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# Intra-axiom redundancies in SNOMED CT

Kathrin Dentler<sup>a,b,\*</sup>, Ronald Cornet<sup>b,c,1</sup>

- <sup>a</sup> Al Department, VU University Amsterdam, de Boelelaan 1081a, 1081HV Amsterdam, The Netherlands
- b Department of Medical Informatics, Academic Medical Center, University of Amsterdam, Meibergdreef 15, 1105 AZ Amsterdam, The Netherlands
- <sup>c</sup> Department of Biomedical Engineering, Linköping University, SE-581 83 Linköping, Sweden

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#### ABSTRACT

Objective: Intra-axiom redundancies are elements of concept definitions that are redundant as they are entailed by other elements of the concept definition. While such redundancies are harmless from a logical point of view, they make concept definitions hard to maintain, and they might lead to content-related problems when concepts evolve. The objective of this study is to develop a fully automated method to detect intra-axiom redundancies in OWL 2 EL and apply it to SNOMED Clinical Terms (SNOMED CT). Materials and methods: We developed a software program in which we implemented, adapted and extended readily existing rules for redundancy elimination. With this, we analysed occurence of redundancy in 11 releases of SNOMED CT (January 2009 to January 2014). We used the ELK reasoner to classify SNOMED CT, and Pellet for explanation of equivalence. We analysed the completeness and soundness of the results by an in-depth examination of the identified redundant elements in the July 2012 release of SNOMED CT. To determine if concepts with redundant elements lead to maintenance issues, we analysed a small sample of solved redundancies.

Results: Analyses showed that the amount of redundantly defined concepts in SNOMED CT is consistently around 35,000. In the July 2012 version of SNOMED CT, 35,010 (12%) of the 296,433 concepts contained redundant elements in their definitions. The results of applying our method are sound and complete with respect to our evaluation. Analysis of solved redundancies suggests that redundancies in concept definitions lead to inadequate maintenance of SNOMED CT.

Conclusions: Our analysis revealed that redundant elements are continuously introduced and removed, and that redundant elements may be overlooked when concept definitions are corrected. Applying our redundancy detection method to remove intra-axiom redundancies from the stated form of SNOMED CT and to point knowledge modellers to newly introduced redundancies can support creating and maintaining a redundancy-free version of SNOMED CT.

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## 1. Introduction

SNOMED Clinical Terms (SNOMED CT) allows for meaning-based recording and retrieval of clinical information, which thereby becomes (re)usable. One of the advantages of SNOMED CT is its large size and coverage, which on the other hand makes defining new and maintaining existing concepts a challenging task.

http://dx.doi.org/10.1016/j.artmed.2014.10.003 0933-3657/© 2014 Elsevier B.V. All rights reserved. Spackman [1] indicated back in 2001 that concept modellers have been uncertain about which elements are inherited from supertypes and therefore do not have to be added explicitly to a concept definition. Such intra-axiom redundancies, i.e. elements that are already entailed by other elements of the concept definition, are harmless from a logical point of view. However, they impede the maintainability of a terminology [2,3], as they misleadingly suggest that new, meaningful information has been added to a concept.

Moreover, redundant elements might lead to content-related problems when concepts evolve. For example, the rolegroup in the subconcept *Thyroid uptake with thyroid stimulation* was redundant in the July 2012 version of SNOMED CT, as it repeated a rolegroup already contained in the definition of the superconcept *Non-imaging thyroid uptake test*, see Example 1.1. In the subsequent version of SNOMED CT, the method *Radionuclide imaging* 

<sup>\*</sup> Corresponding author. Tel.: +31 20 59 87752. E-mail address: k.dentler@vu.nl (K. Dentler).

<sup>&</sup>lt;sup>1</sup> Ronald Cornet is a member of the Technical Committee of the International Health Terminology Standards Development Organisation (IHTSDO), which publishes SNOMED CT. His position at the IHTSDO, however, had no bearing on the research study or results.

