

# The Potential of the Digital Anatomist Foundational Model for Assuring Consistency in UMLS Sources

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## ABSTRACT

*Inconsistent anatomical concept representation can be identified in anatomy textbooks and hard copy term lists, as well as in UMLS source vocabularies and other controlled medical terminologies. In this report we select some examples of inconsistent representations of anatomical concepts, and illustrate how these inconsistencies can be explained and reconciled by the Digital Anatomist Foundational Model<sup>1</sup>. We use this process for gaining a measure of the validity of the logic-based Model.*

## INTRODUCTION

Anatomical concepts are extensively represented in a number of the source vocabularies of UMLS<sup>2</sup>, as well as in other clinical terminology projects<sup>3,4</sup>. The consistency of anatomical concept representation in these sources has not been formally or comprehensively evaluated. A number of published reports<sup>3-6</sup>, as well as experience with using some of the vocabularies, suggest, however, that there is inconsistency in, and confusion about, the classification of anatomical concepts. These inconsistencies may have a bearing on clinical concept representation<sup>6</sup>.

We believe that, since anatomical entities form the physical substrate of concepts in other biomedical domains (e.g., physiology, pathology, general medicine and surgery, and their subspecialties), consistency and standards in anatomical concept representation should facilitate consistency in the representation of clinical concepts. It is with such an objective in mind that we have begun the development of the Digital Anatomist Foundational Model<sup>1,5</sup>.

## INCONSISTENCIES OF CLASSIFICATION

The UMLS Semantic Network represents three Semantic Types ('*Body System*', '*Body Location or Region*', and '*Body Space or Junction*') as conceptual parts of '*Fully Formed Anatomical Structure*'; '*Body Part, Organ or Organ Component*' is a conceptual part of '*Body System*'. The subordinate node of '*Body Part,*

*Organ or Organ Component*' is '*Tissue*'. Anatomical concepts in the source vocabularies are assigned to one or more of these semantic types. There is considerable duplication of a number of anatomical concepts in the different UMLS source vocabularies. Even though the UMLS semantic types are broad, there are inconsistencies in the assignment of a particular concept to anatomical semantic types. There is even more divergent representation of some concepts within and between the vocabularies themselves.

The representation of serous membranes and serous cavities provides good examples for a need to reconcile existing differences<sup>6</sup>. Serous membranes and the spaces they enclose are clinically important concepts with respect to the location of disease processes, as well as to symptoms and signs, and diagnostic and therapeutic procedures. Serous cavities have, in fact, been chosen by a cohort of reports to illustrate approaches for clinical concept representation<sup>7</sup>.

UMLS relies on Dorland's Medical Dictionary's definition of a *Serous Membrane* (tunica serosa): "the membrane lining the external walls of the body cavities and reflected over the surfaces of protruding organs." 'Pleura', 'Pericardium' and 'Peritoneum' are all assigned through an -is a- relationship to the hypernym *Serous Membrane* and *Tissue*. However, 'Pericardium' also inherits (through the term 'Heart and Pericardium') the semantic type of '*Body Part, Organ, Organ Component*', whereas 'Pleura' and 'Peritoneum' do not. Although no direct -is a- relationships are tractable (owing to the failure of some vocabularies to specify relationships), 'Peritoneum' inherits, in addition to *Tissue*, two other semantic types: '*Body Location or Region*' and '*Body Space or Junction*'. The relationship between the serous membranes and the cavity they enclose is also inconsistent: 'Pleura' is a hypernym of 'Pleural Cavity', whereas 'Peritoneal Cavity' is a hypernym for 'Peritoneum'.

Similar discrepancies exist in vocabularies that are not included in UMLS: GALEN assigns 'Pleural cavity' to *Potential Space*<sup>3</sup>, and the READ CODES include

'Pericardium' in an ordered list as a sibling of cardiac chambers<sup>9</sup>. In some UMLS vocabularies, a further confounding element is the inclusion of such compound terms as 'Heart and Pericardium' and 'Abdomen including Peritoneum and Retroperitoneum'. Inheritance through such concepts leads to the inclusion of 'Pericardium' and 'Pleura' in the semantic type of 'Body System', whereas 'Peritoneum' or 'Peritoneal Cavity' are not assigned to a 'Body System'.

