INTERNATIONAL STANDARD

ISO 16278

First edition
2016-03-01

Health informatics — Categorial structure for terminological systems of human anatomy

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

Reference number
ISO 16278:2016(E)

COPYRIGHT PROTECTED DOCUMENT

ISO copyright office
Ch. de Blandonnet 8 • CP 401
CH-1214 Vernier, Geneva, Switzerland
Tel. +41 22 749 01 11
Fax +41 22 749 09 47
copyright@iso.org
www.iso.org

Contents

Page

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 www.iso.org/directives).

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 www.iso.org/patents).

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 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.

3.2.17

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 1 to entry: 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).

3.2.18

anatomical set

material physical 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 to entry: 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 to entry: 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 exits exist between members of an anatomical set.

3.2.19

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.

3.3 Precise goal of the categorial structure (2.10)

The goal of each anatomical terminology used in the terminological systems of healthcare and biomedical science shall be defined by the users and indicates the situations and applications for which the categorial structure is intended and the limits of use.

EXAMPLE Controlled vocabulary production for clinicians or comparison with another terminological system for coding centres.

3.4 List of anatomical relations (2.8)

3.4.1

has_part

anatomical relation (2.8) which holds between an anatomical category A and an anatomical category B (with one to three dimensions both) if and only if

— each instance of A has some instance of B as a proper part and/or

— proper part between two particular entities a and b means there is a complement c which together with b accounts for the whole of a".

EXAMPLE Stomach has_part fundus. Together with body and pyloric antrum fundus accounts for the whole (100 %) of stomach.

3.4.2

A contained_in B

anatomical relation (2.8) that holds between each *anatomical entity* (2.2) in category *A* contained in some anatomical entity in category *B*

EXAMPLE Urinary bladder *contained_in* pelvic cavity.

Note 1 to entry: The former is a body substance or an anatomical structure; the latter is an anatomical space.

Note 2 to entry: *Contained_in* does not imply *part_of*. Although cavity of urinary bladder is *part_of* urinary bladder, urine *part_of* urinary bladder is an invalid assertion.

Note 3 to entry: Imposing such a restricted meaning on the contained_in relation may seem pedantic, because it implies that an assertion such as brain contained_in skull needs to be replaced by two related statements: brain contained_in cranial cavity, cranial cavity part of skull. The purpose of such specificity at the level of terminological representation is to assure that the role of container is constrained to anatomical structures which have anatomical space as one of their part. This constraint will prevent a reasoner from returning results such as right lobe of liver contained_in liver.

3.4.3

A adjacent_to B

anatomical relation (2.8) which holds between each *anatomical entity* (2.2) in category A which is adjacent to some entity in category B

EXAMPLE Examples are the following:

Spleen *adjacent_to* stomach,

Kidney *adjacent_to* quadratus lumborum,

Medial surface of spleen *adjacent_to* posterior surface of stomach,

Posterior surface of kidney *adjacent_to* anterior surface of quadratus lumborum.

Note 1 to entry: Two anatomical entities of the same dimension are adjacent when they are spatially proximate, share no boundary or parts, and are separated by no further anatomical entities of the same dimension.

Note 2 to entry: The relation of adjacency needs to be asserted at different levels of granularity according to context: Examples of various levels of granularity include organism, organ system, organ, organ part, maximal portion of tissue, cell and subcellular organelle.

Note 3 to entry: A*djacent_to* may be qualified with the aid of qualitative anatomical coordinates such as *anterior_to*, *posterior_to*, *superior_to*, *inferior_to*, *medial_to*, *lateral_to* depending upon the value of the trajectory relationship with the body in the standard anatomical position.

3.4.4

A continuous_with B

anatomical relation (2.8) that holds between each *anatomical entity* (2.2) in category *A* and some entity in category B when there is no bona fide boundary (real physical discontinuity) between the related entities and their parts

EXAMPLE 1 Arterial trunk *continuous_with* branch of arterial trunk.

EXAMPLE 2 Thoracic part of oesophagus *continuous_with* abdominal part of oesophagus.

Note 1 to entry: *Continuous_with* like *adjacent_to*, can be qualified: thoracic part of oesophagus *Continuous_with superiorly*, cervical part of oesophagus; thoracic part of oesophagus *continuous_with inferiorly* abdominal part of oesophagus.

