Collaborative Ontology for Neurofibromatosis

Hack for NF 2020

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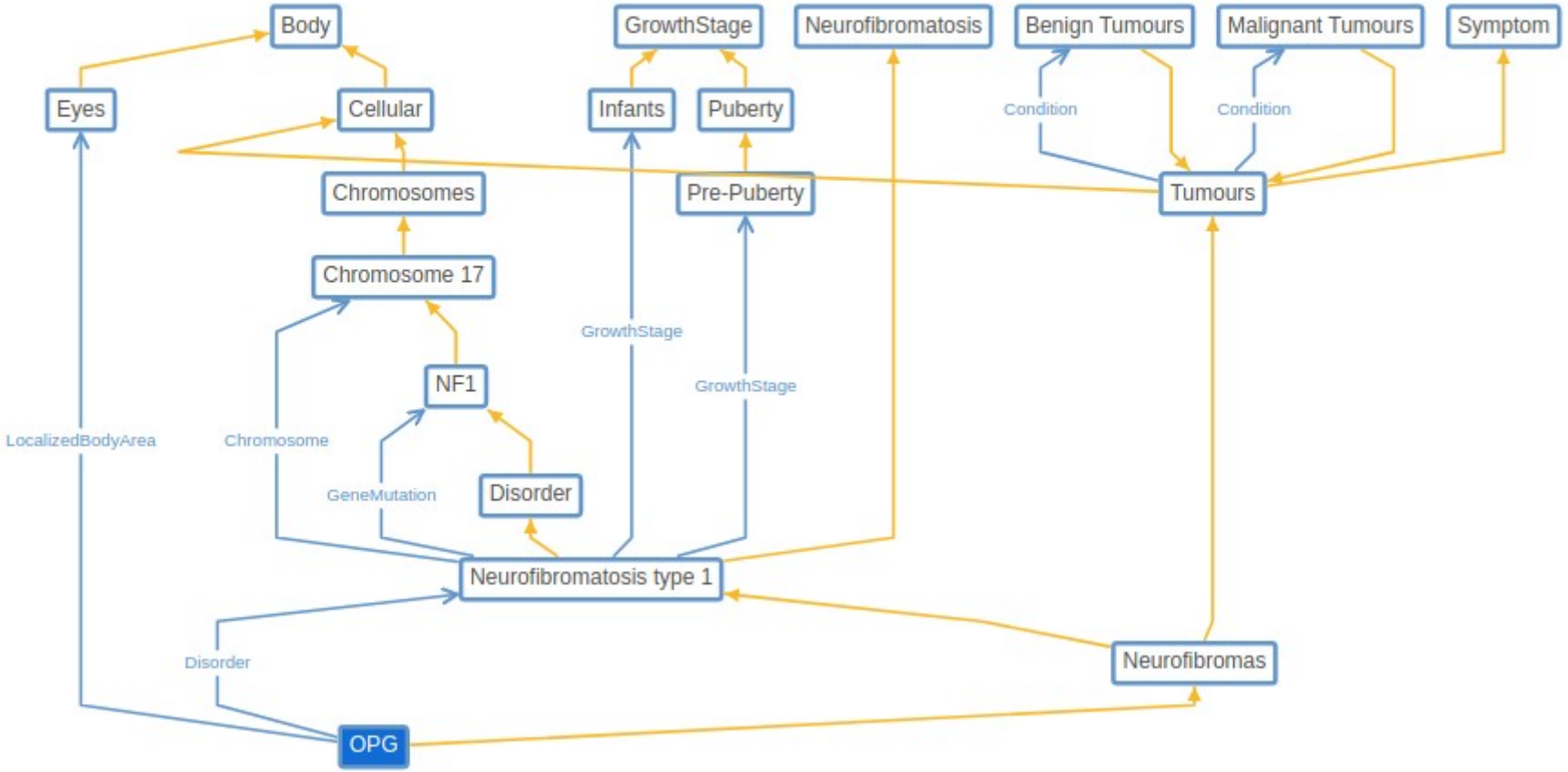
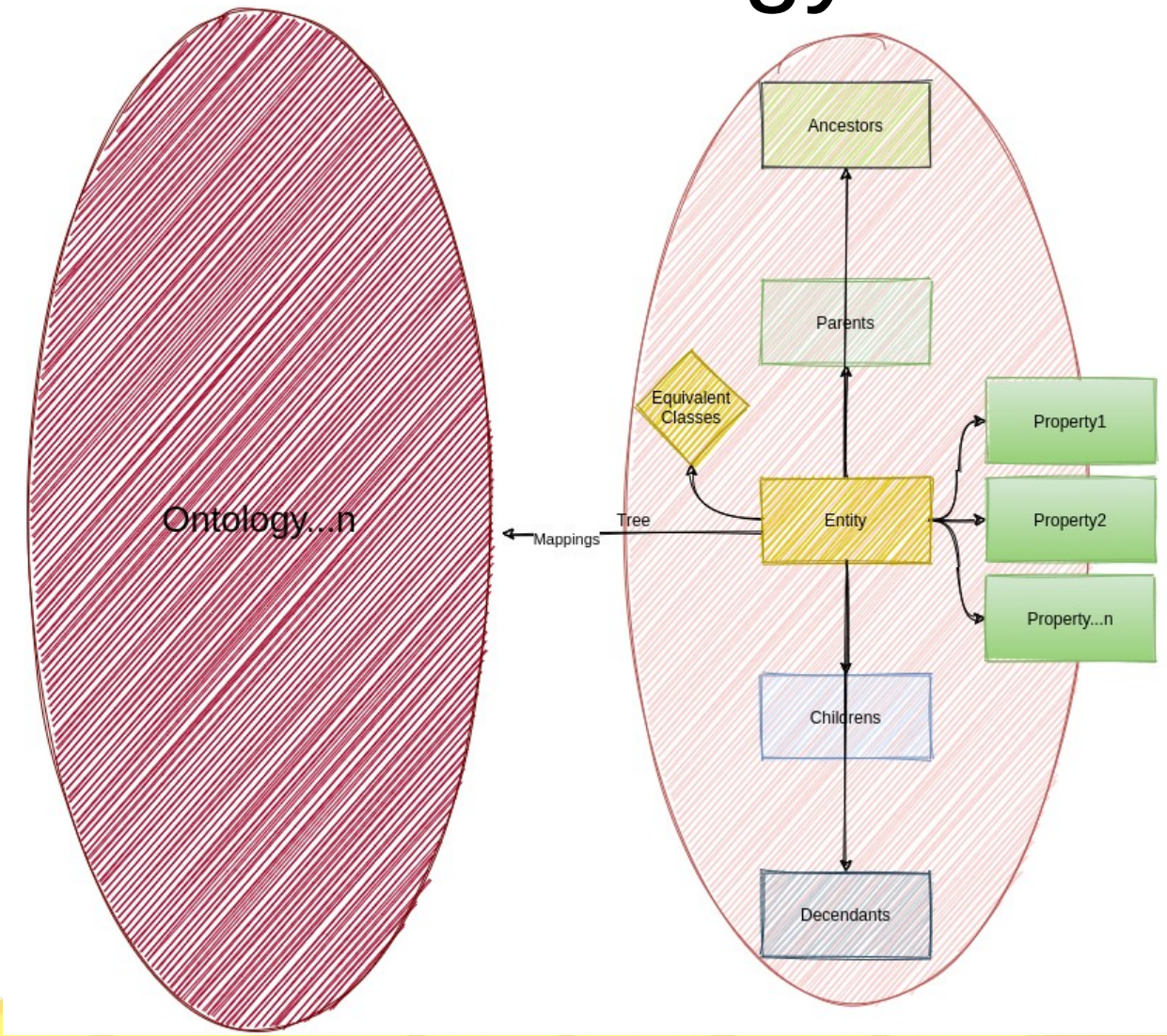
Manual Ontology Attempt

