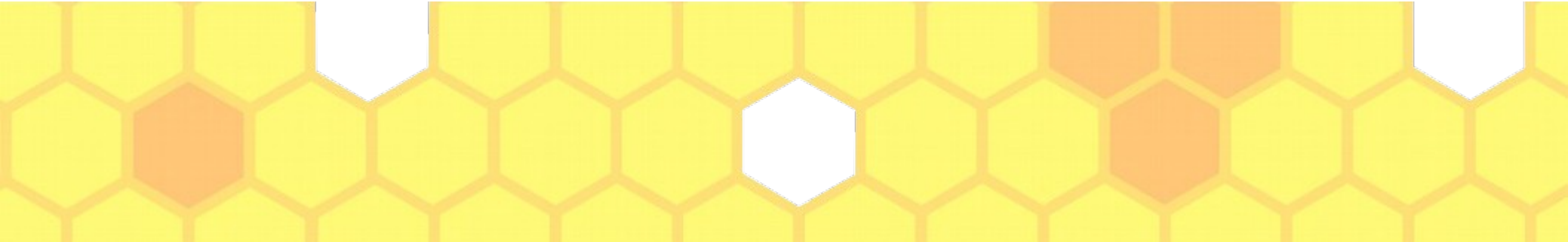




Collaborative Ontology for Neurofibromatosis

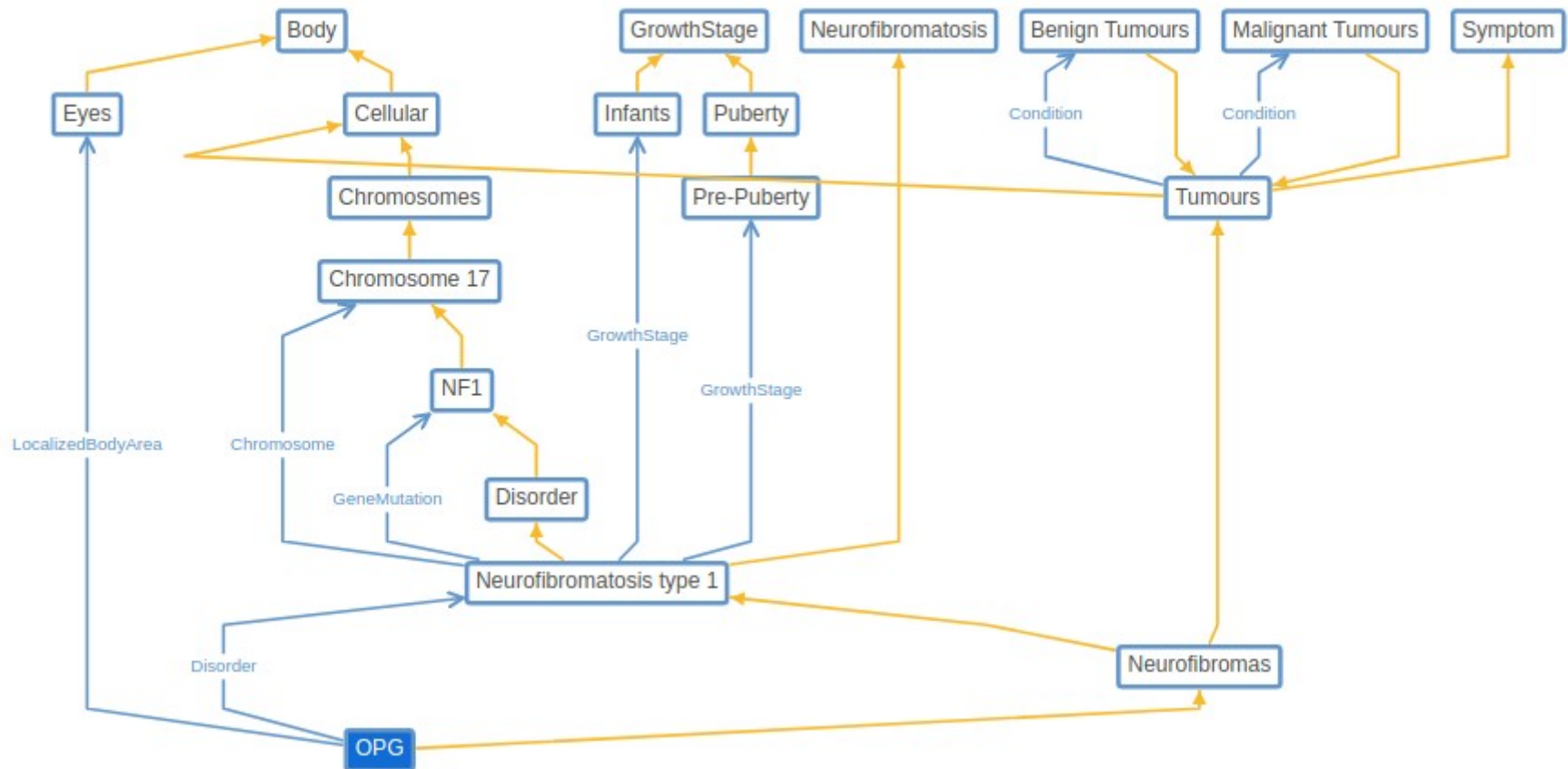
Hack for NF 2020
By: Krishna Udaiwal



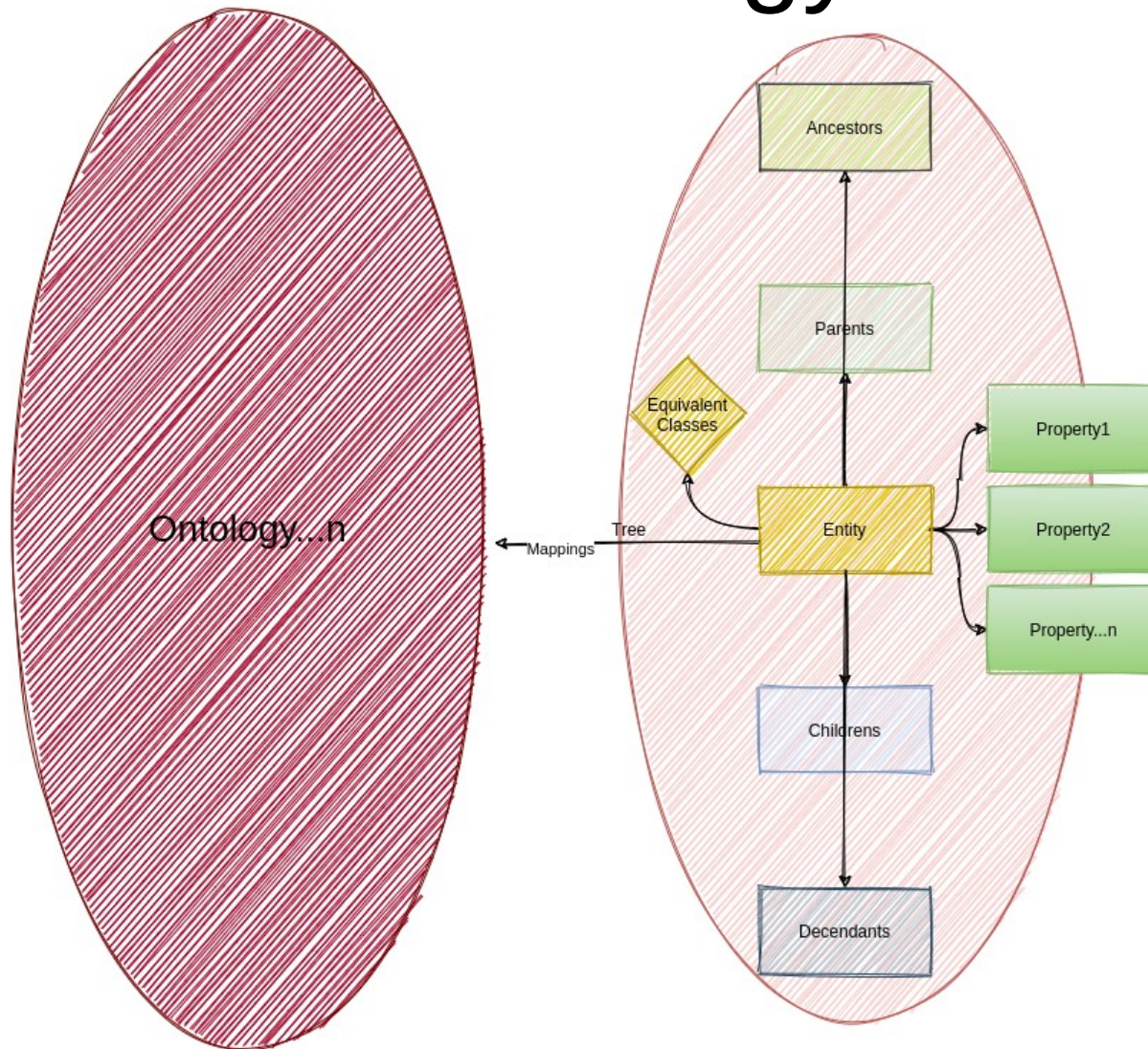
Manual Ontology Attempt

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

The screenshot displays the Protege Ontology Editor interface. The top menu bar includes File, Edit, View, Reasoner, Tools, Refactor, Window, Mastro, Ontop, and Help. The main window shows the 'untitled-ontology-12' with a search bar. The 'Active ontology' tab is selected, showing the 'Ontology header' with the IRI and Version IRI. The 'Ontology metrics' panel on the right lists various counts: Axiom (5658140), Logical axiom count (1154531), Declaration axioms count (505193), Class count (404091), Object property count (1268), Data property count (104), Individual count (98975), and Annotation Property count (789). The 'Class axioms' section shows SubClassOf (783773), EquivalentClasses (106074), DisjointClasses (746), GCI count (54), and Hidden GCI Count (90199). The 'Object