



Knowledge-Based Systems
Laboratory activity: 2022-2023
Ontology title: Orbital Disease Ontology

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Contents

1	1.1 Interogări	3
2	Reusing other ontologies 2.1 Reusing other Ontologies	4
3	Tboxes	6
4	Aboxes	8
5	Queries5.1 Interogari Nrql5.2 Interogari Racer5.3 Evaluare ontologie5.4 Rezultate query-uri	11
6	Query-uri pentru ontologie in limbaj natural	12
7	Design patterns	14
8	Fred	15
9	Verbaliser	19
10	Anexa .1 Racer code	20 20 22

Competency questions

1.1 Interogări

- 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?

Reusing other ontologies

2.1 Reusing other Ontologies

- 1. 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
- 2. Dupa descarcarea ontologiei in format owl am folosit site-ul acesta pentru a genera un rdf folosind EasyRdf converter

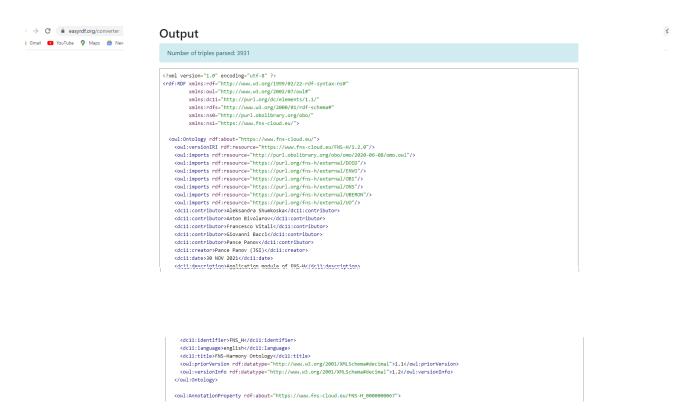


Figure 2.1: EasyRdf

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/)

- 3. 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
- 4. 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 :

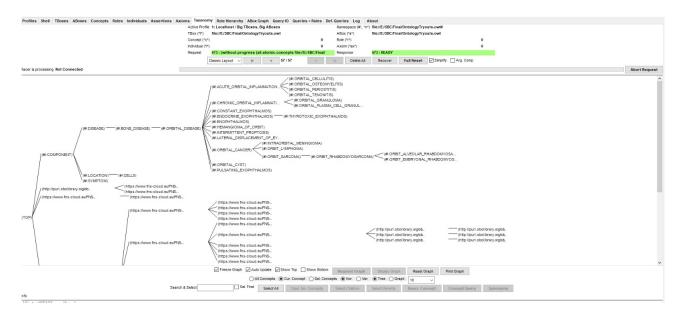


Figure 2.2: OntologyReusing

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.

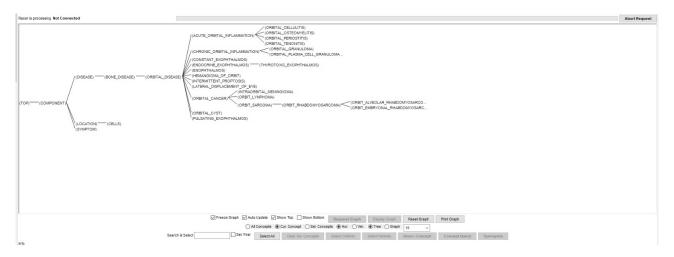


Figure 3.1: Tbox

```
(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 3.2: Roles

```
(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 3.3: Implies Conceps

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.