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was removed from the rolegroup in the superconcept, which makes sense for a concept with the name *Non-imaging thyroid uptake test*. However, the method was not removed from the rolegroup in the subconcept, as shown in Example 1.2, which is apparently incorrect. In this paper, we inventory redundant elements in SNOMED CT concept definitions.

**Example 1.1.** Two concept definitions in the July 2012 version of SNOMED CT. The definition of *Thyroid uptake with thyroid stimulation* contains a redundant element, the rolegroup (RG).

```
Non-imaging thyroid uptake test ☐

Radionuclide study of endocrine function ☐

∃RG(∃Method.Radionuclide imaging ☐

∃Procedure site.Thyroid structure ☐

∃Using substance.Radioactive isotope)

Thyroid uptake with thyroid stimulation ☐

Stimulation test ☐ Non-imaging thyroid uptake test ☐

Radionuclide uptake study ☐

∃RG(∃Method.Radionuclide imaging ☐

∃Procedure site.Thyroid structure ☐

∃Using substance.Radioactive isotope)
```

**Example 1.2.** Definitions of the concepts from Example 1.1 in the January 2013 version of SNOMED CT. The definition of *Non-imaging thyroid uptake test* has been corrected, but the previously redundant rolegroup is left unchanged.

```
Non-imaging thyroid uptake test ☐

Radionuclide study of endocrine function ☐

∃RG(∃Method.Radionuclide imaging ☐

∃Procedure site.Thyroid structure ☐

∃Using substance.Radioactive isotope)

Thyroid uptake with thyroid stimulation ☐

Stimulation test ☐ Non-imaging thyroid uptake test ☐

Radionuclide uptake study ☐

∃RG(∃Method.Radionuclide imaging ☐

∃Procedure site.Thyroid structure ☐

∃Using substance.Radioactive isotope)
```

## 2. Background

# 2.1. SNOMED CT concept definitions and rolegroups

SNOMED CT is based on the lightweight Description Logic  $EL^+$  [4]. Its concepts are defined by conjunctions of other concepts as well as role-value pairs that are represented as exists restrictions ( $\exists$ ). These exists restrictions can be either ungrouped or grouped in so-called *rolegroups* "to add clarity to concept definitions. A rolegroup combines an attribute-value pair with one or more other attribute-value pairs. Rolegroups originated to add clarity to *Clinical finding* concepts which require multiple *Associated morphology* attributes and multiple *Finding site* attributes and to *Procedure* concepts which require multiple *Method* attributes and multiple *Procedure site* attributes." [5]. For an indepth analysis of rolegroups, see [6]. Rolegroups are formally represented as an existential restriction on a role named Rolegroup (RG).

Concepts can be either *primitive*, i.e. specified by *necessary* conditions only (denoted by the subsumption operator  $\sqsubseteq$ ) or *fully defined*, i.e. specified by both *necessary and sufficient* conditions (denoted by the equivalence operator  $\equiv$ ). Example 2.1 presents a concept which is fully defined by the conjunction of one concept and two rolegroups.

**Example 2.1.** Fully defined concept definition of *Brain stem contusion with open intracranial wound*. Rolegroups are used to distinguish this concept from "intracranial contusion with open wound of brain stem".

```
Brain stem contusion with open intracranial wound ≡

Contusion of brain with open intracranial wound □

∃RG(∃Associated morphology.Open wound □

∃Finding site.Intracranial structure) □

∃RG(∃Associated morphology.Open contusion □

∃Finding site.Brainstem structure)
```

#### 2.2. Trivially and non-trivially primitive concepts

For our evaluation, we distinguish *trivially primitive concepts*, that are primitive and subsumed by one concept only, and *nontrivially primitive concepts*, that are primitive and described by the conjunction of one or more concepts and exists restrictions. With regard to Example 2.2, we refer to the concept *Brain tissue structure* as trivially primitive, and to *Structure of lobe of brain* as non-trivially primitive.

