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[purl.org/steschu](http://purl.org/steschu)

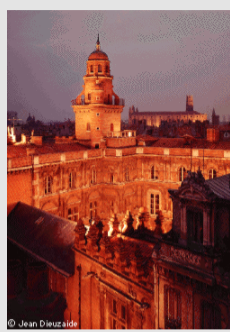


4<sup>e</sup> édition du Symposium sur l'Ingénierie de l'Information Médicale  
Les 23 et 24 Novembre 2017 à Toulouse

Keynote address:

# **Annotating clinical narratives with SNOMED CT**

## **Aspects of Reliability and Semantic Interoperability**



# Eighth International Conference on Principles of Knowledge Representation and Reasoning (KR2002)

April 22-25, 2002; [Toulouse](#), France

Colocated with [AIPS'02](#), with several associated workshops.

Sponsored by [KR, Inc.](#)

KR

The Eighth International Conference on Principles of Knowledge Representation and Reasoning (KR2002) will be held in [Toulouse](#), France from 22 to 25 April 2002. KR2002 will be held in conjunction with [AIPS'02](#).

Explicit representations of knowledge manipulated by inference algorithms provide an important foundation for much work in Artificial Intelligence, including natural language dialogue systems, high level vision, robotics and other knowledge based systems. The KR conferences have established themselves as the leading forum for timely, in-depth presentation of progress in the theory and principles underlying the representation and computational manipulation of knowledge. The traditional very high standard of papers will be maintained at KR2002.

## [Call for Papers](#)

Submission deadline: 11:59pm, Hawaii Time, November 1, 2001

## Information for Authors

Authors should consult <http://www.kr.org/kr-kr02.kr2002instructions.pdf> for instructions on how to format their accepted papers.

## Accepted Papers

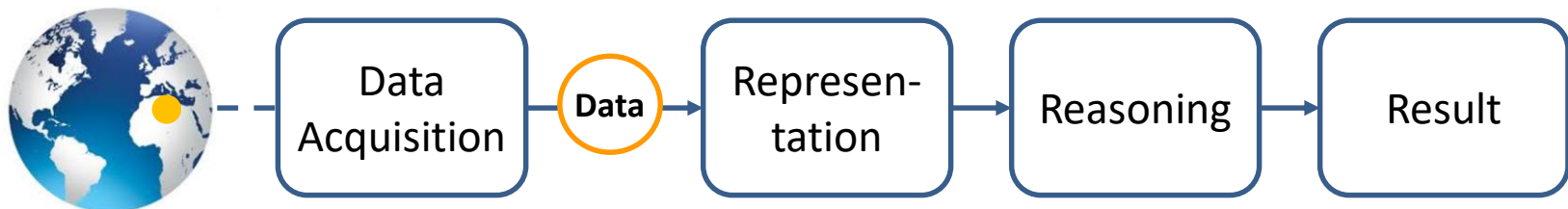
A list of accepted papers is [available](#).

## Associated Workshops

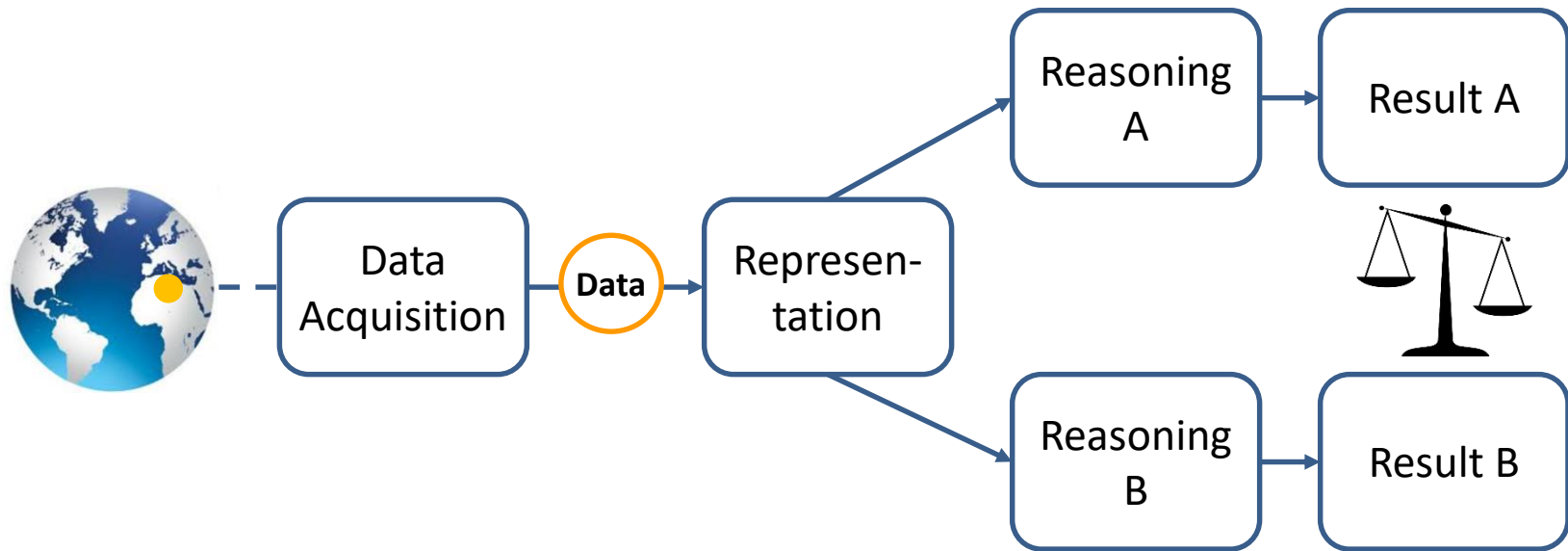
KR2002 will have the following associated workshops:

- the [2002 International Workshop on Description Logics \(DL2002\)](#), which will be held April 19 - 21
- the [Nineth International Workshop on Knowledge Representation meets Databases \(KRDB-2002\)](#), which will be held April 21
- the [Nineth International Workshop on Non-Monotonic Reasoning \(NMR2002\)](#), which will be held April 19 - 21
- the [Workshop on Formal Ontology, Knowledge Representation and Intelligent Systems for the World Wide Web](#), which will be held April 19 - 20
- the [Workshop on Topics for Agent Based Systems \(TABS\)](#), which will be held April 21

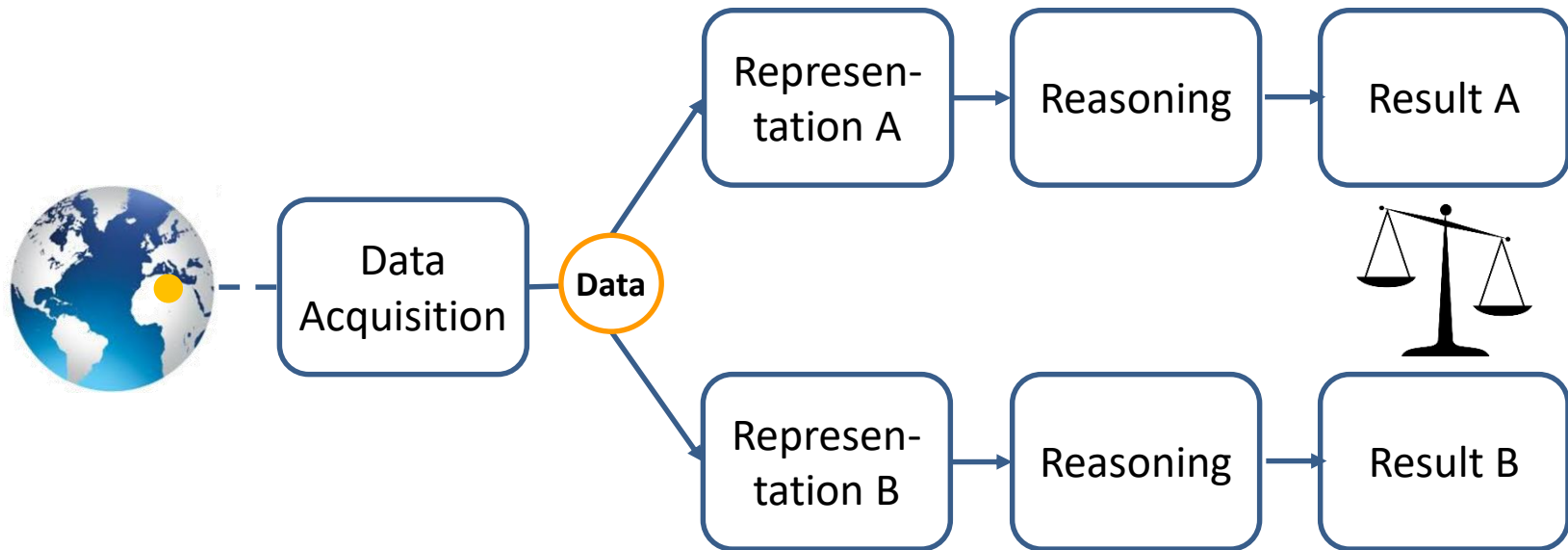
# Typical information engineering workflow



