

**VIENNA
ISMB
ECCB
2011**



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July 15–16
SIGS AND TUTORIALS
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CONFERENCE

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Bio-Ontologies SIG

**Records and situations.
Integrating contextual
aspects in clinical
ontologies**

Representing entities and discourse in biomedicine and health care

Representing entities and discourse in biomedicine and health care

Ontology

- Theory of reality



Representing entities and discourse in biomedicine and health care

Ontology

- Theory of reality



Semantics

- Theory of meaning
of (human language) designations



Representing entities and discourse in biomedicine and health care

Ontology

- Theory of reality



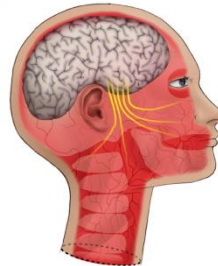
Semantics

- Theory of meaning
of (human language) designations

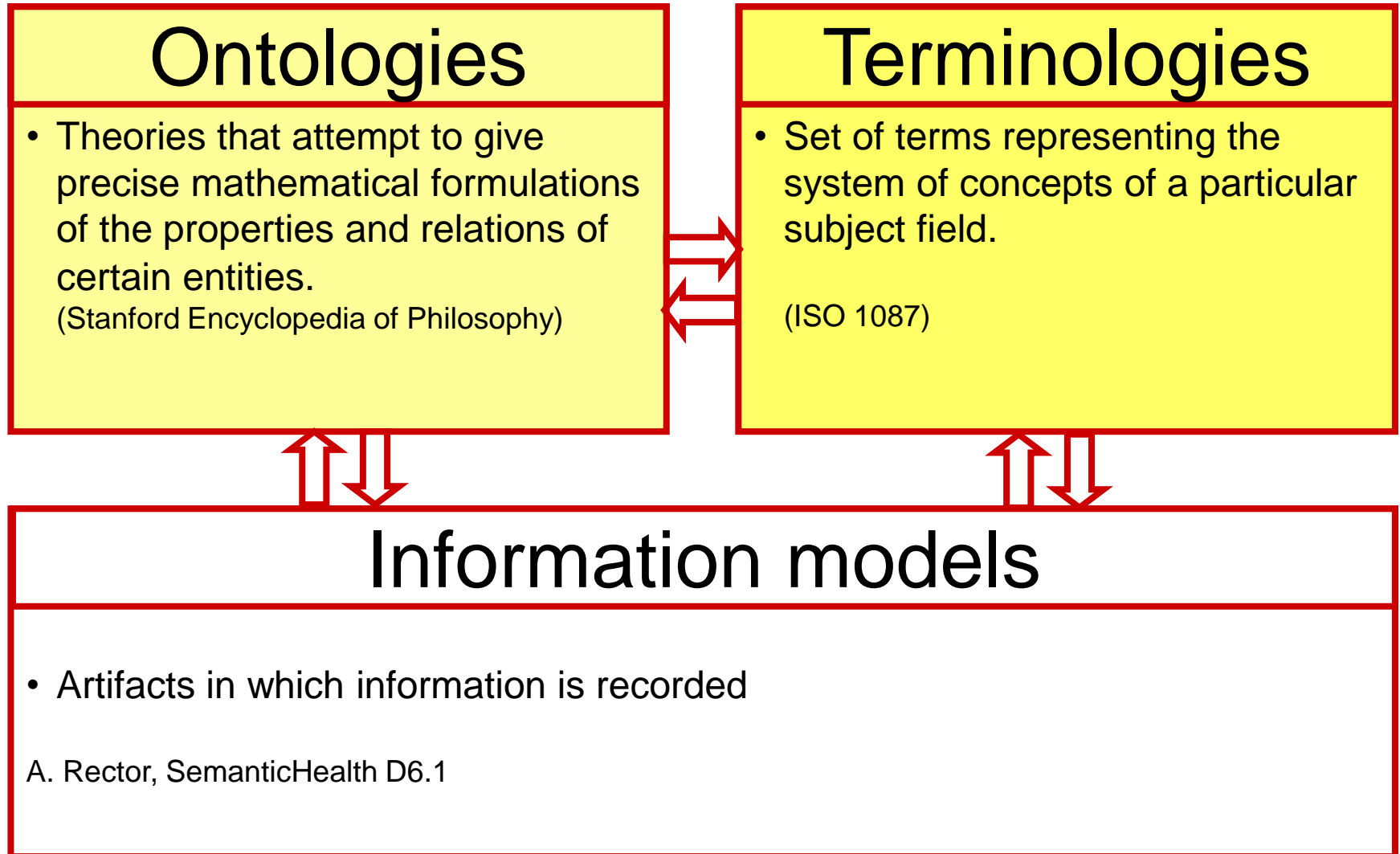


Epistemology

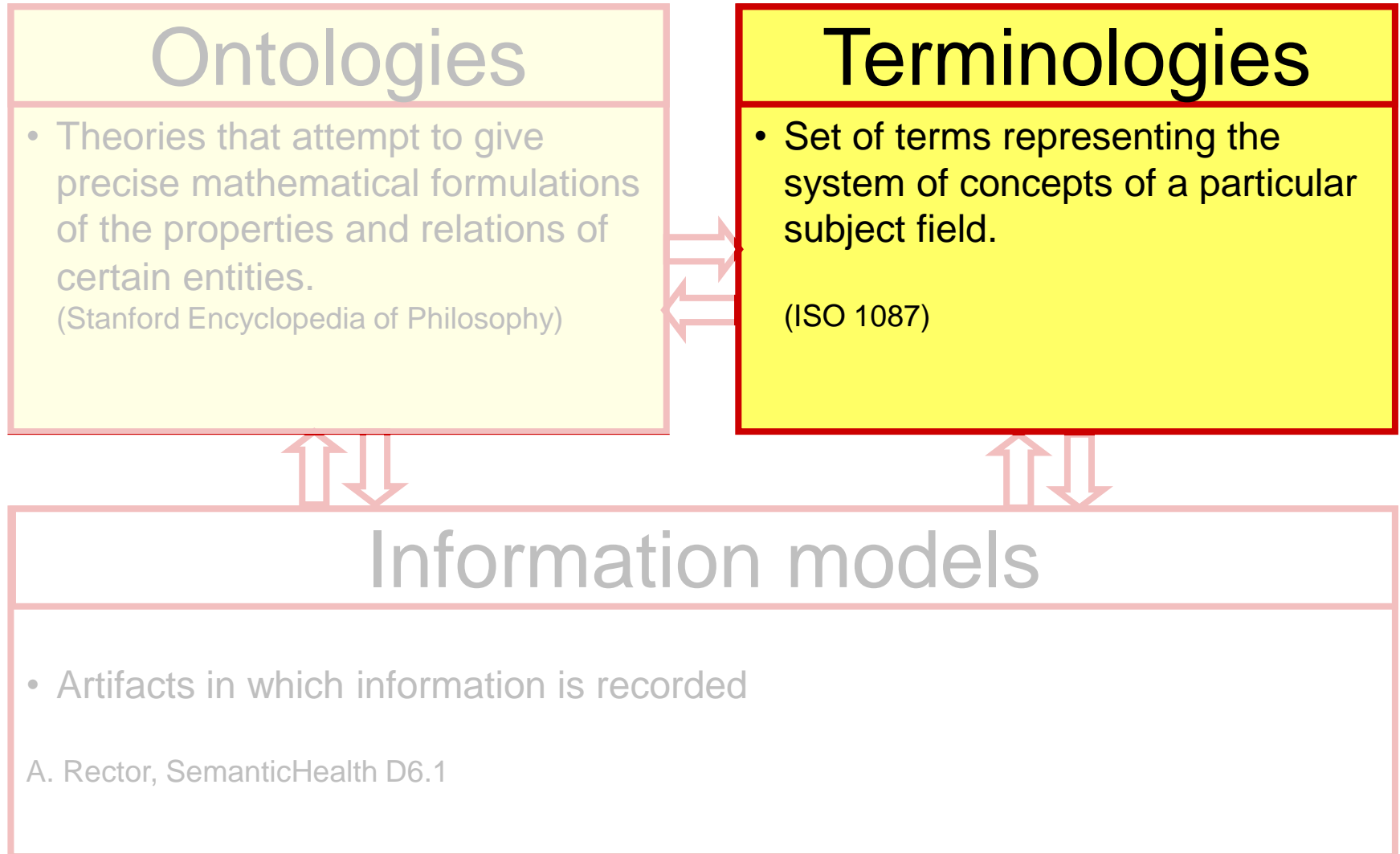
- Theory of knowledge



Representational Artifacts



Representational Artifacts



Biomedical terminologies are sets of terms...

Example SNOMED CT

Domain terms

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

Biomedical terminologies are sets of terms ?

Example SNOMED CT

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

Biomedical terminologies are sets of terms ?

