

Addressing the negation gap in SNOMED CT by reified negated concepts

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Abstract. Despite increasing performance of computer hardware, reasoning with large OWL ontologies still poses some nearly insurmountable challenges. The complexity of sound and complete reasoning makes OWL DL intractable for large ontologies. OWL-EL appears a good compromise and OWL models following this profile have demonstrated good scalability, using specialised reasoners like ELK and Snorocket. SNOMED CT is moving more towards description logics and has chosen OWL-EL as the representational profile for the reasons mentioned above. A major drawback of this is the lack of support of logical negation (NOT) for this profile. Many SNOMED CT concepts suggest negation (e.g. by expressions like “A without B”, “Absence of X” etc). Based on such lexical patterns, we have identified the currently underlying OWL modelling patterns, classified them into distinct categories and for each category manually inspected some of their concepts in order to assess if they were correctly classified. Finally, we discuss several OWL remodelling approaches able to express negation in a tractable way (EL profile) and avoiding wrong inferences.

Keywords. SNOMED CT, OWL DL, negation, ontology design patterns

1. Introduction

SNOMED CT (SCT) is a semantically rich, ontology-based, large clinical healthcare terminology, which provides a standardized way to represent clinical phrases captured by clinicians and enables their automatic interpretation [1]. SCT concepts are defined by following the SNOMED CT concept model, which provides nine main top-level categories (also known as semantic types), among which we find *Clinical finding*, *Procedure*, *Body structure*, etc.

Among other formats, SCT is provided as an OWL ontology, enabling the use of description logic (DL) reasoning which supports advanced classification and querying. Given its large size and the decrease of performance of reasoners over large and expressive OWL ontologies, it implements the EL profile (EL++) [2]. This profile

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provides polynomial time algorithms for all the standard reasoning tasks of description logic but lacks the DL features universal restriction, negation and disjunction. We found circa 6,000 SNOMED CT concepts out of approximately 311,000 total that include negation, being modelled in different ways, many of which lead to unintended inferences like *Absence of finger* implying *Absence of hand*, or *No Back Pain* entailing *No Pain*.

In this work we identified OWL modelling patterns corresponding to lexical patterns that are typical for SNOMED CT concepts that bear some implicit negation. We classified them into several categories and inspected sample concepts of each category in order to assess if they were correctly classified. Finally, different alternatives and steps toward remodelling are proposed.

2. Material and Methods

Within a PostgreSQL database we checked the SNOMED CT January 2019 release for the following lexical negation patterns and identified the number of concepts involved (see Table 1). In total, there are 5,823 SNOMED CT concepts with labels that lexically include negation cues. Most of them were descendants of *Clinical Finding* (4,122), followed by *Situation* (713). The substrings “not” and “without” were the two most frequent lexical cues (3,536).

Table 1. Negation lexical patterns; Number of concepts and their semantic type

Lexical pattern	Concept count	Semantic Type		
		<i>Clinical Finding</i>	<i>Situation</i>	<i>Procedure</i>
“Absence”	354	300	18	2
“Absent”	446	359	70	2
“No”	956	573	236	14
“Lack of”	110	105	0	5
“Not”	1743	1191	323	96
“Without”	1793	1331	15	357
“Non”	41	24	1	3
“Never”	22	0	17	5
“Negative”	350	236	28	23
“Excluded”	8	3	5	0
Total	5823	4122	713	507

For each semantic type we have selected the two most frequent patterns (highlighted in bold in Table 1) and inspected their hierarchies in order to find concepts wrongly classified. For concepts where inappropriate reasoning results were found, alternative modelling patterns are proposed and discussed.

3. Results

A frequent lexical pattern is the substring “not” in the label. Out of 1,191 ones under *Clinical Finding*, 818 were fully defined concepts, i.e. using equivalentTo axioms in OWL. More than half of them (633) corresponded to negated verb phrases with the substring “does not”, most of which denote fully defined concepts. To define a new concept following this pattern, the variable parts (“X”) are substituted by words that represent some activity. Note the property ‘Role group (attribute)’, a unique SCT construct used to group attribute-value pairs for classification purposes [3].

```
'Does not X (finding)' equivalentTo
'Finding related to ability to X (finding)'
and 'Role group (attribute)' some (
  'Has interpretation (attribute)' some 'Does not (qualifier value)' and
  'Interprets (attribute)' some Ability to X (observable entity)'
```

