

# Standard Specification for Quality Indicators for Controlled Health Vocabularies<sup>1</sup>

This standard is issued under the fixed designation E 2087; the number immediately following the designation indicates the year of original adoption or, in the case of revision, the year of last revision. A number in parentheses indicates the year of last reapproval. A superscript epsilon ( $\epsilon$ ) indicates an editorial change since the last revision or reapproval.

#### **INTRODUCTION**

In 1839, William Farr stated in his First Annual Report of the Registrar-General of Births, Deaths, and Marriages in England, "The nomenclature is of as much importance in this department of inquiry, as weights and measures in the physical sciences, and should be settled without delay." Since that time this theme has been heard resounding from an in increasingly large group of scientists (see Appendix X1). Today, the need for controlled vocabularies to support health record systems has been widely recognized (see Specification E 1238, Guide E 1239, Guide E 1384, Specification E 1633, and EN 12017). Controlled vocabularies provide systems with the means to aggregate data. This aggregation of data can be done at multiple levels of granularity and therefore can enhance the clinical retrieval of a problem oriented record, data pertaining to a classification for billing purposes, or outcomes data for a given population. Maintenance of large-scale vocabularies has become a burdensome problem as the size of term sets has escalated (IS 15188). Without a well-structured backbone, large-scale vocabularies cannot scale to provide the level of interoperability required by today's complex electronic health record applications.

The solution rests with standards (1).<sup>2</sup> Over the past ten or more years, Medical Informatics researchers have been studying controlled vocabulary issues directly. They have examined the structure and content of existing vocabularies to determine why they seem unsuitable for particular needs, and they have proposed solutions. In some cases, proposed solutions have been carried forward into practice and new experience has been gained (2). As we prepare to enter the twenty-first century, it seems appropriate to pause to reflect on this experience, and publish a standard set of goals for the development of comparable, reusable, multipurpose, and maintainable controlled health vocabularies (IS 12200, IS 12620) (3).

This specification represents the initial input taken from the ANSI-HISB Framework Paper by Chute, et al (4), the Desiderata from Cimino (3), the ToMeLo Architecture and Terminology Paper by Rossi-Mori and Zanstra, and the Compositionality Paper by Elkin, et al (5). Other useful references include, "GALEN Generalized Architecture for Language, Encyclopedias and Nomenclatures in Medicine: Univ. of Manchester" (6, 7) and "Unified Medical Language System (UMLS) Knowledge Sources" (8).

### 1. Scope

1.1 This specification covers the documentation of the principal notions necessary and sufficient to assign value to a controlled health vocabulary. This specification will serve as a guide for governments, funding agencies, terminology developers, terminology integration organizations, and the purchasers and users of controlled health terminology systems working toward improved terminological development and recognition

of value in a controlled health vocabulary. It is applicable to all areas of health care about which information is kept or utilized. It is intended to complement and utilize those notions already identified by other national and international standards bodies.

1.2 This specification will provide vocabulary developers and authors with the guidelines needed to construct useful, maintainable controlled health vocabularies. These tenets do not attempt to specify all of the richness that can be incorporated into a health terminology. However this specification does specify the minimal requirements, which, if not adhered to, will ensure that the vocabulary will have limited generalizability and will be very difficult, if not impossible, to maintain. This specification will provide terminology developers with a sturdy starting point for the development of controlled health vocabularies. This foundation serves as the

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basis from which vocabulary developers will build robust, large-scale, reliable and maintainable terminologies.

1.3 This specification explicitly does not refer to classifications or coding systems (for example, a simple list of pairs of rubrics and codes) that are not designed to be used clinically.

