Informatica Biomedica

lezione19

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Informatica Biomedica: Lezione 19

Modeling Biological Structure

Semantic Nets and Ontologies
- An Introduction to the Gene Ontology

Ontology
- A Simple Frame System

The UW Foundational Model of Anatomy

Representing Anatomical Relations in the FMA

A Simple Network Interface -

Fonti essenziali:

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- Chapt. 6 of I.J. Kalet, *Principles of Biomedical Informatics*, Academic Press, 2009.

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- From Wikipedia, the free encyclopedia

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- However, there is a lot of knowledge in the annotations, the text information that accompanies each sequence.
- This is generally not in an easily computable form, but it is computationally powerful material.

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Semantic Nets and Ontologies

We need a language that incorporates the idea of classification of entities, particularly hierarchical classification systems that are usually referred to as ontologies.

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from Wikipedia

Ontology

(from the Greek $\nu\tau$: of being and $\lambda\gamma\iota\alpha$, -logia: science, study, theory) is the philosophical study of the nature of being, existence or reality in general, as well as the basic categories of being and their relations.

Traditionally listed as a part of the major branch of philosophy known as metaphysics, ontology deals with questions concerning what entities exist or can be said to exist, and how such entities can be grouped, related within a hierarchy, and subdivided according to similarities and differences.

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- While frames can be used to represent data, the emphasis here is on frame systems as knowledge representation languages.
- They are an example of a variety of knowledge representation systems called slot and filler systems,
- where the basic structural idea is to group together names of attributes and their values (of course the values can be other frames).

Frames for knowledge representation

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- The slots describe the frame with attribute-value pairs [slotname, value]

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- Structure (anatomy) and function (physiology) are the dual framework for understanding
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- They are also the essential framework for describing biological knowledge from molecular biology to ecology and evolution.

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- Its ontological framework can be applied and extended to all other species.

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- the FMA as such, is not designed as an end-user application for anatomy students, teachers or any other particular user group

Why Foundational? (1/2)

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- In other words, it is not possible to represent or describe the content domains of other, non-anatomical, biomedical disciplines without explicitly or implicitly referring to anatomical entities
- For example, the circulation must take for granted the existence of the heart and blood vessels, and the same is true for gastritis and the stomach as well as for dementia and the brain
- This means that anatomy is foundational to non-anatomical biomedical disciplines because they reuse anatomical classes

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4. Metaknowledge (Mk),

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Anatomy taxonomy (At),

Anatomy

classifies anatomical entities according to the characteristics they share (genus) and by which they can be distinguished from one another (differentia).

This is implemented as a class hierarchy

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- These structural relationships express such ideas as containment, constituent parts, connectivity, etc.

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- This is the motivation for the modeling of anatomical transformation.
- This part of the FMA is more a work in progress than the AT and the ASA.

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- These are anatomical axioms

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- Most importantly, the FMA represents relations between entities, not only in terms of a superclasses or subsumption hierarchy (class-subclass relationships) but also other relationships such as composition (various part-of relations), spatial relations, and connectivity (e.g., for the blood vessels and lymphatic systems, upstream and downstream connectivity)

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- In addition to the general relationships already mentioned, there are specialized relationships that apply only to certain subclasses of anatomical entities

