

Forthcoming in Achille Varzi and Laure Vieu (eds.), *Proceedings of FOIS 2004*.  
*International Conference on Formal Ontology and Information Systems*, Turin, 4-6 November 2004

# The Ontology-Epistemology Divide: A Case Study in Medical Terminology

Olivier BODENREIDER<sup>1</sup>, Barry SMITH<sup>2,3</sup>, Anita BURGUN<sup>4</sup>

<sup>1</sup> *US National Library of Medicine, Bethesda, Maryland, USA*

<sup>2</sup> *Institute for Formal Ontology and Medical Information Science, Saarbrücken, Germany*

<sup>3</sup> *Department of Philosophy, University at Buffalo, New York, USA*

<sup>4</sup> *Laboratoire d'Informatique Médicale, Université de Rennes I, France*

**Abstract.** Medical terminology collects and organizes the many different kinds of terms employed in the biomedical domain both by practitioners and also in the course of biomedical research. In addition to serving as labels for biomedical classes, these names reflect the organizational principles of biomedical vocabularies and ontologies. Some names represent invariant features (classes, universals) of biomedical reality (i.e., they are a matter for ontology). Other names, however, convey also how this reality is perceived, measured, and understood by health professionals (i.e., they belong to the domain of epistemology). We analyze terms from several biomedical vocabularies in order to throw light on the interactions between ontological and epistemological components of these terminologies. We identify four cases: 1) terms containing classification criteria, 2) terms reflecting detectability, modality, uncertainty, and vagueness, 3) terms created in order to obtain a complete partition of a given domain, and 4) terms reflecting mere fiat boundaries. We show that epistemology-loaded terms are pervasive in biomedical vocabularies, that the “classes” they name often do not comply with sound classification principles, and that they are therefore likely to cause problems in the evolution and alignment of terminologies and associated ontologies.

## 1 Introduction

### 1.1 Biomedical terminology

The purpose of biomedical terminology is to collect the names of substances, qualities and processes employed in the biomedical domain both by practitioners and in the course of biomedical research. Biomedical terminology reflects not only the various subspecialties of biomedicine (roughly corresponding to specialized subdomains or dimensions of biomedical reality), but also the many purposes for which terminologies are developed. Specialized terminologies include SNOMED CT<sup>1</sup> for clinical medicine, the Foundational Model of Anatomy<sup>2</sup> for anatomical structures, the International Statistical Classification of Diseases and Related Health Problems<sup>3</sup> (or International Classification of Diseases, for short) for health disorders, the Current Procedural Terminology<sup>4</sup> for medical procedures, and the

---

<sup>1</sup> <http://www.snomed.org/>

<sup>2</sup> <http://fma.biostr.washington.edu/>

<sup>3</sup> <http://www.who.int/whosis/icd10/>

<sup>4</sup> <http://www.ama-assn.org/ama/pub/category/3113.html>

Gene Ontology<sup>TM5</sup> for molecular biology. Most terminologies were originally developed to serve a particular purpose. The Medical Subjects Headings<sup>6</sup> (MeSH) is the controlled vocabulary used for indexing the biomedical literature at the US National Library of Medicine, a purpose analogous to that of the Gene Ontology, which is used to ‘annotate’ (characterize, index) genes and gene products. The very names of some terminologies reflect their purpose clearly. This is the case, for example, of the Alternative Billing Concepts terminology. The International Classification of Diseases (ICD-9-CM) evolved out of a terminology for compiling mortality and morbidity statistics but now constitutes a controlled vocabulary used by the insurance industry for reporting claims. We show that in many cases biomedical terms are crafted not only for naming the classes of entities found in biomedical reality, but also to represent additional information. In this paper, we are particularly interested in the intrusion of epistemology into biomedical terminology.

