TECHNICAL SPECIFICATION

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Health informatics — Controlled health terminology — Structure and high-level indicators

Informatique de santé — Terminologie contrôlée relative à la santé — Structure et indicateurs de haut niveau

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Foreword

ISO (the International Organization for Standardization) is a worldwide federation of national standards bodies (ISO member bodies). The work of preparing International Standards is normally carried out through ISO technical committees. Each member body interested in a subject for which a technical committee has been established has the right to be represented on that committee. International organizations, governmental and non-governmental, in liaison with ISO, also take part in the work. ISO collaborates closely with the International Electrotechnical Commission (IEC) on all matters of electrotechnical standardization.

International Standards are drafted in accordance with the rules given in the ISO/IEC Directives, Part 3.

The main task of technical committees is to prepare International Standards. Draft International Standards adopted by the technical committees are circulated to the member bodies for voting. Publication as an International Standard requires approval by at least 75 % of the member bodies casting a vote.

In other circumstances, particularly when there is an urgent market requirement for such documents, a technical committee may decide to publish other types of normative document:

— an ISO Publicly Available Specification (ISO/PAS) represents an agreement between technical experts in an ISO working group and is accepted for publication if it is approved by more than 50 % of the members of the parent committee casting a vote;

— an ISO Technical Specification (ISO/TS) represents an agreement between the members of a technical committee and is accepted for publication if it is approved by 2/3 of the members of the committee casting a vote.

An ISO/PAS or ISO/TS is reviewed after three years with a view to deciding whether it should be confirmed for a further three years, revised to become an International Standard, or withdrawn. In the case of a confirmed ISO/PAS or ISO/TS, it is reviewed again after six years at which time it has to be either transposed into an International Standard or withdrawn.

Attention is drawn to the possibility that some of the elements of this Technical Specification may be the subject of patent rights. ISO shall not be held responsible for identifying any or all such patent rights.

ISO/TS 17117 was prepared by Technical Committee ISO/TC 215, *Health informatics*.

Annex B forms a normative part of this Technical Specification. Annex A is for information only.

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 increasingly large group of scientists (see Annex A). Today, the need for controlled terminologies to support health record systems has been widely recognized (E-1238, E-1239, E-1384, E-1633, ENV 12017). Controlled terminologies provide systems with the means to aggregate data. This aggregation of data can be carried out 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 terminologies has become a burdensome problem, as the size of term sets has escalated. Without a well-structured backbone, large-scale terminologies cannot scale to provide the level of interoperability required by today’s complex electronic health record applications.

The solution rests with standards [7]. Over the past ten or more years, medical informatics researchers have been studying controlled terminology issues directly. They have examined the structure and content of existing terminologies 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.[8] As we have entered the twenty-first century, it seems appropriate to pause to reflect on this experience, and to publish a standard set of goals for the development of comparable, reusable, multipurpose and maintainable controlled health terminologies (ISO 12200, ISO 12620).

This Technical Specification is the first deliverable for the ISO/TC 215 *Health informatics*, Working group 3, *Health concept representation*, that is also working on an International Standard to be the basis for future standards in this area. It will serve as a guide for governments, funding agencies, terminology developers, terminology integration organizations and the purchasers and users of controlled health terminology systems toward improved terminological development and recognition of value in a controlled health terminology. This ISO/TS 17117 on quality indicators of controlled health terminologies is based on previous work in ASTM that naturally could not be harmonized with ISO work already in progress. The present work is therefore published as a Technical Specification at this time with the intent to revise it to be compatible with the planned basic terminology standard and converted to a full International Standard after a maximum of three years.

Health informatics — Controlled health terminology — Structure and high-level indicators

1 Scope

This Technical Specification specifies the principal ideas which are necessary and sufficient to assign value to a controlled health terminology. It is applicable to all areas of healthcare about which information is kept or utilized.

2 Normative references

The following normative documents contain provisions which, through reference in this text, constitute provisions of this Technical Specification. For dated references, subsequent amendments to, or revisions of, any of these publications do not apply. However, parties to agreements based on this Technical Specification are encouraged to investigate the possibility of applying the most recent editions of the normative documents indicated below. For undated references, the latest edition of the normative document referred to applies. Members of ISO and IEC maintain registers of currently valid International Standards.