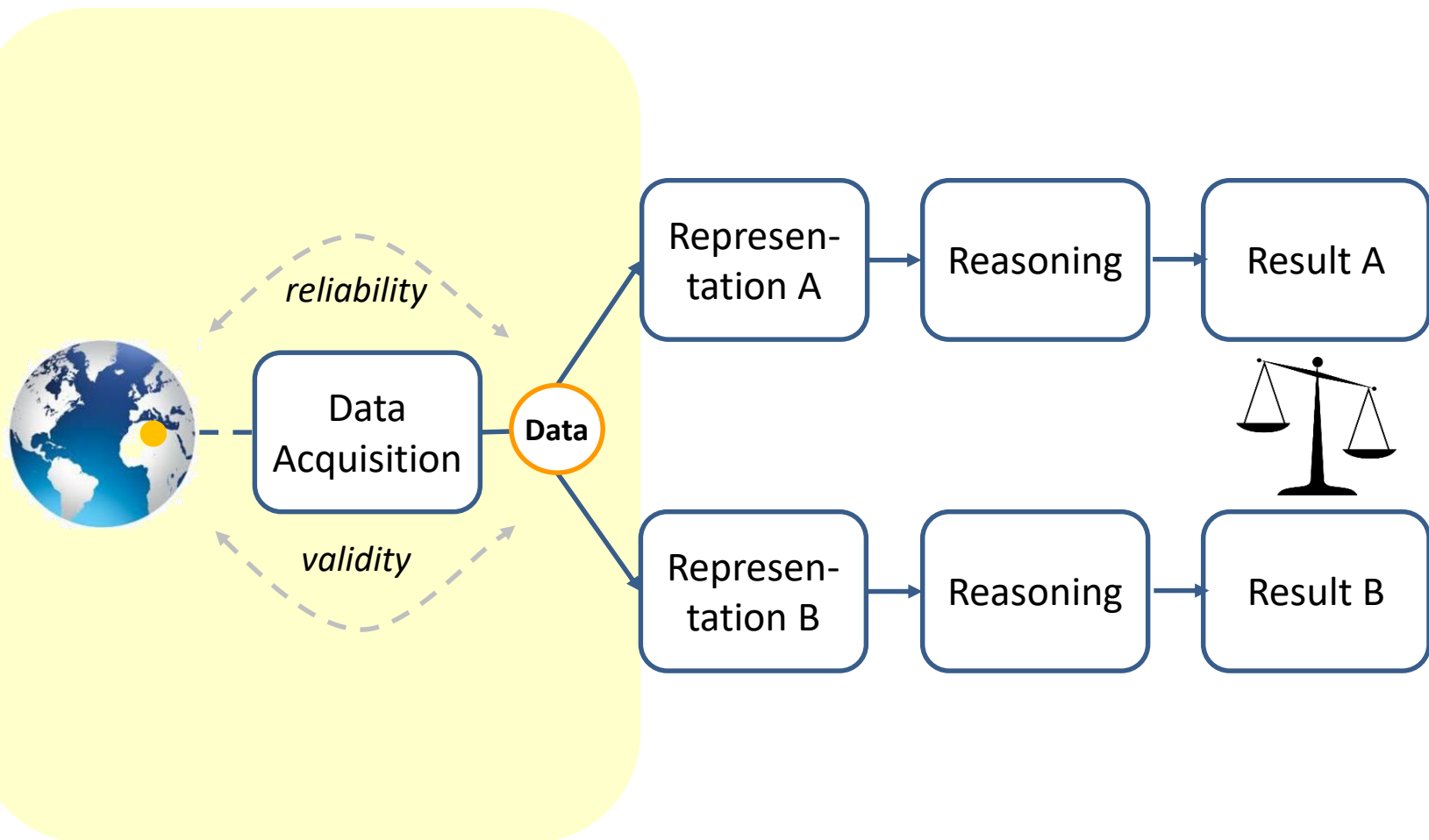
# Typical information engineering workflow



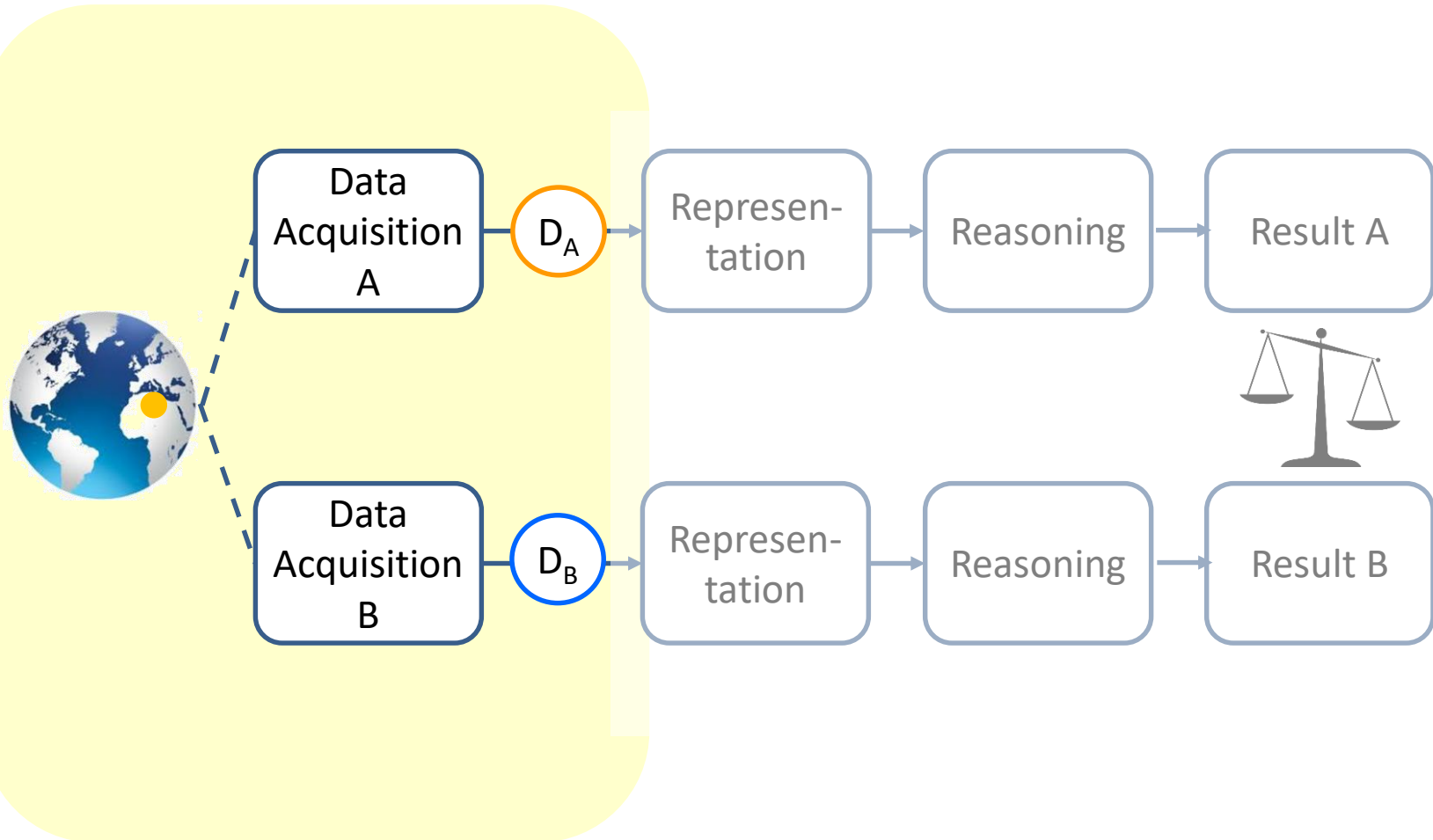
# Typical information engineering workflow



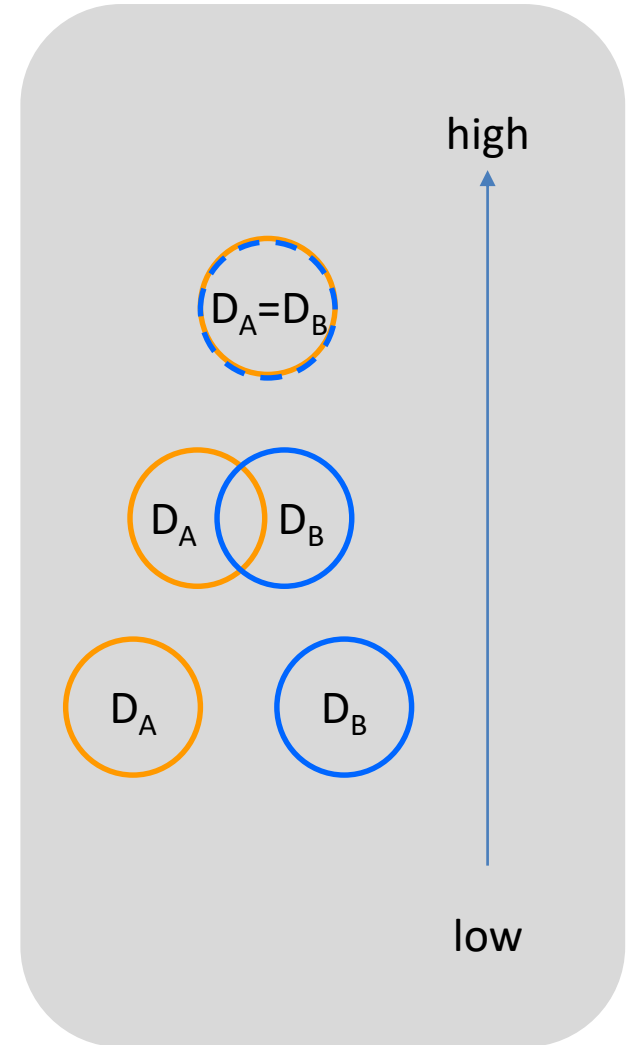
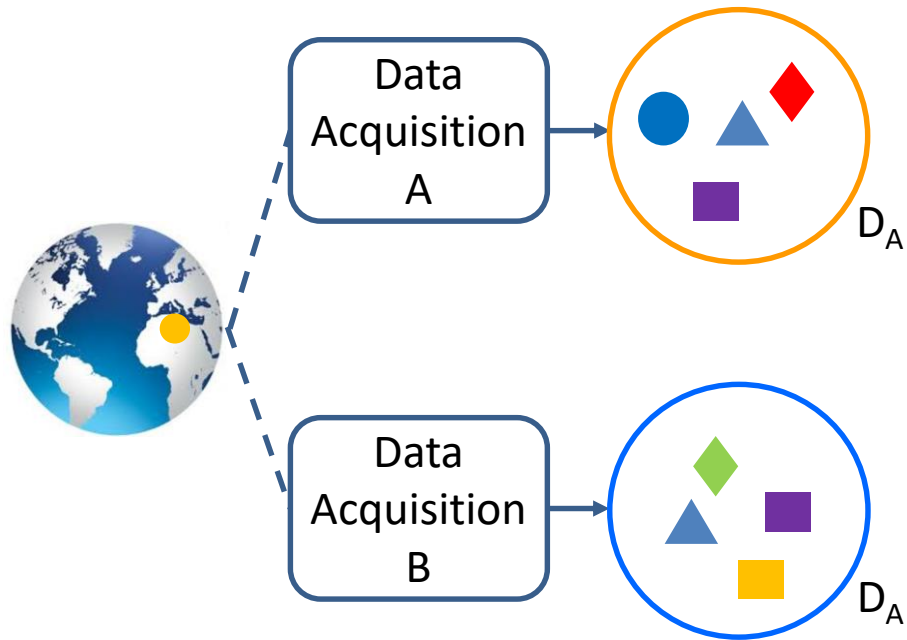
# Typical information engineering workflow



