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17
CLASSIFICATORY
CHALLENGES IN PHYSICAL DISEASE

Mathias Brochhausen

Introduction
Classifying entities into categories is at the core of scientific practice, especially in the life sciences. In medicine, the discipline of classifying diseases is called *nosology*. In this chapter we will give an overview of the theoretical background and current approaches to the nosology of physical diseases, as well as the aims and contexts of disease classifications and their challenges.

In the first section, we will give a brief overview of the role of classification in science and its ontological background. In the second section, we will introduce a number of best practices in creating classifications. In the third section, we will introduce basic methodologies of how to classify diseases. We will also see that disease classifications are done with specific aims and for usage in specific contexts. This is what creates the challenges in providing both useful and consistent classification in medicine. In the fourth section, we will focus on classification criteria formulated from the requirements created by using computer systems to manage biomedical data. The fifth section provides an overview of the *International Classification of Diseases* against the background of the requirements and best practices. In the last section, we will summarize our discussion regarding the challenges in classifying physical diseases and current approaches used in the area of biomedical informatics and documentation to overcome those issues.

We should note that the distinction between psychiatric disease in general and physical disease is disputed, as psychiatric diseases can indeed have physical causes (Szasz 1960). However, for the purpose of analyzing systems of classification, the distinction is useful, because classification systems exist specifically for psychiatric disease.

That nosology is not merely a theoretical exercise but is highly important to medical practice can be seen from the example of Langerhans Cell Histiocytosis (LCH). LCH, which has been called “Histiocytosis X” in the past, is a rare disease characterized by the proliferation of Langerhans cells. Langerhans cells are immature dendritic cells found in the bone, skin, lymph nodes, and other organs (Morimoto et al. 2014). For some time, there has been uncertainty whether LCH is a neoplastic disease (a disease caused by an abnormal tissue that grows by cellular proliferation more rapidly than normal, as do cancers) or a reactive disease (a disease brought about by an aberrant immune response) (Morimoto et al. 2014; Demellawy et al. 2015). Due to these two conflicting classifications of LCH, it remained unclear what the most promising research agenda or therapeutic regimen for that disease was.
The Role of Classification in Science

How is it that the classification of a disease, such as LCH, is so relevant not only for therapeutic purposes but also for how we conduct research to learn more about a disease? The answer to this question is that classifications (e.g., the classification of mechanisms) are extremely relevant to the development of science. In their influential paper “Thinking about Mechanisms,” Machamer, Darden, and Craver analyze mechanisms and break them down into entities and activities. The authors stress the fact that while mechanisms are an important aspect of scientific explanation in all sciences, their role is particularly prominent in the life sciences. According to Machamer et al., specific kinds of mechanisms are interdependent with specific kinds of entities or specific kinds of properties, so in order to explain the phenomena of nature, we refer to mechanisms and kinds of entities that are changed or involved in specific activities (Machamer et al. 2000). One example of this are the pathogenic mechanisms that lead to a disease (e.g., LCH): “The discovery of different kinds of mechanisms with their kinds of entities and different activities is an important part of scientific development” (Machamer et al. 2000: 15). Grouping particulars in kinds that are similar in some ways is a core aspect of the scientific process. When this is done based on the structures of the natural world rather than on human interest, we talk about natural kinds. There are indications that Machamer et al. are talking about natural kinds in the sentence quoted above. In this chapter we will use the term “kind” interchangeably with “type.”

Assuming that scientific classifications like the ones mentioned by Machamer et al. reflect the structures of the natural world, the question is on what grounds we create such classifications. In other words, what determines whether individuals are of the same kind or type?

According to Smith, the answer to this question is that systems of classification should be based on universals (Smith 2006: 292). Universals are repeatables that may be instantiated by different individuals at different times and places (Lowe 2007: 10). One example of a universal is redness. Imagine standing on a street and watching objects pass by. The first object is a red scooter. If someone asked you what color the scooter was, you would say it was red. Next, a red car passes by. If the same person asked you what color the car was, you would probably say it was also red. So, being red is a repeatable: many things in many locations at many times are red. We might say they share the same property—namely, being red. Phrased in the language of universals, what they share is that they all instantiate the universal redness.

