Introduction to Ontologies

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EHS Workshop
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monarch INITIATIVE

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Anschutz Medical Campus

Hello!

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Resources

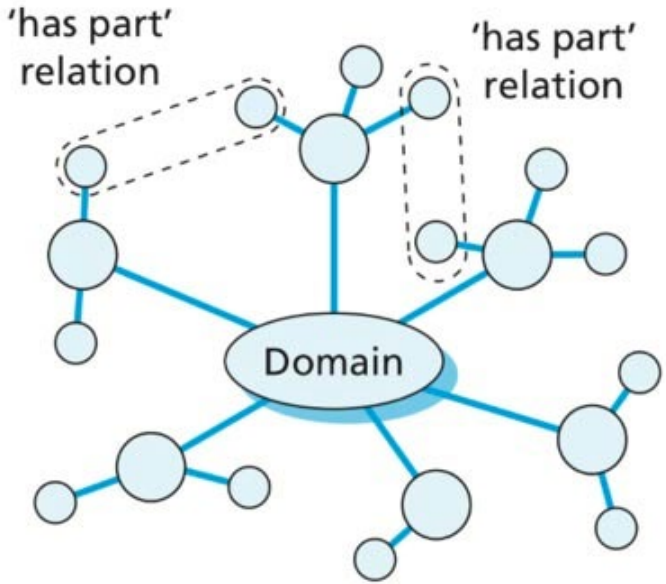
Slides:
bit.ly/ehsworkshop

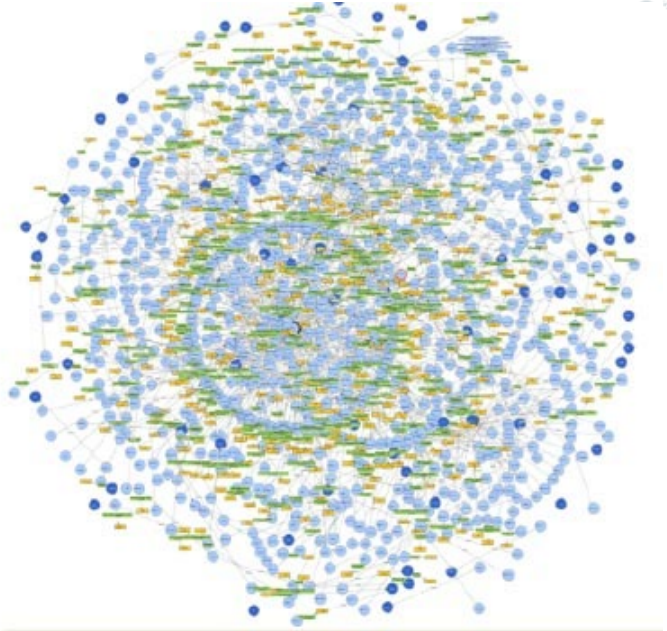
Ontology Resources:
http://obofoundry.org/resources

1.

Introduction to Ontologies

Ontologies are **systematic representations of knowledge** that can be used to **integrate and analyze** large amounts of heterogeneous data





DOI: 10.1056/NEJMra1615014
Image credit: https://kyndi.com/blog/creating-knowledge-and-maximizing-the-value-of-data-with-ontologies/

Terminology vs Ontology

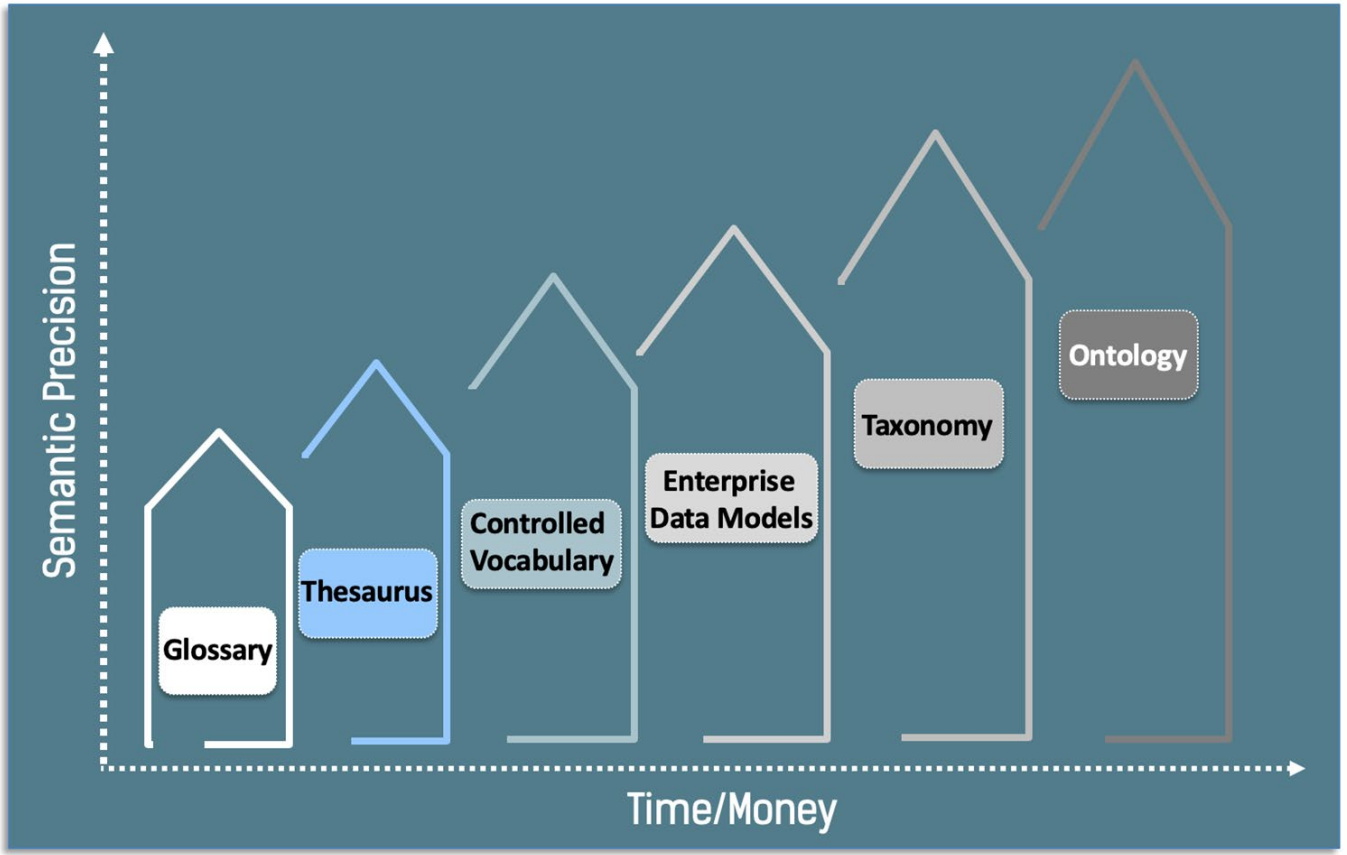
Terminology

A collection of terms, may include synonyms, definitions.

