Appendix

A review of auditing techniques for the Unified Medical Language System

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Systematic Screening Process of the Related Papers

We initially used the keywords “UMLS”, “Auditing”, “Quality Assurance”, “Consistency”, “Inconsistency”, “Ambiguity”, “Redundancy”, “Aligning”, “Errors” in the following query: “(UMLS OR Unified Medical Language System OR Metathesaurus) AND (Auditing OR QA OR Quality Assurance OR Consistency OR Inconsistency OR Ambiguity OR Redundancy OR Aligning OR Errors)”. The recall rate of relevant articles with which the research team is familiar was low.

We then expanded the search query with more keywords as follows: “(UMLS OR Unified Medical Language System) AND (Concepts OR Relationships OR Semantic network OR Semantic type OR Relation OR terms OR Metathesaurus) AND (Auditing OR QA OR Quality Assurance OR Assessing OR Evaluating OR Analysis OR Analyzing OR Consistency OR Inconsistency OR Enriching OR Enhancing OR Matching OR Ambiguity OR Validating OR Modeling OR Redundancy OR Improving OR Aligning OR Mapping)”.

This query resulted in 1211 articles for further screening. As this process may miss relevant articles published in venues that are not indexed or not retrieved by PubMed (i.e., the ones included in ACM Digital Library or Google Scholar but not in PubMed, the ones without certain keywords such as “UMLS”), we sought to retrieve more relevant papers in two ways. First, to look for PubMed papers which did not conform to the above query, we just tried to search with the keyword “UMLS”. About 1500 papers were retrieved and scanning their titles we found few missed papers. Second, to obtain papers not indexed by PubMed, we conducted several queries on Google Scholar, each combining “UMLS” with one keyword, e.g., “auditing”, “QA”, “errors”, and “inconsistencies”. The lists returned were very long, and for each search we scanned the titles of only first 10 pages. Those “quality measures” for the above systematic review, found 25 extra papers, which were not retrieved from PubMed, resulting in 1236 records. In the title screening process (Step 3), co-authors ZH and DW separately coded the relevance of each record based on inclusion and exclusion criteria (see Table 1). They included the records for abstract/full-text screening as long as at least one of the two reviewers coded “Yes” for the records, resulting in 168 records, where, the two coders agreed on including 92, excluding 1068, and disagreed on only 76 for an agreement rate of 93.9%. Of the 76 disagreements one coder was positive on 31 and the other on 45 papers. Hence, the discrepancies were not because one of the coders was more inclusive than the other.
In the abstract/full-text screening (Step 4), the two coders first coded the 168 records separately, and then discussed to resolve the conflicts. The ones that are still questionable were sent to all the authors to review. Finally, 83 articles were included in this review. Figure 2 shows the PRISMA flow chart for identifying the papers for this review, including the distribution of the rejected 85 papers by the various exclusion criteria.

**Discussion of the Systematic Screening Process of the Related Papers**

As reported, the recall for the initial criteria of the systematic review was too low. Based on some known papers missed the criteria was extended, for the price of lowering the precision and many irrelevant papers were retrieved. It was improper to tailor further the query. In Methods, we described how the rest of the 25 papers (mostly in venues not indexed by PubMed) were obtained. Some reviews (e.g., [14]) consider coverage in auditing of terminologies. We excluded it since it is about the content of a terminology and not about the aspects of concepts which was the focus of this review. In contract, we included enrichment and alignment identifying missing concepts in sources, and indirectly completing the Metathesaurus.

We followed the PRISMA approach for selecting articles for this review. We note that by the end of step 3 of the systematic review, both the coders agreed on including 92 papers of 1237 and disagreed on 76 others. Finally, 83 of the 92 were included after reviewing abstracts/full text of the articles, i.e., nine papers were removed during the extensive step 4 of the systematic review but none of the 76 disputed papers were added. This suggests that our retrieval strategy was comprehensive and that our initial screening criteria were reproducible. We observe that 25 of the papers are from sources that are not retrieved from PubMed, demonstrating the importance of expanding the search strategy beyond PubMed to databases that are not limited to biomedical content.
**Appendix Table 1.** Number of errors found/reported/corrected in the UMLS. Results reflect the specific version of the UMLS used in each study.