If being red is based on a universal, is it possible that being a human being or being a bald eagle is based on universals, too? Or moving to the example of diseases: could a case of LCH be an instance of the universal LCH? Smith thinks so and proposes a classification of diseases that is based on universals (Smith 2006: 292). Smith does not only allow universals, such as redness, which we might call property universals, but also holds that kinds are universals (e.g., being a human or being a case of LCH). Smith, in a paper co-authored with Ceusters, points out that classifying based on universals links classifications closely to the empirical world, since in principle, all universals need to be instantiated (Smith and Ceusters 2010: 178). So, while LCH might be a universal (there have been and are actual cases of LCH), unicorn is not (there are no actual unicorns).

In the history of philosophy, classification based on universals has led to the development of a tree-like representation of the kinds of substances that exist. The intention was to give a unique and comprehensive representation of the types of entities that exist. This representation is called the Porphyrian Tree. One example of such a tree is the Linnaean hierarchy of living beings. Is it possible to build a similar classification of all diseases that is both unique and comprehensive? The answer to that question, at least once we get to the level of classifying particular diseases and their subcategories, seems to be “no.”
In *The Disunity of Science*, John Dupré shows why this is so. He argues that for any given domain there might be more than one classification system and that the ways we classify depend on context (e.g., the goals of a particular scientific study). Although he does not deny that there are objective divisions between kinds of things, he holds that the context dependence of classifications allows for what he calls “promiscuous realism” (Dupré 1995). Dupré’s main thesis is “that there are countless legitimate, objectively grounded ways of classifying objects in the world. And these may often cross-classify one another in indefinitely complex ways” (Dupré 1995: 18). In discussing biological species, it becomes clear that Dupré rejects the Aristotelian notion of an unambiguous classification based on some essential property, whether it is morphological or genetic (Dupré 1995: 34). Although he argues against essential properties, Dupré agrees that there are properties on which we can build more informative classifications than others. For example, classifying animals based on their having feathers will be more useful than classifying them based on their being gray (Dupré 1995: 64). Dupré advocates that multiple classifications based on meaningful criteria should be allowed to exist beside one another. The call for pluralism in scientific classification has created an echo in philosophy of science and has led to the formulation of scientific pluralism (Longino 2001; Solomon 2001; Kellert et al. 2006). Scientific pluralism holds “that the multiplicity of approaches that presently characterizes many areas of scientific investigation does not necessarily constitute a deficiency” (Kellert et al. 2006: x), but that the question of whether a unified theory to account for all phenomena is still an open, empirical question.

We will see how methodologies created for medical information science can provide systems of classifications that allow the creation of multiple hierarchies along different lines of properties (e.g., diseases classified based on the anatomical site or system effected, diseases classified based on their causes, etc.).

### Criteria for Well-Designed Classification Systems

If classifications are such a key element of the scientific endeavor, it raises the question of how we can create better classification systems, especially for scientific purposes. Jansen (Jansen 2009) gives an overview of best practices when it comes to building classifications. He starts by giving a classification that he deems to be a bad example, taken from Jorge Luís Borges’s *The Analytical Language of John Wilkins*. Borges claims that, according to an old Chinese dictionary, animals can be classified into (a) those belonging to the Emperor, (b) those that are embalmed, (c) those that are tame, (d) pigs, (e) sirens, (f) imaginary animals, (g) wild dogs, (h) those included in this classification, (i) those that are crazy-acting (j), those that are uncountable (k) those painted with the finest brush made of camel hair, (l) others, (m) those which have just broken a vase, and (n) those which, from a distance, look like flies.

(Borges 1975: 103)

Based on the shortcomings of this taxonomy, Jansen proposed a number of criteria that good classification systems should fulfill. Here are the four criteria we will focus on in this chapter:

1. **Ontological grounding**—In a good classification, classes are based on the properties of their members. This excludes classes like “others” (Jansen 2009: 160). Suppose you are taking it upon yourself to list all birds in a specific area by type, but all you know are silver gulls and bald eagles. You decide to report all birds that are neither a silver gull nor a
bald eagle in a class called “other.” The latter class will contain many different birds (like finches, penguins, ostriches, etc.) that all have nothing in common except being a bird and not being a silver gull or a bald eagle. So, this class does not classify birds by their properties, but merely by the fact that they do not have the properties the members of the class “silver gull” or “bald eagle” have.

