


Open knowledge bases in the age of generative AI



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BOSC/BOKR Invited Talk
ISMB 2025

My history with the BOSC/BOKR community: from bioperl to Open Bio Ontologies

Open software,
tools, and libraries



Open ontologies and
knowledge bases



The Bioperl toolkit: Perl modules for the life sciences

Jason E Stajich ¹, David Block, Kris Boulez, Steven E Brenner, Stephen A Chervitz, Chris Dagdigan, Georg Fuellen, James G R Gilbert, Ian Korf, Hilmar Lapp, Heikki Lehtväslaiho, Chad Matsalla, Chris J Mungall, Brian I Osborne, Matthew R Pocock, Peter Schattner, Martin Senger, Lincoln D Stein, Elia Stupka, Mark D Wilkinson, Ewan Birney

Affiliations + expand

PMID: 12368254 PMCID: [PMC187536](#) DOI: [10.1101/gr.361602](#)

Ontologies for biologists: a community model for the annotation of genomic data.

Ashburner M ¹, Mungall CJ, Lewis SE

Author information ▶

Cold Spring Harbor Symposia on Quantitative Biology, 01 Jan 2003, 68:227-235

<https://doi.org/10.1101/sqb.2003.68.227> PMID: 15338622

Review

Outline

The role of knowledge representation in AI

Knowledge representation as **open team science**

How will **agentic AI** change open knowledge representation?

AI alignment and knowledge representation

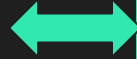
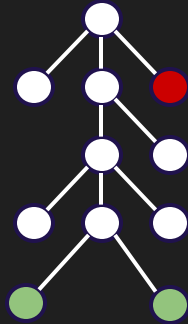
Knowledge representation: ontologies, knowledge bases, and data repositories

Statistics	
Ontologies	1,213
Classes	17,745,672
Properties	36,286
Mappings	93,177,234



BioPortal

ontologies



Knowledge bases
(sensu NIH)

GO, Reactome,
Monarch, MODs,
ClinGen, UniProt,
DrugBank, ...



Data repositories

ENCODE, DCCs,
NMDC, SRA,

The graph of biological knowledge

Phenotypic abnormalities

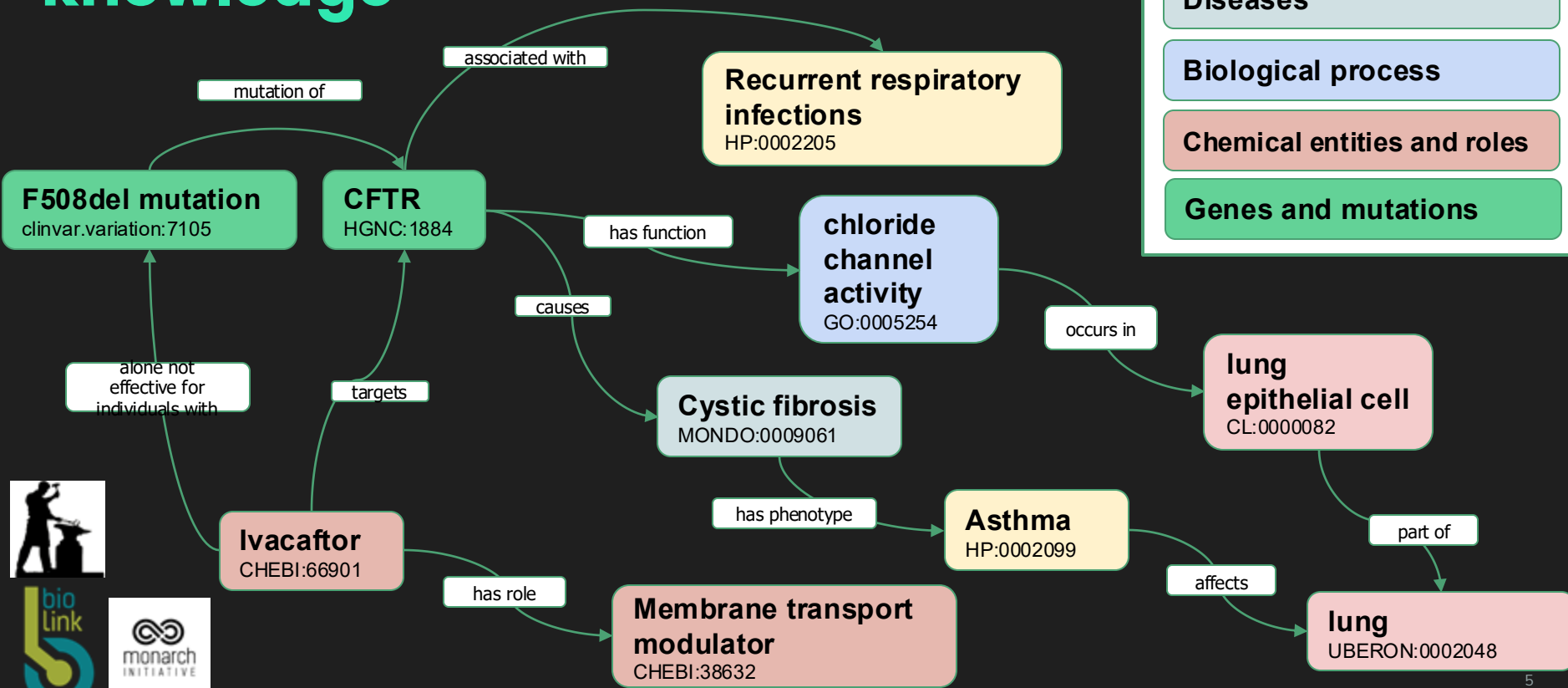
Anatomical entities

Diseases

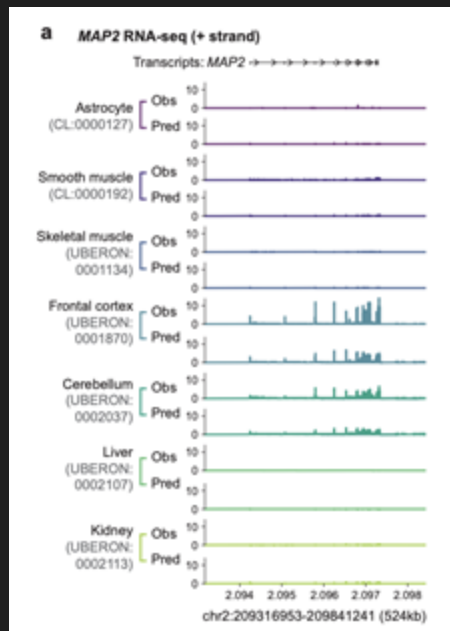
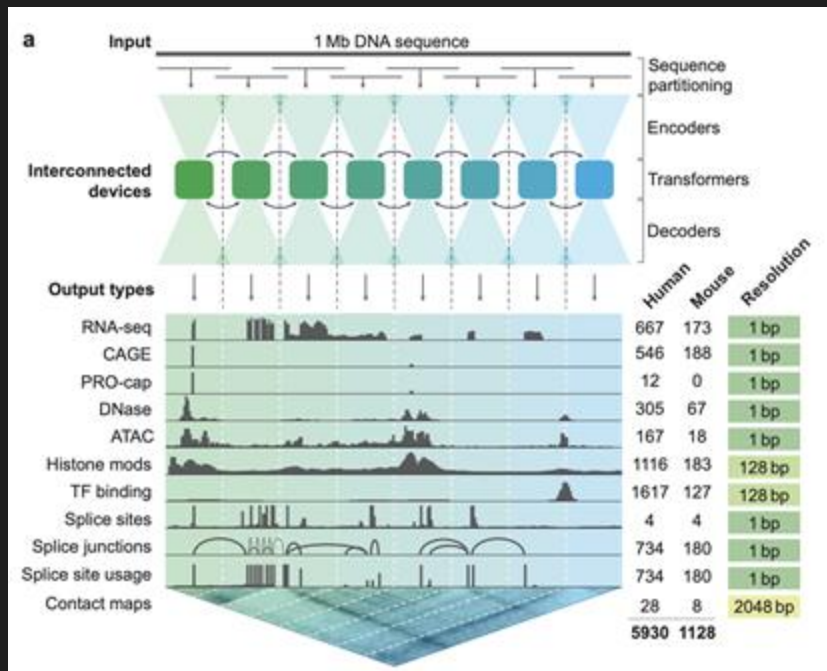
Biological process

Chemical entities and roles

Genes and mutations

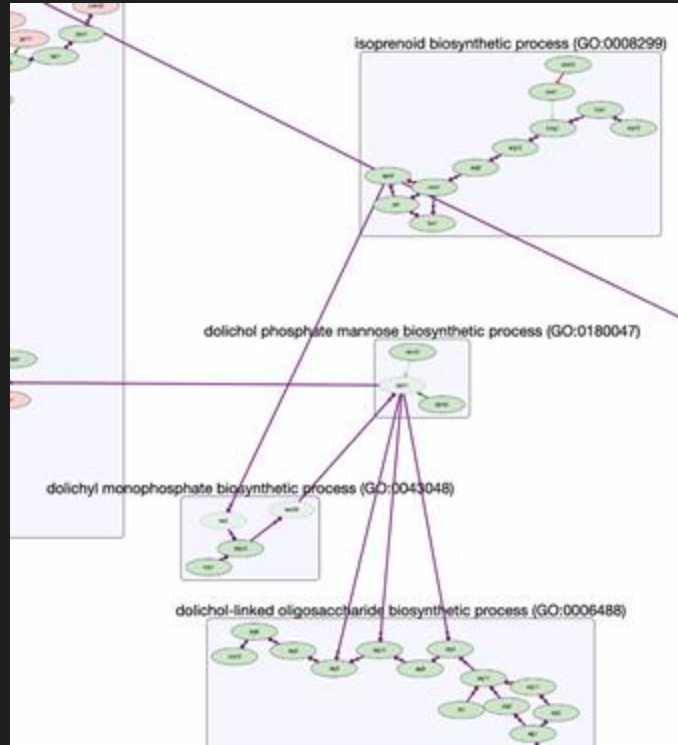


Knowledge representation powers AI



Avsec, Žiga, et al. "AlphaGenome: advancing regulatory variant effect prediction with a unified DNA sequence model." *bioRxiv* (2025): 2025-06.

