

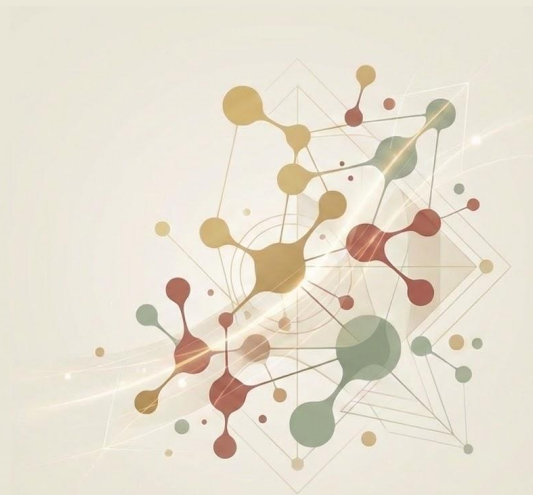
Dismech: a Disease Mechanisms Knowledge Base Built For and With Agentic AI

**Harry Caufield
Lawrence Berkeley National Laboratory
February 13 2026**



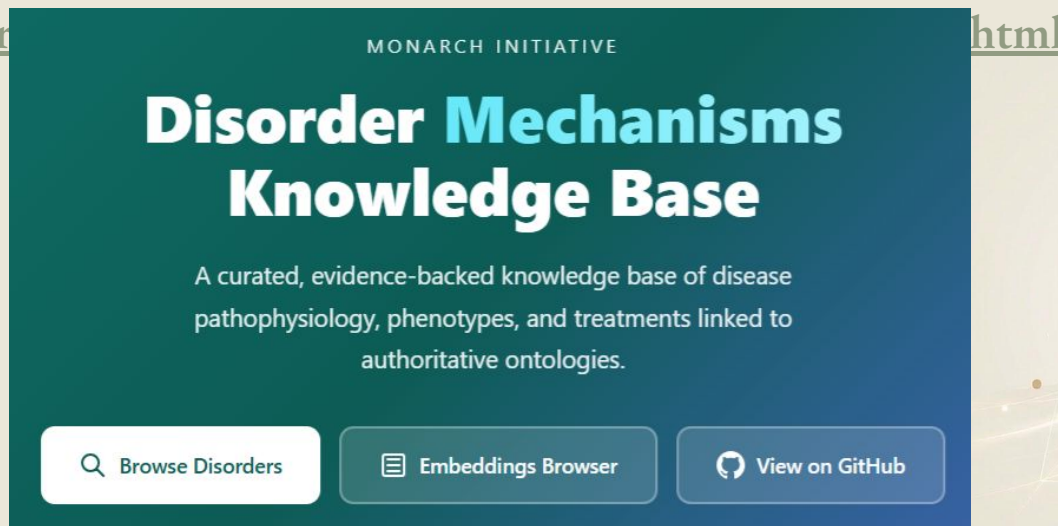
What is Dismech?

- A Disorder Mechanisms Knowledge Base containing structured evidence from scientific literature.
- A proof-of-concept for how agentic AI can support data curation.
- A growing set of examples of how ontologies support reusability of structured data
- A resource built by members of the Monarch Initiative
 - Anyone can curate a new page!



Where is Dismech?


- GitHub: <https://github.com/monarch-initiative/dismech>
- Main site: <https://monarch-initiative.github.io/dismech/>
- Embedding Browser: <https://monarch-initiative.github.io/dismech/app/embeddings/index.html>
- QC Dashboard:
<https://monarch-initiative.github.io/dismech/app/quality-control/index.html>





MONARCH INITIATIVE

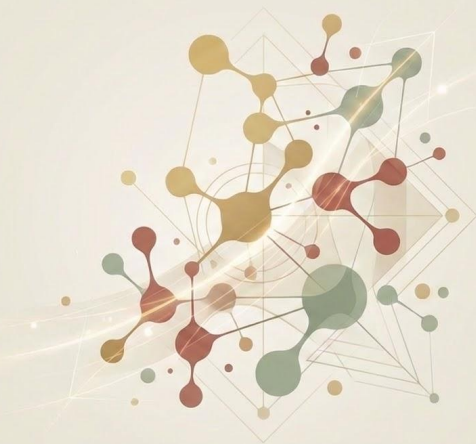
Disorder Mechanisms Knowledge Base

A curated, evidence-backed knowledge base of disease pathophysiology, phenotypes, and treatments linked to authoritative ontologies.

 Browse Disorders

 Embeddings Browser

 View on GitHub



What can I do with Dismech?

Disorder Mechanisms Browser

Explore disease pathophysiology, phenotypes, and mechanisms from the dismech knowledge base

Search disorders, phenotypes, cell types, genes...

CATEGORY

- ☐ Complex 75
- ☐ Genetic 47
- ☐ Mendelian 40
- ☐ Infectious Disease 31
- ☐ Autoimmune 21
- ☐ Infectious 8
- ☐ Cancer 5
- ☐ Neurological Disorder 4

Show 24 more...

DISEASE CLASS

- ☐ Autoimmune Disease 33

496 disorders

Clear filters

22q11.2 Deletion Syndrome

MONDO:0018923

Genetic

22q11.2 deletion syndrome is a chromosomal microdeletion disorder caused by hemizygous deletion of 1.5-3 Mb at chromosome 22q11.2. It is the most common microdeletion syndrome, occurring in approximately 1:4000 live births. The phenotype is highly va...

PATHOPHYSIOLOGY

TBX1 haploinsufficiency and pharyngeal arch development Cardiac neural crest migration defect Thymic hypoplasia and T-cell immunodeficiency
Parathyroid hypoplasia and hypocalcemia

PHENOTYPES

Conotruncal heart defect Hypocalcemia Thymic hypoplasia Velopharyngeal insufficiency Cleft palate +4 more

CELL TYPES

neural crest cell pharyngeal arch mesenchyme cell migratory cardiac neural crest cell cortical thymic epithelial cell medullary thymic epithelial cell +1 more

ASSOCIATED GENES



What can I do with Dismech?