**Example 2.2.** Two primitive concept definitions, *Brain tissue structure*, which is trivially primitive, and *Structure of lobe of brain*, which is non-trivially primitive.

```
Brain tissue structure 
☐ Brain part

Structure of lobe of brain 
☐

Brain part 
☐ Brain tissue structure
```

## 2.3. Redundant elements in SNOMED CT concept definitions

An element that is part of a concept definition, i.e. a concept or an existential restriction, is redundant if it has been stated explicitly even though it is already entailed by another element of the concept definition, i.e., by superconcepts, other existential restrictions, or a combination thereof. Therefore, we define an element to be redundant if it is more general than or equivalent to an element that is contained in the definition of the same concept or a stated superconcept. Redundant elements can be eliminated without affecting the ontology's logical closure. Even more, elimination of redundant elements only changes the syntax of concept definitions, but does not influence their semantics. For example, the concept *Brain part* in the definition of the concept *Structure of lobe of brain* in Example 2.2 is redundant as it subsumes the concept *Brain tissue structure*.

## 3. Materials and methods

We employed all 11 versions of SNOMED CT that were convertible to OWL, i.e. the January 2009 version to the January 2014 version. We converted these versions with the Perl script that is provided with each release of SNOMED CT. This script makes use of two tables: concepts and stated relationships. The latter faithfully represents the information as it was specified by modellers, and has been released since 2009.

We relied on the high-performance reasoner ELK [7] to classify SNOMED CT, and to check for subsumption and equivalence relationships between concepts and roles, while Pellet [8] was used in our evaluation to explain equivalence relationships that were hard to reproduce manually. We relied on the OWL API [9] to carry out all experiments.

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3.1. Method to detect redundant elements in SNOMED CT concept definitions

We exploit the relatively simple structure of SNOMED CT concept definitions to detect intra-axiom redundancies. To this end, we adapted and extended the rules 1 to 3 of redundancy elimination for concept definitions that contain rolegroups as defined by Spackman et al. [10] (and adopted their original numbering). The rules are based on Definition 3.1.

**Definition 3.1.** More general or equivalent exists restriction. An exists restriction is more general than or equivalent to another exists restriction whenever both its role and its value concept subsume or are equivalent to the respective elements in the other exists restriction.

```
\exists R.C \supseteq \exists S.D \Leftrightarrow (R \supseteq S) and (C \supseteq D)
```

In SNOMED CT, all concept definitions are merely conjunctions of ungrouped or grouped exists restrictions and superconcepts. Consequently, the rules define for each of these elements whether they are redundant:

1 An ungrouped exists restriction is redundant when it is more general than or equivalent to an ungrouped exists restriction within the definition of *the same concept or a superconcept*.

```
(\exists R.C \sqcap \exists S.D \sqcap \exists T.E) \equiv (\exists S.D \sqcap \exists T.E) \Leftrightarrow \exists R.C \supseteq \exists S.D
```

2 A rolegroup is redundant when all its exists restrictions are more general than or equivalent to those contained in another rolegroup in the definition of *the same concept or a superconcept*.

```
(RG(\exists R_1.C_1 \sqcap \ldots \sqcap \exists R_n.C_n) \sqcap RG(\exists S_1.D_1 \sqcap \ldots \sqcap \exists S_m.D_m))
\equiv RG(\exists S_1.D_1 \sqcap \ldots \sqcap \exists S_m.D_m)
\Leftrightarrow \forall i = 1, \ldots, n \exists j = 1, \ldots, m | \exists R_i.C_i \supseteq \exists S_i.D_i
```

3 An exists restriction is redundant within a rolegroup when it is more general than or equivalent to another exists restriction in *the same rolegroup*.

```
RG(\exists R.C \sqcap \exists S.D \sqcap \exists T.E) \equiv RG(\exists S.D \sqcap \exists T.E) \Leftrightarrow \exists R.C \supseteq \exists S.D
```

4 A concept is redundant when it is more general than or equivalent to one of the other concepts in the definition of *the same concept* or a superconcept.

```
(C \sqcap D) \equiv D \Leftrightarrow C \supseteq D
```

Note that rule 3 is an exception with regard to our redundancy definition, as it does not concern an element of a concept definition, but an element within an element. To test whether a concept is defined redundantly, these four rules are applied to a concept and all its stated superconcepts. As the rules are independent from each other, the order of their execution should not influence the obtained results.

