Orbital Disease Ontology

Knowledge Based Systems

Marian Theodor[1] Mihai Gligor[2]

[1]Universitatea Tehnica Cluj-Napoca

May 24, 2023

Table of Contents

Table of Contents

Competency questions - Interogari

1. Care sunt bolile care au ca simptom X ?
2. Care sunt simptomele bolii X ?
3. Ce boala apare in locatia X ?
4. Este X o boala ?
5. Este X un simptom ?
6. Este X o locatie a unei boli ?
7. Are X baza materiala in Y ?
8. Este X adiacent lui Y ?
9. Este X o subcategorie a bolii Y?
10. Are boala X o locatie determinata?

Table of Contents

Reusing other ontologies

- Am folosit o ontologie cu tema asemanatoare cu cea dezvoltata de noi. Ontologia este preluata de pe site ul Bioportal (site asemnator cu Ontobee, dar am gasit ontologii mai usor de descarat in format owl) fiind accesibila la linkul acesta : `https://bioportal.bioontology.org/ontologies/FNS-H/?p=classes&conceptid=http`
- Dupa descarcarea ontologiei in format owl am folosit site-ul acesta pentru a genera un rdf folosind EasyRdf converter si am salvat un fisier .rdf (selectati optiunea all files)
  (Exista posibilitatea sa se poata incarca direct in Prodege cu extensia owl.
  (Link catre Prodege: https://protege.stanford.edu/)

Reusing other ontologies

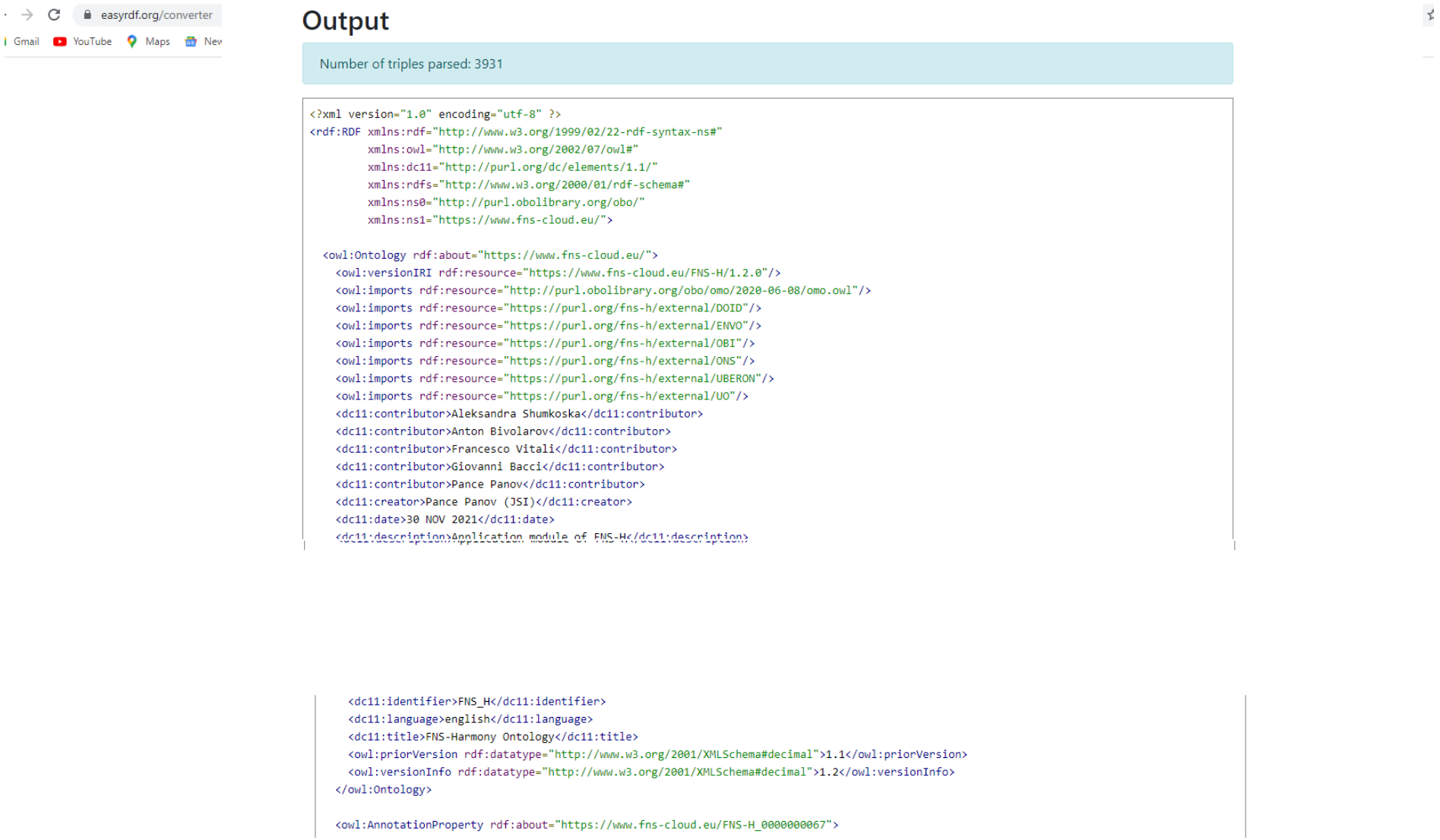


Figure: EasyRdf

Reusing other ontologies

In prodege am facut urmatoarele (poze se pot gasi la capitolul FRED):

1. Am incarcarat ontologia .rdf
2. Am selectat start reasoner
3. Am dat export
4. Am selectat clase si subclase
5. Am selectat locatia si am denumit fisierul Ontologie.owl
6. Am salvat in formatul owl/xml

Reusing other ontologies

Am incarcat fiserul salvat cu owl/xml in racer: L-am salvat in OWL cu comanda (save-kb “Ontoreusing.owl” :syntax :owl) Dupa am adaugat continutul la ontlogia noastra deja exista in owl
Cum arata taxologia :

