Conceptualization of Anatomical Spatial Entities in the Digital Anatomist Foundational Model

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ABSTRACT

Anatomical spatial concepts are indispensable in educational and clinical discourse, yet a system for representing these concepts has not been proposed. Guided by explicit principles and definitions of the Digital Anatomist Foundational Model, we developed an ontology of spaces, surfaces, lines and points that are associated with anatomical structures. Ontologies for Anatomical Structure and Anatomical Spatial Entity were instantiated for the thorax, abdomen, pelvis and perineum. Representing the concepts in -part of-hierarchies as well, provided formative evaluation of the classification. We invite empirical evaluation of the Foundational Model through its use for educational and clinical applications.

INTRODUCTION

Anatomical structures (material objects that constitute the human body) are extensively represented in current clinical terminology projects¹⁻³. However, the representation of spaces and other spatial entities, such as surfaces, edges, lines and points, is much less comprehensive and consistent in these computerbased knowledge sources. Spaces that exist within or between anatomical structures are an important clinical concept domain since they are frequently the location of disease processes that must be distinguished from those that affect anatomical structures. For instance, the radiological appearance of bronchopneumonia in the anterior segment may be similar to that of an interlobar pleural effusion in the horizontal fissure. However, different etiological factors and pathological processes must be considered, depending on whether the radiological opacity is assigned to a 'bronchopulmonary segment' (anatomical structure) or to a subdivision of 'pleural cavity' (anatomical space). This assignment will also influence diagnostic and therapeutic decisions. Therefore, if an anatomy model is to serve as a resource for clinical concept representation, the model must make clear distinctions between anatomical structures and spaces.

An anatomy model must also represent spatial concepts of lower than three dimensions. Surfaces, lines and points that are associated with the body and its component parts are important in anatomical dis-

course both for learning anatomy and for describing the location of clinical findings. Moreover, emerging knowledge-based programs that allow interactive 3-D manipulation of graphical models of human anatomy need to make use of spatial relationships that are specified in terms of object surfaces, edges and points⁴.

Guided by a set of principles, we have formulated a Foundational Model of anatomy, which represents the physical entities that make up the human body and specifies taxonomic and physical relationships that exist among anatomical concepts⁵. The backbone of the Foundational Model is the Anatomy Ontology (Ao), which makes explicit the distinctions between three major classes of concepts: anatomical structures, anatomical spatial entities, and body substances⁶. In a previous report, we have illustrated that the Digital Anatomist Foundational Model can explain inconsistencies found in the representation of anatomical structures by source vocabularies of UMLS, as well as other clinical terminology projects and traditional hard copy sources⁷. To date, we have populated the model with over 25,000 concepts, which suggests that the principles and definitions on which the Foundational Model is based provide reliable support for a consistent and comprehensive representation of anatomical concepts, from the level of tissues to body parts and ultimately the entire human body. In this report we discuss the classification of anatomical spatial entities, supported by definitions, and illustrate their relationships to one another and to anatomical structures. Our objectives are to fill a substantial gap in anatomical knowledge representation and to bring to the attention of clinical knowledge modelers the Digital Anatomist Foundational Model as a resource for clinical concept representation.

METHOD OF APPROACH

We established the Digital Anatomist Foundational Model by enhancing UMLS semantic types and relationships as a starting point, and used the 'thorax' as a proof of concept domain of anatomical information⁶. To date, we have extended instantiation of the Model to the 'abdomen', 'pelvis', and 'perineum', and work is in progress on the remaining body parts. Based on

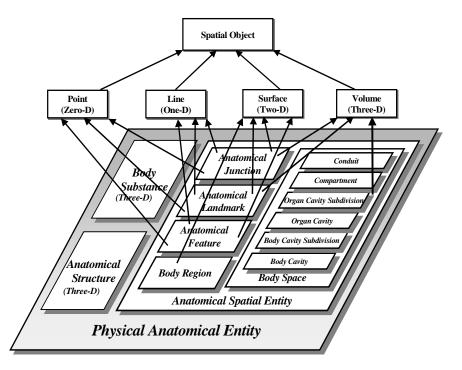


Figure 1 illustrates the *-is a-* relationships of the three major classes of the *Anatomy Ontology* (**Ao**). Subclasses of *Anatomical Spatial Entity* and *Body Space* are correlated with the major classes of the *Spatial Ontology* (**So**).