#### 3.2. Evaluation of our method

To evaluate the results obtained by the application of the four rules of redundancy detection, we assessed the completeness and soundness of its output. In absence of a gold standard, we measured completeness by matching our findings to definitions that are likely to be redundant according to Cornet's and Abu-Hanna's method [11], and soundness by checking whether the logical closure is unchanged after classifying a version of the ontology from which all redundant elements have been removed.

3.2.1. Completeness: comparison of identified redundant concepts to redundant concepts according to Cornet's and Abu-Hanna's method

Cornet's and Abu-Hanna's method [11] detects concepts with equivalent definitions in terminological systems represented in a description logic, to address the problems of redundancy and underspecification. Concepts that become equivalent to any superconcept when applying this method are likely to be defined redundantly [12]. Let us regard Example 3.1, which presents a sample group of equivalent concepts that can be detected by applying this method.

**Example 3.1.** Group of concepts with equivalent concept definitions.

Here, we can make two interesting observations. First, we see three concepts with definitions that obviously become equivalent when making these concepts fully defined. Second, the three concepts become equivalent to their superconcept *Finding of volume of heart sounds*, and thus, they are likely to be defined redundantly. And indeed, four levels up the concept hierarchy, we encounter their common superconcept presented in Example 3.2, which already contains a rolegroup that defines the *Finding site* to be the *Heart structure*.

**Example 3.2.** Explanation for redundancy of concept definitions in Example 3.1.

```
Cardiac finding ☐

Cardiovascular finding ☐

∃RG(∃Finding site.Heart structure)
```

We evaluated the results obtained by the application of the four rules of redundancy detection by checking whether the concepts that are likely to be redundant according to Cornet and Abu-Hanna are indeed contained in the identified set of redundant concepts. In order to detect redundant definitions, we applied the approach proposed by Cornet and Abu-Hanna as follows:

- 1 Replace each non-trivially primitive concept by a fully defined concept with the same definition.
- 2 Classify the ontology.
- 3 For each concept in the ontology, retrieve equivalent concepts from a reasoner.
- 4 Identify concepts that have become equivalent to any stated superconcept, as those are likely to be defined redundantly.
- 5 Identify and exclude indirect redundancies that emerge due to concepts being subsumed by the conjunction of concepts with equivalent definitions such as in Example 3.3 and wrongly

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identified redundancies due to the propagation of equivalence such as in Example 3.4.<sup>2</sup>

**Example 3.3.** Concepts without intra-axiom redundancy: Because *Midwifery personnel* and *Professional midwife* have the same definitions, they become equivalent when applying Cornet's and Abu-Hanna's method. And because *Auxiliary midwife* is being subsumed by the two of them, it also becomes equivalent.

```
Auxiliary midwife ☐
Professional midwife ☐ Midwifery personnel

Professional midwife ☐
Medical, dental, veterinary/related worker ☐
Health visitor, nurse/midwife

Midwifery personnel ☐
Medical, dental, veterinary/related worker ☐
Health visitor, nurse/midwife
```

Cornet's and Abu-Hanna's method does not necessarily retrieve all redundant concepts. For example, a concept can refine its stated superconcept and additionally contain redundant elements. Likewise, redundant elements in fully defined concept definitions are not detected by Cornet's and Abu-Hanna's method. Therefore, the evaluation of the results of the four rules of redundancy detection can only be partial.

