

Using Ontologies:

Tutorial on Finding and Requesting Ontology Terms

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EHLC

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This content: <https://oboacademy.github.io/oobook/lesson/getting-hands-on/>

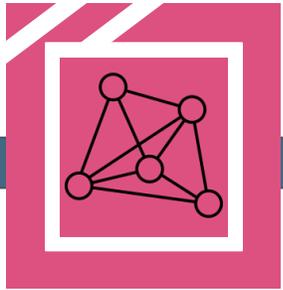
Hello!

I am Nicole Vasilevsky

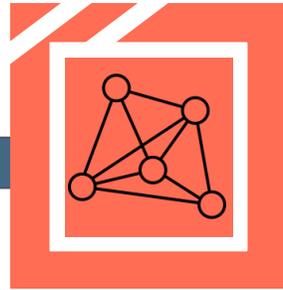
- I am the **Associate Director for Data Science** at the Critical Path Institute
- Formerly part of the **Translational and Integrative Sciences Lab (TISLab)** at Univ. of Colorado and Oregon Health & Science University (OHSU)
- Member of **OBO Foundry** Community, **International Society for Biocuration**
- I'm not an NIH employee – all views expressed are my own



Learning Objectives



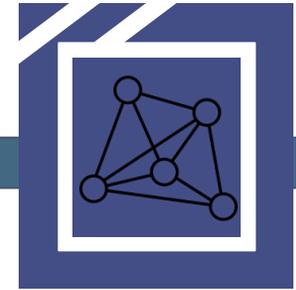
Identify the best ontology or ontologies for annotation of biomedical data



Find ontology terms and chose the best ontology term



Make new term requests or request changes



Understand open source ontology community development

A single, vibrant red poppy flower stands out against a background of tall, golden-brown grass. The flower is in sharp focus, while the grass is blurred, creating a sense of depth. The overall scene is peaceful and natural.

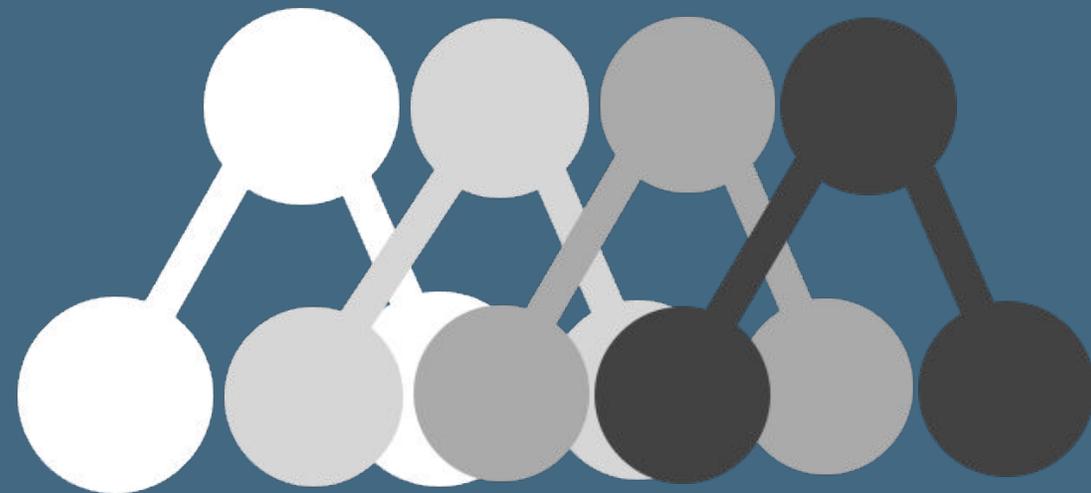
Where to find and choose the right ontology terms