property axioms' section shows SubObjectPropertyOf (917), EquivalentObjectProperties (10), InverseObjectProperties (225), DisjointObjectProperties (12), FunctionalObjectProperty (15), InverseFunctionalObjectProperty (3), TransitiveObjectProperty (110), SymmetricObjectProperty (93), and AsymmetricObjectProperty (2). The 'Ontology imports' panel at the bottom shows a list of imported ontologies, including 'http://livercancer.imbi.uni-heidelberg.de/ccont', 'http://ontology.neuinfo.org/NIF/ftl/NIF-Dysfunction.ttl', and 'http://purl.bioontology.org/ontology/OMIM/

Protege Results

Datatypes | **Individuals** | **Class hierarchy (inferred)**

Classes | **Object properties** | **Data properties** | **Annotation properties**

Class hierarchy: Neurofibroma

- Class hierarchy icons
- Asserted
- Disease
 - Cancer
 - Cardiovascular Disease
 - Connective and Soft Tissue Disorder
 - Cystic Keratinizing Epithelioma
 - Endocrine Disorder
 - Fibroepithelial Polyp
 - Liver and Biliary Tract Disorder
 - Metabolic Disorder
 - Neoplasm
 - Hamartoma
 - Neoplasm by Morphology
 - Neoplasm by Site
 - Breast Neoplasm
 - Cardiovascular Neoplasm
 - Connective and Soft Tissue Neoplasm
 - Endocrine Neoplasm
 - Eye Neoplasm
 - Gastrointestinal Neoplasm
 - Head and Neck Neoplasm
 - Hematopoietic and Lymphoid System Neoplasm
 - Nervous System Neoplasm
 - Benign Nervous System Neoplasm
 - Central Nervous System Neoplasm
 - Cranial Nerve Neoplasm
 - Malignant Nervous System Neoplasm
 - Neuroepithelial, Perineurial, and Schwann Cell
 - Peripheral Nervous System Neoplasm
 - Autonomic Nervous System Neoplasm
 - Benign Peripheral Nerve Granular Cell Tumo
 - Childhood Medialastinal Neurogenic Neoplas
 - Cutaneous Neural Neoplasm
 - Hemangioma of Peripheral Nerve
 - Intraneural Perineuroma
 - Lipomatosis of Nerve
 - Malignant Peripheral Nervous System Neopl
 - Nerve Plexus Neoplasm
 - Nerve Root Neoplasm
 - Neurofibroma**
 - Atypical Neurofibroma
 - Cardiac Neurofibroma
 - Cellular Neurofibroma
 - Diffuse Neurofibroma
 - Epithelioid Neurofibroma
 - Esophageal Neurofibroma
 - Gallbladder Neurofibroma
 - Mediastinal Neurofibroma
 - Melanotic Neurofibroma

