

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

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?

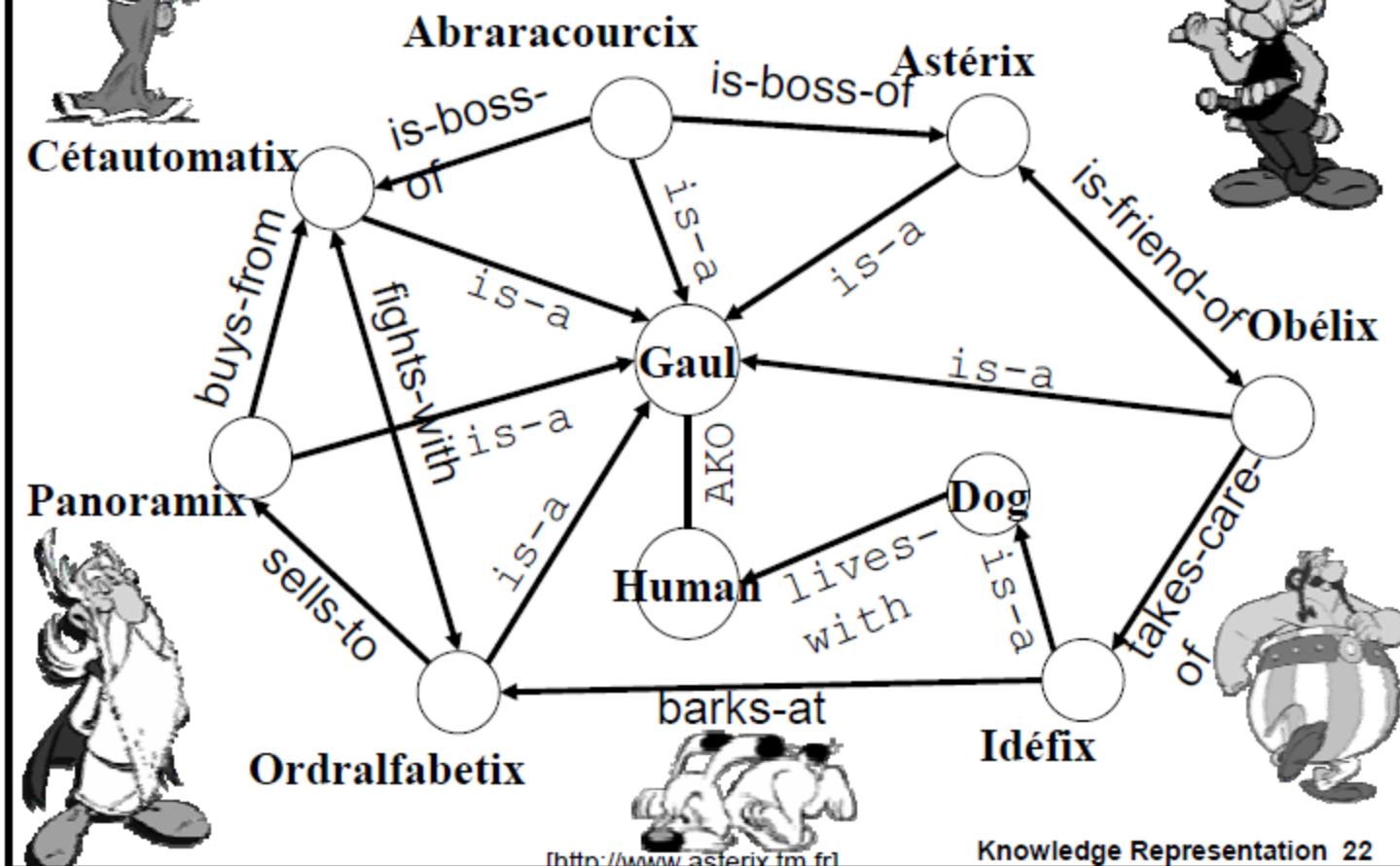
Advantages of Production Rules

- 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

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



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 datamodel for objects ("resources") and relations between them. These datamodels 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 (the OPB):

```
<?xml version="1.0"?>
```

```
<rdf:RDF
```

```
  xmlns:temporal="http://swrl.stanford.edu/ontologies/built-ins/3.3/temporal.owl#"
```

```
  xmlns:rdfs="http://www.w3.org/2000/01/rdf-schema#"
```

```
    (...many more namespaces...)
```

```
  xml:base="http://www.owl-ontologies.com/unnamed.owl">
```

```
    <owl:Ontology rdf:about="">
```

```
      <owl:imports rdf:resource="http://swrl.stanford.edu/ontologies/built-ins/3.3/query.owl"/>
```

```
      <owl:imports rdf:resource="http://www.w3.org/2003/11/swrl"/>
```

```
        (...many more imports...)
```

```
    </owl:Ontology>
```

```
    <owl:Class rdf:ID="Rotational_displacement">
```

```
      <rdfs:label rdf:datatype="http://www.w3.org/2001/XMLSchema#string"
```

```
        >Rotational displacement</rdfs:label>
```

```
      <rdfs:subClassOf>
```

```
        <owl:Class rdf:ID="Solid_displacement"/>
```

```
      </rdfs:subClassOf>
```

