Ontology for the Twenty First Century: An Introduction with Recommendations

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Forward

The material below represents an introduction to ontology, information ontology and ontology design. This introduction is based on the realist approach to ontology and on the associated Basic Formal Ontology (BFO) and principles for best ontology practices that have been developed and researched at IFOMIS in the last four years.

While focusing on work done at IFOMIS, the majority of the discussion is intended to be of a very general nature and to be applicable to a large number of theoretical and applied issues in ontology generally.

The material available here is part of an ongoing project to provide a one-stop resource and introduction for ontology, with special emphasis on principles for best ontology practices. In this spirit we welcome critical comments, suggestions of further resources for inclusion and feedback regarding the accuracy and usefulness of this material.

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Contents

Introduction

A sea of information

In the last fifty years the development of new technologies, the increasing interconnectedness amongst people in all parts of the globe and the continuous increase in the size of scientific, economic and consumer groups, has lead to a veritable explosion in the amount of data that is produced, used and in need of management world-wide. This is especially true in areas such as the biological sciences, medical research and medical practice. In these disciplines thousands of scientists, doctors and clinicians are contributing daily to a massive body of biomedical knowledge and information. Now, like never before in history, the amount of information that is available and being added to daily by the results of new scientific experiments, research and clinical trials constitutes a veritable ocean of extraordinary depth and breadth.

Managing the sea of information

The current and ever-growing stock of biomedical knowledge and information is greater than the knowledge that could realistically and reliably be acquired by any single human individual, even in an entire lifetime. Further, even if it were possible for human individuals to learn or memorize all of currently existing biomedical information, limitations on the speed and power of ordinary human reasoning would prevent such individuals from being able to efficiently retrieve all of the information that they have learned on any given occasion, or to reason about it in systematic ways. Thus, in spite of the fact that massive amounts of data and information are being generated on a daily basis, ensuring the availability of this information to working biomedical researchers and medical practitioners remains a problem. The sea of information is so large and so deep that current researchers and practitioners simply cannot fathom it all. Yet progress in both biological science and medical treatment depends upon current research and treatments making use of the results of previous research and clinical trials. It is thus crucial that the information contained in biomedical textbooks, journals and clinical trial reports be efficiently accessible to and usable by individuals, doctors and research groups other than its original authors, for purposes both of performing research and of treating the actual diseases of actual patients. The problem then, in a nutshell, is to chart the ever-growing sea of information in such a way that its various parts, portions and depths can be efficiently accessed, used, navigated and reasoned about by human individuals.

The obvious solution

The obvious solution to the problem of navigating the sea of information, that is, of managing and making widely available the ever-growing body of biomedical information, has been to make use of the super-human memory and reasoning capacities of computers. In this respect computers have three great virtues. First, they are able to reliably store a tremendous amount of information. Second, they are able to efficiently, reliably and automatically retrieve and reason about the information that they store. Third, information stored in a computerized format can be made instantly accessible to individuals in all parts of the globe via the internet. Thus, if the biomedical knowledge possessed by experts in the various sub-fields of biology and medicine could be organized and stored in interconnected computer repositories, it would be accessible in real time to anyone anywhere in the world, and could be continuously updated in light of new scientific and medical discoveries. Further, the information contained in these databases could, in principle, be used as the basis for certain kinds of automated reasoning that would independently assist in furthering the goals of scientific research and clinical practice. Such a resource would be invaluable for the purposes of both biomedical research and the treatment of patients (imagine a doctor with immediate access to the most current information about all known diseases at the click of a mouse).[1] However, there are a number of obstacles to the achievement of this vision.

Obstacles to the obvious solution

The first obstacle is that the members of the global community of scientific researchers speak different languages, use different terminologies, and format the results of their research in different ways.

The second obstacle is that the computer technology used by these same groups to encode and store their results has, thus far, suffered from many of the same problems. These two obstacles together might be labeled "*the problem of human idiosyncrasy*".

[1] For a discussion of a collaborative effort towards achieving these tasks in the biomedical domain, see Rubin D. L., Lewis Suzanna, Mungall Chris, Misra S., Westerfield M., Ashburner Michael, Sim I., Chute Christopher, Solbrig H., Storey M. A., Smith Barry, Richter J. D., Noy N. F., Musen Mark. "The National Center for Biomedical Ontology: Advancing Biomedicine through Structured Organization of Scientific Knowledge", in: *Omics: A Journal of Integrative Biology*, 10 (2) (2006): 185-198.

A third obstacle is that scientific theories themselves are not merely long lists of true statements about reality, but rather logically interconnected sets of propositions, usually resting on a number of basic concepts and principles. Scientific theories thus have an internal structure and organization that is, as a rule, more detailed and interconnected than are the entries in a standard computer database.

Fourth, the very virtues of computer based implementations (unlimited memory, efficient retrieval and reasoning, widespread availability), in conjunction with the problems already discussed, create a fourth great obstacle to the realization of this ideal. This problem can be summed up by saying that computers are *dumb beasts.* Computers do not understand themselves, their programming or the intended interpretation of the representations that they manipulate. This last fact has a number of implications.[2]

Computers and their discontents

First, unlike human beings, two computers that encode the same information but use different terminology or different organizational principles cannot, for example by sitting down and talking about the matter, come to some kind of agreement or understanding about the common referents of their discrepant terminologies. Rather, the information in two such machines will be separated by a chasm that can be crossed, if at all, only with the help of human intervention. Thus, for two computer-based biomedical information repositories to be compatible or "interoperable", it is essential that they be given either the same terminology, definitions, etc. or an explicit set of instructions for the translation of one terminology into the language of the other and conversely. This might be labeled "*the problem of Babel*".

Second, computers are able to reason only with the information that they have been given. This means that if the original data entered into the computer is vague, ambiguous, contradictory or in some other way unclear, the computer programmed to reason with such data will produce either nothing at all, or else results that are just as vague, ambiguous, contradictory and unclear as the original information it was given, if not more so. This problem could aptly be called "*the problem of nonsense-in-nonsense-out*".

[2] In making this point we do not mean to take a definitive stand on the question of whether or not it is *in principle* possible for a computer or something like a computer to think and reason in the way that human individuals do. We mean only to stress the point that at this time computers, and especially the kinds of databases and information systems being used in the medical and biomedical domains, in fact do not think and understand in the way that human beings do, and that it is this fact about them that leads to many difficulties and problems in realizing the vision of information sharing and use that is under discussion here.

Thirdly and finally, just as a computer can only reason with what it has been given, so it can only represent what it has been given as this has been given. Computers cannot check the information that they contain for factual accuracy to reality. This means that the information contained in a computer database will only be as accurate to the facts of a given scientific or practical domain as the persons constructing this database have taken the time to ensure it to be. This might be called "*the problem of computer-information-solipsism*".

The shape of solutions sketched

There are thus a number of problems in need of a solution before the use of computers to store, manage and reason about complex biomedical information can be expected to have wide-spread and optimal success. The idiosyncrasies of both biomedical researchers and practitioners, as well as of computer programmers and data-base builders need to be overcome. Computer languages and programs with expressive power sufficient to handle the richness of scientific theories need to be developed and implemented. Substantial care needs to be taken at the outset to ensure that the terminology, definitions, etc. that are entered into biomedical information databases are interoperable between databases, internally coherent and well-defined, and accurate to the facts of reality as reflected in the current (and developing) state of knowledge possessed in the biomedical sciences. The basic answer that has been proposed and is being developed as a general response to all of these problems is "ontology".[3]

Ontology

Traditionally, ontology has been defined as the philosophical study of what exists: the study of the kinds of entities in reality, and the relationships that these entities bear to one another.[4] While this study includes the entities dealt with by the specialized sciences (physics, chemistry, biology, etc.), it also has a more general focus, one directed at providing an account of the most general or basic features of reality: an account of the kinds of objects and relations that are common to all scientific domains whatsoever. Examples of such general or common features of reality might include identity, both at a time and across time, qualities such as color

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[3] For a general and recent discussion of many of these points, and for what might be called a summary of many of the things to come in this manual, see Barry Smith and Werner Ceusters. "Ontology as the Core Discipline of Biomedical Informatics: Legacies of the Past and Recommendations for the Future Direction of Research", in: Dodig Crnkovic Gordana, Stuart Susan (eds.). *Computing, Philosophy, and Cognitive Science.* Cambridge Scholars Press, Cambridge, forthcoming (http://ontology.buffalo.edu/medo/Recommendations_2005.pdf).

[4] For more on this see Barry Smith. "Ontology." in L. Floridi (ed.), *Blackwell Guide to the Philosophy of Computing and Information*, Oxford: Blackwell, 2003, 155–166.

and shape, compositional structure involving relationships of part to whole, relationships such as causation and class-membership, etc. Thus, whereas the biologist studies, for example, cells and the physicist studies, for example, atoms, the philosophical ontologist is interested not only in studying (usually with the help of the natural scientist) these entities themselves, but primarily in giving an account of what is common to both cells and atoms, and of the relationships in which these kinds of entities stand to one another, relationships that may well extend across the normal disciplinary boundaries of specialized sciences such as biology and physics. The goal of philosophical ontology is to provide clear, coherent and rigorously worked out accounts of the basic structures to be found in reality.[5]

In recent times use of the term 'ontology' has become prominent in the areas of computer science and information science research and in the applications of these fields to the management of scientific and other kinds of information. Here 'ontology' has the meaning of a standardized or agreed upon terminological framework, of varying generality, in terms of which information repositories of different sorts are to be constructed. The purpose of such ontologies is both to give an articulate internal structure to electronic information repositories, and to make possible the interoperability or inter-translatability of different repositories containing different information, in such a way that the information in both repositories can be understood in terms of a common language. The ontological problem of information repository construction and management is not, however, simply the problem of agreeing on the use of a common vocabulary. Rather, it is the problem of adopting a (sometimes very general) set of basic categories of objects, of determining what kinds of entities fall within each of these categories of objects, and of determining what relationships hold within and amongst the different categories in the ontology. The ontological problem for computer and information science is thus identical to many of the problems of philosophical ontology, and it is becoming more and more clear that success in the former will be achievable, if at all, only by appeal to the methods, insights and theories of the latter. [6]

How ontology can help

[5] For more detailed discussion of ontology and of related issues in metaphysics, see E. J. Lowe, *A Survey of Metaphysics,* Oxford: Oxford University Press, 2002 & *The Four Category Ontology,* Oxford: Oxford University Press, 2006.

[6] For more on 'ontology' in the philosophical and in the information scientific use, see Barry Smith. "Ontology", and also Douglas Mayhew and Dirk Siebert. "Ontology: The Discipline and the Tool." in: Büchel Gregor, Klein Bertin, Roth-Berghofer Thomas (eds.). *Proceedings of the First Workshop on Philosophy and Informatics, Deutsches Forschungszentrum für künstliche Intelligenz*, Cologne, 2004, 57-64.

The ontological approach to solving the problem of computer based biomedical information management addresses the four major obstacles discussed above in roughly reverse order. The ontological approach begins with human researchers attempting to specify in detail the information that is to be computer-implemented. This includes selecting appropriate terminology, defining this terminology in a rigorous, clear and logically coherent fashion, and ensuring, as much as possible, that the information to be implemented is accurate to the facts of reality. Careful attention to these issues ensures that the internal structure of the scientific theory of interest is maintained and implemented. Further, structuring the information to be implemented in terms of general ontological theories, theories about relationships and objects that are common to different scientific domains, helps address the problems of interoperability or "communication" between different computer artifacts.[7]

Also, beginning with a rigorous and logically coherent specification of the theoretical information to be implemented makes it possible to address the problems of human idiosyncrasy. If different groups of researchers use different terminology for the same entities and to express the same scientific truths, it should be possible to inquire into the reasons for this. If the difference in terminology and expression is rooted in theoretical differences, these differences can be made explicit and discussed, in a scientific fashion. If the differences are merely terminological, then in the interest of making scientific discourse more intelligible, it should be possible to get the two groups to accept a single standardized term or set of terminology, at least for the purposes of interacting with common repositories of information for their scientific field.

Finally, the issue of computational idiosyncrasy can be addressed. Given a formal specification of the theoretical information to be encoded, along with a human consensus about its terminology and a solid understanding of the internal structure of the theory, the only questions of computation that are relevant are first that the program or programming language that is selected have a sufficient amount of expressive power to include all pertinent information of the theory, second that it have sufficient reasoning power to support automated information retrieval and integration, and third that it be interoperable with other such systems. In short, once it is clear what the content and goals of the meaningful scientific theories involved are, computational idiosyncrasy should largely disappear.

[7] Such general ontologies are often called "formal" or "top-level" ontologies, and more will be said about them in the sections to follow.

In what follows: towards an *Organon* for the Information Ontology Age

In the following pages the basic elements of the ontological approach to solving the problems of information management will be presented and explained. In addition, specific recommendations and principles will be put forward. These are intended for use by individuals interested in the possibility of constructing information ontologies of their own.

This discussion is intended to take place specifically against the background of the use of computer based ontologies for the purposes of science, scientific research, and the application of scientific results to particular problems, as in medical practice. In the age of computers there are many potential uses for terminologies and ontologies in the organization of electronic information. However, we are here concerned specifically with the application of ontology in the domain of science. Taking science to be the systematic attempt to account for and explain what *actually* exists, its behavior and its nature, our primary focus will be on the construction of ontologies for the purpose of representing what exists, not for keeping track of fictional discourse, daydreams or mythological creatures.

This document is thus written in the spirit of an *Organon,* an instrument for the proper conduct and representation of scientific research. The first *Organon* was written by the Ancient Greek philosopher Aristotle in the 4th Century B.C., and included his works on logic and the theory of science.[8] The second great *Organon,* the *Novum Organum* (1620) of the Englishman Francis Bacon was written as an update, extension and correction of the Aristotelian *Organon* in light of the success and experimental methods of modern natural science almost 2000 years latter.[9] All sciences have some methodological assumptions in common; the principles of logical inference and reasoning are one example, while the complicated structure of scientific verification and falsification, and adherence to principles of theory evaluation such as "simplicity", "explanatory power" and "predictive power" are others. It is possible for any given scientist or scientific community to employ these methods with greater or lesser adequacy, and the goal of an *Organon* is to codify, so far as is possible, the principles for best or most adequate scientific research and representation practices.

The dawn of the computer age, and the increasing use of computers in scientific research, both for representing information, and for acquiring new results, has raised a host of new

[8] The works known as Aristotle's *Organon* can be found in *The Complete Works of Aristotle*, Two Volumes (Jonathan Barnes ed.). Princeton: Princeton University Press, 1984.

[9] Francis Bacon. *Novum Organum* (Urback, P. and Gibson, J. transl. and eds.). Chicago: Open Court, 1994.

questions about the nature of science, the nature and limits of computability and the nature of representation itself. Just as the advent of Modern Science during the Renaissance made it necessary for Francis Bacon to update the *Organon* of Aristotle with a new organum that would take into account the scientific developments of his time, so what is needed at the beginning of the 21st century is another new organum, an "*Organon* for the Information Ontology Age", one that will clarify the basic principles and methods of computer-based and computer-assisted scientific representation and research. This document is not such an *Organon*, but is intended as contribution towards the realization of that task.

Chapter 1:
What does it mean to "build an ontology"?

The questions "what is an ontology" and "how does one use the ontological approach to organize the sea of information" are closely related, and it will become ever clearer as we move forward that answering the second question involves answering the first, and vice versa. The purpose of this section is to provide an initial overview of the answers to these two questions, one that will begin to establish the framework for understanding and applying the specific methods and principles that will be discussed in sections to come.

The following definition of 'ontology' has recently been proposed, and it contains most of the elements that it will be important to discuss here: an ontology is a representational artifact whose representational units are intended to designate universals in reality and the relations between them.[10]

This definition has two parts. The first identifies an ontology as a representational artifact consisting of representational units, while the second has to do with what the representational units in such an artifact are intended to refer to or be about. We will deal with each of these in turn.

Representations

The human world is full of representations and representational artifacts. The key feature common to all representations is that they make reference to or are about something else. Thus a representation is an idea, an image or a description that refers to some entity or entities external to itself. The memory that I have of the Tower Bridge in London is a representation in my mind

[10] Barry Smith, Waclaw Kusnierczyk, Daniel Schober, Werner Ceusters. "Towards a Reference Terminology for Ontology Research and Development in the Biomedical Domain." Forthcoming in *Proceedings of KR-MED 2006.* (http://ontology.buffalo.edu/bfo/Terminology_for_Ontologies.pdf).

that is about or refers to an entity other than itself, namely the actual Tower Bridge that exists on the Thames River in London. Similarly, the thoughts of a scientist as she looks through a microscope at bacteria, namely the thoughts that "these are bacteria", are mental representations that, taken together, point beyond themselves and make reference to the actual existing bacteria that are under investigation. It is, indeed, one of the most basic features of human thought that beliefs, desires and experiences in general point beyond themselves and refer to the objects that they are about.[11] However, representations by themselves are not yet ontologies in the sense in which we are here interested. Ontologies have the important further feature of being representational artifacts.

Representational artifacts

A representational artifact is an entity which makes publicly available pre-existing cognitive representations from the minds of its author or authors. Representational artifacts include things such as signs, books, pictures and diagrams. A key feature of representational artifacts it that they include ledgers or rules for their interpretation. Thus, maps do not simply come color coded, they also come with a key or table that makes it possible to interpet their color coding as representing certain kinds of things (countries, oceans, mountain ranges, etc.), and the words in which these tables and keys are written themselves have publicly available rules for their interpretation as referring to things in the world, namely the semantics of natural language itself.

A simple kind of representational artifact would be if I were to draw a picture of the Tower Bridge in London based on the mental representation that comprises my memory of having once seen it.[12] Whereas my memory is a cognitive representation, the picture that I draw based on it is a representational artifact. I intend for it to refer to the same thing to which my original memory refers to, namely the Tower Bridge, and yet it exists independently of my own mind or thoughts in a form that is publicly observable and inspectable.

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[11] It is important to note that representations need not always be mental or cognitive in nature. A picture of the Tower Bridge in London is also a representation, something that makes reference to an entity other than itself. For more on the directedness or aboutness of mental states in general, and on the distinction between mental and non-mental representations in particular, see John R. Searle. *Intentionality: An essay in the philosophy of mind.* Cambridge: Cambridge University Press, 1993. See especially Searle's distinction between "derived" and non-derived or original intentionality.

[12] It should be noted that everything to be said in what follows would hold good also if, instead of a picture of the Tower Bridge, I produced a detailed handwritten description of it. This document would also be a representational artifact, and all that will be said about the picture of the Tower Bridge would apply equally well to a written description.

Note here that just as my memory of the Tower Bridge can be better or worse, more or less accurate, so also the representational artifact that I create based on this memory can be better or worse, that is, more or less accurate as a representation of the real entity it is intended to refer to. Notice also that once I render my mental representation into the form of a representational artifact, it becomes available to the community at large for inspection and use. If the drawing (representational artifact) I have created is a good one, someone who was previously unfamiliar with the Tower Bridge may gain new knowledge about an object that exists in the world from inspecting it, while if the representation I have created is a bad one, someone else who has also seen the Tower Bridge will be able to criticize it and suggest needed improvements. There are two points of crucial importance to note here.

<u>Representational artifacts normally represent things, not mental representations, concepts or memories</u>

First, when I attempt to create a representational artifact that makes reference to the Tower Bridge by drawing a picture, it is not the mental representation in my head, the actual memory of the Tower Bridge, that I am trying to draw, but rather, the Tower Bridge itself. This is a very important point to keep in mind when constructing an ontology, and there are a number of ways to see that it is true. First, what would it mean for me to attempt to draw a picture (create a representational artifact) *of my memory*, that is, of my mental representation of the Tower Bridge? What would such a picture look like? It is quite arguable that no one really knows what a drawing of a memory would look like. But more importantly for the purposes of ontology, the answer to this question is unimportant.[13]

In creating a drawing of the Tower Bridge (representational artifact) based on my memory of having seen it (cognitive representation), my primary concern is with accurately representing, not my memory of the Bridge, but the Bridge itself (the thing in reality). Should I have an opportunity to see the bridge again in the future, and to compare it with the drawing that I have made, I may well identify a mistake or an absence of detail in the drawing and decide to correct it in order to more accurately represent the bridge, even if my original memory of the bridge contains no such additional information. Additionally, if other people look at my drawing

[13] Except, perhaps to an empirical psychologist. But note that, even in the case of empirical psychology, where researchers might actually be interested in representing facts about memories or concepts as scientific objects, the goal of these researchers will not be to represent their memory of memories or their concepts of concepts, but rather to represent the actually existing neuro-physiological/mental objects in reality that are the objects of empirical psychological research.

of the Tower Bridge and criticize it, they will engage in this criticism by citing facts, not about my memory or my cognitive representations, but about the drawing and about the bridge itself. And, as far as ontology for the domain of natural science is concerned, this holds true in every case. When constructing a representational artifact for use in science, such as an ontology, based on cognitive representations or concepts in the minds of individual subjects, the goal is not to accurately represent in a publicly accessible way the representations or concepts that exist in the individual's minds, but rather the things in reality that these representations are representations of.[14]

It is possible both to *use* and to *mention* representational artifacts, but this distinction must be respected

The second important point to be made in connection with representational artifacts is that there is a fundamental distinction between *using* such artifacts to make reference to things in reality, the entities that they are representations of, on the one hand, and *mentioning* such artifacts by engaging in discourse about them on the other. The construction of coherent functional ontologies requires that this *use-mention* distinction be strictly taken into account. Consider first an example from ordinary language. One can use the words 'The Tower Bridge' to refer to an object in reality, as in "The Tower Bridge is a well-known structure on the Thames River in London". However, one can also mention the words 'The Tower Bridge', as in " 'The Tower Bridge' is a set of words used primarily by speakers of English to refer to a structure on the Thames River in London, and these words are made up of fourteen occurrences of letters from the Latin Alphabet".

Similarly, to return to the example of a drawing of the Tower Bridge, one could use such a drawing in order to explain to someone what the Tower Bridge is, and what its characteristic features are. In this case the representational artifact is being used in order to talk about the

[14] For more on "concepts" and problems with the use of conceptual terminology in information ontology, see Barry Smith. "From Concepts to Clinical Reality: An Essay on the Benchmarking of Biomedical Terminologies", in *Journal of Biomedical Informatics*, forthcoming, Barry Smith. "Beyond Concepts: Ontology as Reality Representation" In Achille Varzi and Laure Vieu (eds.), *Proceedings of FOIS 2004. International Conference on Formal Ontology and Information Systems,* Turin, 4—6 November 2004, Gunnar O. Klein & Barry Smith. "Concepts Systems and Ontologies: Recommendations based on discussions between realist philosophers and ISO/CEN experts concerning the standards addressing "concepts" and related terms." (http://ontology.buffalo.edu/concepts/ConceptsandOntologies.pdf), Ingvar Johansson. "Bioinformatics and biological reality." *Journal of Biomedical Informatics* 39 (2006) 274—287, Barry Smith and Werner Ceusters. "Wüsteria", in *Proceedings of the XIXth International Congress of the European Federation for Medical Informatics* (MIE 2005), (http://ontology.buffalo.edu/concepts/ConceptsandOntologies.pdf).

object that it makes reference to. But one could also mention such a drawing by making it and its properties the explicit theme of discourse and saying things like "this drawing is made of paper", "this drawing was done with a mixture of pencil and pastel" and "this drawing leaves important details of the Tower Bridge out of account". In all such cases discourse *mentions* the representation and is thus explicitly about the representation itself, not about the objects or entities to which the representation refers. It is a very common mistake in the construction of ontologies to mix statements that are meant to be part of the ontology, and thus refer to objects in reality on the one hand, with statements that are about the ontology (representational artifact) itself, and thus are meant to refer to items, entries and components within the ontology, but not to the representation-independent things in reality. Some examples include the following:

In the Medical Subject Headings (Mesh) can be found the following hierarchical relationship "National Socialism *is_a* MeSH Descriptor".[15] Here National Socialism, which is a kind of political movement that existed in the world, is identified as a kind of term in the MeSH database, however the definition of 'National Socialism' in MeSH as "The doctrines and policies of the Nazis or the National Social German Workers party, which ruled Germany under Adolf Hitler from 1933-1945. These doctrines and policies included racist nationalism, expansionism, and state control of the economy. (from Columbia Encyclopedia, 6th ed. and American Heritage College Dictionary, 3d ed.)" makes it clear that the use and mention of the term 'National Socialism' have here been confused. Similarly, in the National Cancer Institute Thesaurus (NCIT), the following definition of 'Conceptual Entities' can be found "An organizational header for concepts representing mostly abstract entities".[16] As a definition there are a number of problems with this. However, what is important here is simply that, once again, the use and mention of a term have been conflated. Whereas we would expect a database that is about things in reality to provide definitions that would tell us facts about the basic features of those things (in this case the basic features of conceptual entities), what we get here is an explanation of how the term 'conceptual entity' is used as a part of the representational artifact that is the NCIT. Once again, statements about things (use) and statements about the words used to mention things (mention) have been conflated. Other examples include the definition of 'mouse' as "name for

[15] MeSH accessed via UMLS Knowledge Source Server Release 2006AC. http://umlsks.nlm.nih.gov/kss/servlet/Turbine/template/admin%2Cuser%2CKSS_login.vm;jsessionid=421388A81A91D5EC4C8759E7D5B3B13A.kss2, Thursday, September 28, 2006.
[16] NCI Thesaurus accessed Via UMLS Knowledge Source Server, Release 2006AC, Thursday, September 28, 2006.

the species *mus musculus*" in BIRNLex,[17] the entry "Bacterium causes Experimental Model of Disease" in the Unified Medical Language System (UMLS),[18] the definition of 'animal' as "a subtype of Living Subject representing any animal-of-interest to the Personnel Management domain" from the HL7 Glossary, [19] and "living subject is_a code system" from the HL7.[20] All of these are examples of ways that databases and ontologies can go wrong when they fail to keep separate the use of the ontology and its terms to refer to things in reality, and the mention of terms and elements of the ontology in discourse that is explicitly about it, its construction and its constituent elements.

The example of a representational artifact that we have focused on so far, a drawing of the Tower Bridge, is admittedly quite simple. More sophisticated representational artifacts include certain aspects of natural language, maps, blue-prints, and various classificatory schemes employed by the natural sciences, notably the periodic table of the elements. However, it is important to recognize that the basic distinction between representing a memory or concept and representing a thing, and between using a representational artifact and mentioning it, apply in these more sophisticated cases as well. It is one thing to talk about (mention) the Periodic Table of the Elements as an important development in the history of human knowledge, and it is quite another thing to use this same Table in order to learn something about the nature of elements in reality. What all of the things mentioned above have in common is that they are codified publicly available representations possessing interpretive rules for understanding how they refer to reality, which can be publicly evaluated for accuracy and usefulness.

To say, then, that an ontology is a representational artifact is to say that it is an explicit and publicly available rendering of a representation or system of representations belonging to an individual or group of individuals. Paradigm examples of ontologies in this sense would be scientific textbooks (or at the least, portions thereof), geographical maps, and certain kinds of information databases. When such a representational artifact is formalized, that is, when such an

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[17] UMLS Semantic Network http://semanticnetwork.nlm.nih.gov/

[18] UMLS Semantic Network http://semanticnetwork.nlm.nih.gov/, Also to be found in the UMLS are "Experimental Model of Disease **affects** Fungus", "Experimental model of disease **is_a** Pathologic Function", "cancer documentation is_a cancer", "living subject is_a information object representing an animal or complex organism", "individual allele is_a act of observation".

[19] Various Contributors eds., HL7 Publishing Technical Committee. Last Published 11/22/2005 8:05 PM. HL7® Version 3 Standard, © 2005 Health Level Seven®, Inc.

[20] HL7 Version 3.0 accessed Via UMLS Knowledge Source Server, Release 2006AC, Thursday, September 28, 2006.

artifact is expressed in a logical or programming language of some sort, it can be called a 'formalized representational artifact'. Formalized representational artifacts have the advantage, normally, of being both rigorously formulated and computer implementable. It is thus ontologies in the sense of formalized representational artifacts that currently hold out the greatest promise of enabling computers to help human researchers and clinicians cope with the ever-growing sea of biomedical information.

Ontological reality, an initial introduction

It is the second part of the above definition, the point that the representational units of an ontology "are intended to designate universals in reality and the relations between them," that is crucial to an initial understanding of what an ontology is. From what has been said so far, it might be thought that any representational artifact at all is or could be considered an ontology. This is false. While there are many kinds of representational artifacts, a representational artifact is an ontology only if the intended referents of its representational units are real universals and real relationships amongst such universals on the side of reality.[21] But what is a universal, and what is meant by talk of relationships amongst universals?

It is a basic assumption of scientific inquiry that nature is structured, ordered and regular, at least to some degree. Though scientists always perform experiments and make observations regarding particular objects, what they are actually interested in are the generalizations about the structure, order and regularity that exists in nature that such experiments and observations make possible. Universals are that which is general or abstract in reality. They are the philosophical explanation of the structure, order and regularity that is to be found in nature, and they are what all members of a natural kind, grouping or species (for example the kind "feline" or "mammal") have in common. Universals are repeatable in the sense that they can be instantiated by more than one object and at more than one time, whereas particulars, such as myself, Tibbles the cat and specific political administrations, are non-repeatable, they can exist only in one place and during one period of time. Because of this, universals do not have a determinate location in space or time. Rather, they exist at all times and in all places where particular entities instantiating them exist.

[21] In this connection it should be noted that while the picture of the Tower Bridge discussed above is indeed a representational artifact, it is not an ontology in the strict sense, since it represents only a single particular thing in reality and not, for example, information about bridges in general.

One way of characterizing a universal is to say that it is that in virtue of which a thing is what it is, and without which that thing would not be the kind of thing that it is. For example, Aristotle, a well-known Ancient Greek philosopher, believed that the universal "human" is characterized by the features of being an "animal" and being "rational". Given this characterization of the universal "human", the following statements should also all be true:

- all humans are rational animals;
- it is in virtue of being a rational animal that any given entity is a human;
- to understand what a rational animal is is to understand what it is for a entity, any entity, to be human; and
- any entity that lacks one or both of these qualities is not a human being;
- the universal "human" has existed, exists now, and will exist in the future, regardless of the knowing activities of scientists, it neither came into existence when Aristotle first thought about it, nor will it go out of existence if everyone stops thinking about it.

Whether or not Aristotle was entirely correct about the nature of human beings is not important here. What is important is that to know the nature of a thing, for example a human being or a chemical element, is to know the nature of the universal(s) that that thing instantiates and that, taken together, comprise the nature of that thing. Thus, the goal of scientific research in any given domain is to discover the nature of the universals that are instantiated by entities in that domain, whether in biology, chemistry, physics, or some other science.

