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Concept Systems and Ontologies: Recommendations for Basic Terminology

Gunnar O. Klein Department of Microbiology, Tumor and Cell Biology, Karolinska Institutet
gunnar.klein@ki.se

Barry Smith Department of Philosophy and Center of Excellence in Bioinformatics and Life Sciences,
University at Buffalo
phismith@buffalo.edu

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Summary

This essay concerns the problems surrounding the use of the term “concept” in current ontology and terminology research. It is based on the constructive dialogue between realist ontology on the one hand and the world of formal standardization of health informatics on the other, but its conclusions are not restricted to the domain of medicine. The term “concept” is one of the most misused even in literature and technical standards which attempt to bring clarity. In this paper we propose to use the term “concept” in the context of producing defined professional terminologies with one specific and consistent meaning which we propose for adoption as the agreed meaning of the term in future terminological research, and specifically in the development of formal terminologies to be used in computer systems. We also discuss and propose new definitions of a set of cognate terms. We describe the relations governing the realm of concepts, and compare these to the richer and more complex set of relations obtaining between entities in the real world. On this basis we also summarize an associated terminology for ontologies as representations of the real world and a partial mapping between the world of concepts and the world of reality.

1. Introduction

In recent years Smith and other realist philosophers such as Ingvar Johansson have been challenging established standardization endeavours in Health Informatics dealing with what are called “concepts”. Bodenreider *et al.*, Schulz and Jansen, Spackman and Reynoso, Ceusters *et al.* and others have also contributed to these discussions [Bodenreider 04, Schulz 06, Spackman 04, Ceusters 06], the results of which are summarized in a series of papers by Smith and his co-workers, in which it is argued that the concept of “concept” is used in much of the work on biomedical terminologies, ontologies and controlled vocabularies in ways that are seriously flawed.¹ The main critique has been that the

term “concept” in these standards is used in multiple ways. It is often unclearly defined (or not defined at all). Where it is defined it is often viewed as signifying the result of some cognitive process, for example in the form of *knowledge in the mind of an expert* or *an entry in a terminological system*. Problems then arise, because attempts are simultaneously made to use the same term for purposes of reasoning among relationships between entities in the real world.

Most recently, prominent former advocates of the concept-based approach have recognized some of the merits of this critique, as for example Cimino, who argues that both concepts and universals ‘be embraced and can co-exist peacefully in controlled terminologies’ [Cimino 06], and Solbrig and Chute, who argue that ‘the use of the term “concept” as the name of a class in a model can introduce serious confusion’ [Solbrig 09].

In what follows we describe an irenic proposal to overcome these confusions. The authors of this communication agree that the term “concept” has been misused in, many influential writings, but we accept that this term can still serve an important role in modern health informatics if only it is

¹ For those involved in terminology standardization as it applies to ISO’s Technical Committee 37 founded by the Austrian terminologist Eugen Wüster and to its successor institutions, an item of specific interest is the paper by Smith, Ceusters and Temmerman entitled “Wüsteria” [Smith 05c]. In this paper it is argued that Wüster’s influence led to a pervasive confusion between concepts and entities in reality. One reason to bring this up here is the recent discussion of the draft EN 15822 *Health informatics – Categorical Structure for Anatomy* in health informatics circles. Another is the growth in significance of the HL7 Reference Information Model, which Smith and Ceusters see as a particularly egregious embodiment of the Wüsterian confusion, with serious consequences in the form of failed health informatics projects involving major investments by national

governments [Smith 06c]. For an overview see: <http://hl7-watch.blogspot.com/>.

properly used. We agree also that there is a need to have a serious and constructive discussion on how to resolve the problems which have arisen through its misuse. We propose a set of terms and definitions that we believe should replace the single term “concept” in those specialist contexts where information structures and semantic interoperability is the concern. We have of course no ambition to change the very frequent and varying uses of the term “concept” in natural language discourse.

While one of us, Klein, is an M.D. and has his main function in Health Informatics – hence the use of many examples from the health area – there is nothing specific to the healthcare domain about the problems and solutions discussed.