Fanconi_Anemia

MONDO:0019391

Genetic

Fanconi anemia is a rare inherited bone marrow failure syndrome characterized by genomic instability, congenital abnormalities, progressive bone marrow failure, and increased susceptibility to malignancies. Most cases result from biallelic pathogenic...

PATHOPHYSIOLOGY

DNA Repair DeficiencyBone Marrow FailureGenomic InstabilityHematopoietic Stem Cell AttritionTranslesion Synthesis Defect+ 14 more

PHENOTYPES

PancytopeniaIncreased Risk of LeukemiaAplastic AnemiaThrombocytopeniaLeukopenia+ 87 more

CELL TYPES

hematopoietic stem cellepithelial cell

ASSOCIATED GENES

FANCAFANCBFANCCFANCEFANCF+ 20 more

TREATMENTS

Hematopoietic Stem Cell Transplantation (HSCT)Androgen TherapyGrowth Hormone TherapySupportive CareCancer Surveillance+ 1 more

ENVIRONMENTAL FACTORS

Protections from DNA-damaging Agents

19 mechanisms**92** phenotypes**25** genes**6** treatments

[View source YAML](#)

What can I do with Dismech?

Fanconi_Anemia

Genetic

MONDO:0019391

Bone Marrow Failure

Congenital Disorder

Fanconi anemia is a rare inherited bone marrow failure syndrome characterized by genomic instability, congenital abnormalities, progressive bone marrow failure, and increased susceptibility to malignancies. Most cases result from biallelic pathogenic variants in genes encoding components of the Fanconi anemia/BRCA DNA interstrand crosslink repair pathway, with recognized exceptions including X-linked FANCB and autosomal dominant RAD51-associated disease (FA-R). Between 21 and 22 complementation groups are cited depending on nomenclature and curation conventions, with FANCA accounting for 60-70% of cases. The phenotypic spectrum is highly variable even within the same complementation group, reflecting genetic modifiers and environmental factors.

