

Viewpoint ■

Clinical Classification and Terminology:

Some History and Current Observations

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Abstract The evolution of health terminology has undergone glacial transition over time, although this pace has quickened recently. After a long history of near neglect, unimaginative structure, and factious development, health terminologies are in an era of unprecedented importance, sophistication, and collaboration. The major highlights of this history are reviewed, together with important intellectual advances in health terminology development. The inescapable conclusion is that we are amidst a major revolution in the role and capabilities of health terminologies, entering an age of large-scale systems for health concept representation with international implications.

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It is easier to speak about health terminology than to write about it, since people may read and wonder exactly what is meant by this word or that term. The “meta-terminology” of terminology is no exception, fraught as it is with notions of concepts, classification, nomenclature, and terminology. The reality is that these words are often used casually, imprecisely, and even interchangeably. Formal distinctions do, of course, exist and warrant some overview.

The abstractions implicit in a concept date at least to Plato’s articulation of a perfect form apart from the shadow of that form in this world. Plato classified things by using strict divisions, a method rejected by Aristotle, who replaced dichotomy with the syllogism¹ or logical premise. Aristotle made this more practical yet, introducing the notion of differentia, which could serve to distinguish or describe form.² Indeed, one can almost deduce that the entire genre of multi-axial classifications were anticipated by Aristotle, if one replaces the modern notion of classifi-

cation axes with attributes about specific differences.¹ Put simply, Aristotle introduced the notion that abstract concepts represent descriptions, or more properly definitions, of things that have been classified by describing their attributes.

But abstract descriptions are not words or terms. Strictly, nomenclature simply means the naming of things. However, to name something implies knowledge of its form, perilously close to an Aristotelian definition. Conventionally, however, nomenclatures can be nothing more than lists of recognized or sanctioned words and have little or no relationship to a system of classification. Indeed, some dreary examples illustrate just how removed from useful organizing schemas some nomenclatures can be.³ However, classifications and nomenclatures can be more helpfully regarded as lying along a continuum, where the first categorizes and aggregates while the second supports detailed descriptions.

Finally, “terminology” invokes the language labels attached to a concept. Here, one can become horribly distracted by eponyms or word origins,⁴ tracing the Greek, Latin, or even Norman routes of our English language.⁵ However, this view is profoundly ethnocentric, since the language labels used for a medical concept by speakers of different languages may well be quite different, rendering the study of which term to use essentially a local concern. This may usefully be distinguished from the concept systemization to which a language label (term) might be attached—hardly a local concern if comparability, consistency, and interoperability are shared goals.

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“Terminology,” then, is a convenient moniker, which can mean everything or very little. Many authors, including I, invoke terminology to subsume the entire problem, from classification to nomenclatures, language labels to concepts. Practically, we mean the naming problem, enabling clinical users to invoke a set of controlled terms that correspond to formal concepts organized by a classification schema.

An Inauspicious Beginning

Modern terminology began with the modest classification developed for the London Bills of Mortality in the latter part of the 16th century. These bills were collected and collated by a cadre of parish clerks, who had essentially no medical training and indeed were hired primarily to chant during ecclesiastical ceremonies.⁶ Despite what must have been substantial misclassification bias, some 70 years later John Graunt made insightful observations⁷ on the patterns of mortality emerging from the three score rubrics that this system (a forerunner of the ICD) comprised. He deduced the modern notions of epidemic and endemic disease patterns and speculated on causality. This writing, in a stroke, invented the disciplines of population-based epidemiology, tables of demographically defined mortality, small-area variations analysis, and modern nosology. What made it inauspicious was that poor John retired a failed draper by trade,⁸ his work on epidemiology and nosology to languish virtually unnoticed for 250 years.

I previously detailed some of the early history of terminology in the years that followed.⁹ Suffice it to say that a thousand flowers bloomed, and most faded. At least two factors contributed to the quandary of hard-toiling terminologists of earlier eras. First, health concept description is actually quite hard. Most clinical characterizations have been and remain largely syndromic or symptomatic. A deep pathophysiologic explanation for disease description has emerged only in my lifetime, while a molecular or genomic basis has appeared only since my elementary-school-aged son was born. As such, many descriptions were overloaded with huge variations or fraught with more morbid modifiers and quaint qualifiers than statistical tabulations could bear.

