Representing Complexity in Part-Whole Relationships within the Foundational Model of Anatomy

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ABSTRACT

The Foundational Model of Anatomy (FMA) is a frame-based ontology that represents declarative knowledge about the structural organization of the human body. Part-whole relationships play a particularly important role in this representation. In order to assure that knowledge-based applications relying on the FMA as a resource can reason about anatomy, we have modified and enhanced currently available schemes of meronymic relationships. We have introduced and defined distinct partitions for decomposing anatomical structures and attributed the part relationships in order to eliminate ambiguity and enhance specificity in the richness of meronymic relationships within the FMA.

INTRODUCTION

In anatomical discourse, the most natural path of reasoning follows part-whole relationships. Although the exploration of part relations has been, and remains, an active area in knowledge representation and linguistic research, and many examples in the literature relate to anatomy, none of the proposed schemes is entirely satisfactory or comprehensive for describing part relationships in the human body (or for that matter, in any vertebrate or even metazoan organism). A number of investigators have addressed this problem.[1-6] A reason for the lack of a generally accepted scheme is that knowledge modelers have used multiple and conflicting contexts in which the human body, and many subdivisions of it, may be decomposed into its parts. The generalizable AI and linguistic contexts may not be sufficiently specific for anatomy[1-3], whereas considering anatomical parts and wholes on the basis of their involvement in physiological functions, diseases and medical procedures is too limiting and conflicts with other contexts.[4-6]

We are addressing this long-standing and difficult problem in the Foundational Model of Anatomy (FMA)[7,8] Our objective in this paper is to illustrate the approach we have taken to modeling complex anatomical part-whole relationships within the human body. We believe the scheme we propose accommodates various views and conventions without sacrificing consistency.

ANATOMICAL PARTS AND WHOLES

All material objects are assembled from pre-existing objects and materials, except for biological organisms (and perhaps the universe), which begin their existence as a whole and elaborate their own parts. The *de novo* genesis of such anatomical parts is regulated, in a highly conserved manner, by the coordinated expression of the organism's own structural genes. This ontogenetic process accounts for the structural complexity of highly evolved forms of Metazoa, such as the Vertebrates. These biological principles also provide the rationale for the FMA's definition of `Anatomical Structure`: ***Material physical anatomical entity which has inherent 3D shape; is generated by coordinated expression of the organism's own structural genes.*** The purpose of the Foundational Model of Anatomy is to represent declarative, canonical knowledge about the anatomical complexity that results from the ontogenesis of the human body.[8]

The first principle of modeling the FMA constrains this representation to a strictly structural context. Consequently, the majority of classes in the *Anatomy Taxonomy* (**AT**) component of the FMA are defined in terms of the structural attributes of the entities to which they refer.[8,9] On the basis of these attributes, sets of anatomical parts are related to one another by the *is-a* relationship in the **AT**. The purpose of this taxonomy is to assure the inheritance of defining attributes by progressively more specialized classes of the hierarchy as one moves away from its root.

The second principle of modeling declares three of these parts, `Organ`, `Cell` and `Biological macromolecule` as units of structural organization of the whole (i.e., the `Body`). Other classes of the **AT** subsume concepts that refer either to aggregates of these units (e.g., `Organ system`, `Anatomical set`, `Anatomical cluster`), or are parts of these units (e.g., `Body part`, `Organ part`, `Cell part`). All these classes are explicitly defined in

English, and the defining structural attributes are formally represented as template slots of metaclasses in Protégé-2000, a frame-based knowledge acquisition system, in which the FMA is implemented.[10]

Since the FMA is to represent knowledge about anatomical structure, its taxonomy also accommodates *descriptors* of the physical entities (substances, objects, spaces, surfaces, lines and points) it models. For example, terms, coordinates, relationships and other non-physical concepts that form an indispensable part of anatomical discourse, are also included in the **AT**. Consequently, the classes `Physical anatomical entity` and `Conceptual anatomical entity` are subsumed by `Anatomical entity`, which is the root of the **AT**. Since nonphysical entities also have parts, the *has-part* attribute, and its inverse, *part-of,* are introduced at the root of the **AT** and are inherited by all **AT** classes. The kind of information that has to be associated with these part relations, however, requires elaboration and specification as one descends along the inheritance hierarchy away from its root. Therefore we distinguish between generic and specific part-whole relationships.

GENERIC PART-WHOLE RELATIONS

The FMA's emphasis on structural relations requires that we introduce greater specificity into these relationships than most current structured vocabularies. Ambiguities that call for clarification include distinguishing part relations from boundary and containment relationships.