Ontology

A classification of knowledge in which terms are textually and logically defined. Provides *semantics* or *meaning* to terms.

Complexity of Vocabulary Types



Ontology use case

Query

Where can I buy light beer?

Website says sells
light beer

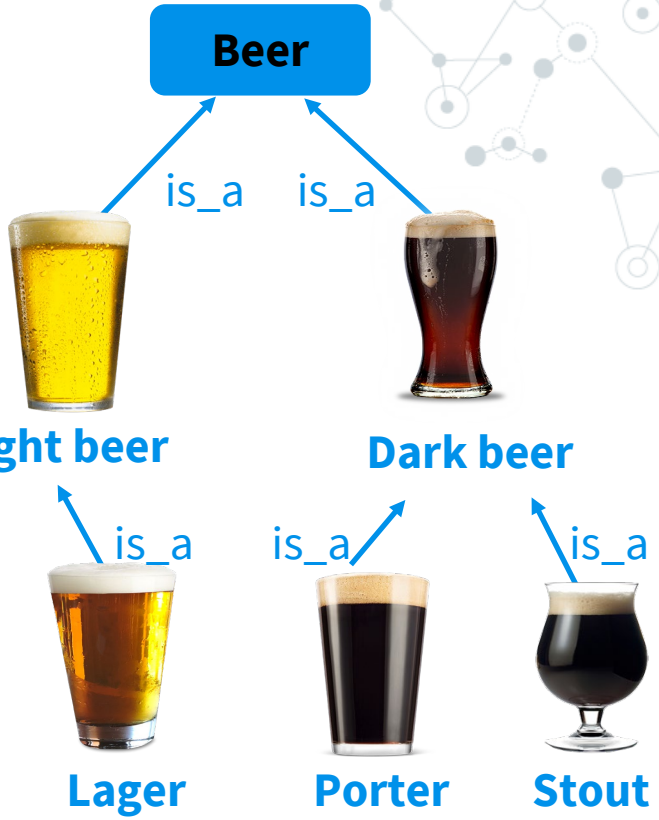
Website says sells
pilsner, lager, pale ale, etc.

Elements of an Ontology

on·tol·o·gy
/änˈtäləjē/

A knowledge classification of a domain, where the relationships between concepts are formally defined and logically related, which allows for computational reasoning

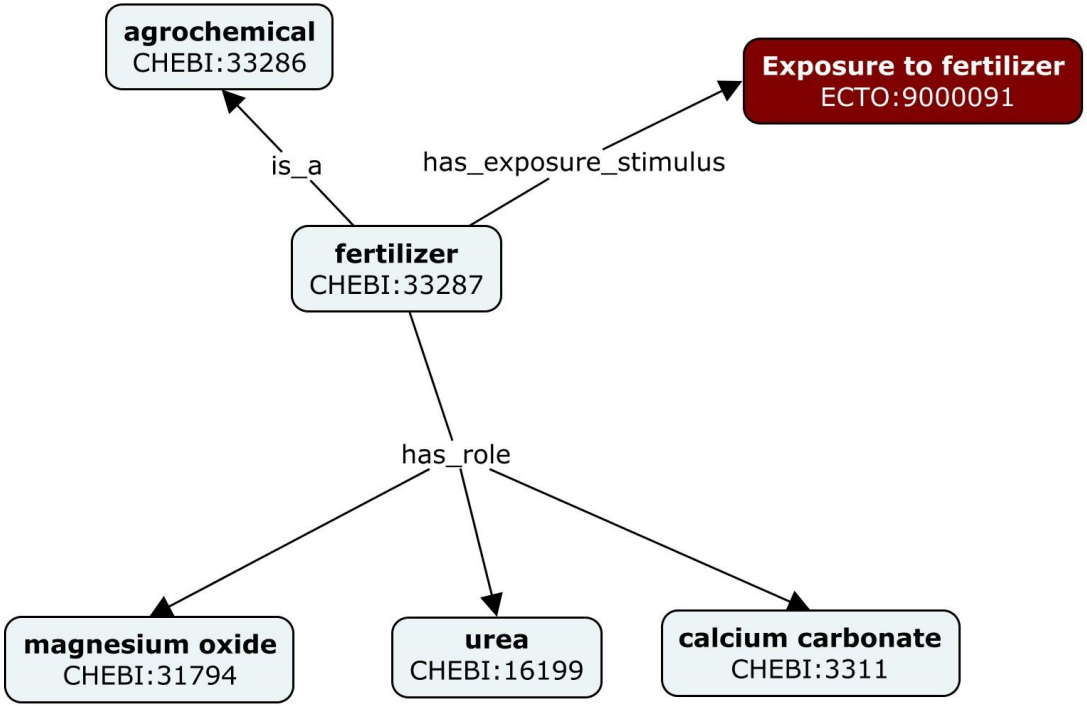
Classes (terms) - concepts in an ontology
Relationships (properties) - how the classes relate to each other
Instances - individuals



Ontology use case

Query

I want to find symptoms when someone was exposed to fertilizer.



