The Copernican Era of Healthcare Terminology: A Re-centering of Health Information Systems

CG Chute
Mayo Foundation, Rochester, MN 55905

ABSTRACT

Health terminology and classifications have been an unseen backwater in healthcare practice and information systems development. Today however, the recognized need for comparable patient data is driving a new discovery about its strategic importance. Consistent patient descriptions and concept-centered data representations are crucial for efficient discovery of optimal treatments, best outcomes, and efficient practice patterns. The fabled linkage of knowledge sources at the time and place of care requires the conceptual intermediary of common terminology.

A brief history overviewing the evolution of health classifications will provide the foundation for considering present and evolving health terminology developments. Their roles in health information systems will be characterized. Discussion will focus on the likely influences of the HIPAA legislation nationally and the new ISO Healthcare Informatics Technical Committee internationally, on terminology adaptation and incorporation.

HISTORICAL PERSPECTIVE

The evolution of interest in healthcare terminologies parallels in part the long evolution of classifications, nomenclatures, and structured vocabularies over the past four centuries. While Platonic notions of abstract ideas or concepts and Aristotelian principles of classifying or ordering the natural world are among the oldest elements of information theory in the Western world, application of these principles to clinical findings, disease, and treatments are surprisingly recent.

The London Bills of Mortality[1] date to the late 16th century, and were the first recognized systems of classifying cause of death as applied to populations. Comprising some 60 odd mortal events, they served as the basis for public health practices, plague control, and later to generate profound new insights into the nature of infectious disease, epidemics, and the causal nature of water born agents in the miasmic conditions of the 19th century. The apocryphal tale of John Snow removing the pump handle from the well during a cholera epidemic was made possible by the carefully kept mortality records of the time[2].

Such public health records came not without effort, debate, passion, and intellectual intrigue focusing upon the humble undertaking of characterizing causes of death. It was to be nearly a century after Snow before the subtleties of disease morbidity were to be tackled. Meanwhile, the vicissitudes of mortality classifications engaged many of the best minds of the day, including our high school biology friend, Linnaeus, whose *Genera Morborum*[3] (1763) is among the many, many forgotten systems which emerged during that era[4].

A Different Problem: A Coinage for Healthcare

That so many systems came and went, most now forgotten entirely, might be our first hint that the problem of naming mortal causes, diseases, and other health categories is neither easy nor straightforward. Consider of oft-used analogy of fiscal information systems and currencies. Electronic medical records and healthcare applications today fall far behind the ubiquitous presence of ATM machines and electronic banking. However, healthcare suffers still for lack of a "currency" on which to base information systems. Health events, findings, and outcomes are more abstract, complex, and less well defined than dollars and cents, or even foreign currency conversions. Medicine lacks still a reliable coinage on which to base its analog of economic models and forecasting, yet somehow we persist in attempting health policy development and best practice mandates, with nary a clue of what we are measuring. .

This is not to assert we have no classifications or nomenclatures, indeed we do. Nor does it imply we have no language with which to communicate clinical findings and actions, claiming such would be absurd. Rather I submit our representations are either informal, or under-specified. Thus, the goal of representing what we see and do for patients in a modern electronic medium is compromised for lack of a formal, fully specifiable description. To complete our coinage example, when the workable but awkward British system of pound, shilling, and pence encountered the computer age, that coinage became decimal-digital in a hurry.

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Heritage of the ICD

The vision driving those who attended the First Statistical Congress of 1853 in Brussels was:

> *Il y a lieu de former une nomenclature uniforme des causes de décès applicable à tous les pays.*[5]

For the first time, an international body engaged in a process to agree upon a common system for organizing health events, in this case a uniform cause of death classification applicable to all countries. The parallels of this meeting 145 years ago and the first meeting of the ISO Technical Committee for Health Informatics (TC215) in Orlando last August are striking.

