

The Ontology of Biological Taxa

Stefan Schulz^{1,*}, Holger Stenzhorn^{1,2}, and Martin Boeker¹

¹Institute of Medical Biometry and Medical Informatics, University Medical Center Freiburg, Stefan-Meier-Str. 26, 79104 Freiburg, Germany

²Digital Enterprise Research Institute (DERI), National University of Ireland, Galway, University Road, Galway, Ireland

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ABSTRACT

Motivation: The classification of biological entities in terms of species and taxa is an important endeavor in biology. Although a large amount of statements encoded in current biomedical ontologies is taxon dependent, there is no obvious or standard way for introducing taxon information into an integrative ontology architecture, supposedly because of ongoing controversies about the ontological nature of species and taxa.

Results: In this article, we discuss different approaches on how to represent biological taxa using existing standards for biomedical ontologies such as the description logic OWL DL and the OBO Relation Ontology. We demonstrate how hidden ambiguities of the species concept can be dealt with and existing controversies can be overcome. A novel approach is to envisage taxon information as qualities that inhere in biological organisms, organism parts, and populations.

Availability: The presented methodology has been implemented in the domain top-level ontology BioTop, openly accessible at <http://purl.org/biotop>. BioTop may help to improve the logical and ontological rigor of biomedical ontologies and further provides a clear architectural principle to deal with biological taxa information.

Contact: stschulz@uni-freiburg.de

1 INTRODUCTION

The classification of biological entities according to their morphological, genetic, evolutionary, and functional characteristics is a fundamental organizing principle since Carolus Linnaeus established conventions for naming living organisms (Ereshefsky, 2001). One century later, the distinction of species received its theoretical underpinning with Charles Darwin's theory of evolution (1859) and was finally demystified by the spectacular advances of molecular biology in the late 20th century. Although these changes have drastically challenged the basic assumptions of Linnaeus' biological theory and have given rise to an ongoing debate about the concept of biological species and taxa (Hey, 2006), his main organizing principle remains the same.

All biology is, in some way, related to the concept of biological taxa. Taxa are hierarchically structured labels or categories used for biological classification, such as *species*, *family*, *class* etc. All organisms, populations, tissues, cells, cell components, and biological macromolecules that are under scrutiny of experimental or

Table 1. Biological taxa with examples

Taxon (Rank)	Asian Elephant	Chimpanzee	Drosophila
<i>Species</i>	<i>Elephas maximus</i>	<i>Simia troglodytes</i>	<i>Drosophila melanogaster</i>
<i>Genus</i>	<i>Elephas</i>	<i>Pan</i>	<i>Drosophila</i>
<i>Subfamily</i>			Drosophilinae
<i>Family</i>	Elephantidae	Hominidae	Drosophilidae
<i>Superfamily</i>	Elephantoidea		
<i>Order</i>	Proboscidea	Primates	Diptera
<i>Class</i>	Mammalia	Mammalia	Insecta
<i>Subphylum</i>	Vertebrata	Vertebrata	
<i>Phylum</i>	Chordata	Chordata	Arthropoda
<i>Kingdom</i>	Animalia	Animalia	Animalia

descriptive biologists are related to some hierarchy of taxa and most biological discoveries have their scope related to one species or taxon. Table 1 gives an exemplary overview of the hierarchical order of taxa. The basic taxon is the *species*. Several species are grouped together by a *genus*. Several genera constitute a *family*, several families an *order*, several orders a *class*, and then several classes a *phylum* or *division*. Finally, the top-most level, the *kingdom* distinguishes between animals and plants. Similar to the several criteria which are discussed to delineate the concept of species, no clear principles exist that govern the division of superordinate taxa. For instance, orders can be further split into superorders and suborders. Even more, the number of taxonomic divisions is variable, and there are also divisions without rank name.

The importance of species and biological taxa is evidenced by many sources. Biological taxa constitute 3,497 out of 24,766 descriptors of MeSH¹, the indexing vocabulary of Medline. In the Open Biomedical Ontologies (OBO) collection² (Smith *et al.*, 2007), 30 out of 66 ontologies are taxon-specific, with taxa ranging from species such as *Homo Sapiens* or *C. elegans*, genera such as *Plasmodium* over families such as *Poaceae* to classes such as *Mammalia*. Due to the sheer number of taxa there is no universal authoritative source, but every important subfield within biology has been independently maintained by curators, so called systematists,

¹ Medical Subject Headings, <http://www.nlm.nih.gov/mesh>

² Open Biomedical Ontologies, <http://www.obofoundry.org>

and for a long time the field of biological systematics has been considered an important research discipline. A converging effort in unifying taxon information for whole biology is the Catalogue of Life³ targeted for complete coverage of all 1.75 million known species by 2011. In the mentioned OBO collection nearly half a million taxon entries of medical interest is available in computer-processable form via the rapidly growing NCBI Taxonomy (Wheeler *et al.*, 2008).

To sum up, biological taxa constitute an overarching and systematic ordering principle that is relevant in practically all biological subject areas.

In this paper we will show how the realm of biological systematics can be embedded into an ontological framework. It is structured as follows: We start with a summary introduction of domain ontologies in general, as well as in the context of the biology, addressing the OBO ontologies and the BioTop biomedical top-domain ontology. Then we provide a formal account of different aspects of the conceptualization of biological taxa and demonstrate how this is implemented in BioTop. Finally we briefly describe our tentative implementation supporting our claim that an overarching ontological framework for biology must have a conclusive and practical account of biological taxa.

2 BIOMEDICAL ONTOLOGIES

2.1 The Foundations of Biomedical Ontology

It is mainly the information explosion in biology and the necessity to process huge amounts of research data which have stimulated the proliferation of biomedical ontologies. Rubin *et al.* (2007) give an overview of the broad range of biomedical information services that can be supported by domain ontologies, with the Gene Ontology (Ashburner *et al.*, 2000) and the OBO collection as the most prominent examples. Whereas this tenet used to be addressed in the past mainly by what had been termed biomedical *terminologies* (with the UMLS⁴ as prototypical example), we have seen – more recently – a steady growth in the usage of the term “ontology”. Due to the lack of a clear notion of what an ontology actually constitutes (Kuśnierczyk, 2006) there is a tendency for either insupportable expectations or general rejection of this term. In this paper, we detach the concept of terminology from the one of ontology subscribing to the following definitions:

According to ISO (2000), a *terminology* is defined as a set of terms representing the system of concepts of a particular subject field. Terminologies relate the senses or meanings of linguistic entities. In contrast, according to Quine (1948), *Ontology* (in singular and uppercase) is the study of what there is. In our understanding, *ontologies* (plural and lowercase) are formal theories that attempt to give precise formulations of the types of entities in reality, of their properties, and of the relations between them (Guarino, 1998). In contradistinction to terminology, formal ontologies strive for describing (as much as possible) what the consensus in a given scientific domain is, independently of human language. Their constituent nodes are referred to as *types*, *kinds*, or *universals*. As they are well suited to hierarchically order and *classify* particular entities (e.g., a given piece of tissue, a cell under a microscope, an amount of biological substance, an animal, a particular population

of bacteria, etc.), they are also referred to as *classes*, a parlance we will use in the following, in accordance with the more recent language use in current biomedical ontology engineering and research⁵.