Image credit <https://pixabay.com/photos/flower-poppy-field-red-flower-100263/>

<https://oboacademy.github.io/oobook/lesson/ontology-term-use/>

OBO Foundry

obofoundry.org

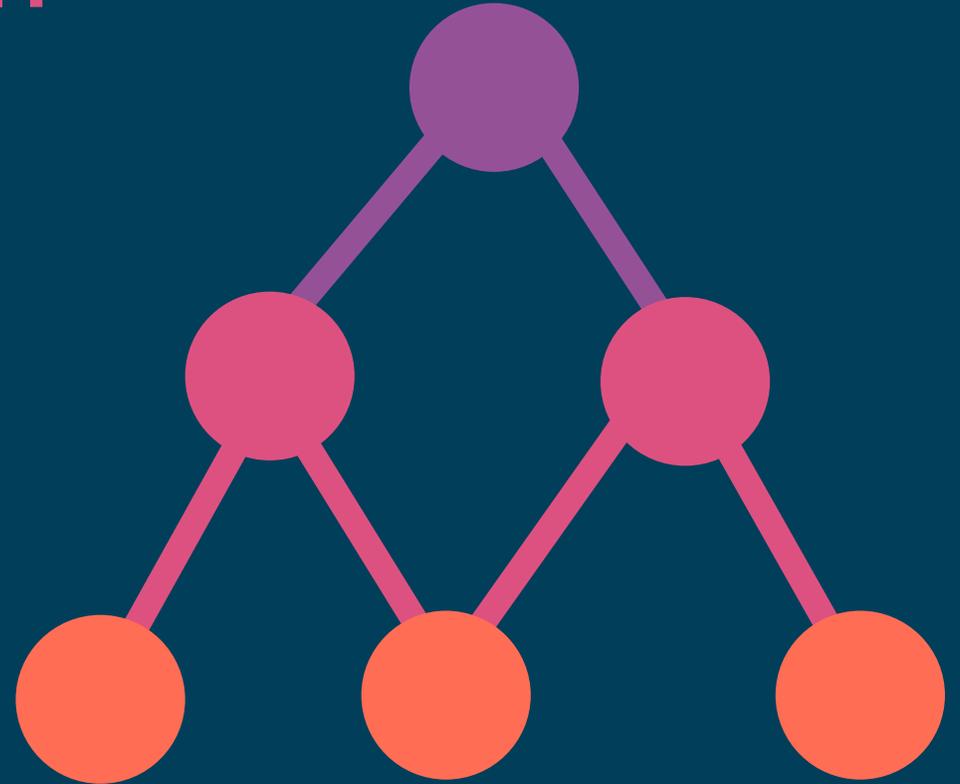


Community of ontology developers that are committed to shared set of principles for ontology development

Meetings on Biweekly Tuesdays 9am PT

Why are Ontologies Useful?

- Provide a logical classification of information in a particular domain or subject area
- Can be used for
 - data annotations
 - structuring disparate data types
 - classifying information
 - inferencing
 - reasoning across data
 - computational analyses



Finding good ontologies

- OBO Foundry
- The Ontology Lookup Service (OLS)
- BioPortal
- Ontobee



- The Ontology Lookup Service (OLS)
- BioPortal
- Ontobee

Finding Terms using Ontology Browsers

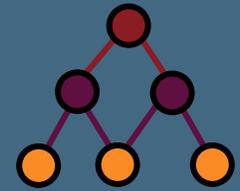


Assessing ontologies for use

- License
- Quality
 - Is there an ontology tracker to report issues?
 - Is it currently active?
 - Community involvement
 - Scientifically sound

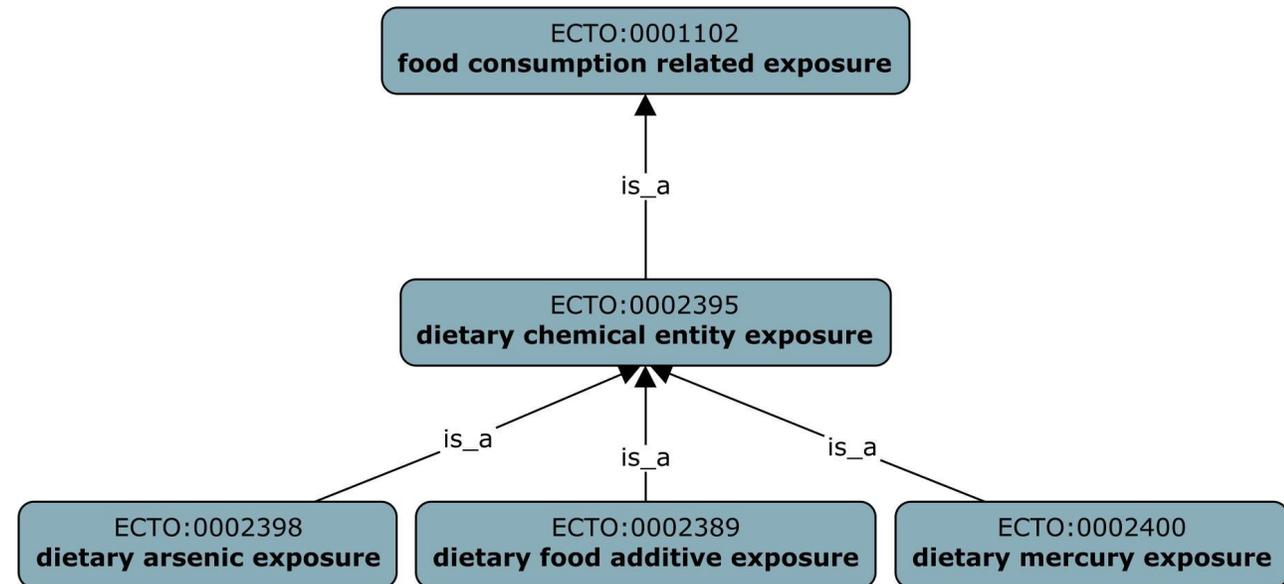


Using Ontology Terms for Annotations



Considerations:

- Ontology terms are classified in a hierarchy
 - The `is_a` relation is like inheritance
 - Children terms inherit the properties and relationships of the parent term
- Ontology terms are textually and logically defined



Dietary chemical entity exposure

A exposure event arising as a result of ingestion in food of chemical entity.