ISO 704, *Terminology work — Principles and methods*

ISO 860, *Terminology work — Harmonization of concepts and terms*

ISO 1087-1, *Terminology work — Vocabulary — Part 1: Theory and application*

ISO 1087-2, *Terminology work — Vocabulary — Part 2: Computer applications*

ISO/IEC 11179-3, *Information technology — Specification and standardization of data elements — Part 3: Basic attributes of data elements*

ISO 12620, *Computer applications in terminology — Data categories*

ISO/IEC 2382-4, *Information technology — Vocabulary — Part 4: Organization of data*

ISO/IEC/TR 9789, *Information technology — Guidelines for the organization and representation of data elements for data interchange — Coding methods and principles*

E-1284, *Standard Guide for Construction of a Clinical Nomenclature for Support of Electronic Health Records*

E-1712, *Standard Specification for Representing Clinical Laboratory Test and Analyte Names*

ENV 12264, *Health Informatics — Categorical Structures of Systems of Concepts — Model for Representation of Semantics*

3 Terms and definitions

For the purposes of this Technical Specification, the following terms and definitions apply.

3.1
terminology
set of terms representing a system of concepts within a specified domain

NOTE This implies a published purpose and scope from which one can determine the degree to which this representation adequately covers the domain specified.

3.2
controlled health terminology
set of terms intended for clinical use

NOTE This implies enough content and structure to provide a representation capable of encoding comparable data, at a granularity consistent with that generated by the practice within the domain being represented, within the purpose and scope of the terminology.

3.3
classification
terminology which aggregates data at a prescribed level of abstraction for a particular domain

NOTE 1 This fixing of the level of abstraction that can be expressed using the classification system is often done to enhance consistency when the classification is to be applied across a diverse user group, such as is the case with some of the current billing classification schemes.

NOTE 2 See Annex A for the history of classification.

3.4
ontology
organization of concepts for which a rational argument can be made

EXAMPLE A hierarchy of qualifiers would be a qualifier ontology.

NOTE Colloquially, this term is used to describe a hierarchy constructed for a specific purpose.

3.5
qualifier
string which, when added to a term, changes the meaning of the term in a temporal or administrative sense

EXAMPLES "History of" or "recurrent".

3.6
modifier
string which, when added to a term, changes the meaning of the term in the clinical sense

EXAMPLES "Clinical stage" or "severity of illness".

3.7
canonical term
preferred atomic or pre-coordinated term for a particular medical concept

3.8
term
word or words corresponding to one or more concepts

4 General

4.1 Basics

The basic characteristics of a terminology influence its utility and appropriateness in clinical applications. Terminologies should be evaluated within the context of their stated scope and purpose and are intended to complement and utilize those notions already identified by other national and international standards bodies.

This Technical Specification explicitly refers only to terminologies that are primarily designed to be used for clinical concept representation or to the aspect of a terminology designed to be used for clinical concept representation. This Technical Specification will also provide terminology developers and authors with the quality guidelines needed to construct useful and maintainable controlled health terminologies. These tenets do not attempt to specify all the richness which can be incorporated into a health terminology. However, this Technical Specification does specify the minimal requirements, which, if not adhered to, will assure that the terminology will have only limited generalizability and will be very difficult, if not impossible, to maintain. Terminologies which do not currently meet these criteria, can be in compliance with this Technical Specification by putting in place mechanisms to move toward these goals. Principles for implementation are specified in Annex B.

This Technical Specification will provide terminology developers with a sturdy starting point for the development of controlled health terminologies. This foundation serves as the basis from which terminology developers will build robust, large-scale, reliable and maintainable terminologies.

4.2 Concept orientation

The basic unit of a terminology shall be a concept, which is the embodiment of some specific meaning and not a code or character string. Identifiers of a concept shall correspond to one and only one meaning and, in a well-ordered terminology, only one concept may have that same meaning, as specified in ISO 860. However, multiple terms (linguistic representations) may have the same meaning if they are explicit representations of the same concept. This implies non-redundancy, non-ambiguity, non-vagueness and internal consistency.

4.2.1 Non-redundancy

Terminologies shall be internally normalized. There shall not be more than one concept identifier in the terminology with the same meaning, as specified in ISO 704 and E-1284. This does not exclude synonymy, rather it requires that this be explicitly represented.

4.2.2 Non-ambiguity

No concept identifier should have more than one meaning. However, an entry term can point to more than one concept.

EXAMPLE MI as myocardial infarction and mitral insufficiency.

NOTE Some authors have referred to entry terms as an interface terminology.

4.2.3 Non-vagueness

Concept names shall be context free.