Note 2 to entry: Qualification here is different from the case of anterior, posterior, etc. qualifications of *adjacency* (3.4.3.). In this case, the qualification pertains to the parts of the entities related. In the case of adjacency, the qualification is of the relation itself.

3.4.5

A attached_to B

anatomical relation (2.8) which holds between each *anatomical entity* (2.2) in category A and some entity in category B when some of the parts of the entity in category A are intermingled with some of the parts of the entity in category B across a portion of their maximal boundary which the related entities share

EXAMPLE Each patellar ligament is attached_to the patella at a narrow area along the lower margin of the latter and also to the tuberosity of the tibia. All these anatomical structures have their own real boundaries, but at its proximal and distal ends the patellar ligament comes in intimate contact with circumscribed areas of each bone, where extensions of its collagen fiber bundles (so called 'Sharpey's fibers') penetrate the bone and intermingle with each bone's own collagen fibers network. The ligament may be separated from the bone only by severing Sharpey's fiber.

The circumference of the tympanic membrane is attached_to bones of the skull forming the external auditory meatus.

The visceral pleura is attached_to the lung proper intermingling its loose connective tissue on its non-serous surface with the fibrous stroma of the lung.

The brachilis muscle is attached_to the humerus.

3.4.6

has_dimension

anatomical relation (2.8) that relates an anatomical entity to the number of its *spatial dimension* (2.3)

EXAMPLE Wall of stomach *has_dimension* 3.

3.4.7

has_shape

anatomical relation (2.8) that relates an anatomical entity to its *three-dimensional shape* (2.4)

EXAMPLE Oesophagus *has_shape* hollow cylinder.

3.4.8

has_boundary

anatomical relation (2.8) which relates categories of anatomical entities of one to three dimensions to categories of immaterial physical anatomical entities of one dimension lower, called bounding anatomical entities

EXAMPLE 1 Cavity of stomach *has_boundary* internal surface of stomach.

EXAMPLE 2 Oesophagus and stomach has_boundary plane of gastro esophageal junction.

EXAMPLE 3 Abdominal cavity *has_boundary* plane of pelvic inlet.

Note 1 to entry: Such bounding entities delimit anatomical entities of one, two or three dimensions from one another. A boundary may be bona fide or fiat. A bona fide boundary of an anatomical structure is a real physical discontinuity. A fiat boundary is a virtual plane or line such as those that demarcate the oesophagus from the stomach.

Note 2 to entry: The practical application of boundary information is critical for example to processes of automatic image segmentation to the analysis of volumetric datasets.

Note 3 to entry: Each stomach can be decomposed into two partitions: one, into fundus, body and pyloric antrum and the other in wall and cavity. In each case, one or more fiat boundaries may be involved. Each decomposition or partition accounts for the whole (100 %) of the partitioned entity. A fiat boundary in the cavity of stomach demarcates the cavity of pyloric antrum from the cavity of the body of the stomach. A fiat boundary in the internal surface of stomach demarcates the internal surface of pyloric antrum from the internal surface of the body of the stomach.

3.5 List of minimal anatomical domain constraints (2.9)

The list shall contain the different anatomical relations (2.8) from 3.4 and the different related anatomical categories from 3.2, which are valid and necessary for the precise goal from 3.3 of an anatomical categorial structure (2.10).

4 Conformance

An anatomical categorial structure (2.10) claiming conformance to this International Standard shall provide the information described by items 3.2, 3.3 and 3.5 and shall be conformant to the following three rules:

— anatomical categorial structure (2.10) claiming conformance to this International Standard shall have as root nodes any anatomical entity (3.2) from one anatomical category (2.7) listed in item 3.2.

— anatomical categorial structure (2.10) claiming conformance to this International Standard shall make precise the level of granularity of the classes of anatomical categories (2.7) used as described by all the items under 3.2.

— anatomical categorial structure (2.10) claiming conformance to this International Standard shall use, when necessary, the anatomical relations (2.8) only as described by all the items under 3.4.

Annex A
(informative)

A reference ontology for biomedical informatics: the Foundational Model of Anatomy

A.1 General

The Foundational Model of Anatomy (FMA), initially developed as an enhancement of the anatomical content of UMLS, is a domain ontology of the concepts and relationships that pertain to the structural organization of the human body. It encompasses the material objects from the molecular to the macroscopic levels that constitute the body and associates with them non-material entities (spaces, surfaces, lines, and points) required for describing structural relationships. The disciplined modelling approach employed for the development of the FMA relies on a set of declared principles, high level schemes, Aristotelian definitions and a frame-based authoring environment. The FMA is applying, as a reference, ontology in biomedical informatics for correlating different views of anatomy, aligning existing and emerging ontologies in bioinformatics ontologies and providing a structure-based template for representing biological functions.