0

MAPPINGS

1

DEFINITIONS

2

INHERITANCE

19

MECHANISMS

0

HISTOPATHOLOGY

92

PHENOTYPES

25

GENES

6

TREATMENTS

24

SUBTYPES

0

DIFFERENTIALS

0

DATASETS

3

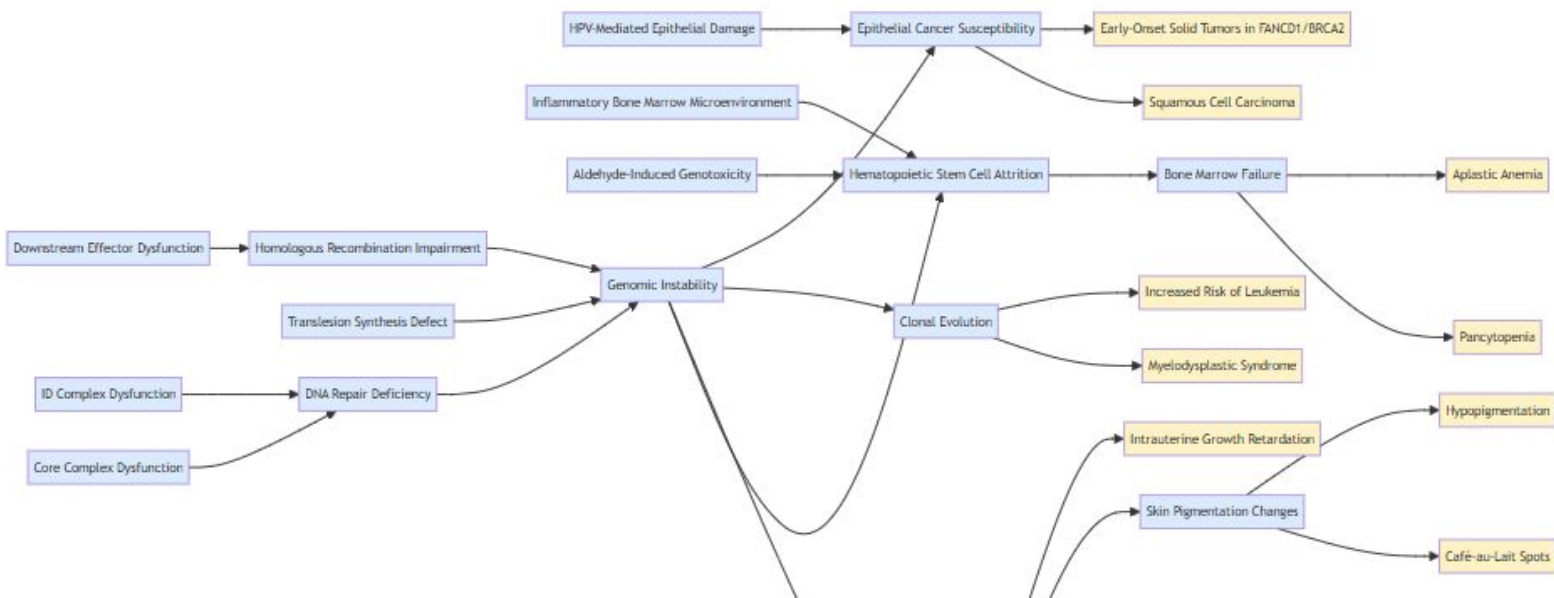
TRIALS



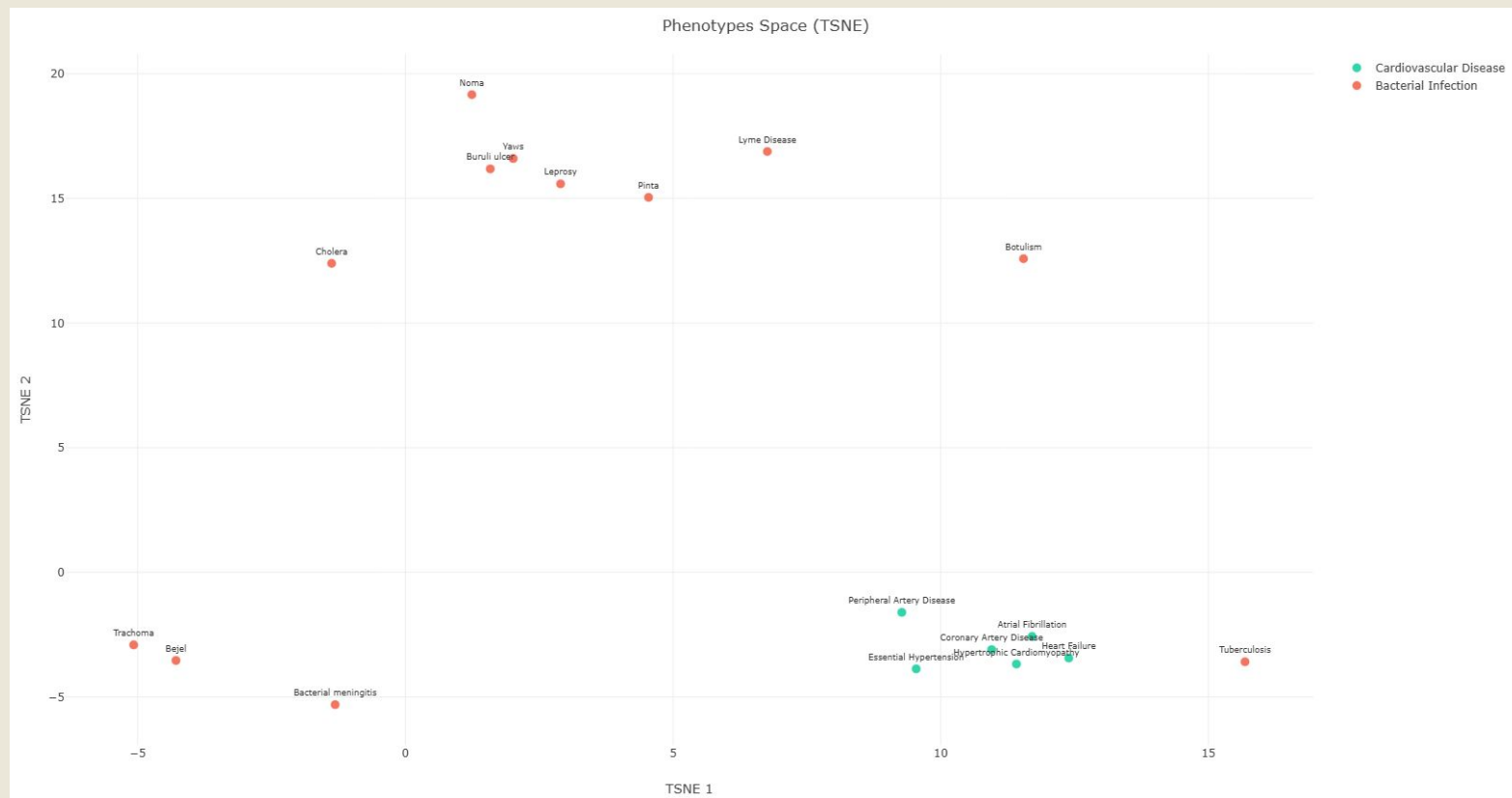
What can I do with Dismech?



Causal Graph



What can I do with Dismech?



What can I do with Dismech?

QC Dashboard - Multi-File Comparison

497 files analyzed (sorted by compliance, lowest first)

FILES ANALYZED

497

AVERAGE COMPLIANCE

68.9%

TOTAL CHECKS

12138/17620

TOTAL VIOLATIONS

207

A bot uses the QC reporting to open PRs for incremental fixes on GitHub.



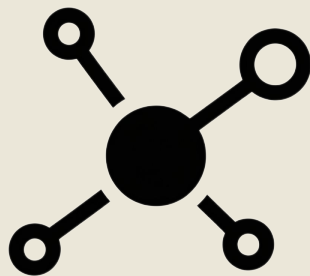
How is Dismech assembled?

Foundational elements:



LinkML

(data models and tools, including reference-validator, term-validator...) interface for ontologies -
Ontology Access Kit, OAK (common Python interface for ontologies -
OntoGPT uses this too)



An existing network of ontologies from OBO Foundry (term grounding, inheritance, and subsetting)



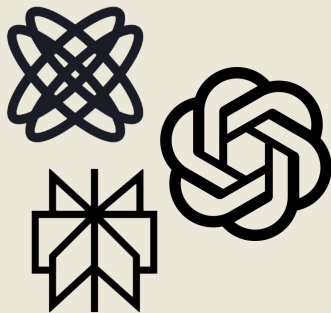
Citation entries from PubMed (source text for evidence)

How is Dismech assembled?