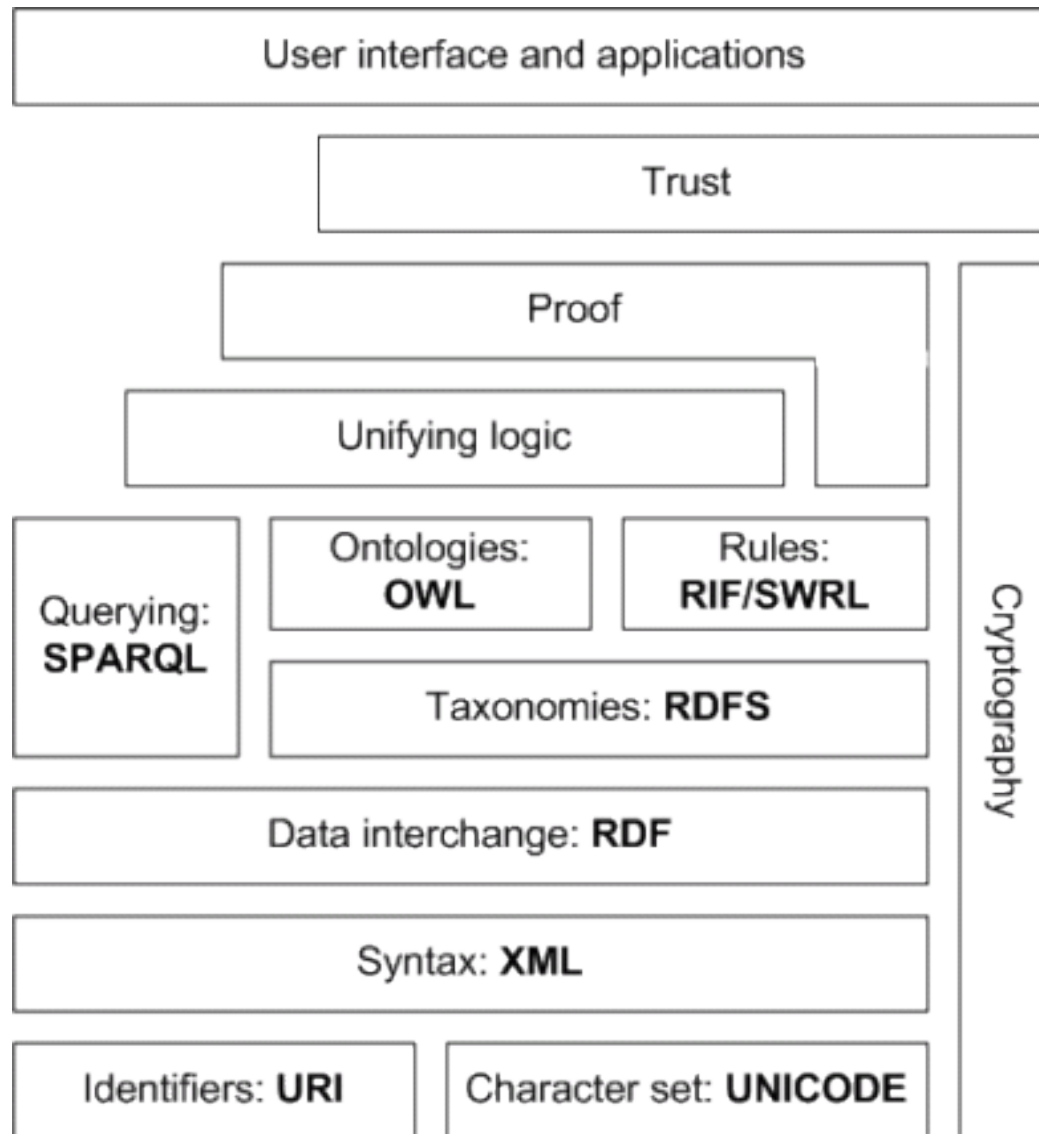
```
      <protege:subclassesDisjoint rdf:datatype="http://www.w3.org/2001/XMLSchema#bool"
```

```
        >true</protege:subclassesDisjoint>
```

```
      <owl:disjointWith>
```

```
        <owl:Class rdf:ID="Bending_displacement"/>
```

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

<i>Slot Name</i>	<i>Filler</i>
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)

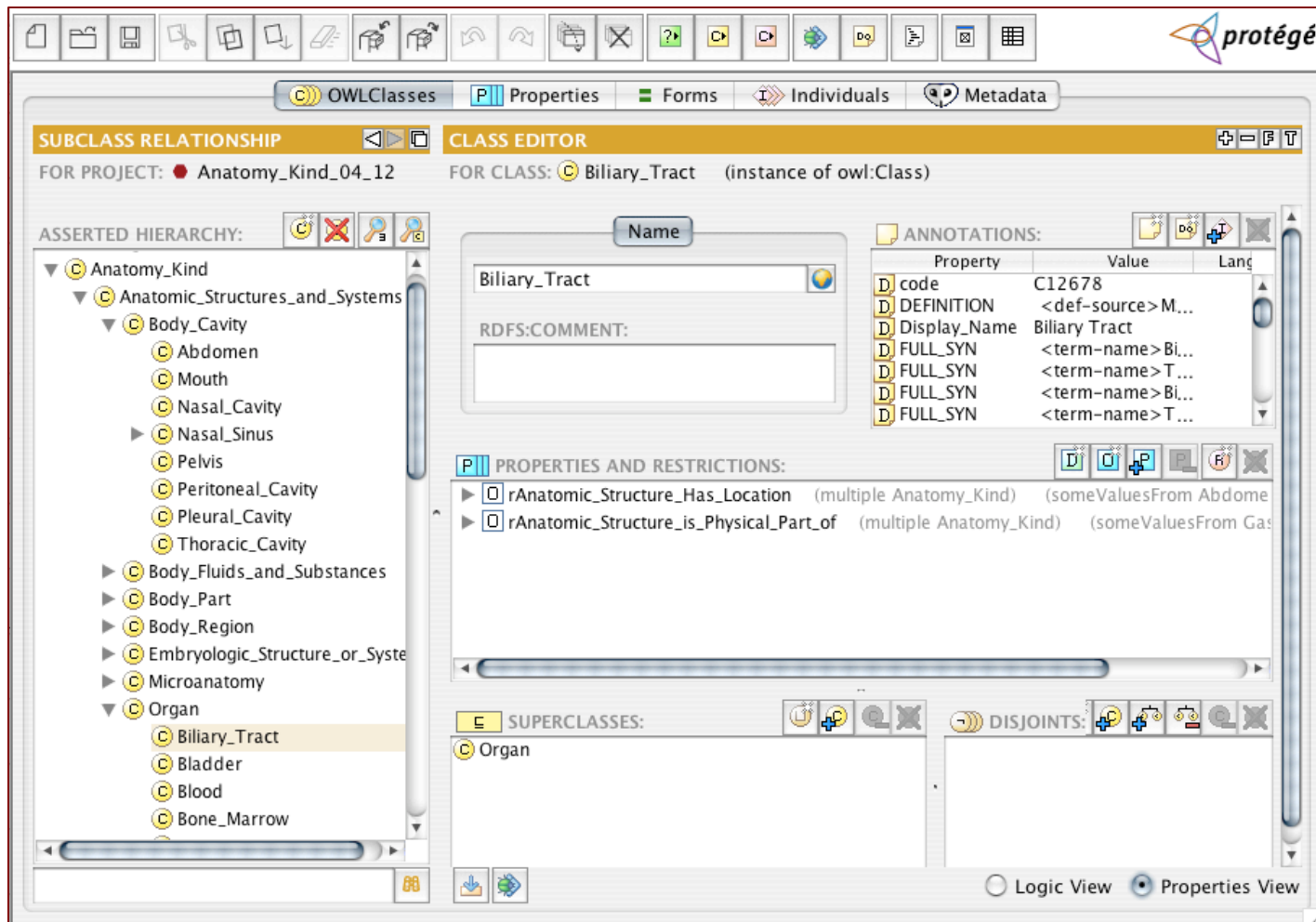
Ontology Tools

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

What is Protégé?