Knowledge representation has explanatory power, not just predictive



Gene Ontology Causal
Activity Models (GO-CAMs)
Representation of causal
biology knowledge



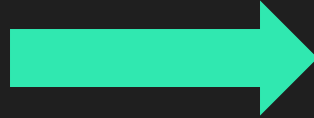
PomBase

<https://pombase.org/gocam/mega-model/connected>
<http://geneontology.org/gocam>

Knowledge representation encompasses modeling, curation, and ontology development

Knowledge
(unstructured and
semi-structured)

Modeling
Curation
Ontology Development



Computable
consistent
representation
of knowledge

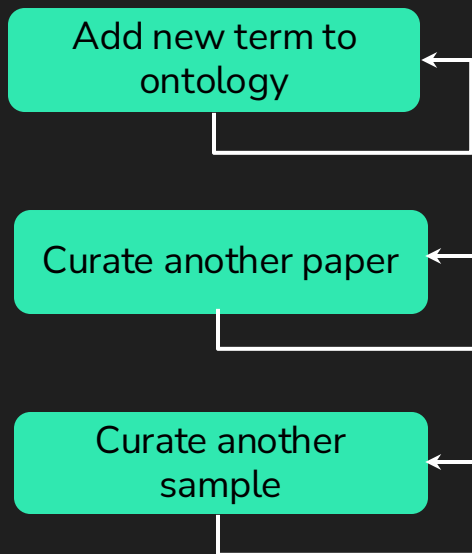
The future of biocuration

[Doug Howe](#) , [Maria Costanzo](#), [Petra Fey](#), [Takashi Gojobori](#), [Linda Hannick](#), [Winston Hide](#), [David P. Hill](#), [Renate Kania](#), [Mary Schaeffer](#), [Susan St Pierre](#), [Simon Twigger](#),
[Owen White](#) & [Seung Yon Rhee](#) 

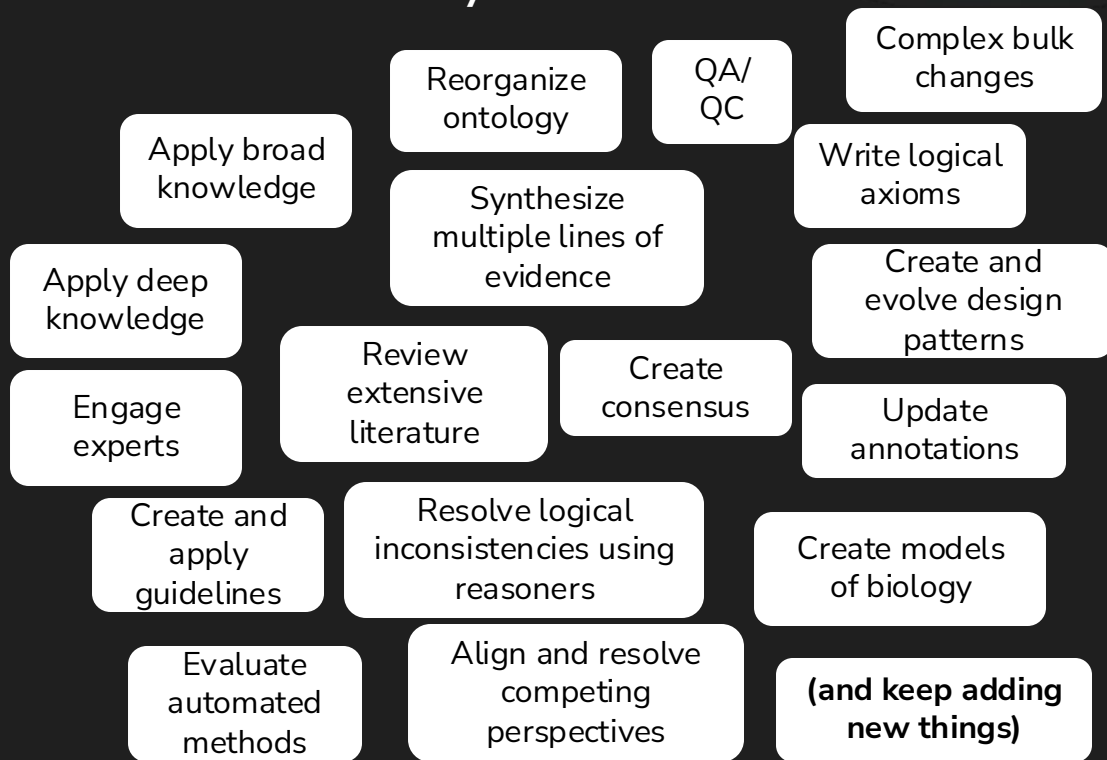
[Nature](#) 455, 47–50 (2008) | [Cite this article](#)

Knowledge Representation: **Tasks**

What people **think** we do

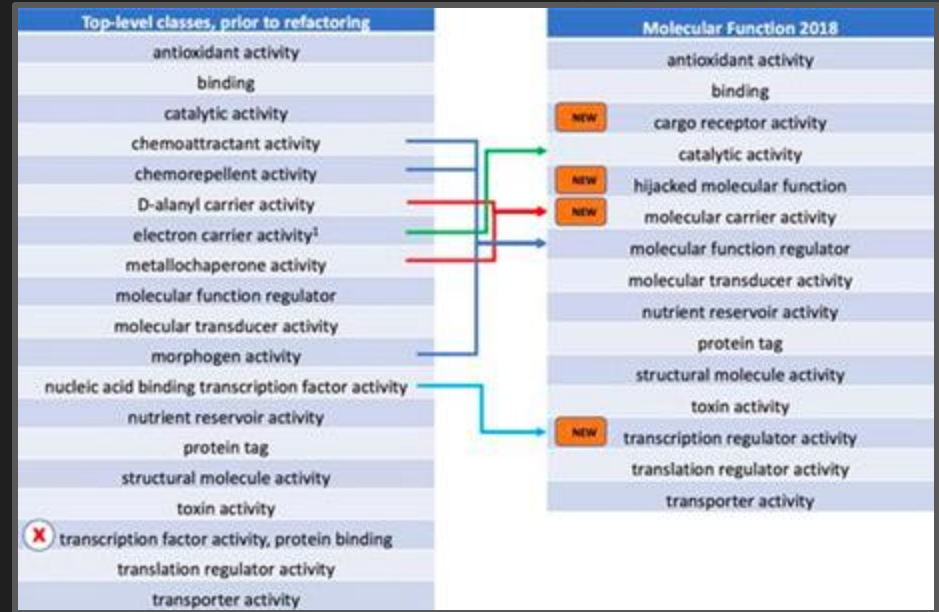


What we **really** do



Knowledge representation is a fluid process

- Change is a constant
 - Scientific knowledge changes
 - Our knowledge representation changes
- **Refactoring** knowledge bases common and **time consuming**
 - (just like with software)



Refactoring the molecular function branch of GO

from:

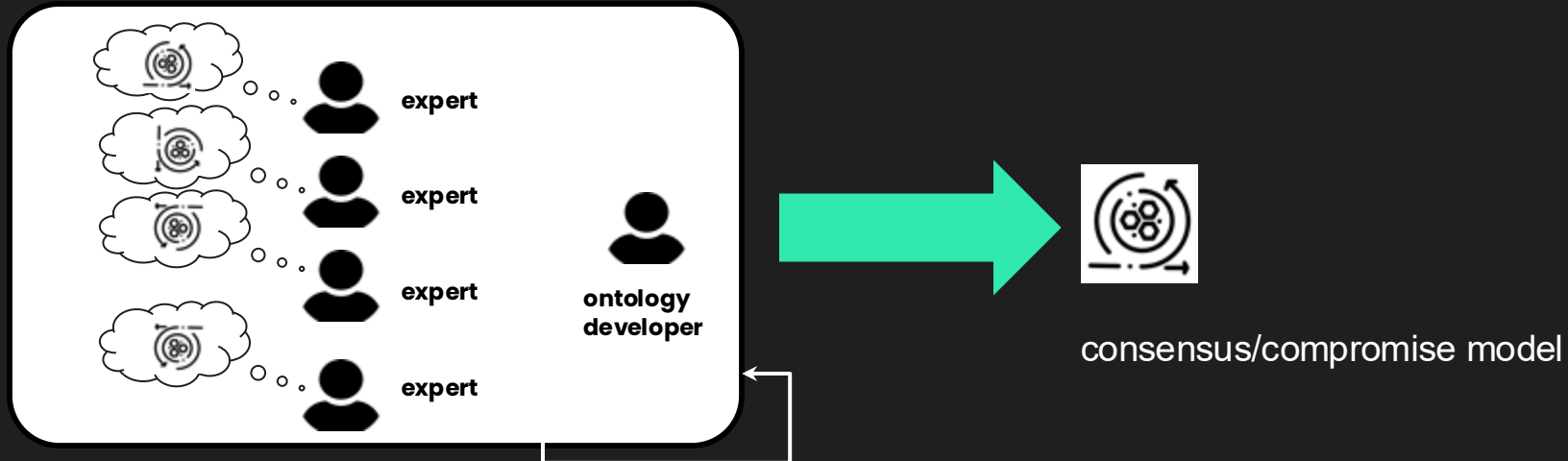
Gene Ontology Consortium. (2019). The gene ontology resource: 20 years and still GOing strong. Nucleic acids research, 47(D1), D330-D338.

Biological knowledge representation is consensus creation

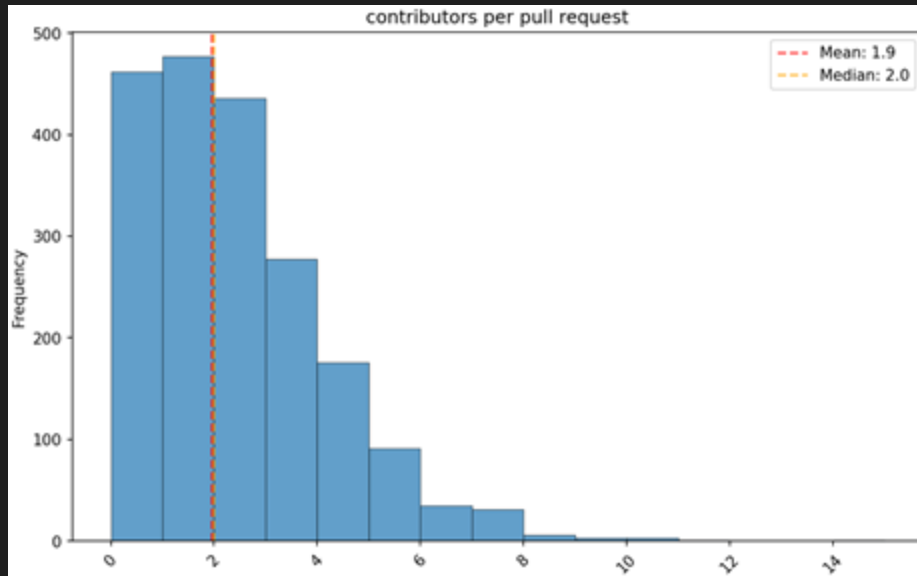
Ontology development is consensus creation, not (merely) representation

[Fabian Neuhaus](#) ✉ and [Janna Hastings](#) ✉ [View all authors and affiliations](#)