| Ref | ASPE | AT | KNW | Free-Text Comment | Numbers of Concepts /Synonyms | Number of Relationships (IS-A and/or semantic relationships) | Number of Semantic Type (ST) Assignments |
|-----|------|----|-----|-------------------|-------------------------------|-------------------------------------------------------------|----------------------------------------|
| Bodenreider et al. 2004 [10] | STA HREL | AH | IEK | Only discusses how their methods can be used to identify semantic type inconsistencies. No numbers provided. | N/A | N/A | N/A |
| Cimino et al. 1998 [11] | CCNS STA HREL LREL | AH AS AS AS | IK IK IK IK | N/A | 3274 redundant concepts | 544 IS-A relationships were incorrect | 1817 concepts with ambiguous STs |
| Mougin et al. 2014 [17] | HREL LREL | AH | IK | Evaluation of multiply-related concepts in UMLS | N/A | 2880 (0.7%) multiply-related concept pairs present contradictory combinations | N/A |
| Mougin et al. 2009 [18] | STA | AH | IEK | N/A | N/A | N/A | 91 concepts with multiple Semantic Groups were inappropriately categorized. |
| Wei et al. 2012 [19] | STA | AH | IEK | N/A | N/A | N/A | 173 concepts with incorrect ST assignment |
| Gu et al. 2016 [20] | STA | AH | IEK | N/A | N/A | N/A | 241 concepts with ST assignment errors |
| Reference           | Method | Levels | Sampling | Synonyms Identified | Unclassified Errors | Notes                                                                 |
|---------------------|--------|--------|----------|---------------------|---------------------|----------------------------------------------------------------------|
| He et al. 2014 [21] | STA    | AS     | IK       | N/A                 | N/A                 | 12161 concepts with redundant ST assignments in 2001                |
| Geller et al. 2013  | STA    | AS     | IEK      | N/A                 | N/A                 | 278 concepts with a wrong ST assignment                             |
| He et al. 2014 [23] | CCNS   | AH     | IEK      | Topological patterns between two sources in the UMLS can be used to identify missing synonyms, missing parents/children in the UMLS. | 35 newly identified synonyms and 6 unclassified errors for a sample of 241 patterns | N/A                                                                    |
| He et al. 2015 [24] | CCNS   | AH     | IEK      | Same as above       | 75 missing synonyms and 5 unclassified errors for a sample of 299 patterns | N/A                                                                    |
| He et al. 2016 [25] | CCNS   | AH     | IEK      | Same as above       | 35 missing synonyms and 1 unclassified error for a sample of 240 patterns | N/A                                                                    |
| Gu et al. 2012 [26] | LREL   | AS     | IK       | N/A                 | N/A                 | 651,701 cases in which two concepts have questionable multiple relationships between them | N/A                                                                    |
| Cimino et al. 2001  | STA    | CCNS   | HREL     | AS                  | IK                  | 38140 pairs of potentially redundant concepts                     | 2868 IS-A relationships were incorrect                                |
| Hole et al. 2000    | CCNS   | AH     | IK       | N/A                 | 4,558 correctly identified synonyms                               | 8082 concepts with exclusive ST assignments                          |
| Huang et al.        | CCNS   | AH     | IK       | N/A                 | a total of 191                                                  | N/A                                                                    |
| Year | Authors | Methodology | Results |
|------|---------|-------------|---------|
| 2007 [29] | | matches for the 315 multiword UMST terms using the general synonym dictionary and preexisting synonyms with the combined “PS/Perfect Matching” approach | |
| Huang et al. 2009 [30] | CCNS AS EK N/A | 203 correctly identified synonyms | N/A N/A |
| Bodenreider et al. 2002 [31] | HREL LREL CCNS AS IEK Mapping gene ontology concepts in UMLS | 1240 (rejected in UMLS) 141,871 (not in UMLS) | 1,259 relationships are missing in the UMLS N/A (in four categories of relationships) |
| Bodenreider 2001 [32] | HREL AS IK N/A | N/A | 13,000 reflexive relationships, 1800 direct circular hierarchical relationships and 120 indirect circular hierarchical relationships N/A |
| Mougin et al. 2005 [33] | HREL AS IK | Compare two methods to obtain the sets of descendants for a given Metathesaurus source concept. The issue of eliminating cycles from the Metathesaurus graph is addressed from a theoretical perspective: the creation of sets of descendants. | N/A N/A |
| Authors                          | HREL | AH | IK  | Evaluation of families of terminologies within the UMLS | N/A | 325,492 redundant hierarchical relationships | N/A |
|---------------------------------|------|----|-----|--------------------------------------------------------|-----|-----------------------------------------------|-----|
| Bodenreider et al. 1998 [35]    | HREL | AH | IEK | Application of UMLS concepts to MAOUSSC               | 148 (31%) | N/A | Eight semantic categories |
| Bodenreider et al. 2002 [36]    | HREL | AH | IEK | Evaluating adjectival modification in SNOMED and UMLS terms | N/A | 715 67% 293 34% 66 26% 43 37% (acute, chronic), (unilateral, bilateral), (primary, secondary), and (acquired, congenital). No paradigmatic relationship represented with the context. | 4 (acute, chronic), (unilateral, bilateral), (primary, secondary), and (acquired, congenital). |
| Chen et al. 2009 [37]           | STA  | AH | IEK | N/A                                                      | N/A | N/A | 64 concepts with wrong ST assignments |
| Schulz and Hahn 2001 [38]       | HREL | AH | IK  | Pathology and Anatomy concepts reengineered to build a knowledge base | N/A | 1 Cycle in anatomy 355 cycles in pathology. 2328 inconsistencies in anatomy | N/A |
| Geller et al. 2003 [39]         | STA  | AH | IEK | N/A                                                      | N/A | N/A | 8622 concepts with redundant ST assignments |
