

Terminologia Anatomica; Considered from the Perspective of
Next-Generation Knowledge Sources

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ABSTRACT

This report examines the semantic structure of *Terminologia Anatomica*, taking one randomly selected page as an example. The focus of analysis is the meaning imparted to an anatomical term by virtue of its location within the structured list. *Terminologia*'s structure expressed through hierarchies of headings, varied typographical styles, indentations and an alphanumeric code implies specific relationships between the terms embedded in the list. Together, terms and relationships can potentially capture essential elements of anatomical knowledge. The analysis focuses on these knowledge elements and evaluates the consistency and logic in their representation. Most critical of these elements are class inclusion and part-whole relationships, which are implied, rather than explicitly modeled by *Terminologia*. This limits the use of the term list to those who have some knowledge of anatomy and excludes computer programs from navigating through the terminology. Assuring consistency in the explicit representation of anatomical relationships would facilitate adoption of *Terminologia* as the anatomical standard by the various controlled medical terminology (CMT) projects. These projects are motivated by the need for computerizing the patient record, and their aim is to generate machine-understandable representations of biomedical concepts, including anatomy. Because of the lack of a consistent and explicit representation of anatomy, each of these CMTs has generated its own anatomy model. None of these models is compatible with each other, yet each is consistent with textbook descriptions of anatomy. The analysis of the semantic structure of *Terminologia Anatomica* leads to some suggestions for enhancing the term list in ways that would facilitate its adoption as the standard for anatomical knowledge representation in biomedical informatics.

Keywords: knowledge representation, structured vocabulary, controlled medical terminology, anatomical terminology, nomenclature, standards

INTRODUCTION

The publication of *Terminologia Anatomica* (Federative Committee on Anatomical Terminology, 1998) coincides with an unprecedented level of interest in anatomy. There are probably more anatomy textbooks and atlases on the market than ever before and a number of journals are dedicated primarily to anatomy. Affordable access to the personal computer, and the powerful authoring programs it supports, have promoted the creation of computer-based anatomy programs by virtually anyone who teaches anatomy. Although the majority of such programs target local student populations, many are intended eventually for national or worldwide distribution. Indeed, electronic publications of anatomy have become perhaps even more numerous and diverse than hard copy textbooks and atlases. Anatomy is also an integral component of both hard copy and electronic publications that relate to radiology, surgery and other fields of clinical medicine. In view of this widespread pursuit of anatomical information in health education and clinical research and practice, there has never been a greater need for standards in anatomical terminology than we are currently facing. *Terminologia Anatomica* has been intended by its authors to provide such standards.

This stated objective invites a critical examination of this publication. Such an examination must take into consideration other terminologies, which have recently been developed and contain large compendia of anatomical terms. Most of these terminologies are primarily clinically oriented; they are regarded as a new generation of knowledge sources for clinical medicine in the information age.

My objectives in this communication are to define *Terminologia Anatomica* and evaluate its semantic structure and semantic specificity in comparison with other terminologies. I embark on this analysis as a scholarly response to the request for suggestions that would benefit subsequent editions of *Terminologia* (Whitmore, 1999). I hope that this analysis will facilitate the evolution of *Terminologia*, so that it becomes a source of knowledge not only for anatomists but also for all who need anatomical information in the clinical and scientific worlds.

WHAT IS TERMINOLOGIA ANATOMICA?

In a broad sense, *Terminologia* is a list of terms that pertain to the anatomy of the human body. If these terms were assembled merely in alphabetical order, the list would serve only for the verification of spelling. *Terminologia*, however, has a much more complex and richer structure, which enhances the intrinsic meaning of each term. These layers of meaning are communicated by the position of a term within the structure of the list. Such a semantic organizational framework places *Terminologia* in the class of *structured vocabularies*.

The structure of a terminology implies semantic relationships between its terms. Consequently, the terms and the relationships specified by the structure of the terminology, in their aggregate, represent some knowledge. Since *Terminologia Anatomica* has a structure, it qualifies as a knowledge source.

Unlike the knowledge expressed through narrative text, the knowledge embedded in the structure of a terminology can be “understood” by computers. Once a structured vocabulary has been rendered navigable by a computer program, it is referred to, by convention, as a *controlled terminology*. The development of controlled terminologies has become a very active field in medical informatics because of the potential these machine-understandable, structured vocabularies have for representing knowledge and assuring standards in the use of biomedical terms. If clinically oriented controlled terminologies are to rely on *Terminologia Anatomica* as a standard for anatomical terms, *Terminologia* must be evaluated not only for the comprehensiveness of its terms but also for its semantic structure. The structure of the current version of *Terminologia* is as yet implicit. The objective of the following analysis is to identify the features of the semantic structure that should be made explicit.

WHAT IS THE SEMANTIC STRUCTURE OF *TERMINOLOGIA ANATOMICA*?

The overall organization of *Terminologia Anatomica* conforms to that of its predecessor, *Nomina Anatomica* (International Anatomical Nomenclature Committee, 1989). Both are divided into two main, unequal parts: a comparatively brief section on *General Anatomy* precedes *Systemic Anatomy*. The latter contains the majority of terms by far, and is subdivided into chapters, each of which is devoted to a so-called system of the body. *Terminologia* incorporates, with minor modifications, all the Latin terms of *Nomina Anatomica*. In other respects, however, *Terminologia* presents a much richer and potentially more expressive structure than its predecessor. This structure is reflected in several features:

1. A hierarchy of headings is presented through the use of different fonts and varied shading of the panels in which the headings appear. Combined with indentations in the list, these headings have the potential for representing various relationships between the terms.
2. The primary Latin terms are associated with their English equivalents in current usage, and any existing eponyms may also be linked to a Latin/English term pair with the use of a special index.
3. An alphanumeric code is associated with the Latin/English term pairs, and in many respects reflects the ranking of the headings and their subordinate terms.
4. A tabular form displays the corresponding terms and code in three columns, which together with the varied fonts of the headings and their shaded panels lends clarity to the complex term list.

It is perhaps best to examine these new features of *Terminologia* and consider what elements of semantic structure they imply. The question to be answered is how implicit elements of meaning in the structured list might be made explicit.

Systemic Organization

All anatomists have an implicit understanding of what is meant by the systemic and regional organizations of anatomy, the contrasting yet complementary approaches to

representing, teaching, learning and retrieving anatomical information. However, definitions of systemic and regional anatomy are hard to find and are difficult to formulate. Although it is generally believed that shared function provides the rationale for a systemic organization, no one would deny that the structures that constitute the upper limb or the hand, both classified as regions rather than systems, also have and share important functions. The lack of explicit, rather than implied definitions may account for some of the inconsistencies evident in the organizational plan of anatomical knowledge sources, including that of *Terminologia Anatomica*. Many of the footnotes in *Terminologia* qualify as definitions. Their intent, however, is to explain changes in the use of terms, rather than to define concepts according to which sections of the terminology are organized. Such definitions are a requirement for the logical and consistent structuring of knowledge sources. Indeed, the need for explicit definitions may be illustrated by some of the differences between *Nomina* and *Terminologia Anatomica*.

Nomina divides the body into seven systems, whereas the Systemic Anatomy section of *Terminologia* is organized into 13 systems, each of which occupies a chapter. Two additional chapters are devoted to the thoracic and abdominopelvic cavities, each of which is equivalent to one of the systems. Intuitively, the organization of *Terminologia* appears more logical. For instance, the digestive, respiratory, urinary and reproductive systems are ranked as equivalent to the skeletal and muscular systems, whereas in *Nomina* they were drawn together under 'Splanchnologia'. *Terminologia* renames the 'Angiologia' chapter of *Nomina* as 'Cardiovascular system'. This new name resolves the inconsistency in *Nomina Anatomica*, which assigned the heart to the 'system of vessels' (Angiologia), rather than to viscera (Splanchnologia). *Terminologia* eliminates splanchnology altogether with the result that the terms 'viscus' or 'viscera' do not appear at all; the adjective 'visceral' is retained, however, in composite terms.

All these changes are the outcome of consensus reached by members of the Federative Committee on Anatomical Terminology. Since they have agreed to retain a systemic organization as the framework for *Terminologia Anatomica*, it seems desirable to answer the following question: By what criteria does a group of anatomical entities qualify as a system?

