International Standard

ISO 17117-1

Health informatics — Terminological resources —

Part 1: Characteristics

Informatique de santé — Ressources terminologiques —

Partie 1: Caractéristiques

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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.

The procedures used to develop this document and those intended for its further maintenance are described in the ISO/IEC Directives, Part 1. In particular, the different approval criteria needed for the different types of ISO documents should be noted. This document was drafted in accordance with the editorial rules of the ISO/IEC Directives, Part 2 (see www.iso.org/directives).

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This document was prepared by Technical Committee ISO/TC 215, Health informatics, in collaboration with the European Committee for Standardization (CEN) Technical Committee CEN/TC 251, Health informatics, in accordance with the Agreement on technical cooperation between ISO and CEN (Vienna Agreement).

This second edition cancels and replaces the first edition (ISO 17117-1:2018), which has been technically revised.

The main changes are as follows:

— The document has been updated to reflect the state of medical and technical practices at a time, and incorporating relevant new normative references.

A list of all parts in the ISO 17117 series can be found on the ISO website.

Any feedback or questions on this document should be directed to the user's national standards body. A complete listing of these bodies can be found at www.iso.org/members.html.

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Introduction

Health terminology is complex and multifaceted. It has been estimated that up to 45 million different terms are needed to adequately describe health-related concepts such as conditions of patients and populations, actions in healthcare, and related concepts, such as medicines, biomedical molecules, genes, organisms, technical methods and social concepts (see ISO 17115). Many formal and less formal terminological resources exist to represent this complexity. These can be called terminological systems, coding systems, formal concept representation systems, classification systems, and others. Specific features of different terminological resources make them more or less useful for particular purposes and technological environments.

The need for formal terminological resources to support health information management has been widely recognized.[13][14][15] Such resources are required for precise data collection, accurate interpretation of data and interoperability among information systems that exchange such data.[14] National governments, healthcare organizations and others are currently concerned with the question of which of the available terminological resources will meet their requirements, i.e. they wish to 'assign value' to specific terminological resources to decide which are suitable for their purposes and healthcare contexts.

A set of criteria to support such evaluations was originally published in ISO/TS 17117:2002[1)]. The main purpose was to enable users to assess whether a terminological resource has the characteristics that will support their specified requirements, since the characteristics of a terminological resource influence its utility and appropriateness in applications. There has been much progress in the study and use of terminological resources since that time and some experience of formal evaluations.[16][17]

As the first part of the entire revision work, this document identifies the characteristics of terminological resources in healthcare (Clause 4) and functions or roles invoked by those characteristics (Clause 5). This document also provides a framework to identify different types of terminological resources using a combination of those characteristics and functions, which is essential for the development of criteria for the categorization of terminological resources in healthcare. Requirements for, and evaluation criteria of, terminological resources in healthcare, addressed in other parts of ISO 17117, are tightly related to the characteristics of terminological resources and functions that they can provide.

The target groups for this document are:

a) organizations wishing to select terminological systems for use in healthcare information systems;

b) developers of terminological systems;

c) developers of terminology standards;

d) those undertaking independent evaluations or academic reviews of terminological resources;

e) terminology Registration Authorities.

1) Withdrawn.

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Health informatics — Terminological resources —

Part 1: Characteristics

1 Scope

This document defines universal and specialized characteristics of health terminological resources that make them fit for the purposes required of various applications. It covers only terminological resources that are primarily designed to be used for clinical concept representation or to those parts of other terminological resources designed to be used for clinical concept representation.

This document helps users to assess whether a terminology has the characteristics or provides the functions that will support their specified requirements. In order to do that, this document focuses on defining characteristics and functions of terminological resources in healthcare that can be used to identify different types of terminological resources for categorization purposes.

NOTE 1 Categorization of healthcare terminological systems according to the name of the system might not be helpful and has caused confusion in the past.

