Ontology Content Patterns as Bridge for the Semantic Representation of Clinical Information

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Three Kinds of Artefacts

Focus on **clinical information representation**

- **EHR Information Models**
  - ISO 13606, openEHR, HL7 RIM, DCM, etc.

- **Medical Terminologies / Ontologies**
  - SNOMED CT, ICD, LOINC, etc.

- **Clinical Models**
  - ISO 13606 / openEHR archetypes, DCMs, HL7 CDA, Intermountain CEM, etc.

**OVERLAPPING AREA**

**ISO-SEMANTIC MODELS**
Methods

ONTOLOGICAL FRAMEWORK

SEMANTIC INFRASTRUCTURE

SEMANTIC PATTERNS
Ontological Framework

Precise categorization by upper level ontology

BioTopLite 2.0

Information entity ontology

epistemic entities

- Clinical datatype entity
- Information Item
- Request record
- ... (other clinical data entities)

SNOMED CT ontology

- Clinical Finding
- Procedure
- Qualifier value
- Disorder
- Evaluation procedure
- Laboratory procedure
- ... (other clinical procedure entities)

FORMALISED IN OWL 2 DL (description logics)
Ontological Framework (II)

Medical Application Systems

HOMOGENEOUS DATA ACCESS POINT

SEMANTIC INFRASTRUCTURE

STRUCTURED HETEROGENEOUS DATA

EHR - OpenEHR

Patient: Franz Müller
DoB: 17/01/1965
Encounter: 23/08/98

Tobacco Use Summary
- Cigarette smoker, 14 /d

EHR - HL7 CDA

Patient: Franz Müller
DoB: 17/01/1965
Encounter: 23/08/2001

Social history
- Heavy cigarette tobacco smoker
Ontological Framework (III)

Patient: Franz Müller
DoB: 17/01/1965
Encounter: 23/08/98

Tobacco Use Summary
- Cigarette smoker, 14 /d

openEHR archetype

EVALUATION[at0000] matches {
  -- Tobacco Use Summary
  data matches {
    ITEM_TREE[at0001] matches {
      -- Tree
      items cardinality matches {0..*; unordered} matches {
        ELEMENT[at0002] occurrences matches {0..1} matches {
          -- Smoking Status
          value matches {
            DV_CODED_TEXT matches {
              defining_code matches {
                [local::
                  at0003, -- Current Smoker
                  at0027, -- Never Smoked
                  at0005] -- Ex-smoker
              }}
          },
          CLUSTER[at0029] occurrences matches {0..*} matches {
            -- Smoking Details
            items cardinality matches {1..*; unordered} matches {
              ELEMENT[at0028] occurrences matches {0..1} matches {
                -- Form
                value matches {
                  DV_TEXT matches {*}}
              },
              ELEMENT[at0030] occurrences matches {0..1} matches {
                -- Typical smoked amount
                value matches {
                  C_DV_QUANTITY <
                  property = <[openehr::382]>
                  list = <
                    ["1"] = <
                      units = <"/d">
                      magnitude = <|>=0.0|>
                      precision = <|0|>
                    >>}}}}}}}}}}}}}}

OWL DL

-- Smoking status
InformationItem
  and isAboutSituation only sct:TobaccoSmokingSituation
  and isOutcomeOf some sct:HistoryTaking

-- Form
InformationItem
  and isAboutSituation only sct:CigaretteTobaccoSmoking
  and isOutcomeOf some sct:HistoryTaking

-- Amount
ObservationResult
  and isAboutQuality some (MassIntake
  and inheresIn some sct:CigaretteTobaccoSmokingSituation
  and projectsOnTo some (ValueRegion
  and isRepresentedBy only (hasValue value 14
  and hasInformationAttribute some sct:PerDay)))
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Ontological Framework (IV)

Primary Care

EHR

Patient: Franz Müller
DoB: 17/01/1965
Encounter: 23/08/2001

Social history
-Heavy cigarette tobacco smoker

--- Current Heavy tobacco smoker
InformationItem
  and isAboutSituation only sct:HeavyCigaretteSmokingSituation
  and isOutcomeOf some sct:Evaluation

HL7 CDA model compliant with ‘Meaningful Use’

Patient: Franz Müller
DoB: 17/01/1965
Encounter: 23/08/2001

Social History
- Began smoking daily on 4/10/2011.