# 2. Referenced Documents

- 2.1 ASTM Standards:
- E 1238 Specification for Transferring Clinical Observations Between Independent Computer Systems<sup>3</sup>
- E 1239 Guide for Description of Reservation/Registration-Admission, Discharge, Transfer (R-ADT) Systems for Automated Patient Care Information Systems<sup>3</sup>
- E 1284 Guide for Construction of a Clinical Nomenclature for Support of Electronic Health Records<sup>3</sup>
- E 1384 Guide for Content and Structure of the Electronic Health Record  $(EHR)^3$
- E 1633 Specification for Coded Values Used in the Electronic Health Record<sup>3</sup>
- E 1712 Specification for Representing Clinical Laboratory Procedure and Analyte Names<sup>3</sup>
- 2.2 Other Standards:
- ISO/DIS 860 International Harmonization of Concepts and Terms
- EN 12017 Medical Informatics—Vocabulary
- EN 12264 Medical Informatics—Categorical Structure of Syntax of Concepts—Model for Representation of Semantics
- ICD-9-CM
- IS 704 Principles and Methods of Terminology
- IS 1087-1 Terminology—Vocabulary—Part 1: Theory and Application
- IS 1087-2 Terminology—Vocabulary—Part 2: Computer Applications
- IS 11179-3 Terminology—Data Registries
- IS 12200 Terminology—Computer Applications-Machine Readable Terminology Interchange Format
- IS 12620 Terminology—Computer Applications—Data Categories
- IS 15188 Project Management for Terminology Standardization
- IS 2382–4 Information Technology—Vocabulary—Part 4: Organization of Data
- ISO TR 9789 Guidelines for the Organization and Representation of Data Elements for Data Interchange—Coding Methods and Principles

## 3. General Information

3.1 Basic characteristics of a terminology influence its utility and appropriateness in clinical applications.

3.1.1 *Concept Orientation* (3)—The basic unit of a vocabulary must be a concept, which is the embodiment of some specific meaning and not a code or a character string. Representations of a concept must correspond to one and only one meaning, and in a well-ordered vocabulary only one concept may have that same meaning (ISO/DIS 860). However, mul-

tiple terms (linguistic representations) may have the same meaning if they are explicit representations of the same concept. This implies non-redundancy, non-ambiguity, and non-vagueness.

3.1.1.1 *Non-redundancy*—Terminologies must be internally consistent. There must not be more than one concept in the terminology with the same meaning (IS 704, Guide E 1284). This does not exclude synonymy; rather, it requires that this be explicitly represented.

3.1.1.2 *Non-Ambiguity*—No concept should have two or more meanings. However an entry term (some authors have referred to this as an "interface terminology") can point to more than one concept (for example, MI as a myocardial infarction and mitral insufficiency).

3.1.1.3 *Non-Vagueness*—Concept names must be context free (some authors have referred to this as "context laden"). For example "diabetes mellitus" should not have the child concept "well controlled," instead the child concept's name should be "diabetes mellitus, well controlled."

3.2 *Purpose and Scope*—Any controlled vocabulary must have its purpose and scope clearly stated in operational terms so that it its fitness for particular purposes can be assessed and evaluated (IS 15188). Where appropriate, it may be useful to illustrate the scope by examples or "use cases" as in database models and other specification tools. Criteria such as coverage and comprehensiveness can only be judged relative to the intended use and scope. For example, a vocabulary might be comprehensive and detailed enough for general practice with respect to cardiovascular signs, symptoms, and disorders, but inadequate to a specialist cardiology or cardiothoracic surgery unit. Conversely, a vocabulary sufficiently detailed to cope with cardiology and cardiothoracic surgery might be totally impractical in general practice.

3.3 *Coverage* (3)—Each segment of the healthcare process must have explicit in-depth coverage and not rely on broad summary categories that lump specific clinical concepts together. For example, it is often important to distinguish specific diagnosis from categories presently labeled Not Elsewhere Classified (NEC), or to differentiate disease severity such as indolent prostate cancer from widely metastatic disease. The extent to which the depth of coverage is incomplete must be explicitly specified for each domain (scope) and purpose as indicated in 3.2.

3.4 *Comprehensiveness* (9)—All segments of the healthcare process, such as physical findings, risk factors, or functional status, must be addressed for all related disciplines, across the breadth of medicine, surgery, nursing and dentistry. This criterion applies because decision support, risk adjustment, outcomes research, and useful guidelines require more than diagnoses and procedures. Examples include existing AHCPR guidelines and the HCFA mortality model. The extent to which the degree of comprehensiveness is incomplete must be explicitly specified for each domain (scope) and purpose as indicated in 3.2.