The principles, anatomical categories and relations adopted for the construction of this International Standard for Categorial Structures of Human Anatomy have been largely derived from the Foundational Model of Anatomy (FMA).

There is no definitions in this International Standard which are in contradiction with the FMA but this International Standard is not establishing the FMA as an IS. In practice, when using the whole FMA, a terminology will be compliant with this International Standard. A terminology using only the Categorial Structure of Human Anatomy will be compliant as well without using the whole FMA.

The following annotated bibliography of the Digital Anatomist Foundational Model of Anatomy (FMA) gives a sound insight within the work done on human anatomy.

A.2 Principles, ontological framework and implementation of the FMA

— Rosse, C., M. Ben-Said, K.R. Eno, J.F. Brinkley 1995 Enhancements of Anatomical Information in UMLS Knowledge Sources. In: Gardner RM, editor. Proc 19th Annu Symp Comput Appl Med Care (SCAMC 95). Philadelphia: Hanley & Belfus, 1995: 873-877.

NOTE 1 The first report on the initial version of the FMA (Digital Anatomist Symbolic Knowledge Base) and its relation to the anatomical content of UMLS.

— Rosse, C., Shapiro, L.G. and Brinkley, JF. 1998. The Digital Anatomist Foundational Model: Principles for Defining and Structuring its Concept Domain. In Chute EG (ed): A paradigm shift in health care information systems: clinical infrastructures for the 21st century. JAMIA Symposium Supplement. ‘98: 820-824

NOTE 2 A preliminary account of principles for guiding the establishment of an ontology of anatomy.

— Rosse C, Mejino JL, Modayur BR, Jakobovits R, Hinshaw KP, Brinkley JF. 1998. Motivation and organizational principles for anatomical knowledge representation: the Digital Anatomist Symbolic Knowledge Base. J. Am. Med. Informatics Assoc.5:17-40.

NOTE 3 Distinguishes two meanings of the term ‘anatomy’: 1. the structural organization of a biological organism and 2, the scientific discipline that studies this organization, thus separating epistemology and reality. Proposes a comprehensive ontology of the physical entities that constitute a human body based on the structural properties by which anatomical entities can be sorted into classes and distinguished from one another.

— Mejino, J.L.V. and Rosse ,C. 1999. Conceptualizations of Anatomical Spatial Entities in the Digital Anatomist Foundational Model. J. Am. Med. Assoc. AMIA ’99 Symp. Suppl. ‘99: 112-116.

NOTE 4 The FMA is the only ontology that explicitly distinguishes among physical entities between those that have or do not have mass and treats anatomical spaces, surfaces, lines and points as universals or classes.

— Michael J, Mejino JLV, Rosse C. 2001. The role of definitions in biomedical concept representation. JAMIA Symposium Supplement. ‘01:463-467.

NOTE 5 Advocates the need for Aristotelian definitions in biomedical ontologies and illustrates the employment of genus and differentiae for establishing classes of anatomical entities in the FMA.

— Rosse C, Mejino JVL. 2003. A reference ontology for biomedical informatics: the Foundational Model of Anatomy. J Biomed Inform. 36:478-500.

NOTE 6 The most recent comprehensive account of the FMA which replaces a concept-based view with a reality-based representation of more than 75 000 multiply located anatomical entities (universals) which exist in the idealized (canonical) instances that they subsume. In addition to the taxonomy component of the FMA, it gives an account of the structural and developmental relationships that exist between anatomical entities.

NOTE 7 Free access to the updated version of the fully implemented FMA. The categorial anatomical structures and definitions proposed in the document derive primarily from the FMA.

A.3 Extensions of the FMA beyond human macroscopic anatomy

— Martin RF, Mejino JLV, Bowden DM, Brinkley JF, Rosse C. 2001. Foundational model of neuroanatomy: its implications for the Human Brain Project. JAMIA Symposium Supplement. ‘01:438-442.

— Martin RF, Rickard K, Mejino JLV, Agoncillo AV, Brinkley JF, Rosse C. The evolving neuroanatomical component of the Foundational Model of Anatomy. In proc, AMIA fall symposium. 2003; 927.