Ontology use case

Query

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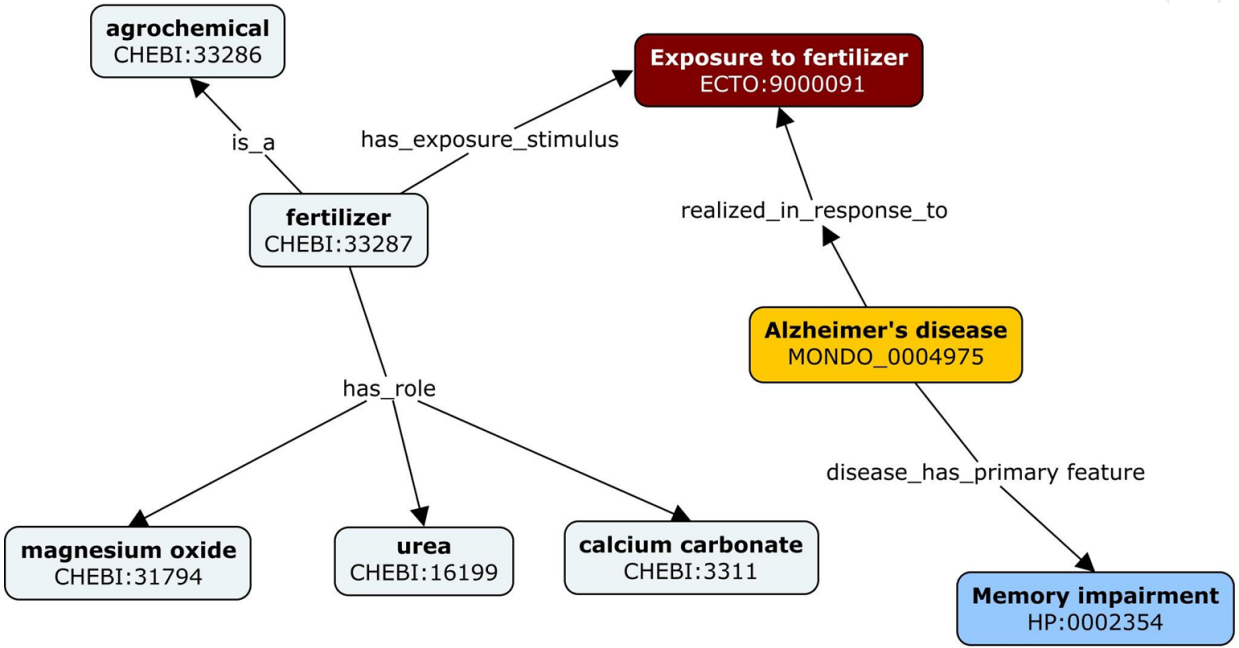
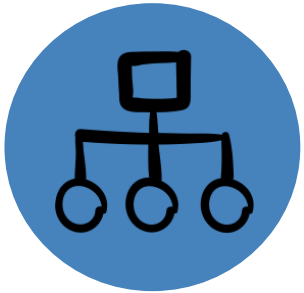
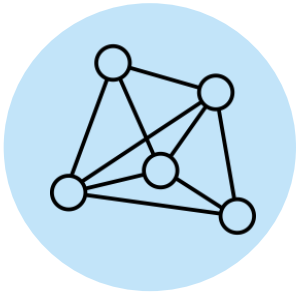


What can you do with an ontology?



Organizing data

Filtering data



Connecting data

Suggesting data

DOI: 10.1056/NEJMra1615014
Image credit: https://kyndi.com/blog/creating-knowledge-and-maximizing-the-value-of-data-with-ontologies/

Examples


GENEONTOLOGY
Unifying Biology


Envo


The
Sequence
ntology


mondo
MONARCH DISEASE ONTOLOGY


DISEASE
ONTOLOGY


SNOMED CT
The global
language of
healthcare


hpo


efo
Experimental Factor Ontology

Ontologies in Everyday Life

amazon

Movies & TV
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Blu-ray
DVD
Movies
Kids & Family
See more
Toys & Games
Action & Toy Figures
Toy Building Sets
Novelty & More
Movie & TV Fan T-Shirts
Women's Novelty Tops & Tees
Men's Novelty Shirts
Movie & TV Fan Accessories
Books
Marvel Comics & Graphic Novels
Superhero Comics & Graphic Novels
Comics & Graphic Novels
Teen & Young Adult Books
Teen & Young Adult Literature & Fiction
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Soundtracks
See All 24 Departments

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Or FREE One-Day Pickup
More Buying Choices
$13.69 (18 used & new offers)

Guardians of the Galaxy (
Prime Video
$3.99 – $17.99 Rent or Buy

Guardians of the Galaxy :
A group of intergalactic criminals a
control of the universe.
Cast


Chris Pratt
Peter Quill


Vin Diesel
Groot

Bra
Coc
Roc

Play trailer

Google

Chris Pratt
American actor

facebook.com/PrattPrattPratt

Christopher Michael Pratt is an American actor. Pratt came to prominence with his television roles, particularly for his role as Andy Dwyer in the NBC sitcom Parks and Recreation, for which he received ... Wikipedia

Born: June 21, 1979 (age 39 years), Virginia, MN
Height: 6' 2"
Spouse: Anna Faris (m. 2009–2018)
TV shows: Parks and Recreation, The O.C., Everwood, MORE
Children: Jack Pratt

Movies
View 25+ more


Guardians of the Galaxy
2014


Jurassic World: Fallen Ki...
2018


Avengers: Infinity War
2018


Passengers
2016


Guardians of the Galaxy V...
2017

2.
Ontology Structure

Ontologies are expressed in formal language
like Web Ontology Language (OWL)

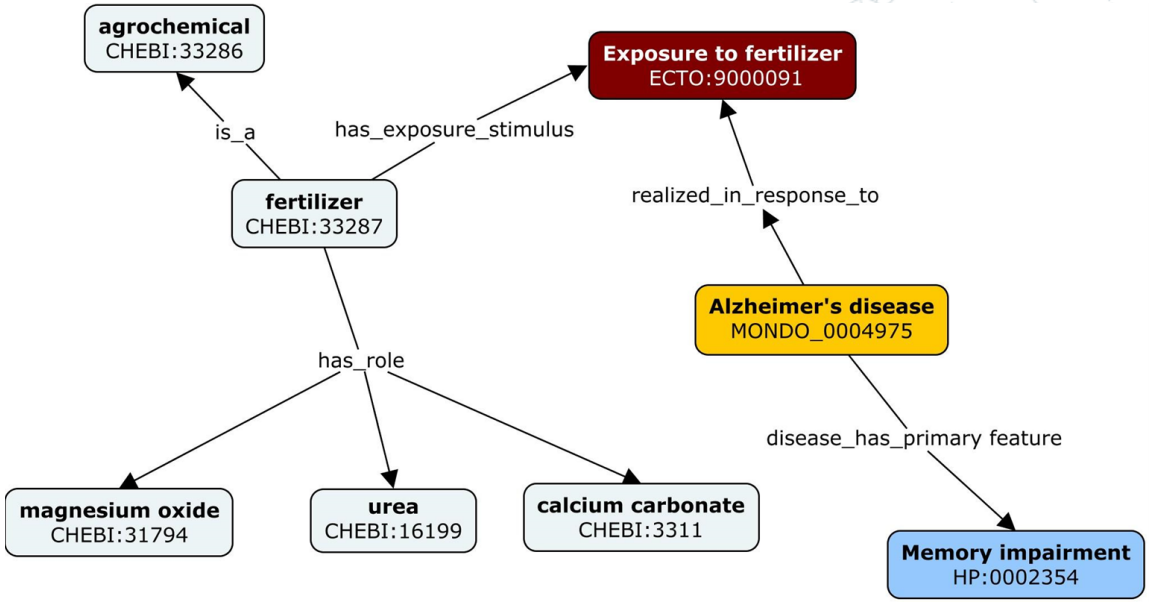
OWL is a semantic web computational logic-based language, designed to represent rich and complex knowledge about things and the relations between them. It also provides detailed, consistent and meaningful distinctions between classes, properties and relationships.

"Why not be inconsistent in at least one aspect of a language which is all about consistency?"

- Guus Schreiber, Why OWL and not WOL?