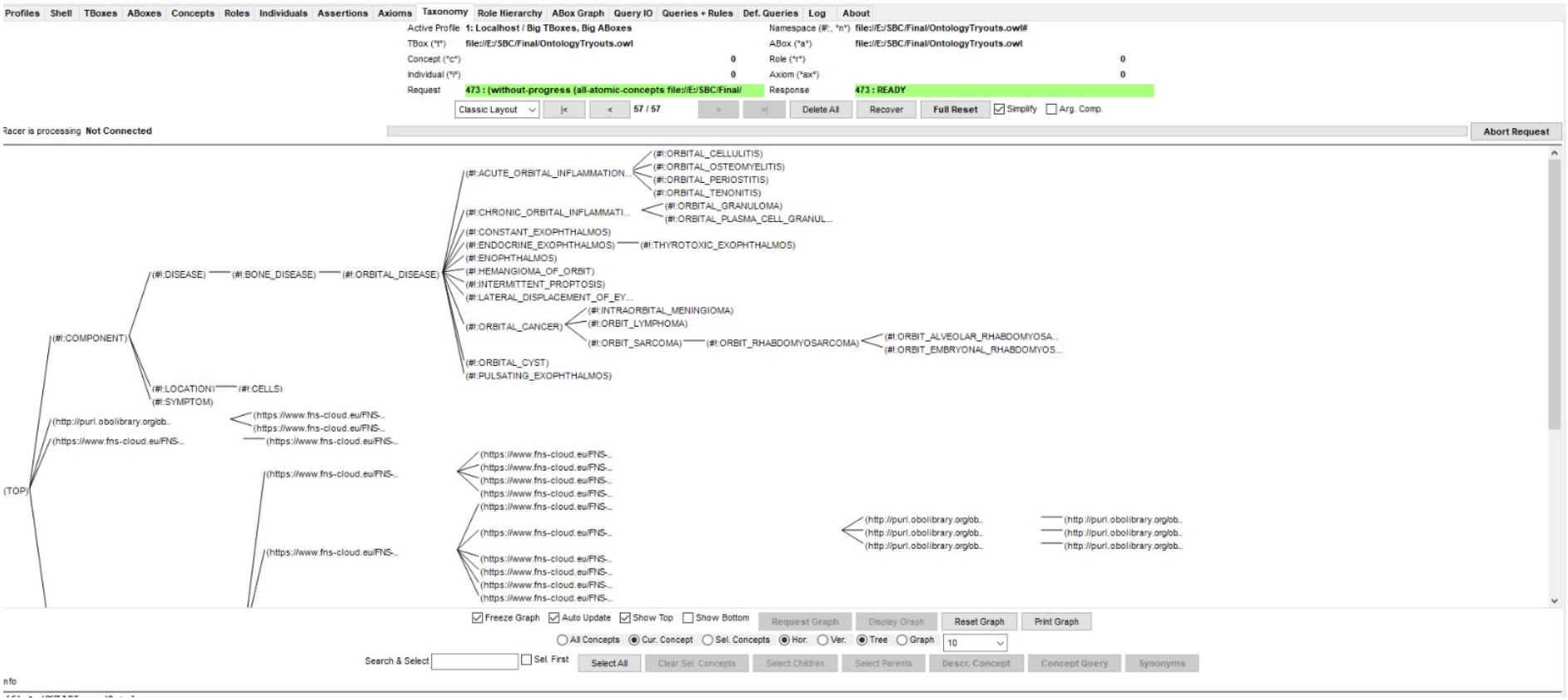


Figure: OntologyReusing

Table of Contents

Tboxes

TBox (Terminology Box) se refera la partea din ontologie ce defineste concepte, subconceptele si relatiile dintre ele. Reprezinta knowledge despre terminologia ontologiei si este de obicei folosita pentru a reprezenta structurile ierarhice dintre concepte (clase si subclase) si axiomele ce le guverneaza. De asemenea, Tbox-urile definesc caracteristicile si proprietatile entitatilor si ce categorii share-uiesc. Exemple de concepte pentru ontologii medicale includ : PatientDisease si Symtpthom. In plus, Cancer poate fi considerat subconcept al conceptului mai broad Disease. De asemenea, in Tbox sunt incluse rolurile (roles) si regulile (rules). Mai jos am atasat taxonomia ontologiei noastre. Pentru a vizualiza taxonomia ontologiei tale, slecteaza load , incarca fiserul .racer , iar la tabul de Taxonomy selecteaza tree 10.

Tboxes

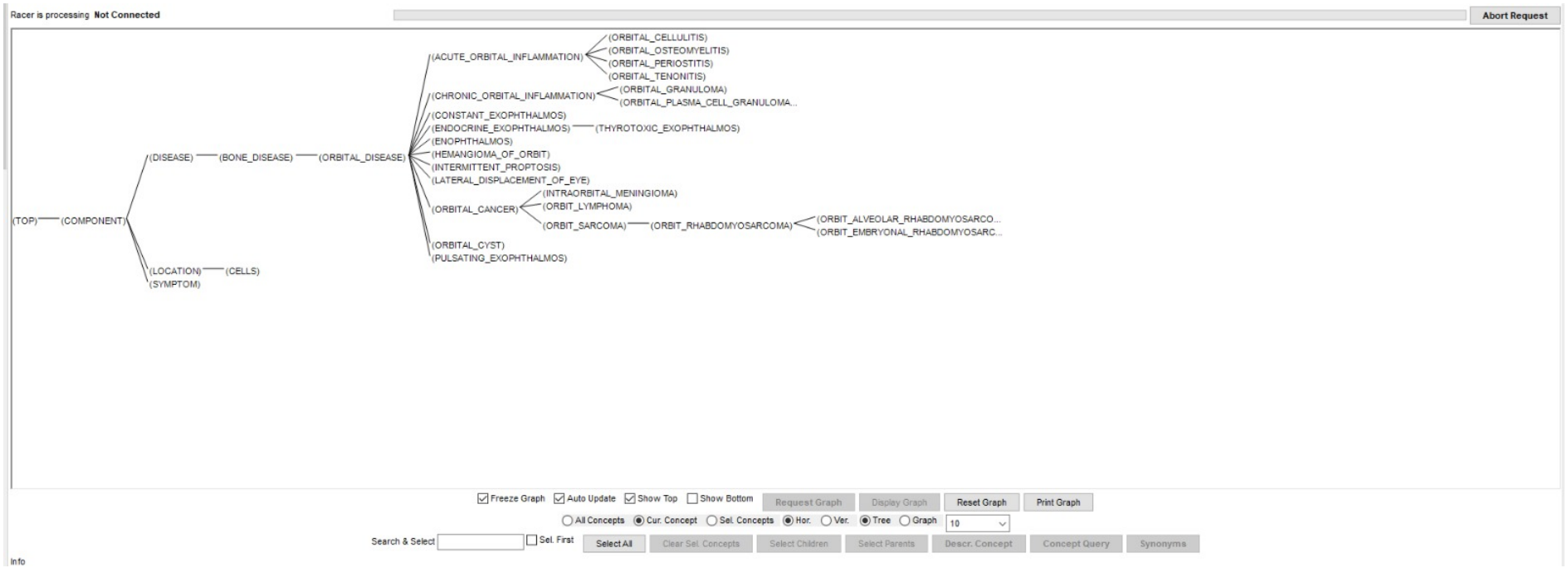


Figure: Tbox

Tboxes

```
(define-primitive-role has-symptom  :domain Disease  :range Symptom)
(define-primitive-role is-different  :domain Disease  :range Symptom)
(define-primitive-role has-location :domain Disease  :range Location)
(define-primitive-role adjacent-to  :domain Disease  :range Location)
(define-primitive-role has-material-basis-in  :domain Disease  :range Cells)
```

Figure: Roles

Tboxes

```
(implies Cells Location)

(implies Bone_disease Disease)
(implies Orbital_disease Bone_disease  )
(implies Endocrine_exophthalmos Orbital_disease )

(implies  Chronic_orbital_inflammation Orbital_disease)
(implies  Acute_orbital_inflammation Orbital_disease)
(implies  Orbital_cancer Orbital_disease)
(implies  Orbit_sarcoma Orbital_cancer)
(implies  Orbit_rhabdomyosarcoma Orbit_sarcoma )
```

Figure: Implies Concepts

Table of Contents

Aboxes

Abox contine informatii despre indivizi si attribute ale acestora .In general aici avem cuvinte cheie precum instance si related, attribute-filler, define-concrete-domain-attribute. La noi in ontologie indivizii sunt locatiile si simptomele ,iar toate bolile sunt definite ca si subconcepte ale conceptului disease.
Observatie: unii indivizi sunt considerati a fi si concepte de catre racer, apartinand ambelor categorii.

