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IHTSDO delivering  
SNOMED CT®  
the global clinical terminology



# -SemanticHealthNet- A Semantic Infrastructure Towards Semantic Interoperability

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SNOMED CT Implementation Showcase

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# Introduction - SemanticHealthNet (SHN) Project

- **3 years EC NoE Project (2012-2015)**
- **Global mission:**
  - Develop and test approaches to **improve semantic interoperability** of health data
  - Create a **virtual organization** to sustain semantic interoperability developments and their adoption across Europe
- **Workpackage 4 mission:**
  - Provide an intermediate semantic layer able to **deal with the unavoidable heterogeneity** which arises when clinical information is represented across or within the same medical domain



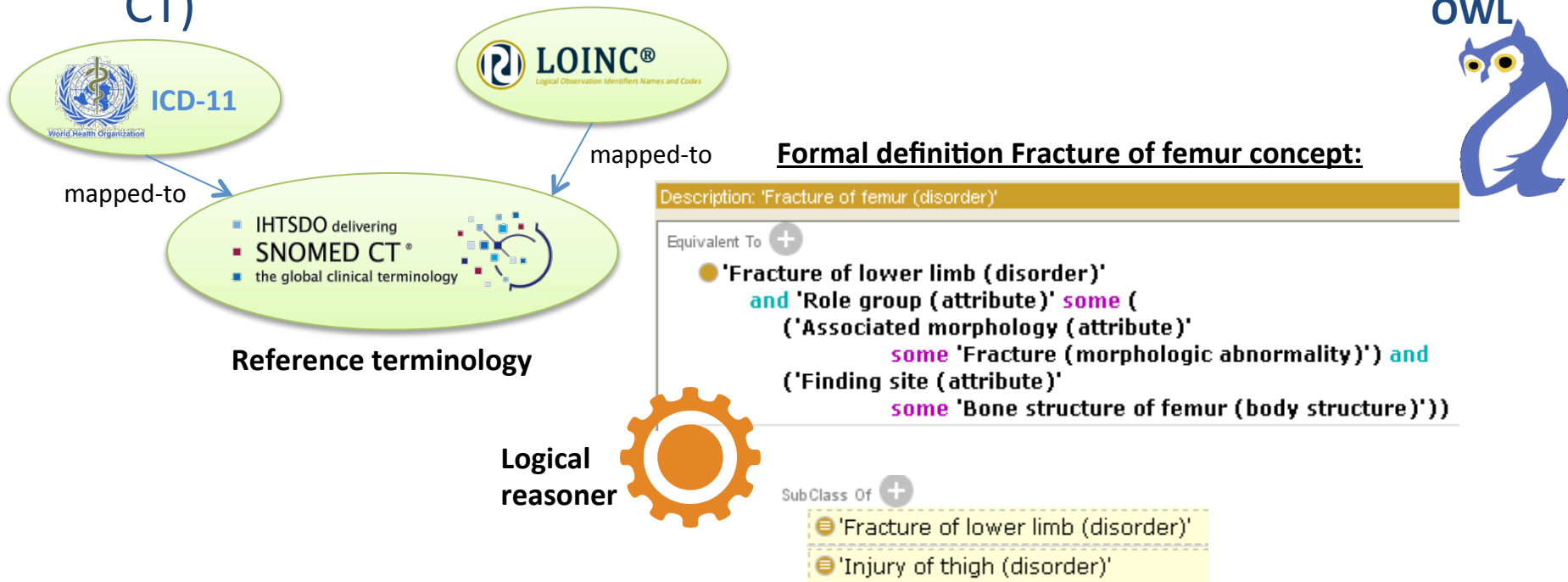
# Workpackage 4 Basic Assumptions

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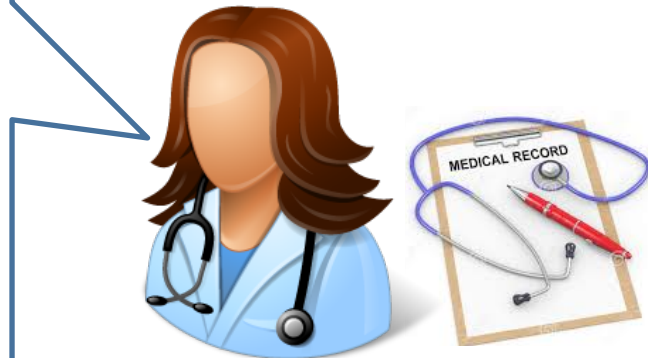
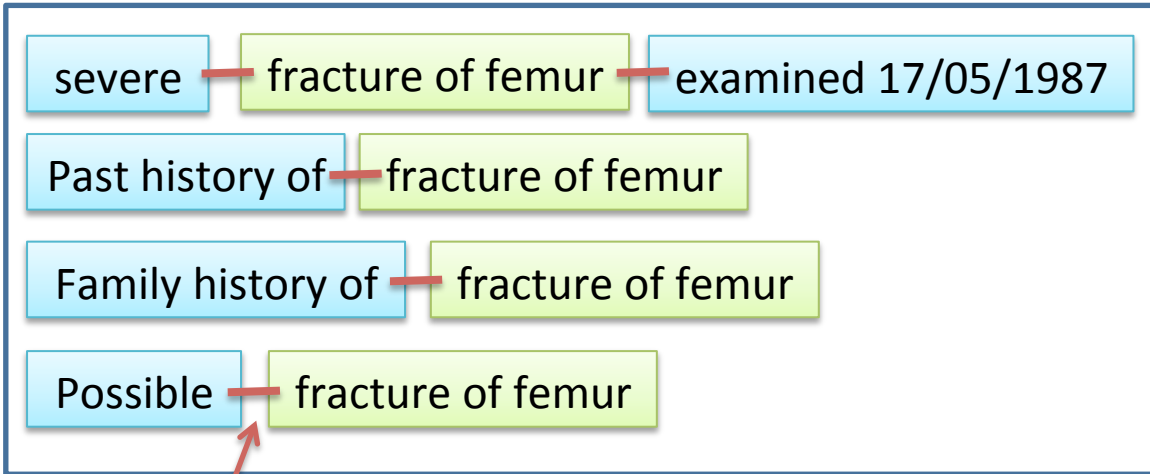
- **Plurality of Information Model approaches exists:**
  - openEHR, EN ISO 13606, HL7 RIM, CIMI, SIAMM, etc.
  - Local schemas are still predominant
  - Information model like structures in existing terminology **context model of SNOMED CT**
  - Free text (out of scope in SHN)
- **Plurality of representations within one specification**
- **WP4's relation to Information Models:**
  - Does not develop “yet another” information model
  - Maintains equidistance and neutrality
  - Looks at content and not at structure

# The role of SNOMED CT in the Project

- Provide agreed formal definitions of medical concepts (i.e. act as **medical domain ontology**)
- **Reference ontology** for representing medical domain concepts (e.g. mappings from ICD-11, LOINC, etc. to SNOMED CT)