Example SNOMED CT

Domain terms

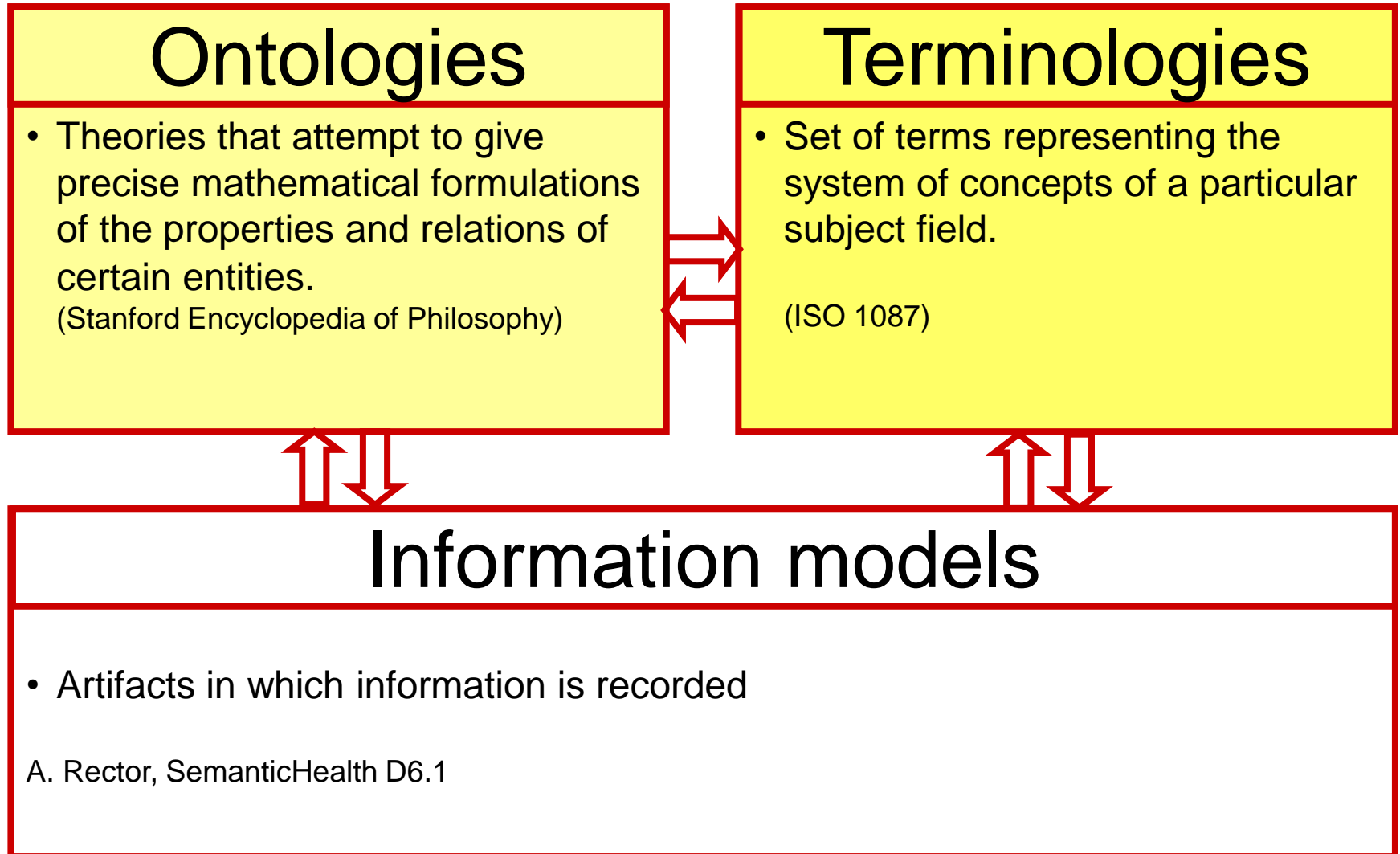
Propositions

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
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