

DO Buzz Newsletter - December 2024



Upcoming Meetings

18th Annual International Biocuration Conference
April 5-9, 2025

Curating the Clinical Genome
June 11 - 13, 2025

ISMB 2025
July 20-24, 2025

CGC Annual Meeting
August 3-6, 2025



Learn about the Disease Ontology Knowledgebase (DO-KB) from our [newest Video Tutorial](#)

- Learn about the Disease Ontology as we walk through a breakdown of our DO-KB Faceted Search, DO-KB SPARQL Sandbox and more!

Highlighted Global Core Biodata Resource!

- We are happy to announce that the Human Disease Ontology Knowledgebase was highlighted as the [Global Core Biodata Resource \(GCBR\) of the Week](#) (August 25th, 2024).



Access the Disease Ontology in Español!

- The Disease Ontology website is now available in Spanish, to expand accessibility of the DO for our global community. To switch to Spanish, simply select the display language in the upper right corner of the [Disease Ontology website](#). We hope this update makes our resources even more valuable and accessible to Spanish-speaking researchers, clinicians, and the broader public!
 - We would love to hear your initial feedback and experiences with the new site. Please [join us](#) in the Disease Ontology's newest [#spanishdiseaseontology](#) channel from the Disease Ontology Slack to share your thoughts and discuss the Spanish website. Looking forward to hearing about your experience!



Featuring the Disease Ontology

- Check out the latest blog featuring the DO, "[Challenges and Insights from a Human Disease Ontology Mapping Project](#)" on the collaboration between SciBite and the Human Disease Ontology Knowledgebase.

Data updates

- The Disease Ontology now annotates the synonyms that are acronyms and changed the cross-reference prefix label from 'OMIM' to 'MIM.'

Join the DO at the ISB 2025 workshop: "AI and Biodata Resources: Implications for Sustainability and Best Practices in Biocuration".

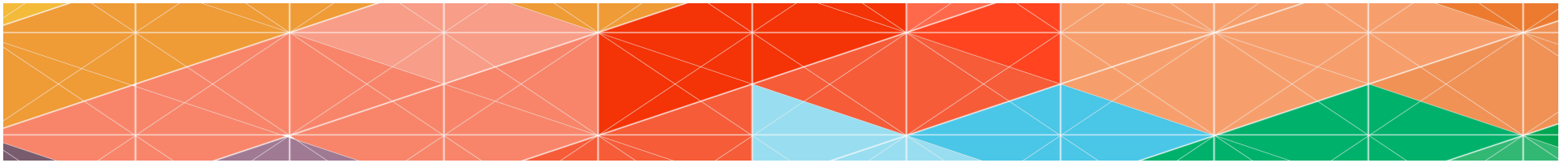
- We look forward to your participation at the 18th Annual International Biocuration Conference! Available in person or remotely, learn more and register [here](#).

The Disease Ontology's recently participated in:

- Pediatric Cancer Diagnosis Ontology Task Force meeting at NIH.
- International Semantic Web Conference and Ontology alignment workshop.

Human Disease Ontology at a Glance

Disease classes	Definitions	Logical axioms	Imports	Cross-references
11,731	9,410	8,664 subClassOf/ 729 equivalentClass	19 sources/ 4,620 classes	21 sources/ 31,212 xrefs



Citations & New Community Resources Using the Disease Ontology

To date, 2,106 works citing the Disease Ontology. For example:

- ◇ TarKG: a comprehensive biomedical knowledge graph for target discovery. doi: [10.1093/bioinformatics/btae598](https://doi.org/10.1093/bioinformatics/btae598)/ PMID: [39392404](https://pubmed.ncbi.nlm.nih.gov/39392404/)
- ◇ CBGDA: a manually curated resource for gene-disease associations based on genome-wide CRISPR. doi: [10.1093/database/baae077](https://doi.org/10.1093/database/baae077), PMID: 39213392
- ◇ The Alzheimer's Knowledge Base: A Knowledge Graph for Alzheimer Disease Research doi: [10.2196/46777](https://doi.org/10.2196/46777) PMID: [38635981](https://pubmed.ncbi.nlm.nih.gov/38635981/)

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

Disclaimer: Article spotlights and community resource lists highlight utilization of the Human Disease Ontology and are not an endorsement of any person(s), resource(s), method(s), or finding(s).

Latest Release Notes

Data releases are available in DO's [GitHub repository](#) ([previous release notes](#)).

Release #147: v2024-12-18 - December

This release of the Human Disease Ontology includes 11,731 disease classes, 9,410 with textual definitions (80.2%).

Classification of CAMRQ3/SCAR34, appendicitis, and childhood leukemias have been revised. Bryant-Li-Bhoj neurodevelopmental syndromes and nonphotosensitive trichothiodystrophies have been added, as have definitions for hydronephrosis and ureteral obstruction. Subtypes have been added, along with revision to existing diseases, for congenital hemolytic anemias, pseudohypoparathyroidisms, combined pituitary hormone deficiencies, and Wieacker-Wolff syndromes (female-restricted added).

Release #146: v2024-11-27 - November

This release of the Human Disease Ontology includes 11,708 disease classes, 9,383 with textual definitions (80.1%).

DO on Social Media



Check out the **Latest Videos** on the Disease Ontology's [YouTube Channel!](#)



Biomedical Ontology
World

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