Although the question whether certain entities really exist are subject to major philosophical disputes, we contend that at any given stage in the development of science, there is a consensus core of scientific understanding of reality, and in our view, it is this which should serve as starting point for developing science-based ontologies. Examples of statements belonging to this consensus core are: that primates are vertebrates, that cells contain cytoplasm, that aspirin tablets contain a derivative of salicylic acid, that ADP is phosphorylated in mitochondria, or that certain biochemical compounds have a clearly delineated composition.

2.2 Top-Level Ontologies

It is widely recognized that the construction of formal ontologies should obey principled criteria. To this end, several top-level ontologies have been devised, such as DOLCE (Gangemi *et al.*, 2002), BFO (Smith *et al.*, 2005), or GOL (Heller *et al.*, 2004). These ontologies mainly coincide in their fundamental division between *continuants* (also called *endurants*, e.g., material objects) and *occurrents* (also called *perdurants*, e.g., events, processes). Orthogonal to this distinction, there is also a coincidence in clearly separating concrete entities or particulars (e.g., “the chimpanzee named Washoe”, “the elephant named Clyde”, or “the 3rd author of this paper”) from the classes they instantiate (e.g., *Chimpanzee*, *Asian Elephant*, *Human*). To this end, we introduce the irreflexive, anti-transitive and asymmetric instantiation relation **instance_of** which relates particulars to classes. In addition, we need a formal relation for subsumption between classes. Here we follow the OBO standard and introduce, for this purpose, the taxonomic subsumption relation *Is_a* by means of **instance_of**⁶ just as proposed by Smith *et al.* (2006):

$$Is_a(A, B) =_{\text{def}} \forall x: (\mathbf{instance_of}(x, A) \rightarrow \mathbf{instance_of}(x, B))$$

In the following discussion we are proposing several possible alternative solutions for an ontological account of species.

2.3 Domain Top-Level Ontologies

Whereas top-level ontologies contain only a restricted set of highly general classes, such as the aforementioned *Continuant*, *Occurrent*, *Function* or *Object*, which are not tied to any particular domain of interest, a domain top-level ontology contains all the classes that are *essentially* needed to describe a certain domain, like *Organism*, *Tissue*, *Cell*, and also *Species* in the case of biology. Those more specific classes are in turn a specialization of the top-level classes as expressed in the formula *Is_a* (*Cell*, *Object*).

³ Catalogue of Life, <http://www.catalogueoflife.org>

⁴ Unified Medical Language System (UMLS): <http://umlsinfo.nlm.nih.gov>

⁵ We follow a general trend and restrict the use of the word *concept* to the realm of terminologies, where it denotes artifacts that represent meanings of linguistic expressions. We avoid it in relation to formal ontologies.

⁶ Throughout this paper, we use capitalized initial letters for the names of relations between universals, as well as for the names of universals. Particulars are highlighted by lower case or by quoted names, bold face is used for relations between particulars.

2.4 BioTop – a Domain Top-Level Ontology

Recently, two separate implementations to encode the top-level of the biomedical domain into ontologies have been created, *viz.*, BioTop⁷ (Stenzhorn *et al.*, 2007) and the Simple Top Bio (Rector *et al.*, 2007). At the moment, efforts set forth by the authors are ongoing to converge these two implementations.

The goal of BioTop is to provide classes and classificatory criteria to categorize the foundational kinds of biology, without any restriction to granularity, species, developmental stages, or states of structural well- or ill-formedness (Schulz and Hahn, 2007). The initial impetus for creating the BioTop ontology was the idea of redesigning and expanding the GENIA ontology (Ohta *et al.*, 2002) in a comprehensive and formally sound way, *i.e.*, to adhere to the fundamental principles of formal rigor, explicitness and precision of ontological axioms. In BioTop's initial development, no definitive commitment existed towards any existing upper ontology, except for the distinction between continuants and occurrents (*cf.* section 2.2).

The primary focus at this stage was set on representing continuants from the area of interest. In the continued development however, the focus was broadened to include the representation of biological processes, functions, and qualities. Additionally, BioTop was aligned with the BFO upper level ontology. BioTop is implemented in OWL DL⁸, an official Semantic Web standard published by the World Wide Web Consortium (W3C). By using this language, our ontology can benefit from a large amount of support tools for editing, automatic classification, etc. OWL DL is also one of the languages accepted by the Open Biological Ontologies (OBO) consortium. The significance of this lies in the fact that, in our view, the high-level BioTop classes can serve as a bridge to link and interface the domain specific ontology classes in the OBO collection. Using such interfacing facility can both potentially reveal overlaps or design errors in OBO ontologies and also create synergetic effects.

2.5 The Difficult Concept of Species

Before we embark on a more general ontological account of biological taxa, we first turn to the most basic taxon, *viz.*, *species*. Both biologists and philosophers disagree on the proper definition of the term “species” and its ontological status (Ereshefsky, 2001). It had been principally the criterion of similarity between organisms and organisms groups that guided Linnaeus' classificatory efforts. Although there are rarely any two individuals with exactly identical characteristics, we made the following observations in regard to the similarity of organisms:

From a diachronic point of view, there are generally significant but relatively minor differences between an organism and its offspring due to sexual or asexual reproduction and spontaneous mutations. However, the distance increases with the number of generations and so today's organisms have little in common with their ancestors. The genetic and phenotypic modifications can be assumed to lie on a mainly continuous scale, and the boundary of the emergence of a new species cannot be drawn by unambiguous criteria, a phenomenon that is ubiquitous in biology (Schulz and Johansson, 2008). No obvious distinguishing feature exists that is

apt to clearly divide the species *Homo sapiens* from *Homo erectus* and nothing indicates any sort of qualitative leap.

As a corollary of this, the parallel evolution of independent lines of organisms increases their genetic and phenotypic distance. Under a synchronic viewpoint, this manifests itself as groups of organisms with clear criteria of species identity. In contrast to the diachronic view, the distinguishing features do not lie on a continuous scale but they are clearly discrete. For instance, the boundary between the species *Homo sapiens* and *Simia troglodytes* (chimpanzee) can be clearly drawn, as there are no organisms existing in the middle.

