Uberon: towards a comprehensive multi-species anatomy ontology

Melissa A Haendel[1], Georgios V Gkoutos[2], Suzanna E Lewis[3], Christopher J Mungall[3]
[1]University of Oregon, Eugene, OR; [2]University of Cambridge, UK; [3]Lawrence Berkeley National Laboratory, Berkeley CA

Abstract

The lack of a single unified species-neutral ontology covering the anatomy of a variety of metazoans is a hindrance to translating model organism research to human health. We have developed an Uber-anatomy ontology to fill this need, filling the gap between the CARO upper-level ontology and species-specific anatomical ontologies.

Introduction

A number of anatomical ontologies (AOs) exist for specific organisms such as human, mouse and fish, many of which utilize the Common Anatomy Reference Ontology (CARO)[1] to structure the highest-level nodes. However, there is as yet a lack of a unified species-neutral ontology containing representations of embryonic and mature anatomical entities that conforms to OBO Foundry principles, similar to the Gene Ontology (GO) for gene function. Other cross species terminologies exist, such as MeSH, Gemina Anatomy and the Minimum Anatomy Terminology, but these resources are not formal ontologies, utilizing a single relation, and therefore do not provide an adequate substrate for reasoning across species.

Results

Uberon is a preliminary multi-species metazoan anatomy ontology created primarily to fulfil two requirements: (1) support translational research by allowing comparison of phenotypes across species and (2) provide logical cross-product definitions for GO biological process terms. The first version of Uberon was generated automatically by aligning existing species-specific anatomy ontologies (ssAOs) and anatomical reference ontologies, and then partially manually curated. Uberon retains reverse *is_a* links to the ssAOs, such that these can be used in cross-species inferencing and queries. A term is generally included in Uberon if it is a generalization over two or more existing species-specific anatomy terms. For example, `UBERON:dorsal_root_ganglion` subsumes `ZFA:dorsal_root_ganglion`, `MA:dorsal_root_ganglion`, and others. Uberon is homology-independent, and thus contains general terms for analogous structures that have evolved multiple times, such as `eye`. Future versions of Uberon may include evolutionary relationships between structures, along the lines of the TAO and

BILA ontologies. Uberon attempts to employ *is_a*, *part_of*, *overlaps*, and developmental relations in the same manner as ssAOs. The current version of the ontology has 2808 terms, and 5110 links between terms, and 9339 links out to other AOs (Table 1), 1643 Wikipedia cross-references, and has been referenced in 682 GO cross-products.

Ontology	Type	Xrefs
FMA	Adult human	2302
MA	Adult mouse	1495
EHDAA	Embryonic human	838
ZFA	Zebrafish	811
TAO	Teleost	755
NIF	Neuroanatomy	701
GAID	Multi-species terms	626
CL	Cell	427
XAO	Xenopus	335
MAT	General	262
FBbt	Drosophila	243
AAO	Amphibian	103
BILA	Bilateria	64
WBbt	C elegans	63
CARO	Upper-level AO	34

Table 1: Number of terms in each AO referenced in Uberon. Each ontology is referenced by its unique ID space (see http://obofoundry.org)

Conclusions

Whilst Uberon is still in its early stages it has so far proven useful as a means of defining terms in the Gene Ontology, and as a means of comparing phenotypic descriptions of genotypic effects across species. Uberon is available from the OBO Foundry site and can be browsed at **http://berkeleybop.org/obo/UBERON**

References

1. Melissa A. Haendel, Fabian Neuhaus, David Osumi-Sutherland, et al. CARO - The Common Anatomy Reference Ontology. In: *Anatomy Ontologies for Bioinformatics, Principles and Practice* Albert Burger, Duncan Davidson and Richard Baldock (Eds.), 2007