From this beginning 145 years ago, arose what we now regard as the ICD. Spearheaded by Jacque Bertillon and the French Government in the latter 19th and early 20th centuries, the system grew to attain worldwide adoption, albeit with an impoverished content never exceeding 200 rubrics in the pre-WHO era. This auspicious beginning for mortality classification aside, the introductions of the early volumes disclaimed:

> The International List of Causes of Death makes no pretension of being a proper nomenclature of diseases or of including a scientific classification of diseases.[6]

This admonition notwithstanding, the pressure to adopt the greatly expanded 6th edition of the ICD, and more particularly, the 7th, 8th, and 9th revisions, to accommodate the needs of morbidity and healthcare, were nearly overwhelming. Nevertheless, the WHO, which had assumed editorial lead for the ICDs in the post-war and post Bertillon period, maintained its focus upon a longitudinally consistent system for mortality classification intended for international statistical tabulation.

What followed were a series of inconsistent, incompatible, and utterly unworkable adaptations of various ICD revisions for morbidity purposes. Their adherence to the 16th century data models of tabular lists after the fashion of the London Bills of Mortality, doomed them to non-expressive, constrained systems, incapable of capturing clinical detail, and unable to accommodate clinical information without substantial bias.[7]

Rise of Multi-Axial Coding

The need for clinically oriented nomenclatures and the dire state of mortality classifications adopted for this purpose did not escape notice. The New York Academy of Medicine convened a forum in 1928, wherein virtually all medical societies, payers, providers, government, and the Commonwealth fund participated in the development of a new model for representing patient diseases and procedures. Later to become known as the Standard Nomenclature of Diseases and Operations[8] (SNDO), this effort yielded the great intellectual innovation of multi-axial coding.

Multi-axial systems compose clinical descriptions from term lists with sharply defined semantic roles. Most simply, an axis of anatomy would contain a hierarchy of body sites and anatomic detail that would combine with a disease etiology axis including, for example, hierarchical details of tumor types such as adenocarcinoma. Thus, a clinician could compose the notion of "adenocarcinoma of the ascending colon" by combining the appropriate elements from the anatomy and etiology axes. The later SNDO included a detailed axis of operations, including a hierarchical placement of radical excision under excision.

The enormous advantage of multi-axial systems was an ability to compose and express clinical concepts that were not anticipated at the time the system was designed or a version implemented. For example, radial prostatectomy could be properly expressed in SNDO decades before the procedure was common. Pituitary procedures could be similarly represented before they were even believed possible.

The success of the SNDO was moderate, particularly in research-oriented, academic medical centers. Pathologists in particular found the system compelling in their cataloging of cases and tissues. Recognizing the need to accommodate microscopic morphology more fully, the Systematized Nomenclature of Pathology[9] (SNOP) was introduced by the College of American Pathologists in 1965. Building upon the widespread admiration of this system, the College tackled the rest of medicine with its ambitious Systematized Nomenclature Of MEDicine (SNOMED), first introduced in 1979[10], and revised in 1982 and 1994[11].

The principles of multi-axial coding also found footing in the ICD-O[12] (O for Oncology) systems of the WHO, which invoke the ICD as an anatomy axis, and the SNOMED cancer morphologies as a detailed histology axis. These systems form the basis for cancer registries, coupled with an extent of disease axis arising from the American Joint Commission on Cancer[13] in the form of the familiar TNM (Tumor, Nodes, Metastasis) designations.

Systems of the Information Age

The flurry of KL-ONE[14] like knowledge representation languages and frame-based systems of the mid-1980's, took information representation

beyond the relational database metaphor[15]. The science of representing concepts abstractly, using formal syntax, restricted domain values, and semantic inheritance, introduced new and valuable mechanisms that could apply to patient conditions and medical knowledge itself. The linkage of Decision Support Systems (DSS) and rule based inference engines to "intelligent" patient records was seen as an obvious strategy to obviate the tedious re-entry of patient descriptions into information support tools.

One of the early problems was a muddling between representing patient findings, conditions, symptoms, and disease with representing medical knowledge *per se*. Fundamentally, both patient data and clinical knowledge require an underlying representation language (akin to a coinage), and ideally these should be common. Nevertheless, those who developed clinical terminologies had little connection with those who developed clinical decision support tools and knowledge engines intended to improve patient care.

Early leaders in this area were the collaborators on the GALEN project (Generalized Architectures for Languages, Encyclopedias, and Nomenclatures in medicine)[16]. Alan Rector early on took great pain to distinguish the models of 1. Terminology (how words mapped to concepts, and concepts could sustain composable units), 2. Information (for example in a record) and 3. Knowledge (such as a textbook of medicine). While all could be expressible in GALEN, there was an explicit recognition that concepts can and must be consistent regardless of the model perspective.