Agentic and automated elements:



Claude and
Claude Code
(agentic framework
and skills)



Deep research agents
including Edison,
OpenAI, and Perplexity
(populating the initial
evidence - retrieved
with Deep Research
Client)

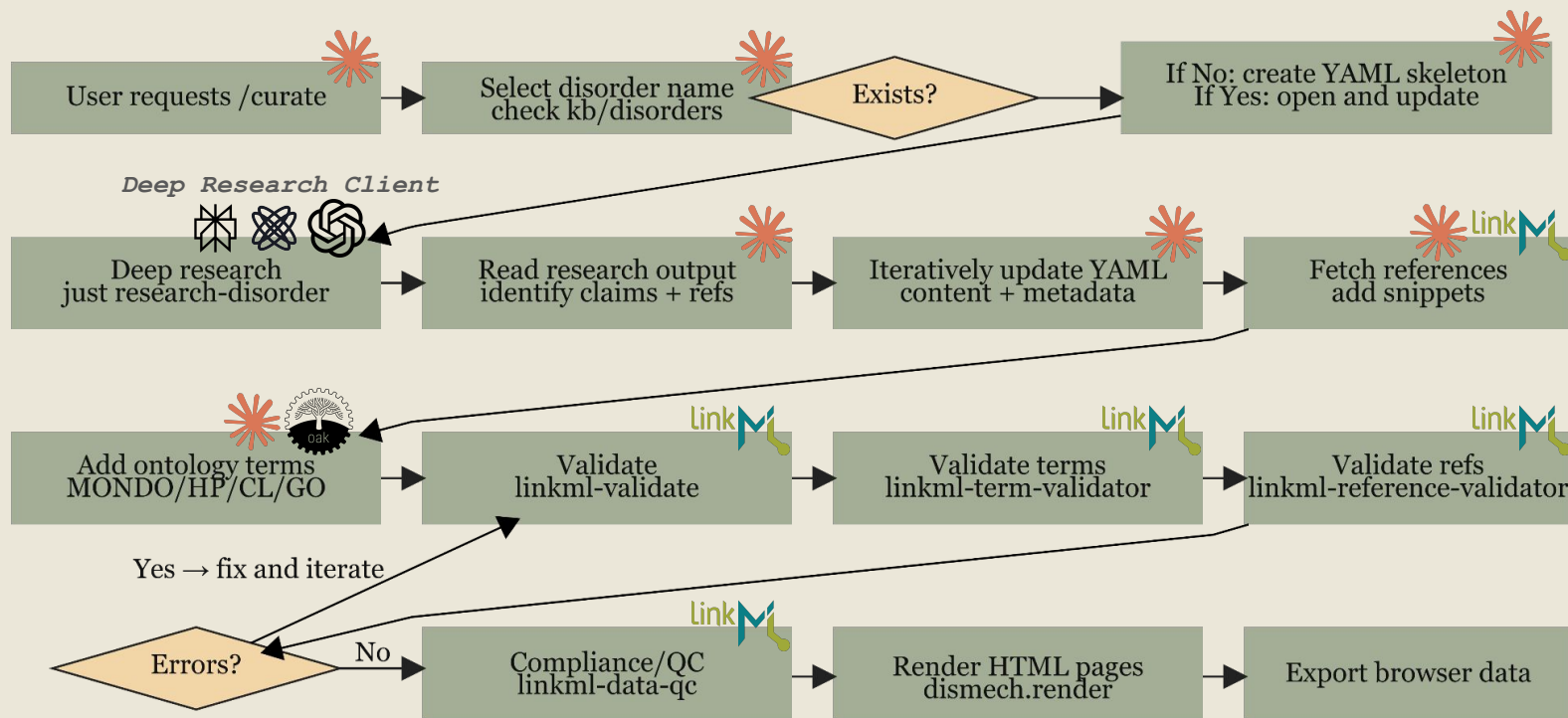


DRAGON-AI Agent
(ontology-aware agent
capable of working
across repositories)



GitHub
Actions
(coordinating agents,
triaging issues, making
regular fixes)

How is Dismech assembled?



How does Dismech use GitHub Actions?

Multiple actions run

- Triggered manually
- Weekly/cron
- In response to manual triggering (@dragon-ai-agent)
- In response to PR

Kinds of actions

- **Deterministic**
 - Standard QC on PRs (includes anti-hallucination checks)
- **AI**
 - Reviews, targeted edits



Example: weekly compliance

Weekly Compliance
[weekly-compliance.yaml](#)

Filter workflow runs

13 workflow runs

Event Status Branch Actor

This workflow has a workflow_dispatch event trigger. [Run workflow](#)

✓ Weekly Compliance Weekly Compliance #13: Manually run by cmungall	main	Today at 2:31 PM 17m 8s	...
✗ Weekly Compliance Weekly Compliance #12: Scheduled	main	Feb 6, 12:53 AM PST 43s	...
✓ Weekly Compliance Weekly Compliance #11: Manually run by cmungall	main	Feb 3, 10:18 AM PST 10m 28s	...
✗ Weekly Compliance Weekly Compliance #10: Scheduled	main	Jan 30, 12:50 AM PST 36s	...
✓ Weekly Compliance Weekly Compliance #9: Manually run by dragon-ai-agent	main	Jan 27, 10:24 PM PST 23m 54s	...
✓ Weekly Compliance Weekly Compliance #8: Manually run by dragon-ai-agent	main	Jan 26, 6:34 PM PST 20m 58s	...
✓ Weekly Compliance Weekly Compliance #7: Manually run by dragon-ai-agent	main	Jan 23, 3:11 PM PST 15m 8s	...