## 1.2 Terms as names for biomedical classes

It is often said that there is nothing that cannot be encountered in the domain of medicine. Deviations are everywhere. Thus anatomy as described in textbooks corresponds to canonical anatomy; it represents some kind of idealized structure to which no actual human body fully corresponds. It is essentially impossible to describe disease manifestations without resorting not only to lists of associated signs and symptoms but also to the frequency distributions of the latter for each particular disease. In addition to the most common, prototypical form of the disease, there are many clinical variants in which some of the common manifestations are missing and other, less frequent manifestations take their place.

In this context of high variability, it is not surprising that names are crafted to represent not only the prototypical classes but also the many possible variants. Thus names are formed that include information identifying specific clinical variants, or information about associated lesions or injuries. The default assumption on the part of those working with, and on, terminologies, is that such specially crafted terms correspond to classes of entities found in biomedical reality in just the same sense as do more straightforward terms such as *meningitis* or *fever*.

This assumption takes many forms, and on the weakest possible reading it consists in the thesis that every term used in clinical practice or in biomedical research is *ipso facto* to be accepted as designating a corresponding ‘class’ or ‘concept’ – whereby the corresponding classes or concepts are then not always conceived as existing on the side of entities in reality but rather as being themselves linguistic entities, correlates of the terms with which they are associated. In what follows, however, we will provide evidence to the effect that only some types of variant terms represent classes (universals) in reality, and that others are in fact disguised assertions about such genuine classes which are formulated as terms merely in order to meet current practical requirements of coding.

Genuine classes are supposed to reflect the categorization principles proposed by Rosch [5] and Tversky [8]. They define resemblance between categories as maximizing the sum of all the common features within a category minus the sum of the measures of all of the distinctive features. Categories must also reflect the perceived world structure.

---

<sup>5</sup> <http://geneontology.org/>

<sup>6</sup> <http://www.nlm.nih.gov/mesh/>

### 1.3 *Ontology vs. epistemology*

Ontology, for the biomedical informatics purposes which are of concern to us here, starts out from the idea that there are invariants in reality – here called ‘classes’ or ‘universals’ – which are captured in the general terms used in the textbooks of biological science and which are instantiated by particular examples or cases of such classes, whether these be organisms or organism parts, qualities, functions, processes, diseases or symptoms. Ontology is then the study of such classes and of the relations between them, for example of the *is\_a* relation which obtains between two classes when it is a matter of scientific law that all instances of the first class are instances of the second, or the *part\_of* relation which obtains between two classes when it is a matter of scientific law that instances of the first exist always as parts of instances of the second.

Epistemology in the strict sense is the study of how cognitive subjects come to know the truth about given phenomena in reality – for example that they instantiate given classes or universals. In the sense that is relevant to our present purposes here, epistemology is the study of biological or medical knowledge. Thus it encompasses the ways in which physicians come to know about the existence of given diseases in given patients.

In this paper, we examine the degree to which biomedical terms are created to represent not instances or classes in reality but rather features reflecting our knowledge or ignorance of such instances or classes. We identify four such cases for which we present examples drawn from medical vocabularies: 1) terms containing classification criteria, 2) terms reflecting detectability, modality, uncertainty, and vagueness, 3) terms created in order to obtain a complete partition of a given domain, and 4) terms reflecting mere fiat boundaries.

## 2 **Terms containing classification criteria**

Compound biomedical terms are often generated from simpler terms by adding qualifiers representing classification criteria. In many cases, such specially created variant terms do not represent classes of entities in reality which are distinct from the classes represented by the corresponding root terms: rather, they represent the same underlying reality but expressed in slightly different ways. Sometimes, variant terms do not make ontological sense at all: they do not represent special classes in reality but are rather such as to convey other sorts of information. In other cases, however, variant terms do refer to corresponding classes, and to classes which are distinct from those referred to by the underlying root terms. For example where the presence or absence of a manifestation is a key element for distinguishing between different diseases or different forms of a disease.