# SNOMED CT - Information Model binding



**Medical records** contain items of information that refer to his clinical situation (described using medical concepts)

**Terminology binding**

**Overlapping area THE BOUNDARY PROBLEM**



**SNOMED CT context model hierarchy**

- [-] SNOMED CT Concept
  - [-] situation with explicit context
    - [-] A/N risk factors
    - [-] critical incident factors
    - [-] critical incident properties
    - [-] disease type AND/OR category not applicable
    - [-] disease type AND/OR category not assigned
    - [-] examination / signs
    - [-] family history with explicit context
    - [-] finding with explicit context
      - [-] caregiver unaware of diagnosis
      - [-] clinical finding absent
      - [-] diabetic foot at risk
      - [-] employment circumstances of partner or spouse
      - [-] family employment circumstance
      - [-] family history unknown

# Existing Terminology Binding Approaches

- **Guidelines specifications-based approaches:** they address the most common overlaps and provide modelling guidelines to resolve ambiguities (e.g. TermInfo, NHS openEHR work)

## HL7 TermInfo guideline rule example:

```
"An Observation class instance in which the Observation.value is a SNOMED CT expression representing a [ <<404684003 | clinical finding ] or a [ <<413350009 | finding with explicit context ] SHALL NOT contain an Act.code which when interpreted with the Observation.value yields a meaning that is substantially different from the meaning implied if the Act.code was "ASSERTION".
```

- For example, an Act.code meaning "Past history" or "Family history" may substantially alter the interpretation of a [ <<404684003 | clinical finding ] and should not be used in this way. Instead the SNOMED CT context model should be used to capture these significant differences in meaning."

It might work for a concrete implementation between an information model and terminology. It does not guarantee interoperability across representations based on other guidelines.