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

An Ontology Editor



A Knowledge-Acquisition Tool

The screenshot shows a software interface for managing Gene Ontology (GO) terms. The top menu bar includes 'Classes', 'Slots', 'Forms', 'Instances', and 'Queries'. The 'Classes' tab is active, showing a hierarchical tree of classes on the left. The 'molecular_function' class is selected. The main area displays the details for 'molecular_function (Gene_Ontology_Metaclass)'. The details are organized into several sections: 'Name' (0003674), 'Term' (molecular_function), 'Synonyms', 'InterPRO ID', 'Documentation', 'Secondary GOIds', 'EC Numbers', 'TC Numbers', 'SwissProt Keyword', 'Definition' (The action characteristic of a gene product), 'Definition Reference' (GO:curators), 'Associated Annotations', and 'Part-Of'. Each section has a 'V' (view) button and a 'C' (create) button. The 'Superclasses' section at the bottom shows 'Gene_Ontology_Entity' as the parent class.

Classes | **Slots** | **Forms** | **Instances** | **Queries**

Relationship | **Superclass** | **V** | **C** | **X**

molecular_function (Gene_Ontology_Metaclass) | **C** | **X**

Name | **Secondary GOIds** | **V** | **C** | **-** | **Definition**

0003674 | | The action characteristic of a gene product.

Term | **EC Numbers** | **V** | **C** | **-** | **Definition Reference**

molecular_function | | GO:curators

Synonyms | **V** | **C** | **-** | **Associated Annotations** | **C** | **+** | **-**

| |

InterPRO ID | **V** | **C** | **-** | **TC Numbers** | **V** | **C** | **-** | **Part-Of** | **V** | **C** | **+** | **-**

| |

Documentation | **SwissProt Keyword** | **V** | **C** | **-** | **Part-Of** | **V** | **C** | **+** | **-**