Previously using Protege Ontology Software, Online, hosted by Stanford
Link: https://webprotege.stanford.edu/#projects/d80b2782-b714-45f9-9cbd-598df356ce5e/



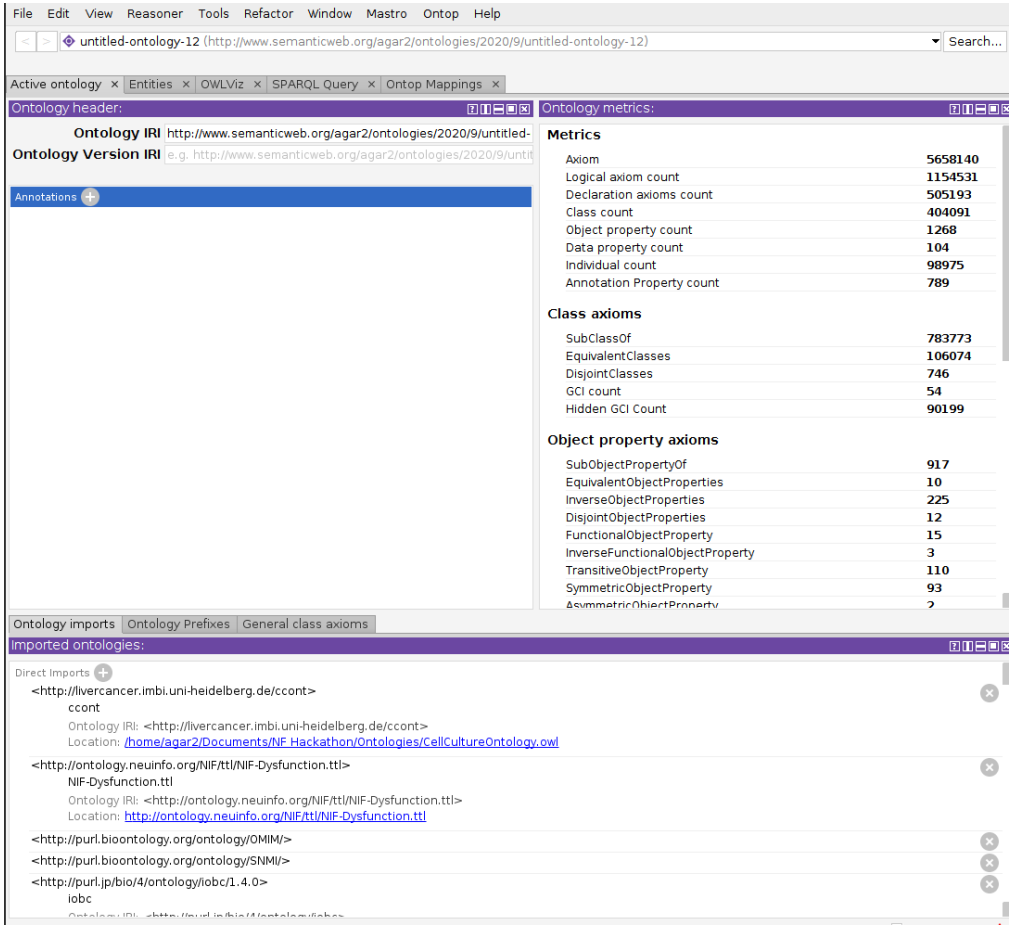
Term to Ontology Scheme



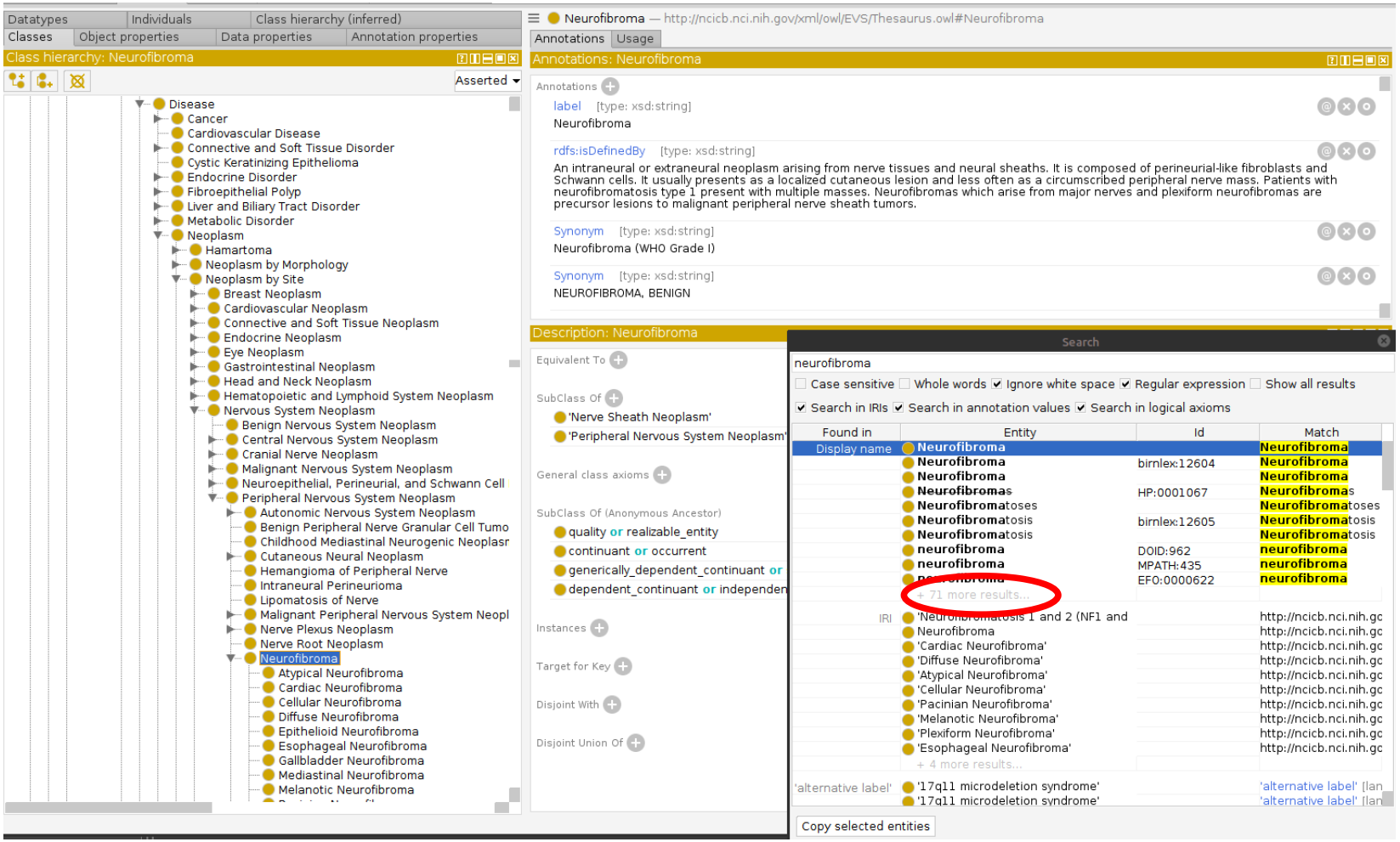
Protege Ontology Editor

Downloaded ~20 ontologies where ‘neurofibroma’ was an entity/class. Issues with loading, found 5.65m axioms if wanted to continue with top-down approach

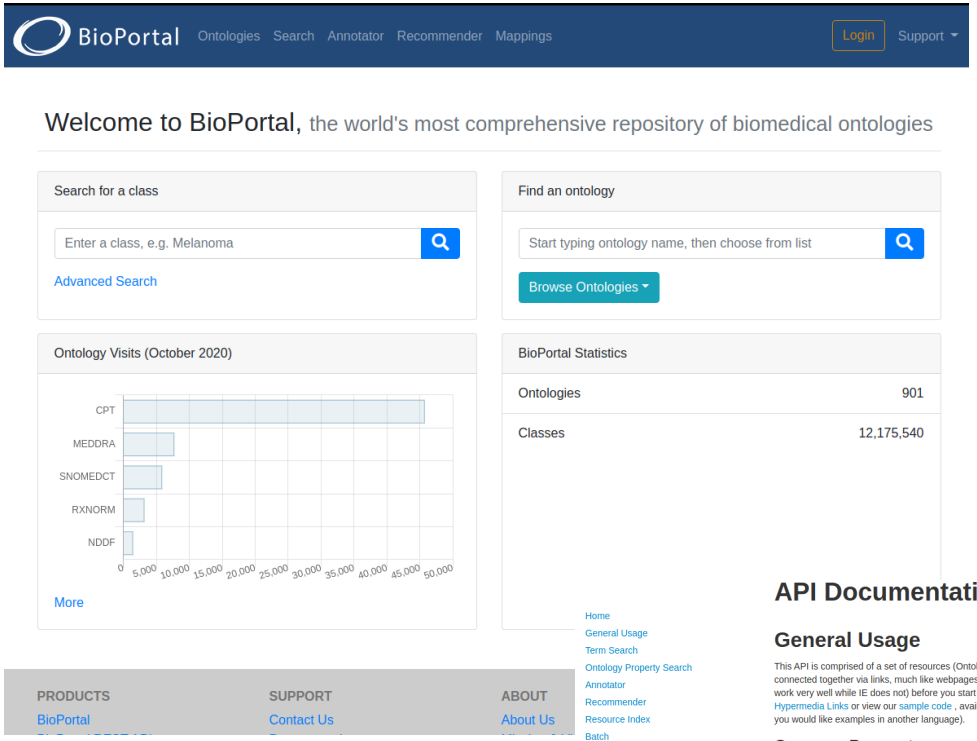
	A	B	C
1	MAPPING TO	ONTOLOGY	SOURCE
2	neurofibroma	BioAssay Ontology	LOOM
3	Neurofibroma	Biomedical Informatics Research Network Project Lexi	LOOM
4	neurofibroma	Cell Culture Ontology	LOOM
5	Neurofibroma	Cigarette Smoke Exposure Ontology	LOOM
6	neurofibroma	Combined Phenotype Ontology	LOOM
