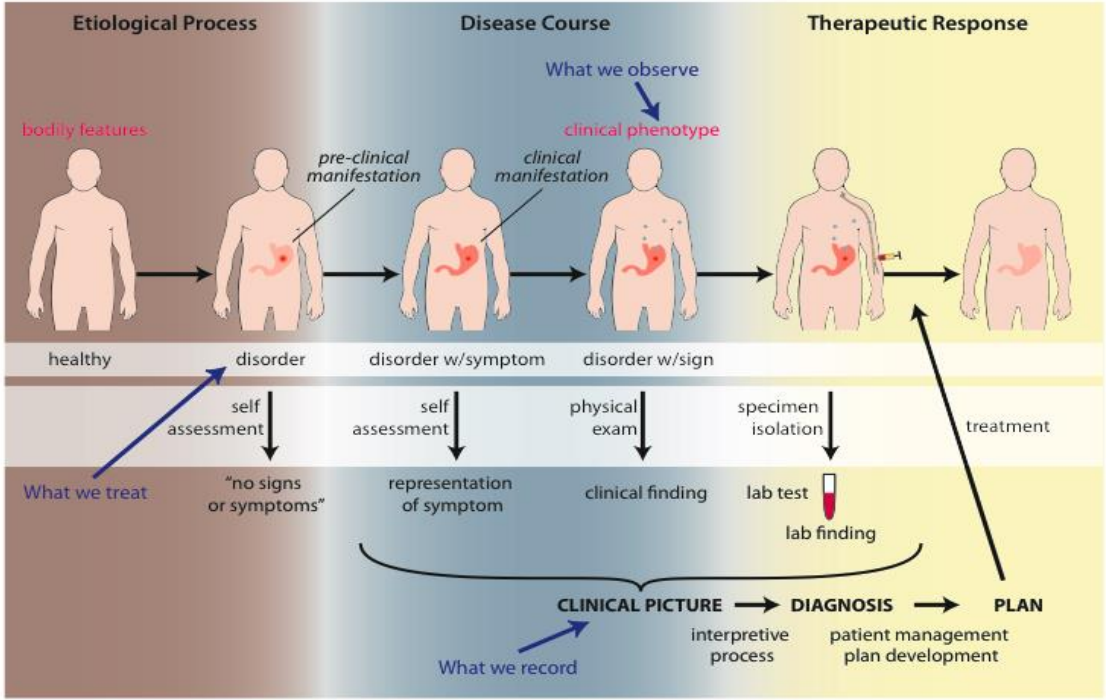
- Clinical perspective
- Pragmatics based
- No upper ontology
- Building blocks: concepts and terms
- Mix of object and subject language

The Basic Formal Ontology (BFO)



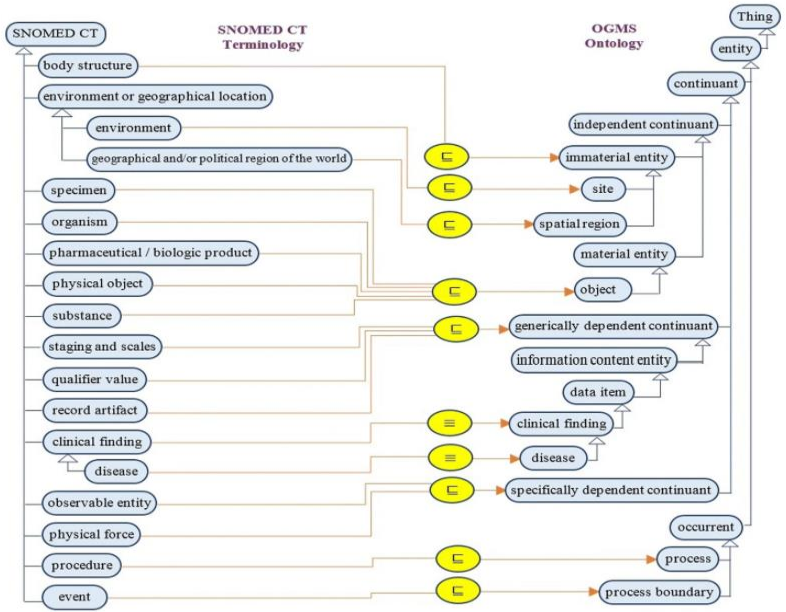
- Ontological realism
- Building blocks: universals and particulars
- Universal top ontology
- ISO-standard ISO/IEC 21838-2:2021
- Fully axiomatized in First Order Logic

Ontology of General Medical Science (OGMS)