2. Structure—In building classifications, we need to take into account that types of things can be represented in a hierarchy: types have subtypes and not all classes in a classification are on the same level (Jansen 2009: 160, 162). Considering silver gulls, we can state that they are a species in the family of gulls. Likewise, we can state that bald eagles are one species under the genus sea eagles. In Borges’s example, we do not find a hierarchy. All classes are on the same level.

3. Disjointedness—Assuming that we are dealing with a hierarchy, all classes on the same level should be disjoint. That means that if A and B are on the same level, no member of A is also a member of B (Jansen 2009: 160). If we think about the two species, silver gull and bald eagle, as classes, then they would be on the same hierarchical level because they are both species of birds. There are no individual birds that belong to both classes. If a bird is a member of the class “silver gull,” then it is not a member of the class “bald eagle.” Jansen’s principle of disjointedness states that this should be the case for all classes on the same hierarchical level.

4. Uniformity—The criteria used for classification are uniform. They refer to properties that exist throughout the entire domain (Jansen 2009: 160–1). This criterion has two parts: first, the classification should be done based on one type of property. In the example of classifying birds introduced above, we started our classification by creating classes based on species membership. Observing finches, penguins, and ostriches, we might be tempted to also introduce classes based on the bird’s preferred mode of locomotion: “flying birds” and “walking birds.” However, these two classes should not be part of the classification that also contains “silver gull” and “bald eagle,” because these classes are based on a different type of property. Second, the classification should be based on a type of property that exists throughout the entire domain. Looking at Borges’s taxonomy, we find those animals that are “painted with the finest brush made of camel hair.” However, not all animals in Borges’s domain are painted. Hence, classifying animals by the types of brushes they are painted with violates the principle of uniformity. (Borges’s example here also contains a category mistake: animals that are painted are not really animals. They are depictions of animals.)

The Aims and Criteria of Classifications of Physical Disease

The classification of diseases is interdependent with the existing system of medicine. Systems of medicine have changed over time, but even at one point in time, multiple systems of medicine can exist in different cultures. In the history of Western medicine, a number of systems of medicine have succeeded each other (e.g., humoral pathology and cellular pathology; Risse 1993). Humoral pathology or humorism is a system of medicine that is based on the assumption that the diseases are the consequence of an imbalance in the proportion of the four bodily fluids: black bile, yellow bile, phlegm, and blood. In his book, *Genesis and Development of a Scientific Fact*, Ludwig Fleck shows how the term syphilis was used to cover multiple different diseases over time. Shifts in the extensions and variations of the usage of the term are linked to shifts in the system of medicine as a whole, from humorism to microbiology and cellular pathology (Fleck 1981). Fleck shows clearly how changes in the system of medicine, while
based on the question of what the nature of disease is, affect and alter the classification of specific diseases.

Because of this influence of historical and scientific context on the classification of disease, looking at the historical development of codified and commonly used classification is relevant to understand which factors shaped where we are now. The earliest classifications of diseases were linked to efforts to unify cause-of-death statistics. This effort was partly motivated by an increased attention to preventing epidemics (Moriyama et al. 2011: 1). It is important to note that the classifications did not develop from clinical operations (e.g., diagnosis, therapy). Another historical aspect that has shaped the look of disease classifications used today is that the development of the classification of disease went hand-in-hand with the development of a disease nomenclature. The two are not the same. A nomenclature, such as the Standard Nomenclature of Diseases and Pathological Conditions, Injuries, and Poisonings for the United States (U.S. Department of Commerce 1920), merely provides a nonhierarchical list of acceptable terms for diagnosis (e.g., “Abscess, Cornea” or “Acetonemia”). It does not provide any grouping of these terms in more general terms, such as sexually transmitted disease, infectious disease, or histiocytosis. As a result, a nomenclature is not particularly useful for epidemic purposes, like monitoring the spread of infectious disease or STDs in a particular area or during a particular time. From the perspective of the criteria established in the previous section, a nomenclature does not fulfill the criteria of structure since it is not hierarchical.