2. Concept – A strange animal with many heads

2.1 The history of the use of the term “concept”

The term “concept” has a long history, going back at least as far as Plato. For present purposes it is important to refer to the great medieval dispute between realists, conceptualists and nominalists over the so-called “problem of universals” [Klima 03].

Realists hold that there are universals – invariant patterns (also called characters or essences, corresponding to common nouns such as ‘electron’ or ‘molecule’ used in the expression of scientific laws) – existing on the side of entities in reality, and that it is in virtue of such universals that particulars – for example these two particular triangular shapes in Figure 1 – manifest relations of similarity to each other:

According to the realist view, such relations of similarity would exist even if there were no cognitive subjects in a position to observe them. Each single universal can be exemplified multiple times by an open-ended plurality of particulars. Universals are further organized into trees, in which universals of greater and lesser generality – called *genera* and *species*, respectively – are linked together via subtype (also known as *is_a*) relations. All universals are distinguished from the particulars in reality which are also called “instances” (see further below on terminology for ontologies).² The relation of similarity between the illustrated instances exists because the *same universal* is instantiated by each of the given individual shapes

Conceptualists, in contrast, hold that there are no universals on the side of entities in reality, but rather only in our minds. One and the same general concept, say *triangle*,



Fig. 1 Two particular triangles are instances of the universal *triangular shape*

can be related to a plurality of triangles in reality. Different cognitive subjects can share the same general concepts, which are in this sense multiply exemplified in different minds. But conceptualists do not believe that a person’s concepts correspond to corresponding universals or invariants on the side of reality. Rather, for the conceptualist all concepts relate to their instances in only *ad hoc* ways; concepts in general are thus treated as if they were all *ad hoc* concepts along the lines of: *things you might take on a holiday* or *things you might need to build a weapon*.

Nominalists, finally, hold that there are universals neither in reality nor in our minds, but rather only general terms (words, expressions). Nominalists thus deny the existence of general concepts which can be shared by a plurality of cognitive subjects. General terms are mere labels for *ad hoc* collections of particular things or events. When different subjects apply the same general term, say “triangle”, to each of the two particulars depicted above, then, according to the nominalist their respective ideas have just as little objective (*de re*³) similarity to each other as do the entities in reality to which general terms are applied.

2.2 Towards disambiguation

Traces of all three of these positions are present in contemporary uses of the term “concept” in ontology and terminology circles. Thus in some contexts the term “concept” refers to what would more properly be called a “universal” in the sense of the realist doctrine; in some contexts it refers to general ideas in people’s minds; and in yet other contexts it refers merely to general terms in some controlled language. Contradictions then arise because such distinct readings are not clearly distinguished in the relevant literature [Smith 04].

To make matters worse, psychological, linguistic and computational uses of the term “concept” have in more recent years also been added to this mix, so that there are today a number of different viewpoints developed for different, yet often related, purposes where the term “concept” has been applied, often unconsciously, with very different meanings. Further confusions arise because the term is sometimes used with a meaning that is left unspecified or with different and

² Confusingly, some medieval realist philosophers used expressions like “general concepts” and “general terms” to refer to universals on the side of reality.

³ The latin term “*de re*” in philosophy signifies “of or regarding the thing” as opposed to “*de dicto*” which means “of or regarding the word (term)”

contradictory meanings in one and the same text. In a systematically multidisciplinary endeavour like health informatics and in other fields of informatics or terminology research matters are made more complicated by the fact that, even where the term “concept” is used in the literature of one specialist community with a clear and consistent meaning, readers from other involved communities will often import their own expectations as to what this term means, in ways which generate now well-documented [Smith 04, Smith 05c] confusions.

Instead of abandoning the term completely, as has been suggested in some circles, we offer here a careful analysis of the different meanings of “concept” and then propose separate terms and definitions to ensure disambiguation. Our proposals thus go beyond the minimal requirement that authors should be careful to provide in every case a clear indication of what they mean by “concept” on all occasions of use. Confusions can be seen to arise even where all of those involved confirm their intention to adhere to this requirement. We thus go further in holding that the different meanings which have come to be associated with the term “concept” should henceforth be marked by systematic use of different terms in order to convey the corresponding different meanings. We realize that these proposals may not be intuitively clear and will at first seem unfamiliar – most of these working in information technology have, after all, been educated against a background in which the term “concept” is so familiar as to be unquestioned. At the same time we are convinced that urgent reforms are required if the multiple problems already identified are to be remedied in a timely manner. Hence if our specific proposals are found to be for whatever reason unsatisfactory, then we invite those involved in terminological research to suggest alternatives.

