

# BioPortal: A Web Repository and Services for Biomedical Ontologies and Data Resources

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# What are we trying to do

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- You've built an ontology, how do you let the world know?
- You need an ontology, where do you go to get it?
- How do you know whether an ontology is any good?
- How do you find resources that are relevant to the domain of the ontology (or to specific terms)?

# BioPortal: A Community-Based Ontology Repository

The screenshot shows the BioPortal website interface. At the top, there is a navigation bar with tabs for 'Browse', 'Search', 'Projects', 'Annotate', 'All Mappings', and 'All Resources Alpha'. On the right side of the navigation bar, there are links for 'Sign In', 'Register', 'Help/About', and 'Send Feedback'. Below the navigation bar, the main content area is titled 'human phenotype ontology'. There are three search boxes: 'Search all ontologies', 'Find an ontology', and 'Search resources'. Below these are three columns of content: 'Most Active Ontologies' (a table), 'Latest Notes' (a list of updates), and 'Latest Mappings' (a list of ontology relationships).

**Most Active Ontologies**

Ontology	Version	Notes	Mappings
<a href="#">Human disease</a>	1.36	0	17732
<a href="#">Mouse adult gross anatomy</a>	1.194	0	3905
<a href="#">NCI Thesaurus</a>	08.12d	9	3798
<a href="#">Foundational Model of Anatomy</a>	3.0	0	1997
<a href="#">Zebrafish anatomy and development</a>	1.21	0	791

**Latest Notes**

- [change namespace from IAO to OBI material entity \(Ontology for Biomedical Investigations\)](#) 06/10/09 whetzel
- [RE:Add mapping to Unit ontology? unit \(Experimental Factor Ontology\)](#) 06/01/09 jamesmalone
- [Add mapping to Unit ontology? unit \(Experimental Factor Ontology\)](#) 04/12/09 whetzel
- [Release notes for version 2009-04-02 entity \(NanoParticle Ontology\)](#) 04/02/09 sobolevnm
- [Missing preferred term field OBI\\_0000577 \(Ontology for Biomedical Investigations\)](#) 04/01/09 whetzel

**Latest Mappings**

- [cellular component \(Biological process\) => Cell component \(Foundational Model of Anatomy\)](#) 04/27/09 matthiassamwald
- [Cell component \(Foundational Model of Anatomy\) => cellular component \(Biological process\)](#) 04/27/09 matthiassamwald
- [Cell \(Foundational Model of Anatomy\) => Cell \(NCI Thesaurus\)](#) 04/16/09 lechatpito
- [Cell \(NCI Thesaurus\) => Cell \(Foundational Model of Anatomy\)](#) 04/16/09 lechatpito
- [tbio:Cell \(Basic Vertebrate Anatomy\) => Cell \(NCI Thesaurus\)](#) 04/16/09 lechatpito

# The National Center for Biomedical Ontology

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- One of the five National Centers for Biomedical Computing launched by NIH
- Collaboration of Stanford, Mayo, Buffalo, Victoria, Medical College of Wisconsin, Washington University, John Hopkins
- Primary goal is to make ontologies accessible and usable
- Research develops technologies for ontology dissemination, indexing, alignment, and peer review

# Key Technology: BioPortal

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- Web accessible repository of ontologies for the biomedical community:
  - <http://bioportal.bioontology.org>
- Online support for ontology
  - Peer review
  - Notes (comments and discussion)
  - Versioning
  - Mapping
  - Search
  - Resources

# The BioPortal Technology

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- BioPortal is a library of biomedical ontologies
- All BioPortal data is accessible through **REST services**
  - BioPortal user interface accesses the repository through REST services as well
- The BioPortal technology is **domain-independent**
  - There are installations of BioPortal for libraries in other domains
  - BioPortal code is open-source

# BioPortal Library

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- 140 ontologies in OWL, RDFS, Protégé frames, and OBO
  - Protégé for OWL, RDFS, frames
  - Mayo Clinic's LexGrid for OBO
- 700,000 classes in the ontologies
- 30,000 concept-to-concept mappings

# A Library of Biomedical Ontologies


BioPortal [Browse](#) [Search](#) [Projects](#) [Annotate](#) [All Mappings](#) [All Resources Alpha](#) [Sign In](#) [Register](#)

[SUBMIT ONTOLOGY](#) [Submit New Ontology](#)

**FILTER BY CATEGORY**

**FILTER BY GROUP**  [Link To This Filter](#)

**FILTER BY TEXT**

 [Subscribe to all ontologies](#)

ONTOLOGY NAME	FORMAT	VERSION	AUTHOR	UPLOADED ON	GROUP	STATUS
<a href="#">ABA Adult Mouse Brain (ABA)</a>	OWL	1.0	Chinh Dang	06/12/2009		<a href="#">Explore</a>
<a href="#">African Traditional Medicine (ATMO)</a>	OBO Format	1.0.1	Ghislain Ateazing	04/23/2009		<a href="#">Explore</a>
<a href="#">Amino Acid (amino-acid)</a>	OWL	1.2	Nick Drummond, Georgina Moulton, Robert Stevens, Phil Lord	04/25/2009		<a href="#">Explore</a>
<a href="#">Amino Acid with Simplified Chinese annotations (Amino Acid-zh_CN)</a>	DWL	vl.2 zh1.1	Nick Drummond, Georgina Moulton, Robert Stevens, Phil Lord. Annotated by: Lin Zhang	04/27/2009		<a href="#">Explore</a>
<a href="#">Amphibian gross anatomy (AAO)</a>	OBO Format	1.8	AmphiAnat list	07/30/2008	OBOFoundry	<a href="#">Explore</a>
<a href="#">Animal natural history and life history (ADW)</a>	PROTEGE	See Remote Site	<a href="http://animaldiversity">Http://animaldiversity</a> Administrators	04/27/2009		
<a href="#">Ascomycete phenotype ontology (APO)</a>	OBO Format	1.2	Fungal_anatomy Administrators	05/02/2009		<a href="#">Explore</a>
<a href="#">Basic Vertebrate Anatomy (basic-vertebrate-gross-anatomy)</a>	OWL	1.1		01/16/2007		<a href="#">Explore</a>
<a href="#">Bilateria anatomy (BILA)</a>	OBO Format	See Remote Site	Thorsten Heinrich	04/13/2009		



