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 - ❄ Winter 2025 ❄

NEW DO PAPER!

Nucleic Acids Research

🎉 **The latest Disease Ontology publication is now available.**

Read our new paper: "The DO-KB knowledgebase 2026 update: expanding programmatic and language access"

CONFERENCES 🌎

- The **16th International Conference on Biological and Biomedical Ontology** (ICBO 2025)
  - The Disease Ontology Team has attended the virtual ICBO 2025, involving discussions on Biological and Biomedical Ontologies in Action for Health, Science, and Sustainability

- Upcoming
  - **Rare Disease day 2026**
    - February 18th, 2025 - National Institute of Health, NIH.
  - **19th Annual International Biocuration Conference**
    - April 20-24th - in Cape Town, South Africa

DATA UPDATES

- Learn the behind the scenes methodology of the Spanish translation of the Disease Ontology in our new YouTube video:
  - We walk you through the standardized workflow we developed for translating the DO, disease data, and website content.
  - If you have not yet explored it, 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 were carefully translated to support accurate multilingual access.

- New **representation of susceptibilities**:
  - Previously the Disease Ontology only included contributes to condition relationships from OMIM susceptibilities to disease. To make these more accessible, the reverse relationship from disease to related OMIM susceptibilities are now included using the has major susceptibility factor 'relation ontology term'.

- New **SPARQL Queries** added:
  - *disease-omim-susc.rq (DOq059)*
    - shows the new relationships between diseases and OMIM susceptibilities (these are the reverse of the relationships from OMIM susceptibilities to diseases).
  - *disease-multifactor-w-gen-susc.rq (DOq060)*
  - *disease-multifactor-w-genetic.rq (DOq061)*
    - show diseases that have some non-genetic disease driver AND either an OMIM susceptibility (#4), or one or more OMIM susceptibility relationships OR MIM cross-reference (as a stand in for a genetic cause/contributor.

Did you know?

To date, the DO has been integrated into more than 450 resources, developed in 48 countries!

November 2025 Release

Disease classes: 11,997

Definitions: 9,713

Logical axioms: 9,272 subClassOf; 727 equivalentClass

Imports: 22 sources; 4,816 classes

Cross-references: 26 sources; 38,567 xrefs

GLOBAL BIODATA COALITION

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DISEASE ONTOLOGY KNOWLEDGEBASE

Community Resources Using the Disease Ontology

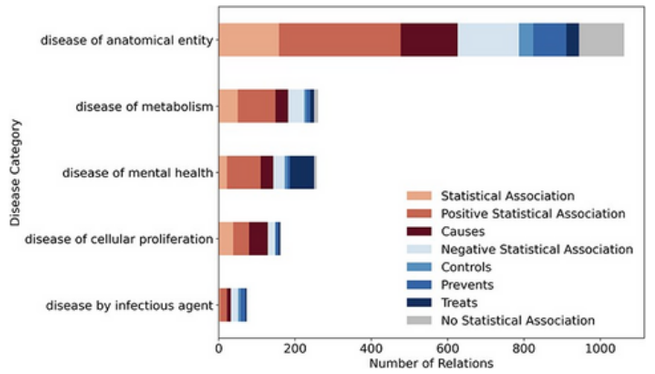
Spotlight: LSSD600 : The first corpus of biomedical abstracts annotated with lifestyle-disease relations

Introducing LSD600, the first corpus specifically designed for LSF-disease.

- Comprising 600 abstracts with 1900 relations of eight distinct types between 5027 diseases and 6930 LSF entities

Notably, all disease entities are standardized using the Disease Ontology, ensuring semantic clarity, interoperability and aligment to FAIR principles.

- Publication: https://pubmed.ncbi.nlm.nih.gov/39824652/
- Database: https://zenodo.org/records/13952449



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 November 2025 Release (v2025-11-25)

This release of the Human Disease Ontology includes 11,997 disease classes, 9,713 with textual definitions (81.0%). Translation files now include 11,824 labels (98.6%), 11,483 synonyms (59.1%), and 6,409 definitions (66.0%) in Spanish.

Newly added diseases include arterionephrosclerosis, chronic pancreatitis, left ventricular failure, Majeed syndrome, muscle dysmorphic disorder, stroke, hemorrhagic stroke, ischemic stroke, oropharyngeal squamous cell carcinoma, paranasal sinus squamous cell carcinoma, poorly differentiated thyroid carcinoma, soft tissue sarcoma, and subcutaneous panniculitis-like T-cell lymphoma.

Disease that have been revised include 'autoimmune interstitial lung, joint, and kidney disease', autosomal dominant Alport syndrome 3A, autosomal dominant hyper-IgE syndromes, bronchiectasis, cerebral infarction, coronary artery disease, C9orf72 frontotemporal dementia and/or amyotrophic lateral sclerosis, complex cortical dysplasia with other brain malformations 14B, COVID-19, Duane retraction syndrome, early-onset epilepsies, Ehlers-Danlos syndrome periodontal types, hereditary fallopian tube carcinoma, hereditary ovarian carcinoma, hyper IgE recurrent infection syndrome 2, immunodeficiency with hyper-IgM type 4, Li-Fraumeni syndrome 2, Lodder-Merla syndrome type 1 with impaired intellectual development and cardiac arrhythmia, mitochondrial DNA depletion syndromes, pontocerebellar hypoplasia type 2A, metal allergy, chloramine T respiratory allergy, disodium cromoglycate allergy, potassium dichromate allergic contact dermatitis, remazole black respiratory allergy, pulmonary eosinophilia, eosinophilic pneumonia, hypereosinophilic syndrome, bone sarcoma, squamous cell carcinoma, head and neck squamous cell carcinoma, nasal cavity squamous cell carcinoma, oropharynx cancer, paranasal sinus cancer, cerebrovascular disease, and ureteral orifice cancer.

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:**