### 2.1 *Variation*

Let us take *febrile seizure* and *afebrile seizure* to illustrate ontologically valid variants. (*Febrile* in this context means ‘fever-related’.) Seizures occur when the normal pattern of neuronal activity becomes disturbed, causing convulsions (among other things). While seizures are one possible manifestation of epilepsy, they are also common in young children exposed to fever (e.g., after receiving immunization shots). Seizures occurring in the context of fever in children are called *febrile seizures*. They can be thought of as a transient overreaction of brain neurons to fever and are distinct from *afebrile seizures* where an underlying inherent condition (e.g., a brain lesion) may cause the seizures. Here, taking account of the presence or absence of a manifestation (fever) in the name of a disorder reflects an ontologically valid distinction as the two kinds of seizures are distinct in their ori-

gin and also have a different prognosis and treatment. Note however that while *afebrile seizure* is a term which serves its epistemological purpose in distinguishing seizure with underlying inherent cause from seizure triggered solely by fever, it can be objected to the given term that it does not capture the (positive) essence or nature of the disease in question, which might more properly be called precisely: seizure with underlying inherent cause.

A similar phenomenon can be observed outside the domain of disorders. Before the recent era of molecular biology, the identification of micro-organisms relied (and still does in many cases rely) on extrinsic (phenotypic) rather than intrinsic (genotypic) characteristics. Besides shape (*coccus, rod, spiral*), one of the most important criteria for identifying bacteria is Gram stain, which is based on the reactions of a bacteria sample upon exposure to crystal violet dye. Gram-positive bacteria (e.g., *Staphylococcus aureus*) appear purple brown under microscopic examination, while Gram-negative bacteria (e.g., *Escherichia coli*) do not. Gram staining is based on the ability of the bacteria cell wall to retain the crystal violet dye during solvent treatment. While this criterion clearly refers to an identification technique (i.e., how we acquire knowledge about given bacteria), the division between Gram-positive and Gram-negative bacteria does in fact correspond to a division in nature on the side of bacteria themselves. The cell walls of Gram-positive microorganisms have a higher peptidoglycan and lower lipid content than do those of Gram-negative bacteria, and this gives the bacteria themselves specific properties (e.g., sensitivity to some antibiotics). In other words, Gram stain reveals differences in constitutional characteristics of bacteria that were simply not known – but were present – when the Danish physician Gram discovered this property in the nineteenth century. This does not, however, imply that the distinction between Gram-positive and Gram-negative bacteria corresponds to a distinction between two *classes* of bacteria.

## 2.2 Conjunction

Here is a completely different case. Tuberculosis is an infectious disease caused by *Mycobacterium tuberculosis*. Although the lungs are most commonly affected, the bacteria may also infect other organs, including the adrenal glands. The name for the corresponding location-based subtype of the disease is *Tuberculosis of the adrenal glands*. Tests are used to diagnose the infection, including direct identification under a microscope and culture of infected fluids. This background helps understand the presence in the medical vocabulary (ICD-9-CM) of a term such as

*Tuberculosis of adrenal glands, tubercle bacilli not found (in sputum) by microscopy, but found by bacterial culture.*

This term is in fact not the name of a special class at all. Rather, it is a sentence-schema, of the form:

[is an instance of] *Tuberculosis of adrenal glands* whereby tubercle bacilli were not found (in sputum) by microscopy but rather by bacterial culture,

and formatted as if it were a class name for coding purposes. For of course the fact of positive or negative identification of *Mycobacterium tuberculosis* by microscopic examination of bodily fluids does not change the disease in any way (it changes neither the corresponding disease class nor the instances by which this class is instantiated). It simply provides information about how a physician obtained knowledge about the disease. This is an example of an epistemological criterion (how the disease was diagnosed) that is introduced into a terminology that is otherwise used for classification purposes.