# Browsing Ontologies

Biomedical Resource Ontology Version 2.7

BRO:Software | [Link Here](#) | [Subscribe](#)

View Ontology Details

Jump To:

- activity:Activity
- area:Area\_of\_Research
- BRO:Deprecated\_Resource
- BRO:Resource

  - BRO:Funding\_Resource
  - BRO:Information\_Resource
  - BRO:Material\_Resource
  - BRO:People\_Resource
  - BRO:Service\_Resource
  - BRO:Software
  - BRO:Training\_Resource

- desc:Biositemaps\_Information\_Model
- area:Deprecated\_Area\_of\_Research
- activity:Deprecated\_Activity

Details Visualization Notes Mappings Resources [alpha](#)

Show Network Neighborhood

```
graph BT; subClassOf1[BRO:Software] -- subClassOf --> BRO_Resource[BRO:Resource]; subClassOf2[Knowledge_Mining_and_Capturing] -- subClassOf --> BRO_Software[BRO:Software]; subClassOf3[Algorithm] -- subClassOf --> BRO_Software; subClassOf4[Software_Distribution] -- subClassOf --> BRO_Software; subClassOf5[Interactive_Tool] -- subClassOf --> BRO_Software;
```

The diagram illustrates the class hierarchy for BRO:Software. It is shown as a subclass of BRO:Resource. Additionally, it is a superclass for several other classes: Knowledge\_Mining\_and\_Capturing, Algorithm, Software\_Distribution, and Interactive\_Tool. The BRO:Software node is highlighted with a green border.

# Ontology Search

**Search all ontologies** Search time: 3.3 s

Search:

Include attributes in search

Contains  Exact Match

Selected Ontologies (111):

Categories:  Filter:

**Ontologies:** [Select All](#) [Select None](#)

- ABA Adult Mouse Brain (ABA)
- African Traditional Medicine (ATMO)
- Amino Acid (amino-acid)
- Amino Acid with Simplified Chinese annotations
- Amphibian gross anatomy (AAO)

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**Matching Concepts** 427 results

Filter:   Exact Matches Only

Concept Name	Ontology	Found In
<a href="#">Melanoma</a>	<a href="#">Cell line ontology</a>	Preferred Name
<a href="#">Melanoma</a>	<a href="#">Galen</a>	Preferred Name
<a href="#">melanoma</a>	<a href="#">Mouse pathology</a>	Preferred Name
<a href="#">Melanoma</a>	<a href="#">NCI Thesaurus</a>	Preferred Name
<a href="#">melanoma</a>	<a href="#">Experimental Factor Ontology</a>	Preferred Name
<a href="#">Malignant melanoma</a>	<a href="#">human phenotype ontology</a>	Synonym
<a href="#">AMELANOTIC MELANOMA</a>	<a href="#">DermLex: The Dermatology Lexicon</a>	Preferred Name
<a href="#">Amelanotic Melanoma</a>	<a href="#">NCI Thesaurus</a>	Preferred Name
<a href="#">Anal Melanoma</a>	<a href="#">NCI Thesaurus</a>	Preferred Name
<a href="#">ANGIOMATOID MELANOMA</a>	<a href="#">DermLex: The Dermatology Lexicon</a>	Preferred Name
<a href="#">Breast Melanoma</a>	<a href="#">NCI Thesaurus</a>	Preferred Name
<a href="#">BULLOUS MELANOMA</a>	<a href="#">DermLex: The Dermatology Lexicon</a>	Preferred Name
<a href="#">Cervical Melanoma</a>	<a href="#">NCI Thesaurus</a>	Preferred Name
<a href="#">CHONDROID MELANOMA</a>	<a href="#">DermLex: The Dermatology Lexicon</a>	Preferred Name

# BioPortal is allowing NCBO to experiment with new models for

- **Dissemination** of knowledge on the Web
- **Integration** and alignment of online content
- Knowledge visualization and **cognitive support**
- **Peer review** of online content

# Notes on Classes

Biomedical Resource Ontology Version 2.7

BRO:Software | [Link Here](#) | [Subscribe](#)

View Ontology Details

Jump To:

- activity:Activity
- area:Area\_of\_Research
- BRO:Deprecated\_Resource
- BRO:Resource

  - BRO:Funding\_Resource
  - BRO:Information\_Resource
  - BRO:Material\_Resource
  - BRO:People\_Resource
  - BRO:Service\_Resource
  - BRO:Software
  - BRO:Training\_Resource

- desc:Biositemaps\_Information\_Model
- area:Deprecated\_Area\_of\_Research
- activity:Deprecated\_Activity

Details Visualization **Notes** Mappings Resources [alpha](#)

**Comment:** Software needs structure, too many top level subclasses DavidStates at 08/09/08 06:56

"binary executable" is not a top level subclass of software, it is a form of software distribution and there are several other subclasses of software distribution (source code, web site, library, toolkit, etc.).

Similarly, "network editor" is just one class of interactive editing tools. Lots of others.

These are just a couple of examples. Software really needs a complete reorganization.

[Reply](#)

**Comment:** RE:Software needs structure, too many top level subclasses PeterLyster at 08/12/08 08:29

The BRO used the initial design principle of: when in doubt make it flat at the top. This is a design principle whose purpose is to get the class names 'on the board and agreed upon' first, i.e., it is a componentization of the design process. This is a way of avoiding getting into debates about hierarchical location too early in the process. We can discuss location in the hierarchy in the future; that is appropriate.

[Reply](#)

**Comment:** RE:Software needs structure, too many top level subclasses PeterLyster at 08/12/08 08:43

I (Peter Lyster) copy marginal notes that I also place in the 'Portals' class. I think this helps to explain the design principles.