An example of a wrongly classified concept is *Does not jump (finding)*. Its inferred definition shows its classification as a subclass of *Does not move (finding)* (subclassOf statement part). This is factually wrong, because from the fact that a patient does not jump you cannot derive that he or she does not move. If we look more deeply in the definition (equivalentTo statement part) it “interprets” *Ability to jump (observable entity)* which is subsumed by *Ability to move (observable entity)* which is correct but causes the wrong classification of *Does not jump (finding)*.

```
'Does not jump (finding)'
subclassOf
  'Does not move (finding)'
  'Finding related to ability to jump (finding)'
equivalentTo
  'Finding related to ability to jump (finding)'
  and 'Role group (attribute)' some (
    'Has interpretation (attribute)' some 'Does not (qualifier value)' and
    'Interprets (attribute)' some 'Ability to jump (observable entity)')
```

Another 323 concepts from the *Situation* hierarchy (238 are non-primitive) have “not” in their label, more specifically concepts under ‘*Clinical finding absent (situation)*’ and ‘*Procedure with explicit context (situation)*’, following (roughly) the corresponding pattern:

```
'Not Clinical finding X (situation)' equivalentTo
'Clinical finding absent (situation)'
and 'Role group (attribute)' some (
  'Associated finding (attribute)' some 'Clinical finding X (finding)' and
  'Finding context (attribute)' some 'Known absent (qualifier value)' and
  'Temporal context (attribute)' some 'Current or specified time (qualifier value)' and
  'Subject relationship context (attribute)' some 'Subject of record (person)')
```

An example is ‘*Joint not swollen (situation)*’. The inferred definition wrongly entails ‘*Swelling absent (situation)*’. Again, a patient might not have swollen joints, but he might have swollen feet. Here the wrong classification occurs due to the axiom “Associated finding” some ‘*Joint swelling (finding)*’, the latter being a subclass of ‘*Swelling of body structure (finding)*’.

‘*Joint not swollen (situation)*’
subclassOf
 ‘*Swelling absent (situation)*’
equivalentTo
 ‘*Clinical finding absent (situation)*’
 and ‘**Role group (attribute)**’ some (
 ‘**Associated finding (attribute)**’ some ‘*Joint swelling (finding)*’ and
 ‘**Finding context (attribute)**’ some ‘*Known absent (qualifier value)*’ and
 ‘**Temporal context (attribute)**’ some ‘*Current or specified time (qualifier value)*’ and
 ‘**Subject relationship context (attribute)**’ some ‘*Subject of record (person)*’)

Under ‘*Procedure with explicit context (situation)*’, we found concepts that represent some contextual information specific to procedures, which syntactically correspond to a verb (**V**) as past participle (**P-ed**) or nominalization (**P-ion**) preceded by “not”, like in “not done”, “not wanted”, “not indicated”, “not suspected”, “not needed”, etc., related to some anatomy term (**A**).

‘**A Not V-ed (situation)**’ equivalentTo
 ‘*Procedure not done (situation)*’
 and ‘**Role group (attribute)**’ some (
 ‘**Associated procedure (attribute)**’ some ‘*V-ion of A (procedure)*’ and
 ‘**Procedure context (attribute)**’ some ‘*Not V-ed (qualifier value)*’ and
 ‘**Temporal context (attribute)**’ some ‘*Current or specified time (qualifier value)*’ and
 ‘**Subject relationship context (attribute)**’ some ‘*Subject of record (person)*’)

In this pattern, in the object property ‘**Procedure context (attribute)**’, restricted by ‘*Qualifier value (qualifier value)*’ concepts, we find negative meaning in descendants of ‘*Context values for action (qualifier value)*’, e.g. ‘*Not done (qualifier value)*’. An example is ‘*Retinae not examined (situation)*’, with the factually wrong entailment ‘*Patient not examined (situation)*’. The latter concept ‘*Patient not examined (situation)*’ is associated with the procedure ‘*Physical examination procedure (procedure)*’ and ‘*Examination of retina (procedure)*’ is a subclass.

‘*Retinae not examined (situation)*’
subclassOf
 ‘*Patient not examined (situation)*’
equivalentTo
 ‘*Procedure not done (situation)*’
 and ‘**Role group (attribute)**’ some (
 ‘**Associated procedure (attribute)**’ some ‘*Examination of retina (procedure)*’ and
 ‘**Procedure context (attribute)**’ some ‘*Not done (qualifier value)*’ and
 ‘**Temporal context (attribute)**’ some ‘*Current or specified time (qualifier value)*’ and
 ‘**Subject relationship context (attribute)**’ some ‘*Subject of record (person)*’)

Out of 96 ‘Procedure (procedure)’ concepts only 4 of them are fully defined, and here the only hint to a negative meaning is the label, without any element with negative polarity in the OWL definition. This is the reason why here no negation-specific OWL pattern can be provided. We analysed the concept ‘*Revision of total prosthetic replacement of shoulder joint not using cement (procedure)*’ and did not find any wrong inference.