# Focus on data acquisition for information engineering

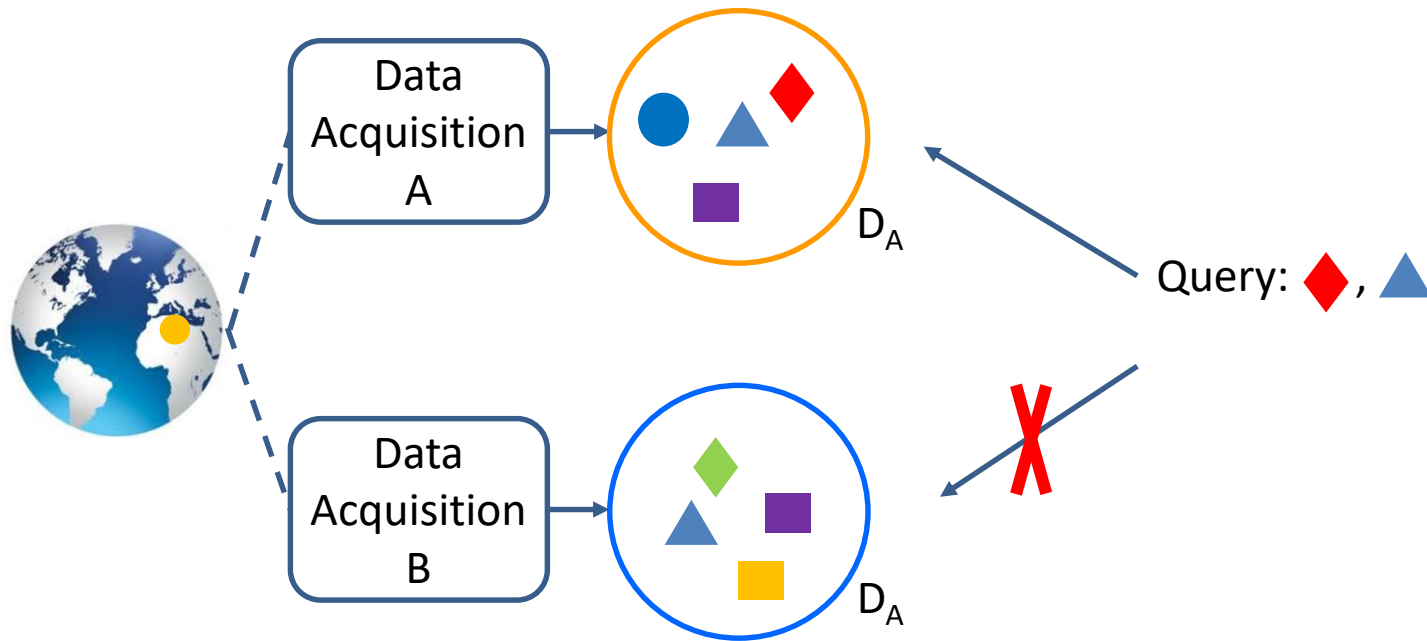


# Data reliability $\rightarrow$ Data interoperability





# Data reliability $\rightarrow$ Data interoperability



# Focus of the talk

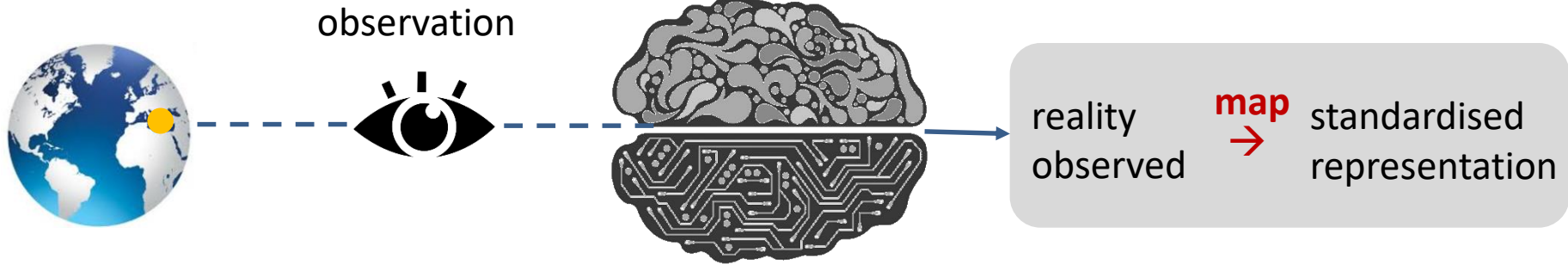
- Analysis of coded extracts from clinical texts
- Inter-annotator agreement (reliability)
- Reasons for inter-annotator disagreement
- Discussion: how to improve agreement

- Assumption:  
better data reliability  
-> better semantic interoperability of clinical data

# Annotating clinical narratives with SNOMED CT

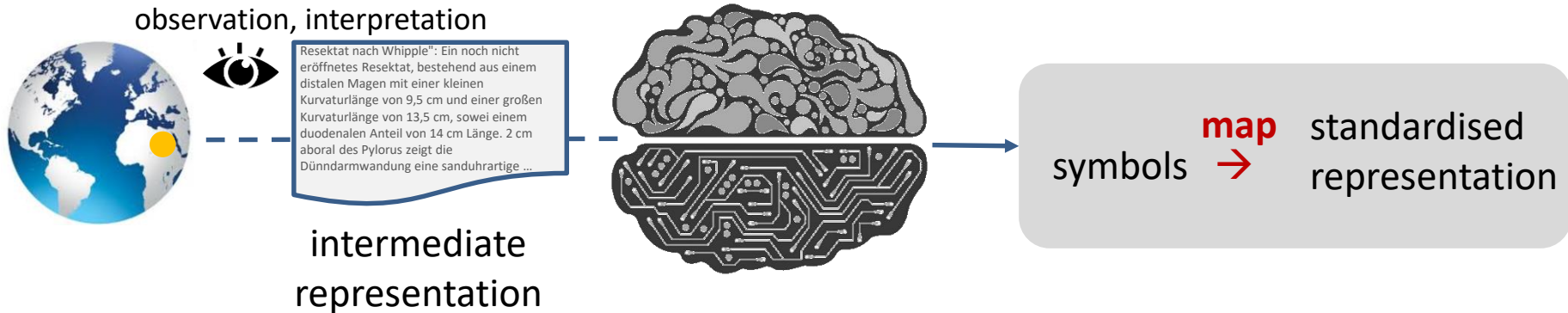
# Annotating clinical narratives with SNOMED CT

## Coding



Vocabularies  
Ontologies

## Annotation



# Annotating clinical narratives with SNOMED CT

Huge clinical  
reference  
terminology

eHealth standard,  
maintained by  
transnational SDO

~300,000  
"concepts"

representable as  
OWL EL

SNOMED CT

preferred terms  
and synonyms in  
several languages

(quasi-)  
ontological  
definitional and  
qualifying axioms

multiple  
hierarchies

covers disorders,  
procedures, body parts,  
substances, devices,  
organisms, qualities...

# Annotation: Sources of complexity

Resektat nach Whipple": Ein noch nicht eröffnetes Resektat, bestehend aus einem distalen Magen mit einer kleinen Kurvaturlänge von 9,5 cm und einer großen Kurvaturlänge von 13,5 cm, sowie einem duodenalen Anteil von 14 cm Länge. 2 cm aboral des Pylorus zeigt die Dünndarmwandung eine sanduhrartige Stenose. Im Magen- und Duodenallumen reichlich zähflüssiger Schleim, sanguinolent;

map

SNOMED CT

## Human language

- words, multiword terms
- syntactic structures
- relations at various levels

## Clinical language

- Compact
- Paragrammatical
- Context-dependent

Best text span to annotate?  
Naïve or analytic annotation?