EXAMPLE “Diabetes mellitus” should not have the child concept “well controlled”, instead the child concept's name should be “diabetes mellitus, well controlled”.

NOTE Some authors have referred to context free as context laden.

4.2.4 Internal consistency

Relationships between concepts should be uniform across parallel domains within the terminology.

EXAMPLE If heart valve structures are specified anatomically, the diagnosis related to each structure is also specified using the same relationships.

4.3 Purpose and scope

Any controlled terminology shall have its purpose and scope clearly stated in operational terms so that its fitness for particular purposes can be assessed and evaluated. 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.

EXAMPLE A terminology 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 terminology sufficiently detailed to cope with cardiology and cardiothoracic surgery might be totally impractical in general practice.

4.3.1 Coverage

Each segment of the healthcare process shall have explicit in-depth coverage, and not rely on broad leaf node categories that place specific clinical concepts together. The extent to which the depth of coverage is incomplete shall be explicitly specified for each domain (scope) and purpose as indicated in 4.3.[9]

EXAMPLE It is often important to distinguish specific diagnosis from categories presently labelled “not elsewhere classified” (NEC), or to differentiate disease severity such as indolent prostate cancer from widely metastatic disease.

4.3.2 Comprehensiveness

The extent to which the degree of comprehensiveness is incomplete shall be explicitly specified for each domain (scope), and purpose as indicated in 4.3. Within the scope and purpose, all aspects of the healthcare process shall be addressed for all related disciplines, such as physical findings, risk factors, or functional status — 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 The existing Agency for Healthcare Research and Quality Guidelines, and the Healthcare Finance Administration (HCFA) mortality model.[10]

4.4 Mapping

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 have the ability to be mapped to those classifications, such as ICD-9. This need for multiple granularities is required for clinical healthcare, as well as is specified in ISO/IEC/TR 9789. The degree to which the terminology is mappable to other classifications shall be explicitly stated.[11]

EXAMPLE An endocrinologist may specify more detail about a patient’s diabetes mellitus than a generalist working in a primary care setting, even though both specialities may be caring for the same patient.

4.5 Systematic definitions

In order for users of the terminology to be certain that the meaning that they assign to concepts is identical to the meaning which the authors of the terminology have assigned to these definitions will need to be explicit and available to the users. Further, as relationships are built into terminologies, multiple authors will need these definitions to ensure consistency in authorship.

EXAMPLE The clinical concept “hypertension” might be defined as a consistently elevated blood pressure and needs to be distinguished from a single “BP > 140/85”.

4.6 Formal definitions

A compositional system should contain formal definitions for non-atomic concepts and formal rules for inferring subsumption from the definitions, as specified in E-1712.

4.7 Explicitness of relations

The logical definition of subsumption should be defined. The formal behaviour of all links/relations/attributes should be explicitly defined. If a looser meaning such as “broader than/narrower than” is used, it should be explicitly stated.

EXAMPLE The primary hierarchical relation should be subsumption as exemplified by logical implication: B is a kind of A means all Bs are As.

4.8 Reference terminologies

The set of canonical concepts, their structure, relationships and, if present, their systematic and formal definitions define the core of the controlled health terminology.

4.9 Atomic reference terminologies

In a reference terminology consisting of only atomic concepts and their systematic definitions, no two or more concepts can be combined to create a composite expression which has the same meaning as any other single concept contained in the atomic reference terminology.

4.10 Colloquial terminologies

The set of terms, which consists of commonly used entry points, maps to one or more canonical terms within the terminology.

NOTE These have been called “entry terms” or “interface terminologies” by different authors.

5 Structure of the terminology model

5.1 Terminology structures

Terminology structures determine the ease with which practical and useful interfaces, for term navigation, entry, or retrieval can be supported, as specified in ISO 704, ISO 1087-1 and ENV 12264.

5.2 Compositional terminologies

5.2.1 Compositionality

Composite concepts are created from atomic and pre-coordinated concepts and shall have the ability to be combined to create compositional expressions[12].

EXAMPLE “Colon cancer” comprises “malignant neoplasm” 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: “kernel concept”, “qualifier (also called “status”) concept”, and “modifier concept”.

NOTE A concept is a notion represented by language, which identifies one idea. However, the term “concept” in this Technical Specification is used to refer to the representation of a concept rather than to the thought itself.

5.2.1.1 Atomic concept

An atomic concept is 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 an entity cannot be meaningfully decomposed. Concepts should be separable into their constituent components, to the extent practical. These should form the root basis of all concepts.