The main focus of this communication is systematic terminological work in fields like biomedicine, where there arises the need to develop systematic representations of real world entities such as disorders or anatomical structures [Smith 07a]. Hence part of our goal is to support also the development of such systematic representations – called “ontologies” – in such a way that they can be used for information integration and alignment and also for automatic reasoning.

The heart of our proposal, simply put, is that “concept” should be used exclusively to refer (1) to *the meaning of a corresponding general term*, this meaning being (2) unique and (3) agreed upon by responsible persons in the given disciplinary field. This view is the position neither of the nominalist nor of the conceptualist but it is proposed here as a resolution to the confusion caused by the different uses of the term and should in principle be acceptable by all the three

philosophical schools mentioned in Section 2.1. This means, in our opinion, that in those areas of health informatics where there already exist terminological standards pertaining to the use of the term “concept” these standards should be revised in future revisions of these standards.⁴

Consider, for example, how our proposal would apply to the treatment of terms such as “mandate” as used for example in EN 13940-1 Health informatics – System of concepts to support continuity of care – Part 1 Basic concepts [EN 07]. When dealing with human constructions such as mandates, agreements, contracts and the like, there is an obvious distinction between the entity referred to on the one hand (i.e. the mandate in question), and the meaning of the term (“mandate”) which is used to refer to this entity. Only the latter, then, would be a candidate for being identified as a concept, in the sense of our proposal.

2.3 On psychological uses of the term “concept”

In natural language, and in some of the work of Eugen Wüster and other influential terminologists, the term “concept” is used to mean what would more properly be called a “mental concept”, “idea” or “thought” (“noesis” in Aristotelian terminology), which may itself be conceived as a certain state of the brain of some individual – a state which may be evoked by the use of a corresponding general term. This idea forms the basis of the famous Semiotic Triangle idea in the specific form which it was given by Ogden and Richards in 1930 (see Figure 2).

Certainly we have no objection to the technical use of “concept” in psychological contexts and in related contexts, for example of psycholinguistics. This technical usage is not, however, an important connotation for purposes of standardization in the domain of terminology systems and ontologies. We mention it only in order to point out the need

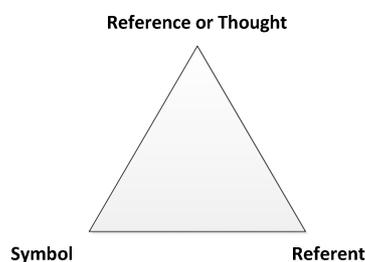


Fig. 2 The semiotic triangle of Ogden and Richards (simplified)

⁴ This refers e.g. to the ISO 17115: 2006 Health informatics – Vocabulary for terminological systems and the EN 12264:2005 Health informatics – Categorical structures for systems of concepts both which refer to the basic ISO/TC 37 standard ISO 1087-1:2000 Terminology work – Vocabulary – Part 1: Theory .and application.

to separate common natural language and technical scientific interpretations from the meaning of “concept” that we believe should be recommended in the context of information systems. This need is all the more urgent given the influence of Wüster’s ideas on the literature of terminology standardization.

3. Recommendations regarding the use of “concept” and related terms

We recommend that the listed terms be used in terminological contexts exclusively in the following ways:

Term: Concept

concept in a terminological system (synonym 1)
agreed meaning of a term (synonym 2)

Definition: meaning of a term as agreed upon by a group of responsible persons

Note 1: The assumption here is that this meaning (for example of a term such as “nephron” or “influenza”) will be agreed upon in virtue of the fact that it is accepted and understood by the members of the relevant community, e.g. within a clinical specialism or scientific discipline.

