DO Buzz Newsletter
October 2023

![Disease Ontology logo]

Upcoming Meetings

[17th Annual Biocuration Conference](#)
March 5-8, 2024

Global Impact

The global impact of the DO, demonstrates the breakdown of [DO's usage worldwide](#) by subcontinent and is updated quarterly. Assessed with Google Analytics, from 2008 to mid-2023 by sub-continent are shown, with "Unknown" representing users for whom Google Analytics could not identify a geographic location. Total sub-continents = 23; Total countries = 201; Total new users = 229,769.
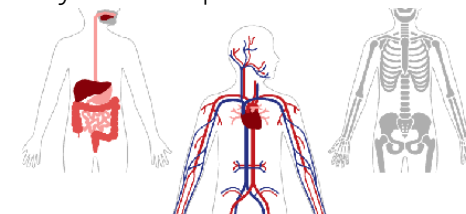
Latest News

◊ Grant announcement:

- Presenting DO's latest collaborative project: "Representing human anatomy for computation and communication: Synergistic development of an anatomical ontology and semantically-augmented anatomical graphics." A NIGMS R01 grant from the University of Kentucky ([Melissa Clarkson, PI](#)).

- The project will create the Foundational Model of Human Anatomy (FMHA) as a derivative of the FMA and will develop a web application for graphically-driven anatomy-based exploration of the Disease Ontology!
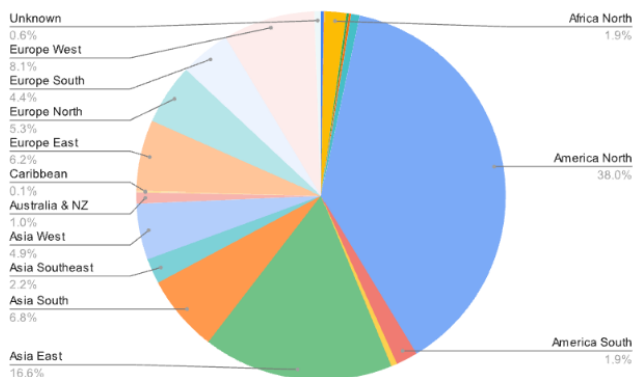
◊ The DO had fruitful conference attendances:

- ISMB/ECCB 2023 conference in Lyon, France with the Presentation "A 20-year journey developing the disease open science ecosystem."

- ICBO-ONTOBRAS Dual Conference 2023 in Brasilia, Brazil on the topic of "The Role of Ontologies in Artificial Intelligence and Machine Learning."
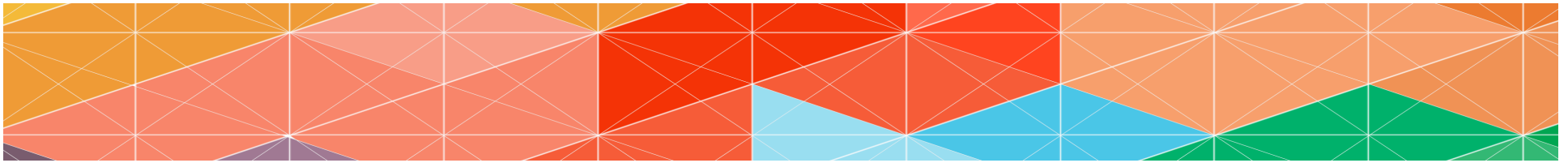
◊ Have you watched our newest youtube videos yet?

- [Machine learning and biocuration](#): Sponsored by [ISB](#)

  - The goals of this workshop are to initiate dialog in order to gain a broader understanding of biocuration and ML needs, to reveal challenges and limitations of developing ML approaches, to foster biocuration support for ML tools, and to facilitate a greater understanding of applications where ML tools are being applied.

- [Panel ML Biocuration:](#)

  - Gaining perspective towards enhancing the intersection of biocuration and machine learning. Panel 2: Biocuration - ML - mapping data between resources.

Human Disease Ontology at a Glance

Disease classes	Definitions	Logical axioms	Imports	Cross-references
11,386	9,022 (79.24%)	8276 subClas-sOf/726 equiva-lentClass	15 sources/ 4,492 xrefs	14 sources/37,574 xrefs

![Pie chart of DO usage by subcontinent: Unknown 0.6%, Europe West 8.1%, Europe South 4.4%, Europe North 5.3%, Europe East 6.2%, Caribbean 0.1%, Australia & NZ 1.0%, Asia West 4.9%, Asia Southeast 2.2%, Asia South 6.8%, Asia East 16.6%, America South 1.9%, America North 38.0%, Africa North 1.9%]

Citations & New Community Resources Using the Disease Ontology

To date, 1,867 works citing the Disease Ontology are listed at lens.org. For example:

◊ BALDR: A Web-based platform for informed comparison and prioritization of biomarker candidates for type 2 diabetes mellitus. doi: 10.1371/journal.pcbi.1011403, PMID: 37590326.

◊ Flame (v2.0): advanced integration and interpretation of functional enrichment results from multiple sources. doi: 10.1093/bioinformatics/btad490, PMID: 37540207

◊ Machine learning and bioinformatics framework integration reveal potential characteristic genes related to immune cell infiltration in preeclampsia. doi: 10.3389/fphys.2023.1078166, PMID: 37389124

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 #132: v2023-10-21
This DO release includes 11,386 diseases, 8,276 SubClassOf and 726 EQ axioms. This release includes the addition of 13 disease terms, 19 definitions, 25 SubClassOf axioms and 1 EQ axiom. New and revised content includes: added synonyms for autosomal recessive spinocerebellar ataxia; the addition of autosomal recessive spinocerebellar ataxia, amyotrophic lateral sclerosis subtypes; revised classifications for schwannomatosis and neurofibromatosis, rhabdomyosarcoma,;the addition of new disease terms: Legius syndrome and Watson syndrome, diphthamide deficiency syndrome and subtypes, renal medullary carcinoma; augmentation of the DO_cancer_slim, updated nomenclature for thyroid cancer and a revised NCIthesaurus slim, produced programmatically based on NCI xrefs or skis mappings.

Release #131 v2023-09-28
This release of the Human Disease Ontology includes 11,373 disease classes, 9,003 with textual definitions (79.2%). Updates include revised mappings based on UMLS 2023AA; the addition of early-onset epilepsy 2 & 3, Yoon-Bellen neurodevelopmental syndrome, NEDDFAC, SCAN3 and updates to other SCAN diseases; revision of viral infectious diseases to incorporate official viral nomenclature changes from ICTV; reactivation of Epstein-Barr virus infectious disease; and various definition updates & corrections.

DO on Social Media

For the latest updates and other information, follow the Disease Ontology on **Twitter**!

🐦 @diseaseontology

• Check out the **Latest Videos** on the Disease Ontology's YouTube Channel!

ISB International Society for Biocuration

Biomedical Ontology World