Knowledge Representations

- How else can we represent knowledge in addition to formal logic?

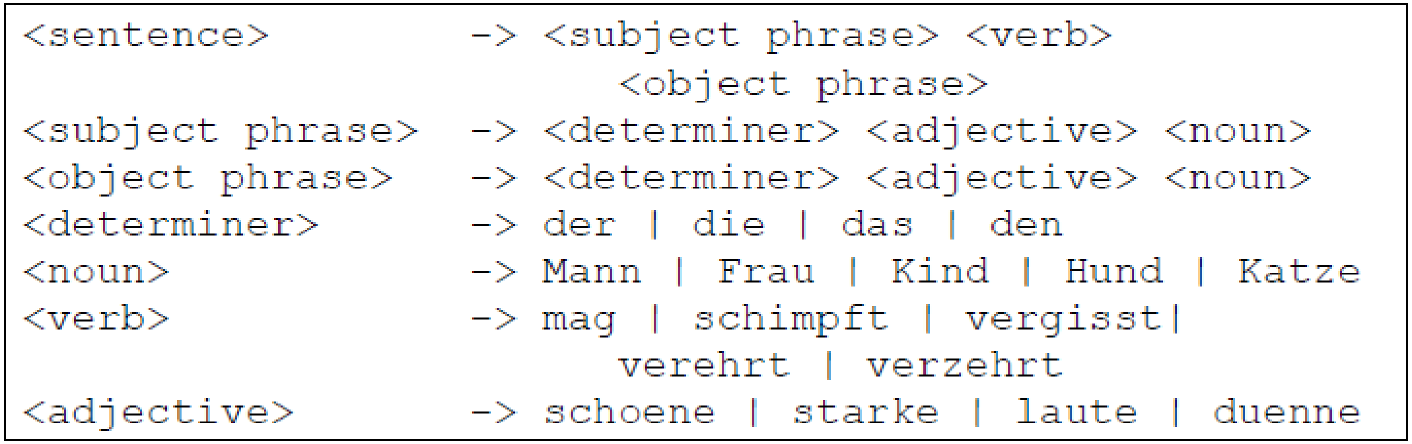
Common Knowledge Representations

- Formal Logic √
- Production Rules
- Semantic Nets
- Schemata and Frames

Production Rules

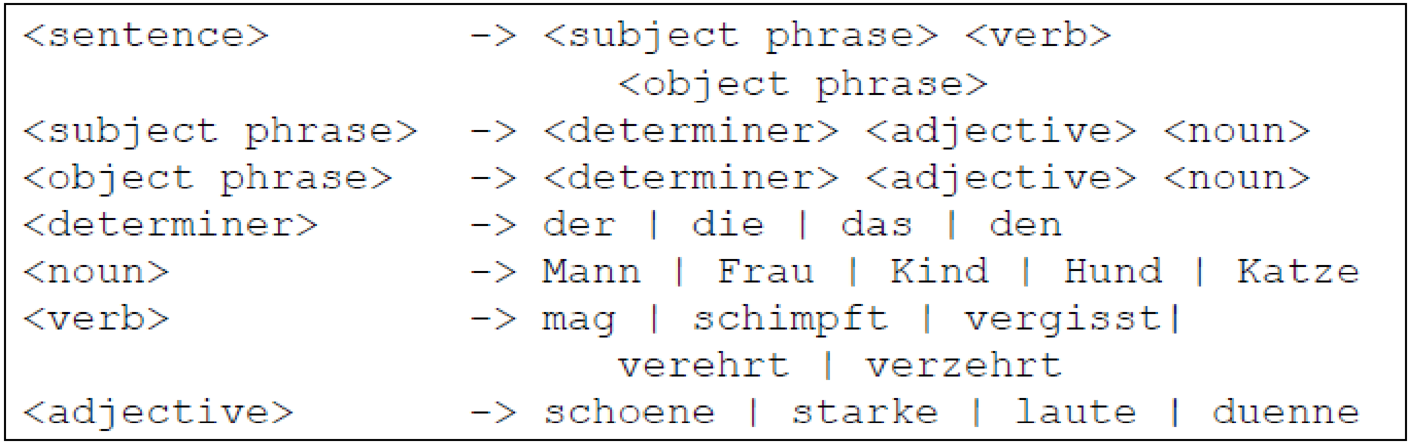
- Frequently used to formulate the knowledge in expert systems.
- BNF is commonly used in Computer Science.

◆ for a subset of the German language

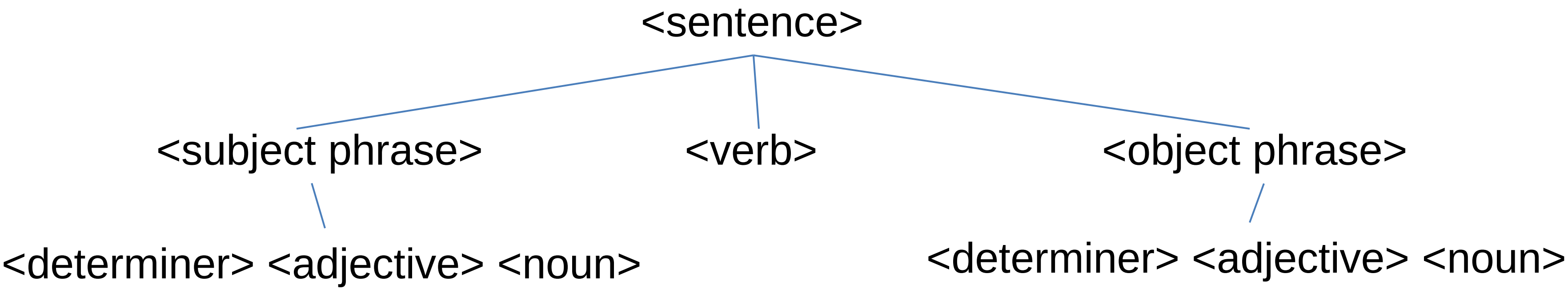


```
<sentence>         -> <subject phrase> <verb>
                        <object phrase>
<subject phrase>   -> <determiner> <adjective> <noun>
<object phrase>    -> <determiner> <adjective> <noun>
<determiner>       -> der | die | das | den
<noun>             -> Mann | Frau | Kind | Hund | Katze
<verb>             -> mag | schimpft | vergisst|
                        verehrt | verzehrt
<adjective>        -> schoene | starke | laute | duenne
```

- for a subset of the German language



```
<sentence>          -> <subject phrase> <verb>
                         <object phrase>
<subject phrase>  -> <determiner> <adjective> <noun>
<object phrase>   -> <determiner> <adjective> <noun>
<determiner>      -> der | die | das | den
<noun>            -> Mann | Frau | Kind | Hund | Katze
<verb>            -> mag | schimpft | vergisst|
                         verehrt | verzehrt
<adjective>       -> schoene | starke | laute | duenne
```



Prolog

- Prolog was developed for AI applications.
- It specifies rules as Horn clauses, a subset of predicate logic.

- Example

```
male( albert ) .
male( edward ) .

female( alice ) .
female( victoria ) .

parents( edward ,  victoria ,  albert ) .
parents( alice ,  victoria ,  albert ) .

sisterof( X , Y )   : -    female( X ) ,
                                parents( X ,  M ,  F ) ,
                                parents( Y ,  M ,  F ) .
```

Prolog Expert System

```
% Automotive Diagnostic Expert System

defect_may_be(drained_battery) :-
    user_says(starter_was_ok, yes),
    user_says(starter_is_ok, no).

defect_may_be(wrong_gear) :-
    user_says(starter_was_ok, no).

defect_may_be(fuel_system) :-
    user_says(starter_was_ok, yes),
    user_says(fuel_is_ok, no).
```

Picture Pattern from my 1974 Thesis

```
PATTERN = CIRCLE $ C1 ¢ CIRCLE $ C2 ¢
    GT(VALU(C2,’RADIUS’), VALU(C1,’RADIUS’)) ¢
    AT(POINT(C2,’TOP’), POINT(C1,’BOT’) ¢
    CIRCLE $ C3 ¢
    GT(VALU(C3,’RADIUS’), VALU(C2,’RADIUS’)) ¢
    AT(POINT(C3,’TOP’), POINT(C2,’BOT’))
```

What is it?

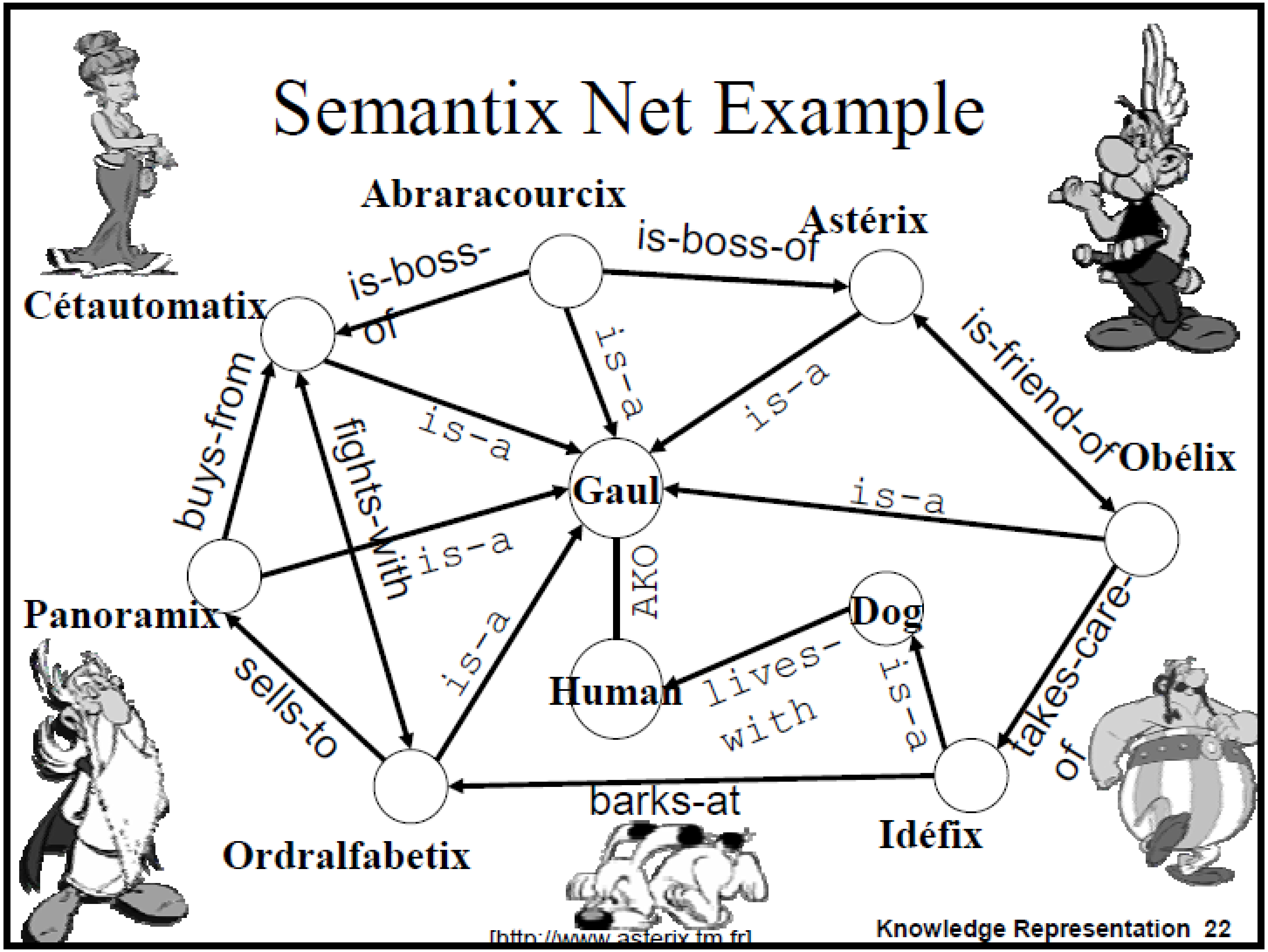
Advantages of Production Rules

- Simpler than full predicate logic
- Still pretty expressive
- Simple backtracking search algorithms
- Easy for programmers to construct the rules
- Humans tend to understand the rules

Disadvantages?

