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The GALEN CORE Model Schemata for Anatomy: Towards a Re-usable Application-Independent Model of Medical Concepts

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Despite underlying much of medicine, anatomical concepts are frequently not separated from other aspects of terminology, leading to duplication and repetition. This paper describes the development of a re-usable terminological model of anatomy using the *GALEN Representation And Integration Language* (GRAIL), which embodies multiple subsumption hierarchies and constrained compositional statements. The model comprises elementary entities sanctioning statements connecting these entities. Of particular importance within the model are partonomic relationships, the degree of specification or uniqueness of a structure, and the way in which abnormality is represented. Although significant problems remain, the model is proving useful in providing a basis for a wider terminological system. The paper discusses progress to date, and prospects for the future.

1. Introduction: Why a model of anatomy

Anatomy is central to medical terminology. The central concept in many medical terms is of a abnormal structure, process or procedure combined with an anatomical site. The GALEN project is funded by the European Community under the initiative on Advanced Informatics in Medicine to develop the foundations for the next generation of re-usable, application-independent multi-lingual systems of medical terminology. It seeks to provide a central resource to support coherent integration of medical informatics systems of diverse types.

GALEN is based on a modelling theory embodied in the GALEN Representation And Integration Language (GRAIL) Kernel[1, 2]. It is developing a Terminology Server based on a Coding Reference (CORE) model which serve as an interlingua for representing other terminologies and models. This paper presents the current state of the GALEN CORE Model's high level model for anatomy, version 1.5.

2. Background: Brief Outline Ideas from the GRAIL Kernel

The GRAIL Kernel is a subsumption language related to systems such as CLASSIC [3], BACK [4], etc. but with important differences. A GRAIL model consists of a subsumption hierarchy of elementary entities and a set of sanctioning statements connecting these entities. The sanctioning statements express the constraints on what composite concepts can be formed. Composite concepts can themselves be the topic of further sanctioning statements. Composite concepts are indicated by the keyword *which,* e.g.:

Entity which *Attribute-Value*
Entity which *<Attribute1-value1 Attribute2-value2 ... AttributeN-valueN>.*

Sanctioning statements are made at three levels: the grammar level, sense-level and necessity-level. Statements at each level must be sanctioned by statements at the next higher level. Roughly speaking, the grammar-level statements sanction queries and operations by the knowledge engineer. They correspond most closely to the type constraints found in other languages. Sense-level statements sanction the generation of new particularisations representing 'sensible' medical concepts. Necessity level statements prevent the generation of entities representing redundant concepts — e.g. “the hand which is a part of the arm”. Statements are written in the form:

TopicEntity levelQualifier Attribute-Value.

Models in the GRAIL Kernel are concerned with what it is sensible to say rather than what is true. Hence all statements about anatomical relations are taken as representing ‘conventional’ or ‘conceptual’ knowledge and are not altered by physical changes. For example, surgical removal of the right middle lobe of a patient’s lung does not make us any less sensible to discuss the that patient’s right middle lobe — for example to say that it is absent —nor is their any physical alteration which will make it more sensible to speak of the “left middle lobe” of the lung. (A special mechanism for dealing with major anatomical abnormalities such as *situs inversus* is beyond the scope of this brief paper.)

3. Methodology

The process of developing the GALEN CORE model has involved a series of limited experiments followed by a systematic development effort. Two broad types of corpora are used as sources: i) existing coding and classification systems and nomenclatures ii) text reports and other ‘raw corpora’ provided by different applications within the project. Of the existing corpora, SNOMED-III [5] has been a particularly important as a reference point and source of concepts, and the ability to cover the concepts in ICD-9 [6] is a basic minimal requirement for the model. In addition GALEN participated in the experiments in representing chest radiograph reports conducted by the CANON group [7] and where possible has attempted to remain compatible with the semantic network set out by the UMLS [8]. A systematic methodology has been developed out of these experiments.

Figure 1: Taxonomy of major elementary categories for anatomical model..