Manual
trigger



Example: weekly compliance

Weekly Compliance
weekly-compliance.yaml

13 workflow runs

This workflow has a workflow_dispatch event

✓ Weekly Compliance
Weekly Compliance #13: Manually run by cmur

✗ Weekly Compliance
Weekly Compliance #12: Scheduled

✓ Weekly Compliance
Weekly Compliance #11: Manually run by cmur

✗ Weekly Compliance
Weekly Compliance #10: Scheduled

✓ Weekly Compliance
Weekly Compliance #9: Manually run by drago

✓ Weekly Compliance
Weekly Compliance #8: Manually run by drago

✓ Weekly Compliance
Weekly Compliance #7: Manually run by drago

Event ▾ Status ▾ Branch ▾ Actor ▾

Run workflow ▾

Use workflow from
Branch: main ▾

Number of poorly performing files to fix
5

A narrative description of areas to focus on. Could be data model based (description, terms); type of assertion (prevalence; treatment); or disease (cancer, genetic)

ALL

Claude model to use
claude-opus-4-5-20251101

Run workflow

Jan 23, 3:11 PM PST

Filter workflow runs

Event ▾ Status ▾ Branch ▾ Actor ▾

Run workflow ▾

Today at 2:31 PM
17m 8s

Feb 6, 12:53 AM PST
43s

Feb 3, 10:18 AM PST
10m 28s

Jan 30, 12:50 AM PST
36s

Jan 27, 10:24 PM PST
23m 54s

Jan 26, 6:34 PM PST
20m 58s

Jan 23, 3:11 PM PST
15m 8s

Manual trigger



Example: weekly compliance

The screenshot displays a workflow management interface for a project named "Weekly Compliance". The interface is divided into three main sections: a list of workflow runs on the left, a central configuration panel, and a detailed view of a specific workflow run on the right.

Workflow Runs List (Left): This section shows a list of 13 workflow runs. Each entry includes a status icon (green checkmark for success, red X for failure), the name "Weekly Compliance", and a description of the run. For example, "Weekly Compliance #13: Manually run by cmungall" is marked as successful, while "Weekly Compliance #12: Scheduled" is marked as failed.

Central Configuration Panel: This panel is used to configure the workflow. It includes a "Run workflow" button at the top right. Below it, there is a section titled "Use workflow from" with a dropdown menu set to "Branch: main". A text input field labeled "Number of poorly performing files to fix" contains the value "5". Below this is a text area for a "Narrative description of areas to focus on. Could be data model based (description, terms); type of assertion (prevalence; treatment); or disease (cancer, genetic)". The text "anatomical locations using uberon" is entered in this field and is highlighted with a blue border. At the bottom, there is a section for "Claude model to" with a dropdown menu set to "claude-opus-4-5" and a "Run workflow" button.

Workflow Run Details (Right): This section shows the details of a specific workflow run. It includes a "Run workflow" button at the top right. Below it, there is a list of workflow runs with columns for "Status", "Branch", and "Actor". The runs are listed in chronological order, with the most recent run at the top. The most recent run is "Today at 2:31 PM" with a status of "17m 8s".

Manual Trigger: A red arrow points to the "Run workflow" button in the right-hand section, labeled "Manual trigger".

5 minutes later...

Filters

is:pr is:open


Labels


Milestones


CS


☐ 23 Open ✓ 227 Closed


Author ▾ Label ▾ Projects ▾ Milestones ▾ Reviews ▾ As


☐  **Add comprehensive cystic fibrosis datasets and models research**


#303 opened 47 minutes ago by github-actions  5 tasks done


☐  **Fix for Unknown records (from history files) shown in the browser and QC dashboard** ✓


#301 opened 2 hours ago by caufieldjh • Approved 


☐  🗓️ **Weekly Compliance Fix: Add UBERON anatomical locations to IgA Nephropathy (2026-02-11)** ✗


#300 opened 3 hours ago by claude  2 tasks done


☐  🗓️ **Weekly Compliance Fix: Add UBERON anatomical locations to Chronic Kidney Disease (2026-02-11)** ✗


#299 opened 3 hours ago by claude  2 tasks done


☐  🗓️ **Weekly Compliance Fix: Add UBERON anatomical locations to Glaucoma (2026-02-11)** ✗


#298 opened 3 hours ago by claude  2 tasks done

☐  🗓️ **Weekly Compliance Fix: Add UBERON anatomical locations to Liver Cirrhosis (2026-02-11)** ✗

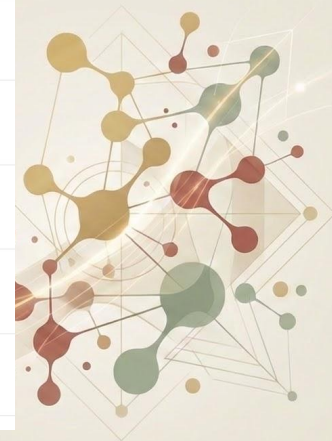
#297 opened 3 hours ago by claude  2 tasks done

☐  🗓️ **Weekly Compliance Fix: Add UBERON anatomical locations to GERD (2026-02-11)** ✗

#296 opened 3 hours ago by claude  2 tasks done

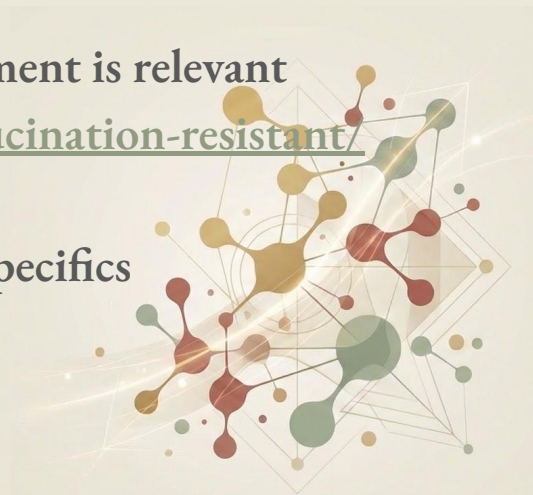
☐  **Add arsenic poisoning** ✓

#293 opened 4 hours ago by caufieldjh • Changes requested



What have we learned from Dismech?