Dietary arsenic exposure

A exposure event arising as a result of ingestion in food of arsenic.

Which is the right ontology to use?

- Start by selecting the appropriate ontology, then search and restrict your search to that ontology
- Recommend using ontologies that are open and interoperable. Focusing on OBO Foundry ontologies is a good place to start
- Make informed decision about which ontology to use
- Maybe the ontology you want to use does not have the term you want, so make a term request to that ontology

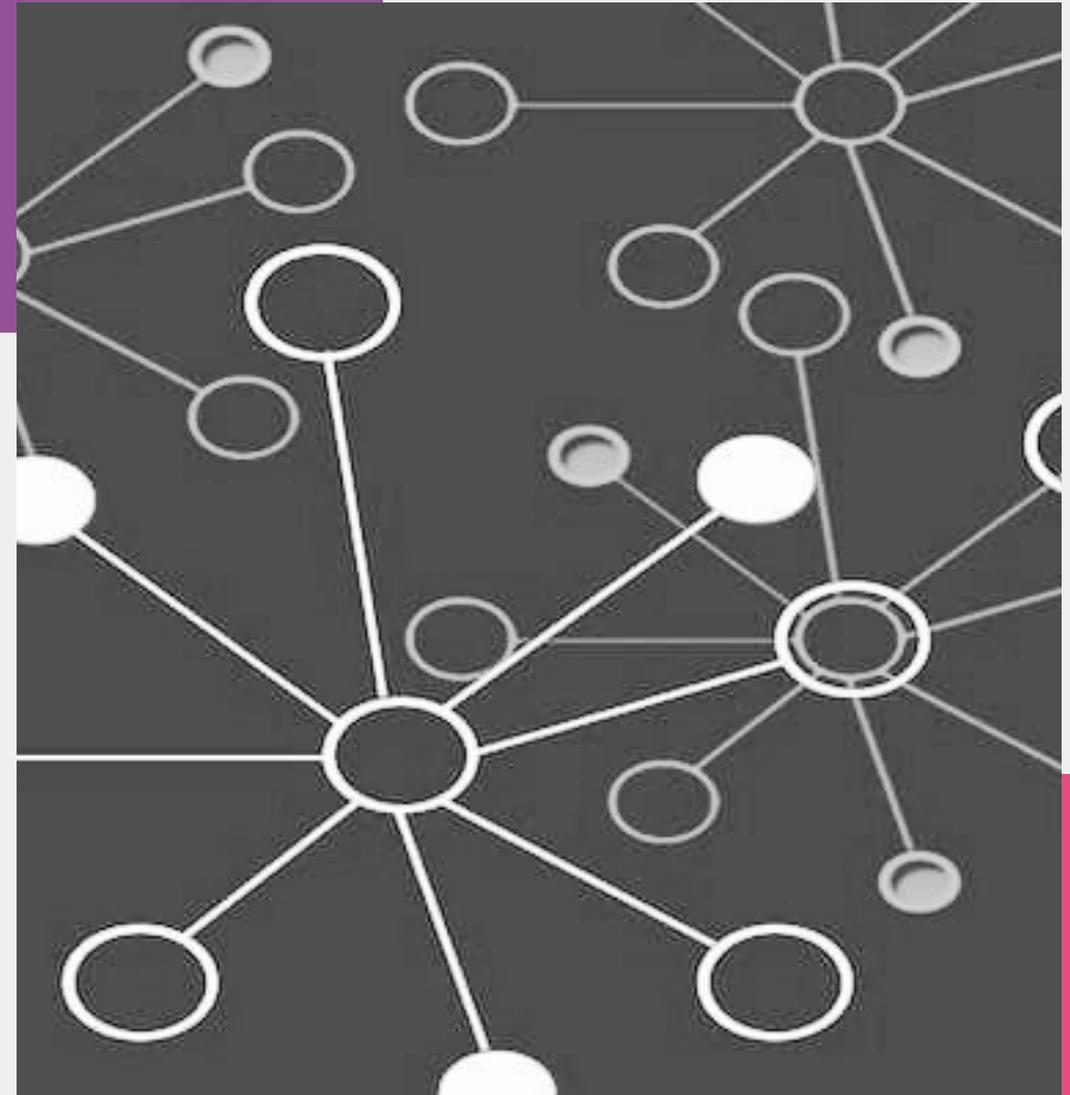


Image credit:
<https://solutionsreview.com/business-process-management/why-is-process-improvement-so-important/>

A young green plant sprout with three leaves is growing from a piece of wood. The wood is resting on a bed of dark soil. The background is a blurred green field.