Semantic Nets

- Graphical representation for propositional information
- Originally developed by Quillian as a model for human memory
- Nodes represent objects, concepts, situations
- Edges represent relationships



Semantix Net Example

[http://www.asterix.tm.fr]

Knowledge Representation 22

Semantic Nets

- Relationships
  - Frequently used: IS-A, A-KIND-OF, PART-OF
  - Can be specified by the designer
- Attributes
  - Can be added to the nodes
- Advantages
  - Easy to encode and understand
- Disadvantages
  - May become large and lead to enormous searches

Related Web Developments

- The Semantic Web: an effort to create a web that uses the concepts from semantic nets.
- It would allow people (and programs) to better understand web content.
- Two main representations at present:
  - RDF (Resource Description Framework) low level, triples (node1, relationship, node2)
  - OWL (web ontology language) adds semantics to RDF

Semantic Web Languages

- **RDF (Resource Description Framework)**
  - Triples: <subject> <property> <object>
  - RDF is a data model for objects ("resources") and relations between them. These data models can be represented in an XML syntax.
- **RDFS (RDF Schema)**
  - A vocabulary for describing properties (subclass, subproperty, domain, range) and classes of RDF resources, with a semantics for generalization-hierarchies of such properties and classes.

- **OWL (Web Ontology Language)**

- OWL adds more vocabulary for describing properties and classes: among others, relations between classes (e.g. disjointness), cardinality (e.g. "exactly one"), equality, richer typing of properties, characteristics of properties (e.g. symmetry), and enumerated classes.

- There are constraints on classes and the types of relationships permitted between them. These provide semantics by allowing systems (reasoners) to infer additional information and provide classification based on the data explicitly provided.

Three “flavors” of OWL:

OWL Full

- OWL Full includes all OWL language constructs without restrictions on how they can be used.
- Not decidable

OWL DL (Description logic)

- OWL DL includes all OWL language constructs, but they can be used only under certain restrictions.
- Decidable
- Most ontologies use OWL DL

OWL Lite (even more restricted)

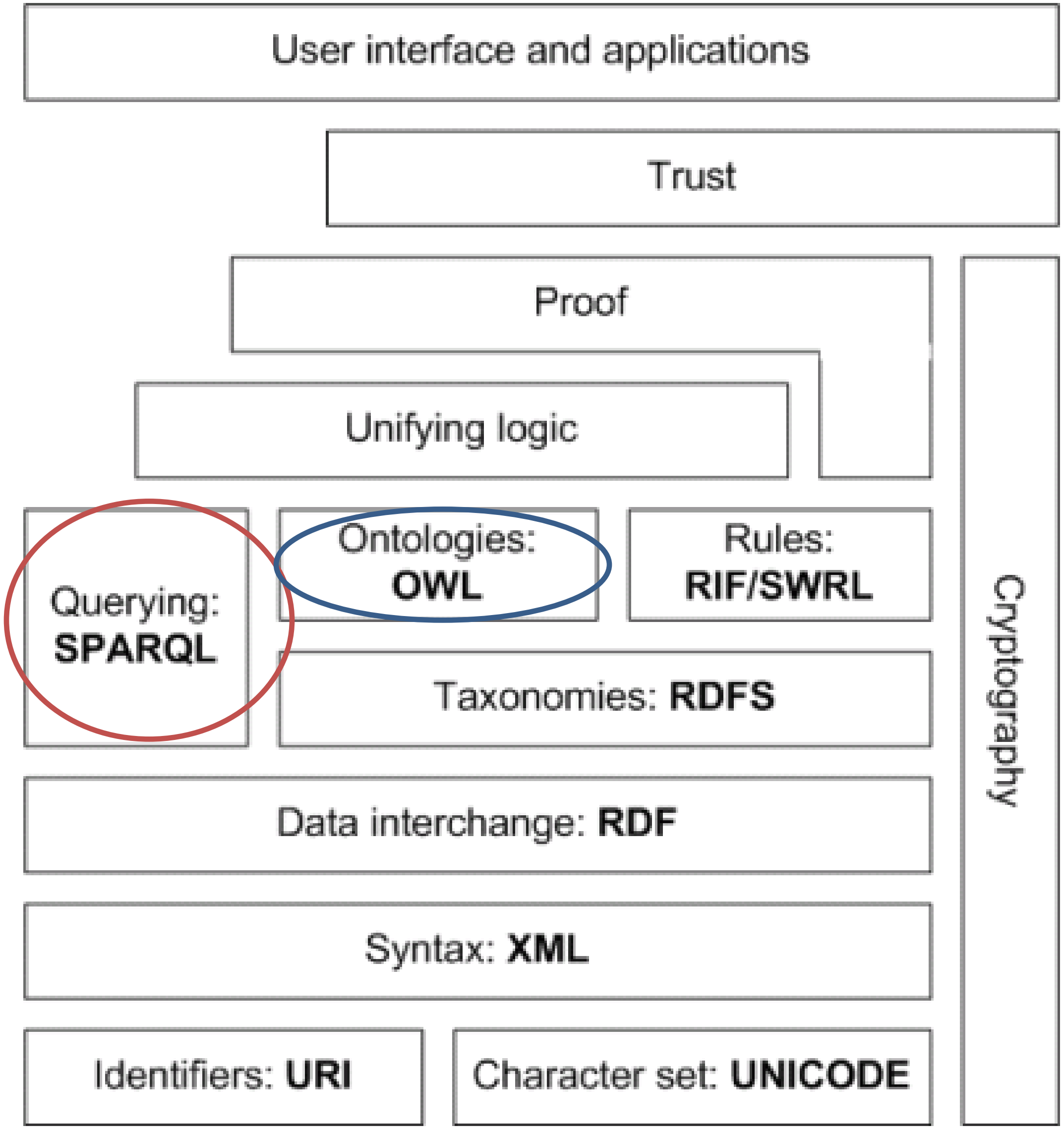
Excerpt of an OWL Ontology:

```
<rdfs:Class rdf:ID="WINE">
    <rdfs:subClassOf rdf:resource="#POTABLE-LIQUID"/>
    <rdfs:subClassOf>
      <daml:Restriction>
        <daml:onProperty rdf:resource="#MAKER"/>
        <daml:minCardinality>
          1
        </daml:minCardinality>
      </daml:Restriction>
    </rdfs:subClassOf>
    <rdfs:subClassOf>
      <daml:Restriction>
        <daml:onProperty rdf:resource="#MAKER"/>
        <daml:toClass rdf:resource="#WINERY"/>
      </daml:Restriction>
    </rdfs:subClassOf>
    <rdfs:subClassOf>
      <daml:Restriction>
        <daml:onProperty rdf:resource="#GRAPE-SLOT"/>
        <daml:minCardinality>
```

My Comments

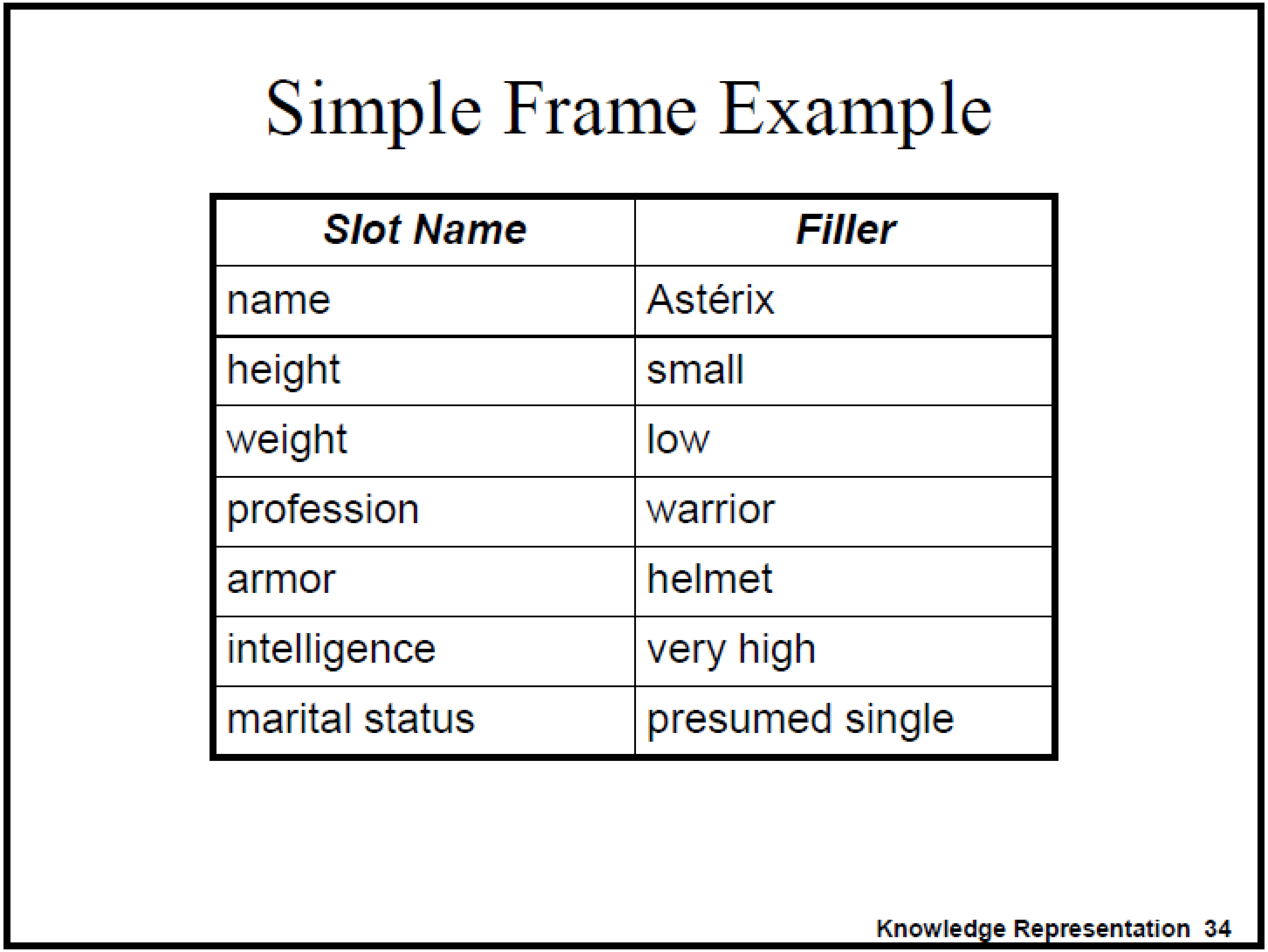
- The international ontology community is sold on OWL, because it is powerful and portable.
- Part of the portability is that it is usually expressed in XML and then interpreted by XML readers.
- Thus, trying to read it as a human is rather difficult; it is for machines.
- But it allows people all over the world to share knowledge bases.

Semantic Web Stack



Frames

- A frame represents related knowledge about a subject
- Frames contain multiple named slots
- Slots contain values of many different kinds
  - rules, facts, images, links to other frames
- Slots can have related procedures that get executed when the value is added, modified or deleted
- Frames can be arranged in a hierarchy or graph



Simple Frame Example

Slot Name	***Filler***
name	Astérix
height	small
weight	low
profession	warrior
armor	helmet
intelligence	very high
marital status	presumed single

Frames

- Advantages
  - Intuitive for many applications
  - Easier to understand than logic
  - Very flexible
- Problems
  - There are inheritance problems, particularly multiple inheritance in graphs

Ontologies

- An ontology is a formal representation of a set of concepts within a domain and the relationships among those concepts.