AnatomicalConcept	IntrinsicallyPathologicalStructure
Structure	[Ulcer Erosion Tumour ..]
BodyStructure	GeneralisedCavity
AnatomicalRegion	ConventionalCavity
BodyPart	[AbdominalCavity ThoracicCavity...]
[Head Neck Thorax Arm ...]	TrueCavity
[Heart Lung Liver ...]	ActualCavity
...	[AnatomicalSinus Lumen ...]
GenericAnatomicalStructure	PotentialCavity
[Wall Angle Membrane...]	[PleuralSpace PeritonealSpace...]

Figure 2: Summary of major selective and descriptive attributes. The A⩾B means that A subsumes B and is used for value sets which are arranged hierarchically.

Selective Attributes:	
hasLaterality	[left right]
hasUpperLowerPosition	[upperPosition middlePosition lowerPosition].
hasMedialLateralPosition	[medial lateral]
hasProximalDistalPosition	[proximal middleProximalDistal distal]
...	
hasOrdinalPosition	[first second third...]
Descriptive Attributes	
hasTopology	[solid hollow ⩾ tubular]
hasSurfaceVisibility	[surfaceVisible internal]
hasSpecificationLevel	[unspecified⩾partiallySpecified⩾wellSpecified⩾UniquelySpecified]
...	

4. Results: Summary of the Schemata

4.1. Anatomic Structure

4.1.1. Elementary Entities: Structures and BodyParts

The first step in developing a GRAIL model is to divide up entities into clean taxonomies according to the 'subsumption' relation — *i.e.* to decide what elementary entities there are and which entities are kinds of other entities. The basic taxonomy for elementary categories is summarised in Figure 1.

4.1.2. Selectors and Descriptors

To express all subsumptions explicitly produces a complex and unmanageable network. GRAIL allows modelers to assign additional characteristics, or 'criteria', to entities. The GRAIL engine classifies entities formally in the subsumption network based on their criteria. There are a series of modifier attributes which are used to express the characteristics by which structures to be further classified along different axes. The most important additional attributes and the corresponding value types are shown in Figure 2.

4.1.3. Degree of Specification

The degree of specification of a category must be modelled explicitly in GRAIL rather than implied by the class-instance distinction as in most subsumption languages. Different applications will require different degrees of specification. However, in anatomy the degree to which a give structure is unique in the body is elementary — the "liver", "right kidney", "left fourth finger" are each uniquely specified although the first is atomic, and the remainder are composites with increasingly complex patterns of selectors. Degree of specification is applied using necessary level statements: *e.g.*

> *(Finger* which <*hasOrdinalPosition OrdinalValueType*
> *hasLaterality lateralityValueType*>*)*
> necessarily *hasSpecificationLevel uniquqelySpecified.*

4.2. Partitive Attributes

4.2.1. Major partitive attributes

Figure 3: Major partitive attributes and informal tests for their use.

HasDivision	Does it divide into similar pieces with similar layers?
hasSurfaceAnatomicalDivision	- in two dimensions?
hasSolidDivision	- in three dimensions?
hasLinearDivision	Can it be divided into segments? Does obstruction of a division obstruct the whole?
hasBlindPouchDivision	Specific to the appendix vermiformix etc.
hasLayer	When the structure is divided, is there still a layer in each division?
hasStructuralComponent	When a structure is divided, does the component (usually) reside in one division?
hasBranch	Is it a branching structure? e.g. arteries, bronchi, nerves, etc.
hasConstituent	For indefinitely many items— cells, molecules, etc.
isMadeOf	For mass items — liquids, tissues, etc.

Most anatomical concepts fit together with part-whole (partitive) relations. In GRAIL there can be many different part-whole relationships, for example the mitral valve is a structural component of the heart but the mucosa is a layer of the stomach. Subsumption and partitive attributes interact, for example 'shaft of the femur" is a part of the "femur", but a "fracture of the shaft of the femur" is a kind of a "fracture of the femur" Details of the mechanism can be found in [2, 9, 10] and a similar mechanism for conceptual graphs is described in [11]. The major partitive attributes are shown in Figure 3 along with an informal test for how each should be used.