Since it is taken for granted that function is the guiding principle, it may seem reasonable to split the urogenital system of *Nomina Anatomica* into separate urinary and genital systems, even though some components of each system are involved in both urinary and genital functions. However, it appears that traditionally regarded systems may not only be split but can also be merged. For instance, on what basis do well established textbooks of anatomy (Williams et al., 1995; Rosse and Gaddum-Rosse, 1997, Moore and Dalley, 1999) include both bones and joints in the skeletal system, whereas both *Nomina* and *Terminologia* represent bones and joints as two separate systems? Distinct functions can readily be specified for bones and joints, yet by themselves neither bones nor joints can be regarded as a system in a functional sense.

The practical benefit of explicitly defining a 'system' in a knowledge source, which is organized according to systems, is that the definition can provide the logical basis for consistently assigning to the appropriate system those anatomical entities which share a

set of inherent properties. Some of the inconsistencies in *Terminologia* may be ascribed to the lack of such definitions, which may be illustrated by the following examples.

Perineal muscles are included in the muscular system (p. 40), and so is the diaphragm. Perineal muscles, however, are also included in the genital system (p. 71), presumably because of their role in genital functions. Yet the function played by the diaphragm in respiration does not qualify it for inclusion in the respiratory system. On the other hand, the ischio-anal fossa and its fat pad (p. 71) take no part in genital functions, yet they are included in the genital system, and in no other system of the terminology.

Another example is the representation of the pleura and pericardium, and the serous cavities associated with them. The pleural cavity, pleura and pericardial cavity are all assigned to the chapter 'Thoracic cavity' (p. 62). The sinuses of the pericardial cavity, as well as the fibrous and serous pericardium (with its parietal and visceral layers), however, are listed under the heart (actually, under the left ventricle, p. 77). Furthermore, they are assigned the same code category as parts of the heart, and are not coded in the thoracic cavity category. On the other hand, the pleura receives coding only under the thoracic cavity and is neither entered nor coded in the section concerned with the lung. The anatomical and functional similarities that exist between the pleura and lung on the one hand, and between the serous pericardium and the heart on the other, argue for representing the relationship between these serous membranes and the viscera in a corresponding manner. *Terminologia* implies a close relationship between the pericardium and the heart by assigning them to the same system. However, *Terminologia* disclaims a similar relationship between the pleura and the lung by assigning them to different systems or chapters.

The kind of inconsistencies and ambiguities illustrated by these examples would be prevented if the principles were explicitly declared which had guided the construction of the terminology. The User Guide states as an introduction to *Terminologia* that "The order of terms follows the anatomy naturally through each system". The cited examples suggest that this principle is not sufficiently stringent to assure consistency in, and a logical semantic structure for the terminology. The problem is that functional processes in which anatomical entities participate do not seem to provide an adequate basis for the logical and consistent classification of the physical objects and spaces of which the body is constituted. This assertion does not negate the importance of functional anatomy. On the contrary, in a knowledge source, multiple functions can and should be associated with any anatomical entity, regardless of the rationale or principles according to which its anatomical terms are classified. Provided the underlying semantic structure of the terminology is sound, that structure can greatly facilitate conceptualizing functional anatomy.

One of the objectives of structured vocabularies and controlled terminologies should be consistency in term assignment within the classification scheme of the terminology. This scheme should group together entities according to their inherent properties, which they share with one another and according to which they may be distinguished from one another. Such a knowledge source will have the potential of supporting reasoning by humans and inference by computer programs.

Hierarchy of Headings

At least nine hierarchical levels or orders of terms may be distinguished in *Terminologia Anatomica*. These organizational levels in the term list are represented through the typographical style of the headings and, at the lowest level, by indentation of a term within the list. The hierarchical interrelations of these different headings become evident at a glance on page 30, for instance, where all but two of the highest order headings appear together (Figure 1).

The meaning of a term is influenced by this complex semantic neighborhood. In other words, a considerable amount of knowledge may be communicated about an anatomical entity by the location of the term that represents that entity within the structured list. For instance, in Figure 1, the term 'Knee joint' identifies a particular anatomical entity. However, it is the location of this term within the list that communicates important elements of knowledge about it.

Before demonstrating these knowledge elements, we should perhaps briefly consider what these elements are. Knowledge may be expressed through stating relationships between entities. In anatomy, there are two major kinds of entities: 1. physical objects of varying size and structural complexity which constitute the body (these may be referred to as anatomical structures); and 2. spatial entities of three or fewer dimensions (spaces, surfaces, lines and points), which are spatially associated with anatomical structures. Relationships that exist between anatomical entities may be conceptualized as three major classes: 1. class inclusion, or taxonomic relationships, which can be expressed as -is a- (e.g., 'Knee joint' -is a- 'Synovial joint of free lower limb'; Fig.1); 2. part-whole relationships, which can be expressed as -part of-, or -has parts- (e.g., 'Knee joint' -has parts- 'Lateral meniscus', 'Medial meniscus'; Fig. 1); 3. spatial association relationships, which describe the location, orientation and adjacencies of anatomical entities in relation to one another, including the body as a whole.

Narrative texts, such as textbooks and journal articles use natural language for describing these entities and relationships. Relationships are often implied rather than explicitly stated because the text usually embeds these entities in one or more contexts, and this reduces the ambiguity of implied relationships. In a structured list, the hierarchy of the headings provides context. Such hierarchies may be expressed in terms of a numerical code, or the code may parallel and enhance the hierarchy expressed through various typographical styles, as is the case in *Terminologia*. In such a structured list the following semantic relationships may be represented: 1. a term may be a *hypernym* (or parent) of another term or terms; 2. a term may be a *hyponym* (or child) of another term; 3. a term may be of equivalent rank with (or sibling of) another term or terms; and 4. a term may be equivalent or *synonymous* with (twin of) another term or terms. The richer the structure of this hierarchy, the greater is the potential for expressing relationships more comprehensively and specifically and thereby representing deeper and more precise knowledge.

Figure 1. A page of *Terminologia Anatomica* selected to illustrate the hierarchical organization of terms through varied typographical styles. Only the two highest hierarchical levels are not illustrated: the name of the major section of *Terminologia* (Systemic Anatomy), and the name of the system (Articular System).