As opposed to universals, particulars are the individual denizens of reality. Particulars instantiate universals, but cannot themselves be instantiated. It is in virtue of instantiating a given universal that two particulars will be similar in some respect. For example, to say of two entities that they are both cats, and that they are similar in virtue of being cats, is to say that they both instantiate the universal "cat". Particulars thus exist in space and time, and come into and pass out of existence. It is possible to causally interact with, directly see with one's eyes, touch and smell particulars, but not universals. For example, the universal cucumber is an abstract entity that is instantiated by and accounts for the similarities amongst all particular cucumbers, but unlike its instances, the universal has never been and cannot be sliced, diced, made into salad or consumed with a cool glass of chardonnay on a warm summer afternoon.

Whereas particulars are fleeting and contingent, universals are what is abiding and permanent in reality. To say that an ontology is concerned with representing universals on the side of reality is thus to say that an ontology is a representational artifact the primary purpose of which is to represent what is essential, law-like and general in reality. The way to begin doing this is to represent, as accurately as possible, the general features that are attributed to reality by the natural sciences, such as biology, chemistry and physics. It is important to distinguish, however, between genuine universals on the one hand and mere classes on the other.[22]

Every universal has a corresponding class, but not every class corresponds to a universal. A class can be defined as a collection of particulars falling under a term in such a way that the term applies to every member of the collection, and every particular to which the term applies is a member of the collection. For example, the class corresponding to the universal “cat” will be designated by the term ‘cat’ and will contain all and only the particular cats that exist in reality. However, there are many classes that do not correspond to any universal, and these fall into two general kinds.

The first are classes designated by arbitrary general terms. One example of an arbitrary general term would be the term ‘grue’, which was created by the philosopher Nelson Goodman to pick out all and only things that are green before a certain time (say the World Cup Tournament of 2006), and then blue at all later times[23]. The problem with this class, and the reason that it does not correspond to any universal, is that there is not any general feature of reality having to do with the possession by an entity of a certain color, such as green or blue, relative to its existing before or after a certain time alone. Thus, even if there are individuals in reality that fall within the extension of this bizarre term, it is highly unlikely that they do so in virtue of instantiating some important common nature or universal. Thus ‘grue’ may well define a class, but it does not correspond to a universal on the side of reality.

The second kinds of classes that do not correspond to universals in reality are classes created by using a general term to make reference to particulars existing at a specific time or in a specific place, such as the class of all women currently living on the north coast of Germany, the class of all athletes over the age of 30, or the class of all individuals currently infected by HIV on the Continent of Africa. There may be important reasons to want to talk about classes such as these, but it is equally important for ontological purposes to recognize that these classes do not correspond to universals. The reason for this is that these classes make explicit reference, not to

[22] For more on this, see Barry Smith, Waclaw Kusnierczyk, Daniel Schober, Werner Ceusters. “Towards a Reference Terminology for Ontology Research and Development in the Biomedical Domain.”
[23] Nelson Goodman. “The New Riddle of Induction”, in *Journal of Philosophy* 63 (1966): 281-331.

general kinds, but to collections or groups of particular entities. Whereas the universal "Oxygen" does not exist at any particular time or place, and is a general law-like feature of reality, the class defined by the expression 'all of the oxygen in this jar' refers only to a particular collection of particular oxygen molecules in a specific location and at a specific time. These particular molecules exist at a certain time and in a certain place, they have a specific temperature and are mixed with a certain ratio of molecules of other sorts, however none of these things is true of the universal "oxygen" (additionally, it is possible for a person to breathe the oxygen molecules in the jar, but it is not possible for any person to breathe the universal "oxygen").

Some examples of arbitrarily defined classes from the International Classification of Disease (ICD 10) include: "other problems with special functions", "tuberculosis of unspecified bones and joints", "tubercle bacilli not found by bacteriological or histological examination, but tuberculosis confirmed by other methods (inoculation of animals)", "other mineral salts, not elsewhere classified, causing adverse effects in therapeutic use".[24]

Universals, classes designated by arbitrarily defined general terms, and classes defined in terms of groups of particulars existing at particular times and places are all important for various purposes. However, for the purposes of constructing an ontology it is essential that the classes designated by each of these things be kept separate, and that primary importance be given to specifying accurate representations of universals. More will be said about these issues in subsequent sections.

The other important notion mentioned in the definition of an ontology that we have given is that of "relationships" holding amongst universals in reality. The general idea of a relationship is familiar from both everyday language and the theories of science. If John and Mary are spouses, then they stand in the relationship of being married (and perhaps also of being in love). If an event such as the collapse of a bridge occurs immediately after some other event, such as an explosion directly underneath the bridge, we say that the explosion stands to the bridge-collapse

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[24] International Classification of Disease 2006 Version, http://www3.who.int/icd/currentversion/fr-icd.htm, accessed Thursday, September 28, 2006. Further examples include: "other general medical examination for administrative purposes", "assault by other specified means", "other accidental submersion or drowning in water transport accident injuring other specified person", "accident to powered aircraft, other and unspecified, injuring occupant of military aircraft, any rank", "other accidental submersion or drowning in water transport accident injuring occupant of other watercraft – crew", "normal pregnancy", "fall on stairs or ladders in water transport injuring occupant of small boat, unpowered", "railway accident involving collision with rolling stock and injuring pedal cyclist", "injury due to war operations by lasers", "nontraffic accident involving motor-driven snow vehicle injuring pedestrian".

in the relationship of cause to effect: the explosion causes the bridge to collapse. For our purposes here, what is important to recognize is that it is not only universals, but also the relationships that exist amongst them (themselves a kind of universal) that comprise the contents of scientific knowledge. It is one thing to know something about the species “feline”, it is another and much better thing to know also how the species “feline” fits into the larger picture of living things in nature, in particular, what its relationship is to other species. Similarly, it is one thing to understand something about the universal “hydrogen”, it is another and much better thing to know how “hydrogen” is related to other element-universals, relations that are captured, for example, in the Periodic Table of the Elements. Often, full knowledge of a given universal requires understanding also the relationships in which it stands to other universals of similar kinds, and conversely. It is for this reason that ontology concerns itself not only with representing universals, but also with representing explicitly the relationships that obtain amongst universals. It is only by doing this that the full internal structure and content of scientific theories can be accurately and completely represented.[25]

As an initial and very general clarification of what is meant by talking of the relations amongst universals that an ontology represents, it is important to distinguish between three general kinds of relationships. Relationships that hold between two universals, relationships that hold between a universal and a particular, and relationships that hold between two particulars.[26]

The paradigm example of a relationship that holds between universals is the “is_a” relationship, as in “feline is_a mammal”. The relationship of “is_a” holds amongst universals in virtue of the fact that they stand in hierarchies of generality. For example, the hierarchy extending from the universal “feline” through the universals “mammal”, “animal” and “organism” can be understood as structured from least to most general in terms of the “is_a” relation. Thus, more concrete universals stand in “is_a” relations to, or are “subsumed_by” more general universals.

[25] It is important to note here that there is a distinction between relations that are particulars or “instance-level” relations, and relations that are universals, just as there is a distinction between particulars and universals more generally. We will not address this issue in any great detail here, but the interested reader is referred to Mertz, D. W. *Moderate Realism and its Logic*. London: Yale University Press, 1996, for a detailed discussion of these matters.

[26] For more on this, see Barry Smith, Werner Ceusters, Bert Klagges, Jacob Kohler, Anand Kumar, Jane Lomax, Chris Mungall, Fabian Neuhaus, Alan L. Rector, and Cornelius Rosse. “Relations in Biomedical Ontologies.” *Genome Biology* 6, no. 6:R46 (2005).

A paradigm example of a relationship between a particular and a universal is the "instantiates" relation, as in "Tibbles instantiates Tabby", where Tibbles is a particular flesh and blood cat, and "Tabby" is the universal for the feline breed of Tabby cats. All particulars stand in the relation of "instantiates" to some universals, but no universal can properly be said to instantiate any particular. Further, no particular stands in an "is_a" relationship to any universal.

Finally, a paradigm example of a relationship holding between particulars is the "part_of" relation, as in "John's left leg part_of John". The relationship "part_of" is a pervasive one, and is one that is very important for many kinds of biomedical science. There are many other relationships that hold amongst particulars, such as "is_cause_of" and "depends_on", and it is important to note that, under specific conditions that will be discussed in more detail later, such relationships can also be construed as obtaining amongst universals.

More will be said about relations in the following sections. For now, what is important to recognize is that there are different kinds of relationships, some of which hold amongst universals, and that fully understanding a given scientific domain requires not only knowing what universals exist in that domain, but also what kinds of relationships hold amongst those universals.

The definition of 'ontology' reconsidered

Given this clarification of the nature of universals and the relationships that obtain amongst them, the definition of an ontology as "a representational artifact whose representational units are intended to designate universals in reality and the relations between them," should be essentially clear. The basic idea is that ontologies are about what is general, structured and law-like in reality. Further, ontologies represent this generality not only by containing general or common terms representing universals, but also by capturing and explicitly representing the relationships that obtain amongst these universals. This differentiates ontologies on the one hand from *terminologies,* representational artifacts containing natural language terms and definitions, but not rendering explicit the structure or relationships amongst the entities referred to by these terms (such as the ULMS), and on the other from *inventories*, representational artifacts designed to keep track of particulars, of what is specific in reality.

What does it mean to construct an ontology?

When one sets out to construct an ontology then, what one is doing is designing a representational artifact that is intended to represent the universals and relations amongst universals that exist, either in a given domain of reality (such as that studied by molecular biology), or across such domains.

Chapter 2: Ontology and other things: the nature and uses of information ontology

This section, still in progress as of now, will provide a general theoretical framework and definition incorporating both realist and conceptualist approaches to ontology. It will then make some general statements applicable to all information ontologies whatsoever, and discuss the differences between information ontologies on the one hand, and things like terminologies, thesauri, taxonomies and etc. on the other. Finally, consideration will be given to some of the specific actual and possible uses of information ontologies.

Some information about the topics of this chapter can be found in the following documents and slides-

- Daniel Schober. "Recommendations." http://msi-ontology.sourceforge.net/recommendations/.
- Stefano Borgo, Nicola Guarino, Laure Vieu "Formal Ontology for Semanticists", http://www.loa-cnr.it/Tutorials/ESSLLI1.pdf

Chapter 3: What are the methods of ontology?

In the introduction, we articulated the problem of managing the sea of biomedical and scientific information, and discussed ontology as the proposed general solution to this problem. In the last sections, an ontology was defined as a representational artifact whose representational units are intended to designate universals in reality and the relations between them; and it was noted that it is when such representational artifacts are regimented and formalized in certain computer tractable ways that they begin to genuinely contribute to the resolution of the "sea of information" problem. Constructing an ontology is the problem of constructing a formalized representational artifact whose representational units designate universals in reality and the relations between them; but what does this process look like? This section sketches an initial outline.

The ontological solution to the problem of the sea of information is essentially a top down one. This means that it begins with theoretical considerations of a very general nature, making the assumption that keeping track of very specific information about organs, genes and diseases requires getting the very general scientific and philosophical details behind this information right, and doing so in a systematic and coherent fashion. It is only once this has been done that the detailed knowledge base of the biomedical sciences can be encoded in such a way as to ensure widespread accessibility and usability. Importantly however, regimenting all of this information in such a way that it can be kept track of and reasoned about by computers requires, for reasons discussed in the introduction, that a maximum amount of clarity and precision be used at each step in the process of identifying and defining universals and the relations amongst them in a given domain. The general method to be followed in constructing an ontology can be summarized in the following steps:

- Explicitly determine and demarcate the subject-matter or domain of the ontology.
- Gather information: Determine what the universals and relations amongst universals dealt with in this subject-matter are.
- Concretize this information in the form of a representational artifact, such as a written document, grid, etc.
- Regiment the information contained in this representational artifact in order to ensure:

i. Logical, philosophical and scientific coherence,

ii. Coherence and compatibility with other relevant ontologies, and

iii. Human intelligibility.

- Formalize the regimented representational artifact in a computer tractable language.
- Implement the representational artifact in some specific computing context.[27]

Determining the subject-matter of an ontology has a number of components. Important among these are dealing with the distinction between formal and material ontologies, gathering information about the domain of reality that is to be represented, and determining what kinds of entities are relevant to a representation of that domain given the purpose that this representation is being designed to fulfill.

Determining what the universals and relations amongst universals dealt with in the subject-matter are has to do both with analyzing the subject-matter itself, and with locating the various entities with which it deals in the context of a more general ontology. Concretizing the information that has been gathered in the form of a representational artifact involves providing a

[27] Compare with the very helpful discussion in Eric Little, "A Proposed Methodology for the Development of Application-Based Formal Ontologies", at http://sunsite.informatik.rwth-aachen.de/Publications/CEUR-WS//Vol-94/ki03rao_little.pdf.

systematic statement, whether in the form of diagrams, a computer file or a written document of some sort, of the entities, relations, terminology etc. that are to be included in the ontology.

Once this has been done, the process of regimentation comes in two steps. The first is to check the universals, relations and domain specific terminology contained in the ontology for logical, philosophical and empirical adequacy, including consistency and human intelligibility. This task is essentially a semantic one which involves making sure that the ontology is maximally effective in representing the universals and relationships that exist in the domain that it is intended to be about. This process should include a good deal of fact-checking, including extensive consultation with domain experts (working biologists, medical practitioners, etc.).

The second step of regimenting the ontology is essentially a syntactical one, and involves translating the terminology, relations, etc. contained in the ontology into a format that is computer tractable, whether this be some fragment of first-order logic, a description logic or something else. Finally, then, comes the process of implementing the formalized representational artifact in some actually functioning computing context.

While the process outlined here is essentially top down in nature, in practice there will be a great deal of interaction and feedback between the different steps of ontology design and construction, with constraints at the computational level and the factual information of the specific scientific domain(s) that are to be modeled influencing decisions that are made about which and how many ontological categories and relations are to be used, and vice versa. The following sections will discuss the steps of this process in greater detail. This will include introducing necessary theoretical notions and clarifications.

Some general principles to be followed in ontology design

Some general principles for ontology construction that underpin the methodology in what follows include:[28]

Realism

'Realism' can be defined as a philosophical position according to which "reality and its constituents exist independently of our (linguistic, conceptual, theoretical, cultural)

[28] For further discussion of these principles, see Pierre Grenon and Barry Smith. "SNAP and SPAN: Towards Dynamic Spatial Ontology." *Spatial Cognition and Computation*, Pierre Grenon, Barry Smith and Louis Goldberg. "Biodynamic Ontology: Applying BFO in the Biomedical Domain." In D. M. PIsanelli (ed.) *Ontologies in Medicine.* Amsterdam: IOS Press, 2004, 20—38, and Barry Smith and Pierre Grenon. "The Cornucopia of Formal Ontological relations." *Dialectica* Vol. 58, 3, (2004), pp. 279—296.

representations thereof."[29] Just as science is the attempt to come to know the general features of reality in the form of universals and relationships obtaining amongst them, so realism is the thesis that the things that scientific knowledge is about are in fact real, mind-independent things. This position has a number of general consequences for ontology. These include the position that ontologies are representations of reality, not representations of people's concepts or mental representations of reality; that science discovers truths about reality, and that the facts that science discovers, such as the nature of universals in the biological or chemical domains, existed long before it occurred to human beings to conceptualize and search for them; and that not all representational schemes are equally good, some are better than others precisely in that they are better representations of reality. There are a number of other principles that go hand in hand with the commitments of realist ontology.[30]

Perspectivalism

Perspectivalism involves the recognition that reality is a complex and variegated phenomenon. While not all representations are good, because some are accurate to the facts of reality and some are not, there are nevertheless many different representations that are equally good (good in the sense of being true) precisely in that they capture different and important features of one and the same reality. These include viewing reality in terms of substances or continuants and their qualities on the one hand, and in terms of occurrents or processes on the other (a distinction that will be more fully developed below), and also viewing reality at different levels of granularity, ranging from the microscopic world of atoms, chemical reactions and molecules, to the macroscopic world of organisms, ecosystems and galaxies.[31]

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[29] Pierre Grenon and Barry Smith. "SNAP and SPAN: Towards Dynamic Spatial Ontology." *Spatial Cognition and Computation,* p. 138.

[30] For more on realism, see Smith Barry: "From Concepts to Clinical Reality: An Essay on the Benchmarking of Biomedical Terminologies", in *Journal of Biomedical Informatics*, forthcoming; Barry Smith. "Beyond Concepts: Ontology as Reality Representation." In Achille Varzi and Laure Vieu (eds.), *Proceedings of FOIS 2004. International Conference on Formal Ontology and Information Systems,* Turin, 4—6 November 2004; Gunnar O. Klein & Barry Smith. "Concepts Systems and Ontologies: Recommendations based on discussions between realist philosophers and ISO/CEN experts concerning the standards addressing "concepts" and related terms." http://ontology.buffalo.edu/concepts/ConceptsandOntologies.pdf; Ingvar Johansson. "Bioinformatics and biological reality." *Journal of Biomedical Informatics* 39 (2006) 274—287.

[31] One noteworthy example of perspectivalism in the history of ontology is Edmund Husserl, who held that there were three separate and irreducible material ontological domains, corresponding to the "psychological", the "social" and the "natural" respectively. Husserl maintained that each of these domains had its own governing essence or universal, and its own characteristic internal laws and relationships, none of which could be reduced or explained away in terms of the kinds of things in the other two domains. Nevertheless, Husserl maintained that there were certain formal ontological relationships and categories that applied to entities in all of these domains, and that universals and relationships from each of these domains were coinstantiated in particular objects in the actual

Fallibilism

Fallibilism involves commitment to the idea that, though our current scientific theories are the best candidates we have for the truth about reality, it may nevertheless be the case, given the (realist) fact that reality exists independently of our ways of conceptualizing it, that portions of our current knowledge are incorrect. The fallibilist maintains that it is a matter of empirical investigation what the facts of reality are, and recognizes that empirical investigation is an on-going, open-ended, experimental process.

Adequatism

The approach being developed here also presupposes the principle of adequetism. This is the position that a good theory of reality must do justice to all of the different phenomena that reality contains. In opposition to the tendency to attempt to reductively explain higher level macroscopic phenomena in terms of "more basic" or fundamental components of reality, adequatism entails that the entities in any given domain of reality be taken seriously on their own terms first. Thus, just as an ontology of physics should be about atoms and sub-atomic particles, and an ontology of chemical reactions should include the existence of various kinds of elements and compounds, so an ontology of biological phenomena should include the existence of, at various levels, cells, organs, biological systems and organisms, as well as populations and environments. The goal of adequatism is to do justice to the vast array of different kinds of entities that exist in the world, in different domains and at different levels of granularity, rather than ignoring them or attempting to explain them away.

The following provides a very general over view of the ontology design process. More will be said about many of the headings below in subsequent sections.

A schematic overview of the ontology design process

- *Explicitly determine and demarcate the domain and extent of the ontology.*

1. Formal & material ontologies
   i. Formal Categories: what formal ontological categories are important for the domain?
   ii. Formal Relations: what formal ontological relations are important for the domain?
2. Granularity: what is the appropriate maximum and minimum level of granularity or complexity that the ontology requires?

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spatial-temporal world. For some discussion of this, see Smith, D. W. "Mind and Body". Printed in Smith, B. & Smith, D. W. eds. *The Cambridge Companion to Husserl*. New York: Cambridge University Press, 1995.

3. Relevance: what is relevant to include in the ontology given i) the domain the ontology is intended to represent and ii) the purpose for which the ontology is to be used?

- *Determine what the universals and relations amongst them are.*

4. Gathering information: what are the important general terms and relations dealt with in the domain? Organize these into an initial list, preferably with some definition and organization into tentative categories.

5. Scientific investigation: How do the terms and relations function in scientific theories of the domain being represented? Are the terms and relations that have been collected an adequate reflection of what is most crucial for understanding the truth about this domain of reality as reflected in current scientific knowledge?

6. Thought experiments and imaginative variation: What are the essential or defining features i) of the domain as a whole, ii) of the particular entities and relations that have been selected as crucial?

- *Concretize the cognitive representations of these things as representational artifacts (bearing in mind that it is the things, not the cognitive representations of them, that are to be represented by these artifacts).*
- *Regiment the representational artifact for-*

7. Logical, philosophical and scientific coherence.

Terminology.

Definition of terminology.

Taxonomy, especially is_a hierarchies.

Categories and relations, align the domain information with relevant formal ontological categories and relations.

8. Coherence and compatibility with other relevant ontologies.

9. Human understandability.

- *Formalize the regimented representational artifact in a computer tractable format.*
- *Implement the formalized regimented representational artifact.*

Chapter 4: Explicitly determine and demarcate the domain and extent of the ontology

The first step in constructing an ontology is to explicitly determine the intended domain of that ontology, to answer the question "what part of reality is this ontology an ontology of?" Providing an explicit statement of the intended subject-matter of an ontology at the outset helps to focus the effort of constructing the ontology by indicating what principles and information need to be included while at the same time ruling out other information as un-important for constructing an ontology of the given domain. For example, the documentation for the Foundational Model of Anatomy, an ontology of human anatomy, reads "The FMA…is strictly constrained to "pure" anatomy, i.e., the structural organization of the body".[32] This statement makes it relatively clear what information is, and what information is not, properly a candidate for inclusion in the FMA, and thus also what terms, ontological categories, universals and relationships might need to be included.

Here we begin by outlining a number of issues that are pertinent to determining the domain of an ontology, including the distinction between formal and material ontologies, and the issues of subject-matter granularity and subject-matter relevance.

Formal and material ontologies

An important distinction to bear in mind when constructing an ontology, and one that will play a role in helping to demarcate the subject-matter of the ontology, is that between formal and material (or domain-specific) ontologies.[33]

A formal ontology is a representation of the categories of objects and of the relationships within and amongst categories that are to be found in any domain of reality whatsoever. The most obvious formal ontological category is "entity"; no matter what science one is considering that science studies entities, and thus the category "entity" applies to the subject matter of that science. A relatively uncontroversial formal ontological relation is the relation "part_of". Even sciences committed to the study of very very small entities acknowledge the existence of at least some parts of those entities, and therefore the relationship "part_of" applies to those domains as well.

A material or domain ontology, by contrast, consists of a representation of the material categories, universals and relationships amongst universals that are to be found in some specific domain of reality, such as genetics, anatomy, plant-biology, cell-biology, physiology, etc. Characteristic of a material ontology is that it will contain many categories, universals and

[32] FMA, http://sig.biostr.washington.edu/projects/fm/AboutFM.html, July 17, 2006.
[33] One of the earliest explicit recognitions of the distinction between formal and material ontologies is to be found in Husserl, E. *Logical investigations Vol. II*. Trans. J. N. Findlay. Amherst: Humanity Books, 1900-01/2000, especially the third and fourth Investigations. The following discussion is also very sympathetic to ideas presented by Alan Rector et. al. "Simple Bio Upper Ontology", http://www.cs.man.ac.uk/~rector/ontologies/simple-top-bio/, especially section "II General Considerations".

relations amongst universals that are not to be found in formal ontologies or in other material ontologies (consider the categories, universals and relationships amongst them that must be dealt with in psychology, physics and sociology respectively).

When one sets out to construct an ontology, it will most often be a material or domain-specific ontology that one is interested in constructing. However, for purposes of managing the sea of biomedical information, the relevance of formal ontology to the construction of material ontologies is two-fold.

First, to the extent that the categories and relations in a formal ontology are well-structured and systematically defined, organizing the universals and relations of a given domain ontology in terms of them will render that domain ontology equally well-structured and hold out at least the possibility that the other universals and relationships in that domain ontology can be well-defined as well. What it means to "organize" a domain ontology in terms of a formal ontology will be discussed in more detail below. However, the basic idea of what this means can be expressed by saying that if, in a formal ontology, the relationship "part_of" has a semantic interpretation such that "if x part_of y and y part_of z, then x part_of z", then in a domain ontology focusing on, for example, anatomy, where a finger part_of arm and arm part_of body, it should be possible to infer that finger part_of body based on the definitions and information explicitly contained in that ontology, and likewise in all similar cases. Thus organizing a material ontology in terms of a formal ontology involves encoding information in the material ontology in a univocal and logically/mathematically precise way based on the fixed meanings of categories and relations in the formal ontology.

Second, using formal ontologies to structure domain ontologies helps to ensure interoperability or communication between and amongst domain ontologies. If the basic set of categories and relations that have been used to structure two different domain ontologies have the same meaning in both ontologies, this will make it easier to bring the information in these ontologies together, and to compare them in various ways. Indeed, one of the primary reasons for interest in formal or "top-level" ontologies in the information science community is the promise that such ontologies hold out of making wide-spread interoperability possible.[34]

[34] For an example of the use of formal ontological principles to improve a domain application, see Simon Jonathan, Fielding James Matthew, Dos Santos Mariana Casella, Smith Barry: "Reference Ontologies for Biomedical Ontology Integration and Natural Language Processing", in *International Journal of Medical Informatics*, forthcoming.

For these reasons it is important, at the outset of designing a domain specific ontology, to consider what formal ontological categories and relations might apply to the domain at hand, and to select a formal ontology with sufficient and sufficiently clear categories and relations to handle the basic kinds of entities to be found in the domain in question. It is important to note that, by definition, a formal ontology should not contain all relations and universals that are pertinent to a given domain. Rather, the specific content of a given domain will be "added on" to or "structured in terms of" a formal ontology based on the best available scientific knowledge about that domain.

There are currently a number of different and competing formal ontologies that have been proposed, consisting of various degrees of expressive power, completeness, and implementability.[35] Aside from these recent developments, the history of philosophy itself contains numerous proposals and much argumentation regarding the basic categories and relations that are to be found in reality.[36] Here we will refrain from exploring all of these positions and the various arguments for and against them. Rather, we will begin by briefly discussing some basic features that are common to all formal ontologies, and then by introducing, explaining and developing the Basic Formal Ontology (BFO), which has been developed at the Institute for Ontology and Medical Information Science in Saarbrücken, Germany. This latter example will then be used as we develop some of the more specific principles for good ontology design and best ontology practices. Importantly, the majority of best ontology practices to be discussed in what follows do not depend in any way on the

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[35] Claudio Masolo et al., *Wonderweb Deliverable D18* (2003 [cited May 6, 2006]); available from http://wonderweb.semanticweb.org/deliverables/documents/D18.pdf, see also Gangemi, A. et. al. "Sweetening Ontologies with DOLCE", http://citeseer.ist.psu.edu/cache/papers/cs/26864/http:zSzzSzwww.ladseb.pd.cnr.itzSzinforzSzOntologyzSzPaperszSzDOLCE-EKAW.pdf/gangemi02sweetening.pdf, and Heller, B. & Herre, H. "Ontological Categories in GOL", *Axiomathes* 14: 57—76, 2004, http://www.onto-med.de/en/theories/gfo/index.html; Alan Rector et. al. "Simple Bio Upper Ontology", http://www.cs.man.ac.uk/~rector/ontologies/simple-top-bio/.

[36] Aristotle. *Metaphysics*. In McKeon, R. ed.*The Basic Works of Aristotle*. New York: The Modern Library, 2001, Nelson Goodman. *Problems and Projects*. New York: Bobbs-Merrill Company, 1972, Husserl, E. *Logical Investigations Vol. II.* Trans. J. N. Findlay. Amherst: Humanity Books, 1900-01/2000, Kant, I. *Critique of pure reason*. Trans. Paul Guyer and Allen W. Wood. Cambridge: Cambridge University Press, 1871/1998, Lowe, E. J. *A Survey of Metaphysics*. Oxford: Oxford University Press, 2002; Mertz, D. W. *Moderate Realism and its Logic*. London: Yale University Press, 1996, Quine, W. V. O. *Word and object*. Cambridge: MIT Press, 1960, Simons, P. *Parts: A Study in Ontology*. Clarendon: Oxford University Press, 1987, Smith, B. ed. *Parts and Moments*. Munchen: Philosophia Verlag, 1982, Varzi, A. 'Mereology', in Edward N. Zalta (ed.), *Stanford Encyclopedia of Philosophy*, Stanford: CSLI (internet publication), 2003. ftp.

categories and relations of BFO in particular, but are applicable to the construction of any domain ontology, based on almost any formal ontology whatsoever.

Basic features of formal ontologies

Formal ontologies specify reality along two essential dimensions. First, they state what the basic categories of reality are. Second, they state the basic relations that hold within and among objects belonging to the basic categories of reality.

The term ‘ontological category’, as it is being used here can be interpreted to mean “universal that applies to every material domain of reality,” in the case of formal ontologies, and “universal of highest generality within a domain” in the case of domain ontologies.[37]

The ontological categories in a formal ontology should include all and only very basic and very general kinds of entities. Examples of ontological categories include the category “universal” and the category “particular” that have already been discussed. Other ontological categories might include “individual”, “collective”, “property”, “dependent entity”, “process”, “event” or “spatial region”. Each of the categories in an ontology should be carefully defined, ideally involving the statement of identity conditions of the form “an x belongs to category y just in case it is a, b, and c…”, and no two categories should be identical or have the classes that they designate overlap (no entity in the world should belong simultaneously to two different formal ontological categories).

A good test of whether or not something is a formal ontological category is to try and find two universals from very different scientific domains, both of which can be correctly understood as belonging to the category in question. Thus the fact that cups, rocks, planets, organisms and perhaps even some social organizations can be described as substances, meaning as independently existing entities that maintain their identity through the loss and replacement of (most kinds of) parts and are also the bearers of qualities or properties of various sorts, makes “substance” a likely candidate for a formal ontological category.

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[37] The use of the term ‘category’ has a long and variegated usage in the history of philosophy, and new uses are being developed daily, both by philosophers and by researchers in the information sciences and in bioinformatics. We will not discuss all of the different possible uses of the term ‘category’ here. For an introduction to the philosophical usage and issues surrounding the term, see Thomasson, A., "Categories", *The Stanford Encyclopedia of Philosophy* (Winter 2005 Edition), Edward N. Zalta (ed.), URL = http://plato.stanford.edu/entries/categories/.

The formal relations contained in an ontology are extremely important. They serve to specify both the internal structure of objects that are to be found within ontological categories and the relationships that obtain between different ontological categories.

An example of a relation performing the first kind of function is the "part_of" relation. For any given object falling within any given ontological category, it is possible to further analyze that object by analyzing its parts.