The following aspects are not covered in this document:

— evaluations of terminological resources;

— health service requirements for terminological resources and evaluation criteria based on the characteristics and functions;

— the nature and quality of mappings between different terminologies;

NOTE 2 It is unlikely that a single terminology will meet all the terminology requirements of a healthcare organization: some terminology providers produce mappings to administrative or classification systems such as the International Classification of Diseases (ICD). The presence of such maps would be a consideration in the evaluation of the terminology.

— the nature and quality of mappings between different versions of the same terminology;

NOTE 3 To support data migration and historical retrieval, terminology providers can provide maps between versions of their terminology. The presence of such maps would be a consideration in the evaluation of the terminology.

— terminology server requirements and techniques and tools for terminology developers;

— characteristics for computational biology terminology.

2 Normative references

There are no normative references in this document.

3 Terms and definitions

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

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ISO and IEC maintain terminology databases for use in standardization at the following addresses:

— ISO Online browsing platform: available at https://www.iso.org/obp

— IEC Electropedia: available at https://www.electropedia.org/

3.1 General terms

3.1.1
concept
unit of knowledge created by a unique combination of *characteristics* (3.1.3)

Note 1 to entry: Informally, the term 'concept' is often used when what is meant is 'concept representation'. However, this leads to confusion when precise meanings are required. Concepts arise out of human individuals and social conceptualizations of the world around them. Concept representations are artefacts constructed of symbols.

Note 2 to entry: Concept representations are not necessarily bound to particular languages. However, they are influenced by the social or cultural context of use often leading to different categorizations.

[SOURCE: ISO 1087:2019, 3.2.7, modified — The notes to entry were changed.]

3.1.2
term
linguistic representation of a *concept* (3.1.1) in a specific *subject field* (3.1.15)

3.1.3
characteristic
abstraction of a property

EXAMPLE 'Being a disease of the nervous system' is a characteristic of the concept 'Functional parkinsonism'.

Note 1 to entry: Characteristics are used for describing *concepts* (3.1.1).

[SOURCE: ISO 1087:2019, 3.2.1, modified — The example was changed.]

3.1.4
term identifier
sequence of letters, numbers or symbols, capable of uniquely identifying a *term* (3.1.2) within the *terminological resource* (3.4.1)

Note 1 to entry: Term identifier shall be unique within the terminological resource.

3.1.5
concept identifier
canonical expression (3.3.5), or sequence of letters, numbers or symbols, capable of uniquely identifying a *concept* (3.1.1) within the *terminological resource* (3.4.1)

Note 1 to entry: Concept identifier shall be unique within the terminological resource, so terms shall not be used as concept identifiers in case polysemy exists.

3.1.6
code
identifier expressed by a series of letters, numbers, or symbols

Note 1 to entry: A code is a *concept identifier* (3.1.5) when used in a *coding system* (3.4.4).

3.1.7
terminological resource identifier
unique permanent identifier of a *terminological resource* (3.4.1) for use in information interchange

Note 1 to entry: This is equivalent to Health Coding Scheme Designator in EN 1068:2005 for registration of coding systems.

Note 2 to entry: Globally unique schemes such as OIDs, UUIDs, and URIs may be used for this purpose.

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3.1.8
terminological resource version identifier
version identifier
identifier assigned to a version under which a *terminological resource* (3.4.1) is published or updated

3.1.9
coding scheme
collection of rules that map the elements in one set, the 'coded set', onto the elements in a second set, 'the code set'

Note 1 to entry: The two sets are not part of the coding scheme.

3.1.10
composite characteristic
representation of a *characteristic* (3.1.3)

EXAMPLE has Cause Bacteria; Location = LeftUpperLobeOfLung.

Note 1 to entry: Typically expressed by a *semantic link* (3.2.5) and a *characterizing concept* (3.1.11).

3.1.11
characterizing concept
concept (3.1.1) that is referenced by a *semantic link* (3.2.5) in a *composite characteristic* (3.1.10)

EXAMPLE 'Bacterium' in the construct 'Disease that has Cause Bacterium'; 'Yellow' in the construct 'Skin Lesion that has Colour Yellow'.