Even under the diachronic perspective, the distinction between groups of organisms with diverging characteristics may be blurred, *e.g.*, by the distinction of subgroups of the same species. And different species may even form hybrids and merge to a new species. All these peculiarities claim for a non-arbitrary conceptualization of what constitutes exactly a species. There are different types of species concepts, from which the concept of “biological species” as a group of organisms that can interbreed and produce fertile offspring (Mayr, 1969), has found the widest acceptance. Nevertheless, this definition provides only necessary but not sufficient criteria. A defined population of organisms (*e.g.*, the Asian elephants living in Thailand) certainly fulfills this criterion although they do not form a species of their own since they can mate and produce fertile offspring with elephants from Cambodia, for instance. Abbreviating the ability of producing fertile offspring by φ , according to the biological species concept, the pertinence of biological organisms to the same species is expressed the predicate σ :

$$\sigma(o_1, o_2) =_{\text{def}} (\exists t: \varphi(o_1, o_2, t)) \vee (\exists o, t_1, t_2: (\varphi(o_1, o, t_1) \wedge \varphi(o_2, o, t_2)))$$

The shortcomings of Mayr's definition are well known (Grene, 2004, ch. 10): Firstly, it only allows the comparison of organisms living at the same time. Secondly, the definition depends on the dispositional criterion φ , the verification of which remains speculative in many cases. Thirdly, the definition fails with infertile individuals, as well as with species in extinction of which only female or male individuals remain. Fourthly, it fails in the numerous cases of asexual reproduction such as bacteria. It is therefore neither easily applicable, nor generally valid, in spite of its theoretical soundness (Hull, 1997). So it is not surprising that other species concepts compete with Mayr's one. The 22 different conceptualizations of species identified and discussed by Mayden (1997) bear witness on the intensive discussions and disagreements among theoretical biologists and philosophers.

For our practical purpose of biomedical ontologies the formalization of species or – more generally – of biological taxa that we propose, is intended to be neutral to the different and conflicting species conceptualizations. It departs from the principle that biological taxa are something that regardless of its existence in nature or its (*fiat*) attribution by biologists has a highly-ranked importance in biology and therefore requires to be accounted for in biomedical ontologies⁹.

In the following, we will analyze the ontological status of biological taxa and propose and critically assess alternative solutions.

⁷ Available at <http://purl.org/biotop>

⁸ Web Ontology Language (OWL): <http://www.w3.org/TR/owl-features>

⁹ The approach should be flexible enough to support even classification schemes that contradict classic taxonomic principles such as *carnivore* and *herbivore*. The authors are aware of the fact that this may challenge some of the philosophical foundations underlying Basic Formal Ontology (BFO).

3 CONCURRENT ACCOUNTS OF BIOLOGICAL TAXA

3.1 Biological Taxa as Meta-Properties

The above restriction to a two-leveled ontological framework (i.e., dividing the world exhaustively into particulars and universals) has often been challenged. Gangemi *et al.* (2001) contend that there is a fundamental difference between instances in an ontology on the one hand and domain entities (particulars, cf. section 2.1) on the other hand. They argue that we can extend a theory *A* (which follows the two level assumption) by a meta-theory *B*. Whereas theory *A* describes domain entities (particulars) that instantiate universals (classes), *B* takes *A*'s universals as instances of so-called meta-properties. Indexing the instantiation relation by theory level (using subscripts in the formulae) we may state in theory *A* that

$$\text{instance_of}_A(x, y)$$

and then place this in the context of theory *B* with

$$\text{instance_of}_B(y, z)$$

To give a concrete example:

$$\begin{aligned} \text{instance_of}_A(\text{"Clyde"}, \textit{Elephas maximus}) \\ \text{instance_of}_B(\textit{Elephas maximus}, \textit{Species}) \end{aligned}$$

Due to the algebraic property of antitransitivity (as claimed by Gangemi *et al.*, 2001), we can then coherently reject the hypothesis that our elephant "Clyde" is an instance of *Species*. There are several arguments against this solution. Let us consider the second-level predications $\text{instance_of}_B(\textit{Elephas maximus}, \textit{Species})$ on the one hand and $\text{instance_of}_B(\textit{Elephas maximus}, \textit{Genus Elephas})$ on the other hand. Whereas the first one asserts that the class *Elephas maximus* is an instance of a *Species*, the second one states that the species class *Elephas maximus* as a member of the genus *Elephas*. In the same right as we have stated

$$\text{instance_of}_B(\textit{Elephas maximus}, \textit{Species})$$

we could then assert in a third-level predication (instance_of_C)

$$\text{instance_of}_C(\textit{Genus Elephas}, \textit{Genus})$$

"Clyde" would then be a second-level instance of *Species* and a second-level instance of *Genus Elephas*, as well as, in virtue of the latter, a third-level instance of *Genus*.

Given $\text{instance_of}_C(\textit{Species}, \textit{Taxon})$ and $\text{instance_of}_C(\textit{Genus}, \textit{Taxon})$, "Clyde" would finally act simultaneously both as third and forth-level instance of *Taxon*. Together with the argument that "Clyde" might also directly instantiate *Genus Elephas* and the fact that some taxonomic levels (such as subfamilies) are sometimes skipped, it is very obvious that this solution leads to an obscure and inconsistent picture.

Another shortcoming of this approach lies in the fact that it lacks a transitive hierarchical relation between taxa of different levels that would be able to express in simple terms (e.g., that all Indian elephants are vertebrates). From a computational viewpoint, there is also an important performance argument. For example, efficient reasoning algorithms which have been developed for description logics (Baader *et al.*, 2003) and are coherent with the Semantic

Web standard OWL DL do not provide support for reasoning capabilities about instances of instances.

3.2 Biological Taxa as Hierarchies of Classes

We could simplify the above approach (and render it well-suited for description logics-based reasoning) by conflating the level of classes with the one of the meta-level classes. Given the definitions above and a division of all entities in either particulars or classes, it may appear straightforward to use the *Is_a* relation for expressing that *Chimpanzees*, *Indian Elephants*, *Humans*, etc. are species, or that *Genus Pan*, *Genus Elephas* and *Genus Homo* are genera:

$$\begin{aligned} \textit{Is_a}(\textit{Elephas maximus}, \textit{Species}) \\ \textit{Is_a}(\textit{Simia troglodytes}, \textit{Species}) \\ \textit{Is_a}(\textit{Genus Elephas}, \textit{Genus}) \\ \textit{Is_a}(\textit{Genus Pan}, \textit{Genus}), \end{aligned}$$

just as

$$\begin{aligned} \textit{Is_a}(\textit{Elephas maximus}, \textit{Genus Elephas}) \\ \textit{Is_a}(\textit{Simia troglodytes}, \textit{Genus Pan}) \end{aligned}$$

The weakness of this solution, however immediately derives from the above definition of the *Is_a* relation.