| |

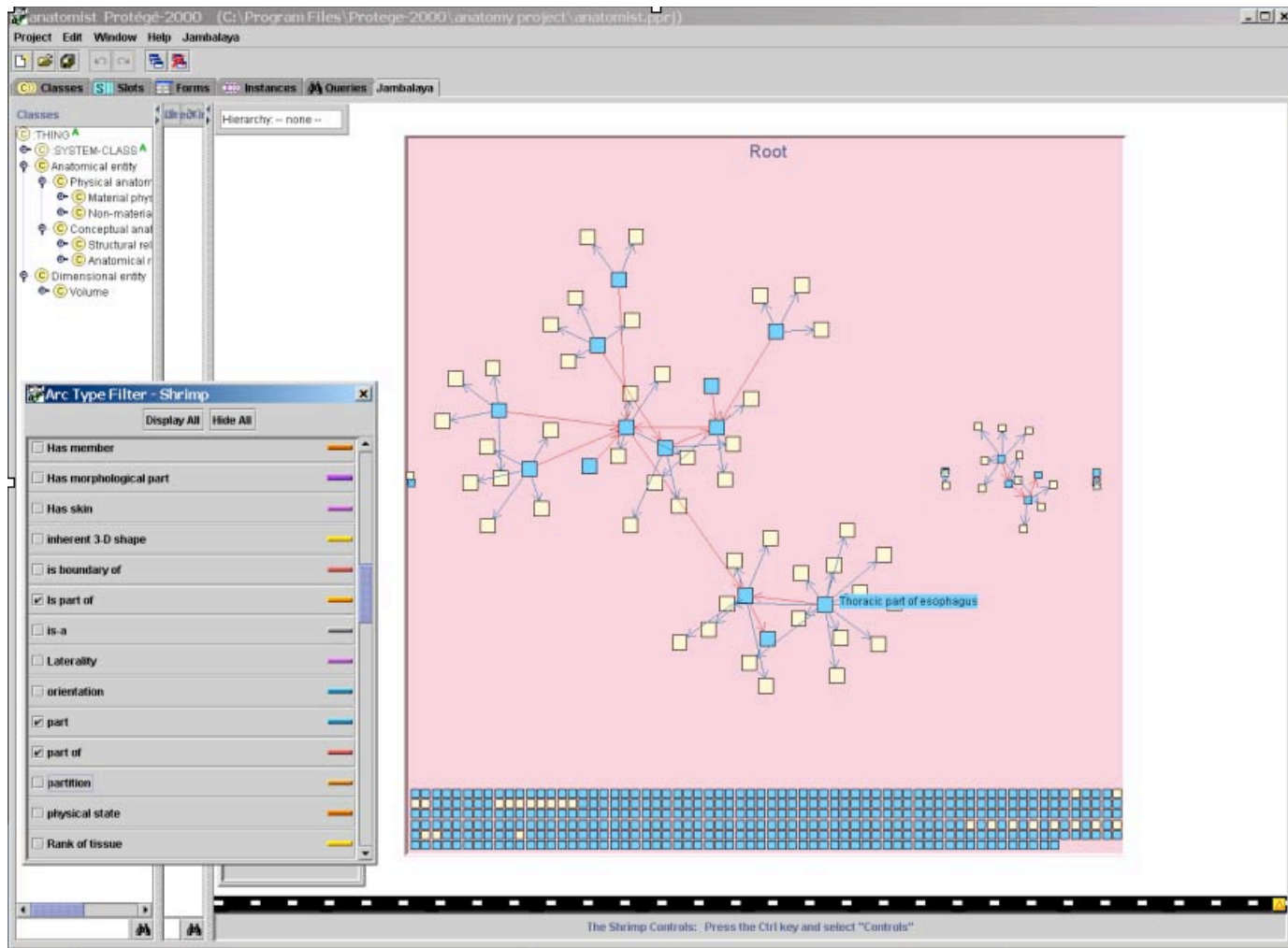
Superclasses | **+** | **-**

Gene_Ontology_Entity

A Platform for Other Applications

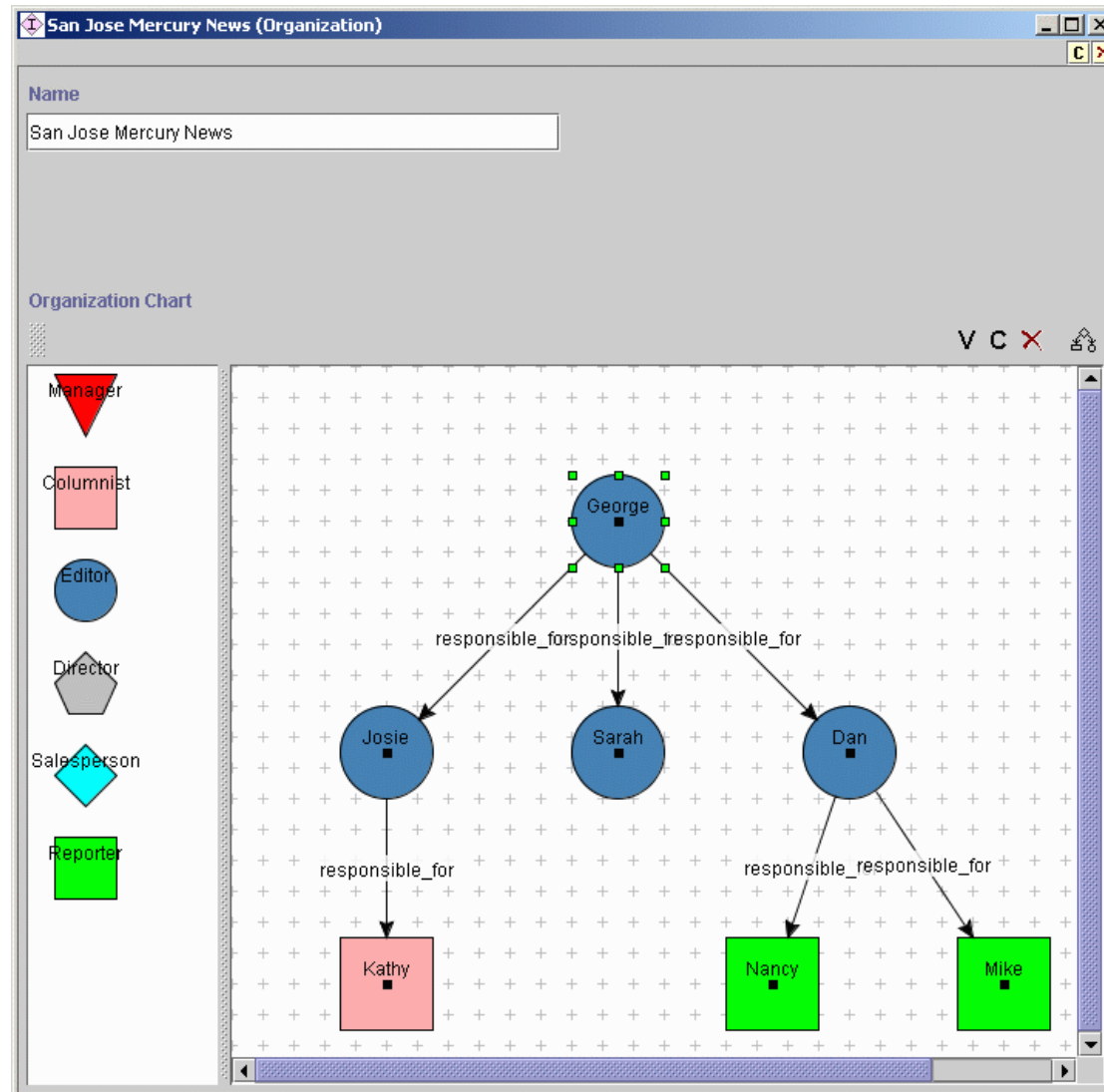
- A **Java API** that enables developers to write plugins for
 - Visualization systems
 - Inferencing systems
 - Scripting facilities
 - Import and export formats
 - User-interface features
 - Means of accessing external data sources
- About **60** plugins currently in the library (developed at Stanford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

Visualization: Jambalaya



Developed at University of Victoria

Graph Widget



Some Applications within SMI 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 (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

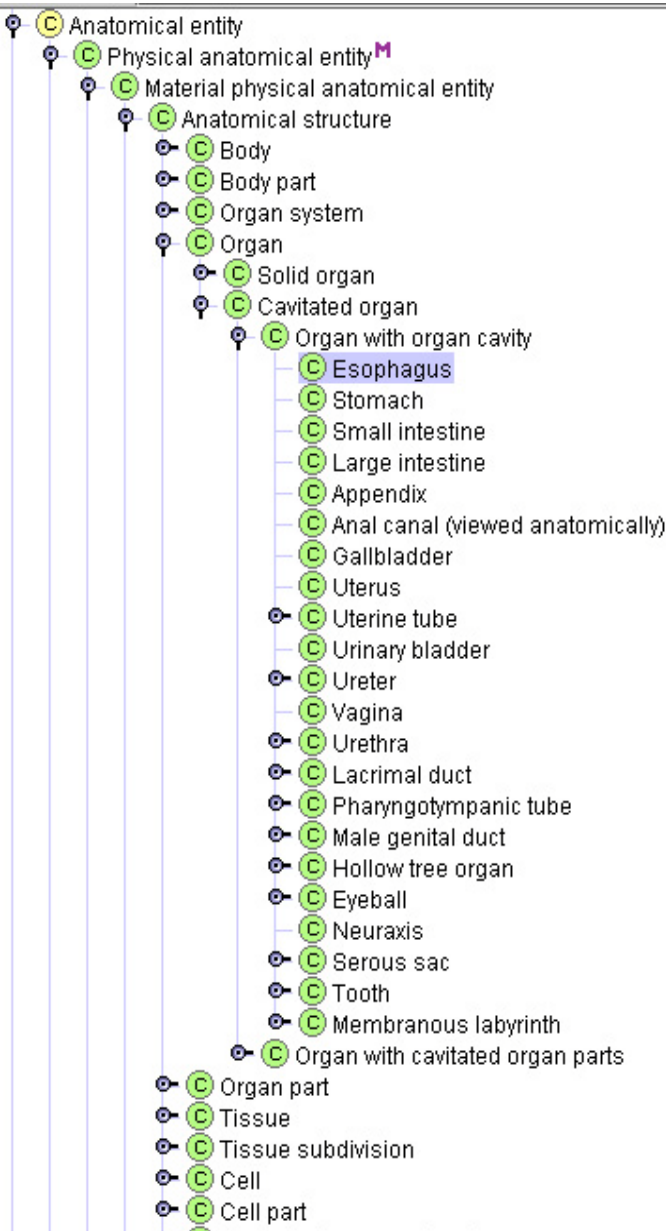
declare the principles

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



Preferred Name

Esophagus

LWDAID

7131

Synonyms

Gullet

Non-English Equivalents

Oesophagus

Definition

Organ with organ cavity which is continuous proximally with the pharynx and distally with the stomach.
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-esophagus