How to make new term requests

Image credit <https://pixabay.com/photos/grow-leaves-trunk-plant-nature-781769/>

<https://oboacademy.github.io/oobook/howto/term-request/>



- Ontologies are stored on GitHub
- Community can contribute to open ontologies
 - Create issues on our tracker
 - Report bugs
 - Request new terms
 - Request changes to classifications, definitions, etc.
 - Comment on tickets, have discussions
 - (*More advanced*): Directly edit ontologies using pull requests

<https://github.com>

Ontology issue trackers



A tracker is a place to put a formal ontology request

1



Trackers have long been used in the software community for keeping track of bugs, feature requests, etc

2



Advantages:

- Open
- Documentation
- Community can comment

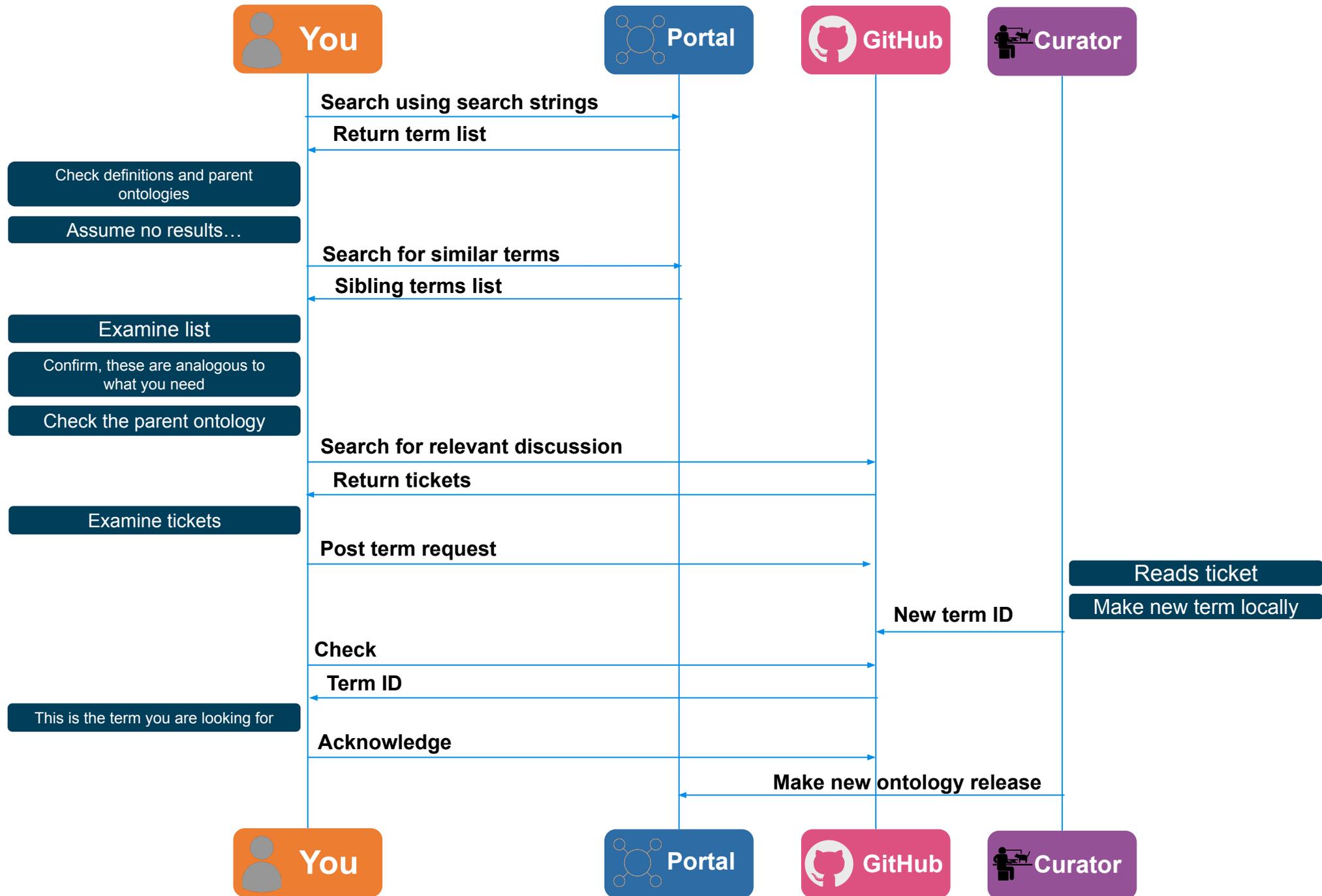
3



Tracker IDs can be referenced in ontology metadata, such as in an editor note or definition annotation

