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### **Semantic Interoperability and Data Mining in Biomedicine**

## **Engineering ontologies: foundations and theories from philosophy and logical theory**

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## 1. The philosophical roots of ontology

Ontology as a branch of philosophy is the science of what is, of the kinds and structures of objects, properties, events, processes and relations in every area of reality. ‘Ontology’ is often used by philosophers as a synonym for ‘metaphysics’ (literally: ‘what comes after the *Physics*’), a term which was used by early students of Aristotle to refer to what Aristotle himself called ‘first philosophy’.<sup>1</sup> The term ‘ontology’ (or *ontologia*) was itself coined in 1613, independently, by two philosophers, Rudolf Göckel (Goclenius), in his *Lexicon philosophicum* and Jacob Lorhard (Lorhardus), in his *Theatrum philosophicum*. The first occurrence in English recorded by the OED appears in Bailey’s dictionary of 1721, which defines ontology as ‘an Account of being in the Abstract’.

The methods of philosophical ontology are the methods of philosophy in general. They include the development of theories of wider or narrower scope and the testing and refinement of such theories by measuring them up, either against difficult counterexamples or against the results of science. These methods were familiar already to Aristotle. Some philosophical ontologists conceived ontology as being based on a special *a priori* insight into the essence of being or reality. Here, however, we prefer to look at the entire history of ontology as an endeavor which has some of the features of an empirical science. Seen from this perspective, ontology is like physics or chemistry; it is part of a piecemeal, on-going process of exploration, hypothesis-formation, testing and revision. Ontological claims advanced as true today may well be rejected tomorrow in light of further discoveries or new and better arguments.

The general label of philosophical ontology applies of course to multiple, often competitive research approaches. Among these, a special relevance has for our purposes the so-called *descriptive* or *realist ontology*. It seeks not explanation but rather a description of reality in terms of a classification of entities that is exhaustive in the sense that it can serve as an answer to such questions as: What classes of entities are needed for a complete description and explanation of all the goings-on in the universe? Or: What classes of entities are needed to give an account of what makes true all truths? Or: What classes of entities are needed to facilitate the making of predictions about the future? Sometimes a division is made – as for example in the case of Husserl and Ingarden – between formal and material (or regional) ontology. Formal ontology is domain-neutral; it deals with those aspects of reality (for example parthood and identity) which are shared in common by all material regions. Material ontology deals with those features (for example mind or causality) which are specific to given domains. If, as we shall argue, ontology must be multi-faceted, then there can be no sum of all material ontologies.

Philosophical ontology seeks a classification that is exhaustive in the sense that all types of entities are included in its classifications, including also the types of relations by which entities are tied together. In striving for exhaustiveness, philosophical ontology seeks a taxonomy of the entities in reality at all levels of aggregation (or, what comes to the same thing, at all levels of granularity), from the microphysical to the cosmological, and including also the middle world (the mesocosmos) of human-scale entities in between. Note that ontology as thus conceived is at odds with the attitude of reductionism, which sees reality in terms of some one privileged level of basic existents. Different schools of reductionism offer

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<sup>1</sup> Sometimes ‘ontology’ is used in a broader sense, to refer to the study of what might exist, where

different approaches to the selection of the basic existents. One large division is that between what we might call substantialists and fluxists, which is to say between those who conceive reality in terms of substances or things and those who favor an ontology centered on process or function or on continuous fields of variation. Most reductionists are nominalists, which is to say that they deny the existence of universals or multiply-exemplified entities and conceive the world as being made up exclusively of individuals.

Reductionists seek to establish the ‘ultimate furniture of the universe’. They seek to decompose reality into its simplest or most basic constituents. They thus favor a criterion of ontological economy, according to which an assay of reality is good to the degree to which it appeals to the smallest possible number of types of entities. The challenge is then to show that all putative references to non-basic entities can be eliminated in favor of entities on the basic level. The idea is that what is true on the basic level explains those phenomena which appear to obtain on the non-basic levels. The striving for explanatory unification supports reductionism.

Descriptive or realist ontology, in contrast, requires a stand-point of adequacy to all levels of reality, both basic and non-basic.<sup>2</sup> Reductionists seek to ‘reduce’ the apparent variety of types of entities existing in reality by showing how this variety is generated, for example through permutations and combinations of basic existents. The history of philosophical ontology is indeed marked by a certain trade-off between generativity on the one hand and descriptiveness on the other. By ‘generativity’ we understand the power of an ontology to yield new categories – and thus to exhaust the domain that is to be covered by ontological investigation – in some recursive fashion. By ‘descriptiveness’ we understand that feature of an ontology which consists in its

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‘metaphysics’ is used for the study of which of the various alternative possibilities is true of reality. See

reflecting, in more or less empirical ways, the traits or features of reality which exist independently of and prior to the ontology itself. It is generativity which gives an ontology its power to extend itself into new domains of entities; it is descriptiveness which ties an ontology to the world beyond.