# Existing Terminology Binding Approaches (II)

- Define **clinical models** and constrain their elements and values to a **set of SNOMED CT values**.
  - **Simpler approaches**: EN ISO 13606, openEHR, etc.
  - More **sophisticated approaches** based on the definition of a set of general clinical models: CIMI, SIAMM
- **Simple approach example (EN ISO 13606):**

```
ENTRY[at0000] matches {-- Past history
```

```
  items matches {
```

```
    ELEMENT[at0001] matches {-- Condition
```

```
      value matches {
```

```
        CODED_TEXT matches {*} } -- terminology binding
```

```
    CLUSTER[at0002] matches {-- Details
```

```
404684003 | clinical finding |
```

Non semantic  
interoperable  
clinical models



```
ENTRY[at0000] matches {-- Entry
```

```
  items matches {
```

```
    ELEMENT[at0001] matches {-- Past history
```

```
      value matches {
```

```
        CODED_TEXT matches {*} } -- terminology binding
```

```
    CLUSTER[at0002] matches {-- Details
```

```
417662000 | past history of clinical finding |
```

# Existing Terminology Binding Approaches (II)

- Sophisticated approach example (CIMI):

```
ENTRY[at0000.1] matches { -- Observation
  link matches {LINK[at0.1] occurrences matches {0..*} -- Associated request}
  data matches {
    → use_archetype CLUSTER [CIMI-CORE-CLUSTER.observable.v1] -- Observable
      use_archetype CLUSTER [CIMI-CORE-CLUSTER.finding.v1] -- Results
      use_archetype CLUSTER [CIMI-CORE-CLUSTER.observe_action.v1] -- Observe action
```

```
CLUSTER[at0000] matches { -- Observable
  item matches {
    ELEMENT[at0001] occurrences matches {1} matches { -- Name
      value matches { TEXT matches {*}}}}
    ITEM[at0002] occurrences matches {0..*} -- Reason
    ITEM[at0003] occurrences matches {0..*} -- Method
    ELEMENT[at0004] occurrences matches {0..1} matches { -- Status
      value matches { CODED_TEXT matches {*}}}}
```

```
CLUSTER[at0000] matches { -- Action
  item matches {
    ELEMENT[at0001] occurrences matches {1} matches { -- Action type
      value matches { CODED_TEXT matches {*}}}}
    ITEM[at0009] occurrences matches {0..*} -- Reason
    ITEM[at0010] occurrences matches {0..*} -- Method
```

- Simple approaches do not address the boundary problem and produce non-interoperable clinical models
- Sophisticated approaches forbid the use of certain SNOMED CT hierarchies (e.g. context model)
- In both the decision of the elements to include in a clinical model is mainly a modeler decision not guided by any formal constraint



