
Health informatics — Vocabulary for terminological systems

*Informatique de santé — Vocabulaire pour les systèmes
terminologiques*



Reference number
ISO 17115:2007(E)

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Published in Switzerland

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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.

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ISO 17115 was prepared by Technical Committee ISO/TC 215, *Health informatics*.

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Introduction

Health terminology is complex and multifaceted, more so than most language domains. It has been estimated that between 500 000 and 45 million different concepts are needed to adequately describe concepts (e.g. conditions of patients and populations), actions in healthcare and related concepts (e.g. biomedical molecules, genes, organisms, technical methods and social concepts).

It is obvious that to adequately represent and especially to process this complexity, simple coding schemes are inadequate and formal multidimensional concept representation systems are required. Several such formal systems exist but systems and their underlying philosophy are described in different ways. The system itself can, for example, be called an ontology, medical entity dictionary, coding and reference model or reference terminology. The differences in terminology are understandable; this kind of work is highly interdisciplinary and integrates knowledge from linguistics, philosophy, informatics and health sciences, and there is room for misunderstanding between disciplines.

This International Standard is based on other standards, with clarifications and examples appropriate to health care in order to inform those working with terminology in health care, and aims to establish a set of basic concepts required to describe formal concept representation systems, especially for health sciences, and to describe representation of concepts and characteristics, for use especially in formal computer-based concept representation systems. These issues have previously been addressed by EN 12264, which is partly replaced by this International Standard. This document is not intended to be exhaustive, but to serve as a basis for related International Standards by reference and associated implementation guides.

Informally, the term "concepts" is often used when what is meant is "concept representations". However, this leads to confusion when precise meanings are required. Concepts arise out of human individual and social conceptualization of the world around them. Concept representations are artefacts constructed of symbols and are often manifest in computer programs. Because they are artefacts, it is possible to be precise about the functioning and capabilities of concept representations. It is more difficult to be clear about the yet poorly understood function of human conceptualization.

Health informatics — Vocabulary for terminological systems

1 Scope

1.1 Main purposes

This International Standard defines a set of basic concepts required to describe formal concept representation systems, especially for health sciences, and describes representation of concepts and characteristics, for use especially in formal computer-based concept representation systems. A main motivation is to make it possible to precisely describe content models described in other International Standards.

The principles established by ISO/TC 37 are extended here into a formal system suited to health informatics.

Potential uses for this International Standard are

- to describe formal definitions, parts of definitions and how they are related, and
- to describe patterns for concept representation in a particular domain.

1.2 Target groups

The target groups for this International Standard are

- developers of concept representation systems for different health care domains,
- developers of standards for concept representation, especially those describing domain concept models,
- information modellers, knowledge engineers, and standards developers building information models for health information systems, such as electronic health records and decision support systems,
- developers of information systems that require an explicit system of concepts for internal organization, data warehouse management and middleware services.

1.3 Topics not considered

This International Standard does not include enumeration of axiomatic concepts and semantic links, or detailed content of health terminology systems (classifications, nomenclatures or reference terminology of health concepts).

2 Terms and definitions

For the purposes of this document, the following terms and definitions apply. Selected background terms and definitions from ISO 1087-1 are provided in Annex A.

NOTE The use of angular brackets <> refers to a category which can be specialized to various concepts as required.

2.1 Specialization

2.1.1

specialize

form a more **specific concept** (A.3.2.16) [by constraining the **extension** (A.3.2.8) of a more **generic concept** (A.3.2.15)]

EXAMPLE 1 Infection that hasCause Bacteria can be specialized to Infection that hasCause.

EXAMPLE 2 Pneumococcus Hepatitis can be specialized to NonA-NonB-hepatitis.

NOTE 1 To specialize is to increase the intension and decrease the extension of a concept. The more **specific concept** (A.3.2.16) has a larger **intension** (A.3.2.9), but a smaller extension than the **general concept** (A.3.2.3). Specialization and generalization can be achieved in many ways, including replacing a semantic link with a more specific semantic link (and vice versa for generalization).

NOTE 2 The more specific concept has a broader **intension**, but a narrower **extension** than the **generic concept**.

NOTE 3 Ways to specialize concepts include

- adding one or more **composite characteristics** (2.2.1),
- replacing the **characterizing concept** (2.2.2) in one or more **characteristics** (A.3.2.4) with a more **specific concept** (A.3.2.16), and
- forming an intersection of two concepts (where the intersection is a specialization of both the "parents").