Second, earlier workers did not have the advantage of modern computer science notions about knowledge representations or the tools and machines to manage such complexity. Subsequent work has shown that this complexity is formidable,¹⁰ which perhaps goes far to explain a long tradition of unremarkable attempts to characterize patients using controlled terminologies.

Recent history is perhaps more remarkable for how terminologies were perceived and used or, arguably, misused throughout the industry. An impartial observer might be excused for concluding that a careful and complete representation of patient findings and conditions was not a major goal of clinical classifications in the late 20th century, as citizen concern, news media, and litigation have highlighted problems of fraud and abuse¹¹ that center on distortions of underlying health information rather than its accurate characterization. The association of patient classifications with billing and other administrative functions now dominates industry perceptions, despite the huge potential for well-formed terminologies to enhance health care efficiency and efficacy.

Some Divergence Along the Way

Most terminologists credit the birth of the Standardized Nomenclature of Diseases (SND), later to become (with Operations) the SNDO,¹² as the beginning of a modern era for clinical descriptions. Conceived during a symposium at the New York Academy of Medicine in 1929, this terminology introduced the notion of multi-axial coding. Having but two axes initially, the principles illustrated by the SND were innovative and highly expressive. The two axes were topology (or anatomy) and etiology (or pathophysiology). By way of example, tracing down the anatomy one could readily locate the prostate with a code of 764; similar tracing in the other axis could readily identify adenocarcinomas with a code of 8091. By mixing these two codes together, the diagnostic phrase for prostatic adenocarcinoma could be composed. It was but a few years later that an axis for operations was added to form the SNDO. Thus, combining the code for radical excision, 14, with the anatomy code 764, enables one to compose the procedure of radical prostatectomy, 764-14.

This hardly seems radical in the face of modern, highly expressive terminologies. However, the innovation lay in the implicit ability to compose and express clinical concepts that had not been anticipated or even considered possible. Consider pituitary resection, a form of intracranial surgery that had not been attempted in the pre-antibiotic era of SNDO's development yet is fully expressible in that system. Admittedly, the unsanctioned use or abuse of such multi-axial compositions can breed more nonsense than sense. Still, the ability to express exponential numbers of clinical phrases in controlled terms is powerful and seductive.

Alas, there is a dark side to this expressive power,

since the members of the CANON Group¹³ pointed out no less than 17 ways to compose appendectomy in SNOMED International¹⁴ (a.k.a. SNOMED 3). Although we might celebrate such diversity in compositional freedom, a difficulty emerges when we try to either retrieve such cases or trigger from them in decision support logic. In either case, we must try to anticipate virtually all permutations and combinations of expression to ensure complete retrieval or accurate applications of decision systems. There are forces to overcome this difficulty, known variously as semantic closure or expression rules. Their effect, when they are well designed, is to ensure that all alternative ways to compose an expression, e.g., the 17 variations on appendectomy, can be normalized to a standard or “canonic” form. These powers lie for the most part in our future, although they are known to many current developers and their harbingers appear in some advanced prototypes in use today.

The direct lineage of SNDO included SNOMED 3, with many stops before and after. In somewhat direct descendancy, an evolutionary ordering would be SND, SNDO, SNOP (pathology), SNOMED (medicine), SNOMED II, SNOMED International, SNOMED RT, and the recently announced product of the merger with the U.K. National Health Service’s Clinical Terms¹⁵ (formerly the Read Codes), SNOMED Clinical Terms.

Divergence of a more insidious kind had run parallel to multiple expressions of composite concepts in a terminology system. More terminology systems were developing. Many of them were multi-axial, and those invariably had axes that overlapped those of other multi-axial systems. Rather than enumerate them all, an example will suffice. The American College of Radiology introduced a coding system for x-ray findings in 1955.¹⁶ What is pertinent is that yet another anatomy axis was introduced, naturally defining most precisely observations made from plain films. It was not, however, compatible or consistent with the SNDO or SNOMED anatomy axes. In fact, all along the SNDO lineage were wide inconsistencies in the anatomy and other axes.

