A Semantic-Web Oriented Representation of Clinical Element Models

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Acknowledgement

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Introduction

Clinical Element Model (CEM):

- Logical models ensure semantic interoperability for:
  - Data representation
  - Data interpretation
  - Data exchange within and across heterogeneous sources and applications

- Represented in CEML/CDL
  - Define syntax and grammar
  - Not semantics
Introduction

Semantic Web

- Explicit and formal semantic knowledge representation
- OWL/RDF:
  - Define relationships
  - Define classes
  - Define constraints
- Consistency checking
- Link to other domain terminologies
- Harmonize with other clinical data modeling languages
- Semantic reasoning
Layer 1: meta-ontology

Layer 2: detailed clinical element models

Layer 3: patient instances

RDF Triple Store
# CEM Abstract Instance Model

<table>
<thead>
<tr>
<th>Property</th>
<th>Type</th>
<th>Cardinality</th>
</tr>
</thead>
<tbody>
<tr>
<td>type</td>
<td>CNE</td>
<td>1</td>
</tr>
<tr>
<td>key</td>
<td>CNE</td>
<td>1</td>
</tr>
<tr>
<td>data</td>
<td>CHOICE&lt;CWE, CO, ST, PQ, IVLPQ, RTOPQ, TS, II, INT, REAL, ED&gt;</td>
<td>0-1</td>
</tr>
<tr>
<td>attrs</td>
<td>SEQUENCE&lt;CEInstance (Attribution)&gt;</td>
<td>0-1</td>
</tr>
<tr>
<td>items</td>
<td>SEQUENCE&lt;CEInstance&gt;</td>
<td>0-1</td>
</tr>
<tr>
<td>quals</td>
<td>SEQUENCE&lt;CEInstance (Component)&gt;</td>
<td>0-1</td>
</tr>
<tr>
<td>mods</td>
<td>SEQUENCE&lt;CEInstance (Modifier)&gt;</td>
<td>0-1</td>
</tr>
<tr>
<td>alt</td>
<td>CHOICE&lt;CWE, PQ, ED, ST&gt;</td>
<td>0-1</td>
</tr>
</tbody>
</table>
Clinical Element Categories

- CEInstance: parent class for all the clinical element classes
- Statement
- Component
- Association
- Modifier
- Attribution
Model Element Value Choice
Properties

- **Type:**
  - unique name
  - URI

- **Key:**
  - refers to a code from a standard terminology
  - annotation property

- **Data:**
  - Represents values
  - data type property
  - Domain: SimpleStatement or SimpleComponent
Properties

- **Item**
  - Associate a clinical element class with its sub-clinical element item
- **Object Property**
  - Domain: Panel, CompoundStatement, Component
  - Range: CEInstance
Properties

- **Att**
  - Declares the who, where, why, and when
  - Object Property
  - Domain: CEInstance, Range: Attribution

- **Qual**
  - Add useful information but not change the meaning
  - Object Property
  - Domain: CEInstance, Range: Component

- **Mod**
  - Affect the semantic interpretation of the instance
  - Object Property
  - Domain: CEInstance, Range: Modifier
Statement

- A complete assertion about a particular aspect of a patient
- Statement, Simple Statement, Compound Statement
### Statement

#### Simple Statement
- **data**: HL7 datatype

#### Compound Statement
- **item**: component
- **item**: component
- **item**: component

<table>
<thead>
<tr>
<th>Statement</th>
<th>The definition defines a complete assertion</th>
<th>super(SimpleStatement, CompoundStatement); DisjointClasses(all the other category-classes)</th>
</tr>
</thead>
<tbody>
<tr>
<td>SimpleStatement</td>
<td>The meaning is conveyed by a single clinical value; defines a complete assertion</td>
<td>subClassOf(restriction(data max 1)); DisjointClasses(all the other category-classes)</td>
</tr>
<tr>
<td>CompoundStatement</td>
<td>The meaning is conveyed by multiple clinical values (components); defines a complete assertion</td>
<td>subClassOf(restriction(item some Component and item only Component)); DisjointClasses(all the other category-classes)</td>
</tr>
</tbody>
</table>
Specialized Statements

- Patients
- Providers
- Encounters
- Contacts
- Activities
- Locations
Component

- Can not persist alone
- Must be item or qual for another CEInstance
- Component, SimpleComponent, CompandComponent

<table>
<thead>
<tr>
<th></th>
<th>Used only within another constraint type, cannot be persisted alone in the EHR</th>
<th>super(SimpleComponent, CompoundComponent); subClassOf(restriction(itemFor some CEInstance or qualFor CEInstance)); DisjointClasses(all the other category-classes)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Component</td>
<td></td>
<td></td>
</tr>
<tr>
<td>SimpleComponent</td>
<td>The meaning is conveyed by a single clinical value; cannot stand alone</td>
<td>subClassOf(restriction(data max 1)); DisjointClasses(all the other category-classes)</td>
</tr>
<tr>
<td>Compound-Component</td>
<td>The meaning is conveyed by multiple clinical values (components); cannot stand alone</td>
<td>subClassOf(restriction(item some Component and item only Component)); DisjointClasses(all the other category-classes)</td>
</tr>
</tbody>
</table>
Association

