DISEASE ONTOLOGY KNOWLEDGEBASE

https://disease-ontology.org/do-kb/

The DO-KB is expanding the disease information ecosystem
- **expanding the representation of the DO diseaseome**
- **a comprehensive network of disease to disease relationships**
- **a disease feature similarity network for differential diagnosis exploration**

DO-KB SPARQL Sandbox

- A data playground for querying the Human Disease Ontology SPARQL endpoint and to perform federated SPARQL queries across connected resources for disease knowledge discovery.

- A linked data representation (RDF triple store) of the Human Disease Ontology's diseases, disease features and mechanisms.

DO-KB Faceted Search Interface

- A new way to explore human disease features and mechanisms, represented here as data facets.

- The DO-KB Faceted Search Interface enables exploration of connectivity across diseases.

The DO-KB's SPARQL endpoint, explore DO data and query disease, genomic, proteomic resources through federated queries.

Mine the DO-KB 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.

DO-KB SPARQL Sandbox

Select one of the provided SPARQL queries or paste in a novel query to retrieve bespoke ML-ready datasets. Searches may be performed against the DO's primary release file, doid.owl or the doid-merged.owl file for exploring annotations, e.g. ECO codes, and axiom relationships.

- The DO's SPARQL endpoint (**https://sparql.disease-ontology.org**) is available for building federated queries.
- Visit our SPARQL Resources page to learn more about using the SPARQL Sandbox and endpoint.
- Our SPARQL Sandbox and endpoint are currently in **beta**. If you discover any issues, please reach out and let us know!

Query

```
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX obo: <http://purl.obolibrary.org/obo/>
PREFIX oboInOwl <http://www.geneontology.org/formats/oboInOwl#>

SELECT ?class ?id ?label
WHERE {
    ?class a owl:Class ;
        oboInOwl:id ?id ;
        rdfs:label ?label ;
        rdfs:subClassOf* obo:DOID_225 .
}
```

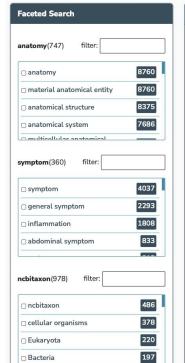Submit Query   Download Results ⬇

Choose a Query:

1. All diseases with their parents (identified by label)
2. Count of diseases in each branch (diseases may be in multiple branches)
3. Syndromes (branch search; returns IRI, ID and label)
4. Diseases with MeSH cross-references
5. Diseases with OMIM cross-references
6. Diseases with PubMed sources (limited to first 10 results)
7. Diseases in DO_infectious_disease_slim with their parents and branches
8. Diseases with their definitions and count of logical axioms
9. Diseases with their Evidence and Conclusion Ontology (ECO) reference types (queries the doid-merged.owl graph)
10. Count of classes imported from other ontologies (queries the doid-merged.owl graph)
11. All diseases with equivalent class axioms (axioms are formatted for readability and may not be displayed in full; queries the doid-merged.owl graph)
12. Extract the disease and supporting import class hierarchies using subClassOf relationships (executes a CONSTRUCT query that returns RDF triples)
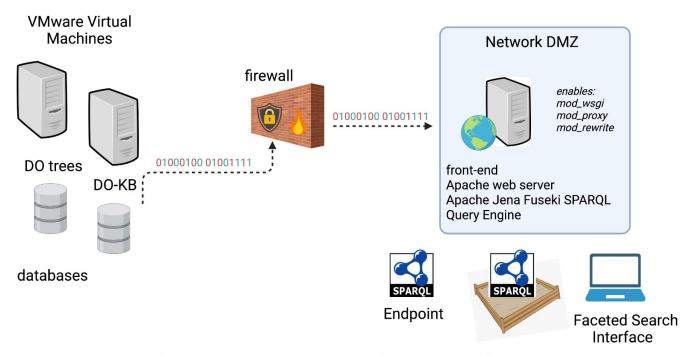
Request New SPARQL Queries

DO-KB Faceted Search Interface

Identify diseases with similar characteristics and explore relationships between diseases through common characteristics or features.

