

DO July 2023 Buzz Newsletter



Celebrating our 20th anniversary!

We are thrilled to celebrate the Disease Ontology's 20th anniversary, marking two decades of advancing disease knowledge, fostering collaboration, and serving as a trusted resource. Established at Northwestern University in 2003 by Rex Chisholm and Warren Kibbe, and joined by Lynn Schriml (TIGR) in 2005, the Disease Ontology moved to the University of Maryland School of Medicine in 2007. With over 11,349 disease terms, our comprehensive and evolving ontology has facilitated research, clinical decision-making and data interoperability. We are grateful for your support and look forward to continuing our commitment to providing valuable resources, driving progress in understanding diseases, and improving human health for many more years to come!



Disease Ontology's launches and more!

- ◇ Introducing the new [Disease Ontology website](#) with [Disease Ontology Knowledgebase \(DO-KB\)](#) and enhanced features!
 - [DO-KB's SPARQL Sandbox](#): a dynamic data playground for exploring the Human Disease Ontology with federated SPARQL queries across connected resources, embark on a journey of groundbreaking disease knowledge discovery.
 - DO-KB's SPARQL endpoint: Uncover the potential of disease-related queries with federated searches that bridge diverse resources. (<https://sparql.disease-ontology.org>)
 - [DO-KB's Faceted Search Interface](#): Dive into a fascinating world of human disease features and mechanisms. Discover a fresh approach that unveils data facets, enabling exploration of disease connectivity. Mine disease-data connections through faceted query and retrieval of DO diseases sharing features or mechanisms, including phenotypes, environmental or genetic drivers, anatomy, variant type, and age of onset.
- ◇ [Disease Ontology Global Impact](#)
 - Launched statistics showcasing the breakdown of DO's usage worldwide including the utilization of 362 biomedical resources and the influx of 229,769 new users from various sub-continent identified from 2008 to mid 2023.
- ◇ New Biocuration Video Series
 - Starting with the captivating inaugural episode titled "[What is Biocuration?](#)"
- ◇ Introducing the inaugural public DO Slack channel
 - Offering a dynamic platform for engaging discussions, fruitful collaborations, and valuable knowledge sharing among our community.
- ◇ Conferences:
 - The Disease Ontology team had a fruitful presence at two significant conferences: the 16th Annual ISB Biocuration conference in Padova, Italy and the Practical Biocuration event at the European Bioinformatics Institute in the UK, where they actively contributed to advancing the field of biocuration. Learn more from our [Youtube videos](#) (ML Biocuration Intro/panel 1/panel 2)

Human Disease Ontology at a glance:

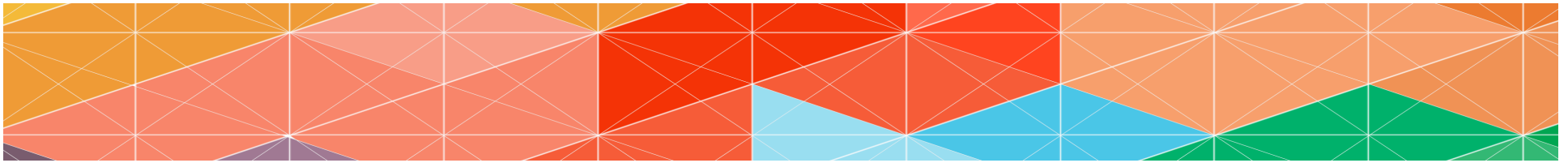
Disease Classes	11,349
Definitions	8,977 (79.1 %)
Logical Axioms	8,223 subClassOf/ 724 equivalentClass
Imports	15 sources / 4,442 classes
Cross-references	14 sources / 37,497 xrefs

Upcoming Meetings

[ISMB Bio-ontology and BOSC 2023](#)
In-person
July 23-27, 2023

[ICBO 2023](#)
Aug 28-Sept 1st, 2023
Brasilia, DF Brazil





Citations & New Community Resources Using the Disease Ontology

To date, 1,818 works citing the Disease Ontology are listed at lens.org. For example:

- ◇ Identification of diagnostic hub genes related to neutrophils and infiltrating immune cell alterations in idiopathic pulmonary fibrosis. doi: [10.3389/fimmu.2023.1078055](https://doi.org/10.3389/fimmu.2023.1078055), PMID: [37334348](https://pubmed.ncbi.nlm.nih.gov/37334348/).
 - DO enrichment analysis to explore the diseases favored by the DEGs (differentially expressed genes)
- ◇ Constructing ferroptosis-related competing endogenous RNA networks and exploring potential biomarkers correlated with immune infiltration cells in asthma using combinative bioinformatics strategy. doi: [10.1186/s12864-023-09400-7](https://doi.org/10.1186/s12864-023-09400-7), PMID: [37259023](https://pubmed.ncbi.nlm.nih.gov/37259023/)
 - Screened differentially expressed lncRNAs (DElncRNAs) between patients and healthy subjects, constructed a competing endogenous RNA (ceRNA) network.
- ◇ MultiDS-MDA: Integrating multiple data sources into heterogeneous network for predicting novel metabolite-drug associations.. doi: [10.1016/j.combiomed.2023.107067](https://doi.org/10.1016/j.combiomed.2023.107067), PMID: [37276756](https://pubmed.ncbi.nlm.nih.gov/37276756/)
 - Computational framework, MultiDS-MDA, for identifying metabolite-drug associations.

A THOROUGH LIST OF ONTOLOGIES, RESOURCES AND METHODOLOGIES THAT USE THE DO IS AVAILABLE AT [HTTPS://DISEASE-ONTOLOGY.ORG/COMMUNITY/USE-CASES](https://disease-ontology.org/community/use-cases).

Latest Release Notes

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

Release #127: v2023-06-29

DO's June 2023 release. Includes 11,349 disease terms, the addition of 35 new diseases, 42 new SubClassOf axioms. This release includes reformatting of gene symbols in definitions, the addition of mtDNA depletion syndrome subtypes 16-20, retinal macular dystrophy and subtypes, sorbitol dehydrogenase deficiency with peripheral neuropathy, hyperphosphatasia with impaired intellectual development syndrome subtypes, revised nemaline classification and COX deficiency classification, new digenic disease annotations.

Release #126: v2023-05-31

This release includes 11,314 disease terms and 8,942 textual definitions (79.0%) with added definitions, xrefs, new DO terms and revised classifications. Included are updates for bradyopsia, NBCCS - nevoid basal cell carcinoma syndrome, leukoencephalopathy with vanishing white matter, developmental and epileptic encephalopathy, hypomyelinating leukodystrophy, epidermolytic hyperkeratosis, autosomal recessive spinocerebellar ataxia, Paget's disease of bone, spastic quadriplegic cerebral palsy, spinal muscular atrophy and various myopathies. Added diseases include acrocardiofacial syndrome, Hengel-Marooftan-Schols syndrome, early onset progressive encephalopathy with brain atrophy and thin corpus callosum, Luo-Schoch-Yamamoto syndrome and Pierpont syndrome, along with many others.

DO on Social Media

- Check out the **Latest Video** on the Disease Ontology's [YouTube Channel](#):  "What is biocuration?"

For the latest updates and other information, follow the Disease Ontology on [Twitter!](#)

- *Are you aware of videos describing use of the DO?* Please let us [know!](#)
Help augment the "[Applications using DO](#)" playlist.

 [@diseaseontology](#)

- *Videos are also available in "The Human Disease Ontology" playlists at:*



[YouTube Channel](#)

or

Biomedical Ontology [YouTube Channel](#)
World