## Ontology

- classes
- relations
- logical constructors
- axioms

## Terminology

- concepts
- preferred terms
- synonyms
- definitions

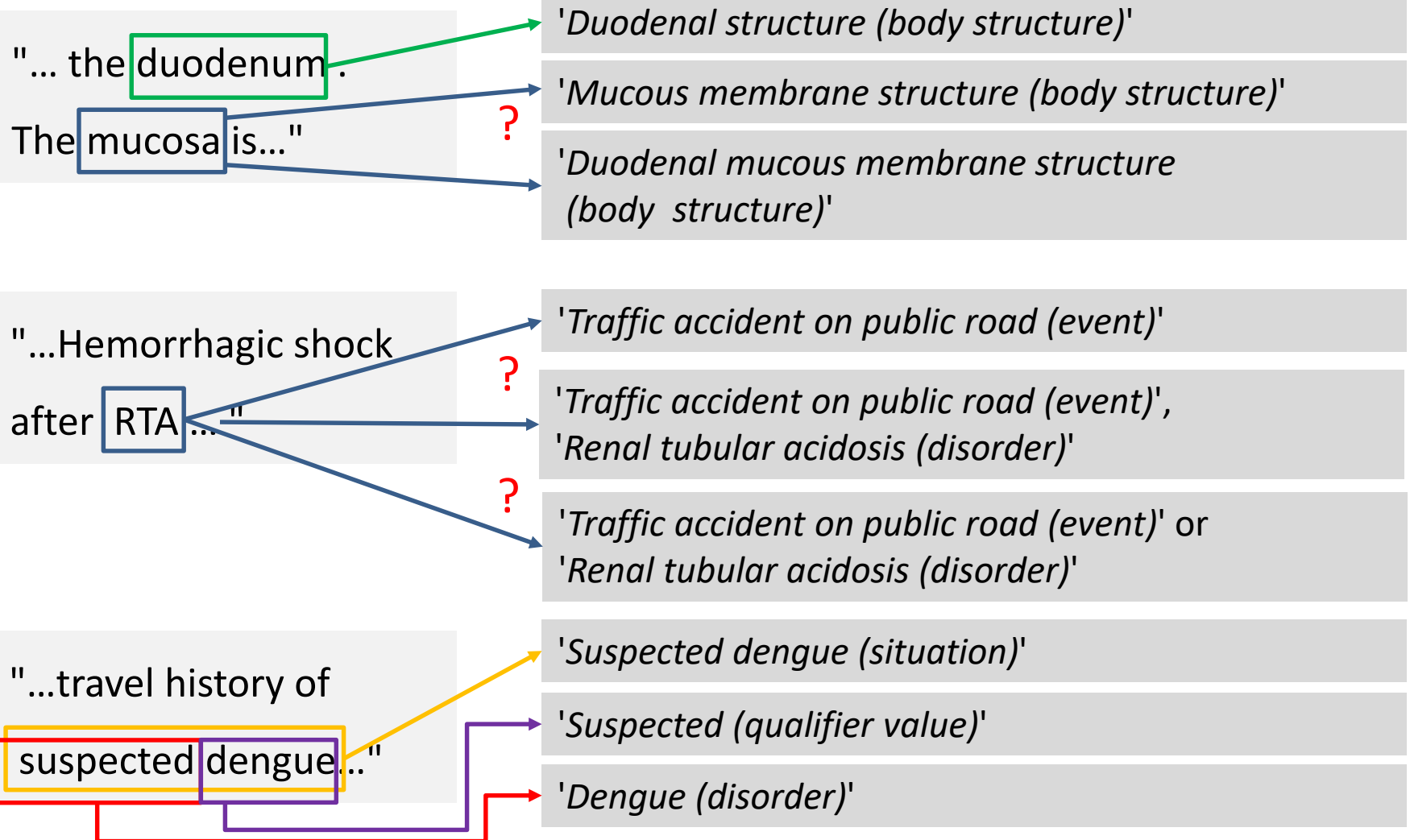
## SNOMED CT

- Ill-defined concepts
- Similar concepts
- Pre-coordination vs. post-coordination
- Complex annotations (> 1 concept / term)

# Examples

## Clinical text

## SNOMED CT concepts (FSNs)



# Coding / Annotation guidelines

- Examples:

1. German coding guidelines for ICD and OPS, 171 pages
2. Using SNOMED CT in CDA models: 147 pages
3. CHEMDNER-patents: annotation of chemical entities in patent corpus: annotation manual 30 pages
4. CRAFT Concept Annotation guidelines: 47 pages
5. Gene Ontology Annotation conventions: 7 pages

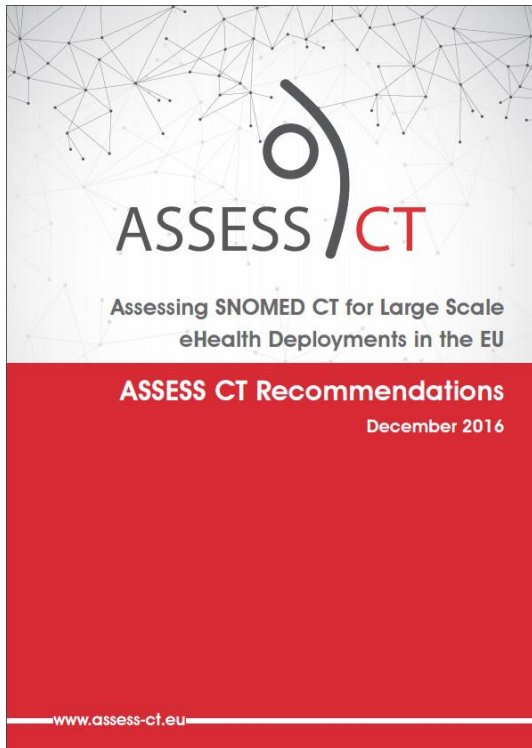
- Complex rule sets, requiring intensive training

1. [http://www.dkgev.de/media/file/21502.Deutsche\\_Kodierrichtlinien\\_Version\\_2016.pdf](http://www.dkgev.de/media/file/21502.Deutsche_Kodierrichtlinien_Version_2016.pdf)
2. <http://www.snomed.org/resource/resource/249>
3. [http://www.biocreative.org/media/store/files/2015/cemp\\_patent\\_guidelines\\_v1.pdf](http://www.biocreative.org/media/store/files/2015/cemp_patent_guidelines_v1.pdf)
4. [http://bionlp-corpora.sourceforge.net/CRAFT/guidelines/CRAFT\\_concept\\_annotation\\_guidelines.pdf](http://bionlp-corpora.sourceforge.net/CRAFT/guidelines/CRAFT_concept_annotation_guidelines.pdf)
5. <http://geneontology.org/page/go-annotation-conventions>



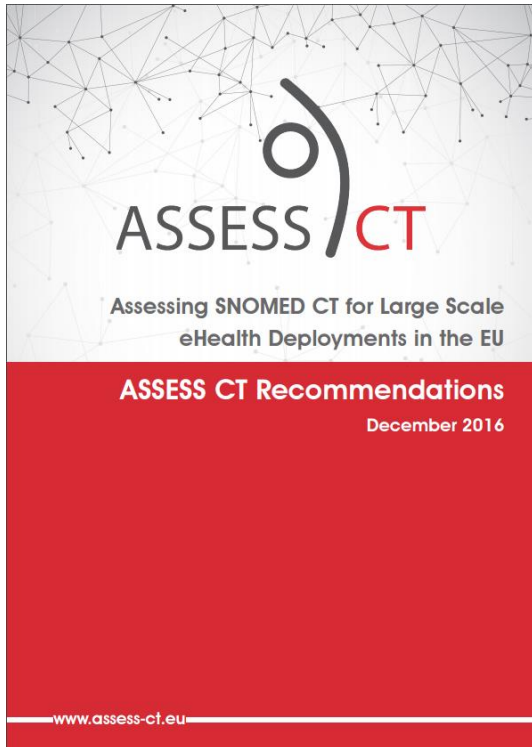
# Annotation experiments in ASSESS-CT

# Annotation experiments in ASSESS-CT



- EU support action on the fitness of SNOMED CT as a EU core reference terminology

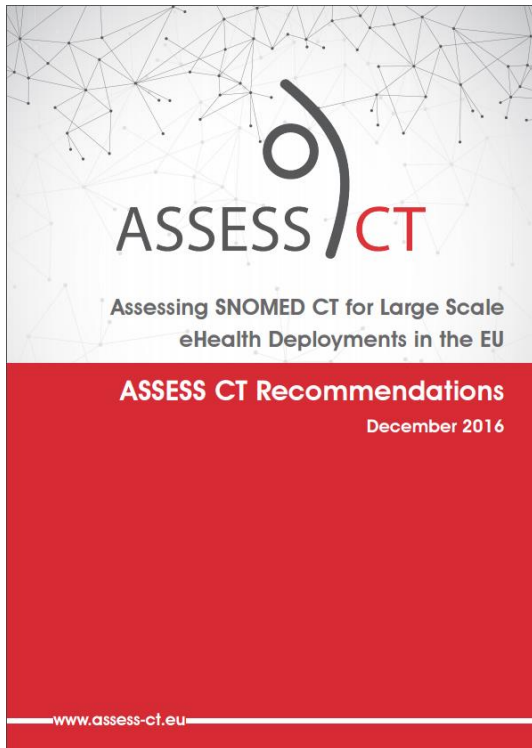
# Annotation experiments in ASSESS-CT



- EU support action on the fitness of SNOMED CT as a EU core reference terminology
- Domain experts annotate 60 samples of clinical documents with SNOMED CT