NOTE 1 The FMA is the only ontology that fully integrates neuroanatomy from the cellular to the macroscopic levels for both the central and peripheral nervous systems with the anatomy of other parts of the body.

— Agoncillo AV, Mejino JLV, Rickard KL, Detwiler LT, Rosse C. Proposed classification of cells in the Foundational Model of Anatomy. Proc AMIA Symp.2003; 775.

NOTE 2 The cellular and subcellular entities represented in the FMA compare in number to those in the gene ontology. Correlation of these two ontologies is being pursued as illustrated by the next reference.

— Gennari, J.H., Silberfein, A., and Wiley, J.C. (2005). Integrating genomic knowledge sources through an anatomy ontology. Proceedings of the Pacific Symposium on Biocomputing 2005, pp. 115-126

— Travillian RS, Rosse C, Shapiro LG. An Approach to the Anatomical Correlation of Species through the Foundational Model of Anatomy. Proc AMIA Symp. 2003; 669-673.

NOTE 3 Extension of the FMA to the anatomy of other mammalian species, particularly to that of the mouse, is being pursued by our own as well as other groups.

— Cook DL, Mejino JLV Jr, Rosse C. Rosse C. Evolution of a Foundational Model of Physiology: Symbolic Representation for Functional Bioinformatics**.** In *Proceedings, MedInfo 2004*, pages 336-340, San Francisco, CA.

NOTE 4 The FMA furnishes the participants in all physiological and pathological processes.

— Kumar A, Yip YL, Smith B, Marwede D, Novotny D. An ontology for carcinoma classification for clinical bioinformatics.Stud Health Technol Inform. 2005;116:635-40

— Rosse C, Kumar A, Mejino JLV, Cook DL, Detwiler LT, Smith B. 2005. A Strategy for Improving and Integrating Biomedical Ontologies. Proc AMIA Symp 2005 To appear

NOTE 5 A high-level or meta-ontology is proposed through the integration of Basic Formal Ontology and the FMA, which encompasses anatomical and pathological continuants as well as physiological and pathological occurrents. The framework of this Ontology of Biomedical Reality (OBR) takes account of organismal entities in the purview of the basic biomedical sciences.

A.4 Relations in the FMA and their influence on other ontologies

— Neal, P.J., Shapiro, L.G. and Rosse, C. 1998. The Digital Anatomist Spatial Abstraction: a scheme for the spatial description of anatomical entities. In Chute CG (ed): A paradigm shift in health care information systems: clinical infrastructures for the 21st century. JAMIA Symposium Supplement. 1998:423-427.

NOTE 1 The first publication that adopts for the domain of anatomy distinctions between boundary and part relations from general spatial theory.

— Mejino JLV Jr, Agoncillo AV, Rickard KL, Rosse C. Representing Complexity in Part-Whole Relationships within the Foundational Model of Anatomy. Proc AMIA Symp. 2003;450-454.

NOTE 2 The FMA accommodates multiple, overlapping ways to decompose anatomical entities into their parts to accord with the different contexts in which anatomy is applied in biomedicine.

— Smith B, Rosse C. The role of foundational relations in the alignment of biomedical ontologies. In *Proceedings, MedInfo 2004*, pages 444-448, San Francisco, CA.

NOTE 3 The relations IS_A and HAS_PART are formalized for instances and classes of the FMA.

— Mejino JLVJ, Rosse C. Symbolic modeling of structural relationships in the Foundational Model of Anatomy. In: Proceedings, First International Workshop on Formal Biomedical Knowledge Representation (KR-MED 2004). Whistler Mountain, Canada; 2004.

NOTE 4 Natural language definitions are provided for a whole suite of structural relationships implemented in the FMA.

— Smith B, Mejino Jr. JLV, Schulz S, Kumar A, Rosse C: Anatomical Information Science. In Cohn AG, Mark DM (editors): Spatial Information Theory; Proceedings of International Conference, COSIT 2005, Ellicottville, NY, USA, September 14-18, 2005. p.149.

NOTE 5 Formalizes and extends the definitions of the FMA's suite of structural relations.

— Smith B, Ceusters W, Klagges B, Köhler J, Kumar A, Lomax J, Mungall C, Neuhaus F, AL Rector, Rosse C.: Relations in biomedical ontologies. *Genome Biology* 2005, 6:R46 doi:10.1186/gb-2005-6-5-r46

NOTE 6 Redefines some of the relations dealt with by the previous two publications in the context of Open Biomedical Ontologies.

