Towards interoperability between anatomy and phenotype ontologies

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Abstract

Achieving interoperability between biomedical ontologies is a highly desired state of affairs. We identify a major problem for the interoperability between biomedical ontologies. The common use of anatomy ontologies together with phenotype ontologies may lead to inconsistencies. We provide a solution to this problem through the use of an extended logical framework for representing ontologies.

Current efforts within the biomedical ontology community focus on achieving interoperability between various biomedical ontologies that cover a range of diverse domains. Achieving this interoperability will contribute to the creation of a rich knowledge base that can be used for querying, as well as generating and testing novel hypotheses. The OBO Foundry principles, as applied to a number of biomedical ontologies, are designed to facilitate this interoperability. However, extensions to the relationship ontology are required to meet the OBO Foundry interoperability goals. Using current approaches, inconsistencies arise when ontologies taking a canonical view of a domain – mostly anatomy ontologies – are combined with ontologies of exceptions or properties – mostly phenotype ontologies.

We have developed a methodology for accurately representing canonical domain ontologies within the OBO Foundry ontologies. This is achieved by adding an extension to the semantics for relationships in the biomedical ontologies that allows for treating canonical information as *default*. In the definition of these relations, a non-monotonic extension of the Web Ontology Language (OWL) is used. Conclusions drawn from default knowledge involving relations defined in this manner may be revoked when additional information becomes available. We show how this extension can be used to achieve interoperability between ontologies, and further allows for the inclusion of more knowledge within them. We apply the formalism to ontologies of mouse anatomy and mammalian phenotypes.

Biomedical ontologies require a new class of relations transcending those already available in that they are used in conjunction with default knowledge. The coverage of default knowledge is necessary in order to ensure interoperability between current and future ontologies.