| 30 Anatomia systemica / Systemic anatomy | | |
|--|---|--|
| A03.5.11.405 | Spatia interossea metacarpi | Interosseous metacarpal spaces |
| A03.5.11.501 | Articulationes metacarpophalangeae | Metacarpophalangeal joints |
| A03.5.11.502 | Ligg. collateralia | Collateral ligaments |
| A03.5.11.503 | Ligg. palmaria | Palmar ligaments |
| A03.5.11.504 | Lig. metacarpale transversum profundum | Deep transverse metacarpal ligament |
| A03.5.11.601 | Articulationes interphalangeae manus | Interphalangeal joints of hand |
| A03.5.11.602 | Ligg. collateralia | Collateral ligaments |
| A03.5.11.603 | Ligg. palmaria | Palmar ligaments |
| A03.6.00.001 Juncturae membri inferioris joints of lower limb | | |
| A03.4.00.001 | JUNCTURAE CINGULI PELVICI | JOINTS OF PELVIC GIRDLE |
| A03.6.01.001 | Syndesmoses cinguli pelvici | Syndesmoses of pelvic girdle |
| A03.6.01.002 | Membrana obturatoria | Obturator membrane |
| A03.6.01.003 | Canalis obturatorius | Obturator canal |
| A03.6.02.001 | Symphysis pubica | Pubic symphysis |
| A03.6.02.002 | Discus interpubicus; Fibrocartilago interpubica | Interpubic disc; Interpubic fibrocartilage |
| A03.6.02.003 | Lig. pubicum superius | Superior pubic ligament |
| A03.6.02.004 | Lig. pubicum inferius | Inferior pubic ligament |
| A03.6.03.001 | Articulatio sacroiliaca | Sacro-iliac joint |
| A03.6.03.002 | Lig. sacroiliacum anterius | Anterior sacro-iliac ligament |
| A03.6.03.003 | Lig. sacroiliacum interosseum | Interosseous sacro-iliac ligament |
| A03.6.03.004 | Lig. sacroiliacum posterius | Posterior sacro-iliac ligament |
| A03.6.03.005 | Lig. sacrotuberale | Sacrotuberous ligament |
| A03.6.03.006 | Processus falciformis | Falciform process |
| A03.6.03.007 | Lig. sacrospinale | Sacrospinous ligament |
| A03.6.03.008 | Foramen ischiadicum majus | Greater sciatic foramen |
| A03.6.03.009 | Foramen ischiadicum minus | Lesser sciatic foramen |
| A03.6.04.001 JUNCTURAE MEMBRI INFERIORIS LIBERI JOINTS OF FREE LOWER LIMB | | |
| A03.6.05.001 | Syndesmosis tibiofibularis | Tibiofibular syndesmosis; Inferior tibiofibular joint |
| A03.6.05.002 | Membrana interossea cruris | Interosseous membrane of leg |
| A03.6.05.003 | Lig. tibiofibulare anterius | Anterior tibiofibular ligament |
| A03.6.05.004 | Lig. tibiofibulare posterius | Posterior tibiofibular ligament |
| A03.6.06.001 Articulationes membri inferioris liberi Synovial joints of free lower limb | | |
| A03.6.07.001 | Articulatio coxae; Articulatio coxofemoralis | Hip joint |
| A03.6.07.002 | Zona orbicularis | Zona orbicularis |
| A03.6.07.003 | Lig. iliofemorale | Ilio femoral ligament |
| A03.6.07.004 | Pars transversa | Transverse part |
| A03.6.07.005 | Pars descendens | Descending part |
| A03.6.07.006 | Lig. ischiofemorale | Ischiofemorale ligament |
| A03.6.07.007 | Lig. pubofemorale | Pubofemorale ligament |
| A03.6.07.008 | Labrum acetabuli | Acetabular labrum |
| A03.6.07.009 | Lig. transversum acetabuli | Transverse acetabular ligament |
| A03.6.07.010 | Lig. capitis femoris | Ligament of head of femur |
| A03.6.08.001 | Articulatio genus | Knee joint |
| A03.6.08.002 | Meniscus lateralis | Lateral meniscus |
| A03.6.08.003 | Lig. meniscofemorale anterius | Anterior meniscofemorale ligament |
| A03.6.08.004 | Lig. meniscofemorale posterius | Posterior meniscofemorale ligament |
| A03.6.08.005 | Meniscus medialis | Medial meniscus |
| A03.6.08.006 | Lig. transversum genus | Transverse ligament of knee |
| A03.6.08.007 | Lig. cruciatum anterius | Anterior cruciate ligament |
| A03.6.08.008 | Lig. cruciatum posterius | Posterior cruciate ligament |
| A03.6.08.009 | Plica synovialis infrapatellaris | Infrapatellar synovial fold |
| A03.6.08.010 | Plicae alares | Alar folds |
| A03.6.08.011 | Lig. collaterale fibulare | Fibular collateral ligament |

How does *Terminologia* exploit the varied styles of its headings to represent knowledge elements of anatomy? In Figure 1, terms appearing in shaded panels, in upper or lower case, represent groups, kinds or classes of anatomical structures, whereas terms in non-shaded parts of the list, shown in boldface or non-boldface, are individual structures, or so called ‘instances’ (explained and defined below). Although relationships are not explicitly stated, it will be clear to readers of the list (with or without knowledge of anatomy) that it is the -is a- relationship that arranges the headings into a hierarchy. The knowledge embedded in this semantic structure allows one to make, among others, the following inferences. Because the ‘Knee joint’ and the ‘Hip joint’ are siblings in the list, they are of the same kind of joint. The immediate hypernym or parent of these two terms identifies the kind or class of joint to which the knee and hip joints belong: ‘Synovial joints of free lower limb’. The free lower limb also has another joint, which is not of the synovial type: ‘Tibiofibular syndesmosis’. Although this term, and also ‘Pubic symphysis’, for instance, do not include the word ‘joint’, it can be inferred from the hierarchy of their hypernyms that each of these terms designates a joint, albeit in different parts of the lower limb; moreover, both these joints are of a different type or class than the knee or hip joints. Despite the distant location of the terms ‘Knee joint’ and ‘Joints of lower limb’, it may also be inferred through the subordination of intermediate headings that the ‘Knee joint’ -is a- joint of the lower limb, although the list makes no such direct statement.

These examples illustrate the power of structured lists for representing knowledge. The advantage is, that unlike narrative texts, a structured list can be “understood” by computers, enabling them to make the same kind of inferences as humans do. This is only possible, however, if the structure of the list is consistent, logical and free of ambiguity. Does *Terminologia* satisfy these requirements? Let us attempt to answer this question in the context of Figure 1.

Do relationships implied by style hold consistently throughout the list? In Figure 1 all terms appearing in shaded panels have an implied -is a- relationship to the term in the black panel. The same relationship evidently holds between ‘Synovial joints of free lower limb’ in the shaded panel and its hyponyms shown in boldface (‘Hip joint’ and ‘Knee joint’). This assumption cannot be generalized, however, to the upper portion of p. 30. The style implies that ‘Pubic symphysis’ and ‘Sacro-iliac joint’, shown in boldface, are ‘Syndesmoses of pelvic girdle’: placing the latter term in a shaded panel assigns it the role of a hypernym. The implied -is a- relationship, in this instance is false. Based on the style, however, a computer scientist or computer program will assume such a relationship, whereas for an anatomist it will be hard to define any relationship.

At the level of the children of boldfaced terms (e.g., hyponyms of ‘Knee joint’), the implied relationship to their hypernym is -has parts-, rather than -is a-. This apparently also holds true for most of the indented terms (e.g., ‘Iliofemoral ligament’ -has parts- ‘Transverse part’, ‘Descending part’), although the consistency of this representation is questionable. For instance, is ‘Alar folds’ part of, or a kind of, ‘Infrapatellar synovial fold’? Are the anterior and posterior meniscofemoral ligaments parts of the lateral meniscus? Such ambiguities make it hard to interpret the relationships that are implied by the various styles of the headings and by the indentation of terms in the list.

Moreover, in other chapters of *Terminologia*, the styles of headings shown in Figure 1 may assume different implied meanings. For instance, in the cardiovascular system, the same style headings as ‘Joints of pelvic girdle’ and ‘Syndesmoses of pelvic girdle’ are subordinated to one another on the basis of the -part of- rather than the -is a- relationship: ‘AORTA’, ‘Ascending aorta’ (p. 79). Based on the experience with the terms in Figure 1, an individual not familiar with anatomy, or a computer program, would infer that the ascending aorta is one of several aortas, rather than a part of the aorta. Also, subordination of terms to boldface headings, or indentation of terms, may imply -is a-, rather than -part of- relationships as discussed above. For instance, this is the case for hyponyms of ‘Muscles of back proper’ (p. 36), whereas in the case of arteries or nerves, these styles imply -branch of- or -has branch- relationships.

These examples suggest that consistency in the use of styles and symbols can be a powerful feature of a terminology if these symbols are used consistently and if their meanings are declared. The very nature of a structured term list is that it endows with meaning both locality and typographical styles in the list, whether the authors of the terminology have intended that or not. If maintaining consistency of symbols is not practical (i.e., the same style or symbol has to stand for -is a-, -part of- or -branch of- relationships), then it becomes imperative that the relationships be made explicit in the list.

Is the representation logical and free of ambiguity? The examples discussed above suggest that a source of ambiguity is the failure to state explicitly the relationship between terms. Ambiguity also arises when non-boldface terms are directly subordinated to terms that appear in shaded panels. The hyponyms of ‘Syndesmoses of pelvic girdle’ and ‘Tibiofibular syndesmosis’ illustrate this problem (Fig.1).

In addition to the boldface terms ‘Pubic symphysis’ and ‘Sacroiliac joint’ (discussed above), ‘Obturator membrane’ and ‘Obturator canal’ are also subordinated to ‘Syndesmoses of pelvic girdle’. Since these latter terms are not in boldface, they presumably hold a different relationship to their hypernym than ‘Pubic symphysis’ and ‘Sacroiliac joint’. What is this relationship? Is the ‘Obturator membrane’ and ‘Obturator canal’ each a kind of syndesmosis of the pelvis, or are they part of one syndesmosis? The ‘Obturator membrane’ may qualify as a syndesmosis but the ‘Obturator canal’ does not. This is difficult to reconcile with the fact that the parent ‘Syndesmoses of pelvic girdle’ is a plural term. Since neither the ‘Pubic symphysis’ nor the ‘Sacroiliac joint’ is a syndesmosis, the logic of the representation in Figure 1 is further confounded.