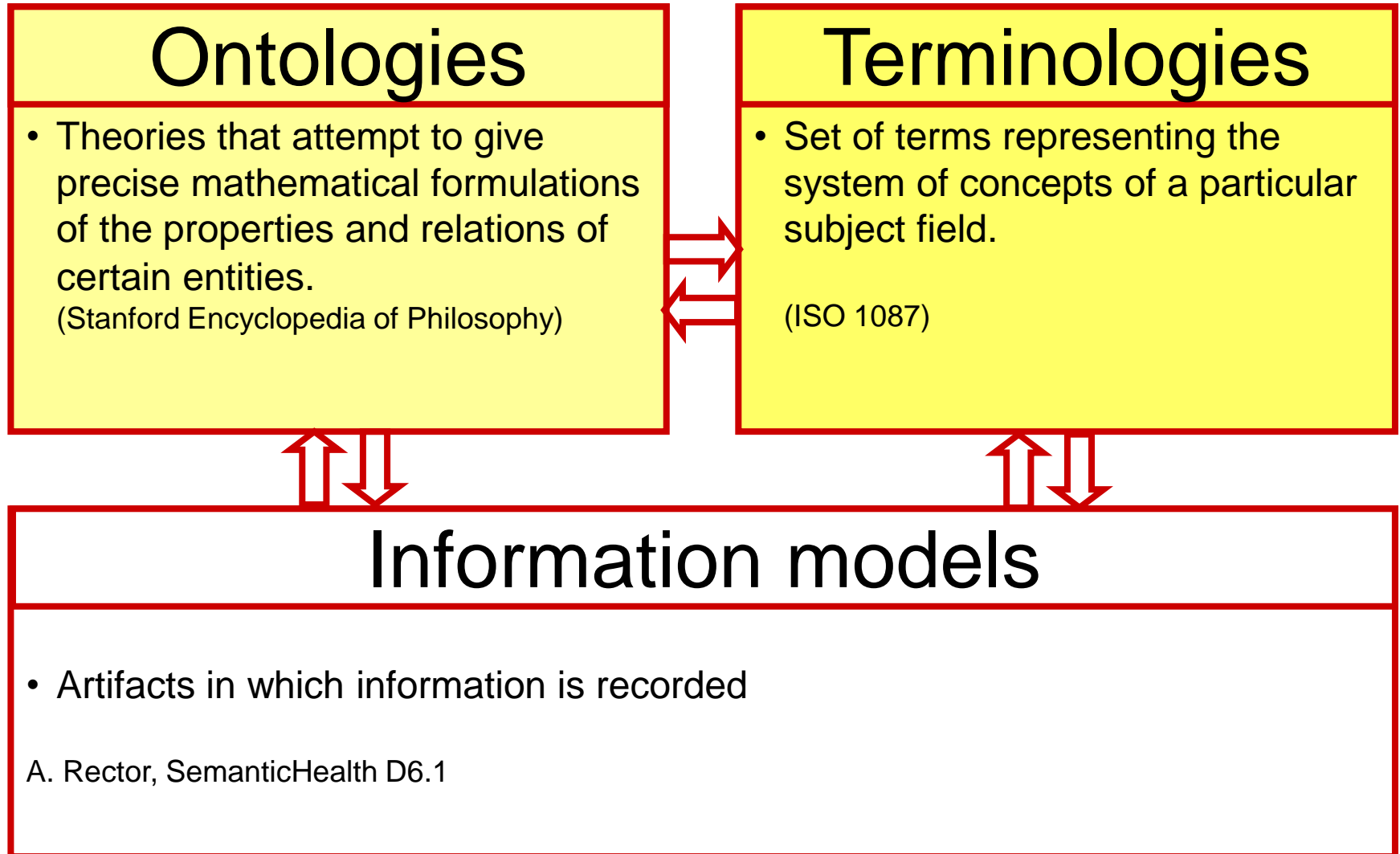
- Context independent
- Context and observer dependent (administrative, clinical contexts)
- "Epistemic intrusion" [1]