So, nomenclatures might answer the need for unification of the terms used in diagnoses, but they do not provide the means to classify the diagnoses according to different kinds of pathological mechanisms. The latter is essential to the scientific endeavor. For example, a nomenclature will contain the term Langerhans cell histiocytosis and may list symptoms and other diagnostic criteria; however, a nomenclature would not inform us that a whole group of diseases share a proliferation of either tissue macrophages or dendritic cells (called histiocytes) and that these are called histiocytosis (Cline 1994). Being able to refer to a group of diseases that share one property is relevant not only for diagnostic purposes (differential diagnosis) but also for scientific purposes. To understand LCH, comparison to other forms of histiocytosis is helpful. However, the example also shows the potential pitfalls when we create classifications: nowadays, histiocytosis is still used sometimes, but it is regarded as an umbrella term (Kumar et al. 2015: 621) that captures multiple diseases that, while similar in some respect (proliferation of macrophages or dendritic cells), are different in other clinically and pathologically relevant ways. The more pressing question recently in LCH research has been whether LCH is a neoplastic or immune-reactive disease (Morimoto et al. 2014; Demellawy et al. 2015).

This example also demonstrates that building classifications of physical diseases is not trivial, nor is there one agreed-upon criterion by which to classify. When classifying disease, we could base a classification on a number of traditional criteria: the location of the disease (anatomical), the cause of the disease (etiology), the mechanism that causes the disease (pathogenesis), or the symptoms of the disease. In working toward one classification of physical disease that can be used for all medical activities, more than one criterion is often used (Snider 2003: 678–9).

The fact that the location of the disease is not an optimal criterion for disease classification became apparent fairly early in the history of medicine. Many diseases are not restricted to one body part or one organ system. For example, tuberculosis does not only affect the lungs and the respiratory system but may also affect the spine (Pott’s disease) and the lymph nodes (scrofula). Similarly, classifying diseases by symptoms into classes like “diseases presenting with chronic airflow obstruction” is problematic due to the large number of
nonspecific symptoms. LCH, for example, shows a multitude of nonspecific symptoms (e.g., of the skin-rashes, etc.).

In the practice of nosology, which criterion is used depends on the particular disease and our knowledge about that disease:

In a disease without a single etiology but with well-defined pathology, such as mitral stenosis, the pathology of the heart valve would be a defining characteristic. If etiology was not single, and pathology was not specific, well-defined pathophysiology would be used as a defining characteristic.

(Snider 2003: 678)

In the example of LCH, where the cause is yet unknown, some progress regarding the pathogenesis has been made (Demellawy et al. 2015).

Over the last decade, there has also been the vision of linking diseases, even those that are not hereditary, to gene loci. Many small steps have been made toward linking common diseases to genes, but the field still needs to undertake efforts to create more comprehensive genetic mappings for human diseases (Altshuler et al. 2008). If such mappings were created, diseases could also be classified based on genes or gene loci that have been associated with that specific disease. With respect to LCH, recent studies have shown an association between mutations of the BRAF gene and the pathogenesis of the disease. This finding led to the re-evaluation of LCH as a neoplastic disease (Demellawy et al. 2015).

In the case of LCH, this shift in classification from autoimmune disease to neoplastic disease has already led to a number of changes that affect patients. First, there now exists a number of clinical trials based on the new understanding of the disease. There have already been successful applications of BRAF inhibitors to cure the disease (Charles et al. 2014; Haroche et al. 2015). Second, those seeking funding for research on LCH are experiencing success with institutions that specialize in research on childhood cancers (e.g., St. Baldrick’s Foundation). Third, the PDQ database, which is run by the National Cancer Institute and provides cancer information for patients and providers, started to include LCH around 2 years ago. Last, since LCH is now considered a neoplastic disease, families have access to assistance from organizations that were established to help families of children with cancer (P.K. Campbell, personal communication, June 30, 2015).

The classification criteria we have seen so far were without exception criteria based on the biomedical reality of disease. The multiple criteria that can be chosen to classify and the fact that the choice among these criteria is often based on pragmatic considerations reflect Dupré’s promiscuous realism: the properties used to classify are actual properties of an objective physical phenomenon. However, some classifications are more based on socio-political assessments and expectations: Johansson und Lynøe (Johansson and Lynøe 2008: 15) give the example of drapetomania. This disease classification was created by surgeon and psychologist Samuel A. Cartwright in the 1850s. Cartwright claimed that slaves who ran away from their owners suffered from a psychiatric disease, drapetomania, which manifested by the afflicted slaves running away from their owners (Johansson and Lynøe 2008: 14–15). It is obvious that socio-political assessments of whether slaves ought to be free and the expectation that they ought to suffer their slavery without complaint or resistance shaped this classification of a “disease.” There are no physical or biological aspects examined for that diagnosis. The only thing wrong with the afflicted persons is that they do not conform to the social and political norms in their environment. Such disease classifications, which are not based on the physical basis of disease, do not fall under Dupré’s call for pluralism, because he stresses the fact that his perspective is a realist one (Dupré 1995: 57–58).
Criteria for Classifications in Biomedical Information Management