Moreover, a comparison with ‘Tibiofibular syndesmosis’ does not help to clear up the ambiguity. Although this term has no boldface children, it is not clear whether the interosseous membrane and anterior and posterior tibiofibular ligaments are part of a single syndesmosis, as the singular form of their hypernym ‘Tibiofibular syndesmosis’ implies, or whether they are each a syndesmosis? Arguably both propositions could be defended. The inconsistency of assigning a single membrane as a hyponym to the plural term ‘Syndesmoses of pelvic girdle’, and several membranes or ligaments as hyponyms to the singular term ‘Tibiofibular syndesmosis’, is confusing.

Is there more than one syndesmosis associated with the pelvic girdle as the parent term 'Syndesmoses of pelvic girdle' implies or is the plural form a typographical error? The children of the 'Sacro-iliac joint' (which is a synovial joint and not a syndesmosis) include the sacrotuberous and sacrospinous ligaments (Fig.1). If the 'Obturator membrane' qualifies as a syndesmosis of the pelvis, so must the sacrotuberous and sacrospinous ligaments. Their sibling relationship with the various sacro-iliac ligaments implies that the sacrotuberous and sacrospinous ligaments are also parts of the sacro-iliac joint. This is an incorrect proposition. Rather, the sacrotuberous and sacrospinous ligaments are syndesmoses and should be subordinated to 'Syndesmoses of pelvic girdle', which would justify the plural form of the parent term.

The representation would be more logical if 'Syndesmoses of pelvic girdle' were shown as a boldface hyponym of 'JOINTS OF PELVIC GIRDLE', equivalent to 'Pubic symphysis' and 'Sacro-iliac joint'. Then 'Obturator membrane', 'Sacrotuberous ligament' and 'Sacrospinous ligament' could be assigned as children of 'Syndesmoses of the pelvic girdle'. Neither the current nor the proposed representation reveals that 'Syndesmoses of the pelvic girdle', 'Pubic symphysis' and 'Sacro-iliac joint' are different types of joints of the pelvic girdle. The terminology should, however, provide this information in order to be consistent with the representation of the joints of the free lower limb. Even if these requirements were satisfied, an explanation would still be called for to justify the inclusion of 'Obturator canal', 'Greater sciatic foramen' and 'Lesser sciatic foramen' under the heading 'Joints of the lower limb', or as part of any of the joints included in the list.

The pursuit of the questions posed in this section about one page of *Terminologia* demonstrates both the power and the challenges of representing anatomical knowledge through a hierarchy of headings. Clarity in the representation of explicit rather than implied relationships between the various types of headings, which represent classes of anatomical entities, is a requirement for assuring the logical and consistent semantic structure of the term list and also for avoiding misleading implications and ambiguity. Equally important is clarity about the role and meaning of terms in the list and the code that is associated with them.

Terms, Concepts and Codes

An anatomical term is a spoken or written expression of a thought that refers to an anatomical entity. Two propositions in this assertion deserve examination: 1. the reciprocal relationship between thought and term; and 2. the nature of the anatomical entities to which the thoughts and terms refer.

Relationship between Thought and Term. In my attempts to illustrate this relationship with specific examples, such as the heart or the little finger, the thought or idea of 'heart' or 'little finger' had to precede the act of writing down these terms. In a reciprocal sense, reading each of these terms engenders corresponding thoughts. In order to conform to the use of terms in some fields of knowledge representation, let us call that thought a 'concept'. The concepts 'heart' and 'little finger' exist regardless of the terms that are

used for communicating these thoughts. For instance, Tarzan would be capable of formulating the concepts 'heart' and 'little finger' and distinguish between the two, even before he met Jane. The concept 'heart' may also be engendered or communicated by the appropriate gestures of a sign language, the symbol ♥, an anatomical drawing or photograph, a radiological image, or an actual anatomical specimen identified in, or removed from, the body of a frog or a human subject.

The purpose of these examples is to illustrate the primary and dominant role of concepts as opposed to that of terms. In other words, the examples emphasize the uniqueness of concepts. Terms are but one of the kinds of symbols for representing anatomical concepts. It is possible, for instance, to construct a multiple-choice examination for evaluating complex and detailed knowledge of anatomy without using any anatomical terms (Rosse et al. 1998). Nevertheless, in comparison with other symbols, terms have many advantages for representing concepts, and provide the most effective and expressive vehicles for this purpose. Therefore, the challenge for knowledge sources in general, and controlled terminologies in particular, is to represent concepts through terms with specificity and consistency. This means that a term should invoke the same thought (concept) in everyone regardless of the context in which the term occurs. The establishment of terminological standards implies also that everyone should use the same term when referring to a particular concept.

The problem is that in anatomy, as in other fields, several terms may invoke the same concept. Such terms are synonyms; e.g., medial ligament of ankle joint and deltoid ligament, both denoted by *Terminologia* code A03.6.10.003. On the other hand, the same term may denote a number of different concepts. Such terms are homonyms; e.g., proximal phalanx, which may refer to a specific segment of a finger or a toe. Both synonyms and homonyms predispose to ambiguity. Such ambiguities may be minimized by the context of narrative text, or by associating the term with a non-verbal representation of the concept, such as a drawing. In a terminology, such contexts can be provided by definitions, the semantic structure of the terminology, and also by some type of numerical code. However, the best representation of the uniqueness of anatomical concepts would be achieved through associating a unique term with each concept.

Uniqueness of Anatomical Concepts. How does *Terminologia Anatomica* represent the uniqueness of anatomical concepts?

The segment of *Terminologia* illustrated in Figure 1 shows three rows in which more than one term appears (see 'Interpubic disc', 'Tibiofibular syndesmosis' and 'Hip joint'). It is made clear that terms in a row refer to the same concept not only by their tabular grouping, but also by assigning the same code to all the terms. The three examples cited illustrate the semantic equivalence of Latin and English language strings, as well as of synonyms in each language. Eponyms, which appear in a separate index rather than in the table, also receive the same code. The grouping of synonyms aided by the tabular form and the code is evidently intended for representing the uniqueness of anatomical concepts; in other words, several terms may refer to the same concept and that concept is implied by the code.

It is more difficult to assure the reciprocal semantic requirement: a term should refer only to one concept. In the majority of instances, *Terminologia* satisfies this requirement. However, there are notable exceptions. For instance, the term ‘Phalanges’ appears in two places and receives two different codes (A02.4.10.001 and A02.5.18.001). In one context the homonym refers to phalanges of the hand and in the other to those of the foot. These are two distinct concepts, as suggested by the specific codes. The terms could also be rendered specific for the respective concepts by appropriate extensions: ‘Phalanges of hand’, ‘Phalanges of foot’. The same pertains to proximal, middle and distal phalanges, which also appear as homonyms associated with different codes. Another homonym is ‘Sacciform recess’, coded A03.5.09.009 and A03.5.10.003, depending on its association with the elbow or distal radio-ulnar joints. In other instances, *Terminologia* restricts the meaning of some of its homonyms. For example, the general term ‘Interosseous membrane’ (A03.0.00.007) is rendered specific for the concepts it designates not only by the code but also by discriminating extensions [e.g., ‘Interosseous membrane of forearm’ (A03.5.06.002) and ‘Interosseous membrane of leg’ (A03.6.05.002)]. Likewise, ambiguity about the meaning of such homonyms as ‘Os’ - referring either to the mouth (A05.1.00.001) or to ‘bone’ - is avoided by appropriate extensions (e.g., Os coxae; A02.5.01.001). The consistent use of such extensions with all homonyms would enhance the specificity of the terminology and would clarify the meaning of a homonym without having to consult the code or the various headings under which the homonym appears.

