

Response to reviewers for the MEDINFO 2019 paper

**Expanding Evolutionary Terminology Auditing with Historic Formal and Linguistic Intensions:
a Case Study in SNOMED CT**

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Decision: accepted as Full Paper & Student Paper

Comments to authors:

SPC member:

The topic of this paper is interesting but as outlined by reviewers 2 and 3, more details should be provided concerning the methods.

AUTH: This has been taken care of. See comments there.

Reviewer 1:

This paper demonstrated the method of quantified evaluation of the adherence to the criterion of concept permanence on SNOMED-CT and presented the issues of SNOMED-CT.

I hope that the specific issue clarified in this research will be disclosed and improve SNOMED-CT.

AUTH: the acceptance of this paper in the proceedings will at least disclose the issues. Whether SNOMED CT will address them is beyond the authors' capacities.

Reviewer 2:

This paper presented a case study on auditing evolutionary concept changes in SNOMED CT, the largest clinical terminology. The study focused on analyzing historical changes with respect to the concepts' formal intensions (or supertypes) and linguistic intensions (or names). The historical changes of concepts have been properly modelled and automatically audited. Intuitive examples have been provided. Overall, the paper is well-written and easy to follow.

The reviewer has the following comments to further improve the paper.

* The method to suggest missing subsumers in a concept's transitive closure lacks of details. It is unclear how a missing subsumer can be identified by the method. More details would be appreciated.

Auth: we explained this better in the paragraph in which we introduced the notion of 'suspiciously gapped' signature.

* For the rule-based string transformation algorithm, it is unclear how the knowledgebase was manually constructed and how reliable it is. How large is the knowledgebase? Does it contain sufficient rules?

Auth: how the KB was constructed deserves a paper on its own and is beyond the scope of this paper. We did add that 99 rules were used.

* The caption of Table 3 is a bit misleading. The table only shows an example of the string transformation algorithm rather than the actual algorithm.

Auth: we changed the caption to 'string transformation algorithm example'

* Page 3:

- Please fix the grammatical issue for this statement: "which are not intended to be identical their semantic similarity."

Auth: we did so and rewrote the sentence as 'But obviously, it renders this algorithm inappropriate for computing the semantic similarity of distinct concepts on the basis of their linguistic intensions'

- "39,771 (=340,639-300868, 11.68%)" : a comma is missing for the number "300868"

Auth: corrected

* Table 2 and Table 3:

- R1, R2, R3 appear in both Table 2 and Table 3, which may confuse the readers. It would be nice to use different symbols denoting them since they don't share the same meanings.

Auth: we changed the symbols in Table 2 using 'S'n and 'V'n rather than 'R'n, standing for 'Set' and 'Value' respectively.

* This study mainly focused on analyzing the subsumption relations of concepts. The reviewer is curious about the evolutionary changes regarding the logical definitions (including other defining relations) of concepts.

Auth: we would gladly accept funding to perform the suggested work.

Reviewer 3:

Authors describe a formal way to quantify the evolution of concept's meaning. Thus, they analyse how the changes in concepts description can lead to different interpretations of their meaning. They compute the formal and linguistic intentions of the concepts and defined signals to find the variation of meaning induced by the modifications. It is an original approach to audit SNOMED CT considering the assumption that the meanings of its concepts represented by a unique code are permanent. Despite the originality of the work, the article needs to be improved.

"The first one is to assess the extent to which the principle of concept adherence is adhered to and whether adherence can be quantified by resorting to SNOMED CT's history mechanism": This part is very ambiguous. It gives the impression that there is confusion between "concept" previously used in the article as a type of SNOMED CT entity and the meaning of each entity.

Auth: there was actually a typo in that sentence; rather than 'concept adherence', we meant 'concept permanence'. That has been corrected. Thanks for noticing this. The sentence now reads: "The first one is to assess the extent to which the principle of concept permanence is adhered to and whether adherence to this principle can be quantified by resorting to SNOMED CT's history mechanism."

“ For ontologies that do not explicitly adhere to a view based on Ontological Realism...” confusing: the meaning of the other ontologies cannot be computed?