  Does that sound familiar?

- It allows deep understanding of and reasoning about a domain.
- UW Medical School has one enormous and now famous ontology: the Foundational Model of Anatomy (FMA)

Ontology Tools

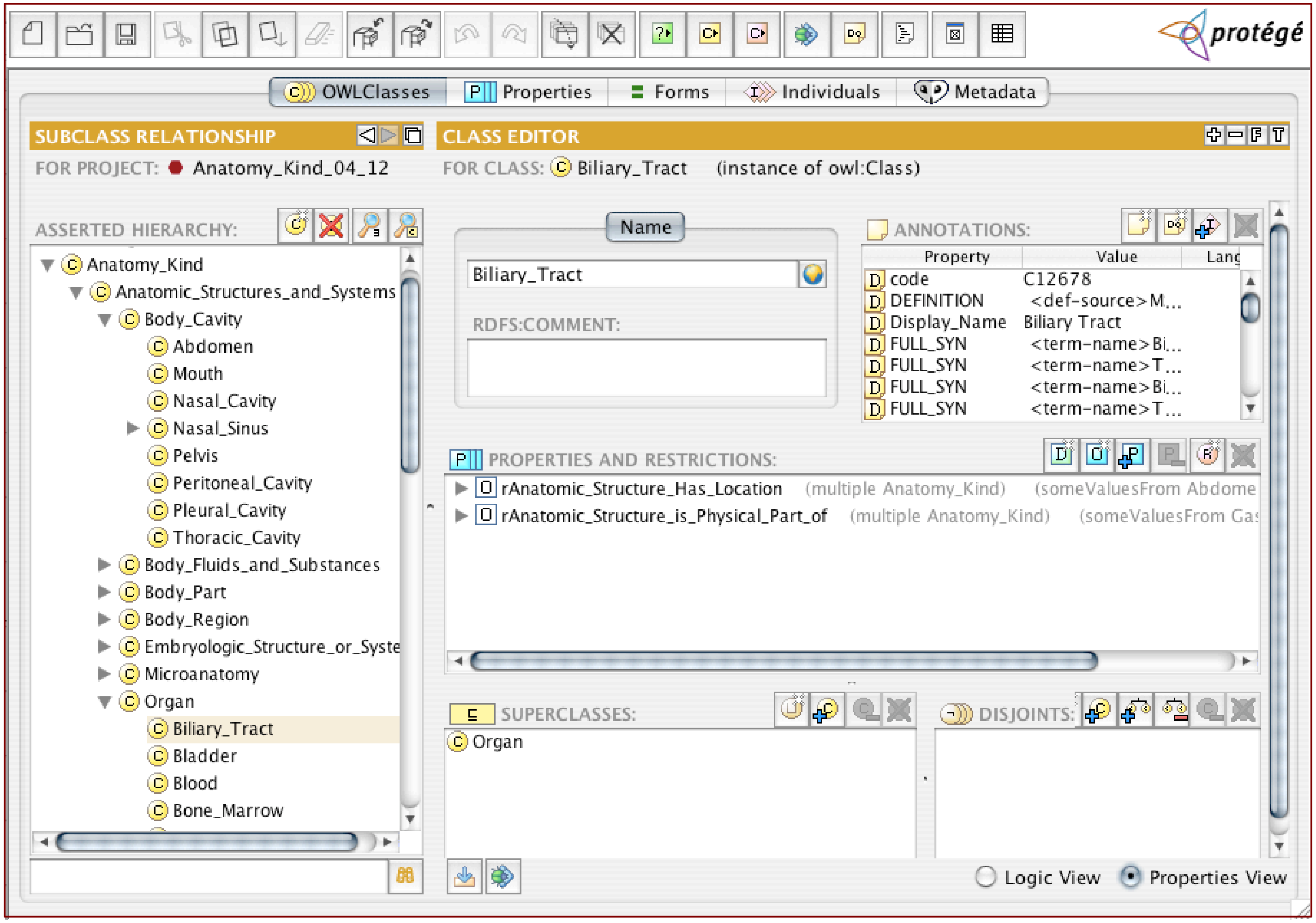
- Ontology-development becomes more accessible with tools.
- Protégé
  - Developed at Stanford Medical Informatics
  - Is an extensible and customizable toolset for
    - constructing knowledge bases
    - developing applications that use these knowledge bases

http://protege.stanford.edu

What is Protégé?

- An ontology editor
- A knowledge-acquisition tool
- A platform for knowledge-based applications

An Ontology Editor



FOR PROJECT: ● Anatomy_Kind_04_12

FOR CLASS: Biliary_Tract (instance of owl:Class)

```
▼ Anatomy_Kind
  ▼ Anatomic_Structures_and_Systems
    ▼ Body_Cavity
        Abdomen
        Mouth
        Nasal_Cavity
      ▶ Nasal_Sinus
        Pelvis
        Peritoneal_Cavity
        Pleural_Cavity
        Thoracic_Cavity
    ▶ Body_Fluids_and_Substances
    ▶ Body_Part
    ▶ Body_Region
    ▶ Embryologic_Structure_or_Syste
    ▶ Microanatomy
    ▼ Organ
        Biliary_Tract
        Bladder
        Blood
        Bone_Marrow
```

Name: Biliary_Tract

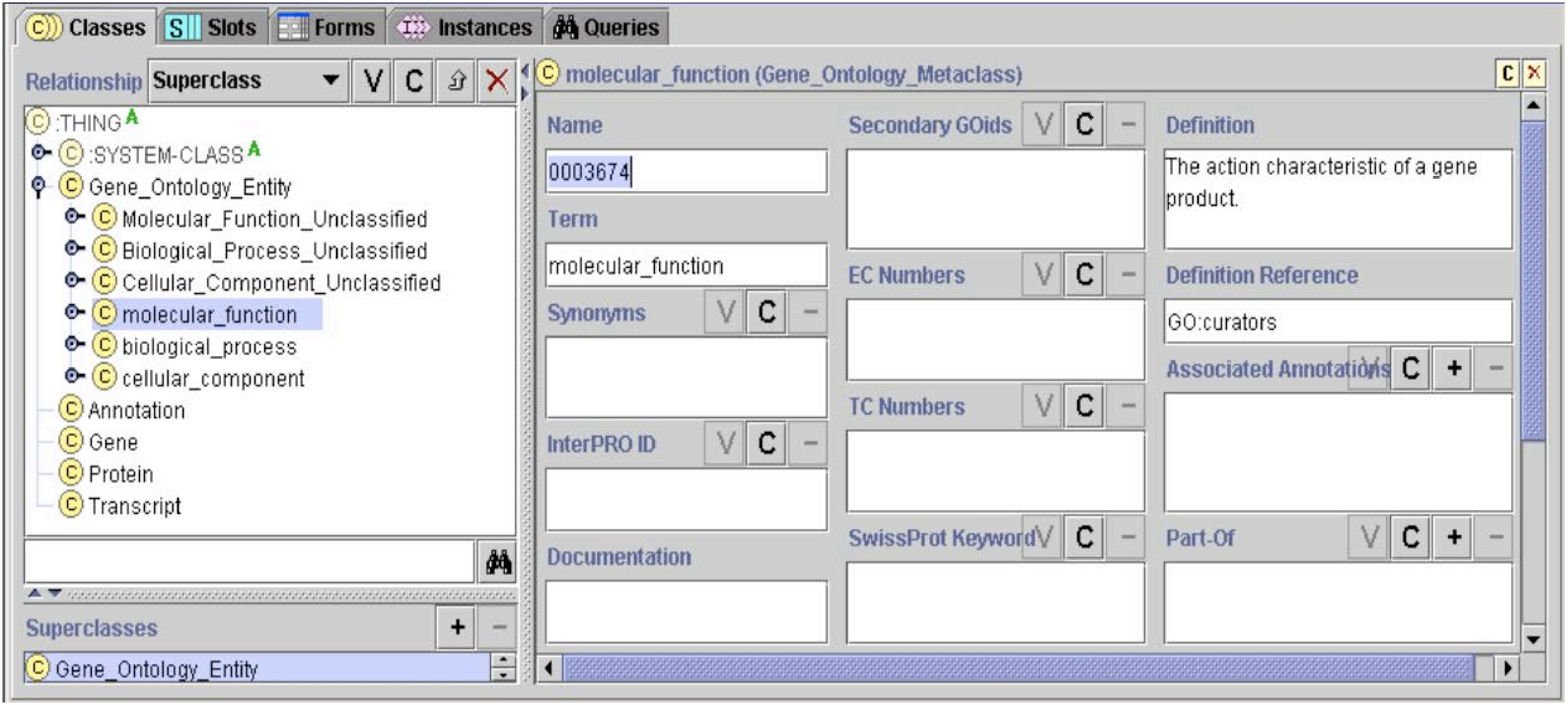
Property	Value	Lang
code	C12678	
DEFINITION	<def-source>M...	
Display_Name	Biliary Tract	
FULL_SYN	<term-name>Bi...	
FULL_SYN	<term-name>T...	
FULL_SYN	<term-name>Bi...	
FULL_SYN	<term-name>T...	

```
▶ rAnatomic_Structure_Has_Location      (multiple Anatomy_Kind)   (someValuesFrom Abdome
▶ rAnatomic_Structure_is_Physical_Part_of   (multiple Anatomy_Kind)   (someValuesFrom Gas
```

SUPERCLASSES: Organ

NCI Thesaurus

A Knowledge-Acquisition Tool



molecular_function (Gene_Ontology_Metaclass)

Name: 0003674

Term: molecular_function

Definition: The action characteristic of a gene product.