Key Features

- **Hierarchical classification:** Terms are arranged in a hierarchy
- **Text definitions:** Terms are defined
- **Synonyms:** alternative terms
- **Logical definitions:** Semantics - relationships between terms are defined, allowing logical inference and sophisticated data queries

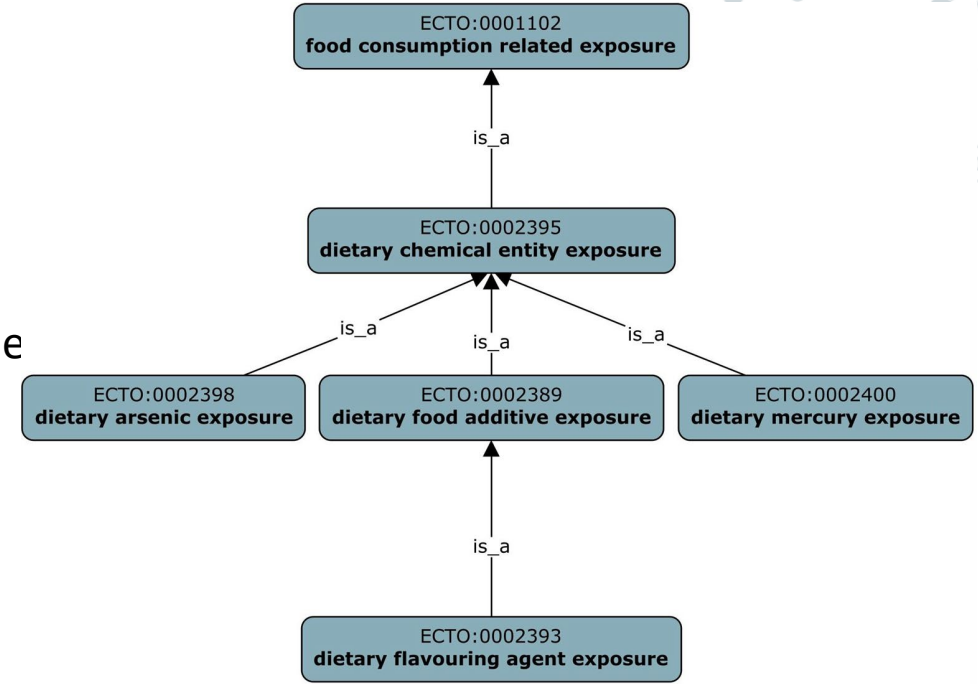


http://www.obofoundry.org/resources

The is_a relationship

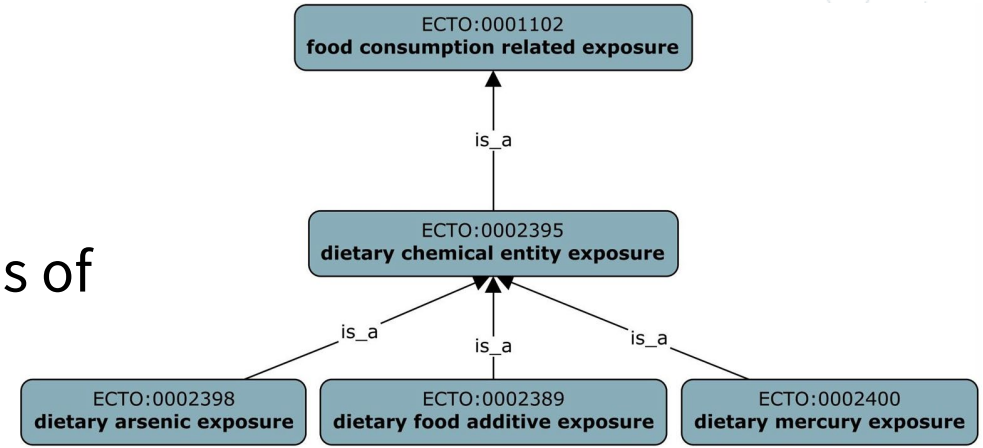
- Terms (classes) in an ontology are often classified via an ‘is_a relationship

- What is a subsumption hierarchy?
- Which class is a superclass of ‘dietary food additive exposure?
- Which class is a subclass of ‘food consumption related exposure’?
- Is ‘dietary flavouring agent exposure’ a type of ‘dietary chemical entity exposure’?
- What is the relationship of ‘dietary arsenic exposure’ to ‘dietary chemical entity exposure’?



The is_a relationship

- The **is_a** relation is like inheritance
- Children terms inherit the properties and relationships of the parent term



