

Tutorial

The Human Disease Ontology (DO) website navigation





Query disease terms in DO's OBO (default view) or OWL trees. Explore related diseases (e.g. shared anatomy, cell of origin, mode of inheritance, sequence structural variant type, symptom).

[Go »](#)[Advanced Search »](#)

Navigation

OBO tree

[View OWL tree](#)

- [-] disease
 - [+] disease by infectious agent
 - [+] disease of anatomical entity
 - [+] disease of cellular proliferation
 - [+] disease of mental health
 - [+] disease of metabolism
 - [+] genetic disease
 - [+] physical disorder
 - [+] syndrome

Welcome

The [Disease Ontology](#) has been developed as a standardized ontology for human disease with the purpose of providing the biomedical community with consistent, reusable and sustainable descriptions of human disease terms, phenotype characteristics and related medical vocabulary disease concepts through collaborative efforts of researchers at Northwestern University, Center for Genetic Medicine and the University of Maryland School of Medicine, Institute for Genome Sciences.

The Disease Ontology semantically integrates disease and medical vocabularies through extensive cross mapping of DO terms to MeSH, ICD, NCI's thesaurus, SNOMED and OMIM.

To get started please visit the [tutorial page](#).

Explore the Human Disease Ontology Project Information & Resources



About

Community

Tools & Resources

Outreach

Curation

Downloads



About

The Disease Ontology Project

DO Team

Scientific Advisory Board

Release Notes on GitHub

How is DO FAIR?

Statistics

DO's Logo

FAQ

Community

Community Calendar

Publications

Collaborators

Newsletter

Outreach

Contact Us

Tutorial

SlideShare

Curation

curation Tools and Documentation
DO SPARQL queries
ROBOT
DO Style Guide
Design Patterns

Downloads
GitHub
OBO Foundry
DO Releases
Imports

Tools & Resources

DO Resources

Related Resources

Citing DO

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[Learn more](#) page.

Basic search: text based search of disease terms



carcinoma



















Go »

Advanced Search »

Navigation

OBO tree

View OWL tree 

-   disease
 -   disease by infectious agent
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 -   disease of mental health
 -   disease of metabolism
 -   genetic disease
 -   physical disorder
 -   syndrome

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OBO Tree

Search doid.obo file

To View Disease Metadata

Select a disease term

or

Term in the OBO Tree

Metadata

View results of query in new tab

Number of disease terms in query result set

carcinoma [Advanced Search »](#)

Navigation View OWL tree

OBO tree


- disease
 - disease by infectious agent
 - disease of anatomical entity
 - disease of cellular proliferation
 - benign neoplasm
 - cancer
 - cell type cancer
 - blastoma
 - carcinoma
 - germ cell cancer
 - malignant adenoma
 - malignant glioma
 - malignant hemangioma
 - malignant mesothelioma
 - melanoma
 - mesenchymal cell neoplasm
 - mixed cell type cancer
 - osteosarcoma
 - sarcoma
 - organ system cancer
 - pre-malignant neoplasm
 - disease of mental health
 - disease of metabolism
 - genetic disease
 - physical disorder
 - syndrome

Displaying results 1 - 19 of 684

ID	Name
DOID:305	carcinoma
DOID:0080199	colorectal carcinoma
DOID:4876	trachea carcinoma
DOID:3459	breast carcinoma
DOID:1520	colon carcinoma
DOID:2871	endometrial carcinoma
DOID:5662	pleomorphic carcinoma
DOID:12342	retroperitoneum carcinoma
DOID:6425	eyelid carcinoma
DOID:4838	myoepithelial carcinoma
DOID:4830	adenosquamous carcinoma
DOID:2762	bone carcinoma
DOID:1791	peritoneal carcinoma
DOID:686	liver carcinoma
DOID:3451	skin carcinoma
DOID:1519	cecum carcinoma
DOID:3737	verrucous carcinoma
DOID:0050922	gastrointestinal carcinoma
DOID:4451	renal carcinoma

[First](#) [« Previous](#) [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [Next »](#) [Last](#)

X – to remove one of the result tabs



carcinoma [Advanced Search »](#)