The important distinction is divisions and structural components. Divisions are roughly self-similar and have the same layers. By contrast, components are discrete parts of a particular structure. When the structure is divided, structural components normally remain in one or the other divisions. Branching structures are dealt with by a separate attribute *hasBranch/isBranchOf* because branching is not usually considered transitive — e.g. we do not speak of the radial artery as a branch of the aorta.

4.2.2. Containment and Cavities

Figure 4: Attributes relating to cavities

forms	Hollow structures and bilayered membranes *form* cavities
contains	Cavities contain substances and structures

The structure of cavities and containment is one of the most difficult. Structures are designated as *solid*, *hollow*, or *tubular* by the attribute *hasTopologicalForm*.. All hollow structures form a *TrueCavity*. The *TrueCavity* formed by a tubular structure is called a *Lumen*. In additional bilayered membranes such as the *Pleura* and *Peritoneum* form *PotentialSpaces*. *TrueCavities* may also occur in solid organs either physiologically, as the ventricles of the brain, or pathologically as the cavity in a granuloma. *Cavities* such as the *AbdominalCavity* which are formed by surface structures and whose boundaries are often vague in common usage (whether or not there is a formal anatomical definition) are designated as *ConventionalCavities*. The attribute *contains/isContainedIn* pertains only to cavities and their contents. *Cavities contain Substances* and *Structures* — *BloodVessels contain Blood;* the *AbdominalCavity contains* the *Liver*.

4.3. Linking Anatomy Function

4.3.1. Functions, processes, states and Locations

There are two fundamental attributes which link normal anatomy and with abnormal anatomy and processes respectively — *hasLocation* and *isFunctionOf*. Processes have numerous *Features* , e.g. *Rate*, *Regularity* etc. which may also have *States*. *States* are linked to features of processes and anatomy by the attribute *affects*. The pattern is illustrated by the expressions for the presence of a peptic ulcer in the stomach and gastric hyperacidity respectively:

presence which *affects Peptic (Ulcer* which *hasLocation Stomach)*

elevation which *affects (Rate* which *isFeatureOf (*

secretion which *<isFunctionOf Stomach hasProduct GastricAcid>)).*

4.3.2. Presence and Absence ; abnormality and pathology.

Figure 5: Summary of key attributes linking anatomy, processes, and states.

affects	Links States including presence and absence to features and structures
isFeatureOf	Links *Features* to *Processes*
hasLocation	*Links* (usually pathological) *Structures* to the Structures in which they are located. (Note that this is distinct from hasStructuralComponent which links the features of normal anatomy.)
hasPathologicalStatus	takes values *[pathological physiological]* to indicate whether or not a given state represents a ‘disease’ or ‘pathology’.
hasAbnormalityStatus	takes values [normal nonNormal] to indicate whether a state is the normal structure or in some way variant.

The use of *presence* above provides a uniform manner of dealing with both the *presence* of abnormal structures and the *absence* of normal structures and places both within a uniform pattern along with states of features of processes such as elevation of rate of secretion of gastric acid.

This uniform representation also opens the way to a uniform treatment of *normal/nonNormal* and *pathological/physiological*. The GALEN CORE Model separates these two aspects as distinct — a state may be abnormal without being pathological and, occasionally, the presence of a normal structure may be pathological — as in an ectopic pregnancy. *IntrinsicallyPathologicalStructures* are important category of structures including ulcers, tumours, lacerations, etc. whose presence is always pathological. A summary of the key attributes and their usages is included in Figure 5.

5 An Example

The overall modelling style will be illustrated by the example of the lung. At the top level is the grammar-level statement which indicates that it is reasonable to describe lobes of the lung as having positions “upper”, “middle”, and “lower”.

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(Lobe which *isSolidDivisionOf -Lung)*
grammatically *hasUpperLowerPosition -[upper middle lower]*.

