# INTERNATIONAL STANDARD



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# Health informatics — Terminological resources —

Part 1: Characteristics

Informatique de santé — Ressources terminologiques —

iTeh STPartie DearacteristiquesEVIEW (standards.iteh.ai)

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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 <a href="https://www.iso.org/directives">www.iso.org/directives</a>).

Attention is drawn to the possibility that some of the elements of this document may be the subject of patent rights. ISO shall not be held responsible for identifying any or all such patent rights. Details of any patent rights identified during the development of the document will be in the Introduction and/or on the ISO list of patent declarations received (see <a href="https://www.iso.org/patents">www.iso.org/patents</a>).

Any trade name used in this document is information given for the convenience of users and does not constitute an endorsement.

For an explanation on the voluntary nature of standards, the meaning of ISO specific terms and expressions related to conformity assessment, as well as information about ISO's adherence to the World Trade Organization (WTO) principles in the Technical Barriers to Trade (TBT) see the following URL: <a href="https://www.iso.org/iso/foreword.html">www.iso.org/iso/foreword.html</a>. (standards.iteh.ai)

This document was prepared by ISO/TC 215, Health informatics.

This first edition of ISO 17117-1/cancels and replaces ISO/TS+17011702002, which has been technically revised.

A list of all the parts of ISO 17117 is available on the ISO website.

### 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 like conditions of patients and populations, actions in healthcare and related concepts, such as medicines, biomedical molecules, genes, organisms, technical methods and social concepts<sup>[3]</sup>. Many formal and less formal terminological resources exist to represent this complexity. These may 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[6][7][8]. Such resources are required for precise data collection, accurate interpretation of data and interoperability among information systems that exchange such data<sup>[7]</sup>. 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 by ISO in 2002 (ISO/TS 17117). 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<sup>[9][10]</sup>. This revision updates the original Technical Specification with a revised scope and purpose commensurate with present and future healthcare and technology contexts, incorporating new definitional standards where relevant.

As the first part of the entire revision work, this document (ISO 17117-1) 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, which will be addressed in the future parts of ISO 17117, are tightly related to the characteristics of terminological resources and functions that they can provide.

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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 refers only to 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. The focus of this document is to define characteristics and functions of terminological resources in healthcare that can be used to identify different types of them for categorization purposes. <u>Clauses 4</u> and <u>5</u> support categorization according to the characteristics and functions of the terminological resources rather than the name.

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

The target groups for this document are dards.iteh.ai)

- a) organizations wishing to select terminological systems for use in healthcare information systems; <u>ISO 17117-1:2018</u>
- b) developers of terminological systems: log/standards/sist/30d2d66d-9612-4b38-
- c) developers of terminology standards;
- d) those undertaking independent evaluations/academic reviews of terminological resources;
- e) terminology Registration Authorities.

This document contains general characteristics and criteria with which systems can be evaluated.

The following considerations are outside the scope of 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. 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 statistical classifications 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. 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. Progress in medical science and in terminology science will necessitate updating of this document in due course.

#### 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.

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

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

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

NOTE Selected terms from ISO 1087-1 are given in <u>Annex A</u> as background to the terms and definitions in Clause 3.

#### 3.1 General terms

#### 3.1.1

#### concept

unit of knowledge created by a unique combination of characteristics

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 individual and social conceptualizations of the world a round them. Concept representations are artefacts constructed of symbols.

#### (standards.iteh.ai)

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 1087-1:2000, 3:2:1://modified]h.ai/catalog/standards/sist/30d2d66d-9612-4b38-

b6bd-d85f055b601c/iso-17117-1-2018

#### 3.1.2 term

linguistic representation of a concept in a specific subject field

[SOURCE: ISO 1087-1:2000, 3.4.3, modified]

#### 3.1.3

#### characteristic

abstraction of a property of an object or of a set of objects

[SOURCE: ISO 1087-1:2000, 3.2.4]

#### 3.1.4

#### term identifier

sequence of letters, numbers or symbols, capable of uniquely identifying a term within the terminological resource

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 within the terminological resource

Note 1 to entry: Concept identifier shall be unique within the terminological resource, so terms shall not be used for the purpose here 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* (<u>3.4.1</u>) 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.