the -is a- relationship, we proposed a hierarchy of semantic types (subclasses) as children of Anatomical Structure and considered the spaces (3-D), surfaces (2-D), lines or edges (1-D), and points (0-D) that are associated with various types of structures. Paralleling the Anatomical Structure ontology, we also developed an inheritance hierarchy for Anatomical Spatial Entity. We specified in narrative text definitions the generalia and differentia on the basis of which anatomical spatial concepts could be grouped together or distinguished from one another in the subclasses of this ontology. We evaluated the first iteration of the Anatomical Spatial Entity ontology by populating its subclasses with instances from the 'thorax'. These first iterations were reviewed and discussed in the context of a graduate course (Knowledge Representation in Anatomy, CS590 BR), and the implementations were revised in a cyclic manner as a result of the discussions. In parallel with establishing the ontologies, we also generated a -part of- hierarchy for both Anatomical Structure and Anatomical Spatial Entity. These -part of- hierarchies have provided an additional validity check on class assignments in the ontologies, calling from time to time for the modification of subclass definitions and assignments.

We are currently pursuing the representation of spatial relationships between the concepts we have entered in **Ao** and the *-part of-* hierarchies⁸. The description of these complex relationships is facilitated by the classi-

fication of anatomical structures and anatomical spatial entities according to their spatial dimensions and their shapes. These parameters are captured by the *Spatial Ontology* (So), distinct from Ao⁸. Mapping the classes of Ao to So (Figure 1) provides for significant economy, as well as consistency, in the description of spatial relationships.

A Java-based authoring program (Model Builder) supports data entry in the Foundational Model (Figure 2). The current version of Model Builder is link- rather than object-centered, and directed acyclic graphs based on one specific relationship are displayed separately. An object-centered version of Model Builder capable of displaying multiple relationships is un-

der development. The formalized description of these representations in description logic is being explored through Protégé⁹.

ANATOMICAL SPATIAL ENTITY ONTOLOGY

Anatomical Spatial Entity is defined as a physical anatomical entity of 3 or fewer dimensions, which is associated with the exterior or interior of anatomical structures. This definition illustrates that the dominant class of **Ao** is Anatomical Structure⁶, although in Figure 1, Anatomical Spatial Entity occupies the larger area. Body Substance, the third major class of the Anatomy Ontology, is defined as a physical anatomical entity and a substance, which is produced or processed by anatomical structures and is contained in a variety of body spaces. The subclass Body Space is one of the children of Anatomical Spatial Entity; it maps to Volume, the 3-D object class of the Spatial Ontology. Siblings of Body Space (to be defined below) map to 0-D, 1-D, 2-D, as well as 3-D classes of the Spatial Ontology (Fig. 1).

Body Space. In a clinical context, this is the most important subclass. *Body Space* is a 3-D *Anatomical Spatial Entity* that is generated by morphogenetic processes and is enclosed by anatomical structures. The classification of body spaces is largely determined by the subclasses of anatomical structures with which they are associated. The organizational unit principle

of the Foundational Model designates *Organ* as the unit of macroscopic anatomy. Therefore, the principal subclasses of *Anatomical Structure* are *Organ*, *Organ Part* (which constitute organs), *Body Part* and *Organ System* (both of which are constituted by organs).