Meanwhile, many adaptations of the large-category classifications springing from the International Classification of Diseases (ICD) were attempted, particularly for use in morbidity, albeit in inconsistent and incompatible ways. Most added a decimal point and digits after the three-number rubric from an ICD version, such as ICD-7¹⁷ or ICD-8.¹⁸ These at least maintained consistency with the underlying three-digit ICD meanings. However, some adaptations eschewed this convention and were essentially inconsistent with everything.¹⁹

Our stage was set, and many actors came and went. The audience thinned, as interest could not be sustained in a cacophony of codes unusable outside their provinces. The great promise of comparable health information languished for a time, just as the seminal contributions of Gaunt were nearly forgotten. Bills and insurance claims seemed to be all that was left over most of the land.

The Promise and the Reality

What many of the players knew, and the audience seemed not to realize, was that comparable patient data are the key to improved effectiveness and efficiency in health care. Patient descriptions are the hub around which patient encounters can be tabulated and inferred, to emerge as new medical knowledge. Similarly, improved care practices and decision support logic are knowledge re-engaged with practice via the linkage of shared terms and concepts.

Nearly 20 years ago,²⁰ Scott Blois depicted a scenario in which we would describe our patient to the “system” and ask whether we had seen any patients like that before. The vision continued with the obvious next question: What happened to those patients? Then, on the basis of these “just-in-time” outcome analyses, tightly tailored to the patient sitting in front of us, what Bayesian probabilities are associated with the various treatment options that we might begin this afternoon? While problems of confidentiality might interfere with the goal of achieving a sample size large enough to provide a useful repository of patient experience, the relevant concern is this: Exactly how do we describe our patient to the system? Furthermore, how are the patients already in there characterized? Clearly, this idealized reuse of clinical experience is highly dependent on consistent and comparable descriptions, the very purpose of clinical nomenclatures.

The difficulty is not hard to see: Few, if any, systems for clinical description could function at this level today. Furthermore, the human entry of such descriptions awaits faster and friendlier interfaces and navigation aids for terminology use, given the enormous time pressures bearing on clinicians everywhere.

Collaboration and Convergence

Perhaps the first shot across the bow for consistency and compatibility was the concerted effort to adopt a standard derivative of the ICD for clinical use. The advent of ICD-9-CM²¹ involved the collaboration and cooperation of the major medical society, associations, payers, government, and industry. It was a great al-

legiance of industry leaders working in common cause for comparable patient data. The enormous success of ICD-9-CM is a testimony to how deeply these commitments for comparability were held.

Although they began somewhat later, similar convergence and cooperation occurred at the other end of the terminology spectrum regarding detailed clinical nomenclatures. In a series of meetings sponsored by the Computer-based Patient Record Institute (CPRI),²² industry representatives of payers, providers, academia, and government began coalescing terminology principles on the path toward establishing comparable content. The meetings were premised on implementation of a framework for terminology development, jointly forged by CPRI and the ANSI Health Informatics Standards Board.²³ While not, of course, entirely responsible for consensus and convergence, these meetings contributed to the trend already under way.

At least three key observations have emerged from those meetings: a definition of clinical terminology, a recognition of a synergistic spectrum running from detailed nomenclatures to highly aggregating classifications, and the separation of thinking about terminologies into phases of use.²⁴ First, the definition for clinical terminology is:

Standardized terms and their synonyms which record patient findings, circumstances, events, and interventions with sufficient detail to support clinical care, decision support, outcomes research, and quality improvement; and can be efficiently mapped to broader classifications for administrative, regulatory, oversight, and fiscal requirements.²²

This definition clarifies expectations, simplifies scope, and affirms the synergies across the terminology spectrum.

Second, the recognition that nomenclatures might complement and not compete with classifications resolves what has been a very long running controversy.²⁵ Many authors have derided one modality or another, failing to recognize that each serves its purpose. It is self-evident that well-defined nomenclatures can be "rolled up" into aggregating classifications, although the rules and logic about how exactly to undertake this are not always obvious or explicit.

Third, the phases of terminology use are now widely regarded as entry terms, reference terminologies, and aggregate or administrative classifications. Entry terms are colloquial expressions or terms (in the strict sense of the word) that are familiar to users and convey sufficient specificity to say what is meant. These

are translated into an underling reference terminology, which is capable of semantic closure and unambiguous representation. Finally, the formal reference terms can be aggregated using explicit inclusion, exclusion, and cross-referencing rules (which are not always readily available in machinable form) into high-level classifications like ICD-9-CM.