NOTE 4 The opposite is **generalize** (2.1.2).

2.1.2

generalize

form a more **generic concept** (A.3.2.15) [that represents a superset of the **extension(s)** (A.3.2.8) of one or more **specific concepts** (A.3.2.16)]

EXAMPLE Infection that hasCause Pneumococcus can be generalized to Infection that hasCause Bacterium.

NOTE 1 To generalize is to decrease the **intension** (A.3.2.9) and increase the extension of a concept. Specialization and generalization can be achieved in many ways, including replacing a semantic link with a less specific semantic link (and vice versa for specialization).

NOTE 2 This can be done by removing one or more **characteristics** (A.3.2.4) or by replacing the **characterizing concept** (2.2.2) in one or more characteristics with a more generic concept.

NOTE 3 The opposite is **specialize** (2.1.1).

2.1.3

level of specialization

property of a **concept** (A.3.2.1) reflecting the number of and detail of **characteristics** (A.3.2.4) in its **intension** (A.3.2.9)

NOTE A **specific concept** (A.3.2.16) has a high level of specialization and a fine granularity; a **generic concept** (A.3.2.15) has low level of specialization and coarse granularity.

2.1.4

generic concept

category

concept (A.3.2.1) in a **generic relation** (A.3.2.21) having the narrower **intension** (A.3.2.9) [and the wider **extension** (A.3.2.8)]

[ISO 1087-1:2000, A.3.2.15]

2.2 Formal representation of characteristics

2.2.1

composite characteristic

qualifier

representation of a **characteristic** (A.3.2.4)

EXAMPLE hasCause Bacteria; Location = LeftUpperLobeOfLung.

NOTE 1 Typically expressed by a **semantic link** (2.2.3) and a **characterizing concept** (2.2.2).

NOTE 2 Can be compared to an attribute-value pair in a **compositional system** (2.5.2).

NOTE 3 A qualifier often denotes **characteristics** with a small simple **characterizing generic concept** (2.3.3), such as laterality (left or right), or severity (low, moderate, high).

2.2.2

characterizing concept

concept (A.3.2.1) that is referenced by a **semantic link** (2.2.3) in a **composite characteristic** (2.2.1)

EXAMPLES "Bacterium" in the construct "Disease that hasCause Bacterium"; "Yellow" in the construct "SkinLesion that hasColour Yellow".

2.2.3

semantic link

formal representation of a directed **associative relation** (A.3.2.23) or **partitive relation** (A.3.2.22) between two **concepts** (A.3.2.1)

EXAMPLES hasLocation (with inverse isLocationOf); isCauseOf (with inverse hasCause).

NOTE 1 This includes all relations except the **generic relation** (A.3.2.21).

NOTE 2 A semantic link always has an inverse, i.e. another semantic link with the opposite direction.

NOTE 3 A semantic link can be part of a **composite characteristic** (2.2.1) where it describes the role of the **characterizing concept** (2.2.2). Similarly, it describes the role of a **characterizing generic concept** (2.3.3) in a **sanctioned characteristic** (A.3.1).

2.3 Sanctioned specialization

2.3.1

sanctioned characteristic

formal representation of a **type of characteristics** (A.3.2.5)

EXAMPLE 1 performedUsing <INSTRUMENT>; hasLocation <BodyPartOrImplantedDevice>.

EXAMPLE 2 "CauseOfInflammation canBe set{ bacteria, virus, parasite, autoimmune, chemical, physical }", where "canBe" is the **semantic link** (2.2.3), and "set{ bacteria, virus, parasite, autoimmune, chemical, physical }" is the **characterizing generic concept** (2.3.3).

NOTE A sanctioned characteristic is typically made up of a combination of a semantic link and a characterizing generic concept, and can be used in **domain constraints** (2.3.2).

2.3.2

domain constraint

sanction rule prescribing the set of **sanctioned characteristics** (2.3.1) that are valid to **specialize** (2.1.1) a **concept** (A.3.2.1) in a certain **subject field** (A.3.1.2)

EXAMPLE "Infection possibly hasLocation SkeletalStructure" describes that an infection in a certain context can be located in a structure that is a kind of skeletal structure.

NOTE 1 The rule describes the set of potential **characteristics** (A.3.2.4) by combining the **semantic link** (2.2.3) and the **characterizing generic concept** (2.3.3) it links to, possibly by enumeration of the concepts in the characterizing generic concept.

NOTE 2 Different levels of sanctioning are possible (e.g. conceivable, sensible, normal, usuallyInTheContextOf, necessary).