7	neurofibroma	Computer Retrieval of Information on Scientific Project	CUI
8	NEUROFIBROMA	Dermatology Lexicon	LOOM
9	Neurofibroma	Disease core ontology applied to Rare Diseases	LOOM
10	neurofibroma	Drug Target Ontology	LOOM
11	neurofibroma	Experimental Factor Ontology	LOOM
12	neurofibroma	Human Dermatological Disease Ontology	LOOM
13	neurofibroma	Human Disease Ontology	LOOM
14	neurofibroma	Hypertension Ontology	LOOM
15	Neurofibroma	Interlinking Ontology for Biological Concepts	LOOM
16	Neurofibrome	MedDRA French	CUI
17	Neurofibroma	Medical Dictionary for Regulatory Activities Terminol	CUI
18	Neurofibroma	Medical Subject Headings	LOOM
19	neurofibroma	Mondo Disease Ontology	LOOM
20	Neurofibroma	National Cancer Institute Thesaurus	LOOM
21	Neurofibroma [Disease/Fin	National Drug File - Reference Terminology	CUI
22	neurofibroma	Natural Products Ontology	LOOM
23	Neurofibroma	Neuroscience Information Framework (NIF) Dysfunctio	LOOM
24	Neurofibroma	Online Mendelian Inheritance in Man	LOOM
25	neurofibroma	Ontology of Consumer Health Vocabulary	LOOM
26	Neurofibroma	Orphanet Rare Disease Ontology	LOOM
27	neurofibroma	Radiology Gamuts Ontology	LOOM
28	neurofibroma	Radiology Lexicon	LOOM
29	Neurofibroma	Read Codes, Clinical Terms Version 3 (CTV3)	CUI
30	Neurofibroma	Robert Hoehndorf Version of MeSH	LOOM
31	Neurofibroma	SNOMED CT	CUI
32	neurofibroma	SNOMED Terminos Clinicos	CUI
33	Neurofibroma, NOS	Systematized Nomenclature of Medicine, International	CUI
34	Neurofibrome	Thesaurus Biomedical Francais/Anglais [French transla	CUI
35	neurofibroma	Zebrafish Phenotype Ontology	LOOM
36			