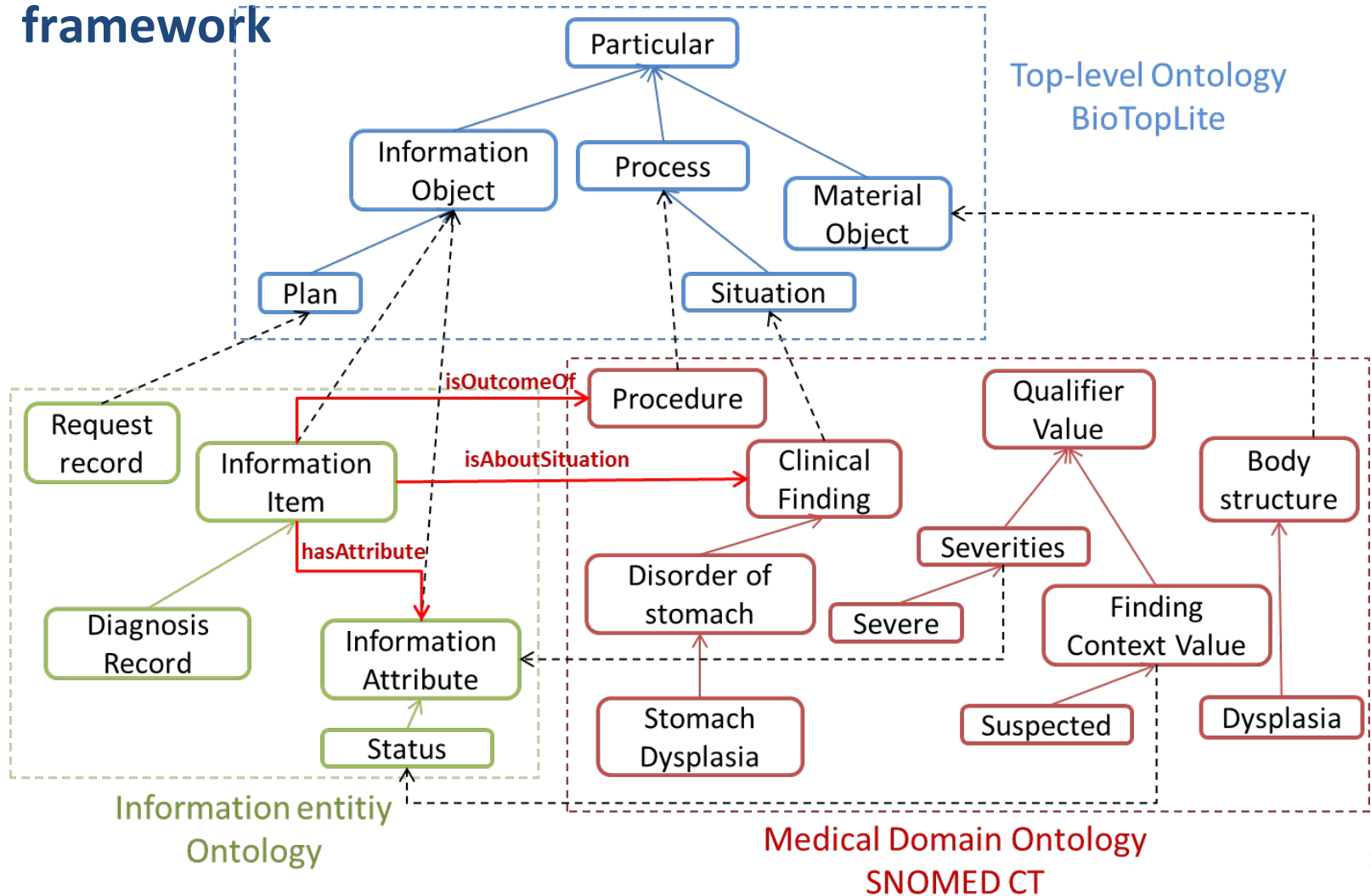


# SHN Terminology Binding Approach

- **Ontology design content patterns:** small fragments of an ontology for modelling an specific use case
- First introduced by Gangemi, Blomqvist and Sandkuhl in **2005**.
- They were devised to **guide and standardize** the way **ontologies are developed**
- Intended to **help non-expert** ontology users
- They **package best practice into reusable blocks** of ontology functionality, to be adapted and specialized by those users in their individual ontology development use cases

# SHN Semantic Patterns

- They are based on the **SemanticHealthNet ontological framework**



# SHN Semantic Patterns

- They are **language-independent** and should be encoded in a high order representation language
- We have represented them as:
  - A set of **RDF** (Subject-Predicate-Object) triples enhanced by cardinality constraint

*shn:InformationItem* 'describes situation' *shn:ClinicalSituation*

*shn:InformationItem* 'results from process' *shn:ClinicalProcess*

- **OWL 2 DL:**

- RDF Subject and Object transform into OWL classes
- RDF Predicate transforms into OWL DL expression