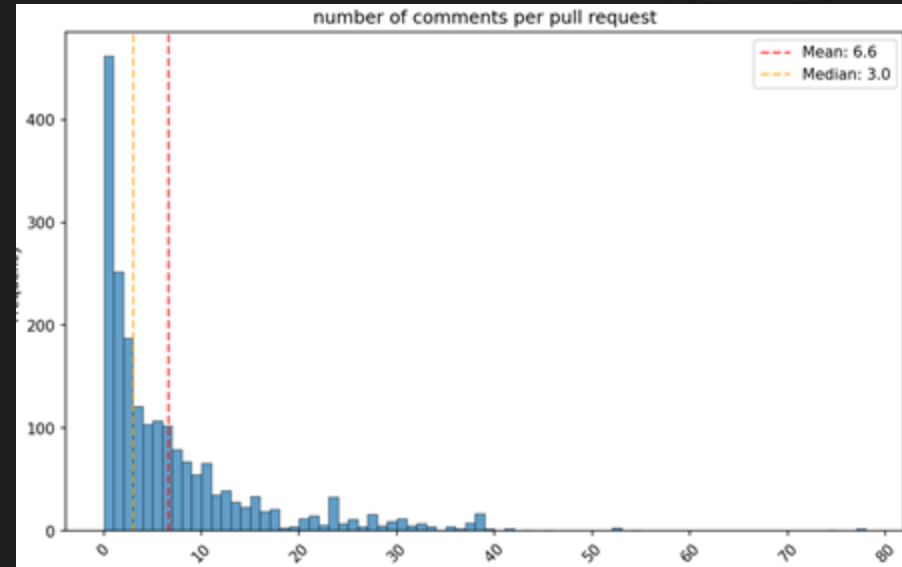
[Volume 17, Issue 4](#) | <https://doi.org/10.3233/AO-220273>



Knowledge Representation: discussion and consensus making



Multiple people involved in each decision

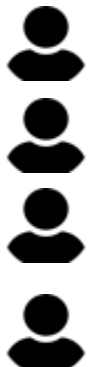


Decisions involve (often intense) discussion

Gene Ontology Pull Requests (PRs) Jan 2024 -> now

Consensus knowledge representation: Mondo Epilepsy Workshops

- Series of open workshops 2024-2025
- Consensus revisions

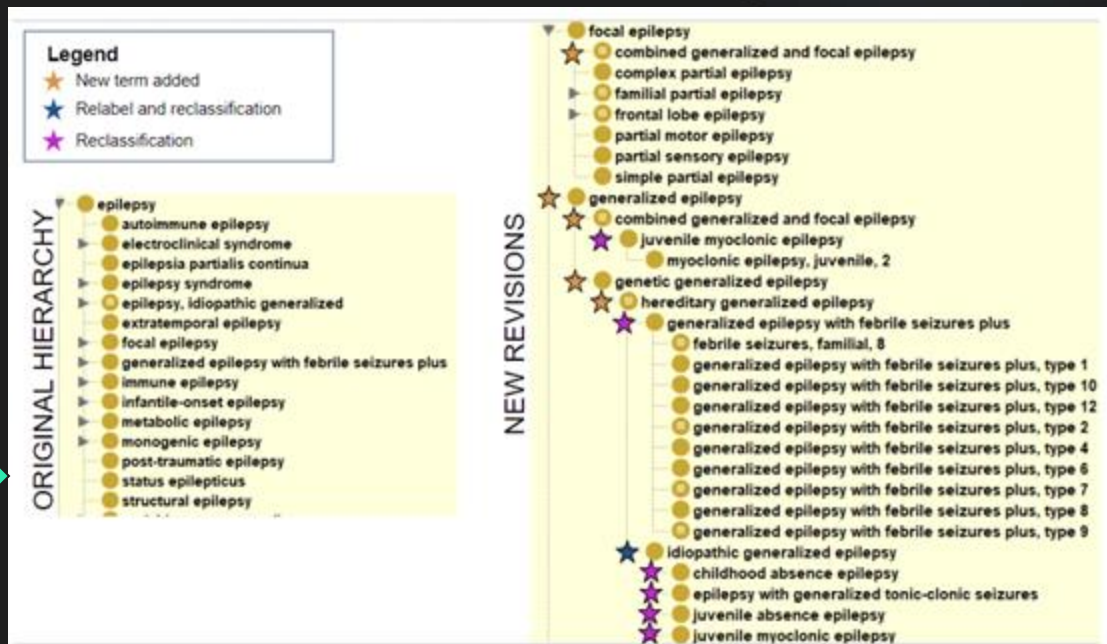
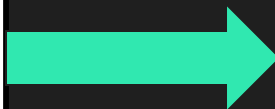


Clinicians

Geneticists

Patient
Advocates

Curators and
ontology developers



Can LLMs alone help with this? (spoiler: NO)

Naive approach: ChatGPT-as-Oracle

- Works for toy examples or small ontologies
- Unable to hold mid-size to large ontologies in memory
- Prone to hallucination
- **Poorly aligned with the real challenges**

make an ontology of immune cell types in obo format

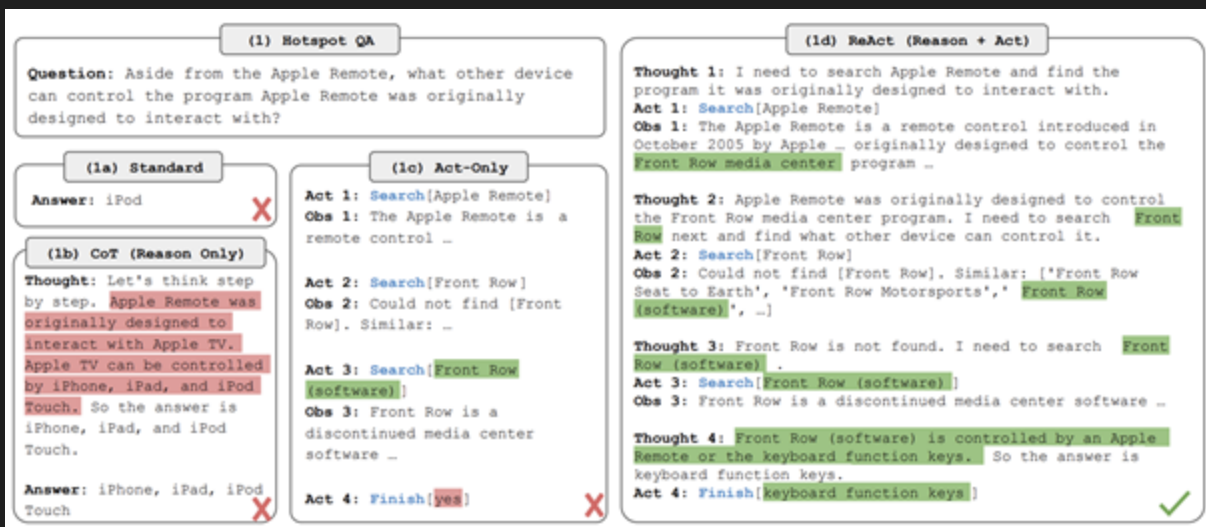
Here's a simple ontology of immune cell types in OBO format, based on established categories (innate vs adaptive, lineage types, etc.), suitable for extension or integration into a broader ontology such as the Cell Ontology (CL).

```
[Term]
id: IMM:0000000
name: immune cell
def: "A cell that is part of the immune system"

[Term]
id: IMM:0000001
name: lymphocyte
is_a: IMM:0000000 ! immune cell
def: "A type of white blood cell involved in the immune response"

[Term]
id: IMM:0000002
name: B cell
is_a: IMM:0000001 ! lymphocyte
def: "A lymphocyte that matures in the bone marrow and produces antibodies"
```

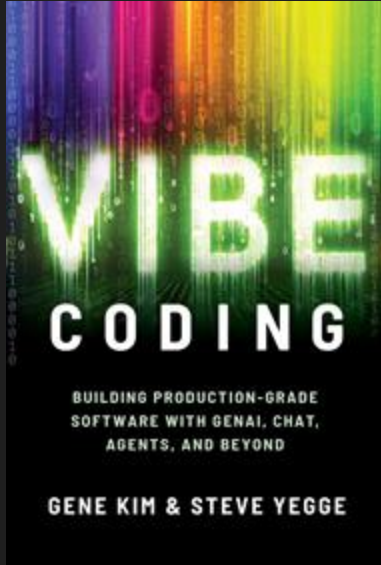
Agentic AI is more aligned with the tasks we really need help with



Yao, S., et al. (2022). ReAct: Synergizing Reasoning and Acting in Language Models. arXiv:2210.03629.

Schick, T., et al. (2023). Toolformer: Language Models Can Teach Themselves to Use Tools. arXiv:2302.04761.

Agentic AI has taken software development by storm



Agentic AI coders

Copy-paste from
ChatGPT

"Autocomplete" tools in
IDE

Why agents for knowledge bases?

Tasks and Tools



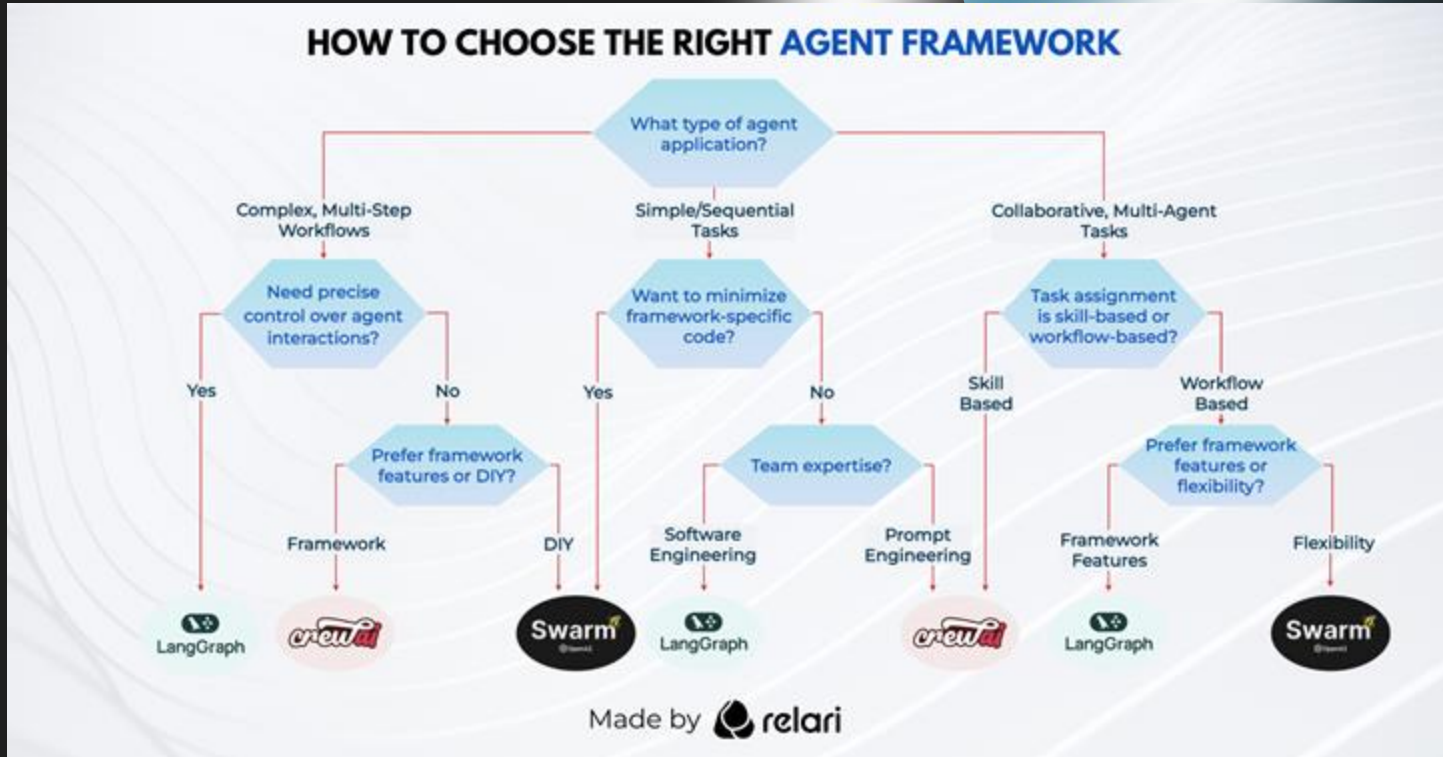
Add
transcriptomically
identified molecular
layer interneuron
subtypes to the cell
ontology