So given that

$$\begin{aligned} \text{instance_of}(\text{"Clyde"}, \textit{Elephas maximus}) \\ \text{instance_of}(\text{"Washoe"}, \textit{Simia troglodytes}) \end{aligned}$$

we can infer that

$$\begin{aligned} \text{instance_of}(\text{"Clyde"}, \textit{Genus Elephas}) \\ \text{instance_of}(\text{"Washoe"}, \textit{Genus Pan}) \end{aligned}$$

as well as that

$$\begin{aligned} \text{instance_of}(\text{"Clyde"}, \textit{Species}) \\ \text{instance_of}(\text{"Washoe"}, \textit{Species}) \\ \text{instance_of}(\text{"Clyde"}, \textit{Genus}) \\ \text{instance_of}(\text{"Washoe"}, \textit{Genus}) \end{aligned}$$

We finally end up with all taxa in a specialization hierarchy, having individual organisms as instances. This neither captures the nature of a biological organism, nor the intended meaning of *Species* or *Genus*, since neither Clyde nor Washoe or any other individual animal is an instance of the class *Species*.

Nevertheless we could consistently do this excluding the terms species, genus, etc. This would reduce the instances of taxa (*Elephant*, *Elephantidae*, *Vertebrates*) to classes of organisms and we would no longer be able to account for the meaning of terms like *Genus* or *Species* in a description logic-based framework. However the resulting assertions such as "Clyde is an instance of Mammalia" (on par with "Clyde is an instance of Elephant") would collide with the plural meaning of the taxon terms.

3.3 Biological Taxa as Populations

Several authors have argued in favor of the inclusion of collectives into an ontological framework (Bittner *et al.*, 2004; Rector *et al.*, 2006; Schulz *et al.*, 2006). BioTop has embraced these aspects by introducing the relation **has_granular_part**, an irreflexive and intransitive subrelation of the OBO Relation Ontology relation **has_part** (Schulz *et al.*, 2006).

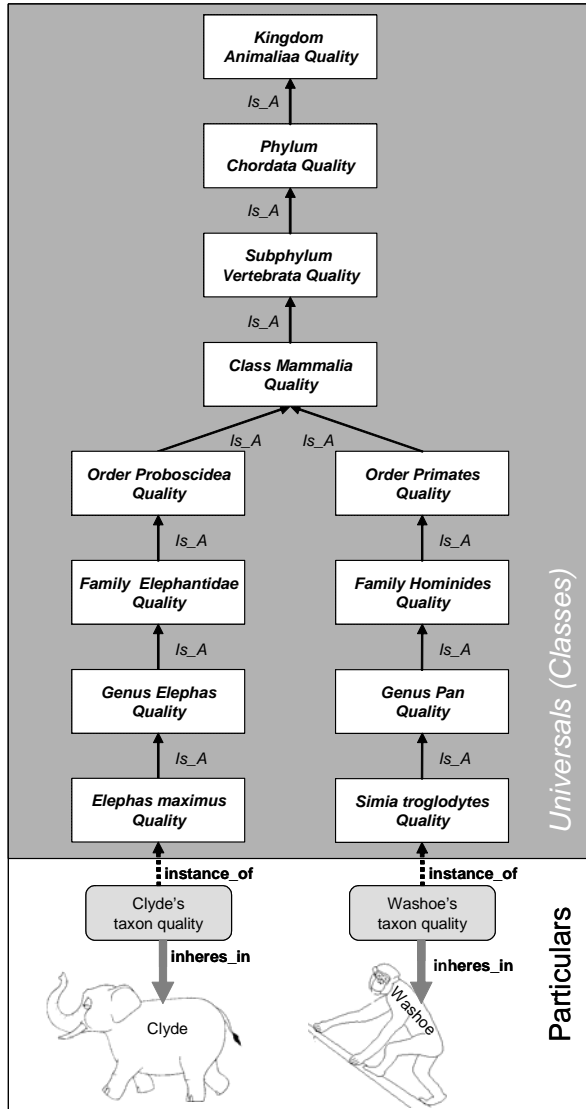


Figure 1: Taxon qualities inhering in individual organisms

This allows us to relate a collective entity to each of its constituent elements, without, however, resort to set theory. For instance,

has_granular_part (“Population of Thai Elephants”, “Clyde”)

asserts that there is a collective entity “Population of Thai Elephants” that is constituted by “granular parts” like our elephant “Clyde” and a number of other individuals similar to “Clyde”. It permits to define collectives in terms of granular parts such as

$$\begin{aligned} \forall x: \text{instance_of}(x, \text{ElephantPopulation}) \leftrightarrow \\ \exists y_1, y_2, \dots, y_n: \text{instance_of}(y_1, y_2, \dots, y_n, \text{Elephant}) \wedge \\ \text{has_granular_part}(x, y_1, y_2, \dots, y_n) \wedge \\ \neg \exists z: (\text{instance_of}(z, \neg \text{Elephant}) \wedge \\ \text{has_granular_part}(x, z)) \end{aligned}$$

Note that “Population of Thai Elephants” is a particular collective and an instance of the universal collective *ElephantPopulation*. The union of all possible instances of *ElephantPopulation*,

viz., “TotalElephantPopulation” would then be the maximal population of elephants every individual elephant is a granular part of.

$$\begin{aligned} \forall x: \text{instance_of}(x, \text{Elephant}) \leftrightarrow \\ \text{has_granular_part}(\text{TotalElephantPopulation}, x) \end{aligned}$$

Yet, “TotalElephantPopulation” is a particular entity. Our proposal here is to consider it as an instance of *Species*. In the same way, we could introduce other populations in different degrees of abstraction such as “TotalVertebratePopulation” which would then be an instance of *Phylum*.

It may be practical for many purposes to equate biological taxa with biological populations although the meaning of *Elephantidae* or *Vertebratae*, in practice, goes further. Especially in molecular biology, species information is not only attributed to whole organisms, but also to organism parts, their constituting cells and derived cell lines. As an example, individual cells from the HELA cell line are considered human cells, but their existence is not dependent on any human population. The interpretation of biological taxa as populations is therefore not adequate for such cases. We can use the OBO relation **derives_from** in order to express that a HELA cell is a human cell:

$$\begin{aligned} \forall x: \text{instance_of}(x, \text{HELA Cell}) \leftrightarrow \\ \exists y: \text{instance_of}(y, \text{Human}) \wedge \\ \text{derives_from}(x, y) \wedge \\ \text{has_granular_part}(\text{“TotalHumanPopulation”}, y) \end{aligned}$$

3.4 Biological Taxa as Qualities

Most top-level ontologies coincide in granting qualities a prominent status. For instance BFO describes the class *Quality* as “A dependent continuant that is exhibited if it inheres in an entity or categorical property. Examples: the color of a tomato, the ambient temperature of air, the circumference shape of a nose, the mass of a piece of gold, the weight of a chimpanzee”¹⁰. DOLCE introduces qualities as “...the basic entities we can perceive or measure: shapes, colors, sizes, sounds, smells, as well as weights, lengths, electric charges” (Masolo, 2003) and also makes reference to the relationship of inherence. The position of the class *Quality* in BFO makes clear that qualities are dependent entities, i.e., they can only exist in dependence on the entities they inhere in.