An example of a relation performing the second kind of function would be the relation "instantiates". It is entities belonging to the ontological category "particular" that "instantiate" entities belonging to the ontological category "universal". Thus "Fido instantiates Golden Retriever", and "this-oxygen-molecule instantiates oxygen". An ontology that includes the categories "particular" and "universal", as well as the relationship of "instantiates" holding between them is both more explicit and more structured than one that only contains these categories. Further, it should be noted that the relationship "instantiates" can be used to help define each of these categories. For example, one could begin to define 'universal' as "an entity that can be instantiated", and one can begin defining 'particular' as "an entity that instantiates universals, but is not instantiated by any other thing." In such cases adding explicit formal relations to a system of ontological categories helps to implicitly define the nature of these categories themselves.

As with formal categories, so with formal relations, a good test of whether or not a relation is formal is to check and see if it applies to entities in many different scientific domains. The fact that cells, buildings, molecules, organisms, environments and football games can all be said to have "parts" in the same sense makes the "part_of" relationship a good candidate for inclusion in a formal ontology.

The dimensions of an ontology

Whereas formal categories and relations are intended to be basic features of any domain ontology whatsoever, the constraints imposed by a formal ontology alone do not yet help to determine what the specific content (in terms of domain specific universals and relations) to be included in a domain ontology should be. The question of what information should, and what information should not, be included in a given domain ontology is left unanswered by formal ontological considerations alone. There are at least two general dimensions along which the appropriate contents of a domain specific ontology need to be determined. These two dimensions might be called the "horizontal" and the "vertical" dimensions of a domain ontology respectively, and we will discuss them here under the headings of "relevance" and "granularity".

Relevance

The "horizontal" demarcation of the contents of an ontology is the problem of determining what and how much existing information about a given domain should be included in that ontology, and can be summarized by describing it as a problem of determining what is *relevant* for the ontology. An ontology should include only that information about reality that is relevant to the domain that it is a representation of.[38] The issue of relevance is a complicated one about which much has been written. Though there is no clear consensual definition of 'relevance' applying to all cases of its use, this does not mean that what is relevant to a given ontology is a merely subjective affair, to be decided purely by the attitudes, purposes or opinions of the ontology designers. Rather it is possible to understand relevance as an objective phenomenon, one tied to basic features of reality and of the various sciences that study those features of reality. In this connection there are two considerations that are especially important to keep in mind when determining what is relevant to a given ontology.

The first is that what is relevant to the ontological representation of a given domain will be determined by reality itself. Domains of reality contain their own internal objective relevancies. What is objectively relevant to a gene ontology, for example, will be determined first and foremost by the nature of genes themselves, what they are, what processes they characteristically initiate or are involved in, etc.

Similarly, an ontology explicitly intended to include the scientific information that we have about cells would be expected to include at least a complete listing of known types of cells (eukaryotic, prokaryotic, etc.) and a complete specification of the characteristic kinds of parts of each of these kinds of cells. However, while it is relevant to such an ontology to include the information that genetic material is located in cells, it is arguably not relevant to such an ontology to include detailed information about the nature of genes, their characteristic expressions or the processes involved in their recombination and transmission; this information would be much more relevant to an ontology devoted specifically to the representation of the

[38] Ceusters, Werner & Smith, Barry. "A realism Based Approach to the Evolution of Biomedical Ontologies." in *Proceedings of AMIA Symposium 2006,* forthcoming.

nature of genes and genetic processes themselves. Importantly, these relevancies are determined by the nature of genes and cells themselves: understanding the kinds of cells there are and the parts and processes that they are involved in, in short understanding the nature of cells, does not require understanding everything that there is to know about genetics. Thus the vast majority of information that is possessed by the science of genetics is not directly relevant to the representation of the ontology of cells, and so should not be included.

The fact that the nature of cells is different from the nature of genes suggests that there should be (at least) two separate domain ontologies, one for cells and one for genes. The way to handle the fact that there are connections between cells and genes is simply to build explicit links and references from the one ontology to the other in those places where the information contained in one comes to an end, and the information in another begins. Assuming that these domain ontologies have been structured in terms of the same formal ontology, and that terminological usage in the two is consistent, it should be possible to connect them and to bring them into alignment in this way.

The second major consideration to be kept in mind regarding relevance is that what is relevant to an ontology will also be determined by the purpose for which that ontology is being designed. Goal-oriented human activities generate their own sets of objectively relevant features of reality, actions, etc. For example, the project of going to the grocery store to buy milk automatically makes the following entities relevant: the store itself, the means of transportation, the transportational route, the milk, some means of payment, and an ordering amongst the series of activities that must be engaged in. The same can be said for scientific investigations (the goal of which is to gain new information about the world) and medical treatments (the goal of which is to heal a patient based on existing biological knowledge and medical techniques), though of course the number of things relevant to these goal oriented activities will be greater and involve more complex relations.

Reference and application ontologies

Recognizing these two kinds of relevancy substantiates the distinction between a reference ontology and an application ontology. A reference ontology is a representational artifact analogous to a scientific theory, in which expressive completeness and adequacy to the facts of reality are of primary importance. An application ontology, on the other hand, is a representational artifact designed to assist in the achievement of some specific goal. Reference ontologies will be constructed and structured primarily based on objective relevance, while application ontologies will be constructed and structured primarily in terms of goal-oriented relevancies. Ideally, application ontologies will make use of (and also be able to reuse) portions of reference ontologies in order to accomplish their particular ends.

When designing an ontology then, it is essential at the very beginning to carefully consider what the domain of the ontology is. This will include asking the question of whether the ontology is primarily intended to be a comprehensive representation of scientific information in a given domain, and thus a reference ontology, or whether it is primarily intended to be used by human beings in order to accomplish certain very specific goals, such as medical treatments, in which case it is an application ontology. In the former case what is relevant to include in the ontology will depend primarily on the nature of the objects that exist in the domain to be represented, while in the latter case what is relevant to the ontology will depend on what portions of reality and what information is objectively salient to achieving the stated objective or goal.[39]

Granularity

In a sense, the issue of determining the appropriate granularity of an ontology is just a sub-part of the problem of determining what is relevant to be represented in that ontology. However, whereas we have described the problem of relevance as the horizontal problem of determining how much and what kind of information to included in an ontology, the problem of granularity is the specifically vertical problem of determining how fine-grained or course-grained the ontology should be. The issue of granularity arises because things in reality, and also the parts of things, come in many different sizes and possess varying degrees of complexity. There is a continuum spanning from the level of sub-atomic particles, atoms and molecules, through the levels of ordinary everyday objects such as animals, rocks, tables and lakes, and on to the level of ecosystems, planets, solar systems, galaxies, and ultimately the universe itself. Things exist at all of these different levels, and as things become bigger and more complex (consisting of ever more and more diverse parts) they also exhibit features not to be found in

[39] See Christopher Menzel. "Reference Ontologies / Application Ontologies: Either / Or or Both/And?" in P. Grenon, C. Menzel, and B. Smith (eds.), *Proceedings of the Workshop on Reference Ontologies and Application Ontologies*, KI 2003, September 16, 2003, Hamburg, Germany. CEUR Workshop Proceedings, vol. 94 (2004), ISSN 1613-0073, & Fielding, Matthew James, Mariana Casella Dos Santos, and Barry Smith: "Reference Ontologies for Biomedical Ontology Integration and Natural Language Processing", in *International Journal of Medical Informatics*, forthcoming.

smaller less complex entities.[40] The problem of granularity for ontology design is the problem of deciding the upper and lower limits of entity size and complexity that are to be represented in a given domain-ontology. Should an ontology of mountains include information about the atoms out of which the mountains are composed, should an ontology of the circulatory system include detailed information about blood cells, and should an ontology of cells include detailed information about genetics? Each of these questions is a question about the appropriate granularity of the respective ontologies mentioned. Importantly, any given ontology should specify both an upper ('largest') and a lower ('smallest') boundary or level of granularity that it will represent.[41]

The appropriate granularity for an ontology should be decided, essentially, along the same lines as should be used in deciding what is relevant for inclusion in the ontology in general. Namely, in the case of reference ontologies by focusing on the nature of the objects to be represented, and in the case of application ontologies by focusing on the objective saliencies determined by the intended goal. There is, however, an important confusion that needs to be avoided when determining the granularity of an ontology.

Determining the granularity of an ontology (the size and complexity of the kind of objects that it represents) is not the same as determining the generality of an ontology (the abstractness of the universals that it represents).

Suppose that there was good reason to develop an ontology of human anatomy that did not represent anything beneath the level of granularity possessed by bodily organs such as the heart, kidneys, etc. In such a case, the ontology would represent universals and relations amongst them for all features of the body down to the level of organs, but nothing further. Now, this ontology would also include a universal representation for "human heart", and importantly "human heart" is (arguably) the most specific (least general) universal that can be discussed before one crosses the universal/particular divide and must begin discussing the existence of particular hearts instantiating this universal. Such an ontology would then be very "specific", in

[40] It is worth noting here that there is no guaranteed relationship between the size of an object and its complexity. Genes are very small and yet very complex, whereas mountains are very big, but relatively simple in their composition and functioning. Nevertheless, the issue of granularity encompasses both the literal question of what sizes of objects should be represented in an ontology and also what level of complexity should be represented, and thus the two issues are mentioned together here.

[41] For more on granularity and the theory of granularity, see Kumar Anand, Smith Barry, Novotny Daniel: "Biomedical Informatics and Granularity", in: Functional and Comparative Genomics, 5 (2004) , 501-508, and the papers at the following link http://ontology.buffalo.edu/smith/articles/vagueness.html.

that it would represent universals falling at the bottom of their respective hierarchies, but it would also have a relatively coarse granularity (insofar as there is more to human anatomy than just the level of the whole body through the level of bodily organs).

Alternatively, there might be reason to develop an ontology of genes that contained only information common to all genes, no matter what particular living thing they are found in. Such an ontology might well leave out of account many features of genes that exist only in particular species of organisms, but not in all living things. This ontology would be very general in the sense that the universals it represented would be higher in the taxonomic hierarchy of gene-universals, but it would (or at least could) still have a very fine grained granularity, insofar as it is representing things that exist at the level of genes, their components, their properties and the processes that they engage in.

From this it should be clear that the task of determining the level of granularity that an ontology will represent (how fine- or course-grained it will be) is separate from the task of determining the level of generality that that ontology will represent (in terms of the abstractness of the universals that it contains).[42]

Chapter 5: Introduction to Basic Formal Ontology and Relations

Fully structuring domain ontologies in a way that makes them computer-tractable and interoperable, as well as in a way that renders the information that they contain as clear, rigorous, and unambiguous as possible, requires the use of formal or "top-level" ontologies. In addition, the important issues of terminology selection, term-definition, and classification in the sense of the construction of tree-like structures organizing the information contained in a domain ontology all benefit from and can be better understood in the context of, an explicitly defined formal ontology. Here we provide an introduction to some of the core features of the Basic

[42] For a conflation of these two issues, see Natalya F. Noy and Deborah L. McGuinness. ``Ontology Development 101: A Guide to Creating Your First Ontology''. Stanford Knowledge Systems Laboratory Technical Report KSL-01-05 and Stanford Medical Informatics Technical Report SMI-2001-0880, March 2001. "Deciding where classes end and individual instances begin starts with deciding what is the lowest level of granularity in the representation. The level of granularity is in turn determined by a potential application of the ontology. In other words, what are the most specific items that are going to be represented in the knowledge base?" http://ksl.stanford.edu/people/dlm/papers/ontology-tutorial-noy-mcguinness.pdf

Formal Ontology (BFO in what follows) that has been developed at the Institute for Ontology and Medical Information Science in Saarbrücken Germany.

A few Initial Remarks

In order to avoid misunderstandings and confusion, a few brief remarks about the Basic Formal Ontology should be made at the outside. Most of these points will be made again and discussed in more detail below.

First, BFO is very general, top-level, and formal: it won't park your car for you. To say this is simply to stress the fact that a top-level ontology by itself is no panacea. Even once such an ontology has been adopted for use, a great deal of work will remain to be done in terms of integrating the top-level ontology with domain specific information.

Second, the BFO categories are categories containing or representing universals primarily. While it is possible to classify particulars in terms of the BFO categories, the primary intended use for BFO is in the structuring of domain ontologies, which, qua ontologies, have the task of organizing and representing the universals that are the objects of general knowledge in scientific domains.

Third, BFO's perspectivalism is serious. The SNAP/SPAN divide (to be discussed below) must be kept in mind, and confusions between continuants and occurrents, between objects and processes should be strenuously avoided. Also, the BFO categories are meant to apply to entities at or within a specific level of granularity. No universal should belong to two or more BFO categories (object, object aggregate, process, etc.) at any given level of granularity, but the converse of this is that failure to carefully respect the level of granularity required for one's domain ontology can lead to instances of multiple inheritance under BFO classes, which should be avoided.

Fourth, BFO is non-committal in a number of ways. Both because of its generality, and because of its perspectivalism, BFO will not necessarily have an answer for every domain specific, or even for every philosophical problem or question. This does not mean that there is no answer, or that an answer within the framework of BFO cannot be found, but it does mean that some questions will have to be addressed and carefully worked through at the time of implementation.

Fifth, many BFO category partitions are not strictly speaking exhaustive, nevertheless they are treated as exhaustive for purposes of domain applications in the context of science. More will be said about this below.

Sixth, BFO is to be extended downward, and to have relationships explicitly added to it, but outward extension of the existing BFO classes is highly unlikely to be necessary.

BFO

BFO has been developed in accordance with the principles of realism, adequatism, fallibilism and perspectivalism that have been discussed above. As a realist ontology, the intended interpretation of the basic categories and relations of BFO is that of real divisions amongst the kinds of entities that exist in the mind-independent world. Adequatism means that BFO has been designed to directly take into account and represent the various kinds in reality, whether fundamental particles of physics, surgical procedures or presidential elections. The principle of fallibilism entails acknowledgement of the possibility that certain basic assumptions may need to be revised in light of future researches, both in the empirical sciences and in ontology. Finally comes BFO's perspectivalism.

Perspectivalism states that while not all perspectives or ways of dividing up reality are good, there are nevertheless many equally good (equally veridical or true) ways of dividing up and representing reality from different perspectives. The most straightforward example of different perspectives regarding the same thing that are nevertheless equally veridical that has been discussed is granularity. It is perfectly legitimate to examine a living organism, such as a rhinoceros, from the perspective of its characteristic genetic material and code. On the other hand, there are scientifically interesting things to be learned about rhinoceroses that can only be investigated by adopting a perspective which takes into account the whole animal, its typical environment, and the various kinds of behavior that it typically engages in. The view of rhinoceroses in terms of their genes does not typically include information about their behavior or their environment, and vice versa. Nevertheless, these are two equally good scientific perspectives to take on one and the same kind of animal. Ontological perspectivalism acknowledges the existence of these different perspectives and requires that both be expressible within a formal ontology. Consistent with this insight BFO recognizes ontologies, perspectives that have been rendered into formalized representational artifacts, at many different levels of granularity.

Aside from granularity, there is another great perspectival divide within BFO, one of equally fundamental importance. This is the distinction between continuant and occurrent entities, or between SNAP and SPAN ontologies.[43] In what follows we will develop the

[43] The following are technical reports and papers on the various features of BFO: Pierre Grenon: "Spatio-temporality in Basic Formal Ontology: SNAP and SPAN, Upper-Level Ontology, and Framework for Formalization", Pierre Grenon: "BFO in a Nutshell: A Bi-categorial Axiomatization of BFO and Comparison with DOLCE", Pierre Grenon, "Nuts in BFO's Nutshell: Revisions to the Bi-categorial Axiomatization of BFO", Barry Smith. "Against Fantology." In M. E. Reicher & J. C. Marek (eds.), *Experience and Analysis.* Wien, 2005, Pierre Grenon and Barry Smith. "SNAP and SPAN: Towards Dynamic Spatial Ontology." *Spatial Cognition and Computation,* Barry Smith and Pierre Grenon, "The Cornucopia of Formal-Ontological Relations," *dialectica* 58, no. 3 (2004), Pierre Grenon, Barry Smith and Louis Goldberg. "Biodynamic Ontology: Applying BFO in the Biomedical Domain." In D. M.

taxonomy of BFO classes that fall under SNAP and SPAN, beginning with the top-level classes and working downwards through the sub-classes, first for SNAP and then for SPAN.[44]

The great divide

BFO recognizes a basic distinction between two kinds of entities: substantial entities or continuants, and processual entities or occurrents. Corresponding to these two kinds of entities are two basic and distinct perspectives that can be taken on the world, neither of which can fully capture or represent the features of reality represented by the other: these are the SNAP and SPAN perspectives or ontologies respectively. Each of these basic perspectives can also be used to represent entities at different levels of granularity, resulting in further perspectival sub-divisions of the basic SNAP and SPAN ontologies. Finally, BFO includes the specification of formal relationships obtaining amongst objects within ontological categories, relationships obtaining between categories within an ontological perspective, and also relationships obtaining between ontological perspectives. Here we will begin by characterizing some of the basic categories of entities recognized by BFO. We will then discuss the perspectivalism of BFO as it relates to these categories and to issues of space, time and change. Finally, we will introduce, define and discuss some of the crucial ontological relations recognized by BFO, and discuss their connection to the rest of the ontology.[45]

PIsanelli (ed.) *Ontologies in Medicine.* Amsterdam: IOS Press, 2004, 20—38, Barry Smith and Pierre Grenon. "The Cornucopia of Formal Ontological relations." *Dialectica* Vol. 58, 3, (2004), pp. 279—296.

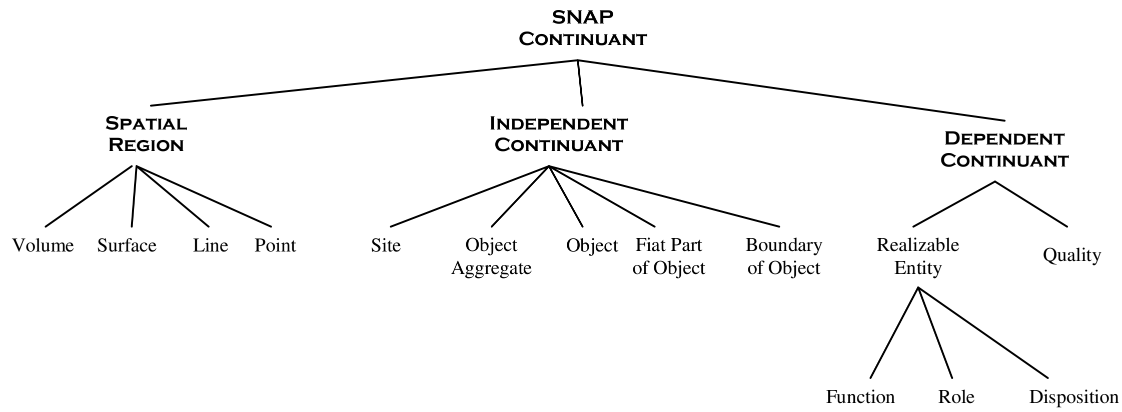
[44] It should be noted here that BFO remains neutral on the question of how many different kinds of ontological perspectives there may be. Here we focus on granularity and SNAP/SPAN as the crucial perspectives in understanding BFO. Another perspectival division in BFO could be said to be that between particulars and universals. As a formal ontology, BFO is primarily interested in representing universals. However, there is no reason in principle why particulars could not be viewed through and organized in terms of the BFO universals as well.

[45] For more on this basic distinction, see Bittner, T., Donnelly, M., & Smith, B. "Endurants and Perdurants in Directly Depicting Ontologies." *AI* Communications, ISSN 0921-7126, IOS Press. See also Partridge, Chris. Note: A Couple of Meta-Ontological Choices for Ontological Architectures." *Technical Report 06/02, LADSEB-CNR Padova, Italy, June 2002,* http://suo.ieee.org/email/msg03740.html. For examples of an ontology with a similar continuant – occurent distinction, see Gangemi, A. et. al. "Sweetening Ontologies with DOLCE", http://citeseer.ist.psu.edu/cache/papers/cs/26864/http:zSzzSzwww.ladseb.pd.cnr.itzSzinforzSzOntologyzSzPaperszS

The BFO categories: SNAP Continuant

The SNAP portion or perspective of BFO represents continuants: entities that endure through time while maintaining their identity. Examples of such entities include a human individual, the color of a ripe apple, the disposition of an organism to bleed, and the Berlin Wall. The SNAP ontology recognizes three major categories of continuants: dependent continuants, independent continuants and spatial regions. The defining feature of independent continuants is that they are the kinds of things in which other continuants, such as qualities and dispositions, can inhere. They are the bearers of qualities and other dependent continuants. The defining feature of dependent continuants is that they are the kinds of things (qualities, roles, functions) that inhere in or are born by something else (namely independent entities). Spatial regions are different from both independent and dependent continuants in that they neither inhere in anything, nor are they themselves bearers of qualities, thus nothing inheres in them. Each of these categories admits of further sub-divisions or sub-categories. Here we will present first the SNAP, and then the SPAN categories of BFO, starting with the most general categories (SNAP Level 1 and SPAN Level 1), and then working downwards through the sub-categories or sub-classes of these (SNAP Level 1+n & SPAN Level 1+n).

zDOLCE-EKAW.pdf/gangemi02sweetening.pdf; for discussion of the issue and a different basic commitment regarding continuants and occurrents, see "Heller, B & Herre, H. "Ontological Categories in GOL", *Axiomathes* 14: 57—76, 2004, http://www.onto-med.de/en/theories/gfo/index.html.



Level 1

SNAP: Continuant

Df: An entity that exists in full at any time in which it exists at all, persists through time while maintaining its identity, and has no temporal parts. (examples: a heart, a person, the color of a tomato, the mass of a cloud, a symphony orchestra, the disposition of blood to coagulate, the lawn and atmosphere in front of the White House)

SNAP continuant has exactly three mutually exclusive and jointly exhaustive sub-classes. These are: spatial region, independent continuant and dependent continuant.

Level 2: sub-classes of Continuant

Spatial Region

Df: An independent continuant entity that is neither a bearer of qualities nor inheres in any other entities. (examples: the sum total of all space in the universe, parts of the sum total of all space in the universe)

According to BFO, SNAP space is an entity in its own right, something like a giant three-dimensional container within which various other continuants and continuant qualities exist and move. This feature of BFO makes it possible to represent the location of entities and of their various parts and qualities both at a time and across time, and also to talk about unoccupied spatial regions. Examples of spatial regions include the space occupied by a tomato at a given time, the space that was occupied by a tomato at one time, but is now empty due to the fact that the tomato has been moved, and the sum total of all space in the universe.

Spatial regions are very unique kinds of entities in SNAP. They are considered entities in the strict sense, however, they are neither concrete material entities like most other independent entities, nor are they dependent in the way that qualities and realizables are. The 'space' of spatial regions is the absolute or total space of physics, and should be carefully distinguished from spaces or places that are relativized to particular continuant entities or aggregates of such entities along with the medium or matter that they enclose; there is a separate category for dealing with such "relativized spaces" in BFO and it is the category "site". As a general rule, unless one is building a domain ontology for the representation of information about physics, information about location and place should be treated in terms of BFO site, not in terms of BFO spatial region.

Independent Continuant

Df: The continuant entities that are the bearers of qualities and realizables; entities in which other entities inhere and which themselves cannot inhere in anything. (examples: an organism, a heart, a symphony orchestra, a chair, the bottom right portion of a human torso, a leg, a person, a symphony orchestra, the lawn and atmosphere in front of the White House)

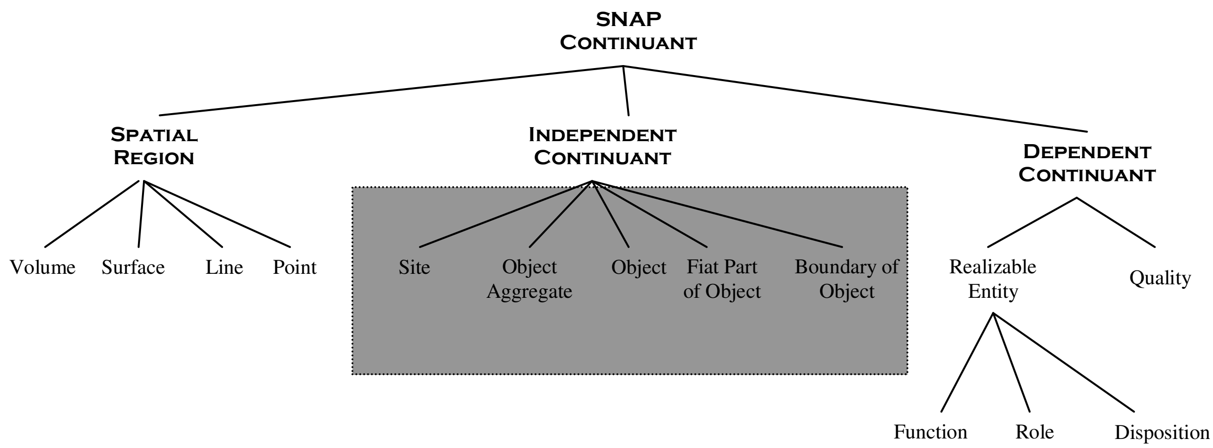
Independent continuant entities come in many forms, but their key feature is that of being the bearers of properties or qualities (SNAP dependent entities). Independent continuant entities are often, but not always (the major exception being "boundary"), existentially independent with regard to other entities. This means that their present and future existence is not strictly dependent on the existence of any other beings or kinds of beings.

Dependent Continuant

Df: The continuant entities that inhere in or are born by other entities. (examples: the mass of a cloud, the color of a tomato, the smell of mozzarella, the liquidity of blood, the disposition of fish to decay, the role of being a doctor, the function of the heart to pump blood)

Dependent continuant entities inhere in other dependent entities and in independent continuant entities. They exhibit a kind of existential dependence insofar as in order for them to exist, some other entitie(s) or kind(s) of entitie(s) in which they inhere must exist as well.

Level 3: Sub-Classes of Independent Continuant



Site

Df: An independent continuant consisting of a characteristic spatial shape in relation to some arrangement of other continuant entities and of the medium which is enclosed in whole or in part by this characteristic spatial shape. Sites are entities that can be occupied by other continuant entities. (examples: a city, a nasal cavity, a blood vein, an environment, sinuses, canals, ventricles, the lumen of the gastrointestinal tract, the location of the battle of Austerlitz, the interior of the aorta, the room in which one is located, the top of one's desk including the things that are on it and the atmosphere surrounding them)

For example, a room, understood as the walls of the room along with the space that they enclose, is a site. However, the space enclosed by a room taken by itself, is also a site, not a spatial region. There are two ways of understanding how this is so.

First, the space (like the air) enclosed by a room (site) moves through absolute space (spatial regions) continuously (as the earth, on which the building containing the room is located, itself turns in and moves through the absolute space of the universe). Thus, while at any given moment the "space in a room" is co-extensive with some spatial region, it is not identical to that spatial region because the site that is the interior of the room remains what it is even after the room, and the site with it, has itself moved so that it occupies a new and numerically distinct spatial region. The site in a room is thus spatial shape that is "relativized" to some other continuant entity or entities, including the walls of the room and also the air which is the filling or medium in the room.[46] Similar things can be said about other sites, such as nasal cavities, conduits, the insides of stomachs, and environments. They consist of a continuant entity or set of continuant entities along with some associated medium-filled spatial shape and structure that is not strictly identical with any particular spatial region.

The point here is not that "sites are spatial regions that can move". Spatial regions by definition cannot move, rather they are that in relation to which things move, while sites are not spatial regions, though they are always located at one, and it is not essential to a site qua site that it be mobile in any way. A nasal cavity would still be a site, even if it was located in a

[46] For more on relativized space, see Maureen Donnelly, "Relative Places", in *Applied Ontology 1,* (2005) 55-75.

numerically identical spatial region for its entire life-time. The point is that sites are continuant entities that have an arrangement of other continuant entities and a medium as parts, and which normally exhibit a characteristic spatial shape as well. However, one way of seeing that sites are different form spatial regions is by understanding that sites can be located at many different spatial regions while still maintaining their identity as sites (your nasal cavity does not become something new or different every time you turn your head).

Second, sites, but not spatial regions, typically consist not only of an arrangement of continuant entities and an associated spatial shape, but also of a medium such as air or fluids that is partially or completely enclosed by the arrangement of continuant entities and the spatial shape that is associated with them. Thus the nasal cavity is a site that consists of the inner membranes and parts of the nose, along with the spatial shape of the cavity itself and the oxygen or (in more unfortunate circumstances) other medium that fills and is partially enclosed by the cavity. Similarly, the cranial cavity is a site that consists of the skull, the brain, and the fluid that almost entirely fills the spaces that are enclosed by the skull walls and the brain taken together. Spatial regions do not, qua spatial regions, ever have any medium inside of them, they are pure space or pure "space shapes", whose definition does not require them to have any continuant entities whatsoever located in them.

As noted previously, site should be kept carefully separate from spatial region.[47] In domains such as the biomedical one, it is, in general, site, not spatial region, that should be used in coding information about location and place.

Object Aggregate

[47] For more on the kinds of entities that will plausibly count as sites, see Barry Smith and Achille Varzi, "The Niche", *Nous*, 33:2 (1999), 198–222, as well as other papers at http://ontology.buffalo.edu/bio/niche-smith.htm; also Casati, R. and Varzi, A. "Holes", *The Stanford Encyclopedia of Philosophy* (Winter 2003 Edition), Edward N. Zalta (ed.), URL = http://plato.stanford.edu/entries/holes/; and Roberto Casati and Achille C. Varzi. Holes and Other Superficialities. A Bradford Book. The MIT Press, Cambridge, Massachussets, 1994.

Df: A independent continuant entity that is a mereological sum of separate objects. (examples: a heap of stones, a group of commuters on the subway, a collection of bacteria, a flock of geese, the patients in a hospital, a symphony orchestra).

Aggregates are continuants that are collections of other separate objects. This means that they possess a degree of unity or connectedness that is weaker than that possessed by objects proper (compare a heap of stones to a stone), that they have non-connected boundaries (there are gaps or spaces between some or all of their parts), and that they have as parts only objects of the SNAP Object sort (see following definition).