- Panel
- Collection
- Sequence
- List
- Semantic Link
- Annotation
- Patient Relationship
Panel

- Represents a common grouping of clinical observations
- A collection of other statements or panels

Panel

| Panel | represents a common grouping of clinical observations | subClassOf(restriction(item only(Panel or Statement))and (item some (Panel or Statement))); DisjointClasses(all the other category-classes) |
Sequence

- Defines an association wherein its associated associations and/or statements are part of some ordered sequence.
List

- Define a list is an association that contains a set of references of some other independent statements or associations.
- Define a new class called List and use owl:unionOf to represent the association of referenced elements.
Semantic Link

- An association that represents a strong semantic relationship between two or more statements/associations

```prolog
:patient1
  a :Patient ;
  :item :_treatment1 .
:_treatment1
  a :Treatment ;
  :treated_for :Headache ;
  :treated_by :Aspirin .
```
Annotation

- An association that adds independent textual annotation information to the referenced statements and/or associations
Modifier and Attribution

- **Modifier**: can only be associated with the mod property
- **Attribution**: can only be associated with the att property

<table>
<thead>
<tr>
<th>Modifier</th>
<th>Similar to Component, except can only used with property <em>mods</em></th>
<th>subClassOf(restriction(modFor some CEInstance)); DisjointClasses(all the other category-classes)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Attribution</td>
<td>Similar to Component, except can only used with property <em>atts</em></td>
<td>subClassOf(restriction(attFor some CEInstance)); DisjointClasses(all the other category-classes)</td>
</tr>
</tbody>
</table>
Example

Description: BloodPressurePanel

1. Panel
   - att max 1 Observed
   - item max 1 DiastolicBloodPressure
   - item max 1 SystolicBloodPressure
   - mod max 1 Subject
   - qual max 1 BloodPressureBodyLocationPrecood

2. (item some (Association or Statement)) and (item only (Association or Statement))

Description: DiastolicBloodPressure

3. Equivalent classes

4. BodyLocationPrecood
   - BloodPressureBodyLocationPrecood
     - Arm
     - Finger
     - Wrist

5. Annotations
   - Preferred Unit
     "MillimetersOfMercury"

6. Inherited anonymous classes
   - data max 1

Description: DiastolicBloodPressureData

7. Class references

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SHARPn.org
## Evaluation

<table>
<thead>
<tr>
<th>Criterion</th>
<th>Score</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Definition of Classes (lexical)</td>
<td>5</td>
<td>The reviewers agreed that the lexical definition of each class is clear.</td>
</tr>
<tr>
<td>Hierarchical Structure (sub classes hierarchy)</td>
<td>4</td>
<td>The reviewers agreed that the sub-class hierarchy faithfully represents the CEM specifications. There is a concern about defining the basic category classes (e.g., SimpleStatement and ComponentStatement) and the specialized classes (e.g., Patient, Activity, etc) as sibling classes under the Statement class since they represent different types of classifications of CEMs. An alternative solution could be to create two parent classes under the Statement class to separate the two groups of classes.</td>
</tr>
<tr>
<td>Definition of Classes (semantic)</td>
<td>5</td>
<td>The reviewers agreed that the semantic definition of each class is correct.</td>
</tr>
<tr>
<td>Semantic Relations (object property definitions)</td>
<td>5</td>
<td>The reviewers agreed that the domain and range defined for each property are correct.</td>
</tr>
</tbody>
</table>
# Evaluation

**Context and Application** *(validation of the usage of the CEM-OWL model in a detailed CEM)*

<table>
<thead>
<tr>
<th>Class Usage</th>
<th>4</th>
<th>The reviewers agreed that defining each detailed CEM (e.g. BloodPressurePanel) as a new class and a subclass of the category it belongs to (e.g. Panel) are appropriate and semantically correct. There was a discussion about the option on defining each detailed CEM as an individual of the category class (e.g., BloodPressurePanel rdf:type Panel). The reviewers agreed that the current way is more appropriately aligned to the three-layer approach used for this representation.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Relation Usage</td>
<td>5</td>
<td>The reviewers agreed that the relations defined in the meta-ontology are sufficient to represent the connection between components in the detailed CEM.</td>
</tr>
<tr>
<td>Cardinality Constraints</td>
<td>5</td>
<td>The reviewers agreed that the cardinality constraints are defined correctly and the OWL cardinality constraints are sufficient to represent CEM cardinality constraints.</td>
</tr>
</tbody>
</table>
Conclusion and Future Directions

- Meta-Ontology: semantically defined the basic classes, properties, their relationships, and constraints
- Convertor: CDL → OWL
- Represent SHARPn normalized data using RDF
- Investigate SWRL/Drools combination for phenotyping