

DL Requirements from Medicine and Biology

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The need for semantically precise domain descriptions has given rise to an increasing number of so-called bio-ontologies covering different fields of biology and medicine. Examples are the Foundational Model of Anatomy (FMA)[5], the Gene Ontology [3] and the Open Biological Ontologies [6]. There are several examples of the conversion of biomedical ontologies into T-Boxes [2, 7, 1, 8] in order to enable terminological reasoning. The biomedical domain, however, exhibits certain peculiarities. Besides the generally large size of biomedical terminology systems (10,000 - 100,000 concepts) the physical composition of organisms (anatomy, biological structure) plays an pivotal role which which may impact the performance of description logics implementations:

- Part-Whole hierarchies constitute an important ordering principle.
- Definitory cycles are common, e.g. $Cell \sqsubseteq \exists haspart.Cytoplasm$ and $Cytoplasm \sqsubseteq \exists partof.Cell$.
- Pairwise disjunction in taxonomies, e.g. $Organ \sqsubseteq \neg Tissue$
- Pairwise disjunction in partonomies, e.g. $\exists partof.Trunk \sqsubseteq \neg \exists partof.Head$
- Role inclusion, e.g. $r \circ s \sqsubseteq r$ [4]

References

- [1] R. Beck and S. Schulz. Logic-based remodeling of the digital anatomist foundational model. *AMIA 2003 – Proc. of the 2003 Symposium of the American Medical Informatics Association*, pages 687–691, 2003.
- [2] A. Gangemi, D. M. Pisanelli, and G. Steve. An overview of the ONION project: Applying ontologies to the integration of medical terminologies. *Data & Knowledge Engineering*, 31(2):183–220, 1999.
- [3] Gene Ontology Consortium. Creating the Gene Ontology resource: Design and implementation. *Genome Research*, 11(8):1425–1433, 2001.
- [4] I. Horrocks and U. Sattler. The effect of adding complex role inclusion axioms in description logics. In *IJCAI'03 – Proc. of the 18th International Joint Conference on Artificial Intelligence*, pages 343–348, 2003.
- [5] C. Rosse and J. L. V. Mejino. A reference ontology for bioinformatics: the Foundational Model of Anatomy. *Journal of Biomedical Informatics*, 36:478–500, 2003.
- [6] OBO. *Open Biological Ontologies (OBO)*. <http://obo.sourceforge.net/>, 2004.
- [7] S. Schulz and U. Hahn. Medical knowledge reengineering – converting major portions of the UMLS into a terminological knowledge base. *International Journal of Medical Informatics*, 64(2/3):207–221, 2001.
- [8] C. J. Wroe, R. Stevens, C. A. Goble, and M. Ashburner. A methodology to migrate the gene ontology to a description logic environment using DAML+OIL. *PSB 2003 – Proc. of the Pacific Symposium on Biocomputing 2003*, pages 624–635, 2003.