Note 2: The current version of the International Standard ISO 1087-1 (dating from 2000) [ISO 00] defines a concept as: a unit of knowledge created by a unique combination of characteristics. We prefer “meaning” to “unit of knowledge”, for a number of reasons. First, there can be agreed meanings for terms like “unicorn” which do not correspond to any unit of knowledge, since there are no corresponding entities in reality about which knowledge could be gained. We also prefer “meaning” to “unit of thought”, in order to avoid the psychological connotations of the latter. Our investigations suggest that the term “characteristic” is subject to the same sorts of problems as have affected the term “concept”.

Note 3: The fact that an identified group of persons (those responsible for the establishment of a given terminological system) share a common understanding of the meaning of a term – typically captured by means of a definition – is important in those contexts where the term “concept” is used today in modern informatics because this is what distinguishes concepts as we shall here understand them from ideas in the minds of individual cognitive subjects. In particular, the existence of agreed meanings on the part of responsible persons is clearly indispensable for the development of formalized standards, including international standards.

Note 4: Philosophers differ as to what is meant by “meaning”. Sometimes this term is defined as meaning: that which remains constant when a word in one language is

translated correctly into another language. Sometimes operational definitions are provided on the basis of the view that persons demonstrate that they share command of a common meaning for a given term when they demonstrate that they have the ability to use this term in the same way – for example, that they have the ability, upon receiving information containing a corresponding term, to associate it with the same referents.

Note 5: The term “concept”, on the reading “agreed meaning of a term”, refers to this meaning itself and thus not to any specification of this meaning in some natural or artificial language or in some formal model for example in the form of a definition. One and the same concept (in the sense of “agreed meaning”) will typically correspond to several alternative ways of expressing this meaning, and thus to different linguistic expressions in the same as well as in different languages. Moreover, definitions are not always required, since some terms must in any given terminological system be specified as primitives. Terms such as “up” or “down” are so well understood by all potential users of a terminology that they need no definition. In other cases meanings must be specified in the context of the pertinent terminological system, and there are two possible ways to do this: by the linguistic representation of a definition, or by a translation of the corresponding general term into some other language in a way which is, in the specific context of the terminological system in question, able to convey a sufficient understanding of the given meaning. The provision of a definition in the form of a statement of necessary and sufficient conditions is of course preferable wherever this is possible. However, sometimes we can only use *ostensive* definitions, as for example in the case of a term like “SARS”, for which we were initially able to specify only that it referred to a syndrome which a certain defined set of individual patient cases shared in common. (Note the way in which an ostensive definition of this sort points to a universal in reality, i.e. to a certain multiply exemplified entity.)

Term: Concept definition

concept definition representation (synonym1)
definition (synonym2)

Definition: specification of a concept (i.e. of the agreed meaning of a term) by means of a descriptive statement or a formal expression which serves to differentiate it from other concepts

Note: There may be more than one definition which captures the same agreed meaning. While the definition of a concept (in the sense of: agreed meaning of a term) will most often take the form of a linguistic expression, such a definition may also be expressed by graphical or other means

Term: Concept system

Definition: collection of representations of concepts structured by means of representations of relations

Note 1: A concept system is a collection of elements (called concept system nodes) which are related together via interconnections representing relations such as *narrower_than* and *broader_than* between the corresponding meanings.

Note 2: Graphical representations such as directed acyclical graphs (DAGs), UML or Venn diagrams may serve the definition of a concept by showing its interrelations with other concepts.

Term: Concept system node

Definition: information element within the structure of a concept system that is a pointer linking one or several synonymous terms with a given concept definition and linked to other such information elements in the representation of relations between the corresponding concepts

Note 1: In many modern concept systems designed for use with information systems, the concept system node has as one key component a numeric concept identifier.

Note 2: Where a concept system exists in graph-theoretical form, the concept system nodes are the vertices of the graph. The edges of the graph then represent relations between the concepts represented by the corresponding nodes.