Narrative text sources are not much help in resolving these inconsistencies because they lack the semantic specificity required for logic-based knowledge representation. Therefore, we have declared a set of principles according to which a consistent classification of anatomical concepts could be developed<sup>1,5</sup>. We hypothesize that inconsistencies, such as we cite above, can be explained and reconciled by the Digital Anatomist Foundational Model, the development of which is guided by the principles we set forth<sup>1</sup>. This report is our first attempt to test this hypothesis.

#### RESOLUTION OF INCONSISTENCIES

We follow the process of making class assignments in the foundational model, when new concepts, such as serous membrane and its hyponyms, are entered. Of the four components of the foundational model (Anatomy Ontology, Anatomical Structural Abstraction, Anatomical Transformation Abstraction, and Metaknowledge)<sup>1</sup>, this process initially involves the Anatomy Ontology (Ao) and the Part-of Network of the Anatomical Structural Abstraction (described in a companion report<sup>10</sup>). First we verify that of the three top level concepts of Ao, serous membrane, satisfies the definition of *Anatomical Structure*, and not those of *Anatomical Spatial Entity* and *Body Substance*<sup>5</sup>. Assignment to one of the subclasses of *Anatomical Structure* is guided by the "Organizational unit principle"<sup>1</sup>, which calls for determining the relationship of serous membrane to *Organ*. Is it an organ; a part of an organ, or an anatomical structure constituted of organs? Answers to these questions are aided by considering instances of *Serous Membrane*, such as the 'Pleura' and the 'Peritoneum'. Neither the 'Pleura' nor the 'Peritoneum' satisfy the definition of *Organ*<sup>5</sup> in that they are not self-contained macroscopic anatomical entities. Next we apply the "Constitutive principle"<sup>1</sup> in order to decide of what organs are these membranes a part of and what parts does each membrane have. Both 'Pleura' and 'Peritoneum' are divisible into a parietal and visceral membrane; these subdivisions, in each instance, enclose between them a cavity, thereby forming a closed sac. The right and left pleural sacs and the peritoneal sac, each satisfy the definition of *Organ*<sup>5</sup>

and can, therefore, be assigned to the Ao organ subclass *Serous Sac*. It follows that *Serous Membrane* and its hyponyms 'Pleura' and 'Peritoneum' must be assigned to the Ao subclass *Organ Part*<sup>5</sup>. What kind of *Organ Part*, is solved again by applying the "Constitutive principle"<sup>1</sup>. What kind of anatomical structures constitute the 'Pleura' and the 'Peritoneum'? Ao designates *Tissue* as the smallest organ part. *Tissue* is defined as an organ part consisting of similarly specialized cells and intercellular matrix, aggregated according to specific spatial relationships<sup>5</sup>. Visceral and parietal pleura, or peritoneum, are constituted by *Mesothelium* (a type of *Epithelium*, which is a subclass of *Tissue*), supported by a membrane composed of dense, irregular *Connective tissue* (a subclass of *Tissue*). *Mesothelium* is the principal tissue of *Serous Membrane* (it is one of its defining attributes); *Connective tissue* is a subsidiary tissue of *Serous Membrane* and it is pervaded by nerves, blood vessels and lymphatics. An organ part is classified as an *Organ Component* if the anatomically most complex structures into which it is divisible are tissues. The organ part is classified as an *Organ Subdivision* if it is divisible into organ components (structures larger and more complex than a tissue).

Although these rules would be better illustrated with organs such as the heart or the liver, in Figure 1 we correlate ontologic (-is a-) and part-of relationships for the 'Left Pleural Sac', in order to illustrate the interaction of these two axes when class assignments are made in the foundational model. We have found it necessary to consider both -is a- and -part-of-relationships in parallel and represent each relationship explicitly.

Before such class assignments are sanctioned, the same process is applied to siblings of 'Left Pleural Sac' (Right Pleural Sac, Peritoneal Sac, Serous Pericardial Sac), in order to assess whether the assumptions hold true for other instances of the organ subclass *Serous Sac*.