Logical  
reasoner



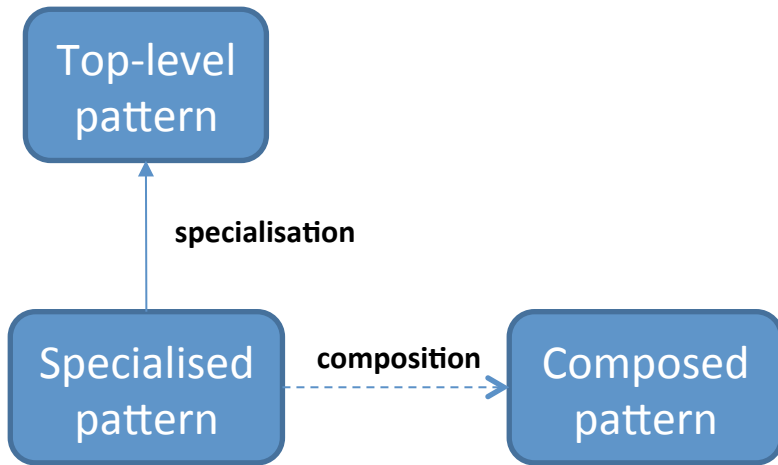
*shn:InformationItem*

and **shn:isAboutSituation** only *shn:ClinicalSituation*

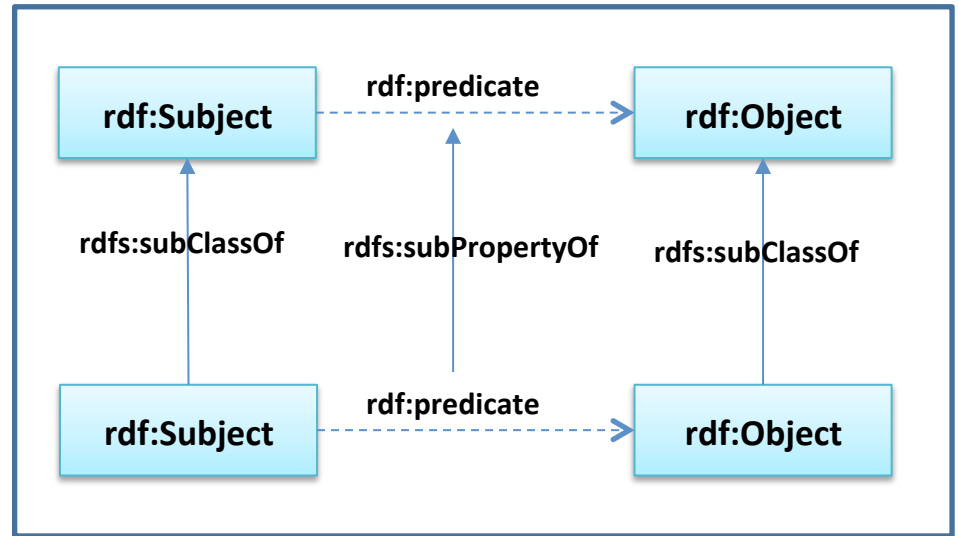
and **bt1:isOutcomeOf** some *shn:ClinicalProcess*

# SHN Semantic Patterns (II)

- They can be **specialised and composed** by following similar principles to object oriented languages



**Pattern Specialisation / Composition**



**RDF Triple Specialisation**

# Top-level Semantic Patterns

## OUR HYPOTHESIS:

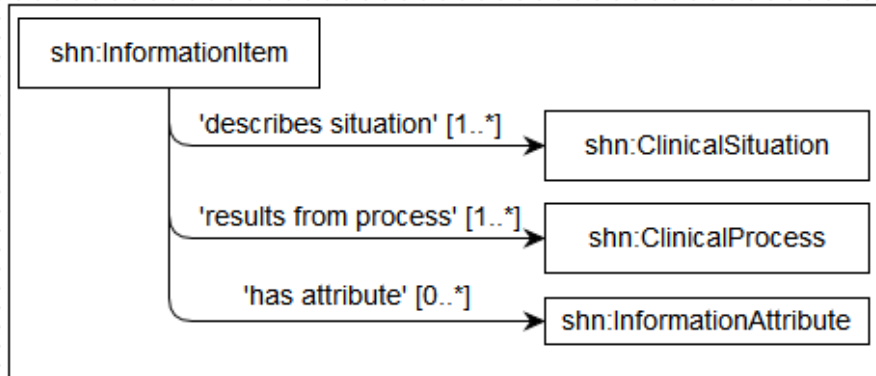
- A limited set of top-level semantic patterns that can be specialized and composed is sufficient to represent a great variety of clinical information.

## 7 top-level patterns extracted from the SHN Heart Failure Summary

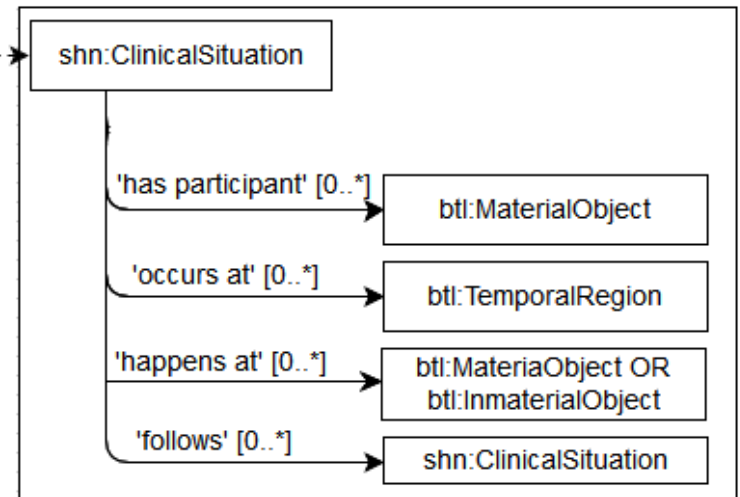
Top-level pattern	Example
<b>OBSERVATION RESULT PATTERN (OB_PT)</b>	record of body weight, height, etc-
<b>INFORMATION CLINICAL SITUATION (I_CS_PT)</b>	Cancer diagnosis, breathlessness symptom, etc.
<b>PLAN CLINICAL PROCESS (P_CP_PT)</b>	request to administer some drug, etc.
<b>CLINICAL PROCESS (CP_PT)</b>	assessment, history taking, etc.
<b>CLINICAL SITUATION (CS_PT)</b>	heart attack, diabetes, cancer, etc.
<b>PAST HISTORY CLINICAL SITUATION (PH_CS_PT)</b>	past history of heart failure, past history of cancer, etc.
<b>FAMILY HISTORY CLINICAL SITUATION (FH_CS_PT)</b>	family history of diabetes, family history of high blood pressure, etc.