Our proposal here is to interpret the relation of a biological object to a given taxon as the ascription of a quality. For example, the quality of belonging to the species *Homo Sapiens* is a quality that inheres in any human organism, tissue, or cell. The quality of belonging to the phylum *Chordatae* is a quality that inheres in any biological object that is part of or derived from an organism the species of which belongs to the phylum *Chordatae*.

Fig. 1 depicts a segment of our proposed subclass hierarchy of taxon qualities. The hierarchy exhibits two organizational principles: generalization vs. specialization on one side, and the relevance to an organizational level on the other. Every instance of a material biological object has one inherent taxon quality.

¹⁰ SNAP Continuant Definitions:
http://www.ifomis.org/bfo/manual/snap.pdf

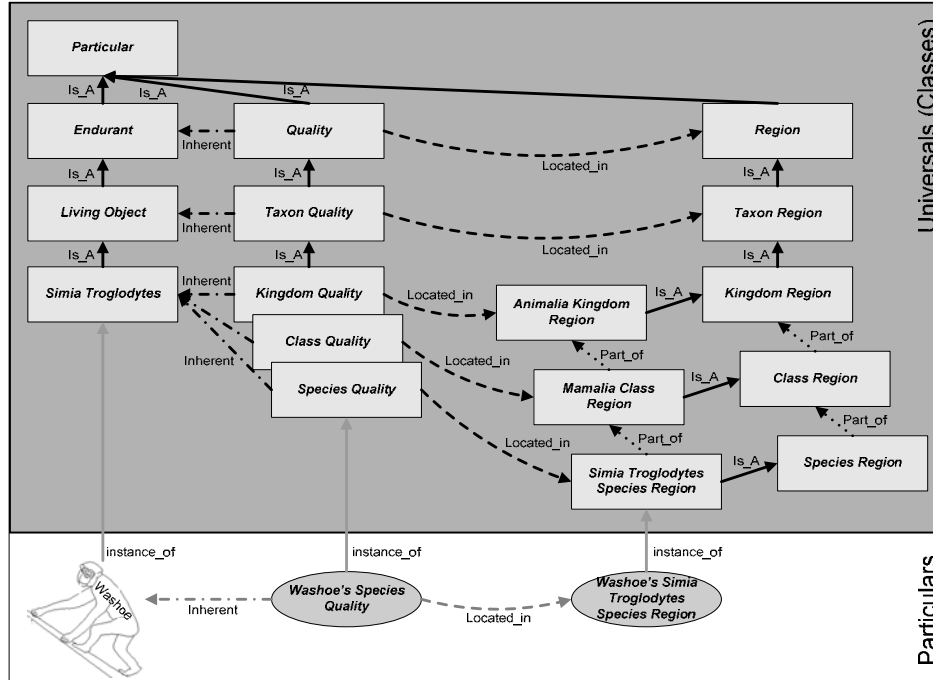


Figure 2: Taxon Qualities inhering in individual organism and their location in Taxon Regions consistent with the DOLCE upper level ontology.

Since, e.g., every human is a hominid, every inhering instance of the class *Homo Sapiens Quality* is also an instance of *Family Hominides Quality* etc. The introduction of qualities is helpful for ontological definitions such as

$$\begin{aligned} \forall x: & \text{instance_of}(x, \text{Human}) \leftrightarrow \\ & \text{instance_of}(x, \text{Organism}) \wedge \\ & \exists y: \text{instance_of}(y, \text{Homo Sapiens Quality}) \wedge \\ & \text{inheres_in}(y, x) \end{aligned}$$

$$\begin{aligned} \forall x: & \text{instance_of}(x, \text{Vertebrate}) \leftrightarrow \\ & \text{instance_of}(x, \text{Organism}) \wedge \\ & \exists y: \text{instance_of}(y, \text{Vertebrate Quality}) \wedge \\ & \text{inheres_in}(y, x) \end{aligned}$$

Based on a hierarchy of qualities, such definitions permit inferences such as that every human is a vertebrate or that every human population is part of some vertebrate population. In addition, it allows for linking organism parts with qualities such as

$$\begin{aligned} \forall x: & \text{instance_of}(x, \text{VertebrateHeart}) \leftrightarrow \\ & \text{instance_of}(x, \text{Heart}) \wedge \\ & \exists y: \text{instance_of}(y, \text{Vertebrate Quality}) \wedge \\ & \text{inheres_in}(y, x) \end{aligned}$$

If the import of the taxon concept should be extended from biological organisms to their parts, as argued in section 3.3 (e.g., *human leukocyte*), the attribution of qualities to organism parts or derivatives can easily be axiomatized by so-called right identity rules (with \otimes being the relation concatenation symbol):

$$\begin{aligned} \text{part_of}(x, y) \otimes \text{inheres_in}(z, y) & \rightarrow \text{inheres_in}(z, x) \\ \text{derives_from}(x, y) \otimes \text{inheres_in}(z, y) & \rightarrow \text{inheres_in}(z, x) \end{aligned}$$

3.5 Biological Taxa as Qualia

An alternative approach to a subclass hierarchy based on the DOLCE upper ontology (Masolo, 2003) is represented in Fig. 2. Since DOLCE is inspired by trope theory (Goodman, 1951), which distinguishes between qualities and their values (i.e., *Qualia*) this proposal introduces another layer of abstraction. Each quality type has an associated quality space (i.e., *Region*) in which it is located.

As in BFO, qualities are dependent entities which are inherent in their respective particulars. Compared to the representation depicted in Fig. 1 only few taxon qualities – one for every taxon – are organized in a flat hierarchy and are related to corresponding value regions. The subsumption hierarchy of taxon qualities of the former approach is represented as a partonomic hierarchy of the *Taxon Regions* in the latter, e.g., the *Species Region* is part of the *Class Region* which is itself part of the *Kingdom Region*. The variety of features is represented as subclasses of the basic *Taxon Regions*, e.g., *Mammalia Class Region Is_a Class Region*.

The main advantages of this approach are a clearer separation of hierarchies and the possibility to make explicit assertions on the specialized *Taxon Regions* without uncontrolled inheritance of restrictions. Its disadvantage lies in a higher complexity.

3.6 Synthesizing Different Taxon Accounts

We have proposed four mutually dependent kinds of ontologically relevant entities that describe different aspects of what is meant by biological taxa on the one hand, and that are expressible in a description logics-based framework on the other.