Attributed Part

related part	anatomical/arbitrary	shared/unshared	partition
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

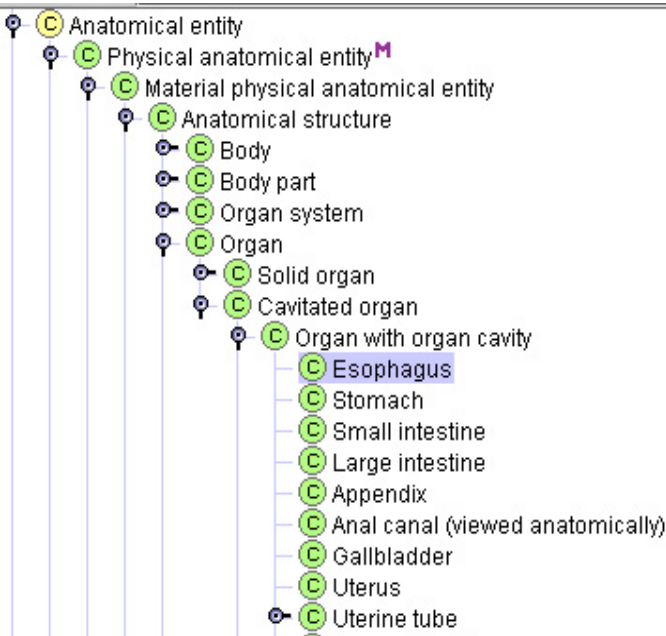
Attributed Continuous With

related object	coordinate	laterality
Pharynx	Superior	
Stomach	Inferior	

Orientation

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

Foundational Model of Anatomy Ontology



Anatomy Taxonomy (AT)
- defines the essence of the entity
through an inheritance hierarchy

Preferred Name

V

LWDAID

Esophagus

7131

Synonyms

V

C

X

S

Gullet

Non-English Equivalents

V

C

X

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 Examples: There is only one esophagus.

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+

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Part

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+

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Upper gastrointestinal tract

Foregut

Wall of esophagus

Lumen of esophagus

Cervical part of esophagus

Thoracic part of esophagus

Abdominal part of esophagus

Broncho-esophagus

Attributed Part

V

C

related part	anatomical/arbitrary	shared/unshared	partition
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

C

related object	coordinate	laterality
Pharynx	Superior	
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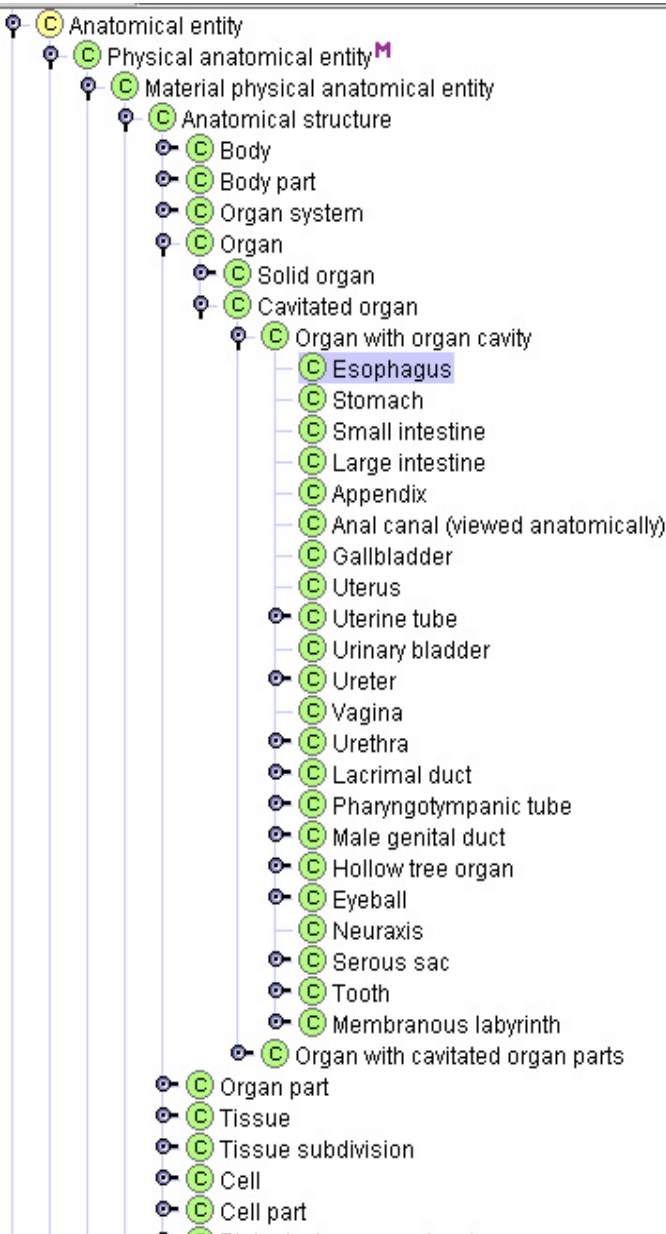
Orientation

V

C

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

Foundational Model of Anatomy Ontology



Preferred Name

V

LWDAID

Esophagus

7131

Synonyms

V

C

X

S

Non-English Equivalents

V

C

X

S

Gullet

Oesophagus

Anatomical Structural Abstraction (ASA)

- declares the spatio-structural properties of an

anatomical entity

Set of viscera

Wall of esophagus

Lumen of esophagus

Cervical part of esophagus

Thoracic part of esophagus

Abdominal part of esophagus

Broncho-esophagus

Part Of

V

+

-

Upper gastrointestinal tract

Foregut

Attributed Part

V

C

related part	anatomical/arbitrary	shared/unshared	partition
Wall of esophagus	Anatomical	Unshared	Partition 1
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Attributed Continuous With

V

C

related object	coordinate	laterality
Pharynx	Superior	
Stomach	Inferior	

Orientation

V

C

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

Foundational Model of Anatomy Ontology

Unifying theory of anatomy

High Level Scheme

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

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)