Nitroglycerin pump spray as required	387404004;385074009;225761000
Amantadine bds	372763006;229799001
Allopurinol 300 ½ tablet every other day (last dose on 20091130)	387135004;385055001;225760004
Mefenamic acid 500 mg up to 3x daily for pain in conjunction with	387185008;258684004;229798009;22253000
simultaneous administration of a drug to protect the stomach e. g.	79970003;416118004;373517009;69695003
Pantoprazole 40mg.	395821003;258684004
Torsemide bds	318034005;229799001
Melperone 50 mg p. m.	442519006;258684004;422133006
§ 7 Intact teeth are in the mouth.	11163003;245543004;123851003
Fractures are visible on the medians of Mandible and Maxilla	263172003;263156006;260528009
the fragments are dislocated.	123735002
Normal mucous membranes in mouth pharynx and on the larynx.	17621005;33044003;71248005
Hyoid and thyroid cartilage are intact.	21387005;52940008;11163003
Fragmental fractures of the two upper vertebrae of the cervical spine.	13321001;207984009;207983003
Otherwise the cervical spine is intact.	122494005;11163003
Oesophagus as well as	262793000;282459005:

# Annotation experiments in ASSESS-CT



- EU support action on the fitness of SNOMED CT as a EU core reference terminology
- Domain experts annotate 60 samples of clinical documents with SNOMED CT

- 1/3 of samples annotated twice
- Support: Webinars, annotation guidelines

Nitroglycerin pump spray as required	387404004;385074009;225761000
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# Principal quantitative results (English)

Concept coverage [95% CI]	SNOMED CT
Text annotations – English	.86 [.82-.88]



Term coverage [95% CI]	SNOMED CT
Text annotations – English	.68 [.64; .70]



Inter annotator agreement Krippendorff's Alpha [95% CI]	SNOMED CT
Text annotations	.37 [.33-.41]



(similar results with alternative annotation task, using non-SNOMED UMLS extract)

# Agreement map: SNOMED annotations



green: agreement – yellow: only annotated by one coder – red: disagreement - white no annotations

# Systematic error analysis

# Systematic error analysis

- Creation of gold standard for SNOMED CT
  - 20 English text samples annotated twice → 208 NPs
  - Analysis of English SNOMED CT annotations by two additional terminology experts
  - Consensus finding, according to pre-established annotation guidelines
- Inspection, analysis and classification of text annotation disagreements
- Presentation of some disagreement cases for SNOMED CT



# Reasons for disagreement

# Human issues

- Lack of domain knowledge / carelessness

Tokens	Annotator #1	Annotator #2	Gold standard
"IV"	'Structure of abductor hallucis muscle (body structure)'	'Abducens nerve structure (body structure)'	'Abducens nerve structure (body structure)'




- Retrieval error (synonym not recognised)

Tokens	Annotator #1	Annotator #2	Gold standard
"Glibenclamide"	'Glyburide (substance)'	–	'Glyburide (substance)'

# Ontology issues (I)

- Logical polysemy ("dot categories")\*

Tokens	Annotator #1	Annotator #2	Gold standard
'Lymphoma'	'Malignant lymphoma (disorder)'	'Malignant lymphoma - category (morphologic abnormality)'	'Malignant lymphoma (disorder)'

 Malignant lymphoma (disorder)  

SCTID: 118600007

118600007 | Malignant lymphoma (disorder) |

- Lymphoma (clinical)
- Malignant lymphoma (clinical)
- Malignant lymphoma (disorder)
- Malignant lymphoma
- Lymphoma




Associated morphology →  
Malignant lymphoma - category

\*Alexandra Arapinis, Laure Vieu: A plea for complex categories in ontologies. Applied Ontology 10(3-4): 285-296 (2015)

# Ontological issues (II)

## ■ Incomplete definitions

Tokens	Annotator #1	Annotator #2	Gold standard
"Motor: normal bulk and tone"	'Skeletal muscle structure (body structure)' <hr/> <i>'Normal (qualifier value)'</i>	'Muscle finding (finding)' <hr/> <i>'Normal (qualifier value)'</i>	'Skeletal muscle normal (finding)'

 Skeletal muscle    
normal (finding)  
SCTID: 298300008  
298300008 | Skeletal muscle  
normal (finding) |  
Skeletal muscle normal  
Skeletal muscle normal (finding)

Finding site → Skeletal muscle  
structure

# Ontological issues (II)

## ■ Incomplete definitions

<b>Tokens</b>	<b>Annotator #1</b>	<b>Annotator #2</b>	<b>Gold standard</b>
"Motor: normal bulk and tone"	<i>'Skeletal muscle structure (body structure)'</i> <hr/> <i>'Normal (qualifier value)'</i>	<i>'Muscle finding (finding)'</i> <hr/> <i>'Normal (qualifier value)'</i>	<i>'Skeletal muscle normal (finding)'</i>

<b>Tokens</b>	<b>Annotator #1</b>	<b>Annotator #2</b>	<b>Gold standard</b>
"Former smoker"	<i>'In the past (qualifier value)'</i> <hr/> <i>'Smoker (finding)'</i>	<i>'History of (contextual qualifier) (qualifier value)'</i> <hr/> <i>'Smoker (finding)'</i>	<i>'Ex-smoker (finding)'</i>

# Ontological issues (III)

## ■ Navigational concepts




<b>Tokens</b>	<b>Annotator #1</b>	<b>Annotator #2</b>	<b>Gold standard</b>
"palpebral fissure"	<i>Finding of measures of palpebral fissure (finding)</i>	<i>Structure of palpebral fissure (body structure)</i>	<i>Measure of palpebral fissure (observable entity)</i>

## ■ Fuzzy, undefined qualifiers

<b>Tokens</b>	<b>Annotator #1</b>	<b>Annotator #2</b>	<b>Gold standard</b>
"Significant bleeding"	<i>'Significant (qualifier value)'</i>	<i>'Severe (severity modifier) (qualifier value)'</i>	<i>'Moderate (severity modifier) (qualifier value)'</i>
	<i>'Bleeding (finding)'</i>	<i>'Bleeding (finding)'</i>	<i>'Bleeding (finding)'</i>

# Interface term (synonym) issues

Tokens	Annotator #1	Annotator #2	Gold standard
"Blood extra- vacation"	'Blood (substance)' <hr/> 'Extravasation (morphologic abnormality)'	'Hemorrhage (morphologic abnormality)'  "extravasation of blood"	'Hemorrhage (morphologic abnormality)'

 Hemorrhage  
(morphologic  
abnormality)  

SCTID: 50960005

50960005 | Hemorrhage  
(morphologic abnormality) |

- Hemorrhage
- Extravasation of blood
- Blood loss
- Bleeding
- Haemorrhage
- Hemorrhage (morphologic  
abnormality)

No attributes

# Interface term (synonym) issues

Tokens	Annotator #1	Annotator #2	Gold standard
"Blood extra- vasation"	<i>'Blood (substance)'</i>	<i>'Hemorrhage (morphologic abnormality)'</i>	<i>'Hemorrhage (morphologic abnormality)'</i>
	<i>'Extravasation (morphologic abnormality)'</i>		
		"extravasation of blood"	

Tokens	Annotator #1	Annotator #2	Gold standard
"anxious"	<i>'Anxiety (finding)'</i>	<i>'Worried (finding)'</i>	<i>'Anxiety (finding)'</i>
		"anxious cognitions"	



# Prevention and remediation of annotation disagreements

# Prevention and remediation of annotation disagreements

- Rationales:
  - More principled SNOMED CT coding of EHR content
  - More principled binding of SNOMED CT codes to clinical models
  - Consistent manual annotations for training corpora and reference standards
  - Improvement of performance of NLP-based annotations

# Preventive measures

# Prevention: annotation processes

- Training with continuous feedback
  - Early detection of inter annotator disagreement triggers guideline enforcement / revision
- Tooling
  - Optimised concept retrieval (fuzzy, substring, synonyms)
  - Guideline enforcement by appropriate tools
  - Postcoordination support (complex syntactic expressions instead of simple concept grouping)

# Prevention: improve SNOMED CT quality

- Fill gaps
  - Add missing equivalence axioms
  - Self-explaining labels, text definitions where necessary
- Preference rules to manage polysemy
- Strengthen ontological foundations
  - Upper-level ontology alignment
  - Better distinction between domain entities and information entities
  - Overhaul problematic subhierarchies, especially qualifiers

# Prevention: improve content maintenance

- Data-driven terminology maintenance
  - Harvest notorious disagreements between annotations from clinical datasets
  - Detect imbalances by analysing concept frequency and co-occurrence between comparable institutions
  - Community processes: crowdsourcing of interface terms by languages, dialects, specialties, user groups (*ASSESS-CT: interface terminologies to be maintained separately from reference terminologies*)