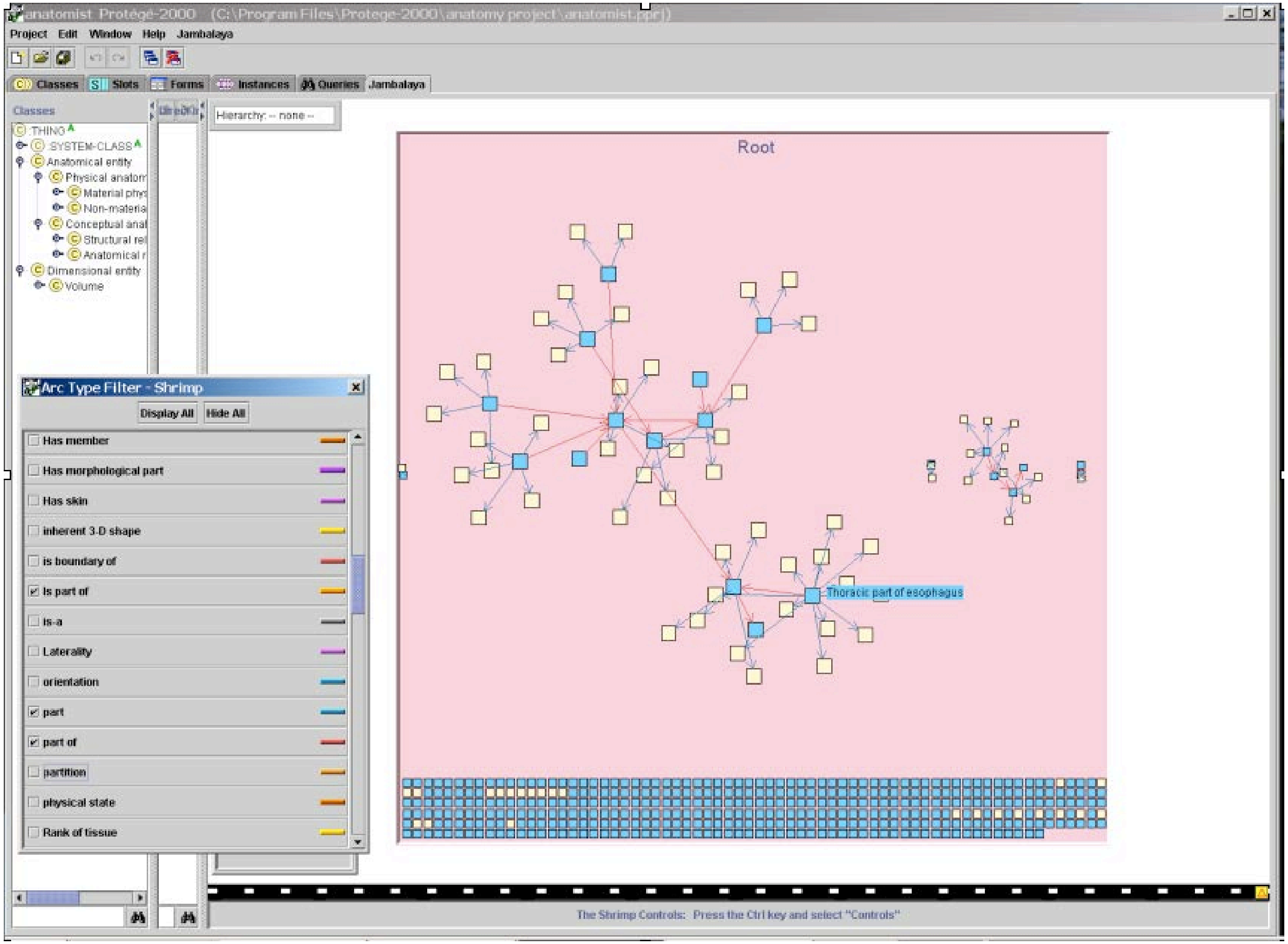
Definition Reference: GO:curators

Gene Ontology

A Platform for Other Applications

- A Java API that enables developers to write plugins for
  - Visualization systems
  - Inferencing systems
  - Scripting facilities
  - Import and export formats
  - User-interface features
  - Means of accessing external data sources
- About 60 plugins currently in the library (developed at Stanford and elsewhere)

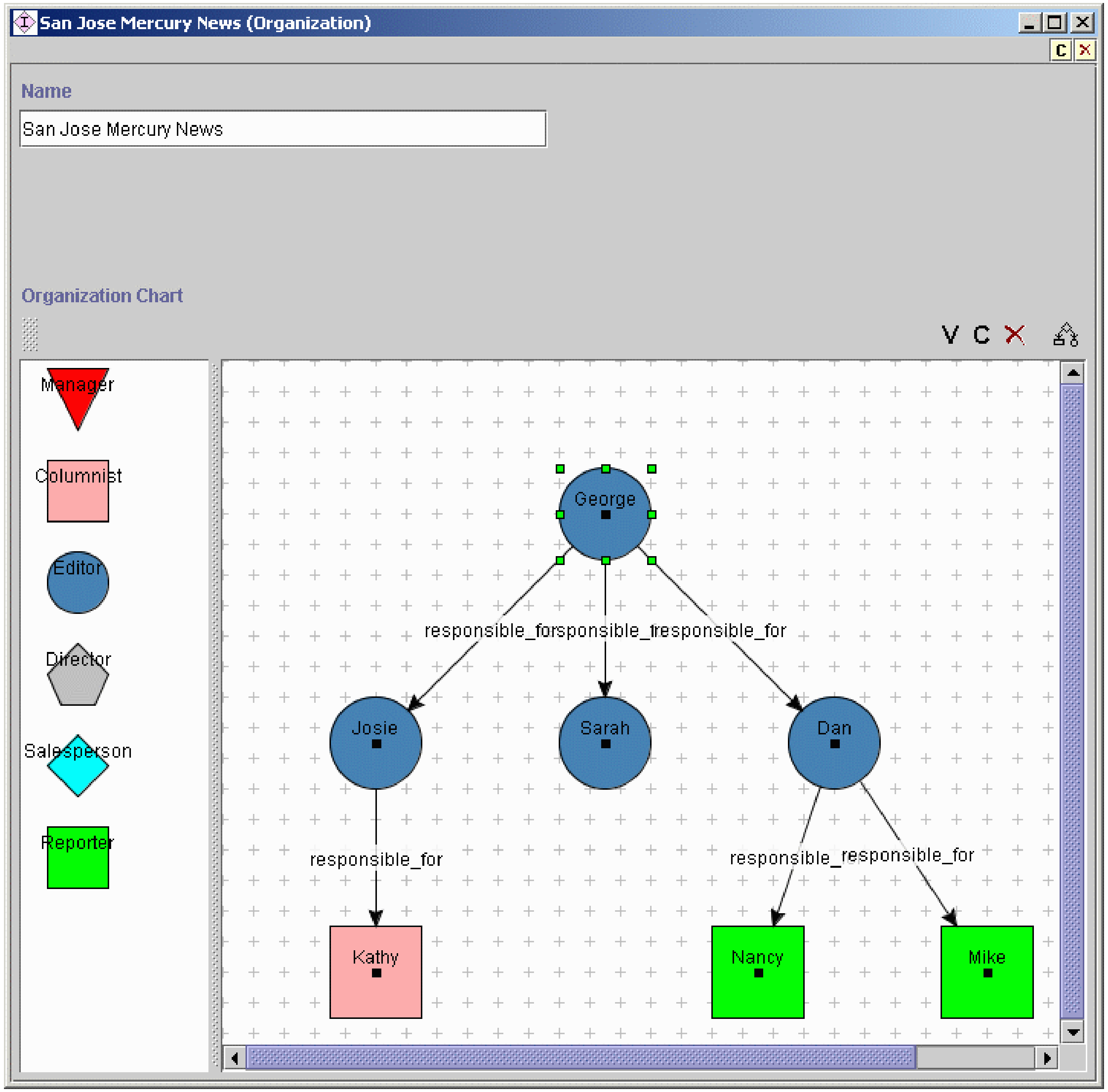
Visualization: Jambalaya


anatomist Protégé-2000 (C:\Program Files\Protege-2000\anatomy project\anatomist.pprj)
Project Edit Window Help Jambalaya
Classes Slots Forms Instances Queries Jambalaya
Classes
:THING
:SYSTEM-CLASS
Anatomical entity
Physical anatom
Material phys
Non-materia
Conceptual anat
Structural rel
Anatomical r
Dimensional entity
Volume
Hierarchy: -- none --
Root
Thoracic part of esophagus
Arc Type Filter - Shrimp
Display All
Hide All
The Shrimp Controls: Press the Ctrl key and select "Controls"

□ Has member
□ Has morphological part
□ Has skin
□ inherent 3-D shape
□ is boundary of
☑ Is part of
□ is-a
□ Laterality
□ orientation
☑ part
☑ part of
□ partition
□ physical state
□ Rank of tissue

Developed at University of Victoria

Graph Widget



San Jose Mercury News (Organization)

Name

San Jose Mercury News

Organization Chart

Some Applications Supported by Protégé:

- Surveillance of data sources for evidence of potential bioterrorism
- Concept-based information retrieval
- Modeling of metabolic pathways
- Automation of guideline-based therapy

What Makes Protégé Different

- Easy-to-use graphical interface
- Scalability
  - currently can handle up to 5 million concepts
- Plugin architecture
  - active international community of plugin developers
- It’s a platform for other applications
  - Integration with Eclipse (Mayo Clinic)
  - A server and a client for (Semantic) Web Services
- Open source

Foundational Model of Anatomy

- Developed here at UW in the Department of Biological Structure by Prof. Cornelius Rosse and Dr. Onard Mejino and other team members (including me)
- Purpose was to provide a comprehensive framework for the study of the human body
- Can be used by students, doctors, medical researchers, even the general public

Foundational Model of Anatomy (FMA) Ontology

Motivating Hypothesis

“A sound ontological framework of biological structure (anatomy) provides a logical, comprehensive and efficient framework for organizing all types of information about biological organisms”

Why Anatomy?

Hypothesis 1: Manifestations of health and disease are attributes of anatomical structures.

Hypothesis 2: Representation of anatomy should facilitate representation of other domains and interoperability between biomedical domains.

Foundational Model of Anatomy (FMA) Ontology

Theory:

The FMA is a *spatio-structural ontology* of the entities and relations which together form the *phenotypic structure* of a biological organism at all salient *levels of granularity*.

High level Objectives of the FMA theory

Foundational Model of Anatomy

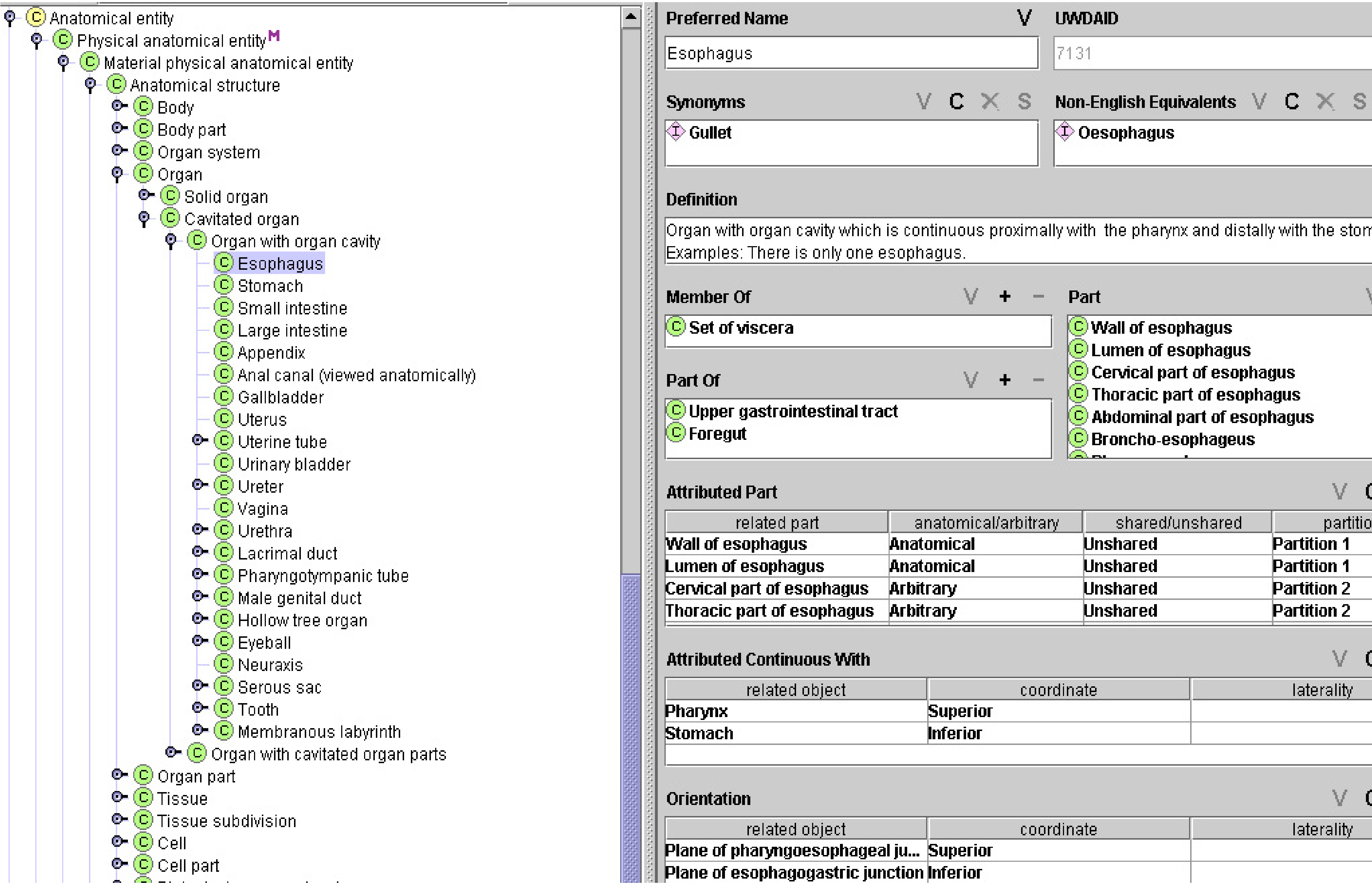
declare the principles

for including entities and relations
that are implicitly assumed
when knowledge of anatomy
is applied in different contexts;

explicitly define

entities and relations
necessary and sufficient for consistently
representing the structure of a
biological organism.