3.5 *Mapping* (10)—Government and payers mandate the form and classification schema for much clinical data exchange. Thus, comprehensive and detailed representations of patient data within computer-based patient records should be

<sup>&</sup>lt;sup>3</sup> Annual Book of ASTM Standards, Vol 14.01.

able to be mapped to those classifications, such as ICD-9-CM. This need for multiple granularities is needed for clinical health care as well (ISO TR 9789). For example, an endocrinologist may specify more detail about a patient's diabetes mellitus than a generalist working in an urgent care setting, even though both may be caring for the same patient. The degree to which the terminology is isolated from other classifications must be explicitly stated.

3.6 Systematic Definitions (4)—In order for users of the vocabulary to be certain that the meaning that they assign to concepts is identical to the meaning which the authors of the vocabulary have assigned, these definitions will need to be explicit and available to the users. Further, as relationships are built into vocabularies, multiple authors will need these definitions to ensure consistency in authorship.

3.7 *Formal Definitions*—A compositional system should contain formal definitions for non-atomic concepts and formal rules for inferring subsumption from the definitions (Specification E 1712).

3.8 *Explicitness of Relations*—The logical definition of subsumption should be defined. The formal behavior of all links/relations/attributes should be explicitly defined. The primary hierarchical relation should be subsumption ("kind of") as defined by logical implication: "B is a kind of A" means "All Bs are As." If a looser meaning such as "broader than/narrower than" is used, it should be explicitly stated.

3.9 *Reference Terminology*—The set of canonical concepts, their structure, relationships, and, if present, their systematic and formal definitions. These features define the core of the controlled health terminology.

3.10 Atomic Reference Terminology—A reference terminology consisting of only atomic concepts and their systematic and formal definitions. In this type of reference terminology, no two or more concepts can be combined to create a composite expression as the same meaning as any other single concept contained in the atomic reference terminology.

3.11 *Colloquial Terminology*—The set of terms that consist of commonly used entry points and which map to one or more canonical terms within the vocabulary. These have been called "entry terms" or "interface terminologies" by different authors.

### 4. Structure of the Terminology Model

4.1 Terminology structures determine the ease with which practical and useful interfaces for term navigation, entry, or retrieval can be supported (IS 704, IS 1087-1, EN 12264). Terminologies that do not currently meet these criteria can be in compliance with this specification by putting mechanisms in place to move toward these goals.

4.2 *Compositionality*—Atomic concepts must be able to be combined to create composite concepts (11). A concept is a notion represented by language, which identifies one idea. For example, "colon cancer" comprises "neoplasm, malignant" and "large bowel" as atomic components. In a compositional system, concept representations can be divided into atomic and composite concept representations. Composite concept representations can be further divided into "named pre-coordinated concept representations" and "post coordinated representation expressions." Within a composite concept, it may be possible to separate the constituents into three categories: the kernel concept, modifier concept, and qualifier (also called "status") concept. These terms are being specifically defined in a document on meta-terminology currently being written under the auspices of ISO TC 215 Working Group 3.

NOTE 1—The term "concept" in this specification is used to refer to the representation of a concept rather than the thought itself.

4.2.1 Atomic Concept—A representation of a concept that is not composed of other simpler concept representations within a particular terminology. In many cases "atomic concepts" will correspond to what philosophers call "natural kinds." Such entities cannot be meaningfully decomposed. Concepts should be separable into their constituent components, to the extent that it is practical. These concepts should form the root basis of all concepts. For example, in the UMLS Metathesaurus, colon is a synonym for large bowel, and cancer is a synonym for neoplasm, malignant. Colon cancer is non-atomic, since it can be broken down into "large bowel" and "neoplasm, malignant." Each of these two more atomic terms has a separate and unique Concept Unique Identifier (CUI).

4.2.2 *Composite Concept*—A concept composed as an expression made up of atomic concepts linked by semantic representations (such as roles, attributes, or links).