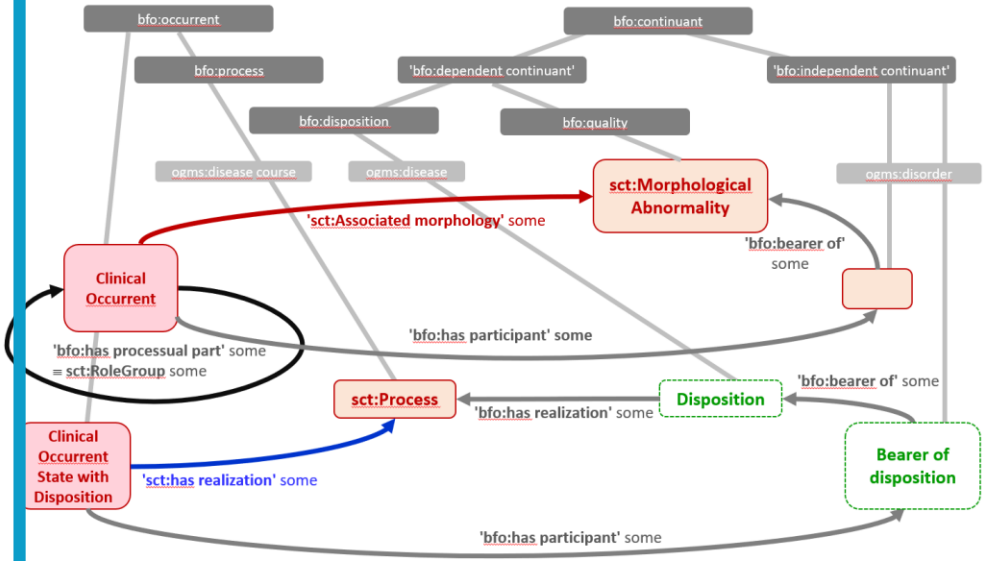


- BFO as top ontology
- Key distinctions:
  - disorder
  - disease
  - disease course
  - diagnosis
  - pathological process
  - bodily feature
  - ...
- Axiomatization in progress

Attempts to align SNOMED CT with BFO/OGMS



El-Sappagh, S., Franda, F., Ali, F. *et al.* SNOMED CT standard ontology based on the ontology for general medical science. *BMC Med Inform Decis Mak* 18, 76 (2018). https://doi.org/10.1186/s12911-018-0651-5



Schulz S. SNOMED CT x BFO: can the gap between legacy terminology and foundational ontology be bridged? ICBO/FOIS; September 16, 2021; Bolzano, Italy2021.

Our solution: one model-theoretic framework

- Domain of discourse contains three most elementary individuals:
  - Ontologically repeatable: universal (from BFO) concept (from SNOMED CT)
  - Ontologically unique: particular (from BFO)
- Basic assumption:
  - Although concepts cannot directly be mapped to universals, their non-repeatable counterparts are reliably describable from both BFO's and SNOMED CT's perspective
- This requires two separate multiplicity relations:
  - For universals: (forall (i u t) (if (instance-of i u t)
    (and (particular i) (universal u) (instance-of t temporal-region t))))
  - For concepts: (forall (x y) (if (individual-of x y) (and (particular x) (concept y))))

Snomed CT → BFO/OGMS translation example:

```
(forall (x y z)          (if          (and
                                      (individual-of x sctid-64572001-disease)
                                      (sctid-363698007-finding-site x y)
                                      (individual-of y sctid-34707002-biliary-tract-structure)
                                      (sctid-116676008-associated-morphology x z)
                                      (individual-of z sctid-70179006-cholangiocarcinoma)
                                      (and          (= x z)
                                                    (exists (rx ry t)
                                                                (and
                                                                (occupies-spatial-region x rx t)
                                                                (occupies-spatial-region y ry t)
                                                                (rcc-overlap rx ry t)
                                                                (instance-of x ogms-disorder t)
                                                                (instance-of y ogms-bodily-component t))
                                                    )
                                      )
                         )
)
```

Predicates in 'pure' SNOMED CT

Logical symbols, constructors and quantifiers

Predicates in 'pure' realism-based language

Take away messages

- Feasibility demonstrated in small domain
- Generalizability to more terminologies for which a direct mapping is not possible because of absent or incompatible ontological commitment
- Translation axioms can only partially be generated from the sources:
  - Automatically for the antecedent part of the axioms
  - Semi-automatically by means of templates for the consequent part
- Can't be done in OWL-DL because of the latter's limitation in the use of binary predicates only.

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