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Bridging the semantics gap between terminologies, ontologies, and information models

Representational Artifacts

Representational Artifacts

Ontology

- Theme of reality

Representational Artifacts

Ontology

- Theory of reality

Semantics

- Theory of meaning
  of (human language) designations

blah blah

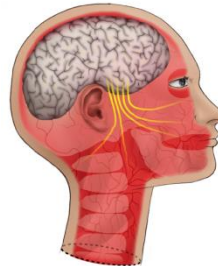# Representational Artifacts

Ontology

- Theory of reality
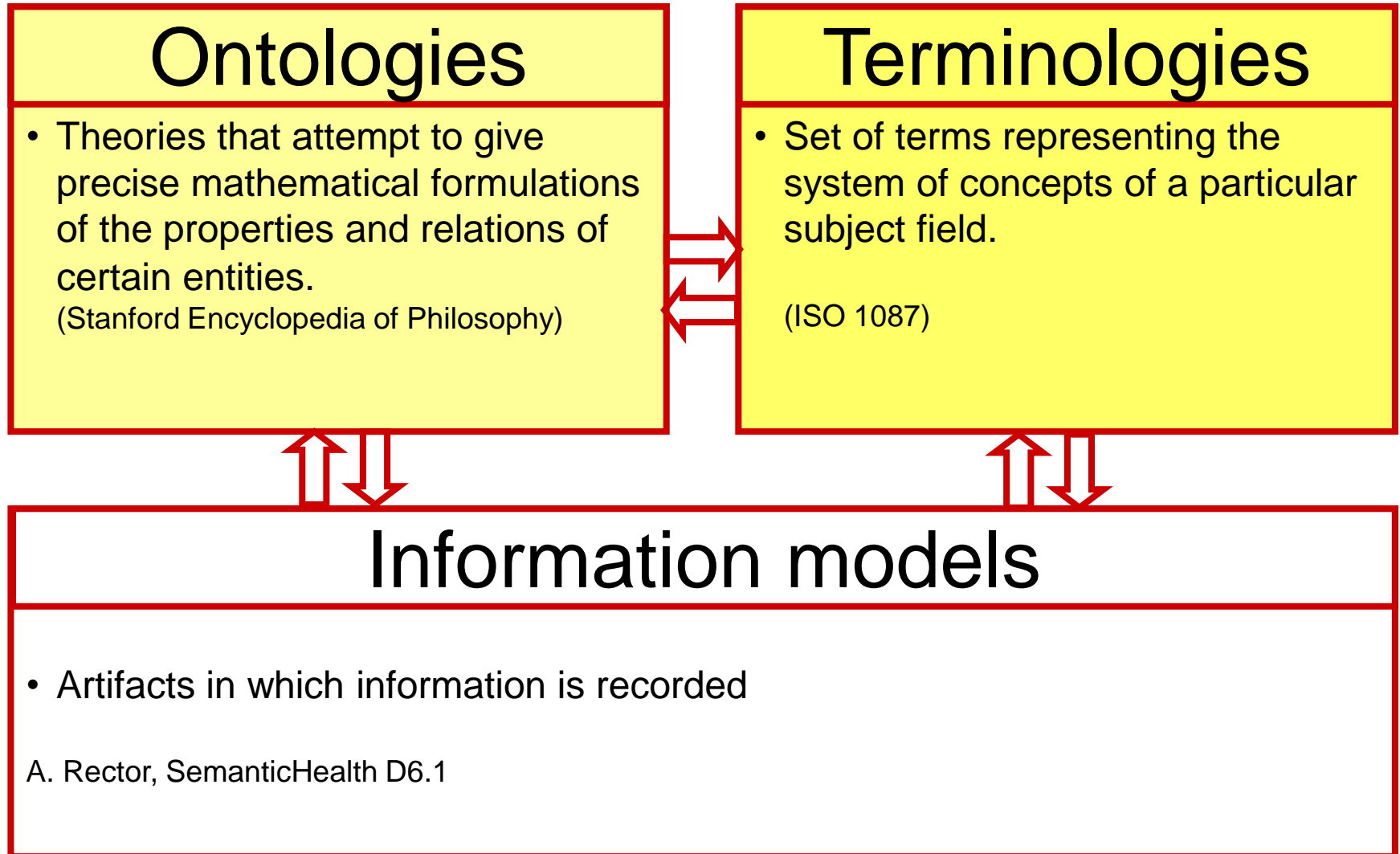

Semantics

- Theory of meaning
  of (human language) designations

blah blah

Epistemology

- Theory of knowledge

Representational Artifacts

Ontologies

- Theories that attempt to give precise mathematical formulations of the properties and relations of certain entities.
(Stanford Encyclopedia of Philosophy)

Terminologies

- Set of terms representing the system of concepts of a particular subject field.

