**Stefan Schulz**
Daniel Schober
Djamila Raufie
Martin Boeker

Medical Informatics
Research Group
University
Medical Center
Freiburg, Germany

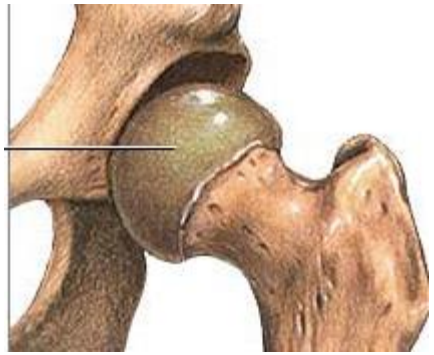
# Pre- and Post-Coordination in Biomedical Ontologies

UNIVERSITÄTS KLINIKUM FREIBURG

# Frege's Principle

- Compositionality: The meaning of a complex expression is determined by its structure and the meanings of its constituents

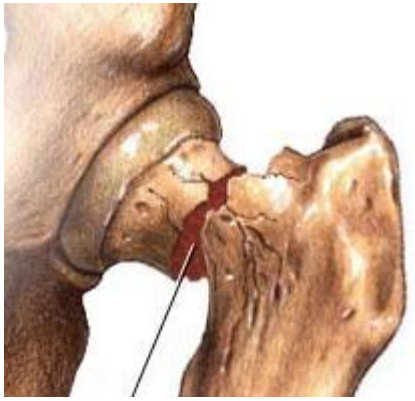
Hip



+

Fracture

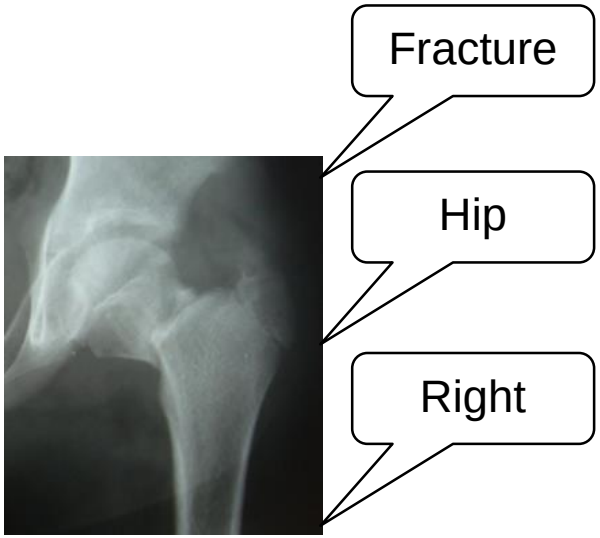
=

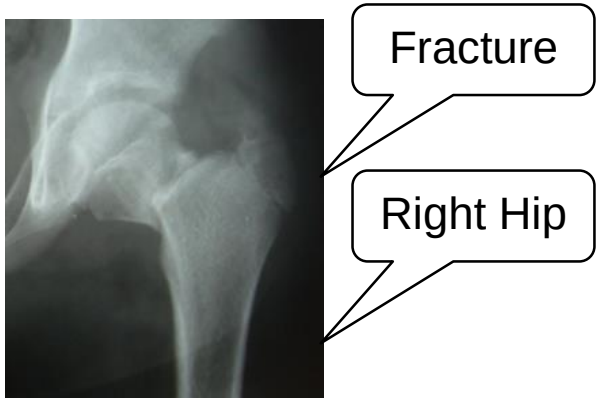


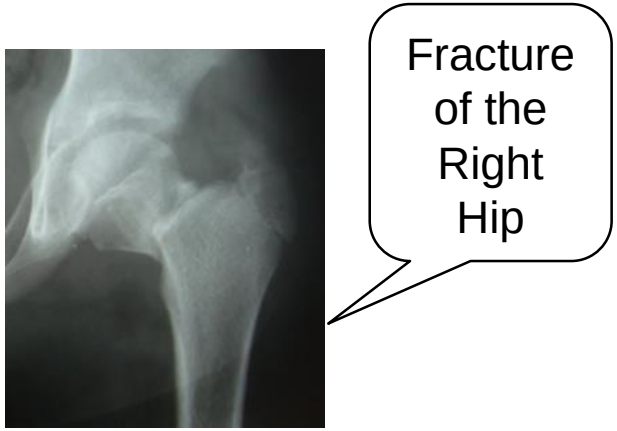
Fracture of the hip

# Degrees of Coordination

Coding, Annotation, using terminologies and ontologies



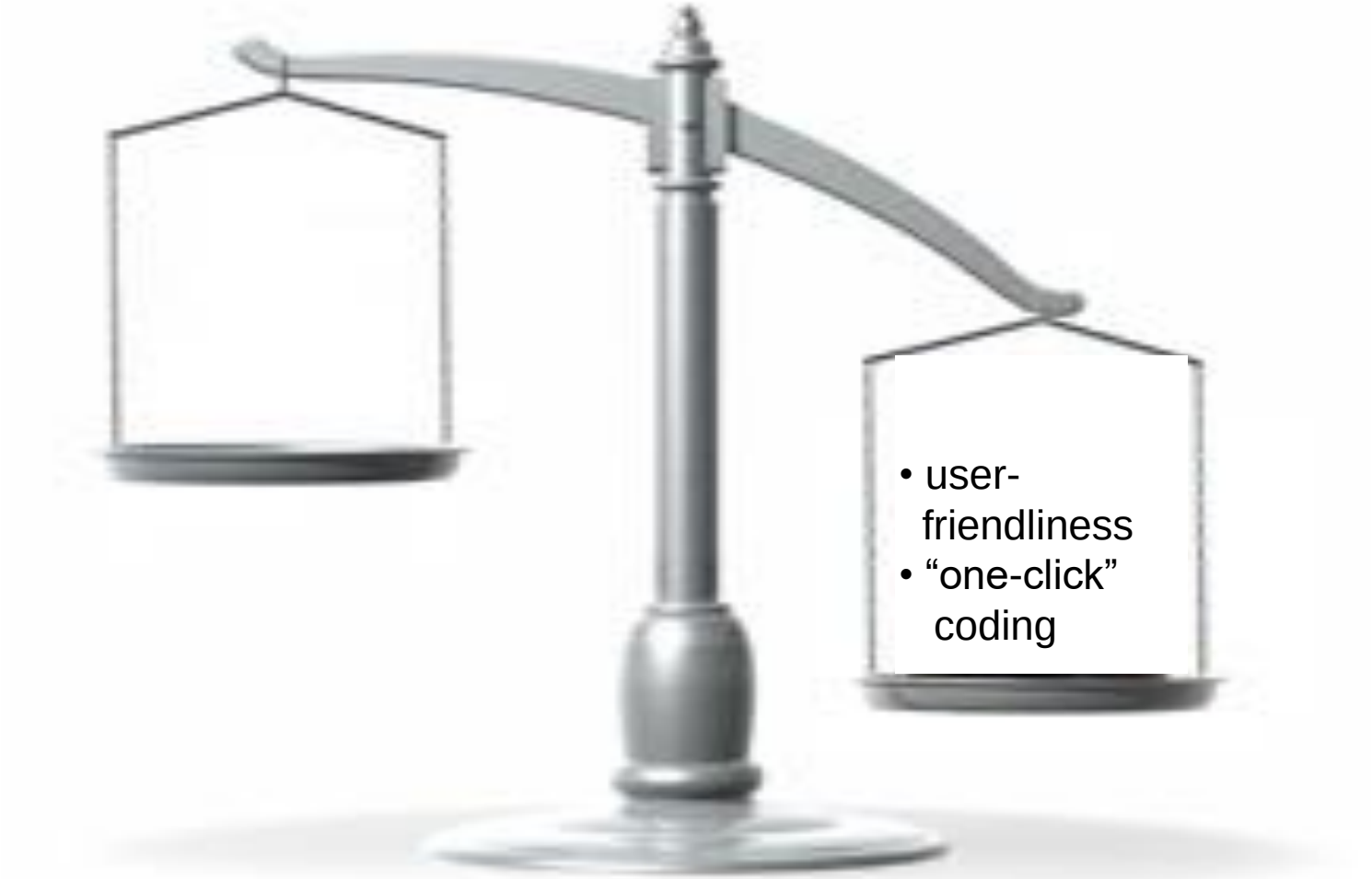




Post-coordination ⟶ Pre-coordination

# Degrees of Coordination


Post-coordination → Pre-coordination

# Degrees of Coordination

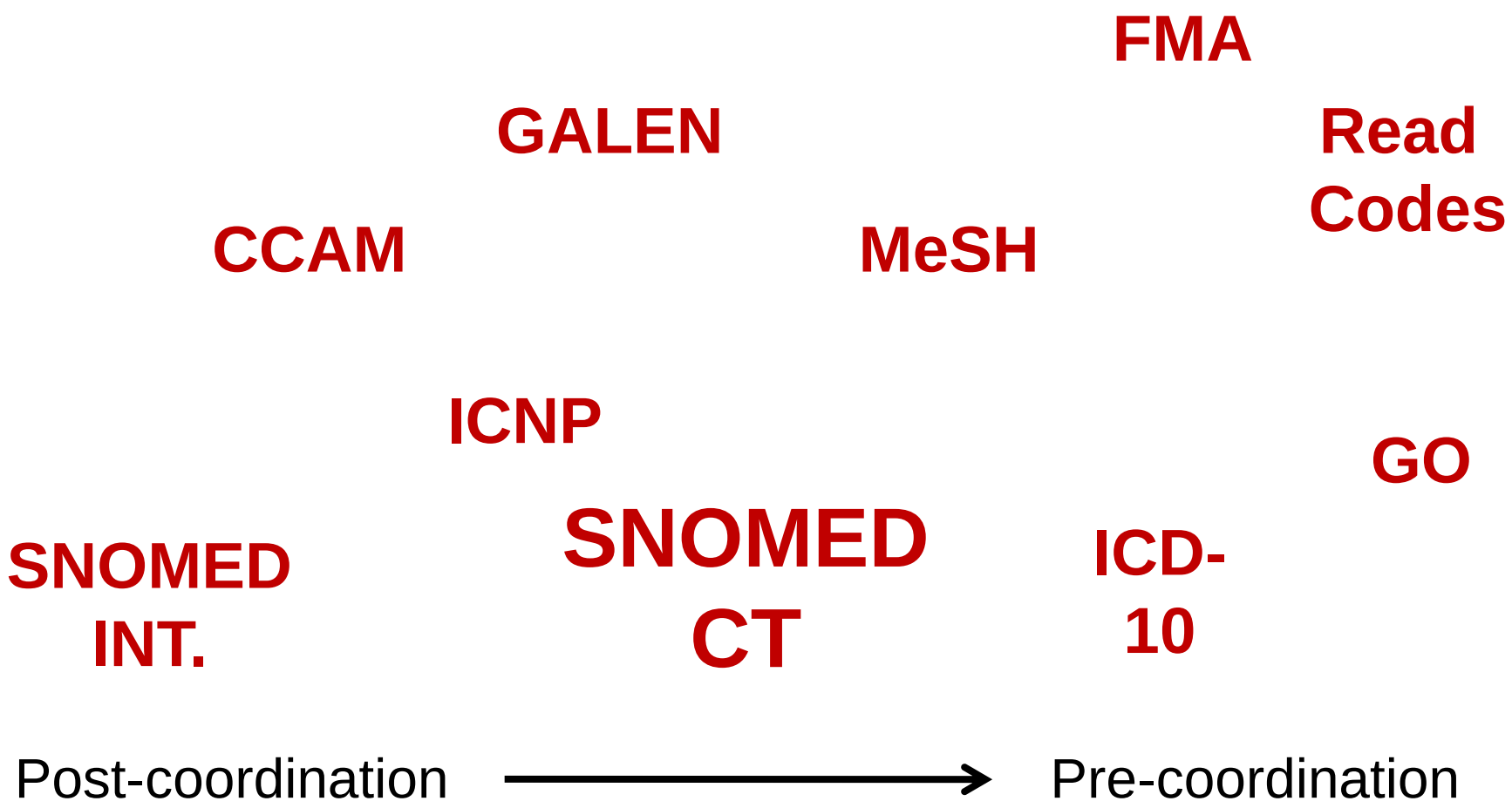



Post-coordination → Pre-coordination

# Limits of precoordination

- Combinatorial explosion
- Example: codes for burns:
  - with 200 different sites
  - with 4 different degrees