Dietary chemical entity exposure

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

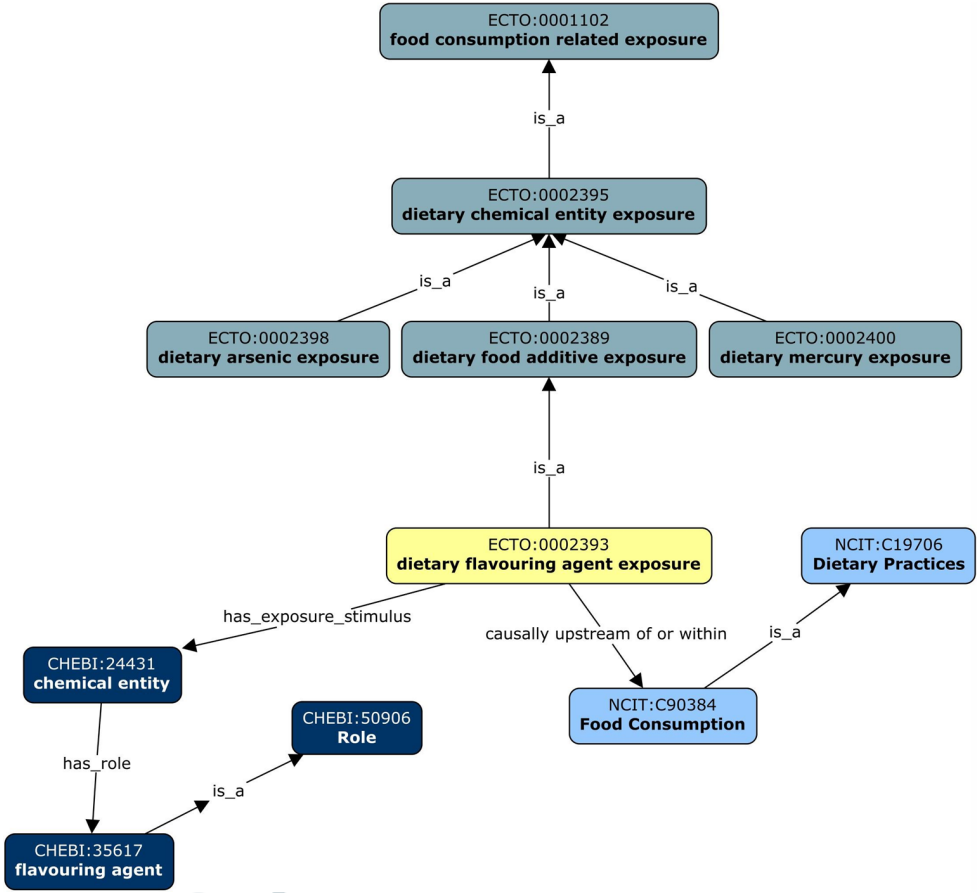
Dietary arsenic exposure

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

Ontologies textually and logically define the relationships between classes

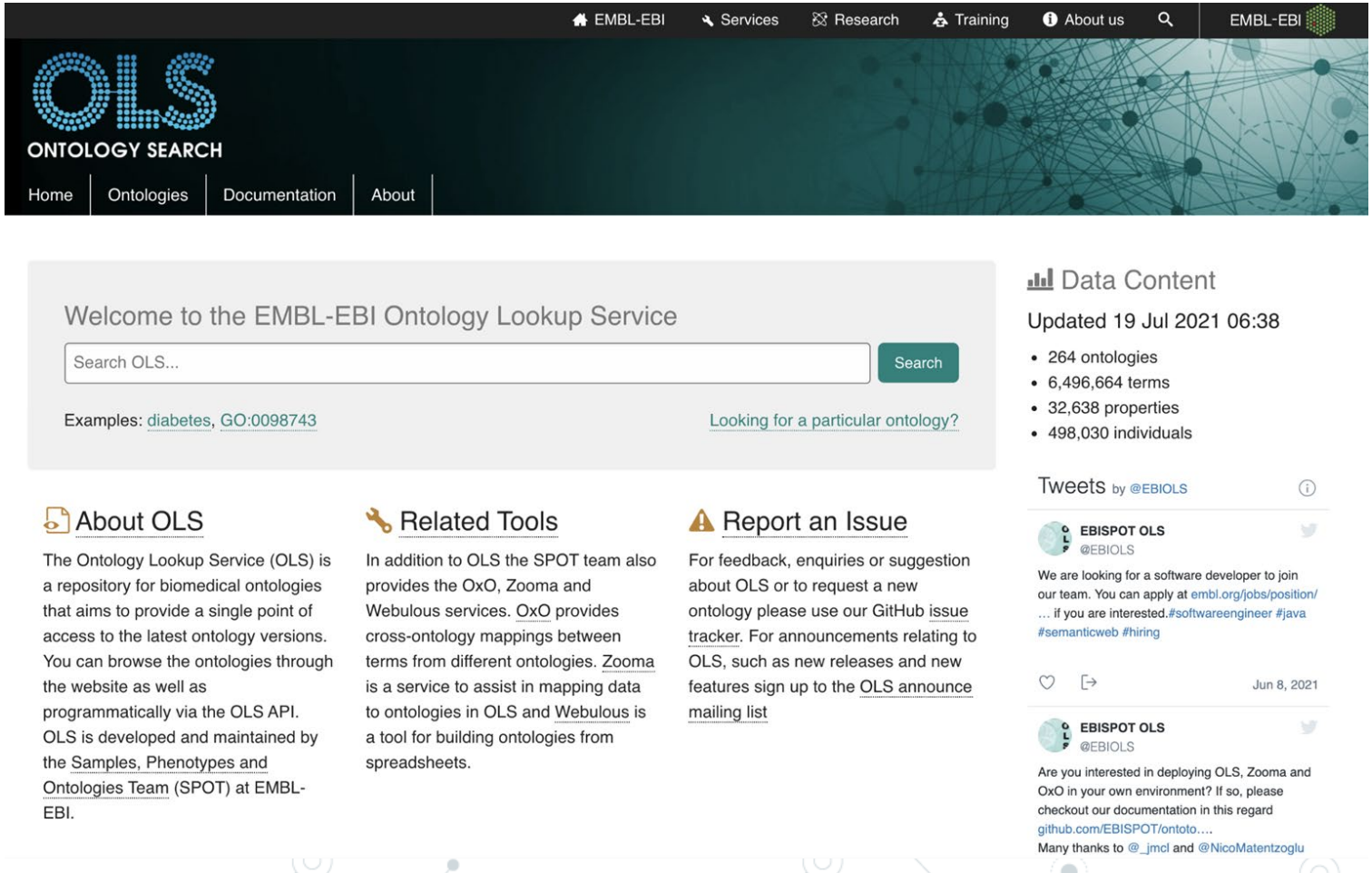
Text (human readable) definition
An exposure event arising as a result of ingestion of food containing chemicals with the role of flavouring agent.

Logical (machine readable) definition
'exposure event'
and ('has exposure stimulus' some
('chemical entity'
and ('has role' some 'flavouring agent')))
and ('causally upstream of or within' some 'Food Consumption')



3.

How to look up ontology terms/hierarchies



https://www.ebi.ac.uk/ols/index

OLS / Environment Exposure Ontology ECTO / ECTO:9000091 Copy

exposure to fertilizer

http://purl.obolibrary.org/obo/ECTO_9000091 Copy

Search ECTO Search

An exposure to fertilizer.

Synonyms: `exposure to fertilizer`

Tree view | Term mappings | Term history

- entity
  - occurrent
    - process
      - exposure event or process
        - exposure to chemical with application
          - exposure to agrochemical
            - exposure to fertilizer
  - exposure event
    - exposure to agrochemical
      - exposure to fertilizer
    - exposure to chemical with application
      - exposure to agrochemical
        - exposure to fertilizer
    - exposure to fertilizer