4

Term search and request workflow



How to request new terms & changes

Step

1

Go to the GitHub tracker and select **Issues**



monarch-initiative / mondo Public template

Edit Pins Unwatch 10 Fork 37 Starred 154

<> Code Issues 212 Pull requests 14 Discussions Actions Projects 13 Wiki Security Insights Settings

master 62 branches 84 tags Go to file Add file <> Code Use this template

sabrinatoro added "baclofen withdrawal syndrome" (#5764) d92343d yesterday 10,414 commits

.github	Removed poetry dependency and triggers QC checks (#5721)	last month
components	release 230104 (#5770)	last week
docs	Add video exclude superclass (#5752)	last month
imports	release 230104 (#5770)	last week
reports	release 230104 (#5770)	last week
scratch	scratch	4 years ago
src	added "baclofen withdrawal syndrome" (#5764)	yesterday
.gitignore	Filling in object_label for *.sssom.tsv file (#5491)	3 weeks ago
.readthedocs.yml	Initial MkDocs files for Read the Docs.	3 years ago

About Mondo Disease Ontology

obofoundry.org/ontology/mondo

owl ontology mondo disease monarchinitiative disease-classification obofoundry

Readme CC-BY-4.0 license 154 stars 10 watching 37 forks

Releases 79

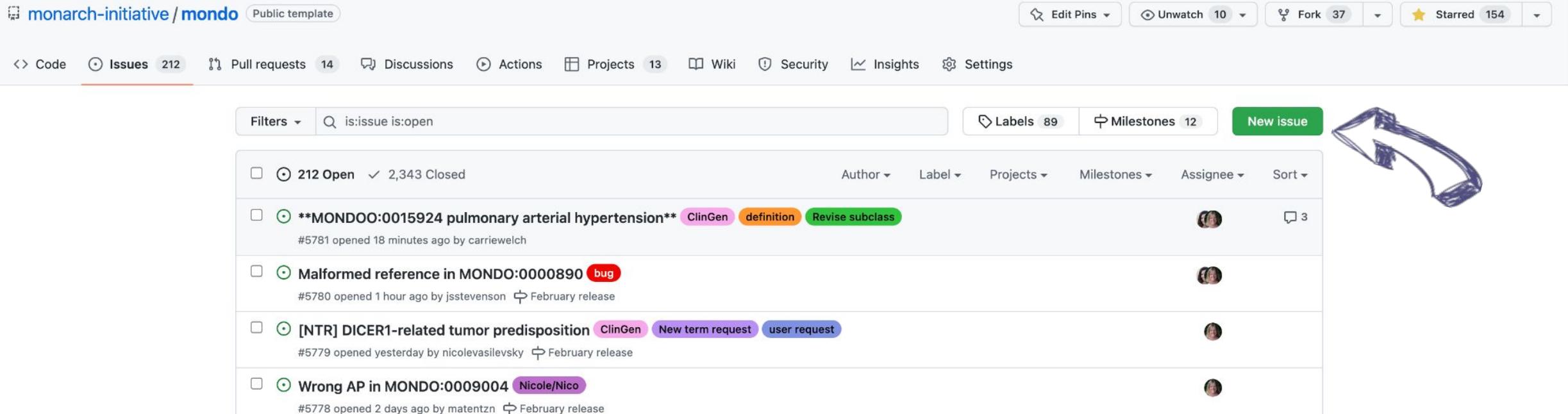
This example: <https://github.com/monarch-initiative/mondo>

How to request new terms & changes

Step

2

Click **New Issue**



monarch-initiative / mondo Public template

Edit Pins Unwatch 10 Fork 37 Starred 154

Code Issues 212 Pull requests 14 Discussions Actions Projects 13 Wiki Security Insights Settings

Filters is:issue is:open Labels 89 Milestones 12 **New issue**

<input type="checkbox"/>	212 Open ✓ 2,343 Closed	Author	Label	Projects	Milestones	Assignee	Sort
<input type="checkbox"/>	**MONDO:0015924 pulmonary arterial hypertension** ClinGen definition Revise subclass #5781 opened 18 minutes ago by carriewelch						3
<input type="checkbox"/>	Malformed reference in MONDO:0000890 bug #5780 opened 1 hour ago by jsstevenson February release						
<input type="checkbox"/>	[NTR] DICER1-related tumor predisposition ClinGen New term request user request #5779 opened yesterday by nicolevasilevsky February release						
<input type="checkbox"/>	Wrong AP in MONDO:0009004 Nicole/Nico #5778 opened 2 days ago by matentzn February release						

This example: <https://github.com/monarch-initiative/mondo>

How to request new terms & changes

Step 3 Pick appropriate template (in some repositories, not all)



Add term - gene related syndrome

New term suggestion for a term that is defined by a gene, such as NAA10-related syndrome

Get started

Add synonym

New synonym suggestion for an existing Mondo term.

Get started

Add term

New term suggestion for Mondo

Get started

Custom issue template

Describe this issue template's purpose here.