Aboxes

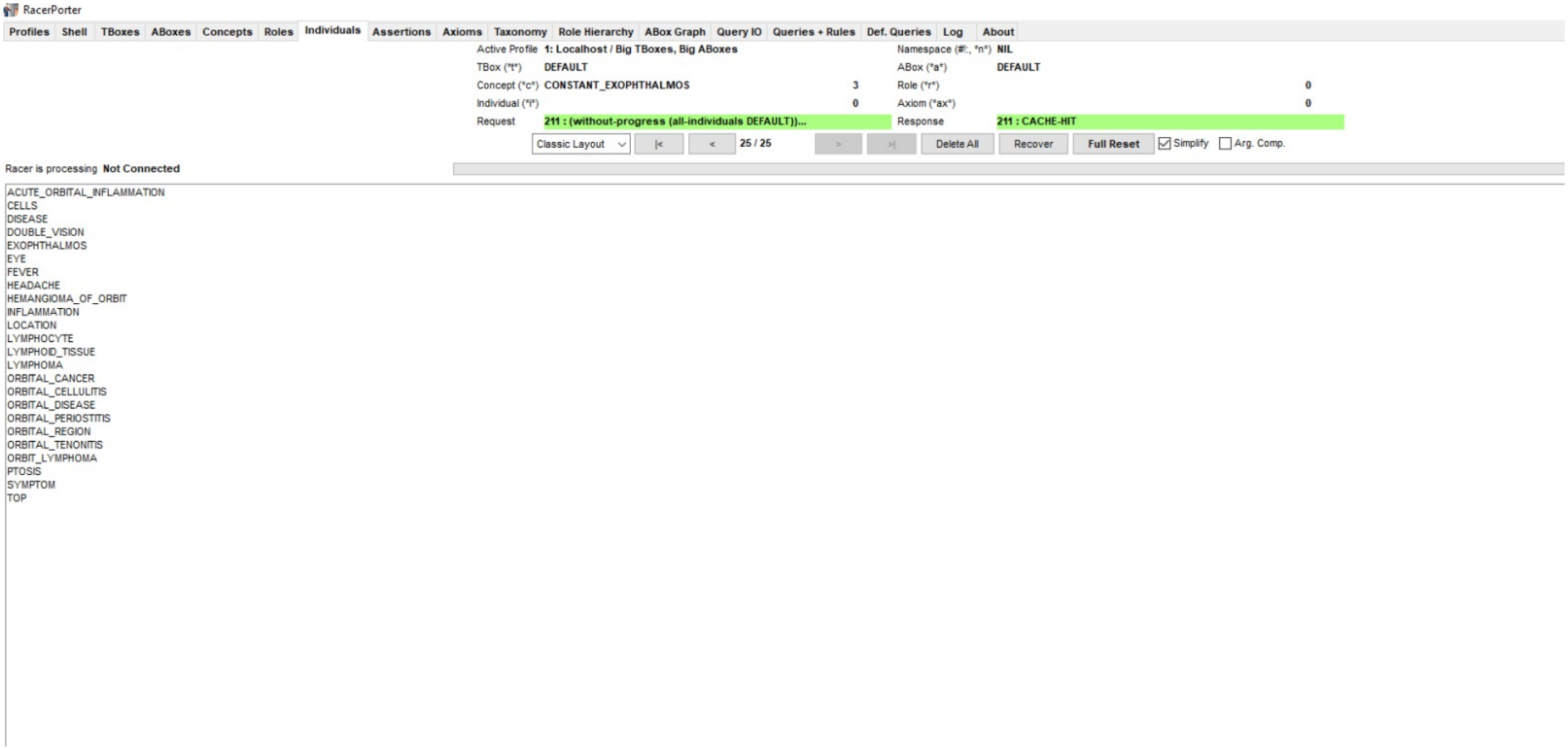


Figure: Indivizi

Aboxes

```
(related  Orbital_cellulitis fever has-symptom)
(related  Orbital_cellulitis ptosis has-symptom)
(related  Orbital_cellulitis headache has-symptom)
(related  Orbital_cellulitis double_vision has-symptom)
(related  Orbital_cellulitis inflammation has-symptom)
(related  Orbital_cancer ptosis has-symptom)
(related  Orbital_cancer exophthalmos has-symptom)
(related  Orbital_tenonitis inflammation has-symptom)
(related  Orbital_periostitis inflammation has-symptom)
(related  Acute_orbital_inflammation inflammation has-symptom)

(related  Hemangioma_of_orbit orbital_region has-location)
(related  Lymphoma lymphoid_tissue has-location)

(related  Orbital_disease eye  adjacent-to)

(related  Orbit_lymphoma lymphocyte  has-material-basis-in)
```

Figure: Related

Aboxes

```
(instance fever Symptom)
(instance ptosis Symptom)
(instance headache Symptom)
(instance double_vision Symptom)
(instance inflammation Symptom)
(instance exophthalmos Symptom)

(instance orbital_region Location)
(instance lymphoid_tissue Location)
(instance eye Location)
(instance lymphocyte Cells)
```

Table of Contents

Queries - Interogari Nrql

Interogari Nrql

1. Care sunt bolile care au ca simptom febra ?
   (retrieve (?x) (and (?x Disease) (?x fever has-symptom)))
2. Care sunt bolile care au ca simptom dureri de cap ?
   (retrieve (?x) (and (?x Disease) (?x headache has-symptom)))
3. Ce boala apare in tesutul limfoid?
   (retrieve (?x) (and (?x Disease) (?x lymphoid tissue has-location)))
4. Care sunt bolile care au ca simptom durerile de cap ?
   (retrieve (?x) (and (?x Disease) (?x inflammation has-symptom)))

Queries - Interogari Racer

Interogari Racer

1. Care sunt bolile care au ca simptom febra ? (individual-fillers fever (inv has-symptom))
2. Care sunt simptomele bolii orbital cellulitis ? (individual-fillers orbital cellulitis has-symptom)

Queries - Evaluare ontologie

Evaluare ontologie

1. Cod pentru determinarea si evaluarea diferitilor factori ai ontologiei

Examples

- (all-atomic-concepts)
- (all-individuals)
- (abox-consistent?)
- ((tbox-cyclic?)
- (tbox-coherent?)
- (realize-abox)
- (classify-tbox)
- (evaluate (length (all-individuals)))
- (evaluate (length (all-atomic-concepts)))
- (evaluate (length (all-roles)))

Queries - Rezultate

Rezultate query-uri

```
[1] ? (RACER-READ-FILE "D:/Faculta/SBC/OrbitalDiseaseOntology.racer")
(FULL-RESET) --> :OKAY-FULL-RESET
(DISABLE-NRQL-WARNINGS) --> :OKAY-WARNINGS-DISABLED
Redundant definition ORBITAL_CANCER for ORBIT_LYMPHOMA ignored.
(RETRIEVE (?X) (AND (?X DISEASE) (?X FEVER HAS-SYMPTOM))) --> (((?X ORBITAL_CELLULITIS)))
(INDIVIDUAL-FILLERS FEVER (INV HAS-SYMPTOM)) --> (ORBITAL_CELLULITIS)
(INDIVIDUAL-FILLERS ORBITAL_CELLULITIS HAS-SYMPTOM) --> (INFLAMMATION DOUBLE_VISION HEADACHE PTOSIS FEVER)
(RETRIEVE (?X) (AND (?X DISEASE) (?X HEADACHE HAS-SYMPTOM))) --> (((?X ORBITAL_CELLULITIS)))
(RETRIEVE (?X) (AND (?X DISEASE) (?X LYMPHOID_TISSUE HAS-LOCATION))) --> (((?X LYMPHOMA)))
(RETRIEVE (?X) (AND (?X DISEASE) (?X INFLAMMATION HAS-SYMPTOM))) --> (((?X ORBITAL_CELLULITIS)) ((?X ORBITAL_TENONITIS)) ((?X ORB»
ITAL_PERIOSTITIS)) ((?X ACUTE_ORBITAL_INFLAMMATION)))
(ALL-ATOMIC-CONCEPTS) --> (TOP BOTTOM ORBITAL_GRANULOMA CHRONIC_ORBITAL_INFLAMMATION ORBITAL_PLASMA_CELL_GRANULOMA ORBIT_SARCOMA »
ENDOCRINE_EXOPHTHALMOS BONE_DISEASE ORBIT_EMBRYONAL_RHABDOMYOSARCOMA THYROTOXIC_EXOPHTHALMOS HEMANGIOMA_OF_ORBIT ORBIT_RHABDOMYOS»
ARCOMA LATERAL_DISPLACEMENT_OF_EYE ORBIT_ALVEOLAR_RHABDOMYOSARCOMA CONSTANT_EXOPHTHALMOS ORBIT_LYMPHOMA SYMPTOM ENOPHTHALMOS ORBI»
TAL_CANCER PULSATING_EXOPHTHALMOS LOCATION INTRAORBITAL_MENINGIOMA DISEASE ORBITAL_CYST ORBITAL_PERIOSTITIS ORBITAL_DISEASE ORBIT»
AL_OSTEOMYELITIS INTERMITTENT_PROPTOSIS ORBITAL_TENONITIS CELLS COMPONENT ACUTE_ORBITAL_INFLAMMATION ORBITAL_CELLULITIS)
(ALL-INDIVIDUALS) --> (ORBIT_LYMPHOMA LYMPHOCYTE ORBITAL_DISEASE EYE LYMPHOMA LYMPHOID_TISSUE HEMANGIOMA_OF_ORBIT ORBITAL_REGION »
ACUTE_ORBITAL_INFLAMMATION INFLAMMATION ORBITAL_PERIOSTITIS ORBITAL_TENONITIS ORBITAL_CANCER EXOPHTHALMOS PTOSIS ORBITAL_CELLULIT»
IS DOUBLE_VISION HEADACHE FEVER CELLS LOCATION TOP SYMPTOM DISEASE)
(ABOX-CONSISTENT?) --> T
(TBOX-CYCLIC?) --> NIL
(TBOX-COHERENT?) --> T
(EVALUATE (LENGTH (ALL-INDIVIDUALS))) --> 24
(EVALUATE (LENGTH (ALL-ATOMIC-CONCEPTS))) --> 33
(EVALUATE (LENGTH (ALL-ROLES))) --> 18
(EVALUATE (LENGTH (ALL-RULES))) --> 0

[1] > :OKAY
```

Figure: Rezultate query

Design Patterns

Ontology design patterns sunt soluții de modelare care rezolvă diverse probleme în proiectarea ontologiei. Există șase tipuri de astfel de modele: structurale, corespondență, conținut, raționament, prezentare și lexico-sintactice. In ontologia noastra, am folosit doua patternuri structurale un pattern de continut si unul de presentare :

Design Patterns - Pattern-uri structurale

N-arry relation design pattern. Pentru conceptul de Disease am definit urmatoarele relatii.