- Agents can support consistent curation of a knowledge resource
- Agents can be very confident about their mistakes
- Skills are very helpful in guiding agent activities
 - But must be explicit about how to use external tools and services
- Exact text evidence is key
 - But of variable value in providing users with why a statement is relevant
 - See <https://ai4curation.io/aidocs/how-tos/make-ids-hallucination-resistant/>
- Humans drive the agent
 - Provide high-level guidance so the agent can handle the specifics
 - Give feedback to agents with GitHub comments



Objective: full provenance for every assertion

Phenotypes

92

Blood (6) Cardiovascular (7) Digestive (8) Ear (5) Endocrine (6) Eye (6) Genitourinary (13) Head and Neck (10) Integument (4) Limbs (9) Metabolism (1) Musculoskeletal (8) Nervous System (5) Growth (2) Other (2) Expand all

▼ BLOOD 6

Pancytopenia VERY_FREQUENT Pancytopenia (HP:0001876)

Decreased levels of red blood cells, white blood cells, and platelets. Over 80% of FA patients develop bone marrow failure. Typically presents in the first decade, often initially with thrombocytopenia or leukopenia. Notably, patients with FANCD1/BRCA2 mutations may not develop BMF. FA-S (biallelic BRCA1) patients also characteristically lack bone marrow failure.

SHOW EVIDENCE (1 REFERENCE)

PMID:35596788 SUPPORT HUMAN CLINICAL

"Fanconi anemia is a genetic disorder that is characterized by bone marrow failure, as well as a predisposition to malignancies including leukemia and squamous cell carcinoma (SCC)."

CONTEXT-SPECIFIC ANNOTATIONS (4)

VERY_FREQUENT

Over 80% of FA patients develop bone marrow failure.

SHOW EVIDENCE (1 REFERENCE)

GENETIC FA-D1 FANCD1/BRCA2 EXCLUDED

Neither FANCD1/BRCA2 patient in the Israeli cohort developed BMF. One was transplanted for AML before age 6 months; the other had no complications by age 17.

SHOW EVIDENCE (1 REFERENCE)

GENETIC FA-S BRCA1 HOMOZYGOUS EXCLUDED

FA-S (biallelic BRCA1) patients characteristically lack bone marrow failure, distinguishing them from most other FA complementation groups.

SHOW EVIDENCE (1 REFERENCE)

Objective: full provenance for every assertion

- category: Hematologic

name: Increased Risk of Leukemia

description: Significantly elevated risk of developing leukemia, predominantly acute myeloid leukemia (AML) with an incidence of 13% by age 50. Acute lymphoblastic leukemia (ALL) also reported, especially in FANCD1/BRCA2 patients.

frequency: FREQUENT

notes: The incidence of AML is 13% by age 50 years. Patients with FANCA mutations develop cancer at a significantly older age (mean 18.5 years) compared to non-FANCA groups (mean 5.2 years, $P=0.001$). Patients with FANCC mutations show a trend toward more MDS. FANCG patients may have less MDS and cancer overall. All FANCA patients in the Israeli cohort developed their first cancer after age 10, while all non-FANCA patients developed cancer by age 10.

evidence:

- reference: PMID:24037726

supports: SUPPORT

evidence_source: HUMAN_CLINICAL

snippet: "It is characterized by phenotypes including progressive bone marrow failure (BMF), developmental abnormalities, and increased occurrence of leukemia and cancer."

explanation: Clinical description confirming that increased leukemia occurrence is a defining phenotype of FA.

92

Phenotypes

Blood (6) Cardiovascular (7) Digestive (8) Ear (5) Endocrine (6) Eye (6) Genitourinary (13) Head and Neck (10) Integument (4) Limbs (9) Metabolism (1) Musculoskeletal (8) Nervous System (5) Growth (2) Other (2) Expand all

▼ BLOOD 6

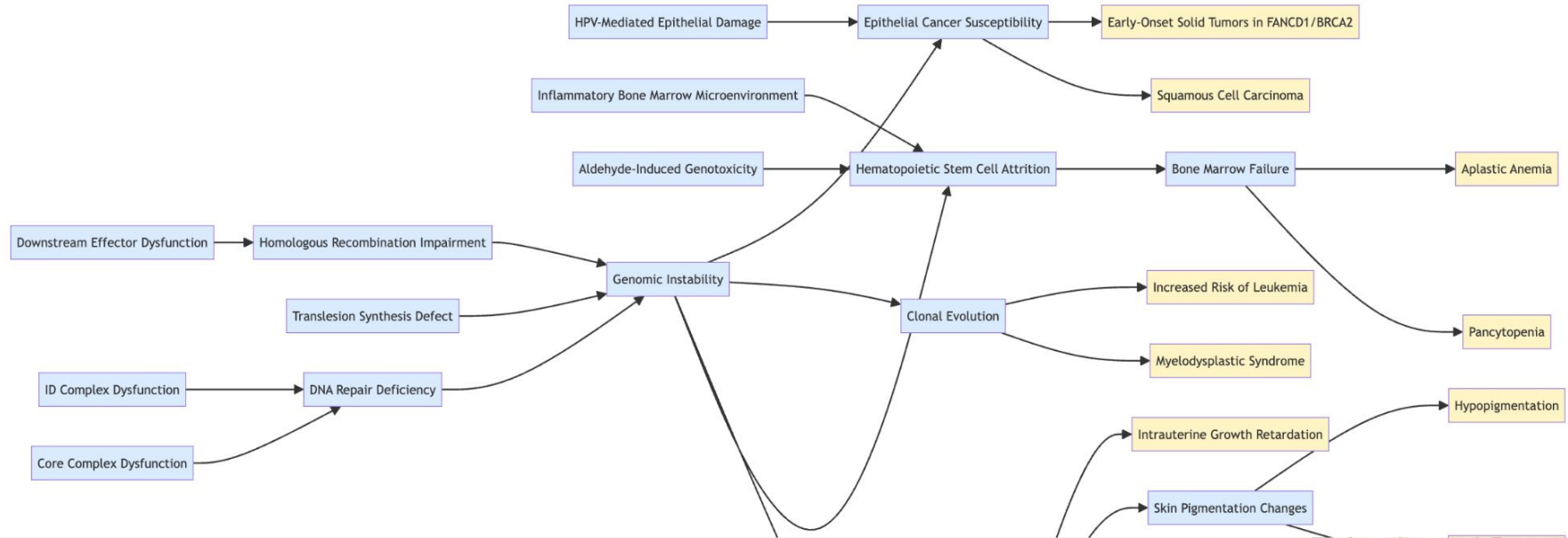
Pancytopenia VERY_FREQUENT **Pancytopenia** (HP:0001876)