Navigation

OBO tree View OWL tree

- [-] disease
 - [-] disease by infectious agent
 - [-] disease of anatomical entity
 - [-] disease of cellular proliferation
 - [-] benign neoplasm
 - [-] cancer
 - [-] cell type cancer
 - [-] blastoma
 - [-] carcinoma
 - [-] germ cell cancer
 - [-] malignant adenoma
 - [-] malignant glioma
 - [-] malignant hemangioma
 - [-] malignant mesothelioma
 - [-] melanoma
 - [-] mesenchymal cell neoplasm
 - [-] mixed cell type cancer
 - [-] osteosarcoma
 - [-] sarcoma
 - [-] organ system cancer
 - [-] pre-malignant neoplasm
 - [-] disease of mental health
 - [-] disease of metabolism
 - [-] genetic disease
 - [-] physical disorder
 - [-] syndrome

Welcome Search: Carcinoma Carcinoma

Metadata

ID	DOID:305
Name	carcinoma
Definition	A cell type cancer that has_material_basis_in abnormally proliferating cells derives_from epithelial cells. http://en.wikipedia.org/wiki/Carcinoma
Xrefs	CSP2005:2000-5801 MESH:D002277 MESH:D009375 NCI2004_11_17:C6927 NCI:C2916 NCI:C3709 SNOMEDCT_US_2018_03_01:68453008 SNOMEDCT_US_2018_03_01:71298006 UMLS_CUI:C0007097 UMLS_CUI:C0553707 UMLS_CUI:C1368683
Alternateids	DOID:2428 DOID:6570
Subsets	DO_cancer_slim DO_FlyBase_slim NCIthesaurus
Synonyms	epithelial cancer [EXACT] epithelioma [EXACT] malignant Epithelioma [EXACT]
Parent Relationships	is_a cell type cancer

[Add an item to the term tracker](#)

Disease Term Metadata

DOID - Disease Ontology unique ID

Name - Disease Term name

Definition - Textual definition

Xrefs - Clinical Vocabulary cross references

Alternateids - merged DOIDs

Subsets - DO term included in one of DO Slim files

Synonyms - synonymous disease terms

Parent Relationships

-- direct is_a parent term in DO

Advanced Searches

Match : enables Boolean AND, OR or NOT matching across search fields

Expanded search capabilities with the addition of Boolean Advanced searches

- Enabling a more sophisticated querying across DO definitions, Xrefs, disease names and subsets.
- The Advanced Search functionality allows the user to do one or more search variables, example: intersection of DO records that contain both OMIM and NCI thesaurus Xrefs.



Advanced Search

Name

Match: AND

Definition ❌

Match: AND

Xrefs ❌

Match: AND

Subset ❌

- DO_AGR_slim
- DO_FlyBase_slim
- DO_MGI_slim
- DO_cancer_slim
- DO_rare_slim
- NCIthesaurus
- TopNodes_DOcancerslim

AND
 OR
 NOT

Search specific metadata fields:
Definition, Name, Synonym, Subset, DOID, Alternate ID, Definition and Xref.

DO OWL Tree

Search doid.owl file

To View Disease Metadata

Submit a search term from any of the import files or the DO

Identify all diseases annotated with X-linked inheritance

Or

Identify all diseases 'located_in' the lung

Click on the + to expand or collapse a branch

Double click on the + to expand all of the sibling terms



X-linked inheritance | [Go »](#) | [Advanced Search »](#)

Navigation

OWL tree | [View OBO tree](#)

- doid.owl
 - anatomy
 - cell
 - chebi
 - disease
 - food material
 - inheritance pattern
 - mitochondrial inheritance
 - monogenic inheritance
 - allosomal inheritance
 - X-linked inheritance**
 - Y-linked inheritance
 - autosomal inheritance
 - multifactorial inheritance
 - ncbitaxon
 - omim_susceptibility
 - phenotype
 - sequence
 - symptom
 - transmission process

Advanced Search | Advanced Search | Advanced Search | Search: Epithe | **Search: X-lin**

Displaying results 1 - 19 of 22

ID	Name
GENO:0000146	X-linked dominant inheritance
GENO:0000149	X-linked recessive inheritance
GENO:0000936	X-linked inheritance
GENO:0000939	co-dominant X-linked inheritance
GENO:0000938	incomplete X-linked dominant inheritance
GENO:0000937	complete X-linked dominant inheritance
GENO:0000941	Y-linked inheritance
DOID:0050566	X-linked nonsyndromic deafness
DOID:0050953	X-linked hereditary ataxia
DOID:0080009	X-linked dominant disease
DOID:0080012	X-linked recessive disease
DOID:0111225	centronuclear myopathy X-linked
DOID:0050735	X-linked monogenic disease
DOID:0090057	X-linked dystonia-parkinsonism
DOID:0060309	syndromic X-linked intellectual disability
DOID:0070189	X-linked spermatogenic failure 1
DOID:0070195	X-linked chronic granulomatous disease
DOID:0070159	hereditary sensory neuropathy X-linked
DOID:0070185	X-linked spermatogenic failure 2

DO OWL Tree

The DO OWL tree includes both direct is_a and inferred disease parent classes.