| Gu et al. 1999 [40]             | STA  | AH | IEK | N/A                                                      | N/A | N/A | 8622 concepts with redundant ST assignments |
| Source                          | Type | AH | IEK | ST   | Description                                                                                                                                 |
|--------------------------------|------|----|-----|------|--------------------------------------------------------------------------------------------------------------------------------------------|
| Gu et al. 2000 [41]            | STA  | AH | IEK | N/A  | N/A                                                                                                                                       | 8622 concepts with redundant ST assignments |
| Cimino et al. 2003 [42]        | HREL | AS | IK  | N/A  | N/A                                                                                                                                       | 17,022 (24.3%) of the parent–child is-a relationships in the UMLS Metathesaurus could not be explained based on the semantic types of the concept. |
| Gu et al. 2004 [43]            | STA  | AH | IEK | N/A  | N/A                                                                                                                                       | 1739 erroneous concepts                     |
| Gu et al. 2007 [44]            | STA  | AH | IEK | N/A  | N/A                                                                                                                                       | 57 erroneous concepts                       |
| Gu et al. 2002 [45]            | STA  | AH | IEK | N/A  | N/A                                                                                                                                       | 292 concepts with incorrect classification   |
| Peng et al. 2002 [46]          | STA  | AS | IK  | N/A  | N/A                                                                                                                                       | 12657 concepts with redundant ST assignments |
| Chen et al. 2009 [47]          | STA  | AH | IEK | N/A  | N/A                                                                                                                                       | 379 erroneous concepts                       |
| Fan et al. 2007 [48]           | STA  | AS | IEK | N/A  | N/A                                                                                                                                       | 223 concepts evaluated for semantic classification |
| Huang et al. 2010 [52]         | CCNS | AS | IEK | N/A  | N/A                                                                                                                                       | 28 pairs of duplicate concepts              |
| Erdogan et al. 2010 [55]       | CCNS | AH | IK  | N/A  | N/A                                                                                                                                       | 81,512 concepts are inconsistent due to the |
following reason: the semantic group of the parent differs from that of the source concept, and no ancestor of the concept is inconsistent.

| Authors          | CCNS | AH | IK | N/A | 81,512 concepts are inconsistent based on semantic groups. | N/A | N/A |
|------------------|------|----|----|-----|----------------------------------------------------------|-----|-----|
| Erdogan et al. 2010 [56] | CCNS | AH | IK | N/A | 81,512 concepts are inconsistent based on semantic groups. | N/A | N/A |
| Liu et al. 2001 [57]    | CCNS | AS | EK | N/A | 5,579 discovered ambiguous synonyms (abbreviations) | N/A | N/A |
| Merrill 2009 [58]      | CCNS | MN | IEK | N/A | This paper advances a detailed exploration of the complex relationships among terms, concepts, and synonymy in the UMLS (Unified Medical Language System) Metathesaurus, and proposes the study and understanding of the Metathesaurus from a model-theoretic perspective. No quantitative results were reported | N/A | N/A |
| Gu et al. 2012 [62]    | STA  | AH | IEK | N/A | 359 concepts with error | N/A | N/A |
| Morrey et al. 2009 [63] | ALL  | AS | IK | N/A | The average number of inconsistencies using the tool was 44. | N/A | N/A |
| Ochs et al. 2011 [64]  | ALL  | AS | IK | Description of the tool | N/A | N/A | N/A |
| Reference                  | Tag | Category | Concept Type | Relationships | Note                                                                 |
|---------------------------|-----|----------|--------------|---------------|----------------------------------------------------------------------|
| Halper et al. 2011 [67]   | HREL | AS       | IK           | N/A           | 456 cycles of three concepts parent-child relationships. For one sample of 40 cycles, 45 relationships were wrong, and 3 relationships were missing. |
|                           |     |          |              |               | N/A                                                                |
| Chen et al. 2009 [68]     | STA | AH       | IEK          | N/A           | 915 concepts missed the assignments to the intersection STs EMD ∩ NP |
|                           |     |          |              |               | N/A                                                                |
| Chen et al. 2012 [69]     | STA | AH       | IEK          | N/A           | 554 concepts were added or reassigned EMD                            |
|                           |     |          |              |               | N/A                                                                |
| Geller et al. 2009 [70]   | STA | AS       | IK           | 38,323 parent-child pairs exist semantic inversion and 544,441 cases exist lack of ancestry. In two samples of 100 parent-child pairs, 84% with semantic inversion and 40% with lack of ancestry were found to be wrong. | N/A |
|                           |     |          |              |               | N/A                                                                |
| Gu et al. 2018 [71]       | STA | AH       | IEK          | N/A           | 19 concepts with error                                              |