- (define-primitive-role has-symptom :domain Disease :range Symptom)
- (define-primitive-role has-location :domain Disease :range Location)
- (define-primitive-role adjacent-to :domain Disease :range Location)
- (define-primitive-role has-material-basis-in :domain Disease :range Cells)

Partition design pattern. Pentru demonstrarea acestui pattern am considerat un concept numit Component care este impartit in subconcepte diferite intre ele care compun ontologia noastra (Disease, Symptom, Location).

- (equivalent Component (or Disease Symptom Location))
- (disjoint Disease Symptom Location)

Design Patterns - Pattern de continut

PartOf design pattern. Pentru demonstrarea acestui pattern am considerat entitatea Component si am definit partile ei componente.

- (define-primitive-role subNodeOf :transitive t :inverse superNodeOf)
- (define-primitive-role directSubNodeOf :parent subNodeOf)
- (related Disease top directSubNodeOf)
- (related Symptom top directSubNodeOf)
- (related Location top directSubNodeOf)

Design Patterns - Pattern de prezentare

1. Pentru realizarea acestui pattern, am incercat sa respectam anumite conventii de scriere pentru a realiza un cod cat mai usor de citit indiferent de expertiza celui care vizualizeaza. Rolurile si instantele existente incep cu litera mica, iar conceptele cu litera mare. In cazul in care acestea sunt compuse din mai multe cuvinte aceste sunt separate pintr-o linie, in cazul rolurilor, si printr-un underline in cazul conceptelor si instantelor pentru a fi mai usor de citit.

Table of Contents

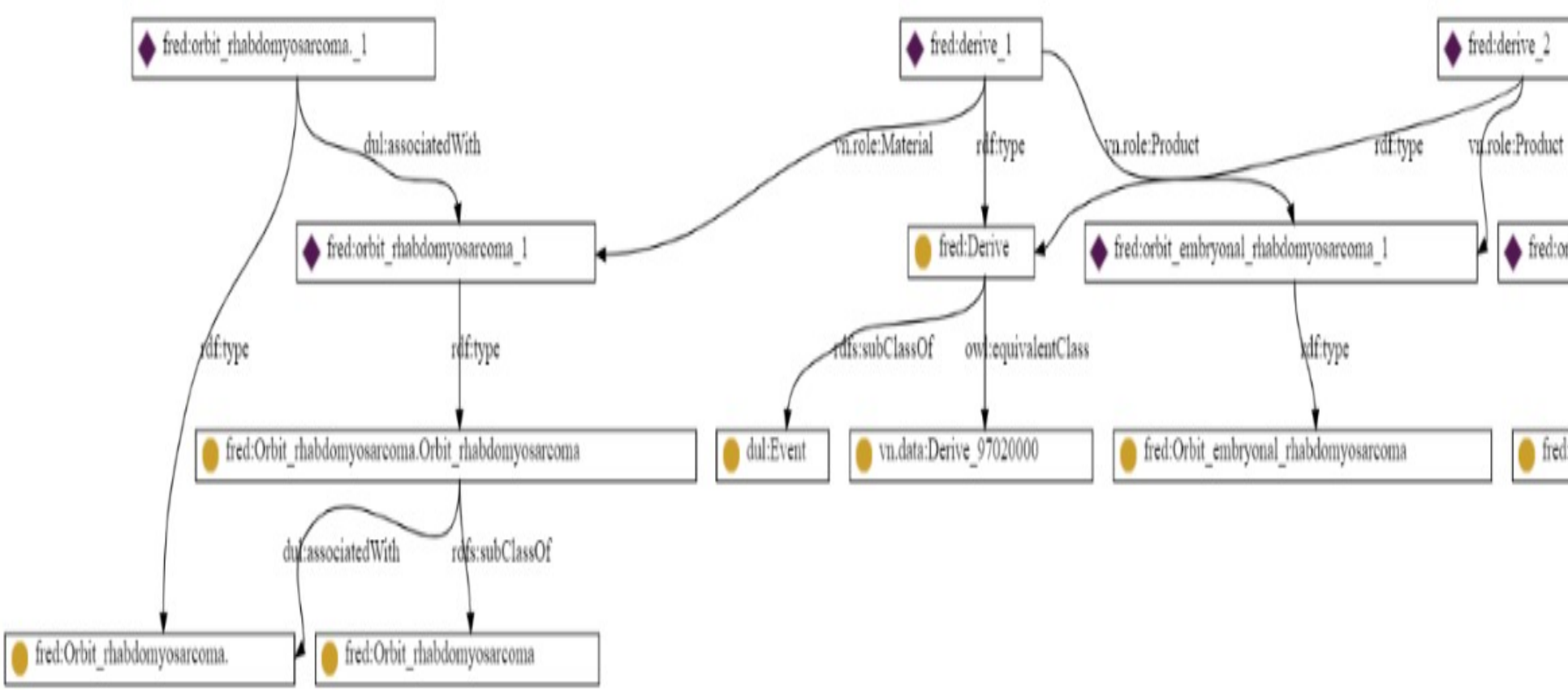
Step 1: Despre Fred

Prin intermediul FRED, utilizatorii pot defini clase, proprietăți și relații între entități, precum și reguli și constrângeri pentru a modela cunoștințele specifice domeniului în care este folosită ontologia. Acesta oferă o interfață intuitivă și ușor de utilizat, care permite utilizatorilor să creeze ontologii fără a avea nevoie de cunoștințe avansate în programare sau în limbajul de interogare a ontologiilor, cum ar fi SPARQL.