Note 3: When SNOMED CT, for example, uses the term “concept”, then we believe that what it means is “concept system node” in the terminology advanced here. Certainly SNOMED officially defines “concept” as meaning: “unit of thought”. When we inspect its actual practice, however, which includes for example the use of what are called “navigational concepts” such as “infectious organism”, then we discover that concepts in SNOMED CT are used as pointers which allow the capture of relationships of synonymy between terms (which SNOMED CT calls “descriptions”) and also certain “association” relationships with other concept nodes as well as with associated attributes. This use of “concept” to mean what we are calling “concept system node” is also the most common (if often not always clearly formulated) usage in information model standards for Electronic Health Records or other health informatics standards such as the HL7 RIM or CEN message standards.

4. Recommendations regarding treatment of relations between concepts

ISO 1087-1 [ISO 00] and many other works on concepts specify a number of different types of *relations* between

concepts. A concept, on our proposal, is the meaning of a term. Thus no concept is any kind of real world entity of the sort to which reference might be made using the corresponding general term (e.g. “nephron” or “influenza”). The concept *influenza* is not a disease. The concept *influenza* also cannot stand in any relations such as *caused_by* or *treated_by*. Thus there are in fact rather few relations which should be used to link concepts, effectively only namely the relations *narrower_than* and its inverse *broader_than* which obtains between superordinate and subordinate concepts. In ISO 1087-1 this is defined as:

“generic relation” = def. relation between two concepts (3.2.1) where the intension (3.2.9) of one of the concepts (3.2.1) includes that of the other concept (3.2.1) and at least one additional delimiting characteristic (3.2.7)”

This generic relation between concepts can be called a **semantic relation**, in order to stress the fact that it is a relation which has agreed meanings as its relata. In what follows we shall refer to it by means of the compound phrase “*is_a* (is narrower in meaning than)”.

What ISO1087-1 calls “partitive” and “associative relations” (such as *part_of* or *causes*) are not appropriate for concept systems – since they are not relations which hold between meanings. Such relations should be used, rather, in ontologies (see below), in which real world entities and their interrelations are taken into account.

5. Terminology for ontologies

5.1 Entities

If concept systems are systems of meanings, then we need a supplementary terminology for those representation systems which relate to real world entities, both those investigated by the natural sciences (for example cells or electrons) and those existing in administrative domains (such as mandates or documents recording lab results). Unfortunately this terminology – the terminology of ontology – is not yet established in a consistent way in informatics and terminology circles.

What we propose here reflects an emerging consensus in ontological research; but we also provide alternative synonyms (in parentheses) to serve as guidance for the wider community. Our remarks supplement the proposals advanced in [Smith 06b], which are in turn being adopted by the ontologies developed by the OBO Foundry (<http://obofoundry.org>), a consortium of influential ontology groups in the bio-sciences.

5.2 Instances and types

The entities in reality are of two kinds: “instances” and “types”, for short: see Figure 3. Alternative terms for what we here call “type” are “universal”, which is used frequently in realist philosophy and modern ontology, and also: “class”, “kind”, “category”, “genus”, “species”, “taxon”.

What we here propose to call “instance” has in the ontology literature also been called “individual” or “particular”. These terms can be regarded as synonyms in this context, but we prefer the term “instance” since it draws attention to the fact that the entities in question are *instances of* corresponding *types*. Thus the particular cell in this Petri-dish is an *instance of* the type *cell*, as also of the type *B-lymphocyte*, and so on.

5.3 Continuants and occurrents

Entities (instances and types) can be further classified according to the following scheme focusing on their persistence, dividing them into two main kinds *occurrents* and *continuants*, Figure 4.

5.4 Relations between real-world entities

Individual instances can have various relations to other instances. For example Mary’s heart is part of Mary; Mary’s run is part of Mary’s morning work-out, and so on. In some cases *all* instances of a given type stand in such relations to correlated instances of some other type. Thus all instances of the type *influenza* are also instances of the type *infectious disease*. All instances of the type *adult* are identical to some instance of the type *child* existing at some earlier time. All