**Example 3.4.** Example for wrongly identified redundancy. The concepts *Pancreatic function outside reference range* and *Measurement finding outside reference range* would be equivalent if all involved concepts were fully defined.

```
Pancreatic function outside reference range 
Measurement finding outside reference range 
∃RG(∃Has interpretation.Outside reference range 
∃Interprets.Pancreatic function test)

Measurement finding outside reference range 
Measurement finding 
∃RG(∃Has interpretation.Outside reference range 
∃Interprets.Measurement procedure)

Pancreatic function test 
Measurement procedure 
Measurement procedure 
∃RG(∃Has Method.Measurement - action)

Measurement procedure 
Procedure by method 
∃RG(∃Has Method.Measurement - action)
```

# 3.2.2. Soundness: preservation of logical closure

Deleting redundant parts of concept definitions should not affect the logical closure, and therefore a change in the concept hierarchy would indicate the removal of a non-redundant part of a concept definition. Thus, we deleted all identified intra-axiom redundancies and checked whether the computed concept hierarchy obtained from classifying the manipulated version is the same as the one obtained from classifying the original version by bidirectional comparison of both versions to the official SNOMED CT distribution.

**Table 1**Detected concepts with redundant elements. The examples in column 'example' refer to the examples disseminated along the paper.

Rule	Concepts	Example and explanation
1 (ungrouped exists restriction)	7874	Example 4.1: The ungrouped exists restriction $\exists Has\ active\ ingredient.$ Thymoxamine (substance) is redundant, as it is already contained in the superconcept Thymoxamine (product).
2 (rolegroup)	26,599	Example 2.1: The first rolegroup is redundant, as it is more general than the second one, because open wound subsumes open contusion, and Intracranial structure subsumes  Brainstem structure
3 (grouped exists restriction)	6	Example 4.2: The exists restriction ∃Associated morphology. Traumatic abnormality in the first rolegroup is redundant, as Traumatic abnormality subsumes Closed traumatic abnormality.
4 (concept)	531	Example 2.2: The concept <i>Brain part</i> is redundant as it subsumes the concept <i>Brain tissue structure</i> .

#### 3.3. Comparison of SNOMED CT releases

We generated sets of concepts with redundant elements for each of the SNOMED CT releases since January 2009. To determine changes, we compared each release with its successor. Four types of changes were distinguished: new concepts with redundant elements, existing concepts with new redundant elements, existing concepts with removed (solved) redundant elements, and concepts with redundant elements which were inactivated.

To assess whether redundancies cause maintenance issues, we took a small sample of concepts with redundant elements. For each of the changes between versions of SNOMED CT, we randomly selected two concepts for which redundancies were solved. Hence, a total of 20 concepts were manually reviewed to assess whether the changes made to the concept definitions were adequate.

# 4. Results

4.1. Redundant elements in concept definitions in the July 2012 version

Applying the four rules of redundancy detection on the July 2012 version of SNOMED CT, 35,010 (12%) of the 296,433 concepts were identified to contain redundant elements in their definitions. Table 1 gives an overview of the results, only regarding the first explanation for these redundancies (the rules were applied in the same order as they are presented in this paper). 11,858 of these concepts are fully defined, and 23,152 non-trivially primitive.

**Example 4.1.** Parenteral form thymoxamine.

```
Parenteral form thymoxamine (product) ≡

Thymoxamine (product) □

∃Has active ingredient.Thymoxamine (substance)

Thymoxamine (product) ⊑

Alpha blocking vasodilator □ Alpha 1 adrenergic blocking agent □

∃Has active ingredient.Thymoxamine (substance)
```

<sup>&</sup>lt;sup>2</sup> These cases could be prevented by applying the method only on one superconcept - subconcept pair at a time instead of the entire SNOMED CT. We did not apply this method because it is not feasible even with very fast classification.

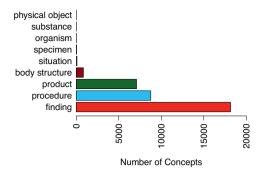


Fig. 1. SNOMED CT categories of concepts with redundancies.