A rule of *Dimensionality Consistency* enforces the distinction between boundary and partonomy relationships in the FMA.[11] Part-whole relationships are valid only for entities of the same dimension; boundary relationships are valid for entities that differ by one in their dimensionality. Accordingly, the following are valid assertions: `Right ventricle` -*has part*- `Wall of right ventricle`, `Cavity of right ventricle` (3D anatomical entities); `Surface of heart` -*has part*- `Diaphragmatic surface of heart` (2D entities); and `Surface of heart` -*bounds*- `Heart` (2D and 3D entities, respectively).

A rule of *Containment/Part Distinction* constrains the *contains* relationship to the class `Anatomical space`, and its inverse, *contained-in,* to `Body substance` and `Anatomical structure`. Whereas all these classes inherit the generic part attribute from the metaclass of `Anatomical entity`, the *contains* attribute is introduced as a template slot only in the metaclass `Anatomical space`; the inverse *contained-in* attribute is introduced in metaclasses `Body substance` and `Anatomical structure`. Therefore, in accord with the rules of *Dimensionality Consistency* and *Containment/Part Distinction*, the following are valid assertions: `Biceps brachii` -*contained in*- `Anterior compartment of arm`; `Anterior compartment of arm` -*part of*- `Arm`; `Biceps brachii` -*part of*- `Arm`. Although this example suggests transitivity across containment and part relations, another example negates such an assumption: `Blood` –*contained in*- `Cavity of right ventricle`; `Cavity of right ventricle` –*part of*- `Right ventricle`; but `Blood` -*part of*- `Right ventricle` is an invalid assertion. Thus, in an anatomical context, keeping containment and part relations independent of one another serves the purpose of specificity and clarity.

As long as it conforms to the *Dimensionality Consistency* and *Containment/Part Distinction* rules, the simple, or "generic" part-whole relationship adequately expresses all that needs to be said about the parts of such classes of the **AT** as `Conceptual anatomical entity`, `Anatomical surface`, `Anatomical line` and `Body substance`, and with some qualifications also `Anatomical space`. However, when we address part-whole relations in the class `Anatomical structure`, specifications must be introduced in the generic part-whole relationship and these refinements must accordingly be inherited by the frames of the concepts subsumed by the class `Anatomical structure`.

SPECIFIC PART-WHOLE RELATIONS

Several attempts have been reported to refine and modify the categories of meronymic relationships originally proposed by Winston et al.,[1] but the basic taxonomy they advanced remains the foundation of all subsequent schemes. The six kinds of parts they propose rely on functional as well as structural elements, and therefore do not correspond exactly to the anatomical parts we define strictly in terms of structure. Although 'Portion/Mass' and 'Member/Collection' are modeled in the FMA, in the present context they do not merit as much attention as 'Component/Integral-Object' and 'Stuff/Object'. These two meronymic categories are distinguished from one another by the properties of separability and the specificity of their structural relations.

Component objects can be separated from one another without altering the identity of the whole or the part (like separating the handle from the main part of the cup, or wheels from the car), and they exhibit specific patterns of spatial or structural relationships to one another, which cannot be rearranged; whereas stuff objects (like porcelain in a cup or the steel in a wheel) lack both these attributes. Other authors substitute countability for separability.[4] As the section *Anatomical Parts and Wholes* implies, anatomical structures are not like cups or cars, and they are not made of parts like porcelain or steel. The task of matching **AT** classes of `Anatomical structure` with component or stuff object is far from straightforward. The representation of the partonomy of anatomical structures, at all levels of the **AT**, must distinguish parts established by gene expression from those defined by arbitrary criteria, and also designate parts that are shared or unshared by higher level structures. Moreover, all such parts can be viewed in different contexts and these views must be accommodated in a generalizable resource such as the FMA. It is these modeling challenges that have motivated us to modify and extend the meronymic categories of Winston et al., in order to express the richness of part-whole relationships that characterize anatomical structures. In order to avoid confusion in the classification we have implemented, we redefined and renamed these categories in terms of the different partitions current in anatomical and clinical discourse, and further specified these categories by attributes. Before describing partitions and attributed part relations, however, we summarize the rules we established for assuring the consistent and comprehensive representation of all parts of an anatomical structure in any meronymic relationship or partition.

Rules for Part-Whole Relationships. For any general or specific meronymic relationship to be valid for anatomical structures, it has to conform to five rules: 1. *Dimensionality Consistency*; 2. *Containment/Part Distinction*, both defined in the section on general parts; 3. *Partition Consistency*, which specifies that any given decomposition of an anatomical structure should be constrained to a single defined context; 4. *Transitivity*, specified by Winston et al.; and 5. *Completeness of Set of Parts*, which requires that any partition of an anatomical structure into its parts must account for the whole (100%) of that structure. In other words, the specific partition will not be valid unless it can account for the whole. Component and stuff objects can be regarded as two of such possible partitions, although as already intimated, they require elaboration and translation into an anatomical context.