- The totality of organisms belonging to one taxon (e.g., all grampositive bacteria, all primates, or all humans). This entity is a particular that instantiates the class *Maximal Biological Population*. For each taxon there is one such instance.
- Population classes, the instances of which are defined as parts of some instance of *Maximal Biological Population*. For example, “Elephant Population in Thailand” is an instance of the class *Elephas Maximus Population*, the latter being a subclass of *Elephas Population* and so on. For each taxon there is one such population class.
- Taxon quality classes that are instantiated by each and every particular object to which a taxon can be ascribed. There is one such taxon quality class for each taxon. Because taxon classes are arranged in an *Is_a* hierarchy, the quality of a subordinate taxon is also the quality of a superordinate taxon. For example, an instance *tq_{Clyde}* of *Elephas Maximus Quality* can be ascribed to the elephant “Clyde”. *tq_{Clyde}* is equally an instance of *Genus Elephas Quality*, of *Family Elephantidae Quality*, and so on.
- Taxon quality regions that are represented by a mereological inclusion hierarchy. In contrast to the third approach, every taxon-relevant entity has an inherent quality instance from each taxonomic level.

4 IMPLEMENTATION

We extended BioTop by the notion of biological taxa following the quality approach discussed in section 3.4.

```

bfo:Entity
  bfo:Continuant
    bfo:DependentContinuant
      bfo:SpecificallyDependentContinuant
        bfo:Quality
          biotop:ContinuantQuality
            biotop:TaxonQuality
  
```

The class *biotop:TaxonQuality* has the following restrictions¹¹:

```

biotop:TaxonQuality implies
  ∃ inheres_in.( ∃ has_part. biotop:NucleicAcid) AND
  ∀ inheres_in.( ∃ has_part. biotop:NucleicAcid)
  
```

So we claim the existence of genetic information as a limiting and necessary condition for those entities biological taxa can be ascribed to.

In the inverse direction we claim the inherence of taxon qualities to the classes *biotop:Cell*, *biotop:Organism*, *biotop:Tissue*, *biotop:OrganismPart*, *biotop:NucleicAcid*, e.g.,

```

biotop:Cell implies ∃ inv_inheres_in. biotop:TaxonQuality
  
```

The class *biotop:TaxonQuality* is then the interface to a specialized ontology such as the NCBI taxon ontology. For demonstration purposes we created *taxdemo*, a small example ontology¹².

```

taxdemo:TaxonQuality ≡ biotop:TaxonQuality
taxdemo:KingdomAnimaliaQuality
taxdemo:PhylumChordataQuality
taxdemo:ClassMammaliaQuality
taxdemo:OrderPrimatesQuality
taxdemo:FamilyHominidaeQuality
taxdemo:GenusHomoQuality
taxdemo:HomoSapiensQuality
  
```

In parallel, the taxonomic ranks (*TaxonQuality*, *KingdomQuality*, etc.) are indirectly represented as a second hierarchy.

```

taxdemo:TaxonQuality ≡ biotop:TaxonQuality
taxdemo:KingdomQuality
taxdemo:KingdomAnimaliaQuality
taxdemo:KingdomBacteriaQuality
taxdemo:KingdomVirusesQuality
taxdemo:PhylumQuality
taxdemo:PhylumChordataQuality
taxdemo:ClassQuality
taxdemo:ClassMammaliaQuality
taxdemo:OrderQuality
taxdemo:OrderPrimatesQuality
taxdemo:OrderProboscideaQuality
taxdemo:FamilyQuality
taxdemo:FamilyHominidaeQuality
taxdemo:FamilyElephantidaeQuality
taxdemo:GenusQuality
taxdemo:GenusHomoQuality
taxdemo:GenusPanQuality
taxdemo:GenusElephasQuality
taxdemo:SpeciesQuality
taxdemo:HomoSapiensQuality
taxdemo:ElephasMaximusQuality
  
```

This allows us to define population as a plurality of organism of the same species as follows:

```

taxdemo:Population IMPLIES
  ∃ has_granular_part.biotop:Organism AND
  =1 inv_inheres_in. taxdemo:SpeciesQuality
  
```

These criteria are not met by mixed groups of individuals, e.g., a group of different primates which coincide only at the level of *taxdemo:OrderQuality*

The flexibility of our approach becomes obvious when we use taxon information for parts of organisms. For instance, the class *HumanLeukocyte* can be defined as

```

taxdemo:HumanLeukocyte EQUIVALENT TO
taxdemo:Leukocyte AND
  ∃ inv_inheres_in. taxdemo:HomoSapiensQuality
  
```

If we define

```

taxdemo:AnimalCell EQUIVALENT TO
taxdemo:Cell AND
  ∃ inv_inheres_in. taxdemo:KingdomAnimaliaQuality
  
```

¹¹ For the Description Logics notation cf. Baader *et al.*, 2003, or http://en.wikipedia.org/wiki/Description_logic

¹² Available at <http://purl.org/biotop>

then *taxdemo:HumanLeukocyte* can be classified as *taxdemo:AnimalCell*, provided that the ontology supports:

```
taxdemo:HomoSapiensQuality Is_a
taxdemo:KingdomAnimaliaQuality
```

together with

```
taxdemo:Leukocyte Is_a biotop:Cell
```

It is obvious that this kind of reasoning can be of great advantage for biological fact retrieval from databases or for semantically-enriched information extraction from texts.

From a computational perspective, however, we acknowledge that there still is a bottleneck with regard to the use of inverses (such as **inheres_in** vs. **inv_inheres_in**) and qualified number restrictions (such as =1) in description logics reasoners¹³.

We admit that the meaning of the taxonomic rank classes *SpeciesQuality*, *GenusQuality*, *KingdomQuality* etc. is somewhat counterintuitive, since every instance of *SpeciesQuality* is also an instance of *GenusQuality* and so on.¹⁴ They are therefore not suited to comprehensively represent the meaning of *Species* as disjoint from *Genus*, *Kingdom* etc. Such a reading would require the meta-class representation as discussed in 3.1, discarded due to computational reasons. In our framework, the only way to have an instantiable *Species* (*Genus*, *Kingdom*) class would be to collect all *maximal* populations (cf. section 3.3) with identical species-(genus-, kingdom-) level qualities as instances of *Species* (*Genus*, *Kingdom*) which, again, would only partially match the meaning of *Species* (*Genus*, *Kingdom*). We refrained from implementing the solution discussed in section 3.5 because its more differentiated approach to the representation of qualities is not supported by the BFO upper ontology, currently in use for BioTop.

5 RELATED WORK

Literature on the ontology of taxa roughly falls into two categories: The conceptualization of the nature of species on the one hand, and the ontological status of taxa on the other. In both cases, the focus lies mainly on species whereas higher taxa are seldom addressed.