(ISO 1087)

Information models

- Artifacts in which information is recorded

A. Rector, SemanticHealth D6.1

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Medical terminologies are sets of terms

Term
Operation on heart
Gallstones
Natural death
Helicobacter blood test
Asphyxia
Nose
Heart disease
Diabetes mellitus
Tuberculosis of lung

- Context independent
- Observer independent
- Can be represented by simple description logics axioms

Source: SNOMED CT

Connecting terminologies with ontologies

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A. Rector, SemanticHealth D6.1
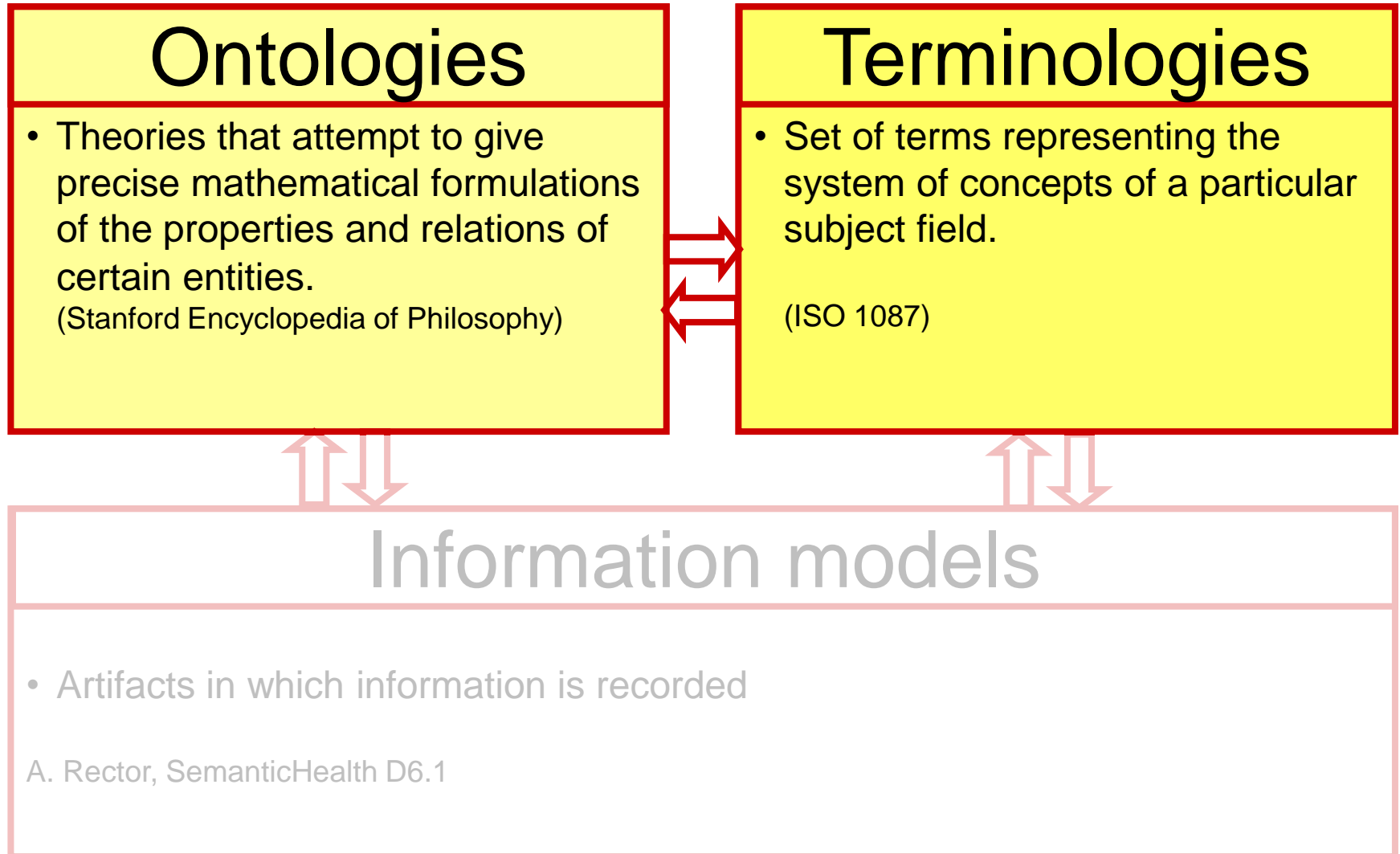
Simple description logic EL (as used in SNOMED CT)