Graph view -

Reset tree

Show all siblings

Term information -

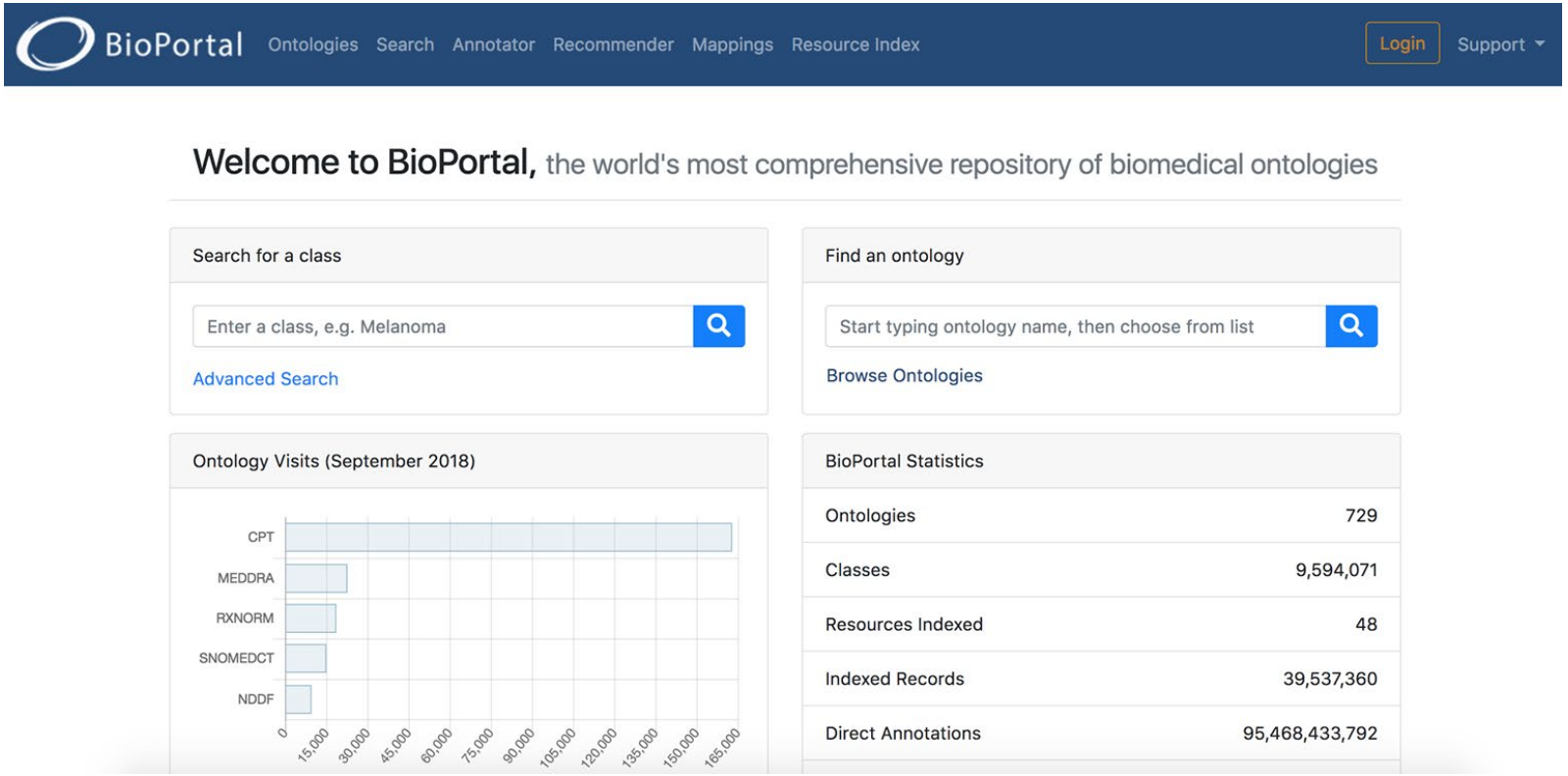
Term relations -

Equivalent to:

- exposure event and *has exposure stimulus* some fertilizer

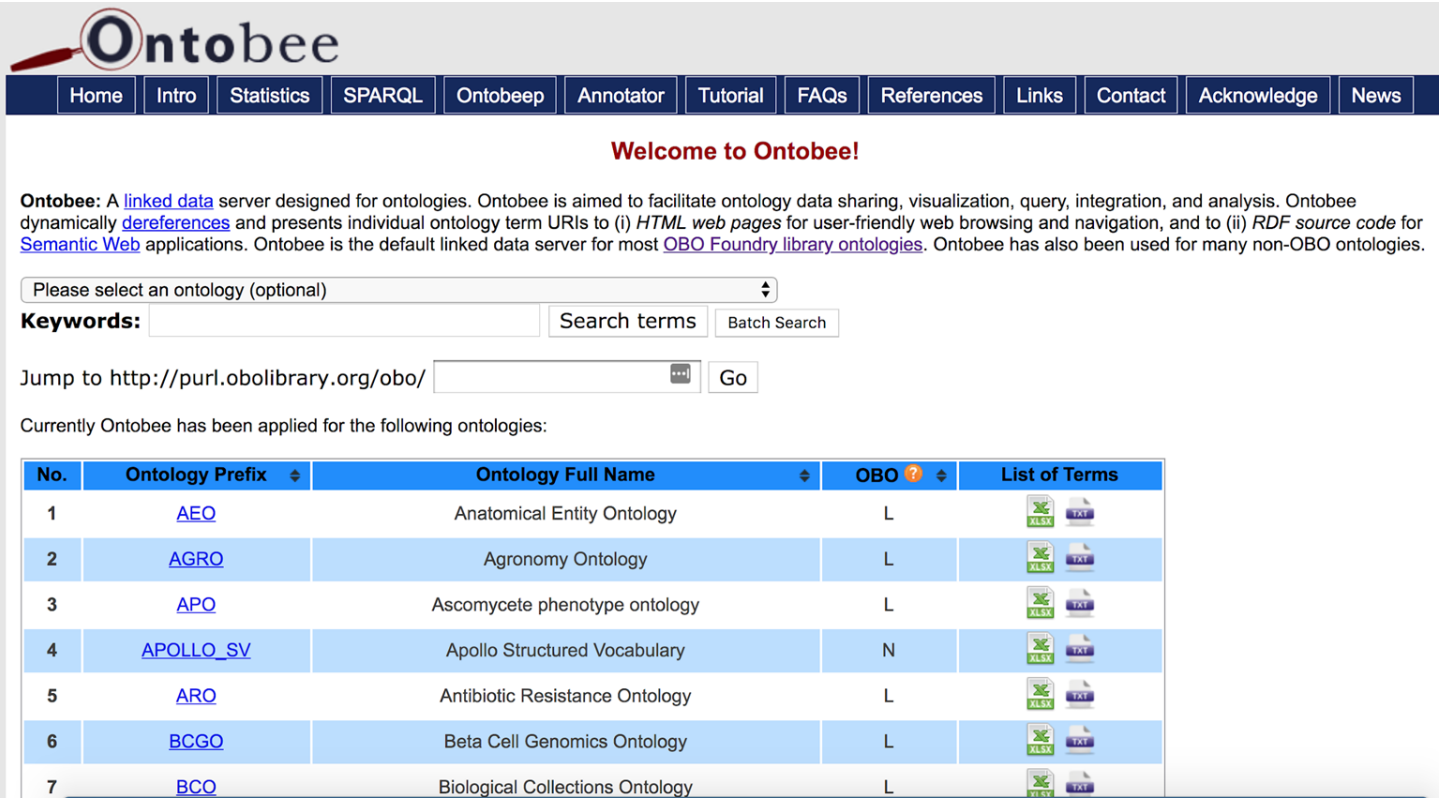
Subclass of:

- exposure event
- exposure to agrochemical
- *has exposure stimulus* some fertilizer