Closely related to the previous example are many cases where the presence of associated manifestations, lesions, or injuries are used to define classes. Consider terms such as:

- *Closed skull fracture without intracranial injury*
- *Open skull fracture without intracranial injury*
- *Closed skull fracture with intracranial injury*
- *Open skull fracture with intracranial injury*

A fracture of the skull may be *open* (when the broken bone penetrates the skin) or *closed*. A fracture of the skull can in either case also be associated with intracranial injury. The presence of skin penetration by the fractured bone need not imply a different sort of fracture, but it does imply a change in the total disease phenomenon of which the fracture forms a part. For example, open fractures are more likely to get infected, due to the breach created in the skin barrier. Here, skin penetration has a direct and predictable consequence on the evolution of the fracture. Therefore, the term open fracture represents more than the simple conjunction of a fracture and skin laceration occurring at different locations and the presence or absence of skin penetration by the fractured bone is, therefore, a classification criterion that is valid from the point of view of ontology.

In contrast, the presence of concomitant intracranial injury does not change the fracture itself and, therefore, a fracture without intracranial injury does not differ from the fracture when taken alone. The presence of intracranial injury may indeed affect the prognosis of that fracture, making a fracture of the skull with intracranial injury worse than an isolated fracture. On the other hand, a brain concussion (i.e., a head trauma without fracture) with intracranial injury might have the same prognosis as a skull fracture with intracranial injury. Thus, there is no ontological difference between a skull fracture and a skull fracture without intracranial injury. Where a skull fracture *with* intracranial injury is most properly conceived as a conjunction of a skull fracture and a (possibly related) intracranial injury, the term *Skull fracture without intracranial injury* merely conveys the information that the presence of possible intracranial injury in the context of a skull fracture has been adequately checked and ruled out. The added precision brought by this latter term therefore relates to what is known about a given case of skull fracture rather than to the reality of the fracture itself.

Although affecting primarily the subdomain of disorders (e.g., *Gallbladder calculus without mention of cholecystitis*, *Tuberculin skin test reactor without active tuberculosis*), this phenomenon is also encountered with procedures (e.g., *Adenoidectomy without tonsillectomy*, *Repair of malunion of humerus without graft*).

### **3 Terms reflecting detectability, modality, uncertainty, and vagueness**

#### *3.1 Detectability*

Many diseases start with a latent phase. Tumors often grow from a single cell in which the functions regulating cell proliferation have been altered. At this early stage, most tumors are not detectable by the techniques currently available. It often takes months if not years before the tumor has grown large enough that its presence becomes apparent to, say, a radiologist. Similarly, the diagnosis of diseases, abnormalities, and manifestations is sometimes fortuitous: they may be discovered either by chance or during an investigation focused on other, quite different matters. Some special terms, now, are coined to mark the way in which a disease is discovered. Thus an *asymptomatic cholelithiasis* names a condition – the presence of gallstones – whose diagnosis is made when gallstones are discovered during an abdominal ultrasound, CT scan, or X-Ray exam prescribed for another problem (e.g., the

presence of blood in urine) and in the absence of symptoms of cholelithiasis (e.g., jaundice).

Along the same lines, a *subclinical seizure* is a seizure that can be detected by EEG, but has no clinical manifestations. These examples illustrate what we call “threshold classes”, i.e., classes created for the purpose of representing the early stage or a milder form of a disease. In this case, although inevitably emphasizing how the physician came to discover the disease, detectability essentially represents the severity of the disease, the disease classes below some detection threshold often representing milder forms. Such class terms are ontologically valid in the same way in which, for example, *embryo* or *fetus* are ontologically valid class terms in biology. (They represent phase sortals [10].) They correspond to a partition of the domain of diseases along the dimension of severity. Because in the prototypical case diseases are for obvious reasons above some threshold of detectability, the partitioning of diseases by severity is however largely incomplete.