‘Revision of total prosthetic replacement of shoulder joint not using cement (procedure)’

subclassOf

‘Prosthetic uncemented total shoulder replacement (procedure)’

‘Revision of total prosthetic replacement of shoulder joint (procedure)’

equivalentTo

‘Uncemented total replacement of joint (procedure)’

and **‘Role group (attribute)’** some (

‘Revision status (attribute)’ some *‘Revision - value (qualifier value)’* and

‘Method (attribute)’ some *‘Surgical insertion - action (qualifier value)’* and

‘Direct device (attribute)’ some *‘Total shoulder replacement prosthesis (physical object)’* and

‘Procedure site - Indirect (attribute)’ some *‘Entire glenohumeral joint (body structure)’*)

‘Prosthetic uncemented total shoulder replacement (procedure)’ is a subclass of the not fully defined concept *‘Uncemented total replacement of joint (procedure)’* which lacks any formal representation of the property “uncemented”.

Another frequent lexical cue is “without”. Most of the concepts with “without” in their label are from *Clinical Findings*, more specifically among those with the semantic tag *Disorder* (1223). 951 are not fully defined, which is conclusive because the formalisation of the meaning of “without” would require the negation operator. Among these concepts we inspected concepts corresponding to the lexical patterns “without complication”, “without infection”, “without disease/finding”. We did not find any wrong classification result regarding the use of “without” in *Clinical Findings* besides the fact that negation is not explicitly modelled. Therefore, no specific ontology pattern can be given. See the following example:

‘Open wound without complication (disorder)’

subclassOf

‘Open wound (disorder)’

and **‘Role group (attribute)’** some

‘Associated morphology (attribute)’ some *‘Open wound (morphologic abnormality)’*)

Many of the *Procedure* concepts with “without” in their label and most of the fully defined ones (103 out of 136) follow the pattern “without contrast”. As it happens with concepts under *Clinical Findings*, the meaning of “without” is not explicitly modelled. We did not find any wrong classification result regarding the use of “without” in *Procedure* apart from the fact that it is not explicitly modelled, e.g. in:

‘Imaging procedure without contrast (procedure)’ **subclassOf**

‘Procedure (procedure)’

and **‘Role group (attribute)’** some (**‘Method (attribute)’** some *‘Imaging - action (qualifier value)’*)

Together with “not”, most of the negated *Situation* concepts use “no”. Among the most common ones are the ones under *Clinical Finding Absent* hierarchy. The following concept ‘No history of migraine (situation)’ is wrongly classified under ‘No history of cardiovascular system disease (situation)’ and under ‘No pain (situation)’. The ontology pattern is the same as the “Absent-Pattern” introduced above.

'No history of migraine(situation)'

subClassOf

'No history of cardiovascular system disease (situation)'

'No pain (situation)'

equivalentTo

'Finding with explicit context (situation)'

and **'Role group (attribute)'** some (

'Associated finding (attribute)' some *'Migraine (disorder)'* and

'Finding context (attribute)' some *'Known absent (qualifier value)'* and

'Temporal context (attribute)' some *'All times past (qualifier value)'* and

'Subject relationship context (attribute)' some *'Subject of record (person)'*)

Table 2 provides an overview of lexical negation patterns and their distribution.

Table 2: Lexical negation patterns; Number of concepts and their semantic type

Lexical Cue	N° total concepts (non-primitive)	Semantic Type	Modelling pattern name	N° total concepts (non-primitive)
“Not”	1191 (576)	Clinical Finding	“Does not”	633 (558)
	323 (238)	Situation	Clinical finding absent	74 (70)
			Procedure with explicit context	228 (168)
“Without”	96 (4)	Procedure	Only modelled textually	96 (4)
“Without”	1331 (591)	Clinical Finding	“Without complication”	125 (59)
			“Without infection”	209 (198)
	357 (136)	Procedure	“Without contrast”	116 (103)
“No”	236 (216)	Situation	Clinical Finding absent	201(194)
			No Family History	9 (6)
			No history of	24 (23)

4. Discussion

Our analysis demonstrated obvious shortcomings of the current modelling of SNOMED CT content with a negative polarity. Expressing this content in OWL-EL, i.e. a language that does not provide negation operators, is problematic. Workarounds that use OWL syntax, but ignore its semantics are inappropriate and lead to improper reasoning results, as e.g. criticised in the case of the NCI Thesaurus [4].