# Remediation of annotation disagreements

# Remediation of annotation disagreements

- Exploit ontological dependencies / implications

Concept A	Concept B	Dependency
'Mast cell neoplasm (disorder)'	'Mast cell neoplasm (morphologic abnormality)'	A subclassOf <b>AssociatedMorphology</b> some B
'Isosorbide dinitrate (product)'	'Isosorbide dinitrate (substance)'	A subclassOf <b>HasActiveIngredient</b> some B
'Palpation (procedure)'	'Palpation - action (qualifier value)'	A subclassOf <b>Method</b> some B
'Blood pressure taking (procedure)'	'Blood pressure (observable entity)'	A subclassOf <b>hasOutcome</b> some B
'Increased size (finding)'	'Increased (qualifier value)'	A subclassOf <b>isBearerOf</b> some B
'Finding of heart rate (finding)'	'Heart rate (observable entity)'	A subclassOf <b>Interprets</b> some B



# Experiment

- Gold standard expansion:
  - Step 1: include concepts linked by attributive relations:
    - $A \text{ subclassOf } \text{Rel some } B$
  - Step 2: include additional first-level taxonomic relations:
    - $A \text{ subclassOf } B$

Language of text sample	Gold standard expansion	F measure
English	no expansion	0.28
	expansion step 1	0.28
	expansion step 2	0.29

- only insignificant improvement
- possibly due to missing relations in SNOMED CT (see "former smoker" and "skeletal muscle normal" examples)
- just a side issue... requires more investigation

# Conclusions

# Conclusions

- Poor agreement hampers SNOMED CT use:
  - Clinical decision support, cohort building, content retrieval, summarisation, analytics,... (but not specific for SNOMED CT → ACCESS CT)
- Prevention & Remediation:
  - Education, tooling, guidelines
  - Large-scale SNOMED CT content and structure improvement
  - High coverage local *interface terminologies*, representing real language of clinicians

# Outlook

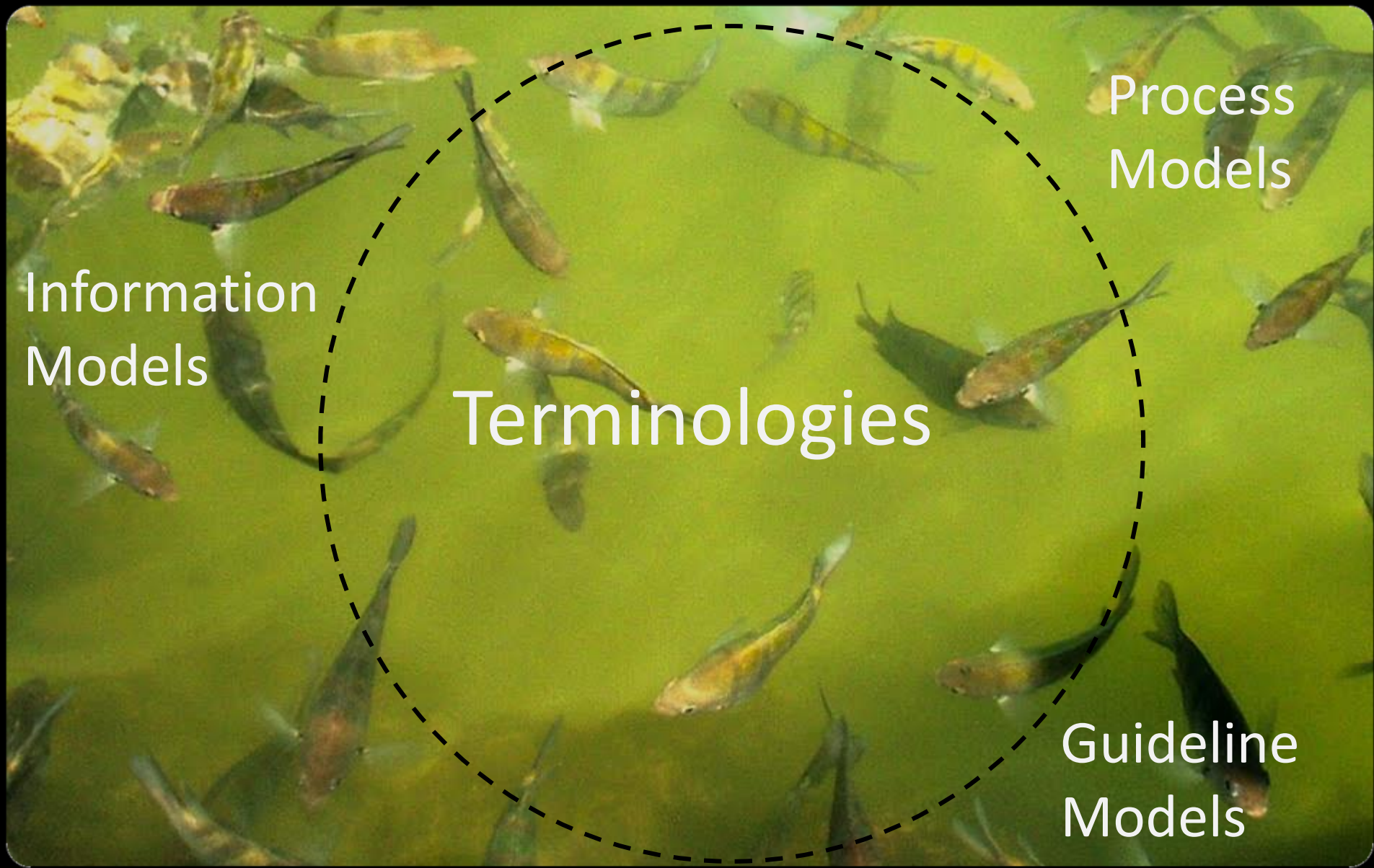
- "Learning systems" for improvement terminology content / structure / tooling.
- "Clinical big data": pooling of non-re-identifiable annotations from multiple institutions
- Community efforts for interface terminology creation and maintenance
- Post processing of SNOMED CT annotations:  
Stream of codes → text knowledge graph

# Thanks for your attention

- Slides will be made accessible at [purl.org/steschu](https://purl.org/steschu)
- Acknowledgements: ASSESS CT team:  
Jose Antonio Miñarro-Giménez, Catalina Martínez-Costa, Daniel Karlsson, Kirstine Rosenbeck Gøeg, Kornél Markó, Benny Van Bruwaene, Ronald Cornet, Marie-Christine Jaulent, Päivi Hämäläinen, Heike Dewenter, Reza Fathollah Nejad, Sylvia Thun, Veli Stroetmann, Dipak Kalra
- Contact: [stefan.schulz@medunigraz.at](mailto:stefan.schulz@medunigraz.at)



# Ecosystem of semantic assets





Information  
Models

Reference  
Terminologies

...describe and standardize a neutral,  
language-independent sense

- The meaning of domain terms
- The properties of the objects that these terms denote
- Representational units are commonly called "concepts"
- RTs enhanced by formal descriptions = "Ontologies"

Guideline  
Models



Information  
Models

Core  
Reference  
Terminology

- Systems of non-overlapping classes in single hierarchies, for data aggregation and ordering.
- aka classifications, e.g. the WHO classifications
- Typically used for health statistics and reimbursement

Aggregation  
Terminologies  
(Classifications)