|                           |     |          |              |               | N/A                                                                |
| Mejino et al. 1998 [72]   | STA | MN       | IEK          | This report selected some examples of inconsistent representations of anatomical concepts, and illustrated how these inconsistencies can be explained and reconciled by the Digital Anatomist Foundational Model. | N/A |
|                           |     |          |              |               | NA                                                                 |
| Author et al.     | Methodology | Application                                                                 | Results                                               |
|------------------|-------------|-----------------------------------------------------------------------------|--------------------------------------------------------|
| Morrey et al.    | STA AH IEK  | N/A                                                                         | 1,668 concepts with redundant ST assignments          |
| Morrey et al.    | STA AH IEK  | The methodology has several potential applications, including its use to derive a pre-coordinated guide for ST assignments to new UMLS chemical concepts, as a tool for auditing existing concepts, inter-terminology mapping, and to serve as an upper-level network for ChEBI. | N/A                                                   |
| Fan et al. 2007  | STA AS EK   | N/A                                                                         | 218 concepts evaluated for semantic classification     |
| Fan et al. 2007  | STA AS EK   | N/A                                                                         | 100 concepts evaluated for semantic classification     |
| Fan et al. 2007  | STA AS IEK  | N/A                                                                         | 223 concepts evaluated for semantic classification     |
| Fan et al. 2008  | STA AS IEK  | N/A                                                                         | ~50,000 concepts were semantically reclassified       |
| He et al. 2017   | STA MN EK   | N/A                                                                         | 328 concepts with error                               |
| Pisanelli et al. | HREL AS IK  | N/A                                                                         | 100 cycles                                            |
| Study                 | Methodology | Analysis | Integration Type | Results                                                                 | Notes                     |
|----------------------|-------------|----------|------------------|-------------------------------------------------------------------------|---------------------------|
| Xing et al. 2016     | HREL        | AS       | IK               | N/A                                                                     | 2,096 redundant IS-A relationships | N/A |
|                      |             |          |                  |                                                                         |                           |
| Cui et al. 2015      | HREL        | AS       | IK               | COHeRE method for evaluation of relation consistency                  | 138,987 concept pairs were found to have inconsistent relationships across multiple source vocabularies. | 10 semantic types |
|                      |             |          |                  |                                                                         |                           |
| Chen et al. 2009     | HREL        | AH       | IEK              | N/A                                                                     | 40 missing IS-A relationships | N/A |
|                      |             |          |                  |                                                                         |                           |
| Mary et al. 2003     | HREL        | AH       | IEK              | Comparison of UMLS to ADM                                              | for the 3,386 couples, propose at least one new relationship in 45 % (1546 / 3386) of cases. | N/A |
|                      | LREL        |          |                  |                                                                         |                           |
| Vizenor et al. 2009  | LREL        | AH       | IK               | Comparison of Metathesaurus and Semantic Network                       | 37% of 90 associative relationships that have a frequency of 1000 or more are inconsistent with relations in the Semantic Network | N/A |
|                      | STA         |          |                  |                                                                         |                           |
| He et al. 2017       | CCNS        | AH       | IEK              | Topological patterns between two sources in the UMLS can be used to identify missing synonyms, missing parents/children in the UMLS. | 9 missing synonyms for a sample of 100 patterns | N/A |
|                      |             |          |                  |                                                                         |                           |
| He et al. 2016       | CCNS        | AH       | IEK              | Same as above                                                          | N/A                       | N/A |
|                      |             |          |                  |                                                                         |                           |
| He et al. 2018       | CCNS        | AH       | IEK              | Same as above                                                          | 15 missing synonyms for a sample of 100 patterns | N/A |
|                      |             |          |                  |                                                                         |                           |
| Reference                  | Methodology | Alignment Type | Description                                                                                                                                                                                                 | N/A Value | N/A Value |