2.3.3

characterizing generic concept

characterizing category

value domain

formal category (2.5.3) whose specialization by a **domain constraint** (2.3.2) is allowed to be used as **characterizing concept** (2.2.2) in a particular context

EXAMPLE <INFECTIOUS_ORGANISM> = {bacterium, virus, parasite}, in the context of "Infection that hasCause INFECTIOUS_ORGANISM".

NOTE The context includes a **superordinate concept** (A.3.2.13) and a **semantic link** (2.2.3).

2.4 Formal concept representation

2.4.1

compositional concept representation

intensional definition (A.3.3.2) of a **concept** (A.3.2.1) using as **delimiting characteristics** (A.3.2.7) one or more **composite characteristics** (2.2.1)

NOTE This allows inference and subsumption within a **compositional system** (2.5.2). It is usually expressed in a formalism, such as description logic.

2.4.2

axiomatic concept representation

axiom concept representation present in a **formal system** (2.5.1) without a **formal definition** (2.4.3)

EXAMPLES Liver; Incision act; Pain.

NOTE This often represents a "natural kind" from the perspective of a particular terminology system; i.e. something that "just exists". It may have a definition or description outside the system but, by choice, this is not represented in the system.

2.4.3

formal definition

definition within a **formal system** (2.5.1)

NOTE 1 This can be done by a **compositional concept representation** (2.4.1) or a formal **extensional definition** (A.3.3.3).

NOTE 2 It is usually automatically processable and governed by explicit rules.

2.4.4

concept name

canonical expression

term (A.3.4.3) which uniquely designates a **concept** (A.3.2.1) within a **concept system** (A.3.2.11)

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 It is the preferred expression to represent a **concept** (A.3.2.1) in a given terminology system.

NOTE 2 It is unique within the system and unambiguous.

2.4.5

categorial structure

minimal set of **domain constraints** (2.3.2) for representing **concept systems** (A.3.2.11) in a **subject field** (A.3.1.2)

2.4.6

precoordinated concept representation

compositional concept representation (2.4.1) within a **formal system** (2.5.1), with an equivalent single unique identifier

EXAMPLE Problem = Fracture that hasLocation Femur. This is an example of how a precoordinated concept is represented.

NOTE The identifier (code, term, etc.) may be within or outside the terminology system in question.

2.4.7**post-coordinated concept representation**

compositional concept representation (2.4.1) using more than one **concept** (A.3.2.1) from one or many **formal systems** (2.5.1), combined using mechanisms within or outside the formal systems

EXAMPLE Problem.Main = "Fracture", Problem.Location = Femur within a template for a problem description.

NOTE Combining concepts from disparate terminologies can cause problems with overlapping and/or conflicting concepts. Typically, the mechanisms for making **compositional concept representations** (2.4.1) are specified in an information model (e.g. as templates for a certain type of concept).

2.5 Terminology and information models, concept systems**2.5.1****formal [concept representation] system**

set of machine processable definitions in a **subject field** (A.3.1.2)

2.5.2**compositional system**

system that supports the creation of **compositional concept representations** (2.4.1)

2.5.3**formal category**

generic concept (2.1.4) represented by a **formal definition** (2.4.3)

NOTE This implies that the generic concept's **extension** (A.3.2.8) can be determined algorithmically and includes extensionally defined **concepts** (A.3.2.1) and formal **intensional definitions** (A.3.3.2).

2.6 Specified concepts**2.6.1****mapping**

assigning an element in one set to an element in another set through **semantic correspondence** (2.6.2)

NOTE It is the relation with the best semantic correspondence between an element in one set and an element in another set.

2.6.2**semantic correspondence**

measure of similarity between two concepts

NOTE The opposite is semantic distance.

2.6.3**instance of a concept**

member of the **extension** (A.3.2.8) of a **concept** (A.3.2.1)

2.6.4**focus concept representation**

specified representation of the **concept** (A.3.2.1) of interest within a **formal system** (2.5.1)

EXAMPLE "Moderately severe inflammation caused by pneumococci located in the upper lobe of the left lung, ascertained by plain film pulmonary X-ray and sputum culture" in the context of a diagnosis with confirmatory evidence.

NOTE It includes context information, enabling independent use.

2.6.5

generic relation

subtype relation

relation between two **concepts** (A.3.2.1) where the **intension** (A.3.2.9) of one of the concepts includes that of the other concept and at least one additional **delimiting characteristic** (A.3.2.7)

[ISO 1087-1:2000, A.3.2.21]

NOTE All individuals in the **extension** (A.3.2.8) of the second are included in the extension of the first.