Get started

fix xref

Report issue about incorrect xref on term.

Get started

.. .

Get started

This example: <https://github.com/monarch-initiative/mondo>

How to request new terms & changes

Step 4

Fill in the information that is requested on the template below each header (if applicable).

Otherwise, add as much information as possible.

Issue: Add term

New term suggestion for Mondo. If this doesn't look right, [choose a different type](#).



[NTR]

Write

Preview

H B I

****Preferred term label:****

****Synonyms****

****Definition (free text, please give PubMed ID, if applicable, in format PMID:#####)****

****Parent term (use [OLS](https://www.ebi.ac.uk/ols/ontologies/mondo), or your favorite ontology browser)****

****Children terms (if applicable)**** Should any existing terms that should be moved underneath this new proposed term?

****Your nano-attribution (ORCID)****

If you don't have an ORCID, you can sign up for one [\[here\]\(https://orcid.org/\)](https://orcid.org/)

Attach files by dragging & dropping, selecting or pasting them.

Styling with Markdown is supported

Submit new issue

Remember, contributions to this repository should follow its [contributing guidelines](#).

This example: <https://github.com/monarch-initiative/mondo>

How to request new terms & changes

Step 5

Click **Submit new issue**

Best practices for being a good open scientist:

- See your request through to the end
- Feel free to check in with the ontology curator regarding the status of your request

Issue: Add term

New term suggestion for Mondo. If this doesn't look right, [choose a different type](#).



[NTR]

Write

Preview

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****Preferred term label:****

****Synonyms****

****Definition (free text, please give PubMed ID, if applicable, in format PMID:#####)****

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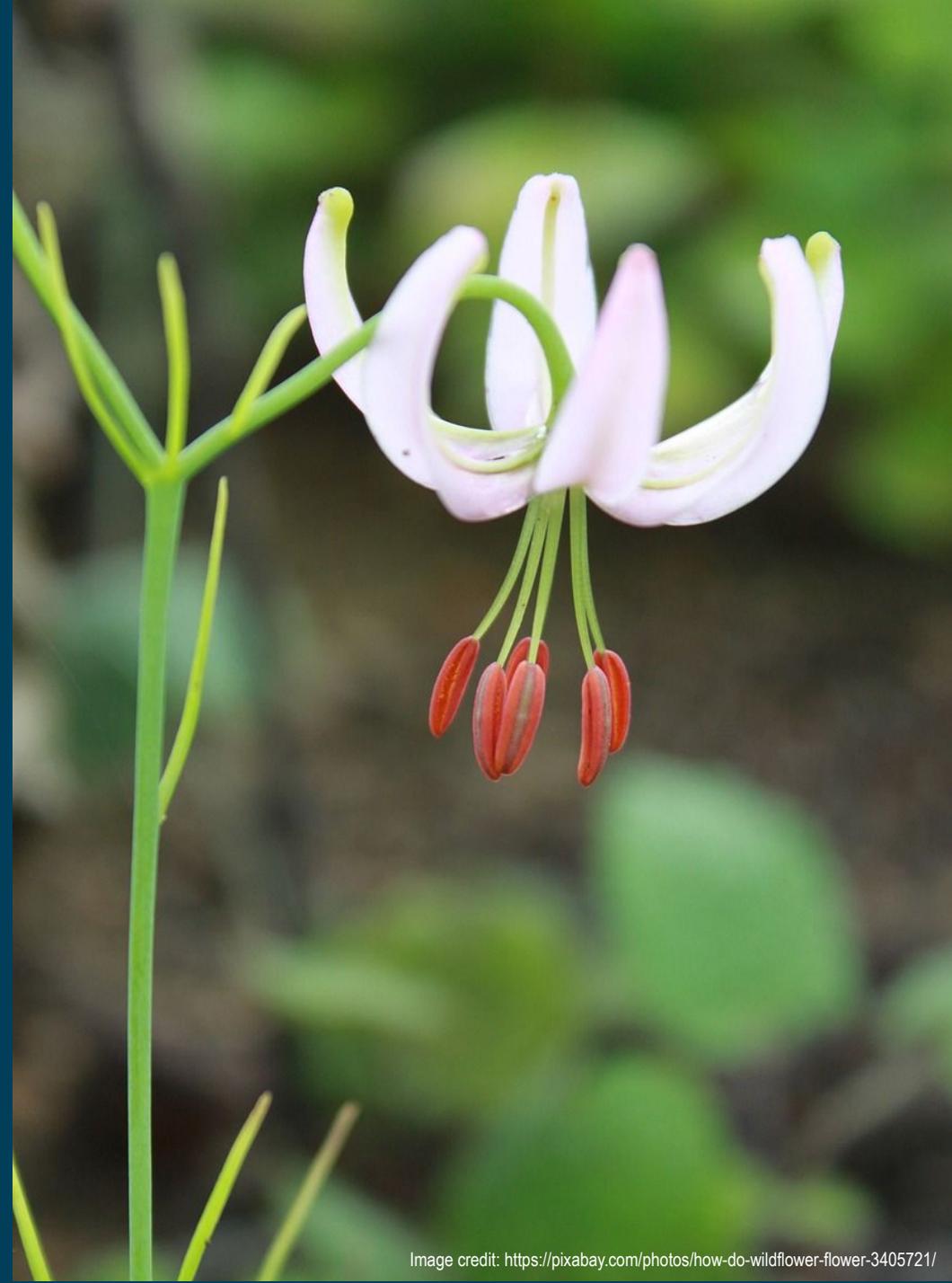