DL Constructor		Meaning	Example
$\sqcap$	$E \sqcap F$	Intersection between E and F	*Acid $\sqcap$ Organic Molecule*
$\exists$	$\exists r.G$	Existential restriction of the relation r by the filler G	*$\exists$has-locus.Lung*
$\sqsubseteq$	$A \sqsubseteq B$	B subsumes A	*Lung $\sqsubseteq$ Organ*
$\equiv$	$C \equiv D$	C and D are equivalent	*Organic Acid $\equiv$ Acid $\sqcap$ Organic Molecule*

- Allows the expression of
  - Class hierarchies, e.g. *Hepatitis B $\sqsubseteq$ Hepatitis*
  - Necessary and sufficient conditions, e.g.
    *Hepatitis $\equiv$ Inflammatory disease $\sqcap$ $\exists$ **has-location**.Liver*
  - Pre/postcoordination of complex expressions, e.g.
    *Fracture of left femur $\equiv$ Femur Fracture $\sqcap$ $\exists$ **has-laterality**.Left*
- But not:
  - Negations, cardinality constraints, value restrictions

Medical terminologies are collections of terms

Domain terms   *Domain terms ??*

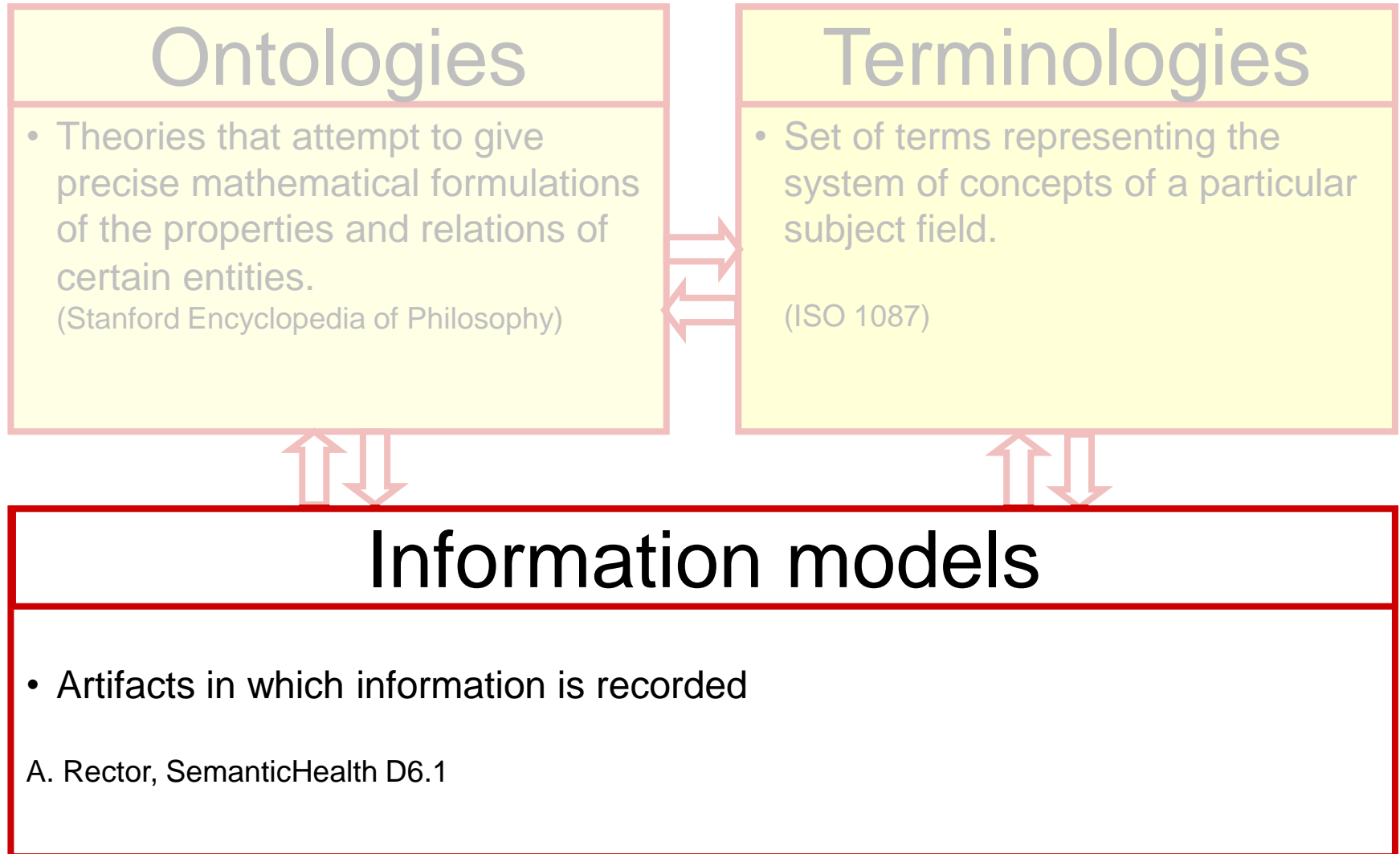|	
Operation on heart	Operation on heart, rescheduled
Gallstones	Suspected Gallstones
Natural death	Natural death with probable cause suspected
Helicobacter blood test	Helicobacter blood test negative
Asphyxia	Poor condition at birth without known asphyxia
Nose	Absent Nose
Heart disease	Heart disease excluded
Diabetes mellitus	Newly diagnosed diabetes
Tuberculosis of lung	Tuberculosis of lung, confirmed histologically