A particular kind of difficulty arises when *Terminologia* assigns two different codes to a homonym and the semantic structure of the headings of these terms fails to imply two distinct meanings. For instance, ‘Palm’ (A01.1.00.028; Latin equivalents ‘Palma, Vola’) is a hyponym of ‘Parts of human body’ (no code assigned; p.2), and also of ‘Regions of upper limb’ (A01.2.07.001). The latter entry for ‘Palm’ is coded A01.2.07.022, and is associated with the synonym ‘Palmar region’ (Latin equivalents are ‘Palma, Vola, Regio palmaris’). Are there one or two concepts to which these distinctly coded, separate entries refer?

Depending on the context, the term ‘palm’ may actually engender more than two thoughts or concepts, each of which refers to a distinct anatomical entity: 1. The anterior surface of hand (as in “Placed the coin in the palm of your hand”), a two-dimensional entity. 2. A compartment (as in “The lumbrical muscles are located in the palm of the hand), a three-dimensional entity. 3. A subdivision of the hand, complementary to that of the back of the hand (as in “Dissect the palm of the hand before the back of the hand”). As the examples illustrate, the distinctions between these three concepts are provided by the contexts in which the term ‘palm’ appears. It is not clear whether the two entries for ‘Palm’ in *Terminologia* refer to any particular one of these concepts, or to all three. Yet, being included in an anatomical rather than a botanical terminology, no one would associate the homonym ‘palm’ with a tree prevalent in the tropics.

Homonyms should present less of a challenge in anatomy than in most other fields of the biomedical sciences. The reason is that the majority of anatomical terms are associated with distinct physical objects. This should make it relatively easy for the authors of *Terminologia* to assure that both its terms and codes are unique and specific for the concepts or thoughts that refer to discrete objects and spaces that constitute the human body.

The Nature of Named Anatomical Entities. What are the anatomical entities to which the concepts and terms of *Terminologia* refer? As noted in an earlier section, in most instances, the terms appearing as various types of headings refer to groups of anatomical entities. Returning to the example of Figure 1, the term ‘Joints of lower limb’ (A03.6.00.001) designates a group of entities, yet it corresponds to one concept or thought: all the joints of the limb. The same is true of each of the subgroups of joints designated by the subordinate headings in Figure 1. A similar hierarchy of concepts exists in other sections of *Terminologia*; e.g., ‘Spinal nerves’ (A14.2.02.002) and their subgroups. These hierarchies may be thought of as inverted trees, their progressively finer branches issuing from a trunk and terminating in leaves, which are the terms of the lowest order or rank in the terminology. These terms, which have no hyponyms or children, are the ‘leaf concepts’ or ‘instances’ of the terminology.

What are the anatomical entities that correspond to these leaf concepts? One would expect them to be concrete objects and spaces, such as a tibia picked out from a bone box, or an epiploic foramen, into which one inserts a finger. In fact, however, these leaf concepts are as abstract as the higher order concepts and terms of *Terminologia*; they do not refer to concrete objects and spaces. Even the term ‘right tibia’ (not present in *Terminologia*) stands for a group of objects that are remarkably similar, yet distinguishable from one another (e.g., your and my right tibia and that of cadaver No. 33). A distinct thought is associated with each of these tibias, and it is these leaf concepts that are the true, concrete instances in anatomical knowledge, as well as in the everyday practice of medicine. In medical records, these anatomical instances are distinguished from one another by the patient’s name and number, which may be regarded as extensions of their hypernym; e.g., ‘Right tibia of John Doe’. Such an instance is a physical object, in other words, a concrete anatomical structure. *Terminologia* clearly stops considerably short of such a level of granularity and specificity. Its leaf concepts are in fact classes. These leaf concepts, therefore, accommodate both normal and abnormal variations. The variations, however, remain unspecified, which is one factor that constrains the scope of the terminology. It also assigns a broad meaning to leaf concepts, which do not distinguish between bilaterally or serially occurring anatomical entities.

Another factor in constraining the size of *Terminologia* is the compositional approach it has adopted. The User Guide cites the example of the leaf concept ‘Deep nodes’, a child of ‘Popliteal nodes’, which in turn is a hyponym of ‘Lymph nodes of lower limb’. From these entries the required term ‘Deep popliteal lymph nodes’ is to be composed by the user of the terminology. A human user will undoubtedly infer that such nodes exist in the right and left lower limbs, and can generate the more narrow classes of ‘right deep popliteal lymph nodes’ and ‘left deep popliteal lymph nodes’. Only an intelligent computer program can make such an inference. In other instances, however, such inferences may not be obvious to a non-expert user. Consider for instance, ‘Lateral abdominal cutaneous branch’, a hyponym of ‘Intercostal nerves’. How many such branches of each nerve, and how many intercostal nerves on both left and right sides are there? The price paid for concision is that such elementary knowledge elements remain unspecified by the compositional approach. This may restrict the usefulness of the

terminology to those who are familiar with its field. An alternative is an enumerative approach, which explicitly represents each intercostal nerve and each branch of each nerve. Such a terminology will be much larger, but it will include many knowledge elements that are not intuitively evident to its non-expert users.

The Code. *Terminologia* employs a context-dependent alphanumeric code, which is hierarchical and fixed. In preceding sections I make frequent reference to the meaning implied by this code. Figure 1 illustrates the salient properties of the code. Both the advantages and disadvantages of this coding system are revealed by this example.

In the code associated with the heading ‘Joints of lower limb’, the letter ‘A’ presumably distinguishes the “gross” or “topographical” anatomy section of the terminology from its histology and embryology sections, which are as yet anticipated and will presumably be designated by the letters ‘H’ and ‘E’, respectively. The next two digits, ‘03’ stand for the ‘Articular system’; the ‘Skeletal system’ is coded ‘02’ and the ‘Muscular system’ ‘04’. The next number (‘6’) in the code designates the class ‘Joints of lower limb’; the preceding class of the same rank (Joints of upper limb) is numbered ‘5’, and there are no classes beyond ‘6’ in the ‘03’ category. For the moment let us disregard the next entry (Joints of pelvic girdle), and turn to ‘Syndesmoses of pelvic girdle’ coded ‘01’ after the integer ‘6’, implying that this is the first subclass in code category ‘6’. The next triplet of digits should then designate the members of the ‘01’ subclass. The first hyponym of ‘Syndesmoses of pelvic girdle’, however, is numbered ‘002’ rather than ‘001’, because this number is assigned to the term designating the subclass itself. It would be more logical to code ‘Syndesmoses of pelvic girdle’ A03.6.01.000 than A03.6.01.001, and reserve the latter code for the first member of this class, which now has to be coded ‘002’ rather than ‘001’. The same is true for ‘Joints of lower limb’, which should be coded A03.6.00.000, rather than A03.6.00.001.

It is evident that overall the component sections of the code follow the sections and classes of the term list, but the code nevertheless remains less expressive than the style of headings and the indentation of terms. However, implementing a relatively trivial change, such as suggested above for the last group of numbers in the code, would render the system more expressive and would thereby communicate more knowledge.

One of the problems with a fixed hierarchical code is illustrated by the entry ‘Joints of pelvic girdle’, which is coded ‘A03.4’, rather than ‘A03.6’, as is its hypernym. Moreover, the descendants of the ‘4’ group or class resume the code category of ‘6’. The ‘A03.4’ class, when it first appears in the terminology, is preceded by ‘A03.3’ ‘Thoracic joints’, and is succeeded by ‘A03.5’ ‘Joints of upper limb’. While it is logical to include ‘Joints of pelvic girdle’ among the joints of both the trunk and the lower limb, it is disconcerting to encounter a discrepancy in the sequence of the code. This discrepancy is made the more obvious by the fact that the hyponyms of ‘Joints of pelvic girdle’ are not presented in the code category of A03.4, where the term is coded, but in the apparently anomalous code category of ‘A03.6’. Similar discrepancies between the code and the semantic structure of the term list are mentioned in an earlier section in connection with subordinating the pericardium to the left ventricle.

The fixed hierarchical code makes it problematic to represent concepts in more than one place in a terminology. It also makes it difficult to reclassify the concepts according to different viewpoints (e.g., regional instead of systemic) and to add new classes to, or subdivide and merge existing classes.

Consideration of the uniqueness of concepts in a previous section has led to the conclusion that the coding system of *Terminologia* is intended to provide concept rather than term identifiers. It follows that the coding system has to furnish a unique identifier for each unique concept. *Terminologia* meets this primary requirement in the majority of instances. There are exceptions, however, as suggested earlier by the discussion of homonyms, particularly in reference to the 'Palm'. More than one code is assigned to the same term when the homonym appears in different sections of the terminology.