EXAMPLE In SNOMED-RT, “colon” is a synonym for “large bowel” and “cancer” is a synonym for “neoplasm, malignant”. Therefore, the term “colon cancer” is non-atomic as it can be broken down into “large bowel” and “neoplasm, malignant”. Each of these two atomic terms has a separate and unique concept identifier, as does the pre-coordinated term “colon cancer”.

5.2.1.2 Composite concept

A composite concept is composed as an expression made up of atomic concepts linked by semantic relations (such as roles, attributes or links).

5.2.1.2.1 Pre-coordinated concept

Such an entity 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 terminology (that is the terminology should be normalized for content, as indicated in 5.2.2).

EXAMPLE The term “colon cancer” is non-atomic, however it has a single unique identifier, which means to the SNOMED-RT that it represents a “single” concept. It has the same status in the terminology as the site “large bowel” and the diagnosis “neoplasm, malignant”.

5.2.1.2.2 Post-coordinated concept

A post-coordinated concept is a composite concept, which is not pre-coordinated and therefore shall 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 terminology to more completely represent a user’s query.

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.

5.2.1.3 Types of atomic and pre-coordinated concepts

Unique concept representations can be classified within a terminology 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.

5.2.1.3.1 Kernel concept

This is an atomic or pre-coordinated concept, which represents one of the one or more main concepts within a pre-coordinated or post-coordinated composition.

5.2.1.3.2 Modifiers and qualifiers

Constituents of a composite concept that refine the meaning of a kernel concept are known as modifiers or qualifiers.

EXAMPLE 1 “Stage 1a” in the expression “having colon cancer stage 1a” or “brittle, poorly controlled” in the expression “brittle, poorly controlled diabetes mellitus” are examples of qualifiers and modifiers.

In general, these concepts are expressed as a link plus a value (“attribute-value-pair”). Terminologies shall support a logical structure that can support temporal duration and trend. Attributes shall be themselves elements of a terminology, and fit into a practical model that extends a terminology.

EXAMPLE 2 Cancers may be further defined by their stage and histology, have been symptomatic for a specifiable time, and may progress over a given interval.

Attributes are required to capture important data features for structured data entry and pertinent to secondary data uses such as aggregation and retrieval. Kernal concepts can be refined in many ways, including a clinical sense, a temporal sense and by status terms, such as “recurrent”.

5.2.2 Normalization of content

Normalization is the process of supporting and mapping alternative words and shorthand terms for composite concepts. All pre-coordinated concepts shall 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 (i.e. when there is no pre-coordinated concept for this notion in the terminology). This functionality is critical to define explicitly equivalent meaning, and to accommodate personal, regional and discipline specific preferences. Additionally, the incorporation of terms as synonyms, represented in a language other than that primarily used in the host terminology, can achieve a simple form of multilingual support.

5.2.3 Normalization of semantics

In compositional systems, there exists the possibility of representing the same concept with multiple potential sets of atoms, which may be linked by different semantic links. In this case, the terminology 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.

EXAMPLE The concept represented by the term “Laparoscopic Cholecystectomy” might be represented in the following two dissections:

Surgical Procedure: Excision”{Has Site Gallbladder}, {Has Method Endoscopic}

and

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

5.2.4 Multiple hierarchies

Concepts should be accessible through all reasonable hierarchical paths, i.e they shall allow multiple semantic parents. 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), as a concept of interest can be found by following intuitive paths (i.e. users should not have to guess where a particular concept was instantiated).[13]

EXAMPLE One example of multiple semantic parentage is “stomach cancer” which can be viewed as a “neoplasm” or as a “gastrointestinal disease”.

5.2.5 Consistency of view

A concept in multiple hierarchies shall be the same concept in each case. The previous example of stomach cancer in 5.2.4 shall not have changes in nuance or structure when arrived at via the cancer hierarchy as opposed to the gastrointestinal disease hierachy. Inconsistent views could have catastrophic consequences for retrieval and decision support, by inadvertently introducing variations in meaning which may be unrecognized and therefore be misleading to users of the system.[14]

5.2.6 Explicit uncertainty

Notions of “probable”, “suspected”, “history of” or differential possibilities such as a differential diagnosis list, shall 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. These criteria are listed separately, as many current terminological systems fail to address this area adequately.

5.2.7 Representational form

The representational form of the identifiers within the terminology should be meaningless. Computer coding of concept identifiers shall 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 concepts 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 well this principle.