Source: SNOMED CT

Terms vs. Propositions

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- Context independent
- Observer independent
- Can be represented by simple description logics axioms

- Context and observer dependent (administrative, clinical contexts)
- Epistemic contexts
- Cannot be represented in simple DL

Representational Artifacts

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Terms vs. propositions ≈ Ontologies vs. Information models

Domain Ontologies	Information Models
Contain classes that have **physically existing domain entities** (particulars) as members	Classes have **information entities** as members
Represent real-world particulars in terms of their inherent properties	Represent **artifacts** that are build to collect or annotate **information**
Can exist **independently of information models** as long as only the existence of particular things is recorded	Are required to record beliefs or states of knowledge **about real things** or types of things (as represented by ontologies)
Relatively context independent	**Context dependent**

- Language is misleading:
    - HL7 **Act Class**: "*a record of something that is being done, has been done, can be done, or is intended or requested to be done*"; i.e. members of this class are not concrete acts, but information entities about acts.

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"Tonsillectomy"

"Planned Tonsillectomy"

denotes

denotes

isAbout

real process

information artifact

Can Information Models be expressed by the same logical framework as ontologies?

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- Example SNOMED CT: "inbuilt" information model ("situation in specific context")

  - *Absent nose $\sqsubseteq$ Congenital malformation $\sqcap$*
    $$\exists \textbf{FindingSite}. \textit{Nasal Structure}$$

  - *Suspected Gallstones $\equiv \exists$ rg.*
    ($\exists$ **Associated finding**.*Gallstone* $\sqcap$
    $\exists$ **Finding context**.*Suspected* $\sqcap$
    $\exists$ **Temporal context**. *Current or Specified* $\sqcap$
    $\exists$ **Subject relationship context**. *Subject of record*)

  - *Rescheduled operation of heart $\equiv$*
    *Operation on heart $\sqcap \exists$ **Priority**. Rescheduled.*

Can Information Models be expressed by the same logical framework as ontologies?