The first line of scientific discussion is characterized by numerous publications which started with the seminal book of Mayr (1942), who compared several approaches to delineate the nature of species¹⁵ and propagated the popular concept of species as a group of organisms that interbreed and produce fertile offspring. Hull (1997) casts doubt on the monistic assumption that there is one single and ideal way to define species and hypothesizes a trade-off between theoretical significance and practical applicability of species concepts. He classifies the existing species concepts into three categories, *viz.*, (i) similarity-based (which, of course, hinges on some unambiguous notion of phenic or genetic resemblance), (ii) biological and evolutionary (which includes Mayr's and other proposals such as Hennig, 1966) centering around the behavior (i.e., mating, reproduction) of biological organisms, and (iii) phylogenetic focusing the historic development of species.

Mayden (1997) performed an extensive literature review and identified 22 distinct species concepts. In contradistinction to Hull, he propagates the cladistics-based evolutionary significant unit ("Evolutionary Species Concept", Simpson, 1961), rooted in the philosophical principle of identity: "*An evolutionary species is an entity composed of organisms that maintains its identity from other such entities through time and over space and that has its own independent evolutionary fate and historical tendencies*". According to Hey (2006), this concept of species is the most acceptable and most compatible with other species concepts that are rather criterion-based detection protocols than theoretically underpinned concepts. He argues that no criterion that presumes to delineate natural boundaries can overcome the generic vagueness (Hull 1965) of species concepts. Our approach advocates neutrality towards the conceptualization of species and is apt to coexist with both monistic and pluralistic approaches. We are aware of the fact that in the latter case species qualities with multiple parents may be taken into account, due to different categorizations according to conflicting species concepts.

The second line of discussion is on more abstract grounds, and scrutinizes the ontological nature of species, regardless of the species concepts subtleties as exposed above. A fundamental question in here is whether species – seen as single evolving lineage that act as units of evolution – are classes or individuals, the latter being advocated by Ghiselin (1974) and Hull (1978), with the consequence that every single organism is a spatiotemporal *part* of its species. This theory comes close to our view of species as the totality of organisms belonging to one specific species, which can be generalized from species to taxa. We prefer this mereological approach over the set-theoretical one (also pointed out by Ereshefsky, 2007), because the view of a group of organisms as mathematical sets (that are not localized in space and time) is rather counterintuitive. The conceptualization of species as universals or natural kinds conflicts with the fact that there are relatively few "essential" properties that are shared by all individuals of a species (including developmental stages and malformations). Boyd's (1999) *Homeostatic Property Cluster Theory* tries to overcome this, but is still too much committed to similarity-based criteria according to Ereshefsky (2007). The approach pursued in this paper, *viz.*, introducing theory-neutral species qualities – that are extensible to general taxon qualities – seems to be rather novel.

6 CONCLUSION

We have proposed an ontological approach to biological taxa in the context of the domain top-level ontology BioTop¹⁶. It is essentially based upon the assumption that every biological organism, population, or biological matter has some inherent taxon quality. Since it does not raise further reaching ontological claims, our approach largely bypasses the ongoing dispute on species concepts. This enables us to delineate biological populations in terms of shared taxon qualities and to formulate taxon-specific axioms in the framework of description logics.

Our proposal is fully embedded into the standards of Open Biological Ontology and is in line with a major top-level ontology, BFO. Our account of taxon qualities (i.e. the preference of the

¹³ See frequently updated list at <http://www.cs.man.ac.uk/~sattler/reasoners.html>

¹⁴ An instance of *HomoSapiensQuality* would be an instance of *KingdomQuality*, too.

¹⁵ For an overview of earlier approaches see Hey (2006)

¹⁶ BioTop, together with a tentative taxon-specific extension is available at <http://purl.org/biotop>.

bfo:Entity
bfo:Continua
nt
bfo:Depen
dentCon-
tinuant

er approach described in section 3.4 over the more complex (found in section 3.5) also demonstrates how fundamental ontology design decisions depend on the choice of the underlying level model.

ur approach represents taxon qualities as a simple *is_a* hierarchy the import of subsets of existing taxonomy databases such as the NCBI taxonomy is straightforward and scalable. These data can automatically be transformed into an OWL subtype hierarchy and linked to the BioTop node TaxonQuality.