3.1.12
characterizing generic concept
characterizing category
value domain
formal category whose specialization by a *domain constraint* (3.1.14) is allowed to be used as *characterizing concept* (3.1.11) in a particular context

EXAMPLE 1 <INFECTIOUS_ORGANISM> = {bacterium, virus, parasite}, in the context of 'infection that has Cause INFECTIOUS_ORGANISM'.

EXAMPLE 2 has Cause Bacteria; Location = LeftUpperLobeOfLung.

Note 1 to entry: The context includes a superordinate concept and a *semantic link* (3.2.5).

3.1.13
sanctioned characteristic
formal representation of a type of *characteristics* (3.1.3)

EXAMPLE 1 performed Using <INSTRUMENT>; hasLocation <BodyPartOfImplantedDevice>.

EXAMPLE 2 'Cause Of Inflammation can Be set {bacteria, virus, parasite, autoimmune, chemical, physical}', where 'can Be' is the semantic link, and 'set {bacteria, virus, parasite, autoimmune, chemical, physical}' is the characterizing generic concept.

Note 1 to entry: A sanctioned characteristic is typically made up of a combination of a *semantic link* (3.2.5) and a *characterizing generic concept* (3.1.12), and can be used in *domain constraints* (3.1.14).

3.1.14
domain constraint
sanction rule prescribing the set of *sanctioned characteristics* (3.1.13) that are valid to specialize a *concept* (3.1.1) in a particular *subject field* (3.1.15)

EXAMPLE 'Infection possibly has Location Skeletal Structure' describes that an infection in a certain context can be located in a structure that is a kind of skeletal structure.

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Note 1 to entry: The rule describes the set of potential *characteristics* (3.1.3) by combining the *semantic link* (3.2.5) and the *characterizing generic concept* (3.1.12) it links to, possibly by enumeration of the concepts in the characterizing generic concept.

Note 2 to entry: Different levels of sanctioning are possible (e.g. conceivable, sensible, normal, usually In The Context Of, necessary).

[SOURCE: ISO 17115:2020, 3.1.5]

3.1.15
subject field
domain
field of special knowledge

Note 1 to entry: The borderlines of a subject field are defined from a purpose-related point of view.

[SOURCE: ISO 17115:2020, 3.1.7]

3.2 Relations between concepts

3.2.1
generic relation
generalization-specialization relation
genus-species relation
relation between two *concepts* (3.1.1) where the intension of one of the concepts includes that of the other concept and at least one additional delimiting *characteristic* (3.1.3)

Note 1 to entry: A generic relation exists between the concepts ‘word’ and ‘pronoun’, ‘vehicle’ and ‘car’, ‘person’ and ‘child’.

Note 2 to entry: This relation is equivalent to ‘parent-child’ or ‘is-a’ relation. The child concept has the same intension as the parent concept and at least one additional delimiting characteristic. Also the same in X ‘is-a’ Y.

Note 3 to entry: The use of the term ‘relation’ from an English perspective means that this is describing the concept (i.e. this concept is a generic relation of another concept) not the relationship. However, the ‘generic relation’ here means the relationship itself, not a concept.

[SOURCE: ISO 17115:2020, 3.2.1]

3.2.2
partitive relation
whole-part relation
relation between two *concepts* (3.1.1) where one of the concepts constitutes the whole and the other concept a part of that whole

Note 1 to entry: A partitive relation exists between the concepts ‘week’ and ‘day’, ‘molecule’ and ‘atom’.

Note 2 to entry: This relation is different from *generic relation* (3.2.1). For example, a day is part of a week, but is not a specialization of a week, i.e. it is not a type of week.