Central to the abstractions of GALEN, and indeed knowledge representation languages in general, is the notion of a single concept. The venerable MeSH[17] system of the National Library of Medicine supported a concept-based representation, wherein a biomedical literature search term could appear in several different MeSH trees, or hierarchies. Additionally, "Entry Terms" corresponded to what we would consider today as synonyms for these controlled concepts.

A concept-based model of terminologies was made more explicit still, with the introduction of the Unified Medical Language System (UMLS) Metathesaurus in 1990[18]. A seminal principle in the design and implementation of the Metathesaurus is the incorporation of multiple terminologies into a common structure, wherein they are linked (using relational tables) to common concepts. The abstract representation of the concept is by way of a number, or CUI (Concept Unique Identifier). This not only simplifies the maintenance and updating of disparate terminology additions to the UMLS, but additionally affords a mechanism to easily accommodate different natural languages (e.g. French, Spanish, or Russian) into the Metathesaurus[19].

The problems of representing terminologies, their models, and abstractions, in a generalizable and interchangeable fashion, were studied in a series of papers by the CANON group[20]. Settling upon Conceptual Graph notation[21] as a compromise for human and machine readable terminology structure interchange, the group focused on the exchange of semantically interoperable chest x-ray reports. Technology options for interoperable, object-based terminologies have today expanded to include ASN.1, Java, and the rapidly evolving XML standard of W3C[22].

Closely coupled to adequate structure is adequate content; put simply, there have to be goods on the shelves, no matter how fancy the shelves. Here, two systems have emerged with formidable substance, SNOMED[11] and the UK NHS Clinical Terms[23] (formerly Read Codes). Both systems have considerable coverage of the clinical world, though neither is what would be regarded as exhaustively complete[24]. However, both have embraced radically more sophisticated structures and architectures to facilitate interoperability[25,26].

Distributed Terminology Development

Harnessing the energy, interest, and time of many groups and organizations in the expansion toward more complete clinical content of terminologies is an obvious mechanism for development. All too often, however, such distributed strategies impart redundant, overlapping, and inconsistent content; worse the hierarchical "description" of a term by virtue of defining parents and characteristic children can conflict between derivative expansions of a common starting point.

Keith Campbell studied this problem thoughtfully in his Stanford doctoral thesis[27], and has implemented his model for the Convergent Medical Terminology (CMT) project. The CMT arose from within Kaiser-Permanente (KP), and has attracted support from the KP-Mayo project, the College of American Pathologists, and the VA. Today, the project is a primary technology for ensuring comparable patient data within KP, and the foundation for the content expansion of SNOMED RT (Reference Terminology)[26].

The underlying principle enabling the resolution of overlap and conflicts which emerge in the decentralized development of the CMT, is a dependence upon "description logic" to define terms.

Each new term or edit introduces a local change from a known reference version of the terminology. This change is expressed by defining a term by its parents. Conflicts are then detectable by machine, and can be algorithmically resolved under some logical circumstances, or flagged for human review in others.

As always, an example better illustrates these principles. Starting from:

```
Pneumonia
    Is_a Disease
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one group might refine this to be

```
Pneumonia
    Is_a Disease
    In_the Lungs
```

while another might choose

```
Pneumonia
    Is_a Disease
    Kind_of Infectious Disease
```

Clearly, both are true and can be algorithmically resolved as:

```
Pneumonia
    Is_a Disease
    Kind_of Infectious Disease
    In_the Lungs
```

While seemingly simple, this powerful idea has proven enormously effective in facilitating the wide scale expansion of the CMT by many, distributed modelers throughout the country[28].

Crucial to the practical conduct of distributed terminology development has been the emergence of sophisticated editing tools that are semantically enabled. Campbell developed a lexically-based navigator with Lexical Technology, based on Lexical's Metaphrase[29] family of terminology server resources. Similarly, a second-generation tool to manage description logic inferencing and conflict resolution arising from distributed editing was created by Eric Mays and Ontyx from the principles underpinning the original KREP environment[30].

THE ROLE OF TERMINOLOGY IN HEALTH CARE

Fiscal Origins

While the London Bills of Mortality may have had negligible impact on present healthcare classifications, their descendants in the form of the ICD's have a dominant effect. In particular, the clinical modification of these systems has fostered an entire industry, replete with associations and government support agencies which focus upon their development and use. However, the driving need for this has been the rise of third party payers, who reasonably need some characterization of illness and care on which to base appropriate payment. The evolution to prospective payment has deepened our dependence upon these classifications.

But for the exigencies of these fiscally driven administrative descriptions, it is unlikely that our representation of symptoms, findings, diagnoses, and interventions would have been premised upon a venerable disease mortality classification. The urgencies which drove payment classifications prompted the hasty adoption of the ICD as the only viable classification available, and has captured the focus of clinical information processing ever since.

The principle concerns surrounding continued adherence to these systems are two-fold: 1) they lack a detailed clinical specificity for broad application to other uses of patient data such as knowledge support or outcomes research; and 2) distortions and idiosyncratic emphases arise from a tendency to optimize reimbursement at the occasional cost of accuracy.

The Clinical Concept

Information system development is re-centering its attention upon the commonality among admitting conditions, working diagnoses, discharge diagnoses, and clinical outcomes. These represent point-times along the longitudinal healthcare process and should not be artificially separated. Similarly, patient information lives along another continuum from detailed, specific findings and conditions to broad groupings for specific purposes; these historically have been represented as a counterpoint between nomenclatures and classifications.

This recognition of a fundamental set of clinical concepts and descriptions, transcending the specific focus of administrative and financial groupings, constitutes a revolution akin to the Copernican recognition of the Sun as the center of our solar system. This new foundation for patient information will underlay virtually all practical clinical information systems in the future, and some in the present.

What we lack, however, is a consistent clinical nomenclature to serve the purpose of this common foundation. While many candidates have been noted, a clear consensus by all interested parties, including provider, payers, systems vendors, government, and of course patient interests has yet to emerge. Much work has appeared which attempts to characterize this[31,32,33]. A National Summit meeting last November made some progress toward establishing an agenda for building interlocking clinical terminologies[34].

Electronic Health Records

The availability of excellent clinical terminologies is not sufficient; they must be pragmatically incorporated into our healthcare delivery process. The era of abstracting or coding data from a larger paper or electronic source is clearly behind us; we can no longer afford the overhead nor forgo the advantages of using well defined information in real time.

Many components of emerging electronic health records should be founded upon the availability of a common terminology server, which can logically and semantically link patient descriptions, needs, requirements, indications, warnings, and knowledge support. These servers should facilitate the direct entry of controlled terms and composite descriptions. This would constitute coded information, quality controlled by the care provider directly and the rigors of the clinical process that will quickly highlight inaccurate or incomplete descriptions.

Thus, the emphasis should not be upon a single terminology or classification, but the suite of terms, tools, and resources needed to bring consistent patient descriptions to the point of care. Corresponding systems should be reflected at the stage of inquiry, which parallel the data entry tasks completely.

The Foundation of Comparable Information

We are emerging into a new information world, with profound impact upon our systems of healthcare. Underpinning this future must be a robust system of clinically describing what we see, do, learn, and discover. These descriptions in turn must be transmissible between systems, enterprises, knowledge resources, and user communities.

More profoundly, we are on the cusp of a realistic ability to learn from our collective experiences on an unprecedented scale and thoroughness. While anecdote, folklore, and vaguely described clinical experience were the backbone of our practice, we can look forward to tapping repositories of consistently collected patient information to engage in outcome analyses on demand. These inquiries can be specifically tailored, just as Blois had envisioned[35], matching the immediate decisions and concerns of the patient with whom we are planning care.

CONCLUSIONS

We can profit from a long and rich tradition of health descriptions, classifications, and nomenclatures. Nevertheless, the realities of a complex health information environment will benefit from common and consistent systems for describing patient findings, diagnoses, and interventions. Such systems are beginning to emerge, as interlocking suites of tools and content based upon logical foundations of description. Concept representation has achieved a central position in Informatics thinking, development, and future needs. Our tasks and opportunities are clear, we need only engage the challenges.

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