6 Maintenance

6.1 Basics

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

6.1.1 Context-free identifiers

Unique codes attached to concepts shall not be tied to hierarchical position or other contexts; their format shall not carry meaning. Because health knowledge is being constantly updated, the categorization of health concepts is likely to change. For this reason, the “code” assigned to a concept shall not be inextricably bound to a hierarchy position in the terminology, so that the code need not change when concepts are hierarchically reorganized. Changing the “code” may make historical patient data confusing or erroneous.

EXAMPLE “Peptic ulcer disease” is now understood as an infectious disease, but this was not always so.

NOTE This notion of context-free identifiers is the same as non-semantic identifiers.[15]

6.1.2 Persistence of identifiers

Codes shall not be reused when a concept is 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 can be dangerous to the management of individual patients whose data might be subsequently misinterpreted.

NOTE This encompasses the notion of concept permanence.

6.1.3 Version control

Updates and modifications shall be referable to consistent version identifiers. Usage in patient records should carry this version information. This is true because the interpretation of coded patient data is a function of terminologies that exist at a point in time.

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 may be hidden from ordinary review, as specified in ISO 12620, ISO 1087-2, ISO 11179-3 and ISO/IEC 2382-4.[16], [17]

6.1.3.1 Editorial information

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

6.1.3.2 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, their future interpretation and aggregation are dependent upon that term being carried and cross-referenced to subsequent terms.

EXAMPLE Human T-cell leukemia virus — type III (HTLV III) to human immunodeficiency virus (HIV).

6.1.4 Recognize redundancy

Authors of these large-scale terminologies will need mechanisms to identify redundancy when it occurs. This is essential for the safe evolution of any such terminology. This implies normalization of concepts and semantics (see 5.2.2 and 5.2.3), but specifically addresses the need for terminology systems to provide the tools and resources necessary to accomplish this task.

6.1.5 Language independence

It would be desirable for terminologies to support multilingual presentations. As healthcare confronts the global economy and multiethnic practice environments, routine terminology maintenance shall incorporate multilingual support. While substantially lacking the power and utility of machine translation linguistics, this simplistic addition will enhance understanding and use globally. Have there been translations? What is the expected cost of translation?

6.1.6 Responsiveness

The frequency of updates, or sub-versions, should be sufficiently short to accommodate new codes and repairs quickly, ideally in the order of weeks.

7 Evaluation

7.1 Basics

As we seek to understand quality in the controlled terminologies that are created or used, a standard criterion for the evaluation of these systems is needed. All evaluations shall reflect and specifically identify the purpose and scope of the terminology being evaluated.[18]

7.1.1 Measures of purpose and scope

Important dimensions along which purpose and scope should be defined to address the following issues.

7.1.1.1 Clinical area

What is the clinical area of use of the terminology, the disease area of patients addressed and/or the expected profession of users? Within what parts of healthcare is the terminology intended to be used and by whom?

7.1.1.2 Primary use

What is the primary intended usage of the terminology?

EXAMPLE Some areas of usage include: 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.

7.1.1.3 Persistence and extent of use

While some terminologies are intended, at least initially, primarily for a specific study or a specific site, others are not. If intended to be persistent, what are the means of updating or change management, etc.?

7.1.1.4 Degree of automatic inferencing intended

Developers should define whether or not and to what degree automatic inferencing is intended. Developers should define whether or not classification is intended to be automatic. Developers should define whether or not it is intended that validation on input be possible and within what limits. Developers should define whether or not post-coordinated expressions are to be accepted and if so, what can be inferred about them and what restrictions shall be placed on them (i.e. is formal sanctioning required?).

7.1.1.5 Transformations (mappings) to other terminologies

What transformations (mappings) are supported and for what intended purpose? What is the sensitivity and specificity of the transformations?

EXAMPLE Transformation for purposes of bibliographic retrieval may require less precision than transformation for clinical usage.

7.1.1.6 User/developer extensibility

Is it intended that the terminology be extended by users or application developers? If so, within what limits? If not, what mechanisms are available for meeting new needs as they arise?

7.1.1.7 Natural language

Is natural language input or output supported (for analysis or input)? To what level of accuracy?

7.1.1.8 Other functions

What other functions are intended?

EXAMPLE Linkage to specific decision support systems, linkage to post-marketing surveillance, etc.

7.1.1.9 Current status

To what extent is the system intended to be “finished” or work in progress? If different components of the terminology are at different stages of completion, how is this indicated?