The ambiguous use of the term 'Palm' is not an isolated instance. The same problem exists for almost all the corresponding terms entered under 'Parts of the body' and the various body regions (pp. 2-6). For instance, what is the difference between 'Forehead' (A01.1.00.002) and 'Frontal region' (A01.2.01.002), or 'Occiput' (A01.1.00.003) and 'Occipital region' (A01.2.01.004); and toward the end of the list, between 'Hand' (A01.1.00.025) and 'Hand region' (A01.2.07.017), or 'Foot' (A01.1.00.040) and 'Foot region' (A01.2.08.022)? Since each member of all these pairs receives a different code, the difference between the concepts to which these codes refer should be made explicit. If there is no difference, then there should be only one coded entry. A footnote on page 3 states that the term 'regions' may "be restricted to areas of the surface of the body or be three dimensional". This statement equates, rather than distinguishes so called parts and regions of the body, and therefore augments rather than resolves the ambiguity. These examples illustrate the influence a fixed, context-dependent code mandates; namely, a different meaning should be associated with a term if the term receives more than one distinct code. Thus the code imposes a meaning on the terms. The distinct coding of repeated occurrences of a term implies that each iteration of the term refers to a unique concept.

The disadvantages imposed by a fixed, context-dependent coding systems have been recognized by knowledge modelers (Schulz et al., 1997), and context-free identifiers have been advocated (Cimino, 1998). Terminologies that separate their term hierarchies from the code of concept identifiers retain the ability to represent knowledge through the semantic structure of the term lists. At the same time they also provide flexibility for regrouping their terms and accommodating new terms and classes without disrupting the logic of their representation scheme.

HOW IS ANATOMY REPRESENTED IN OTHER TERMINOLOGIES?

Anatomical concepts and terms are an integral part of not only anatomy texts and atlases but of all discourse pertaining to the human body. Therefore, a logical and consistent representation of anatomical concepts is a requirement for knowledge sources in essentially all fields of the biomedical sciences. The need for structured vocabularies or controlled terminologies has become particularly evident since computerizing the patient record (Shortliffe, 1999) has assumed a national priority in the United States and other

parts of the world. Until recently *Nomina Anatomica* provided the only available structured list of anatomical terms, which could be reused by clinical terminology initiatives. Others have commented on the shortcomings of *Nomina Anatomica* with respect to serving as a resource of anatomical terms for the emerging terminology projects (e.g., Message et al., 1996.). In view of these shortcomings, knowledge modelers were forced to construct their own structured anatomical term lists based on their understanding of narrative text sources. The products of these efforts fall into four categories: 1. *ad hoc* attempts that needed selected anatomical terms for modeling particular disease processes (e.g., Yalcinalp et al. 1990; Horn 1991) or using anatomy for illustrating various knowledge representation approaches (e.g., Niggeman et al. 1990; Lucas 1993); 2. controlled medical terminology projects that incorporate several or all of the basic and clinical biomedical sciences; 3. the Unified Medical Language System (UMLS) of the National Library of Medicine (McCray and Nelson, 1995), which provides a high level scheme for correlating various biomedical terminologies; 4. controlled terminologies that are limited to anatomy, and are complementary or serve as alternatives to *Terminologia* (Pommert et al. 1994; Rosse et al. 1998). Reciprocal relevance exists between *Terminologia* and the terminologies that fall into categories 2-4.

Controlled Medical Terminologies (CMT). In order to evaluate the potential of *Terminologia* for standardizing the representation of anatomical concepts, it may be informative to examine some of the features of the four more widely used CMTs with a focus on their current representation of anatomical information. These selected terminologies include *Medical Subject Headings* (MeSH), developed primarily for indexing the medical literature (National Library of Medicine, 1999); *Systematic Nomenclature of Medicine* (SNOMED), generated under the auspices of the American College of Pathologists (Spackman et al., 1997); the *Read Codes* (RCD), developed for the National Health Service of the United Kingdom (Robinson et al., 1997); *General Architecture for Languages Encyclopedias and Nomenclatures in Medicine* (GALEN), sponsored by the European Union (Rector et al., 1993); and UMLS, which correlates and provides access to these and many other terminologies (McCray and Nelson 1995), with the exception of GALEN. These knowledge sources contain over 100,000 concepts, several thousand of which pertain to anatomy. They are all under active development and rely on a professional editorial and maintenance staff. As a rule, substantial changes are implemented in their new editions. Each of these CMTs has its own distinct semantic structure and represents anatomy in different ways. These differences are perhaps best illustrated by comparing the representation of a particular anatomical concept, such as the pericardium, in the various terminologies. Figure 2 is a collage of these representations as they are displayed through the UMLS browser, accessible on-line.

In Figure 2, panel A identifies the components of UMLS (McCray and Nelson 1995). The other panels were reached through the 'Metathesaurus'. These panels display the results of a search for the term 'Pericardium'. Panel B integrates and enhances the information about the term 'Pericardium', as it appears in the various CMTs accessible through UMLS. The Metathesaurus represents concepts, and associates with each concept a preferred name (Concept name), as well as a numerical unique identifier (UI; 'C' in the code designates 'concept'; term identifiers, not shown in the panel, are headed by the letter 'T'). The example concept is classified by UMLS as 'Tissue' (Semantic type), and a narrative text definition of the concept is provided. In addition to synonyms, the

| | |
|---|---|
|  <p>Users are responsible for compliance with UMLS copyright restrictions.</p> <p style="text-align: right;">A</p> | <p style="text-align: center;">ANCESTORS</p> <p>MSH99 Anatomy (MeSH Category) [A] Tissues [A10] Membranes [A10.615] Serous Membrane [A10.615.789] Pericardium [A10.615.789.470]</p> <p>MSH99 Anatomy (MeSH Category) [A] Cardiovascular System [A7] Heart [A7.541] Pericardium [A7.541.795]</p> <p style="text-align: right;">C</p> |
| <p style="text-align: center;">BASIC CONCEPT INFORMATION</p> <hr/> <p>Concept Name:Pericardium</p> <hr/> <p>UI: C0031050</p> <hr/> <p>Semantic Type: Tissue</p> <hr/> <p>Definition (MSH99): The fibroserous sac surrounding the heart and the roots of the great vessels.</p> <hr/> <p>Synonyms : Pericardial sac 39 PERICARDIUM</p> <hr/> <p>Sources: LCH90, MSH99, RCD98, SNM2, UWDA97, SNMI98, AOD95, CSP98</p> <hr/> <p>Other Languages : PERICARDE - French Perikard - German Herzbeutel - German Pericardium - German Pericardio - Italian PERICARDIO - Portuguese PERIKARD - Russian PERICARDIO - Spanish</p> <p style="text-align: right;">B</p> | <p>RCD98 Anatomical concept [X79tP] Human body structure [Xa16M] Body system structure [Xa1gC] Cardiovascular structure [X74dF] Cardiac structure [Xa061] Cardiac wall structure [XM0PW] Pericardium [7N414]</p> <p>RCD98 Read thesaurus [.....] Anatomical concept [X79tP] Human body structure [Xa16M] Body region structure [Xa1gD] Trunk structure [Xa1gJ] Body cavity structure [Xa1Va] Thoracic cavity structure [X74Xz] Intrathoracic cardiovascular structure [Xa1IX] Cardiac structure [Xa061] Pericardium [7N414]</p> <p style="text-align: right;">D</p> |
| <p style="text-align: right;">B</p> | <p>SNMI98 TOPOGRAPHY CARDIOVASCULAR SYSTEM HEART AND PERICARDIUM PERICARDIUM Pericardium, NOS [T-39000]</p> <p style="text-align: right;">E</p> |

Figure 2. Anatomical concept representation viewed through the Web-based browser of the Unified Medical Language System (UMLS) of the National Library of Medicine. **A.** UMLS logo, with different component programs of UMLS represented by clickable tabs around the circumference. Panels B-E were reached by clicking 'Metathesaurus'. **B.** Basic concept information illustrated by 'Pericardium'. **C-E.** Ancestors of the concept 'Pericardium' represented in three source vocabularies of UMLS: C. Medical Subject Headings (MSH99), D. Read Codes (RCD98) and E. Systematic Nomenclature of Medicine (SNMI98).