— Donnelly M, Bittner T, Rosse C. A formal theory for spatial representation and reasoning in biomedical ontologies. Artif Intell Med. 2005 To appear

NOTE 7 Evaluates spatial representation of anatomy in the FMA and GALEN and proposes and extension of formal spatial theory to anatomy.

A.5 Querying the FMA

— Mork P, Brinkley JF, Rosse C. 2003. OQAFMA querying agent for the Foundational Model of anatomy: providing flexible and efficient access to a large semantic network. J Biomed Inform 2003;36:501-517.

— Distelhorst G, Srivastava V, Rosse C, Brinkley JF. A Prototype Natural Language Interface to a Large Complex Knowledge Base, the Foundational Model of Anatomy. Proc AMIA Symp. 2003: 200-204.

— Detwiler LT, Mejino JLV, Rosse C, Brinkley JF. Efficient web-based navigation of the Foundational Model of Anatomy. Proceedings AMIA Symposium. 2003; 829.

— Detwiler LT, Chung E, Li A, Mejino JLV Jr, Agoncillo AV, Brinkley JF, Rosse C. A Relation-Centric Query Engine for the Foundational Model of Anatomy. In *Proceedings, MedInfo 2004*, pages 341-345, San Francisco, CA.

A.6 Evaluation of the FMA

— Shapiro LG, Chung E, Detwiler T, Mejino JLV, Agoncillo AV, Brinkley JF, Rosse C. 2005. Processes and problems in the formative evaluation of an interface to the Foundational Model of Anatomy knowledge base. J Am Med Inform Assoc. 2005;12:35-46.

— Smith B, Köhler J, Kumar A: On the application of formal principles to life science data: A case study in the Gene Ontology. *DILS 2004: Data Integration in the Life Sciences*. 2004; 124-139.

NOTE 1 Contrasts the FMA's sound ontological structure with shortcomings of the Gene Ontology.

— Zhang S, Bodenreider O. Law and order: Assessing and enforcing compliance with ontological modeling principles. *Computers in Biology and Medicine* 2005: To appear

NOTE 2 Evaluates the FMA as a case study and finds it compliant with 10 ontological principles.

A.7 Uses and selected applications of the FMA

A.7.1 Research in ontologies, informatics and computer science

— Mejino, J.L. and Rosse, C. 1998. The Potential of the Digital Anatomist Foundational Model for assuring consistency in UMLS sources. In Chute EG (ed): A paradigm shift in health care information systems: clinical infrastructures for the 21st century. JAMIA Symposium Supplement. '98:825-829

— Agoncillo, A., Mejino JLV, 1999. Rosse C. Influence of the Digital Anatomist Foundational Model on Traditional Representations of Anatomical Concepts. J. Am. Med. Assoc. AMIA '99 Symp. Suppl. '99: 2-6.

— Noy NF, Mejino JLV Jr., Musen MA, Rosse C. 2004. Pushing the envelope: challenges in frame-based representation of human anatomy. Data & Knowledge Engineering 2004;48:335-359.

NOTE 1 Challenges presented by the FMA for an ontology authoring program and the solution of these challenges through Protégé.

— Peter Mork, Philip A. Bernstein: Adapting a Generic Match Algorithm to Align Ontologies of Human Anatomy. ICDE 2004: 787-790

NOTE 2 The FMA serves as a test-bed for developing a generic model matching algorithm.

— Songmao Zhang, Peter Mork, Olivier Bodenreider: Lessons learned from aligning two representations of anatomy. KR-MED 2004: 102-108

NOTE 3 A case study in ontology alignment using the anatomy component of GALEN and the FMA

— Songmao Zhang, Olivier Bodenreider: Investigating Implicit Knowledge in Ontologies with Application to the Anatomical Domain. Pacific Symposium on Biocomputing 2004: 250-261

NOTE 4 A case study of the FMA.

— Rickard, K. L. and Mejino, J. L. V. and Martin, R. F. and Agoncillo, A. V. and Rosse, C. (2004) Problems and Solutions with Integrating Terminologies into Evolving Knowledge Bases. In Proceedings, MedInfo 2004, pages 420-424, San Francisco, CA.

NOTE 5 Conceptual and technical challenges in integrating Terminologia Anatomica, a hard copy terminology with the FMA.