[SOURCE: ISO 17115:2020, 3.2.2]

3.2.3
hierarchical relation
relation between two *concepts* (3.1.1) which may be either a *generic relation* (3.2.1) or a *partitive relation* (3.2.2)

EXAMPLE Relations such as ‘stomach’ ↔ ‘organ’ and ‘stomach’ ↔ ‘body’ are hierarchical relations, but the former is a generic relation and the latter is a partitive relation.

Note 1 to entry: Partitive relations have several sub-types, such as ‘constitutional part of’, ‘regional part of’, ‘grouped by’, and so on, which are also hierarchical types of relations.

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Note 2 to entry: A *characteristic* (3.1.3) which is explicitly identified in the *terminological resource* (3.4.1) may be used to determine a hierarchy, and it includes declaration of directionality. There may be more than one such characteristic in which case the terminological resource can be said to have 'poly-hierarchy'. The characteristic shall have a value in the two concepts at either end of the relation which can be compared to determine the hierarchy.

[SOURCE: ISO 1087:2019, 3.2.12, modified — The admitted term "hierarchical concept relation" was removed; "relation between two concepts which may be either" was added in the definition; the example and the notes were added.]

3.2.4
associative relation
associative concept relation
pragmatic relation
non-hierarchical *concept* (3.1.1) relation

EXAMPLE The relation between disease 'X' and virus 'Y' is not a *hierarchical relation* (3.2.3) but an associative one. The relation exists under a certain theme of interest (e.g. 'a disease and its causative agent") and explicitly recognized by virtue of experience.

[SOURCE: ISO 1087:2019, 3.2.23, modified— The example was changed.]

3.2.5
semantic link
formal representation of a directed *associative relation* (3.2.4) or *partitive relation* (3.2.2) between two *concepts* (3.1.1)

EXAMPLE hasCause (with inverse isCauseOf).

Note 1 to entry: This includes all relations except the *generic relation* (3.2.1)

Note 2 to entry: A semantic link always has an inverse, i.e. another semantic link with the opposite direction.

[SOURCE: ISO 17115:2020, 3.2.5]

3.3 Formal concept representation

3.3.1
axiomatic concept representation
atomic concept representation
concept representation that is not composed of other simpler concept representations within a *formal (concept representation) system* (3.4.7)

EXAMPLE 'Liver', 'Incision act', 'Pain'.

Note 1 to entry: In many cases, axiomatic concept representations will correspond to what philosophers call 'natural kinds'. Such an entity cannot be meaningfully decomposed. These should form the basis of all concept representations.

Note 2 to entry: A *coding system* (3.4.4) may include axiomatic concept representations as well as *compositional concept representations* (3.3.2) required for different use cases.

3.3.2
compositional concept representation
composite concept representation
intensional definition of a *concept* (3.1.1) using as delimiting characteristics one or more *composite characteristics* (3.1.10)

Note 1 to entry: Each *characterizing concept* (3.1.11) in a composite characteristic may be *axiomatic concept representation* (3.3.1) or another compositional concept representation.

Note 2 to entry: This allows inference of subsumption within a *formal (concept representation) system* (3.4.7). It is often expressed in a formalism, such as description logic.

Note 3 to entry: Compositional concept representation can be further divided into *pre-coordinated concept representation* (3.3.3) and *post-coordinated concept representation* (3.3.4).

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3.3.3
pre-coordinated concept representation
compositional concept representation (3.3.2) predefined within a *formal (concept representation) system* (3.4.7), with an equivalent single unique *concept identifier* (3.1.5)

EXAMPLE In SNOMED CT, 'cancer of colon' is predefined and has a single unique identifier, which means to the SNOMED CT that it represents a 'single' concept. However, 'colon' is a synonym for 'colon structure' and 'cancer' is a synonym for 'malignant neoplastic disease in SNOMED CT. Therefore, 'colon cancer' is non-atomic as it can be broken down into compositional concept representation (e.g. 'cancer of colon' = 'malignant neoplastic disease' < Finding_Site: 'colon structure'>.).