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REFERENCES

- Ashburner, M., Ball, C., Blake, J., et al. (2000). Gene Ontology: Tool for the Unification of Biology. *Gene Ontology Consortium. Nature Genetics*, 25, 25-29.
- Baader, F., Calvanese, D., McGuinness, D.L., Nardi, D. and Patel-Schneider, P.F., eds. (2003). *The Description Logic Handbook. Theory, Implementation, and Applications*, Cambridge, U.K.: Cambridge University Press.
- Bittner, T., Donnelly, M., and Smith, B. (2004) Individuals, Universals, Collections: On the Foundational Relations of Ontology. In: *Formal Ontology in Information Systems. Proceedings of the 3rd International Conference – FOIS 2004*, 37 – 48.
- Boyd, R. (1999). Homeostasis, species, and higher taxa, in Wilson, R. (ed.), *Species: New Interdisciplinary Essays*, 141 – 185. Cambridge: MIT Press.
- Darwin, C. (1859). *On the Origin of Species*. In: *The Works of Charles Darwin*. Edited by Paul H. Barrett and R. B. Freeman. Cambridge: Cambridge University Press.
- Ereshefsky, M. (2001). *The Poverty of the Linnaean Hierarchy: A Philosophical Study of Biological Taxonomy*. Cambridge: Cambridge University Press.
- Ereshefsky, M. (2007). Species. *The Stanford Encyclopedia of Philosophy* (Summer 2007 Edition). In: Zalta, N. (ed.), <http://plato.stanford.edu/archives/sum2007/entries/species>, last accessed Jan 11th, 2008.
- Gangemi, A., Guarino, N., Masolo, C., Oltramari, A. (2001). In: Gómez-Pérez, A., Gruninger M., Stuckenschmidt, H., Uschold, M. (eds.). *Ontologies and Information Sharing: Proceedings of the IJCAI-01 Workshop on Ontologies and Information Sharing*; 26 – 33.
- Gangemi, A., Guarino, N., Masolo, C., Oltramari, A. and Schneider, L. (2002). Sweetening ontologies with Dolce. In: Gómez-Pérez A., Benjamins, R. V. (eds.). *Knowledge Engineering and Knowledge Management: Ontologies and the Semantic Web. Proceedings of the 13th International Conference – EKAW 2002*; Berlin: Springer; 166-181.
- Ghiselin, M. (1974). A Radical Solution to the Species Problem, *Systematic Zoology*, 23: 536 – 544.
- Goodman N. (1951). *The Structure of Appearance*. Cambridge, MA: Harvard University Press.
- Grene, M., and Depew, D. (2004). *The Philosophy of Biology: An Episodic History*, Cambridge University Press, 2004.
- Guarino, N. (1998). Formal Ontology in Information Systems. *Proceedings of FOIS'98*, Trento, Italy, 6 – 8 June 1998. Amsterdam, IOS Press, 3 – 15.
- Guarino, N. and Giarretta, P. (1998). Towards Very Large Knowledge Bases: Knowledge Building and Knowledge Sharing., chapter *Ontologies and Knowledge Bases: Towards a Terminological Clarification*, pages 25 – 32. IOS Press, 1998.
- Heller, B. and Herre, H. (2004). Ontological Categories in GOL. *Axiomathes*, 14, 57 – 76. Kluwer Academic Publishers.
- Hennig, W. (1966). *Phylogenetic Systematics*. Urbana: University of Illinois Press.
- Hey, J. (2006). On the Failure of Modern Species Concepts. *Trends in Ecology & Evolution*, 21 (8), 447 – 450.
- Hull, D. (1965). The Effect of Essentialism on Taxonomy: Two Thousand Years of Stasis. *British Journal for the Philosophy of Science*, 15: 314 – 326; 16: 1 – 18.
- Hull, D. (1978). A Matter of Individuality. *Philosophy of Science*, 45: 335 – 360.
- Hull, D. (1997). The ideal species concept – and why we cannot get it. Claridge M.F., Dawah H.A. and Wilson M.R. (eds.). *Species: The Units of Biodiversity*, Special Vol. 54. Systematics Association, Chapman and Hall Ltd, London, 357 – 380.
- International Organization for Standardization (ISO) (2000): ISO 1087 – 1: Terminology work – Vocabulary – Part 1: Theory and applications, Geneva, Switzerland.
- Kuśnierczyk, W. (2006). Nontological Engineering. *Formal Ontology in Information Systems. Proceedings of the 4th International Conference FOIS 2006*, 39 – 50.
- Masolo, C., Borgo, S., Gangemi, A., Guarino, N., Oltramari, A. (2003). WonderWeb Deliverable D18 Ontology Library. Infrastructure for the Semantic Web. <http://wonderweb.semanticweb.org>, last accessed Jan 11th, 2008.
- Mayden, R.L. (1997). A hierarchy of species concepts: the denouement of the species problem. In: Claridge, M.F., Dawah, H.A. and Wilson, M.R.(eds.). *Species: The Units of Biodiversity*. Special Vol. 54. Systematics Association, Chapman and Hall Ltd, London, 381 – 424.
- Mayr, E. (1942). *Systematics and the Origin of Species*, Columbia University Press.
- Mayr, E. (1969). *Principles of Systematic Zoology*. New York: McGraw – Hill.
- Ohta, Tomoko, Yuka Tateisi, Hideki Mima and Jun'ichi Tsujii. (2002). GENIA Corpus: an Annotated Research Abstract Corpus in Molecular Biology Domain. *Proc. of the Human Language Technology Conference (HLT 2002)*. pp73 – 77.
- Quine, O. (1948). *On What There Is*. Review of *Metaphysics*.
- Rector, A., Rogers J, Bittner T. (2006). Granularity, scale and collectivity: when size does and does not matter. *Journal of Biomedical Informatics*, 39 (3): 333 – 349.
- Rector, A., Stevens, A. and Rogers, J. (2007). Simple Bio Upper Ontology. [<http://www.cs.man.ac.uk/~rektor/ontologies/simple-top-bio>] – Last accessed: Jan 15, 2008.
- Rubin, D., Shah, N., Noy, N. (2008). Biomedical ontologies: a functional perspective. *Briefings in Bioinformatics*, 9 (1): 75 – 90.
- Schulz, S., Kumar, A., Bittner, T. (2006). Biomedical Ontologies: What part-of is and isn't. *Journal of Biomedical Informatics*, 39 (3): 350 – 361.
- Schulz, S., Beißwanger, E., Hahn, U., Wermter, J., Stenzhorn, H. and Kumar, A. (2006). From GENIA to BioTop – Towards a Top – level Ontology for Biology. *4th International Conference on Formal Ontology in Information Systems (FOIS 2006)*, Baltimore, USA, November 2006, 103 – 114.
- Schulz, S. and Hahn U. (2007). Towards the Ontological Foundations of Symbolic Biological Theories. *Artificial Intelligence in Medicine*, 39 (3): 237 – 250.
- Schulz S. and Johansson I. (2008). Continua in biological systems. *The Monist*, accepted for publication.
- Simpson, G.G. (1961). *Principles of Animal Taxonomy*, Columbia University Press.
- Smith, B., Ceusters, W., Klagges, B., Köhler, J., Kumar, A., Lomax J., Mungall C., Neuhaus F., Rector A.L., Rosse C. (2005). Relations in Biomedical Ontologies, *Genome Biology*. 2005; 6 (5).
- Smith, B., Ashburner, M., Rosse, C., Bard, J., Bug, W., Ceusters, W., Goldberg, L.J., Eilbeck, K., Ireland, A., Mungall, C.J.; OBI Consortium, Leontis, N., Rocca – Serra, P., Ruttenberg, A., Sansone, S.A., Scheuermann, R.H., Shah, N., Whetzel P.L., Lewis, S. (2007). The OBO Foundry: Coordinated Evolution of Ontologies to Support Biomedical Data Integration. *Nature Biotechnology* 25 (11): 1251 – 1255.
- Stenzhorn, H., Beißwanger, E., Schulz, S. (2007). Towards a Top - Domain Ontology for Linking Biomedical Ontologies. In: K.A. Kuhn, J.R. Warren, T.-Y. Leong (eds.). *MEDINFO 2007 – Proceedings of the 12th World Congress on Health (Medical) Informatics – Building Sustainable Health Systems*. Amsterdam: IOS Press, 2007; 1225 – 1229.
- Wheeler, D.L., Barrett, T., Benson, D.A., Bryant, S.H., Canese, K., Chetvernin, V., Church, D.M., Dicuccio, M., Edgar, R., Federhen, S., Feolo, M., Geer, L.Y., Helmberg, W., Kapustin, Y., Khovayko, O., Landsman, D., Lipman, D.J., Madden, T.L., Maglott, D.R., Miller, V., Ostell, J., Pruitt, K.D., Schuler G.D., Shumway, M., Sequeira, E., Sherry, S.T., Sirotkin, K., Souvorov, A., Starchenko, G., Tatusov, R.L., Tatusova, T.A., Wagner, L., Yaschenko, E.(2008). Database resources of the National Center for Biotechnology Information. *Nucleic Acids Res.* 36 (Database issue): D13 – 21. Epub 2007 Nov 27.