### **Example 4.2.** Closed skull fracture with intracranial injury.

```
Closed skull fracture with intracranial injury ≡

Fracture of skull □

∃RG(∃Finding site.Intracranial structure □

∃Associated morphology.Traumatic abnormality □

∃Associated morphology.Closed traumatic abnormality) □

∃RG(∃Associated morphology.Fracture, closed □

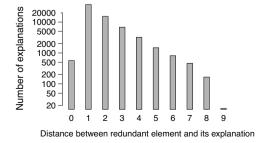
∃Finding site.Bone structure of cranium)

Explanation:
Closed traumatic abnormality □ Traumatic abnormality
```

Fig. 1 shows the SNOMED CT categories that the concepts with redundant elements belong to. Most concepts belong to the categories *finding*, *procedure* and *product*. We found that 18% of all concepts in the category *finding*, 16% of all concepts in the category *procedure* and 41% of all concepts in the category *product* contained redundant elements. In all other categories, the percentage of concepts with redundant elements was less than 3%.

Fig. 2 depicts the distances between redundant concepts and the concepts containing the explanation for the redundancy. A distance of 0 is interesting as it makes a concept redundant with regard to its own definition. But also long distances are interesting: an element is introduced, not repeated for some concepts down the hierarchy, but then it is. The concept Measurement of Human T-lymphotropic virus 1 recombinant glycoprotein 21 antibody and Human T-lymphotropic virus 2 recombinant glycoprotein 21 antibody is one of the concepts with the longest distance to its explanation (9 levels up in the hierarchy).

An exhaustive search for all redundant elements and all explanations resulted in 65,336 explanations: 13,808 for rule 1, 50,680 for rule 2, 6 for rule 3 and 842 for rule 4. The maximum number of explanations was 16 for the concept *Late congenital syphilitic meningitis*. The concept with the most (6) redundant elements was Diphtheria + tetanus + pertussis + poliomyelitis + recombinant hepatitis B virus + recombinant haemophilus influenzae type B vaccine.



**Fig. 2.** Distances between redundant concepts and the concepts containing the explanation.

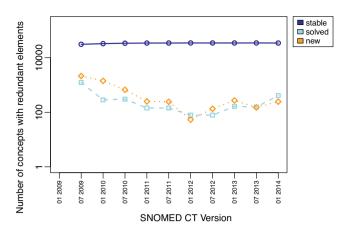


Fig. 3. Development of concepts with redundant elements according to versions

#### 4.2. Evaluation of our method

#### 4.2.1. Completeness

Applying Cornet's and Abu-Hanna's method, 45,975 concept definitions have been identified for which at least one other concept with a logically equivalent definition exists. Of these, 12,823 definitions of non-trivially primitive concepts are equivalent to the definition of at least one of their stated superconcepts.

12,094 of these redundancies have been confirmed to be redundant by our method to detect intra-axiom redundancies. 698 out of the 729 non-confirmed redundancies were subsumed by the conjunction of concepts, like in Example 3.3. For the remaining 31 non-confirmed redundancies, we successfully generated explanations with Pellet based on the manipulated version of SNOMED CT. A manual revision confirmed that all explanations contained further axioms that have been re-defined from being primitive to fully defined, like the explanation given in Example 3.4. Therefore, the results of our method are complete with regard to Cornet's and Abu-Hanna's method.

# 4.2.2. Soundness

We generated the logical closure of both the original and the manipulated OWL versions of SNOMED CT, and compared the computed class hierarchies to the one contained in the official distribution. The OWL versions and the database table contained exactly the same set of 438,554 subclass axioms and "is-a" relations, respectively.

# 4.3. Comparison of SNOMED CT releases from January 2009 to January 2014

Fig. 3 shows the development of concepts with redundant elements over time. For each version, the plot shows the concepts with redundant elements that have not changed since the previous version (stable), as well as concepts with solved or new redundancies.