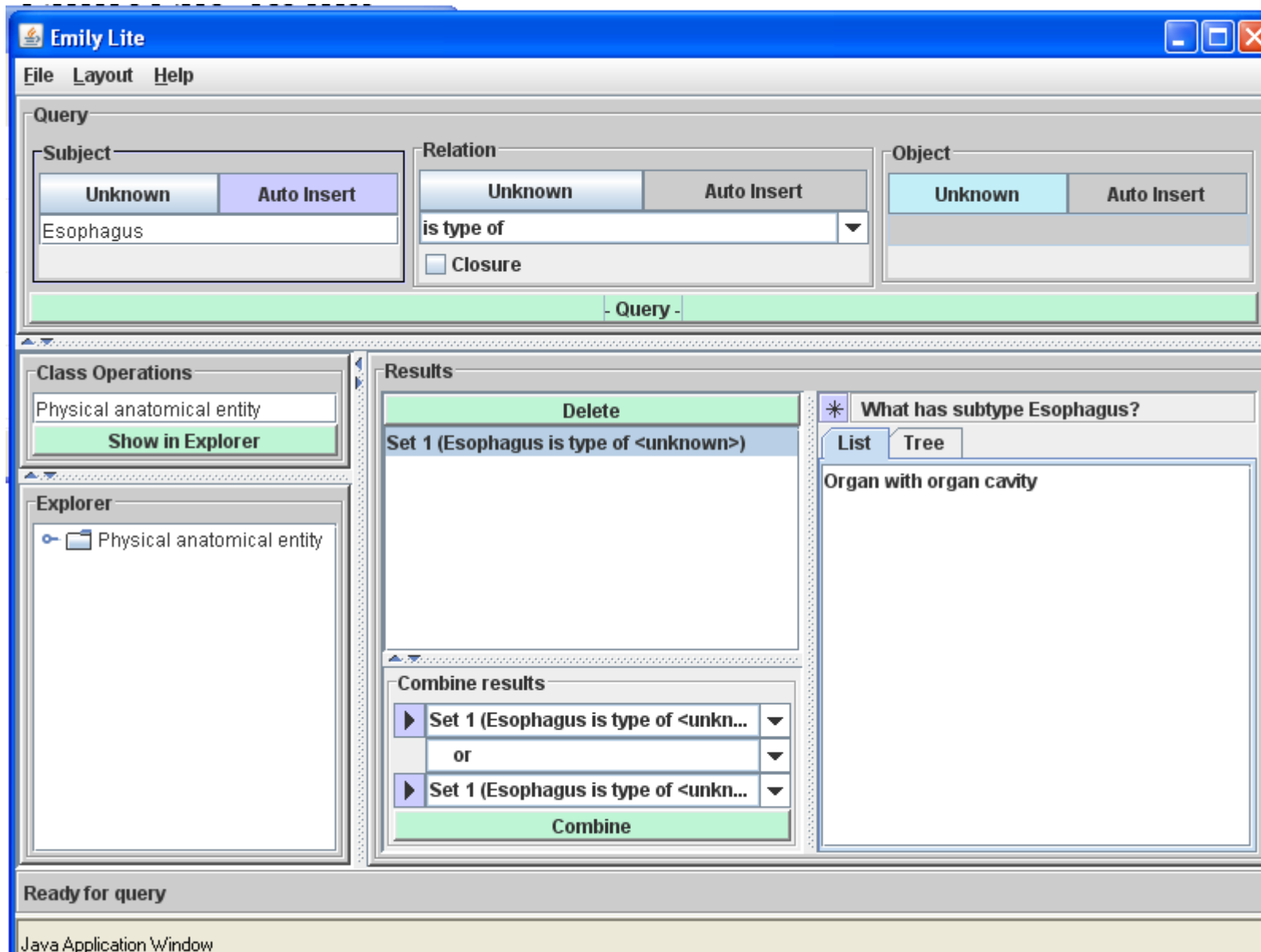
<http://fme.biostr.washington.edu:8089/FME/index.html>

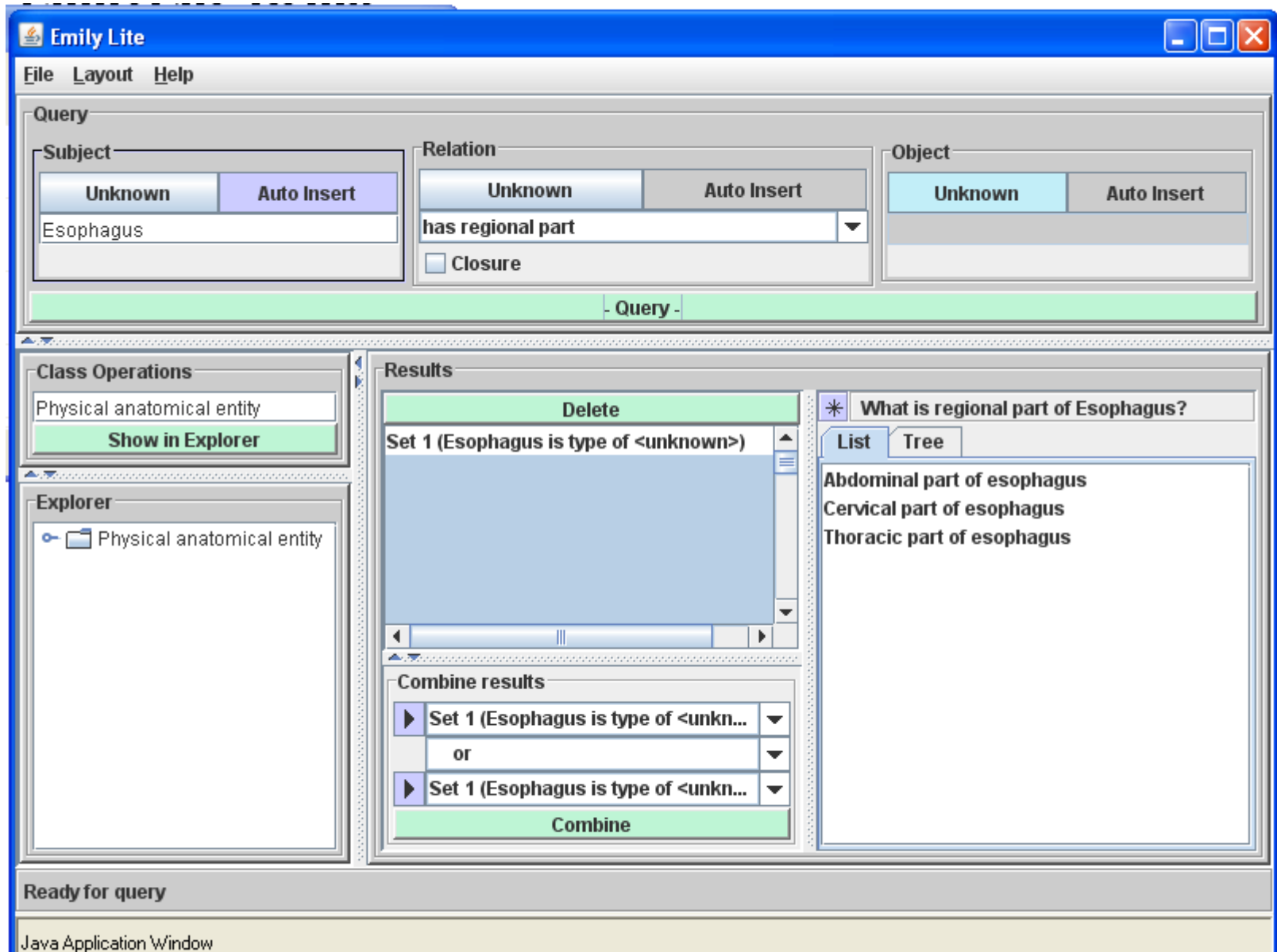
allows browsing through the frames following links.