3.3.4
post-coordinated concept representation
compositional concept representation (3.3.2), which is not pre-coordinated and therefore is represented using more than one *concept* (3.1.1) from one or many *compositional systems* (3.4.5), combined using mechanisms within or outside the compositional systems

EXAMPLE 1 Problem.Main = Fracture, Problem.Location = Femur (within a template for a problem description).

EXAMPLE 2 Some common *terminological resources* (3.4.1), such as IETF BCP-47 for language tags, explicitly construct post-coordinated concept representations from disparate coding systems for language, script, region, and so on.

Note 1 to entry: Combining concepts from disparate terminologies can cause problems with either overlapping or conflicting concepts, or both, because there might be various ways to form compositional concept representations for the same concept. Typically, the mechanisms for making compositional concept representations across disparate terminological resources are specified in an information model (e.g. as templates for a certain type of concept).

3.3.5
canonical expression
concept name
term (3.1.2) which uniquely designates a *concept* (3.1.1) within a *terminology* (3.4.2)

EXAMPLE 1 Machine readable: <Inflammation that <hasCause Bacteria hasLocation Lung>>(with compositional characteristics sorted alphabetically after semantic link) instead of <pulmonaryInfection that hasCause Bacteria>

EXAMPLE 2 General language: Inflammation that has cause bacteria and has location lung (with compositional characteristics sorted alphabetically after semantic link) instead of pulmonary infection that has cause bacteria.

Note 1 to entry: It is unique within the system and unambiguous.

3.3.6
categorial structure
minimal set of *domain constraints* (3.1.14) for representing *terminologies* (3.4.2) in a *subject field* (3.1.15)

[SOURCE: ISO 17115:2020, 3.1.1, modified — The admitted term "reference terminology model" was removed.]

3.4 Terminological resources (in health domain)

3.4.1
terminological resource
controlled set of *terms* (3.1.2) in health domain

Note 1 to entry: Usually designed and controlled for use with computers for a specific purpose in the health domain, such as data entry, aggregation, retrieval and analysis. Accordingly, it has usability characteristics in health domain as described in Clause 4.

Note 2 to entry: It has a *generic relation* (3.2.1) to the terms *terminology* (3.4.2), *classification system* (3.4.3), *coding system* (3.4.4), *compositional system* (3.4.5), *nomenclature* (3.4.6), and *formal (concept representation) system* (3.4.7).

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3.4.2
terminology
terminological system
concept representation system
<health> structured human and machine-readable representation of health *concepts* (3.1.1) and relationships

Note 1 to entry: Every terminology shall be organized by either *hierarchical relations* (3.2.3) or *associative relations* (3.2.4), or both. Typically, most terminologies are organized by hierarchical relations.

Note 2 to entry: Every terminology shall have term representations of health concepts for human-readability.

Note 3 to entry: It is used directly or indirectly to describe health concepts such as health conditions and healthcare activities, and to allow their subsequent retrieval for analysis.

3.4.3
classification system
statistical classification
classification
terminological resource (3.4.1) which has characteristics of mutual exclusiveness and exhaustiveness to aggregate data to a pre-prescribed level of specialization for a specific purpose

Note 1 to entry: Both of the two additional characteristics are indispensable to provide data aggregation function (5.4), and accordingly, to suit the use case for statistical analysis and data aggregation.

Note 2 to entry: It usually includes *hierarchical relations* (3.2.3) as well as definitions and rules for use but might not, as in the example simple classification system of gender: Male, Female.

Note 3 to entry: It does not necessarily consist of a pre-defined exhaustive set of mutually exclusive categories. Classification systems are allowed to have explicit rules to enable mutual exclusiveness.

Note 4 to entry: All classification systems are suited to statistical use as they group things and have concepts to represent other and unspecified.

3.4.4
coding system
combination of a set of *concepts (coded concepts)* (3.1.1), a set of *code* (3.1.6) values, and at least one *coding scheme* (3.1.9) mapping code values to coded concepts

Note 1 to entry: Coded concepts are typically represented by *terms* (3.1.2) but can have other representation. Code values are typically numeric or alphanumeric.