We found that 24% (1325/5589) of all new concepts with redundant elements have been newly added, and that 26% (773/3014) of the concepts with solved redundant elements are due to inactivated concepts. 14% of the concepts with solved redundant elements remained unchanged, and thus have been solved due to a change in a superconcept.

The analysis of our random sample showed that at least two of these (i.e., 10%) were changed inadequately. For example, in the July 2012 version the concept *Drug-induced impotence* redundantly defined the *finding site*, *Erectile structure of penis*, which was already defined in the ancestor *Male erectile disorder*. In the January 2013 release, this finding site was no longer part of the definition of *Male* 

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*erectile disorder* (which makes sense), but it was still part of the definition of *Drug-induced impotence*, which is inconsistent.

#### 5. Related and future work

Campbell et al. [13] proposed a semantics-based conflict identification method for the distributed development of logic-based terminologies. Conflicts that can be detected are *multiply-defined term conflicts* and *non-unique definition conflicts*. Multiply-defined terms refer to the same term, but do not have the same definitions. They can be sub-classified into semantically-conflicting definitions and semantically equivalent definitions. When the definitions are semantically equivalent, it is possible to automatically adopt the simplest or the most comprehensive definition. Non-unique definitions are non-synonymous terms that received identical definitions by different editors.

Other (automated) auditing methods to ensure the quality aspect non-redundancy [14] focused mainly on the detection of equivalent concepts. Cimino has developed a method to identify multiple synonymous concepts and applied it to the 2001 UMLS Metathesaurus [15]. Grimm and Wissmann [3] provide methods to compute non-redundant ontologies, and Entendre [16] makes users aware of redundancies.

The IHTSDO<sup>3</sup> describes methods to convert concepts into normal forms, some of which imply the elimination of redundancies, and Peng et al. [17] have proposed a method to identify redundant classifications, i.e. unnecessary, simultaneous assignments to sub- and superconcepts. The Ecco tool [18] facilitates the analysis of ontology differences by applying methods to syntactically or semantically detect effectual changes as well as ineffectual changes such as adding or deleting intra-axiom redundancies.

An interesting direction of future work would be to generalise our method. In principle, our definition of a redundant element could be operationalised directly by checking whether an element is more general than or equivalent to an element that is contained in the definition of the same concept or a stated superconcept.

#### 6. Discussion and conclusions

Our results show that 35,010 (12%) of all 296,433 SNOMED CT concepts of the July 2012 version were defined redundantly. These redundancies unnecessarily impede the work of concept modellers, and ultimately the quality of a terminology. Redundant elements in concept definitions are introduced and solved in comparable amounts in all versions of SNOMED CT between January 2009 and January 2014. On average, about three quarters of the introduced redundancies are caused by changes in definitions of existing concepts, rather than by creating new concepts. This indicates that redundancy is easily overlooked and hard to detect. Our own experience confirms that manual search for the causes of redundancies can be a tedious task. Therefore, we suggest to remove them from the stated relationships. To reach this goal,

the four rules of redundancy detection would have to be applied once to SNOMED CT in its entirety. Further redundancies should be avoided by pointing concept modellers to newly introduced redundancies in the definitions of the concepts they are currently working on, and explaining why these elements are redundant. As shown in Fig. 2, redundancy is generally caused by nearby superconcepts, so that the explanations will most probably be intuitive. For this task, the four rules of redundancy detection could be applied as a background process of terminology editing tools to the concepts that are currently being edited. In order to support these goals, we make both our tools and our results freely available.