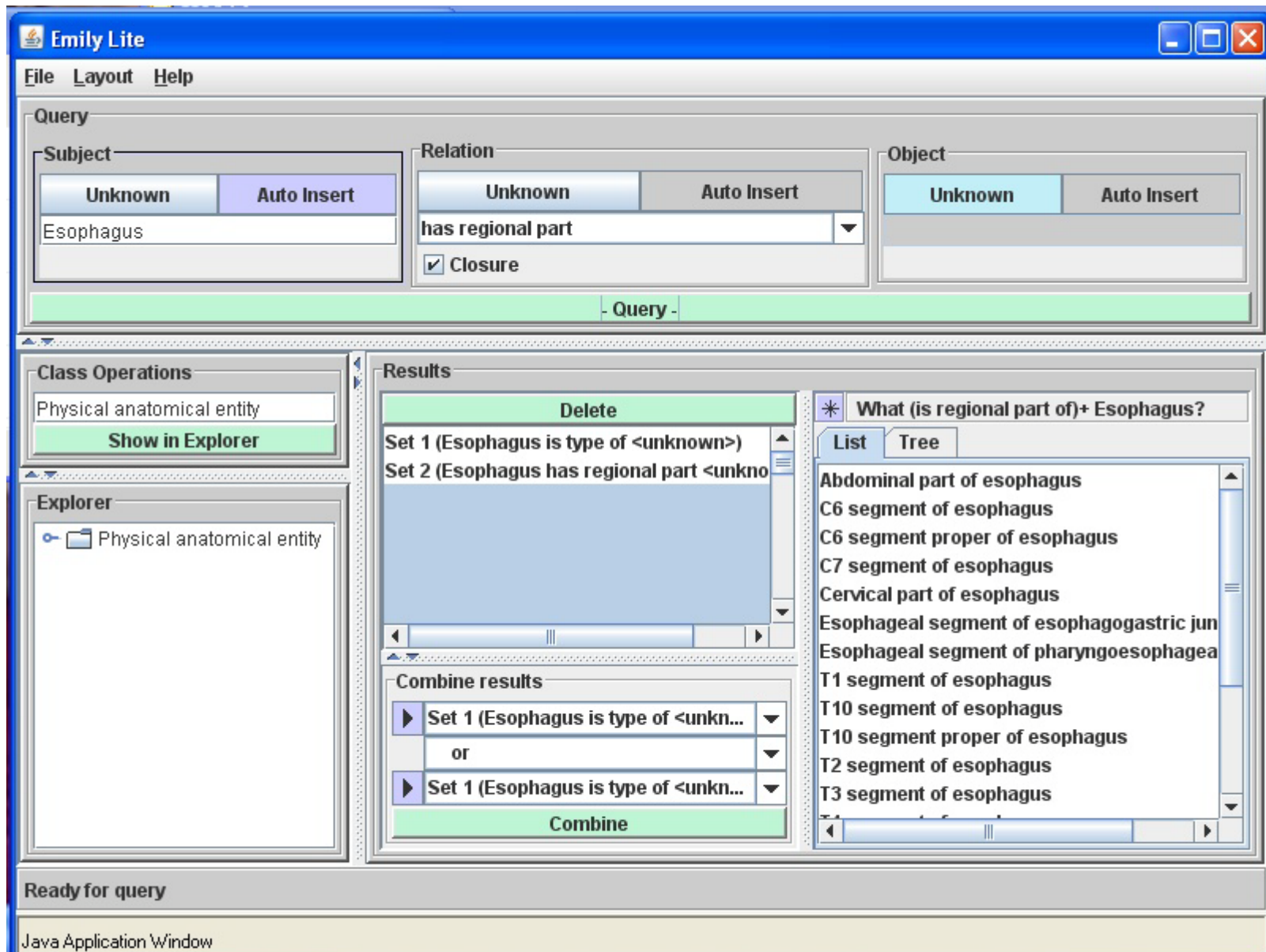
- Emily-Lite

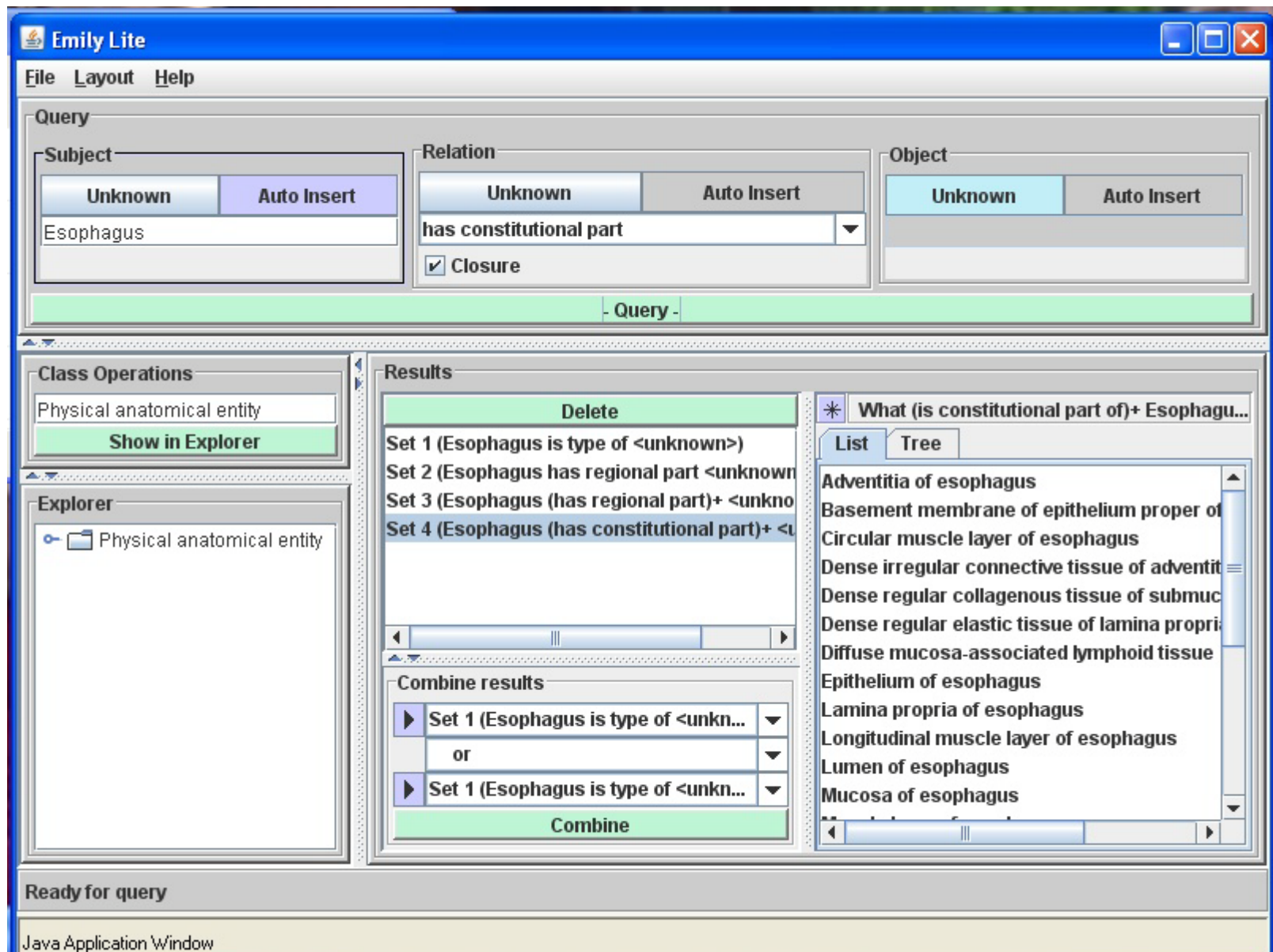
<http://fma.biostr.washington.edu/emilylite/>

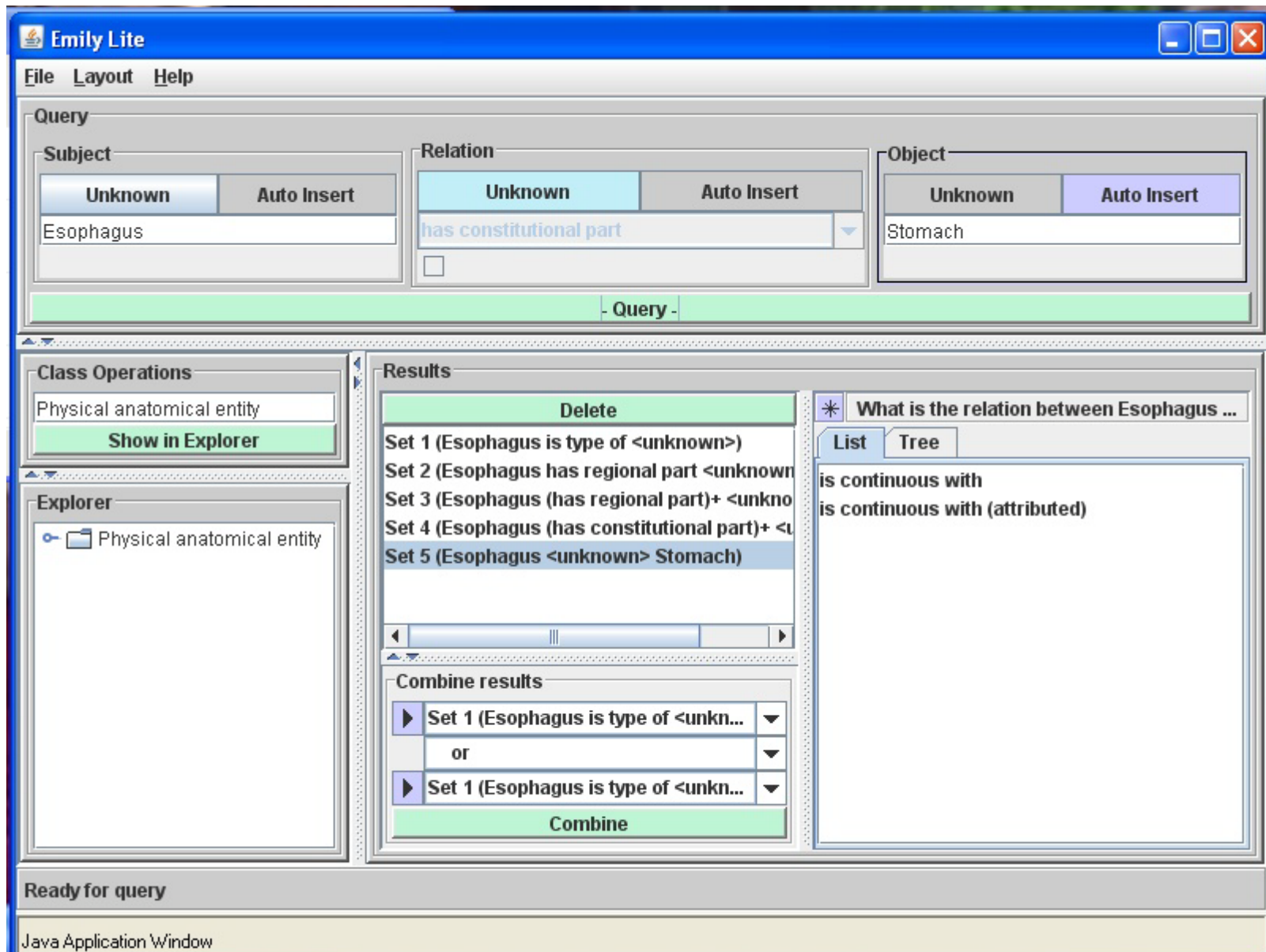
allows queries about entities and their relationships.











Emily Lite
File Layout Help

Query

Subject

Unknown Auto Insert

Esophagus

Relation

Unknown Auto Insert

has constitutional part

☐

Object

Unknown Auto Insert

Heart

- Query -

Class Operations

Physical anatomical entity

Show in Explorer

Explorer

Physical anatomical entity

Results

Delete

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)

Combine results

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

Combine

What is the relation between Esophagus and Heart?

List Tree

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

Ready for query

Java Application Window

Auto Insert

Object

Unknown

Auto Insert

Heart

* What is the relation between Esophagus and Heart?

List

Tree

☒ 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