#### 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 maps 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.

[SOURCE: ISO 17115:2007, 2.7.2, modified]

### (standards.iteh.ai)

#### 3.1.10

#### composite characteristic

representation of a *characteristic* (3.1.3) ISO 17117-1:2018 https://standards.tich.av/catalog/standards/sist/30d2d66d-9612-4b38-

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).

[SOURCE: ISO 17115:2007, 2.2.1]

#### 3.1.11

#### characterizing concept

concept 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".

[SOURCE: ISO 17115:2007, 2.2.2]

#### 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.

[SOURCE: ISO 17115:2007, 2.3.3]

#### 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).

[SOURCE: ISO 17115:2007, 2.3.1]

#### 3.1.14

#### domain constraint

sanction rule prescribing the set of *sanctioned characteristics* (3.1.13) that are valid to specialize a concept in a certain subject field

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.

Note 1 to entry: The rule describes the set of potential characteristics 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). (standards.iteh.ai)

[SOURCE: ISO 17115:2007, 2.3.2]

ISO 17117-1:2018

**3.2 Relations between concepts** ndards.iteh.ai/catalog/standards/sist/30d2d66d-9612-4b38b6bd-d85f055b601c/iso-17117-1-2018

#### 3.2.1

#### generic relation

#### generalization-specialization relation

relation between two concepts where the intension of one of the concepts includes that of the other concept and at least one additional delimiting characteristic

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 1087-1:2000, 3.2.21, modified]

#### 3.2.2

#### partitive relation

#### whole-part relation

relation between two concepts 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 1087-1:2000, 3.2.22, modified]

### 3.2.3

#### hierarchical relation

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

EXAMPLE Relations such as 'stomach'  $\leftrightarrow$  'organ' and 'stomach'  $\leftrightarrow$  'body' are hierarchical relations, but the former is a generic relation and the latter is a partitive relation.

Note 1 to entry: *Partitive relations* (3.2.2) have several sub-types, such as "constitutional part of", "regional part of", "grouped by", and so on, which are also hierarchical types of relations.

Note 2 to entry: A characteristic which is explicitly identified in the terminological resource may be used to determine a hierarchy, and 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 must have a value in the two concepts at either end of the relation which can be compared to determine the hierarchy.

#### [SOURCE: ISO 1087-1:2000, 3.2.20, modified]

#### 3.2.4 associative relation pragmatic relation

relation between two concepts having a non-hierarchical thematic connection by virtue of experience

EXAMPLE The relation between disease "X" and virus "Y" is not a hierarchical but an associative relation. 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 1087-1:2000, 3,2.23, modified] ARD PREVIEW

#### 3.2.5 semantic link

### (standards.iteh.ai)

formal representation of a directed *associative relation* (3.2.4) or *partitive relation* (3.2.2) between two concepts ISO 17117-1:2018

EXAMPLE https://standards.iteh.ai/catalog/standards/sist/30d2d66d-9612-4b38hasCause (with inverse isCauseOf)01c/iso-17117-1-2018

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 17115:2007, 2.2.3]

#### 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 may include axiomatic concept representations as well as compositional concept representations required for different use cases.

[SOURCE: ISO 17115:2007, 2.4.2, modified]

#### 3.3.2

#### compositional concept representation composite concept representation

intensional definition of a concept 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* (3.1.10) 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).

[SOURCE: ISO 17115:2007, 2.4.1, modified]

#### 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

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\_Sife: "colon structure" > .) A RD PREVIEW

# [SOURCE: ISO 17115:2007, 2.4.6, modified]tandards.iteh.ai)

#### 3.3.4

#### post-coordinated concept representation ISO 17117-1:2018

compositional concept representation (3.3.2)/which is anot pre-coordinated and therefore shall be represented using more than one concept from one dr/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, 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 overlapping and/ or conflicting concepts, 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).

[SOURCE: ISO 17115:2007, 2.4.7, modified]

#### **3.3.5 canonical expression concept name** *term* (3.1.2) which uniquely designates a concept within a *terminological system* (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.