Entering the cavity of serous sacs in the foundational model follows a similar process. To our knowledge, the Digital Anatomist Foundational Model is the first attempt to systematically and comprehensively classify spatial anatomical entities. We define *Anatomical Spatial Entity* as a spatial entity of three or fewer dimensions, which is associated with the exterior or interior of anatomical structures<sup>5</sup>. Having assigned *Serous Sac* to *Organ*, dictates that the cavity of the sac (*Serous Cavity*) be classified as *Organ Cavity*. Figure 1 correlates the ontologic and -part-of- relationships of

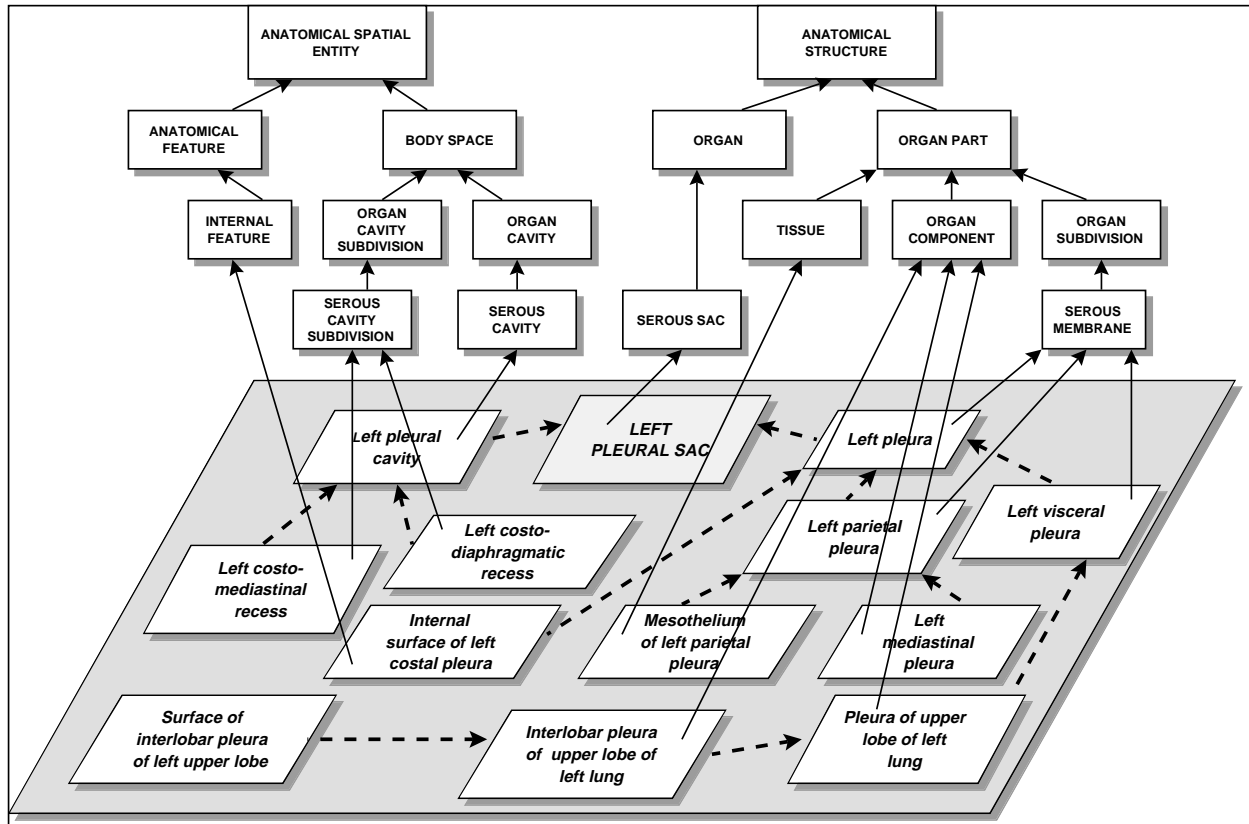


Figure 1. Ontologic (-is a-, solid lines) and meronymic (-part of-, interrupted lines) relationships for 'Left Pleural Sac'. Part-of relationships of 'Left Pleural Sac' are shown in the plane of the shaded quadrangle, whereas semantic types of the concepts are displayed above this plane.

the 'Left Pleural Cavity' with those of the 'Left Pleural Sac'.

The anatomy ontology of Digital Anatomist Foundational Model has been integrated with the UMLS Metathesaurus, and **Ao** subclasses *Organ*, *Organ Subdivision* and *Organ Component* are assigned to UMLS semantic type *Body Part*, *Organ*, *Organ Component*, and **Ao** subclasses 'Organ Cavity', 'Organ Cavity Subdivision' to UMLS semantic type *Body Space or Junction*.