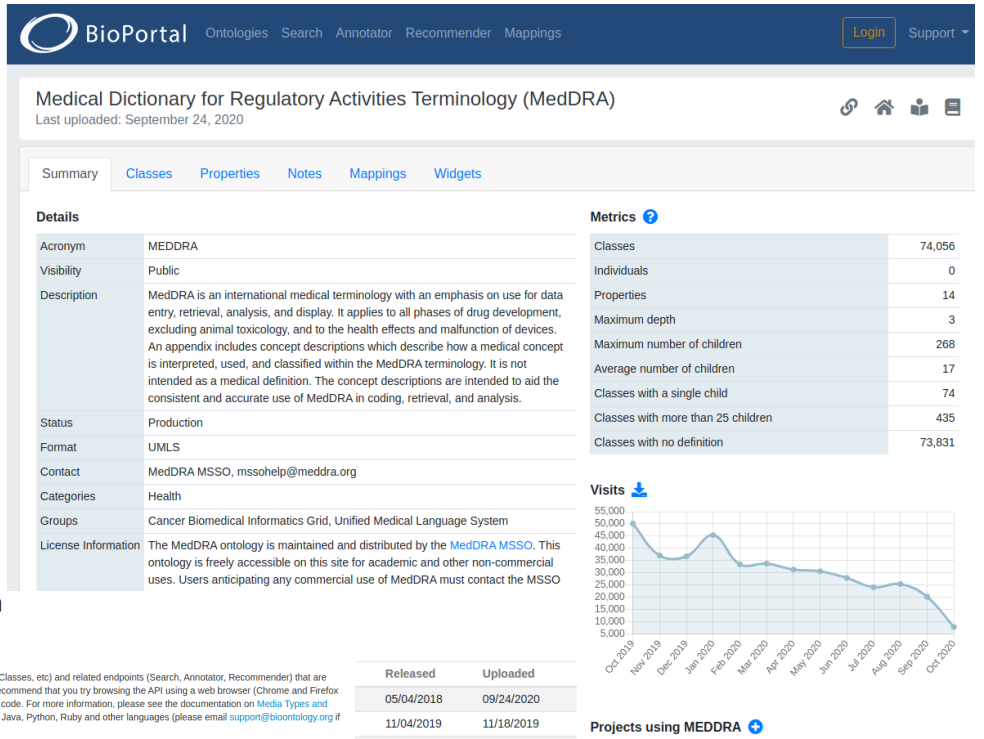


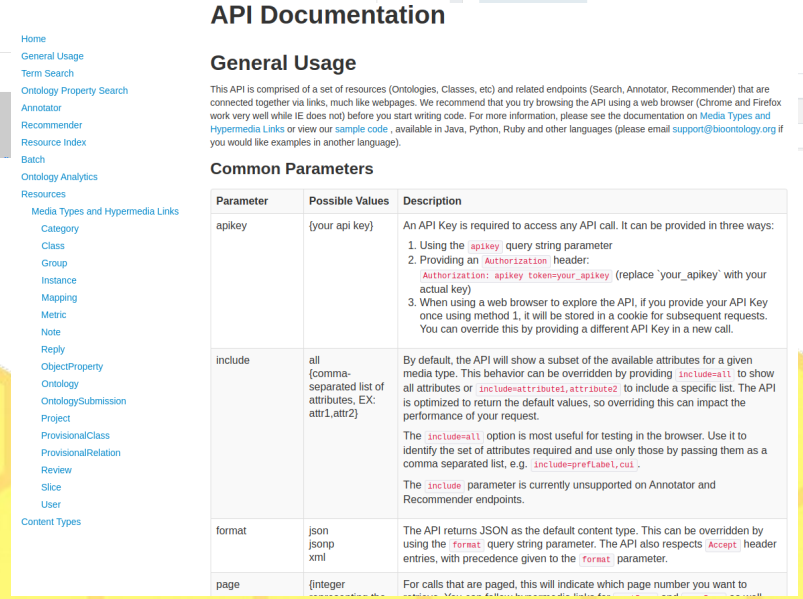
Protege Results



BioPortal Ontologies Repo







OWL (RDF/XML format)

Ontologies > pato.owl

```
1  <?xml version="1.0"?>
2  <rdf:RDF xmlns="http://purl.obolibrary.org/obo/pato.owl#"
3       xml:base="http://purl.obolibrary.org/obo/pato.owl"
4       xmlns:dc="http://purl.org/dc/elements/1.1/"
5       xmlns:go="http://purl.obolibrary.org/obo/go#"
6       xmlns:obo="http://purl.obolibrary.org/obo/"
7       xmlns:owl="http://www.w3.org/2002/07/owl#"
8       xmlns:rdf="http://www.w3.org/1999/02/22-rdf-syntax-ns#"
9       xmlns:xml="http://www.w3.org/XML/1998/namespace"
10      xmlns:xsd="http://www.w3.org/2001/XMLSchema#"
11      xmlns:foaf="http://xmlns.com/foaf/0.1/"
12      xmlns:pato="http://purl.obolibrary.org/obo/pato#"
13      xmlns:rdfs="http://www.w3.org/2000/01/rdf-schema#"
14      xmlns:swrl="http://www.w3.org/2003/11/swrl#"
15      xmlns:swrla="http://swrl.stanford.edu/ontologies/3.3/swrla.owl#"
16      xmlns:swrlb="http://www.w3.org/2003/11/swrlb#"
17      xmlns:terms="http://purl.org/dc/terms/"
18      xmlns:subsets="http://purl.obolibrary.org/obo/ro/subsets#"
19      xmlns:oboInOwl="http://www.geneontology.org/formats/oboInOwl#">
20     <owl:Ontology rdf:about="http://purl.obolibrary.org/obo/pato.owl">
21         <owl:versionIRI rdf:resource="http://purl.obolibrary.org/obo/pato/releases/2020-08-02/p
22         <obo:IAO_0000700 rdf:resource="http://purl.obolibrary.org/obo/PATO_0000001"/>
23         <dc:description rdf:datatype="http://www.w3.org/2001/XMLSchema#string">An ontology of p
24         <dc:title rdf:datatype="http://www.w3.org/2001/XMLSchema#string">PATO - the Phenotype A
25         <terms:license rdf:resource="https://creativecommons.org/licenses/by/3.0/"/>
26         <oboInOwl:default-namespace rdf:datatype="http://www.w3.org/2001/XMLSchema#string">qual
27         <oboInOwl:hasOBOFormatVersion rdf:datatype="http://www.w3.org/2001/XMLSchema#string">1.
28     </owl:Ontology>
29
30
31
32     <!--
33     ///////////////////////////////////////////////////////////////////////////////////////
34     //
35     // Annotation properties
```