More tangible evidence for closer cooperation appears in the adoption by reference within SNOMED of codes from LOINC (Logical Observations Identifiers, Names, and Codes)²⁶ for laboratory tests and findings. Similarly, the National Library of Medicine has hosted many fruitful encounters to organize the formal reference of SNOMED elements in CPT-5 construction as well as facilitate the mappings of SNOMED content into ICD-9-CM and ICD-10 classifications.

Without exception, the most extraordinary convergence is the ongoing effort to derive a new work from the rich combined content and structure of SNOMED RT and the U.K. NHS Clinical Terms, which will be called SNOMED Clinical Terms. Preliminary evaluations show less than 30 percent full redundancy of lexical content, ensuring that the merger will yield something at least additively greater than the parts. More important, the redundant processes, development, infrastructure, and fragmenting market share will all be replaced by what is evolving as a highly synergistic and collaborative enterprise.

Previously competing terminologies are redefining their roles as complementary entry terminologies. Similarly, highly specialized terminologies for application in narrow domains are beginning to serve as interlocking extensions of larger terminologies. This has been most apparent in the suite of SNOMED microglossaries, the first of which appeared in the radiology community,²⁷ thereby resolving the insidious proliferation of redundant axes, such as anatomy, that had threatened the goal of establishing comparable patient information.

Convergence and openness have not been limited to terminology content. Two major terminology-oriented software firms, Lexical Technologies and Ontyx, have merged to form a new organization named Apelon (obviously all is in a name). The venerable European GALEN effort, centered in Manchester, England, is now open software, sharing the very core program code.²⁸

Finally, while redundant standard terminologies proliferated, so too did standards organizations concerned about terminology issues. During the last 18 months, the newly formed ISO TC 215 on Health Informatics has established a Working Group (no. 3) on

health terminologies.²⁹ Progress within this working group has fostered the emergence of standards about standards, such as a meta-vocabulary, a foundation model for health terminology, good terminology development indicators, semantic links, and models for nursing adaptation of meta-standards. Although it is unlikely that any new terminologies will emerge from this working group soon, the international forum affords an opportunity for agreements on first-order principles about terminologies.

Tasks for the Future

The typical conclusion of a researcher is that yet more research needs to be done. Likewise, the convergence of common and comparable systems to uniquely characterize patient findings, events, and outcomes is far from complete—more convergence needs to occur. However, within the existing body of major nomenclatures and classifications, this process is well under way, if not yet widely regarded as nearing completion.

Among the CPRI terminology recommendations was the recurring assertion that a coordinating body, serving the public interest, should be regarded as the single adjutant in the community. No such body has emerged, although newly formed structures within the ANSI Health Information Standards Board might serve that purpose. Regardless, the placement of terminology efforts among the constellation of interlocking entry, reference, and aggregating systems must continue.

The greatest challenge remains the problem of semantic normalization or closure among the myriad ways in which a concept can be composed across and within systems. The problem becomes tightly coupled with that of reconciling the role of semantic representation in the information model of health systems with the expression of modified meaning in a terminology. An often-used example contrasts placing a diagnosis in a “family history” field of a medical record with modifying that diagnosis with a “family history” qualifier from the terminology. These are, of course, conceptually identical, if expressively variant.

Finally, the genomic revolution and the advent of molecularly based diagnostics and therapies opens an entirely new dimension in the clinical naming problem. To date, the nomenclatures in bioinformatics have been ad hoc, haphazard, and at best completely divorced from patient record systems. Increasingly, however, those concerned with medical records and terminology must confront the problems associated with expressing diseases as variations in DNA sequences. Furthermore, some methods must be devel-

oped to cope with the geometric explosions of disease variants attributable to single nucleotide polymorphisms—including their co-occurrences and permutations—that exist within all of us. The clinical consequences of these findings are only beginning to be recognized and are far from being understood.

Such challenges aside, progress in the past few years alone toward intelligent, conceptually consistent, and collaborative approaches in health information has been heartening. We have turned away from increasing fragmentation and have embraced cooperative consolidation. The ultimate beneficiaries of such efforts will, of course, be those patients whose care is enhanced or made more effective as a consequence. Eventually, that should be all of us.

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