Inconsistencies pertaining to the 'Pericardium' are more complex to resolve. This complexity is related to the fact that UMLS source vocabularies and other terminologies either fail to make a distinction between the 'fibrous pericardium' and the 'serous pericardial sac', or do not specify the relationship between these two concepts. Figure 2 shows the -part of-relationships of the pericardium as a hierarchy. In order to appreciate the distinction between the 'Fibrous Pericardium' and the 'Pericardial Sac', it would be necessary to represent spatial adjacency relationships

between parts of these two anatomical structures. However, until such relationships are implemented in the structural abstraction component of the Digital Anatomist Foundational Model<sup>10</sup>, the model displays only -part of- relationships as a directed acyclic graph.

The heart and the roots of the great vessels are surrounded by the 'Pericardial Sac', which is a *Serous Sac*, constituted by the 'Serous Pericardium' (a *Serous Membrane*) and the 'Pericardial Cavity' (a *Serous Cavity*). Subdivisions of the 'Visceral Serous Pericardium' are adjacent (adherent) to, and surround, the heart and the great vessels, whereas the 'Parietal Serous Pericardium' is adjacent (adherent) to the inner surface of the 'Fibrous Pericardium' and the superior surface of the diaphragm. The 'Fibrous Pericardium' surrounds the 'Pericardial Sac' everywhere, except inferiorly.

In **Ao**, the 'Fibrous Pericardium' is classified as an *Organ*, assigned to the organ subclass *Membrane(organ)*. It is not a *Serous Membrane*, since its organ parts do not include *Mesothelium*, the

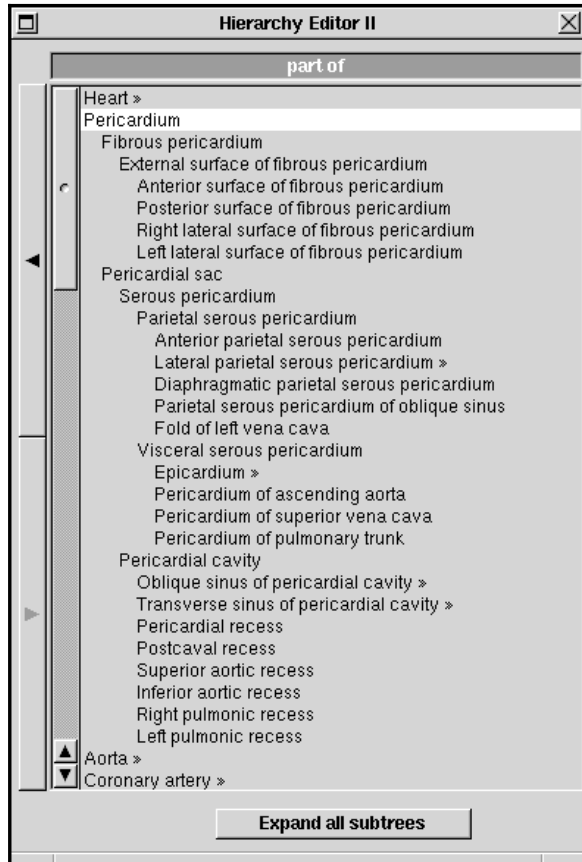


Figure 2 . Screen capture from the authoring program Knowledge Base Manager showing the -part of-hierarchy for 'Pericardium'. The symbol >> indicates that the hierarchy node has at least one generation of children that is not shown. Tab indentation indicates immediate offspring of a node.

principal tissue of *Serous Membrane*; it has an embryological origin that is distinct from the embryological derivation of the 'Pericardial Sac'.

The explanation for the inconsistencies in classifying serous membranes and serous cavities, may largely be found in the inadequacy of the definitions on which the classifications have been based<sup>8</sup>. There are serous membranes that do not line the "external walls" of the body cavities (e.g. parietal serous pericardium, synovial membrane of joints) and many cavities in the body are not lined by serous membranes (e.g. maxillary sinus, tympanic cavity). Serous membranes surround cavities into which viscera do not protrude (e.g., bursa, synovial joint), and serous membranes are associated with viscera that are suspended in, rather than protrude into a cavity (e.g. liver and spleen in abdominal cavity, heart

and lung in thoracic cavity). Were *Serous Membrane* defined according to the anatomical structures that constitute it and the structures that it in turn constitutes, *Serous Membrane* could not be assigned as a hyponym of *Tissue*. The definition of *Tissue* is concordant in the UMLS Semantic Network and the Digital Anatomist Foundational Model.