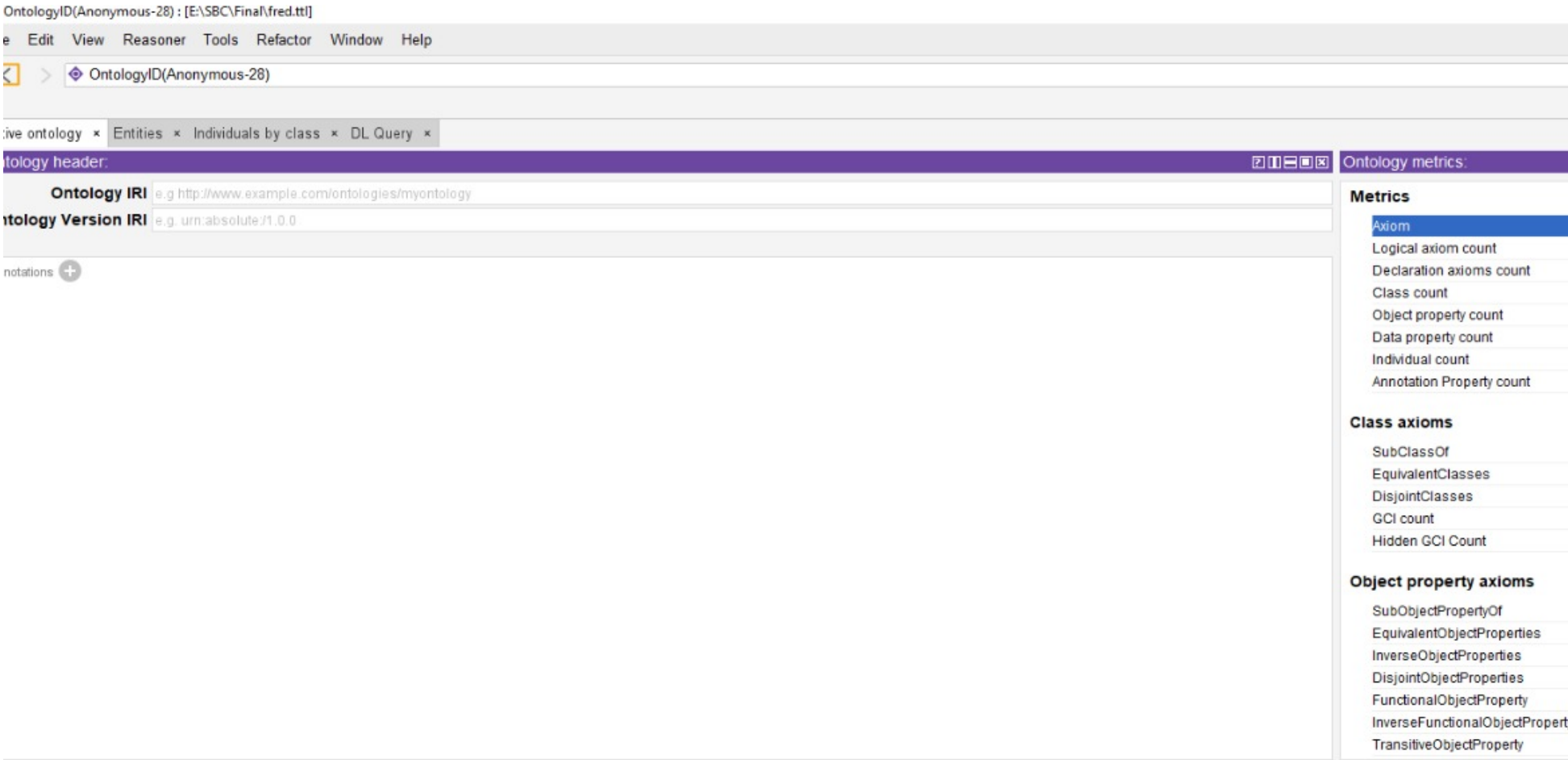
FRED tool pune la dispoziție și funcționalități pentru gestionarea ontologiilor, inclusiv importul și exportul de ontologii în diferite formate standard, precum RDF sau OWL. De asemenea, oferă facilități de vizualizare și explorare a ontologiilor create, ajutând utilizatorii să înțeleagă și să verifice structura și conținutul ontologiei lor.

Step 1: Translating the myth in DL using Fred

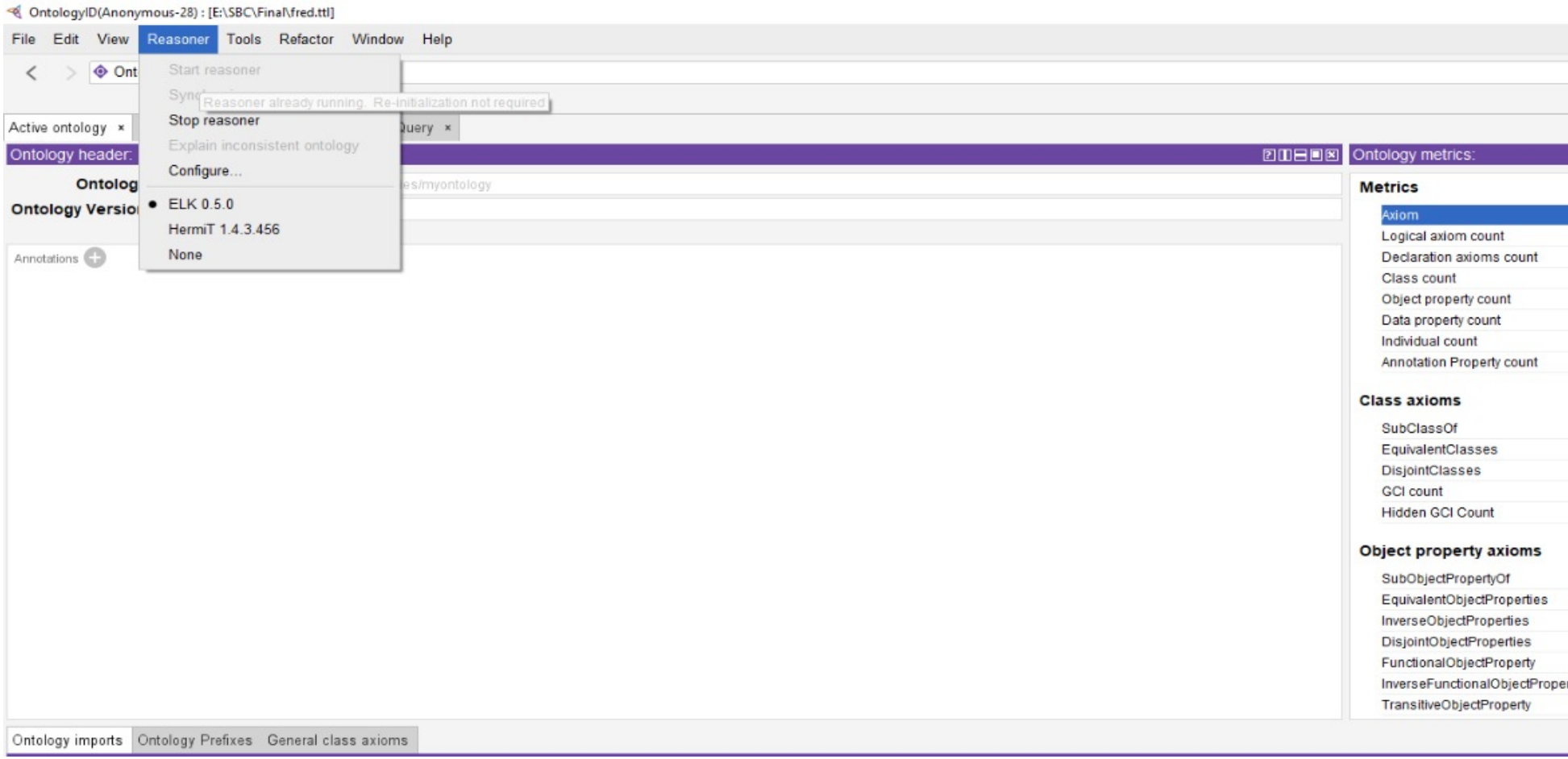
1. Accesați http://wit.istc.cnr.it/stlab-tools/fred/demo/
2. Obțineți reprezentarea grafică a sentence-ului si salvați-o ca nume.png
3. Obțineți formalizarea textului în sintaxa turtle si salvați-o ca nume.ttl