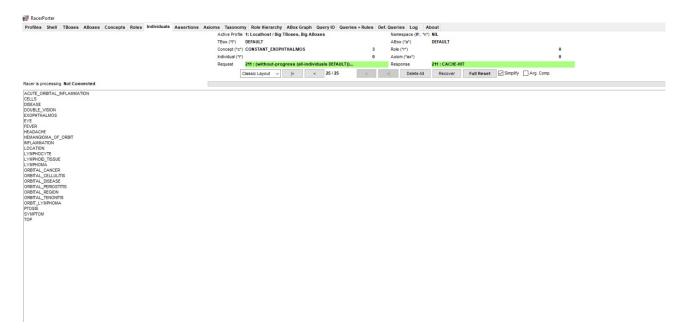


Figure 4.1: Indivizi

```
(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 Orbital_disease eye adjacent-to)
(related Orbital_disease eye adjacent-to)
```

Figure 4.2: Related

```
(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)
```

Figure 4.3: Instance

Queries

5.1 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))
```

5.2 Interogari Racer

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

5.3 Evaluare ontologie

1. Cod pentru determinarea si evaluarea diferitilor factori ai ontologiei

```
(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)))
(evaluate (length (all-rules)))
```

5.4 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 (2X) (AND (2X DISEASE) (2X LYMPHOID_TISSUE HAS-LOCATION))) --> (((2X LYMPHOMA)))
(RETRIEVE (2X) (AND (2X DISEASE) (2X INFLAMMATION HAS-SYMPTOM))) --> (((2X ORBITAL_CELLULITIS)) ((2X ORBITAL_TENONITIS)) ((2X ORB
ITAL PERIOSTITIS)) ((2X 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_RHABDOMYOSARCOMA
ARCOMA LATERAL_DISPLACEMENT_OF_EYE ORBIT_ALVEOLAR_RHABDOMYOSARCOMA CONSTANT_EXOPHTHALMOS ORBIT_LYMPHOMA_SYMPTOM ENOPHTHALMOS ORBIT
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 5.1: Rezultate query

Query-uri pentru ontologie in limbaj natural

1. Urmatoarele query-uri folosite in ontologie au fost generate prin intermediul ChatGPT. Acestea au fost formulate in limbaj natural, dupa care au fost incarcate impreuna cu ontologia in chat, urmand ca acesta sa creeze codul necesar.

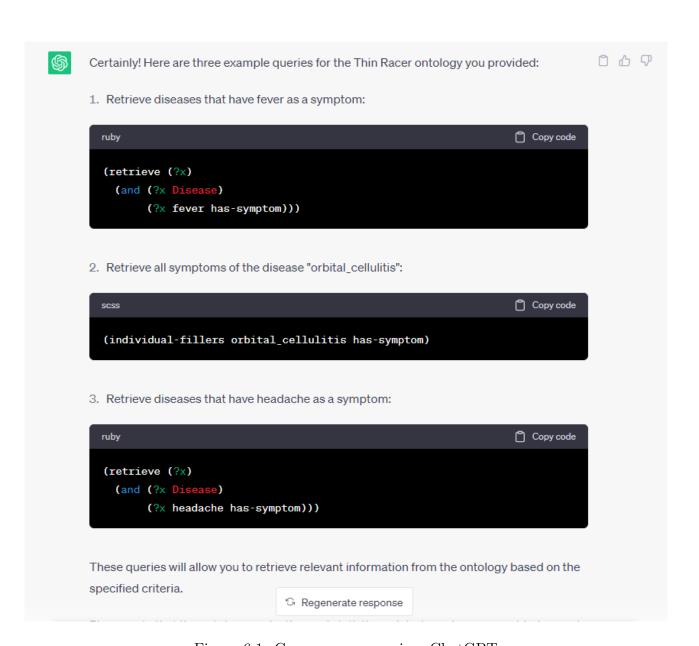


Figure 6.1: Generare query-uri cu ChatGPT

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 :

Pattern-uri structurale:

1. 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)
```

2. 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)
```

Pattern de continut:

1. 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)
```

Pattern de prezentare:

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.