### 3.2 Modality

The presence of modality indicators in medical terms is a completely different issue. Take for example the following three terms for an abscess of the ovary and Fallopian tube:

- *Definite tubo-ovarian abscess*
- *Probable tubo-ovarian abscess*
- *Possible tubo-ovarian abscess.*

Here, *definite*, *probable*, and *possible* clearly refer to modality, not detectability. In other words, these qualifiers reflect the confidence of the physician at the time the diagnosis is posed, i.e., an epistemological feature that does not reflect the nature or severity of the disease being diagnosed. Here again, because of the uncertainty inherent to the diagnostic process, such features are found mostly in the subdomain of diseases. Other examples of terms exhibiting modality markers include:

- *Diseases of possible viral origin*
- *Probable suicide*
- *Basal cell tumor, uncertain whether benign or malignant*
- *Diarrhea of presumed infectious origin.*

In some cases, terms even reflect the degree of confidence a physician has towards several alternative possible diagnoses:

- *Atypical squamous cells of uncertain significance, probably benign*
- *Atypical squamous cells of uncertain significance, probably malignant*
- *Atypical squamous cells of uncertain significance suggestive of an intraepithelial lesion.*

These examples present a particularly clear form of a phenomenon seen in almost all biomedical terminologies, namely the expression via single terms of information which should more properly be conveyed in the form of complete sentences.

### 3.3 Vagueness, underspecification, and other hedges.

Vagueness is frequently encountered in medicine, and it is frequently important (for example for legal reasons) that clinical coding systems capture vagueness in explicit fashion in their constituent terms. Once again, however, we should beware of drawing ontological conclusions from the existence of terms of given sorts.

Vagueness arises for example in the presence of preliminary or incomplete diagnosis, but it is present for many other reasons also. Many class names exhibit underspecification markers such as *unspecified* and *not otherwise specified* (abbreviated *NOS*). Examples of such terms include:

- *Open fracture of unspecified cervical vertebra*
- *Concussion with loss of consciousness of unspecified duration*
- *Replacement of unspecified heart valve*
- *Poisoning by unspecified drug or medicinal substance*
- *Colostomy, not otherwise specified*
- *Chemical element, NOS.*

In the examples above, *unspecified cervical vertebra* refers to one of the seven cervical vertebrae and *unspecified heart valve* refers to the mitral, tricuspid, aortic, or pulmonary heart valve. Further specification for *colostomy* could be in terms of permanence (permanent vs. temporary) or localization (transverse colostomy vs. sigmoidostomy).

Markers expressing vagueness and other types of hedges are pervasive in biomedical vocabularies [3]. The issue here is not so much the existence of vagueness but rather how vagueness is represented. Going back to the examples above, there might well be circumstances where it is not known which of the four heart valves was replaced. In this case, the valve replaced simply needs to be referred to as *heart valve* and there is no need to creating a spurious class term such as *unspecified heart valve*. Similarly, in the absence of further information about the permanence or localization of a colostomy, every particular instance of colostomy still shares the characteristics common to colostomies in general (i.e., it is an artificial opening from the colon on the abdomen wall). Thus the class *Colostomy, not otherwise specified*, too, shares all the characteristics of the class *Colostomy* – but it has no additional characteristics either. *Not otherwise specified* expresses the – quite trivial – fact that further information could be gained but is not currently available about this particular instance. Thus again, it is an epistemological rather than an ontological feature which is here expressed.

#### **4 Terms created in order to obtain a complete partition of the domain**

Medical terminologies such as the International Classification of Diseases (ICD) aim at providing a coding system for all possible health problems. In other words, ICD sets out to provide a complete partition of the domain of health problems. At the same time however it also aims to be as concise as possible, offering only of the order of 20,000 classification slots (i.e., nodes in the classification tree), which means that it is impossible to represent even all standard forms of diseases, let alone their clinical variants. The trade-off adopted by the World Health Organization in developing ICD is to provide slots for the most frequent problems (corresponding essentially to genuine biological classes), while reserving part of the 20,000 slots to groupings of the less frequent diseases (corresponding to its own rules of thumb for constructing artificial classes) by means of terms involving ‘other’.