Annotations: Neurofibroma

Annotations +

label [type: xsd:string]
Neurofibroma

rdfs:isDefinedBy [type: xsd:string]
An intraneural or extraneural neoplasm arising from nerve tissues and neural sheaths. It is composed of perineurial-like fibroblasts and Schwann cells. It usually presents as a localized cutaneous lesion and less often as a circumscribed peripheral nerve mass. Patients with neurofibromatosis type 1 present with multiple masses. Neurofibromas which arise from major nerves and plexiform neurofibromas are precursor lesions to malignant peripheral nerve sheath tumors.

Synonym [type: xsd:string]
Neurofibroma (WHO Grade I)

Synonym [type: xsd:string]
NEUROFIBROMA, BENIGN

Description: Neurofibroma

Equivalent To +

SubClass Of +

- 'Nerve Sheath Neoplasm'
- 'Peripheral Nervous System Neoplasm'

General class axioms +

SubClass Of (Anonymous Ancestor)

- quality or realizable_entity
- continuant or occurrent
- generically_dependent_continuant or
- dependent_continuant or independent

Instances +

Target for Key +

Disjoint With +

Disjoint Union Of +

Search

neurofibroma

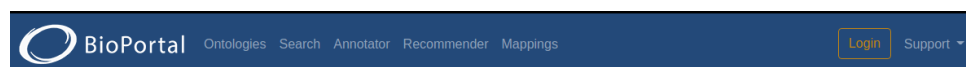
☐ Case sensitive ☐ Whole words ☒ Ignore white space ☒ Regular expression ☐ Show all results

☒ Search in IRIs ☒ Search in annotation values ☒ Search in logical axioms

Found in	Entity	Id	Match
Display name	Neurofibroma		Neurofibroma
	Neurofibroma	birnlex:12604	Neurofibroma
	Neurofibroma		Neurofibroma
	Neurofibromas	HP:0001067	Neurofibromas
	Neurofibromatoses		Neurofibromatoses
	Neurofibromatosis	birnlex:12605	Neurofibromatosis
	Neurofibromatosis		Neurofibromatosis
	neurofibroma	DOID:962	neurofibroma
	neurofibroma	MPATH:435	neurofibroma
	neurofibroma	EFO:0000622	neurofibroma
	71 more results...		
IRI	'Neurofibromatosis 1 and 2 (NF1 and Neurofibroma		http://ncicb.nci.nih.gov
	'Cardiac Neurofibroma'		http://ncicb.nci.nih.gov
	'Diffuse Neurofibroma'		http://ncicb.nci.nih.gov
	'Atypical Neurofibroma'		http://ncicb.nci.nih.gov
	'Cellular Neurofibroma'		http://ncicb.nci.nih.gov
	'Pacinian Neurofibroma'		http://ncicb.nci.nih.gov
	'Melanotic Neurofibroma'		http://ncicb.nci.nih.gov
	'Plexiform Neurofibroma'		http://ncicb.nci.nih.gov
	'Esophageal Neurofibroma'		http://ncicb.nci.nih.gov
	+ 4 more results...		
'alternative label'	'17q11 microdeletion syndrome'		'alternative label' [lan
	'17q11 microdeletion syndrome'		'alternative label' [lan