Object (Substance)

Df: A independent continuant entity that is spatially extended, maximally self-connected and self-contained (the parts of a substance are not separated from each other by spatial gaps), and possesses an internal unity; the identity of substantial objects is independent of that of other entities and can be maintained through time and through loss and gain of parts and qualities. (examples: an organism, a chair, a cell, a lung, an apple)

Objects (in the philosophical sense of 'substance' here) are perhaps the most familiar and immediately accessible entities that the world contains. When asked to name some entities, a person is likely to respond with a list something like the following: a bus, an apple, a desk, a cat, a building, a tree, a pencil, etc. These entities all have certain features in common that qualify them for inclusion in the category of object (substance). Whereas a pile of rocks or the pieces of a dismembered chicken possess very little structure and internal unity as entities, it is characteristic of BFO objects to possess a high degree of unity. This unity is to be understood, on the one hand, in terms of the topological notion of being self-connected and self-contained,[48] and on the other by the fact that these entities have identity conditions that are not dependent on the existence of other entities (in the way in which the identity conditions of an instance of color or mass entail that there also exist some other entity that it is the color or mass of). BFO objects are characteristically independent entities that maintain their identity through time while gaining and losing qualities or parts, and that exist in their entirety at any moment in which they exist at all. Each of these features of substance will be addressed in turn.[49]

An independent entity is an entity that can exist and be what it is regardless of what other objects exist. Thus a doorknob is an independent entity because it can be removed from a door and still exist as a doorknob, a rock is an independent entity because it can be moved from one place to another, and the objects around it can be destroyed, removed or replaced, and it will remain just what it is, a rock. Organisms are also independent entities, insofar as their existence as organisms involves a certain unity, capacity for locomotion, and independence of any exact location.[50] In each of these cases the thing's being what it is, the identity conditions of the thing

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[48] Grenon, P. & Smith, B. "A Formal Theory of Substances, Qualities and Universals", in Achille Varzi and Laure Vieu (eds.), *Proceedings of FOIS 2004. International Conference on Formal Ontology and Information Systems*, Turin, 4—6 November 2004; Barry Smith, "Mereotopology: A Theory of Parts and Boundaries", *Data and Knowledge Engineering*, 20 (1996), 287-303.

[49] This listing of features is arguably not a complete definition of what it is for a thing to be a substance. However, the features here listed are almost universally acknowledged to be characteristics of what is meant by 'substance', and so we focus on them here. For more on the notion of "substance" and competing definitions, see Neuhaus, F., Grenon, P. & Smith, B. "A Formal Theory of Substances, Qualities and Universals", in Achille Varzi and Laure Vieu (eds.), *Proceedings of FOIS 2004. International Conference on Formal Ontology and Information Systems*, Turin, 4—6 November 2004, Barry Smith and Berit Brogaard, "Sixteen Days," *Journal of Medicine and Philosophy* 28, no. 1 (2003).; E. J. Lowe, *The Possibility of Metaphysics: Substance, Identity and Time* (Oxford: Oxford University Press, 1998).; Robinson, H.., "Substance", *The Stanford Encyclopedia of Philosophy* (Winter 2004 Edition), Edward N. Zalta (ed.), http://plato.stanford.edu/entries/substance/.

[50] Organisms are of course highly complex entities, and there are a number of senses in which they are dependent on other organisms, on environmental niches, etc. However, the primary issue here is simply the question of whether or not an entity can exist as what it is independently of other entities. In the case of organisms, the fact that an organism exists at a given moment does not entail anything about what else must exist at that exact moment (even though there are many things that probably exist, none of them are strictly entailed by the simple fact of an organism's existing), and in this sense their existence is independent of that of any other specific entities.

are, as it were, internal to the thing itself, requiring only the existence of the thing and not of any other things in order for it to be the kind of thing that it is.

The feature of objects having to do with their ability to gain and lose parts and qualities is best illustrated by the case of organisms. During her lifetime an average human being will lose and replace a tremendous number of blood, bone and tissue cells, never mind the loss of hair, dead skin cells and other bodily matter. In spite of all of this changing and replacing of bodily parts, it is still the same person that exists at different points in her life. Aging by 5 years, visiting the Barber or losing a finger will all involve the loss, gain and/or re-organization of the compositional parts of a human being to some extent, but none of these things by itself results in a new human being coming into existence, or in an entity's failing to continue to be the kind of entity that it is, in this case a human being. Similarly, human beings gain and lose a number of qualities as they age, grow and mature. At one time Jill's hair might be blond, at another time brown; at one time Jill may be fit and athletic, while at another time she may be some what out of condition, or even downright sedentary; at one time Jill may believe that philosophy is a useless speculative endeavor with no real connection to or implications for human life, while at another time she may come to recognize the foundational role that has been and still is played by philosophy in the history and existence of Western Culture. In each of these cases Jill gradually loses one quality (hair-color, physical fitness, a belief) and takes on another in its stead. But once again, in spite of these changes, Jill remains identical to herself, and she also remains an instance of the kind human. It is a characteristic feature of BFO objects that they are able to undergo both of these kinds of changes in time while maintaining their identity.

The final basic feature of BFO objects, that they exist in total or are wholly present at every moment in which they exist at all, is primarily intended to contrast substances with entities belonging to the BFO category of processes, and so will be further clarified shortly. However, the idea is that for continuant objects, all of the parts that they have exist, along with them, at any moment at which they exist. Thus, right now Jill has all of her bodily parts and qualities intact and functioning, and a complete inventory of all of the parts of Jill need make reference only to what exists in the present moment. A heart attack, on the other hand is an event or process, an entity that exists, but that occurs in time and has as parts a beginning, a middle and a (however tragic) end. Thus a complete inventory of all of the parts and qualities of a heart attack must make reference, not only to what exists in the present moment, but also to what has existed at different points in time. Objects are thus independent entities that exist through loss and gain of parts and qualities in time as self-identical, and that are wholly present at any time at which they exist. Examples include organisms, cells, atoms, rocks, planets, and books.

Fiat part of object

Df: A independent continuant entity that is part of a object but is not demarcated by any physical discontinuities. (examples: upper and lower lobes of the left lung, the dorsal and ventral surfaces of the body, the east side of Copenhagen, the lower right portion of a human torso)[51]

Fiat parts are to be contrasted with bona fide parts. The bona fide parts of an object are, roughly, those that are connected to it in such a way that there is a natural discontinuity between them. Thus the head, wings and legs of a chicken, along with the bones of its skeleton and its internal organs, are all bona fide parts of the chicken. On the other hand, a two inch wide slice cut from the chicken's wing, or a spherical mass of biomatter arbitrarily removed from its inside, will be fiat parts of the chicken. Tissue samples, parts of objects demarcated for specific

[51]For more on fiat parts and fiat objects see Barry Smith, "Fiat Objects" (expanded version, Topoi 20: 2, 2001, 131–148), and the other papers at http://ontology.buffalo.edu/smith/articles/fiat.htm.

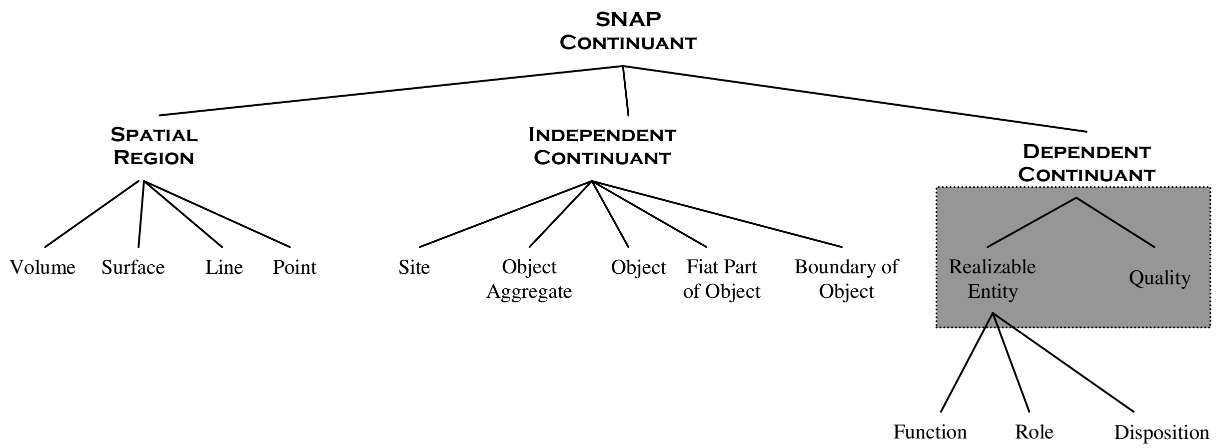
purposes, and certain kinds of relatively unified masses of stuff, such as a piece of skin, a pound of meat or a fragment of bone all count as fiat parts of objects.

Boundary of object

Df: A independent continuant entity that is a lower-dimensional part of some other continuant entity. (examples: the surface of skin, the surface of the earth, the surface of the interior of the stomach, the outer surface of a cell)[52]

In physical entities boundaries are often a closed two-dimensional surface (inner or outer) of a thing. Another way of thinking about boundaries is that they are those privileged parts of objects that exist at exactly the point where the object is separated off from the rest of the existing entities in the world. While the theoretical definition of a boundary is rather complicated, the intuitive idea of what a boundary is can, for the most part, be trusted as a guide in the case of identifying domain specific entities as BFO boundaries.

Level 3: Sub-classes of Dependent Continuant



Realizable entity

[52] See Barry Smith. "Boundaries: An Essay in Mereotopology." In L. Hahn, ed., *The Philosophy of Roderick Chisholm* (Library of Living Philosophers), LaSalle: Open Court, 1997, 534-561.

Df: Realizable entities are dependent continuants that inhere in continuant entities and are not exhibited in full at every time in which they inhere in an entity or group of entities. The exhibition or actualization of a realizable entity is a particular manifestation, functioning, or process that occurs under certain circumstances. (examples: the role of being a doctor, the function of the reproductive organs, the disposition of blood to coagulate, the disposition of metal to conduct electricity)[53]

Realizable entities are entities whose lives contain periods of actualization, when they are manifested as transformations or processes in their bearers, and also periods of dormancy, when they exist and inhere in their bearers, but are not manifested. Some realizables (such as the fragility of a vase) can only be manifested once in their lifetime (for example, in the event of the vase's breaking).

Quality

Df: A dependent continuant that is exhibited if it inheres in an entity or entities at all (a categorical property). (examples: the color of a tomato, the ambient temperature of air, the circumference of a waist, the shape of a nose, the mass of a piece of gold, the weight of a chimpanzee)

As opposed to SNAP objects, qualities are a paradigmatic kind of dependent entity. What all dependent entities of this sort have in common is that they inhere in other entities; thus in order for them to exist some other entity or entities must also exist. Examples of qualities include the elasticity of skin, the mass of a kidney, the rationality of a person, the color of blood, the shape of a hand, etc. Notice that in each of these cases the quality is mentioned in relationship to some other substantial entity, such as skin, a kidney, a hand, etc. This is because of the dependent nature of qualities. There cannot be color without it being the color of something and there cannot be mass without it being the mass of something. In particular, it is BFO independent continuants that qualities depend on or "inhere_in". It is also often the qualities of objects, or at

[53] For more on realizable entities, see Ludger Jansen. "The Ontology of Tendencies and Medical Information Science." In Ingvar Johansson and Bertin Klein (Eds.), *WSPI 2006: Contributions to the Third International Workshop on Philosophy and Informatics.* Saarbrücken, May 3—4, 2006. http://home.arcor.de/metaphysicus/Texte/tendencies%20WSPI2006%20proc.pdf#search=%22Ludger%20Jansen%2C%20tendencies%22; Fara, Michael, "Dispositions", *The Stanford Encyclopedia of Philosophy (Fall 2006 Edition)*, Edward N. Zalta (ed.), URL = <http://plato.stanford.edu/archives/fall2006/entries/dispositions/>.

least certain of them, those that are characteristic or essential, that are made use of in classifying them.[54]

Level 3: Sub-classes of Spatial Region

volume/three dimensional

Df: a spatial region with three dimensions. (examples: a cube-shaped part of space, a sphere-shaped part of space)

surface/two dimensional

Df: a spatial region with two dimensions. (examples: the surface of a cube-shaped part of space, the surface of a sphere-shaped part of space, the surface of a rectilinear planar figure-shaped part os space)

line/one dimensional

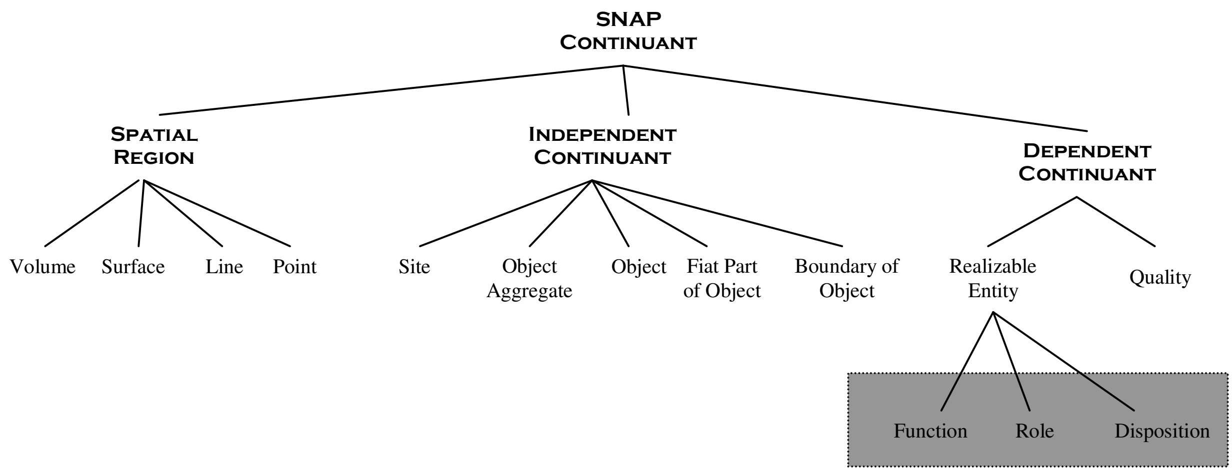
Df: a spatial region with one dimension. (examples: the part of space that is a line stretching from one end of absolute space to the other, an edge of a cube-shaped part of space)

point/zero dimensional

Df: a spatial region with no dimensions.

Level 4: Sub-classes of Realizable Entity

[54] For discussion of some features of qualities, see Johansson, I, "Determinables as Universals", *The Monist* 1, 2000, pp. 101-121, Johansson Ingvar: "Qualities and the Endurant-Perdurant Distinction", in: Roth-Berghofer Thomas et al. (eds.): Proceedings of the 3rd Conference on Professional Knowledge Management (WM 2005), Kaiserslautern, 2005, 543-550.



Function

Df: A realizable entity the manifestation of which is an essentialy end-directed activity of a continuant entity in virtue of that continuant entity being a specific kind of entity in the kind or kinds of contexts that it is made for. (examples: the function of a birth canal to enable transport, the function of the heart in the body to pump blood, the function of reproduction in the transmission of genetic material, the digestive function of the stomach to nutriate the body, the function of a hammer to drive in nails, the function of a computer program to compute mathematical equations, the function of an automobile to provide transportation, the function of a judge in a court of law)

A functioning is characteristically an activity or process, a doing of something. However, the ground of a functioning, and the fact about the world that makes a process a functioning rather than just a happening, is that it is a process or activity brought about by the realization of an essential dependent feature of a kind of object, which is aimed at or directed towards the realization of that object's essential goal or goals. The function of a kind of entity must be characterized in relation to the natural or intended context(s) in which that entity exists. Thus the function of a heart in the context of a living organism is to pump blood, the function of a hammer in the context of building things is to be used to drive in nails, and the function of reproduction in the context of a biological population is the transmission of genetic material. The function of a thing is what it is to do or what it is meant or intended to do and when a thing has a function it is not possible to remove this function and still have the thing remain what it is (a hammer made out of fragile glass is not properly speaking a hammer, any more than a heart on the table in a laboratory is a heart). Only organisms, certain tool-like products of organisms, intentionally designed artifacts, and certain kinds of social groups or institutions have functions.[55]

Role

Df: A realizable entity the manifestation of which brings about some result or end that is not essential to a continuant entity in virtue of the kind of thing that it is, but that can be served or participated in by that kind of continuant entity in some kinds of natural, social or institutional contexts. (Examples: the role of a person as a surgeon, the role of an artificial heart in pumping blood, the role of a chemical compound in an experiment, the role of a drug in the treatment of a disease, the role of a patient relative as defined by a hospital administrative form, the role of a woman as a legal mother in the context of system of laws, the role of a biological grandfather as

[55] Peter Godfrey-Smith. "Function", in Jaegwon Kim & Ernest Sosa (eds.) *A Companion to Metaphysics,* Malden: Blackwell, 1995, pp. 187—88. For IFOMIS writings that include discussions of biological function, see Johansson, Ingvar. "Functions, Function Concepts, and Scales." *The Monist* 86, 2004, 96—115, < http://hem.passagen.se/ijohansson/functions1.htm >; Johansson, I., Smith, B., Tsikolia, N. *et al*. 2005. Functional Anatomy. A Taxonomic Proposal. *Acta Biotheoretica*, 53(3), 153-66, < http://ontology.buffalo.edu/medo/Functional_Anatomy.pdf >; Smith, B. and Grenon, P. 2004. The Cornucopia of Formal-Ontological Relations. *Dialectica*, vol. 58, no. 3; Smith, B., Munn, K. and Papakin, I. 2004. Bodily Systems and the Spatial-Functional Structure of Human Body, in D. Pisanelli and M. Domenico (eds.) *Ontologies in Medicine: Proceedings of the Workshop on Medical Ontologies*. Rome, October 2003, Amsterdam: IOS Press, 39-63, < http://ontology.buffalo.edu/medo/OBS.pdf >. For a concise overview, synopsis and extension of the views put forward in these papers, as well as a contrast with other views of biological function, see Díaz-Herrera, Patricia. "What is a Biological Function?" forthcoming in *Proceedings of FOIS 2006.* For a general overview of positions and issues regarding teleology and function in biology, see Allen, Colin, "Teleological Notions in Biology", *The Stanford Encyclopedia of Philosophy (Summer 2004 Edition)*, Edward N. Zalta (ed.), URL = <http://plato.stanford.edu/archives/sum2004/entries/teleology-biology/>. On artifactual function, see Hilpinen, Risto, "Artifact", *The Stanford Encyclopedia of Philosophy (Fall 2004 Edition)*, Edward N. Zalta (ed.), URL = http://plato.stanford.edu/archives/fall2004/entries/artifact/; for more detailed treatments, see Dipert, R., 1993, *Artifacts, Art Works, and Agency*, Philadelphia: Temple University Press, and Thomasson, A., 2003, 'Realism and Human Kinds', *Philosophy and Phenomenological Research* 67, 580-609.

legal guardian in the context of a system of laws, the role of a tree in maintaining stability in an ecosystem, the role of ingested matter in digestion, etc.)

The roles that a continuant object does or can realize involve that object contributing mediately or immediately to a series of events that, in a particular context, brings something about, causes something to happen or can lead to the achievement of certain goals. What is crucial for understanding roles is that they are non-essential dependent realizable features that a continuant can take on, but that are not essential to the identity of that continuant. A heart in the context of the human body has the function of pumping blood, but in the context of an attack upon a human body by a hungry lion, that same heart can play the role of dinner for the lion. Similarly, the regular interactions amongst genes can, if the genes are assigned certain representational roles, be used to represent and solve certain kinds of mathematical equations; in this case the genes and relationships amongst them play the role of representational units in a computer program, but this is not their function. A gene would still be a gene and a heart a heart, even if the first was never used as a representational unit in a computer program, and the second was never eaten by a lion. In certain social contexts a person can play the role of being a lawyer, but it is not essential to persons that they be lawyers. Note that the role of "being a lawyer" is a social artifact and has a function in its appropriate social contexts, however the function is of the role, not of the entity that takes on the role. Thus even though it is the function of a lawyer to take certain kinds of actions in certain social contexts, it is not the function of the person who plays the role of lawyer to take certain kinds of actions in certain social contexts except in a derivative and secondary sense of 'function', one mediated by a thing's playing certain kinds of roles which themselves have associated functions.

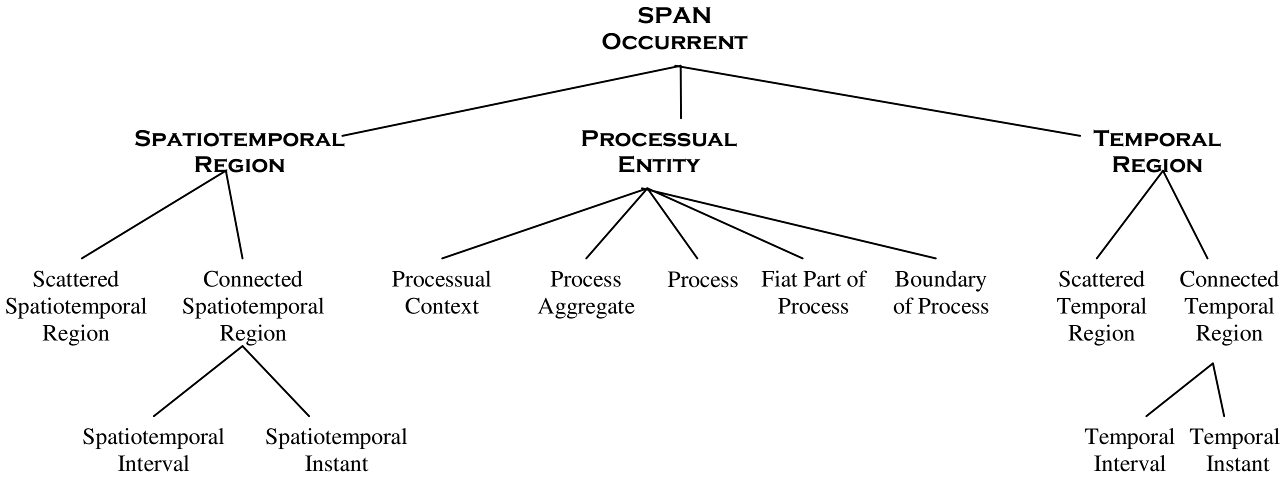
Disposition

Df: A realizable entity that essentially causes a specific process or transformation in the object in which it inheres, under specific circumstances and in conjunction with the laws of nature. A general formula for dispositions is: X (object) has the disposition D to (transform, initiate a process) R under conditions C. (examples: the disposition of vegetables to decay when not refrigerated, the disposition of a vase to brake if dropped, the disposition of blood to coagulate, the disposition of a patient with a weakened immune system to contract disease, the disposition of metal to conduct electricity)

Dispositions are essentially defined by their manifestations, to be fragile is to break under certain circumstances and to be soluble is to dissolve in liquid under certain circumstances. Dispositions will always be identical with some quality or qualities, but may be identical with different qualities or arrangements under different circumstances (thus a vase is fragile because of the structural properties of the material that composes it, whereas a banana that has been dipped in liquid nitrogen is fragile because of the additional qualities that are added to its normal structural properties by being frozen). Alternatively, whether or not a given quality or collection of qualities is a disposition has to do with both the conditions in which those qualities are placed and the obtaining of the laws of nature. Matches normally have the disposition to be flammable, and this disposition will be identical with some of the qualities of matches. However, a match possessing the normal set of qualities that make it flammable, plus the additional quality of being wet, will no longer have the disposition. An alteration in the condition of the match can lead to the loss of its disposition. Similarly, if the match has a set of qualities that constitute the disposition of being flammable given the laws of nature as we know them, transfer of this match and its qualities to an alternative universe where the laws of nature that govern our own universe do not hold might also lead to the loss (non-existence) of this disposition in the match.[56]

The BFO categories: SPAN Occurrent

As opposed to SNAP, the SPAN portion or perspective of BFO represents occurrents: entities that happen, unfold, or develop in time. Examples of such entities include the process of respiration, a five mile run through the Black Forest, a whole human life in the 19th century, the development of an embryo, the functioning of a heart. The characteristic feature of occurrents, or processual entities, is that they are extended both in space (they occupy a definite spatial location at every time during which they exist), and also in time (understanding what a processual entity is, its identiy, requires knowing about how it has been at different times).[57]



Level 1

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56 Stephen Mumford. "Conditionals, Functional Essences and Martin on Dispositions." *The Philosophical Quarterly,* Vol. 46, No. 182. (Jan., 1996), pp. 86—92. The treatment of disposition given here is also consistent with Ludger Jansen's characterization of dispositions as a kind of realizable entity in Ludger Jansen. "The Ontology of Tendencies and Medical Information Science." In Ingvar Johansson and Bertin Klein (Eds.), *WSPI 2006: Contributions to the Third International Workshop on Philosophy and Informatics.* Saarbrücken, May 3—4, 2006. http://home.arcor.de/metaphysicus/Texte/tendencies%20WSPI2006%20proc.pdf#search=%22Ludger%20Jansen%2C%20tendencies%22; see also Brian P. McLaughlin. "Dispositions", in Jaegwon Kim & Ernest Sosa (eds.) *A Companion to Metaphysics,* Malden: Blackwell, 1995, pp. 121—24.

57 For a highly abstract discussion of the 4-Dimensionalist ontological perspective of SPAN, see Theodore Sider, *Four-Dimensionalism: An Ontology of Persistence and Time* (Oxford: Oxford University Press, 2001).

Occurrent

Df: an entity that has temporal parts and that happens, unfolds or develops in time. Sometimes also called 'perdurant' entities. (examples: the life of an organism, a surgical operation as processual context for a nosocomical infection, the spatiotemporal setting occupied by a process of cellular meiosis, the most interesting part of Van Gogh's life, the spatiotemporal region occupied by the development of a cancer tumor)

Level 2: Sub-classes of Occurrent[58]

Spatiotemporal region

Df: an occurrent entity at or in which processual entities can be located. (examples: the spatiotemporal region occupied by a human life, the spatiotemporal region occupied by the development of a cancer tumor, the spatiotemporal setting occupied by a process of cellular meiosis)

Just as the SNAP representation of substances views space as a self-subsistent container within which substances and their qualities exist, so the SPAN representation of processes views the combination of space and time together as such a container, within which processes unfold. The SPAN process ontology views the entirety of space and time, past, present and future, as existing in its entirety at every moment. Processes, then, have a beginning, a duration, and an end in this space time. An often-used analogy is to think of processes, such as John's life, the running of the Boston Marathon or the re-combination of DNA in a fern plant deep in the Amazon Jungle, as temporally extended continuums or "worms" that are stretched out in and

[58] It is worth pointing out that there is no SPAN class corresponding to SNAP dependent entity, or to its subclasses continuant entity and realizable entity. The reason for this is that BFO treats properties and qualities of processes, such as the rate of John's heartbeat or the effectiveness of a surgical procedure as entirely explainable in terms of the qualities and realizable features of continuant entities. Thus the rate of a heartbeat (apparent process quality) will be understood in terms of the state (SNAP quality or realizable) taken on by the muscles of the heart and circulatory system at each of a series of times, while the effectiveness (apparent process quality) of a surgical procedure will be understood in terms of the states (SNAP quality or realizable) taken on by the various parts and organs of a patient during the period and as a result of a particular surgery.

through the single unified eternal container that is the entirety of space-time, as God would see it were he interested in thinking about processes. As with substantial entities, so with processual entities, every process occupies some region of space-time at every moment in which it exists, and it is possible to talk about empty regions of space-time. Spatiotemporal regions are different from temporal regions. This point will be explained below under the definition of 'temporal region'.

Processual entity

Df: an occurrent entity that exists in time by occurring or happening, has temporal parts, and always depends on some SNAP entity or entities. (examples: the life of an organism, the process of meiosis, the course of a disease, the flight of a bird, the process of cell division)

The key feature of processual entities is that they have temporal as well as spatial parts. Whereas John, considered as a substance, exists along with all of his parts at every instant in which he exists at all, the processual entity we call John's life can be divided up into parts in many different ways, notably into his childhood, his adolescence, his adulthood and his old-age. These are temporal parts of John's life; a complete account of the life of John would require discussion of all of them, but they cannot all, on pain of contradiction, exist at the same instant of time. Thus the entity that is John's life is a process that is stretched out in time and has temporal as well as spatial parts (the time he spent in Seattle, the time he spent in Stockholm, etc).

To borrow an idea from Ted Sider, a contemporary philosopher, to hug all of John considered as a three dimensional SNAP object requires only that one exist at the same time as John and give him a big hug, whereas to hug the person John in the sense of the process that is "John's Life" (SPAN), one "must hold him in an interpenetrating total embrace from his birth until his death; only thus would you have access to all his past and future temporal parts."[59] John exists in his entirety at every time in which he exists at all, but John's life is only partially present at any time at which it exists, it has parts that are spread out in time, and understanding it requires understanding the existence of all of these different parts and their existence at different times.

Processual entities, such as John's life, can also have other processual entities as parts, such as the functioning of John's respiratory and circulatory systems, the (unfortunate) development of John's cancer, and the curing of John's lingering psychological disorders. Each of these processes is a part of the process that is John's life, and like this life, each of these processes also has its own extended existence in time, and hence its own temporal parts.

Importantly, unlike substances which can gain and lose parts and qualities while maintaining their identity, the identity of processes is entirely determined by the identity of their parts and of their location in space-time. This means that if two processes differ with regard to even one temporal part, or one sub-process-part, then these two processes are non-identical.

Temporal region

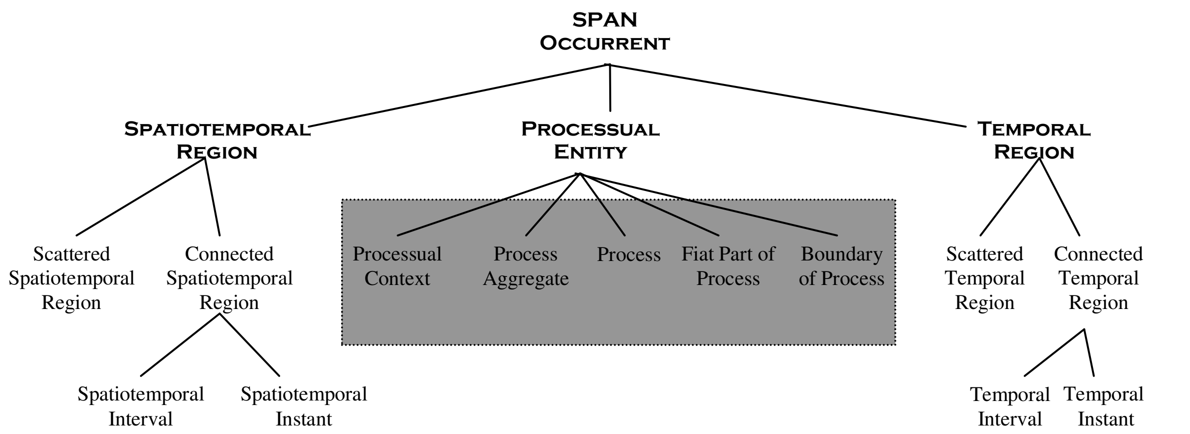
Df: an occurrent entity that is part of time. (examples: the time it takes to run a marathon, the duration of a surgical procedure, the moment of death)

The time of temporal region is the time that is common to both SNAP and SPAN perspectives in the sense that the temporal indexes associated with each SNAP view will be temporal regions, and can also be used to index or locate process parts at a particular time. The time of temporal region is different from the time of spacetime region in that it has no 'space-like' parts. The four dimensional regions of spacetime are where processual entities unfold through their successive parts, but continuant entities do not exist in this kind of space, and hence

[59] Ibid.p. 3

also not in this kind of time. Temporal region is thus a neutral kind of time that can be used to index both processual entities existing in SPAN spacetime regions and continuant entities existing in SNAP spatial regions. For most purposes of representing time in a domain ontology it is temporal region rather than spacetime region that should be used.