Definitions formulated according to foundational principles of a domain are a requirement for assuring clarity and consistency in classifying the domain's concepts. Digital Anatomist definitions meet this requirement. Applying them to *Serous Membrane* and *Serous Cavity* eliminates inconsistencies and ambiguities in the classification of these concepts. These definitions are: 1. *Serous Sac* is an *Organ* constituted by one or more contiguous serous membranes which enclose a *Serous Cavity*; together with other organs, a *Serous Sac* constitutes a *Body Part* or an *Organ System*; Examples: Bursa, Pleural Sac, Sac of Processus Vaginalis. 2. *Serous Membrane* is an *Organ Component* constituted by *Mesothelium* (principal tissue) and irregular connective tissue (subsidiary tissue); it constitutes the lining of synovial joints, or with other contiguous serous membranes, a *Serous Sac*; Examples: Synovial Membrane, Parietal Peritoneum, Interlobar Pleura, Epicardium. 3. *Serous Cavity* is an *Organ Cavity* enclosed by one or more contiguous serous membranes and is filled with *Serous Fluid* (a *Body Substance*); Examples: Pericardial Cavity, Cavity of Processus Vaginalis, Synovial Joint Cavity.

## DISCUSSION

We believe the Digital Anatomist Foundational Model is well suited for coherently and consistently modelling the structure of anatomical knowledge, because it 1. declares the constraints (principles) for including concepts and relationships in the model; 2. explicitly defines the concepts; and 3. makes explicit the assumptions about relationships between concepts. Perhaps the most important criterion for classifying physical anatomical entities is to determine the relation they have to *Organ*. Applying this criterion to serous membranes, such as 'Pleura', leads to the concept of *Serous Sac*. Because of the anatomical structures that constitute a *Serous Sac*, and the structures that the sac constitutes, *Serous Sac* must be assigned as a member of the subclass *Organ*.

Although the concepts *Serous Sac*, 'Pleural Sac', 'Peritoneal Sac', do appear in most standard anatomy textbooks, the emphasis tends to be on the *Serous Membrane* (Pleura, Peritoneum), rather than the sac as

such. None of the sources, however, regards any of the serous sacs as *Organ*. The explanation for this is that *Organ* has not been either explicitly or consistently defined by these sources.

Classifying *Serous Sac* as *Organ* and *Serous Membrane* as *Organ Part*, clears up not only the confusion about the relationship between a *Serous Membrane* and a *Serous Cavity*, but it also eliminates ambiguities about the relationship of instances of these membranes and cavities to more complex anatomical structures. Anatomical structures constituted of organs are *Organ System* and *Body Part*. Provided these structures are considered in terms of the Digital Anatomist definitions<sup>5</sup>, 'Pleural Sac', as an *Organ*, may be related to both the *Respiratory System* and the *Thorax*. Although these two relationships of 'Pleural sac' will be permitted by different rules, (which take into account the differential defining attributes of *Organ System* and *Body Part*), both relationships may be represented by the -part-of- link. The representation will be consistent up and down the complexity and size range of anatomical structures, from the level of 'Tissue' (e.g., Mesothelium of Pleura), to a *Body Part* (*Thorax*), or indeed, the entire human body.

### CONCLUSIONS

We have confirmed the hypothesis that inconsistencies identified in UMLS and other knowledge sources with respect to anatomical concept representation can be explained and reconciled by the Digital Anatomist Foundational Model. Using serous membranes and serous cavities as examples, we have shown that applying the principles and definitions of the model, a representation can be formulated for these concepts which is consistent and comprehensive from the level of tissues to body parts and the entire human body. We regard the outcome of our analysis as a measure for the validity of the model. We suggest, therefore, that the Digital Anatomist Foundational Model is a good candidate for adoption as a standard for anatomical concept representation. We believe, moreover, that adoption of the model by controlled medical terminology projects would rectify current inconsistencies in their anatomy sections, and would contribute to assuring consistency in the representation of clinical concepts.

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