What is an Anatomy Ontology?

The tremendous anatomical diversity of life, across frequently divergent embryonic and adult forms and across multiple levels of organization, presents an enormous but well-understood communication challenge for biologists. Perhaps no society knows this better than the American Association of Anatomists which, since its inception in 1888, have been instrumental in initiating and establishing standardized anatomical terminology (see http://www.anatomy.org/content/history and in 1955 constituted a permanent Committee on Anatomical Nomenclature. Researchers require, reference, supplement and develop sets of standardized terms as evidenced by frequent reference (1,277 times; 283 in last ten years) to ‘terminology’, ‘vocabulary’ or ‘nomenclature’ in papers published in The Anatomical Record (est. 1906). In this age of cross-disciplinary and computational research, however, in which linking evolutionary change, developmental anatomy, and genetics is not only crucial for new breakthroughs but within the realm of possibilities, communication among biologists is more important than ever before. Once again, the American Association of Anatomists which in 1990 made the decision to change the focus of the American Journal of Anatomy to developmental anatomy and started to publish Developmental Dynamics, is well positioned to advance cross-disciplinary terminology standards.

It is important to consider the recent study by Diogo et al. (2012) of the head and neck muscles of the serval and tiger in this context. They, in the tradition of many investigators before them, provide a useful and important addition to the literature on the comparative anatomy of mammals. The characterization of their work, however, as “a Proposal of a Mammalian and a Veterinary Muscle Ontology” warrants closer consideration from the perspective of bioinformatics.

In information-rich fields of science, the term “ontology” has drifted from its original philosophical meaning (the study of what exists) and has come to mean a specific kind of computational tool. This is exemplified in the life sciences by the highly influential Gene Ontology (GO; Ashburner et al., 2000), a collection of thousands of terms used for the annotation of genes and gene products. These terms are organized into a large network structure that can be operated on by computer algorithms to assist in the interpretation of biological data.

In addition to the GO, there are already a number of anatomy ontologies, in which terms such as “masseter,” “muscle,” “jaw,” and “pharyngeal arch” form a network connected via formally defined relationships such as “part of,” “subtype of,” and “develops from.” These ontologies are used to facilitate the ability to answer questions such as “what genes are expressed in muscles of the jaw?” In the past 25 years, particularly since the conception of the semantic web (Berners-Lee et al., 2001), many diverse anatomy ontologies (AOs) have been constructed, from the Foundational Model of Anatomy (FMA; Rosse and Mejino, 2008) to the Hymenoptera Anatomy Ontology (HAO; Seltmann, et al., 2012). The FMA has over 80,000 terms in the domain of human anatomy and the HAO is the largest multi-species anatomy ontology for arthropods with references to over a thousand publications and over three thousand illustrations. In the vertebrate domain, some recent efforts have focused on multi-species AOs.
(e.g., VSAO, Dahdul et al., 2012; Uberon, Mungall et al., 2012) that are modules or include
modules to bridge across many ontologies. Other efforts focus on the representation of
phenotypic diversity in AOs and linking phenotype to genes (Balhoff et al., 2010; Deans et al.,
2012; Mabee et al., 2012). A Common Anatomy Reference Ontology was developed to
standardize the structure of AOs (CARO; Haendel et al., 2008). The power of such tools for
understanding the genetic basis of development and evolutionary change is just beginning to be
realized (Mabee et al., 2007a,b).

Diogo et al. (2012), in the centuries-old tradition of comparative anatomy, present an
organization for the terminology of head and neck muscles, an updated nomenclature, and
testable hypotheses of homology. Despite their title, however, they did not contribute to an
ontology. Because AOs are increasingly important to modern data-rich anatomical research
and central to data interoperability we urge our colleagues to contribute terms and definitions to
community ontologies and to reserve the use of the term “ontology” for the sense in which it is
being used by the wider disciplines of biology, medicine, bioinformatics, and computer science.
We also urge the AAA terminology committee to promote and advance the development of and
use of anatomy ontologies by authors. We hope that morphologists, developmental biologists,
and other readers and contributors to *The Anatomical Record* will work with the growing
community of anatomists and bioinformaticians who are versed in the semantics of AOs (see
e.g., Bioportal.org, ontobee.org, PhenotypeRCN.org, Uberon.org) to make anatomical facts and
knowledge as accessible to computers as they have been to human experts.

We make the following specific recommendations for anatomists wishing to produce anatomy
ontologies:

- These ontologies should be expressed using a standard, machine-processable format,
such as the OBO or OWL languages (Bard, 2005).
- The ontology should consist of a set of terms (a controlled vocabulary) corresponding to
anatomical entities of interest.
- The terms in the ontology should be connected via well-defined relationship types
(Smith, 2005) such as “part of,” “is a,” and “develops from.”
- As much as possible, the terms in the ontology should have concise definitions, to
ensure they are used consistently across applications.
- Development of the ontology should follow standard, community-defined principles and
practices, such as those laid down by the Open Biological Ontologies Foundry (Smith,
2007).

Our intent is not to suggest that every morphological description be presented in computable
form. However, there are ongoing efforts by some publishers to make knowledge accessible by
marking up articles with ontology terms at the time of publication. (Note that these efforts would
require more work on the part of the publishers, not the authors.) By doing this, morphological
descriptions can be moved to datastores such as the Phenoscape Knowledgebase
(Phenoscape KB; Mabee et al., 2012) so that meticulous descriptions of anatomy, as
exemplified by Diogo et al. (2012), become available immediately to be used to answer
questions across all of the biological disciplines, from phylogenetics to model organisms, developmental biology, and genetics. Perhaps the American Society of Anatomists could take the lead by promoting communication through the use of ontologies in *The Anatomical Record*, and facilitate interdisciplinary discovery by working with its publisher to be among the first to mark-up every new paper with ontology terms.

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LITERATURE CITED