— Jakobovits RM, Rosse C, Brinkley JF. 2002. WIRM: An Open Source Toolkit for Building Biomedical Web Applications. J. Am. Med. Informatics Assoc;9:557-570.

A.7.2 Research in biomedical imaging

— Brinkley, J.F. and Rosse, C. 1997. The Digital Anatomist distributed framework and its applications to knowledge-based medical imaging. J. Am. Med. Informatics Assoc.4:165-183.

— Brinkley JF, Rosse C. 2002. Imaging informatics and the Human Brain Project: the role of structure. Year Book of Medical Informatics. Haux R, Kulikowski C. editors: Year Book of Medical Informatics '02. Schattauer and the International Medical Informatics Association. Stutgart. 131-148.

— Brinkley JF, Rosse C. 2002. Imaging and the Human Brain Project: a review. Methods of Information in Medicine 41:245-260.

A.7.3 Design of information systems

— Brinkley, J.F., Bradley, S.W., Sundsten, J. W. and Rosse, C. 1997. The Digital Anatomist Information System and Its Use in the Generation and Delivery of Web-Based Anatomy Atlases. Computers in Biomed. Res. 30:472-503.

— Brinkley, J.F., Wong, B.A., Hinshaw, K.P. and Rosse, C. 1999 Design of an Anatomy Information System IEEE 19:3 pp 38-48.

— Brinkley, J.F. and Rosse, C. 1998. Requirements for an on-line knowledge-based anatomy information system. In Chute EG (ed): A paradigm shift in health care information systems: clinical infrastructures for the 21st century. JAMIA Symposium Supplement. '98:892-896

A.7.4 Clinical informatics

— Kalet, I.J., Wu, J., Lease, M., Austin-Seymour, M.M., Brinkley, J.F., and Rosse, C. 1999 Anatomical information in radiation treatment planning. J. Am. Med. Assoc. AMIA '99 Symp. Suppl. '99: 291-295.

— Jakobovits R, Brinkley JF, Rosse C, Weinberger E. 2001. Enabling clinicians, researchers and educators to build custom web-based biomedical information systems. JAMIA Symposium Supplement. '01:279-283

A.7.5 Education

— The Digital Anatomist Interactive Atlases: http://sig.biostr.washington.edu/projects/da/

NOTE The Digital Anatomist Interactive Atlases integrate the FMA with 3D graphical models and other images of anatomy enabling knowledge-based navigation and interactivity. The atlases are used in 95 countries and experience an average of 20,000 hits per day.

— Wong, B.A., Rosse, C., and Brinkley, J.F. Semi-automatic Scene Generation using the Digital Anatomist Foundational Model. J. Am. Med. Assoc. AMIA '99 Symp. Suppl. 1999: 637-641

— Campbell B, Rosse C, Brinkley JF. 2001. The virtual anatomy lab: a hands-on anatomy learning environment. In Westwood JD , Miller Hoffman H, Mogel JT, Stredney D, Robb RA. (editors) Medicine meets virtual reality 2001. IOS Press Amsterdam. pp 85-87.

A.7.6 Sources of anatomical information

— Hollinshead WH. Anatomy for surgeons, vols. 1–3. 3rd ed. Philadelphia: Harper and Row; 1982.

— Rosse C, Gaddum-Rosse P. In: Hollinshead's textbook of anatomy. 5th ed. Philadelphia: Lippincott-Raven; 1997. p. 902.

— Williams PL, Bannister LH, Berry MM, Collins P, Dyson M, Dussec JE, Ferguson MWJ. In: Gray_s anatomy. 38th ed. New York: Churchill Livingstone; 1995. p. 2092

— Rector AL, Gangemi A, Galeazzi E, Glowinski A J, Rossi-Mori A. The GALEN CORE model schemata for anatomy: towards a re-usable application-independent model of medical concepts. Proceedings of Medical Informatics in Europe MIE 94, pp 229-233, 1994.

— GALEN-IN-USE: http://www.opengalen.org/download/specifications/irconfig.rtf

— Federative Committee on Anatomical Terminology (FCAT). Terminologia Anatomica. Stuttgart: Thieme, 1998

— Rosse C, Terminologia Anatomica; Considered from the Perspective of Next-Generation Knowledge Sources. 2001. Clin. Anat. 14:120-133.