We adopted the design principle of (i) initially align the BRO top level with NIFSTD (**Data Resource; Bibliographic**

# Users create notes in order to

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- discuss class definitions
- suggest changes and corrections
- request new items
- provide additional information about a class (e.g., references, supporting documentation)

# Projects in BioPortal

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- Users can describe their **ontology-based projects** in BioPortal
- Projects are linked to ontologies they are using
- Ontology reviews can be done in the context of projects

# Reviewers Provide

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- General review and rating
- **Usage** information
  - Which applications have successfully used the ontology?
  - What problems were encountered?
- **Coverage**
  - Does it cover the domain properly?
  - Are there major gaps?
  - Are some parts developed better than others?
- **Concept-specific comments**
  - Are there problems with specific concepts?
  - What alternative definitions should be used?

# An Experiment: Community-Based Annotation as Peer Review

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- Potentially makes ontology evaluation a democratic process
- Assumes users' application of ontologies will lead to insights not achievable by inspection alone
- Assumes end-users will be motivated to comment on and engage in dialog about ontologies in the repository



# Ontology Evaluation

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- Ontologies are not like journal articles:
  - It is difficult to judge methodological soundness simply by inspection
  - We may wish to use an ontology even though some portions
    - Are not well designed
    - Make distinctions that are different from those that we might want

# Computable ontology metrics

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- Logical consistency checking
  - e.g., use a reasoner to determine if an ontology is consistent
- Structural consistency based on meta-properties
- Rules of thumbs and heuristics
- Statistical information
  - number of classes and properties
  - connectedness, fan-out, etc.
  - cycles

# Not all useful metrics are computable

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- Many aspects of ontology quality are *subjective*
- The most useful information for the user selecting an ontology:
  - *who has used an ontology for a similar task and how well did it work?*

# Some Ontology Metrics are Subjective

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What is a “good” feature in some setting, can be a “bad” feature in another setting

- High level of **axiomatization**:
  - **good** if you want to perform reasoning
  - can be **bad** because of the high computational and cognitive cost if you don't need the axioms
- **Organizing** anatomy concepts based primarily on their structure rather than function
  - can be **good** if you need to understand which organs a wound goes through
  - **not appropriate** if you need to understand spread of disease

# Community-Based Evaluation

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*Which ontology from the library is appropriate for my task?*

- The only people who know the answer to these question are
  - (maybe) ontology authors
  - other users of the ontology
- Allow users to provide ratings for ontologies

# Conflicting Sources of Metadata

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- Authors and users can contradict one another
  - Quality of documentation?
  - References (e.g., positive and negative analyses of the ontology)
  - ...
- Metadata schema must enable diversity of views on some metadata values

# Ontology Mappings

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- BioPortal ontologies overlap in content
  - The same is true for almost any ontology library
- A **mapping** between two terms in different ontologies indicates a relationship between them
  - Usually a **similarity** relationship
  - For example, *nostril* in NCI Thesaurus is similar to *naris* in Mouse Anatomy Ontology

# Mappings in BioPortal

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- Mappings in BioPortal are **concept to concept** mappings
- Mappings are created by users or uploaded in bulk
- Bulk uploads are usually the results of automatic or semi-automatic mapping
- There is detailed **metadata** for provenance of mappings
- ~30,000 mappings in BioPortal now
  - The number will dramatically increase (to millions) in the coming weeks



# Why do you need mappings?

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- Annotation of resources with terms from different ontologies
- Information integration
- Ontology integration
- ..... *many other uses*

# Using BioPortal Mappings

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- Mappings are used for query expansion in information extraction
- Mappings can be used as navigation mechanism, linking one ontology to another
- Mappings may indicate which ontologies are “important”
  - If everyone tries to map their ontology to NCI Thesaurus, NCI Thesaurus must be an important ontology
- Users can download mappings satisfying a set of criteria in RDF
  - For example, download all mappings between NCI Thesaurus and Gene Ontology based on UMLS

# Viewing all mappings for an ontology

CONCEPT	MAPS TO
<a href="#">NCI Thesaurus : Pelvic Bone --&gt;</a>	<a href="#">Mouse adult gross anatomy : pelvis bone (1)</a> Mapped By TerryHayamizu <a href="#">Mouse adult gross anatomy : hip bone (1)</a> Mapped By SongmaoZhang <a href="#">Mouse adult gross anatomy : pelvic girdle bone (1)</a> Mapped By SongmaoZhang
<a href="#">NCI Thesaurus : Adipose Tissue --&gt;</a>	<a href="#">Mouse adult gross anatomy : adipose tissue (2)</a> Mapped By TerryHayamizu , SongmaoZhang <a href="#">Mouse adult gross anatomy : fat (1)</a> Mapped By TerryHayamizu <a href="#">Mouse adult gross anatomy : fat pad (1)</a> Mapped By SongmaoZhang
<a href="#">NCI Thesaurus : Sural Artery --&gt;</a>	<a href="#">Mouse adult gross anatomy : external sural artery (2)</a> Mapped By TerryHayamizu , SongmaoZhang <a href="#">Mouse adult gross anatomy : sural artery (1)</a> Mapped By SongmaoZhang <a href="#">Mouse adult gross anatomy : superficial sural artery (1)</a> Mapped By SongmaoZhang
<a href="#">NCI Thesaurus : Spermatic Artery --&gt;</a>	<a href="#">Mouse adult gross anatomy : spermatic artery (2)</a> Mapped By TerryHayamizu , SongmaoZhang <a href="#">Mouse adult gross anatomy : testicular artery (1)</a> Mapped By SongmaoZhang <a href="#">Mouse adult gross anatomy : internal spermatic artery (1)</a> Mapped By SongmaoZhang
<a href="#">NCI Thesaurus : Myocardium --&gt;</a>	<a href="#">Mouse adult gross anatomy : heart myocardium (1)</a> Mapped By TerryHayamizu <a href="#">Mouse adult gross anatomy : myocardium (2)</a> Mapped By TerryHayamizu , SongmaoZhang

View Ontology Details

Jump To:

- ⊖ Nerve Sheath
- ⊖ Synovial Membrane
- ⊖ Muscle Tissue
  - ⊕ Smooth Muscle Tissue
    - ⊖ Muscle Layer
  - ⊖ Striated Muscle Tissue
    - ⊕ Skeletal Muscle Tissue
      - ⊖ Visceral Striated M
      - ⊖ Myocardium
- ⊕ Adipose Tissue
- ⊕ Skin Tissue
- ⊕ Blood Vessel Tissue
  - ⊕ Endothelium
- ⊕ Parathyroid Gland Tissue
- ⊕ Salivary Gland Tissue
  - ⊖ Parenchyma
- ⊕ Epithelial Tissue
- ⊕ Embryonic Tissue
- ⊕ Splenic Tissue
- ⊕ Nerve Tissue, Neuroepithelial Tissue
- ⊕ Intestinal Wall Tissue
- ⊕ Endocrine Reproductive Tissue
- ⊕ Other Anatomic Concept
- ⊕ Body Part

Details Visualization Notes **Mappings** Resources [alpha](#)

"New Point-to-Point Mapping"

MAPPING TO	SOURCE	MAPPED BY	MAPPED ON	NOTES
Myocardium (Galen)	<a href="#">Prompt</a>	ngriff	02/12/08	<a href="#">View Notes</a>
<a href="#">heart myocardium (Mouse adult gross anatomy)</a>	NCICB	TerryHayamizu	04/23/08	<a href="#">View Notes</a>
<a href="#">myocardium (Mouse adult gross anatomy)</a>	NCICB	TerryHayamizu	04/23/08	<a href="#">View Notes</a>
<a href="#">cardiac muscle tissue (Mouse adult gross anatomy)</a>	NCICB	TerryHayamizu	04/23/08	<a href="#">View Notes</a>
<a href="#">myocardium (Mouse adult gross anatomy)</a>	NLM	SongmaoZhang	04/23/08	<a href="#">View Notes</a>

# Annotator

Use ontologies to annotate *your* data

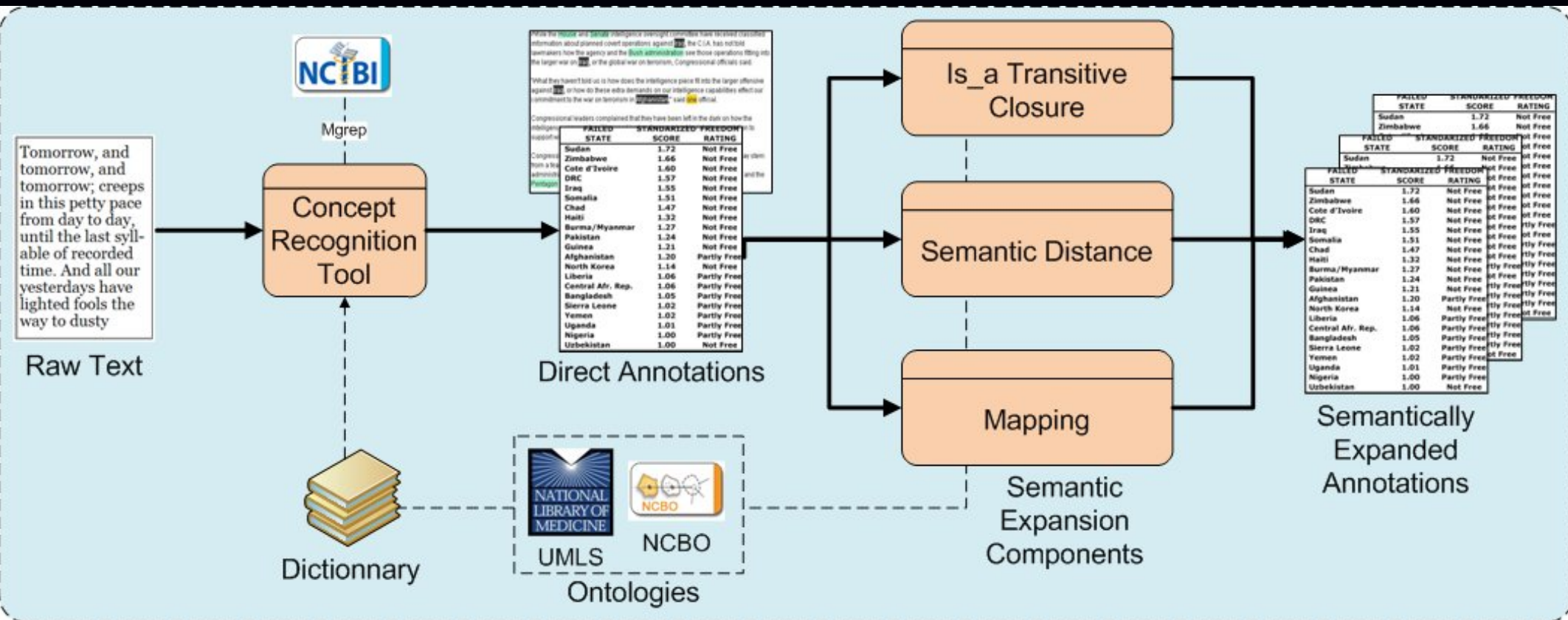
- Give your text as input
- Select your parameters
- Get your results...
  - in text, XML or OWL

The screenshot displays the 'Open Biomedical Annotator' web interface. At the top, there are input fields for 'Ontologies' (set to SNOMEDCT) and 'Semantic Types' (7,T033,T200,T026,T029,T023,T038,T017,T047,T048,T191,T019,T121,T195,T020,T050), each with a 'Choose...' button. Below these are radio buttons for 'Annotate Text' (selected) and 'Get Annotations By Resource Element Alpha', along with an 'Options Change...' button. The 'Text' input field contains the sentence: 'Melanoma is a disease of the melanocytes affecting the bowel and the eye'. An 'Annotate' button is located at the bottom right of this section.