Auth: we changed the sentence as follows: “Since SNOMED CT is an ontology that does not explicitly adhere to a view based on Ontological Realism [12], the meaning of a SNOMED CT concept can be thought of as ...”

The historical signature of concepts is very important. However, it is introduced in a wordy sentence that is difficult to understand.

Auth: we shortened the sentence a bit

The notions of “supertype” and “supertype relations” are introduced without explanation.

Auth: (1) ‘Supertype’ is a commonly used synonym for ‘subsumer’. Nevertheless, we replaced every occurrence of ‘supertype’ in the paper by ‘subsumer’.

(2) We never intended the phrase ‘concept supertype relation’ to be read as ‘(concept)(supertype relation)’. We therefore changed each occurrence thereof in ‘concept-subsumer pair’ and defined such pair by means of the following sentence: “Concepts C1 and C2 form a concept-subsumer pair if and only if C2 is in the transitive closure set of C1.”

Variables, used for the analysis process, should be clearly listed. The contribution of these variables must also be discussed.

Auth: They were, although we agree that the word ‘include’ might have caused the confusion. We replaced it by ‘are’.

Methodology must be more systematized and many aspects need to be clarified:

- The algorithm of the linguistic intentions must be explained, the rules described and some appropriate examples provided.

Auth: This is explained, and an example is provided in Table 3.

- Explain the sampling strategy for formal intensions analysis.

Auth: we explained that our random samples were generated using a random number generator. The population from which they were selected was already specified.

- The mechanism of providing a missing subsumer must be described and better explained. (For example, I do not understand why a 87290003: Congenital anomaly of head (disorder) must be subsumed by 204223000: Ear, face and neck congenital anomalies (disorder). Or a “disorder” be subsumed by a “finding”. These subsumptions seem incorrect to me).

Auth: We added the sentence “Decisions for whether a supertype is truly missing were based on SNOMED CT’s editorial guidelines [9].” We not only agree with this reviewer that a disorder being subsumed by a finding is a mistake, we even consider it plain nonsense as obviously disorders exist whether or not they are found. But that is unfortunately not how SNOMED CT perceives that part

of reality. We made it clear in the introduction that it was our goal to assess the extent to which SNOMED CT authors follow SNOMED CT guidelines, not the extent to which SNOMED CT's view differs from our – or this reviewer's – view on reality. Thus whenever a disorder concept does not have an existing corresponding finding concept in its transitive closure, there is a missing subsumer per SNOMED CT's own concept model.

- Explain why providing a subsumer and not proposing the deletion (and eventually the replacement) of the concepts for which the algorithm has recognized a change in the meaning?

Auth: this question is not clear to us since we do not provide subsumers at all, nor propose anything. We just highlight, as exemplified in Tables 1 and 5, that there were subsumers in earlier versions which seem to be correct, but disappeared in later versions from being subsumers, despite still existing as concepts in these later versions. What we also highlight in Table 1 is what the possible cause might be of this unjustified disappearance of multiple subsumers: the justified deletion of a subsumer (for example because of duplication as is the case in Table 1) which is subsumed by the missing subsumers.

- It is said that the meaning of a concept is a COMBINATION of three aspects. Only two of them are described because the third was not accessible. Explain why the methodology does not use a combination of the two computed intensions?

Auth: because there is obviously no point in doing so: a change in one aspect is sufficient for a change in the whole. Since formal intension and linguistic intension are completely independent of each other, it is impossible that a change in the linguistic intension of a concept would compensate some change in the formal intension such that there would be 'no change' in the COMBINATION at all. Furthermore, it was already clearly specified in the first paragraph of the method section that 'whether two concepts have the same meaning can then in theory be determined by applying appropriate similarity functions to EACH of the three aspects followed by an assessment of whether the similarities are sufficiently high'. Anyhow, we refrained from mentioning 'combination'.

Corrections are needed: "What is, for instance, the scope of 'partial' in a super-type? A 'resection of stomach fundus' (Table 5), whether complete or partial wrt. the fundus is for sure partial wrt. the stomach."

Auth: we don't see what sort of correction would be required here. Perhaps the reviewer did not understand 'wrt.' as abbreviation for 'with respect to'? We thus reformulated the sentence without changing the intended meaning.