There are several possibilities to partly or fully tackle the problem. However, each of these solutions would require some major re-modelling. Following, we sketch and discuss some of the ones that we foresee:

1. OWL-EL is extended to OWL-DL, i.e. the negation operator is allowed as a logical constructor in the SNOMED CT description logics framework. The drawback here is that even if added to a minor number of axioms, a dramatic drop in reasoning performance would ensue, for all that we know about the theoretical complexity of OWL-EL and the practical experiences with it. In order to avoid a dramatic drop in reasoning, a combination of OWL reasoners and ontology modularization could be used to improve the reasoning performance. In [5] a combination of an EL and more expressive reasoner is used for ontology classification based on the identification of the minimal non-EL subontology. In [6], a similar approach is followed by implementing the MORE reasoner, as a combination of a fully-fledged DL reasoner and a less expressive one.
2. The model of meaning of the SNOMED CT situation hierarchy is revised in the sense that it is taken seriously what it actually is, *viz.* an information model inside SNOMED CT. As such, it should be seen rather as a frame-like model, like openEHR [7] and HL7 FHIR [8], refraining from any formal semantics. Then it could remain structurally unchanged, just excluding it from any transformation into OWL. However, this is just a partial solution, given that SNOMED CT Situation concepts account for only 12% of all negations.
3. Reifying negations. This consists in representing negation within the concept label and avoid using the OWL construct ‘not’. This approach already partly exists, as shown with the example *Imaging procedure without contrast (procedure)*. Such primitive concepts, located in a high place in the hierarchy could then just be co-ordinated with more specific ones like *Computed tomography of facial bones without contrast (procedure)*. On the downside, this does - within OWL-EL - not allow any reference to the concept *Contrast media (substance)* unless a new relation is introduced. And whereas the contrast media are arranged in taxonomic order, hierarchies of reified negated hierarchies would have to be constructed in parallel, cf. Fig. 1. As long as the size of such hierarchies is small, it may still be acceptable. However, if large subhierarchies are expected in the scope of negations, it would lead to a kind of upside-down duplication of large parts of SNOMED CT. Such “ghost hierarchies” would entail significant additional maintenance load, unless they are constructed automatically.

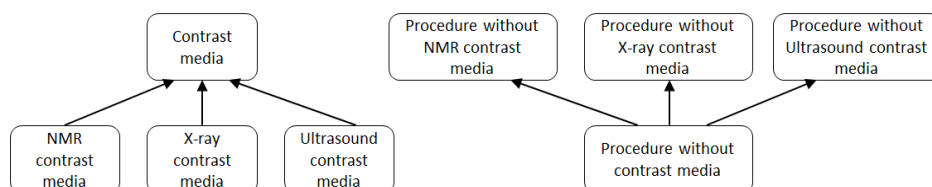


Figure 1. Inverted hierarchies for reified SNOMED CT concepts with negative polarity

5. Conclusion and further work

SNOMED CT Content with more or less explicit negative meaning cannot be neglected. Using ten lexical patterns, 5,823 concepts with negative meaning were identified, mostly in the subhierarchies *Clinical Finding*, *Procedure* and *Situation*. In the best case, negative polarity is implicitly contained in primitive (i.e. not fully defined concepts); in the worst case a negative connotation is ignored in definitional axioms and leads to wrong inferences. A remediation of this situation should start with a systematic scrutiny of primitive concepts in the qualifier value hierarchy that incorporate a negative polarity, like “does not”, “not done”, “absent”. This is necessary because using them in definitions leads to paradoxical results, because their implicit negation is not paralleled by any formal negation: “absent pain” is not a subtype of pain like “severe pain”, despite the same syntactic structure. “Absent pain” implies, e.g. “absent back pain” (among thousand other kinds of pain), whereas “back pain” implies “pain”. There is hardly an alternative to using logical negation to express the missing of something, using a pattern like ‘*Clinical condition*’ and not (**includes** some *Pain*)”. Using more expressive reasoning only with small fragments of the ontology containing negation would be an alternative to explore. Another option would be adding such expressions to SNOMED CT that would be ignored by an OWL-EL reasoner, but at least the wrong inference (that no back pain entails no pain) would be avoided. The expected inferences could be added by subclass statements of the type ‘*Absence of pain*’ subclassOf ‘*Absence of back pain*’. Such axioms could be created by batch processes when generating the OWL version, however, at the price of large additional “upside - down” subclass hierarchies. Future work will concern experimental evaluation of the approaches discussed above.

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