Level 3: Sub-classes of Processual entity



Processual context (setting)

Df: An Occurrent entity consisting of a characteristic spatial shape inhering in some arrangement of other occurrent entities. Processual contexts are characteristically entities at or in which other occurrent entities can be located or occur. (examples: a surgical operation as processual context for a nosocomical infection, a routine check-up as setting for the finding of a tumor, a clinical trial as processual context for the discovery of a new treatment or drug)

Process Aggregate

Df: A processual entity that is a mereological sum of processes and possesses non-connected boundaries. (examples: the beating of the hearts of each of seven individuals in a room, the playing of each of the members of an orchestra, a process of digestion and a process of thinking taken together)

Process

Df: a processual entity that is a maximally connected spatio-temporal whole, and has bona fide beginnings and endings corresponding to real discontinuities. (examples: the life of an organism, the process of sleeping, the process of cell-division, the functioning of the heart)

Fiat part of process

Df: a processual entity that is part of a process, but that does not have bona fide beginnings and endings corresponding to real discontinuities. (examples: chewing during a meal, the middle part of a rainstorm, the worst part of a heart-attack, the most interesting part of Van Gogh's life)

Temporal boundary of process (event)

Df: a processual entity that is the fiat or bona fide instantaneous temporal boundary of a process. (examples: the forming of a synapse, the onset of REM sleep, the detaching of a finger in an industrial accident, birth, death, the final separation of two cells at the end of cell-division, the incision at the beginning of a surgery)

<u>Level 3: Sub-Classes of Temporal region</u>

Scattered temporal region

Df: a temporal region every point of which is not mediatley or immediately connected with every other point of which. (examples: the time occupied by the individual games of the World Series, the time occupied by the individual liaisons in a romantic affair).

Connected temporal region

Df: a temporal region every point of which is mediately or immediately connected with every other point of which. (examples: the WWII years, the time from the beginning to the end of a heart attack, the time taken up by an occurrence of cellular meiosis)

Level 3: Sub-Classes of Space time region

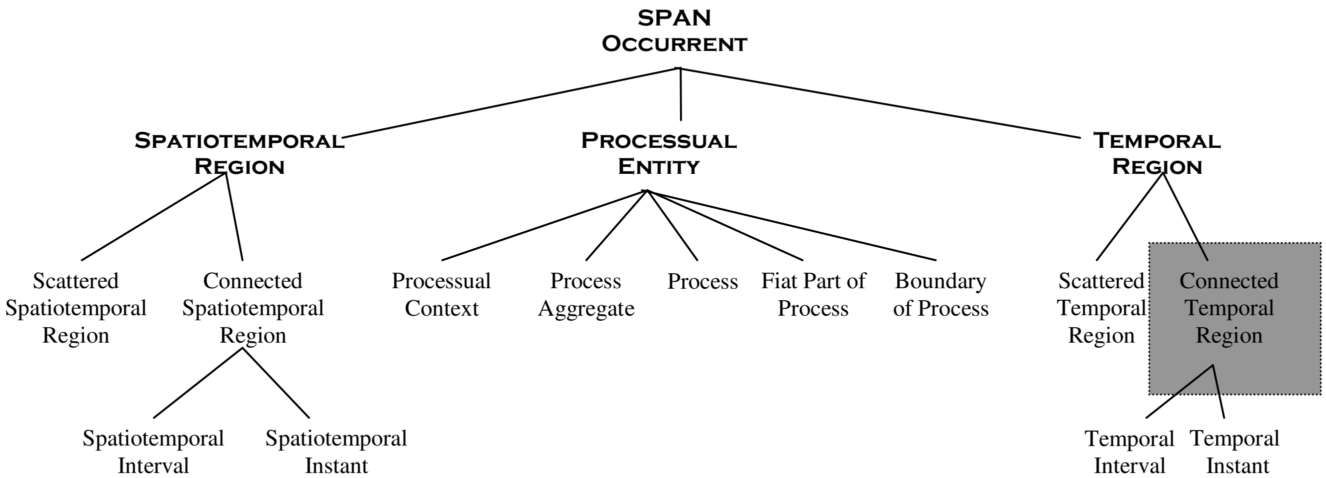
Scattered space time region

Df: a space time region that has spatial and temporal dimensions, and every spatial and temporal point of which is not connected with every other spatial and temporal point of which. (example: the space and time occupied by the individual games of the World Series, the space and time occupied by the individual liaisons in a romantic affair)

Connected space time region

Df: a space time region that has temporal and spatial dimensions such that all points within the spacetime region are mediately or immediately connected to all other points within the same space time region.. (example: the spatial and temporal location of an individual organism's life, the spatial and temporal location of the development of a fetus)

Level 4: Sub-Classes of Connected temporal region



Temporal interval

Df: a connected temporal region lasting for more than a single moment of time. (examples: any continuous temporal duration during which a process occurs)

Temporal instant

Df: a connected temporal region comprising a single moment of time. (examples: right now, the moment at which a finger is detached in an industrial accident, the moment at which a child is born, the moment of death)

Level 4: Sub-Classes of Connected space time region

Spatiotemporal interval

Df: a connected space time region that endures for more than a single moment of time. (examples: the space time region occupied by a process, or by a fiat part of a process)

Spatiotemporal instant

Df: a connected space time region at a specific moment. (examples: the space time region occupied by a single instantaneous temporal slice (part) of a process)

BFO perspectivalism revisited

At this point the SNAP SPAN perspectivalism of BFO can be more clearly stated. SNAP ontologies represent some portion of space and its continuant occupants, including parts and dependent qualities of these objects at a given specific instant of time. A SNAP ontology is thus a representation of some portion of the world of substances frozen at an instant of time (every SNAP ontology thus includes a time-index or a reference to a particular moment in time).

SPAN ontologies, by contrast, represent all or some portion of space-time and the processes or 'spatio-temporal extended worms' that occupy it. Whereas time is external to SNAP ontologies, functioning as an index for locating the ontology itself relative to other SNAP views, time is internal to and represented by SPAN ontologies, where it functions as a dimension along which objects are extended and determined. SNAP ontologies thus correspond to anatomy, the study of the static three-dimensional structures found in the body, while SPAN ontologies correspond to physiology, the study of the operation of processes in and the functional movements of the body. Once again, the time in terms of which SNAP and SPAN entities are to be located and related to one another is the time of the SPAN class temporal region.

Time and change

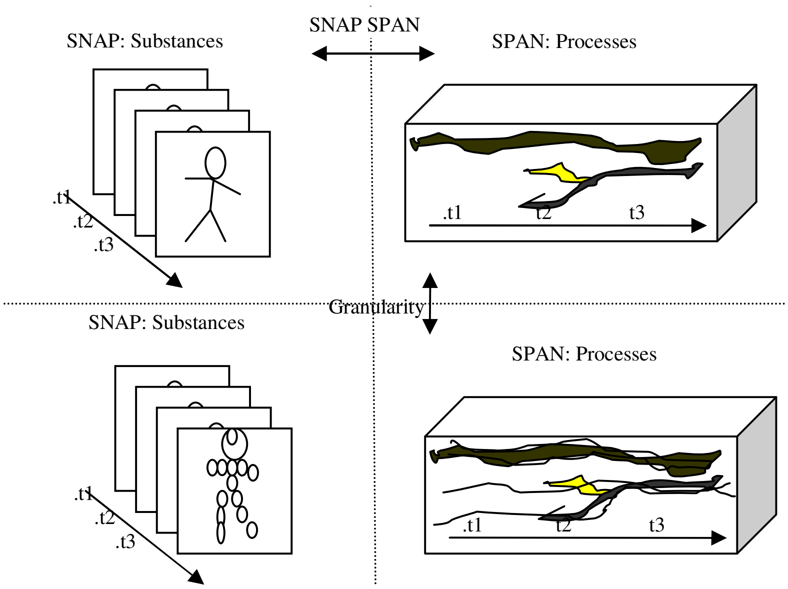
The passage of time and the occurrence of change in SNAP ontologies can be represented by lining up chronologically a series of SNAP representations of the world taken at different times, and then observing the differences and similarities possessed by objects represented in these ontologies. For instance, one might have a SNAP ontology including a representation of Jill from last year when she had blond hair, and another SNAP ontology including a representation of Jill from this year, when her hair is brown. One could then point to the difference in hair-color as a change, but nevertheless identify the substance in which this hair-color inheres, namely Jill, as identical in the two cases, due to underlying biological and/or psychological continuities represented in the two cases.

Alternatively, time and change are intrinsically represented within every single instance of a SPAN ontology in the form of morphological transformations, separations and conjoinings of the temporally extended processes (worms) that are represented. SPAN captures the flowing and continuous nature of processes by representing change in terms of a constant and connected succession of temporal parts or stages, each blending into the next. Nevertheless, it is in principle always possible to choose any two instants of time in a SPAN ontology, and to compare the process parts existing at those instants of time in order to determine whether or not they are identical, or whether or not change has occurred. Thus the transformation of Jill's hair from blond to brown can be represented and identified as a SPAN process, where the part of this process where her hair is blond, and the part where it is brown, can be identified and compared, as well as connected by intermediate stages (such as the process of dying the hair a new color, or of allowing it to return, naturally by growing, to its normal color).

Granularity and perspectives

Both SNAP and SPAN ontologies are veridical perspectives on the world. However every SNAP and every SPAN ontology will represent reality only within a certain range of granularity, one possessing both an upper and a lower limit of the size and complexity of objects represented. Thus the major SNAP and SPAN perspectives admit of determination into full perspectives or ontologies along the lines of granularity. It is only once a representational artifact has been specified both in terms of whether it is a SNAP or a SPAN representation, and in terms of the level of granularity that it represents, that it can be an ontology in the full sense of the term. The BFO ontology is thus "perspectival" along two major dimensions, as indicated in diagram 1.

Diagram 1: The Perspectives of BFO



Universals and particulars

The categories and relations recognized by the SNAP and SPAN ontologies can be used to talk about both universals and particulars. However, given our definition of an ontology as a representational artifact whose representational units are intended to represent universals and relationships amongst universals on the side of reality, it is primarily universals and the relationships amongst them that are relevant. Biological species, cells and canonical anatomical structures are all candidate universals for inclusion in the category of SNAP object universals, while universals such as redness, mass and elasticity (each of which admits of more specific determinations, e.g. bright red) are candidates for inclusion in the category of quality universals. Determining which BFO category a given universal belongs in can be carried out by appeal to the characteristic features of the BFO categories themselves.

Thus, given a universal that current science suggests actually exists on the side of reality, the first question is whether its instances are continuant SNAP entities or occurrent SPAN entities. If the universal in question has SNAP instances, then the next question is whether these instances are dependent or independent objects, and so on until the appropriate formal ontological category has been located. It should be noted that formal ontological relationships that obtain between different ontological categories will also obtain amongst instances of the universals belonging to those categories. For example, if every quality is dependent_on some SNAP object for its existence, then every instance of the quality universal red is dependent_on some instance of some universal from the category of SNAP object, and likewise in other cases. Thus placing the universals to be found in a given domain into the categories of a formal ontology itself initiates the process of determining what kinds of relationships must hold between the different universals in that domain.

The Exhaustiveness and Downward Expansion of the BFO classes

The BFO classes are defined so as to be mutually exclusive relative to a given level of granularity, though the same thing might count as a SNAP object at one level of granularity, and as an aggregate of objects at another. For example, at a level of granularity dealing with groups as its primary focus of interest, a symphony orchestra could be considered a SNAP object, while at a level of granularity focusing on individual persons, the same symphony orchestra would be considered a SNAP aggregate of objects composed of persons as SNAP objects. Similarly, a human individual might be considered a SNAP aggregate of objects (individual cells) at one level of granular focus, and a SNAP object (organism) at another. However, regardless of the level of granularity being dealt with, something must be said about the exhaustiveness of the BFO classes.

The axioms expressing the basic features of BFO do make it in principle possible, in certain cases, that BFO could recognize kinds of entities that are not explicitly identified in the BFO taxonomies or definitions that have been given here.[60] An example would be the entity kind "aggregate of two separate objects, a fiat part of a third object and the boundary of a

[60] See footnote 1 for references to papers and technical reports that discuss these axioms.

fourth". Due to the mereological principles underlying some of the axioms of BFO, it is technically possible that such entity types could be included as BFO classes.[61] Indeed, an exhaustive partition of SNAP independent continuant would have to include classes for all the different combinatorial possibilities that could be used to compose a mereological sum or aggregate. Since entities of this sort lack salience and are systematically irrelevant for a principled analysis of the ontology of scientific domains, they are intentionally not included in any existing definition sets, taxonomies or implementations of BFO. However, the fact that they are available in the axiomatization of BFO means that the taxonomy that has been provided here is not in every case strictly exhaustive.

In spite of this lack of literal exhaustiveness, the top-level BFO classes (and for that matter, the top-level classes of any formal ontology) should be implemented and used *as if* they represent an exhaustive partition of their domain, this includes using "union of" and "disjoint with" statements in OWL implementations. The reason for this is that the purpose of a top level ontology is to provide a stable framework within which to structure and organize domain-specific information. If the top-level ontology is not treated as exhaustive of its domain, then it will not be genuinely functioning as a top-level ontology. Further, if it is left open to the end-users of a top-level ontology to add new top-level classes willy-nilly, then one of the very difficulties that top-level ontologies are meant to address, the problem of ensuring interoperability, will quickly re-assert itself.

Consistent with the principle of *falliblism*, we acknowledge that it is possible that future research in ontology and in the natural sciences, as well as continued attempts at specific domain implementations, will reveal the need for an expansion or addition of classes under some of the

[61] For discussions of axiomatic mereology or "part-whole" theory, see Varzi, A. 'Mereology', in Edward N. Zalta (ed.), *Stanford Encyclopedia of Philosophy*, Stanford: CSLI (internet publication), 2003. ftp, and Simons, P. *Parts: A Study in Ontology*. Clarendon: Oxford University Press, 1987.

top-level classes of BFO (such as SNAP Independent Continuant or SPAN Processual Entity). Our position, however, is that alterations to the top-level ontology should not be handled by allowing end-users the ability to willy-nilly make aleterations, but rather by a careful scientific review process involving collaboration between end-users and ontology developers that will introduce such changes, if at all, only in new and updated versions of the top-level ontology, versions that are complete with documentation providing detailed evidence and principled reasons for why the addition (or for that matter deletion) of a class has occurred.

The primary direction for expansion of BFO, first by providing further formal ontological classes, and then moving on to classes of a more domain specific nature (geography, medicine, etc.), is thus downward. The question of which classes are the most ontologically and scientifically correct to be used in a downward expansion of BFO is an open one, and one that the developers of BFO, as well as domain users of BFO (or any other top-level ontology) will have to address. Discussion of some possible ways in which the sub-classes of SNAP realizable entity could be extended are discussed in the appendix to this chapter.

BFO relations

As has already been noted and discussed in earlier sections, specification of universals and ontological categories alone is not enough to adequately capture all of the important scientific information about a given domain. Rather, the relationships that obtain amongst the universals and categories in an ontology also need to be represented. There are two dimensions along which relationships can be discussed in BFO.

BFO perspectivalism and relations

First, the perspectival nature of the BFO ontology leads to the possibility of discussing three distinct kinds of relationships: intra-ontological relationships, trans-ontological relationships and meta-ontological relationships.

In the context of BFO an intra-ontological relationship is one that obtains between entities represented within a single SNAP or SPAN ontological perspective. For example, the relationship part_of that obtains between a hand (substantial entity) and the body of which it is a part (substantial entity) within SNAP ontology.

Trans-ontological relationships are relationships that obtain between entities that are represented in different ontologies. The most perspicuous such relation recognized by BFO is the participates_in relationship that obtains between a substance from some SNAP ontology, and a process in some SPAN ontology. The underlying idea behind this relation (and other similar ones) is that processes do not exist without substances that participate in them (imagine the functioning of John's respiratory system (process) without the existence of John (substance)) and on which they depend.

Finally, meta-ontological relationships are relationships that hold between two distinct ontological perspectives, or between an object within an ontological perspective and an ontological perspective itself. An example of the first meta-ontological relation would be the relationship earlier_than that obtains between two SNAP ontologies, one of which represents entities at an earlier point in time than the other. An example of the second kind of meta-ontological relation would be the relation has_constituent obtaining between either a SNAP or SPAN ontology and the actual objects that they represent.[62] In what follows we will focus

[62] For more on the different kind of relationships that can be analyzed within BFO see Barry Smith and Pierre Grenon. "The Cornucopia of Formal Ontological relations." Dialectica Vol. 58, 3, (2004), pp. 279—296.

primarily on intra- and trans-ontological relations, as these are more immediately crucial for domain-specific ontology design in general.

Relations for BFO and everyone

The second dimension along which relationships can be discussed is in terms of whether they are relationships that hold between two universals, relationships that hold between a universal and a particular, and relationships that hold between two particulars. These distinctions were already touched on earlier, and they will form the framework for our discussion of ontological relations here. The primary structure of the discussion of ontological relations that follows is taken more or less directly from the multi-authored OBO relations paper.[63]

To avoid systematic confusion

Relations are a crucial part of any ontology at any level. Thus it is absolutely crucial that relations be strictly defined, and that their definitions be strictly adhered to when entering domain specific information into an ontology. To put it simply, at every point in developing an ontology, from its initial conception through its construction to its use, 'is_a' should mean "is_a" and 'pat_of' should mean "part_of", and *nothing else*.

Allowing deviation in the interpretation of relationships obtaining amongst ontological categories, universals and/or classes amounts to allowing systematic ambiguities into one's ontology, ambiguities that will confound automated reasoning and consistency checkers, while

[63] Barry Smith, Werner Ceusters, Bert Klagges, Jacob Kohler, Anand Kumar, Jane Lomax, Chris Mungall, Fabian Neuhaus, Alan L. Rector, and Cornelius Rosse. "Barry Smith et al., "Relations in Biomedical Ontologies," review of Reviewed Item, *Genome Biology* 6, no. 6:R46 (2005).Relations in Biomedical Ontologies." *Genome Biology* 6, no. 6:R46 (2005). More on-line information about "Relations in Biomedical Ontologies." *http://obo.sourceforge.net/relationship/*. For still more on relations, see Brochhausen Mathias: "The Derives_From Relation in Biomedical Ontologies", in: (MIE 2006) Studies in Health Technology and Informatics, 124 (2006), 769-774; Stefan Schulz, Anand Kumar, Thomas Bittner: Biomedical ontologies: What *part-of* is and isn't. Journal of Biomedical Informatics 39(3): 350-361 (2006), Schulz Stefan, Daumke Philipp, Smith Barry, Hahn Udo: "How to Distinguish Parthood from Location in Bioontologies", in: Proceedings of AMIA Symposium 2005, Washington D.C., 2005, 669-673, Bittner Thomas, Donnelly Maureen, Smith Barry: "Individuals, Universals, Collections: On the Foundational Relations of Ontology", in Varzi Achille C., Vieu Laure (eds.): Formal Ontology and Information Systems. Proceedings of the Third International Conference (FOIS 2004), IOS Press, Amsterdam, 2004, 37-48.

simultaneously making human interpretation of the information represented in the ontology a difficult and subjective affair.[64]

Primitive and defined

No ontology can explicitly define all of its categories, universals and relations. To attempt to do so would involve an infinite regress. For example, the difference between independent and dependent entities (substances and qualities) can be spelled out in terms of their being entities of certain sorts possessing certain kinds of identity conditions. However, these definitions then themselves presuppose things, such as the notion of an "entity" and the notion of "identity". It is, however, hard to imagine what a good definition of 'entity' in general or 'identity' in general might look like, and even if there were such a definition, it would necessarily make reference to new entities or relations that themselves lack definitions. Therefore, unless we wish to continue providing definitions forever, it is necessary at some point to accept certain relations and categories as primitive and not further definable. Ideally these should be categories and relations that have a certain amount of intuitive plausibility and understandability, such as "entity", "part_of" and "instance_of".

Once the primitive or undefined categories and relations of an ontology have been decided on, it is possible to appeal to these in the process of defining other relationships and categories.

Relations of interest and their definitions

For purposes of ontology construction the primary relationships we are interested in are those that obtain between universals or classes, including but not limited to the is_a or "class-

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[64] For a discussion of the ambiguity of *part_of* and *is_a* in the Gene Ontology, see Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

subsumption” relation. However, it is important to address and define relationships that obtain between particulars and also relationships that obtain between universals and particulars, both because these may sometimes be important to represent in an ontology, and also because definitions of such relations can themselves help to play a role in defining the explicit relations that do obtain between universals or classes.

The kinds of things relations relate

When adding a relationship to any ontology, whether formal or domain specific, it is crucial that it be clear what kinds of entities these relationships hold amongst. Minimally, it should be specified whether the relationship is of the universal-universal, universal-particular or particular-particular form. However, given the presence of a formal ontology such as BFO, it should also be possible to specify what categories of objects a given relationship holds amongst. For example, the relationship “instantiates” always holds between a particular and a universal, as in Fido instantiates Labrador Retriever. The relationship “part_of” on the other hand, can hold either between two particulars or between two universals, but not between a universal and a particular. Fido is not a part_of the universal Labrador Retriever. However, Fido’s tail is “part_of” Fido (particular-particular), while canine pre-molar part_of dog (universal-universal).

Representing a relationship as obtaining between kinds of entities that it was not intended to obtain between can amount to changing the meaning of that relationship and can introduce systematic ambiguity into one’s ontology. For example, to say that dog part_of mammal (universal-universal) is to identify an is_a relationship between universals as a part_of relationship. This can only lead to confusion because, as will become clear below, the part_of and is_a relations, even when both obtain amongst classes or universals, have fundamentally different meanings or semantics. Similar problems arise for the already discussed Fido part_of

Labrador Retriever (particular-universal) where the part_of relation is mistaken for the relation "instantiates".

Some conventions

We follow the authors of the OBO relations paper in adopting the following conventions for representing relations. The upper-case variables C, C^1…Cn will be used to represent SNAP continuant classes or universals, while lower-case variables c, c^1…cn will be used to represent particular SNAP continuants. Similarly, upper-case variables P, P^1…Pn will be used to represent SPAN occurrent classes and universals, while lower-case variables p, p^1…pn will be used to represent particular SPAN occurrents.

We also adopt the following convention for representing relations. A relation that holds between two classes or universals will be represented in *italics*, as in C^1 *is_a* C^2. A relation holding between an instance and a class will be represented in boldface type, as in c^1 **instance_of** C^1. A relation obtaining between two particulars will also be written in bold type, as in c^1 **part_of** c^2. We thus have the following conventions for the three kinds of relations that have been discussed so far-

i. <class/universal, class/universal> P *is_a* P^1

ii. <instance, class/universal> c^1 **instance_of** C^1

iii. <instance, instance> c^1 **part_of** c^2

These conventions make it possible to distinguish the different basic kinds of relations, and to determine (based on the variables P, C, p, c located on either side) the basic ontological category that the entities being related belong to (SNAP or SPAN, continuants or occurrents), and whether the entities involved are particulars (c, p) or universals (C, P). Consider the case of Fido and the relationship "part_of" discussed in the previous section. Fido's tail part_of Fido would be represented using these conventions as c^1 **part_of** c^2. Whereas a relationship between universals, as in canine premolar part_of dog, will be represented as C^1 *part_of* C^2. Notice too that saying that the part_of relationship does not obtain between particulars and universals amounts to saying that all of the following statements, given our conventions, are nonsensical-

i) C^1 **part_of** c^1 (the universal Labrador Retriever is part of Fido),

ii) c^1 **part_of** C^2 (Fido is part of the universal Labrador Retriever),

iii) P^1 **part_of** p^1 (the universal "dog life" is part of Fido's life),

iv) p^1 **part_of** P^1 (Fido's life is part of the universal "dog life").

While somewhat technical, adopting conventions such as these, and abiding by them in a rigorous fashion amounts to a first major step in the direction of constructing coherent ontologies that will support various kinds of automated retrieval and reasoning.[65]

The formal properties of relations

There are a number of relatively straightforward and well-understood properties of relationships, including reflexivity, symmetry, transitivity, and their various failures and negations, *that should always be taken into account when defining relations in any ontology*.

To say that a relationship R is reflexive is to say that any thing T that bears the relation R to something else, E, also bears that relationship to itself. The relationship "is as tall as" is reflexive, because when John is as tall as Jill, he also stands in this same relationship to himself, "John is as tall as John".

[65] For more on the conventions here discussed see Barry Smith, Werner Ceusters, Bert Klagges, Jacob Kohler, Anand Kumar, Jane Lomax, Chris Mungall, Fabian Neuhaus, Alan L. Rector, and Cornelius Rosse. "Smith et al., "Relations in Biomedical Ontologies," review of Reviewed Item, no.Relations in Biomedical Ontologies" *Genome Biology* 6, no. 6:R46 (2005); for more on properties of relations, see Tidman, P. & Kahane, H. *Logic and Philosophy: A Modern Introduction*. New York: Wadsworth Publishing Co, 1999, or any similar introduction to logic textbook with a chapter on the properties of relations.

To say that a relationship R is symmetric is to say that if a thing T bears the relationship R to something else, E, then E also bears the relationship R to T. The relationship "is next to" is symmetric, because of John is next to Mary, then necessarily Mary is also next to John. The relationship "is next to" will always hold in both directions between any two things with regard to which it holds at all, and thus is symmetric.

Finally, to say that a relationship R is transitive is to say that if a thing T bears the relationship R to another thing E, and E bears the same relationship R to some third thing H, then T also bears the relation R to H. A simple example of a transitive relation is "is taller than". If John is taller than Mary, and Mary is taller than Steve, then John is taller than Steve.

Any relationship can be defined as possessing or not possessing each of these properties in an explicit fashion, *and this should be done*, *because the basic logical properties of a relationship go a long ways towards establishing what its correct meaning or interpretation is*. For example, the "part_of" relationship obtaining between continuant particulars, as in c^1 **part_of** c^2 comes with a certain intuitive interpretation. Whatever its meaning is, we know that it must be understood in such a way that Fido's tail **part_of** Fido, Texas **part_of** the United States, and these water molecules **part_of** this cloud are all true. However, when we add that the **part_of** relation is reflexive (everything is trivially part of itself), transitive (if c^1 **part_of** c^2 and $\mathbf{c^2}$ **part_of** c3, then c^1 **part_of** c3) and antisymmetric (if c^1 **part_of** c^2 and c^2 **part_of** c^1, then $c^1=c^2$), then we have said something definite and concrete about the nature of this relationship, and we have provided a way of systematically distinguishing it from all other relationships. Further, once this has been done we know that we should never represent a relationship as a "part_of" relation unless it has at least these basic logical properties, and we also know how to begin 'explaining' to a computer how to reason with this relation. All of this comes from explicitly determining the formal logical properties of a given ontological relationship, and consistently abiding by the specified interpretation at all times.

Summary and next steps

In the preceding we have stressed the importance of rigorously defining and consistently applying relations in a formal ontology. This includes explicitly identifying whether the relation holds between universals, particulars or universals and particulars, keeping track of the kinds of entities that the relationship holds amongst (continuants, processes, qualities, etc), adopting clear conventions for demarcating these features of different relationships, and clearly defining the logical properties of relationships, such as reflexivity, symmetry, and transitivity.

In what follows we will, again following the work in the OBO relations paper, introduce some primitive relationships and then use these to define others. By the end of this process we will have a set of clearly defined relationships amongst universals that can be used for purposes of ontology construction, and we will show how these relationships line up with the formal ontological categories of BFO (SNAP object, quality, spatial region, process, spatio-temporal region) that have already been discussed. In this process a number of principles for best ontology practices will be introduced and discussed, and the relationship between categories and relations in a formal ontology should become relatively clear.

Preliminaries to the primitive relationships

In introducing the following relationships we will add two more conventions for representing entities to the list that was begun earlier. First, we will use variables r, r^1…rn to represent three-dimensional space regions, and second we will use variables t, t^1…tn to represent instants of time.

For our purposes here we will also use the upper-case variables C and P to refer, indiscriminately, to both classes and universals, though this practice should be avoided in the design of actual domain ontologies.

It should also be noted that relationships involving reference to continuant particulars will always include a time-index. This is included to do justice to the fact that continuants always exist in total at a particular time, and that they may alter or change the relationships that they have across time while continuing to exist. Additionally, this time indexing of particular continuant relations is consistent with the time index that is attached to SNAP ontologies.

It must also be noted at the outset that separate relationships will be defined for the purposes of relating entities of different kinds, even if the semantics of the relationships themselves are the same. Thus we will introduce a **part_of** relation holding specifically between particular continuants (SNAP entities), and a separate **part_of** relation obtaining between particular occurrents (SPAN entities). The reason that these two relations must be defined separately is that, even though **part_of** itself will have the same logical properties (reflexive, transitive, antisymmetric) in the two cases, it is nevertheless false to say of any substance/continuant that it is **part_of** some occurrent/process, and vice versa. This is because the sense in which John's hand **part_of** John is very different from the sense in which one might try to say that John is part of his life. And since every relationship should have one well-defined meaning, the more basic sense of part (hand **part_of** John) is here opted for. Thus, while almost everything can be legitimately divided up into parts, it is rarely correct to identify part relations as obtaining across ontological categories (between substances and spatial regions, or between processes and substances), and thus it is necessary to define multiple relationships, relationships that apply to the **part_of** relations that apply within ontological categories (of continuants c and processes p), rather than just one relationship whose intended interpretation is that it hold both within and across all such categories.