[1] Bodenreider O, Smith B, Burgun A (2004). The Ontology-Epistemology Divide: A Case Study in Medical Terminology. Int. Conf. on Formal Ontology and Information Systems (FOIS 2004). Amsterdam: IOS-Press, 185-195.

Representational Artifacts



Representational Artifacts



Classical view: Terms vs. propositions \approx Ontologies vs. Information models

Domain Ontologies	Information Models
Contain classes that have physically existing domain entities (particulars) as members	Classes have information artifacts 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:
 - A suspected gallstone is not a gallstone
 - An absent nose is not a nose
 - A rescheduled operation is not an operation
 - A planned tonsillectomy is no tonsillectomy

Information on
gallstones, noses,
operations etc

Terms vs. propositions \approx

Ontologies vs. Information models

Domain Ontologies	Information Models
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Relatively context independent	Context dependent

“Tonsillectomy of patient #123”

denotes



real process

“Planned Tonsillectomy”

denotes

OPERATING ROOM PLANNER				
Procedure & Remarks	Surgeon	Attending	Anesthetist	An.Type

information artifact

isAbout

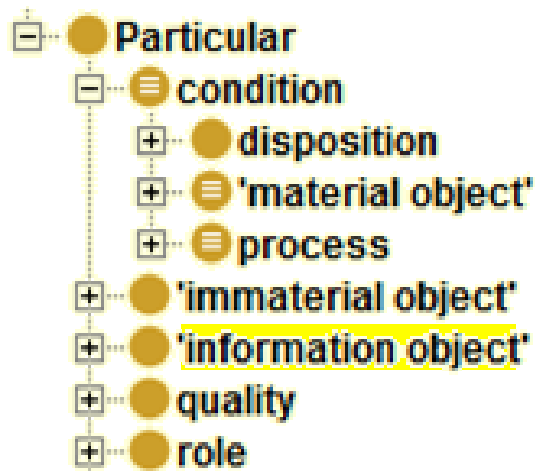


type of process

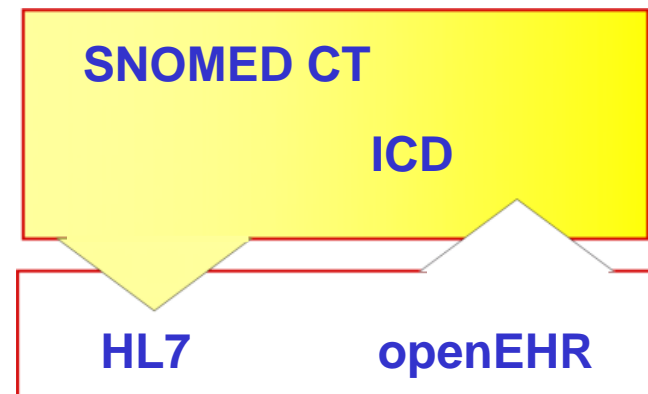
Crisp boundary or gradient ?