Metathesaurus provides a number of equivalents of the term in several languages other than English. This example illustrates that through integrating information from eight different vocabularies (listed by their acronyms as ‘Sources’), the Metathesaurus provides thorough semantic and lexical documentation for an anatomical concept. Panels C-E display the sequential hypernyms (“ancestors”) of the term ‘Pericardium’ in three of eight UMLS terminologies. The UMLS browser displays hyponyms by indentations, although this may not be the case in the individual CMTs.

Of these CMTs, only MeSH provides a narrative text definition, which is based on Dorland’s Medical Dictionary (Dorland’s 1988), and is adopted by UMLS. UMLS provides its own definitions for high level semantic types, according to which concepts in the various source vocabularies are correlated. In the case of the example concept, there is a discrepancy between the definition and the semantic type assignment. The UMLS definition of ‘Tissue’ is “an aggregation of similarly specialized cells united in the performance of a particular function”. The definition of the pericardium as a sac is not consistent with that of ‘Tissue’, since the pericardial sac consists of more than one tissue. Moreover, the pericardial sac consists of more than one type of pericardium. The definition accounts only for one of several concepts to which the term ‘Pericardium’ may refer. It is consistent with the definition provided to regard the ‘Pericardial sac’ as a synonym of ‘Pericardium’. Yet, the definition excludes the concepts that refer to the various membranes that constitute the sac; each is a kind of pericardium, and each in turn is a kind of membrane.

Although none of the displays in panels C-E specifies the relationships between hypernyms and hyponyms, each of the term lists implies that the ‘Pericardium’ is regarded as a membrane rather than a sac. Both MeSH (Fig.2. C) and the Read Codes (Fig.2. D) present a dual classification scheme. In MeSH one code relates the term ‘Pericardium’ to ‘Membranes’, and another code to ‘Heart’ and ‘Cardiovascular System’. In the first instance the implied relationship is -is a-, in the second, -part of-. The Read Codes retain the same alphanumeric identifier for ‘Pericardium’ in both schemes, and trace a dual ancestry to ‘Body region structure’ and ‘Body system structure’. By classifying anatomical entities in parallel as a ‘body region structure’ and a ‘body system structure’, the consistency of the -is a- relationship is maintained. In fact, this classification merges the -is a- and -part of- relationships (Schulz et al., 1997), with the result that ‘Pericardium’ is represented as part of the heart. Pericardium is included in the ‘topography’ or T axis of SNOMED, which is organized according to systems (Fig. 2. E). Although in its latest edition SNOMED distinguishes between -is a- and -part of- relationships, this has not been as yet implemented for its T axis. The implied relationship in Fig.2. E is -part of-.

The examples illustrate that as far as anatomy is concerned, authors of CMTs are wrestling with two problems: 1. how to distinguishing between -is a- and -part of- relationships; and 2. what classes to establish for the classification of anatomical entities.

Whereas it is true that the visceral layer of the serous pericardium may be regarded as part of the heart wall, or of the heart itself, this relationship does not hold for the pericardial sac, or the fibrous pericardium, or for that matter for the parietal layer of the serous pericardium. The ‘Pericardium’ is subordinated to a variety of hypernyms in the

CMTs. Some agreement exists in the systemic classification schemes, but since the implied relationships are -part of-, rather than -is a-, the hypernyms of 'Pericardium' along the systemic axis do not qualify as classes. Unfortunately, it is not possible to verify how 'Pericardium' fits in the anatomy model of GALEN (Rector et al., 1994), since GALEN is not freely accessible. Although GALEN allows part-whole relationships among anatomical structures, its scheme does not correlate with the CMTs shown in Figure 2. It introduces classes of anatomical concepts, which are hard to correlate with other classifications. For example, spaces are classified as structures and the criteria according to which 'ConventionalCavity', 'TrueCavity', ActualCavity, and 'PotentialCavity' may be distinguished from one another, are not declared (as far as one may judge from published reports).

It is interesting to note that the information represented in Figure 2 by the different CMTs, and also by GALEN, is entirely compatible with anatomy textbooks. The relationships implied by textbook descriptions accommodate the divergent representations in these sources. The examples cited above illustrate that textbook descriptions of a concept such as the 'Pericardium' lend themselves to different interpretations when the relationships of this concept have to be modeled in machine-understandable form. In other words, available textbooks do not provide a consistent and logical representation of anatomical concepts, on which authors of controlled medical terminologies could rely. Nor does, in its current state, *Terminologia Anatomica*. The divergent results yielded by attempts to represent anatomical concepts by clinically motivated CMTs illustrate that there is a real need for a logical and consistent knowledge source, which could serve as a standard for anatomical concept representation.

CAN TERMINOLOGIA ANATOMICA BECOME THE STANDARD?

This is the problem that has motivated the authors of *Terminologia* and also the analysis I present in this paper. Before drawing conclusions from my analysis, it may be instructive to look for lessons in the history of *Terminologia Anatomica*. An excellent account of this history is included in *Terminologia*. The first publication of *Nomina Anatomica* in 1895 was intended by its authors, an international group of anatomists, "to serve as a world-wide official standard vocabulary for all health sciences". That the publication of *Terminologia Anatomica*, more than a century later, remains to be motivated by the very same objective suggests that the intervening editions of *Nomina Anatomica* had not entirely attained their goal. Such a conclusion is indeed borne out by the current usage of anatomical terms and by the difficulties encountered in modeling anatomical knowledge.

Knowledge modelers now recognize that it is very difficult to enforce the use of a standard terminology. Terminological standards are at the root of some of the problems faced by computerizing the patient record. Emphasis in the efforts for reaching such standards has shifted, however, from the representation of terms to that of concepts. Indeed, an underlying principle of UMLS is to standardize concept rather than term representation (McCray and Nelson, 1995). This principle provides for the persistence of concepts and standardizes the meaning of varied and changing terms by associating them with a persistent concept. The concept domain of anatomy is perhaps better suited for standardizing its representation than most of the other biomedical sciences, because anatomy describes the physical objects and spaces that constitute the body. Might it

increase the heterogeneity of anatomical terms if *Terminologia*, emulating UMLS, shifted its focus from terms to concepts? That need not be the case. By adopting the approach of UMLS and declaring one of the terms that denote a concept the “preferred name”, a convergence of term usage can in fact be encouraged.

Terminologia Anatomica must be readily navigable by computer programs, without necessarily resorting to a human user who understands anatomy. Unless such navigability is assured, the evolving controlled medical terminologies, and other computer-based applications that require anatomical knowledge, will be barred from efficiently mapping the terms of *Terminologia* to their own systems. The possibility for implementing such programmatic mapping would assure the best guarantee that *Terminologia* would indeed become the standard for anatomical knowledge as we enter the most information-intensive century in the history of anatomy. The release of *Terminologia* on CD-ROM as well as in hard copy was projected in its promotion literature. At the time of writing, more than a year after the publication of the hard copy, the electronic publication is still being awaited. The shortcomings I identified in the consistency and semantic structure of *Terminologia*, however, will impose some limitations on the extent to which computer programs could mine *Terminologia* for anatomical knowledge, even if it were available on CD-ROM.

In order to transform *Terminologia Anatomica* into a contemporary knowledge source that can meet such needs, it may be necessary for the Federative Committee on Anatomical Terminology to reevaluate the potential and scope of *Terminologia*. In doing so, it may prove helpful to learn from current trends and experiences in contemporary knowledge modeling. Although *Terminologia* can exert the greatest potential impact on clinically motivated CMTs, the comparisons I present with such CMTs suggest that plans for the evolution of *Terminologia* should look beyond the current status of CMTs. They all have shortcomings. Authors of next-generation anatomical knowledge sources should, therefore, learn from these shortcomings and aim to overcome them. Being focussed on a well-definable concept domain such as anatomy, anatomists should lead the way in representing deep knowledge within a discipline through a logical and consistent scheme.