Current Heavy tobacco smoker

‘Meaningful Use’:
> = 10 cigarettes / day
Semantic Patterns

emiesfriendly **abstractions** that provide a more intuitive and closer to natural language representation

**Prevent** modelers from a in-depth **ontology and description logics** modeling knowledge and skills

Represent **recurring content modelling tasks** by following a standardized structure
- E.g. who does what, when and where?, which objects take part in a certain event?, etc.

**Defined wrt. a reference ontology** (particular view on it)

They can be **specialised** and **composed**
Semantic Patterns (II)

- Represented as a set of **constrained** Subject–Predicate–Object (SPO) triples.
Semantic Patterns
OWL DL Representation

Use of DL reasoning for:
- Detection of iso–semantic information across models
- Advanced query of clinical information

OWL 2 DL transformation:
- SUB and OBJ into OWL DL classes
- Predicates into OWL DL expressions

<table>
<thead>
<tr>
<th>Predicate</th>
<th>OWL DL expression</th>
</tr>
</thead>
<tbody>
<tr>
<td>'describes situation'</td>
<td>SUBJ subClassOf shn:isAboutSituation only OBJ</td>
</tr>
<tr>
<td>'results from process'</td>
<td>SUBJ subClassOf btl:isOutcomeOf some OBJ</td>
</tr>
<tr>
<td>'has attribute'</td>
<td>SUBJ subClassOf btl:hasInformationAttribute some OBJ</td>
</tr>
<tr>
<td>'has observed value'</td>
<td>SUBJ subClassOf btl:Quality and btl:projectsOnto some OBJ</td>
</tr>
<tr>
<td>'has units'</td>
<td>SUBJ subClassOf btl:isRepresentedBy only</td>
</tr>
<tr>
<td></td>
<td>(shn:hasInformationAttribute some OBJ)</td>
</tr>
</tbody>
</table>

\[ shn:InformationItem \text{`describes situation`} shn:ClinicalSituation \]

\[ shn:InformationItem \text{and} shn:isAboutSituation \text{only} shn:ClinicalSituation \]
Advanced Query of Clinical Information

--- Smoking status
InformationItem
  and isAboutSituation only sct:TobaccoSmokingSituation
  and isOutcomeOf some sct:HistoryTaking

--- Current Heavy tobacco smoker
InformationItem
  and isAboutSituation only sct:HeavyCigaretteSmokingSituation
  and isOutcomeOf some sct:Evaluation

--- Form
InformationItem
  and isAboutSituation only sct:CigaretteTobaccoSmoking
  and isOutcomeOf some sct:HistoryTaking

--- Amount
ObservationResult
  and isAboutQuality some (MassIntake
  and inheresIn some sct:CigaretteTobaccoSmokingSituation
  and projectsOnTo some (ValueRegion
  and isRepresentedBy only (hasValue value 14
  and hasInformationAttribute some sct:PerDay)))

Hospital
Patient: Franz Müller
DoB: 17/01/1965
Encounter: 23/08/98

Tobacco Use Summary
- Cigarette smoker, 14 /d

Primary Care
Patient: Franz Müller
DoB: 17/01/1965
Encounter: 23/08/2001

Social History
- Heavy cigarette tobacco smoker

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## Advanced Query of Clinical Information (II)

| #Q1 | $\text{shn:InformationItem}$ and $\text{btl:isOutcomeOf}$ some $\text{sct:HistoryTaking}$ and $\text{shn:isAboutSituation}$ only $\text{sct:TobaccoSmokingSituation}$ |

---

### Smoking status

InformationItem

- isAboutSituation only $\text{sct:TobaccoSmokingSituation}$
- isOutcomeOf some $\text{sct:HistoryTaking}$