  - with / without loss of tissue
  - with / without infection
  - with 5 different mechanisms
- → 16 000 codes
- If also one code for adjacent sites (e.g “burn of wrist and forearm”…)
- → >> 100 000 codes

# Biomedical Terminologies



# Problem with compositional terminologies

| | |
|---|---|
| D5-46210 | Acute appendicitis |

| | |
|---|---|
| D5-46100 | Appendicitis |
| G-A231 | Acute |

| | |
|---|---|
| M-41000 | Acute inflammation |
| G-C006 | In |
| T-59200 | Appendix |

| | |
|---|---|
| G-A231 | Acute |
| M-40000 | Inflammation |
| G-C006 | In |
| T-59200 | Appendix |

- Equivalence between synonymous expressions cannot be automatically checked
- Lack of relations and nesting of expressions: creates ambiguity
- Nonsensical compositions possible

# Solution: simple description logics(OWL-EL)

*Appendicitis* equivalentTo
*Inflammation* and
**hasLocation** some *Appendix*

*AcuteAppendicitis* equivalentTo
*AcuteInflammation* and
**hasLocation** some *Appendix*

*AcuteAppendicitis* equivalentTo
(*Inflammation* and
**hasQuality** some *Acute*) and
**hasLocation** some *Appendix*

*AcuteKidmonia* equivalentTo
(*AcutePneumonia* and
**hasLocation** some *Kidney*)

- Equivalence between synonymous expressions **can** be automatically checked
- Relations and nesting of expression
- Nonsensical compositions still possible

# Observations

- Most modern biomedical ontologies exhibit a mixture of precoordinated classes with classes for postcoordination
- Classification of expressions of different degrees of compositionality supported by inexpressive DL (OWL-EL)
  - equivalence: equivalentTo (≡)
  - subsumption: subClassOf (⊑)
  - conjunction: “and” (⊓)
  - existential restriction: “some” (∃)
- Persisting deficits:
  - user-friendly guidance for post-coordination by constraints and patterns
  - plausibility checking of post-composed expressions relies on users’ domain knowledge
  - knowledge-intensive reasoning services not supported by OWL-EL

# Case study: pneumonia ontology

# Case study: pneumonia ontology

- Anatomical localization:
  - the parts of the lung and its tissues
- Disease course
  - acute or chronic
- Etiological characteristics
  - infections, physical, chemical...
- Pre-existing conditions, of which the pneumonia is a complication
- Environmental characteristics
- Where it was acquired (community or hospital)

# OWL-EL axioms

using BioTop upper domain ontology: http://purl.org/biotop

*Pneumonia* equivalentTo *Inflammation* and

**hasParticipant** some *LungTissue*

*LobalPneumonia* equivalentTo *Pneuonia* and

**hasLocus** some *LungLobe*

*AcutePneumonia* equivalentTo *Pneumonia* and

**bearerOf** some AcutenessQuality

*BacterialPneumonia* equivalentTo *Pneumonia* and

**hasAgent** some BacteriaPopulation

# Limitations

- OWL-EL does not prevent to define, e.g.
  - Pneumomia located in the kidney
  - Pneumomia being simultaneously acute and chronic
  - Pneumonia caused by elephants
  - Pneumonia as a complication of ingrown nail
- Open world semantics + OWL-EL: no constraints
- Needed:
  - Disjoint categories, e.g. for enforcing non-overlapping of toplevel categories, e.g. Pneumonia is a process, therefore it is no material object
  - Allowed values, e.g. caused-by restricted to micro-organisms