Plan:

- > Review existing ontology structure using **OLS/BioPortal**
- > Download and review relevant publications using **NCBI API**
- > Check out the ontology from the repo using **git**
- > Make edits to the ontology using **text editing tools**
- > Align with design patterns for transcriptomics terms
- > Run OBO standard checks using **ROBOT**
- > Reason over the ontology using **Whelk**
- > Everything OK? **If not keep iterating?**
- > Commit changes and make a pull request using **GitHub tools**

Building agents may seem daunting...



We already adapt software engineering tools and strategies for KR



Formal Validation and Linting

We use OWL Reasoners to validate ontologies, analogous to type checking for programs



Containerized CI/CD

Ontologies are managed in GitHub, with PRs validated using the Ontology Development Kit



Development Environments

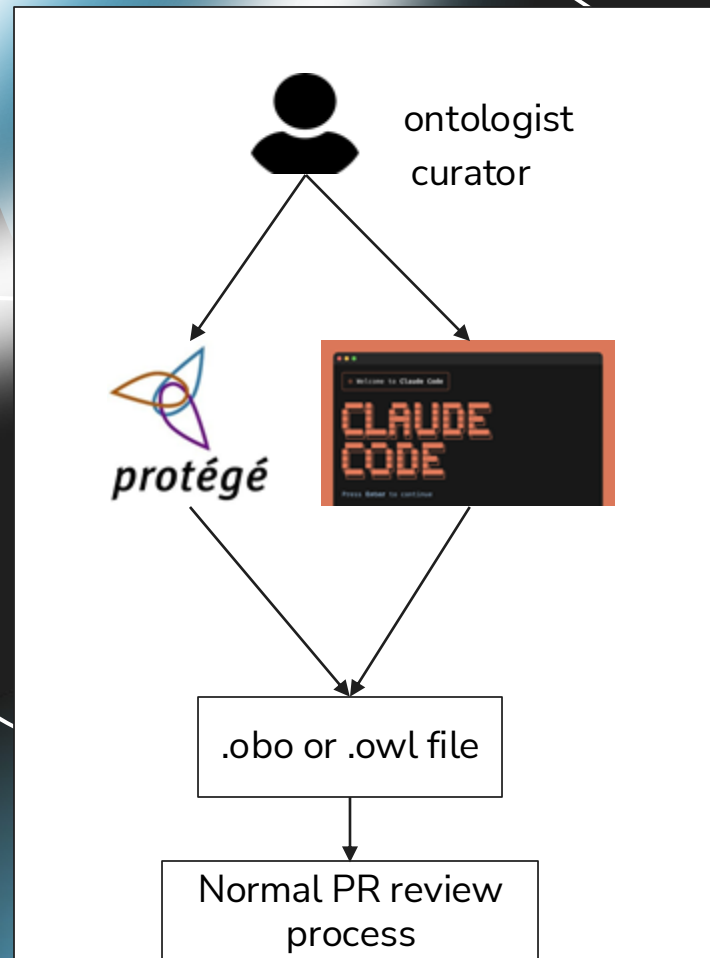
Ontologies are developed using Ontology Development Environments (ODEs) such as Protege, analogous to software IDEs





Reuse what is out there: The Semantic Coding Agent pattern


We favor a **lightweight approach** using **off the shelf coding agents**, customized using **simple text instructions** and **command line tools**

We call this the **Semantic Coding Agent pattern**



Everyone is a agent programmer

 master [uberont / CLAUDE.md](#) 

 **cmungall** Merge branch 'master' into Claude_NT_Ed_meta

Preview

Code

Blame

148 lines (117 loc) · 6.55 KB

UBERON Ontology Project Guide

This includes instructions for editing the uberont ontology.

Project Layout

- Main development file is `src/ontology/uberont-edit.obo`
- individual terms checked out in `terms`
- ODK and ontology documentation can be found in `/docs/`

Querying ontology

- To look at a specific term if you know the ID:
 - `obo-grep.pl -r 'id: UBERON:0004177' src/ontology/uberont-edit.obo`
- All mentions of an ID
 - `obo-grep.pl -r 'UBERON:0004177' src/ontology/uberont-edit.obo`

Relationships

All terms should have at least one `is_a` (this can be implicit by a logical definition, see below relationships).

Logical definitions

These should follow genus-differentia form, and the text definition should mirror the logical d

```
[Term]
id: UBERON:0001742
name: epiglottic cartilage
def: "The most superior of the laryngeal cartilages, which is found at the root
subset: uberont_slim
xref: EMAPA:37522 {source="MA:th"}
xref: FMA:55112
xref: MA:0001763
xref: NCIT:C32526
xref: SCTID:278990001
xref: UMLS:C0458526 {source="ncithesaurus:Epiglottic_Cartilage"}
intersection_of: UBERON:0001739 ! laryngeal cartilage
intersection_of: part_of UBERON:0000388 ! epiglottis
relationship: composed_primarily_of UBERON:0001996 ! elastic cartilage tissue
```

The reasoner can find the most specific `is_a`, so it's OK to leave this off.

Agents love tools. We have tools

Custom	robot	Reasoning, Validation, General Ontology Processing
	artl-mcp	Full text lookup
	obo-scripts	Tools for obo format files (search, extraction)
	OAK	Tools for ontology querying
	linkml	Schema operations
	ols-mcp	External ontology search
	dosdp-tools	Design pattern compliance
Builtin	Web search	Generic web search (including literature)
	File edit	Targeted editing of files (including ontology files)
	Python	Arbitrary ad-hoc scripting
	Unix tools	Arbitrary command line processing and querying



Multiple ways to invoke agents

Technical /
Powerful

Easy to use /
Less flexible

Local Command Line Agent Tools

- Claude Code
- Codex
- Gemini-CLI

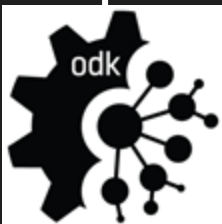
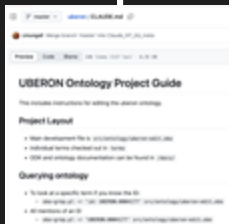
Agent Desktop Agent UIs

- Goose
- Claude Desktop

GitHub Agents

- @Copilot
- @dragon-ai-agent

Instructions.md



Tools (mcp, cli)

Documenting patterns

The screenshot shows the AI4Curators - AI Guides website. The header is blue with a logo, the title 'AI4Curators - AI Guides', a search bar, and a GitHub link. The left sidebar contains a navigation menu with categories like Home, Curator how-tos, Administrator how-tos, Tutorials, Reference, and FAQ. The main content area features the article 'How to integrate AI into your Knowledge Base' with a table of contents on the right. The article text includes tips on using simple MCPs or command line tools, and mentions frameworks like ODK-AI.

AI4Curators - AI Guides

Search

GitHub

Table of contents

Tip 1: No fancy frameworks needed - just simple MCPs or command line tools

Tip 2: Keep AI instructions checked in at the root of your GitHub repo

Tip 3: Train curators to use simple tool-enabled AI applications (e.g Goose)

Set up GitHub actions

Document and Train

Continuous evaluation.

How to integrate AI into your Knowledge Base

This is a high level advanced guide for maintainers of knowledge bases with some high level pointers on how to start effectively using AI to enhance curation.

Tip 1: No fancy frameworks needed - just simple MCPs or command line tools

Intimidated by the growing number of agent frameworks? Don't worry, you don't need most of these.

All you really need are a handful of simple MCPs servers or command line tools. These can be hooked up to generic frameworks.

The command line tools could be wrapped in a Docker container; this is the strategy for ODK-AI.

You should rely on existing tools for doing things like literature search - you only need to write MCPs that are specific to read/write/validation on your KB.

Current agent deployments

Ontologies

- GO
- UBERON
- CL
- MONDO
- FBbt (Fly anatomy)
- OPTIMO

Semantic Schemas

- Biolink
- Chemrof

Registries

- KG-Registry

Annotations + KGs

- Monarch pathophysiology annotations
- MaterialsKG

Experimental: only trained ai-controller can invoke

Agents can handle **simple** **new term requests**

NTR: episcleral vein #1680

Open #3582

paola-scibite opened on Nov 20, 2020

Preferred term label:
episcleral vein

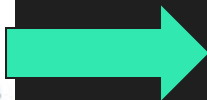
Synonyms

Definition (free text, please give PubMed ID)
A vein located between the bulbar conjunctiva and the sclera.
https://en.wikipedia.org/w/index.php?title=Episcleral_layer&oldid=87081125
FMA:51800

Parent term (use <https://www.ebi.ac.uk/ols/ontologies/uberon>)
subclass of UBERON:0009141 craniocervical region vein (or possibly UBERON:0010292 episcleral layer of eyeball)

Your nano-attribution (ORCID)
<https://orcid.org/0000-0002-2825-0621>

Thanks!
Paola



Add episcleral vein term (UBERON:7770009) #3582

Open Copilot wants to merge 3 commits into [master](#) from [copilot/fia-3582](#)

Conversation 3 Commits 3 Checks 4 Files changed 3

Copilot AI commented [last week](#) · edited ·

This PR adds a new term "episcleral vein" to the UBERON ontology as requested in issue [#1680](#).