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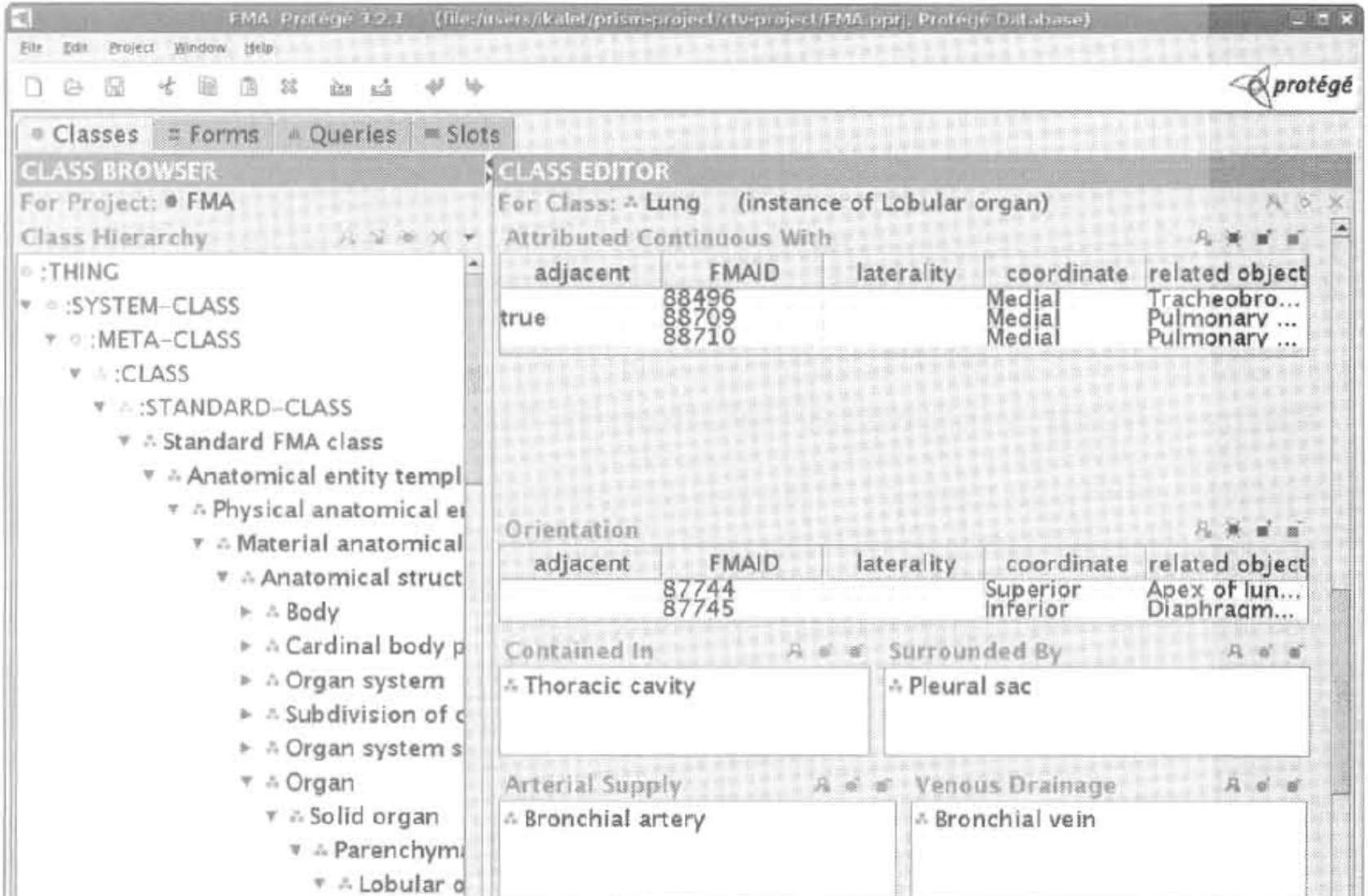
- For example, arterial supply, venous drainage, and lymphatic drainage are relationships between types of vessels and types of organs
- In total there are nearly 200 relationship types in the FMA
- The FMA is organized as a class hierarchy, as seen in the Protégé screen capture below

A part of the FMA class hierarchy

A screen capture of a part of the FMA class hierarchy, displayed in the Protégé class browser window.



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- What is not commonplace is the rich model of relationships in the FMA
- These are among the many pieces of information that are associated with the FMA classes

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- not all anatomical entities have the same attributes, and so in the model, not all the instances of the anatomical entity class should have the same collection of slots, aside from the question of variations in their values
- For example, it makes sense for every entity to have a name, an ID, synonyms, and a description

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- These must be defined as template slots by additional metac1asses, where needed

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- A full-featured frame system is necessary

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- It was developed by the Structural Informatics Group at the University of Washington

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- The initial purpose of the FME was to provide a simple and intuitive interface to the FMA for domain experts, in the field of anatomy, participating in the evaluation of the FMA

Knowledge bases downloads

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- See Web enabled interface to FME