# Pattern Specialisation / Composition

## Information - clinical situation pattern



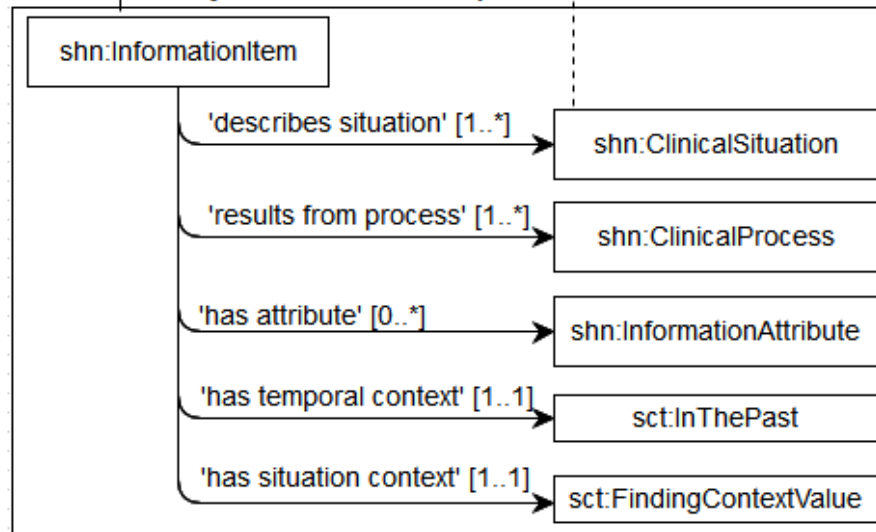
## Clinical situation pattern

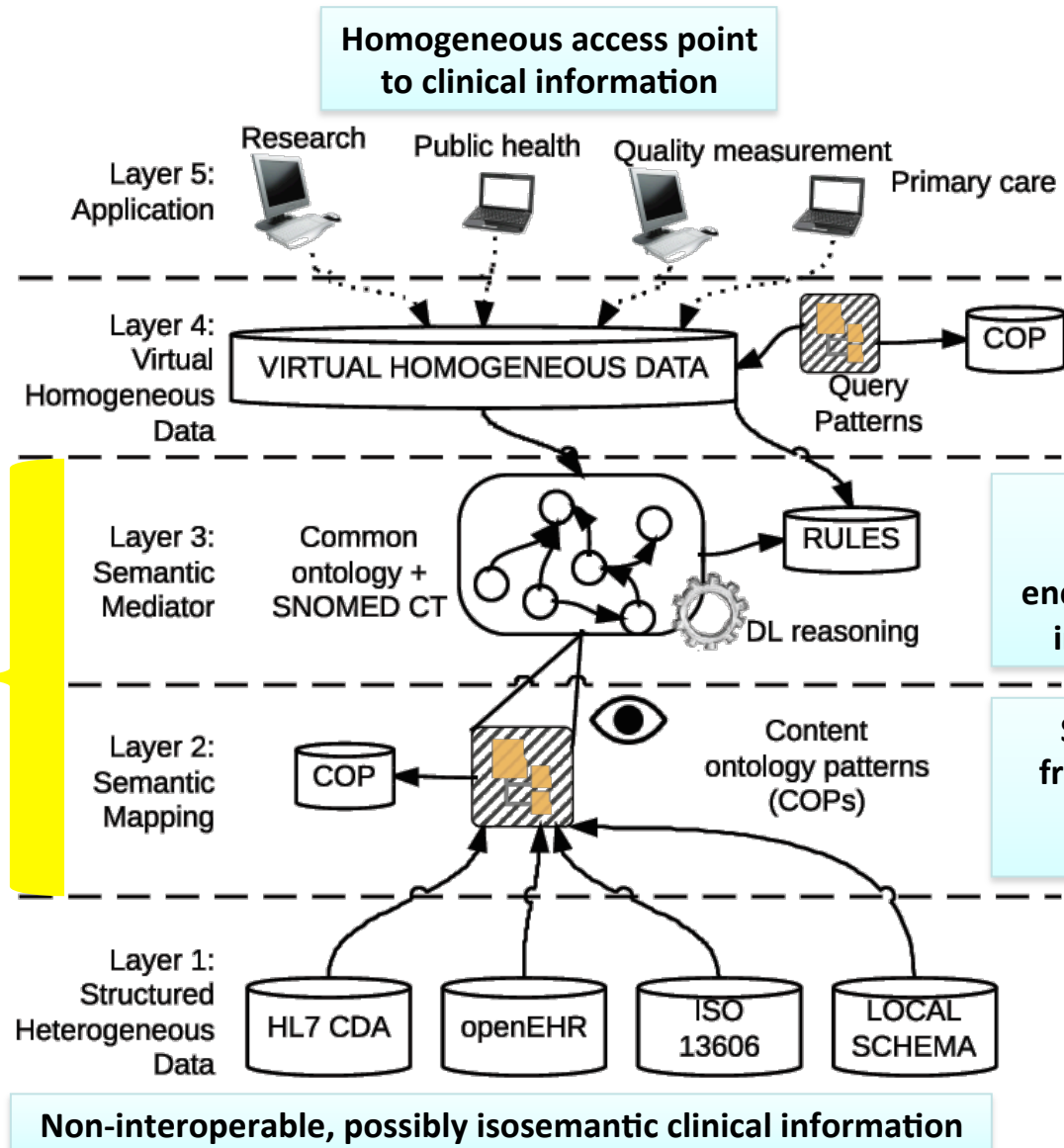


*pattern specialisation*

*pattern composition*

## History - clinical situation pattern





**SEMANTIC INFRASTRUCTURE**

Description logics formal ontology encompassing clinical and informational entities

Semantic patterns as frame-like user-friendly representation

# SHN Semantic Patterns role

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- Allow the consistent use of SNOMED CT within EHR clinical models (i.e. **address the boundary problem**)
- Enable **semantic interoperability** across heterogeneously structured clinical models within or not the same EHR specification
  - Enable **querying** over data heterogeneously structured and encoded
- Provide **advanced clinical information exploitation** capabilities
- **Guide** the **development** of new clinical models
- Detect **semantic inconsistencies** across existing clinical models



# Semantic Interoperability Example



GP Consultation

**General Questions**

History of diabetes mellitus ?

YES  NO

General questionnaire



Specialist

Past History : Diabetes mellitus | 732110... ▾

Cause: Pregnancy | 289908002 | ▾