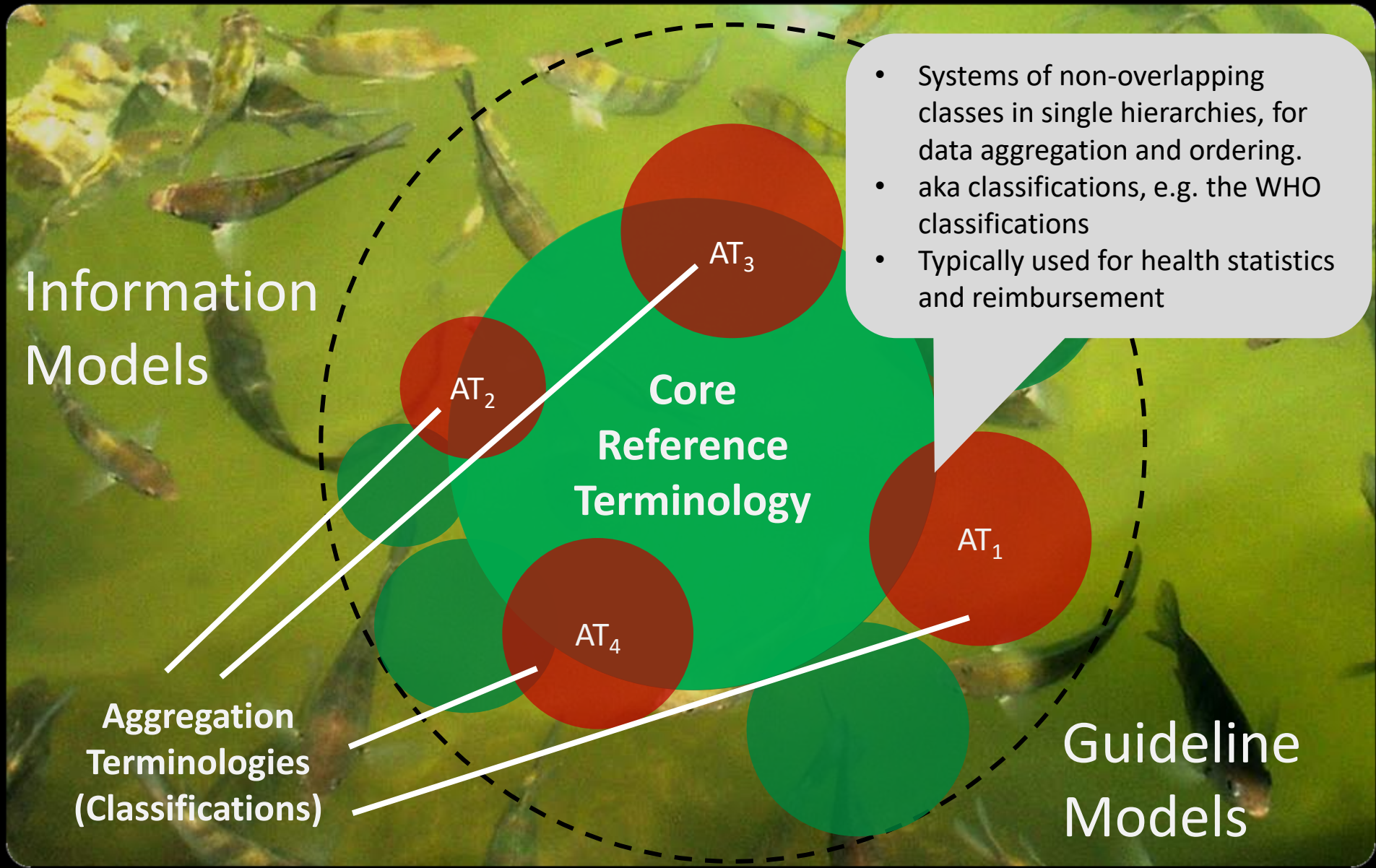
Guideline  
Models

AT<sub>2</sub>

AT<sub>3</sub>

AT<sub>1</sub>

AT<sub>4</sub>



- Reference and aggregation terminologies represent / organize the domain
- They are not primarily representations of language
- They use human language labels as a means to univocally describe the entities they denote, independently of the language actually used in human communication

- Systems of non-overlapping classes in single hierarchies, for data aggregation and ordering.
- aka classifications, e.g. the WHO classifications
- Typically used for health statistics and reimbursement

The diagram features a central green circle labeled "Core Reference Terminology". Surrounding it are four overlapping red circles labeled AT<sub>1</sub>, AT<sub>2</sub>, AT<sub>3</sub>, and AT<sub>4</sub>. A dashed black circle encloses the central green circle and the four red circles. The background is a greenish-yellow image of water with fish. The text "Guideline Models" is located in the bottom right corner.

## Core Reference Terminology

Guideline Models



Information  
Models

**User Interface  
Terminology  
(language specific)**

- Collections of terms used in written and oral communication within a group of users
- Terms often ambiguous.
- Entries in user interface terminologies to be further specified by language, dialect, time, sub(domain), user group.

Guideline  
Models

**User Interface Terminology  
(e.g. Portuguese)**

[chemistry]

"Ca"

"Cálcio"

[oncology]

"Ca"

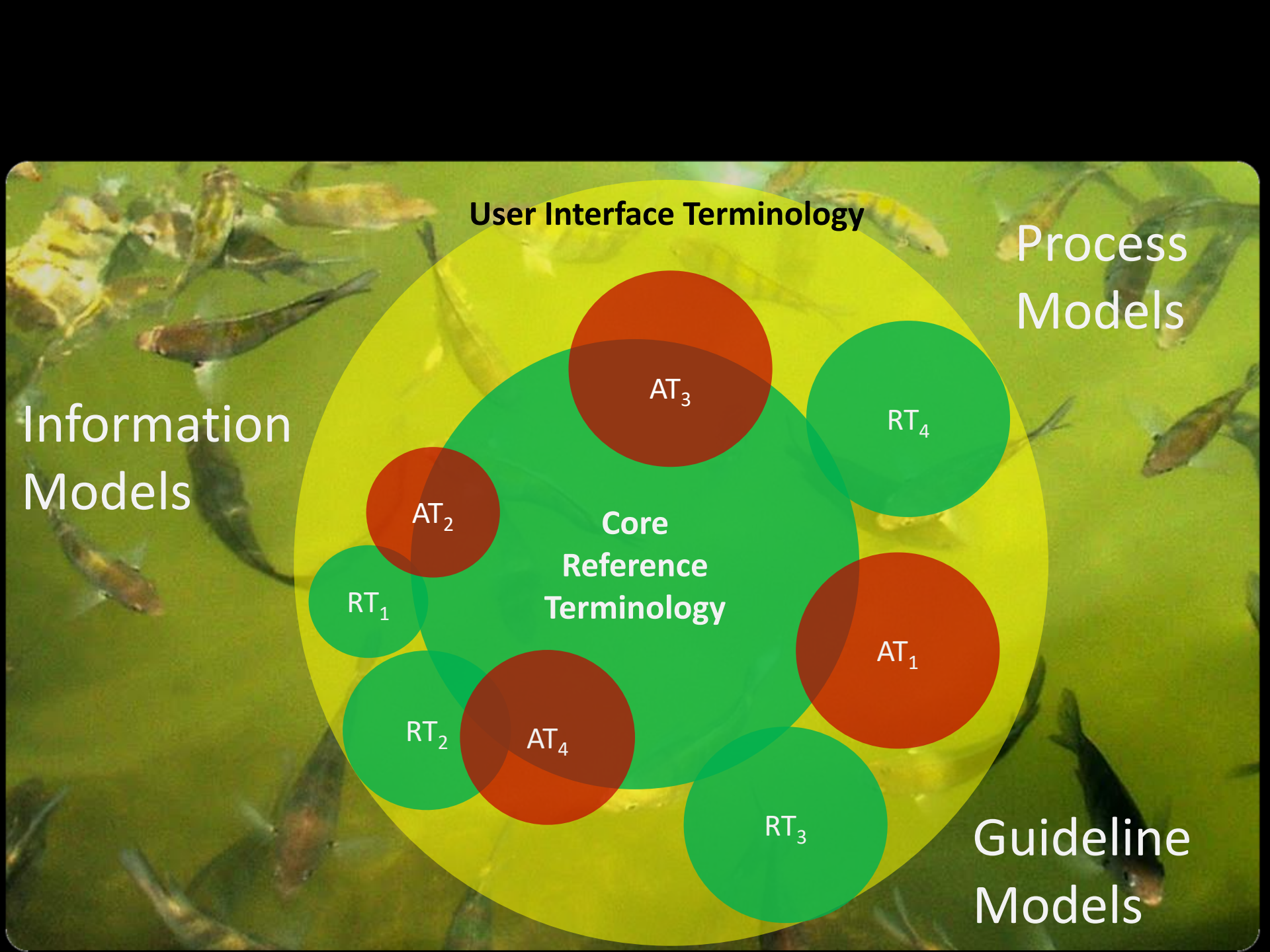
"Câncer"

"Carcinoma"

**Reference  
Terminology**

5540006 |  
Calcium  
(substance) |

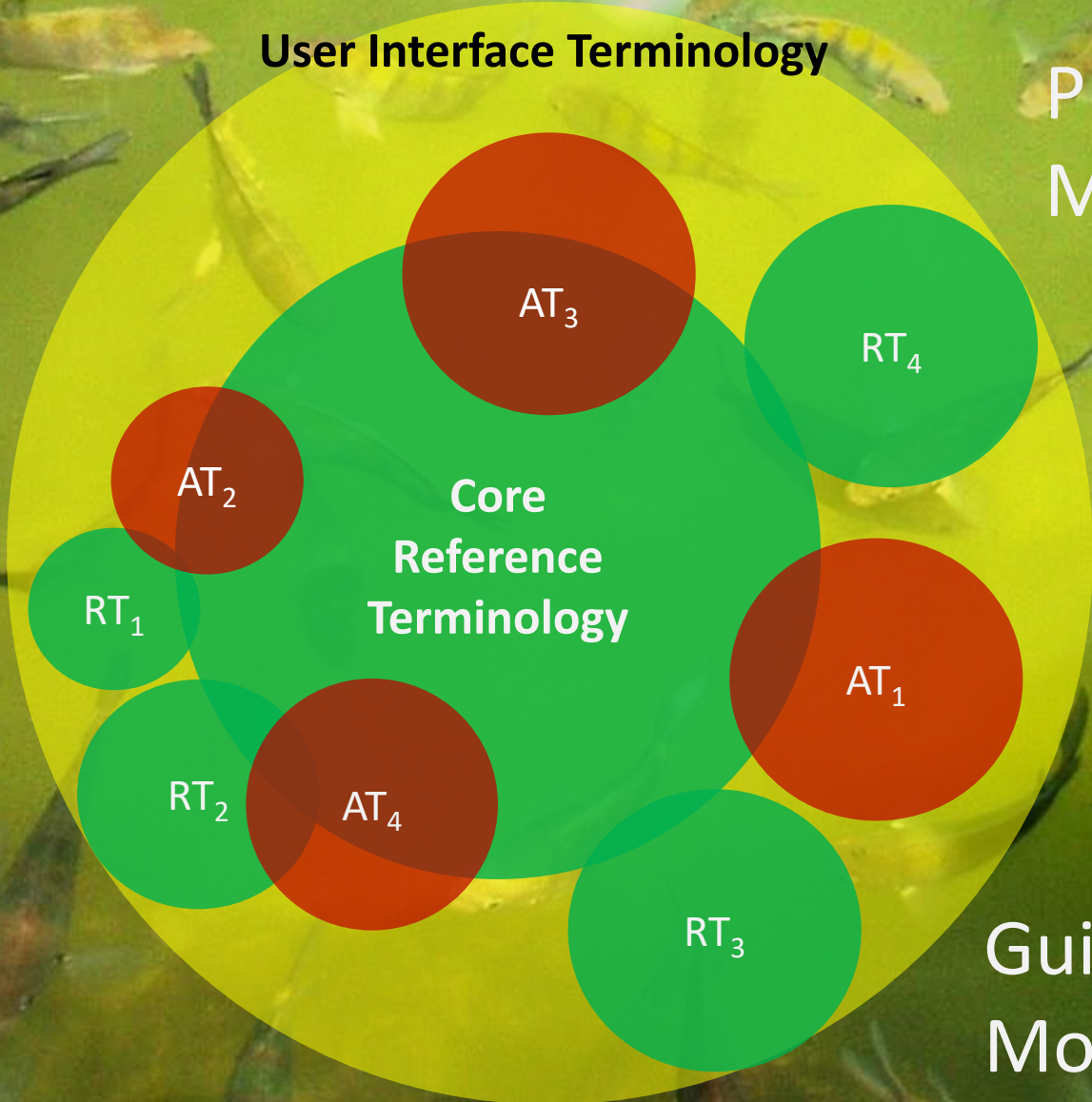
68453008 |  
Carcinoma  
(morphologic  
abnormality) |