Some primitive relations of interest

Our primitive relations and their definitions are then the following:

c **instance_of** *C* **at** *t*

We take this as a primitive relation obtaining between a continuant instance and a class which it instantiates at a specific time. For example: Fido **instance_of** Labrador Retriever **at** the present.

p **instance_of** *P*

We take this as a primitive relation obtaining between a process instance and a class which it instantiates, one holding independently of time. For example: John's Life **instance_of** Human Life. Assuming that John has existed at any time at all, this statement will be true regardless of when it is uttered.

c **part_of** c^1 **at** t

We take this as a primitive relation obtaining between two continuant instances and a time at which the one is part of the other. For example: this cell nucleus **part_of** this cell **at** the present.

p **part_of** p^1

We take this as a primitive relation of parthood, holding independently of time between process instances (one a subprocess of the other). For example: this tumor's growth **part_of** Mary's Life

r **part_of** r^1

We take this as a primitive relation of parthood, holding independently of time, between spatial regions (one a subregion of the other). For example: the spatial region occupied by Central Park **part_of** the spatial region occupied by New York City.

c^1 **inheres in** c^2 **at** t

We take this as a primitive relation obtaining specifically between a dependent continuant and an independent continuant at a particular time. For example: the shape of John's body **inheres_in** John **at** July 26, 2006.

c **located_in** *r* **at** *t*

We take this as a primitive relation between a continuant instance, a spatial region which it occupies, and a time. For example: John **located_in** the dinning room **at** dinner time.

p **has_participant** *c* **at** *t* –

We take this as a primitive relation between a process, a continuant, and a time. For example: John's life **has_participant** John **at** January [1], 1984.

<u>Comments on the primitive relations</u>

The primitive relations here introduced, and primitive relations in general, have a number of features worth commenting on.

First, it should be noted that the relationships introduced so far are of the particular-particular and particular-universal form only; none of them satisfy the universal-universal format required for representation in scientific domain ontologies. The reason for this, which will become clear below, is that it has proven easier to define universal-universal relations in terms of previously accepted primitive relations of the particular-particular and universal-particular sort.

Second, the formal properties of these relationships (reflexivity, symmetry, transitivity, etc.) have not yet been specified. All that has been done so far is an introduction of the relationship, a specification of what kinds of entities it holds amongst, and an intuitive characterization of it in terms of examples.

The logical properties of primitive relationships are usually specified via the adoption of certain axioms or postulates in one's theory of relations. For example, having accepted instance-level continuant part hood (c^1 **part_of** c^2 at t), it is possible to define its logical properties by explicitly adopting axioms that assign them (indeed, this is usually what is done). Our purpose here is only to give an outline of what a theory of relations for purposes of ontology construction looks like, not to develop a full axiomatic theory, and so we will refrain from discussing axioms for relations except when such discussion is crucial for clarifying the matters at hand.[66]

Some defined relationships

Defined relationships that are of interest to us here include the following:

C is_a C^1

is_a understood as a relationship between classes or universals is the crucial relationship used in ontologies for the purpose of representing the hierarchical or taxonomic structure obtaining amongst universals of greater and lesser generality. A typical example of an *is_a* relation obtaining between universals would be eukaryotic cell *is_a* cell.

We follow the OBO relations paper in defining the *is_a* relations amongst classes in terms of the primitive relations already accepted that obtain amongst particulars and particulars and universals. Here in particular we make use of the already defined **instance_of** relation. This procedure of relying on primitive relations between particulars and between particulars and universals in defining relationships that obtain between classes or universals will be used throughout the following section, and has some important implications for the construction of domain specific ontologies in terms of formal relations that we will discuss in the next section.

We define the relationship *is_a* for continuants in the following way. Continuant class C *is_a* continuant class C^1 just in case any particular continuant c that is an **instance_of** C at a given time t, is also an **instance_of** C^1 at the same time t. Intuitively, this is to say that the class

[66] For some suggested axioms, see Barry Smith, Werner Ceusters, Bert Klagges, Jacob Kohler, Anand Kumar, Jane Lomax, Chris Mungall, Fabian Neuhaus, Alan L. Rector, and Cornelius Rosse. Relations in Biomedical Ontologies" *Genome Biology* 6, no. 6:R46 (2005).

Canine *is_a* Mammal just in case at any time any thing (c) exists and is an **instance_of** Canine, that thing is also an **instance_of** Mammal at that time. To say Canine *is_a* Mammal is just to say that all instances of Canine are also instances of Mammal. A similar definition of *is_a* can be given for process classes.

P *is_a* P^1

Here we say that a process class P *is_a* process class P^1 just in case every particular process p that is an **instance_of** P is also an **instance_of** P^1, regardless of time. We write 'regardless of time' because of the fact that processes have all of their parts and properties essentially, regardless of the time at which they are instantiated by particulars. Thus, for any given process particular, it either is or is not an **instance_of** a process class, regardless of time. Thus Lung Cancer Development *is_a* Cancer Development just in case every **instance_of** Lung Cancer Development is also an **instance_of** Cancer Development.

The next crucial class-level relationship that we will define in terms of our already accepted primitive relations is the class level *part_of* relation. As with *is_a* we will define versions of *part_of* that apply to both continuant classes and process classes.

C part_of C^1

A class C *part_of* a class C^1 just in case for every instance c of C at some time t there is some instance c^1 of the class C^1 such that c **part_of** c^1 **at** t. Thus to say that Human Heart *part_of* Human Circulatory System is to say that for every particular human heart instance, there is some time in which some instance of Human Circulatory System exists, and that particular heart is **part_of** that instance of Human Circulatory System at that time. Notice that this definition requires that in order for one class to be a *part_of* another *every* single **instance_of** the first class must at some time be **part_of** some **instance_of** the other, but this does not require that every instance of the first class always be **part_of** some instance of the other, nor does it require that the **instance_of** the first class be **part_of** any particular **instance_of** the other. Thus the fact that some particular hearts are currently not **part_of** any circulatory system (for example, those hearts that are sitting in jars of formaldehyde in laboratories), and the fact that some particular hearts have been **part_of** two different circulatory systems (as in a heart transplant) are both consistent with the class-level definition of *part_of* that has been provided here. A similar definition, once again, is available for Process classes.

P *part_of* P^1

A process class P is *part_of* a process class P^1 just in case every particular process p **instance_of** P is **part_of** some particular process p^1 **instance_of** P^1. Thus Third Trimester *part_of* Human Fetal Development just in case every particular third trimester is part of some particular human fetal development. Note here that it is consistent with this definition, for example, that not every Human Fetal Development has a Third Trimester as part (since some fetal developments end prior to the third trimester). All that is important is that the relationship hold in the direction stated, that is Third Trimester *part_of* Human Fetal Development.

The next two class-level defined relationships we will introduce, '*inheres_in*' and '*located_in*', will be defined in such a way that they apply to continuant or SNAP entities only. In the case of *inheres_in* this is necessary because this is a relationship obtaining only between specific kinds of substantial entities, namely between dependent and independent entities (our examples here are qualities and substances) respectively. In the case of *located_in* we focus on continuants only for convenience. It is also possible, and in a full development of the BFO ontology and associated required relations would be desirable, to define a *located_in* relationship for processual or SPAN entities.

C *inheres_in* C^1

The class C *inheres_in* the class C1 just in case for every particular continuant c **instance_of** C, at all times t that c exists there exists some particular continuant c^1 **instance_of** C^1 such that c **inheres_in** c^1 **at** t, and at any two times at which the same instance c of C exists, that instance inheres in the same instance c^1 of C^1 at those times. The *inheres_in* relation is importantly different from other relations that we have defined so far insofar as its intended interpretation is that of a kind of dependence relation obtaining specifically between dependent and independent objects. In terms of the examplary ontological categories that we have discussed so far, this relationship is conceived of as obtaining between dependent universals and SNAP independent universals specifically. So, for example, every quality (such as the quality of red/a certain redness) depends on or *inheres_in* some object (such as an apple, a jacket or a red squirrel) at every time at which that instance of that quality exists at all, and it inheres in the same object at all times at which it exists. An example more specific to our definition would be Rational *inheres_in* Human Being. For this relational statement to be true every instance of rationality must **inhere_in** the same particular human being at every time at which that instance of rationality exists at all.

C *located_in* C^1

Defining the class-level relationship *located_in* comes, following the OBO relations paper, in two steps. First we will use the instance level primitive relation *c* **located_in** *r* **at** *t* (obtaining between a continuant instance, a spatial region and a time) to define an instance level relation *c* **located_in** c^1 at *t* (obtaining between two continuant particulars and a time), and then use this newly defined relation to define the class level relationship C *located_in* C^1.

c **located_in** c^1 at *t*

One particular continuant c is **located_in** another particular continuant c^1 **at** a time t just in case there are two spatial regions r and r^1 such that the particular continuant c is **located_in** r at t and the particular continuant c^1 is **located_in** r^1 **at** t, and the region r is **part_of** the region r^1 **at** t. Intuitively, a kidney is **located_in** a torso at a time just in case the kidney is **located_in** a specific spatial region r **at** that time, and the torso is **located_in** a specific spatial region r^1, and the first spatial region r is **part_of** the second spatial region r^1 **at** that time.

Given this definition we can define the *located_in* relation for classes as follows-

C *located_in* C^1

A continuant class *C* is *located_in* a continuant class C^1 just in case for all particular continuants *c* **instance_of** C at some time t, there is some particular continuant c^1 **instance_of** C^1 at t such that *c* **located_in** c^1 **at** *t*. Thus Kidney *located_in* Torso means that for every instance of Kidney there is some time t such that an instance of Torso exists at t and the instance of Kidney is **located_in** the instance of Torso at that time. The basic features of the *located_in* relation are thus similar to those of the *part_of* relation, though these two still have importantly different meanings.

The final relation we, following the OBO relations paper, will define here is different from the relationships that have gone before insofar as it is what we have earlier called a trans-ontological relation: from the perspective of BFO this relation relates entities that are to be found in different ontologies or ontological perspectives, namely occurrents and continuants. There are arguably many such relationships: however the one that we will focus on here is the *has_participant* relationship obtaining between process classes and continuant classes. The idea behind this relationship is that because no process can exist without some substance that it is a process of or relative to, every process has some substantial continuant that participates in it.

Thus John participates in his life, his running and his respiratory processes, while a fetus participates in its development through the trimesters of pregnancy.

P has_participant C

A process class P *has_participant* a continuant class C just in case for every particular process p **instance_of** P there is some continuant c **instance_of** C and some time t at which c exists such that the process particular p **has participant** the continuant c **at** t. For example, Third Trimester *has_participant* Fetus because every instance of the process Third Trimester is such that there is a continuant particular, namely a fetus, and a time at which that fetus exists, such that at that time that third trimester instance **has_participant** that particular fetus at that time.

Summary and comments

In the preceding we followed the conventions, established in the OBO relations paper, of using primitive relations between particulars and between particulars and universals in order to define class-level relationships between universals of various sorts. We have specifically focused on the relations *is_a, part_of, inheres_in, located_in* and *has_participant* both because these relations are relatively common in existing ontologies, and because they bear a direct relevance to the BFO ontology that is here being presented. There are two important points that should be made now regarding the class/universal level relations here defined.

All-some structure

Relationships defined as obtaining between classes or universals *must* adhere to the rule of possessing all-some structure. If a class C bears some *relationship* to a class C^1, then all relevant instances of c must bear the relevant instance level relationship to some instance of C^1 at all relevant times (where the specifics of what 'relevant' means here will be determined by the definition of the relationship in question). This point can be captured simply by saying that relationships obtaining amongst universals and classes should not admit of exceptions, if they do then these relationships don't actually hold.

Consider the class-level definition of *part_of*, A class C *part_of* a class C^1 just in case for every instance of c at some time t there is some instance c^1 of the class C^1 such that c **part_of** c^1 **at** t. What this basically says is that for one class to be *part_of* another all instances of that class must at some time be **part_of** some instance of the other class. An example already given above of a case where the *part_of* relation holds is the following: to say that Human Heart *part_of* Human Circulatory System is to say that for every particular human heart instance, there is some time in which some instance of Human Circulatory System exists, and that particular heart is **part_of** that instance of Human Circulatory system at that time. However, consider the following inference cancerous tumor **part_of** John, therefore Cancerous Tumor *part_of* Human Being. While the first assertion is, or at least could be, a true instance-level assertion about a particular tumor and the individual it is a part of, the second assertion fails to conform to the all-some structure, because there are some instances of Cancerous Tumor that are not **part_of** any human being whatsoever; for example, cancerous tumors found in non-human animals.

The *is_a* relation has similar features. We have said continuant class C *is_a* continuant class C^1 just in case any particular continuant c that is an **instance_of** C at a given time t, is also an **instance_of** C^1 at the same time t. In other words, all instances of C must also be instances of C^1. Thus Prokaryotic Cell *is_a* Cell is in perfect order as it stands, while Cancer *is_a* Terminal Disease fails to respect the all-some structure, insofar as not all instances of cancer are terminal.

The rule to be learned for the construction of domain ontologies is that identifying relationships between domain classes and universals should always be carried out in accordance with the all-some structure laid down by the formal or top-level relationship in question, such as *part_of, is_a,* etc.

Inverse and reciprocal relations

For any given ontological relationship that has been defined in an ontology, it is possible to ask about whether or not there is another relationship that, as it were, goes in the other direction. Answering this question definitively, and including an explicit definition of the answer (in the form of a new relation) within an ontology, can often be very helpful in providing more rigorous structure to an ontology as a whole (examples will be given below). The "directionality" of a relationship is in large part determined by the logical properties (symmetry, reflexivity, etc.) of that relationship. Without going into too much detail on these matters here, we will, following the OBO relations paper, take the time to discuss two general sorts of relationships that "go in the other direction". The first are "inverse" relations, while the second are "reciprocal" relations.

The "inverse" of a relation R is defined as that relation which obtains between each pair of relata of R when taken in reverse order. That is to say, if C *is_a* C^1, what is the relationship between C^1 and C going, as it were, in the other direction. In the case of inverse relations this new relationship can usually be defined straightforwardly by appeal to the definition of the original relationship in question. Thus, the intuitive inverse of the *is_a* relation can be defined directly as the *has_subclass* relation, by saying that if C^1 *has_subclass* C, then C *is_a* C^1. For example, if Prokaryotic Cell *is_a* Cell, then Cell *has_subclass* Prokaryotic Cell. However, in many cases, in fact for most of the relations between classes that we have defined above, it is not possible to directly define an inverse relation. This makes it necessary to define what have been called "reciprocal relations".

A “reciprocal” relation might be characterized as “the relation that ‘goes in the other direction’ relative to a given relation such as *part_of* when no straightforward inverse relation can be defined”. An example used in the OBO paper is the following. While Human Testis *part_of* Human is a true biological relational assertion conforming to the “all-some” structure, the intuitive inverse relation *has_part* cannot simply be defined directly in terms of *part_of.* This would amount to saying that just as Human Testis *part_of* Human, so Human *has_part* Human Testis. However, the second statement fails to conform to the all-some structure required for definitions of relationships amongst classes, insofar as not all humans have some part that is a testis (in fact roughly half of all human beings, the females, do not have such parts). Thus a slightly more complicated relation has to be defined, which has been called, following GALEN, a “reciprocal” relation. Thus *has_part* is not the inverse but the reciprocal of *part_of,* and it is defined in the following way- C *has_part* C^1 just in case for every instance of c that exists at a time t there is some instance c^1 of C^1 that also exists at t such that c^1 **part_of** c **at** t.

The benefit of paying attention to and explicitly defining inverse and reciprocal relations in an ontology is as follows. In the case of *is_a* and *has_subclass*, explicitly representing both of these relationships as relationships in an ontology will make it possible to reason both “up” and “down” the hierarchies of universals. For a given universal class, say Homo Sapiens, it will thus be possible both to query regarding all of the classes to which Homo Sapiens stands in the *is_a* relation, and also all of those classes that Homo Sapiens stands in the *has_subclass* relation to, thus making it possible to explore in a systematic fashion the universals that are explicitly related to the universal Homo Sapiens. Similarly, one could use the explicitly defined *part_of* and *has_part* relations with regard to a universal such as Human Hand both to explore what other entities Hand is characteristically *part_of,* and also to explore what the characteristic parts of the human hand themselves are (via the *has_part* relation).

<u>Putting it all together</u>

At this point we have introduced the BFO categories and the class/universal level formal relations *is_a, part_of, inheres_in, located_in* and *has_participant.* What is crucial to recognize now is the way in which formal categories and formal relations fit together and complement one another.

<u>Every relation specified is connected with specific categories of entities</u>

Each of the relations that we have discussed above can be associated with specific categories from BFO. The relations in an ontology thus help to exhibit, at the most general level, the way in which the different kinds of entities recognized by an ontology relate to one another and are held together.

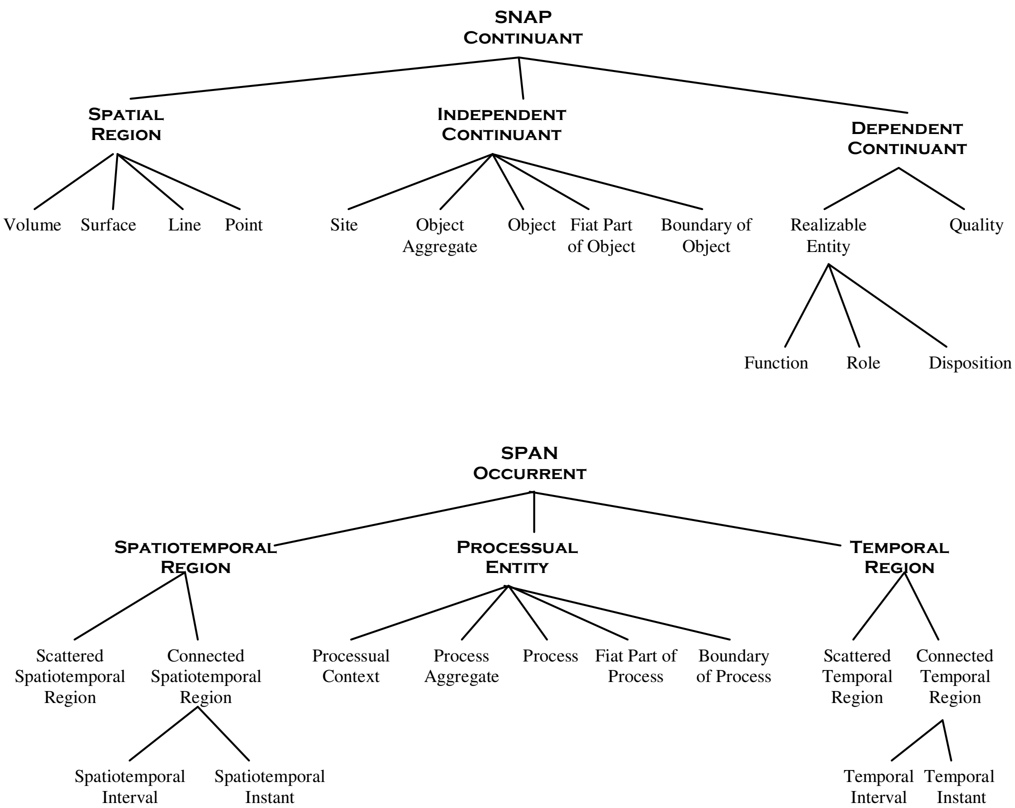
<u>The relations can be used to help define categories of entities</u>

As has already been noted, well defined formal relations can be used to help more precisely define the nature of the categories that they relate. For example, a universal can be defined, in part, as an entity that is not an **instance_of** anything, while a particular can be defined, in part, as an entity that is an **instance_of** other entities, but does not itself have any entities standing in the **instance_of** relation to it. Similarly, a substance can be partially defined as an entity to which other entities stand in the relation of **inhere_in_at**, but which does not itself **inhere_in** any other entity **at** any time. Such definitional support provides structure to the formal ontology, and can help to guide the decision of which domain-specific universals to identify as falling within different ontological categories.

<u>The three kinds of relation</u>

The explicit inclusion of the three kinds of relations, universal-universal, universal-particular and particular-particular makes it possible that a domain specific reference ontology might be used, in conjunction with well formulated information about specific particulars in the world, to perform reasoning about real world entities. The paradigm case of this would be a software tool that could connect particularized electronic healthcare records to domain-specific ontologies of biological and medical information in such a way as to facilitate diagnosis, treatment and research in the biomedical sciences.[67]

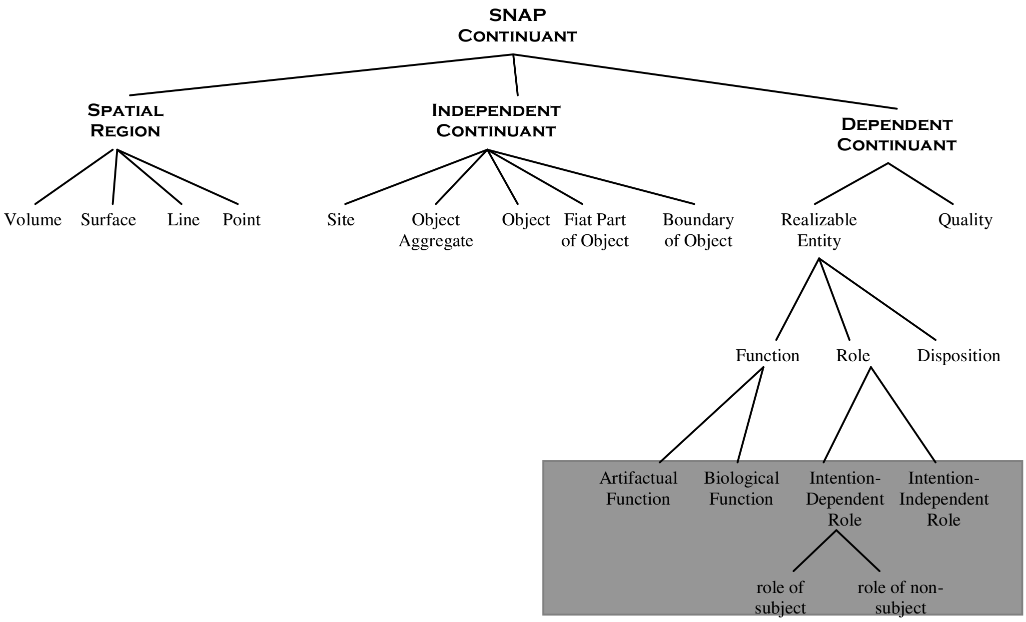
Basic Formal Ontology



Chapter 5: Introduction to Basic Formal Ontology and Relations

[67] See Werner Ceusters and Barry Smith, "Tracking Referents in Electronic Health Records", *Medical Informatics Europe* (MIE 2005), Geneva, Stud Health Technol Inform. 2005;116:71–76, and the other papers at http://ontology.buffalo.edu/referent-tracking/.

Appendix 1: Possible Downward Extensions of BFO Classes



<u>SNAP Level 5: Sub-Classes of Role</u>

intention independent

Df: A realizable entity that is not essential to a continuant entity in virtue of the kind of thing that it is, but that can be played by that kind of continuant entity in some kinds of natural contexts independently of all human thought, intention and intervention. (examples: the role of carbohydrate molecules in the digestive process, the role of a tree as nesting place for a bird, the role of an antelope as dinner for a lion, the role of a boulder in initiating an avalanche, the role of a beaver dam in creating a wetland area, the role of a tree in maintaining an oxygen carbon-dioxide balance in an ecosystem, the role of an ecosystem in stabilizing global climate patterns, the role of the earth in keeping the moon from spinning off into the sun)

Comments: Intention dependent roles are either *played* or *not played.*

intention dependent

Df: A realizable entity that is not essential to a continuant entity in virtue of the kind of thing that it is, but that can be played by that kind of continuant entity in some kinds of intentional contexts such as social or institutional contexts. (examples: the role of a document in hospital administration, the role of a drug in treatment of a patient, the role of a woman as lawyer, the role of a cat as house pet, the role of a public official in the context of a government, the role of an implanted artificial heart in the context of a living organism, the role of a surgical instrument in making a hair line incision just beneath the right nipple, the role of a child as legally guarded by a specific individual)[68]

Comments: Bearers of intention dependent roles are either used or not used correctly, or are either responsibly or irresponsibly fulfilling their roles.

SNAP Level 5: Sub-Classes of Function

biological function

Df: A realizable entity the manifestation of which is an essential end-directed activity of a living non-intentionally dependent continuant entity, or of the non-intentionally dependent products of such a living entity, in virtue of that living continuant entity being a specific kind of entity in the non-intentionally dependent kind or kinds of contexts that it is made for. (examples: the function of a birth canal to enable transport, the function of the heart in the body to pump blood, the function of reproduction in the transmission of genetic material, the digestive function of the stomach to nutriate the body, the function of a beaver's dam to generate a living space for it, the function of a spider's web to catch it's food)[69]

Comments: with regard to the performing of its biological function, a thing is either *healthy* or *ill.* Only living organisms have biological functions in virtue of their basic ends or goals. The natural parts and sub-parts of living organisms have functions only in virtue of the role that they play in bringing about the functioning of the organism of which they are parts. The biological function of the heart, to pump blood, is thus defined by the role that it plays in maintaining the

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[68] The notion of intentionality being appealed to here is essentially that of Brentano, F. (1973) Psychology from an Empirical Standpoint. New York: Humanities Press; Chisholm, R. M. 1984. The primacy of the intentional. Synthese 61:89-110; Crane, T. 2001. Elements of mind. Oxford: Oxford University Press and Searle, J. R. 1983. Intentionality: An essay in the philosophy of mind. Cambridge: Cambridge University Press, while the notions of intention-dependent and intention-independent and their use in theories of social reality receives treatment in Schutz, A. & Luckmann, T. (1973) The Structure of the Life-Word (Zaner & Engelhardt transl.). Evanston: Northwestern University Press; and more recently and directly in Searle, J. R. The Construction of Social Reality. New York: The Free Press, 1995.

[69] For IFOMIS writings that include discussions of biological function, see Johansson, Ingvar. "Functions, Function Concepts, and Scales." *The Monist* 86, 2004, 96—115, < http://hem.passagen.se/ijohansson/functions1.htm >; Johansson, I., Smith, B., Tsikolia, N. *et al*. 2005. Functional Anatomy. A Taxonomic Proposal. *Acta Biotheoretica*, 53(3), 153-66, < http://ontology.buffalo.edu/medo/Functional_Anatomy.pdf >; Smith, B. and Grenon, P. 2004. The Cornucopia of Formal-Ontological Relations. *Dialectica*, vol. 58, no. 3; Smith, B., Munn, K. and Papakin, I. 2004. Bodily Systems and the Spatial-Functional Structure of Human Body, in D. Pisanelli and M. Domenico (eds.) *Ontologies in Medicine: Proceedings of the Workshop on Medical Ontologies*. Rome, October 2003, Amsterdam: IOS Press, 39-63, < http://ontology.buffalo.edu/medo/OBS.pdf >. For a concise overview, synopsis and extension of the views put forward in these papers, as well as a contrast with other views of biological function, see Díaz-Herrera, Patricia. "What is a Biological Function?" forthcoming in *Proceedings of FOIS 2006.* For a general overview of positions and issues regarding teleology and function in biology, see Allen, Colin, "Teleological Notions in Biology", *The Stanford Encyclopedia of Philosophy (Summer 2004 Edition)*, Edward N. Zalta (ed.), URL = <http://plato.stanford.edu/archives/sum2004/entries/teleology-biology/>.

survival of the organism that it is a natural part of. The non-intentionally produced tools of living organisms have a biological function only in a derivative sense, relative to the organisms that produced them. Thus a beaver dam has a function relative to the kind beaver, while a spider web has a function relative to the kind spider.

artifactual function

Df: A realizable entity the manifestation of which is an essential end-directed activity of an intention-dependent continuant entity in virtue of that continuant entity being a specific kind of entity in the kind or kinds of intentionally dependent contexts that it is made for. (examples: the function of a hammer in the context of building something to drive in nails, the function of a government to protect the rights of its citizens, the function of a treatment to heal a patient, the function of a CD to encode and play back music, the function of a lawyer in the context of a particular government and set of laws, the function of a legal mother in providing basic life sustaining services for her legal children).[70]

Comments: with regard to the performing of its artifactual function, a thing is either *functional* or *broken*. Only intentionally dependent entities that have been designed according to some plan for the sake of achieving a particular purpose have functions. The parts and sub-parts of such entities normally have a function only with regard to the role that they play in the functioning of the entity as a whole. One important exception to this is arguably human persons. In virtue of being a person an individual has a certain biological end or function, but in virtue of playing a role in a society that is directed towards the achievement of some collective end or other individual persons can also play a role in bringing about the functioning of a society or social group.

SNAP Level 6 Subclasses of intention-dependent role

role of non-subject

Df: A realizable entity that is not essential to a non-subject continuant entity in virtue of the kind of thing that it is, but that can be played by that kind of continuant entity in some kinds of intentional contexts such as social or institutional contexts. Roles of non-subjects are the bearers of derived intentionality (the role of a document in hospital administration, the role of a drug in

[70] Hilpinen, Risto, "Artifact", *The Stanford Encyclopedia of Philosophy (Fall 2004 Edition)*, Edward N. Zalta (ed.), URL = http://plato.stanford.edu/archives/fall2004/entries/artifact/; for more detailed treatments, see Dipert, R., 1993, *Artifacts, Art Works, and Agency*, Philadelphia: Temple University Press; and Thomasson, A., 2003, 'Realism and Human Kinds', *Philosophy and Phenomenological Research* 67, 580-609.

treatment of a patient, the role of a cat as house pet, the role of an implanted artificial heart in the context of a living organism, the role of a surgical instrument in making a hair line incision just beneath the right nipple)

Comment: A non-subject entity can be used to realize its role either *correctly* or *incorrectly.*

role of subject

Df: A realizable entity that is not essential to a subject/agent in virtue of the kind of thing that it is, but that can be played by that kind of continuant entity in some kinds of intentional contexts such as social or institutional contexts. Roles of subjects are the bearers of both original and derived intentionality. (examples: the role of a woman as lawyer, the role of a public official in the context of a government, the role of a child as legally guarded by a specific individual

Comments: Subjects of intention dependent roles of subjects incur duties and obligations that they would not have were they not the subjects of such roles, they are therefore *responsible* or *irresponsible* for performing the duties associated with their roles.

Chapter 6: Information gathering: Determine what the universals and relations amongst them in the domain are

Once the domain or scope of an ontology has been decided on, what is needed is a systematic survey of information pertaining to the intended domain to be represented. For example, building an ontology for the domain of cellular processes requires surveying and integrating the most up to date, advanced and authoritative textbooks, research and researchers in the area of cellular processes, and similarly for the development of any other domain specific ontology.

The end result of such a survey should be the achievement of provisionally definitive answers to the following questions: what are the domain universals and relations that need to be represented, what is or are the best domain-specific term or terms that should be used in representing these universals and relations, what are the explicit upper and lower bounds of the ontology, and how much formal ontological apparatus is needed for dealing with the domain to be modeled?