Note 2 to entry: Coding systems without organization by either *hierarchical relations* (3.2.3) or *associative relations* (3.2.4), or both, such as ISO 3166-1 for country codes, are not terminologies.

3.4.5
compositional system
compositional terminology
terminology (3.4.2) that supports the creation of *compositional concept representation* (3.3.2)

Note 1 to entry: The definition implies that compositional system has an ability of post-coordination. It does not preclude that a compositional system also includes axiomatic and pre-coordinated concepts.

Note 2 to entry: Pre-defined concepts in a compositional system are not necessarily represented as compositional concept representation, since pre-defined concepts may include *axiomatic concept representations* (3.3.1).

3.4.6
nomenclature
compositional system (3.4.5) structured systematically according to pre-established naming rules

EXAMPLE 1 Naming rules for a family of molecules such as '—ane', '—ene', 'halo—', 'hydroxyl—' in prefix or suffix is an example of pre-established naming rules in the compositional concept creation.

EXAMPLE 2 In SNOMED CT, 'fully specified names' use a number of pre-established naming rules.

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3.4.7
formal (concept representation) system
compositional system (3.4.5) with a set of machine-processable definitions in a *subject field* (3.1.15)

Note 1 to entry: Each definition of a pre-defined concept shall be an *axiomatic concept representation* (3.3.1), a *compositional concept representation* (3.3.2) or a formal extensional definition.

4 Characteristics of terminological resources in the health domain

4.1 Basics

Clause 4 and Clause 5 support categorization according to the characteristics and functions of the terminological resources rather than the name.

The characteristics of a terminological resource influence its utility and appropriateness in clinical applications. Terminological resources 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 document explicitly refers only to terminological resources that are primarily designed to be used for health concept representation or to the aspect of a terminological resource designed to be used for health concept representation. This document will also provide terminological resource developers and users with the basic characteristics and functions invoked by those characteristics that should be taken into account on the occasion of assessing whether a terminological resource meets their requirements. These tenets do not attempt to specify all the richness that can be incorporated into a healthcare terminological resource. However, this document does specify the minimal characteristics, which will ensure that the terminological resource can provide the functions that are indispensable for the requirements on it. Figure 1 illustrates how those components are related to each other.

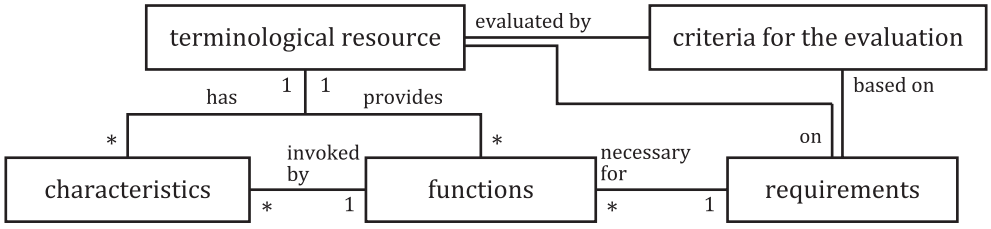


Figure 1 — Relations among the components in ISO 17117

This document will also provide terminology developers with a sturdy starting point for the development of healthcare terminological resources. This foundation serves as the basis from which terminology developers will build robust, large-scale, reliable and maintainable terminological resources.

Some classes of terminological resources defined in Clause 3 may have overlaps. For example, classification systems are typically coding systems. Clause 4 defines characteristics for the terminological resources defined in Clause 3. To assess whether a terminological resource meets the applicable requirements, users and developers shall pay considerable attention to the characteristics the terminological resource has and the functions invoked by those characteristics, rather than its class name.

Annex A gives relations between characteristics, functions, requirements and evaluation criteria of terminological resources. Relations among terminological resources are given in Annex B.

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