The Disease Ontology values your feedback! Take a few moments to complete our:

<----2025 Annual DO-KB User Survey

We want to know what disease information matters most to you. Your input will help shape our data, tools, and future updates.

DO Buzz Newsletter

Spring 2025

CONFERENCES

The Disease Ontology Team has recently attended and presented at:

- The 18th Annual International Biocuration Conference at Stowers Institute
  - Workshop, 'AI and Biodata Resources: Implications for Sustainability and Best Practices in Biocuration'
  - Talk: “A Standardized Workflow for Translating Ontology and Website Content”,
  - Poster Presentation: “Disease Ontology Expansion: Honoring Contributors, Updating Data & Resources, and Strengthening FAIR Data”
- Rare Disease day poster
  - Check out our poster, prepared for the FDA-NIH Rare Disease Day 2025, to explore how the Disease Ontology supports rare diseases data accessibility.
- 9th Disease Maps Community Meeting in Leuven, Belgium.
  - Talk: “DO-KB: a semantic approach driving mechanistic-based modeling to advance disease knowledge”

Biocuration 2025
Disease Ontology team

DATA UPDATES

- **Spanish Website and Disease Data Now Available**
  - The Disease Ontology is developing a standardized workflow for translating ontology and website content. The website and data trees are now accessible in Spanish. You can switch between English and Español using the language tab. All disease names, definitions, and synonyms have been carefully translated to ensure accurate multilingual access. Learn more about this process from our most recent talk. Disease Ontology Español
- **New Disease Ontology API Launched**
  - We have launched a powerful new RESTful API, built on SwaggerUI, that allows users to explore and interact with the DO without needing to write code. The API supports advanced and faceted searches, term lookups, and programmatic access to our full disease dataset.
- **Ongoing Curation & 2025 Data Releases**
  - The DO continues to expand with new disease terms, updated definitions, cross-references, and logical axioms. In 2025, major updates include: language-tagged content (en, es), Spanish DO files (doid-es.owl, doid-international.owl), acronym annotations, transition from OMIM to MIM prefixes, and targeted curation in areas such as phenotypic series, glycosylation disorders, viral diseases, and childhood cancers (DO_childhood_cancer_slim).

Human Disease Ontology at a Glance

Disease classes: 11,870

Definitions: 9,554

Logical axioms: 8,863 subClassOf; 726 equivalentClass

Imports: 22 sources; 4,767 classes

Cross-references: 23 sources; 38,372 xrefs

COMMUNITY & INITIATIVES

- 🎉 **DO Contributor Registry Launched**
  - We are excited to announce the launch of the DO Contributor Registry! This initiative highlights and thanks the many researchers, clinicians, and institutions who contribute to the development and maintenance of the ontology. Your input helps ensure the DO remains a trusted, comprehensive resource for the global research community.
- **Acronym Annotations Added**
  - To improve data discovery and interoperability, acronyms are now formally annotated as synonyms in the DO. This enhancement helps users find terms more easily and ensures broader recognition of commonly used disease abbreviations.
- **Nosology Program Launch**
  - The Disease Ontology Knowledgebase has launched a new DO based Nosology Program at the University of Maryland School of Medicine. This initiative uses the Disease Ontology to explore the complexities of disease classification and serves as a valuable tool for both research and biomedical education.
- 📣 **Follow us on Bluesky!** Bluesky
- We’ve launched an official presence on the Bluesky social platform! Follow us for updates, behind the scenes insights, and ways to engage with the Disease Ontology community.

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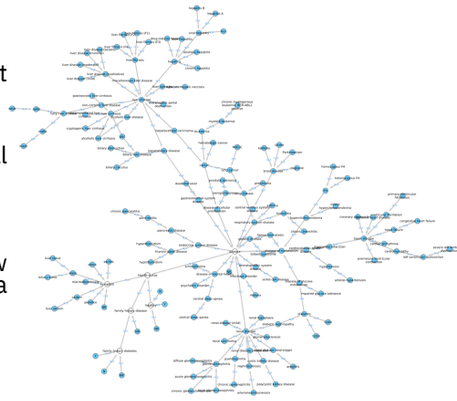
Community Resources Using the Disease Ontology

Collabortion Spotlight: **PK-DB Integrates the Disease Ontology**



PK-DB (Pharmacokinetics Database) is an open, FAIR-compliant platform for managing and analyzing pharmacokinetic data to support reproducibility and drug development.

We are excited to spotlight PK-DB's integration of the Disease Ontology. PK-DB has successfully mapped over 90% of their internal disease terms to DOID, greatly enhancing the structure, consistency, and clarity of their disease annotations.

The Disease Ontology plays a key role in providing structuring terms that helped PK-DB connect related disease concepts and clearly distinguish between diseases and symptoms. This structured mapping is now reflected in their disease overview (health_status_disease.png) and embedded in their comprehensive knowledge graph (all_ontologies.png), where DOID serves as a central component of their metadata framework.

We are thrilled to see this impactful use of DO and look forward to continued collaboration with the PK-DB team!

Learn more about PK-DB and Matthias' work at: 🌐 Website: livermetabolism.com 🔗 ORCID: 0000-0003-1725-179X
💼 LinkedIn: linkedin.com/in/matthias-koenig 🐦 X (Twitter): @konigmatt

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A thorough list of Ontologies, Resources and Methodologies that use the DO is available at https://disease-ontology.org/community/use-cases.

Latest Release Notes

Data releases are available in DO's GitHub repository

DO April 2025 Release (v2025-04-29)

This release of the Human Disease Ontology includes 11,870 disease classes, 9,554 with textual definitions (80.5%). Terms with Spanish translation have increased to include 10,447 labels (88%), 11,714 synonyms (60.8%), and 1,280 definitions (13.4%).

Diseases revised and updated include viral infectious diseases, various congenital disease subtypes, citrullinemias, adiaspiromycosis, sickle cell diseases, Christianson syndrome, autosomal dominant intellectual developmental disorder 43, autosomal recessive nonsyndromic deafness 61, combined oxidative phosphorylation deficiency 21, congenital malabsorptive diarrhea 4, progressive multifocal leukoencephalopathy, retinitis pigmentosa 96, and spermatogenic failure 14.

Newly added diseases include subtypes of autosomal recessive limb-girdle muscular dystrophy, autosomal recessive nonsyndromic deafness, immunodeficiency, mucopolysaccharidosis, retinitis pigmentosa, and visceral heterotaxy.

For the latest updates and other information, follow the Disease Ontology on:

- **Twitter:** @diseaseontology
- **Facebook:** @diseaseontology
- **Youtube:** @DiseaseontologyOrgDOID
- **Linkedin:** Disease Ontology LinkedIn
- **BlueSky:** @diseaseontology.bsky.social
- **Slack:**