Consult domain literature and domain experts

The best candidates for the truth about a given scientific domain are the theories currently held by scientists who study that domain. Thus if the domain to be modeled is cell biology, then the most current theories about cells, and researchers currently working on the study of cell biology, are what should be consulted. The activity of domain-specific ontology construction is usually an interdisciplinary one, and the ideal case is that such an endeavor include a number of competent domain experts. In every case the goal of consulting domain literature and experts is to gain as thorough and systematic a knowledge of the domain to be represented as is possible.

Determine the domain universals and relations

More specifically, the goal of consulting domain literature and experts is to determine what the universals and universal-level relationships relevant to a given domain are. Thus, in an ontology of bodily organs, universals would include "heart", "kidney", "lungs", etc., while relations of interest might include the relation of *located_in* between "heart" and "chest-cavity" and the relation *part_of* between the "respiratory system" and "lung".[71] Determining the domain universals and relations amounts to determining which invariant features and regularities in the domain play central roles in scientific explanations of that domain. E.g. no theory of the circulatory system would be complete without mention of the heart, veins, arteries and the various connections that these bear to one another, thus in such a theory of circulation these would be the domain universals and relations of interest.

Generate a terminology

Along with consultation of domain experts and literature and the determination of domain universals and relations should go the process of actively constructing a domain terminology, a set of terms characteristically used to talk about the entities in that domain. Such a terminology may be initially acquired from existing thesauri or domain handbooks, or it may be manually constructed as the consultation of domain literature and experts progresses. The initial domain terminology should include for each term-

- a listing of common synonyms or alternative expressions
- a clear concise natural language definition of the term
- an initial statement of the most likely ontological category to which the entities referred to by the term belong (continutant-occurrent, object-process…).

The terms and definitions in the initial terminology do not represent the final state of the terms and definitions that will be included in the domain ontology. They are rather a first-draft or gloss for the sake of getting the relevant information organized and assembled in a single place.

[71] Being careful all the while to keep separate instances and instance level relations on the one hand, and universals and universal level relations on the other, as has been discussed in chapters 1 and 5.

Further principles for terminology selection and regimentation will be discussed in the next section.

Determine the granularity required

While the question of the upper and lower boundaries of the granularity of entities to be represented by the ontology should already have been initially considered at the point where the domain of the ontology was first specified, once the process of consultation of domain literature and experts has been completed, it should be possible to provide a rather precise statement of the levels of granularity to be represented by the ontology.

If there was any question as to whether or not detailed information about genetic material should be included in a domain ontology about cells, a careful survey of current literature and theories in the area of cell biology should render the answer to this question clear. Thus, final consideration of the exact scope of the ontology should take place at the point where the domain universals, relations and corresponding terminology have been determined, but before these things have been regimented in a systematic fashion.

Determine the amount of formal ontological apparatus required

Similar to the issue of granularity, the question of which formal ontological categories and relations are necessary for the structuring of a given domain ontology will depend in large part on the contents of that domain ontology. Thus, while initial consideration of the issue of formal ontological categories and relations should take place when the scope of the ontology is initially stated, a final determination of what formal ontological categories and relations are relevant should take place after a relatively thorough survey of domain specific information has taken place. In terms of the BFO formal ontology previously developed, for example, an ontology of anatomy will require only the use of categories and relations pertaining to SNAP or continuant ontological perspectives, while for the purposes of representing domain specific information about physiology it seems clear that categories from both SNAP and SPAN would be required.[72]

Chapter 7: Systematize and Regiment the Domain Information[73]

[72] Compare with the very helpful discussion in Eric Little, "A Proposed Methodology for the Development of Application-Based Formal Ontologies", at http://sunsite.informatik.rwth-aachen.de/Publications/CEUR-WS//Vol-94/ki03rao_little.pdf.

[73] The material here largely supplements and is supplemented by the on-line material to be found in Daniel Schober. "Recommendations." http://msi-ontology.sourceforge.net/recommendations/.

Once the domain information has been assembled in the form of a terminology with definitions recognizing the important domain universals and relations, and the appropriate scope, granularity and formal ontological apparatus required for the ontology have been determined, the next step is to regiment the domain information in a systematic and coherent fashion. The goal of this regimentation is to develop a representational artifact that is as logically coherent, unambiguous and true to the facts of reality as possible.

In order to overcome the obstacles to the use of computers in organizing the sea of biomedical information, the problems of *human idiosyncrasy, of Babel,* of *non-sense-in-non-sense-out,* and of *computer-information-solipsism* that were discussed in the introduction, it is necessary to establish a maximum amount of clarity, consistency and coherence in the domain information that is to be represented. The regimented representational artifact that will result from this process will itself serve as the basis for generating a formalized representational artifact for purposes of computer implementation.

There are three major and interrelated facets of regimentation for domain ontologies. These are terminological regimentation, definition of terminology, and location of terms in taxonomic hierarchies based on *is_a* relations. We will treat each of these issues in turn, though it should be noted at the beginning that there is a large degree of overlap and interdependency amongst principles for best practices in these areas.[74]

<u>Terminology</u>

[74] For a general overview of the kinds of thinking behind the contents of this chapter it is a good idea to simply consult the chapters on definitions and classification to be found in most standard introductory logic text books, such as Kelley, D. *The Art of Reasoning (Third Edition)*. New York: Norton, 1998. The basic ideas here being discussed are as old as Aristotle, see especially Aristotle. *Posterior Analytics*. In McKeon, R. ed. *The Basic Works of Aristotle*. New York: The Modern Library, 2001, along with the other logical works in the same or similar volumes (Prior Analytics, Topics, etc.). For a summary of Aristotle's logic, see Smith, Robin, "Aristotle's Logic", *The Stanford Encyclopedia of Philosophy (Fall 2004 Edition)*, Edward N. Zalta (ed.), URL = <http://plato.stanford.edu/archives/fall2004/entries/aristotle-logic/>. For a systematic attempt to formulate the Aristotelian approach to biological classification, see Barry Smith. "The Logic of Biological Classification and the Foundations of Biomedical Ontology." In Dag Westerstahl (ed.), *Invited Papers from the 10th International Conference in Logic Methodology and Philosophy of Science,* Oviedo, Spain, 2003. Elsevier-North-Holland, 2004. For a historically and philosophically informed discussion of Aristotelian principles as they relate to parts of the information ontology domain, see Derek Rayside and Gerard T. Campbell, "An Aristotelian Understanding of Object-Oriented Programming" (paper presented at the 15th ACM Conference on Object-Oriented Programming, Systems, Languages, and Applications (OOPSLA 2000), Minneapolis, Minnesota USA, 2000), Derek Rayside and Gerard T. Campbell, "Aristotle and Object-Oriented Programming: Why Modern Students Need Traditional Logic" (paper presented at the 31st ACM/SIGCSE Technical Symposium on Computer Science Education, Austin, Texas, 2000).

The process of gathering domain-specific information should result in the construction of a lexicon or terminology. However, if the goal is to use this domain specific information for purposes of representation in a computer-based ontology, then a more rigorous formalization of this terminology is needed. In general, the syntax of the terminology selected for inclusion in a domain ontology should be formulated in terms of clear and explicitly stated conventions, and should be such as to be familiar to and easily recognizable by potential users of the ontology, especially, but in some cases not limited to, domain experts. Regimenting a terminology thus involves both the explicit statement of (as well as ruthlessly consistent adherence to) syntactic conventions for the writing of terms and explicit consideration of the intended audience or user-base for the ontology. Some specific principles along these lines include the following.

Terminological Conservativism:

Don't reinvent the 'wheel': There are already a sufficient number of words in the world and in the biological and medical communities to ensure that the creation of new highly specialized terms for purposes of inclusion in a domain ontology will rarely, if ever, be necessary. A simple principle to follow in selecting terms for a domain ontology is to stay as close as possible to the actual use of people working in the field the domain ontology is about. In selecting terms for inclusion in a domain ontology, terms that are widely used and well-known by domain experts should be

a. given preference over highly specialized and little used terms, and
b. given the same meaning that they currently have in their use by domain experts.

Creating new terms to represent things that a community of domain experts are already familiar with under a different name, or using a familiar term with a new and different meaning, are both likely to lead to confusion, both in the encoding of information into the ontology, and in its interpretation by end-users. The terminological choices of domain ontology builder(s) should be as respectful as possible of the current terminology, usage and practice of contemporary domain experts and potential users of the ontology.

Singular Nouns:

For the sake of intelligibility: the general terms in an ontology should be formulated in the singular, and the ontology's documentation should pay careful attention to the distinction between singular and plural nouns and to the requirement of noun-verb agreement. Thus 'cat', not ‘cats’, and ‘eukaryotic cell’, not ‘eukaryotic cells’. There are a number of reasons why this convention should be adopted.

First, it is crucial that some syntactical standard or other be adopted and rigorously ruthlessly adhered to for the encoding of common nouns, in order to ensure that they always appear similar to human users, and in order to ensure maximal tractability for machines. In this respect rendering all such terms in the singular is as good a decision as any. Additionally, ensuring grammatical intersubstitutability of terms with their corresponding definitions (something that will be further discussed below) will be much easier if all terms have a standard grammatical format.

There is also a more principled reason for representing the common nouns or universal terms in an ontology in the singular. This is that the common nouns in an ontology always refer either to universals or to defined classes. In either case, however, the reference of these terms is singular. There is only one universal “feline”, even if it has many instances, and there is only one defined class “all the debutants in Texas in 1984”, even if it has many members. Thus it makes sense to use singular rather than plural terms to refer to entities such as universals and classes, and to do this with ruthless consistency when constructing a domain ontology.[75]

Common nouns in lower case:

For the sake of intelligibility: represent terms referring to universals or classes in all lowercase letters. Thus ‘cat’, not ‘Cat’ or ‘CAT’, and ‘eukaryotic cell’, not ‘Eukaryotic Cell’ or ‘EUKARYOTIC CELL’. As with the convention regarding use of singular nouns, this convention is proposed largely because some convention or other must be adopted and rigorously consistently adhered to. However, in English capital letters are normally used to indicate either a proper name (Tom, Seattle, Jupiter) or an acronym (the U.N., the E.U., the U.K.), whereas common nouns normally do not involve capital letters of any sort. It is thus more consistent with English usage to use all lower case letters for the encoding of general terms.[76]

[75] Barry Smith. “Against Idiosyncrasy in Ontology Development.” Forthcoming in B. Bennett and C. Fellbaum (Eds.), *Formal Ontology and Information Systems,* (FOIS 2006), Baltimore November 9—11, 2006.

[76] There are of course other languages with other grammatical rules for capitalization of nouns. Should it prove more intelligible to use capital letters in an ontology the natural language of which is, for example, German, then by all means this convention can be altered. Again, what is crucial is that the convention used by an ontology be explicitly stated and consistently adhered to throughout.

Avoid Acronyms:

For the sake of intelligibility: don't use acronyms as part or all of any term. Thus, instead of 'ATP' or 'atp' write 'adenosine triphosphate', and instead of 'dna' or 'DNA' write 'deoxyribonucleic acid'**.** Using an acronym rather than the term for a universal or class increases the chances of confusion on the part of domain expert-users, while rendering use of the ontology by non-domain experts nearly impossible. In the worst case, an ontology whose terminology is filed with acronyms will be equivalent to an ontology intended to be used by speakers of French that is written in Russian. The ontology itself will not be understandable or usable without appeal to some further interpretative or translational guide.

Univocity:

For the sake of intelligibility: Terms should have the same meaning on every occasion of their use. In an ontology, 'cell' should mean cell, 'cancer' should mean cancer, and similarly in all other cases. The principle of univocity in ontology terminology development is difficult to maintain because ordinary language regularly violates it. For example, the English word 'bank' can mean both "a financial institution" and "a stretch of earth directly connected and running parallel to a river". The reasons for avoiding such ambiguity in the context of ontology design are quite straightforward.

First, if a single term is used in more than one way in a given context, human participants in discourse regarding that context are likely to become confused.

Second, if a single term is used in more than one way within an ontology that is intended to be computer implemented, it is likely that the computer will not be able to keep the two uses distinct, thus leading to both computational errors and user confusion.[77]

It should be noted here that what the principle of univocity specifically says is that every term in an ontology should have one meaning, not that no two terms should have the same meaning (as in the case of synonyms). It is often important to include not only a term and its meaning, but also other terms that are synonymous with this term, in an ontology. However, this is entirely different than giving one and the same term multiple meanings.

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[77] For a discussion of the violation of the principle of univocity regarding the relations *part_of* and *is_a* in the Gene Ontology, see Smith, B., Köhler, J., & Kumar, A. On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology; see also Barry Smith. "Against Idiosyncrasy in Ontology Development." Forthcoming in B. Bennett and C. Fellbaum (Eds.), *Formal Ontology and Information Systems,* (FOIS 2006), Baltimore November 9—11, 2006.

One example of violation of the principle of univocity comes from the National Cancer Institute Thesaurus' term 'disease progression', which includes three different possible interpretations: (1) "Cancer that continues to grow or spread"; (2) "Increase in the size of a tumor or spread of cancer in the body"; (3) "The worsening of a disease over time. This concept is most often used for chronic and incurable diseases where the stage of the disease is an important determinant of therapy and prognosis".[78] While the relationship between the meaning of the first two definitions of the term 'disease progression' might be debatable, the first two and the third definitions are clearly different. In definitions (1) and (2) 'disease progression' is something that involves only cancer, whereas in the third definition 'disease progression' involves the worsening of any disease whatsoever over time, a 'disease progression' is identified as a "concept", not as a thing or a process, and use of the term usually implies the presence of an incurable or chronic disease. Imagine two different doctors from different hospitals attempting to talk about the 'disease progression' in a particular patient, while using these different definitions: the first doctor will think that the patient has cancer, while the second will think that the patient is afflicted by an unspecified disease that involves a concept and is most likely chronic or incurable.

Universal/Instance Univocity:

For the sake of intelligibility: Terms/expressions referring to Universals, and terms/expressions referring to instances should be clearly demarcated. For example, the common noun 'dog' can be plausibly understood as referring to a type or universal "dog". The term 'dog' which occurs in the sentence 'The accident was caused by a dog unintentionally ejected from a motor vehicle due to failure to use restraining harness' can be plausibly understood as referring to a single particular dog. However, these two uses of the term 'dog' should be kept clearly separate in an ontology. There are a number of different ways to do this. One simple way would be to abide by the conventions we have already put forward for representing common nouns, using 'dog' to refer to the universal dog, while using a capital letter, proper names or alphanumeric strings to refer to particular dogs, as in 'Dog', 'Fido' or

[78] NCI Thesaurus accessed via UMLS Knowledge Source Server Version 2006AC, Thursday, September 28, 2006.

'#d437' (importantly, by using one of these conventions consistently, not by mixing all three together!).[79]

Definition of Terms

Whereas regimenting the terminology of an ontology is primarily the task of adopting and ruthlessly consistently abiding by syntactical conventions for the naming of various kinds of entities within the ontology, regimenting the definitions of terms in an ontology is a semantic task, one that has to do with providing a definitive statement of the nature of the things that the terms refer to. In a scientific ontology we are not interested in the *lexical* or *conventional* definition of a term, a definition that reports the meaning that all or most members of a given language community attribute to a term (as can be found in a dictionary), but rather in the *real* definition of a term, that is, in a scientific statement of the basic nature of the kind of thing that that term refers to. Providing definitions is similar to selecting the terminology in a domain ontology insofar as it is important that the definitions be as clear, consistent and accurate as possible, while also being organized in terms of a coherent and consistently applied set of conventions. In the following we put forward a number of principles for the formulation of domain-ontology definitions.

Essential Features:

Use essential features in defining terms: The definition of a term referring to a universal or kind should be stated in terms of the essential features of the entities that are instances of that kind. The essential features of a thing are those features without which the thing would not be the kind of thing that it is. For example, it is arguably not essential to a thing's being an instance of the universal human that it have precisely two legs, ten fingers, an appendix or blond hair. On the other hand, if an entity is unable to engage in any kind of communication, to think in a (somewhat) rational way, or to have certain kinds of self-reflective or self-aware thoughts, then this might be grounds for maintaining that this thing is indeed not an instance of the universal human. Thus Aristotle defined 'human' as "an animal that is rational". Taking rationality and being an animal to be essential features of human beings means saying that while many other features of a thing may change (such as hair color, skin color, body parts, height, weight, strength, taste in food, etc.), develop or be eliminated altogether, a thing cannot lack either the

[79] Barry Smith. "Against Idiosyncrasy in Ontology Development." Forthcoming in B. Bennett and C. Fellbaum (Eds.), *Formal Ontology and Information Systems,* (FOIS 2006), Baltimore November 9—11, 2006.

feature of being an animal or the feature of being rational and still be an instance of the universal human being.

For natural objects, such as those studied by chemistry, biology and physics, the essential features of a thing are usually the features of that thing that play a prominent role in scientific explanation of its existence and behavior. Thus a good Aristotelian definition of 'water' would be water is a molecular compound consisting of hydrogen and oxygen, or water is the molecular compound H20.

For artifacts, objects created by humans to be used in various contexts, the essential features usually have to do with the purpose or use for which the artifact was created. Thus a knife is a tool for cutting things, while a chair is furniture that can accommodate a normal sitting human being.

Definitions in a domain ontology should always be in terms of the essential features of the entities under consideration. What is essential to the domain as a whole will, as a general rule, be determined by the statement of the intended scope of the ontology. Thus the statement in the Foundational Model of Anatomy documentation that "The FMA…is strictly constrained to "pure" anatomy, i.e., the structural organization of the body",[80] already provides some clues as to what the essential features of entities in this domain will be.

Specifically, in a domain ontology of anatomy, the essential features of entities will include their location in the body, and the spatial and physical relationships that they stand in to other bodily parts, organs and structures, as well as the internal organization of the entities under consideration. For example, the FMA defines 'heart' as an "Organ with cavitated organ parts, which is continuous with the systemic and pulmonary arterial and venous trees".[81] While quite sophisticated, this definition makes reference to precisely the sorts of essential features of entities that one would expect to find in an anatomy ontology, the internal structure and parts of the entity ("Organ with cavitated organ parts"), and reference to the relationship the entity bears to other bodily parts and structures ("continuous with the systemic and pulmonary arterial and venous trees").

In addition to considering the nature of the domain under investigation, another way to go about determining what the essential features of entities instantiating a domain universal are is

[80] FMA, http://sig.biostr.washington.edu/projects/fm/AboutFM.html, accessed July 17, 2006.
[81] FMA, http://fme.biostr.washington.edu:8089/FME/index.html, August 3, 2006.

the following. In the light of available scientific knowledge, attempt to imagine the subtraction and variation of the features that the entity has, at each step checking to see whether or not the variation in features leads to the intuitive conclusion that the entity is no longer an instance of the universal referred to by the term that we are interested in defining. If the removal or drastic variation of a feature of the entity in imagination leads to the conclusion that the entity is no longer an instance of the universal under consideration, then it is highly likely that that feature is one of the essential features of entities of this kind, and should be included in the definition of the term referring to the universal in question. To take a simple example, when engaging in this procedure regarding instances of the kind chair, it is possible to imagine chairs with all different features of size, shape and color; however, the second that one imagines a thing in which it is impossible for a normal human being to sit, it is clear that whatever the entity being imagined is, it is not an instance of chair, and so the feature of being a thing upon which a normal human being can sit is at least a necessary condition for something's being a chair.

A final point is that with regard to defined classes that do not refer to universals on the side of reality, the essential features to be used in the definition just are the features mentioned in the arbitrary designation of the class. Thus the 'essential' features of the class of all people suffering from HIV on the African continent just are "to be suffering from HIV" and to be "on the African continent".

Some examples of definitions that fail to utilize essential features of the things being defined include the definition of 'water' from the International Classification of Nursing Procedures as "a type of Nursing Phenomenon of Physical Environment with the specific characteristics: clear liquid compound of hydrogen and oxygen that is essential for most plant and animal life influencing life and development of human beings",[82] the definition of 'living subject' as "a subtype of Entity representing an organism or complex animal, alive or not" from the HL7 RIM, [83] and the definition of 'person' as "A living subject representing a single human being [sic] who is uniquely identifiable through one or more legal documents", from the HL7 Glossary.[84]

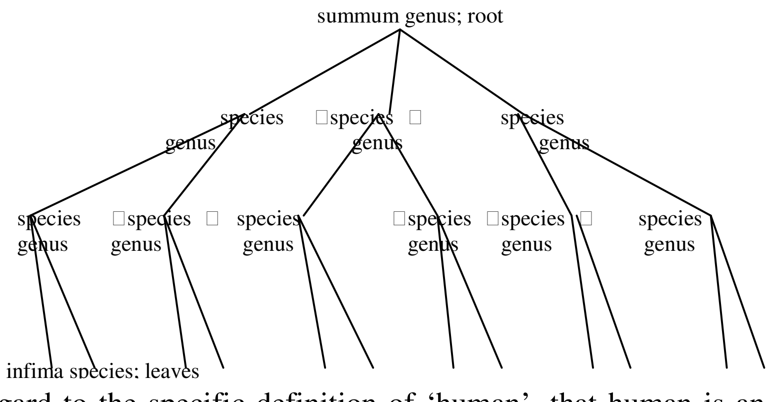
[82] International Classification of Nursing Procedures (ICNP), http://www.icn.ch/icnp.htm, accessed May, 2006.
[83] HL7 Version 3.0 accessed via Knowledge Source Server Version 2006AC, Thursday, September 28, 2006.
[84] Various Contributors eds., HL7 Publishing Technical Committee. Last Published 11/22/2005 8:05 PM. HL7® Version 3 Standard, © 2005 Health Level Seven®, Inc.

Aristotelian structure:

Use Aristotelian structure when formulating definitions: Consider again Aristotle's definition of 'human': a human is an animal that is rational. This definition has the basic form 'An A is a B that Cs/is (a) C'; an A (human) is a B (animal) that Cs (is rational). This basic format should be used to structure the definitions that are provided for terms anywhere in a domain ontology. The advantages of using this structure are that A, B, and C will always occupy the same places in the definition, and they can always be interpreted in similar ways, regardless of the specific domain in which terms are being defined.

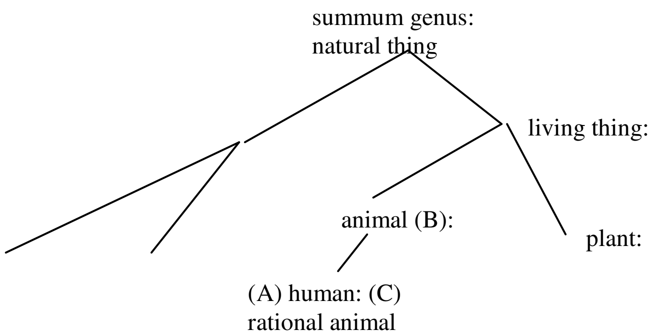
The traditional Aristotelian definition structure should be understood in the following way: A is the term that is being defined ('human', 'chair', 'cell', etc.), B refers to the genus of the original term, the next highest class/universal in the hierarchy of classes/universals in which the term is located, and C refers to the differentia of the universal designated by A. The differentiae of A are the essential features of A, those features that any entity must possess in order to be an instance of A, and those features that distinguish entities of kind A from all other entities. Thus in the Aristotelian definition of human, a human (A) is an animal (B: genus) that is rational (C: differentia). The structure of Aristotelian definitions can be understood against the background of species-genus hierarchies, the taxonomies that universals naturally form in which the higher levels of the hierarchy represent universals of greater generality (genus) relative to the lower levels (species) in the hierarchy. This basic structure is represented in diagram 2.

Diagram 2.



With regard to the specific definition of 'human', that human is an animal that is rational, the structure that this definition is based on can be seen in diagram 3.

Diagram 3.



This diagram shows how the Aristotelian definitional structure maps onto the hierarchy of greater and lesser generality amongst universals, a hierarchy that is normally represented as a taxonomy structured by *is_a* relations. More will be said about taxonomic structures and classification in the next section, however, it should already be clear that one advantage of consistently using the Aristotelian definitional structure is that it can be used to explicitly locate the place in *is_a* hierarchies of the universals referred to by the terms being defined.

The Aristotelian definitional structure thus represents a consistent format for the representation of definitions that can be used regardless of ontological domain, and that is inherently directed at explicitly representing the location of the term defined in an *is_a* hierarchy based on the informational content and structure of the definition alone. These are all advantages of the Aristotelian definitional structure, and reasons why it should be utilized in a systematic fashion while constructing domain ontologies.

Aristotelian approach:

Define the terms in an ontology from the top down: Terms in an ontology should be defined by beginning with the most general universals, and then by systematically working 'downwards' towards the least general. This procedure is highly consistent with the principle requiring the use of Aristotelian structure in definitions. Beginning with an undefined or primitive top node or root term, terms on the next level down will be defined by saying that an A is a B (top level node and genus) that Cs/ is (a) C (differentia). This procedure can be reiterated as many times, and at as many different levels as necessary, but starting from the most general level keeps things simple at the beginning, and gives the ontology developer a better perspective from which to assess the comprehensiveness of the ontology that she is building.

A more general consideration in favor of the top down approach comes from the point, already discussed earlier, that an ontology should have a well defined and delimited domain to which it is intended to apply, one that is determined, as much as possible, by the actual unity of scientific and practical domains of research in reality. Thus beginning with the more general entities and relations in an ontology and working downwards ensures ruling out, form the beginning, consideration of entities that are not relevant to the domain one's ontology is intended to represent.[85]

Positivity:

Don't use negatives: Definitions in a scientific domain ontology are intended to convey the essential information about their subject-matter to a user. Utilizing negative predicates (non-physical, non-environmental, non-cellular), or negative characterizations (not a part of the heart, not a breathing thing) involves providing much less information about the entities referred to by the term being defined than would be provided if only positive characterizations were given. Compare, for example, a definition of heart as "an organ that is *not* part of the nervous system", with the definition from the FMA, the heart is an "Organ with cavitated organ parts, which is continuous with the systemic and pulmonary arterial and venous trees". The first, negative, definition of the heart ensures only that the heart is not the brain, while leaving entirely open the possibility that it is the lungs, the kidneys or any number of other organs that are "not part of the nervous system". Thus, while negative definitions do provide *some* information about the entities being defined, positive definitions are much more exact and provide much more information, and they should be preferred and formulated whenever possible in the construction of domain ontologies.[86]

Intelligibility:

Keep it simple: The terms used in a definition should be simpler (more intelligible, more scientifically, logically or ontologically basic) than the term to be defined. Definitions of terms are given in order to explain to people who do not know the meaning of the term what that meaning is. It is generally the case that a person who does not know the meaning of a term, especially a technical term, also does not know the meaning(s) of terms more abstract or

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[85] Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

[86] Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

complex than the original term. Thus a definition that uses such abstract or complex terms in defining the original term is unlikely to serve its purpose.

A few examples from a standard logic textbook are the following: defining 'maturity' as "the stage of psychological development in which a person becomes well-adjusted" or 'death' as the "cessation of one's participation in finitude".[87] The problem with both of these definitions is that understanding them requires understanding other terms, such as 'psychological development', 'well-adjusted', 'finitude' whose meanings are more complicated than the terms being defined. In scientific contexts it is inevitable that definitions will involve a certain degree of complexity and specialized terminology, however this should be kept to an absolute minimum in ontology design. Further, when specialized and potentially obscure terminology is used in the definition of a given term, the ontology should either itself include or at the very least include references to clear definitions of this terminology itself.[88]

Some examples of definitions that violate the principle of intelligibility in actual ontologies include the old BIRNLex definition of 'mouse' as a "common name for the species *mus musculus*",[89] and the old 'GO:0007512: adult heart development', which was defined as "generation and development of the heart of a fully developed and mature organism".[90]

Non-Circularity:

Avoid circularity in the definition of terms: A definition is circular if the term to be defined, or a near synonym of that term, occurs in the definition itself. For example, defining 'plant cell' as "a cell that is found in plants" or 'surgical tool' as a device that is used by surgeons. These definitions are circular because they provide no more information about the nature of the things the terms refer to than the terms themselves provide. Since definitions are intended to explain the meaning of a term to someone who does not already understand that term's meaning, using the term itself or some very similar expression in its own definition defeats the purpose of providing a definition in the first place.[91]

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[87]Tutorial Web-Site for Kelley, D. *The Art of Reasoning*, http://www.wwnorton.com/college/phil/logic3/ch3/index.htm, accessed August 4, 2006.

[88] Smith, B., Köhler, J., & Kumar, A. On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology."

[89] BIRNLex, http://137.110.143.4:8080/BIRNLex/. Note: this definition has now been fixed.

[90] Smith, B., Köhler, J., & Kumar, A. On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology. Note: this definition has now been fixed.

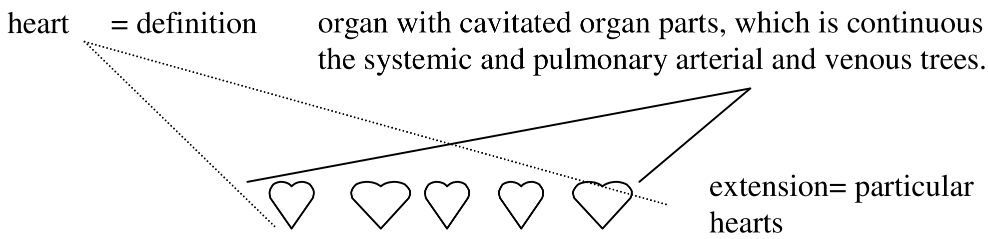
[91] See Kohler J, Munn K, Ruegg A, Skusa A, Smith B. "Quality Control for Terms and Definitions in Ontologies and Taxonomies." *BMC Bioinformatics*. 2006 Apr 19;7(1):212.

Some (now repaired) examples include the old GO definition of 'hemolysis' as "the causes of hemolysis",[92] and the old BIRNLex definition of 'eyeball' as "the eyeball and its constituent parts".[93]

Term-Definition Intersubstitutability:

In all extensional contexts a defined term should be intersubstitutable with its definition in such a way that the result is both grammatically correct and truth-preserving. The basic idea behind this principle is that wherever a term refers to a thing the definition of that term should also successfully refer to that thing. Thus in the FMA the extension of the term 'heart' should be identical with the extension of its definition 'organ with cavitated organ parts, which is continuous with the systemic and pulmonary arterial and venous trees'.[94] This relationship can be seen in diagram 4.

Diagram 4.



The intersubstitutability of a term and its definition with regard to the truth-value of sentences in which they occur is important both for preserving truth across inference in automated reasoning contexts and for ensuring intelligibility for human users of ontologies. Additionally, the intersubstitutability of a term and its definition with regard to preservation of grammatical correctness is important for the sake of intelligibility to human users. If replacing a term with its definition results in a grammatically incorrect expression, this will substantially impede the human usability of an ontology.[95]

Werner Ceusters and Barry Smith. "A Realism-Based Approach to the Evolution of Biomedical Ontologies." forthcoming in *Proceedings of AMIA Symposium. 2006.* http://ontology.buffalo.edu/bfo/Versioning.pdf, and also Barry Smith. "Against Idiosyncrasy in Ontology Development." Forthcoming in B. Bennett and C. Fellbaum (Eds.), *Formal Ontology and Information Systems,* (FOIS 2006), Baltimore November 9—11, 2006.