|---------------------------|-------------|----------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------|------------|
| Keloth et al. 2018 [91]   | CCNS        | AH             | IEK Same as above                                                                                                                                                                                           | N/A        | N/A        |
|                           |             |                | 35 concepts not agreed to be imported. Detailed analyzes for disagreement not reported.                                                                                                                    |            |            |
| Keloth et al. 2019 [92]   | CCNS        | AH             | IEK Same as above                                                                                                                                                                                           | N/A        | N/A        |
|                           |             |                | For a sample size of 150, 35 concepts not agreed to be imported and 24 unclassified errors.                                                                                                                   |            |            |
| Vizenor et al. 2006 [93]  | HREL        | AH             | IEK Methods for aligning SN relationships and Metathesaurus relationships. These methods can be used to find errors in relationships.                                                                   | N/A        | Aligned 80 Metathesaurus relationships with Semantic Network relationships |
|                           | LREL        |                |                                                                                                                                                                                                          |            |            |
| Schulz et al. 2009 [94]   | STA         | AH             | IEK Aligning SN with BioTop                                                                                                                                                                                | N/A        | N/A        |
|                           |             |                | 133 inconsistent multiple ST combinations.                                                                                                                                                                 |            |            |
| Bodenreider et al. 2005 [101] | CCNS     | AH             | IEK Used UMLS synonyms for finding anchor concepts for aligning different source vocabularies. The correction in sources in turn leads to correction in UMLS.                                                      | 639 concepts in NCI Thesaurus aligned with Adult Mouse Anatomical Dictionary | N/A        |
| Winnenburg et al. 2013 [102] | CCNS     | AH             | IEK Same as above                                                                                                                                                                                           | 221 lexical mappings and 343 instance-based mappings between ATC and MeSH    | N/A        |
| Zhang and Bodenreider 2005 [103] | CCNS     | AH             | IEK Same as above                                                                                                                                                                                           | 715 mappings from direct alignment and 703 from indirect alignment         | N/A        |
| Authors and Year | CCNS | AH | IEK | Description | MA-NCI | MA-FMA | NCI-FMA | N/A | N/A |
|------------------|------|----|-----|-------------|--------|--------|---------|-----|-----|
| Zhang and Bodenreider 2006 [104] | CCNS | AH | IEK | Same as above | Aligned 3029 concepts in FMA and GALEN | N/A | N/A |
| Zhang and Bodenreider 2006 [105] | CCNS | AH | IEK | Same as above | Direct Mappings MA-NCI: 715, MA-FMA: 1,353, NCI-FMA: 2,173. Indirect mappings FMA: 703, NCI: 771, MA: 741 | N/A | N/A |
| Zhang and Bodenreider 2007 [106] | CCNS | AH | IEK | Same as above | 1,338 matches between Adult Mouse Anatomical Dictionary and NCI Thesaurus | N/A | N/A |
| Zhang et al. 2007 [107] | CCNS | AH | IEK | Same as above | 337 and 336 matches using two techniques for aligning FMA and GALEN | N/A | N/A |
| Zhang et al. 2004 [108] | CCNS | AH | IEK | Paper on comparing different alignment techniques and lessons learned. | N/A | N/A | N/A |
| Zhang et al. 2007 [109] | CCNS | AH | IEK | Same as above | N/A | N/A | N/A |
| Zhang et al. 2007 [110] | CCNS | AH | IEK | Same as above | N/A | N/A | N/A |
| Reference | Authors | Year | Type | Description | N/A 1 | N/A 2 | N/A 3 |
|-----------|---------|------|------|-------------|-------|-------|-------|
| Bodenreider et al. 2008 [111] | CCNS AH IEK | Paper describing challenges in the process of alignment. | N/A | N/A | N/A |
| Jiménez-Ruiz et al. 2012 [112] | CCNS AH IEK | Using UMLS to align two terminologies and find unsatisfiable classes. For exam, SCT-NCI: 20K and FMA-NCI: > 14K | N/A | N/A | N/A |
| Jiménez-Ruiz et al. 2011 [113] | CCNS AH IEK | Reasoner detected > 15000 unsatisfiable classes when integrating FMA-NCI | N/A | N/A | N/A |
| Jiménez-Ruiz et al. 2010 [114] | CCNS AH IEK | Seems to be a poster abstract. They found 5015 unsatisfiable entities when integrating FMA-NCI. 16,764 when integrating FMA-SCT; 76025 when integrating SCT-NCI | N/A | N/A | N/A |
| Rudniy et al. 2010 [115] | CCNS AS IEK | Did not report errors. Use UMLS dataset as testbed for an algorithm for integration | N/A | N/A | N/A |

List of abbreviations for Appendix Table 1: Ref – Reference, ASPE – Aspect, AT – Automation level, KNW – Knowledge source, STA – Semantic Type Assignment, HREL – Hierarchical Relationships, LREL – Lateral Relationships, CCNS – Concepts, Concept Names, and Synonyms, AS – Automated Systematic, AH – Automated Heuristic, MN – Manual, IK – Intrinsic Knowledge, EK – Extrinsic Knowledge, IEK – Intrinsic and Extrinsic Knowledge

References

10. Bodenreider O, Burgun A. Aligning knowledge sources in the UMLS: methods, quantitative results, and applications. Stud Health Technol Inform. 2004;107(0 1):327.