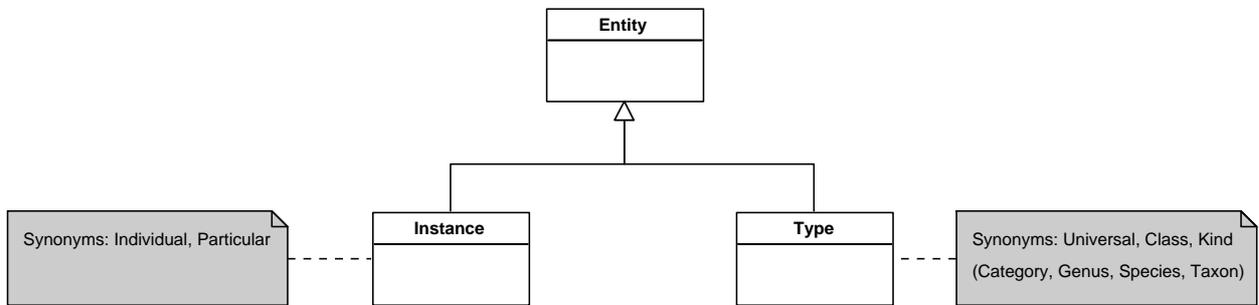


Fig. 3 The two basic kinds of entities

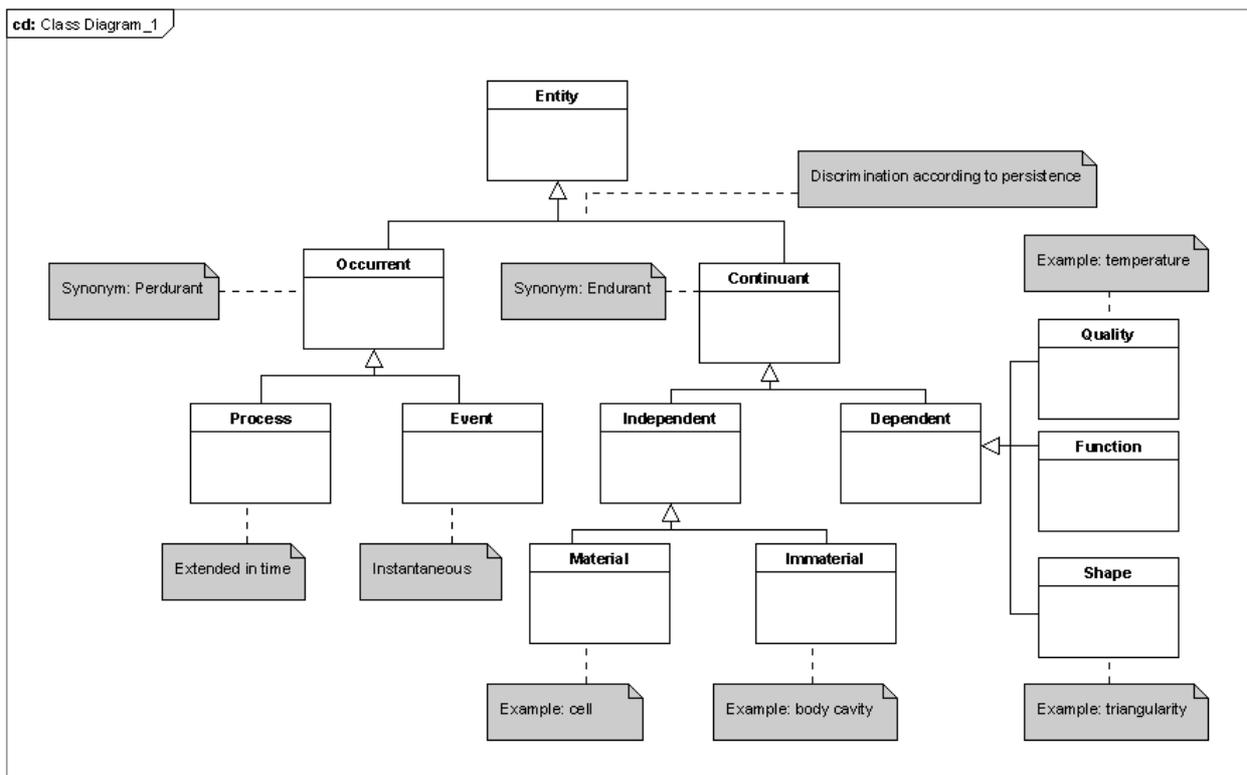


Fig. 4 A top level ontology of entities (provided for purposes of illustration)

instances of the type *nucleus* are adjacent to some instance of the type *cytoplasm*, and so on. Such relations are themselves such as to obtain universally, in the sense that they hold of all instances of any given type (namely, in each case, of the type first mentioned). They often do not hold when inverted (thus it is not the case that all instances of the type *cytoplasm* are adjacent to some instance of the type *nucleus*) [Smith 05a].