4.2.2.1 *Pre-coordinated Concept*—An entity that can be broken into parts without loss of meaning (can be meaningfully decomposed) when the atomic concepts are examined in aggregate. These are representations, which are considered single concepts within the host vocabulary. Ideally, these concepts should have their equivalent composite concepts explicitly defined within the vocabulary (that is, the vocabulary should be normalized for content). For example, colon cancer is non-atomic, however it has a single CUI, which means to the Metathesaurus that it represents a single concept. It has the same status in the vocabulary as the site "large bowel" and the diagnosis "neoplasm, malignant."

4.2.2.2 Post-coordinated Concept—A composite concept is not pre-coordinated and therefore must be represented as an expression of multiple concepts using the representation language. This is the attempt of a system to construct a set of concepts from within a controlled vocabulary to more completely represent a user's query. For example, the concept "bacterial effusion, left knee" is not a unique term within the SNOMED-RT terminology. It represents a clinical concept that some patient has an infected left knee joint. As it cannot be represented by a single concept identifier, to fully capture the intended meaning a system would need to build a representation from multiple concept identifiers or lose information to free text.

4.2.3 *Types of Atomic and Pre-coordinated Concepts*—We can classify unique concept representations within a vocabulary into at least three distinct types: kernel concepts, modifiers, and qualifiers (which contain status concepts). This separation allows user interfaces to provide more readable and therefore more useful presentations of composite concepts.

4.2.3.1 *Kernel Concept*—An atomic or pre-coordinated concept, which represents one of the one or more main concepts within a pre-coordinated or post-coordinated composition.

4.2.3.2 Refining Kernel Concept-Constituents of a composite concept refine the meaning of a kernel concept. For example, "stage 1 a" in "having colon cancer stage 1a," or "brittle, poorly controlled," in "Brittle, poorly controlled diabetes mellitus." In general, these concepts are expressed as a link plus a value ("attribute-value pair"). Terminologies must support a logical structure that can support temporal duration and trend. Attributes must be themselves elements of a terminology and fit into a practical model that extends a terminology. For example, cancers may be further defined by their stage and histology if they have been symptomatic for a specifiable time and if they may progress over a given interval. Attributes are required to capture important data features for structured data entry and are pertinent to secondary data uses such as aggregation and retrieval. Kernel concepts can be refined in many ways, including a clinical sense, a temporal sense, and by status terms (for example, "recurrent").

4.3 Normalization of Content—Normalization is the process of supporting and mapping alternative words and shorthand terms for composite concepts. All pre-coordinated concepts must be mapped to or logically recognizable by all possible equivalent post-coordinated concepts. There should be mechanisms for identifying this synonymy for user created ("new") post-coordinated concepts as well (that is, when there is no pre-coordinated concept for this notion in the vocabulary). This functionality is critical to define explicitly equivalent meaning and to accommodate personal, regional, and discipline-specific preferences. Additionally, the incorporation of non-English terms as synonyms can achieve a simple form of multilingual support.

4.4 Normalization of Semantics—In compositional systems, there exists the possibility of representing the same concept with multiple potential sets of atoms that may be linked by different semantic links. In this case the vocabulary needs to be able to recognize this redundancy/synonymy (depending on your perspective). The extent to which normalization can be performed formally by the system should be clearly indicated. For example, the concept represented by the term "laparoscopic cholecystectomy" might be represented in the following two dissections:

4.4.1 "Surgical Procedure: Excision" {Has Site Gallbladder}, {Has Method Endoscopic} and

4.4.2 "Surgical Procedure: Excision" {Has Site Gallbladder}, {Using Device Endoscope}.

4.5 *Multiple Hierarchies* (12)—Concepts should be accessible through all reasonable hierarchical paths (that is, they must allow multiple semantic parents). For example, stomach cancer can be viewed as a neoplasm or as a gastrointestinal (GI) disease. A balance between number of parents (as siblings) and number of children in a hierarchy should be maintained. This feature assumes obvious advantages for natural navigation of terms (for retrieval and analysis), so a concept of interest can be found by following intuitive paths (users should not have to guess where a particular concept was instantiated).