11. Cimino JJ. Auditing the Unified Medical Language System with semantic methods. J Am Med Inform Assoc. 1998;5(1):41-51.
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.
17. Mougin F, Grabar N. Auditing the multiply-related concepts within the UMLS. J Am Med Inform Assoc. 2014;21(e2):e185-e93.
18. Mougin F, Bodenreider O, Burgun A. Analyzing polysemous concepts from a clinical perspective: application to auditing concept categorization in the UMLS. J Biomed Inform. 2009;42(3):440-51.
19. Wei D, Halper M, Elhanan G. Using SNOMED semantic concept groupings to enhance semantic-type assignment consistency in the UMLS. Proceedings of the 2nd ACM SIGHIT International Health Informatics Symposium: ACM; 2012. p. 825-30.
20. Gu H, Chen Y, He Z, Halper M, Chen L. Quality assurance of UMLS semantic type assignments using SNOMED CT hierarchies. Methods Inf Med. 2016;55(02):158-65.
21. He Z, Morrey CP, Perl Y, Elhanan G, Chen L, Chen Y, et al. Sculpting the UMLS Refined Semantic Network. Online J Public Health Inform. 2014;6(2):e181.
22. Geller J, He Z, Perl Y, Morrey CP, Xu J. Rule-based support system for multiple UMLS semantic type assignments. J Biomed Inform. 2013;46(1):97-110.
23. He Z, Geller J, Elhanan G. Categorizing the relationships between structurally congruent concepts from pairs of terminologies for semantic harmonization. AMIA Summits on Translational Science Proceedings. 2014;2014:48.
24. He Z, Geller J, Chen Y. A comparative analysis of the density of the SNOMED CT conceptual content for semantic harmonization. Artif Intell Med. 2015;64(1):29-40.
25. He Z, Chen Y, de Coronado S, Piskorski K, Geller J. Topological-pattern-based recommendation of UMLS concepts for National Cancer Institute thesaurus. AMIA Annu Symp Proc. 2016;618-627.
26. Gu H, Elhanan G, Halper M, He Z. Questionable relationship triples in the UMLS. Proceedings of 2012 IEEE-EMBS International Conference on Biomedical and Health Informatics: IEEE; 2012. p. 713-6.
27. Cimino JJ. Battling Scylla and Charybdis: the search for redundancy and ambiguity in the 2001 UMLS metathesaurus. Proc AMIA Symp. 2001:120-4.
28. Hole WT, Srinivasan S. Discovering missed synonymy in a large concept-oriented Metathesaurus. Proc AMIA Symp. 2000:354-8.
29. Huang KC, Geller J, Halper M, Cimino JJ. Piecewise synonyms for enhanced UMLS source terminology integration. AMIA Annu Symp Proc. 2007:339-43.
30. Huang KC, Geller J, Halper M, Perl Y, Xu J. Using WordNet synonym substitution to enhance UMLS source integration. Artif Intell Med. 2009;46(2):97-109.
31. Bodenreider O, Mitchell JA, McCray AT. Evaluation of the UMLS as a terminology and knowledge resource for biomedical informatics. Proc AMIA Symp. 2002:61-5.
32. Bodenreider O. Circular hierarchical relationships in the UMLS: etiology, diagnosis, treatment, complications and prevention. Proc AMIA Symp. 2001:57-61.
33. Mougin F, Bodenreider O. Approaches to eliminating cycles in the UMLS Metathesaurus: naive vs. formal. AMIA Annu Symp Proc. 2005:550-4.
34. Bodenreider O. Strength in numbers: exploring redundacy in hierarchical relations across biomedical terminologies. AMIA Annu Symp Proc. 2003:101-5.
35. Bodenreider O, Burgun A, Botti G, Fieschi M, Le Beux P, Kohler F. Evaluation of the Unified Medical Language System as a medical knowledge source. J Am Med Inform Assoc. 1998;5(1):76-87.