Domain Ontologies	Information Models
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Relatively context independent	Context dependent

ontologically



technically

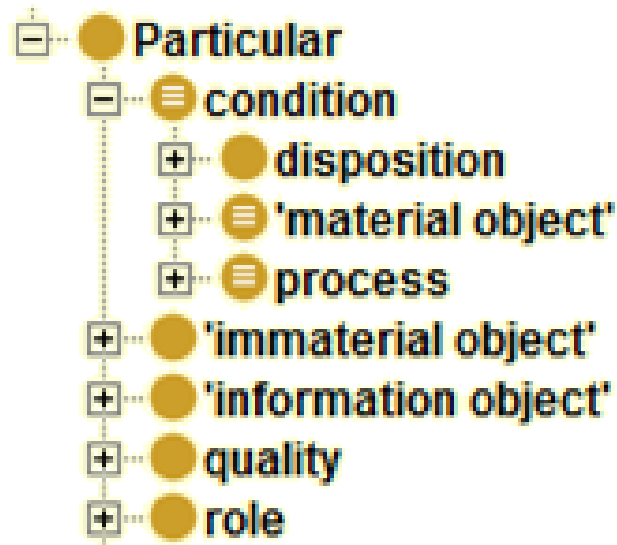


Proposal

- Our proposal:
 - refrain from a “canonic” division between ontologies and information models
 - common ontological framework which accommodates both based on the BioTop upper level ontology
 - interoperability between different representational flavors (ontology / information model combination)
 - example use case and competency questions

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

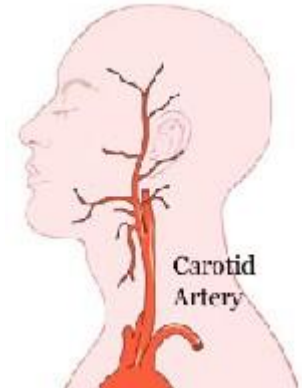
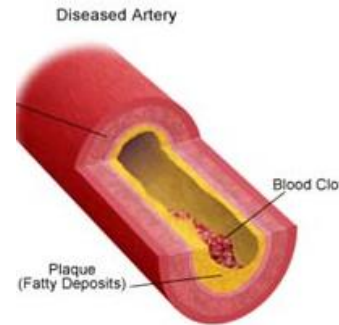
- Information objects as extension of a domain ontology
- Example: BioTop upper ontology (<http://purl.org/biotop>)