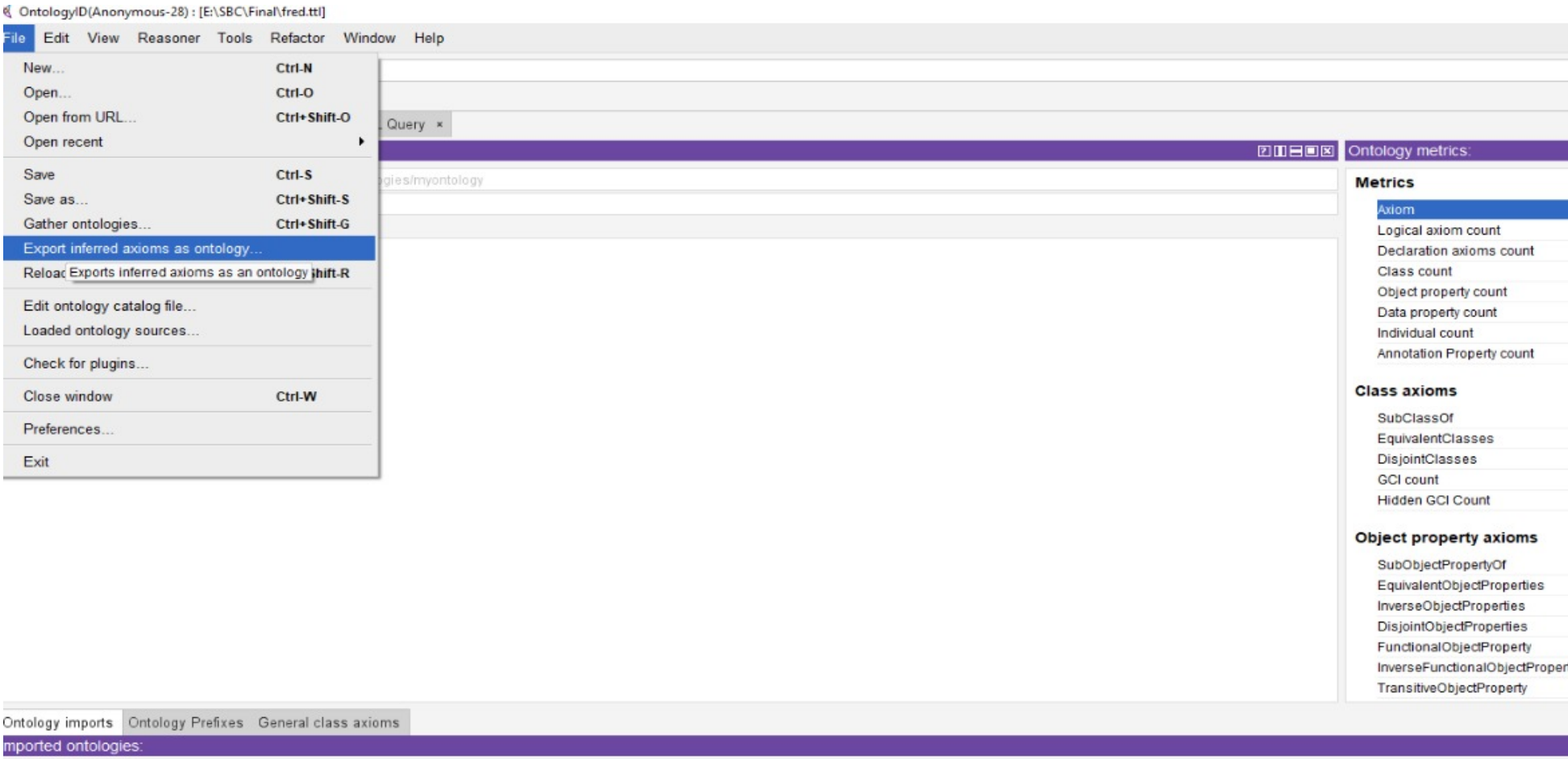
Step 2: Protege



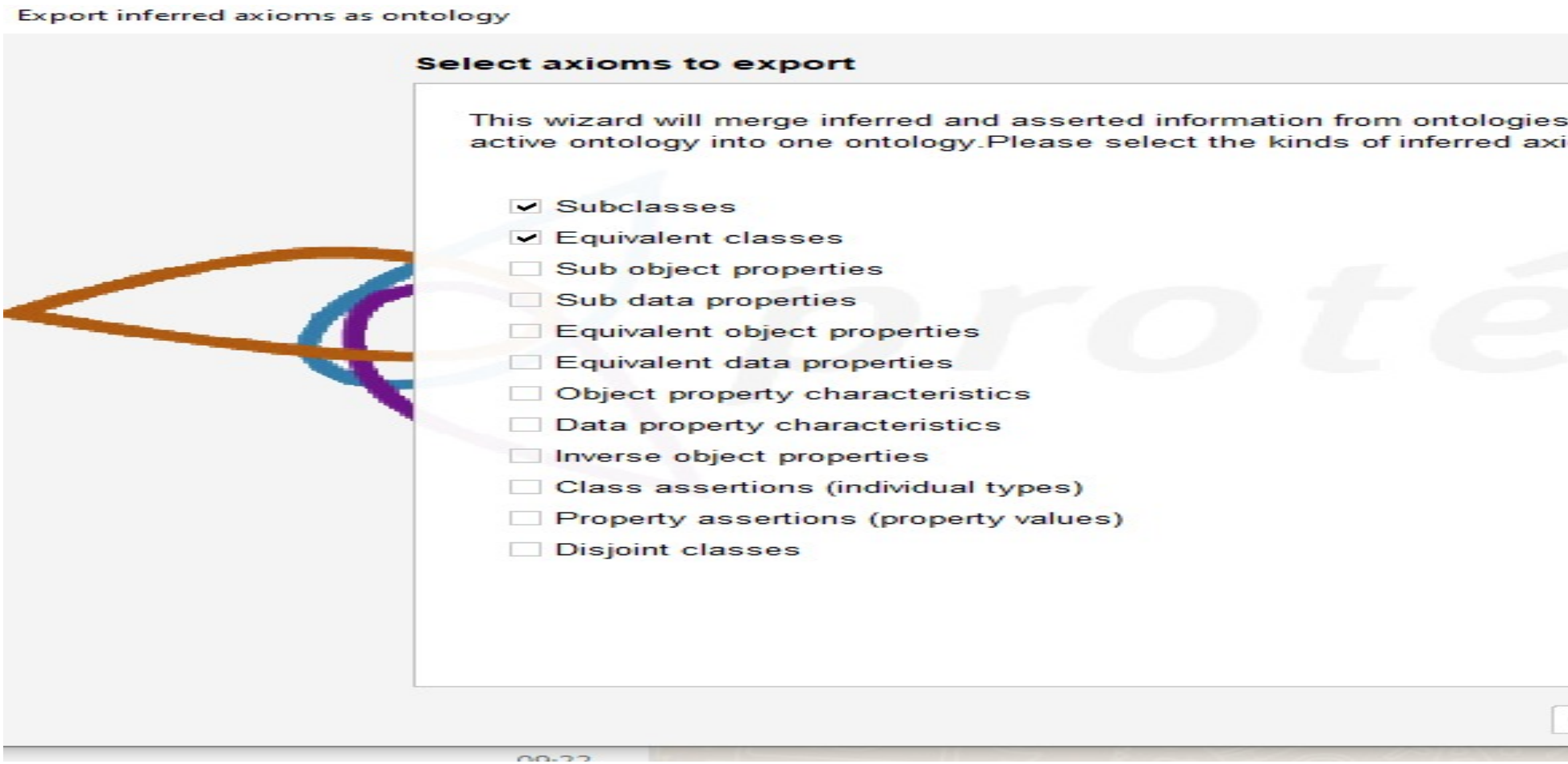
Step 2: Protege



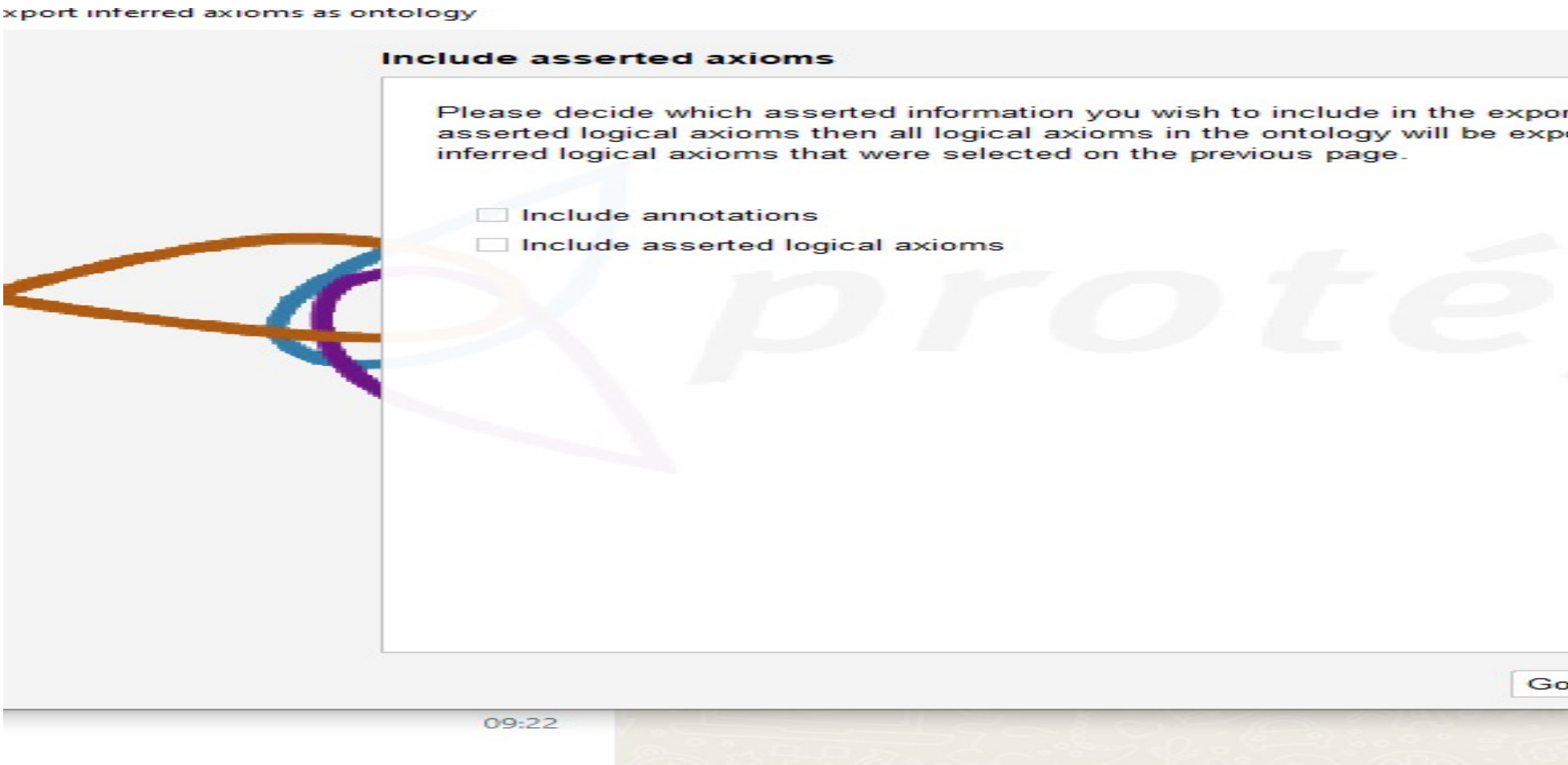
Step 2: Protege



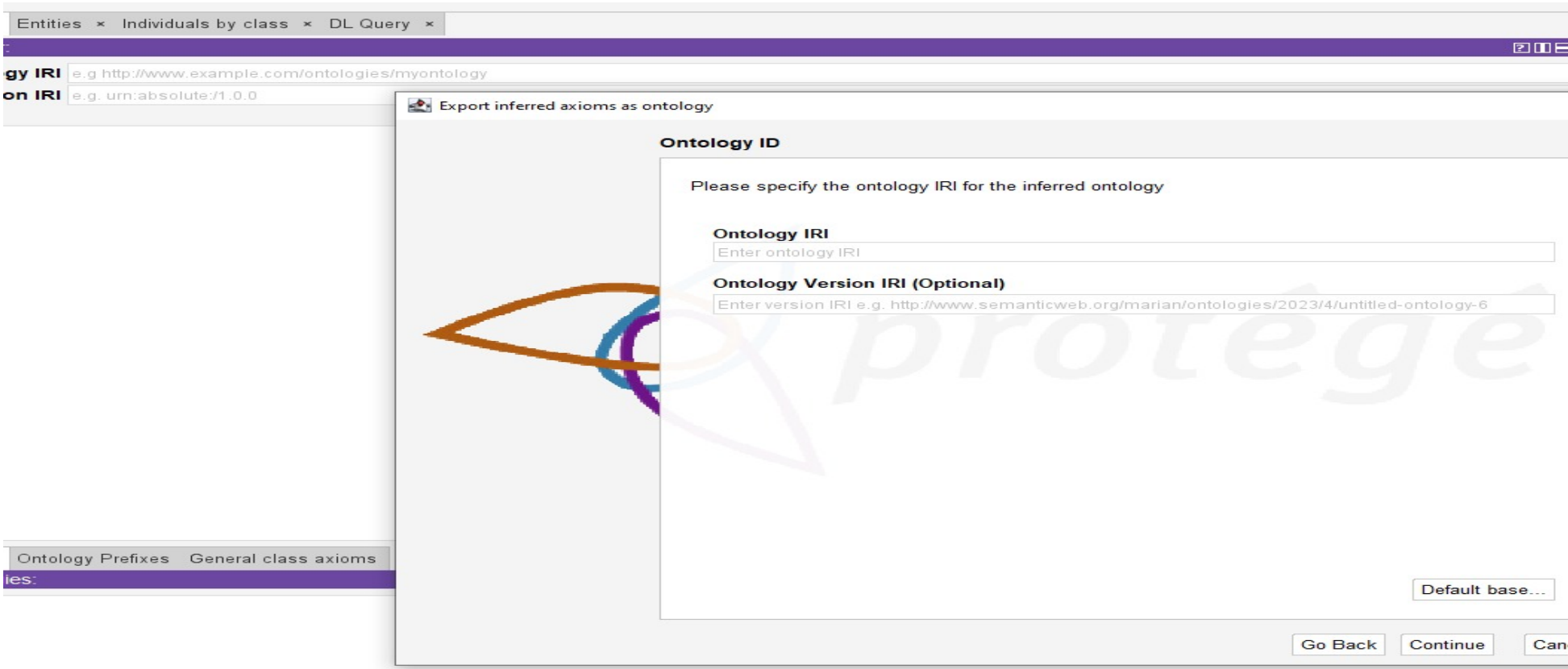
Step 2: Protege



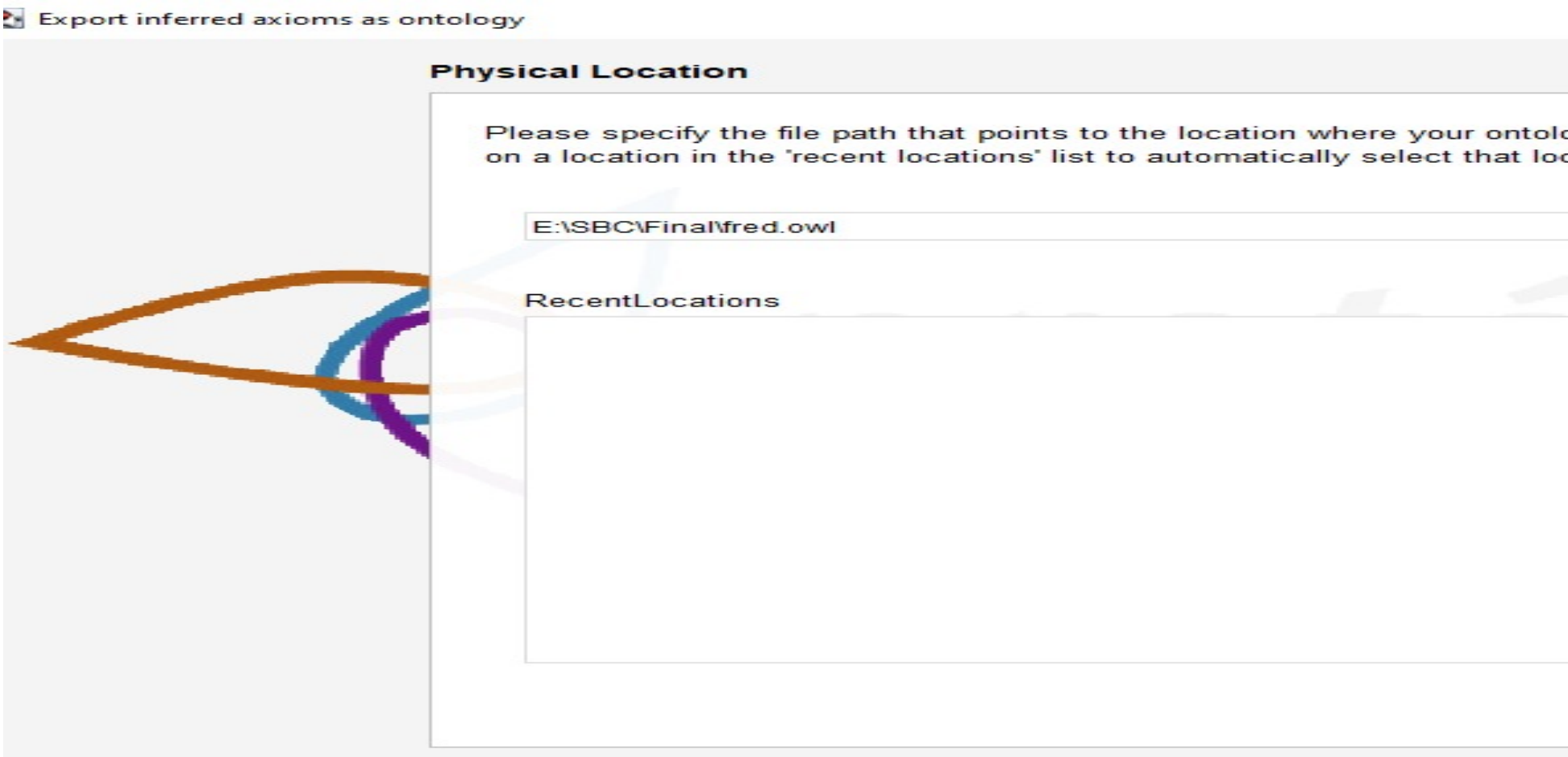
Step 2: Protege



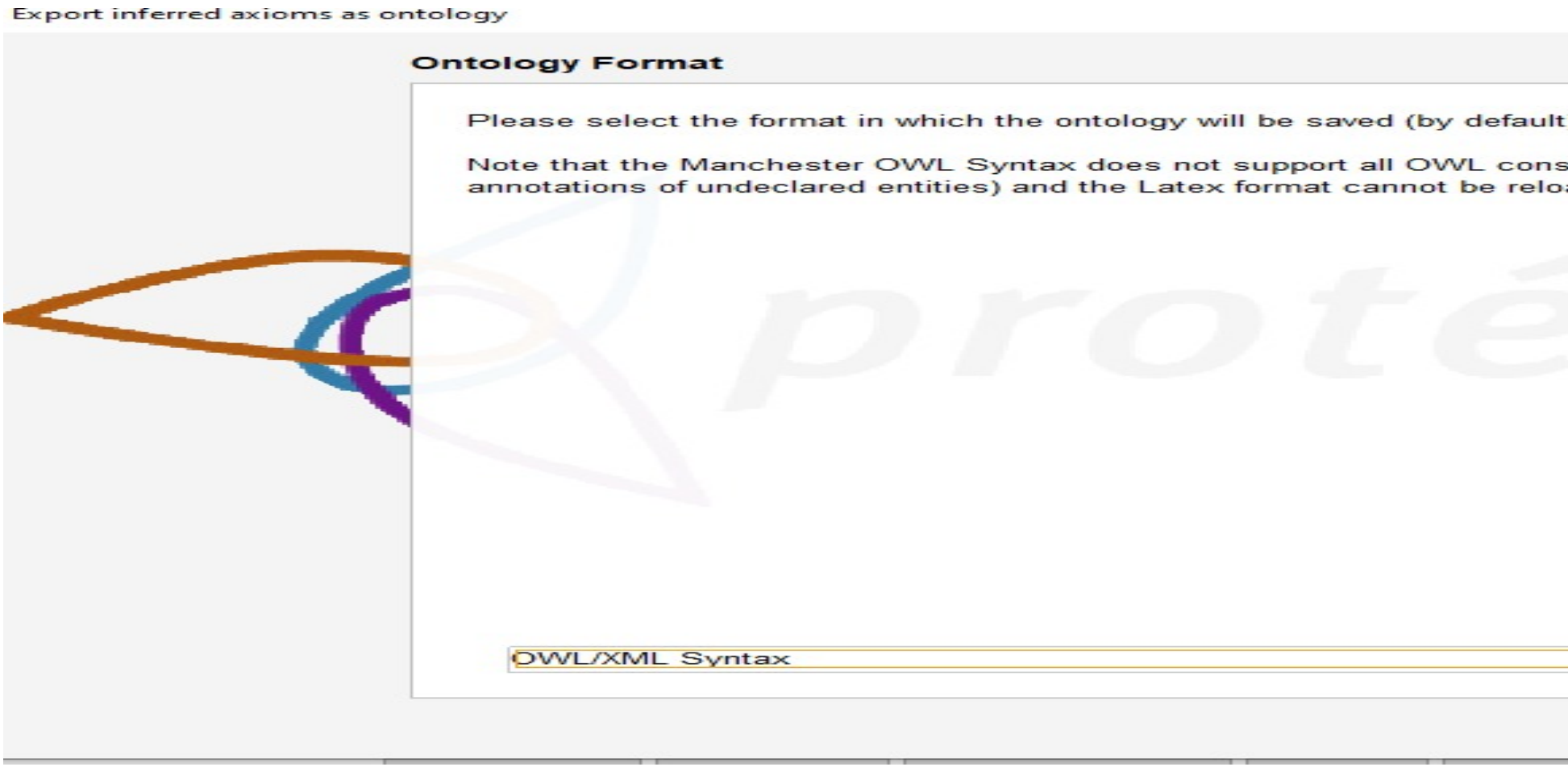
Step 2: Protege



Step 2: Protege



Step 2: Protege



Step 3: Simplify the ontology

1. Am incarcat fiserul salvat cu owl/xml in racer
2. L-am salvat in OWL cu comanda (save-kb “fred.owl” :syntax :owl)
3. Dupa am adaugat continutul la ontlogia noastra deja existenta in owl

Table of Contents

Verbalizer

Pasul 1: Traducerea în DL folosind Fred

1. Accesați http://wit.istc.cnr.it/stlab-tools/fred/demo/
2. Obțineți reprezentarea grafică a sentence-ului si salvați-o ca nume.png
3. Obțineți formalizarea textului în sintaxa turtle si salvați-o ca nume.ttl