Let us examine, for example, the representation of *Cystic fibrosis* in ICD-10. As illustrated in Table 1, this class has four subclasses. *Cystic fibrosis with pulmonary manifestations* and *Cystic fibrosis with intestinal manifestations* correspond to two frequent clinical forms of cystic fibrosis. *Cystic fibrosis, unspecified* is another example of a class whose name exhibits underspecification markers (see 3.3 above). *Cystic fibrosis with other manifestations* is created for the purpose of representing those clinical forms not covered by the first two cases (e.g., cystic fibrosis which affects the reproductive system) and thus to complete the classification at minimal cost in extra terminological resources.

Table 1 – *Cystic fibrosis* in ICD-10

E84	Cystic fibrosis
E84.0	Cystic fibrosis with pulmonary manifestations
E84.1	Cystic fibrosis with intestinal manifestations
E84.8	Cystic fibrosis with other manifestations
E84.9	Cystic fibrosis, unspecified

The issue with such artificial classes created in order to obtain a complete partition of a given domain even though the number of classificatory slots is limited is that their definitions are relative to (and thus vary with changes in definitions of) other classes. Thus for example the instances of *Cystic fibrosis with other manifestations* are instances of *Cystic fibrosis* that are instances of neither *Cystic fibrosis with pulmonary manifestations* nor *Cystic fibrosis with intestinal manifestations*. Such terminological practice brings instabilities also in the sense that if a new ‘with ... manifestations’ subclass of *cystic fibrosis* is introduced at some later stage in addition to the *with pulmonary manifestations* and *with intestinal manifestations* subclasses, then the meaning and extension of *Cystic fibrosis with other manifestations* will itself change even though the term itself remains the same.

It often occurs that a plurality of clinical vocabularies all define what is purported to be the same class but in ways which make their respective definitions relative to the definitions of other classes provided within the corresponding host vocabulary. As a consequence, the putatively identical classes in the separate vocabularies are subject to a spurious differentiation of a sort which blocks alignment of the data coded in their terms. For example, Clinical Terms Version 3 (CTV3) also represents *Cystic fibrosis*, but its subclasses, shown in Table 2, are slightly different from those of ICD.

Table 2 – *Cystic fibrosis* in Clinical Terms V. 3

C370	Cystic fibrosis
C3702	Cystic fibrosis with pulmonary manifestations
C3703	Cystic fibrosis with intestinal manifestations
C3700	Cystic fibrosis with no meconium ileus
XaBDdb	Cystic fibrosis with other manifestations
C370z	Cystic fibrosis NOS

All subclasses present in ICD are also present in CTV3, but CTV3 has an extra subclass: *Cystic fibrosis with no meconium ileus*. If we assume for the purpose of this demonstration that this additional subclass is ontologically valid, then its instances will be included in *Cystic fibrosis with other manifestations* in ICD but not in CTV3.

Terms like *Cystic fibrosis with other manifestations* are examples of a quite general phenomenon: because they are introduced simply in order to complete a partition they are likely to have different sets of instances depending on which classification they belong to. Other examples of this same phenomenon include:

- *Certain adverse effects not elsewhere classified*
- *Other prostate disorders*
- *Female infertility of other origin*
- *Unclassified epileptic seizures*
- *Removal of other device from thorax*
- *Toxic effect of other pesticides*



To make matters worse, underspecification features are sometimes combined with terms created for the purpose of obtaining a complete partition, leading to class names which are (from the ontological perspective, at least) absurd, such as

*Other specified respiratory tuberculosis, not otherwise specified.*

## 5 Issues related to normality and to fiat boundaries

Many classes found in biomedical vocabularies aim at representing not the reality of instances to which the classification is applied, but rather the information as interpreted and used in some decision-making process (e.g., in the diagnosis of a disease). The fact in reality might be, for example, that a given individual has a height of 4 feet (122 cm). The corresponding interpreted information is, in the context of a seven year-old boy: *normal height*, and in the context of an adult: *dwarfism*. The definition of normality for size (and for many other biological characteristics, such as visual acuity or the amount of hemoglobin per deciliter of whole blood) is determined statistically by reference to a population. The problem here is that normality is thus made relative to population, so that there are almost as many definitions of medical terms involving a normality component as there are populations. Average size, for example, is different in North America and in Asia. Additionally, in the case of size and hemoglobin, there are variations within a given population related to age and gender. In addition to varying across geographic regions, classes whose definitions are relative to a given population will also necessarily vary over time to reflect changes in this population. In other words, such classes will have different sets of instances depending on the part of the world and time in history considered.