Copy selected entities

BioPortal Ontologies Repo

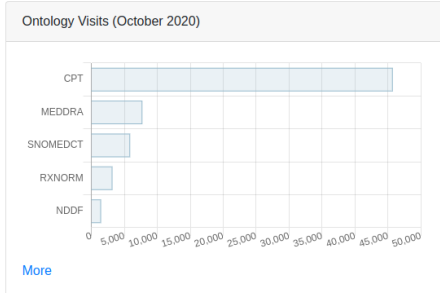


Welcome to BioPortal, the world's most comprehensive repository of biomedical ontologies

Search for a class

Enter a class, e.g. Melanoma

[Advanced Search](#)



Find an ontology

Start typing ontology name, then choose from list

[Browse Ontologies](#)

BioPortal Statistics

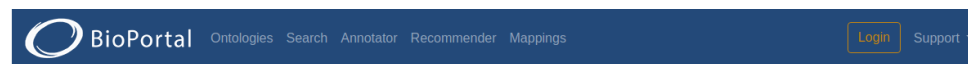
Ontologies	901
Classes	12,175,540

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[Class](#)
[Group](#)
[Instance](#)
[Mapping](#)
[Metric](#)
[Note](#)
[Reply](#)
[ObjectProperty](#)
[Ontology](#)
[OntologySubmission](#)
[Project](#)
[ProvisionalClass](#)
[ProvisionalRelation](#)
[Review](#)
[Slice](#)
[User](#)
[Content Types](#)



Medical Dictionary for Regulatory Activities Terminology (MedDRA)

Last updated: September 24, 2020



[Summary](#) [Classes](#) [Properties](#) [Notes](#) [Mappings](#) [Widgets](#)

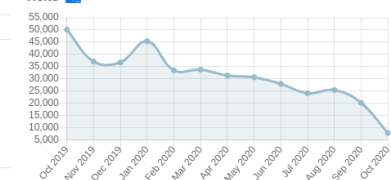
Details

Acronym	MEDDRA
Visibility	Public
Description	MedDRA is an international medical terminology with an emphasis on use for data entry, retrieval, analysis, and display. It applies to all phases of drug development, excluding animal toxicology, and to the health effects and malfunction of devices. An appendix includes concept descriptions which describe how a medical concept is interpreted, used, and classified within the MedDRA terminology. It is not intended as a medical definition. The concept descriptions are intended to aid the consistent and accurate use of MedDRA in coding, retrieval, and analysis.
Status	Production
Format	UMLS
Contact	MedDRA MSSO, mssohelp@meddra.org
Categories	Health
Groups	Cancer Biomedical Informatics Grid, Unified Medical Language System
License Information	The MedDRA ontology is maintained and distributed by the MedDRA MSSO . This ontology is freely accessible on this site for academic and other non-commercial uses. Users anticipating any commercial use of MedDRA must contact the MSSO

Metrics

Classes	74,056
Individuals	0
Properties	14
Maximum depth	3
Maximum number of children	268
Average number of children	17
Classes with a single child	74
Classes with more than 25 children	435
Classes with no definition	73,831

Visits



Projects using MEDDRA

API Documentation

General Usage

This API is comprised of a set of resources (Ontologies, Classes, etc) and related endpoints (Search, Annotator, Recommender) that are connected together via links, much like webpages. We recommend that you try browsing the API using a web browser (Chrome and Firefox work very well while IE does not) before you start writing code. For more information, please see the documentation on [Media Types and Hypermedia Links](#) or view our [sample code](#), available in Java, Python, Ruby and other languages (please email support@bioontology.org if you would like examples in another language).