The bottom section is divided into two panes. The left pane, titled 'Ontologies (1)', has a filter 'UMLS and BioPortal Ontologies' and a list with one checked item: 'SNOMED Clinical Terms, 2008\_07\_31'. The right pane, titled 'Annotation Tag Cloud (5)', shows 'Annotation statistics':  
Expanded annotations generated from the is\_a transitive closure (ISA\_CLOSURE): 0  
Expanded annotations generated from mappings (MAPPING): 0  
Direct annotations generated from concept recognition on the given text (MGREP): 5  
Below the statistics, the words 'Melanoma', 'Eye', and 'Disease' are displayed in large, bold, blue font, each underlined. To the right of these words are two smaller underlined links: 'Entire eye' and 'Intestines'.

At the bottom of the interface, there is a footer with the text: 'Learn more about the [NCBO Annotator web service](#) | Learn more about the [NCBO Biomedical Resources index](#)'.

# NCBO Annotator Web service workflow



HTML page

Code

Specific UI

Contact: [jonquer@stanford.edu](mailto:jonquer@stanford.edu)

The service is currently deployed with the basis URL: [http://ncbolabs-dev2.stanford.edu:8080/OBS\\_v1/](http://ncbolabs-dev2.stanford.edu:8080/OBS_v1/).  
 Documentation is available here: [http://obs.bioontology.org/docs/OBS\\_v08/](http://obs.bioontology.org/docs/OBS_v08/).

Test the Open Biomedical Annotator here:

longestOnly  use  false [default: false]  
 (i.e., do you want the concept recognizer to match the longest sentence only or not.)

wholeWordOnly  use  false [default: true]  
 (i.e., do you want the concept recognizer to match whole word only.)

scored  use  false [default: true]  
 (i.e., do you want the annotations to be scored by relevance.)

localOntologyIDs: \_\_\_\_\_ [default: all]  
 (i.e., list of ontologies you want to use separated with comma (without space) e.g., SNOMEDCT,NCIM,ISH.  
 You can find the list of localOntologyIDs available here: [obs/ontologies](#))

localSemanticTypeIDs: \_\_\_\_\_ [default: all]  
 (i.e., list of UMLS semantic types you want to use separated with comma (without space) e.g., T047,T048,T191.  
 You can find the list of localSemanticTypeIDs available here: [obs/semanticTypes](#))

levelMin: 0 [default: 0]  
 levelMax: 2 [default: 0]  
 (i.e., the level you want the tree-transitive-closure expansion component to be restricted to.)

mappingTypes: \_\_\_\_\_ [default: all]  
 (i.e., list of mapping types you want to use for the mapping expansion component separated with comma (without space) e.g., inter-cul,from-ctrl.  
 You can find the list of mapping types available here: [obs/mappingTypes](#). Note if you use the keyword null in your list, it will disable the mapping expansion component.

text (i.e., the text you want to annotate) [default: empty]

text: \_\_\_\_\_

Be careful to remove end-of-lines characters from your text.

Format (i.e., the format of the results.) [default: none]  
 asText  
 asTreeDelimited (annotations only)  
 asXML

Annotate

```

1 use LWP::UserAgent;
2 $ua = new LWP::UserAgent;
3
4 # make request
5 my $req = new HTTP::Request POST => 'http://ncbolabs-dev2.stanford.edu:8080/OBS_v1/c
6 $req->content_type('application/x-www-form-urlencoded');
7 # set parameters
8 $req->content("longestOnly=true&wholeWordOnly=true&scored=true&localOntologyIDs=SNOM
9
10 # send request and get response.
11 my $res = $ua->request($req);
12 # If you want to get a large result. It is better to write to a file directly.
13 # my $res = $ua->request($req,'file_name.txt');
14
15 # Check the outcome of the response
16 if ($res->is_success) {
17     print "Call successful\n";
18     print $res->decoded_content;
19 }
20 else {
21     print $res->status_line, "\n";
22 }
  
```

Console  
 terminated: Cmd [Program] C:\WINDOWS\system32\cmd.exe  
 Call successful  
 10.0 SNOMEDCT/C0002395 Alzheimer's disease [X]Dementia in Alzheimer's disease  
 6.0 SNOMEDCT/C0014544 Epilepsy Epilepsy NOS (disorder) /// Epileptic attack

Biomedical Annotator

Ontologies: All Ontologies Selected  
 Entity Types: T047,T048,T191,T025,T999  
 Annotate Text  Get Annotations by Resource

Options: Change...

Text: Melanoma is a malignant tumor of melanocytes which is found in the bowel and the eye.

Annotate

Annotation Tag Cloud (363)

UMLS and BioPortal Ontologies

Select All Select None

MAPPING 1083  
 ISA\_CLOSURE 283  
 MGREP 129

Melanoma Neoplasms Malignant neoplasms

monoatomic indium eve Melanocytes elemental indium hypocranium

compound.organ skin Melanoma Skin surface.structure surface.structure ocular.ligne

solid.compound.organ indium(3+) indium(1+) Neoplasm by Special Category Organ

Malignant tumor of unknown origin or ill-defined site surface.structure Neoplasm and/or hamartoma

dermal.system accretion Organ Malignant neoplasm of other and unspecified sites organ.system

larval.lateralia organ.system organ.system organism.subdivision base unit organ.system organism.subdivision

animal.component organism.subdivision organ.system melanoma skin skin eye skin melanoma An