36. Bodenreider O, Burgun A, Rindflesch TC. Assessing the consistency of a biomedical terminology through lexical knowledge. International journal of medical informatics. 2002;67(1-3):85-95.
37. Chen Y, Gu HH, Perl Y, Geller J, Halper M. Structural group auditing of a UMLS semantic type's extent. J Biomed Inform. 2009;42(1):41-52.
38. Schulz S, Hahn U. Medical knowledge reengineering—converting major portions of the UMLS into a terminological knowledge base. International journal of medical informatics. 2001;64(2-3):207-21.
39. Geller J, Gu H, Perl Y, Halper M. Semantic refinement and error correction in large terminological knowledge bases. Data & Knowledge Engineering. 2003;45(1):1-32.
40. Gu H, Perl Y, Geller J, Halper M, Liu LM, Cimino JJ. Modeling the UMLS using an OODB. Proc AMIA Symp. 1999:82-6.
41. Gu H, Perl Y, Geller J, Halper M, Liu LM, Cimino JJ. Representing the UMLS as an object-oriented database: modeling issues and advantages. J Am Med Inform Assoc. 2000;7(1):66-80.
42. Cimino JJ, Min H, Perl Y. Consistency across the hierarchies of the UMLS Semantic Network and Metathesaurus. J Biomed Inform. 2003;36(6):450-61.
43. Gu H, Perl Y, Elhanan G, Min H, Zhang L, Peng Y. Auditing concept categorizations in the UMLS. Artif Intell Med. 2004;31(1):29-44.
44. Gu HH, Hripcsak G, Chen Y, Morrey CP, Elhanan G, Cimino J, et al. Evaluation of a UMLS Auditing Process of Semantic Type Assignments. AMIA Annu Symp Proc. 2007:294-8.
45. Gu HH, Min H, Peng Y, Zhang L, Perl Y. Using the metaschema to audit UMLS classification errors. Proc AMIA Symp. 2002:310-4.
46. Peng Y, Halper MH, Perl Y, Geller J. Auditing the UMLS for redundant classifications. Proc AMIA Symp. 2002:612-6.
47. Chen L, Morrey CP, Gu H, Halper M, Perl Y. Modeling multi-typed structurally viewed chemicals with the UMLS Refined Semantic Network. J Am Med Inform Assoc. 2009;16(1):116-31.
48. Fan J-W, Xu H, Friedman C. Using contextual and lexical features to restructure and validate the classification of biomedical concepts. BMC bioinformatics. 2007;8(1):264.
49. Huang K-C, Geller J, Elhanan G, Perl Y, Halper M. Auditing SNOMED Integration into the UMLS for Duplicate Concepts. AMIA Annu Symp Proc. 2010:321-325.
50. Erdogan H, Bodenreider O, Erdem E. Finding semantic inconsistencies in UMLS using answer set programming. Proceedings of the Twenty-Fourth AAAI Conference on Artificial Intelligence. 2010:1927-1928.
51. Erdogan H, Erdem E, Bodenreider O. Exploiting UMLS semantics for checking semantic consistency among UMLS concepts. Stud Health Technol Inform. 2010;160(0 1):749.
52. Liu H, Lussier YA, Friedman C. A study of abbreviations in the UMLS. Proc AMIA Symp. 2001:393-7.
53. Merrill GH. Concepts and synonymy in the UMLS Metathesaurus. J Biomed Discov Collab. 2009;4:7.
54. Gu HH, Elhanan G, Perl Y, Hripcsak G, Cimino JJ, Xu J, et al. A study of terminology auditors' performance for UMLS semantic type assignments. J Biomed Inform. 2012;45(6):1042-8.
55. Morrey CP, Geller J, Halper M, Perl Y. The Neighborhood Auditing Tool: a hybrid interface for auditing the UMLS. J Biomed Inform. 2009;42(3):468-89.
56. Ochs C, Geller J, Perl Y. A relationship-centric hybrid interface for browsing and auditing the umls. Journal of Integrated Design and Process Science. 2011;15(4):3-25.
57. Halper M, Morrey CP, Chen Y, Elhanan G, Hripcsak G, Perl Y. Auditing hierarchical cycles to locate other inconsistencies in the UMLS. AMIA Annu Symp Proc. 2011:2011:529-36.
58. Chen Y, Gu H, Perl Y, Halper M, Xu J. Expanding the extent of a UMLS semantic type via group neighborhood auditing. J Am Med Inform Assoc. 2009;16(5):746-57.