We have seen how the need for better statistics about mortality and morbidity drove the development of early disease classifications, showing that the purpose behind creating a disease classification is often rooted in the need for medical documentation. The advent of the computer in assisting with medical documentation changed the practice of how data was captured, accumulated, and interpreted. The use of computer systems and the move towards an Electronic Health Record created new requirements regarding standardization, in particular. Thus, shared vocabularies and terminologies became even more important (Shortliffe and Blois 2013).

In 1998, Cimino published a paper outlining the desiderata for controlled medical vocabularies. Controlled vocabularies are organized lists of terms relevant to a specific domain (e.g., disease) compiled to index publications or other material relevant to the domain. Typically, controlled vocabularies are organized hierarchically and thus can be seen as classifications. Although Cimino’s paper is not exclusively about the classification of disease, he is aiming at controlled vocabularies that often contain a classification of disease. Cimino’s motivation is rooted in the challenges that arose from biomedical informatics, and thus, his criteria present a proposal for a best practice of modeling medical data. However, some of them are relevant to classifications of disease in general:

1. Polyhierarchy—Although strict hierarchies are more manageable, most users of medical classifications agree that allowing multiple hierarchies is desired. It allows catering to heterogeneous purposes of use (Cimino 1998: 396–7).
2. Formal definitions—Controlled vocabularies should not only provide textual definitions, but formal definitions should link the term to other terms in the vocabulary (e.g., linking Pneumococcal Pneumonia to Streptococcus pneumonia with a caused by relation; Cimino 1998: 397).
3. Rejection of “Not Elsewhere Classified”—“Not Elsewhere Classified” (NEC) classes are classes that contain all individuals that did not fit the criteria of any other class that is a subclass to the same superclass as the NEC. Although the use of such classes might seem understandable from the pursuit of comprehensiveness, the members of these classes do not necessarily share any property amongst each other, except the fact that they didn’t fit into any other class (Cimino 1998: 397).

We see that Cimino’s rejection of NEC classes is in line with Jansen’s criterion of “ontological grounding.”

It is obvious that Cimino and Jansen disagree greatly regarding polyhierarchy. Jansen rejects the idea that a class can belong to more than one superclass. Allowing for belonging to more than one higher-level group is called multiple inheritance (Jansen 2009: 165) and is likely to exist in polyhierarchies. Cimino, however, considers multiple inheritance to be one of the desiderata embraced by the scientific community in the field of biomedical informatics: “General consensus, seems to favor allowing multiple hierarchies to coexist in a vocabulary. . .” (Cimino 1998: 397).

Smith criticizes the fact that, in this paper, Cimino treats linguistic relations that may hold between linguistic expressions (e.g., terms in writing—is narrower in meaning than, is synonymous, etc.) exactly like the relations relevant in an ontological view. The latter are relations that hold between the entities that the terms stand for, their referents. The relation between diabetic patient and diabetes mellitus is a relation between the actual patient and one actual instance of the disease; it is not a relation between the terms “diabetic patient” and “diabetes
mellitus” (Smith 2006: 289). Linguistic relations, by contrast, hold between the linguistic expressions. The term “diabetic patient” is narrower in meaning than the term “patient.”

Failure to differentiate between statements about linguistic expressions and statements about their referents are called use-mention mistakes. Use-mention mistakes are relevant because they lead to incorrect inferences. Consider the following sentences:

(1) Pertussis is a highly contagious bacterial disease.
(2) “Pertussis” has nine letters.

Knowing that pertussis is also known as whooping cough, we might want to replace “pertussis” with “whooping cough.” Although sentence (1) is still true if we do so, sentence (2) is not. This is why in sentence (2) we marked that we are talking about the term and not about its referent by using quotation marks. In sentence (1) we use the linguistic expression “pertussis” to refer to a bacterial disease. In sentence (2) we merely mention the linguistic sign, without being interested in what it refers to.

This section shows how the need for more standardized classifications for the medical domain links the practice of providing such classification (including classifications of disease) to the question of what is the best practice in creating classifications. It also shows that while there is some concurrence between the foundational rules for classifications established below, many practical concerns exist, which create some dissent.