A library of ontologies and ontology related services

http://bioportal.bioontology.org/

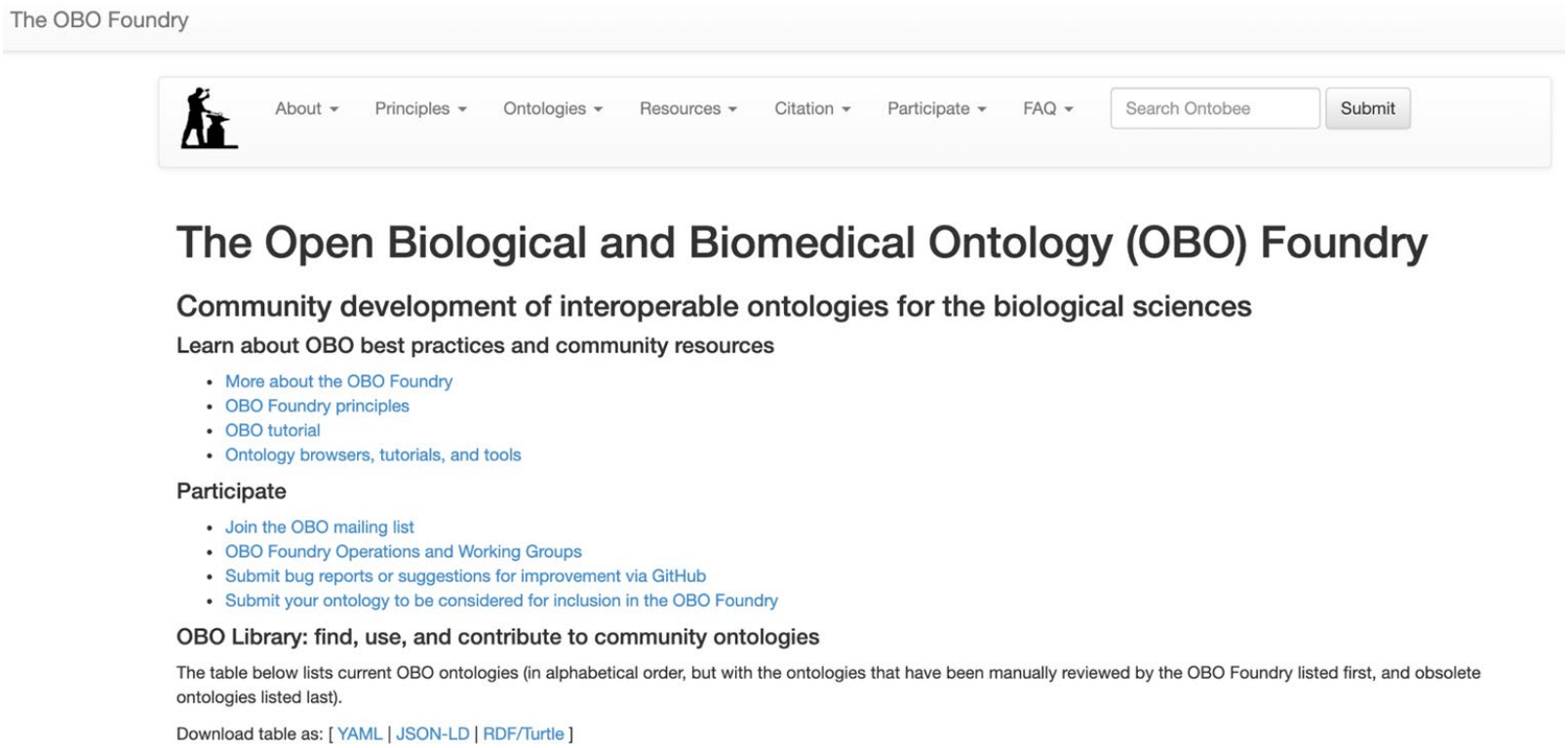


Query ontologies, and provides RDF supporting remote query of each ontology term and the Semantic Web

http://www.ontobee.org

Which is the right ontology to use?

- There are multiple ontologies that exist, 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 are 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



A community of ontologists committed to a shared set of principles to build open biomedical ontologies.

http://obofoundry.org/

3.

Managing Ontologies in GitHub

- Free, open-source online platform for hosting for software development projects
- Provides version control using Git
- Widely used in the ontology development community to:
  - store ontology files
  - version control
  - issue tracker

Sign up for free at: www.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

ecto

EnvironmentOntology / **environmental-exposure-ontology**

Watch 8

Code | Issues 68 | Pull requests 5 | Actions | Projects | Wiki | Security | Insights | Settings

master | 12 branches | 17 tags | Go to file | Add file | Code

laurenechan Make sure pipeline fails when duplicate ids in patterns (#193) ... 08dcac3 on Jun 11 **524** commits

.github/workflows	Make sure pipeline fails when duplicate ids in patterns (#193)	last month
imports	merge fork (#151)	4 months ago
mappings	Mappings 2021 (#169)	3 months ago
patterns	Release	4 months ago
reports	merge fork (#151)	4 months ago
src	Make sure pipeline fails when duplicate ids in patterns (#193)	last month
subsets	new release	3 years ago
.gitignore	Make sure pipeline fails when duplicate ids in patterns (#193)	last month