Severity: Mild | 255604002 | ▾

Detailed Gynaecologist form

## Clinical study about endocrine diseases



- Patients with history of some **endocrine disease**
- Patients with history of **diabetes mellitus**
- Patients with history of **mild** diabetes mellitus
- Patients with history of **gestational diabetes**



# Semantic Interoperability Example (II)

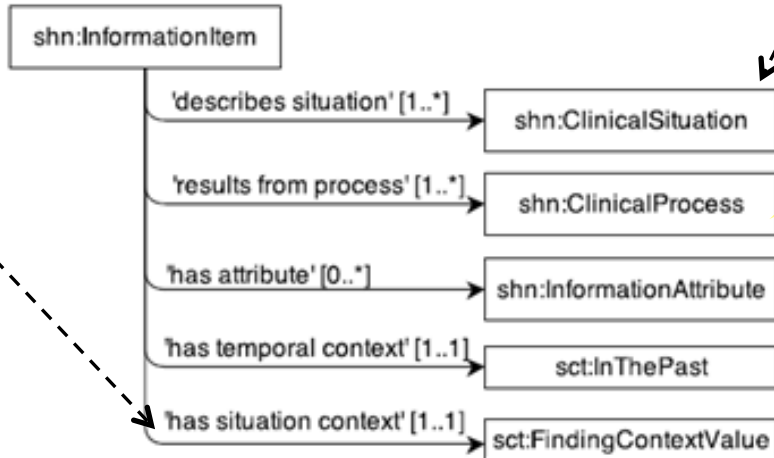
## General questionnaire – ISO 13606 Representation

```
ENTRY[at0000] matches { -- Question group
  items matches {
    CLUSTER[at0001] matches { -- Question group
      items matches {
        CLUSTER[at0002] matches { -- Question
          items matches {
            ELEMENT[at0003] matches { -- Answer
              value matches {
                BL matches {True, False}
              }
            }
          }
        }
      }
    }
  }
}
```

## SNOMED CT Terminology Binding

```
'History of clinical finding in subject (situation)'
and 'Role group (attribute)' some (
  'Associated finding (attribute)'
  some 'Diabetes mellitus (disorder)' and
  'Finding context (attribute)'
  some 'Known present (qualifier value)' and
  'Temporal context (attribute)'
  some 'In the past (qualifier value)' and
  'Subject relationship context (attribute)'
  some 'Subject of record (person)')
```