**User Interface Terminology**

**Process Models**

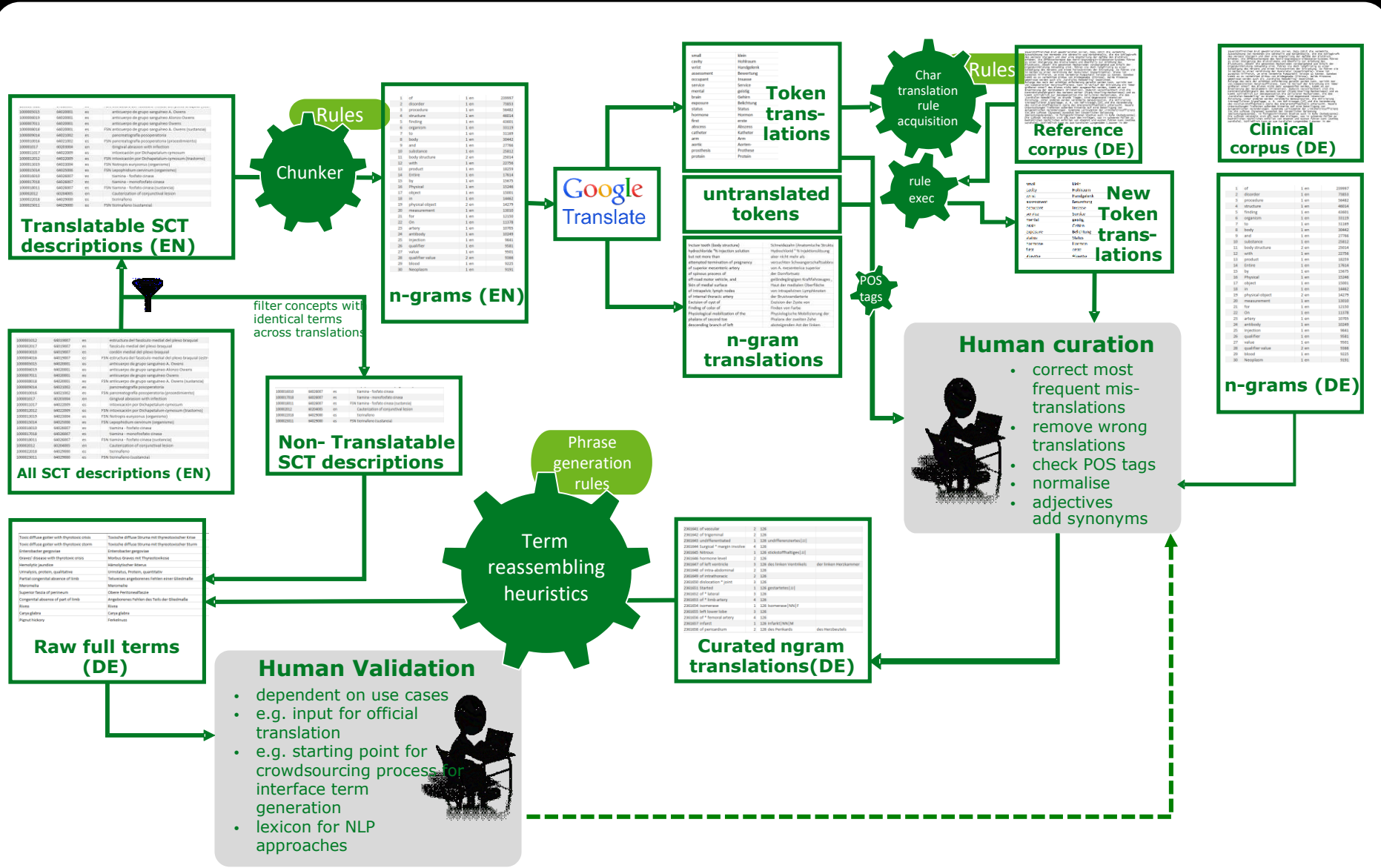
**Information Models**



**Core Reference Terminology**

**Guideline Models**

# MUG-GIT: Creation of German Interface Terminologie for SNOMED CT



# ngram – core vocabulary

vaginal	1	1478	vaginales JJ	Scheiden-	
fluoroscopic guidance	2	1477	Durchleuchtungskontrolle NN F		
disc	1	1476	Scheibe NN F		
lower limb	2	1473	unteres JJ Extremität NN F	Bein NN N	
brain	1	1468	Gehirn NN N	Hirn NN N	Encephalon NN N
preparation	1	1464	Zubereitung NN F	Aufbereitung NN F	Präparation NN F
method	1	1463	Verfahren NN N	Methode NN F	
of bone	2	1462	des Knochens	_Knochen_	
Red	1	1455	rotes JJ		
Monitoring	1	1453	Überwachung NN F	Monitoring NN N	
Computed	1	1453	berechnetes JJ	Computer-	
phalanx	1	1449	Phalanx NN F		
subsp.	1	1449			
anastomosis	1	1447	Anastomose NN F	Anastomosierung NN F	
vessel	1	1446	Blutgefäß NN N	Gefäß NN N	
Computed tomography	2	1443	Computertomographie NN F		
uterus	1	1436	Uterus NN M	Gebärmutter NN F	
difficulty	1	1432	Schwierigkeit NN F		
elbow	1	1429	Ellbogen NN M	Cubitus NN M	Ellbogengelenk NN N
high	1	1429	hohes JJ		
food	1	1423	Lebensmittel NN N	Speise NN F	Nahrungsmittel NN N
Observation	1	1423	Beobachtung NN F		
using fluoroscopic	2	1422			
unable	1	1421	unfähiges JJ		
Peripheral	1	1419	peripheres JJ		
unable to	2	1418	unfähig zu		
Vascular	1	1417	vaskuläres JJ	Gefäß-	
using fluoroscopic guidance	3	1416	mit Durchleuchtungskontrolle		
Benign neoplasm	2	1415	gutartiges JJ Neubildung NN F	gutartiges JJ Neoplasie NN F	benignes JJ Neoplasie NN F

# Machine-generated Interface terms

20170315_240011_002	126952004	Neoplasm of brain	Gehirneubildung
20170315_240011_003	126952004	Neoplasm of brain	Neubildung des Hirns
20170315_240011_004	126952004	Neoplasm of brain	Hirnneubildung
20170315_240011_005	126952004	Neoplasm of brain	Neoplasie des Gehirns
20170315_240011_006	126952004	Neoplasm of brain	Gehirneoplasie
20170315_240011_007	126952004	Neoplasm of brain	Neoplasie des Hirns
20170315_240011_008	126952004	Neoplasm of brain	Hirnneoplasie
20170315_240011_009	126952004	Neoplasm of brain	Neoplasma des Gehirns
20170315_240011_010	126952004	Neoplasm of brain	Gehirneoplasma
20170315_240011_011	126952004	Neoplasm of brain	Neoplasma des Hirns
20170315_240011_012	126952004	Neoplasm of brain	Hirneoplasma
20170315_241010_001	126953009	Neoplasm of cerebrum	Neubildung des Großhirns
20170315_241010_002	126953009	Neoplasm of cerebrum	Neoplasie des Großhirns
20170315_241010_003	126953009	Neoplasm of cerebrum	Neoplasma des Großhirns
20170315_242015_001	126954003	Neoplasm of frontal lobe	Neubildung des Frontallappens
20170315_242015_002	126954003	Neoplasm of frontal lobe	Neubildung des Lobus frontalis
20170315_242015_003	126954003	Neoplasm of frontal lobe	Neoplasie des Frontallappens
20170315_242015_004	126954003	Neoplasm of frontal lobe	Neoplasie des Lobus frontalis
20170315_242015_005	126954003	Neoplasm of frontal lobe	Neoplasma des Frontallappens
20170315_242015_006	126954003	Neoplasm of frontal lobe	Neoplasma des Lobus frontalis
20170315_243013_001	126955002	Neoplasm of temporal lobe	Neubildung des Temporallappens
20170315_243013_002	126955002	Neoplasm of temporal lobe	Neubildung des Lobus temporalis
20170315_243013_003	126955002	Neoplasm of temporal lobe	Neoplasie des Temporallappens
20170315_243013_004	126955002	Neoplasm of temporal lobe	Neoplasie des Lobus temporalis
20170315_243013_005	126955002	Neoplasm of temporal lobe	Neoplasma des Temporallappens