4.5.1 Consistency of View (13)—A concept in multiple hierarchies must be the same concept in each case. The example of stomach cancer in 4.5must not have changes in

nuance or structure when arrived at via the cancer hierarchy as opposed to GI diseases. Inconsistent views could have catastrophic consequences for retrieval and decision support by inadvertently introducing variations in meaning that may be unrecognized and therefore be misleading to users of the system.

4.6 *Explicit Uncertainty*—Notions of "probable," "suspected," "history of," or differential possibilities (that is, a differential diagnosis list) must be supported. The impact of certain versus very uncertain information has obvious impact on decision support and other secondary data uses. Similarly, in the case of incomplete syndromes, clinicians should be able to record the partial criteria consistent with the patient's presentation. This criterion is listed separately as many current terminological systems fail to address this adequately.

4.7 *Representation*—Computer coding of concept identifiers must not place arbitrary restrictions on the terminology, such as numbers of digits, attributes, or composite elements. To do so subverts meaning and content of a terminology to the limitations of format, which in turn often results in the assignment of a concept to the wrong location because it might no longer "fit" where it belongs in a hierarchy. These reorganizations confuse people and machines alike, as intelligent navigation agents are led astray for arbitrary reasons. The long, sequential, alphanumeric tags used as concept identifiers in the UMLS project of the National Library of Medicine exemplify this principle.

### 5. Maintenance

5.1 Technical choices can impact the capacity of a terminology to evolve, change, and remain usable over time.

5.2 Context Free Identifiers (14)—Unique codes attached to concepts must not be tied to hierarchical position or other contexts; their format must not carry meaning. Because health knowledge is being updated constantly, how we categorize health concepts is likely to change (for example, peptic ulcer disease is now understood as an infectious disease, but this was not always so). For this reason, the code assigned to a concept must not be inextricably bound to a hierarchy position in the terminology, so that we need not change the code as we update our understanding of, in this case, the disease. Changing the code may make historical patient data confusing or erroneous. This notion is the same as non-semantic identifiers.

5.3 *Persistence of Identifiers*—Codes must not be reused when a term becomes obsolete or superseded. Consistency of patient description over time is not possible when concepts change codes; the problem is worse when codes can change meaning. This practice not only disrupts historical analyses of aggregate data, but it can be dangerous to the management of individual patients whose data might be subsequently misinterpreted. This encompasses the notion of concept permanence.

5.4 Version Control (15)—Updates and modifications must be referable to consistent version identifiers. Usage in patient records should carry this version information. Because the interpretation of coded patient data is a function of terminologies that exist at a point in time (16) (for example, AIDS patients were coded inconsistently before the introduction of the term AIDS), terminology representations should specify the state of the terminology system at the time a term is used. Version information most easily accomplishes this, and it may be hidden from ordinary review (IS 15188, IS 12620, IS 1087-2, IS 11179-3, IS 2382/4).

5.4.1 *Editorial Information*—New and revised terms, concepts, and synonyms must have their date of entry or effect in the system, along with pointers to their source or authority, or both. Previous ways of representing a new entry should be recorded for historical retrieval purposes.

5.4.2 *Obsolete Marking*—Superseded entries should be so marked, together with their preferred successor. Because data may still exist in historical patient records using obsolete terms, future interpretation and aggregation are dependent upon a term being carried and cross-referenced to subsequent terms (for example, HTLV III to HIV).

5.5 *Recognize Redundancy*—Authors of these large-scale vocabularies will need mechanisms to identify redundancy when it occurs. This is essential for the safe evolution of any such vocabulary. This implies normalization of concepts and semantics, but specifically addresses the need for vocabulary systems to provide the tools and resources necessary to accomplish this task.

5.6 Language Independence—It would be desirable for terminologies to support non-English presentations. As health care confronts the global economy and multi-ethnic practice environments, routine terminology maintenance must incorporate multilingual support. While substantially lacking the power and utility of machine translation linguistics, this simplistic addition will enhance understanding and use in non-English speaking areas. Questions that need to be addressed: Have there been translations? What is the expected cost of translation?