Faceted Search

anatomy(747)   filter: [____]

☐ anatomy	8760
☐ material anatomical entity	8760
☐ anatomical structure	8375
☐ anatomical system	7686
☐ multicellular anatomical	

symptom(360)   filter: [____]

☐ symptom	4037
☐ general symptom	2293
☐ inflammation	1808
☐ abdominal symptom	833

ncbitaxon(978)   filter: [____]

☐ ncbitaxon	486
☐ cellular organisms	378
☐ Eukaryota	220
☐ Bacteria	197

Search Results

Show [50 ▼] entries

DOID	Name
DOID_0001816	angiosarcoma
DOID_0002116	pterygium
DOID_0014667	disease of metabolism
DOID_0040001	shrimp allergy
DOID_0040002	aspirin allergy
DOID_0040003	benzylpenicillin allergy
DOID_0040004	amoxicillin allergy
DOID_0040005	ceftriaxone allergy
DOID_0040006	carbamazepine allergy
DOID_0040007	abacavir allergy
DOID_0040008	isoniazide allergy
DOID_0040009	lidocaine allergy
DOID_0040010	mepivacaine allergy
DOID_0040011	phenobarbital allergy
DOID_0040012	phenytoin allergy
DOID_0040013	ranitidine allergy
DOID_0040014	corticosteroid allergy
DOID_0040015	sulfonamide allergy
DOID_0040016	sulfamethoxazole allergy
DOID_0040017	suprofen allergy
DOID_0040018	thiopental allergy
DOID_0040019	D-mannitol allergy
DOID_0040020	cefotaxime allergy
DOID_0040021	cephalosporin allergy
DOID_0040022	amodiaquine allergy
DOID_0040023	cefaclor allergy
DOID_0040024	ceftazidime allergy

VMware Virtual Machines

DO trees

DO-KB

databases

firewall

01000100 01001111

01000100 01001111

Network DMZ

enables:
mod_wsgi
mod_proxy
mod_rewrite

front-end
Apache web server
Apache Jena Fuseki SPARQL
Query Engine

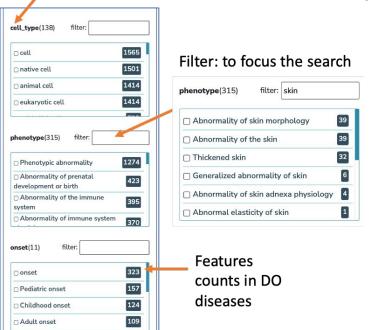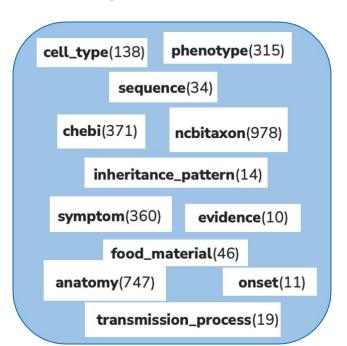SPARQL
Endpoint

SPARQL

Faceted Search
Interface

Augmenting DO technology to launch DO-KB. DO servers are VMware Virtual Machines running **Ubuntu 22.04**. DO server (Python-Django powered web application) and DO-KB serer driven by a Python-Flask framework. **VMware vSphere** client to monitor the health of our front-end server. **Elasticsearch** back-end service for the Faceted Search. DO-KB "**OWL Flattener**" program using Java and the Java OWL API package to convert the hierarchical DO OWL file into a flat list of documents

DO-KB Faceted Search Interface

- A **Facet:** a features or characteristic of a disease, defined in the DO via **ontology imports**
- Disease to Disease connections are defined by these features
- Facet boxes: (DO imports)
  - Select one or more features to retrieve DO diseases sharing those features or mechanisms
    - As facets are selected, the counts refresh, to show additional related features

Filter: to focus the search

Features counts in DO diseases

Identify diseases with similar characteristics and explore relationships between diseases through common characteristics or features

Faceted Search

phenotype

☑ Abnormality of metabolism/homeostasis `12`

symptom

☑ ascites `12`

(fluid collects in spaces within the abdomen)

anatomy

☑ immune system `12`

Search Results

Download Results ⬇

Show `50` ⬍ entries   Search: [   ]

DOID ▲	Name ⬍
DOID_0040090	autoimmune gastritis
DOID_0060031	autoimmune disease of gastrointestinal tract
DOID_0070358	primary biliary cholangitis 1
DOID_0070359	primary biliary cholangitis 2
DOID_0070360	primary biliary cholangitis 3
DOID_0070361	primary biliary cholangitis 4
DOID_0070362	primary biliary cholangitis 5
DOID_0080742	autoimmune cholangitis
DOID_10608	celiac disease
DOID_12236	primary biliary cholangitis
DOID_2048	autoimmune hepatitis
DOID_8577	ulcerative colitis