Malignant sex A sex eye skin eye eye eye tumor A bowel sex eye eye sex melanoma

bowel eye sex eye eye eye skin skin eye eye eye eye eye eye eye melanoma

bowel eye sex eye eye eye skin eye skin eye eye malignant eye sensor.organ

solid.solid.tumor Epithelial Cells NAMEDResourcePart sensor.organ ret.stain visual.system food.organ

central.nervous.system gastrointestinal.system skin.cancer BODY LOCATION Organ of Special Sense cancer face

```

<text-melanoma<:text>
<annotations>
  <obs.common.beans.AnnotationBean>
    <score>122</score>
    <concept>
      <localConceptID>AOD.C0025202</localConceptID>
      <preferredName>melanoma</preferredName>
      <synonyms>
      <isTopLevel>false</isTopLevel>
      <localOntologyID>AOD</localOntologyID>
      <localSemanticTypeID>
        <string>T191</string>
        <string>T000</string>
      </localSemanticTypeID>
      <concept>
      <context class="obs.common.beans.MappingContextBean">
        <contextName>MAPPING</contextName>
        <isDirect>false</isDirect>
        <mappedConceptID>COSTAR.C0025202</mappedConceptID>
        <mappingType>inter-cul</mappingType>
      </context>
      <obs.common.beans.AnnotationBean>
        <score>122</score>
        <concept>
          <localConceptID>OMIM.C0024202</localConceptID>
          <preferredName>Melanoma</preferredName>
          <synonyms>
          <string>Malignant melanoma</string>
          </synonyms>
          <isTopLevel>false</isTopLevel>
          <localOntologyID>OMIM</localOntologyID>
          <localSemanticTypeID>
            <string>T191</string>
            <string>T000</string>
          </localSemanticTypeID>
          <concept>
  
```


# An example

- “Melanoma is a malignant tumor of melanocytes which are found predominantly in skin but also in the bowel and the eye”.
  - NCI/C0025201, Melanocyte in NCI Thesaurus
  - 39228/DOID:1909, Melanoma in Human Disease
- Is\_a closure expansion
  - 39228/DOID:191, Melanocytic neoplasm, direct parent of Melanoma in Human Disease
  - 39228/DOID:0000818, cell proliferation disease, grand parent of Melanoma in Human Disease
- Mapping expansion
  - FMA/C0025201, Melanocyte in Foundational Model of Anatomy, concept mapped to NCI/C0025201 in UMLS.



# NCBO Resource Index

THE NATIONAL CENTER FOR BIOMEDICAL ONTOLOGY








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**Malignant\_Skin\_Neoplasm** ([Link To Concept](#))


Details Visualization Marginal Notes Mappings **Resources**

 ClinicalTrials.gov	ClinicalTrials.gov provides regularly updated information about federally and privately supported clinical research in human volunteers. ClinicalTrials.gov gives you information about a trial's purpose, who may participate, locations, and phone numbers for more details. The information provided on ClinicalTrials.gov should be used in conjunction with advice from health care professionals. Before searching, you may want to learn more about clinical trials.	Annotations:796
 Gene Expression Omnibus DataSets	A gene expression/molecular abundance repository supporting MIAME compliant data submissions, and a curated, online resource for gene expression data browsing, query and retrieval.	Annotations:9
 ArrayExpress	ArrayExpress is a public repository for microarray data, which is aimed at storing MIAME-compliant data in accordance with MGED recommendations. The ArrayExpress Data Warehouse stores gene-indexed expression profiles from a curated subset of experiments in the repository.	Annotations:16
 ARRS GoldMiner	ARRS GoldMiner provides instant access to images published in selected peer-reviewed radiology journals. This new, web-based system allows viewers to search for images by findings, anatomy, imaging technique, and patient age and sex.	Annotations:131
 NextBio	NextBio's data and literature search engine makes massive amounts of disparate biological, clinical and chemical data from public and proprietary sources searchable, regardless of data type and origin, empowering researchers to quickly understand their own experimental results within the context of other research.	Annotations:0

Left sidebar (partial): e\_Disorder, Cancer-Related\_Condition, Differentiation-Induced\_Abnormalities, Disorder\_by\_Site, Reproductive\_System\_Disorder, Eye\_Disorder, Nervous\_System\_Disorder, Breast\_Disorder, Endocrine\_Disorder, Skin\_Disorder, Cutaneous\_Precancerous\_Condition, Non-Neoplastic\_Skin\_Disorder, Hair\_Disorder, Skin\_Vascular\_Disorder, Skin\_Neoplasm, Dermal\_Neoplasm, Epithelial\_Skin\_Neoplasm, Cutaneous\_Hematopoietic\_Disorder, Malignant\_Skin\_Neoplasm, Scrotal\_Neoplasm, Melanocytic\_Skin\_Neoplasm

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 + [Malignant\\_Skin\\_Neoplasm](#)  
 + [Scrotal\\_Neoplasm](#)  
 + [Melanocytic\\_Skin\\_Neoplasm](#)

## Malignant\_Skin\_Neoplasm (Link To Concept)

Details Visualization Marginal Notes Mappings **Resources**

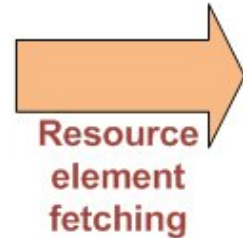
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<a href="#">Cutaneous malignant melanoma</a> ID: 1375      Annotation Context: closure		
<a href="#">Transcription factor FoxM1 inactivation effect on breast cancer cell</a> ID: 1477      Annotation Context: closure		
<a href="#">Melanoma progression</a> ID: 1989      Annotation Context: closure		
<a href="#">Melanoma, cutaneous malignant, classification</a> ID: 2      Annotation Context: closure		
<a href="#">Ductal carcinoma in situ to invasive ductal carcinoma progression (HG-U133A)</a> ID: 2045      Annotation Context: closure		
<a href="#">Ductal carcinoma in situ to invasive ductal carcinoma progression (HG-U133 2.0)</a> ID: 2046      Annotation Context: closure		
<a href="#">Non-melanoma skin cancer</a> ID: 2046		