5.7 *Responsiveness*—The frequency of updates, or subversions, should be sufficiently short to accommodate new codes and repairs quickly. Ideally it should occur weekly.

### 6. Evaluation

6.1 As we seek to understand quality in the controlled vocabularies that we create or use, we need standard criteria for the evaluation of these systems. All evaluations should reflect and specifically identify the purpose and scope of the vocabulary being evaluated. (17)

6.2 *Purpose and Scope*—Important dimensions along which scope should be defined include:

6.2.1 *Clinical Area of Use, Disease Area of Patients, and Expected Profession of Users*—What parts of health care is it intended to be used in and by whom?

6.2.2 *Primary Use*—Includes: reporting for remuneration, management planning, epidemiological research, indexing for bibliographic, Web-based retrieval, recording of clinical details for direct patient care, use for decision support, linking of record to decision support, etc.

6.2.3 *Persistence and Extent of Use*—Some vocabularies are intended, at least initially, primarily for a specific study or a specific site. If a vocabulary is intended to be persistent, there should be a means of updating or some kind of change management.

6.2.4 *Degree of Automatic Inferencing Intended*—Is it intended that classification be automatic? Is it intended that validation on input be possible and, if so, within what limits?

If post-coordinated expressions are to be accepted, what can be inferred about them and what restrictions must be placed on them?

6.2.5 *Transformations (Mappings) to Other Vocabularies*— What transformations/mappings are supported for what intended purpose? For example, transformation for purposes of bibliographic retrieval may require less precision than transformation for clinical usage. What is the sensitivity and specificity of the mappings?

6.2.6 *User/Developer Extensibility*—Is it intended that the vocabulary be extended by users or applications developers? If so, within what limits? If not, what mechanisms are available for meeting new needs as they arise?

6.2.7 *Natural Language Input or Output*—Are they supported for analysis or input? To what level of competence are they supported, for example, stilted telegraphic presentation, idiomatic presentation, etc.?

6.2.8 *Other Functions*—What other functions are intended? Examples include linkage to specific decision support systems, linkage to post-marketing surveillance, etc.

6.2.9 *Current Status*—To what extent is the system intended to be finished or a work in progress? If different components of the terminology are at different stages of completion, how is this indicated?

6.3 Measures of Quality (Terminological Tools):

6.3.1 Interconnectivity (Mapping):

6.3.1.1 To what extent is the vocabulary mappable to other coding systems or reference terminologies?

6.3.1.2 To what extent can the vocabulary accommodate local terminological enhancements?

6.3.1.3 Can the vocabulary server respond to queries sent over a network (LAN, WAN)?

6.3.2 Precision and Recall:

6.3.2.1 What are the vocabulary's precision and recall for mapping diagnoses, procedures, manifestations, anatomy, organisms, etc., against an established and nationally recognized standard query test set? This should be evaluated only within the intended scope and purpose of the vocabulary system.

6.3.2.2 Is a standard search engine used in the mapping process?

6.3.3 Usability:

6.3.3.1 Has the usability of the vocabulary been verified?

6.3.3.2 How have interface considerations been separated from vocabulary evaluation?

6.3.3.3 Is there support for user interfaces? Has an effective user interface been built? Is there a proof of concept? Has the vocabulary been shown to have an effective user interface for its intended use? If not, what questions or issues are outstanding? What is the evidence for speed of entry, accuracy, comprehensiveness, and the like in practice with different approaches?

6.3.3.4 Is there support for computer interfaces and system implementers? Is there a demonstrated proof of concept implementation in software? Can it be shown to be usable for the primary purpose indicated? Have there been cases where interfaces failed?

6.3.4 *Feasibility*—If it is intended for use in an EPR (Electronic Patient Record), what are the options for information storage? Has feasibility been demonstrated?

6.4 *Measures of Quality (Study Design)*—Generalizability (applicability) of any study design reported (evaluating reported evaluations).

6.4.1 What is the vocabulary's healthcare/clinical relevance?

6.4.2 What was the gold standard used in the evaluation?

6.4.3 If published population rates are used for comparison, was the study population comparable to the population from which the rates were derived?