EXAMPLE A generic relation exists between the concepts 'internal organ' and 'heart', 'surgical deed' and 'appendectomy', 'inflammatory disease' and 'pericarditis'.

2.7 Terminological systems

2.7.1

classification

exhaustive set of mutually exclusive **categories** (2.1.4) to aggregate data at a pre-prescribed **level of specialization** (2.1.3) for a specific purpose

EXAMPLE ICD 10.

2.7.2

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"

[ISO 2382-4]

NOTE The two sets are not part of the coding scheme.

2.7.3

coding system

combination of a set of **concepts** (A.3.2.1) [coded concepts], a set of code values, and at least one **coding scheme** (2.7.2) mapping code values to coded concepts

NOTE Coded concepts are typically represented by **terms** (A.3.4.3) but can have other representation. Code values are typically numeric or alphanumeric.

2.7.4

reference terminology

set of atomic level designations structured to support representations of both simple and compositional concepts independent of human language (within machine)

NOTE 1 Reference terminology is designed to uniquely represent **concepts** (A.2.3.1).

NOTE 2 The terminology lists the concepts and specifies their structure, relationships and, if present, their systematic and **formal definitions** (2.4.3).

2.7.5

clinical terminology

terminology required directly or indirectly to describe health conditions and healthcare activities

NOTE 1 Health conditions include symptoms, complaints, illness, diseases, disorders, etc.

NOTE 2 It is used in, for example, medical records, clinical communication, and medical science.

Annex A (normative)

Selected definitions from ISO 1087-1:2000

The following terms and definitions are selected from ISO 1087-1:2000. They are included here as background to the key terms and definitions in this International Standard. For consistency, the numbering in this annex reflects the numbering in ISO 1087-1:2000, with the addition of "A."

A.3.1 Language and reality

A.3.1.1

object

anything perceivable or conceivable

NOTE Objects may be material (e.g. an engine, a sheet of paper, a diamond), immaterial (e.g. conversion ratio, a project plan) or imagined (e.g. a unicorn).

A.3.1.2

subject field

domain

field of special knowledge

NOTE The borderlines of a subject field are defined from a purpose-related point of view.

A.3.2 Concepts

A.3.2.1

concept

unit of knowledge created by a unique combination of **characteristics** (A.3.2.4)

NOTE Concepts are not necessarily bound to particular languages. They are, however, influenced by the social or cultural background often leading to different categorizations.

A.3.2.2

individual concept

concept (A.3.2.1) which corresponds to only one **object** (A.3.1.1)

EXAMPLES Individual concepts are: 'Saturn', 'the Eiffel Tower'.

NOTE Examples of individual concepts are usually represented by **appellations** (A.3.4.2).

A.3.2.3

general concept

concept (A.3.2.1) which corresponds to two or more **objects** (A.3.1.1) which form a group by reason of common properties

EXAMPLES General concepts are 'planet', 'tower'.

A.3.2.4

characteristic

abstraction of a property of an **object** (A.3.1.1) or of a set of **objects** (A.3.1.1)

NOTE Characteristics are used for describing **concepts** (A.3.2.1).

A.3.2.5

type of characteristics

category of **characteristics** (A.3.2.4) which serves as the criterion of subdivision when establishing **concept systems** (A.3.2.11)

NOTE The type of characteristics "colour" embraces characteristics "being red, blue, green", etc. The type of characteristics "material" embraces characteristics "made of wood, metal", etc.

A.3.2.6

essential characteristic

characteristic (A.3.2.4) which is indispensable to understanding a **concept** (A.3.2.1)

A.3.2.7

delimiting characteristic

essential characteristic (A.3.2.6) used for distinguishing a **concept** (A.3.2.1) from related **concepts** (A.3.2.1)

NOTE The delimiting characteristic "support for the back" may be used for distinguishing the concepts 'stool' and 'chair'.

A.3.2.8

extension

totality of **objects** (A.3.1.1) to which a **concept** (A.3.2.1) corresponds

A.3.2.9

intension

set of **characteristics** (A.3.2.4) which makes up the **concept** (A.3.2.1)

A.3.2.10

concept field

unstructured set of thematically related **concepts** (A.3.2.1)

NOTE Concept fields may be used as a starting point for establishing **concept systems** (A.3.2.11).