This grammar-level statement sanctions the sense-level statements which describe which positions for left and right lobes:

(Lobe which *isSolidDivisionOf-'RightLung')*
sensibly *hasUpperLowerPosition-[upper middle lower].*

(*Lobe* which *isSolidDivisionOf -'LeftLung'*
sensibly *hasUpperLowerPosition-[upper lower*].

These sense level statements sanction the generation of particularisations such as that representing “the right middle lobe of the lung”:

(Lobe which *<isSolidDivisionOf-'RightLung'*
hasUpperLowerPosition-middle>) name *RightMiddleLobe*

Since *hasUpperLowerPosition-middle* is only sanctioned for the *RightLung*, an attempt to create the entity representing the “left middle lobe” of any but a congenitally abnormal lung generates an error .

In addition the sense-level statements sanction the ‘necessity-level’ statements which say that, conceptually, all right lungs have upper, middle and lower lobes and that all left lungs have upper and lower lobes, and that hence the “right lung which has a middle lobe” is just the “right lung”.

'RightLung' necessarily *hasSolidDivision*
(Lobe which hasUpperLowerPosition-[upper middle lower])

'LeftLung necessarily *hasSolidDivision*
(Lobe which *hasUpperLowerPosition-[upper lower])*

6 Conclusion: Problems and Prospects

This paper provides a brief introduction to the anatomical structure of the GALEN CORE Model version 1.5. Current experience indicates that the basic schema of partitive attributes is effective. Separating pathological status and abnormality as distinct attributes has proved a major simplification, and the use of meta knowledge to describe the level of specification needed for different applications is a significant step forwards towards re-usable systems. The uniform structure of presence and absence has so far proved useful, but raises a series of problems which have yet to be fully resolved. The model is currently being tested and a more complete report will be published in the near future.

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References

1. Rector A, Nowlan W, Glowinski A. Goals for Concept Representation in the GALEN project. In: Safran C, (ed). SCAMC-93. Washington DC: McGraw Hill, 1993: 414-418.
2. Rector A, Nowlan W. The GALEN Representation and Integration Language (GRAIL) Kernel, Version 1. In: The GALEN Consortium for the EC AIM Programme. (Available from Medical Informatics Group, University of Manchester), 1993:
3. Brachman RJ, McGuinness DL, Patel-Schneider PF, Resnick LA, Borgida A. Living with Classic: When and how to use a KL-ONE-like language. In: Sowa J, ed. Principles of Semantic Networks: Explorations in the representation of knowledge. San Mateo, CA: Morgan Kaufmann, 1991: 401-456.
4. Nebel B. Computational Complexity of Terminological Reasoning in Back. Artificial Intelligence 1988;34:371-383.
5. Côté R, Rothwell D. SNOMED 3.Chicago: College of American Pathologists, 1993
6. WHO. International Classification of Diseases, Version 9.Geneva: WHO, 1980

7. Rector A, Glowinski A, Nowlan W, Rossi-Mori A. Medical Concept Models and Medical Records: An approach based on GALEN and PEN&PAD. Journal of the American Medical Informatics Society 1994 (in press);1(3).
8. McCray AT, Hole W. The Scope and Structure of the First Version of the UMLS Semantic Net. In: Miller RA, (ed).SCAMC-90. Washington DC: IEEE Computer Society Press, 1990: 126-30.
9. Rector A, Nowlan W, Kay S. Unifying Medical Information Using An Architecture Based on Medical Descriptions. In: Miller RA, (ed).SCAMC-90. Washington DC: IEEE Computer Society Press, 1990: 190-194
10. Rector A, Nowlan W, Kay S. Conceptual Knowledge: The Core of Medical Information Systems. In: Lun K, Degoulet P, Pierre T, Rienhoff O, (ed). Seventh World Congress on Medical Informatics, MEDINFO-92. Geneva: North-Holland Publishers, 1991: 1420-1426.
11. Bernauer J, Goldberg H. Compositional classification based on conceptual graphs. Artificial Intelligence in Medicine Europe (AIME-93). Munich: , 1993: .