---

### Current Heavy tobacco smoker

InformationItem

- isAboutSituation only $\text{sct:HeavyCigaretteSmokingSituation}$
- isOutcomeOf some $\text{sct:Evaluation}$

---

**DL reasoning**

**SNOMED CT ontology**
Advanced Query of Clinical Information (III)

<table>
<thead>
<tr>
<th>#Q2</th>
<th>shn:InformationItem and btl:isOutcomeOf some shn:Evaluation and shn:isAboutSituation only sct:HeavyTobaccoSmokingSituation</th>
</tr>
</thead>
<tbody>
<tr>
<td>#Q3</td>
<td>shn:InformationItem and btl:isOutcomeOf some shn:Evaluation and shn:isAboutSituation only sct:HeavyTobaccoSmokingSituation and shn:isAboutSituation only sct:CigaretteTobaccoSmokingSituation</td>
</tr>
</tbody>
</table>

```
-- Amount ObservationResult
  and isAboutQuality some (MassIntake
  and inhesesIn some sct:CigaretteTobaccoSmokingSituation
  and projectsOnto some (ValueRegion
  and isRepresentedBy only (hasValue value 14
  and hasInformationAttribute some sct:PerDay)))
```

```
-- Current Heavy tobacco smoker InformationItem
  and isAboutSituation only sct:HeavyCigaretteSmokingSituation
  and isOutcomeOf some sct:Evaluation
```

**Local system rules. Vary across institutions**

**DL reasoning**

**SNOMED CT ontology**
### Advanced Query of Clinical Information (IV)

| #Q4 | $\text{shn:ObservationResult}$ and $\text{shn:isAboutQuality}$ only ($\text{shn:MassIntake}$ and $\text{btl:inheresIn}$ some $\text{sct:CigaretteTobaccoSmokingSituation}$ and $\text{btl:projectsOnto}$ some ($\text{btl:ValueRegion}$ and $\text{btl:isRepresentedBy}$ only ($\text{shn:hasInformationAttribute}$ some $\text{sct:PerDay}$ $\text{shn:hasValue}$ some int[>15]))) |

---

- **Amount**
  - $\text{ObservationResult}$ and $\text{isAboutQuality}$ some ($\text{MassIntake}$ and $\text{inheresIn}$ some $\text{sct:CigaretteTobaccoSmokingSituation}$ and $\text{projectsOnto}$ some ($\text{ValueRegion}$ and $\text{isRepresentedBy}$ only ($\text{hasValue}$ value 14 and $\text{hasInformationAttribute}$ some $\text{sct:PerDay}$)))

- **Current Heavy tobacco smoker**
  - $\text{InformationItem}$ and $\text{isAboutSituation}$ only $\text{sct:HeavyCigaretteSmokingSituation}$ and $\text{isOutcomeOf}$ some $\text{sct:Evaluation}$

---

*The query does not retrieve any record.*

| $\text{shn:ObservationResult}$ and $\text{shn:isAboutQuality}$ some (mass_intake and 'inheres in' some $\text{sct:CigaretteTobaccoSmokeSituation}$ and 'projects onto' some ('value region' and 'is represented by' only ($\text{shn:hasInformationObjectAttribute}$ some $\text{sct:PerDay}$ and $\text{shn:hasValue}$ some int[>= 10]))) |

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*Local system rules. Vary across institutions*
Conclusions

- The use of **EHR standards and terminologies** does not guarantee semantic interoperability

- SemanticHealthNet proposes an **intermediate semantic layer** able to deal with the present clinical information heterogeneity

- We hypothesize that clinical models information can be represented by the **specialisation and composition of a set of limited top-level semantic patterns**

- The proposed infrastructure faces **two main kinds of challenges** (human & computational)

- **Compromises** between **performance and functionality** might be required
Danke!

ONTOLOGICAL FRAMEWORK

SEMANTIC PATTERNS

SEMANTIC INFRASTRUCTURE