

DO Buzz Newsletter - March 2024



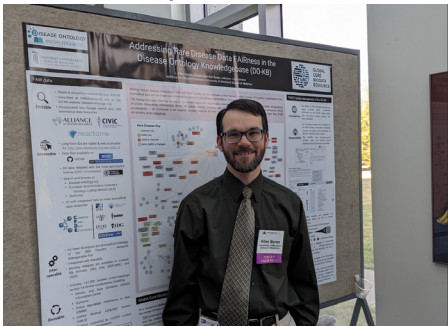
Upcoming Meetings

[Curating the Clinical Genome](#)
2024

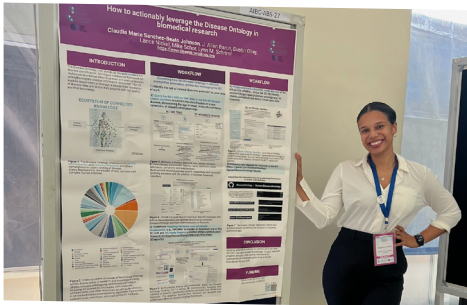
May 9th 2024, May 10th 2024.



Lynn Schriml



J.Allen Baron



Claudia Marie Sanchez-Beato Johnson

Latest News

◇ Learn about our newest publication! 📖🌐

Our latest paper, “**The DO-KB Knowledgebase: a 20-year journey developing the disease open science ecosystem,**” is now officially in press!

- Check it out in the [2024 Database Issue from Nucleic Acids Research](#). PMID: [37953304](#)



◇ We are a **Global Core Biodata Resource!** 🌐📊

We are proud to share that the Disease Ontology Knowledgebase (DO-KB) has been recognized as a [Global Core Biodata](#) Resource by the Global Biodata Coalition! This acknowledgment highlights the fundamental importance of biodata resources to the wider biological and life sciences community, emphasizing our commitment to the long-term preservation of biological data. The Disease Ontology’s long-term dedication to quality service, governance, and global research impact continues to drive our mission forward.



**GLOBAL
CORE
BIODATA
RESOURCE**

◇ **Conferences:** 📅🌐

The DO team attended instrumental conferences showcasing the Disease Ontology’s continued impact across disease and biomedical research.

◇ Rare Disease Day at NIH

The Disease Ontology actively participated in Rare Disease Day at NIH on February 29th, 2024, focusing on rare diseases. J. Allen Baron presentation on “[Addressing Rare Disease Data FAIRness in the Disease Ontology Knowledgebase \(DO-KB\)](#)”.

◇ 17th Annual International Biocuration Conference

- At the 17th Annual International Biocuration Conference from March 5th to 8th, 2024, themed “Enabling the path from Data to Knowledge” in Faridabad, India, our team made significant contributions.
- Lynn Schriml talks on the ‘[Pillars of resource sustainability for the Human Disease Ontology Knowledgebase \(DO-KB\)](#)’ and ‘[Empowering the translation of semantic Disease Ontology \(DO\) data to knowledge in DO-KB,](#)’
- Claudia Marie Sánchez-Beato Johnson presentation on ‘[How to actionably leverage the Disease Ontology in biomedical research](#)’.

◇ The Allied Genetics Conference TAGC 2024

- The Allied Genetics Conference TAGC 2024 held from March 6th to 10th, 2024, in Washington DC.
- J. Allen Baron’s presentation on ‘[Empowering disease knowledge representation with the Disease Ontology Knowledgebase \(DO-KB\)](#)’.

Human Disease Ontology at a Glance

Disease classes	Definitions	Logical axioms	Imports	Cross-references
11,511	9,160	8 , 3 9 3 subClassOf/727 equivalentClass	19 sources/ 4,502 xrefs	21 sources/ 37,842 xrefs



Citations & New Community Resources Using the Disease Ontology

To date, 1,925 works citing the Disease Ontology are listed at lens.org. For example:

- ◇ Predicting gene disease associations with knowledge graph embeddings for diseases with curtailed information. doi: [0.1101/2024.01.11.575314](https://doi.org/0.1101/2024.01.11.575314).
- ◇ The African Human Microbiome Portal: a public web portal of curated metagenomic metadata. doi: [10.1093/bioinformatics/btad490](https://doi.org/10.1093/bioinformatics/btad490), PMID: [38204360](https://pubmed.ncbi.nlm.nih.gov/38204360/)
- ◇ iLncDA-RSN: identification of lncRNA-disease associations based on reliable similarity networks. doi: [10.3389/fgene.2023.1249171](https://doi.org/10.3389/fgene.2023.1249171), PMID: [37614816](https://pubmed.ncbi.nlm.nih.gov/37614816/)

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 #136: v2024-02-28

This release of the Human Disease Ontology includes 11,511 disease classes, 9,160 with textual definitions (79.6%). Diseases that have been revised and/or expanded include CACNA1C-related diseases, long QT syndromes, LAL-D, ARVC, autosomal recessive distal hereditary motor neuropathy 8, Pick's disease, hyper IgM syndromes, hypoparathyroidism, parasitic protozoa infectious diseases, and Canavan disease. New diseases include 'syndromic X-linked intellectual developmental disorder bain type', SPATCCM, ACM subtypes, 'neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy', 'developmental delay, dysmorphic facies, and brain anomalies', and 'infantile hypotonia with psychomotor retardation and characteristic facies-3'.

Release #135:v2024-01-31

This release of the Human Disease Ontology includes 11,501 disease classes, 9,146 with textual definitions (79.5%). Diseases that have been revised and expanded include aniridia, osteosarcomas, distal spinal muscular atrophies, distal hereditary motor neuropathies, peeling skin syndromes, pyridoxine-dependent epilepsies, and Graves disease. New diseases include foveal hypoplasias, developmental dysplasias of the hip, familial focal epilepsies with variable foci, dystonia subtypes, Sifrim-Hitz-Weiss syndrome, cepacia syndrome, round cell sarcoma subtypes, Borrelia miyamotoi disease, PLACK syndrome, familial multiple lipomatosis, and retinal macular dystrophy 2. A number of definitions have additionally been updated

DO on Social Media



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



Biomedical Ontology
World

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