Organ Cavity is defined as a body space that is enclosed by the maximal set of subdivisions of one organ and contains one or more body substances. This definition enforces specificity and calls for the evaluation of the traditional use of some anatomical terms. For instance, there is general agreement that the 'cervix of uterus' is part of the 'uterus'. Yet the 'cervical canal' is not regarded as part of the 'uterine cavity'. Conventional usage limits the term 'uterine cavity' to the cavity enclosed by the 'body of uterus' and excludes from it the cavity enclosed by the 'cervix of uterus'. Consistent with the principles and definitions of the Foundational Model, we therefore propose to represent 'cavity of uterus' as Organ Cavity, and both 'cavity of body of uterus' and 'cervical canal' as Organ Cavity Subdivision, because each of these latter two spaces is enclosed by fewer than the maximal set of organ parts of the 'uterus'. Associating the term 'uterine cavity' as a synonym with 'cavity of body of uterus', rather than with 'cavity of uterus'

assures that a search for the term 'uterine cavity' will lead the user to the concept that the term has traditionally designated. The *-part of-* hierarchy for the 'uterus', displayed in Figure 2 by Model Builder, makes the relationship of all these concepts clear and explicit. Acceptance of this representation by anatomy teachers, students, clinicians and knowledge modelers is ultimately required for the validation of the foundational model of the 'uterus'.

Other subclasses of *Body Space* shown in Fig. 1 are distinguished from *Organ Cavity* and *Organ Cavity Subdivision* by their defining attributes: they are enclosed by the subdivisions of two or more, rather than the same organ; instead of body substances, they contain such anatomical structures as diverse organs or subdivisions of more than one organ.

Body Cavity is an instance, rather than a subclass, of Body Space. We assign a specific meaning to this term and define it as a Body Space that is embryologically derived from the intraembryonic celom, is located in the trunk, is enclosed by the Body Wall (which consists of several organs), and contains serous sacs, viscera and other organs. In terms of this definition there is only one Body Cavity. The 'thoracic

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Figure 2. Screen capture from the Java-based Model Builder, showing the -part of- hierarchy for the uterus.

cavity', 'abdominopelvic cavity', 'abdominal cavity' and 'pelvic cavity' are instances of Body Cavity Subdivision. They all satisfy one subclass (semantic type) definition: a body space that is part of the body cavity, is enclosed by a body wall subdivision (e.g., thoracic wall) and is demarcated from another body cavity subdivision by an anatomical structure (e.g., diaphragm) or a conduit (e.g., pelvic inlet). It contains one or more serous sacs, viscera and other organs, and together with other body cavity subdivisions, constitutes the Body Cavity. The 'cranial cavity' and the 'vertebral canal' (regarded by some anatomy texts as the 'dorsal body cavity') meet the definition of Compartment, as do other body spaces such as the 'mediastinum', 'intercostal space', 'ischiorectal fossa' and 'anterior compartment of forearm'. Compartment is a body space that is surrounded by subdivisions of two or more organs (other than those of the body wall), and contains two or more organs or organ subdivisions that are members of diverse organ subclasses. Conduit shares some of these defining attributes, but can be distinguished from Compartment in that it connects two or more body spaces with one another and may contain one or more body substances or subdivisions of diverse organs. 'Inguinal canal', 'carpal tunnel', 'pharyngotympanic tube', 'superior thoracic aperture' (thoracic inlet), 'intervertebral foramen (canal)' and 'foramen magnum' are some instances of this subclass.

Body Region. We restrict this concept to 2-D anatomical spatial entities (Fig.1) that refer to surface areas of the body, such as 'precordium', 'epigastrium' and 'right upper abdominal quadrant (region)'. The term 'body region' continues to be used in anatomical literature interchangeably with body parts such as 'abdomen', 'upper limb' and 'head' 10. Another ambiguity arises through using the same anatomical term to designate a body region and a compartment. For instance, depending on the context, 'palm of hand' may refer to part of the 'surface of the hand' (a 2-D concept which is a body region), or to a Compartment which is a 3-D concept and contains such anatomical structures as the 'palmar aponeurosis', lumbrical muscles, and superficial and deep palmar arches. We disallow homonyms and distinguish between these two concepts by adding to the term the extension (region) or (compartment) as appropriate.