Verbalizer

Pasul 2: Protege

1. Am incarcarat ontologia .ttl
2. Am selectat start reasoner
3. Am dat export
4. Am selectat clase si subclase
5. Am selectatlocatia si am denumit fisierul Ontologie.owl
6. Am salvat in OWL/XML

Verbalizer

Pasul 3: Online tool

Am pus formatul RDF/XML in : `http://attempto.ifi.uzh.ch/site/docs/owl_to_ace.html` si a generat varianta finala in natural language:

Verbalizer

```
Every LymphocyteOrbital is an Orbit.
Every LymphocyteOrbital is an Orbital.
Every LymphocyteOrbitalCancer is an OrbitalCancer.
Every Material is a Quality.
Every MaterialBasis is a Basis.
Every Orbit is a Quality.
Every OrbitAlveolar is an Alveolar.
Every OrbitAlveolarRhabdomyosarcoma is an Alveolar_rhabdomyosarcoma.
Every OrbitAlveolarRhabdomyosarcoma is an AlveolarRhabdomyosarcoma.
Every OrbitEmbryonalRhabdomyosarcoma is an Embryonal_rhabdomyosarcoma.
Every OrbitEmbryonalRhabdomyosarcoma is an EmbryonalRhabdomyosarcoma.
Every OrbitLymphoma is a Lymphoma.
Every OrbitLymphoma is a Lymphoma.
Every OrbitRhabdomyosarcoma is a Rhabdomyosarcoma.
Every OrbitRhabdomyosarcoma is a Rhabdomyosarcoma.
Every OrbitRhabdomyosarcoma. is a Rhabdomyosarcoma..
Every OrbitRhabdomyosarcoma.Orbit is a Rhabdomyosarcoma.Orbit.
Every OrbitRhabdomyosarcoma.OrbitRhabdomyosarcoma is a Rhabdomyosarcoma.Or
Every OrbitSarcoma is a Sarcoma.
Every Orbital is a Quality.
Every OrbitalCancer is a Cancer.
Every OrbitalCancer is a Cancer.
Every OrbitalMeningioma is a Meningioma.
Every OrbitalMeningioma is a Meningioma.
Every Rhabdomyosarcoma.Orbit is an Orbit_(anatomy).
Every Rhabdomyosarcoma.Orbit is an Orbit.
Every Rhabdomyosarcoma.Orbit is an Orbital.
Every Rhabdomyosarcoma.OrbitRhabdomyosarcoma is an OrbitRhabdomyosarcoma.
Every SymptomExophthalmo is an Exophthalmo.
```