Foundational Model of Anatomy Ontology



- Anatomical entity
  - Physical anatomical entity M
    - Material physical anatomical entity
      - Anatomical structure
        - Body
        - Body part
        - Organ system
        - Organ
          - Solid organ
          - Cavitated organ
            - Organ with organ cavity
              - Esophagus
              - Stomach
              - Small intestine
              - Large intestine
              - Appendix
              - Anal canal (viewed anatomically)
              - Gallbladder
              - Uterus
              - Uterine tube
              - Urinary bladder
              - Ureter
              - Vagina
              - Urethra
              - Lacrimal duct
              - Pharyngotympanic tube
              - Male genital duct
              - Hollow tree organ
              - Eyeball
              - Neuraxis
              - Serous sac
              - Tooth
              - Membranous labyrinth
            - Organ with cavitated organ parts
        - Organ part
        - Tissue
        - Tissue subdivision
        - Cell
        - Cell part

Preferred Name V: Esophagus

UWDAID: 7131

Synonyms V C ✕ S: Gullet

Non-English Equivalents V C ✕ S: Oesophagus

Definition

Organ with organ cavity which is continuous proximally with the pharynx and distally with the stom
Examples: There is only one esophagus.

Member Of V + −: Set of viscera

Part Of V + −: Upper gastrointestinal tract; Foregut

Part V: Wall of esophagus; Lumen of esophagus; Cervical part of esophagus; Thoracic part of esophagus; Abdominal part of esophagus; Broncho-esophageus

Attributed Part V

related part	anatomical/arbitrary	shared/unshared	partitio
Wall of esophagus	Anatomical	Unshared	Partition 1
Lumen of esophagus	Anatomical	Unshared	Partition 1
Cervical part of esophagus	Arbitrary	Unshared	Partition 2
Thoracic part of esophagus	Arbitrary	Unshared	Partition 2

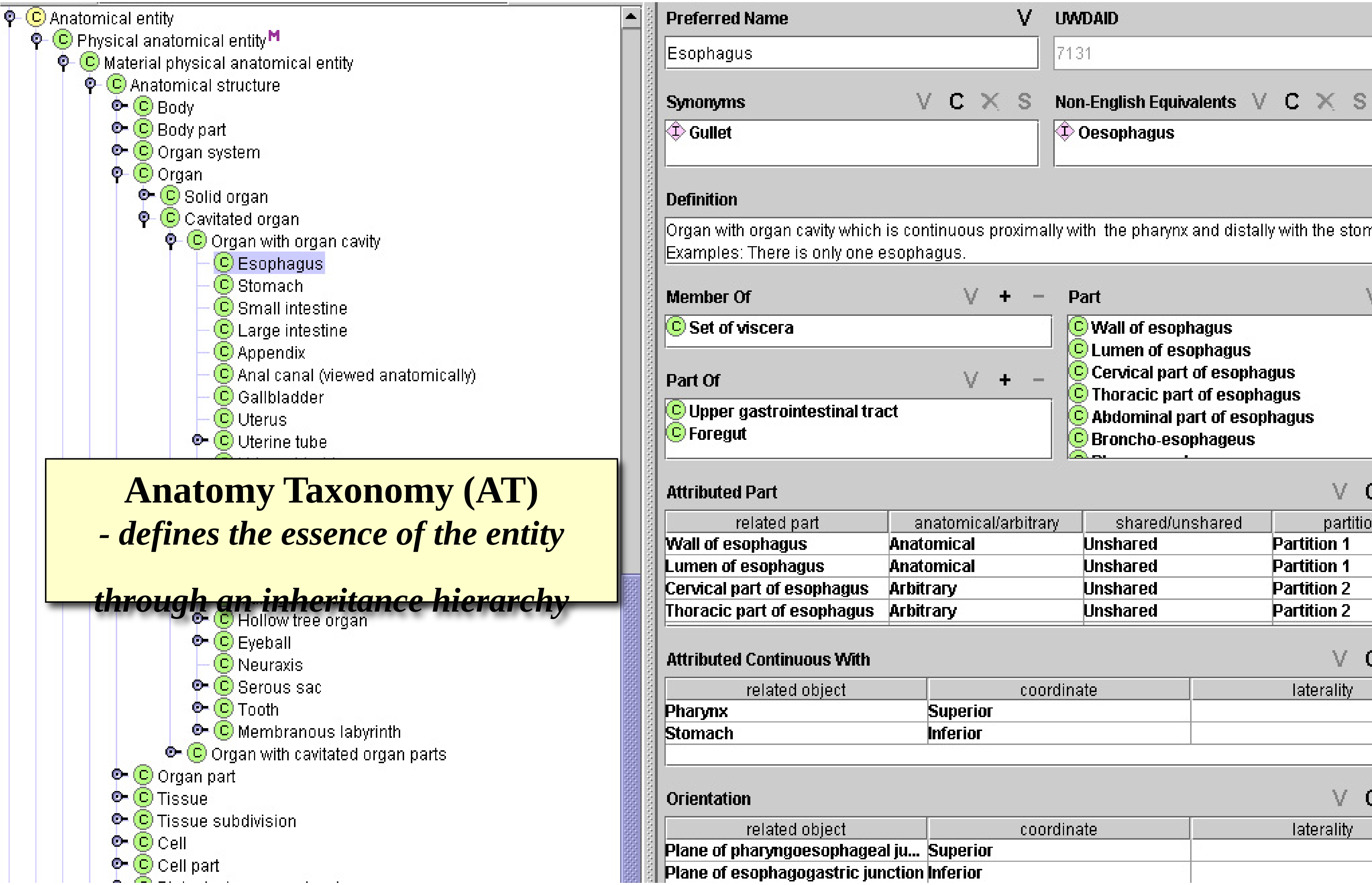
Attributed Continuous With V

related object	coordinate	laterality
Pharynx	Superior	
Stomach	Inferior	

Orientation V

related object	coordinate	laterality
Plane of pharyngoesophageal ju...	Superior	
Plane of esophagogastric junction	Inferior	

Foundational Model of Anatomy Ontology



- Anatomical entity
  - Physical anatomical entity M
    - Material physical anatomical entity
      - Anatomical structure
        - Body
        - Body part
        - Organ system
        - Organ
          - Solid organ
          - Cavitated organ
            - Organ with organ cavity
              - Esophagus
              - Stomach
              - Small intestine
              - Large intestine
              - Appendix
              - Anal canal (viewed anatomically)
              - Gallbladder
              - Uterus
              - Uterine tube
              - Hollow tree organ
              - Eyeball
              - Neuraxis
              - Serous sac
              - Tooth
              - Membranous labyrinth
            - Organ with cavitated organ parts
        - Organ part
        - Tissue
        - Tissue subdivision
        - Cell
        - Cell part

Preferred Name: Esophagus

UWDAID: 7131

Synonyms: Gullet

Non-English Equivalents: Oesophagus

Definition

Organ with organ cavity which is continuous proximally with the pharynx and distally with the stom
Examples: There is only one esophagus.

Member Of: Set of viscera

Part Of: Upper gastrointestinal tract; Foregut

Part: Wall of esophagus; Lumen of esophagus; Cervical part of esophagus; Thoracic part of esophagus; Abdominal part of esophagus; Broncho-esophageus

Attributed Part

related part	anatomical/arbitrary	shared/unshared	partitio
Wall of esophagus	Anatomical	Unshared	Partition 1
Lumen of esophagus	Anatomical	Unshared	Partition 1
Cervical part of esophagus	Arbitrary	Unshared	Partition 2
Thoracic part of esophagus	Arbitrary	Unshared	Partition 2

Attributed Continuous With

related object	coordinate	laterality
Pharynx	Superior	
Stomach	Inferior	

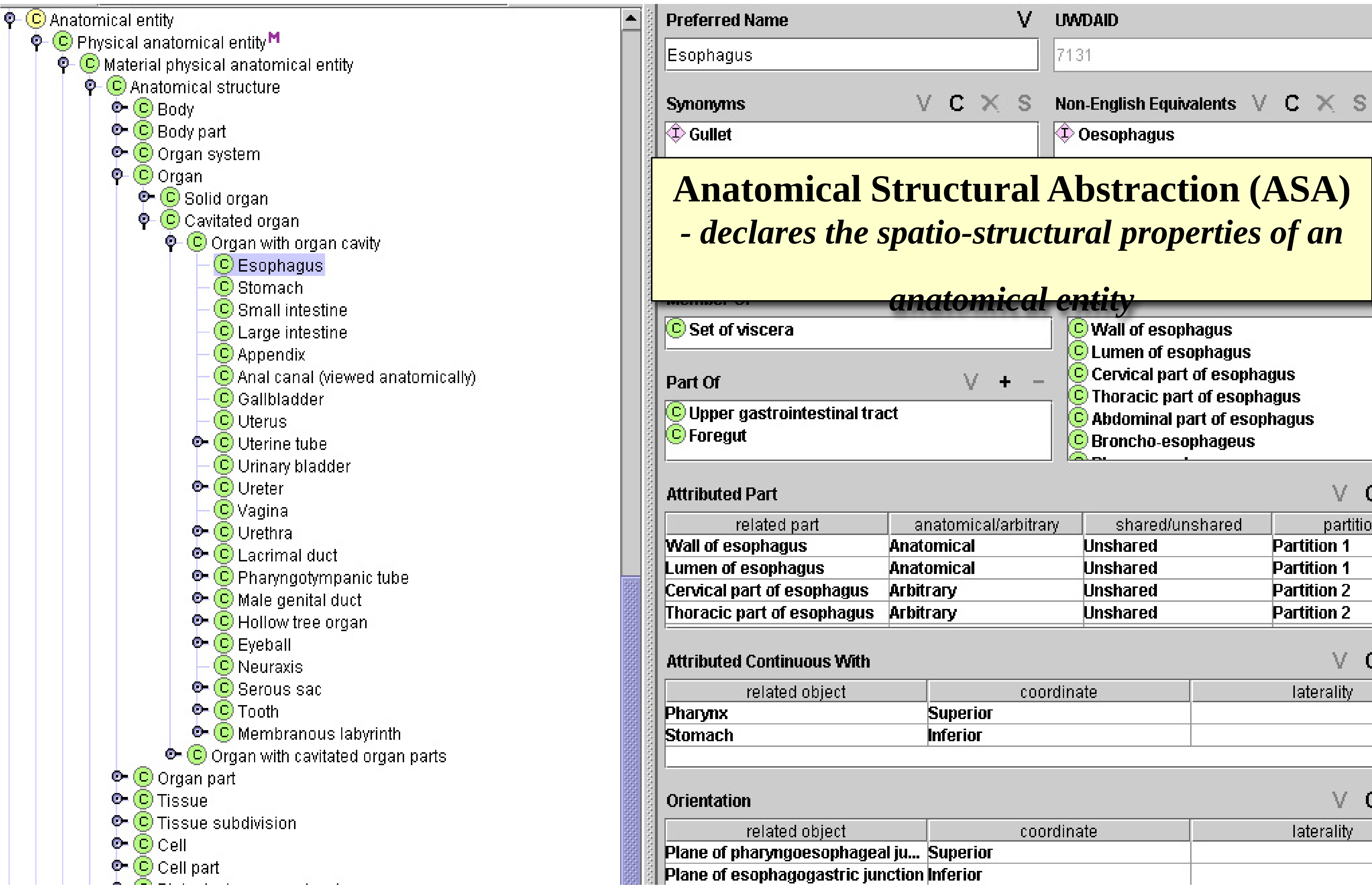
Orientation

related object	coordinate	laterality
Plane of pharyngoesophageal ju...	Superior	
Plane of esophagogastric junction	Inferior	