7.1.2 Measures of quality, terminological tools

7.1.2.1 Interconnectivity (mapping)

7.1.2.1.1 Terminology and other coding systems

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

7.1.2.1.2 Terminology and terminological enhancements

To what extent can the terminology accommodate local terminological enhancements?

7.1.2.1.3 Terminology and networking

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

7.1.2.2 Precision and recall

7.1.2.2.1 Terminology

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

7.1.2.2.2 Search engine

Is a standard search engine used in the mapping process?

7.1.2.3 Usability

7.1.2.3.1 Validation of usability

Has the usability of the terminology been verified?

7.1.2.3.2 Interface considerations

How have interface considerations been separated from terminology evaluation?

7.1.2.3.3 Prototypes

Has an effective user interface been built? Has the terminology been shown to have an effective user interface for its intended use? If not, what are the questions or issues outstanding? Is there evidence for speed of entry, accuracy, comprehensiveness in practice etc. with different approaches? If not, is there proof of concept?

7.1.2.3.4 Application programmer interfaces

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

7.1.2.4 Feasibility

If the terminology is intended for use in an electronic patient record (EPR), what are the options for information storage? Has feasibility been demonstrated?

7.1.3 Other measures of quality

The generalizability (applicability) of any study design reported (evaluating reported evaluations) should have the ability to be evaluated.

7.1.3.1 Healthcare/clinical relevance

What is the terminology's healthcare/clinical relevance?

7.1.3.2 Gold standard

What was the gold standard used in the evaluation?

7.1.3.3 Study population

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

7.1.3.4 Specific aims

Were the specific aims clear?

7.1.3.5 Blinding

Was the study appropriately blinded?

7.1.3.6 Randomization

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

7.1.3.7 Test location

7.1.3.7.1 Independence

Was the test location different from the developer's location?

7.1.3.7.2 Appropriate for study design

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

7.1.3.7.3 Principal investigator associations

Was the principal investigator associated with a:

— university;

— academic medical centre;

— corporation or company;

— hospital;

— government agency;

— primary care centre (health maintenance organization);

— private practice;

— academic organization?

7.1.3.7.4 Principal investigator

Was the principal investigator independent of the terminology being evaluated? Does the principal investigator have a track record of publication in this field of study? Have there been any conflicts of interest in performing this research?

7.1.3.8 Project completion

Was the project completed in a reasonable period of time?

7.1.3.9 Sample size

Power: was the sample size sufficient to show the anticipated effect, should one exist? Statistics: who reviewed the statistical methods?

7.1.3.10 Personnel

7.1.3.10.1 Training level

What is the average level of training and experience of the study personnel?

7.1.3.10.2 Reviewers

Variability: what is the inter-reviewer variability? Type: what was the type of reviewer (physician, nurse, other clinician, coder, knowledge engineer) used in the study? Independence: were the reviewers blinded to the other reviewers' judgments (i.e. reviewer independence)?

Annex A
(informative)

History of classification

A.1 History of classification

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-seventeenth century.[19] The most widely accepted standard for representing patient conditions, ICD-9-CM[20], is an intellectual descendent of this tradition. ICD-9-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.

The first edition of Physicians' Current Procedural Terminology (CPT) appeared in 1966. In the United States, CPT is the coding system used by Medicare and virtually all third-party payers, including workers compensation and Medicaid. As part of the Medicare Part B physician payment schedule, CPT codes are associated with the Resource Based Relative Value Scale (RBRVS) and used to determine payment for services. The CPT code set is Level I of the Health Care Financing Administration Common Procedure Coding System (HCPCS). The CPT code set, currently in its fourth edition, contains numeric modifiers, notes, guidelines and an index designed to provide explanatory information and facilitate the correct usage of the coding system. The American Medical Association (AMA) is currently working to develop the next generation of CPT (CPT-5). Similar limitations exist in the maintenance environment of ICD-10 — the 10th revision of the International Statistical Classification of Diseases and Related Health Problems, which is being adopted as the national standard for diagnosis coding in an increasing number of countries.