[92] Gene Ontology, http://www.geneontology.org/. Note: this definition has now been fixed.

[93] BIRNLex, http://137.110.143.4:8080/BIRNLex/. Note: this definition has now been fixed.

[94] FMA, http://fme.biostr.washington.edu:8089/FME/index.html, August 3, 2006.

[95] For an example from GO, see Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

Context-independence:

Don't leave the definition of a term open to interpretation: It should not be up to the end user of a domain ontology to decide or interpret whether or not the term 'heart' in the ontology means "human heart" or "canine heart", nor should it be up to the user to decide whether 'cell' means "animal cell", "plant cell" or "cell in general". This information should be explicitly included, either in the term itself (say 'plant cell' rather than just 'cell'), or in the definition of the term.

Scientific theories are intended to express the truth about reality in their respective domains, full stop. Thus a scientific definition, ideally, is not just sometimes or partially true, but is true, period. Definitions of scientific terms should attempt to capture this fact. Conversely, rendering the definitions of many scientific terms context independent will involve including more information about context in these terms themselves and/or in their definitions. For example, anatomy is the study of the physical structures present in organisms in general, whereas human anatomy, mouse anatomy, etc. are particular sub-fields of anatomy in general. Thus terms and definitions within these sub-fields, in order to be as context free as possible, should include the fact that they are definitions within a sub-field. The following definition of 'cell' as "structural and physicological unit of a living organism; it (i.e., plant cell) consists of protoplast and cell wall" from the Plant Ontology, violates this principle because it implicitly characterizes the term 'cell', which one would normally expect to refer to the general universal "cell", as applying only to cells within a specific domain, namely "plant cells".[96] Here it would be much better if, instead of 'cell', the term 'plant cell' was used.

Modularity:

A set of definitions should be modular: Modularity is not a feature of a single definition, but rather a property that a set of definitions has if it has been structured in a certain way. A set of definitions satisfies the requirement of modularity if they are organized into levels, with level 0 (root or most general) terms picked out as undefined primitives, and terms on levels n+1 for every n greater than 0 being defined by appeal exclusively to logical and ontological constants together with already defined terms taken from levels less than n+1. In practice what this rather complicated principle means is just that the terms in an ontology should be defined in Aristotelian fashion, from the most general to the most particular, while making use of the

[96] Plant Ontology, http://www.plantontology.org/. Note: this definition has now been fixed.

Aristotelian definitional structure, and abiding by the other principles that have already been discussed.

The principle of modularity is thus closely related to the fact that a system of well-defined terms regarding a specific domain should in normal cases form a hierarchically structured taxonomy. More specifically, if all or most of the terms being defined refer to universals on the side of reality, then the hierarchy amongst universals from more specific (cat, fern, human) to more general (mammal, plant, organism) should be reflected in the definitions of the terms that refer to these universals. The principle of modularity is explicitly intended to ensure that terms lower down in a taxonomic hierarchy inherit all properties and characteristics from their parents, and following this principle helps to ensure logical consistency in the definition of terms, clear demarcations amongst levels of abstractness within the ontology, and the possibility of automated reasoning.[97]

Term-definition/taxonomic-location transparency:

is_a should be built in: Ideally, each term's definition will represent the location in a term hierarchy to which that term belongs. The principle of term-definition/taxonomic-location transparency essentially summarizes and requires that ontology construction abide by the principles that have already been discussed.

If the principle of modularity and the Aristotelian top-down approach to term definition have been adhered to strictly, then the principle of term-definition/taxonomic location transparency will also be satisfied.

Alternatively, violation of the principle of term-definition/taxonomic location transparency by a system of defined terms suggests that the principle of modularity or the Aristotelian approach or both have been violated.

An ontology that adheres to this principle will be humanly intelligibly and, as a general rule, computationally tractable insofar as all of the terms defined in the ontology will also stand in clear relationships to one another.[98]

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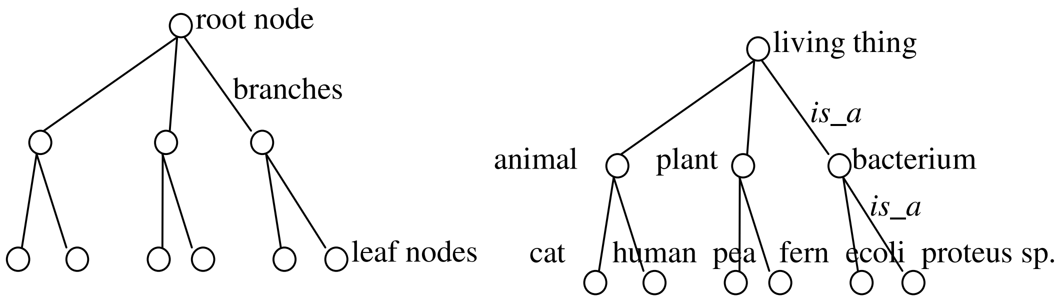
[97] Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

[98] Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

Taxonomy

Considered literally, a taxonomy is a tree-like structure consisting of nodes and branches, usually with a root node, leaf nodes, and intermediate nodes connected to each other, and to the root and leaf nodes by branches. Taxonomies are normally used to represent the hieararchical relationships amongst defined classes or universals in terms of the *is_a* relationship. Thus, a taxonomy is effectively an explicit representation of the structure possessed by a well-defined set of terms in a domain ontology.

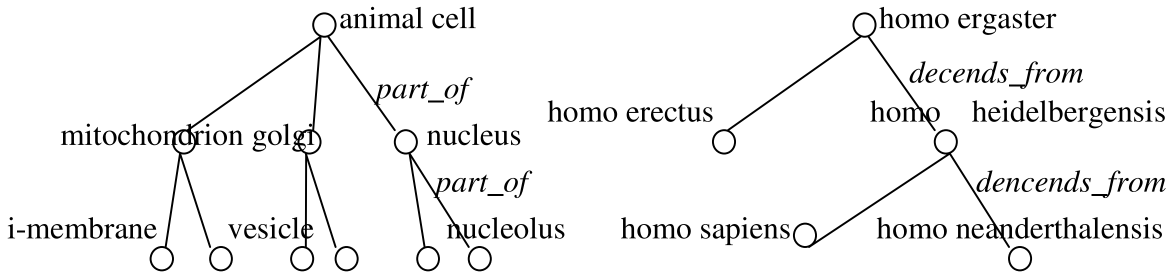
Diagram 5.



Importantly, taxonomic structures can be generated amongst universals and defined classes in terms of a number of different relationships. For example, the *part_of* relationship can be used to generate a taxonomic structure amongst universals. In such a case, the taxonomy generated might better be referred to as a partonomy. However, a partonomy is not a taxonomy based on the *is_a* relation. *Is_a* and *part_of* have different meanings, and therefore must be kept strictly separate during the process of ontology design. Similarly, the relationship *descended_from* can be used to generate taxonomic structures amongst biological species universals, as in phylogenetic trees.[99] Once again, such a *descends_from* relationship is different from either *is_a* or *part_of,* and taxonomies based on these various relationships should be kept rigorously separate.

[99] We did not define the *descends_from* relation or anything like it in the earlier section on ontological categories and relations; however this could be done using the same basic strategy of starting with a primitive instance-level relation, and then defining the relationship amongst universals in terms of it.

Diagram 6.



In what follows we will focus exclusively on taxonomies amongst universals structured by the *is_a* relation.[100] This discussion thus focuses on taxonomies understood as consisting of "finitely many universals arranged in a tree-like hierarchy."[101]

Classification:

Taxonomy is closely related to the issue of classifying entities. Indeed, a taxonomy just is one of the most common kinds of classifications of entities. But what is a classification? Relative to ontology, there are two major senses of the term 'classification'.

The first has to do with identifying entities as instances of a given kind. This can happen in two ways. Particulars can be identified as instances of universals that they instantiate (as in "this cat is an instance of the kind cat"), and universals themselves can be identified as belonging to formal ontological categories (as in "the universal cat belongs in the formal ontological category of object or substance"). In this sense of 'classification', classifying an entity just involves recognizing what type of entity it is, either at the domain level, or at the level of formal ontological categories.

The second sense of 'classification' is the one that is directly related to taxonomies, though it always presupposes that some amount of classification in the first sense of the term has already taken place. In this second sense, a classification is a systematic organization of entities belonging to a given ontological category based on the relationships that these entities stand in both within and across ontological categories. To make this characterization concrete, consider the universals "eukaryotic cell" and "cell". It is clear based on current biological knowledge that a eukaryotic cell *is_a* cell. However, fully understanding the import of this *is_a* relation rests on

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[100] Much of the following discussion can be usefully compared with the more technically oriented proposals of Rector, A. L. "Modularisation of Domain Ontologies Implemented in Description Logics and related formalisms including OWL." http://www.w3.org/TR/webont-req.

[101] Neuhaus, F., Grenon, P. & Smith, B. "A Formal Theory of Substances, Qualities and Universals", in Achille Varzi and Laure Vieu (eds.), *Proceedings of FOIS 2004. International Conference on Formal Ontology and Information Systems*, Turin, 4—6 November 2004.

two things. First, the universals eukaryotic cell and cell are both substantial universals; instances of both universals are entities that persist as identical through time, gain and lose qualities and parts, and are wholly present at any time at which they exist at all. However, the universal eukaryotic cell is differentiated from the universal cell in virtue of its having as part a cell nucleus, such that eukaryotic cell *has_part* cell nucleus is true.

Similarly, plant cell *is_a* eukaryotic cell. Both plant cell and eukaryotic cell are substantial universals, but plant cell is differentiated from eukaryotic cell by the possession of a cell wall as part, thus plant cell *has_part* cell wall is true. So, what is going on here is that substantial universals (cell, eukaryotic cell, plant cell) are being classified in terms of their standing in a specific ontological relationship (*has_part*) to other substantial universals (cell nucleus, cell wall). Notice that this also fits the Aristotelian definitional structure perfectly, "a eukaryotic cell (A) is a cell (B) that has a nucleus (C)".

Similarly, the Aristotelian definition of human, "a human is an animal that is rational" classifies substantial universals into an *is_a* hierarchy based on their standing in the *inheres_in* relation to universals belonging to the ontological category of qualities. Thus rationality *inheres_in* human, and differentiates the universal human from other substantial universals for kinds of animals.

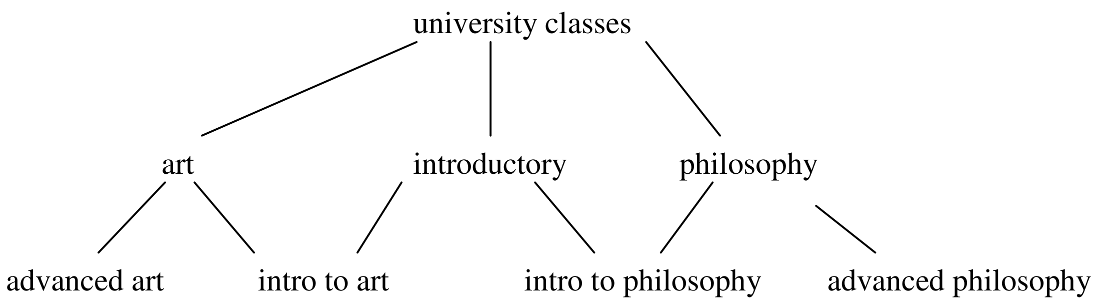
So, in this second (primary) sense, a classification is a systematic organization of entities belonging to a given ontological category based on the relationships that these entities stand in both within and across ontological categories. It is taxonomies in this sense that the universals in a good ontology should be organized in terms of. However, there is one more important feature of such classifications, and this is the principle in terms of which the classification has been generated.

The Principle of Classification:

Every classification should be structured according to the semantics for the *is_a* relation that we have already discussed, and should identify distinct universals or classes only when these can be distinguished both from the universals or classes at the next level up in the hierarchy, and from other universals or classes at the same level, by some ontological feature or relation (such as possession of a characteristic part or quality). However, in addition to these features, classifications that form *is_a* hierarchies are also normally structured in terms of a guiding principle or criterion of classification, one that further determines the specific meaning of the location of universals in the hierarchy, and determines, in part, which universals are relevant for inclusion in the hierarchy.

For example, it is possible to classify the classes in a university in terms of their subject matter, i.e. history, chemistry, biology, philosophy, etc. Alternatively, it is possible to classify the classes in a university in terms of their difficulty level, i.e. introductory, intermediate, advanced, graduate, etc. Each of these 'ways of classifying' university classes amounts to adopting a principle for the classification of university classes. What is important is that, for any given classification, the principle that is being used be specified as clearly as possible at the outset, and then consistently adhered to throughout. Further, two different principles should not be applied at the same level in the same hierarchy. Thus, a classification that attempted to simultaneously classify university classes both by subject matter and by difficulty level would end up looking something like the following:

Diagram 7.



The problem with this hierarchy is that saying A *is_a* B is ambiguous. For example, 'art *is_a* university class' can mean either that art is a university class of a particular kind *or* that art is a university class of a specific difficulty level. In other words, the relation *'is_a'* in this hieararchy is ambiguous. And while it may be clear from context in this particular case which meaning the '*is_a'* relation should be given in each particular context, in a more complicated case such as biological or medical science, such ambiguities, especially if they are perpetrated through an entire hierarchy of *'is_a'* relations, are likely to lead to a great deal of confusion, at least for human users, and often also for automated reasoning.

More sophisticated principles of classification include similarity and difference of anatomical structure (one principle) or similarity and difference of genetic code (a second distinct principle) for organisms, atomic number for the elements in the periodic table, and kind of patient treated by (one principle) or kind of procedure performed by (a second distinct principle) for the classification of doctors in a hospital.

Once more, what is crucial is that the principle being appealed to in classifying the entities in an *is_a* hierarchy be both explicitly identified and consistently adhered to from the beginning.

An Explanatory Note:

Up until this point the principles for best ontology practice that have been discussed have been intended to apply to all kinds of entities and domains whatsoever. The scope of the following recommendations, however, should be understood as limited in the following ways. Traditional philosophical discussions of classification and principles for best classification practices have focused primarily on the classification of substantial or objectual entities in terms of their characteristic qualities and or parts.[102] The following principles for best ontology practice, though arguably having wider application as well, are primarily offered in the spirit of these traditional treatments of classification.[103]

Recent work by Ingvar Johansson has suggested that the principles for best ontology practice with regard to classification may well depend on the kinds of entities being classified.[104] In particular, Johansson's work suggests that some of the principles that will be discussed below do not apply in a straightforward fashion when one is interested in classifying processes in certain ways. Until recently, theories of taxonomy and classification have been given little explicit treatment in philosophy, with most philosophers either ignoring them or presuming that they are perfectly well understood. The burgeoning field of information ontology in conjunction with Johansson's recent work has begun to challenge these assumptions. A full working out of the theory of classification as it applies to ontological entities of all kinds is, at present, beyond the scope of this work. Thus what follows should be taken as sound advice for the classification of most kinds of ontological entities (substances, continuant objects and qualities especially), but

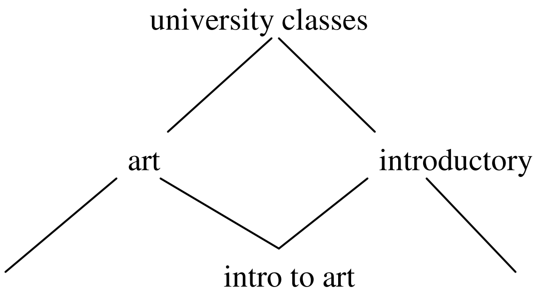
[102] See, for example Kelley, D. *The Art of Reasoning (Third Edition).* New York: Norton, 1998, chapter 1. For a rigorous logical treatment of the issue of biological classification that operates on the same Aristotelian assumptions,
[103] see Barry Smith. "The Logic of Biological Classification and the Foundations of Biomedical Ontology." In Dag Westerstahl (ed.), *Invited Papers from the 10th International Conference in Logic Methodology and Philosophy of Science,* Oviedo, Spain, 2003. Elsevier-North-Holland, 2004; Neuhaus, F., Grenon, P. & Smith, B. "A Formal Theory of Substances, Qualities and Universals", in Achille Varzi and Laure Vieu (eds.), *Proceedings of FOIS 2004. International Conference on Formal Ontology and Information Systems*, Turin, 4—6 November 2004.
[104] Ingvar Johansson. "Four Kinds of "Is_A" Relations." In Ingvar Johansson and Bertin Klein (Eds.), *WSPI 2006: Contributions to the Third International Workshop on Philosophy and Informatics.* Saarbrücken, May 3—4, 2006.

the proviso must be added that future work should be expected to produce more nuanced and perhaps also more varied principles applying to entities belonging to different ontological categories.

Use Single Inheritance:

No diamonds: Based on the forgoing discussion, an important principle for the classification of universals and classes in an ontology is to use single, not double- or multiple-, inheritance. In a classification the relationship of inheritance is the relationship that a less abstract class stands in to the more abstract class that is directly above it in a classificatory hierarchy. Thus cat stands in the inheritance relationship to mammal, plant cell to eukaryotic cell, and eukaryotic cell to cell. Saying that a classification should use single inheritance means saying that every universal or class included in the classification should stand in an inheritance relationship to exactly one universal or class at the next highest level. When this principle is violated, taxonomies take on a diamond-like structure, as can be seen in the outtake from diagram 7 pictured in diagram 8.

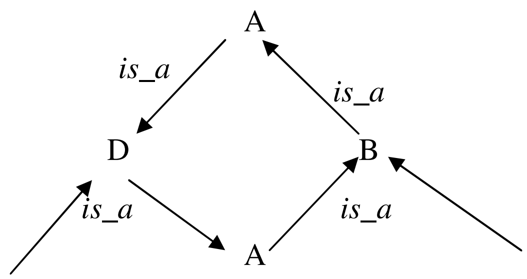
Diagram 8.



Such diamonds or 'multiple inheritance' should be avoided within a single classification in an ontology for a number of reasons. First, as already discussed, they lead to an ambiguity in how the '*is_a'* relation is to be interpreted in such classifications.

Second, allowing such diamonds into a classificatory scheme can lead to the existence of loops within the classification, such that A *is_a* B, B *is_a* C, C *is_a* D, and D *is_a* A, as illustrated in diagram 9. Allowing the existence of such loops in a classification amounts to adopting circular definitions for all or most of the terms located in the loop, and can lead to both human and computer confusion and errors. For example, a computer that is supposed to navigate through the classification hierarchy based on the *is_a* relations amongst universals can become trapped in such a loop, continuously reasoning in a circle until either a human being manually stops the program, or the matter in the computer degenerates, or the world comes to an end.

Diagram 9.



The following is an example of a loop that can be found in the UMLS:

"Topographic regions: General terms
Physical anatomical entity
Anatomical spatial entity
Anatomical surface
Body regions
Topographic regions"[105]

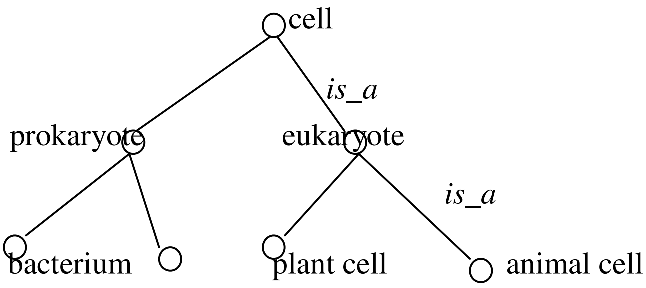
A third problem with multiple inheritance is that it can lead to double-counting and hence to double-naming of entities in a classification. For example, the class "Intro to Art" from diagrams 7 & 8 inherits its properties from both the class "Introductory classes" and the class "Art classes"; if care is not taken, it can easily happen that a class inheriting from multiple super-classes will be taken as itself identifying multiple classes (double or multiple counting), each of which requires a separate name (hence double or multiple naming). If not explicitly recognized, this state of affairs can quickly lead to both human and computer confusion.

A fourth and final problem with allowing multiple inheritance into an ontology is the following: Each classification should involve one and only one principle of classification in order to avoid the possibility of ambiguity in its interpretation, as has already been discussed. Ideally, the principles used for organizing universals in a scientific classification will be as closely related as possible to the essential features of the entities that instantiate those universals. Thus good candidate principles for the classification of biological species include similarity and difference of anatomical structure, on the one hand, and similarity and difference of genetic material on the other. It is largely for biological scientists to decide which of these features is more essential in the identification and classification of biological species. However, what is

[105] See Bodenreider O. Circular hierarchical relationships in the UMLS: etiology, diagnosis, treatment, complications and prevention. Proc AMIA Symp 2001:57-61.

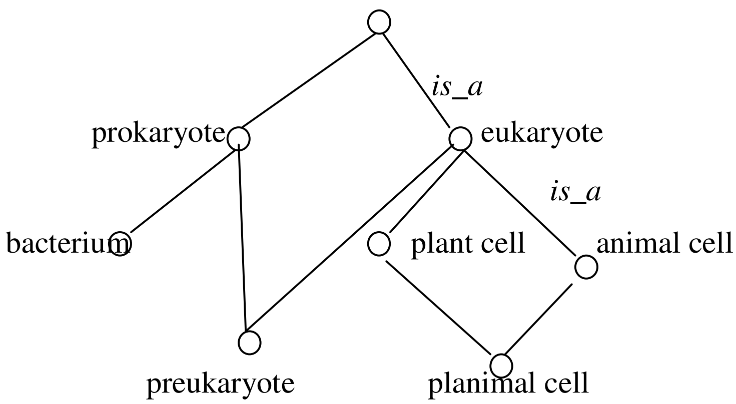
important here is that if the classification of universals in a domain is carried out by specifying essential features and, if the Aristotelian procedure and definitional structure (an A (term) is a B (genus) that Cs (differentia)) is adhered to, then universals at each level should be identified specifically in terms of features that *distinguish* them from all other universals or classes at that level. This means that no two universals at the same level should have any instances in common, and in turn means that no sub-universals or sub-classes of these universals should have any instances in common. Consider the classification of kinds of cells in diagram 10.

Diagram 10.



The classification in diagram 10 is consistent with the following Aristotelian structured definitions: “Prokaryote is a cell that lacks a nucleus”, “Eukaryote is a cell in which the genetic material is organized into a membrane-bound nucleus”, “Plant cell is a eukaryote cell that has a large central vacuole and a cell wall”, and “Animal cell is a eukaryote cell that has a small central vacuole and lacks a cell wall.” Now, given these definitions, suppose we were to allow the two instances of multiple inheritance from diagram 11 to occur.

Diagram 11.



The Aristotelian definition of ‘preukaryote’ would be “a prokaryote and eukaryote that has genetic material that is organized into a membrane-bound nucleus and lacks a nucleus”, while a 'planimal cell' would be 'a eukaryote cell that has a large central vacuole and a cell wall, and has a small central vacuole and lacks a cell wall". Now, the classes 'preukaryote' and 'planimal cell' should strike us as absurd. Their definitions contain manifest contradictions, and they seem like totally arbitrary and contrived denizens of the domain of cells. Note, however, that these classes were derived simply by allowing multiple inheritance into a classification that was well structured and organized according to a single principle along the lines of essential features of the universals in its domain, namely the kinds of cells and their essential differences in terms of possessing or not possessing certain kinds of parts (cell walls, a nucleus, etc.). Allowing multiple inheritance into a good classification leads to manifest absurdity, while a classification that straightforwardly yields multiple inheritance is probably a bad classification to begin with, either using a non-essential principle of classification for its classes and universals, or ambiguously attempting to apply multiple principles at the same time. While there are certainly cases of classification that are more complicated than cell biology, and where the choice of a principle of classification is not entirely clear, these cases are not different in kind, but only in degree of complexity from the case here discussed.[106]

Note: Importantly, it is often possible to classify a single universal in more than one way. For example, doctors can be classified in terms of the kinds of patients that they treat on the one hand, or in terms of the kinds of procedures that they perform on the other, and a term such as 'pediatric surgeon' could be classified in both of these ways (and would probably occupy a rather different place in the two classifications). However, in such cases, the answer is not to allow one taxonomy with multiple inheritance, but rather to construct two separate classifications, and use the definitions of the terms that appear in them, as well as the formal ontology (categories and relations) serving as the background of the domain ontology, to spell out the important relations between these two (or more) separate and diamondless classifications.[107]

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[106] Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm; Barry Smith. "The Logic of Biological Classification and the Foundations of Biomedical Ontology." In Dag Westerstahl (ed.), *Invited Papers from the 10th International Conference in Logic Methodology and Philosophy of Science,* Oviedo, Spain, 2003. Elsevier-North-Holland, 2004.

[107] Bittner, Smith, "Normalizing Medical Ontologies using Basic Formal Ontology", in *Kooperative Versorgung, Vernetzte Forschung, Ubiquitäre Information* (Proceedings of GMDS Innsbruck, 26—30 September 2004), Niebüll: Videel OHG, 199—201.
http://ontology.buffalo.edu/medo/gmds2004Norm.pdf#search=%22Bittner%2C%20Smith%2C%20%E2%80%9CN

Joint Exhaustiveness:

Don't leave relevant universals out: When classifying kinds of entities in a given domain, as much care as possible should be taken to ensure that all relevant universals are included at each level in a taxonomy. An ideal classification would include all existing domain universals along with identifying and differentiating information for each, at each level in the hierarchy of organization. This does not mean that the designers of an ontology should sit around waiting for new scientific information (of which there is always more) before completing their ontology and making it available for use, but it does mean that all relevant domain universals that are discussed in contemporary domain literature and by contemporary domain experts should be included.

Mutual Exclusivity:

No shared sub-classes: No two universals or classes in a classification should have any sub-classes in common. In keeping with the above example, in a classification in which eukaryotic cell and prokaryotic cell occur at the same level as separate universals, they should have no sub-classes or sub-universals (no multiple inheritance) in common. The requirement that the universals or classes at each level of a classification be mutually exclusive is a straightforward consequence of the prohibition of multiple inheritance, and conversely.

Class Positivity:

Complements of classes are not themselves classes: The complement of a class is the class containing all of the entities that do not belong in that class. Thus the complement of the class "dog" is the class "non-dog". As a general rule of thumb, class-compliments should be avoided when selecting the classes for and constructing the classification hierarchies in an ontology. The only thing that all of the members of a class-complement are guaranteed to have in common is the fact that there is some other class to which they all do not belong. Thus saying, of a given sub-class that it is subsumed by, for example, the class of all "non-conifer trees" is providing very little information about that sub-class. Further, class-complements rarely pick out genuine universals on the side of reality. Thus including many class complements in one's ontology is likely to render it less accurate to the facts of reality, and so less useful for both scientific and practical purposes.

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ormalizing%20Medical%20Ontologies%20using%20Basic%20Formal%20Ontology%22; see also Rector, A. L. "Modularisation of Domain Ontologies Implemented in Description Logics and related formalisms including OWL." http://www.w3.org/TR/webont-req.

There are exceptions to this rule, including some of the examples given above. For example, prokaryotic cells are distinguished form eukaryotic and from all other cells precisely by the fact that they lack a cell nucleus. This is, in effect, negative information used to define a class. However, in this particular case there is overwhelming scientific evidence to the effect that, at this level of generality, dividing up cell universals in this way does lead to a principled and exhaustive classification of all kinds of cells. In such cases including negative information, or even featuring it in the definition of a term and the demarcation of a class may be unavoidable. However, even in these cases every effort should be made to include some positive information about the kinds of entities being defined and classified as well.

Class Objectivity:

Which classes exist is not a function of the current state of biological knowledge. Genuine classes, that is, the universals treated by natural science in any given domain, are discovered, not invented or created. This fact suggests a certain kind of general attitude or mind-set that should be taken towards the identification of classes in an ontology, namely, one that seriously takes into account the best available scientific information about reality in any given domain, and attempts to systematically organize that information according to its most essential characteristics.[108]

Class-Univocity:

As with terms, so with classes: No distinctions without differences. Every class should be clearly distinct from every other class in the ontology with respect to at least one property or characteristic. The best that having two classes characterized by exactly the same set of properties can achieve is redundancy. Failure to adhere to the principle of class-univocity can also be problematic insofar as assigning different names to classes that are the same will be likely to lead to human and computational confusion.

Hierarchical Structure:

Classifications have a hierarchical structure: Given the discussion that has gone before, the point should already be quite clear, that the terms in a classificatory hierarchy should be divided into predetermined levels. A classification hierarchy should include terms both for more specific and for more general universals. Thus besides including "red", "human" or "H20" a

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[108] Barry Smith, Jacob Köhler and Anand Kumar. "On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology" in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

good classification should also include, as separate entries, “color”, “animal” or “compound”. Explicitly including this information will generate a division of terms into levels of generality, as can be seen in the examples discussed above. Hierarchically structured classifications are also a straightforward result of strictly adhering to the Aristotelian structure and method for good definitions, of identifying entities in terms of essential characteristics, and of classifying entities consistently in terms of a single principle of classification.[109]

Regimentation: Summary

The process of regimenting the domain information for an ontology thus includes the following steps, all to be carried out in terms of the principles that have been put forward above. First, select the exact terms and the format of these terms that are to be included in the ontology, based on domain information that has already been gathered. Second, provide clear, scientifically accurate and logically coherent definitions for each of these terms. Third, explicitly recognize the place or places of each of these terms in a hierarchical classification of the domain information. When this task has been consistently carried out, the domain information should be ready for the last great step in ontology development: formalization and computer implementation.

[109] Barry Smith, Jacob Köhler and Anand Kumar. “On the Application of Formal Principles to Life Science Data: A Case Study in the Gene Ontology” in *Data Integration in the Life Sciences, First International Workshop*, DILS 2004, Leipzig, Germany, March 25-26, 2004, Proceedings, Editor: Erhard Rahm.

Future Chapters

It should be once more emphasized that the above materials are drafts still representing work in progress. They have been made available due to overwhelming demand, and in the hopes of providing a point of reference for future discussions of information ontology, and of the Basic Formal Ontology in particular. In this spirit comments, suggestions and criticisms are all welcome and, if received, will be incorporated into future versions of the document.

It is hoped that future chapters of this document will be able to deal directly with the specifics of applying formal ontologies to domain information, both theoretically, and in specific computer implementation contexts (OWL, Protégé etc.). Work on these things is currently underway and will hopefully be available in the near future.

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