Decreased levels of red blood cells, white blood cells, and platelets. Over 80% of FA patients develop bone marrow failure. Typically presents in the first decade, often initially with thrombocytopenia or leukopenia. Notably, patients with FANCD1/BRCA2 mutations may not develop BMF. FA-S (biallelic BRCA1) patients also characteristically lack bone marrow failure.

SHOW EVIDENCE (1 REFERENCE)

SHOW EVIDENCE (1 REFERENCE)

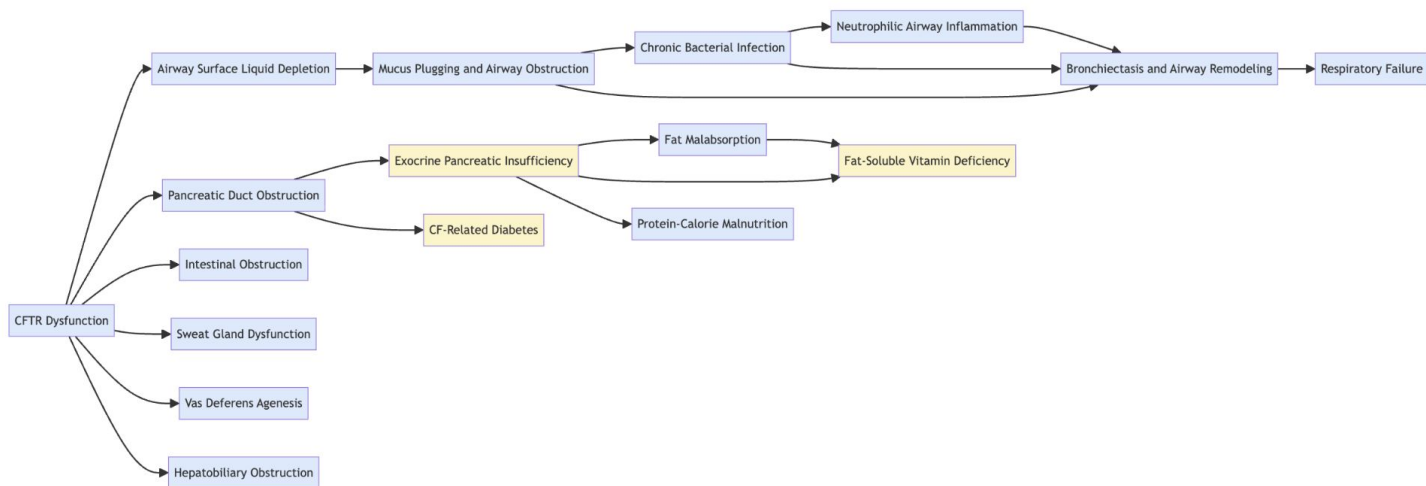
Objective: Mechanistic explanation of every phenotype



Objective: Explain every phenotype



Causal Graph



How do I contribute?

This needs documentation.

Short answer:

- Clone the repo, start Claude Code, run `/curate disease_name_here`
- *Or* use your favorite agentic coding framework and point it at the skills directory.

Add more instructions for new agent managers #190

Open



cmungall opened 2 weeks ago

Member ...

There is a lack of docs on how to get started using an agent to curate a disease.

All workflows are heavily claude code oriented. In fact agent managers should just clone the repo and ask the agent "give me a tour" or rather than old fashioned reading the docs. But there needs to be a bit more bootstrapping.

- README.md - add a link to CONTRIBUTING.md for people who want to contribute by being agent managers
- CONTRIBUTING.md - add a basic preamble about how this repo requires claude code to contribute. Can link to ai4curation/aidocs
- `.claude/commands/curate.md` - this should have a very small amount of instructions for how to have the agent 'manage up' to their manager. E.g. currently it's assumed that `just` is installed. If it's not, the agent typically tries some weird workaround. It should just say "hey, human, I need `just`, want me to install this for you?"
 - it's all a bit open ended which deep research provider to use and for the agent and their human to figure this out. Let's just say falcon (aka edison) is recommended for most users. The agent should instruct the human to go to <https://platform.edisonscientific.com/> and get a key
- some of this could go in CLAUDE.md

<https://github.com/monarch-initiative/dismech/blob/main/.claude/skills/initiate-new-disorder-creation/SKILL.md>

Create sub-issue



<https://github.com/monarch-initiative/dismech/issues/190>

Always give feedback on GitHub - it helps the agent improve

wants to merge 1 commit into `main` from `weekly-compliance-2026-02-11-IgAN`

kb/disorders/IgA_Nephropathy.yaml

```
67 + term:
68 +   id: UBERON:0002320
69 +   label: glomerular mesangium
60 70 biological_processes:
61 71 - preferred_term: Immune Complex Formation
62 72 term:
@@@ -87,6 +97,15 @@ pathophysiology:
87 97 IgA immune complex deposition activates mesangial cells, causing
88 98 proliferation, matrix expansion, and release of inflammatory mediators
89 99 including cytokines and complement components.
100 + locations:
101 + - preferred_term: Glomerular Mesangium
102 + term:
103 +   id: UBERON:0002320
104 +   label: glomerular mesangium
105 + - preferred_term: Renal Glomerulus
```

Comment on line R105



cmungall now

Member ...

not wrong, but redundant with more specific term above



Write a reply

Resolve comment

Finish your review

Write

Preview

H

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📎

↩

Leave a comment

📎 Paste, drop, or click to add files

☐ Comment

Submit general feedback without explicit approval.