- Information entities as an extension of a domain ontology
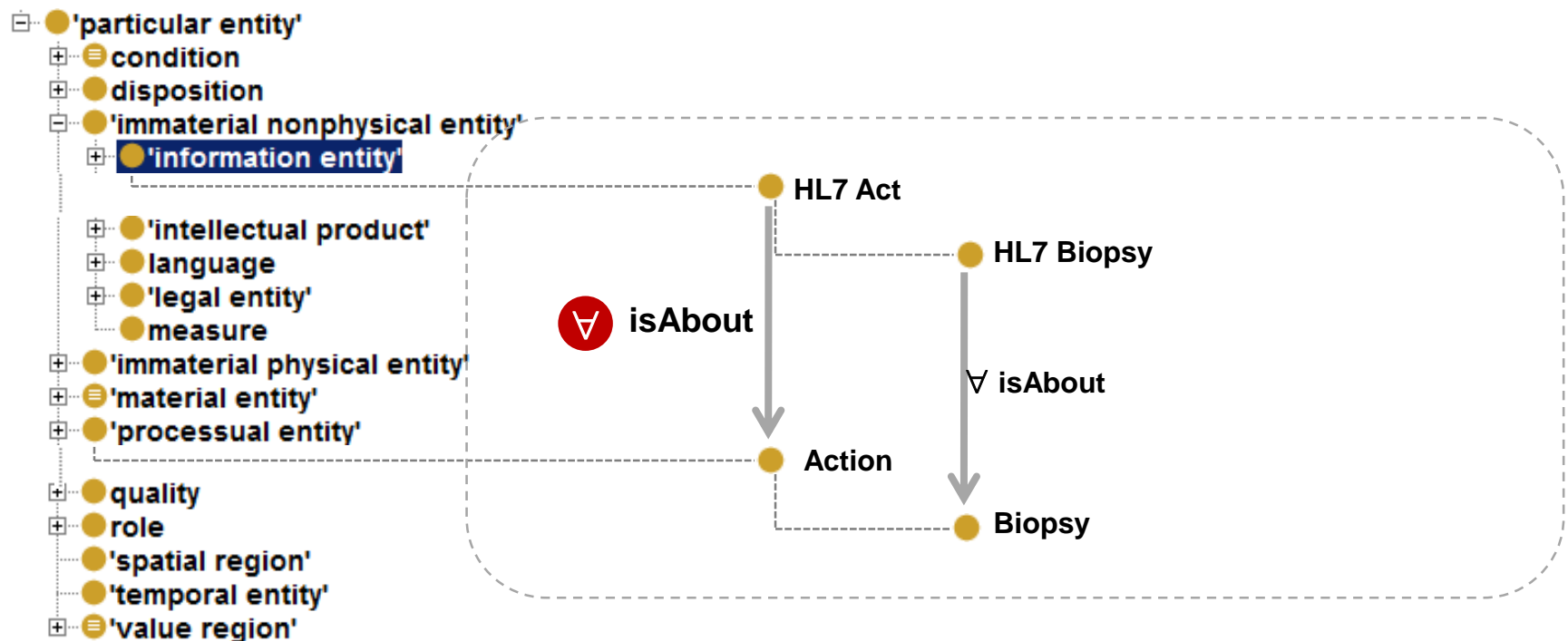- Example: BioTop upper ontology


```
'particular entity'
    condition
    disposition
    'immaterial nonphysical entity'
        'information entity'
        'intellectual product'
        language
        'legal entity'
        measure
    'immaterial physical entity'
    'material entity'
    'processual entity'
    quality
    role
    'spatial region'
    'temporal entity'
    'value region'
```

Can Information Models be expressed as ontologies?