New Term Details

UBERON:7770009 - episcleral vein

- Definition: "A vein located between the bulbar conjunctiva and the sclera."
- Parent class: UBERON:0009141 (craniocervical region vein)
- Part of: UBERON:0010292 (episcleral layer of eyeball)
- References: FMA:51800, Wikipedia:Episcleral_layer
- Contributor: Paola Roncaglia (<https://orcid.org/0000-0002-2825-0621>)

Changes Made

- Added new term to `src/ontology/uberon-edit.sbo` following existing patterns
- Created corresponding term file `series/UBERON_7770009.sbo`
- Included proper metadata: contributor ORCID, creation date, issue tracker reference
- Used next available ID in the 777xxx range (UBERON:7770009)
- Verified term relationships and references are valid

Validation

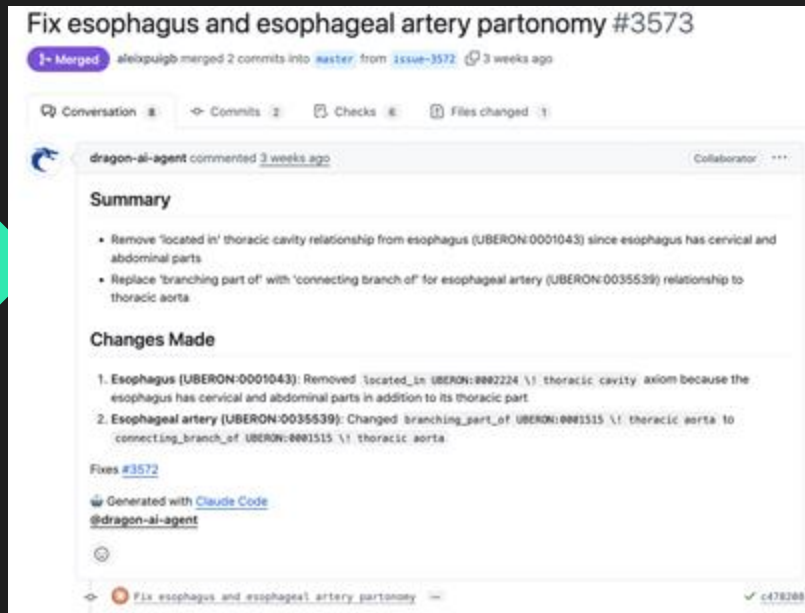
The new term has been validated using the existing `sbo-grep.pl` tool and confirmed to:

- Be properly formatted according to OBO standards
- Have valid parent and part_of relationships
- Include all required metadata fields
- Be searchable within the ontology

Fixes [#1680](#).

- Note that most of the intellectual work is done **up-front by the curator**
- The agent takes care of the mechanical and time consuming part

Agents can handle **simple relationship changes**



- Note that most of the intellectual work is done **up-front by the curator**
- The agent takes care of the mechanical and time-consuming part

Agents can perform deep research

Update alpha gal syndrome #9259



omungall opened 3 weeks ago

Member

Looking at

```
[Term]
id: MONDO:0100001
name: alpha-gal syndrome
def: "An IgE antibody response to a mammalian oligosaccharide epitope, galactose- $\alpha$ -1,3-galactose (alpha-gal)
synonym: "AGS" EXACT ABNOVATION [https://www.cdc.gov/ticks/alpha-gal/index.html]
synonym: "allergic galactose- $\alpha$ -1,3-galactose disease" EXACT [https://orcid.org/0000-0001-5286-3432]
synonym: "alpha-gal allergy" EXACT [https://www.cdc.gov/ticks/alpha-gal/index.html]
synonym: "red meat allergy" EXACT [https://www.cdc.gov/ticks/alpha-gal/index.html]
synonym: "tick bite meat allergy" EXACT [https://www.cdc.gov/ticks/alpha-gal/index.html]
xref: MEDGEN:177844 (source="MONDO:equivalentTo", source="MEDGEN:177844", source="MONDO:MEDGEN")
xref: UMLS:C5442181 (source="MONDO:equivalentTo", source="MEDGEN:177844", source="MONDO:MEDGEN")
is_a: MONDO:0005271 (source="PMID:25747720") ! allergic disease
property_value: http://purl.org/dc/elements/1.1/date "2018-03-13T04:40:23Z" xsd:dateTime
property_value: http://purl.org/dc/terms/creator https://orcid.org/0000-0001-5286-3432
property_value: IAO:0000233 "https://github.com/monarch-initiative/mondo/issues/182" xsd:anyURI
```

This is missing information eg causative agent, symptoms. Also the definition isn't quite right - a disease is not a response.

@dragon-ai-agent please do some research on this ticket. Rather than make a PR right away, I would like you to first help plan what to do. Please post findings directly to this ticket in real time as you find them. We can then discuss them further.

First check up to date scientific resources for the latest on this disease. Check the URLs that are used in this term to support synonyms etc, they may be no longer active.

In order to figure out how best to model this term, you should consult both the editors documentations (in /docs/) and the design pattern docs (in /src/patterns/design-patterns/). You can also look for precedent in the ontology, or for similar issues in this tracker. Find which patterns might be appropriate (and if you can't find a pattern you expect, report it). Again, post findings or even general questions to me to this issue tracker as you go along.

Also look for the latest info on phenotypes/symptoms, and potential etiology, from the right ontology terms or database links.



dragon-ai-agent 3 weeks ago

Starting research on alpha-gal syndrome (MONDO:0100001). I'll post my findings here as I discover

Initial Assessment

Looking at the current term definition: "An IgE antibody response to a mammalian oligosaccharide α -1,3-galactose (alpha-gal)."

You're absolutely right - this definition is problematic because:

1. A disease is not a response - it should describe the condition/syndrome itself
2. Missing key information about causative agent (tick bites)
3. Missing symptom information
4. May need updated scientific understanding

Research Plan

I'll investigate:

1. Current scientific literature and updates
2. Verify URLs used for synonyms
3. Review editor documentation and design patterns
4. Look for similar terms in ontology for
5. Identify appropriate phenotypes/symptoms
6. Check for related issues in tracker

Starting now...



dragon-ai-agent 3 weeks ago

Design Pattern Analysis

Found two relevant patterns for allergic diseases:

1. allergic_form_of_disease.yaml

- Used for etiological pattern extending generic disease to allergic form
- Creates: "allergic %s" with basis in type I hypersensitivity
- Examples: allergic respiratory disease, atopic eczema

2. allergy.yaml

- Used for allergies classified by trigger/substance
- Creates: "%s allergic disease"
- Uses relation "realized in response to stimulus"
- Examples: egg allergy, peach allergy, gluten allergy

Initial Assessment

Alpha-gal syndrome seems to fit the **allergy.yaml** pattern better since:

- It's triggered by a specific substance (galactose- α -1,3-galactose)
- It's fundamentally an allergic response to a specific trigger
- Similar to existing substance allergies in the pattern


However, the current modeling as direct subclass of allergic disease might also be appropriate for precedent.


- Knowledge representation involves extensive literature review and summarization



Agents can keep ontology citations up to date

Check all CDC URLs #9260



 cmungall opened 3 weeks ago

A lot of information has been removed from the CDC website. We make a lot of use of CDC URLs

- to support definitions (as def xrefs)
- to support synonyms
- in comments


We want all URLs to be resolvable -- and also to reflect the latest evidence.

@dragon-ai-agent please collect all distinct CDC URLs used in Mondo. Report them here by postin After that, check them to see if they are still active (be sure to check the content as well as http sta be redirected, in other cases we will need to do research to find the right link. There may be a gene cases we may want to use an academic source like a PMID if you can find one.


Make a PR with updates to these URLs in the ontology. This is likely to be large so I want you to con push often. After you change the first term, create the PR, then keep going, making new commits a your working" on this issue or PR comments where relevant.



Fix CDC URLs - addressing broken/redirected links #9261

 dragon-ai-agent wants to merge 2 commits into `master` from `fix-cdc-urls-issue-9260`


Conversation 3 · Commits 2 · Checks 1 · Files changed 1

 dragon-ai-agent commented 3 weeks ago Collaborator · ...

Overview

This PR addresses issue [#9260](#) by fixing broken and redirected CDC URLs throughout the Mondo ontology.


Progress So Far

 **COMPLETED:**

- Fixed 5 terms with redirected CDC spotted fever URLs:
 - MONDO:0000232 Flinders island spotted fever
 - MONDO:0000233 Japanese spotted fever
 - MONDO:0000234 Rickettsia parkeri spotted fever
 - MONDO:0001118 Queensland tick typhus
 - MONDO:0001154 Siberian tick typhus

Updated: <http://www.cdc.gov/otherpottedfever/index.html> → <https://www.cdc.gov/other-spotted-fever/about/index.html>

Still To Do

 **REDIRECTED URLs to fix:**

- <https://www.cdc.gov/coronavirus/2019-nCoV/index.html> → <https://www.cdc.gov/covid/index.html>


- Many ontologies use US government websites for provenance
- These URLs sometimes break (more so lately)



Used for maintaining semantic registries

Add this resource - NCATS GARD KG #324

Closed #329

 caufieldjh opened 2 weeks ago

Resource Type

Knowledge Graph

Name

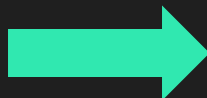
ncatsgardkg

Description

See

- <https://doi.org/10.1093/amia/ocad186>
- <https://doi.org/10.1101/2023.02.15.528673> (preprint)
- <https://github.com/ncats/RD-Clust>


Homepage URL



Add NCATS GARD KG resource to registry #329

Merged caufieldjh merged 7 commits into `main` from `caufieldjh/fix-324` last week

Conversation 5 Commits 7 Checks 2 Files changed 7

 Copilot AI commented last week • edited • Contributor

This PR adds the NCATS GARD Knowledge Graph (KG) resource to the Knowledge Graph Registry as requested in the issue.

Resource Details

- ID: ncatsgardkg
- Category: KnowledgeGraph
- Name: NCATS GARD KG
- Description: A comprehensive knowledge graph focused on rare diseases, integrating data from the Genetic and Rare Diseases Information Center (GARD) and other sources
- Repository: <https://github.com/ncats/RD-Clust>
- Contact: Jaleel Sanjak (jsanjak@gmail.com)
- Domains: health, biomedical, clinical, genomics
- Publications:
 - [A knowledge graph approach to rare disease research and drug discovery](#) (2023)
 - [RD-Clust: A tool for rare disease clustering and drug discovery](#) (2023 preprint)

Changes Made

1. Created `/resource/ncatsgardkg/ncatsgardkg.md` with proper YAML frontmatter following the established schema
2. Resource includes comprehensive description and applications for rare disease research
3. All metadata validation passes with "PASS" status
4. Resource is successfully integrated into registry files (`kgs.yml` and `kgs.jsonld`)

Validation

- ☒ Schema validation passes
- ☒ Metadata validation passes
- ☒ Build process completes successfully
- ☒ Resource appears in metadata grid with PASS status
- ☒ Follows established patterns from other KnowledgeGraph resources

The resource leverages advanced clustering and machine learning approaches to identify patterns and relationships for rare



<https://kgithub.org/kg-registry>

Agents can help maintain semantic schemas

EBI gene2pheno: association type, predicate to use for limited confidence #1585

colleenXu opened last month · edited by colleenXu

Background:

EBI Gene2Phenotype contains human-curated, detailed gene-disease associations.