# Pneumonia: pre-coordination requirements

- Taxonomic hierarchies:

  *BacterialPneumonia* subClassOf *BacterialInflammation*

- Relation axioms and hierarchies

  TransitiveProperty (**partOf**)

  **partOf** subPropertyOf **hasLocus**

- Mereotopologic axioms

  *Pneumonia* equivalentTo *Inflammation* and
  **hasParticipant** some *LungTissue*

  *LungTissue* subClassOf **partOf** some *Lung*

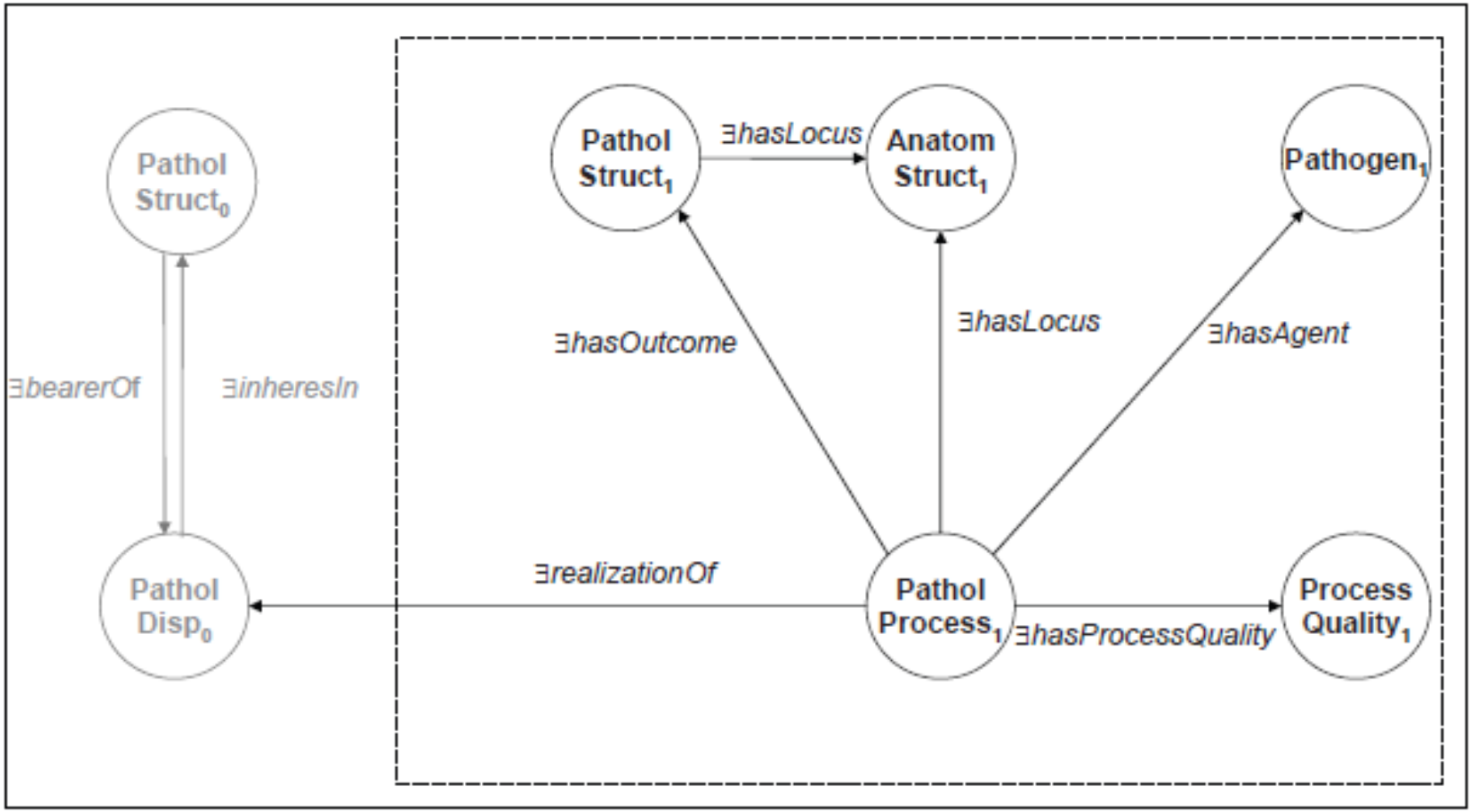
  ---

  *Pneumonia* subClassOf **hasLocus** some *Lung*

# Pneumonia post-coordination requirements

- Support and guide user to compose own post-coordinated compositions
- Post-coordinated expressions to be
  - Valid: allow only meaningful compositions prevent nonsensical coordinations
  - Expressive: enable user to create unambiguous, clearly delineated compositions
  - Reliable: support for compositions that are consistent between different modelers
- Post-coordination needs to
  - restrict users' choices
  - embed coordination axioms, provided by ontology design patterns and upper level ontologies

# Ontology design pattern for infectious diseases



# Sample pattern (I)

- Disease processes can only be located in anatomical regions that have a certain type of tissue:
  - *Pneumonia* subClassOf **hasLocus** only (locusOf some *LungTissue*)
- Tissues only occur in certain body parts / regions
  - *LungTissue* subClassOf **hasLocus** some *Lung*
  - *LungTissue* subClassOf **hasLocus** only (**locusOf** some *Lung*)
- Organs are located in certain regions that do not overlap
  - *Lung* subClassOf **hasLocus** only (**locusOf** some *Thorax*).
  - *Thorax* subClassOf **locusOf** only (not **hasLocus** some (*Abdomen* or *Extremity*)

# Sample patterns (II)

- A secondary disease is a pathological process which is the realization of a pre-existing disposition which inheres in a pathological structure, which exists as congenital disorder or outcome of a former pathological process