## Gene Expression Omnibus



e.g., element GDS1989

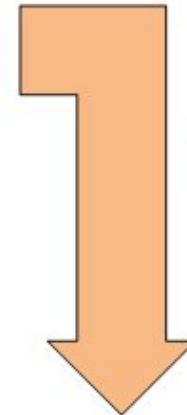
Eutils Web Service API



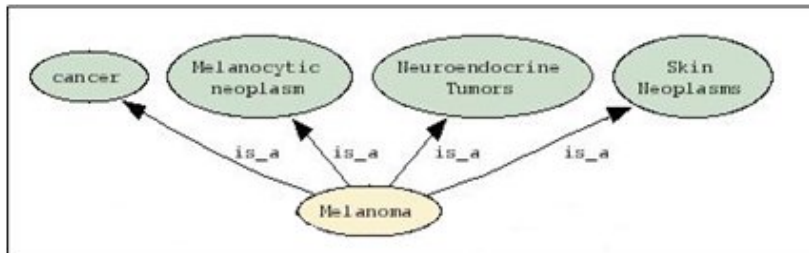
**Resource  
element  
fetching**

```
<title>
  Melanoma progression
</title>

<description>
  Analysis of tissue specimens representing benign
  nevus, atypical nevus, melanoma in situ, vertical growth
  phase (VGP) melanoma, and metastatic growth phase
  (MGP) melanoma. Results identify expression signatures
  that distinguish benign and atypical nevi and melanomas in
  situ from VGPs and MGPs.
</description>
```



**Annotation**



**172 closure annotations and 171 useful.**

Examples:

Cancer, concept (DOID:162) in ontology *Human disease*

Skin Neoplasms, concept (DOID:3165) in ontology *Human disease*

**Transitive  
closure**



```
<title>
  Melanoma progression
</title>

<description>
  Analysis of tissue specimens representing benign
  nevus, atypical nevus, melanoma in situ, vertical growth
  phase (VGP) melanoma, and metastatic growth phase
  (MGP) melanoma. Results identify expression signatures
  that distinguish benign and atypical nevi and melanomas in
  situ from VGPs and MGPs.
</description>
```

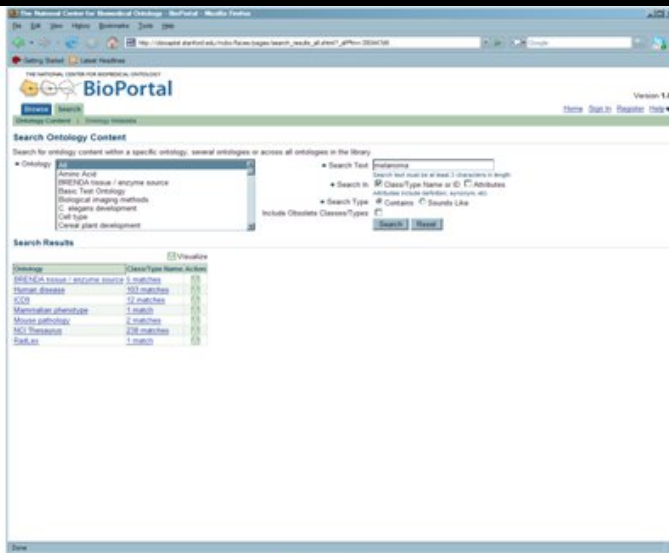
**23 direct annotations  
(4 title, 19 description)**

Example:

*Melanoma*, concept (DOID:1909) in  
ontology *Human disease*.

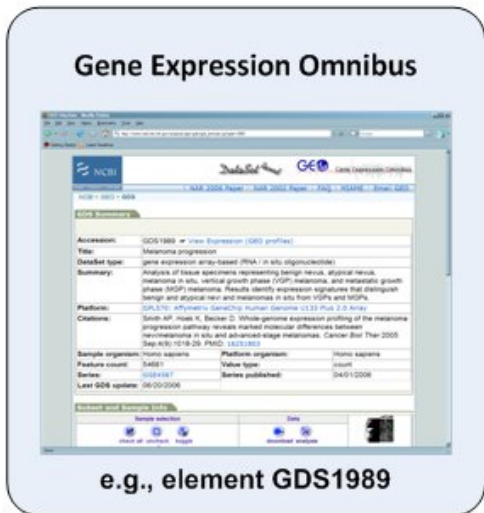


User search



Bioportal search for "melanoma"

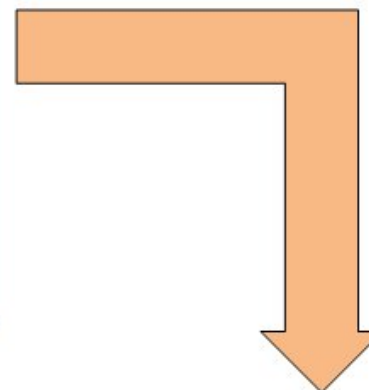
362 matches in all Bioportal ontologies. Example: Melanoma, concept (DOID:1909) in ontology Human disease.



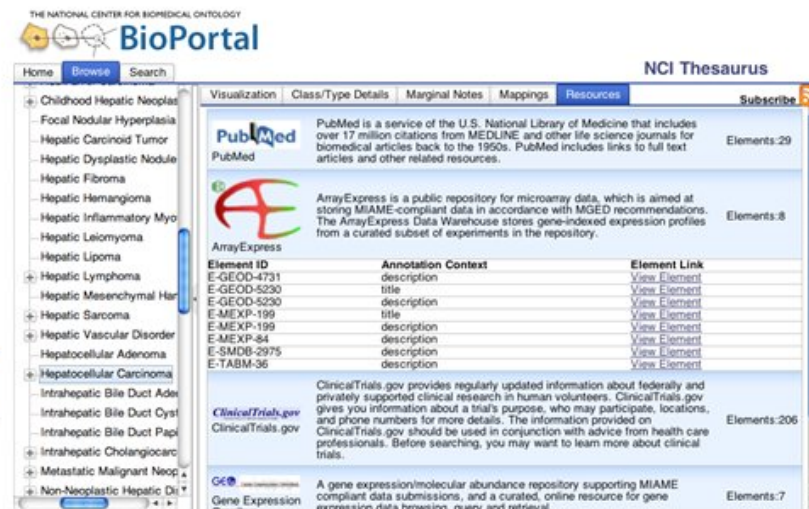
e.g., element GDS1989



Link to the original resource element



OBS index request



Onrez index results display

- 227 PubMed elements,
- 3 ArrayExpress elements,
- 969 ClinicalTrials.gov elements,
- 10 ARRS GoldMiner elements,
- 3 Gene Expression Omnibus elements.

