

Phenotype ontologies: are homology relations central enough? A reply to Deans *et al.*

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In a recent *TREE* article, Deans *et al.* [1] propose that formalized, ontology-based descriptions of phenotypes can revolutionize taxonomy and enhance its relevance to other biological domains. We welcome their cause and clear discussion of the benefits and challenges related to this vision. Nonetheless, the representation of homology relations in such ontologies deserves critical examination, due to major implications for ontology design and subsequent ability to accommodate systematic and evolutionary research. We observe that many use cases mentioned in [1] have motivations and benefits for biological research domains outside of phylogenetic systematics.

With regards to homology, Deans *et al.* accept an apparently widespread stance according to which phenotype classes are defined based on structural equivalence [2–4]. In Uberon [2], which is a phenotype ontology that is applicable to a wide taxonomic range, classes are differentiated on the basis of structural differences among their constituents, irrespective of the relative recency of shared ancestry. Hypotheses concerning descent are formulated using an optional layer of representation on top of these ‘ancestry-neutral’ classes. The expectation is that systematists will thus have the ability to represent alternative hypotheses of homology, whereas other user domains will benefit from the stability and minimum of conceptual commitment of such an ontology. This adherence to multi-contextual classes is eerily similar to previous and ultimately unsuccessful attempts to ‘dial down’ the theory dependency of homology concepts in systematics [5].

The adherence to structural equivalence within the Open Biomedical Ontologies (OBO) framework [6] is based on a specific metaphysical doctrine that holds, roughly, that general terms in science refer to Aristotelian universal kinds. Each kind forms ‘an invariant pattern in reality which is multiply exemplified in an indefinitely extendable range of different instances’ [7]. The OBO framework hierarchically organizes universals described by scientific theories, similar to all universals, into abstract classes that apply to all forms of being and are represented in an upper level ontology. The deeper metaphysical issues are not likely to be resolved soon [8–10]; nevertheless, it is already apparent that the top-down method of OBO is poorly suited for accommodating classes whose reality is highly dependent upon inference and classification schemes adopted in specific biological subdisciplines [11].

Homology relations involve precise hypotheses about evolutionary character identity, transformation, and

hierarchical context. They are essential to the construction of reliable classifications and evolutionary narratives stretching from the root of the tree of life to its species-level tips. The evolving contextual contingency of such relations is a challenge for phenotype ontology construction [2–4], and the narrow scope of homology relations limits reasoning across multiple domains [2,12]. Using the top-down approach of OBO has considerable advantages for interoperability, but this may come at the risk of relegating phylogenetic inferences to a lower level. Where are the use cases that ontologically represent, for example, the nature and occurrence of phenotypic synapomorphies and correctly inferred homoplasious conditions at incremental phylogenetic levels and across multiple analyses?

The example of the Uberon ontology class ‘epithelial tube’ (see <http://www.ontobee.org/>) illustrates this issue. Uberon includes many disparate structures under this class that reflect no homology assessments that are specific enough to support phylogenetic reasoning, such as arthropod oviduct, heart tube, mammary duct, nephron tubule, or neural tube. Conversely, certain anatomical components of, for example, the modern mammalian ear have undergone significant structural transformation, yet their posited homologous relations are of great value for phylogenetic inference. We suggest that, although homology and homoplasy propositions for ‘purely’ systematic or ‘purely’ evolutionary developmental inference frameworks are not identical, both share compatible underlying conceptualizations, neither of which are transparently embedded in the OBO approach.

Mungall *et al.* [2] cite several alternatives to OBO that incorporate homology relations explicitly. We feel that it is prudent to weigh the benefits of maximum interoperability more openly against the costs of compromising other virtues of formal ontology design [8–11]. Faithfulness to the phenomena of domain sciences demands attention. The integration of domains under a common rubric is warranted only to the extent that it reflects causal relations in nature, as opposed to strictly instrumental values. The OBO approach is considered ‘realist’ on grounds that terms are represented as universals, that is, realities revealed by science. Strange, then, that this paradigm is capable of obscuring homology relations that are widely understood to be among the principal realities discovered by phylogenetic biology.

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<http://dx.doi.org/10.1016/j.tree.2012.08.001> Trends in Ecology & Evolution, March 2013, Vol. 28, No. 3