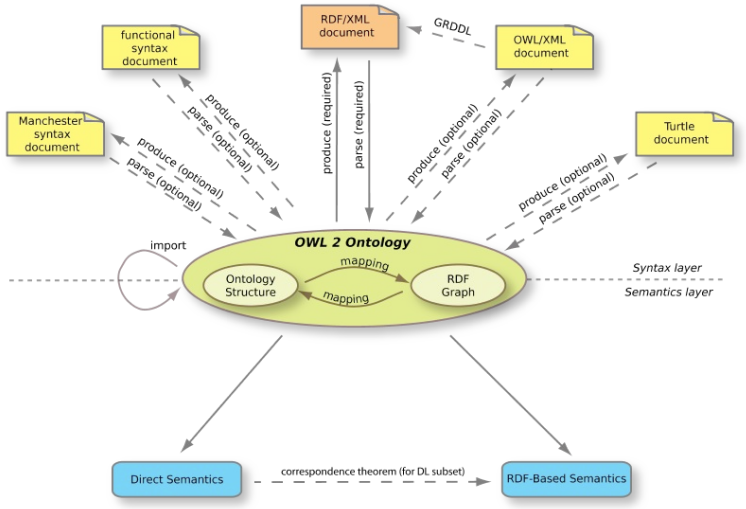
RDF/XML level data

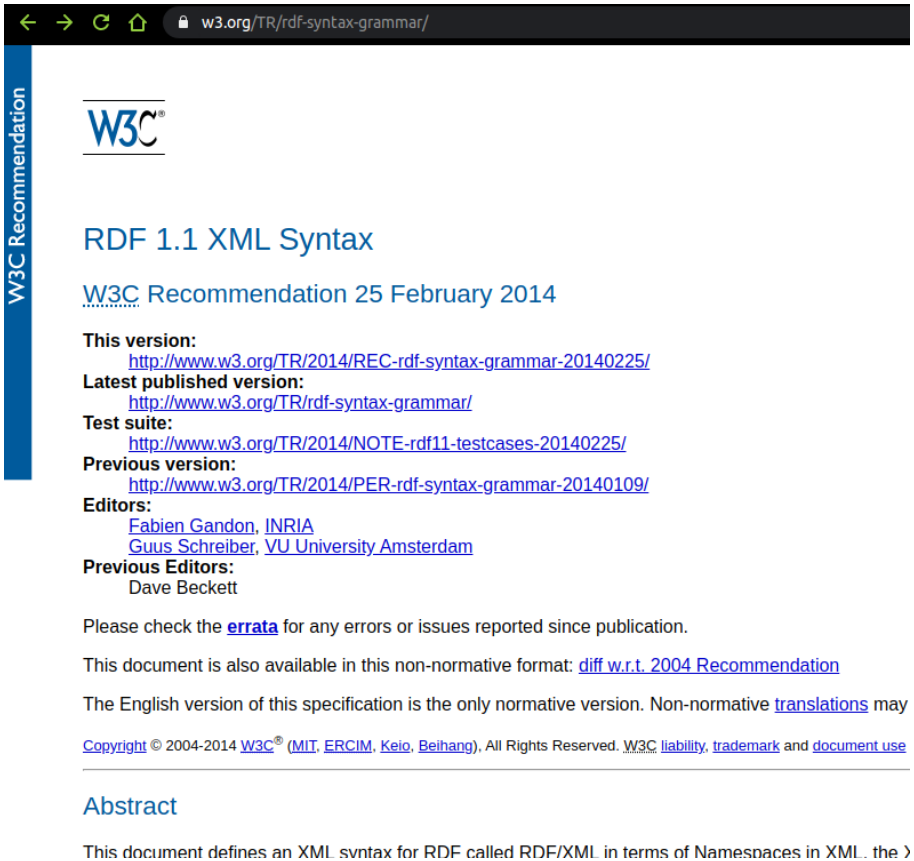
w3.org/2002/07/owl

```
@prefix dc: <http://purl.org/dc/elements/1.1/> .
@prefix grddl: <http://www.w3.org/2003/g/data-view#> .
@prefix owl: <http://www.w3.org/2002/07/owl#> .
@prefix rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#> .
@prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#> .
@prefix xml: <http://www.w3.org/XML/1998/namespace> .
@prefix xsd: <http://www.w3.org/2001/XMLSchema#> .

<http://www.w3.org/2002/07/owl> a owl:Ontology ;
     dc:title "The OWL 2 Schema vocabulary (OWL 2)" ;
     rdfs:comment """
  This ontology partially describes the built-in classes and
  properties that together form the basis of the RDF/XML syntax of OWL 2.
  The content of this ontology is based on Tables 6.1 and 6.2
  in Section 6.4 of the OWL 2 RDF-Based Semantics specification,
  available at http://www.w3.org/TR/owl2-rdf-based-semantics/.
  Please note that those tables do not include the different annotations
  (labels, comments and rdfs:isDefinedBy links) used in this file.
  Also note that the descriptions provided in this ontology do not
  provide a complete and correct formal description of either the syntax
  or the semantics of the introduced terms (please see the OWL 2
  recommendations for the complete and normative specifications).
  Furthermore, the information provided by this ontology may be
  misleading if not used with care. This ontology SHOULD NOT be imported
  into OWL ontologies. Importing this file into an OWL 2 DL ontology
  will cause it to become an OWL 2 Full ontology and may have other,
  unexpected, consequences.
   """ ;
     rdfs:isDefinedBy
          <http://www.w3.org/TR/owl2-mapping-to-rdf/>,
          <http://www.w3.org/TR/owl2-rdf-based-semantics/>,
          <http://www.w3.org/TR/owl2-syntax/> ;
     rdfs:seeAlso   <http://www.w3.org/TR/owl2-rdf-based-semantics/#table-axiomatic-classes>,
                    <http://www.w3.org/TR/owl2-rdf-based-semantics/#table-axiomatic-properties> ;
     owl:imports <http://www.w3.org/2000/01/rdf-schema> ;
     owl:versionIRI <http://www.w3.org/2002/07/owl> ;
     owl:versionInfo "$Date: 2009/11/15 10:54:12 $" ;
     grddl:namespaceTransformation <http://dev.w3.org/cvsweb/2009/owl-grddl/owx2rdf.xsl> .


owl:AllDifferent a rdfs:Class ;
     rdfs:label "AllDifferent" ;
     rdfs:comment "The class of collections of pairwise different individuals." ;
     rdfs:isDefinedBy <http://www.w3.org/2002/07/owl#> ;
     rdfs:subClassOf rdfs:Resource .

owl:AllDisjointClasses a rdfs:Class ;
     rdfs:label "AllDisjointClasses" ;
     rdfs:comment "The class of collections of pairwise disjoint classes." ;
     rdfs:isDefinedBy <http://www.w3.org/2002/07/owl#> ;
     rdfs:subClassOf rdfs:Resource .
```