Fred

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

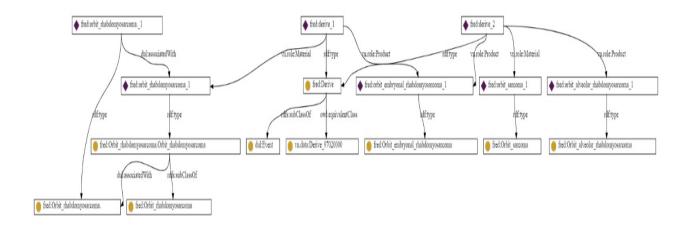
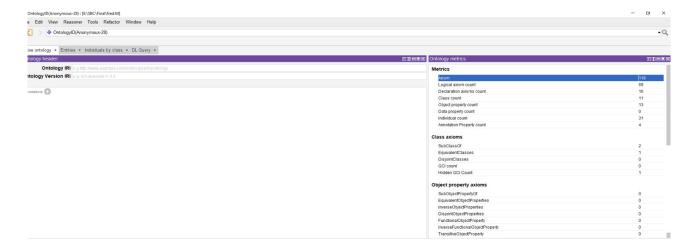
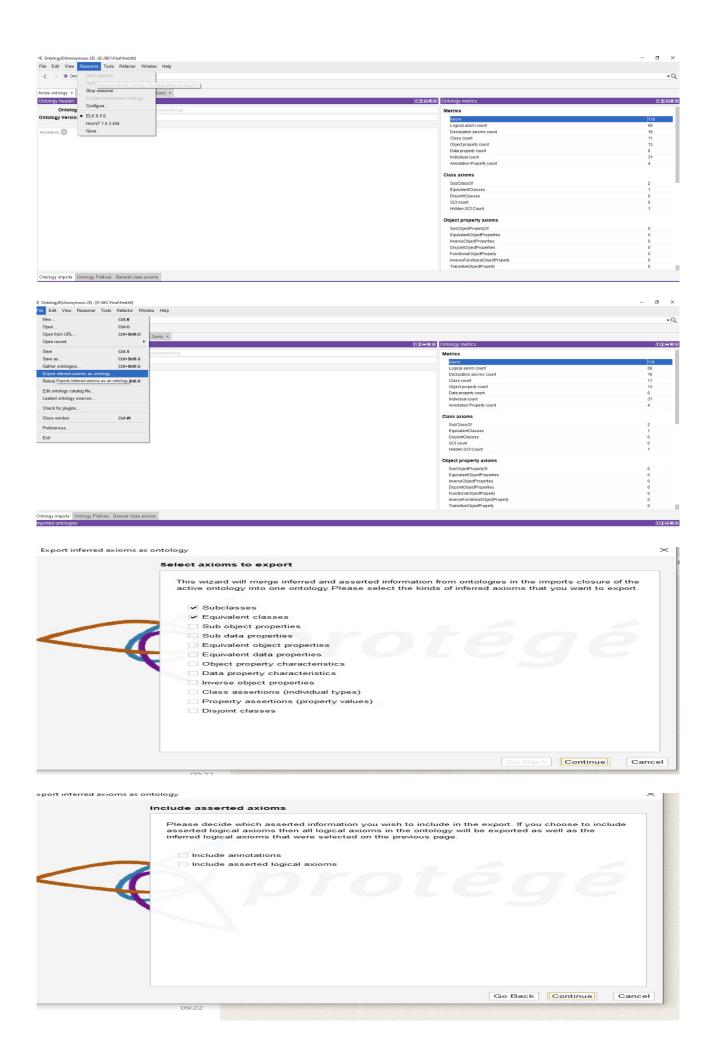
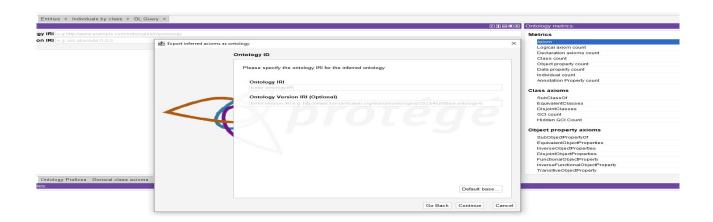