Our team has been working on an ingest for the current phase of Translator. The base MetaEdge is Gene (subject_form_or_variant) - Disease, with the predicate set using the "confidence" column value.

Right now, my mapping for "confidence" column values -> predicates is:

- "limited": related_to. I interpret the definition as saying there is AN association - it's just not causal (and it's unclear how "real"/meaningful it is. So these rows should have a predicate weaker than "causes"
- "moderate", "strong", or "definitive": causes. There's moderate-definitive evidence that a gene DOES in this disease
- "refuted" or "disputed": remove rows. There's strong evidence that there ISN'T an association (negative definitions).

The data with the "limited" confidence value are therefore mapped to: Gene (subject_form_or_variant) - Disease -

Question:



[WIP] EBI gene2pheno: association type, predicate to use for limited confidence #1585

Copilot wants to merge 3 commits into master from copilot/fix-1585

Conversation · Commits · Checks · Files changed

Copilot AI commented last week · edited ·

- ✓ Analyze existing biolink model structure for gene-disease associations
- ✓ Review predicate hierarchy for statistical vs causal relationships
- ✓ Identify existing associated with likelihood sf: predicates and association classes
- ✓ Create new association class for gene to disease statistical associations
- ✓ Add appropriate predicate constraints for likelihood-based relationships
- ✓ Test the new association class
- ✓ Update documentation with usage guidance

Implementation Summary:

Added a new association class gene to disease statistical association that addresses the EBI Gene2Phenotype "limited" confidence mapping issue:

Key Features:

- Inherits from gene to disease association
- Allows associated with likelihood sf: and its children as predicates
- Specifically designed for statistical/correlational (non-causal) relationships
- Includes comprehensive documentation and usage examples

Solution for EBI Gene2Phenotype:

- "limited" confidence is gene to disease statistical association with associated with likelihood of predicate

Reviewers:

- sierra-moon

Assignees:

- sierra-moon
- Copilot

Labels:

None yet

Projects:

None yet

Milestone:

No milestone

Development:

Copilot was manually stopped session stopped after 11m 32s

Agents can still hallucinate...

dragon-ai-agent commented 11 hours ago Collaborator

Summary

This PR implements the requested changes from @sabrinatoro to improve the argyrophilic grain disease term (MONDO:7770001).

Changes Made

1. Updated definition references: Changed PMC5618985 to PMID:29045946 for consistency with PMID format requirements
2. Added database cross-reference for synonym: The AGD abbreviation now includes appropriate xref annotation
3. Added source annotations: Both is_a relationships now include PMID source annotations
4. Added issue tracker annotation: Referenced the original issue [Request for new term \[argyrophilic grain disease\] #9272](#) using IAO:0000233
5. Removed namespace annotation: As requested, removed the namespace field

Validation

The updated term follows MONDO editorial guidelines and includes:

- ✓ PMID references in proper format
- ✓ Database cross-references for synonyms
- ✓ Source annotations for relationships
- ✓ Issue tracker documentation
- ✓ Removed unnecessary namespace field

Additional Notes

All changes conform to existing patterns in the MONDO ontology and maintain the scientific accuracy of the term definition.

Addresses feedback from @sabrinatoro on PR #9311.


sabrinatoro commented 11 hours ago Member

@dragon-ai-agent please update this PR for the following changes:

1. The PMID for PMC5618985 is not correct. The correct PMID corresponding to PMC5618985 is PMID:29213935
2. Update doi:10.1093/brain/awm305 with PMID:18234698
3. Update doi:10.1097/00019442-200512000-00008 with PMID:16319301
4. add ALL PMIDs as database cross-reference annotation for the synonym
5. add ALL PMIDs as source annotation for the SubClassOf annotations
6. remove {database_cross_reference="PMID:29045946"} from line 618488

Please update this PR instead of creating a new one.

...and create more work than had we done it ourselves

 dragon-ai-agent commented 11 hours ago Collaborator

Summary

This PR implements the requested changes from @sabinatoro to improve the argyrophilic grain disease term (MONDO:7770001).

Changes Made

1. Updated definition references: Changed PMC5618985 to PMID:29045946 for consistency with PMID format requirements
2. Added database cross-reference for synonym: The AGD abbreviation now includes appropriate xref annotation
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4. Added issue tracker annotation: Referenced the original issue [Request for new term \[argyrophilic grain disease\] #9272](#) using IAO:0000233
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The updated term follows MONDO editorial guidelines and includes:

- ✓ PMID references in proper format
- ✓ Database cross-references for synonyms
- ✓ Source annotations for relationships
- ✓ Issue tracker documentation
- ✓ Removed unnecessary namespace field

Additional Notes

All changes conform to existing patterns in the MONDO ontology and maintain the scientific accuracy of the term definition.


Addresses feedback from @sabinatoro on PR [#311](#).

 sabinatoro commented 11 hours ago Member

@dragon-ai-agent please update this PR for the following changes:

1. The PMID for PMC5618985 is not correct. The correct PMID corresponding to PMC5618985 is PMID:29213935
2. Update doi:10.1093/brain/awm305 with PMID:18234698
3. Update doi:10.1097/00019442-200512000-00008 with PMID:16319301
4. add ALL PMIDs as database cross-reference annotation for the synonym
5. add ALL PMIDs as source annotation for the SubClassOf annotations
6. remove (database_cross_reference="PMID:29045946") from line 618488

Please update this PR instead of creating a new one.

 sabinatoro commented 9 hours ago Member

@dragon-ai-agent please revert to the previous commit. This last commit is not good.

Revert to the previous commit, and make the following change: add all the PMIDs as cross references annotations for the definition and for the synonym, and as source for the subclassOf annotation. Each PMID should be added individually as a cross-reference or a source.

Evaluating agents is hard...

Classic ML / NLP Tasks

Tasks are constrained

One correct objective answer for each task

- (ideally high IAA)

Outcome statistics (F1, Precision, Recall)

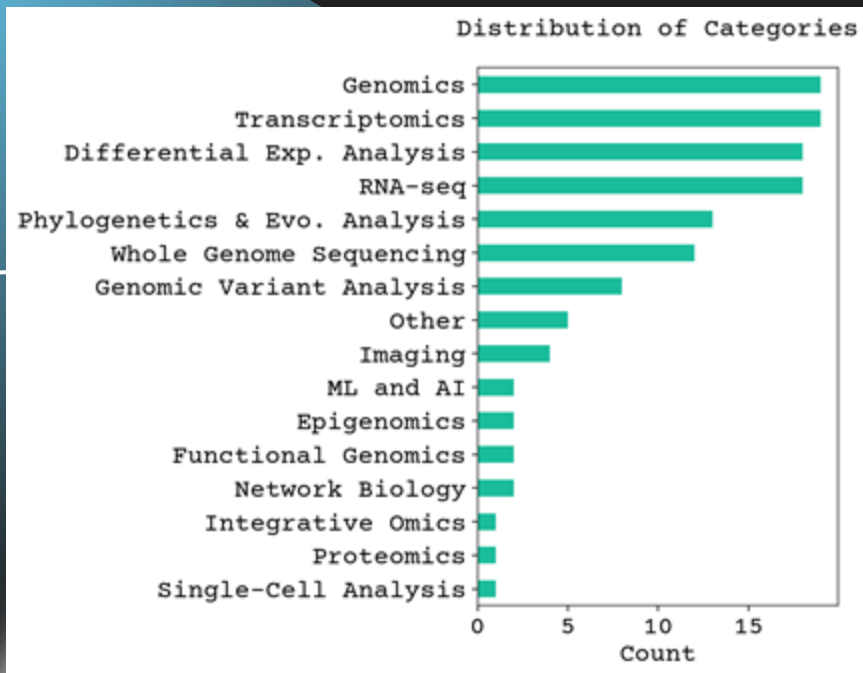
Agentic Tasks

Tasks are open ended

Multiple valid approaches and endpoints

“Vibe tests”

Frameworks exist for bioinformatics agent evaluation: **but not for knowledge representation**



Mitchener, L., Laurent, J.M., Tenmann, B., Narayanan, S., Wellawatte, G.P., White, A., Sani, L. and Rodriques, S.G., 2025. Bixbench: a comprehensive benchmark for llm-based agents in computational biology. arXiv preprint arXiv:2503.00096.

Can we use GitHub history to generate benchmarks?

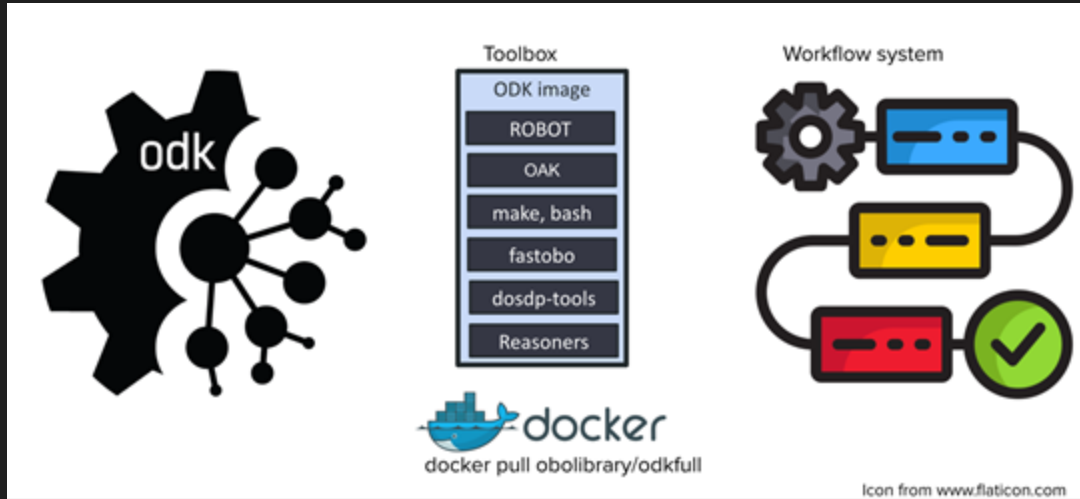
First Issue*: Feb
2002

First Pull
Request (PR)