This example: <https://github.com/monarch-initiative/mondo>

Unfamiliar with GitHub?

We have a 'how to' guide for that

<https://oboacademy.github.io/oobook/lesson/contributing-to-obo-ontologies/#use-github>



**Semantic
Engineering
Open
Science
Best
Practices**



<https://oboacademy.github.io/oobook/howto/open-science-engineer/>

Principle of Collaboration

- Open ontologies need community contributions
- The OBO Academy has a lot of self-paced learning material
- Join Slack channels and email lists
- Reduce work for others as much as possible by communicating clearly
- Be positive and generous with gratitude and attribution
- Promote truly open communication



Image credit: <https://pixabay.com/photos/people-network-social-media-3108155/>

Principle of Upstream Fixing

- Report an bugs or errors to the source ontology
- Make term requests for any missing terms



Image credit: <https://pixabay.com/photos/insect-ladybug-entomology-7278221/1>

Principle of No Ownership

- Ensure that you see your issues through to the end
- Feel free to nudge ontology curators

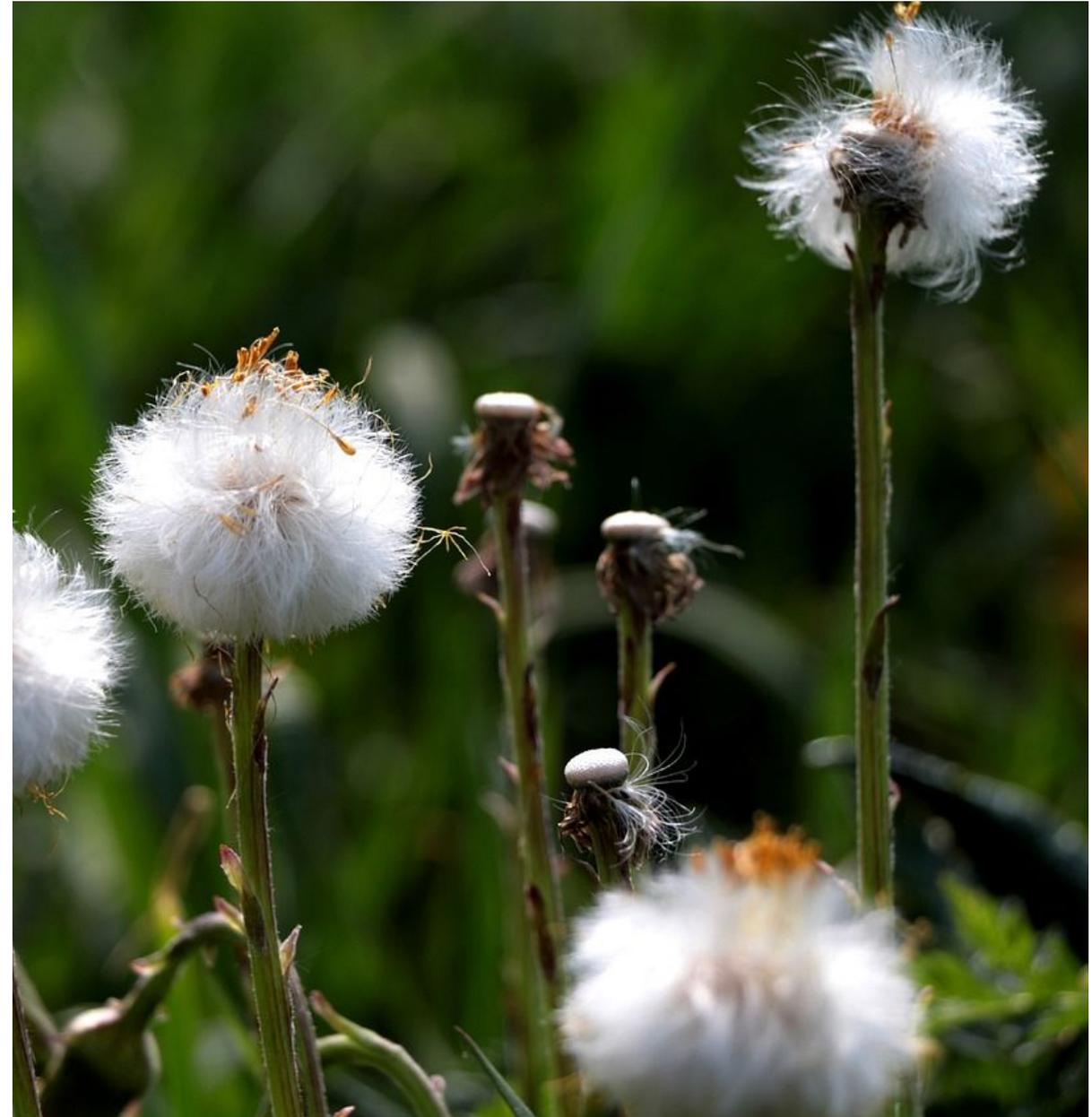


Image credit: <https://pixabay.com/photos/coltsfoot-fluff-finished-blooming-5122700/>



Image credit: <https://pixabay.com/photos/bird-wildlife-animal-nature-7299650/>

Let's get hands on

URL:

<https://oboacademy.github.io/oobook/lesson/getting-hands-on/#exercise>

Short URL:

<https://bit.ly/3GUOvFR>

Summary



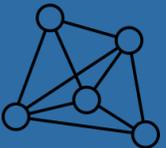
Community contributions are welcome

Our ontologies rely on your expertise and contributions



Many ontologies exist

Make informed decisions when selecting which ontology to use



Ontology terms are clearly defined

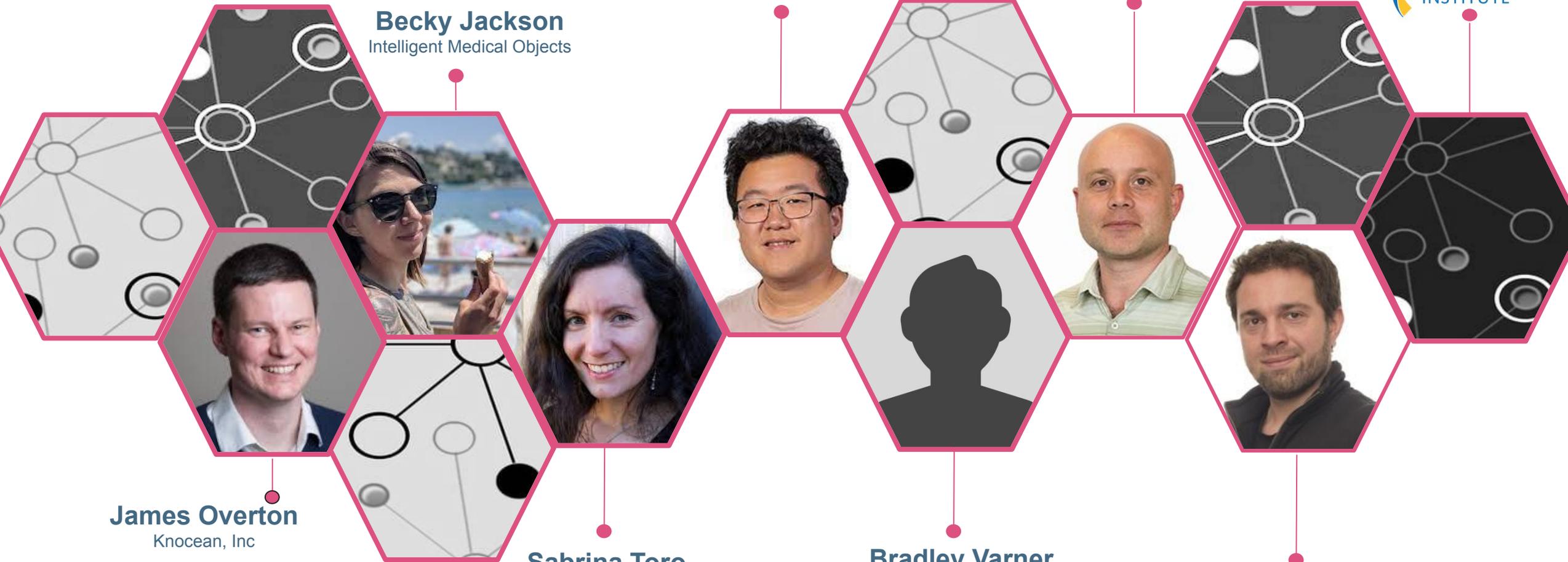
Take this into consideration when selecting terms



Ask for new terms

When the term you need doesn't exist

Acknowledgements



Shawn Tan
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Knocean, Inc

Sabrina Toro
University of Colorado

Bradley Varner
European Bioinformatics Institute

Nico Matentzoglou
Semanticy

Thanks!

You can find me at:

✉ nvasilevsky@c-path.org

<https://oboacademy.github.io/obook/>