The representation of such universal relations between types is the ideal to be approximated to on the realist conception of ontology. Before formulating our proposed definition of “ontology”, however, we need to say something further about universals and their extensions in reality, and also about those general terms – such as “tall Finnish spy” or “diabetic patient in Leipzig on March 2, 1997” – which, on the view here in question, do not refer to types.

We introduce first of all the technical term “collection of instances”, which refers to something that is itself an instance containing other instances as its members. Some of the problems we face turn on the fact that general terms are sometimes used to refer to types, and sometimes to the corresponding collections of instances. (Thus in referring to the protein Lmo-2 we may be referring to the scattered collection of all Lmo-2 molecules or to the corresponding molecule type [Schulz 06]).

We propose that the term “class” be used to refer to collections of instances which are *maximal* in the sense that they comprehend all and only the entities to which a given general term applies. Where the general term in question refers to a type, then the corresponding maximal collection is the *extension* of this type. This class contains all and only those instances which as a matter of fact instantiate the corresponding type at the given time.

Clearly, now, the totality of classes is wider than the totality of extensions of types, since it includes also more or less *ad hoc* or *defined* classes designated by terms like “employee of Swedish bank”, “daughter of Finnish popstar” and so on.

5.5 Recommendations concerning terminology for ontologies

We can now define the term “ontology” in a way which is, we believe, in close conformity with a common consensus use of this term on the part of those working on ontologies in support of natural science and in associated clinical and translational research, as follows:

Term: ontology

Definition: a representational artifact, comprising a taxonomy as proper part, whose representational units are intended to designate some combination of types, classes, and certain relations between them

Term: realism-based ontology

Definition: an ontology built out of terms which are intended to refer exclusively to types and which correspond to that part of the content of a scientific theory that is captured by its constituent general terms and their interrelations.

A realism-based ontology is then a type system and relates to types in something like the way in which a concept system relates to concepts. Much of biomedical knowledge, for example as contained within textbooks, is about the systematization of the universally obtaining relations between types instantiated by real-world instances. Data in medical records, on the other hand, is often a matter of instance-level relations between the corresponding instances, for instance data to the effect that traces of this chemical are located in this blood sample here and now. Where designations of types (for example via clinical codes) are used in expressions of such data, then this is to classify the corresponding instances.

6. Correspondence between concept systems and ontologies

Some concepts – and very many of those concepts used in biomedical terminology systems (more properly called “concept systems” in light of the above) – have a relation to entities in the real world which is closely analogous to the relation between types and their instances. Mary is an *instance* of the type *human being*, but Mary also *falls under* the concept *human being*. The type *human being* stands in an *is_a* (is a subtype of) relation to the type *mammal*. But the concept *human being* stands in an *is_a* (is narrower in meaning than) relation to the concept *mammal*.

In spite of this parallelism, however, the term “concept” should still never be used in place of “type” (“universal”) as thus defined, because the parallelism is only partial. First, there are concepts (understood in the sense of our proposal), for example those associated with terms like:

- case of pneumonia in Russian fiction
- fractured lip
- surgical procedure not performed because of patient request
- absent scrotum

which correspond to no real-world entities on the instance level. Second, there are concepts such as:

- non-rainy day
- non-mammal
- relative of possible smoker
- other metalworker in New Zealand
- person admitted before 9a.m.
- mixture of water and alcohol containing zero amount

of alcohol

- leukemia without mention of remission⁵

which refer to no real-world entities on the level of types. Rather, at best, they refer to *ad hoc* or *defined* classes of one or other sort.