59. Chen Y, Gu H, Perl Y, Geller J. Overcoming an obstacle in expanding a UMLS semantic type extent. J Biomed Inform. 2012;45(1):61-70.
60. Geller J, Morrey CP, Xu J, Halper M, Elhanan G, Perl Y, et al. Comparing inconsistent relationship configurations indicating UMLS errors. AMIA Annu Symp Proc. 2009;2009:193-7.
71. Gu H, He Z, Wei D, Elhanan G, Chen Y. Validating UMLS Semantic Type Assignments Using SNOMED CT Semantic Tags. Methods Inf Med. 2018;57(01/02):43-53.
72. Mejino Jr J, Rosse C. The potential of the digital anatomist foundational model for assuring consistency in UMLS sources. Proc AMIA Symp. 1998:825-829.
73. Morrey CP, Chen L, Halper M, Perl Y. Resolution of redundant semantic type assignments for organic chemicals in the UMLS. Artif Intell Med. 2011;52(3):141-51.
74. Morrey CP, Perl Y, Halper M, Chen L, Gu HH. A chemical specialty semantic network for the Unified Medical Language System. J Cheminform. 2012;4(1):9.
75. Fan JW, Friedman C. Semantic classification of biomedical concepts using distributional similarity. J Am Med Inform Assoc. 2007;14(4):467-77.
76. Fan JW, Xu H, Friedman C. Using distributional analysis to semantically classify UMLS concepts. Stud Health Technol Inform. 2007;129(Pt 1):519-23.
77. Xing G, Zhang GQ, Cui L. FEDRR: fast, exhaustive detection of redundant hierarchical relations for quality improvement of large biomedical ontologies. BioData Min. 2016;9:31.
101. Bodenreider O, Hayamizu TF, Ringwald M, De Coronado S, Zhang S. Of mice and men: aligning mouse and human anatomies. AMIA Annu Symp Proc. 2005:61-5.
102. Winnenburg R, Rodriguez L, Callaghan FM, Sorbello A, Szarfman A, Bodenreider O. Aligning Pharmacologic Classes Between MeSH and ATC. VDOS+ DO@ ICBO. 2013.
103. Zhang S, Bodenreider O. Alignment of multiple ontologies of anatomy: Deriving indirect mappings from direct mappings to a reference. AMIA Annu Symp Proc. 2005:864-8.
104. Zhang S, Bodenreider O. NLM Anatomical Ontology Alignment System Results of the 2006 Ontology Alignment Contest. Proceedings of the 1st International Conference on Ontology Matching-Volume 225: CEUR-WS. org; 2006. p. 153-64.
105. Zhang S, Bodenreider O. Aligning multiple anatomical ontologies through a reference. Proceedings of the 1st International Conference on Ontology Matching-Volume 225: CEUR-WS. org; 2006. p. 201-5.
106. Zhang S, Bodenreider O. Hybrid alignment strategy for anatomical ontologies results of the 2007 ontology alignment contest. Proceedings of the 2nd International Conference on Ontology Matching-Volume 304: CEUR-WS. org; 2007. p. 139-49.
107. Zhang S, Mork P, Bodenreider O, Bernstein PA. Comparing two approaches for aligning representations of anatomy. Artif Intell Med. 2007;39(3):227-36.
108. Zhang S, Mork P, Bodenreider O. Lessons learned from aligning two representations of anatomy. KR-MED. 2004. p. 102-8.
109. Zhang S, Bodenreider O. Experience in Aligning Anatomical Ontologies. Int J Semant Web Inf Syst. 2007;3(2):1-26.
110. Zhang S, Bodenreider O. Lessons learned from cross-validating alignments between large anatomical ontologies. Stud Health Technol Inform. 2007;129(Pt 1):822.
111. Bodenreider O. Issues in mapping LOINC laboratory tests to SNOMED CT. AMIA Annu Symp Proc. 2008:51-5.
112. Jiménez-Ruiz E, Grau BC, Horrocks I. Exploiting the UMLS metathesaurus in the ontology alignment evaluation initiative. E-LKR. 2012.
113. Jiménez-Ruiz E, Grau BC, Horrocks I, Berlanga R. Logic-based assessment of the compatibility of UMLS ontology sources. Journal of biomedical semantics. 2011;2(1):S2.
114. Jimenez-Ruiz E, Grau BC, Horrocks I, Llavori RB. Towards a UMLS-based silver standard for matching biomedical ontologies. Proceedings of the 5th International Workshop on Ontology Matching (OM-2010); 2010.
115. Rudniy A, Geller J, Song M. Shortest Path Edit Distance for Enhancing UMLS Integration and Audit. AMIA Annu Symp Proc. 2010;2010:697-701.