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Health informatics — Categorial structure for terminological systems of human anatomy

Informatique de santé — Structure catégorielle des systèmes terminologiques de l'anatomie humaine

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Foreword

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For an explanation on the meaning of ISO specific terms and expressions related to conformity assessment, as well as information about ISO's adherence to the WTO principles in the Technical Barriers to Trade (TBT) see the following URL: Foreword - Supplementary information

The committee responsible for this document is ISO/TC 215, *Health informatics*.

Introduction

This International Standard specifies a categorial structure for terminologies 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 the following:

— 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;

— polysemy 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 Standards (ISO) 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 Standardization 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.

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

Health informatics — Categorial structure for terminological systems of human anatomy

1 Scope

This International Standard defines the characteristics required to synthetically describe the organization 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, and

— enable the systematic evaluation of human anatomy within terminological systems.

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 the following:

— designers of specialized 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.

This International Standard does not include categorial structure that might 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 standardize the end user classification of human anatomy terminology or to conflict with the concept systems embedded in national practice and languages.

2 Terms and definitions

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

2.1
human anatomy
biological science that concerns the discovery, analysis and representation of the structural organization of the human body

Note 1 to entry: 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 organization of the human body. Although encompassed by the definition of anatomical structure (3.2.9), biological macromolecules do not come under the purview of the science human anatomy.

2.2
anatomical entity
entity that constitutes the structural organization of a particular human body

2.3
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.

2.4
three-dimensional shape
shape of an anatomical entity of spatial dimension with value 3

EXAMPLE Hollow cylinder.

2.5
terminology
set of designations belonging to one special language

[SOURCE: ISO 1087-1:2000]

2.6
anatomical term
verbal designation of an *anatomical entity* (2.2)

2.7
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 to entry: 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 to entry: Each anatomical entity instantiates some anatomical category.

2.8
anatomical relation
relation between two or more anatomical categories derived from corresponding relations between instances of the respective categories

EXAMPLE 1 A is_a B defined to obtain when every entity in category A is at the same time an entity in category B.

EXAMPLE 2 B has_part A defined to obtain when every entity in category B has some entity in category A as part.

Note 1 to entry: Other examples of anatomical relations manifesting this every-some structure are: contained_in adjacent_to, and attached_to.

Note 2 to entry: The definition is adapted from the representation of types of characteristics in EN 12264 and authorised by an *anatomical domain constraint* (2.9).

2.9
anatomical domain constraint
rule prescribing the set of representations of *anatomical relations* (2.8) that are valid to specialize an *anatomical category* (2.5) in a certain domain

Note 1 to entry: The definition is adapted from domain constraint in EN 12264.

2.10
anatomical categorial structure
minimal set of *anatomical domain constraints* (2.9) for representing *anatomical entities* (2.2) in a precise domain to achieve a precise goal

Note 1 to entry: The definition is adapted from the categorial structure in EN 12264.

3 Categorial structure for terminologies of human anatomy description

3.1 Principles

The categorial structures for terminologies of human anatomy are in conformity with the categorial structure as prescribed by EN 12264:2005, Clause 4.

To describe an anatomical categorial structure (2.10), the following information shall be provided:

a) anatomical categories (2.7) that organize the anatomical entities (2.2) and the anatomical relations (2.8)dividing their representation in the domain;

b) precise goal of the anatomical categorial structure (2.10);

c) list of the representations of anatomical relations (2.8) authorized by anatomical domain constraints (2.9);

d) list of minimal anatomical domain constraints (2.9) required by the goal of the anatomical categorial structure (2.10).

3.2 Anatomical categories (2.7)

3.2.1

physical anatomical entity

anatomical entity that has a *spatial dimension* (2.3)

EXAMPLE Organ, surface, apex of the orbit.

3.2.2

immaterial physical anatomical entity

physical anatomical entity that has no mass

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

3.2.3

anatomical space

immaterial physical anatomical entity which has a *spatial dimension* (2.3) of value 3

EXAMPLE Thoracic cavity.

3.2.4

anatomical surface

immaterial physical anatomical entity which has a *spatial dimension* (2.3) of value 2

EXAMPLE Diaphragmatic surface of heart.

3.2.5

anatomical line

immaterial physical anatomical entity which has a *spatial dimension* (2.3) of value 1

EXAMPLE Inferior margin of liver.

3.2.6

anatomical point

immaterial physical anatomical entity which has a *spatial dimension* (2.3) of value 0

EXAMPLE Apex of this heart.

3.2.7

material physical anatomical entity

physical anatomical entity that has a mass

EXAMPLE Liver, cell nucleus, portion of blood.

3.2.8

body substance

material physical anatomical entity that has no *inherent shape* (2.4)

EXAMPLE Portion of blood, portion of cytosol.

3.2.9

anatomical structure

material physical anatomical entity that has an *inherent shape* (2.4) and is generated by a coordinated expression of the organism's own structural genes

EXAMPLE Thorax, tibia, hepatocyte.

Note 1 to entry: Post-surgical anatomy (e.g surgically created stomas, stumps, vascular and intestinal anastomoses) is not an anatomical structure. When used, it shall be defined in the categorical structure needing it, e.g. for surgical procedures.

3.2.10

cell

anatomical structure that consists of cytoplasm surrounded by a plasma membrane

EXAMPLE Leukocyte, hepatocyte.

3.2.11

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.

3.2.12

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.

3.2.13

portion of tissue

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

EXAMPLE Portion of smooth muscle, portion of endothelium.

3.2.14

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

EXAMPLE Head, neck, trunk, upper limb.

Note 1 to entry: Together, all cardinal body parts constitute the body.

3.2.15

body region

sub volume of a *cardinal body part* (3.2.14) demarcated by at least one fiat boundary

EXAMPLE Epigastrium, femoral triangle.

3.2.16

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 to entry: 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 to entry: 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 to entry: 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.