The normality of biological characteristics (and deviations therefrom) is central to the diagnostic process in medicine. Therefore, classes created to express abnormal findings are pervasive in biomedical vocabularies. Examples of such classes include *Precocious puberty*, *Enlarged liver*, and *Decreased libido*. In some cases, the degree of abnormality is made precise, as in:

- *Cerebral spinal fluid protein increased, slight*
- *Cerebral spinal fluid protein increased, marked.*

Beyond normality, this observation can be generalized to notions such as *survivability of the fetus outside the womb at 32 weeks of gestation*, whose variation over time and across geographical locations ranges almost from zero to 90% survival.

## 6 Discussion

Consider, in light of the above, the history of viral hepatitis [6]. *Epidemic jaundice*, now called *hepatitis A*, was known already to the Ancient Greeks. It is transmitted through infected feces. Another form of hepatitis is transmitted by contact with the blood of infected patients, which is why it was named *serum hepatitis* before getting its current name: *hepatitis B*. Contamination after transfusion of contaminated blood was frequent before the mid-1970s, when blood banks started testing for the hepatitis A and B viruses which had been discovered a few years earlier. While the number of transfusion-transmitted hepatitis cases dropped dramatically with the initiation of such testing, the disease did not completely disappear. This observation led physicians to hypothesize that other hepatitis viruses may be responsible for these other cases, named *non-A non-B hepatitis*. The hepatitis C virus was discovered in the early 1990s, and additional D, E, F, and G hepatitis viruses have also been identified. Along the way, terms were crafted in order to name these diseases relatively to what was known at the time, i.e., by exonerating known viruses from

causing the disease. In addition to *non-A non-B hepatitis*, names such as *non-A non-B non-C hepatitis* and *non-A non-B non-C non-D non-E hepatitis* can be found in the literature, if not in biomedical terminology.

While the terminology of hepatitis – in involving the use of names essentially reflecting non-features rather than defining characteristics – is far from ideal, it has not caused classification problems. Each non-feature term can be thought of as the complement of other terms at a certain depth in the hierarchy. At a given level, the corresponding classes are jointly exhaustive and mutually exclusive. The first level under *hepatitis* consists of the three subclasses *hepatitis A*, *hepatitis B*, and *non-A non-B hepatitis*. In turn, *non-A non-B hepatitis* is subdivided into *hepatitis C* and *non-A non-B non-C hepatitis*. Although each kind of viral hepatitis is expected to be a direct subclass of hepatitis rather than being classified further down the hierarchy, an organization of this sort represents a viable if not ideal alternative.

As a matter of fact, many examples of a similar classification scheme based on a particularly important binary distinction and its complement can be found in the biomedical domain, including:

- *Non-Hodgkin lymphoma*
- *Non-invasive medical procedure*
- *Non-opioid analgesics*
- *Non-steroidal anti-inflammatory drugs*
- *Non-insulin dependent diabetes mellitus*

Although these classes emphasize absent features (i.e., the features present in the complement class), we argue that they essentially correspond to valid, genuine classes for which no specific positive name or names have as yet been crafted. That we council the search for such positive names reflects our adherence to the so-called ‘sparse theory of universals’ [2], which argues that it is a mistake to suppose that we can use mere logical combinations to discover universals existing in reality. The needed positive denominations have indeed already been produced in the case of the subclasses of diabetes mellitus: *Type I* (for *insulin dependent*) *diabetes mellitus* and *Type II* (for *non-insulin dependent*) *diabetes mellitus*. In this case, however, the nomenclature based on numbering may still be considered less than fully adequate.