All ontologists must find a way to combine as best they can the indispensable virtues of both generativity and descriptiveness. Philosophical ontology can then be enhanced by taking over elements from the methodology of reductionism, for example through the use of the axiomatic method illustrated also in the work of Lesniewski, Woodger, Goodman and others in formal mereology and illustrated also in Part 2 of Carnap's *Introduction to Symbolic Logic* (1958). Indeed in the course of the twentieth century a range of formal tools became available to ontologists for the formulation of their theories and for the evaluation of their formal qualities. Ontologists nowadays have a choice of formal frameworks (deriving from formal logic, as well as from algebra, category theory, mereology, set theory, topology) in terms of which their theories can be formulated. These new formal tools allow philosophical ontologists to express intuitive principles and definitions in a clear and rigorous fashion, and they can allow also for the testing of theories for formal consistency and completeness through the application of the methods of formal semantics.

It is the work of philosophical ontologists such as Aristotle, Ingarden (1964), Chisholm (1996)<sup>3</sup> which will be of primary importance for us here. Their work rests upon the realist presupposition that a single consistent ontological theory can comprehend the whole of reality at least at some high level of generality and abstractness. The taxonomies they propose are in many ways comparable to scientific taxonomies such as those produced by Linnaeus in biology or by Dalton in chemistry,

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Ingarden (1964).

though radically more general than these. All three of the mentioned philosophers are realist about universals, and all three transcend the dichotomy between substantialists and fluxists, since they accept categories of both things and processes, as well as other categories distinct from both of these.

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<sup>2</sup> Though contrast Mäki 2001, pp. 502 f.

<sup>3</sup> See also Simons 1987, Johansson 1989, Mulligan (ed.), 1992, Searle 1995.

## 2. Formal ontology: the logic of being

The term 'formal ontology' was first used by Husserl in his *Logical Investigations* (1913/1921), where formal ontological relations are in addition described as being 'independent of the peculiarity of any material of knowledge'.

This means that they are such as to apply, in principle, to any domain of reality whatsoever. Husserl himself provides a list of formal ontological categories, which includes items such as: object, state of affairs, unity, plurality, and so on.

These concepts are, like the concepts of formal logic, able to form complex structures in non-arbitrary, law-governed ways, so that by grasping the corresponding laws, we are able to grasp the properties of given structures in such a way as to establish in one go the properties of all formally similar structures. This holds in formal ontology just as it holds in formal logic and in mathematics (Smith 1989).

Formal ontology has been recently defined as "the systematic, formal, axiomatic development of the *logic of all forms and modes of being*" (Cocchiarella 1991).

Although the genuine interpretation of the term "formal ontology" is still a matter of debate, this definition is in our opinion particularly pregnant, as it takes into account both the meanings of the adjective "formal": on one side, this is synonymous of "rigorous", while on the other side it means "related to the forms of being"

Therefore, what formal ontology is concerned in is not so much the bare existency of certain individuals, but rather the rigorous description of their forms of being.

In practice, formal ontology can be intended as the theory of a priori distinctions:

- among the entities of the world (physical objects, events, regions, quantities of matter...);

- among the meta-level categories used to model the world (concepts, properties, qualities, states, roles, parts...)

In its current shape, formal ontology can be seen as the confluence between a school of thought which has addressed metaphysical problems within the mainstream of analytic philosophy, and another school more closely related to phenomenology, in the tradition of Brentano and Husserl.

The former school includes a multitude of philosophers, which roughly agree on the idea of "descriptive metaphysics" proposed by Strawson (Strawson 1959, Aune 1991)

The latter sees the philosophers of the so-called "school of Manchester" (Smith 1982, Smith and Mulligan 1983, Simons 1987, Mulligan 1992) as its principal defenders.

A fundamental role is played in formal ontology by *mereology* and *topology* (intended as the theory of the connection relation).

