INTERNATIONAL STANDARD

ISO 17115

First edition
2007-07-01

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

Contents

Page

Foreword

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International Standards are drafted in accordance with the rules given in the ISO/IEC Directives, Part 2.

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.

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

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 concept**s (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 **concept**s (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 constraint**s (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.