terminology binding



8410007  
| history taking |

History - clinical situation pattern



# Semantic Interoperability Example (III)

## Gynaecologist form – ISO 13606 Representation

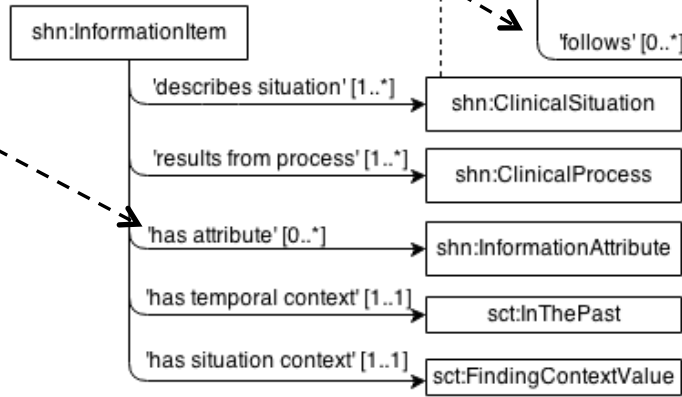
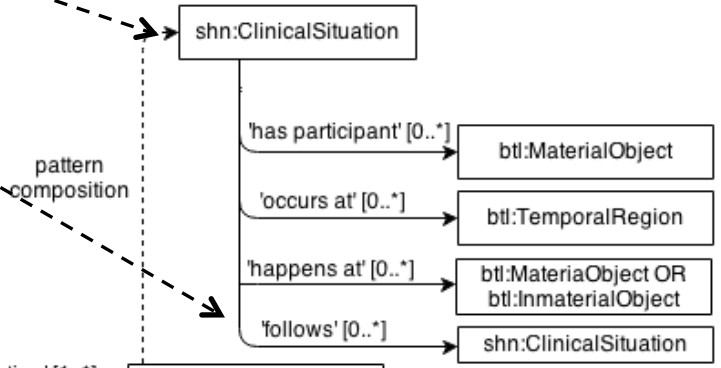
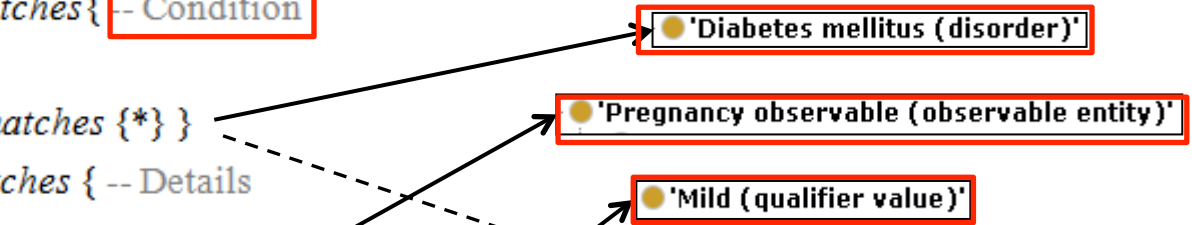
```

ENTRY[at0000] matches {-- Past history
  items matches {
    ELEMENT[at0001] matches {-- Condition
      value matches {
        CODED_TEXT matches {*} }
      CLUSTER[at0002] matches { -- Details
        items matches {
          ELEMENT[at0001] matches {-- Cause
            value matches {
              CODED_TEXT matches {*} } }
          ELEMENT[at0001] matches {-- Severity
            value matches {

```

## SNOMED CT Terminology Binding

terminology binding



# Challenges

## ✓ “Non-technical” challenges:

- ✓ Get more evidence that clinical models information can be sufficiently represented by semantic patterns
- ✓ Get more evidence that a limited number of top-level patterns is sufficient to derive more specific patterns by specialisation / composition mechanisms
- ✓ Engage clinical modelers community to help in testing the content coverage of the patterns

## ✓ Technical challenges:

- ✓ Provide different representations for different tasks
  - ✓ Closer to user representation
  - ✓ Logical representation (OWL DL)
  - ✓ RDF representation (RDF Shapes, SPIN)
  - ✓ UML-like representation
  - ✓ Etc.
- ✓ Solve performance issues related with the use of OWL DL
- ✓ Grow libraries of patterns
- ✓ Provide tools that facilitate clinical modelers engagement

# Conclusion

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- ✓ We need to be able to use **terminologies consistently within EHR information models** to achieve semantic interoperability
- ✓ We need **methods** that allow their consistent use independently of the particular EHR representation (we need to **focus on the content** and not on the structure!)
- ✓ **Semantic patterns** allow setting the **focus** on the content (**information meaning**)
- ✓ Semantic patterns were motivated by our experiences of **representing semantically clinical information**
- ✓ For getting **more evidence** of their usefulness we need the engagement of the community in order to see if this is **something else than theoretical research**

# Thanks for your attention



**Questions?**

**Comments?**