Anatomical Feature and Landmark. The boundaries of anatomical structures and spaces are formed by surfaces (e.g., posterior surface of stomach is posterior boundary of stomach and anterior boundary of lesser sac of peritoneum), which in turn are bounded by lines (e.g., posterior surface of stomach by greater curvature and lesser curvature of stomach), and the lines start and terminate at points, most of which have

anatomical names. These concepts are widely used for describing anatomical structures and they are rather difficult to classify. We have grouped them together in the subclass *Anatomical Feature*, which we define as an anatomical spatial entity of two or fewer dimensions, which is a modulation of the external or internal surfaces of body parts, organs, and organ parts. For the time being we have related anatomical features to the appropriate anatomical structures by the *part of-* link, but this representation will be modified as the *Topology Network* of the *Anatomical Structural Abstraction* becomes implemented in the Foundational Model⁸.

Many of these anatomical features serve as nodes of a qualitative coordinate system in the body utilized for the physical exam and various diagnostic and invasive procedures, as well as for the measurement of body parts, organs, organ parts and the spaces associated with them. This coordinate system is augmented by arbitrary planes, lines and points (sagittal plane, midclavicular line, McBurney's point). We have grouped together these concepts in the subclass Anatomical Landmark (Fig. 1), which we define as a body location that is an organ part or anatomical feature, visible or palpable on an exterior or interior surface of the body, or a line or plane that may be defined with reference to such visible or palpable organ parts or anatomical features. In Ao, instances of Anatomical Landmark are usually also assigned to other subclasses, whereas in So they may be mapped to subclasses of Real or Virtual Point, Real or Virtual Line, Real or Virtual Surface⁸. For instance, 'lesser curvature of stomach' maps to Real Line, whereas 'midclavicular line' maps to Virtual Line because, unlike the curvature, the 'midclavicular line' is not a visible or palpable anatomical entity.

Anatomical Junction. In this subclass we have grouped together the various kinds of continuities and junctions through which the physical integrity of the body as a structured object is assured. Anatomical Junction is an anatomical spatial entity of zero to three dimensions where two or more anatomical structures, body spaces, surfaces or lines meet and establish physical continuity with one another or with the body's exterior, or intermingle their organ components. Junctions of body spaces like orifices (ostium of left coronary artery, external cervical os), branching points of nerves, blood vessels and ducts, and junctions of anatomical structures, like plexuses of nerves (e.g., brachial plexus) and vessels, raphès, and decussations, all satisfy the constraints of this definition. The specificity and description of various subclasses of Anatomical Junction in the Ao are enhanced by mapping them to 0-D to 3-D object classes of **So**.

CONCLUSIONS AND DISCUSSION

Many anatomical terms that denote spatial concepts about the body have never been explicitly defined, related to one another, or adequately represented. Confusion exists, for instance, about the relationship of anatomical structures and spaces^{2-3, 10}, as well as potential and real spaces^{2, 11-12}. A comprehensive and consistent systematization of anatomical spatial entities is lacking from available knowledge sources; yet these concepts are clinically and educationally important. In this paper, we present an ontology of anatomical spatial concepts, which is based on an ontology of anatomical structures. These two ontologies are component graphs of Ao; the subclasses of each can be mapped to So, which classifies anatomical concepts in terms of their dimension and shape. These three interacting ontologies are integrated in the Digital Anatomist Foundational Model⁵, which is intended to provide a consistent and comprehensive description of the physical organization of the human body.

The ontology of anatomical spatial entities organizes anatomical spaces, surfaces, lines and points according to attributes that they manifest in relation to the hierarchy of subclasses of Anatomical Structure. We have also represented the -part of- and -has part- relationships of these concepts comprehensively and transitively in directed acyclic graphs. Guided by explicit principles and definitions, we have instantiated over 4500 spatial concepts for the foundational models of the 'thorax', 'abdomen', 'pelvis' and 'perineum'. This volume of data entry, coupled with the parallel representation of -is a- and -part of- relationships, provide the first measure of validity for a logical and consistent conceptualization of anatomical spatial concepts. These representations await empirical evaluation as integral components of the Digital Anatomist Foundational Model. Our purpose is to make this knowledge available source for the development of educational and clinical applications, and thereby involve knowledge modelers in the evaluation of the Model. These evaluations, and their influence on the Model, should lead to the establishment of standards in anatomical concept representation.

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