6.4.4 Was the study appropriately blinded?

6.4.5 Was the test set selection randomized or shown in some sense to be a representative sample of the end user population?

6.4.6 Test Location:

6.4.6.1 Was it different from the developer's location?

6.4.6.2 How was the test site suited to the study design? (This includes tools, resources, etc.)

6.4.6.3 With which of the following was the principal investigator associated?

University Academic Medical Center Corporation Hospital Government Agency HMO Private Practice

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Academic Organization

6.4.6.4 Was the principal investigator independent of the vocabulary being evaluated? Was the principal investigator an appropriate individual to direct this research (in terms of credentials, backing from academic or professional bodies, and expertise)? Did the investigator have any conflicts of interest in performing this research?

6.4.7 Sample Size:

6.4.7.1 Was the sample size of sufficient size to show the anticipated effect, should one exist?

6.4.7.2 Who reviewed the statistical methods?

6.4.7.3 Were the specific aims clear?

6.4.8 Personnel:

6.4.8.1 Were the study personnel appropriate?

6.4.8.2 Were there sufficient resources to complete the project in a reasonable period of time?

6.4.9 Reviewers:

6.4.9.1 Number of reviewers if hand review was necessary.

6.4.9.2 Type of reviewer (physician, nurse, other clinician, coder, knowledge engineer).

6.4.9.3 Were the reviewers blinded to the other reviewer's judgments? (Was there independence?)

#### APPENDIX

#### (Nonmandatory Information)

### **X1. HISTORY OF CLASSIFICATION**

X1.1 The present coding practices rely on data methods and principles for terminology maintenance that have changed little since the adoption of the statistical bills of mortality in the mid-17<sup>th</sup> century. (18) The most widely accepted standard for representing patient conditions, ICD9-CM (19), is an intellectual descendent of this tradition. ICD9-CM relies overwhelmingly on a tabular data structure with limited concept hierarchies and no explicit mechanism for synonymy, value restrictions, inheritance or semantic and non-semantic linkages. The maintenance environment for this healthcare classification is a word processor, and its distribution is nearly exclusively paper-based.

X1.2 Significant cognitive advances in disease and procedure representation took place in 1928 at the New York Academy of Medicine, resulting in industry-wide support for what became the Standard Nomenclature of Diseases and Operations. The profound technical innovation was the adoption of a multiaxial classification scheme. (9,12) Now a pathologic process (Inflammation) could be combined with an anatomic site (Oropharynx Component: Tonsil) to form a diagnosis (Tonsillitis). The expressive power afforded by the compositional nature of a multiaxial terminological coding system tremendously increased the scope of tractable terminology, and, additionally, the level of granularity that diagnosis could be encoded about patients. (12,20)

X1.3 The College of American Pathology (CAP) carried the torch further by creating the Systematized Nomenclature of Pathology (SNOP), and subsequently the Systemized Nomenclature of Medicine (SNOMED). In these systems, the number, scope, and size of the compositional structures has increased to the point where an astronomical number of terms can be synthesized from SNOMED atoms. One well-recognized limitation of this expressive power is the lack of syntactic grammar, compositional rules, and normalization of both the concepts and the semantics. Normalization is the process by which the system knows that two compositional constructs with the same meaning are indeed the same (for example, that the term "colon cancer" is equivalent to the composition of "malignant neoplasm" and the site "large bowel"). These are issues addressed by CAP in their efforts to make SNOMED a robust reference terminology for health care. (12,20)

X1.4 Other initiatives of importance are the Clinical Terms v3 (Read Codes), which are maintained and disseminated by the National Health Service in the United Kingdom, and the Galen effort which expresses a very detailed formalism for term description. The Read Codes are composed of a large corpus of terms, now in its third revision, that is hierarchically

designed and is slated for use throughout Great Britain. A development of interesting note is the newly signed agreement of CAP and the NHS to merge the content of SNOMED-RT and Clinical Terms Version 3 into a derivative work (SNOMED—Clinical Terms {SNOMED-CT}).

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