XML Syntax - IRIs

OWL/XML
RDF/XML
Turtle
Manchester





https://www.w3.org/TR/owl2-overview/OWL2-structure2-800.png

‘CrossOntology Term Parser’ function

Available via SearchviaCombinedOntology.Rmd

```
> str(combined_output)
List of 14
 $ id          : chr [1:204] "http://purl.bioontology.org/ontology/MEDDRA/10029267" "http://purl.bioontology.org/ontology/SNOMEDCT/8908400
2" "http://purl.bioontology.org/ontology/SNOMEDCT/404029005" "http://purl.bioontology.org/ontology/RCD/Xa99R" ...
 $ prefLabel   : chr [1:114] "Neurofibroma" "neurofibroma" "NEUROFIBROMA" "Plexiform neurofibroma" ...
 $ obsolete    : logi FALSE
 $ subClassOf  : chr [1:150] "http://purl.bioontology.org/ontology/MEDDRA/10029210" "http://purl.bioontology.org/ontology/SNOMEDCT/4001930
07" "http://purl.bioontology.org/ontology/SNOMEDCT/402875000" "http://purl.bioontology.org/ontology/RCD/X77pm" ...
 $ definition  : chr [1:49] "An intraneural or extraneural neoplasm arising from nerve tissues and neural sheaths. It is composed of perin
eu"| __truncated__ "A moderately firm, benign, encapsulated tumor resulting from proliferation of SCHWANN CELLS and FIBROBLASTS tha"| __tr
uncated__ "moderately firm, benign, encapsulated tumor resulting from proliferation of Schwann cells and fibroblasts that "| __truncated__
 "A moderately firm, benign, encapsulated tumor resulting from proliferation of schwann cells and fibroblasts tha"| __truncated__ ...
 $ synonym     : chr [1:231] "Neurofibroma, no ICD-O subtype" "Neurofibroma (disorder)" "neurofibroma" "Neurofibroma (WHO Grade I)" ...
 $ cui         : chr [1:33] "C0027830" "C0206728" "C1096349" "C1275263" ...
 $ semanticType: chr [1:4] "http://purl.bioontology.org/ontology/STY/T191" "http://purl.bioontology.org/ontology/STY/T061" "http://purl.bi
oontology.org/ontology/STY/T033" "http://purl.bioontology.org/ontology/STY/T047"
 $ label       : chr [1:65] "Neurofibroma" "neurofibroma" "RID4462" "神経線維腫" ...
 $ prefixIRI   : chr [1:80] "ncit:C3272" "RID4462" "efo:EFO_0000622" "C0027830" ...
 $ notation    : chr [1:101] "10029267" "89084002" "404029005" "Xa99R" ...
 $ xref        : chr [1:33] "SNOMEDCT_US_2016_03_01:89084002" "MESH:D009455" "DOID:962" "UMLS:C0027830" ...
 $ @id         : chr [1:204] "http://purl.bioontology.org/ontology/MEDDRA/10029267" "http://purl.bioontology.org/ontology/SNOMEDCT/8908400
2" "http://purl.bioontology.org/ontology/SNOMEDCT/404029005" "http://purl.bioontology.org/ontology/RCD/Xa99R" ...
 $ @type       : chr "http://www.w3.org/2002/07/owl#Class"
>
```

Next Steps

- Create Protege (RDF/XML) readable mappings
  - Potentially use OWLAPI java service for industry standard mapping with OWL namespace
- Ontology source dependant rankings matrix
- Output custom RDF graph via R
- Trace out Ontology tree per unique connections
- Add Natural Language Processing engine to potentially parse connections from various sources (ex. Journal Articles, transcripts of visitations, etc)