Figure 8.1: Reprezentarea grafica a propozitiei

Step 2: Protege











Step 3: From OWL/XML syntax to OWL

- 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

Verbaliser

Urmam aceeasi pasi ca si in Fred:

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

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

- 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

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:

```
Every Lymphocyteorbital is an Orbital.

Every Lymphocyteorbitalian orbital.

Every Lymphocyteorbitalian orbital.

Every Material is a Quality.

Every Material is a Quality.

Every Orbitalian orbital orbital.

Every Orbitalian orbitalian orbital.

Every Orbitalian orbitalian orbital.

Every Orbitalian orbitalian orbitalian.

Every Orbitalian orbitalian orbitalian.

Every Orbitalian orbitalian orbitalian.

Every Orbitalian or
```

Anexa

.1 Racer code

```
1 (full-reset)
2 (disable-nrql-warnings)
6 (define-primitive-role has-symptom :domain Disease :range Symptom)
7 (define-primitive-role is-different :domain Disease
                                           :range Symptom)
8 (define-primitive-role has-location :domain Disease :range Location)
9 (define-primitive-role adjacent-to :domain Disease :range Location)
10 (define-primitive-role has-material-basis-in :domain Disease :range Cells)
 15 ;;;;;;;;;;;;PartOf Design pattern;;;;;;;;;;;;;;
16 (define-primitive-role subNodeOf :transitive t :inverse superNodeOf)
17 (define-primitive-role directSubNodeOf :parent subNodeOf)
18 (related Disease top directSubNodeOf)
19 (related Symptom top directSubNodeOf)
20 (related Location top directSubNodeOf)
21 (related Cells Location directSubNodeOf)
 27 (instance fever Symptom)
28 (instance ptosis Symptom)
29 (instance headache Symptom)
30 (instance double_vision Symptom)
31 (instance inflammation Symptom)
32 (instance exophthalmos Symptom)
34 (instance orbital_region Location)
35 (instance lymphoid_tissue Location)
36 (instance eye Location)
 (instance lymphocyte Cells)
41 (equivalent Component (or Disease Symptom Location))
```

```
42 (disjoint Disease Symptom Location)
  45
46 (implies Intermittent_proptosis Orbital_disease)
47 (implies Orbital_cyst Orbital_disease)
48 (implies Pulsating_exophthalmos Orbital_disease)
49 (implies Enophthalmos Orbital_disease)
50 (implies Constant_exophthalmos Orbital_disease)
  (implies Lateral_displacement_of_eye Orbital_disease)
  (implies Hemangioma_of_orbit Orbital_disease)
  (implies Thyrotoxic_exophthalmos Endocrine_exophthalmos)
  (implies Orbital_plasma_cell_granuloma Chronic_orbital_inflammation)
  (implies Orbital_granuloma Chronic_orbital_inflammation)
  (implies Orbital_cellulitis Acute_orbital_inflammation)
59 (implies Orbital_tenonitis Acute_orbital_inflammation)
60 (implies Orbital_osteomyelitis Acute_orbital_inflammation)
  (implies Orbital_periostitis Acute_orbital_inflammation)
63 (implies Intraorbital_meningioma Orbital_cancer)
64 (implies Orbit_lymphoma Orbital_cancer)
65 (implies Orbit_lymphoma Orbital_cancer)
  (implies Orbit_alveolar_rhabdomyosarcoma Orbit_rhabdomyosarcoma)
  (implies Orbit_embryonal_rhabdomyosarcoma Orbit_rhabdomyosarcoma)
  (implies Cells Location)
71 (implies Bone_disease Disease)
72 (implies Orbital_disease Bone_disease
  (implies Endocrine_exophthalmos Orbital_disease )
75 (implies
          Chronic_orbital_inflammation Orbital_disease)
          Acute_orbital_inflammation Orbital_disease)
76 (implies
77 (implies Orbital_cancer Orbital_disease)
78 (implies Orbit_sarcoma Orbital_cancer)
79 (implies Orbit_rhabdomyosarcoma Orbit_sarcoma )
80
82 (related Orbital_cellulitis fever has-symptom)
83 (related Orbital_cellulitis ptosis has-symptom)
84 (related Orbital_cellulitis headache has-symptom)
85 (related Orbital_cellulitis double_vision has-symptom)
86 (related Orbital_cellulitis inflammation has-symptom)
87 (related Orbital_cancer ptosis has-symptom)
88 (related Orbital_cancer exophthalmos has-symptom)
89 (related Orbital_tenonitis inflammation has-symptom)
90 (related Orbital_periostitis inflammation has-symptom)
91 (related Acute_orbital_inflammation inflammation has-symptom)
93 (related Hemangioma_of_orbit orbital_region has-location)
94 (related Lymphoma lymphoid_tissue has-location)
95
  (related Orbital_disease eye adjacent-to)
  (related Orbit_lymphoma lymphocyte has-material-basis-in)
98
99
100
```

```
104
;1_____Returneaza bolile care au ca simptom febra
106 (retrieve (?x)
   (and (?x Disease)
   (?x fever has-symptom)))
108
110 (individual-fillers fever (inv has-symptom) )
;2_____Returneaza toate simptomele bolii orbital_cellulitis
(individual-fillers orbital_cellulitis has-symptom )
114
116 ;3_____Returneaza bolile care au ca simptom dureri de cap
117 (retrieve (?x)
   (and (?x Disease)
119
   (?x headache has-symptom)))
120
;4_____Ce boala apare in tesutul limfoid?
122 (retrieve (?x)
123 (and (?x Disease)
124 (?x lymphoid_tissue has-location)))
126 ;5_____Returneaza toate bolile care au ca simptom inflamatia
127 (retrieve (?x)
128 (and (?x Disease)
129 (?x inflammation has-symptom)))
133 (all-atomic-concepts)
134 (all-individuals)
136 (abox-consistent?)
137 (tbox-cyclic?)
138 (tbox-coherent?)
140 (realize-abox)
  (classify-tbox)
(evaluate (length (all-individuals)))
144 (evaluate (length (all-atomic-concepts)))
145 (evaluate (length (all-roles)))
146 (evaluate (length (all-rules)))
```