In comparison with its predecessor, there are notable improvements in the semantic scheme of the current edition of *Terminologia*. If subsequent editions continue this trend, *Terminologia Anatomica* may well be more successful in influencing the usage of anatomical terms than previous attempts have been during the span of the past century. Controlled medical terminologies, which aspire to encompass the entire concept domain of clinical medicine, are looked upon as knowledge sources that can provide standards for the computer-understandable representation of biomedical concepts. *Terminologia Anatomica* should, therefore, aspire to become an indispensable resource to these evolving and ambitious initiatives in knowledge representation.

DISCUSSION AND CONCLUSIONS

The Federative Committee on Anatomical Terminology has done an admirable job of updating and enhancing *Nomina Anatomica*, which was intended over a century ago as the standard for anatomical terminology. *Terminologia Anatomica* enjoys the advantage

of having been sanctioned by an international body of anatomists, which has been engaged in its preparation for almost a decade. The same body is currently preparing complementary sections of *Terminologia*, which will be concerned with histology and embryology.

The analysis I present in this communication focuses not so much on the terms themselves, but on the enhanced meaning of terms imposed on them by the structure of the vocabulary, in which the terms are embedded. Whether or not intended by its authors, this structure implies definable elements of knowledge. My intention was to examine the extent to which knowledge elements that can be represented through a structured term list are consistent, logical and free of ambiguity in the current, hard copy edition of *Terminologia*.

Establishing a logical and consistent semantic structure for a terminology that represents the critical knowledge elements of a domain is a tall order. *Terminologia Anatomica* has made impressive strides toward such an objective by ordering a large number of its terms in a rich hierarchy. In comparison with *Nomina Anatomica*, *Terminologia* has substantially enhanced the amount of knowledge that can be extracted from its structure. However, the potential and opportunities offered by this structure have not yet been exploited. The focus seems to have been primarily on the terms and associated code. In subsequent editions the emphasis should shift to concepts and the relationships that exist between concepts. Defining relationships presents perhaps the most exciting challenge, since relationships, rather than terms, are the essence of knowledge.

Consideration of concepts and the relationships that exist between them will enforce the rethinking of such traditional aspects of anatomy as the systemic and regional descriptions of the body. Can one discover a logical knowledge organization scheme that accommodates and reconciles these two complementary yet often conflicting views of anatomy? There are a number of fundamental issues that call for more rigorous attention by the authors of *Terminologia*. Perhaps the most important is the establishment of an inheritance hierarchy or ontology, which groups together and distinguishes from one another anatomical entities on the basis of their inherent properties. Such an ontology requires the definition of classes of anatomical entities in terms of inherent properties or defining attributes. Anticipating the need for a knowledge source in the information age will require to consider, and explicitly represent, elements of knowledge that seem superfluous in current narrative text sources, either because these knowledge elements are too abstract or too self-evident to need articulation in words. On the other hand, a machine-understandable, structured terminology will also require greater stringency and specificity than narrative texts. Therefore, the creation of a new knowledge source calls for considering anatomical entities both in broader (i.e., generalizable) and narrower (i.e., specific) terms than traditional sources have demanded.

I have approached the analysis of *Terminologia* from such a dual perspective, because I consider it imperative that a vocabulary such as *Terminologia* should be navigable by computer programs. Therefore, I readily accept the charge that my analysis, focused chiefly on one page of *Terminologia* (Fig.1), may seem to split hairs and may be too pedantic. Yet other pages of *Terminologia* could be, and should be, subjected to similar scrutiny. Moreover, it may not have escaped attention that, in various degrees, similar

shortcomings may also be discovered in other terminologies, which are accessible through UMLS (Fig. 2). These observations indicate that modeling knowledge in the biomedical disciplines is a relatively new and actively evolving field. Anatomy must not only be a part of this activity but should lead the way, because anatomy can provide the foundation for concept representation in other biomedical fields. The challenge in anatomical knowledge representation is to discover methods for modeling the structural organization of the physical objects and spaces that constitute the human body. We anatomists will be assisted in meeting this challenge if we combine the knowledge of our discipline with the experience of those whose primary interest is in developing methods for representing knowledge.

REFERENCES

Cimino J.J. 1998. Desiderata for controlled medical vocabularies in the twenty-first century. *Methods Inf Med* 37:394-403

Dorland's illustrated medical dictionary. 1988. 27th Ed. Philadelphia: Saunders. 1887 p.
 Federative Committee on Anatomical Terminology. 1998. *Terminologia Anatomica*. Stuttgart: Thieme . 292 p.

Horn W. 1991. Utilizing detailed anatomical knowledge for hypothesis formation and hypothesis testing in rheumatological decision support. *Artif Intell Med* 3:21-39.

International Anatomical Nomenclature Committee. 1989. *Nomina Anatomica*. 6th Ed. Edinburgh: Churchill Livingstone. 94 p.

Lucas P. 1993. The representation of medical reasoning models in resolution-based theorem provers. *Artif Intell Med* 5:395-414.

McCray AT, Nelson SJ. 1995. The representation of meaning in the UMLS. *Meth Inform Med* 34:193-201.

Message MA, Anderson RH. 1996. Towards a new terminology for clinical anatomy, with special reference to the heart. *Clin Anat* 9:317-29.

Moore KL, Dalley AF. 1999. *Clinically Oriented Anatomy*. 4th Ed., Philadelphia: Lippincott, Williams & Wilkins Publishers. 1164p.

National Library of Medicine, 1999. *Medical Subject Headings – Annotated Alphabetic list*. U.S. Department of Health and Human Services, Public Health Service; National Library of Medicine, Bethesda, MD. 1265 p.

Niggemann J. 1990. Representation of neuroanatomical knowledge: the description language ADL. In: Czap H, Nedobity W, editors. *Proceedings of the 2nd International Congress on Terminology and Knowledge Engineering: Applications (TKE 90)*. Frankfurt: INDEKS Verlag;200-9.

Pommert A, Schubert R, Riemer M, Schiemann T, Tiede U, Höhne KH. 1994. Symbolic modeling of human anatomy for visualization and simulation. In: *Visualization in Biomedical Computing*. Bellingham (WA): Proceedings series, SPIE - The International Society for Optical Engineering; 2359:412-23.

Rector AL, Nowlan WA, Glowinski A. 1993. Goals for concept representation in the GALEN project. In: Safran C. editor: *Proceedings of the 17th Annual Symposium on Computer Applications in Medical Care (SCAMC 93)*. New York: McGraw Hill. 414-418.

Rector AL, Gangemi A, Galeazzi E, Glowinski AJ, Rossi-Mori A. 1994. The GALEN CORE model schemata for anatomy: Towards a re-usable application-independent model of medical concepts. In: Barahona P, Veloso M, Bryant J, editors. Proceedings of the 12th International Congress of the European Federation for Medical Informatics (MIE 94), Lisbon. IOS Press:229-33.

Robinson D, Comp D, Schulz E, Brown P, Price C. 1997. Updating the Read Codes: user-interactive maintenance of a dynamic clinical vocabulary. *J Am Med Inform Assoc* 4:465-472.

Rosse C, Gaddum-Rosse P. 1997. *Hollinshead's textbook of anatomy*. 5th Ed., Philadelphia: Lippincott-Raven. 902p.

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 Inform Assoc* 5:17-40.

Rosse C, Solomon C. and Schaad DC. 1998. Assessment of spatial reasoning in clinical anatomy. *Clin Anat* 11; p. 136.

Schulz EB, Price C, Brown PJB. 1997. Symbolic anatomic knowledge representation in the Read Codes Version 3: structure and application. *J Am Med Inform Assoc* 4:38-48.

Shortliffe EH. 1999. The evolution of electronic medical records. *Acad Med* 74:414-419.

Spackman KA, Campbell KE, Cote RA. 1997. SNOMED-RT: a reference terminology for health care. In: Masys DR, editor. Proceedings of the 1997 AMIA Annual Fall Symposium. Philadelphia: Hanley & Belfus. 640-644.

Whitmore I. 1999. Terminologia Anatomica: new terminology for the new anatomist. *Anat Rec (New Anat.)* 257:50-53.

Williams PL, Bannister LH, Berry MM, Collins P, Dyson M, Dussec JE, Ferguson MWJ. 1995. *Gray's Anatomy*. 38th Ed. New York: Churchill Livingstone. 2092p.

Yalcinalp LÜ, Sterling L. 1990. Diagnosing jaundice expert system. *Comput Math Appl* 20:125-40.

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