

DRAFT INTERNATIONAL STANDARD

ISO/DIS 16278

ISO/TC 215

Secretariat: ANSI

Voting begins on:
2014-01-30

Voting terminates on:
2014-06-30

Health informatics — Categorical structure for terminologies systems of human anatomy

Informatique de santé — Structure catégorielle des terminologies d'anatomie humaine

ICS: 35.240.80

iTeh STANDARD PREVIEW
(standards.iteh.ai)
Full standard:
<https://standards.iteh.ai/catalog/standards/sist/2066b088-f0a7-434d-9def-709a76d3142d/iso-16278-2016>

ISO/CEN PARALLEL PROCESSING

This draft has been developed within the International Organization for Standardization (ISO), and processed under the **ISO lead** mode of collaboration as defined in the Vienna Agreement.

This draft is hereby submitted to the ISO member bodies and to the CEN member bodies for a parallel five month enquiry.

Should this draft be accepted, a final draft, established on the basis of comments received, will be submitted to a parallel two-month approval vote in ISO and formal vote in CEN.

To expedite distribution, this document is circulated as received from the committee secretariat. ISO Central Secretariat work of editing and text composition will be undertaken at publication stage.

THIS DOCUMENT IS A DRAFT CIRCULATED FOR COMMENT AND APPROVAL. IT IS THEREFORE SUBJECT TO CHANGE AND MAY NOT BE REFERRED TO AS AN INTERNATIONAL STANDARD UNTIL PUBLISHED AS SUCH.

IN ADDITION TO THEIR EVALUATION AS BEING ACCEPTABLE FOR INDUSTRIAL, TECHNOLOGICAL, COMMERCIAL AND USER PURPOSES, DRAFT INTERNATIONAL STANDARDS MAY ON OCCASION HAVE TO BE CONSIDERED IN THE LIGHT OF THEIR POTENTIAL TO BECOME STANDARDS TO WHICH REFERENCE MAY BE MADE IN NATIONAL REGULATIONS.

RECIPIENTS OF THIS DRAFT ARE INVITED TO SUBMIT, WITH THEIR COMMENTS, NOTIFICATION OF ANY RELEVANT PATENT RIGHTS OF WHICH THEY ARE AWARE AND TO PROVIDE SUPPORTING DOCUMENTATION.



Reference number
ISO/DIS 16278:2013(E)

© ISO 2013

iTeh STANDARD PREVIEW
(standards.iteh.ai)

Full standard:
<https://standards.iteh.ai/catalog/standards/sist/2066b088-f0a7-434d-9def-709a76d3142d/iso-16278-2016>

Copyright notice

This ISO document is a Draft International Standard and is copyright-protected by ISO. Except as permitted under the applicable laws of the user's country, neither this ISO draft nor any extract from it may be reproduced, stored in a retrieval system or transmitted in any form or by any means, electronic, photocopying, recording or otherwise, without prior written permission being secured.

Requests for permission to reproduce should be addressed to either ISO at the address below or ISO's member body in the country of the requester.

ISO copyright office
Case postale 56 • CH-1211 Geneva 20
Tel. + 41 22 749 01 11
Fax + 41 22 749 09 47
E-mail copyright@iso.org
Web www.iso.org

Reproduction may be subject to royalty payments or a licensing agreement.

Violators may be prosecuted.

Contents

Page

Foreword	Error! Bookmark not defined.
Introduction	4
1 Scope	5
1.1 Main purpose	5
1.2 Target groups	5
1.3 Topics outside the scope	6
2 Normative references	6
3 Terms and definitions	6
4 Categorical structure for terminologies of human anatomy description	8
4.1 Principles	8
4.2 Anatomical categories (3.7)	8
4.3 Precise goal of the categorical structure (3.8)	11
4.4 List of anatomical relations (3.8)	11
4.5 List of minimal anatomical domain constraints (3.9)	13
5 Conformance	13
Annex A (informative) A reference ontology for biomedical informatics: the Foundational Model of Anatomy	14
A.1 General	14
A.2 Principles, Ontological Framework and Implementation of the FMA	14
A.3 Extensions of the FMA beyond human macroscopic anatomy	15
A.4 Relations in the FMA and their influence on other ontologies	16
A.5 Querying the FMA	16
A.6 Evaluation of the FMA	17
A.7 Uses and Selected Applications of the FMA	17
A.7.1 Research in ontologies, informatics and computer science	17
A.7.2 Research in biomedical imaging	18
A.7.3 Design of information systems	18
A.7.4 Clinical Informatics	18
A.7.5 Education	18
A.7.6 Sources of Anatomical Information	19
Bibliography	20

Introduction

This International standard specifies a categorial structure for terminological systems of human anatomy. Computer-based processing and the interchange of medical or clinical information requires various kinds of terminological systems to represent that information, such as controlled vocabularies, classifications, nomenclatures, terminologies and thesauri, with or without coding schemes.