Partitions. We define a partition as the decomposition of the entire body or any anatomical structure in a given context, meaning a particular viewpoint. Right side and left side of the heart are functional or clinical partitions, whereas subdividing the heart into cardiac chambers or into its walls and cavities are two overlapping anatomical partitions. Yet another kind of partition relates the heart itself to the cardiovascular system. We rank ordered the different contexts according to which anatomical structures can be decomposed into primary and secondary partitions. We begin with primary partitions of anatomical structures, relate them to component and stuff objects, and then cite examples of secondary partitions that overlap with the primary ones.

<u>Primary Partitions.</u> Except for `Cell`, `Cell part`, `Biological macromolecule` and `Acellular anatomical structure`, entities in all other subclasses of `Anatomical structure` are constituted, in the ultimate analysis, by cells and body substances which fill anatomical spaces enclosed within and among anatomical structures. In so far as we can sensibly say that an anatomical structure "is partly" cells, spaces and body substances, we should regard these physical anatomical entities as stuff objects. However, body substances (e.g., blood) can be separated from an anatomical structure without altering its identity. Thus the second of Winston et al.'s criteria for stuff object remains unfulfilled.

A further conflict arises in that, in accord with Winston et al.'s criteria, 'cell' must be regarded as component object, and not as stuff object. Consistent with the rule of transitivity, all parts constituted by cells should be considered as component objects, which is a generalization too broad to be useful. For example, both the lung parenchyma and a lobe of the lung are partly constituted by cells, yet parenchyma and lobe are different kinds of parts of the lung. Moreover, both the fundus of the stomach and smooth muscle in its wall are partly made up of cells, yet they are not only distinct from one another, but fundus is a different kind of part than a lobe, and the same is true for smooth muscle and parenchyma. Since the component and stuff object distinction seems to conflict with the ontogenetically self-elaborated structural organization of the body, we replaced them in the FMA with *constitutional part* and *regional part* (Figure 1). We define these parts before illustrating them with examples.

Constitutional part is a primary partition of an anatomical structure into its compositionally distinct anatomical elements. In the context of the whole, an element is any relatively simple component of which a larger, more complex anatomical structure is compounded; i.e., the partition is compositional rather than spatial.

Regional part is a primary partition that spatially subdivides an anatomical structure into sets of diverse constitutional parts that share a given location within the whole; i.e., the partition is spatial rather than compositional. Entities in all subclasses of `Anatomical structure` have both constitutional

Taxonomy	Attribute	Metaclass level
Generic part		Anatomical entity
Specific part		
Constitutional part	Shared Unshared	Anatomical structure
Regional part	Anatomical Shared Unshared Arbitrary Shared Unshared	Anatomical structure
Systemic part		Body, Body part Body part subdivision Organ system Organ system subdivision
Member/Collection		Anatomical set
Place/Area		Anatomical surface
Portion/Mass		Not yet implemented

Figure 1. Taxonomy of part-whole relationships with their attributes and the level of their introduction into the metaclass hierarchy.

and regional parts, as illustrated by the following examples:

Neuron -*has constitutional part*-Plasma membrane, Protoplasm;

Neuron-*has regional part*-Cell body, Axon, Dendrite;

Lung -*has constitutional part*- Visceral pleura, Lung parenchyma, Intrapulmonary airway, Vasculature, Neural network;

Lung -*has regional part*- Lobe of lung;

Lobe of lung -*has constitutional part*- Visceral pleura, Lung parenchyma etc.;

Lobe of lung -*has regional part*- Bronchopulmonary segment;

Hand -*has constitutional part*- Skin of hand, Skeleton of hand, Musculature of hand, Vasculature, Neural network;

Hand -*has regional part*- Digit, Hand proper.

<u>Secondary Partitions.</u> The primary constitutional and regional partitions may be viewed in a variety of ways. Such an alternative view is the subdivision of the body in accord with functional systems, which is a widely used partition both in anatomy and clinical medicine. The FMA, however, defines `Organ system` in a structural context and constrains the parts of such a system to organs and their primary regional parts. For example, in a clinical context, the `Upper Gastrointestinal system` includes the `Esophagus, Stomach` (organs) and the `Duodenum` (a regional part of the organ `Small intestine`). The inverse relationship *has-systemic-part* is inserted as a template slot in the metaclasses of the `Body, Body part, Organ system`, as well as the metaclasses of `Body part subdivision` and `Organ system subdivision` (Figure 1). For example, not only the `Trunk` (a body part) but also `Thorax, Abdomen, Pelvis` (its subdivisions) have systemic parts, as do subdivisions of an organ system, as illustrated above by the upper GI tract. When organ systems such as the cardiovascular or nervous systems extend into organs constituting their vasculature and neural networks, we discontinue the systemic designation, since these structures meet the definition of constitutional organ parts. As previous examples indicate, vasculature is a constitutional part of organs and body parts. Because of its wide usage, the FMA explicitly represents *systemic part* relations as a secondary partition and these relations can be displayed transitively in a partonomy hierarchy, as can constitutional and regional parts.