The International Classification of Disease and the Classification of Physical Disease

In this section, we will examine the classification of physical disease in The International Statistical Classification of Diseases and Related Health Problems, or International Classification of Diseases (ICD) for short. It is provided and curated by the World Health Organization (WHO). The ICD is not the only classification of disease commonly used today. There are others, such as the Systemized Nomenclature of Medicine (SNOMED), Medical Subject Headings (MeSH), and the National Cancer Institute Thesaurus (NCIT). Although these all provide slightly different categorizations of diseases, they all follow the basic classificatory criteria outlined in the section above on basic methodologies. They demonstrate once more the multiplicity of uses that motivates the creation of disease classification. MeSH, for example, was created for cataloguing medical libraries (Rogers 1963) and still bears the mark of being a library science–inspired artifact. Today, it is still very useful for annotating medical literature (including journal articles) with keywords.

There are multiple non-identical versions of ICD-10. All examples of classes and codes given in this subsection have been interactively retrieved using the standards browser for ICD-10 (http://apps.who.int/classifications/icd10/browse/2015/en).

Historically, the ICD was developed for classifying causes of death for the purpose of statistical and epidemiological analysis. Since 1948, the ICD has increasingly been aimed to be useful in both morbidity and mortality statistics (Moriyama et al. 2011). Currently, ICD-10 classifies diseases and other relevant phenomena in 22 chapters. Chapters I to IV organize diseases based on etiology, while VI to XIV list diseases anatomically by the organ system that is affected. Chapter V lists mental diseases and behavioral disorders. Chapters XV and XVI deal with diseases related to pregnancy, childbirth, and the perinatal period. Congenital malformations, deformations, and chromosomal abnormalities are listed in Chapter XVII. Chapter XVIII is entitled Symptoms, signs and abnormal clinical and laboratory finding, not elsewhere classified. Unlike the preceding chapters of ICD-10, this chapter does not comply with the best
practices we have identified, as it contravenes both Jansen’s rule of ontological grounding and
Cimino’s call for rejecting NEC classes. We will see below that NEC classes or terms are not
uncommon in ICD-10. The subsequent chapters classify injuries and poisoning (Chapter XIX),
occaurrences such as accidents, intentional self-harm, operations of war (Chapter XX), and
factors like tobacco use, stress, and health care encounters (Chapter XXI). Under the heading
Codes for special purposes, ICD-10 provides unassigned codes for future use and reassignment
(Moriyama et al. 2011: 21). Each chapter is classified in categories (coded with a letter and
two numerals). Those categories are further specified into subdivisions (coded with the code
of their category plus an additional number). Each subdivision comes with a list of terms called
inclusion terms. These identify terms that are to be understood as included in the subdivision.
The inclusion terms listed may be synonymous with the subheading itself or may refer to dif-
ferent, more specific disease entities (World Health Organization 2012: 19–20).

Obviously, the ICD-10 is not only a classification of physical diseases, as demonstrated by
the inclusion of mental diseases, injuries, and events leading to harm. With respect to the
classification of physical disease, it is also obvious that ICD-10 does not classify based on one
criterion alone. We find classes based on causes of disease and chapters organized according
to the location of the disease with respect to major organ systems. The ICD-10 is understood
by its creators to be a variable-axis classification, which combines five main axes (epidemic
disease, constitutional/general disease, local disease by site, developmental disease and inju-
ries) into one classification. “It has stood the test of time and, though in some ways arbitrary,
is still regarded as a more useful structure for general epidemiological purposes than any of the
alternatives tested” (World Health Organization 2012: 14). This statement once more stresses
the focus of ICD-10 on statistical usage over clinical usage.

Looking at the ICD-10 and focusing on what kind of hierarchy it provides, it is obvious that
the classification methodology used yields a hierarchy where each category is subsumed under
one and only one chapter and each subdivision is a subdivision of one and only one category.
This is called a monohierarchy. The WHO has identified the fact that ICD-10 provides only
one hierarchy as a key shortcoming. Changing this to provide multiple different hierarchies
would not only fulfill Cimino’s call for polyhierachy, but it also responds to the need for mul-
tiple context-based classifications noted by Dupré. The development of ICD-11, which started
in 2007, will provide a disease classification that can be used for Electronic Health Records and
is based on a shared logical model that allows for multiple context-specific mono-hierarchical
classification (Rodrigues et al. 2014). We will see in the last section that this strategy of using
formal logic to provide a multi-hierarchical structure of sets, while not providing a decision
regarding how physical diseases ought to be classified, covers much if not all of the actual uses
of disease classes.