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.[10], [13] Now a pathologic process (e.g. inflammation) could be combined with an anatomic site (e.g. oropharynx component: tonsil) to form a diagnosis (e.g. 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 our patients.[13]

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 have 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 (e.g. 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 healthcare.[13], [21]

Other initiatives of importance are the Clinical Terms v3 (Read Codes), which are maintained and disseminated by the National Health Service (NHS) in the United Kingdom and the Galen effort, which expresses a very detailed formalism for term description. The Read Codes are a large corpus of terms, which is now in its third revision that is hierarchically designed and is slated for use throughout Great Britain. The Unified Medical Language System and its metathesaurus have been developed in cooperation with the United States National Library of Medicine and provides an interlingua between many existing healthcare terminologies. 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 (announced in April 1999), which will be named SNOMED Clinical Terms.

Annex B
(normative)

Principles for implementation

B.1 General

B.1.1 Basics

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

B.1.2 Concept orientation

Is the terminology concept oriented? This shall be the case.[9] To how many meanings can one identifier correspond?

B.1.2.1 Non-redundancy

Can concepts be redundantly instantiated within the terminology? This shall not be the case.

B.1.2.2 Non-ambiguity

Can concepts be ambiguous? This shall not be the case.

B.1.2.3 Non-vagueness

Are concept definitions independent of their context? This shall be the case.

B.1.2.4 Internal consistency

Are the relationships used in the terminology applied consistently? This shall be the case.

B.1.3 Purpose and scope

What is the purpose of the terminology? What is the scope of the terminology? These are to be stated in operational terms (i.e. what functions is the terminology intended to serve?).

B.1.3.1 Coverage

What is the intended coverage of the terminology?[22]

B.1.3.2 Comprehensiveness

What is the degree of comprehensiveness (expressed in per cent completion) of the terminology within the intended area of coverage? What studies can be referenced to support this assertion? (Use the criteria given in clause 4 for assessing the validity and generalizability of the study referenced.)[23]

B.1.4 Mapping

Is the terminology mappable to classifications or other terminologies? If so, which ones? If it is partially mappable to some classifications or other terminologies, to what extent is this true (expressed in per cent completion)? (Use the criteria given in clause 4 to assess the validity and generalizability of the study referenced.)[24]

B.1.5 Systematic definitions

Are the meanings of each specific concept within the terminology made available for the users? These should be provided.

B.1.6 Formal definitions

Does your terminology support formal definitions? If so, to what extent (expressed in per cent completion) is it fully defined? What studies can be referenced to support this assertion? (Use the criteria given in clause 4 to assess the validity and generalizability of the study referenced.) It is essential that reference terminologies support formal definitions.

B.1.7 Explicitness of relations

Does your terminology support formal subsumption? To what extent are the hierarchies automatically generated by the description logic (expressed as a percentage of all the concepts contained in the terminology)? This is a desirable characteristic.

B.1.8 Reference terminologies

Is the terminology intended to be used as a reference terminology?

B.1.9 Atomic reference terminologies

Is there an explicit mechanism for identifying the atomic portion of the reference terminology? Is it intended that pre-coordinated terms can be used within compositional expressions? This should be a goal of all reference terminologies.

B.1.10 Colloquial terminologies

Specifically, what is the association between the colloquial terms and the reference terminology? How are these two terminologies maintained so as not to create ambiguous or redundant instantiation of data? This is necessary for all reference terminologies intended to be used clinically.

B.2 Structure of the terminology model

B.2.1 Terminology structures

Terminology structures determine the ease with which practical and useful interfaces, for term navigation, entry, or retrieval can be supported, as specified in ISO 704, ISO 1087-1 and ENV 12264.

B.2.1.1 Compositional terminologies

B.2.1.1.1 Compositionality

Does the terminology support the creation of compositional expressions? How is a compositional expression created? If this is governed by rules, they are to be elaborated. If so, can equivalence be identified between arbitrary compositional expressions and by what method?

B.2.1.1.1.1 Atomic concept

Do the authors of the terminology make explicit which concepts are atomic?

B.2.1.1.1.2 Composite concept

A concept composed as an expression is made up of atomic concepts linked by semantic relations (such as roles, attributes or links).

Pre-coordinated concept: do the authors of the terminology make explicit which concepts are pre-coordinated? This shall be true for all compositional terminologies.

Post-coordinated concept: does the terminology support the creation of post-coordinated expressions?

B.2.1.1.1.3 Types of atomic and pre-coordinated concepts

Unique concept representations can be classified within a terminology 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.

Kernel concept: does the terminology identify kernel concepts separately? This should be identified by compositional terminologies.

Modifiers and qualifiers are terms which refine the meaning of a kernal concept. Does the terminology identify modifiers and qualifiers within the terminology? If so, how are they used? This should be identified by compositional terminologies.