Parent Classes are defined by Equivalent To Logical Axioms

Child Classes are defined by SubClass Of Logical Axioms

The screenshot shows the Disease Ontology (DO) website. At the top, the logo reads "DISEASE ONTOLOGY". A search bar contains the text "lung" with a "Go »" button and a link to "Advanced Search »". Below the search bar, there are two tabs: "Welcome" and "Search: Lung". The "Lung Disease" tab is active.

On the left side, there is a "Navigation" panel with a tree view of the ontology. The "OWL tree" is selected, and the "lung disease" class is highlighted. An orange circle highlights the "Navigation" header, and an orange arrow points from the text "Child Classes are defined by SubClass Of Logical Axioms" to the "lung disease" class in the tree.

The right side of the page shows the metadata for the "lung disease" class. The metadata is organized into sections:

- Metadata:** ID: `DOID:850`, Name: `lung disease`. Buttons for "Submit Comment" and "Visualize" are present.
- Definition:** A lower respiratory tract disease in which the function of the lungs is adversely affected by narrowing or blockage of the airways resulting in poor air flow, a loss of elasticity in the lungs that produces a decrease in the total volume of air that the lungs are able to hold, and clotting, scarring, or inflammation of the blood vessels that affect the ability of the lungs to take up oxygen and to release carbon dioxide. Links: <http://www.nlm.nih.gov/medlineplus/ency/article/000066.htm>, <http://www.niehs.nih.gov/health/topics/conditions/lung-disease/index.cfm>.
- Xrefs:** UMLS_CUI:C0024115, MESH:D008171, SNOMEDCT_US_2018_03_01:19829001, ICD10CM:J98.4, NCI:C3198.
- Alternateids:** `DOID:11895`, `DOID:29`, `DOID:11894`, `DOID:766`.
- Subsets:** NCIthesaurus.
- Parent Relationships:** is_a [lower respiratory tract disease](#).
- Equivalent Logical Relationships:** [disease](#) and located in some [lung](#).

At the bottom right, there is a link: "Add an item to the term tracker".

DO OWL Tree

The OWL tree shows all disease that share a common parent.

Example:
pleural disease

(common parent, pleural disease defined by Logical Axiom to UBERON term: pleura)

Child Classes are defined by SubClass Of Logical Axioms

DISEASE ONTOLOGY

pleura [Advanced Search »](#)

Navigation OWL tree View OBO tree

- doid.owl
 - disease
 - disease of anatomical entity
 - respiratory system disease
 - lower respiratory tract disease
 - bronchial disease
 - lung disease
 - pleural disease
 - benign pleural mesothelioma
 - congenital chylothorax
 - malignant pleural mesothelioma
 - neurilemmoma of the pleura
 - pleural empyema
 - pleural lipoma**
 - pleural tuberculosis
 - pleurisy
 - pneumothorax
 - pulmonary blastoma
 - respiratory system cancer
 - disease of cellular proliferation
 - cancer
 - organ system cancer
 - respiratory system cancer
 - lung cancer

Welcome | Search: Lung | **Pleural Lipoma** | Search: Pleura

Metadata	
ID	DOID:10195
Name	pleural lipoma
Definition	A respiratory system benign neoplasm that derives_from fat cells and is located_in the pleura. https://www.ncbi.nlm.nih.gov/pubmed/29222220
Xrefs	NCI2004_11_17:C6644 UMLS_CUI:C1335434 NCI:C6644
Subsets	NCIthesaurus
Synonyms	Lipoma of Pleura [EXACT]
Parent Relationships	is_a lipoma is_a respiratory system benign neoplasm is_a pleural disease
Subclass Logical Relationships	located in some pleura derives_from some fat cell

[Add an item to the term tracker](#)