The specific terminological issues in the field of health informatics are:

- large number of different terminological systems are available in different clinical specialties;
- large overlap among the subject fields involved;
- large number of codes and rubrics, typically in the order of magnitude of 10 000 to 100 000 entries, in commonly used terminological systems;
- increasing need for re-use of coded data in different health-care contexts;
- poly-sema across different clinical specialties and sometimes within them.

The integration of computer-based medical records and administrative information systems in Electronic Health Records (EHR) require rationalization in the field, and a uniform way to represent the meaning of medical concepts to ensure that the receiver EHR of a message will catch the meaning introduced by the sender EHR and not only the string of characters embedded in it.

It is not possible to impose a rigid uniform standardized natural language clinical terminology on healthcare professional providers. Nevertheless standards need to be provided for guiding the development of terminologies in the different sub domains of healthcare to allow semantic interoperability between them. To this end a domain specific semantic representation has been developed (EN 12264) and applied in a series of specific initiatives including European Pre standards (ENV), European Standards (EN) and international ISO standards on various subject fields to describe a set of categorial structures in partially overlapping subject fields: Human anatomy is central to medical terminology (surgical procedures, carcinoma staging, annotation of radiological findings, disease, clinical laboratory and so forth) and also to many scientific and bio-informatics study beyond the scope of clinical medicine . In the US the University of Washington has developed in the public domain an anatomical terminology for EHR named the Digital Anatomist Foundational Model of Anatomy (FMA for short), a reference ontology for biomedical informatics.

International standardisation efforts by CEN and ISO related to Electronic Health Records and semantic interoperability have resulted in a number of categorial structures which are a step towards supporting healthcare terminological systems with a full concept system or ontology that in turn will support multipurpose uses and safe communication. In the present categorial structure standard, several of the definitions of basic terms related to categorial structures have been updated to comply with the most recent version of ISO 17115:2007.

Adequate field testing in several countries, revision and integration have provided the comprehensive basis for this International standard.

1 Scope

1.1 Main purpose

This International standard defines the characteristics required to synthetically describe the organisation and content of human anatomy within a terminological system. It is intended primarily for use with computer-based applications such as clinical electronic health records, decision support and for various bio-medical research purposes.

This International standard will serve to

- facilitate the construction of new terminological systems in a regular form which will increase their coherence and expressiveness;
- facilitate maintenance of human anatomy within terminological systems;
- increase consistency and coherence of existing terminological system;
- allow systematic cross-references between items of human anatomy in different types of terminological systems;
- facilitate convergence among human anatomy within terminological systems;
- make explicit the overlap for human anatomy between different health care domains terminological systems;
- provide elements for negotiation about integration of different terminological systems into information systems between the respective developers;
- enable the systematic evaluation of human anatomy within terminological systems.

1.2 Target groups

The International standard itself is not suitable or intended for use by, individual clinicians or hospital administrators.

The target groups for this International standard are:

- designers of specialised standard healthcare terminological categorial structures;
- developers of healthcare terminological systems including classifications and coding systems;
- producers of services for terminological systems and designers of software including natural language processing;
- information modellers, knowledge engineers, and standards developers building models for health information management systems;
- developers of information systems that require an explicit representation of healthcare terminological systems;
- developers of marked-up standards for representation of healthcare documents.

1.3 Topics outside the scope

This International Standard does not include categorial structure that may be necessary for the description of developmental anatomy during the human life cycle, which includes prenatal development, post-natal growth and aging.

This International Standard has been developed for use as an integrated part of computer-based applications and for the electronic healthcare record. It would be of limited value for manual use.

It is not the purpose of this International Standard to standardise the end user classification of human anatomy terminology or to conflict with the concept systems embedded in national practice and languages.

2 Normative references

The following referenced documents are indispensable for the application of this document. For dated references, only the edition cited applies. For undated references, the latest edition of the referenced document (including any amendments) applies.

EN 12264 2005 *Health Informatics - Categorial structure for systems of concepts*

3 Terms and definitions

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

3.1 Human anatomy
biological science that concerns the discovery, analysis and representation of the structural organisation of the human body

NOTE Human anatomy thus defined encompasses the material objects from the granularity level of the whole human body to that of cell parts, portions of body substances, and non-material entities such as surfaces, spaces, lines and points, that form the phenotypic organisation of the human body. Although encompassed by the definition of anatomical structure (4.2.9), biological macromolecules do not come under the purview of the science human anatomy.