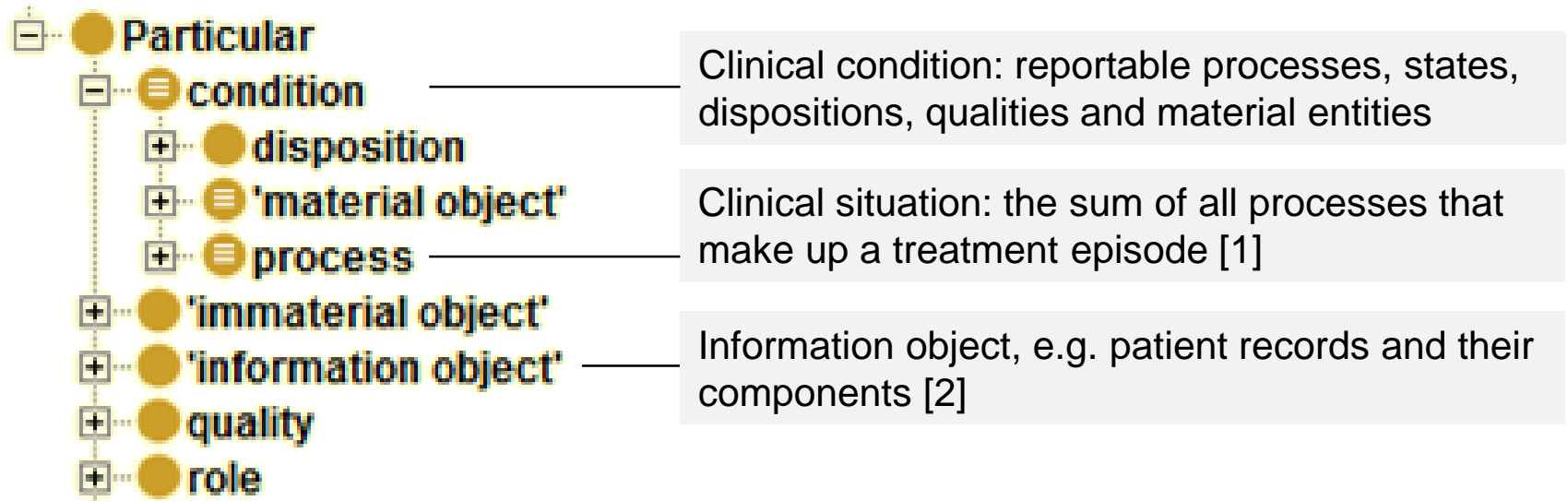
Running example.

"Stenosis of the left carotid artery"

- 1. Stenosis of artery
 - can be on the carotid artery
 - can be left or right
- 2. Proposition on (1.):
 - known whether present or absent
 - unknown whether present or absent
 - on patient him/herself or on a relative, e.g. parent
 - asserted as a future risk



Upper level



[1] 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.

[2] Ruttenberg, A., Courtot, M., The IAO Community: The Information Artifact Ontology (2010)
<http://code.google.com/p/information-artifact-ontology/>

Competing information Model Representations

"Mention of stenosis of right carotid in a patient's health record"

Information model templates

Postcoordination at information model level

Attribute	Value
Finding Context	
Disorder	Stenosis
Location	Carotid artery
Laterality	Left

Attribute	Value
Finding Context	
Disorder	Stenosis of carotid artery
Location	
Laterality	Left

Precoordination at ontology level

Attribute	Value
Finding Context	
Disorder	Stenosis of the Left Carotid Artery
Location	
Laterality	

Equivalent representations, no commitment to existence of pathologic entity

RecordEntry and

(**isAbout** **only** (*Situation* and
(**includes** some (*LivingHuman* and
(**bearerOf** some *SubjectOfRecordRole*) and
(**locusOf** some (*Stenosis* and
(**hasLocus** some (*CarotidArtery* and
bearerOf some *LeftLaterality*))))))))))

*"Mention of stenosis of right carotid in
a patient's health record"*



equivalent

StenosisOfLeftCarotidArtery equivalent
Stenosis and (**hasLocus** some
(*CarotidArtery* and
bearerOf some *LeftLaterality*))

RecordEntry and (**isAbout** **only** (*Situation* and
(**includes** some (*LivingHuman* and
(**bearerOf** some *SubjectOfRecordRole*) and
(**locusOf** some *StenosisOfLeftCarotidArtery*))))))

"Stenosis of the left carotid artery", "known present" vs. "known absent"

Attribute	Value
Finding Context	known present
Disorder	Stenosis
Location	Carotid artery
Laterality	Left

"Mention in the health record that patient has a stenosis of right carotid"

Attribute	Value
Finding Context	known absent
Disorder	Stenosis
Location	Carotid artery
Laterality	Left

"Mention in the health record that patient has no stenosis of right carotid"