Common Parameters

Parameter	Possible Values	Description
apikey	{your api key}	An API Key is required to access any API call. It can be provided in three ways: 1. Using the <code>apikey</code> query string parameter 2. Providing an <code>Authorization</code> header: <code>Authorization: apikey token=your_apikey</code> (replace 'your_apikey' with your actual key) 3. When using a web browser to explore the API, if you provide your API Key once using method 1, it will be stored in a cookie for subsequent requests. You can override this by providing a different API Key in a new call.
include	all (comma-separated list of attributes, EX: attr1,attr2)	By default, the API will show a subset of the available attributes for a given media type. This behavior can be overridden by providing <code>include=all</code> to show all attributes or <code>include=attribute1,attribute2</code> to include a specific list. The API is optimized to return the default values, so overriding this can impact the performance of your request. The <code>include=all</code> option is most useful for testing in the browser. Use it to identify the set of attributes required and use only those by passing them as a comma separated list, e.g. <code>include=prefLabel1,cat1</code> . The <code>include</code> parameter is currently unsupported on Annotator and Recommender endpoints.
format	json jsonp xml	The API returns JSON as the default content type. This can be overridden by using the <code>format</code> query string parameter. The API also respects <code>Accept</code> header entries, with precedence given to the <code>format</code> parameter.
page	{integer}	For calls that are paged, this will indicate which page number you want to

OWL (RDF/XML format)

```
Ontologies > pato.owl
1 <?xml version="1.0"?>
2 <rdf:RDF xmlns="http://purl.obolibrary.org/obo/pato.owl#"
3   xmlns: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/pato-2020-08-02.owl" />
22   <obo:IA0_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

```
< http://www.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#> .

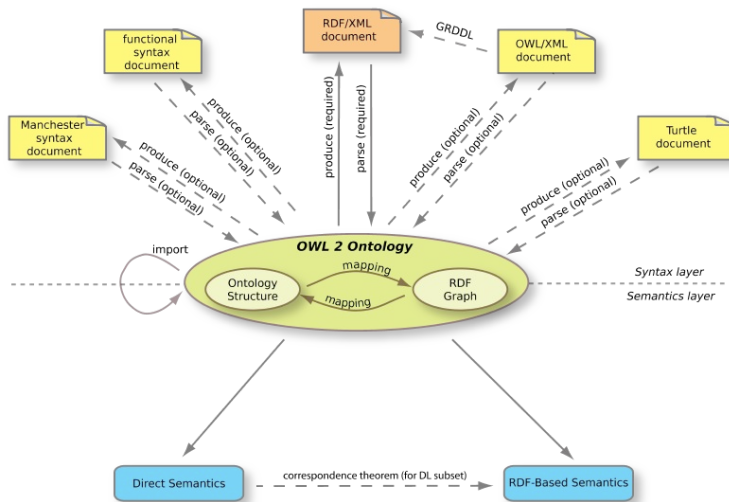
<http://www.w3.org/2002/07/owl> a owl:Ontology ;
  dc:title "The OWL 2 Schema vocabulary (OWL 2)" ;
  rdfs:comment """
This ontology partially describes the built-in classes and
properties that together form the basis of the RDF/XML syntax of OWL 2.
The content of this ontology is based on Tables 6.1 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/owl2rdf.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



← → ↻ 🏠 w3.org/TR/rdf-syntax-grammar/

W3C Recommendation

W3C

RDF 1.1 XML Syntax

W3C Recommendation 25 February 2014

This version:
<http://www.w3.org/TR/2014/REC-rdf-syntax-grammar-20140225/>

Latest published version:
<http://www.w3.org/TR/rdf-syntax-grammar/>

Test suite:
<http://www.w3.org/TR/2014/NOTE-rdf11-testcases-20140225/>

Previous version:
<http://www.w3.org/TR/2014/PER-rdf-syntax-grammar-20140109/>

Editors:
[Fabien Gandon](#), [INRIA](#)
[Guus Schreiber](#), [VU University Amsterdam](#)

Previous Editors:
Dave Beckett

Please check the [errata](#) for any errors or issues reported since publication.

This document is also available in this non-normative format: [diff w.r.t. 2004 Recommendation](#)

The English version of this specification is the only normative version. Non-normative [translations](#) may

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Abstract

This document defines an XML syntax for RDF called RDF/XML in terms of Namespaces in XML. the ›

<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
ontology.org/ontology/STY/T033" "http://purl.bioontology.org/ontology/STY/T047"
 $ label      : chr [1:65] "Neurofibroma" "neurofibroma" "RID4462" "神經線維腫" ...
 $ 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)