3.2 anatomical entity
entity that constitutes the structural organisation of a particular human body

3.3 spatial dimension
number of dimensions of the entity in space

EXAMPLE 1 Entities with spatial dimension of value 3 are organs, cells and body cavity.

EXAMPLE 2 Entities with spatial dimension of value 2: the plane of the esophagogastric junction and the surface of the parietal part of the head.

EXAMPLE 3 Entities with spatial dimension of value 1: pectinate line, linea aspera and superior nuchal line.

EXAMPLE 4 Entities with spatial dimension of value 0: the pointed extremity of petrous part of temporal bone, pointed extremity of the orbit and the pointed extremity of the sacrum.

NOTE TO BE REMOVED BEFORE PUBLICATION: there is a proposal to remove the category *spatial dimension* as it is considered too restrictive for many anatomical terminologies. Please comment on this proposal.

3.4**three-dimensional shape**

shape of an anatomical entity of spatial dimension with value 3

EXAMPLE Hollow cylinder.

3.5**terminology**

set of designations belonging to one special language

[ISO 1087-1:2000]

3.6**anatomical term**

verbal designation of an anatomical entity (3.2)

3.7**anatomical category**

type of anatomical entity shared by all the individual instances in existence in the present, past and future

EXAMPLE The anatomical category liver is instantiated by this liver and all individual livers in existence in the present, past and future.

NOTE 1 Anatomical categories may be more or less general. Where one anatomical category is subsumed by another, the *is_a* relation is asserted to obtain between the more specific or subsumed category and the more general or subsuming anatomical category.

NOTE 2 Each anatomical entity instantiates some anatomical category.

3.8**anatomical relation**

relation between two or more anatomical categories derived from corresponding relations between instances of the respective categories

EXAMPLE A is_a B defined to obtain when every entity in category A is at the same time an entity in category B.

B has_part_ A defined to obtain when every entity in category A has some entity in category B as part.

Other examples of anatomical relations manifesting this every-some structure are: contained_in adjacent_to, attached_to

NOTE The definition is adapted from the representation of types of characteristics in EN 12264 and authorised by an anatomical domain constraint (3.9).

3.9**anatomical domain constraint**

rule prescribing the set of representations of anatomical relations (3.8) that are valid to specialise an anatomical category (3.5) in a certain domain

NOTE The definition is adapted from domain constraint in EN 12264.

3.10**anatomical categorial structure**

minimal set of anatomical domain constraints (3.9) for representing anatomical entities (3.2) in a precise domain to achieve a precise goal

NOTE The definition is adapted from the categorial structure in EN 12264.

4 Categorical structure for terminological systems of human anatomy description

4.1 Principles

The categorical structures for terminological systems of human anatomy are in conformity with the categorical structure as prescribed by clause 4 of EN 12264:2005.

To describe an anatomical **categorical structure** (3.10), the following information shall be provided:

- a) **anatomical categories** (3.7) that organise the **anatomical entities** (3.2) and the **anatomical relations** (3.8) dividing their representation in the domain;
- b) precise goal of the **anatomical categorical structure** (3.10);
- c) list of the representations of **anatomical relations** (3.8) authorised by **anatomical domain constraints** (3.9);
- d) list of minimal **anatomical domain constraints** (3.9) required by the goal of the **anatomical categorical structure** (3.1).

4.2 Anatomical categories (3.7)

4.2.1

Physical anatomical entity

anatomical entity that has a spatial dimension (3.3)

EXAMPLE Organ, surface, apex of the orbit.

4.2.2

Immaterial physical anatomical entity

physical anatomical entity that has no mass

EXAMPLE Anatomical space, anatomical surface (diaphragmatic surface of left ventricle).

4.2.3

Anatomical space

immaterial physical anatomical entity which has a spatial dimension (3.3) of value 3

EXAMPLE Thoracic cavity.

4.2.4

Anatomical surface

immaterial physical anatomical entity which has a spatial dimension (3.3) of value 2

EXAMPLE Diaphragmatic surface of heart.

4.2.5

Anatomical line

immaterial physical anatomical entity which has a spatial dimension (3.3) of value 1

EXAMPLE Inferior margin of liver.

4.2.6

Anatomical point

immaterial physical anatomical entity which has a spatial dimension (3.3) of value 0

EXAMPLE Apex of this heart.

4.2.7**Material physical anatomical entity**

physical anatomical entity that has a mass

EXAMPLE Liver, cell nucleus, portion of blood.