At the organ level, additional secondary partitions have been proposed by various investigators or conventions based on different morphological or developmental criteria. Many of these partitions are eponymous and we designate them collectively as *Variant Views.* For example the liver, in addition to its lobes, can also be subdivided into so called segments as well as sectors, based on the intrahepatic arborization of the hepatic arterial and venous trees, respectively. Similarly, the prostate has been partitioned in three different ways. Secondary partitions of both the liver and prostate are in current use in different surgical or anatomical contexts. Many of such variant views have been implemented in the FMA.

Attributed Part Relations. In addition to partitions, there is a need to further specify constitutional and regional parts of anatomical structures, which may be achieved by attributing these relationships. We have partially implemented two sets of inverse attributes, *anatomical/arbitrary* and *shared/unshared*, and plan to add another inverse attribute, *mandatory/optional*.

<u>Anatomical and Arbitrary Parts.</u> Constitutional parts are genetically determined, whereas regional parts are defined not only by genetically regulated developmental processes (e.g., lobe, cardiac chamber, finger), but also by arbitrary landmarks (e.g., abdominal aorta, epigastrium). In order to represent this distinction, we associate the attributes *anatomical* or *arbitrary* with regional parts at all levels in the **AT**. For example, cell body and the cell appendage are *anatomical* regional parts, whereas the apical or basal portions of an epithelial cell are *arbitrary* regional parts. The lung provides an example with respect to organ parts: the lower lobe is an *anatomical* part, whereas the base of the lung, to which physicians listen, is an *arbitrary* part. Anatomical regional parts are demarcated from one another mainly by real boundaries (if not in the fully developed state, then during their ontogeny), whereas arbitrary regional parts are demarcated by virtual boundaries. Anatomical and arbitrary parts are

analogous with previously proposed *natural* and *fiat* elements.[2]

<u>*Shared and Unshared Parts.*</u> Although inherent 3D shape is a defining attribute of entities in the class `Anatomical structure`, the nature of continuities established between anatomical structures are such that certain parts of one structure become shared by another. The tracheobronchial tree and right and left lungs each meet the definition of `Organ`. However, since a part of the tracheobronchial tree is embedded in the right and left lungs, a distinction needs to be made between the parts of the tree that are shared and unshared. Tree organs (e.g., `Vascular tree`, `Neural tree`) and serous sacs (e.g., `Pleural sac`, `Peritoneal sac`) always share some of their parts with another organ subclass. The attributes *shared* and *unshared* can be associated with constitutional as well as with regional parts and these attributes can specify partonomic relationships at any level of the **AT**. For example, the diaphragm is a shared part of the thorax and abdomen. Likewise, the cadherin cell adhesion molecule is a shared part of the cell membrane and the cytoskeleton.

<u>*Mandatory and Optional Parts.*</u> Schulz et. al.[6] have emphasized the need for declaring whether parts are mandatory (essential) or optional (non-essential), as a requirement for supporting inference. We will associate these attributes with anatomical structures, but this information has not yet been implemented.

TAXONOMY OF PART-WHOLE RELATIONSHIPS

Figure 1 summarizes the meronymic relationships in the FMA. These relations constitute the class `Anatomical part relationship`, a subclass of `Conceptual anatomical entity`. Attributes (*anatomical/arbitrary*, *shared/unshared*) are implemented as template slots of the subclasses of `Specific anatomical part`. These attribute slots are then inherited by the instances of `Specific anatomical part` classes. These classes describe the specific type of part relationship of any given anatomical structure.

CONCLUSIONS AND DISCUSSION

Relying on some of the modeling principles of the Foundational Model of Anatomy and rules we have formulated, we have proposed a taxonomy of part-whole relationships that can capture the richness and specificity of such relationships in a symbolic model of the structural organization of the human body. These part-whole relationships and their attributes have been extensively, though not comprehensively, instantiated in the FMA.

We call this model *foundational*, because we intend it to accommodate, rather than replace, all current sensible naming conventions and views of the structural organization of the human body. To realize this goal we elaborated on the specificity and granularity of meronymic relations and translated them into an anatomical context. The purpose of the Foundational Model of Anatomy is to serve as a reference ontology in diverse fields and support the development of knowledge-based applications that require anatomical information and call for anatomical reasoning.

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