#### References

- [1] Spackman KA. Normal forms for description logic expressions of clinical concepts in SNOMED RT. In: Proceedings of the AMIA symposium. 2001. p. 627–31.
- [2] Schlobach S, Cornet R. Logical Support for Terminological Modeling. In: Fieschi M, Coeira E, Li Y-CJ, editors. Studies in health technology and informatics, vol. 107. IOS Press; 2004. p. 439–43.
- [3] Grimm S, Wissmann J. Elimination of redundancy in ontologies. In: Antoniou G, Grobelnik M, Simperl E, Parsia B, Plexousakis D, De Leenheer PJ, Pan J, editors. The semantic web: research and applications. Berlin, Heidelberg: Springer; 2011. p. 260–74.
- [4] Baader F, Lutz C, Suntisrivaraporn B. Is tractable reasoning in extensions of the description logic el useful in practice? Proceedings of the methods for modalities workshop. Kluwer Academic Publishers; 2005.
- [5] International Health Terminology Standards Development Organisation. SNOMED CT Editorial Guide, Tech. rep. Copenhagen, Denmark: International Health Terminology Standards Development Organisation; 2014.
- [6] Cornet R, Schulz S. Relationship groups in SNOMED CT. In: Adlassnig K-P, Blobel B, Mantas J, Masic I, editors. Studies in health technology and informatics, vol. 150. IOS Press; 2009. p. 223–7.
- [7] Kazakov Y, Krötzsch M, Simančík F. Concurrent classification of EL ontologies. In: Aroyo L, Welty C, Alani H, Taylor J, Bernstein A, Kagal A, Noy N, Blomqvist E, editors. The semantic Web – ISWC 2011. Berlin Heidelberg: Springer; 2011. p. 305–20.
- [8] Sirin E, Parsia B, Grau BC, Kalyanpur A, Katz Y. Pellet: a practical OWL-DL reasoner. Web Semant: Sci Serv Agents World Wide Web 2007;5(2):51–3.
- [9] Horridge M, Bechhofer J. The OWL API: A Java API for OWL ontologies. Semantic Web 2011;2(1):11–21.
- [10] Spackman KA, Dionne R, Mays E, Weis J. Role grouping as an extension to the description logic of Ontylog, motivated by concept modeling in SNOMED. In: Proceedings of the AMIA symposium. 2002. p. 712–6.
- [11] Cornet R, Abu-Hanna A. Auditing description-logic-based medical terminological systems by detecting equivalent concept definitions. Int J Med Informa 2008;77(5):336–45.
- [12] Cornet R, Abu-Hanna A. Two DL-based methods for auditing medical terminological systems. In: Proceedings of the AMIA symposium. 2005. p. 166–70.
- [13] Campbell KE, Cohn SP, Chute CG, Shortliffe EH, Rennels G. Scalable methodologies for distributed development of logic-based convergent medical terminology. Methods Inform Med 1998;37(4-5):426–39.
- [14] Zhu X, Fan J-W, Baorto DM, Weng C, Cimino JJ. A review of auditing methods applied to the content of controlled biomedical terminologies. J Biomed Inform 2009;42(3):413–25.
- [15] Cimino JJ. Battling Scylla and Charybdis: the search for redundancy and ambiguity in the 2001 UMLS metathesaurus. In: Proceedings of the AMIA symposium. 2001 p. 120-4
- [16] Denaux R, Thakker D, Dimitrova V, Cohn AG. Entendre: interactive semantic feedback for ontology authoring. In: Tech rep, Demo Track ISWC 2011. 2011.
- [17] Peng Y, Halper MH, Perl Y, Geller J. Auditing the UMLS for redundant classifications. In: Proceedings of the AMIA symposium. 2002. p. 612–6.
- [18] Gonçalves RS, Parsia B, Sattler U. Ecco: A Hybrid Diff Tool for OWL 2 ontologies. In: 9th OWL experiences and directions workshop. 2012.

 $^{\rm 4}\,$  It should be noted that applying the four rules of redundancy detection to the

entire SNOMED CT is computationally feasible but not in an interactive environment (about 6 h on a laptop equipped with a 2.8 GHz Intel Core 2 Duo processor and 8 GB of physical memory). However, analysing only one concept is sufficiently fast to be executed as a background process.

<sup>&</sup>lt;sup>5</sup> https://github.com/kathrinrin/redundancies (Accessed: 1 October 2014).

<sup>&</sup>lt;sup>3</sup> http://www.ihtsdo.org (Accessed: 1 October 2014).