- Information entities as an extension of a domain ontology

- Example: BioTop upper ontology
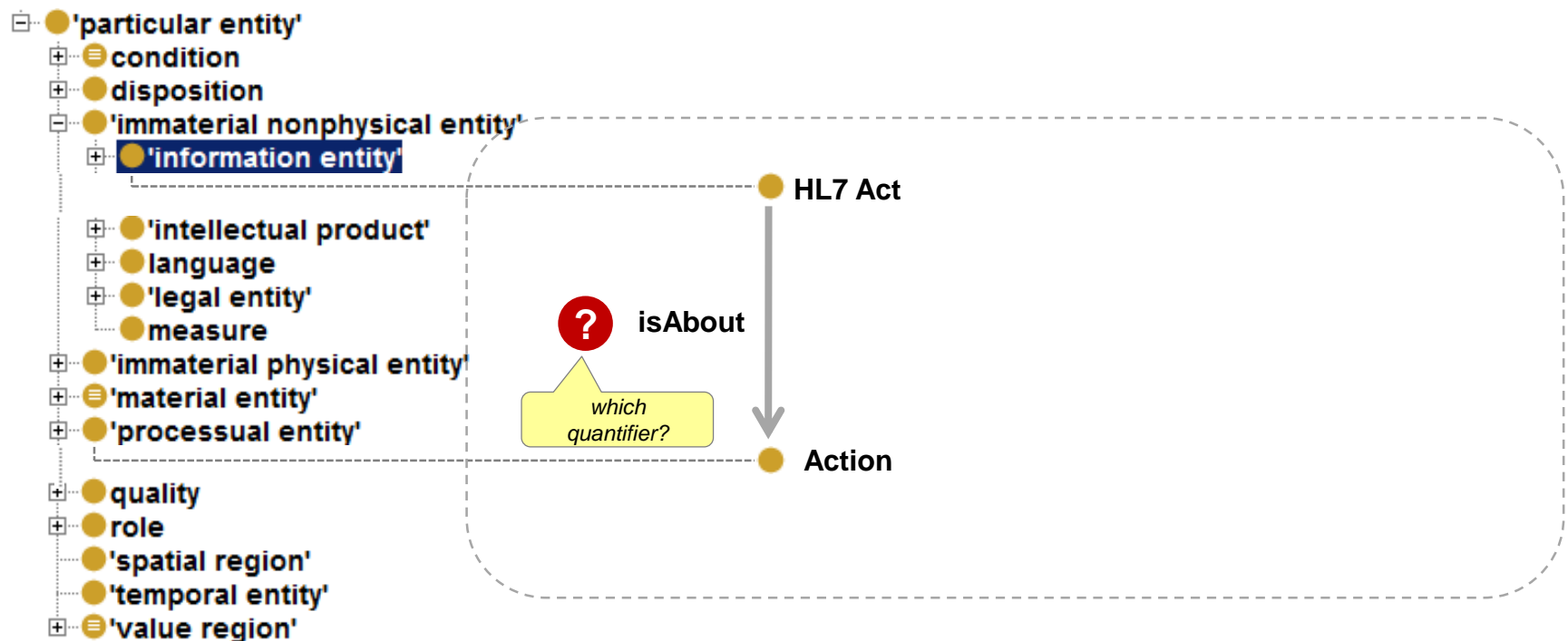
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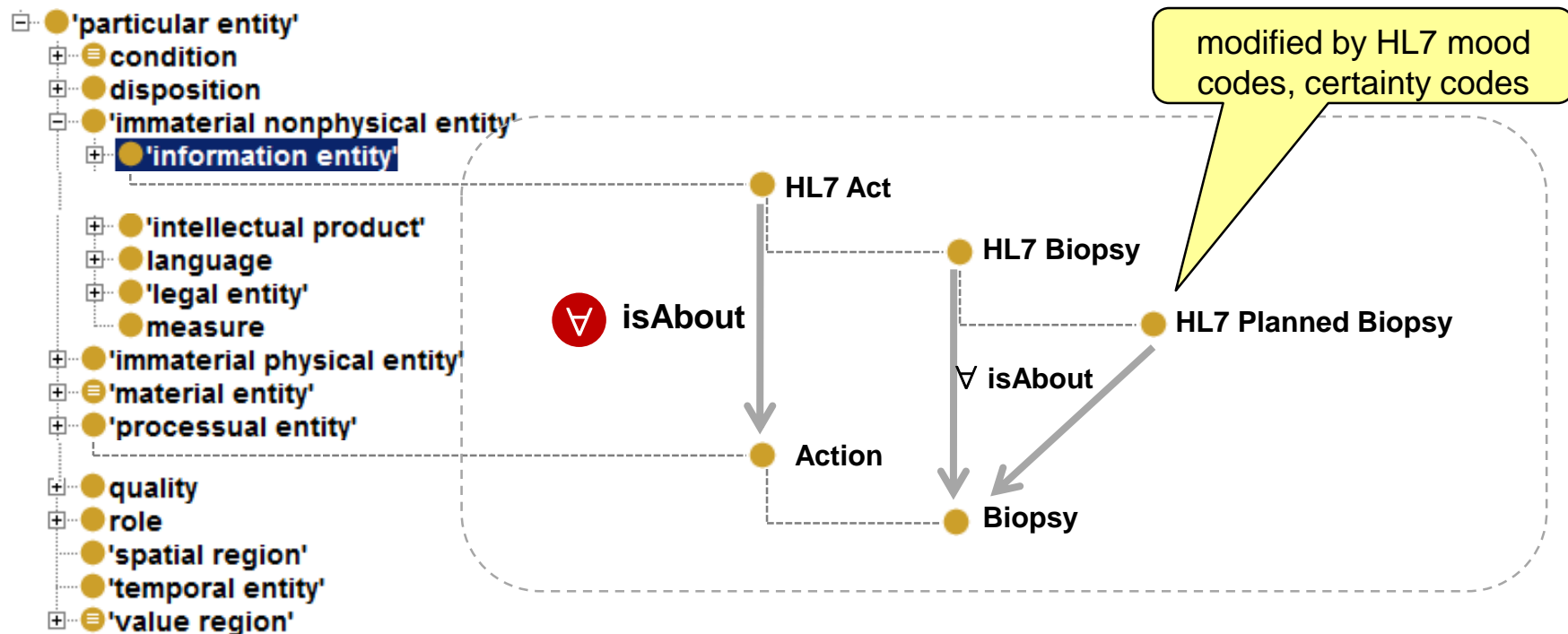
Extended description logics