Listing 1: Orbital Disease Ontology

.2 Competency questions Racer and Nrql and evaluation

```
;1_____Returneaza bolile care au ca simptom febra
(retrieve (?x)
(and (?x Disease)
(?x fever has-symptom)))

(individual-fillers fever (inv has-symptom))

;2_____Returneaza toate simptomele bolii orbital_cellulitis
(individual-fillers orbital_cellulitis has-symptom))
```

```
3;3_____Returneaza bolile care au ca simptom dureri de cap
14 (retrieve (?x)
15 (and (?x Disease)
  (?x headache has-symptom)))
18 ;4_____Ce boala apare in tesutul limfoid?
19 (retrieve (?x)
20 (and (?x Disease)
21 (?x lymphoid_tissue has-location)))
23 ;5_____Returneaza toate bolile care au ca simptom inflamatia
24 (retrieve (?x)
25 (and (?x Disease)
26 (?x inflammation has-symptom)))
29 (all-atomic-concepts)
30 (all-individuals)
32 (abox-consistent?)
33 (tbox-cyclic?)
34 (tbox-coherent?)
36 (realize-abox)
37 (classify-tbox)
39 (evaluate (length (all-individuals)))
40 (evaluate (length (all-atomic-concepts)))
41 (evaluate (length (all-roles)))
42 (evaluate (length (all-rules)))
```

Listing 2: Competency questions Racer and Nrql and evaluation