About

Modular environmental exposures ontology

environment | health | owl | ontology | obo | exposure | obofoundry

Readme

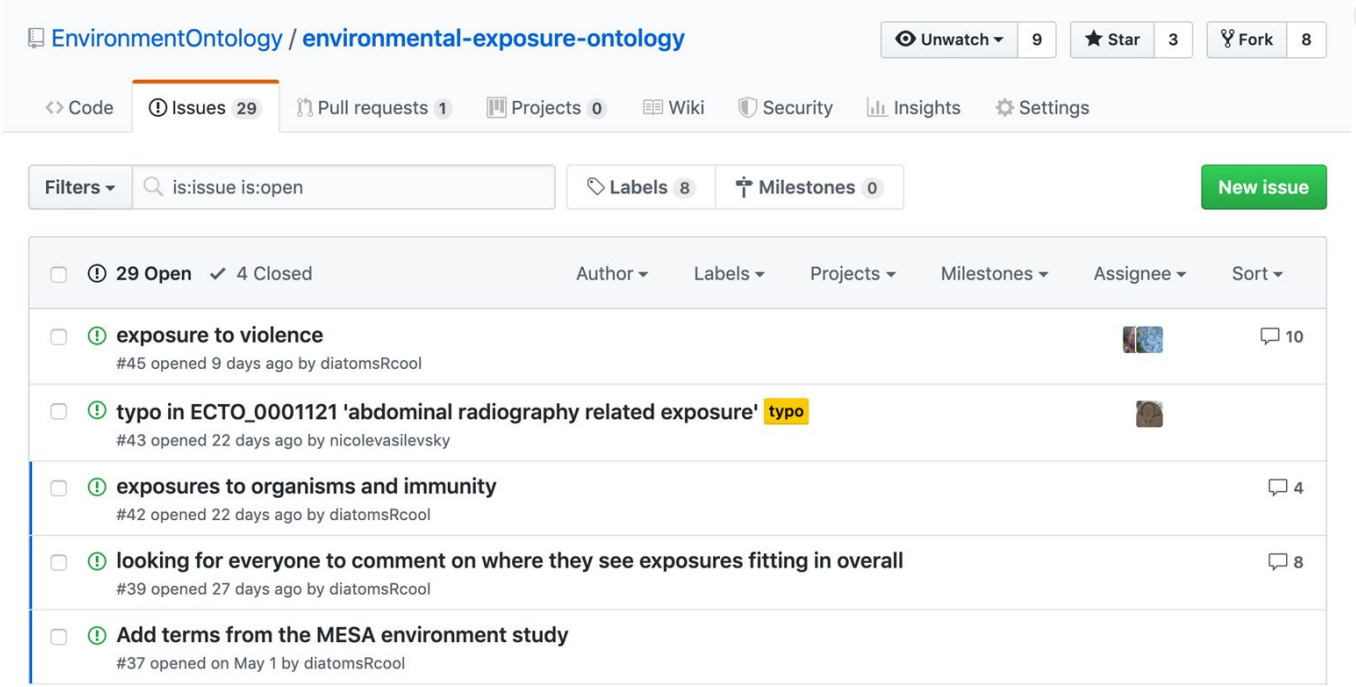
View license

Releases 17

v2021-03-17 Latest
on Mar 18

https://github.com/EnvironmentOntology/environmental-exposure-ontology

ECTO GitHub Issue Tracker



https://github.com/EnvironmentOntology/environmental-exposure-ontology/issues

How do you write a tracker request?

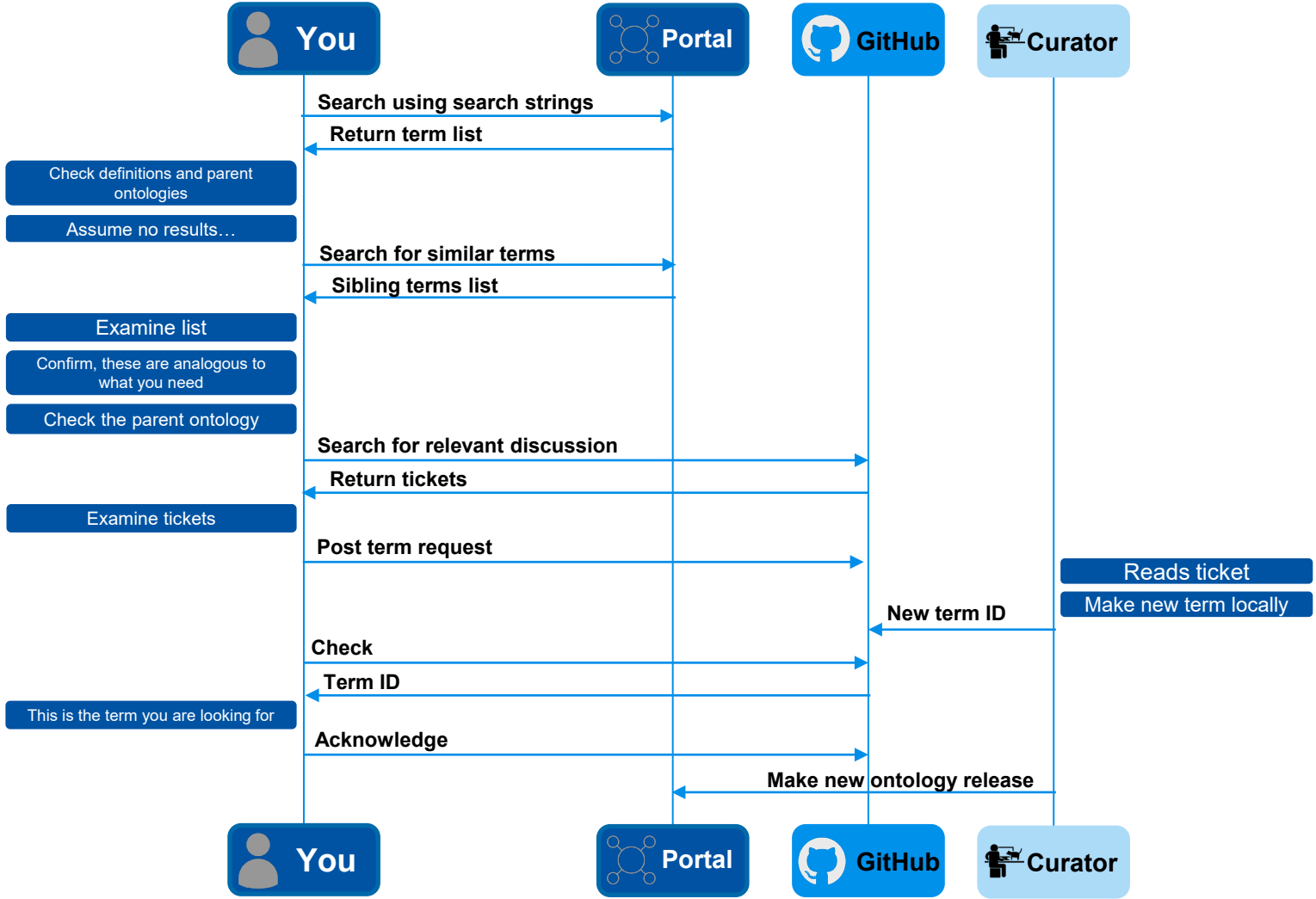
- Provide as much information as possible, in order to facilitate the change you are requesting and for future reference
- For new terms, or term rearrangements, provide the intended hierarchy – both parent/subclass as well as any other relations required
- Provide text definitions for all new or edited terms
- Provide attribution for the definitions
- Provide suggested synonyms

4.

Participate in Ontology Communities

Expert contributions to ontologies

Term search and request workflow



Adapted from:
https://douroucouli.wordpress.com/2021/07/03/how-select-and-request-terms-from-ontologies/

Summary

- Add value to your data by annotating your data with external ontologies
- Semantic enrichment aids in computational analysis
- Participate in ontology communities, your contributions are appreciated and valued
- Join OBO Foundry Slack: obo-communitygroup.slack.com

Acknowledgements

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Nico Matentzoglu
Semanticly

Anne Thessen
University of Colorado

Some content was adapted from:
BDK14: Introduction to OWL2 and data reasoning | https://github.com/OHSUBD2K/BDK14-Ontologies-101

Thanks!

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