Anatomy Taxonomy (AT)

- defines the essence of the entity through an inheritance hierarchy

Foundational Model of Anatomy Ontology



- Anatomical entity
  - Physical anatomical entity M
    - Material physical anatomical entity
      - Anatomical structure
        - Body
        - Body part
        - Organ system
        - Organ
          - Solid organ
          - Cavitated organ
            - Organ with organ cavity
              - Esophagus
              - Stomach
              - Small intestine
              - Large intestine
              - Appendix
              - Anal canal (viewed anatomically)
              - Gallbladder
              - Uterus
              - Uterine tube
              - Urinary bladder
              - Ureter
              - Vagina
              - Urethra
              - Lacrimal duct
              - Pharyngotympanic tube
              - Male genital duct
              - Hollow tree organ
              - Eyeball
              - Neuraxis
              - Serous sac
              - Tooth
              - Membranous labyrinth
            - Organ with cavitated organ parts
        - Organ part
        - Tissue
        - Tissue subdivision
        - Cell
        - Cell part

Preferred Name: Esophagus

UWDAID: 7131

Synonyms: Gullet

Non-English Equivalents: Oesophagus

Member Of: Set of viscera

Part Of:
- Upper gastrointestinal tract
- Foregut

- Wall of esophagus
- Lumen of esophagus
- Cervical part of esophagus
- Thoracic part of esophagus
- Abdominal part of esophagus
- Broncho-esophageus

Attributed Part

related part	anatomical/arbitrary	shared/unshared	partitio
Wall of esophagus	Anatomical	Unshared	Partition 1
Lumen of esophagus	Anatomical	Unshared	Partition 1
Cervical part of esophagus	Arbitrary	Unshared	Partition 2
Thoracic part of esophagus	Arbitrary	Unshared	Partition 2

Attributed Continuous With

related object	coordinate	laterality
Pharynx	Superior	
Stomach	Inferior	

Orientation

related object	coordinate	laterality
Plane of pharyngoesophageal ju...	Superior	
Plane of esophagogastric junction	Inferior	

Anatomical Structural Abstraction (ASA)

- declares the spatio-structural properties of an anatomical entity

Foundational Model of Anatomy Ontology

Unifying theory of anatomy

High Level Scheme

$$\textbf{FMA} = (\textbf{At}, \textbf{ASA}, \textbf{ATA}, \textbf{Mk})$$

where:

At	= Anatomy taxonomy
ASA	= Anatomical Structural Abstraction
ATA	= Anatomical Transformation Abstraction
Mk	= Metaknowledge (principles, rules, axioms)

Exploring the FMA

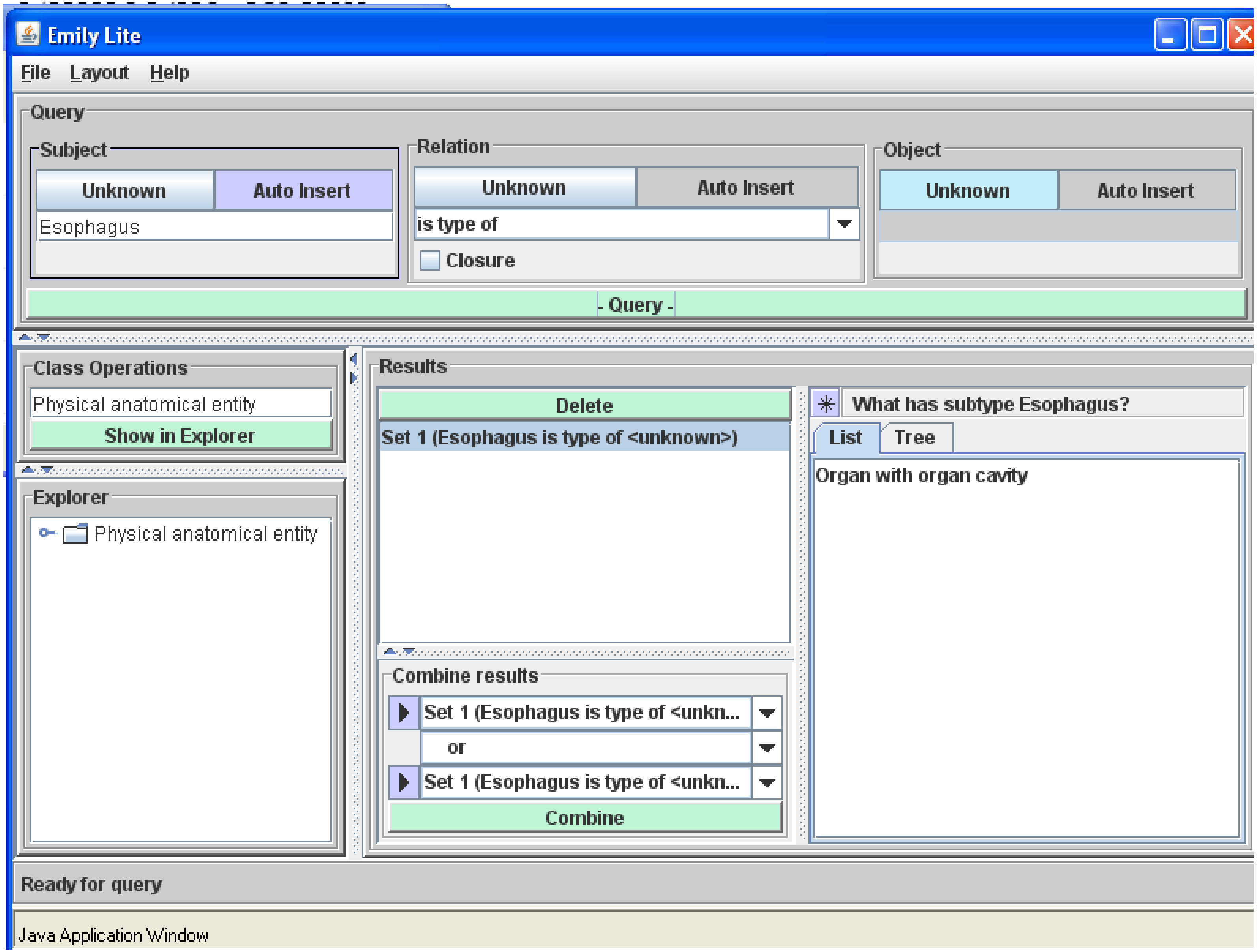
- The Foundational Model Explorer (FME)

http://fme.biostr.washington.edu/FME/index.html

allows browsing through the frames following links.

- Emily Lite

allows relational queries



Subject: Esophagus

Relation: is type of

□ Closure

Class Operations: Physical anatomical entity

Explorer: Physical anatomical entity

Set 1 (Esophagus is type of <unknown>)

What has subtype Esophagus?

Organ with organ cavity

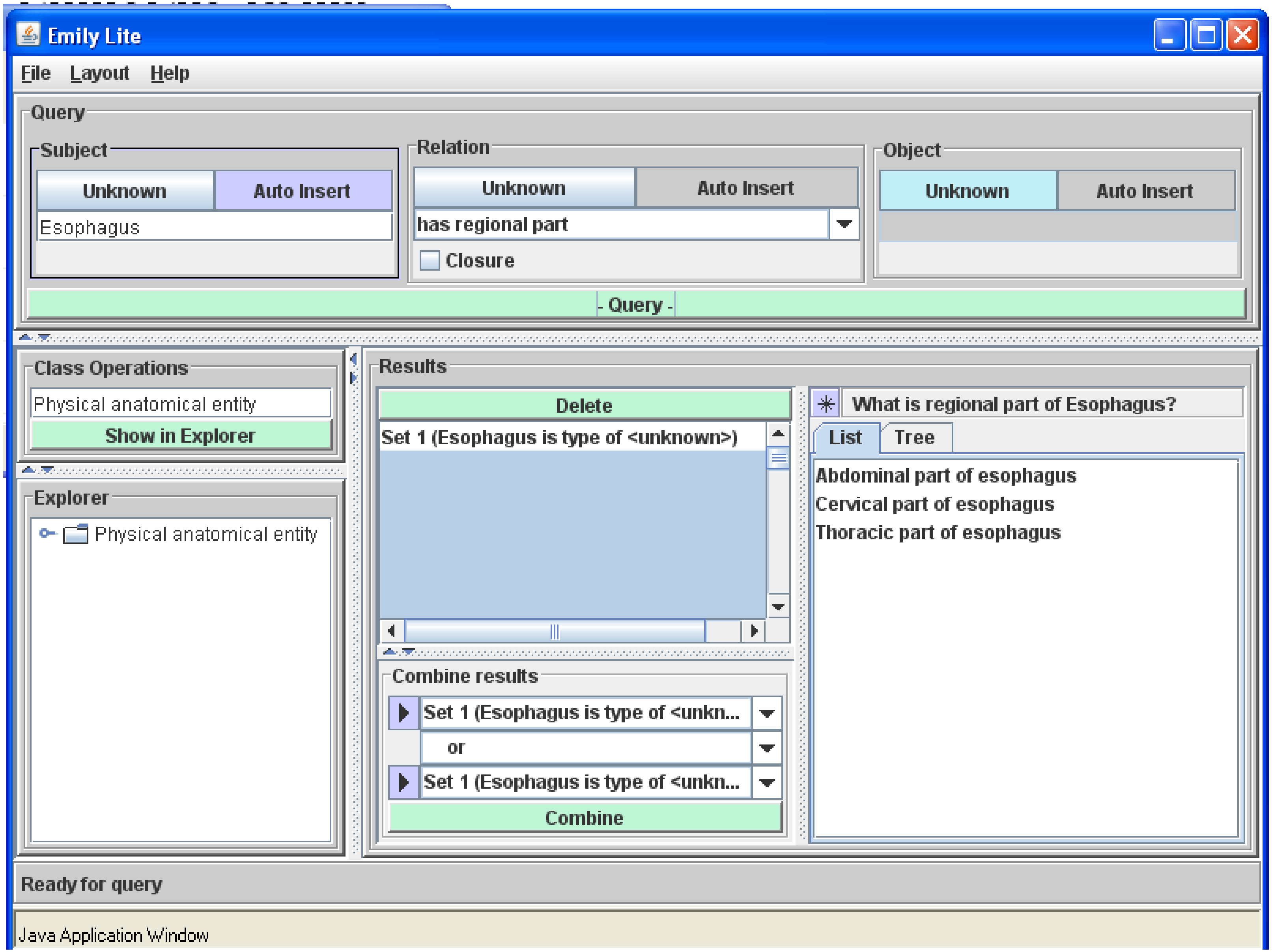
Set 1 (Esophagus is type of <unkn...

or

Set 1 (Esophagus is type of <unkn...