As an example of the current classifications in ICD-10, let us look at LCH. LCH is classified
under C96: Other and unspecified malignant neoplasms of lymphoid, hematopoietic and related tissue.
(We should briefly note that this subcategory uses “other” as a classification criterion, which
we have seen should not be done.) LCH is subdivided into three classes along the distinctions
between unisystem vs. multisystem and unifocal vs. multifocal:

- C96.0 Multifocal and multisystemic (disseminated) Langerhans-cell histiocytosis
  [Letterer-Siwe disease]
- C96.5 Multifocal and unisystemic Langerhans-cell histiocytosis
- C96.6 Unifocal Langerhans-cell histiocytosis

Pulmonary LCH, which is typically classified separately in the clinico-pathological classifica-
tion, is subsumed under C96.6 as an inclusion term with its alternative name Eosinophilic
granuloma. Clinico-pathological classifications are those classifications that are usually found in pathology textbooks (Kumar et al. 2015: 621). They combine pathological considerations with clinically relevant considerations, to facilitate decision making by the health care provider. C96.6 also subsumes the inclusion term Langerhans cell histiocytosis NOS (“not otherwise specified”). This is again a clear violation of the criteria of omitting NEC terms. It raises the question of how these inclusion terms can work properly in a classification of entities. All LCH instances for which we do not have enough information to unambiguously categorize them go under 96.6 and are in the same class as instances of unifocal LCH. Such a classification clearly is not helpful for the clinical and pathological assessment of the situation. Although it is obvious how this classification promotes the statistical need to capture everything, it is also clear that clinical interpretation of statistics thus created may be prone to error. Since all cases for which we do not have sufficient information to subsume under 96.0 or 96.5 are counted as 96.6, we cannot be sure that only unifocal LCH cases are captured by that code. There might even be more multifocal cases than unifocal cases.

The Challenges in Classification of Physical Disease and How to Overcome Them

Our brief survey of classifications of physical disease shows that practical requirements seem to collide with best practice of how classifications should be done. Not only does conforming to general guidelines of how to build classifications such as Jansen’s appear to be a challenge, but so does conforming to domain-specific criteria such as Cimino’s.

The question we need to answer from the perspective of medicine remains: how can we build classifications in such a way that their creation and maintenance follows transparent and commonly acceptable criteria, while still ending up with classifications that allow us to switch perspectives based on the context of use? Multiple authors have stressed the need for multiple hierarchies based on context (Dupré 1995; Cimino 1998).

From the perspective of creating and maintaining a classification, rules like those formulated by Jansen, or Ceuster’s and Smith’s principle of instantiation, certainly provide advantages. However, these approaches have been criticized for overburdening the field of medical classifications with an uncalled-for commitment to metaphysical realism that can lead to impractical situations (Cimino 2006: 300–1). Facing this criticism, Smith and Ceusters have clarified that the actual ontological commitments of those creating the classifications are not relevant. Although they both are metaphysical realists—they think that universals are real (Smith and Ceusters 2010: 140–1)—in their eyes, the commitment to universals can be treated merely as a part of a methodology that allows building classifications and ontologies for practical use in a more transparent and coordinated manner. Using that methodology does not require that whoever uses it believes in the existence of universals. Smith and Ceusters stress the practical advantages of building and maintaining a single affirmed hierarchy based on “is a” relations, but they concede that multiple inheritance and, thus, multiple hierarchies can be inferred based on other relations and inclusion criteria for classes (Smith and Ceusters 2010: 147).

Although the ability to represent multiple, context-dependent hierarchies corresponding to multiple contexts and purposes is crucial, having a concise, formal methodology to create and maintain disease classifications is also indispensable. This means that instead of sticking to traditional mono-hierarchical classifications like ICD-10, we should strive for classifications that are based on formal logical definitions based on properties of the represented entities. This is certainly possible, and work toward making this a reality is on the way, but it is important to note that scientific progress will continuously make reclassifications necessary, as we have seen from the example of LCH.
## References


**Further Reading**