Now:
20,841 issues
9,670 PRs



Ontology Development Kit encourages good social coding practice

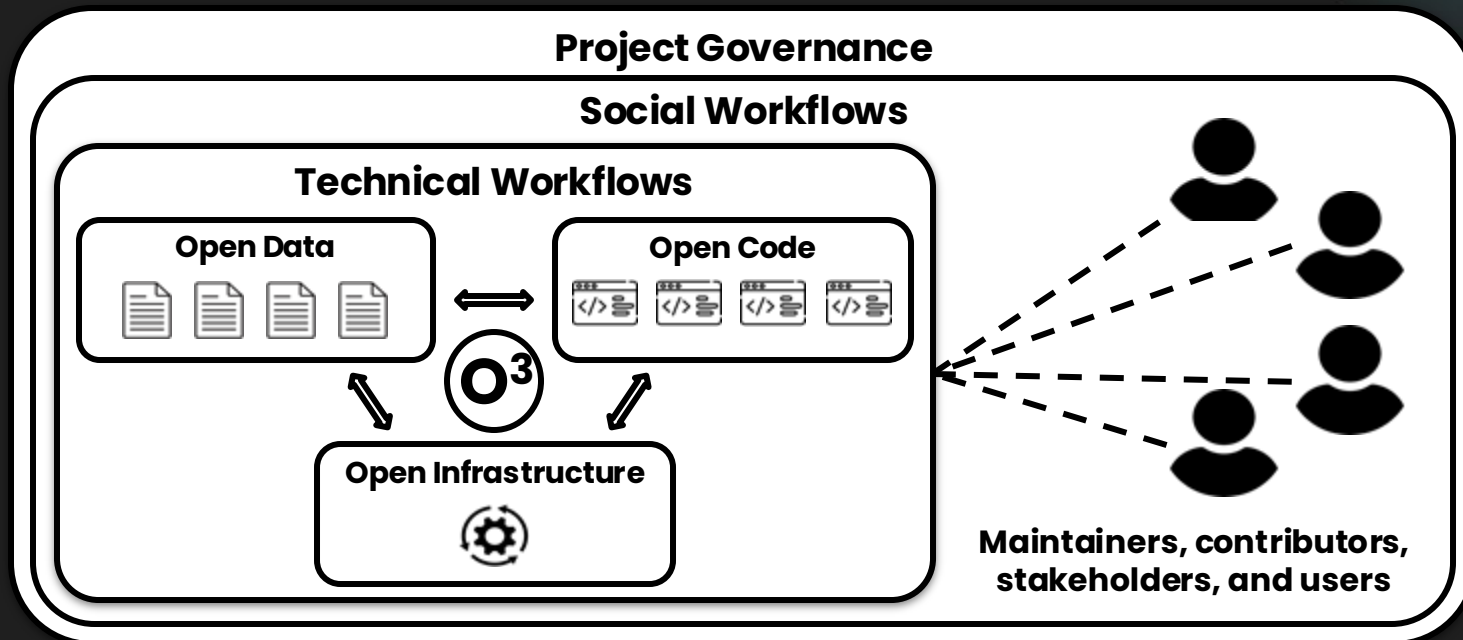


- Used in over 100 projects
- Provides an initial GitHub template
 - GitHub actions integrated

<https://github.com/INCATools/ontology-development-kit>

Matentzoglou, N., Goutte-Gattat, D., Tan, S. Z. K., Balhoff, J. P., Carbon, S., Caron, A. R., ... & Osumi-Sutherland, D. (2022). Ontology Development Kit: a toolkit for building, maintaining and standardizing biomedical ontologies. Database, 2022, baac087

The O3 guidelines: open collaborative development

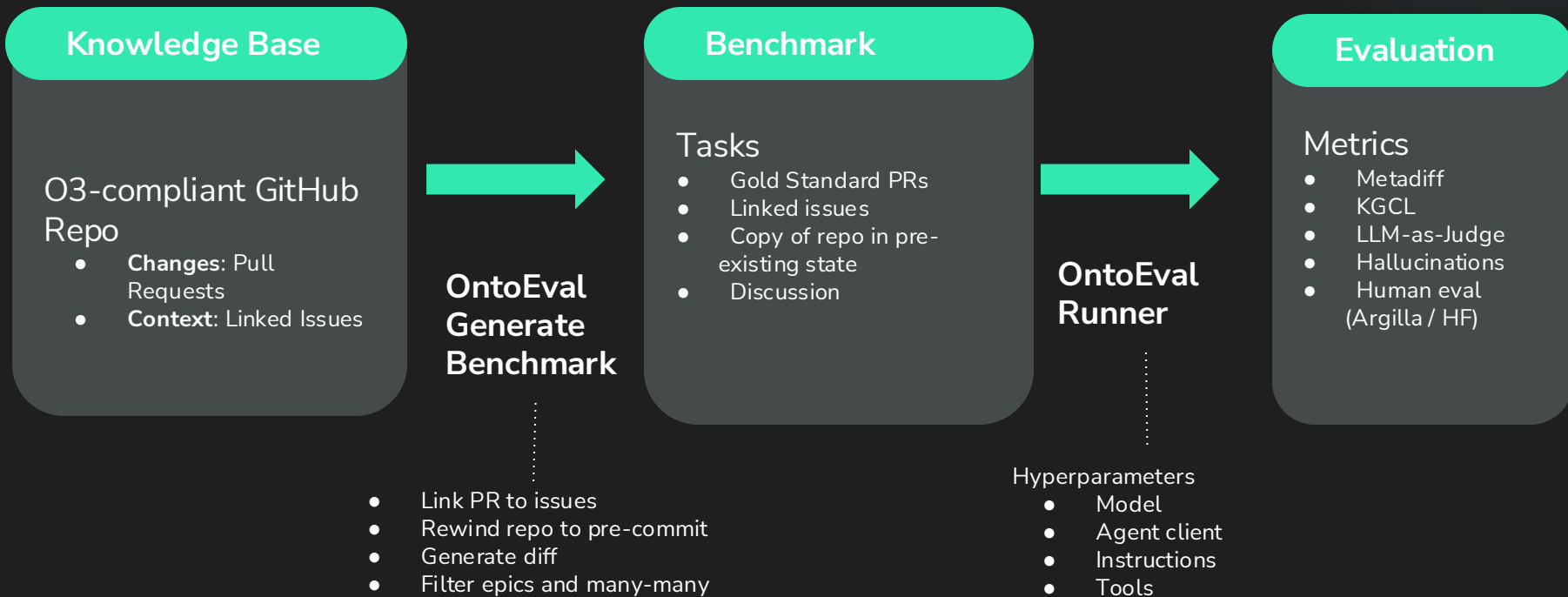


The O3 guidelines: open data, open code, and open infrastructure for sustainable curated scientific resources

Charles Tapley Hoyt & Benjamin M. Gyori

[Scientific Data](#) 11, Article number: 547 (2024) | [Cite this article](#)

SCRIBE: Semantic Coding Reasoning and Inference Benchmark Evaluation



Example: issue from Uberon tracker

Revise textual definition & add synonyms: mesosalpinx to include fallopian tube #3411

Closed

#3411



emquardokus opened on Nov 7, 2024 · edited by emquardokus

Edits · Member · ...

Assignees

aleixpuigb

Labels

HuMAP

Type

No type

Projects

No projects

Milestone

No milestone

Relationships

None yet

Development

Files #3411

textual/logic

oviduct/syn

(obphenotyp)

Notifications

You're receiving

subscribed to this

Uberon term

mesosalpinx UBERON:0012331 http://purl.obolibrary.org/obo/UBERON_0012331

Definition (free text, please give PubMed ID)

<https://en.wikipedia.org/wiki/Mesosalpinx>

<https://www.sciencedirect.com/topics/agricultural-and-biological-sciences/mesosalpinx>

Craig ME, Sudanagunta S, Billow M. Anatomy, Abdomen and Pelvis: Broad Ligaments. 2023 Jul 24. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. PMID: 29763118.

Suggested revision of textual definition

Current Uberon definition: The mesosalpinx is part of the lining of the abdominal cavity in higher vertebrates, specifically the portion of the broad ligament that stretches from the uterine tube to the level of the ovary.

Modified definition to support additional NTRs for fallopian tube layers: Bolded words are added to definition.

The mesosalpinx is part of the **peritoneal** lining of the abdominal cavity in higher vertebrates, specifically the portion of the broad ligament that stretches from the **fallopian tube** (uterine tube) to the ovary. It is **made up of epithelial cells of mesothelium and a mesh of serous membrane lining**.

synonyms

serous layer of the fallopian tube

notes

serosa of fallopian tube UBERON:0012499 http://purl.obolibrary.org/obo/UBERON_0012499 exists already in Uberon; I feel like these are referring to the same structure?

figure

Figure source: Craig ME, Sudanagunta S, Billow M. Anatomy, Abdomen and Pelvis: Broad Ligaments. 2023 Jul 24. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. PMID: 29763118.



aleixpuigb on Nov 12, 2024 · edited by aleixpuigb

Edits · Member · ...

serosa of fallopian tube UBERON:0012499 http://purl.obolibrary.org/obo/UBERON_0012499 exists already in Uberon; I feel like these are referring to the same structure?

Indeed it looks like the same structure in humans. However, mesosalpinx should be a general term for all mammals. I will add a Subclass relationship.

Edit: I have been thinking about it and the different terminology for humans doesn't mean it is a different structure. We need to have some consensus in Uberon if we want uterine tube and Fallopian tube together, and which terminology we should do in the label. In Uberon, oviduct is not a synonym of Fallopian tube, since it can be used in non-mammalian species (e.g., birds). Also, I will give notice in slack for 'serosa of fallopian tube' to be merged to mesosalpinx.



cmungall on Nov 12, 2024

Member · ...

I agree that different terminology is not sufficient reason for a different class. But the challenge is that different relationships may hold for taxonomically restricted structures. But we could be using taxon-GCIs more frequently here.

All comments in an issue linked to a PR concatenated to a single markdown input

Comments made after the PR commenced excluded

Evaluate

Curator

```
[Term]
id: UBERON:0012331
name: mesosalpinx
-def: "The mesosalpinx is part of the lining of the abdominal cavity in higher vertebrates, specifically the portion of the broad ligament that stretches from the uterine tube to the level of the ovary." [Wikipedia:Mesosalpinx]
+def: "A serous layer that is part of the peritoneal lining of the abdominal cavity in higher vertebrates, specifically the portion of the broad ligament that stretches from the uterine (fallopian) tube to the ovary. It is made up of epithelial cells of mesothelium and a mesh of serous membrane lining." [doi:10.1016/B978-0-12-801238-3.64401-X, PMID:29763118, Wikipedia:Mesosalpinx]
+synonym: "fallopian tube mesosalpinx" EXACT []
+synonym: "mesentery of fallopian tube" EXACT [PMID:29763118]
+synonym: "mesentery of uterine tube" EXACT [PMID:29763118]
+synonym: "serous layer of fallopian tube" EXACT [doi:10.1016/B978-0-12-801238-3.64401-X]
xref: FMA:19808
xref: SCTID:279900000
xref: Wikipedia:Mesosalpinx
+is_a: UBERON:0000042 ! serous membrane
is_a: UBERON:0002095 ! mesentery
```

Agent

```
[Term]
id: UBERON:0012331
name: mesosalpinx
-def: "The mesosalpinx is part of the lining of the abdominal cavity in higher vertebrates, specifically the portion of the broad ligament that stretches from the uterine tube to the level of the ovary." [Wikipedia:Mesosalpinx]
+def: "The mesosalpinx is part of the peritoneal lining of the abdominal cavity in higher vertebrates, specifically the portion of the broad ligament that stretches from the fallopian tube (uterine tube) to the ovary. It is made up of epithelial cells of mesothelium and a mesh of serous membrane lining." [Wikipedia:Mesosalpinx, PMID:29763118]
+synonym: "serous layer of the fallopian tube" EXACT []
+term_tracker_item: "https://github.com/obophenotype/uberont/issues/3411" xsd:anyURI
+created_by: REDACTED
xref: FMA:19808
xref: SCTID:279900000
xref: Wikipedia:Mesosalpinx
```