"Stenosis of the left carotid artery", "known present" vs. "known absent"

RecordEntry and **isAbout** some *Situation* and
(**isAbout** only (*Situation* and
(**includes** some (*LivingHuman* and
(**bearerOf** some *SubjectOfRecordRole*) and
(**locusOf** some (*Stenosis* and
(**hasLocus** some (*CarotidArtery* and
bearerOf some *LeftLaterality*)))))))))

*"Mention in the
health record
that patient has
a stenosis of
right carotid"*

RecordEntry and **isAbout** some *Situation* and
(**isAbout** only (*Situation* and
(**includes** some (*LivingHuman* and
(**bearerOf** some *SubjectOfRecordRole*) and
not (**locusOf** some (*Stenosis* and
(**hasLocus** some (*CarotidArtery* and
bearerOf some *LeftLaterality*)))))))))

*"Mention in the
health record
that patient has
no stenosis of
right carotid"*

Stenosis of the left carotid artery in family history

RecordEntry and **isAbout** some *Situation* and
(**isAbout** only (*Situation* and
(**includes** some (*LivingHuman* and
(**bearerOf** some *ParentRole*) and
(**locusOf** some (*Stenosis* and
(**hasLocus** some (*CarotidArtery* and
bearerOf some *LeftLaterality*))))))))))

"Mention in the health record that a parent has a stenosis of right carotid"

Querying the ontology

<http://purl.org/steschu/BO2011>

"Possible Disorder of Artery"

Query:

Query (class expression)

```
RecordEntry
and (isAbout only
(Situation
and (includes some
(LivingHuman
and (LocusOf some
(Disorder
and (hasLocus some Artery))))))
```

Execute

Add to ontology

Ancestor classes (5)

● InformationEntity

● QUERIES

☰ RecordEntry

☰ RecordEntryAboutDisorder

● Thing

Super classes (1)

☰ RecordEntryAboutDisorder

Sub classes (1)

☰ RecordEntryAboutStenosisOfCarotid

Descendant classes (7)

☰ ConfirmedRecordEntryAboutStenosisOfCarotid

☰ ConfirmedRecordEntryAboutStenosisOfCarotidOfParent

☰ ConfirmedRecordEntryAboutStenosisOfLeftCarotid_PostCoord

☰ ConfirmedRecordEntryAboutStenosisOfLeftCarotid_PreCoord

☰ RecordEntryAboutStenosisOfCarotid

☰ RecordEntryAboutStenosisOfLeftCarotid_PostCoord

☰ RecordEntryAboutStenosisOfLeftCarotid_PreCoord

Querying the ontology

<http://purl.org/steschu/BO2011>

"No stenosis of Artery"

Query:

Query (class expression)

```
RecordEntry
and (isAbout some
(Situation
and (includes some
(LivingHuman
and (not (LocusOf some
(Stenosis
and (hasLocus some Artery))))))))
```

Execute Add to ontology

Ancestor classes (8)

- ConfirmedRecordEntry
- InformationEntity
- NegatedRecordEntryAboutStenosisOfCarotid
- NegatedRecordEntryAboutStenosisOfLeftCarotid
- QUERIES
- RecordEntry
- RecordEntryAboutNonExistingStenosisOfLeftCarotid
- Thing

Super classes (1)

- NegatedRecordEntryAboutStenosisOfCarotid

Sub classes (1)

- NegatedRecordEntryAboutStenosis

Descendant classes (2)

- NegatedRecordEntryAboutDisorder
- NegatedRecordEntryAboutStenosis

Conclusion

- BioTop top classes and relations sufficient for expressing some important features of clinical information models
- Equivalence between different flavors of encoding can be computed
- Representation of different epistemic states (known present, known absent, mention of)
- Challenges
 - Scalability
 - DL Reasoning

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- DFG, grant agreement JA 1904/2-1, SCHU 2515/1-1 GoodOD (Good Ontology Design).
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