Thus only some subset of the nodes in a given biomedical concept system will be mappable in a 1-1 way to corresponding nodes in a type system or realism-based ontology.

6.1 Types, concepts and relations

Types and *concepts* should be kept clearly separate also because of the different ways in which they are connected by relations.

Certainly some of the nodes in concept systems stand in *is_a* (is narrower in meaning than) relations in ways which are isomorphic to the *is_a* (is subtype of) relations which hold between the nodes in a corresponding ontology. But there are many more *is_a* relations of the former type than of the latter. There are also many non-*is_a* (is subtype of) relations connecting types in ontologies that have no application to concept systems, including all the familiar relations *part_of*, *transformation_of*, *located_in*, *derives_from*, *adjacent_to*, *participates_in*, and so on.

Concept systems are thus simple hierarchies, whose nodes are joined together exclusively by *is_a* (is narrower in meaning than) relations. Ontologies typically manifest much more complex graph-theoretic structures, in which many further relational edges are included. On the other hand concept systems may be much richer, since they may include many nodes which correspond to no types on the side of reality.

7. Conclusion

There is room, as we hope is now clear, for both concept systems and ontologies, and we anticipate that, because they address different sorts of purpose, both sorts of information artifact will be needed in the future. We hope, however, that the current confusions which pervade the field of information standards will, in light of our remarks in the above, finally be addressed, so that these different purposes can be addressed more successfully in the future.

⁵ Odd terms such as “Leukemia without mention of remission” appear in coding systems in order to ensure non-redundancy of coding. Without such terms, patients coded with “leukemia” and patients coded with “leukemia with remission” would be counted twice. The problem is that the terms in question do not correspond to the way clinicians and biologists think about the corresponding phenomena on the side of the patient. Moreover, they introduce an inherent element of fragility in the coding, since what is and is not mentioned in a given record will of course change with time.

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[担当委員 : 来村 徳信]

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Authors' Profiles



Klein, Gunnar O.

Gunnar O Klein is a medical informatics researcher at the Centre for Medical Terminology, Dept of Microbiology, Tumour and Cell Biology, Karolinska Institutet, which is the medical university of Stockholm Sweden. He has been a leader of standardization of Health Informatics, as a chairman of the European Committee CEN/TC 251 1997–2006 and as a founder and contributor to the international ISO/TC 215 committee for health informatics. In ISO he is currently leading the work on a standard for Metadata of clinical knowledge resources. He has also been working for the co-operation between all relevant standards organisations for Health information and the World Health Organization through the eHealth Standardization Co-ordination Group which he chairs. He has also been a member of several European union research projects related to interoperability of health records, including the Network of Excellence Semantic interoperability and Data Mining where Klein and Smith first started to work together. In addition to the work in medical informatics, Klein has been conducting research in Tumour Immunology and has been a practising physician.



Smith, Barry

Barry Smith is a prominent contributor to both theoretical and applied research in ontology. He is the author of some 450 scientific publications on ontology and related topics, and editor of *The Monist: An International Quarterly Journal of General Philosophical Inquiry*. His research has been funded by the National Institutes of Health, the US, Swiss and Austrian National Science Foundations, the Volkswagen Foundation, and the European Union. In 2002 he received in recognition of his scientific achievements the Wolfgang Paul Award of the Alexander von Humboldt Foundation. Smith is SUNY Distinguished Professor and holder of the Julian Park Chair of Philosophy in the University at Buffalo (New York, USA). He is also Research Director of the Institute for Formal Ontology and Medical Information Science in Saarbrücken, Germany. He studied at Oxford and Manchester, and has held faculty positions in Sheffield, Manchester, Liechtenstein and Leipzig. Smith's primary research focus is the application of ontology in biomedicine and biomedical informatics. He is a Coordinating Editor of the OBO (Open Biomedical Ontologies) Foundry initiative, a principal scientist of the National Center for Biomedical Ontology, a member of the Scientific Advisory Board of the Gene Ontology Consortium and a PI of the Protein Ontology and Infectious Disease Ontology projects. He also collaborates with Hernando de Soto, Director of the Institute for Liberty and Democracy in Lima, Peru, on the ontology of property rights and social development.