4.2.8**Body substance**

material physical anatomical entity that has no inherent shape (3.4)

EXAMPLE Portion of blood, portion of cytosol.

4.2.9**Anatomical structure**

material physical anatomical entity that has an inherent shape (3.4) and is generated by a coordinated expression of the organism's own structural genes

EXAMPLE Thorax ,tibia, hepatocyte.

NOTE Post surgery anatomy (e.g surgically created stomas, stumps, vascular and intestinal anastomoses) is not an anatomical structure . When useful it shall be defined in the categorical structure needing it: e.g for surgical procedures.

4.2.10**Cell**

anatomical structure that consists of cytoplasm surrounded by a plasma membrane

EXAMPLE Leukocyte, hepatocyte.

4.2.11**Organ**

anatomical structure that consists of a maximal collection of cardinal organ parts so connected to one another that together they constitute a self-contained unit of macroscopic anatomy, morphologically distinct from other such units

EXAMPLE Heart , tibia, urinary bladder.

4.2.12**Cardinal organ part**

anatomical structure that consists of two or more portions of tissue, spatially related to one another in patterns determined by coordinated gene expression; together with other contiguous cardinal organ parts it constitutes an organ

EXAMPLE Upper lobe of right lung, shaft of humerus, left ventricle, head of pancreas.

4.2.13**Portion of tissue**

anatomical structure that consists of a directly connected collection of similarly specialised cells and intercellular matrix, aggregated according to genetically determined spatial relationships

EXAMPLE Portion of smooth muscle, portion of endothelium.

4.2.14**Cardinal body part**

anatomical structure that has as its parts the most complete set of diverse subclasses of organ and cardinal organ parts spatially associated with either the skull, a segment of the vertebral column or a complete set of bones of the appendicular skeleton; it is partially surrounded by skin and forms a distinct morphological subdivision of the body; together all cardinal body parts constitute the body

EXAMPLE Head, neck, trunk, upper limb.

4.2.15

Body region

sub volume of a cardinal body part (4.2.14) demarcated by at least one fiat boundary

EXAMPLE Epigastrium, femoral triangle.

4.2.16

Organ systems

anatomical structure that consists of organs predominantly of the same anatomical category, which are interconnected by zones of continuity

EXAMPLE Alimentary system, musculoskeletal system.

NOTE 1 Each musculo-skeletal system is comprised of instances of the classes *muscle* (organ), *bone* (organ), *joint*, and *ligament* (organ), which together form an interconnected anatomical structure.

NOTE 2 Subdivisions of a musculoskeletal system are its skeletal system and articular system, which consist of collections of bones and joints, respectively; the joints interconnecting the bones and vice versa.

NOTE 3 Several of the commonly known systems of the body satisfy this criterion but the endocrine and immune systems do not. Therefore, they are Body systems but not Organ systems. The rationale for subdividing the body into systems is usually claimed to be function. Organ systems have organs as their direct and connected parts. There are many other systems in the body that are not constituted by organs. Some are anatomical structures, others are not.

4.2.17

Anatomical cluster

anatomical structure that consists of a heterogeneous collection of organ parts grouped together in a predetermined manner, but which do not constitute the whole or a subdivision of either a body part or an organ system

EXAMPLE Joint, adnexa of the uterus, root of the lung, renal pedicle, back.

NOTE Such clusters can be composed of cells (e.g., splenic cord consists of erythrocytes, reticular cells, lymphocytes, monocytes, and plasma cells), cardinal organ parts (e.g., tendinous or rotator

cuff consists of the fused tendons of several muscles), as well as of organs (e.g., lacrimal apparatus consists of a lacrimal gland, lacrimal sac, and nasolacrimal duct, each of which is an organ).

4.2.18

Anatomical set

material anatomical entity that consists of the maximum number of discontinuous members of the same class

EXAMPLE Set of cranial nerves, ventral branches of aorta, set of mammary arteries, thoracic viscera, dental arcade.

NOTE 1 Anatomical sets have members, rather than parts (e.g., each instance of oculomotor *nerve* is a member of some instance of *set of cranial nerves*).

NOTE 2 Membership in an anatomical set is often regarded as a kind of part relation. In anatomy, the distinction between part and membership relations is that there is direct continuity of a part with its respective whole, whereas no direct continuity exists between members of an anatomical set.

NOTE 3 In an anatomical set the meaning of set is different from the meaning of a set in mathematics.

4.2.19

Anatomical junction

anatomical structure in which two or more anatomical structures are in physical continuity with one another or intermingle their component parts

EXAMPLE Suture, commissure of the mitral valve, gastroesophageal junction, synapse.