Example: Melanoma progression element (GDS1989)

# Open Biomedical Resources

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- Cancer-Related Condition
- Differentiation-Induced Abnormalities
- Order by Site
- Reproductive System Disorder
- Eye Disorder
- Nervous System Disorder
- Breast Disorder
- Endocrine Disorder
- Skin Disorder
  - + Cutaneous Precancerous Condition
  - + Non-Neoplastic Skin Disorder
  - + Hair Disorder
  - + Skin Vascular Disorder
  - Skin Neoplasm
    - + Dermal Neoplasm
    - + Epithelial Skin Neoplasm
    - + Cutaneous Hematopoietic Neoplasm
    - + Malignant Skin Neoplasm
    - + Scrotal Neoplasm
    - + Melanocytic Skin Neoplasm

## Malignant Skin Neoplasm [\(Link To Concept\)](#)

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# Recap: BioPortal Features

- ~~Ontology repository~~
  - metadata about ontologies
  - ontology browsing and visualization
- Ontology search
- Peer review and comments
  - threaded discussion on classes
  - ontology reviews on different dimensions
- Concept-to-concept mappings
- NCBO resource index
  - biomedical resources annotated with ontology terms
  - semantic annotation
- Annotator

# BioPortal technology is domain-independent

- Other installations of BioPortal:
  - Marine Metadata Initiative repository
  - Open Ontology Repository


Marine Metadata Interoperability  
**Ontology Registry and Repository**  
alpha

Other MMI Ontology Products: [voc2rdf](#) :: [VINE](#)

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Ontology Name	Format	Version	Author	Uploaded On	
<a href="#">AGU Index Terms</a>	OWL-DL	20090604T210443	AGU	06/04/2009	<a href="#">Explore</a>
<a href="#">ARGO Instruments</a>	OWL-DL	20090605T194103	ARGO	06/05/2009	<a href="#">Explore</a>
<a href="#">ARGO Parameters</a>	OWL-DL	20090605T194513	ARGO	06/05/2009	<a href="#">Explore</a>

## OOR Open Ontology Repository

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Filter by Group  [Link To This Filter](#)

Filter Ontologies

# BioPortal Technology in Your Project in Your Project

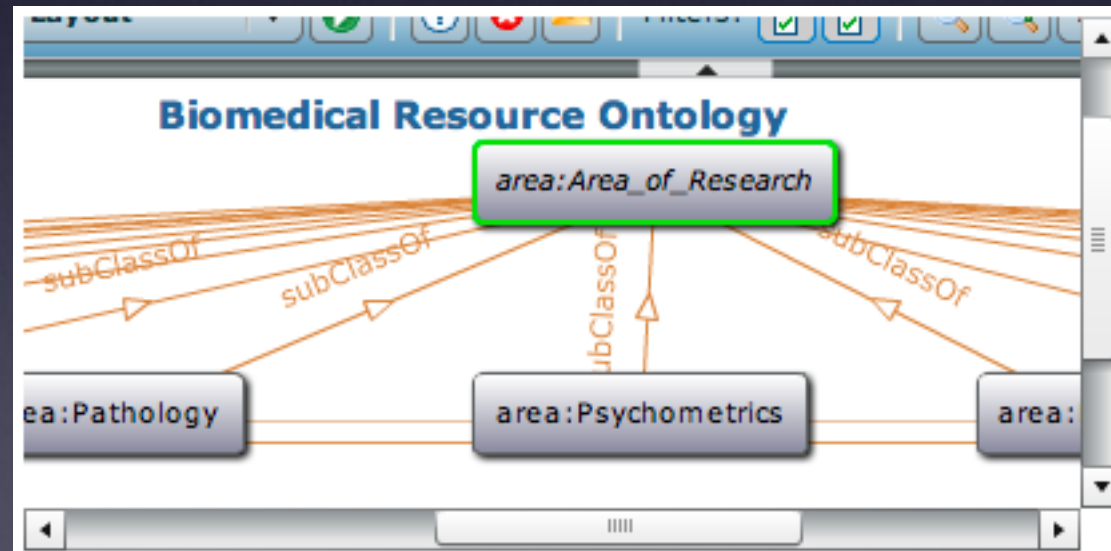
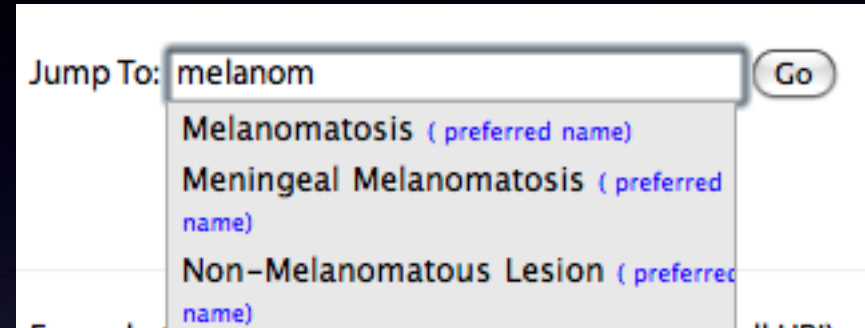
---

- **Ontology Web Services** ( REST services )
  - ontology metadata
  - information about concepts
  - search
  - hierarchical information
- **Annotator** service
  - identify biomedical concepts in your text
- **Ontology widgets**



# Ontology Widgets

- User interface components with “BioPortal inside”:
  - **term-selection** widget for a specific ontology
  - **form fields** with auto-complete from a specific BioPortal ontology
  - **RSS feed** for an ontology
  - **Visualization** widget



# The Challenges Ahead

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- More attention to workflow issues in ontology development and use
- Critical assessment of the role of technology in assisting ontology development
- Measuring the effects of our work, when the most important outcomes are social and interactive

# Thank you

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- Mark Musen
- Nigam Shah
- Trish Whetzel
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- Nick Griffith
- Cherie Youn
- Clement Jonquet
- Benjamin Dai
- Peggy Storey
- Chris Callendar
- Sean Falconer
- Chris Chute
- Pradip Kanjamala
- Jyoti Pathak
- Jim Buntrock
- *and many others*