Bibliography

[1] ISO 704, *Terminology work — Principles and methods*

[2] ISO 1087-1:2000, *Terminology work — Vocabulary — Part 1: Theory and application*

[3] ISO 2788[1)], *Documentation — Guidelines for the establishment and development of monolingual thesauri*

[4] ISO 10241 (all parts), *International terminology standards — Preparation and layout*

[5] ISO 15225:2000/Amd 1:2004[2)], *Nomenclature — Specification for a nomenclature system for medical devices for the purpose of regulatory data exchange*

[6] ISO 17115, *Health informatics — Vocabulary for terminological systems*

[7] ISO 18104, *Health informatics — Categorial structures for representation of nursing diagnoses and nursing actions in terminological systems*

[8] ISO/IEC 2382-4[3)], *Information technology — Vocabulary — Part 4: Organization of data*

[9] ISO/IEC/TR 9789:1994, *Information technology — Guidelines for the organization and representation of data elements for data interchange — Coding methods and principles*

[10] EN 1614:2006, *Health informatics — Representation of dedicated kinds of property in laboratory medicine*

[11] EN 1828:2002, *Health informatics — Categorial structure for classifications and coding systems of surgical procedures*

[12] ENV 12611:1997, *Medical informatics — Categorial structure for system of concepts — medical devices*

[13] EN 12264:2005, *Health informatics — Categorial structures for systems of concepts*

[14] ENV 13940:2001, *Health informatics — System of concepts to support continuity of care*

[15] Federative Committee on Anatomical Terminology (FCAT). Terminologia Anatomica. Thieme, Stuttgart, 1998

[16] Martin R.F., Mejino J.L.V., Bowden D.M., Brinkley J.F., Rosse C. Foundational model of neuroanatomy: its implications for the Human Brain Project. Proc. AMIA Symp. 2001, ••• pp. 438–442

[17] UWDA; University of Washington Digital Anatomy, A reference ontology for biomedical informatics.

[18] The Foundational Model of Anatomy (FMA). http://fme.biostr.Washington.edu:8089/FME/index.htm

[19] Hollinshead W.H. ed. Anatomy for surgeons. Harper and Row, Philadelphia, **Vol. 1–3**, 1982

[20] Rosse C., & Gaddum-Rosse P. Hollinshead_s textbook of anatomy. Lippincott-Raven, Philadelphia, Fifth Edition, 1997, pp. 902.

1) Withdrawn.

2) Withdrawn.

3) Withdrawn.

[21] Williams P.L., Bannister L.H., Berry M.M., Collins P., Dyson M., Dussec J.E. Gray_s anatomy. Churchill Livingstone, New York, Thirty eighth Edition, 1995, pp. 2092.

[22] Martin R.F., & Bowden D. Primate brain maps. Elsevier, Oxford, 2000

[23] Agoncillo A., Mejino J.L.V., Rosse C. Influence of the Digital Anatomist Foundational model on traditional representations of anatomical concepts. Proc. AMIA Symp. 1999, ••• pp. 2–6

[24] Mejino J.L.V., Agoncillo A.V., Rickard K.L., Rosse C. Representing complexity in part-whole relationships within the Foundational Model of Anatomy. Proc. AMIA Symp. 2003, ••• pp. 450–454

[25] Neal P.J., Shapiro L.G., Rosse C. The Digital Anatomist spatial abstraction: a scheme for the spatial description of anatomical entities. Proc. AMIA Symp. 1998, ••• pp. 423–427

[26] Rosse C., & Mejino J.L.V. Journal of Biomedical Informatics 36 (2003) 478–500 499.

[27] Smith B., Ceusters W., Klagges B., Köhler J., Kumar A., Lomax J. Rector Al, Rosse C, Relations in biomedical ontologies. Genome Biol. 2005, **6** p. R46

[28] Bodenreider O., & Burgun A. Characterizing the definitions of anatomical concepts in WorldNet and specialized sources, Proc First Global WorldNet Conf 2002:223–30.

[29] Rosse C., & Mejino J.L.V. J. Biomed. Inform. 2003, **36** pp. 478–500

[30] Rector A.L., Gangemi A., Galeazzi E., Glowinski A.J., Rossi-Mori A. The GALEN CORE model schemata for anatomy: towards a re-usable application-independent model of medical concepts. Proceedings of Medical Informatics in Europe MIE. 1994, **94** pp. 229–233

[31] GALEN-IN-USE. http://www.opengalen.org/download/specifications/irconfig.rtf

ICS 35.240.80

Price based on 17 pages