It is important for a number of reasons that classes denoted by biomedical terms represent as closely as possible the genuine classes which exist in reality. Having variant names for the same class – names incorporating epistemological admixtures – may be acceptable (and possibly desirable) as long these terms do not cause confusion by being held to denote distinct classes. This is the case with the many examples of terms denoting the absence of association between two entities (e.g., *Adenoidectomy without tonsillectomy*, simply corresponding to *Adenoidectomy*). Here, again, there is no special class in reality that is instantiated by individual cases of *Adenoidectomy without tonsillectomy*. Certainly there are sets (in the mathematical sense) of the corresponding instances, but biomedical terminologies and the associated ontologies are interested not in purely contingent relations between sets of instances (as illustrated, for example, by a set-inclusion relation such as that between *animal owned by the Emperor* and *mammal weighing less than 200 kg.*) Rather, they are interested in those sorts of relations which are captured in scientific laws, and this means relations holding between genuine classes in reality [7].

Moreover, class names are often the primary (if not the sole) feature used for aligning biomedical vocabularies. Improper alignment of classes brought about by spurious naming conventions is thus likely to result in inadequate integration of the clinical and research databases in which these classes are contained.

## 7 Conclusions

In this study, we analyze the epistemological features of biomedical terminology and their relations to ontological features. This study is therefore complementary to various other approaches developed to identify ontological distinctions, such as Guarino and Welty's meta-properties [9] and Pustejovsky's qualia structure [4]. Like these approaches, our analysis recognizes the necessity of making these distinctions explicit. In contrast, the influence of information and library sciences on terminology development often results in products in which such distinctions are, if not hidden, at least simply referred to as Ranganathan's "facets" [1]. In the future, we plan to develop a method for identifying epistemological features systematically and to refine the definition of genuine classes in biomedicine.

## Acknowledgments

Barry Smith is supported by the Wolfgang Paul Program of the Alexander von Humboldt Foundation. The authors would like to thank Dr James Cimino for helpful discussions.

## References

- [1] Besser, H. Faceted classifications and thesauri. <http://besser.tsoa.nyu.edu/impact/f95/Papers-projects/Papers/perles.html>
- [2] Lewis, D. New work for a theory of universals. *Australasian Journal of Philosophy*, 61. 343-377, 1983.
- [3] McCray, A.T., Bodenreider, O., Malley, J.D. and Browne, A.C. Evaluating UMLS strings for natural language processing. *Proceedings of the AMIA Annual Symposium*. 448-452, 2001.
- [4] Pustejovsky, J. *The generative lexicon*. MIT Press, Cambridge, Mass., 1995.
- [5] Rosch, E. and Lloyd, B.B. Principles of categorization. in Rosch, E. and Lloyd, B.B. eds. *Cognition and categorization*, L. Erlbaum Associates, Hillsdale, N.J.; New York, 1978, 27-48.
- [6] Schmid, R. History of viral hepatitis: a tale of dogmas and misinterpretations. *J Gastroenterol Hepatol*, 16 (7). 718-722, 2001.
- [7] Smith, B. The Logic of Biological Classification and the Foundations of Biomedical Ontology. in Westerståhl, D. ed. *Invited Papers from the 10th International Conference in Logic Methodology and Philosophy of Science, Oviedo, Spain, 2003*, Elsevier-North-Holland, 2004, (to appear).
- [8] Tversky, A. and Gati, I. Studies of similarity. in Rosch, E. and Lloyd, B.B. eds. *Cognition and categorization*, L. Erlbaum Associates, Hillsdale, N.J.; New York, 1978, 81-99.
- [9] Welty, C. and Guarino, N. Supporting ontological analysis of taxonomic relationships. *Data & Knowledge Engineering*, 39 (1). 51-74, 2001.
- [10] Wiggins, D. *Sameness and substance*. Harvard University Press, Cambridge, Mass., 1980.