DL Constructor		Meaning	Example
$\neg$	$\neg A$	Negation of A	$Base \sqsubseteq \neg Acid$
$\forall$	$\forall r.G$	Value restriction of the relation r by the filler G	$Hand \sqsubseteq$ $\forall has\text{-}Laterality.$
$\sqcup$	$A \sqcup B$	of A with B	$(Left \sqcup Right)$

Examples (I)

- ***Absent nose***:
  - *Human ⊓ ¬ hasPart. Nasal Structure*

- ***Planned heart operation:***
  - *Plan ⊓ ∀ isAbout. Operation on heart*

- ***Rescheduled heart operation***:
  - *Plan ⊓ (∀ isAbout. Operation on heart) ⊓ ∃ participantOf.Rescheduling*

Examples (II)

- Diagnosis of Gallstones

  Diagnosis $\sqcap \forall$ **isAbout**.*Gallstones*

- Confirmed diagnosis of Gallstones

  Diagnosis $\sqcap \forall$ ***isAbout***.*Gallstones* $\sqcap \exists$ *isAbout. Gallstones*

- Suspected diagnosis of Gallstones

  Diagnosis $\sqcap \forall$ ***isAbout***. *Gallstones* $\sqcap \exists$ *hasQuality.Suspected*

- Gallstones not diagnosed

  Gallstones $\sqcap \neg\exists$ *inv(**isAbout**).Diagnosis*

- A false diagnosis of gallstones

  Diagnosis $\sqcap \forall$ *IsAbout.* $\perp$

use referent tracking

Important references

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- Ceusters W, Smith B: Strategies for referent tracking in electronic health records.
  Journal of Biomedical Informatics, 2006, 39 (3), 362–378

- Rector AL, Brandt, S. Why Do it the hard way? The case for an expressive description logic for SNOMED. Journal of the American Medical Informatics Association 2008; 15: 744–751.

- IAO Information Artifact Ontology.
  http://code.google.com/p/information-artifact-ontology/

- BioTop Domain Upper Level Ontology
  http://purl.org/biotop

Conclusions

- Common logical framework encompassing ontologies and information models is possible

- Representing information model entities using SNOMED style simple description logics (OWL-EL) produces incorrect axioms, leading to false conclusions

- More expressive logic (OWL-QL) is necessary to represent most propositions typical for medical information models

- Strict upper level distinction between information entities and real world entities

- Using the **isAbout** relation to bridge information entities with classes of real world entities

- Important: appropriate use of quantifiers to prevent inadequate existential claims

Acknowledgements

Terminologies /
Ontologies stray
the terrain of
information
models

**TermInfo Draft
Standard
for Trial Use
(DSTU):**

Information
models
stray the
terrain of
Terminologies /
Ontologies