A.3.2.11

concept system

system of concepts

set of **concepts** (A.3.2.1) structured according to the relations among them

A.3.2.12

concept diagram

graphic representation of a **concept system** (A.3.2.11)

A.3.2.13

superordinate concept

broader concept

concept (A.3.2.1) which is either a **generic concept** (A.3.2.15) or a **comprehensive concept** (A.3.2.17)

A.3.2.14

subordinate concept

narrower concept

concept (A.3.2.1) which is either a **specific concept** (A.3.2.16) or a **partitive concept** (A.3.2.18)

A.3.2.15

generic concept

concept (A.3.2.1) in a **generic relation** (A.3.2.21) having the narrower **intension** (A.3.2.9)

A.3.2.16

specific concept

concept (A.3.2.1) in a **generic relation** (A.3.2.21) having the broader **intension** (A.3.2.9)

A.3.2.17

comprehensive concept

concept (A.3.2.1) in a **partitive relation** (A.3.2.22) viewed as the whole

A.3.2.18

partitive concept

concept (A.3.2.1) in a **partitive relation** (A.3.2.22) viewed as one of the parts making up the whole

A.3.2.19**coordinate concept**

subordinate concept (A.3.2.14) having the same nearest **superordinate concept** (A.3.2.13) and same criterion of subdivision as some other **concept** (A.3.2.1) in a given **concept system** (A.3.2.11)

A.3.2.20**hierarchical relation**

relation between two **concepts** (A.3.2.1) which may be either a **generic** (A.3.2.21) or a **partitive relation** (A.3.2.22)

A.3.2.21**generic relation**

genus/species relation

relation between two **concepts** (A.3.2.1) where the **intension** (A.3.2.9) of one of the concepts includes that of the other concept and at least one additional **delimiting characteristic** (A.3.2.7)

NOTE A generic relation exists between the concepts 'word' and 'pronoun', 'vehicle' and 'car', 'person' and 'child'.

A.3.2.22**partitive relation**

part-whole relation

relation between two **concepts** (A.3.2.1) where one of the concepts constitutes the whole and the other concept a part of that whole

NOTE A partitive relation exists between the concepts 'week' and 'day', 'molecule' and 'atom'.

A.3.2.23**associative relation**

pragmatic relation

relation between two **concepts** (A.3.2.1) having a non-hierarchical thematic connection by virtue of experience

NOTE An associative relation exists between the concepts 'education' and 'teaching', 'baking' and 'oven'.

A.3.2.24**sequential relation**

associative relation (A.3.2.23) based on spatial or temporal proximity

NOTE A sequential relation exists between the **concepts** (A.3.2.1) 'production' and 'consumption', etc.

A.3.2.25**temporal relation**

sequential relation (A.3.2.24) involving events in time

NOTE A temporal relation exists between the **concepts** (A.3.2.1) 'spring' and 'summer', 'autumn' and 'winter'.

A.3.2.26**causal relation**

associative relation (A.3.2.23) involving cause and its effect

NOTE A causal relation exists between the **concepts** (A.3.2.1) 'action' and 'reaction', 'nuclear explosion' and 'fall-out'.

A.3.3 Definitions**A.3.3.1****definition**

representation of a **concept** (A.3.2.1) by a descriptive statement which serves to differentiate it from related concepts

A.3.3.2

intensional definition

definition (A.3.3.1) which describes the **intension** (A.3.2.9) of a **concept** (A.3.2.1) by stating the **superordinate concept** (A.3.2.13) and the **delimiting characteristics** (A.3.2.7)

NOTE The following is an example of an intensional definition for the concept 'incandescent lamp':

incandescent lamp

electric lamp in which a filament is heated by an electric current in such a way that it emits light

A.3.3.3

extensional definition

description of a **concept** (A.3.2.1) by enumerating all of its **subordinate concepts** (A.3.2.14) under one criterion of subdivision

EXAMPLES

Family 18 in the Periodic Table

helium, neon, argon, krypton, xenon and radon

noble gas

helium, neon, argon, krypton, xenon, or radon

A.3.4 Designations

A.3.4.1

designation

designator

representation of a **concept** (A.3.2.1) by a sign which denotes it

NOTE In terminology work, three types of designations are distinguished: symbols, **appellations** (A.3.4.2) and **terms** (A.3.4.3).

A.3.4.2

appellation

name

verbal **designation** (A.3.4.1) of an **individual concept** (A.3.2.2)

A.3.4.3

term

verbal **designation** (A.3.4.1) of a **general concept** (A.3.2.3) in a specific subject field

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ICS 01.040.35; 35.240.80

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