B.2.1.1.2 Normalization of content

Is the content of the terminology normalized? What studies can be referenced to support this assertion? (Use the criteria given in clause 7 for assessing the validity and generalizability of the study referenced.) This shall be accomplished for all compositional terminologies.

B.2.1.1.3 Normalization of semantics

Are the semantics of the terminology normalized? What studies can be referenced to support this assertion? (Use the criteria given in clause 7 for assessing the validity and generalizability of the study referenced.) For compositional expressions, is it possible to represent the same concept with different semantics? This shall be accomplished for all compositional terminologies.

B.2.1.1.4 Multiple hierarchies

Are multiple hierarchies supported? Are they present within the current version of the terminology?[25]

B.2.1.1.5 Consistency of view

Is a consistency of views into the terminology maintained? This shall be the case for terminologies that support multiple hierarchies.[26]

B.2.1.1.6 Explicit uncertainty

Does the terminology support the input of explicit uncertainty and incomplete syndromes? This should be a feature of compositional terminologies.

B.2.1.1.7 Representational form

Does the representational form of the concept identifier place restrictions on the terminology? If so, what are the restrictions? This shall not be the case.

B.3 Maintenance

B.3.1 Basics

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

B.3.1.1 Context-free identifiers

Does the terminology support context free identifiers? This shall be the case.[27]

B.3.1.2 Persistence of identifiers

Are codes ever reused for different concepts? If so, when can this occur? This shall not be the case.

B.3.1.3 Version control

Are the terminology's codes tied explicitly to the version of the terminology? This shall be the case.[28]

B.3.1.3.1 Editorial information

When the terminology is revised, is the date of the update and the source or authority of the information leading to the update recorded? This shall be the case.

B.3.1.3.2 Obsolete marking

Have obsolete markings been included in the terminological entries? This shall be the case.

B.3.1.4 Recognize redundancy

Does the terminology recognize redundancy? This shall be the case. If so, how is this accomplished?

B.3.1.5 Language independence

Is the terminology presently multilingual? This should be the case. If not, does it have the capacity to become multilingual? If so, an explanation is to be given.

B.3.1.6 Responsiveness

What is the frequency of updates to the terminology? Is it less than or equal to 12 weeks? This should be the case.

B.4 Evaluation

B.4.1 Basics

As we seek to understand quality in the controlled terminologies that are created or used, a standard criteria for the evaluation of these systems is needed. All evaluations shall reflect and specifically identify the purpose and scope of the terminology being evaluated.[29] These criteria stipulate the methods for evaluating studies, which make

claims regarding controlled terminologies. These criteria are also useful as a guide to individuals or organizations wishing to perform valid and useful evaluations of one or more controlled health terminologies.

B.4.1.1 Measures of purpose and scope

Important dimensions along which purpose and scope should be defined to address the following issues.

B.4.1.1.1 Clinical area

What is the clinical area of use of the terminology, the disease area of patients addressed and/or the expected profession of users? Within what parts of healthcare is the terminology intended to be used and by whom?

B.4.1.1.2 Primary use

What is the primary intended usage of the terminology?

EXAMPLE Some areas of usage include: 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.

B.4.1.1.3 Persistence and extent of use

Is the intent of the terminology to persist and evolve? If intended to be persistent, what are the means of updating or change management, etc.?

B.4.1.1.4 Degree of automatic inferencing intended

Is the terminology intended to support automated classification? Is it intended that validation on input be possible, and within what limits? Are post-coordinated expressions to be accepted and, if so, what can be inferred about them and what restrictions shall be placed on them?

B.4.1.1.5 Transformations (mappings) to other terminologies

What transformations (mappings) are supported and for what intended purpose (e.g. transformation for purposes of bibliographic retrieval may require less precision than transformation for clinical usage)? What is the sensitivity and specificity of the transformations?

B.4.1.1.6 User/developer extensibility

Is it intended that the terminology be extended by users or application developers? If so, within what limits? If not, what mechanisms are available for meeting new needs as they arise?

B.4.1.1.7 Natural language

Is natural language input or output supported (for analysis or input)? To what level of accuracy?

B.4.1.1.8 Other functions

What other functions are intended?

EXAMPLE Linkage to specific decision support systems, linkage to post-marketing surveillance, etc.

B.4.1.1.9 Current status

To what extent is the system intended to be “finished” or work in progress? If different components of the terminology are at different stages of completion, how is this indicated?