*Mereology* is the theory of parthood relations: of the relations of part to whole and the relations of part to part within a whole (Varzi, 2004). Its roots can be traced back to the early days of philosophy, beginning with the Presocratic atomists and continuing throughout the writings of Plato (especially the *Parmenides* and the *Thaetetus*), Aristotle (especially the *Metaphysics*, but also the *Physics*, the *Topics*, and *De partibus animalium*), and Boethius (especially *In Ciceronis Topica*). Mereology has also occupied a prominent role in the writings of medieval ontologists and scholastic philosophers such as Garland the Computist, Peter Abelard, Thomas Aquinas, Raymond Lull, and Albert of Saxony, as well as in Jungius's *Logica Hamburgensis* (1638), Leibniz's *Dissertatio de arte combinatoria* (1666) and *Monadology* (1714), and Kant's early writings (the *Gedanken* of 1747 and the *Monadologia physica* of 1756). As a formal theory of parthood relations, however, mereology made its way into modern philosophy mainly through the work of Franz Brentano and of his pupils,

especially Husserl's third *Logical Investigation* (1901). The latter may rightly be considered the first attempt at a rigorous formulation of the theory, though in a format that makes it difficult to disentangle the analysis of mereological concepts from that of other ontologically relevant notions (such as the relation of ontological dependence). It is not until Lesniewski's *Foundations of a General Theory of Manifolds* (1916, in Polish) that the pure theory of part-relations as we know it today was given an exact formulation. And because Lesniewski's work was largely inaccessible to non-speakers of Polish, it is only with the publication of Leonard and Goodman's *The Calculus of Individuals* (1940) that this theory has become a chapter of central interest for modern ontologists and metaphysicians.

Despite the possibility for *mereology* and *topology* to collapse one in the other in the case of a purely extensional domain limited to spatial or temporal entities, they need to be kept separate in order to characterize an entity independently of its spatio-temporal extension (Varzi 1994).

A standard reference for such issues is (Simons 1987), which presents in an accessible way the original formalizations of mereology made by Lesniewski and Goodman, discussing their limits and their possible extensions.

### **3. Building axiomatic ontologies for semantic interoperability**

Ontologies are nowadays considered as the basic infrastructure for achieving semantic interoperability. This hinges on the possibility to use shared vocabularies for describing resource content and capabilities, whose semantics is described in a (reasonably) unambiguous and machine-processable form. Describing this semantics, i.e. what is sometimes called the intended meaning of vocabulary terms, is exactly the job ontologies do for semantic interoperability, and, in particular, for the Semantic Web.

But what kinds of ontologies do we need? This is still an open issue. In most practical applications, ontologies appear as simple taxonomic structures of primitive or composite terms together with associated definitions. These are the so-called lightweight ontologies, used to represent semantic relationships among terms in order to facilitate content-based access to the (Web) data produced by a given community. In this case, the intended meaning of primitive terms is more or less known in advance by the members of such community. Hence, in this case, the role of ontologies is more that of supporting terminological services (inferences based on relationships among terms – usually just taxonomic relationships) rather than explaining or defining their intended meaning.

On the other hand, however, the need to establishing precise agreements as to the meaning of terms becomes crucial as soon as a community of users evolves, or multi-cultural and multilingual communities need to exchange data and services. As recently reported by the Harvard Business Review <sup>1</sup>, this problem may have been “one of the main reasons that so many online market makers have foundered. The

transactions they had viewed as simple and routine actually involved many subtle distinctions in terminology and meaning”.

To capture (or at least approximate) such subtle distinctions we need an explicit representation of the so-called ontological commitments about the meaning of terms, in order to remove terminological and conceptual ambiguities. A rigorous logical axiomatisation seems to be unavoidable in this case, as it accounts not only for the relationships between terms, but – most importantly – for the formal structure of the domain to be represented. This allows one to use axiomatic ontologies not only to facilitate meaning negotiation among agents, but also to clarify and model the negotiation process itself, and in general the structure of interaction.

We should immediately note that building axiomatic ontologies for these purposes may be extremely hard, both conceptually and computationally. However, this job only needs to be undertaken once, before the interaction process starts. The quality of meaning negotiation may drastically affect the trust in a service offered by the Semantic Web, but not the computational performance of the service itself. Thus, for example, a product procurement process involving multiple agents with distributed lightweight ontologies may be carried out in an efficient way by using simple terminological services, but the risk of semantic mismatch can be minimized only if the agents rely on explicit, axiomatised ontologies, which serve to ensure mutual compatibility of the respective models in such a way as to check the extent of real agreement.

Axiomatic ontologies come in different forms and can have different levels of generality, but a special relevance is enjoyed by the so-called foundational ontologies, which address very general domains.

Foundational ontologies are ultimately devoted to facilitate mutual understanding and inter-operability among people and machines. This includes understanding the reasons for non-interoperability, which may in some cases be much more important than implementing an integrated (but unpredictable and conceptually imperfect) system relying on a generic shared “semantics”.

In conclusion, we see the role and nature of foundational ontologies (and axiomatic ontologies in general) as complementary to that of lightweight ontologies: the latter can be built semi-automatically, e.g. by exploiting machine learning techniques; the former require more painful human labour, which can gain immense benefit from the results and methodologies of disciplines such as philosophy, linguistics, and cognitive science.

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