

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.

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

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-merqed.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: <a href="http://www.w3.org/2000/01/rdf-schema#">http://www.w3.org/2000/01/rdf-schema#">http://www.w3.org/2000/01/rdf-schema#</a>
PREFIX obo: <a href="http://www.geneontology.org/formats/obolnOwt#">http://www.geneontology.org/formats/obolnOwt#</a>

SELECT ?class ?id ?label
WHERE {
    ?dass a owt:Class;
    obolnOwtid ?id;
    rdfs:label ?label;
    rdfs:subClassOf* obo:DOID_225.
}
```

Submit Query

Download Results 🕹



Choose a Query:

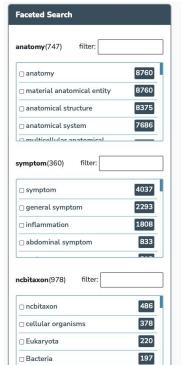
- All diseases with their parents (identified by label)
- 2. Count of diseases in each branch (diseases may be in multiple branches)
- Syndromes (branch search; returns IRI, ID and label)
- 4. Diseases with MeSH cross-references
- 5. Diseases with OMIM cross-references
- Diseases with PubMed sources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

Request New SPARQL Queries

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

DO-KB Faceted Search Interface

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



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	

What is SPARQL?

- SPARQL: SPARQL Protocol And Rdf Query Language
- An RDF (Resource Description Framework) Query Language
- A semantic query language for databases

R D F

What can you do with SPARQL?

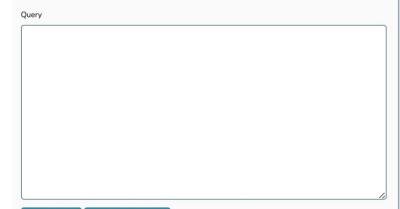
- Enables knowledge to be represented in a machine-readable way
- Used to retrieve and manipulate data stored in RDF format.
- Codifies (stores) relationships between semantic data, in the form of subject-predicate-object 'triples'. E.g., Baltimore is in Maryland
- Use SPARQL to create complex gueries with SELECT statements
- A SPARQL endpoint which allows you to query against the data set

service	endpoint
PRO	https://sparql.proconsortium.org/virtuoso/sparql
UniProt	http://sparql.uniprot.org/sparql
DisGeNet	http://rdf.disgenet.org/sparql/>
MeSH	http://id.nlm.nih.gov/mesh/sparql
Orphanet	https://www.orpha.net/sparql

DO-KB SPARQL Sandbox

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Submit Query

Download Results 🕹

The DO-KB's SPARQL Sandbox provides programmatic access to the DO-KB Knowledgebase for exploring DO diseases, disease features, cross references and mechanisms, and exploring disease-data connections to other SPARQL endpoints.

Choose a Query:

- All diseases with their parents (identified by label)
- 2. Count of diseases in each branch (diseases may be in multiple branches)
- 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)
- Diseases in DO_infectious_disease_slim with their parents and branches
- 8. Diseases with their definitions and count of logical axioms
- Diseases with their Evidence and Conclusion Ontology (ECO) reference types (queries the doid-merged.owl graph)
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- 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

- Select a SPARQL query to retrieve MLready datasets
- SELECT (table formatted results) or CONSTRUCT (RDF results) queries
- Edit queries to refine your search
- Suggest new SPARQL queries to DO-KB
- Utilize the DO-KB SPARQL Endpoint to build your own federated gueries
- Coming Soon:
 - Federated queries to UniProt, MeSH, Protein Ontology

Query execution time: 2.22 second(s).

id label		
DOID:0050120	hemophagocytic lymphohistiocytosis	OMIM:PS267700
DOID:0050156	idiopathic pulmonary fibrosis	OMIM:178500
DOID:0050158	desquamative interstitial pneumonia	OMIM:263000
DOID:0050167	autoimmune polyendocrine syndrome type 1	OMIM:240300

Structuring a SPARQL Query

PREFIX rdfs: http://www.w3.org/2000/01/rdf-schema#>

PREFIX owl: http://www.w3.org/2002/07/owl#>

PREFIX obolnOwl: http://www.geneontology.org/formats/obolnOwl#

SELECT ?id ?label ?parent WHERE {

?class a owl:Class .

?class obolnOwl:id ?id .

?class rdfs:label ?label .

?class rdfs:subClassOf ?parent class .

?parent_class rdfs:label ?parent .

Building a SPARQL Query

Abbreviations used as shortcuts in the query

SELECT : output variables

Conditions that define the query

OWL file, data elements

[ID] obolnOwl:id ?id [label] rdfs:label ?label

[Synonym] obolnOwl:hasExactSynonym obolnOwl:hasOBONamespace

[Slim/Subset] obolnOwl:inSubset

[Alt ID (merges)] obolnOwl:hasAlternativeId