☒ Approve

Submit feedback and approve merging these changes.

☐ Request changes

Submit feedback suggesting changes.

Cancel

Submit review 🗑️ ↩

Hint: sometimes Claude reviews will have links to fix

Minor Suggestions for Enhancement

1. Evidence Source Classification - One Potential Issue

Location: kb/disorders/Kummell_Disease.yaml:175-179

The evidence item for PMID:36428981 is marked as `COMPUTATIONAL` :

```
evidence_source: COMPUTATIONAL
snippet: "HIF-1α/RANKL/Notch1 pathway bidirectionally regulates..."
```



Issue: This reference is a **review article** about HIF-1alpha in bone metabolic diseases, not a computational/in silico study. According to CLAUDE.md guidelines, COMPUTATIONAL should be for "in silico modeling, docking, simulations, ML predictions, network/pathway inference without wet-lab confirmation."

Recommendation: This should likely be classified as `OTHER` (for review/synthesis) or removed from evidence if it doesn't directly support the hypoxia mechanism in Kummell disease specifically. The snippet describes HIF-1alpha's general role in bone metabolism but doesn't specifically demonstrate it in KD.

[Fix this →](#)

2. Treatment Term Specificity

Location: kb/disorders/Kummell_Disease.yaml:374-378 and similar

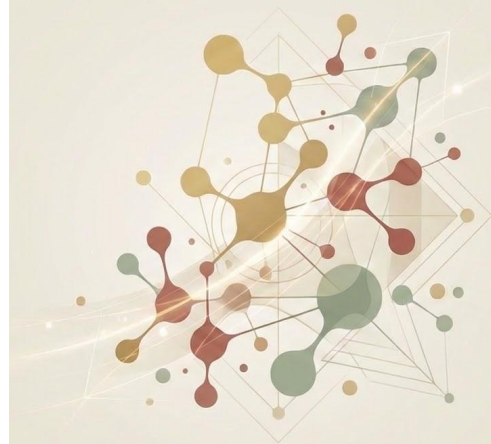
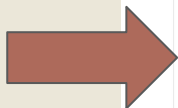
All surgical treatments (PVP, PKP, and spinal fusion) use the generic term MAXO:0000004 (surgical procedure). While correct, there may be more specific MAXO terms available:

- Consider checking for more specific terms for "minimally invasive spine surgery" or "percutaneous vertebral augmentation"
- The generic term is acceptable, but greater specificity would be ideal if available

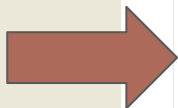
3. Clinical Trial Phase Formatting

Location: kb/disorders/Kummell_Disease.yaml:700

```
phase: PHASE_III
```



Hint: sometimes Claude reviews will have links to fix



Minor Suggestions for Enhancement

1. Evidence Source Classification - One Potential Issue

Location: kb/disorders/Kummell_Disease.yaml:175-179

The evidence item for PMID:36428981 is marked as COMPUTATIONAL :

```
evidence_source: COMPUTATIONAL
snippet: "HIF-1α/RANKL/Notch1 p
```

Issue: This reference is a review article. According to CLAUDE.md guidelines, review articles should be marked as 'OTHER' for predictions, network/pathway inference, etc.

Recommendation: This should likely be marked as 'OTHER' to directly support the hypoxia mechanism in bone metabolism but doesn't specifically demonstrate it in Kummell disease.

[Fix this →](#)

2. Treatment Term Specificity

Location: kb/disorders/Kummell_Disease.yaml:180-181

All surgical treatments (PVP, PKP, and others) are correct, but for more specificity, consider the following:

- Consider checking for more specific terms like "vertebral augmentation"
- The generic term is acceptable, but more specific terms are preferred.

3. Clinical Trial Phase Formatting

Location: kb/disorders/Kummell_Disease.yaml:182-183

```
phase: PHASE_III
```

Claude Code

Research preview

In kb/disorders/Kummell_Disease.yaml at line 177, change evidence_source from COMPUTATIONAL to OTHER for PMID:36428981, since this is a review article about HIF-1alpha in bone metabolism, not a computational/in silico study. Or consider whether this evidence item should remain, as it describes the general role of HIF-1alpha in bone diseases but doesn't specifically demonstrate it in Kummell disease.

☒ </> Code

Opus 4.6



dismech main

Default

Search

⌘K



Links

- Dismech:
 - GitHub - <https://github.com/monarch-initiative/dismech>
 - Main site - <https://monarch-initiative.github.io/dismech/>
 - Embedding Browser - <https://monarch-initiative.github.io/dismech/app/embeddings/index.html>
 - QC Dashboard - <https://monarch-initiative.github.io/dismech/dashboard/index.html>
- LinkML tools:
 - Reference Validator - <https://github.com/linkml/linkml-reference-validator>
 - Term Validator - <https://github.com/linkml/linkml-term-validator>
 - Embeddings Explorer - <https://github.com/linkml/linkml-embeddings-explorer>
 - Browser (faceted interface for your own schema) - <https://github.com/linkml/linkml-browser>
- Deep Research Client - <https://github.com/monarch-initiative/deep-research-client>



Thanks!

Technical support from the Monarch Initiative:

- At LBNL (Chris Mungall, PI)
- At UNC School of Medicine (Melissa Haendel, PI)