  - *Pneumonia* subClassOf only
    **realizationOf** (*PathologicalDisposition* and only
    **inheresIn** (*LungInfarction* or *LungEdema*))
- A disease typically predisposes an organism to develop signs and symptoms
  - *Pneumonia* subClassOf hasOutput some
    (*PathologicalStructure* and **hasLocus** some
    (*Organism* and **bearerOf**
    some (*PathologicalDisposition* and
    only **hasRealization**
    (*Cough* or *Chills* or *Fever*))))

# Additional post-coordination pattern examples

A secondary disease is a pathological process which is the realization of a **pre-existing disposition** which inheres in a pathological structure, which exists as congenital disorder or outcome of a former pathological process

lung infarction or lung edema as a cause of pneumonia (second disease)

→

Pneumonia subClassOf only

realization-of (PathologicalDisposition and only inheresIn.(LungInfarction or LungEdema))

A pneumonia process predisposes an organism to develop **signs and symptoms** like fever, chills, or cough

Pneumonia subClassOf hasoutput.some (PathologicalStructure and hasLocus Organism and bearer-of some (PathologicalDisposition and only has-realization. Cough or Chills or Fever))

# Conclusions

- Pre-coordinated ontologies:  subsumption, class inclusion, equivalence, existential restrictions (OWL-EL)
- Support for post-coordination user guidance: value restrictions, negation, disjunction (OWL RL)
- Sources:
  - Expressive top level ontologies
  - Ontology design patterns
- Problem: expressiveness → lack of scalability
- Possible solutions: use EL functionality only for reasoning, additional RL functionality for e.g. GUI support, use weak negation, new reasoners…

# References / Acknowledgements

- Pneumonia.owl: http://purl.org/biotop/src/pneumonia.zip
- BioTop.owl: http://purl.org/biotop/biotop.owl
- DebugIT project (EU FP7): http://www.debugit.eu