Ready for query

Java Application Window



Esophagus

has regional part

□ Closure

Physical anatomical entity

Physical anatomical entity

Set 1 (Esophagus is type of <unknown>)

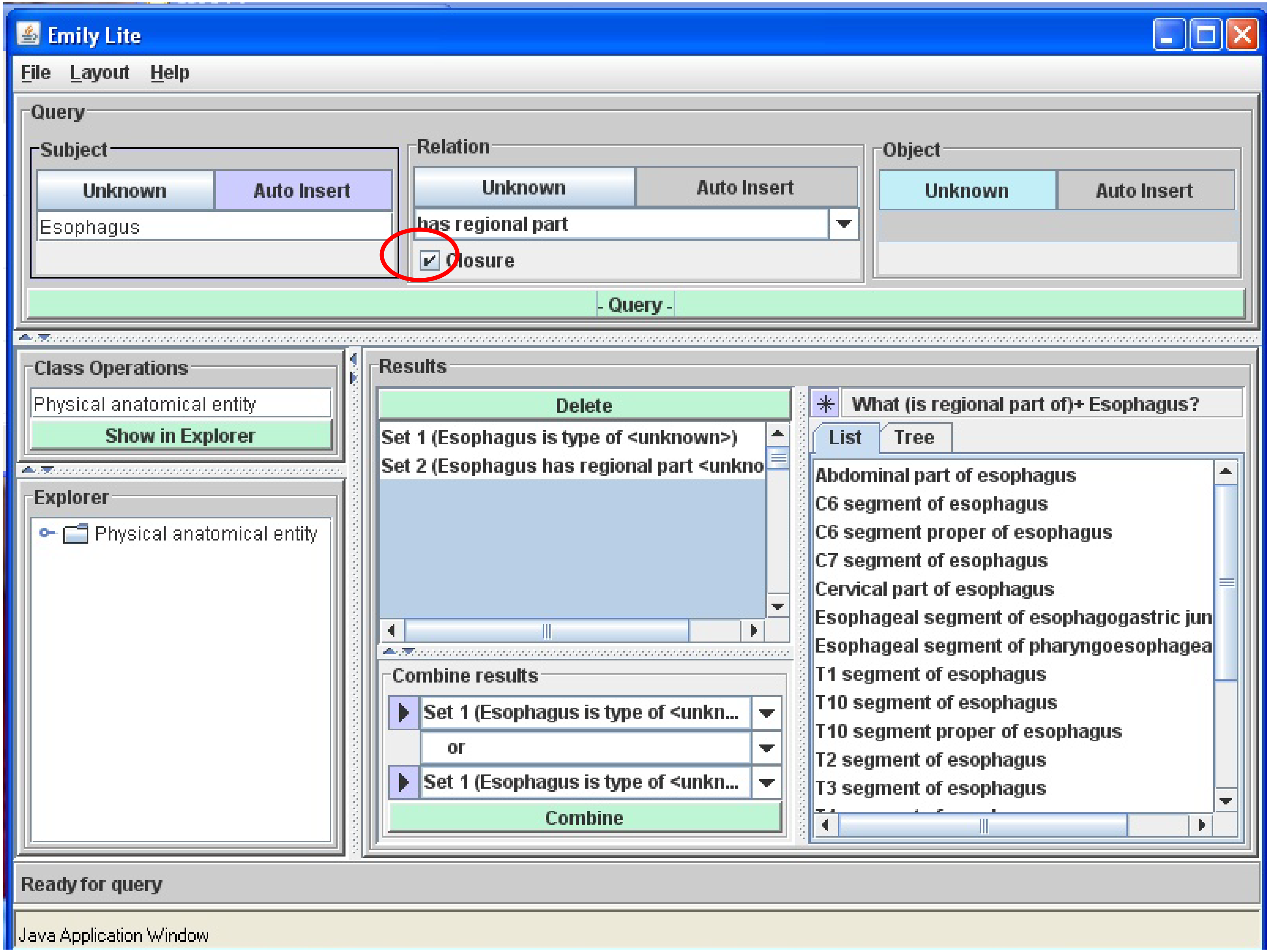
What is regional part of Esophagus?

Abdominal part of esophagus
Cervical part of esophagus
Thoracic part of esophagus

Set 1 (Esophagus is type of <unkn...
or
Set 1 (Esophagus is type of <unkn...

Ready for query

Java Application Window



Subject: Esophagus

Relation: has regional part

☑ Closure

Class Operations: Physical anatomical entity

Explorer: Physical anatomical entity

Results:

Set 1 (Esophagus is type of <unknown>)
Set 2 (Esophagus has regional part <unkno

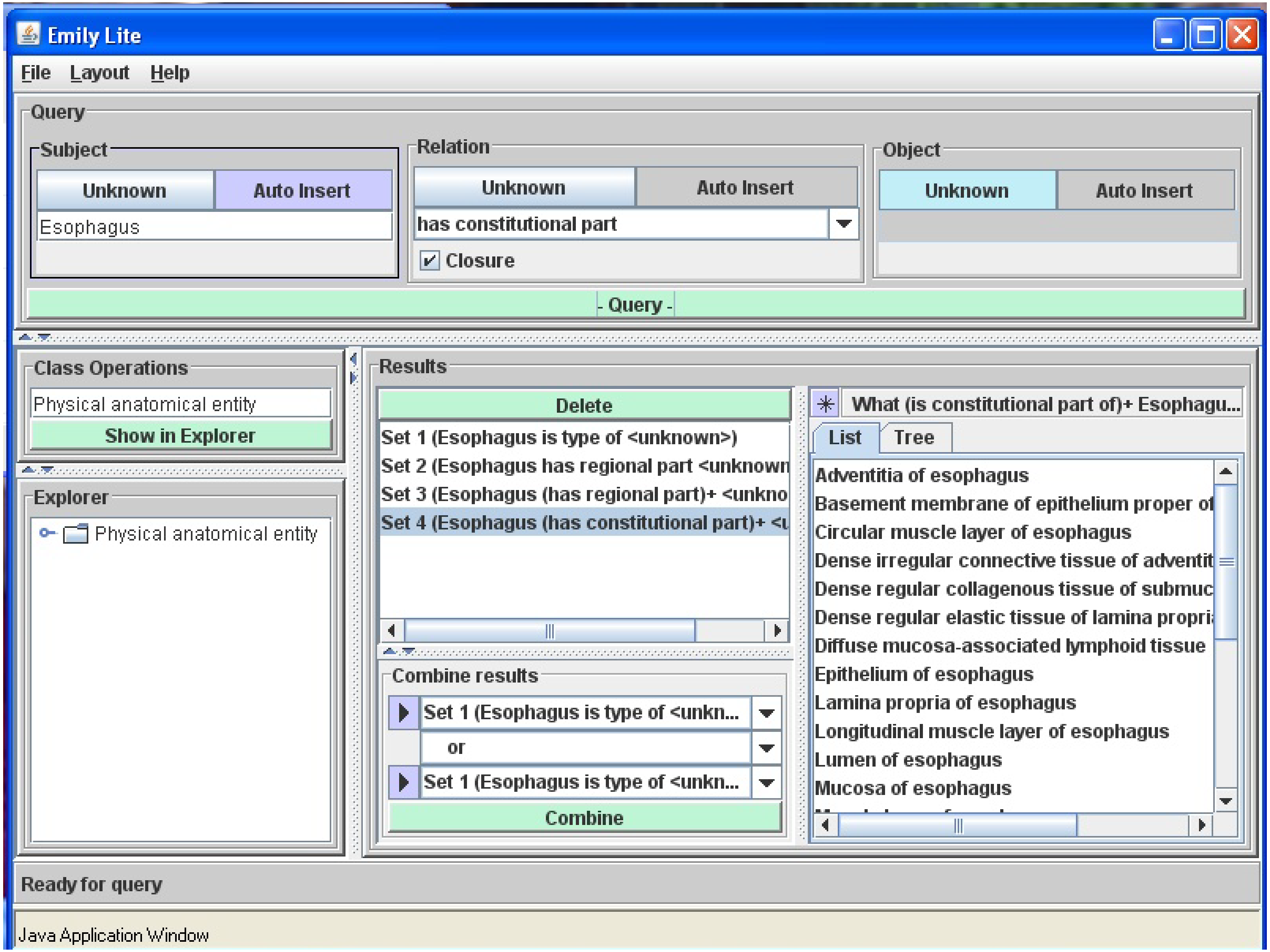
Combine results:

Set 1 (Esophagus is type of <unkn...
or
Set 1 (Esophagus is type of <unkn...

What (is regional part of)+ Esophagus?

Abdominal part of esophagus
C6 segment of esophagus
C6 segment proper of esophagus
C7 segment of esophagus
Cervical part of esophagus
Esophageal segment of esophagogastric jun
Esophageal segment of pharyngoesophagea
T1 segment of esophagus
T10 segment of esophagus
T10 segment proper of esophagus
T2 segment of esophagus
T3 segment of esophagus

Ready for query



Subject: Esophagus

Relation: has constitutional part

☑ Closure

Class Operations: Physical anatomical entity

Explorer: Physical anatomical entity

Results:

- Set 1 (Esophagus is type of <unknown>)
- Set 2 (Esophagus has regional part <unknown
- Set 3 (Esophagus (has regional part)+ <unkno
- Set 4 (Esophagus (has constitutional part)+ <u

What (is constitutional part of)+ Esophagu...

- Adventitia of esophagus
- Basement membrane of epithelium proper of
- Circular muscle layer of esophagus
- Dense irregular connective tissue of adventit
- Dense regular collagenous tissue of submuc
- Dense regular elastic tissue of lamina propri
- Diffuse mucosa-associated lymphoid tissue
- Epithelium of esophagus
- Lamina propria of esophagus
- Longitudinal muscle layer of esophagus
- Lumen of esophagus
- Mucosa of esophagus

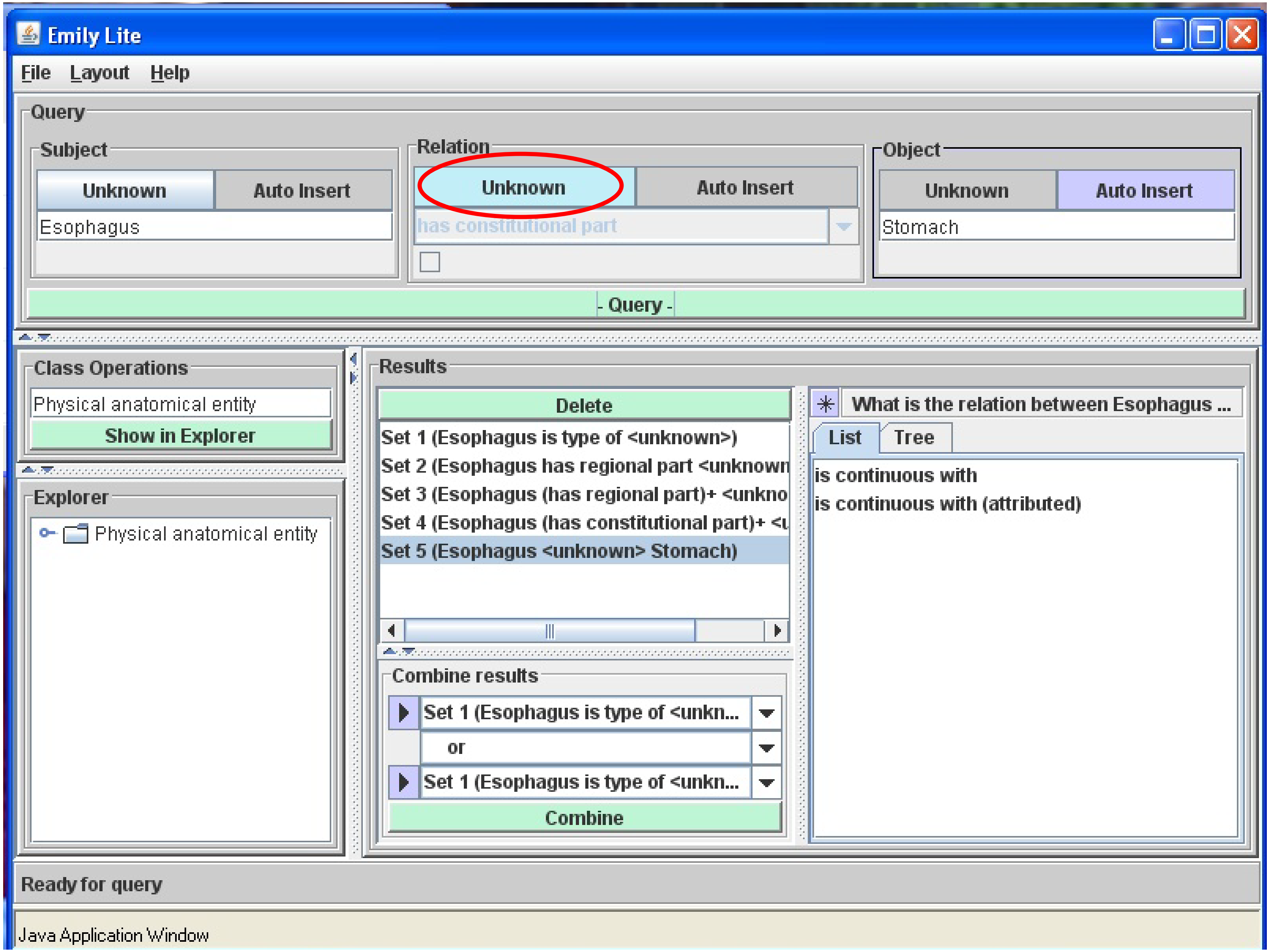
Combine results:

Set 1 (Esophagus is type of <unkn...

or

Set 1 (Esophagus is type of <unkn...

Ready for query



Subject: Esophagus

Relation: has constitutional part

□

Object: Stomach

Class Operations: Physical anatomical entity

Explorer: Physical anatomical entity

Results

Set 1 (Esophagus is type of <unknown>)
Set 2 (Esophagus has regional part <unknown
Set 3 (Esophagus (has regional part)+ <unkno
Set 4 (Esophagus (has constitutional part)+ <u
Set 5 (Esophagus <unknown> Stomach)

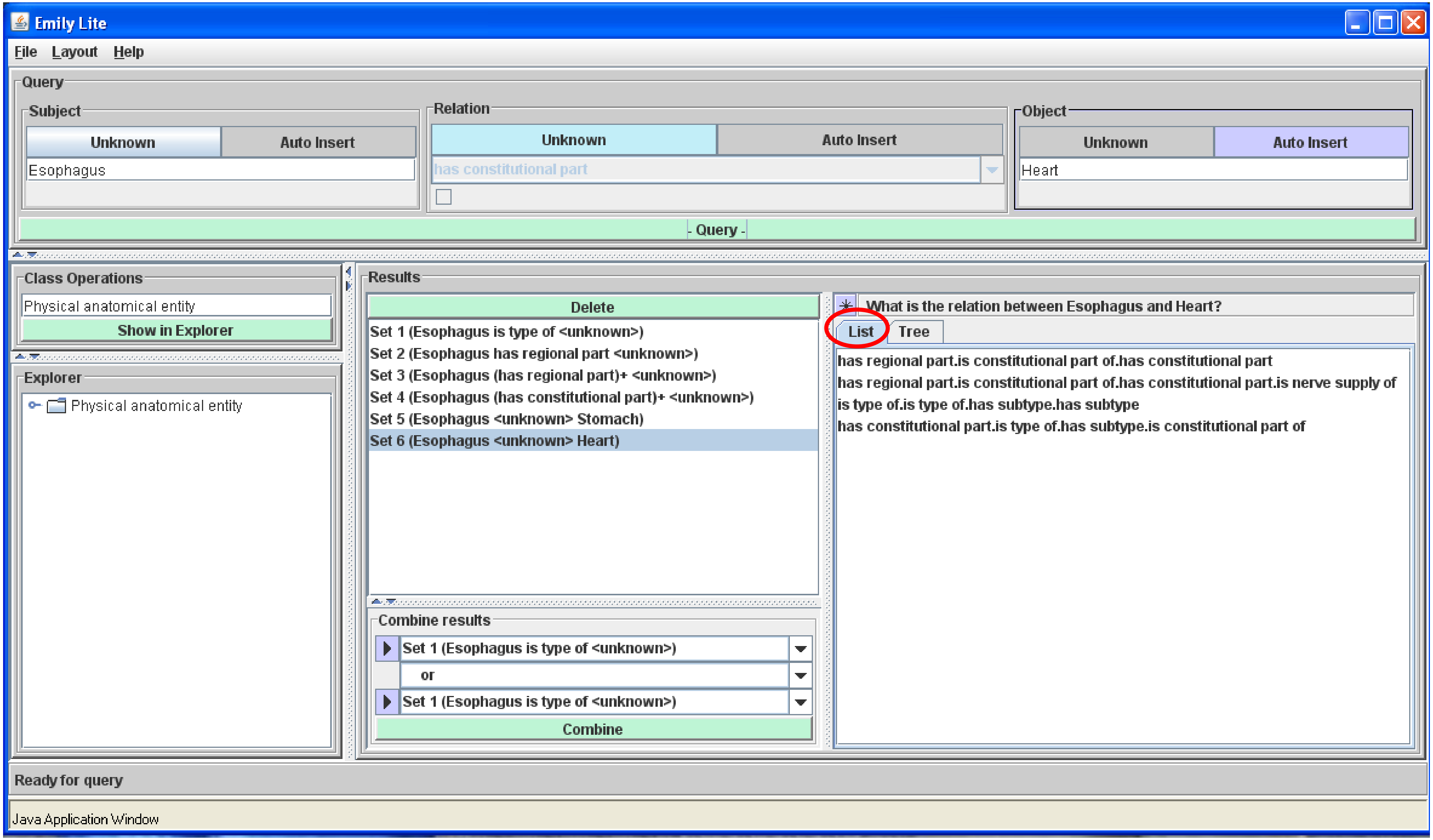
What is the relation between Esophagus ...

is continuous with
is continuous with (attributed)

Combine results

Set 1 (Esophagus is type of <unkn...
or
Set 1 (Esophagus is type of <unkn...

Ready for query



Subject: Esophagus

Relation: has constitutional part ☐

Object: Heart

Class Operations: Physical anatomical entity

Explorer: Physical anatomical entity

Results:

- Set 1 (Esophagus is type of <unknown>)
- Set 2 (Esophagus has regional part <unknown>)
- Set 3 (Esophagus (has regional part)+ <unknown>)
- Set 4 (Esophagus (has constitutional part)+ <unknown>)
- Set 5 (Esophagus <unknown> Stomach)
- Set 6 (Esophagus <unknown> Heart)

What is the relation between Esophagus and Heart?

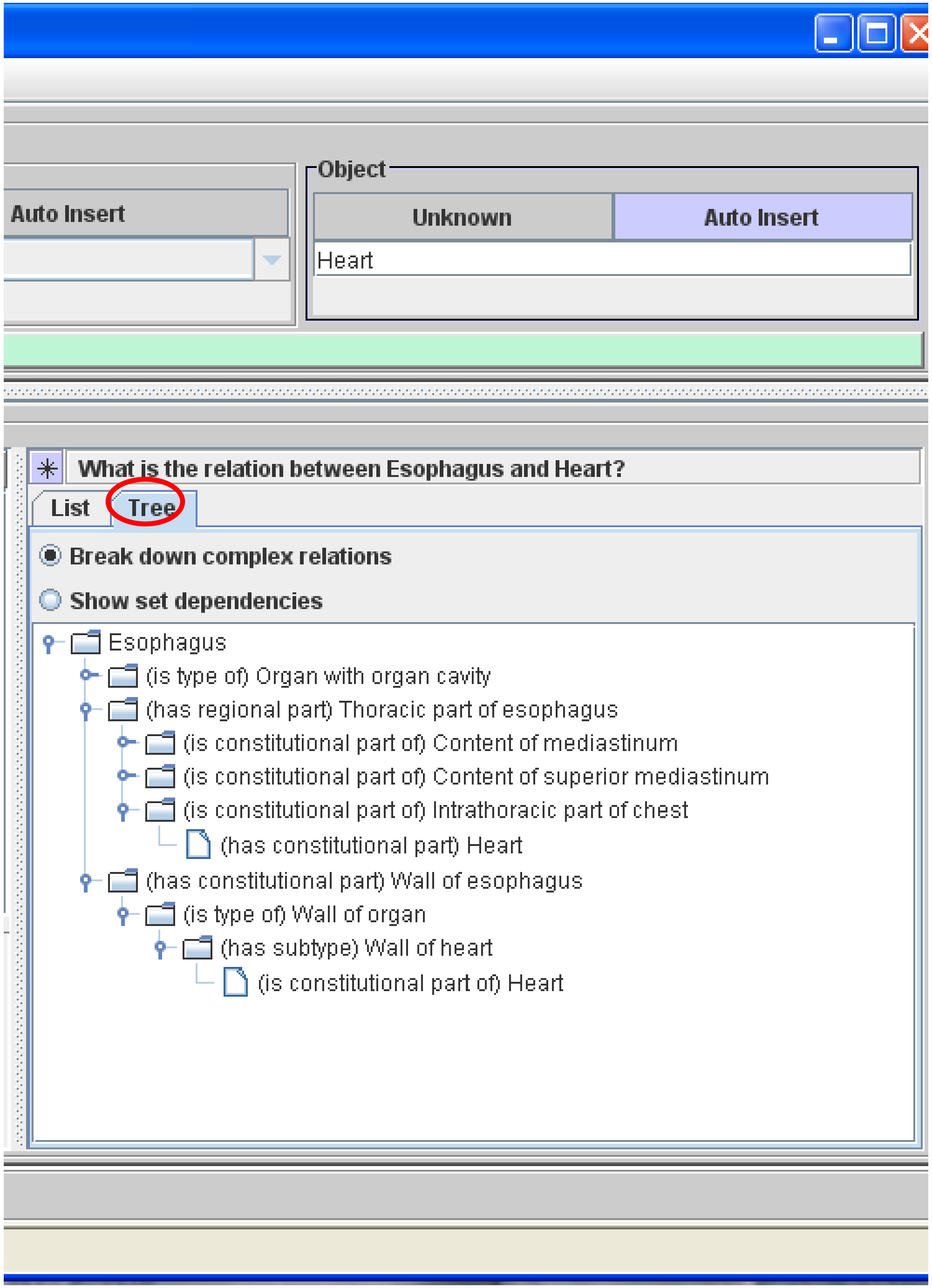
```
has regional part.is constitutional part of.has constitutional part
has regional part.is constitutional part of.has constitutional part.is nerve supply of
is type of.is type of.has subtype.has subtype
has constitutional part.is type of.has subtype.is constitutional part of
```

Combine results:

- Set 1 (Esophagus is type of <unknown>)
- or
- Set 1 (Esophagus is type of <unknown>)

Ready for query

Java Application Window



Object: Heart

What is the relation between Esophagus and Heart?

● Break down complex relations

○ Show set dependencies

- Esophagus
  - (is type of) Organ with organ cavity
  - (has regional part) Thoracic part of esophagus
    - (is constitutional part of) Content of mediastinum
    - (is constitutional part of) Content of superior mediastinum
    - (is constitutional part of) Intrathoracic part of chest
      - (has constitutional part) Heart
  - (has constitutional part) Wall of esophagus
    - (is type of) Wall of organ
      - (has subtype) Wall of heart
        - (is constitutional part of) Heart