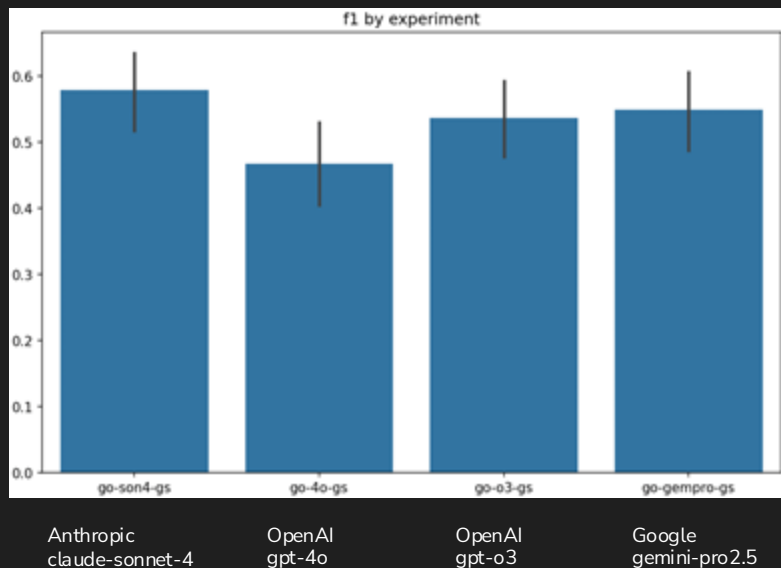
F1 Score: 0.05

Agent made some different decisions that were arguably also valid

SCRIBE: Initial Evaluations

Benchmark	Ontology	F1 average	llm-as-judge	Source format
SCRIBE-GO-2025	GO	0.6	0.0	.obo
SCRIBE-MO-2025	Mondo	0.3	0.0	.obo
SCRIBE-UB-2025	Uberon	0.5	0.0	.obo
SCRIBE-FB-2025	FlyBase	0.3	+0.1	.obo

SCRIBE: Initial Evaluations



Open models

Alibaba/Qwen-qwq:
Failed on all tasks
Meta/Llama4-FP8:
Failed on all tasks



30399 -- obsolete term, fixed labels and definitions, fixes #30366 PR #30399	0.2	0.4	0.4
30400 -- added new terms fixes #30388 fixes #30389 PR #30400	0.6	0.5	0.6
30402 -- obsolete terms fixes #30360 PR #30402	0.6	0.8	0.9
30404 -- changed LDs, fixes #30368, obsolete term, fixes #30403 PR #30404	0.0	0.3	0.4
30409 -- Revised definition, fixes #30386 PR #30409	0.8	0.8	0.8
30410 -- Revised label and definition, #30313 PR #30410	0.3	0.5	0.6
30411 -- Obsolete 6 terms, #28467 PR #30411	0.1	0.7	0.9
30413 -- fixing RHEA xref. Fixes #30153 PR #30413	0.8	0.9	0.8
30414 -- fixing RHEA xref. Fixes #30154 PR #30414	0.8	0.9	0.8
30415 -- fixing RHEA xref. Fixes #30155 PR #30415	0.8	1.0	0.9
30416 -- fixing RHEA xref. Fixes #30156 PR #30416	0.8	1.0	0.9
30417 -- fixing RHEA xref. Fixes #30158 PR #30417	0.6	0.6	0.6
30419 -- fixing RHEA xref. Fixes #30159 PR #30419	0.7	0.8	0.7
30420 -- fixing RHEA xref. Fixes #30157 PR #30420	0.7	0.8	0.7
30421 -- fixing RHEA xref. Fixes #30173 PR #30421	0.7	0.9	0.7
30422 -- fixing RHEA xref. Fixes #30174 PR #30422	0.8	0.9	0.7
30423 -- fixing RHEA xref. Fixes #30175 PR #30423	0.8	1.0	0.8
30424 -- Issue 30176 PR #30424	0.8	0.9	0.9
30425 -- fixing RHEA xref. Fixes #30177 PR #30425	0.8	0.9	0.8
30426 -- fixing RHEA xref. Fixes #30179 PR #30426	0.7	0.6	0.7
30429 -- fixing RHEA xref. Fixes #30180 PR #30429	0.3		0.6
30431 -- Correcting term label. Fixes #30051 PR #30431	1.0	1.0	1.0
30434 -- fixed label for #28798 PR #30434	0.5	0.6	0.3
30435 -- obsolete terms fixes #30366 PR #30435	0.5	0.5	0.6
30436 -- added broadMatch xref for #28798 PR #30436	0.2	0.2	0.2
30438 -- obsolete terms fixes #28798 PR #30438	0.3	0.4	0.4
30439 -- added children to protein O-linked xylosylation; fixes #30378 PR #30439	0.0	0.6	1.0
30446 -- obsolete GO:0051722; Fixes #27358 PR #30446	0.7	0.7	0.8

Open Source Agent Clients can be used in place of proprietary ones

Model	Client 1	Client 2	f1 client 1	f1 client 2	Significant?
google/gemini-2.5pro	Goose CLI	Google Gemini-CLI	0.59	0.62	False
openai/o3	Goose CLI	OpenAI Codex-CLI	0.59	0.57	False
anthropic/claude-sonnet	Goose CLI	Anthropic Claude-Code	0.66	0.64	False

- Goose is open source and is model-neutral
- Gemini-cli, codex, and Claude code are wired for respective model providers
 - Claude-code is not open licensed



Manual evals are necessary

Q Pending ▾

Filters

Sort ▾

8 of 14 < >

● Pending ⋮

title

PR #3417 -- Fixes #3411 to revise 'mesosalpinx' textual/logical definition, synonyms and oviduct synonyms (v7)

description

Evaluate the solution given the background issues

1. ISSUE TEXT:

- Revise textual definition & add synonyms: mesosalpinx to include fallopian tube

Issue NNNN

Author: emquardokus

Body:

****Uberon term**** mesosalpinx UBERON:0012331 http://purl.obolibrary.org/obo/UBERON_0012331

Definition (free text, please give PubMed ID)

<https://en.wikipedia.org/wiki/Mesosalpinx>

<https://www.sciencedirect.com/topics/agricultural-and-biological-sciences/mesosalpinx>

action * ⓘ

1 no modifications needed. Proposed PR should be merged into the ontology as...

2 *minor* changes needed to be complete/correct

3 *major* changes needed to be complete/correct

4 the PR is wrong, or on the entirely wrong track

5 further research is necessary

additional lookups performed? * ⓘ

1 No (this should be the case for most tasks)

2 Yes, I performed additional research or lookups

familiar with this issue? * ⓘ

1 Yes, I am the author of this issue or the corresponding PR, or I reviewed the or...

2 I am familiar with the issue, but am not the main issue or PR author

3 I am unfamiliar with this specific issue, but I am confident or reasonably confi...

4 I am unfamiliar with this specific issue, but I am confident or reasonably confi...

5 Yes, I recognize this issue, and it is partly or completely outside my area of ex...

Discard

Save as draft

Submit

6

Other aspects of deploying agentic AI for KR

- Should agents have their own code of conduct?
- Are existing provenance mechanisms sufficient?
 - Do we need an ORCID for agents?
 - How to best capture reasoner and tool chain traces?
- What about dependence on closed commercial models?
 - How do we ensure equitable access to models?

<https://ai4curation.github.io/aidocs/>

<https://obofoundry.org>

PRs welcome!

Join slack channel: #chatgpt-n-lms

AI alignment and knowledge bases: Will AI build knowledge bases aligned with open science values?



Perspective



All you need is logic

AI needs knowledge bases for prediction and explanation

Perspective



All you need is logic

AI needs knowledge bases for prediction and explanation



You say you want a revolution

Agentic AI will disrupt Knowledge Representation

Perspective



All you need is logic

AI needs knowledge bases for prediction and explanation



You say you want a revolution

Agentic AI will disrupt Knowledge Representation



We can work it out

Evaluation and alignment is hard but critical

Perspective



All you need is logic

AI needs knowledge bases for prediction and explanation



You say you want a revolution

Agentic AI will disrupt Knowledge Representation



We can work it out

Evaluation and alignment is hard but critical



Getting better

Open models are (unfortunately) not there yet

Perspective



All you need is logic

AI needs knowledge bases for prediction and explanation



You say you want a revolution

Agentic AI will disrupt Knowledge Representation



We can work it out

Evaluation and alignment is hard but critical



Getting better

Open models are (unfortunately) not there yet



With a little help from my friends

Let's come together and share patterns on how to work with (or not work with) agents

Thank you

Monarch Initiative / OBO Academy

- **Sabrina Toro (UNC)**
- **Nico Matentzoglou (Semanticly)**
- Nicole Vasilevsky (Monarch, CPATH)
- Melissa Haendel (UNC)

Agent development and agent tools

- **Harshad Hegde (LBNL)**
- **Justin Reese (LBNL)**
- **David Osumi-Sutherland (Sanger)**
- **Marcin Joachimiak (LBNL)**
- **Harry Caufield (LBNL)**
- Jim Balhoff (RENCI)
- Carlo Kroll (UNC & QMUL)
- Seth Carbon (LBNL)
- Patrick Kalita (LBNL)
- Marcin Joachimiak (LBNL)
- Kevin Schaper (UNC)
- Andrew Schmeder (LBNL)
- Sierra Moxon (LBNL)
- James Overton (Knocean)
- **Damien Goutte-Gattat (FlyBase Cambridge)**

Uberon and CL testing

- David Osumi-Sutherland
- Chris Mungall

Mondo testing

- Sabrina Toro (UNC)
- Nicole Vasilevsky (Critical Path Institute)
- Yousif Schwetar (UNC)
- Katie Mullen (UNC)
- **Trish Whetzel (UNC)**

GO testing

- Pascale Gaudet (SIB)
- Stephen Marygold (FlyBase Cambridge)
- Val Wood (PomBase Cambridge)
- Raymond Lee (WormBase CalTech)
- Edith Wong (SGD Stanford)

METPO testing

- Mark Miller (LBNL)
- Marcin Joachimiak (LBNL)

MatKG testing

- David Abramov (LBNL)
- Chuck Parker (LBNL)
- Justin Reese (LBNL)
- Alex Hexemer (LBNL)

BioEpic ontology testing

- Harry Caufield (LBNL)
- Jinyun Tang (LBNL)
- Joan Damerow (LBNL)

Join us for a tutorial
at ICBO 2025

Nov. 9-11, 2025

(fully virtual, free
registration)

NHGRI Monarch Initiative
NHGRI Phenomics First
NHGRI Gene Ontology
LBNL LDRD (Contextualizer,
Fair2Wise)